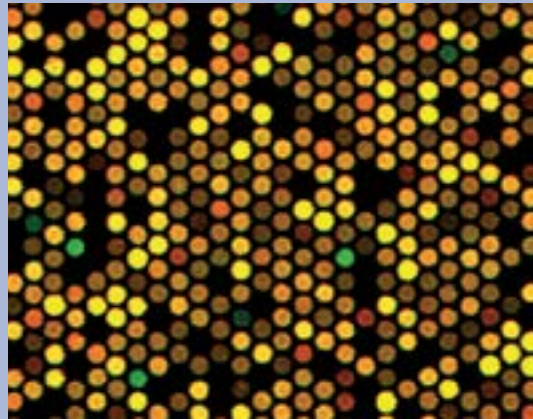
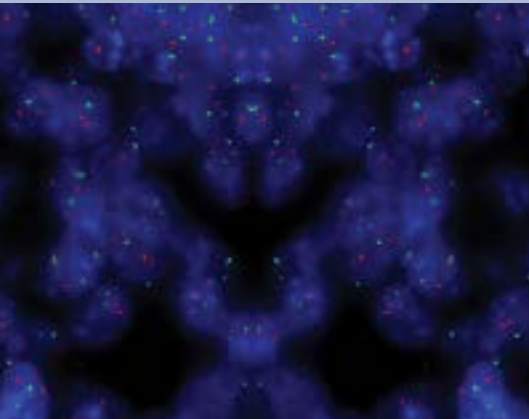




A Sysmex Group Company




Cytocell

CytoSure[™] 

SureSeq[™] 

Oxford Gene Technology

Product Catalog

USA & Canada
SECOND EDITION

- FISH
- Next Generation Sequencing (NGS)
- Arrays

Oxford Gene Technology (OGT) offers high-quality integrated technologies to detect a complete range of genetic aberrations. Learn more in each of these catalog sections. To access additional product information and technical resources, visit www.ogt.com.

CytoCell® FISH Probes for Constitutional Cytogenetics and Hematology/Pathology Cytogenetics

Comprehensive range of high quality, directly-labeled DNA probes for fluorescence *in situ* hybridization (FISH). Includes a wide range of accessories and custom FISH probes. Go to page 4.



SureSeq™ Next Generation Sequencing (NGS) Products for Hematology and Solid Tumor Cancer Research

An expanding portfolio of NGS panels for cancer research, including myPanel™ custom panel content, library preparation products for the accurate detection of genetic variants, optimized for use with Interpret NGS analysis software, a complimentary, powerful and easy-to-use analysis solution. Go to page 141.



CytoSure™ Next Generation Sequencing (NGS) and Array Products for Cytogenetics and Rare Disease Research

A broad range of NGS and array products for constitutional cytogenetics, rare disease and cancer research, including CytoSure Constitutional NGS, a transformative next-generation sequencing panel for intellectual disability (ID) and developmental delay (DD) research. Go to page 169.





A Sysmex Group Company

Oxford Gene Technology - A Sysmex Group Company

Welcome to the new edition of the OGT product catalog. This expansive volume presents our full portfolio of fluorescence *in situ* hybridization (FISH) probes, next generation sequencing (NGS) products and arrays.

We're proud to partner with leading researchers by providing tools that contribute to improving the future of clinical care. Whether searching for genetic variants with our SureSeq NGS panels, analyzing the variations linked to rare disease and reproductive health using CytoSure arrays and NGS solutions, or selecting from our extensive range of Cytocell FISH probes, our customers know they can rely on OGT for the most advanced and accurate tools available.

As part of the Sysmex group since 2017, we've reinforced our initiatives toward personalized medicine with the development of new products, investing in our technical teams and facilities, and expanding product distribution across the globe. Customers choose OGT not only for the quality of our solutions and the range of products – both catalog and customized – but also for the product support available. We are renowned for our application expertise, and have an experienced network of specialists to support our complete range of products.

We know that the decision to choose the best genetic analysis products is critical to your success. We thank you for choosing OGT.

John Anson
CEO, Oxford Gene Technology

Cytocell AML/MDS range of FISH probe kits: For *In Vitro* Diagnostic Use. Rx only. Other Cytocell® FISH probes: Analyte Specific Reagent. Analytical and performance characteristics are not established. Product availability may vary from country to country and is subject to varying regulatory requirements.

myProbes®: For USA - Analyte Specific Reagent. Analytical and performance characteristics are not established.
myProbes®: For Canada - For Research Use Only. Not for use in diagnostic procedures.
CytoSure™, SureSeq™: For Research Use Only (RUO); Not for Use in Diagnostic Procedures.

Contents

Cytocell FISH Probes for Constitutional Cytogenetics and Hematology/Pathology Cytogenetics

Hematology

12	AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual Fusion	IVD	40	IGH Proximal <i>Plus</i>
13	CBFβ (CBFB)/MYH11 Translocation, Dual Fusion	IVD	41	IGH Distal <i>Plus</i>
14	Del(5q) Deletion	IVD	41	IGH <i>Plus</i>
15	Del(7q) Deletion	IVD	42	BCL2 <i>Plus</i>
16	Del(20q) Deletion	IVD	42	IGH Probe Green
17	EVI1 (MECOM) Breakapart	IVD	43	CCND1 <i>Plus</i>
18	MLL (KMT2A) Breakapart	IVD	43	CCND3 <i>Plus</i>
19	P53 (TP53) Deletion	IVD	44	cMYC (MYC) <i>Plus</i>
20	AML1 (RUNX1) Breakapart		44	FGFR3 <i>Plus</i>
21	AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual fusion		45	MAF v2 Probe Red
22	ATM Deletion Probe		45	MAFB <i>Plus</i>
23	BCR/ABL (ABL1) Translocation, Dual Fusion		46	MYEOV <i>Plus</i>
24	BCR/ABL (ABL1) <i>Plus</i> Translocation, Dual Fusion		46	IGK Breakapart
25	BCL6 Breakapart Probe		47	IGL Breakapart
26	CBFB Proximal Probe Red		47	MECOM Probe Red
26	CBFB Distal Probe Green		48	MLL (KMT2A) Breakapart
27	CBFB/MYH11 Translocation, Dual Fusion		48	MLL (KMT2A)
28	CSF1R/RPS14 (5q32-q33) Probe Red		49	AFF1
29	CKS1B/CDKN2C (P18) Amplification/Deletion Probe		49	MLLT1
29	cMYC (MYC) Breakapart		50	MLLT3
30	CRLF2 Distal		50	MLLT4 (AFDN)
30	CRLF2 Proximal		51	MYB Deletion
31	CUX1 (7q22) Probe Green		51	NUP98 Proximal Probe Red
31	13q14.3 Deletion Probe		52	NUP98 Distal Probe Green
32	D13S319 <i>Plus</i> Deletion Probe		52	P16 (CDKN2A) Deletion
33	D13S25 Deletion Probe		53	P53 (TP53) Deletion
33	Del(5q) Deletion		54	P2RY8 Distal
34	Del(7q) Deletion		54	P2RY8 Proximal
34	Del(20q) Deletion		55	P53(TP53)/ATM Probe Combination
35	E2A (TCF3) Breakapart Probe		56	PDGFRB Breakapart
36	E2A (TCF3)		57	FAST PML
36	PBX1		57	FAST RARα (RARA)
37	HLF		58	PML/RARα (RARA) Translocation, Dual Fusion
37	EGR1/CDC25C (5q31) Probe Green		59	RARα (RARA) Distal
38	EVI1 (MECOM) Breakapart		59	RARα (RARA) Proximal
38	EZH2 (7q36) Probe Red		60	RUNX1 Probe Green
39	FIP1L1/CHIC2/PDGFRα Deletion/Fusion		60	TERT (5p15.33) Probe Aqua
40	IGH Breakapart Probe		61	TET2 Probe Red
			61	USP46 (4q12) Probe Green
			62	Chromosome 7 Alpha Satellite Probe Aqua
			62	Chromosome 9 Satellite III Probe Aqua
			63	TCL1 Breakapart
			63	TCRAD Breakapart
			64	TCRB (TRB) Breakapart
			65	TEL/AML1 (ETV6/RUNX1) Translocation, Dual Fusion

Cytocell FISH probes are ASR unless otherwise indicated. See product page for disclaimers.
 ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.
 IVD: *In Vitro* Diagnostic Use. Rx only.
 RUO: For Research Use Only, not for use in diagnostic procedures.

66	TLX1 Breakapart
66	TLX3 Breakapart
67	ETV6 Proximal Probe Green
67	ETV6 Distal Probe Red
68	MPO Probe Red
68	TP53 Probe Green
69	NUP214 Probe Red
69	DEK Probe Green
70	TAS2R1 (5p15.31) Probe Green
71	Alpha Satellite 12 <i>Plus</i>
71	Chromosome 15 Alpha Satellite Probe Red

Hematopathology

75	BCL2 Breakapart
75	BCL6 Breakapart
76	CCND1 Breakapart
76	IGH Breakapart
77	IGH/BCL2 Translocation, Dual Fusion
78	IGH/CCND1 Translocation, Dual Fusion
79	IGH/MALT1 Translocation, Dual Fusion
80	IGH/MYC Translocation, Dual Fusion
81	IGK Breakapart
81	IGL Breakapart
82	MALT1 Breakapart
82	MYC Breakapart
83	P16 (CDKN2A) Deletion
83	P53 (TP53) Deletion
84	RB1 Deletion

Pathology

87	Tissue Pretreatment Kit
88	1p36
88	1q25
89	19p13
89	19q13
90	ALK Breakapart
90	CDKN2A Probe Gold
91	CHOP (DDIT3) Breakapart
91	Centromere 17 Probe Aqua
92	Centromere 7 Probe Green
92	Centromere 3 Probe Red
93	C-MET (MET) Amplification
93	EGFR Amplification
94	EML4 Breakapart
94	ERG Control

95	ETV6 Proximal Probe Green
95	ETV6 Distal Probe Red
96	EWSR1 Breakapart
97	EWSR1/ERG Translocation, Dual Fusion
98	FOXO1 Proximal Probe Green
98	FOXO1 Distal Probe Red
99	FUS Proximal Probe Red
99	FUS Distal Probe Green
100	FLI1/EWSR1 Translocation, Dual Fusion
101	FGFR1 Breakapart/Amplification
101	HER2 (ERBB2) Amplification
102	MDM2 Amplification
102	MPO Probe Red
103	N-MYC (MYCN) Amplification
103	PAX3 Breakapart
104	PAX7 Breakapart
104	RET Proximal
105	RET Distal
105	ROS1 Breakapart
106	ROS1-GOPC (FIG) Proximal
106	ROS1-GOPC (FIG) Distal
107	SRD (CHD5) Deletion
107	SYT (SS18) Breakapart
108	TMPRSS2 Breakapart
108	TFE3 Proximal Probe Red
109	TFE3 Distal Probe Green
109	TOP2A Amplification/Deletion
110	ZNF217 Amplification
110	TP53 Green

Microdeletion

113	Angelman (UBE3A/D15S10) Region
114	Cri-du-chat Region/Sotos Region
115	DiGeorge II (10p14)
116	DiGeorge/VCFS TUPLE1 Region and 22q13.3 Region
117	DiGeorge/VCFS N25 Region and 22q13.3 Region
118	DiGeorge TBX1 Region and 22q13.3 Region
119	Kallmann (KAL1) Region/STS Region
120	Prader-Willi/Angelman (SNRPN) Region
121	Saethre-Chotzen Region/Williams-Beuren Region
122	SHOX
123	Smith-Magenis (RAI1) Region/Miller-Dieker Region
124	SRY
125	Williams-Beuren Region
126	Wolf-Hirschhorn Region

Cytocell FISH probes are ASR unless otherwise indicated. See product page for disclaimers.
 ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.
 IVD: *In Vitro* Diagnostic Use. Rx only.
 RUO: For Research Use Only, not for use in diagnostic procedures.

Satellites

- 129 Satellite Enumeration Probes
- 130 Blue Labeled Satellite Enumeration Probes
- 130 Dual Labeled Satellite Probe Sets
- 130 Acro-P-Arm Probe

Subtelomere Specific Probes

- 133 Aquarius® Subtelomere Specific Probes

Custom FISH Probes

- 136 myProbes® Custom FISH Probes (RUO in Canada)

FISH Accessories

- 138 FISH accessories and ancillary items
- 140 Ordering guide

SureSeq NGS Products for Hematology and Solid Tumor Cancer Research **RUO**

- 143 SureSeq myPanel™ NGS Custom Cancer Panels
- 145 SureSeq myPanel NGS Custom AML Panel
- 147 SureSeq myPanel NGS Custom Breast Cancer Panel
- 149 SureSeq myPanel NGS Custom Colorectal Cancer Panel
- 151 SureSeq myPanel NGS Custom Melanoma Cancer Panel
- 153 SureSeq myPanel NGS Custom Prostate Cancer Panel

SureSeq

- 155 SureSeq CLL + CNV Panel
- 157 SureSeq Core MPN Panel
- 159 SureSeq Ovarian Cancer Panel
- 161 SureSeq Myeloid Panel
- 163 Interpret Software
- 165 SureSeq NGS Library Preparation Kit
- 167 SureSeq FFPE DNA Repair Mix

CytoSure NGS and Array Products for Cytogenetics and Rare Disease **RUO**

CytoSure Next Generation Sequencing (NGS)

- 171 CytoSure Constitutional NGS
- 173 CytoSure NGS Comprehensive FH Panel

Constitutional Arrays

- 175 CytoSure Constitutional v3 and Constitutional v3 + LOH Arrays

CytoSure Disease-Focused Arrays

- 177 CytoSure Medical Research Exome Array
- 179 CytoSure Disease-Focused Arrays
- 180 CytoSure Autism Research Array
- 180 CytoSure Cardiomyopathy Research Array
- 181 CytoSure Epilepsy Research Array
- 181 CytoSure Eye Disease Research Array
- 182 CytoSure NMD Research Array

- 183 CytoSure Cancer +SNP Arrays
- 186 CytoSure Custom Designed aCGH Arrays
- 187 CytoSure Interpret Software
- 189 CytoSure Genomic DNA Labelling kits
- 191 CytoSure Sample Tracking Spike-ins
- 192 Accessories

Educational Resources

FISH Probes

- 195 FISH glossary: Chromosome basics, guide to chromomaps and common terms explained
- 200 Recommended FISH protocol for AML/MDS FDA-cleared IVD Cytocell FISH probes
- 202 AML/MDS FDA-cleared IVD Cytocell FISH probes: Guide to expected abnormal clinical results

Next Generation Sequencing

- 206 Choosing the best enrichment assay
- 208 One-day hybridization protocol
- 211 Comprehensive results from a single NGS assay: panel and software highlights

Arrays

- 212 Basics of array aCGH
- 214 Troubleshooting for arrays

Indices of FISH probes

- 218 Aquarius Hematology Probe Range Summary
- 221 Aquarius Tissue Pretreatment Kit Summary
- 221 Aquarius Hematopathology Probe Range Summary
- 221 Aquarius Pathology Probe Range Summary
- 223 Aquarius Microdeletion Probe Range Summary
- 224 Aquarius Satellite Enumeration Probe Range Summary
- 224 Acro-P-Arm Probe
- 225 Aquarius Subtelomere Specific Probe Range Summary
- 226 FISH Accessories and Ancillary Products
- 227 Index by Chromosome
- 233 Index by Gene Name

Cytocell FISH probes are ASR unless otherwise indicated. See product page for disclaimers.
 ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.
 IVD: *In Vitro* Diagnostic Use. Rx only.
 RUO: For Research Use Only, not for use in diagnostic procedures.



Overview: Cytocell Products

Cytocell FISH probes are high quality, directly labeled DNA probes for fluorescence *in situ* hybridization (FISH). Our portfolio includes a broad range of catalog FISH probes and well as myProbes® custom products. We offer these product ranges from within the USA and Canada:

Aquarius Range

The Aquarius® range of Cytocell FISH probes consists of directly labeled FISH probes for constitutional cytogenetics and hematology/pathology cytogenetics. We also offer a range of FDA-cleared IVD FISH probe test kits specifically designed to detect chromosomal rearrangements reported in acute myeloid leukemia (AML) and myelodysplastic syndromes (MDS). Satellite and subtelomere specific probes are also available.

myProbes

myProbes® is a custom FISH probe design and manufacture service, which utilizes Cytocell's proprietary BAC clone collection.

FISH Accessories

We supply reagents and materials to ensure that your FISH experiment is hassle free. Our range includes: filters, slides, counterstains, hybridization solutions, hybridization chambers, Tissue Pretreatment Kit, slide surface thermometers, and porcelain wash jars. See our FISH accessories section for details.

Trademarks

Aquarius, Cytocell and myProbes are registered trademarks of Cytocell Ltd.

Quality Management

Cytocell Ltd operates a Quality Management System that has been approved by BSI to ISO 9001:2015, ISO 13485:2016, and the full MDSAP audit criteria. The scope of this approval is applicable to the design, development and manufacture of DNA FISH probes, ancillary products and *in vitro* diagnostic kits and reagents for the detection of chromosomal abnormalities in life science research and diagnostic use. For more information or to see our certificates online visit:

<https://www.ogt.com/about-us/quality-assurance/>

Regulatory Status

Cytocell Aquarius® AML/MDS range of FISH probe kits: Refer to individual test kit Package Insert for the specific intended use and limitations. For *In Vitro* Diagnostic Use. Rx only. For sale in the US only. These products have not been licensed in accordance with Canadian law.

Other Cytocell® and myProbes® FISH probes: Analyte Specific Reagent. Analytical and performance characteristics are not established. For Canada: myProbes are RUO - For Research Use Only.

Product availability may vary from country to country and is subject to varying regulatory requirements. Please contact your local representatives for availability.



FM 723073



MD 722804



Cytocell FISH probes are developed and produced in the UK

Additional OGT product information and legal notices

This document and its contents are © Oxford Gene Technology IP Limited. All rights reserved. OGT™, CytoSure™, SureSeq™ and Oxford Gene Technology are trademarks of Oxford Gene Technology IP Limited. CytoSure, and SureSeq: For Research Use Only; Not for Use in Diagnostic Procedures. Product availability may vary from country to country and is subject to varying regulatory requirements. Please contact your local representatives for availability.

Product notices:

Tissue Pretreatment Kit (LPS 100): This product is provided under an agreement between Life Technologies Corporation and CytoCell Ltd and is available for human diagnostics or life science use only.

SureSeq NGS Library Preparation Kit was jointly developed between Oxford Gene Technology and Bioline Reagents Limited. SureSeq FFPE DNA Repair Mix can only be purchased in conjunction with SureSeq NGS panels, not as a standalone product.

CytoSure Arrays: This product is provided under an agreement between Agilent Technologies, Inc. and OGT. The manufacture, use, sale or import of this product may be subject to one or more of U.S. patents, pending applications, and corresponding international equivalents, owned by Agilent Technologies, Inc. The purchaser has the non-transferable right to use and consume the product for RESEARCH USE ONLY AND NOT for DIAGNOSTICS PROCEDURES. It is not intended for use, and should not be used for the diagnosis, prevention, monitoring, treatment or alleviation of any disease or condition, or for the investigation of any physiological process, in any identifiable human, or for any other medical purpose.

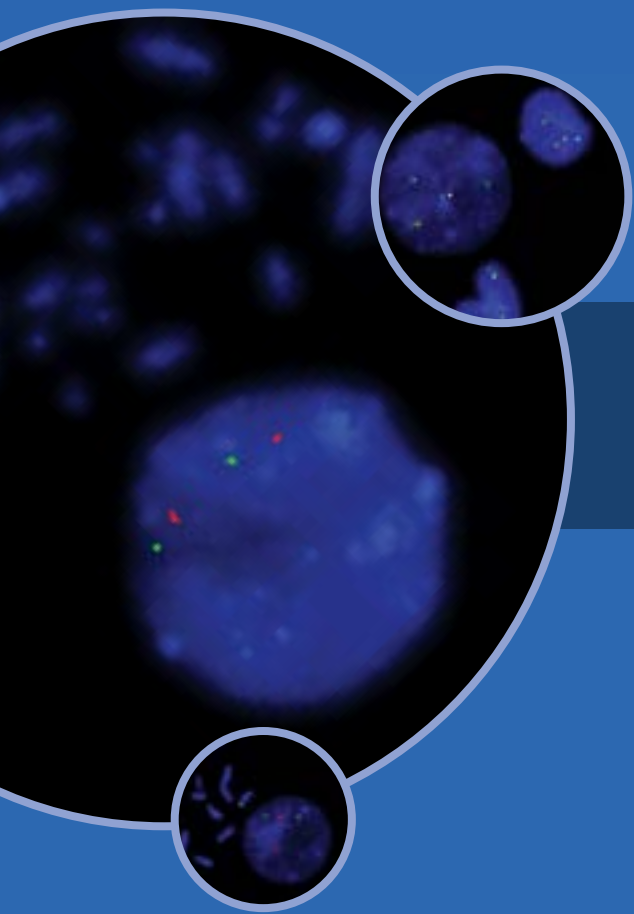
Disclaimer:

All product and company names are trademarks™ or registered® trademarks of their respective holders. Use of them does not imply any affiliation with or endorsement by them. All specifications are subject to change without notice.

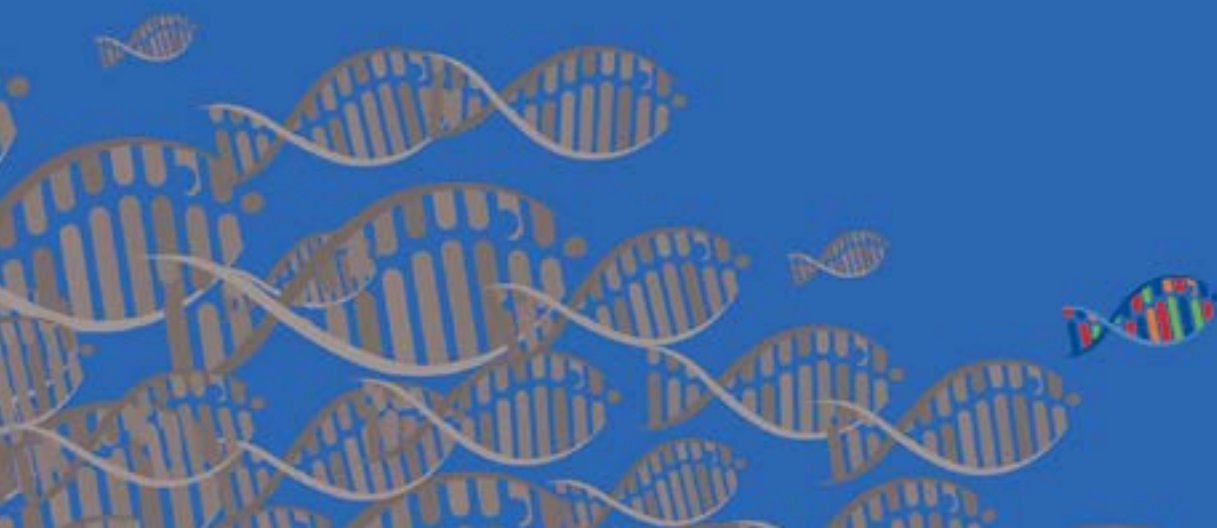
Every attempt has been made to ensure that the information in this catalog is correct at time of going to print. Although every care has been taken in preparing this publication, no responsibility or liability will be accepted by CytoCell Ltd, Oxford Gene Technology IP Limited or its employees for its accuracy or completeness.




Cytocell
aquarius



Hematology



Hematology

The Cytocell hematology range of FISH probes now includes FDA-cleared FISH probe test kits specifically designed to detect chromosomal rearrangements reported in acute myeloid leukemia (AML) and myelodysplastic syndrome (MDS).

Contents

FDA-cleared Class II IVD FISH Probe Kits for AML and MDS*

12	AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual Fusion	IVD	AML	
13	CBFβ (CBFB)/MYH11 Translocation, Dual Fusion	IVD	AML	
14	Del(5q) Deletion	IVD	AML	MDS
15	Del(7q) Deletion	IVD	AML	MDS
16	Del(20q) Deletion	IVD	MDS	
17	EVI1 (MECOM) Breakapart	IVD	AML	MDS
18	MLL (KMT2A) Breakapart	IVD	AML	MDS
19	P53 (TP53) Deletion	IVD	AML	MDS

Refer to our Hematology key to determine the associated disease for the products indicated:*

AML Acute Myeloid Leukemia

MDS Myelodysplastic Syndrome

* The Cytocell Aquarius AML/MDS range of FISH probe test kits are fluorescence *in situ* hybridization (FISH) tests used to detect common chromosomal rearrangements in fixed bone marrow specimens from patients with acute myeloid leukemia (AML) or myelodysplastic syndrome (MDS). The tests are indicated for the characterization of patient specimens consistent with World Health Organization guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are to be interpreted by a qualified pathologist or cytogeneticist. The tests are not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic. Refer to individual test kit Package Insert for the specific intended use and limitations. For *In Vitro* Diagnostic Use. Rx only.



Contents

Hematology FISH Probes (ASR)**	
20	AML1 (RUNX1) Breakapart
21	AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual Fusion
22	ATM Deletion
23	BCR/ABL (ABL1) Translocation, Dual Fusion
24	BCR/ABL (ABL1) <i>Plus</i> Translocation, Dual Fusion
25	BCL6 Breakapart Probe
26	CBFB Proximal Probe Red
26	CBFB Distal Probe Green
27	CBFB/MYH11 Translocation, Dual Fusion
28	CSF1R/RPS14 (5q32-q33) Probe Red
29	CKS1B/CDKN2C (P18) Amplification/Deletion Probe
29	cMYC (MYC) Breakapart
30	CRLF2 Distal
30	CRLF2 Proximal
31	CUX1 (7q22) Probe Green
31	13q14.3 Deletion
32	D13S319 <i>Plus</i> Deletion
33	D13S25 Deletion
33	Del(5q) Deletion
34	Del(7q) Deletion
35	Del(20q) Deletion
35	E2A (TCF3) Breakapart
36	E2A (TCF3)
36	PBX1
37	HLF
37	EGR1/CDC25C (5q31) Probe Green
38	EVI1 (MECOM) Breakapart
38	EZH2 (7q36) Probe Red
39	FIP1L1/CHIC2/PDGFRA Deletion/Fusion
40	IGH Breakapart
40	IGH Proximal <i>Plus</i>
41	IGH Distal <i>Plus</i>
41	IGH <i>Plus</i>
42	IGH Probe Green
42	BCL2 <i>Plus</i>
43	CCND1 <i>Plus</i>
43	CCND3 <i>Plus</i>
44	cMYC (MYC) <i>Plus</i>
44	FGFR3 <i>Plus</i>
45	MAF v2 Probe Red
45	MAFB <i>Plus</i>
46	MYEOV <i>Plus</i>
46	IGK Breakapart
47	IGL Breakapart
47	MECOM Probe Red
48	MLL(KMT2A) Breakapart
48	MLL(KMT2A)
49	AFF1
49	MLLT1
50	MLLT3
50	MLLT4 (AFDN)
51	MYB Deletion
51	NUP98 Proximal Red
52	NUP98 Distal Green
52	P16 (CDKN2A) Deletion
53	P53 (TP53) Deletion
54	P2RY8 Distal
54	P2RY8 Proximal
55	P53 (TP53)/ATM Probe Combination
56	PDGFRB Breakapart
57	<i>FAST</i> PML
57	<i>FAST</i> RAR α (RARA)
58	PML/RAR α (RARA) Translocation, Dual Fusion
59	RAR α (RARA) Distal
59	RAR α (RARA) Proximal
60	RUNX1 Probe Green
60	TERT (5p15.33) Probe Aqua
61	TET2 Probe Red
61	USP46 (4q12) Probe Green
62	Chromosome 7 Alpha Satellite Probe Aqua
62	Chromosome 9 Satellite III Probe Aqua
63	TCL1 Breakapart
63	TCRAD Breakapart
64	TCRB (TRB) Breakapart
65	TEL/AML1 (ETV6/RUNX1) Translocation, DF
66	TLX1 Breakapart
66	TLX3 Breakapart
67	ETV6 Proximal Probe Green
67	ETV6 Distal Probe Red
68	MPO Probe Red
68	TP53 Probe Green
69	NUP214 Probe Red
69	DEK Probe Green
70	TAS2R1 (5p15.31) Probe Green
71	Alpha Satellite 12 <i>Plus</i>
71	Chromosome 15 Alpha Satellite Probe Red

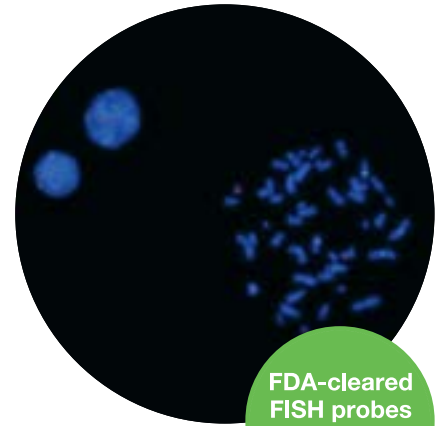
**ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



AML1/ETO (RUNX1/ RUNX1T1) Translocation, Dual Fusion*

AML with a RUNX1-RUNX1T1 fusion resulting from a t(8;21)(q22;q22) translocation is a recognized disease entity according to the World Health Organization (WHO) classification of myeloid neoplasms and acute leukemia¹. The translocation is commonly observed in patients with AML FAB type M2, most commonly in children and young adults² and is a good prognostic indicator^{3,4,5}. The t(8;21) breakpoint mainly occurs in the intron between exons 5 and 6, just before the transactivation domain. The fusion protein created contains the DNA-binding domain of RUNX1 fused to the transcription factor RUNX1T1².

In addition to the reciprocal t(8;21) translocation creating the RUNX1-RUNX1T1 fusion, variant translocations have also been reported. These variant rearrangements may be cryptic and easily overlooked by G-banding; however, FISH can indicate the presence of such rearrangements².



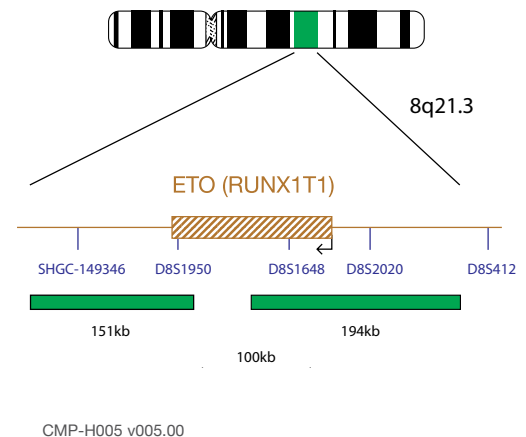
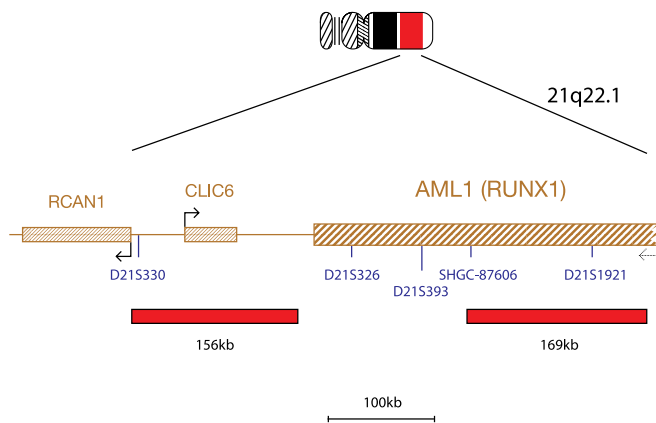
FDA-cleared
FISH probes

Cytocell

AML

REFERENCES

1. Swerdlow, *et al* (eds). WHO Classification of Tumours of Haematopoietic and Lymphoid Tissue, Lyon, France, 4th edition, IARC, 2017
2. Reikvam HH, *et al*. J Biomed Biotechnol. 2011;2011:1-23.
3. Grimwade D, *et al*. Blood. 2001;98(5):1312-20.
4. Harrison CJ, *et al*. J Clin Oncol. 2010;28(16):2674-81.
5. Grimwade D, *et al*. Blood. 2010;116(3):354-65.



* For sale in the US only. This product has not been licensed in accordance with Canadian law.

The AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual Fusion FISH Probe Kit is a fluorescence *in situ* hybridization (FISH) Test used to detect rearrangement involving the AML1 (RUNX1) region on chromosome 21 at location 21q22.1 and the ETO (RUNX1T1) region on chromosome 8 at location 8q21.3 in fixed bone marrow specimens from patients with acute myeloid leukemia (AML). The test is indicated for characterization of patient specimens consistent with World Health Organization (WHO) guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are intended to be interpreted by a qualified pathologist or cytogeneticist. The test is not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic.

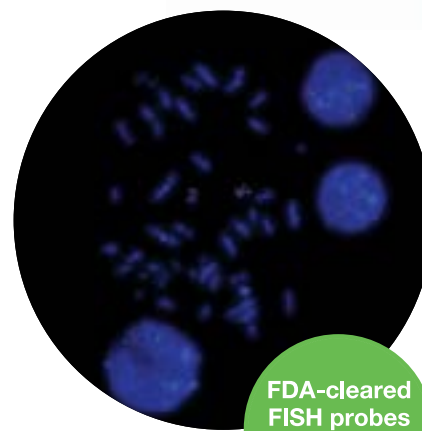
The device has not been specifically validated in patients with <20% blast count. For *In Vitro* Diagnostic Use. Rx only.

Reporting and interpretation of FISH results should be consistent with professional standards of practice and should take into consideration other clinical and diagnostic information. This kit is intended as an adjunct to other diagnostic laboratory tests and therapeutic action should not be initiated on the basis of the FISH result alone. Failure to adhere to the protocol may affect the performance and lead to false results. Each lab is responsible for establishing their own cut-off values. Each laboratory should test sufficiently large number of samples to establish normal population distribution of the signal levels and to assign a cut-off value. The product is for professional use only and is intended to be interpreted by a qualified Pathologist or Cytogeneticist. Product availability may vary from country to country and is subject to varying regulatory requirements.

CBFβ (CBFB)/MYH11 Translocation, Dual Fusion*

The CBFB (*core-binding factor beta subunit*) gene is located at 16q22, while the MYH11 (*myosin heavy chain 11*) gene is located at 16p13.1. The inversion *inv(16)(p13.11q22.1)* and the translocation *t(16;16)(p13.11;q22.1)* give rise to the CBFβ-MYH11 fusion gene.

Acute myeloid leukemias with *inv(16)(p13.11q22.1)* or *t(16;16)(p13.11;q22.1)* form a recognized disease entity according to the World Health Organization (WHO) classification of myeloid neoplasms and acute leukemia¹. These rearrangements are frequently found in patients with a myelomonocytic subtype with increased bone marrow eosinophils, AML FAB (French-American-British classification) type M4Eo. Cases of therapy-related AML may also have this rearrangement^{1,2}.



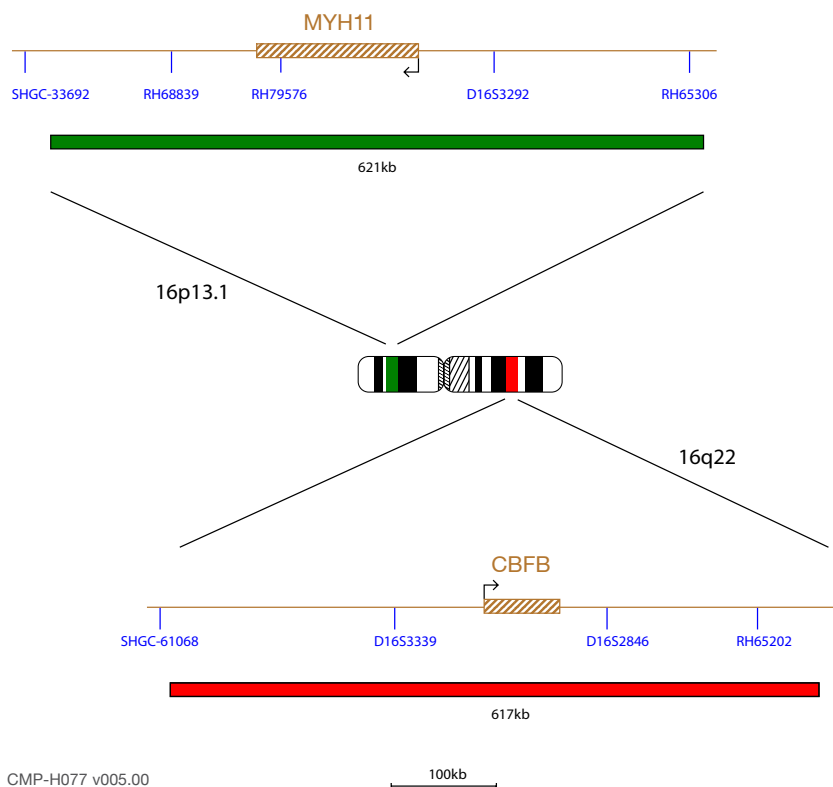
FDA-cleared
FISH probes

Cytocell

AML

REFERENCES

1. Swerdlow, et al (eds). WHO Classification of Tumours of Haematopoietic and Lymphoid Tissue, Lyon, France, 4th edition, IARC, 2017
2. Hernandez JM, et al. Haematologica. 2000;85(5):481-5.



CMP-H077 v005.00

* For sale in the US only. This product has not been licensed in accordance with Canadian law.

The Cytocell Aquarius CBFβ (CBFB) /MYH11 Translocation, Dual Fusion FISH Probe Kit is a fluorescence *in situ* hybridization (FISH) Test used to detect rearrangement of the chromosome 16 causing the CBFβ-MYH11 (CBFB-MYH11) fusion in fixed bone marrow specimens from patients with acute myeloid leukemia (AML). The test is indicated for characterization of patient specimens consistent with World Health Organization (WHO) guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are intended to be interpreted by a qualified pathologist or cytogeneticist. The test is not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic.

The device has not been specifically validated in patients with <20% blast count. For *In Vitro* Diagnostic Use. Rx only.

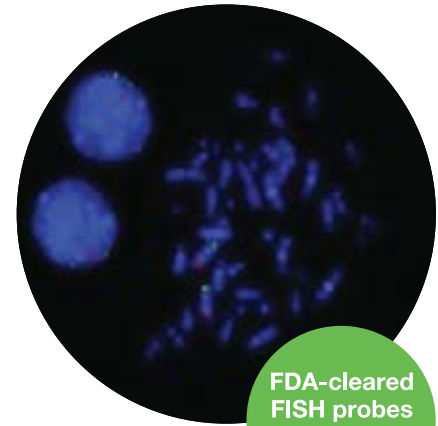
Reporting and interpretation of FISH results should be consistent with professional standards of practice and should take into consideration other clinical and diagnostic information. This kit is intended as an adjunct to other diagnostic laboratory tests and therapeutic action should not be initiated on the basis of the FISH result alone. Failure to adhere to the protocol may affect the performance and lead to false results. Each lab is responsible for establishing their own cut-off values. Each laboratory should test sufficiently large number of samples to establish normal population distribution of the signal levels and to assign a cut-off value. The product is for professional use only and is intended to be interpreted by a qualified Pathologist or Cytogeneticist. Product availability may vary from country to country and is subject to varying regulatory requirements.

Del(5q) Deletion*

Deletions of the long arm of chromosome 5 are one of the most common karyotypic abnormalities reported in myelodysplastic syndrome (MDS) and acute myeloid leukemia (AML) with myelodysplasia related changes^{1,2}.

EGR1 (*early growth response 1*), a tumor suppressor gene at 5q31.2, has been shown to act through haploinsufficiency to initiate the development of MDS/AML³. Loss of 5q31.2, the region detected by this probe set, which includes the EGR1 gene, have been linked to a more aggressive form of MDS and AML and is often accompanied by additional cytogenetic abnormalities and a poorer prognosis^{2,4,5}.

This probe can also detect some deletions that are associated with 5q- syndrome². However, the probe does not cover the critical deleted region for 5q33 and is not intended for the detection of all deletions associated with 5q- syndrome.

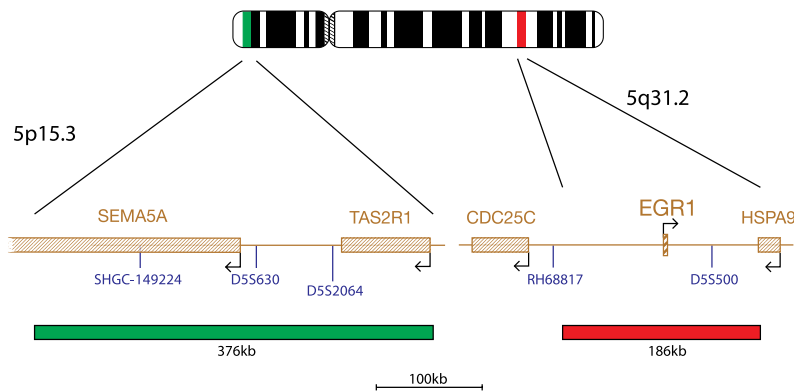


FDA-cleared
FISH probes

Cytocell

REFERENCES

1. Swerdlow, *et al* (eds). WHO Classification of Tumours of Haematopoietic and Lymphoid Tissue, Lyon, France, 4th edition, IARC, 2017
2. Ebert BL. Best Pract Res Clin Haematol. 2010;23(4):457–61.
3. Joslin JM, *et al*. Blood. 2007;110(2):719–26.
4. Fang J, *et al*. Cell Rep. 2014;8(5):1328–38.
5. Boultonwood J, *et al*. Blood. 2010;116(26):5803–11.



CMP-H017 v007

* For sale in the US only. This product has not been licensed in accordance with Canadian law.

The Del(5q) Deletion FISH Probe Kit is a fluorescence *in situ* hybridization (FISH) Test used to detect deletions within the long arm of chromosome 5 at location 5q31.2 in fixed bone marrow specimens from patients with acute myeloid leukemia (AML) or myelodysplastic syndrome (MDS). The test is indicated for characterization of patient specimens consistent with World Health Organization (WHO) guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are intended to be interpreted by a qualified pathologist or cytogeneticist. The test is not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic.

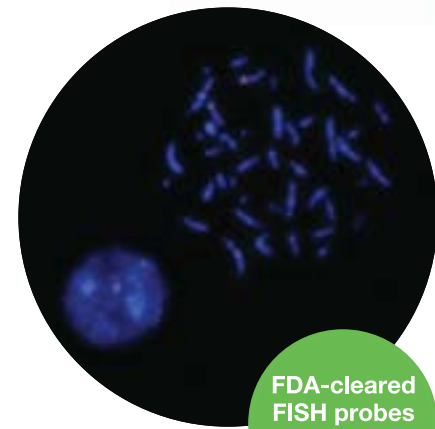
For *In Vitro* Diagnostic Use. Rx only.

Reporting and interpretation of FISH results should be consistent with professional standards of practice and should take into consideration other clinical and diagnostic information. This kit is intended as an adjunct to other diagnostic laboratory tests and therapeutic action should not be initiated on the basis of the FISH result alone. Failure to adhere to the protocol may affect the performance and lead to false results. Each lab is responsible for establishing their own cut-off values. Each laboratory should test sufficiently large number of samples to establish normal population distribution of the signal levels and to assign a cut-off value. The product is for professional use only and is intended to be interpreted by a qualified Pathologist or Cytogeneticist. Product availability may vary from country to country and is subject to varying regulatory requirements.

Del(7q) Deletion*

Monosomy of chromosome 7 and deletions of the long arm of chromosome 7 are recognized recurrent chromosomal aberrations frequently seen in myeloid disorders, including myelodysplastic syndrome (MDS) and acute myeloid leukemia (AML)¹. Furthermore, these abnormalities occur in MDS and AML that develop in patients with constitutional disorders (e.g., Fanconi anemia, Kostmann syndrome, neurofibromatosis type 1, and familial monosomy 7)².

The presence of monosomy 7 or del(7q) as karyotypic change is associated with a poorer outcome in myeloid malignancies^{1,3}. Deletions of chromosome 7 are typically large with heterogeneity in the breakpoints in myeloid diseases, making it difficult to map the common deleted regions (CDRs)⁴.



FDA-cleared
FISH probes

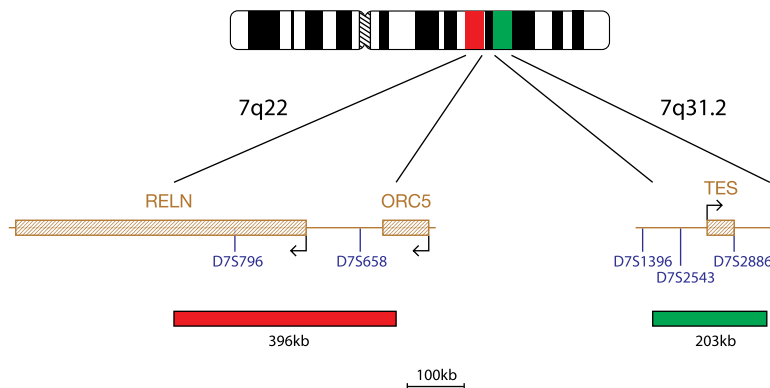
Cytocell

AML

MDS

REFERENCES

1. Jerez A, et al. Blood. 2012;119(25):6109–17.
2. Fischer K, et al. Blood. 1997;89(6):2036–41.
3. Trobaugh-Lotrario D, et al. Bone Marrow Transplant. 2005;35(2):143–9.
4. McNerney ME, et al. Biochem J. 2013;444(3):975–83.



CMP-H018 v006

* For sale in the US only. This product has not been licensed in accordance with Canadian law.

The Del(7q) Deletion FISH Probe Kit is a fluorescence *in situ* hybridization (FISH) Test used to detect deletions within the long arm of chromosome 7 at locations 7q22 and 7q31.2 in fixed bone marrow specimens from patients with acute myeloid leukemia (AML) or myelodysplastic syndrome (MDS). The test is indicated for characterization of patient specimens consistent with World Health Organization (WHO) guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are intended to be interpreted by a qualified pathologist or cytogeneticist. The test is not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic.

In Vitro Diagnostic Use. Rx only.

Reporting and interpretation of FISH results should be consistent with professional standards of practice and should take into consideration other clinical and diagnostic information. This kit is intended as an adjunct to other diagnostic laboratory tests and therapeutic action should not be initiated on the basis of the FISH result alone. Failure to adhere to the protocol may affect the performance and lead to false results. Each lab is responsible for establishing their own cut-off values. Each laboratory should test sufficiently large number of samples to establish normal population distribution of the signal levels and to assign a cut-off value. The product is for professional use only and is intended to be interpreted by a qualified Pathologist or Cytogeneticist. Product availability may vary from country to country and is subject to varying regulatory requirements.

Del(20q) Deletion*

Deletions of the long arm of chromosome 20 are recognized as recurrent chromosomal abnormalities associated with myelodysplastic syndromes (MDS)¹.

The prognosis for MDS where del(20q) is the sole abnormality is good; however, the presence of secondary abnormalities may be indicative of disease progression².



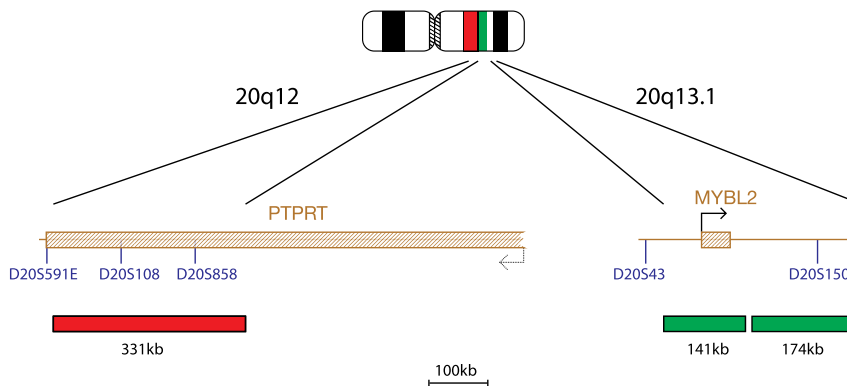
FDA-cleared
FISH probes

Cytocell

MDS

REFERENCES

1. Brezinova J, *et al.* Cancer Genet Cytogenet. 2005 Jul;160(2):188–92.
2. Liu Y-C, *et al.* Cancer Genet Cytogenet. 2006 Nov;171(1):9–16.



CMP-H019 v006

* For sale in the US only. This product has not been licensed in accordance with Canadian law.

The Del(20q) Deletion FISH Probe Kit is a fluorescence *in situ* hybridization (FISH) Test used to detect deletion within the long arm of chromosome 20 at locations 20q12 and 20q13.1, in fixed bone marrow specimens from patients with myelodysplastic syndrome (MDS). The test is indicated for characterization of patient specimens consistent with World Health Organization (WHO) guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are intended to be interpreted by a qualified pathologist or cytogeneticist. The test is not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic.

For *In Vitro* Diagnostic Use. Rx only.

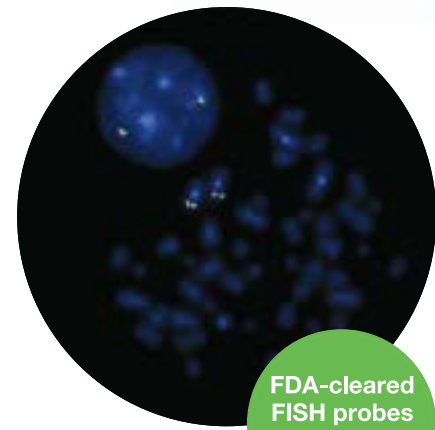
Reporting and interpretation of FISH results should be consistent with professional standards of practice and should take into consideration other clinical and diagnostic information. This kit is intended as an adjunct to other diagnostic laboratory tests and therapeutic action should not be initiated on the basis of the FISH result alone. Failure to adhere to the protocol may affect the performance and lead to false results. Each lab is responsible for establishing their own cut-off values. Each laboratory should test sufficiently large number of samples to establish normal population distribution of the signal levels and to assign a cut-off value. The product is for professional use only and is intended to be interpreted by a qualified Pathologist or Cytogeneticist. Product availability may vary from country to country and is subject to varying regulatory requirements.

EVI1 (MECOM) Breakapart*

The MECOM (*MDS1 and EVI1 complex locus*) oncogene at 3q26.2 is often rearranged in hematological malignancies of myeloid origin. MECOM encodes a zinc finger protein that is inappropriately expressed in the leukemic cells of AML and MDS patients¹. This deregulated expression is often due to a chromosomal rearrangement involving 3q26.2, with the two most common aberrations being the t(3;3)(q21;q26.2) and inv(3)(q21q26.2)¹. The breakpoints for the translocations and inversions vary considerably. Inversion breakpoints are found centromeric to, and include the MECOM gene, covering about 600kb. The majority of breakpoints in 3q26.2 translocations are telomeric to the MECOM gene and cover a region including the telomeric end of the MDS1 gene and the MYNN gene².

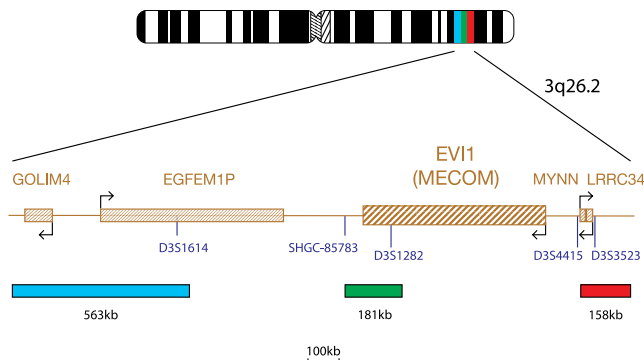
Chromosome rearrangements involving the 3q26.2 region are associated with myeloid malignancies, aberrant expression of MECOM gene and an unfavorable prognosis². AML with inv(3)(q21q26.2) or t(3;3)(q21;q26.2) is a recognized disease entity according to the World Health Organization (WHO) classification of myeloid neoplasms and acute leukemia. This is a transformed or *de novo* AML with a very aggressive clinical course and aberrations that involve MECOM at 3q26.2 and RPN1 (ribophorin I) at 3q21³.

MECOM has also been shown to be rearranged in therapy-related disease via the t(3;21)(q26.2;q22) translocation, resulting in a MECOM-RUNX1 fusion^{3,4}. MECOM rearrangements are very heterogeneous and may be difficult to detect by conventional cytogenetics, making FISH a useful tool for their detection.



REFERENCES

1. Soderholm J, *et al.* Leukemia. 1997;11:352-358.
2. Bobadilla D, *et al.* Br J Haematol. 2007;136(6):806-813.
3. Swerdlow, *et al.*, (eds.) WHO Classification of Tumours of Haematopoietic and Lymphoid Tissue, Lyon, France, 4th edition, IARC, 2017
4. Pedersen-Bjergaard J, *et al.* Leukemia. 2008;22(2):240-248.



CMP-H021 v008

* For sale in the US only. This product has not been licensed in accordance with Canadian law.

The EVI1 (MECOM) Breakapart FISH Probe Kit is a fluorescence *in situ* hybridization (FISH) Test used to detect rearrangement involving the EVI1 (MECOM) region on chromosome 3 at location 3q26.2, in fixed bone marrow specimens from patients with acute myeloid leukemia (AML) or myelodysplastic syndrome (MDS). The test is indicated for characterization of patient specimens consistent with World Health Organization (WHO) guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are intended to be interpreted by a qualified pathologist or cytogeneticist. The test is not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic.

For In Vitro Diagnostic Use. Rx only.

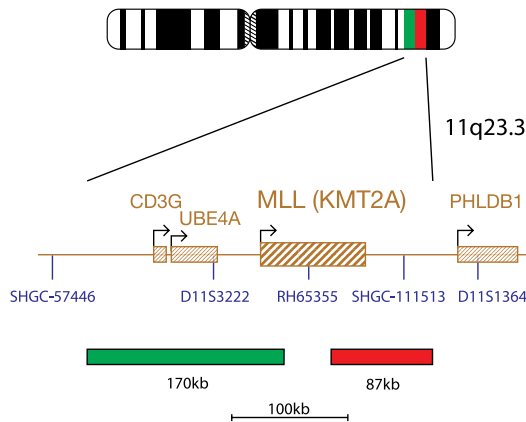
Reporting and interpretation of FISH results should be consistent with professional standards of practice and should take into consideration other clinical and diagnostic information. This kit is intended as an adjunct to other diagnostic laboratory tests and therapeutic action should not be initiated on the basis of the FISH result alone. Failure to adhere to the protocol may affect the performance and lead to false results. Each lab is responsible for establishing their own cut-off values. Each laboratory should test sufficiently large number of samples to establish normal population distribution of the signal levels and to assign a cut-off value. The product is for professional use only and is intended to be interpreted by a qualified Pathologist or Cytogeneticist. Product availability may vary from country to country and is subject to varying regulatory requirements.

MLL (KMT2A) Breakapart*

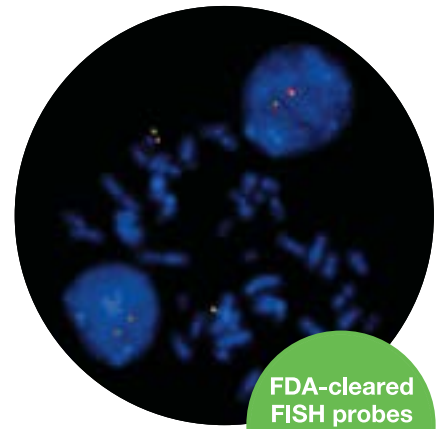
The KMT2A (*lysine methyltransferase 2A*) gene at 11q23.3 encodes for a histone methyltransferase, which functions as an epigenetic regulator of transcription¹.

KMT2A rearrangements are reported frequently in patients with AML and have also been reported in patients with therapy related MDS, albeit at a lower frequency^{2,3,4,5,6}. Historically, KMT2A rearrangements in acute leukemia were associated with a poorer outcome, but recent studies have shown that the prognosis is highly dependent on the fusion partner and may differ between children and adults⁷. Because this is a breakapart probe, it cannot be used to determine the fusion partner.

Probe Specification MLL, 11q23.3, Red
 MLL, 11q23.3, Green



CMP-H036 v006



FDA-cleared
FISH probes

AML MDS

Cytocell

REFERENCES

1. Tomizawa D. *Pediatr Int.* 2015;4:811-9.
2. Wright RL, *et al.* *Crit Rev Oncol Hematol.* 2014;91(3):283-91.
3. Van der Burg M, *et al.* *Leukemia.* 2004;18(5):895-908.
4. Grossmann V, *et al.* *Leukemia.* 2013. p.1933-6.
5. Super HJ, *et al.* *Blood.* 1993 Dec;82(12):3705-11.
6. Schanz J, *et al.* *J Clin Oncol.* 2012;30(8):820-9.
7. Tamai H, *et al.* *J Clin Exp Hematop.* 2010;50(2):91-8.

* For sale in the US only. This product has not been licensed in accordance with Canadian law.

The MLL (KMT2A) Breakapart FISH Probe Kit is a fluorescence *in situ* hybridization (FISH) Test used to detect rearrangement of the MLL (KMT2A) region on chromosome 11 at location 11q23.3 in fixed bone marrow specimens from patients with acute myeloid leukemia (AML) or myelodysplastic syndrome (MDS). The test is indicated for characterization of patient specimens consistent with World Health Organization (WHO) guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are intended to be interpreted by a qualified pathologist or cytogeneticist. The test is not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic.

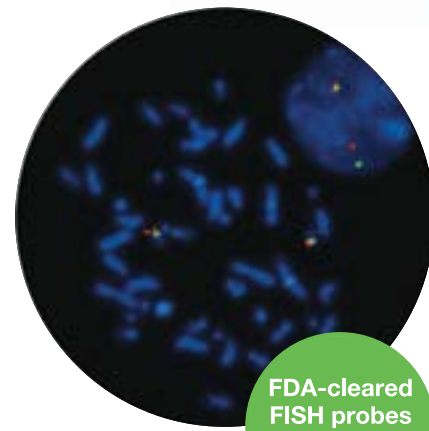
For *In Vitro* Diagnostic Use. Rx only.

Reporting and interpretation of FISH results should be consistent with professional standards of practice and should take into consideration other clinical and diagnostic information. This kit is intended as an adjunct to other diagnostic laboratory tests and therapeutic action should not be initiated on the basis of the FISH result alone. Failure to adhere to the protocol may affect the performance and lead to false results. Each lab is responsible for establishing their own cut-off values. Each laboratory should test sufficiently large number of samples to establish normal population distribution of the signal levels and to assign a cut-off value. The product is for professional use only and is intended to be interpreted by a qualified Pathologist or Cytogeneticist. Product availability may vary from country to country and is subject to varying regulatory requirements.

P53 (TP53) Deletion*

The TP53 (*tumor protein p53*) gene at 17p13 is a tumor suppressor gene that has been shown to be deleted in a wide range of human malignancies. In acute myeloid leukemia (AML) and myelodysplastic syndrome (MDS), TP53 loss is associated with a poor outcome and is often seen as a marker of disease progression or secondary disease^{1,2}.

Probe Specification P53, 17p13, Texas Red
D17Z1, 17p11.1-q11.1, FITC green



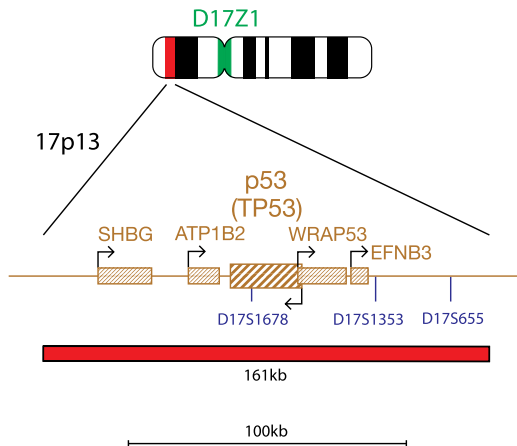
FDA-cleared
FISH probes

Cytocell

AML MDS

REFERENCES

1. Grimwade D, et al. *Blood*. 2010;116(3):354.
2. Seifert H, et al. *Leukemia*. 2009;23(4):656-63.



CMP-H039 v006

* For sale in the US only. This product has not been licensed in accordance with Canadian law.

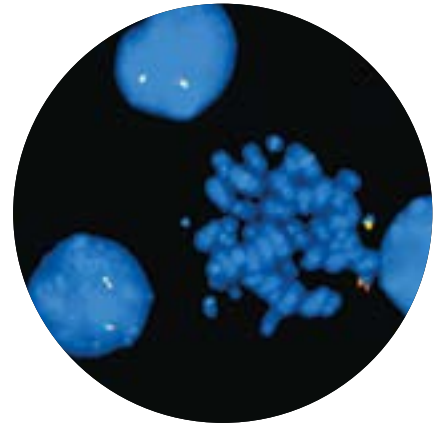
The P53 (TP53) Deletion FISH Probe Kit is a fluorescence *in situ* hybridization (FISH) Test used to detect deletion of the P53 (TP53) region on chromosome 17 at location 17p13 in fixed bone marrow specimens from patients with acute myeloid leukemia (AML) or myelodysplastic syndrome (MDS). The test is indicated for characterization of patient specimens consistent with World Health Organization (WHO) guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are intended to be interpreted by a qualified pathologist or cytogeneticist. The test is not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic.

For *In Vitro* Diagnostic Use. Rx only.

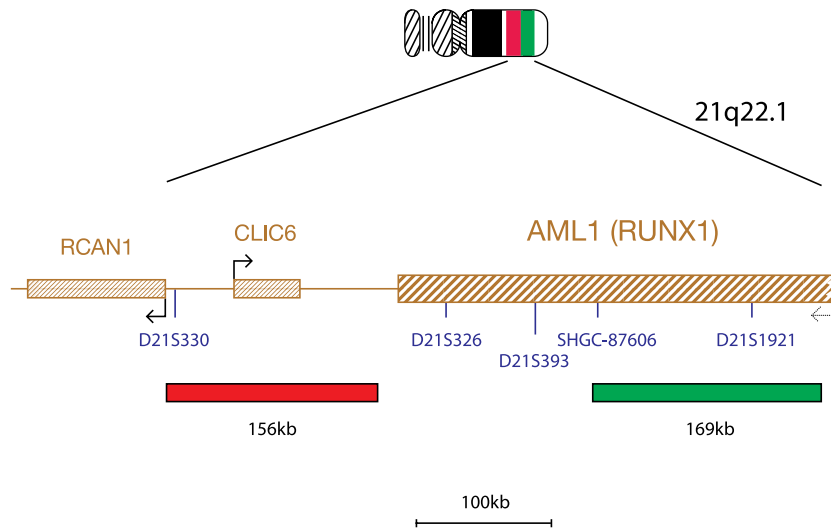
Reporting and interpretation of FISH results should be consistent with professional standards of practice and should take into consideration other clinical and diagnostic information. This kit is intended as an adjunct to other diagnostic laboratory tests and therapeutic action should not be initiated on the basis of the FISH result alone. Failure to adhere to the protocol may affect the performance and lead to false results. Each lab is responsible for establishing their own cut-off values. Each laboratory should test sufficiently large number of samples to establish normal population distribution of the signal levels and to assign a cut-off value. The product is for professional use only and is intended to be interpreted by a qualified Pathologist or Cytogeneticist. Product availability may vary from country to country and is subject to varying regulatory requirements.

AML1 (RUNX1) Breakapart**

The AML1 product consists of a 156kb probe, labeled in red, located centromeric to the AML1 (RUNX1) gene, including the CLIC6 gene and a 169kb green probe, covering part of the AML1 (RUNX1) gene, including markers SHGC-87606 and D21S1921.



Probe Specification
 AML1, 21q22.12, Red
 AML1, 21q22.12, Green



CMP-H003 v006.00

** For sale in Canada only.

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

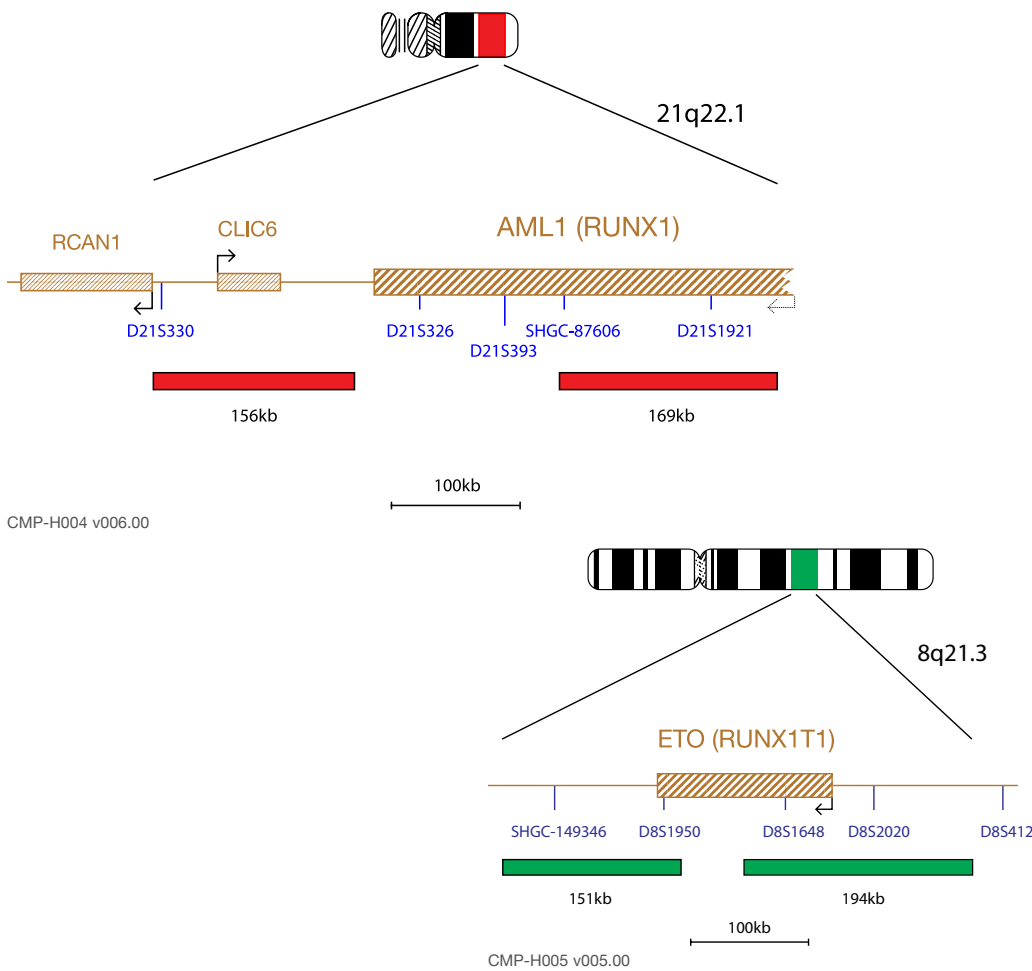


AML1/ETO (RUNX1/ RUNX1T1) Translocation, Dual Fusion**



The AML1 component consists of a 156kb probe, labeled in red, located centromeric to the AML1 (RUNX1) gene that spans the CLIC6 gene and a 169kb probe covering part of the AML1 (RUNX1) gene, including markers SHGC-87606 and D21S1921. The ETO (RUNX1T1) component, labeled in green, consists of a 151kb probe covering the centromeric part of the gene and the flanking region and a 194kb probe covering the telomeric part of the gene and the flanking region.

Probe Specification AML1, 21q22.1 Red
 ETO, 8q21.3, Green



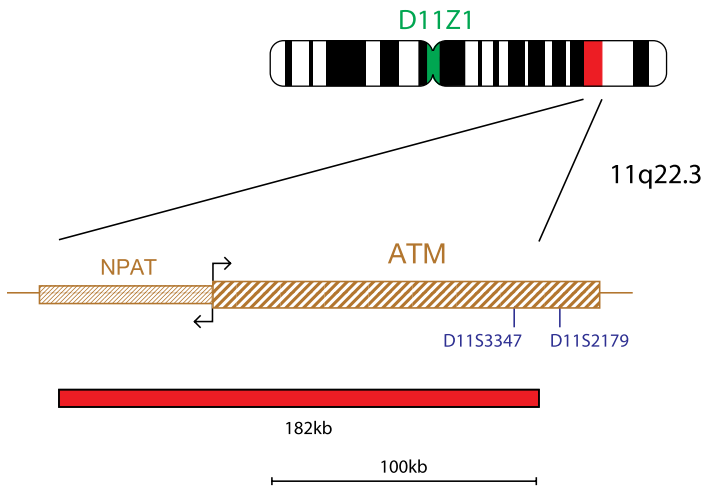
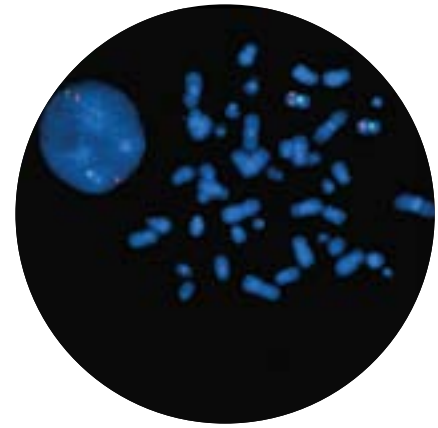
** For sale in Canada only.

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

ATM Deletion

The ATM probe is 182kb, labeled in red, and covers the telomeric end of the NPAT gene and the centromeric end of the ATM gene to just beyond the D11S3347 marker. The probe mix also contains a control probe for the 11centromere (D11Z1) labeled in green.

Probe Specification ATM, 11q22.3, Red
 D11Z1, 11p11.1-q11.1, Green

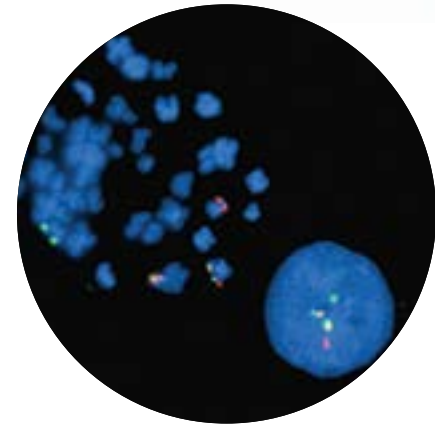


CMP-H006 v004.00

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

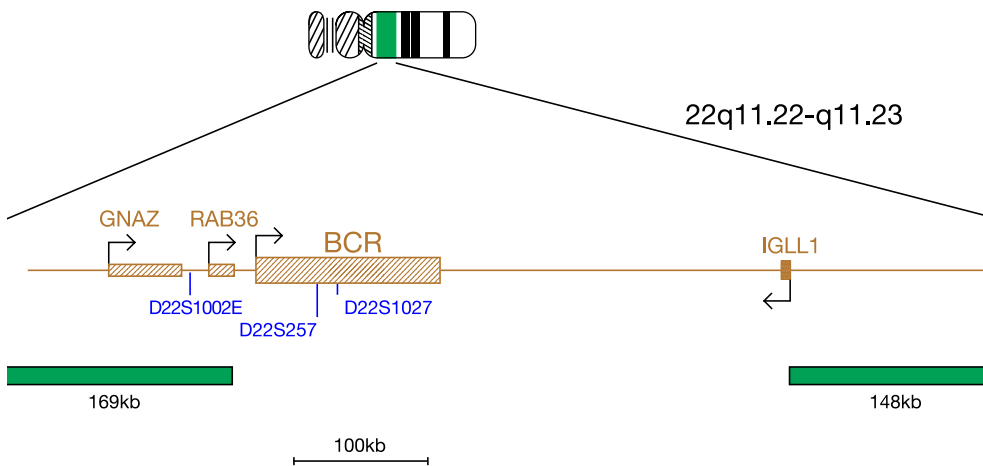


BCR/ABL (ABL1) Translocation, Dual Fusion

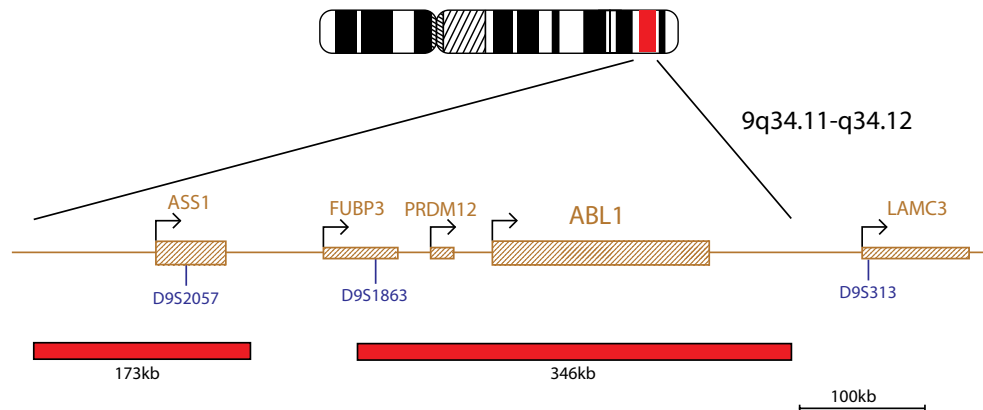


The BCR/ABL1 probe mix contains a 169kb green probe centromeric to the BCR gene and covers the genes GNAZ and RAB36. A second green probe covers a 148kb region that includes the telomeric end of the IGLL1 gene and the flanking region beyond. A red probe covers a 346kb region that includes the ABL1 gene. There is an additional red probe that covers a 173kb region and spans the whole ASS1 gene.

Probe Specification ABL1, 9q34.11-q34.12, Red
BCR, 22q11.22-q11.23, Green



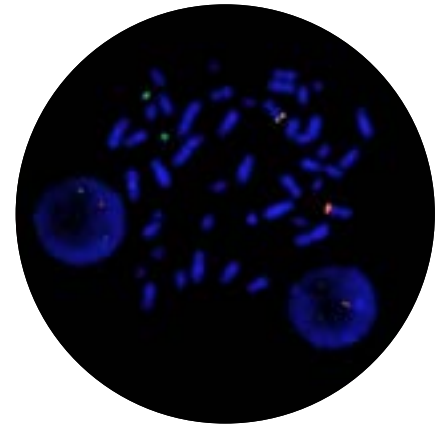
CMP-H008 v003.00



CMP-H009 v002.00

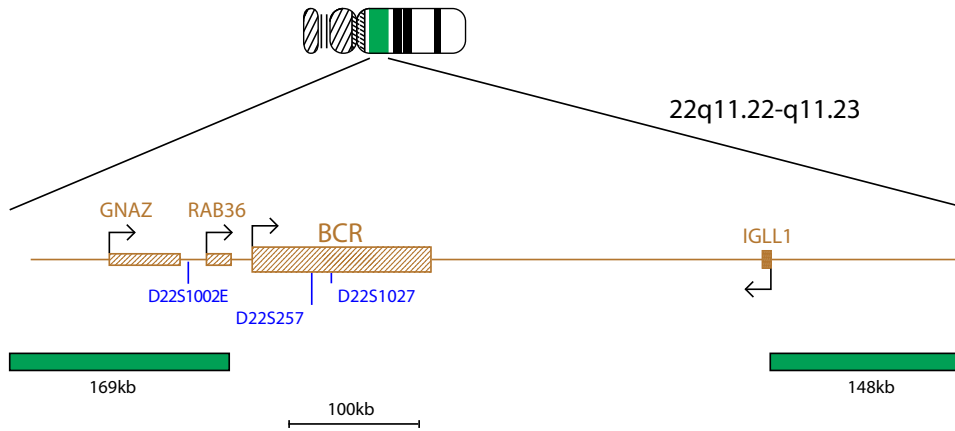
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

BCR/ABL (ABL1) *Plus* Translocation, Dual Fusion

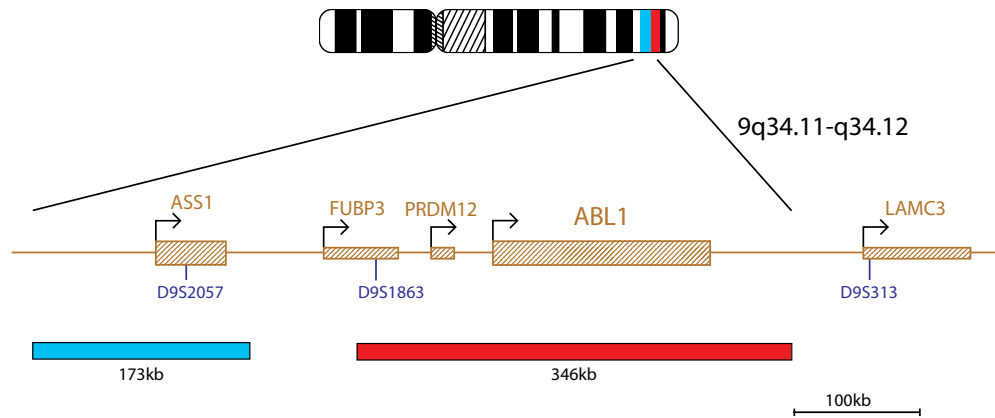


The BCR/ABL1 probe mix contains a 169kb green probe centromeric to the BCR gene and contains the genes GNAZ and RAB36. A second green probe covers a 148kb region telomeric to the BCR gene and covers part of the IGLL1 gene. A red probe covers a 346kb region that spans the ABL1 gene. There is an additional blue probe that covers a 173kb region and spans the whole ASS1 gene.

Probe Specification
 ABL1, 9q34.11-q34.12, Red
 BCR, 22q11.22-q11.23, Green
 ASS1, 9q34.11-q34.12, Blue



CMP-H008 vs003.00



CMP-H010 v002.00

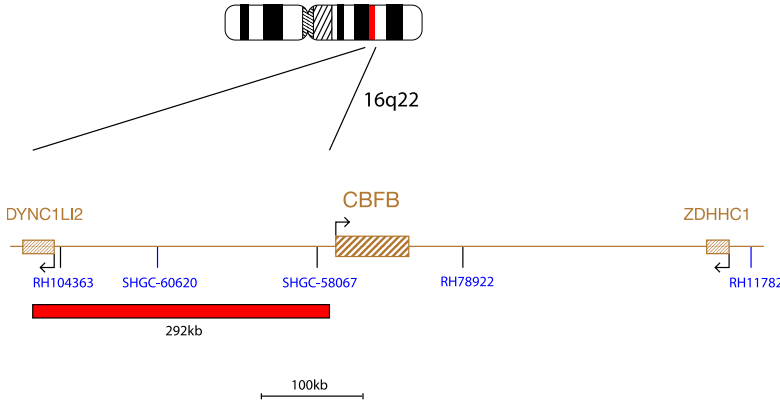
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



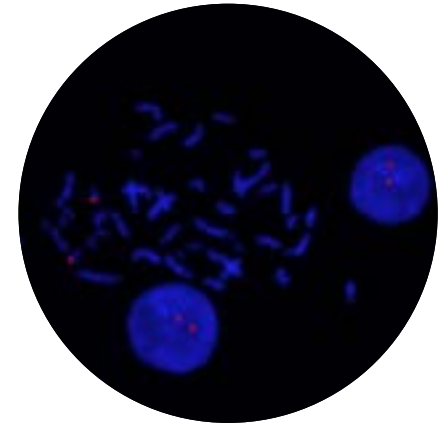
CBFB Proximal Probe Red

The CBFB Proximal probe, labeled in red, consists of a 292kb probe proximal to the CBFB gene, covering RH104363, SHGC-60620 and SHGC-58067 marker.

Probe Specification CBFB Proximal, 16q22.1, Red



CMP-H116 v003.00

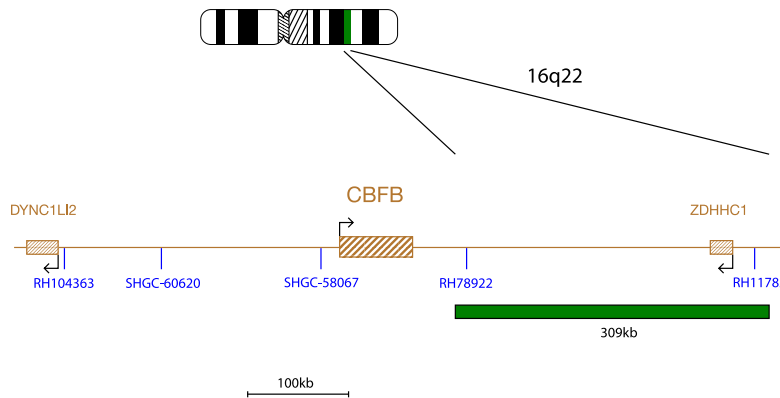


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

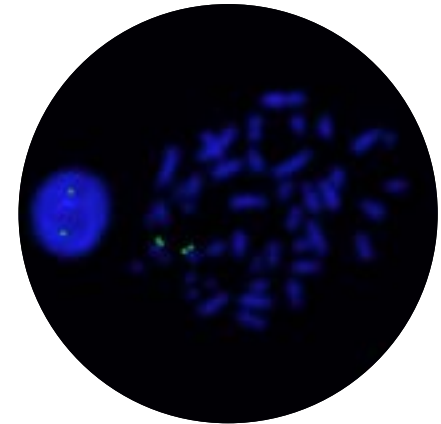
CBFB Distal Probe Green

The CBFB Distal probe, labeled in green, consists of a 309kb probe distal to the CBFB gene, covering RH78922 and RH11782 markers.

Probe Specification CBFB Distal, 16q22.1, Green



CMP-H117 v003.00



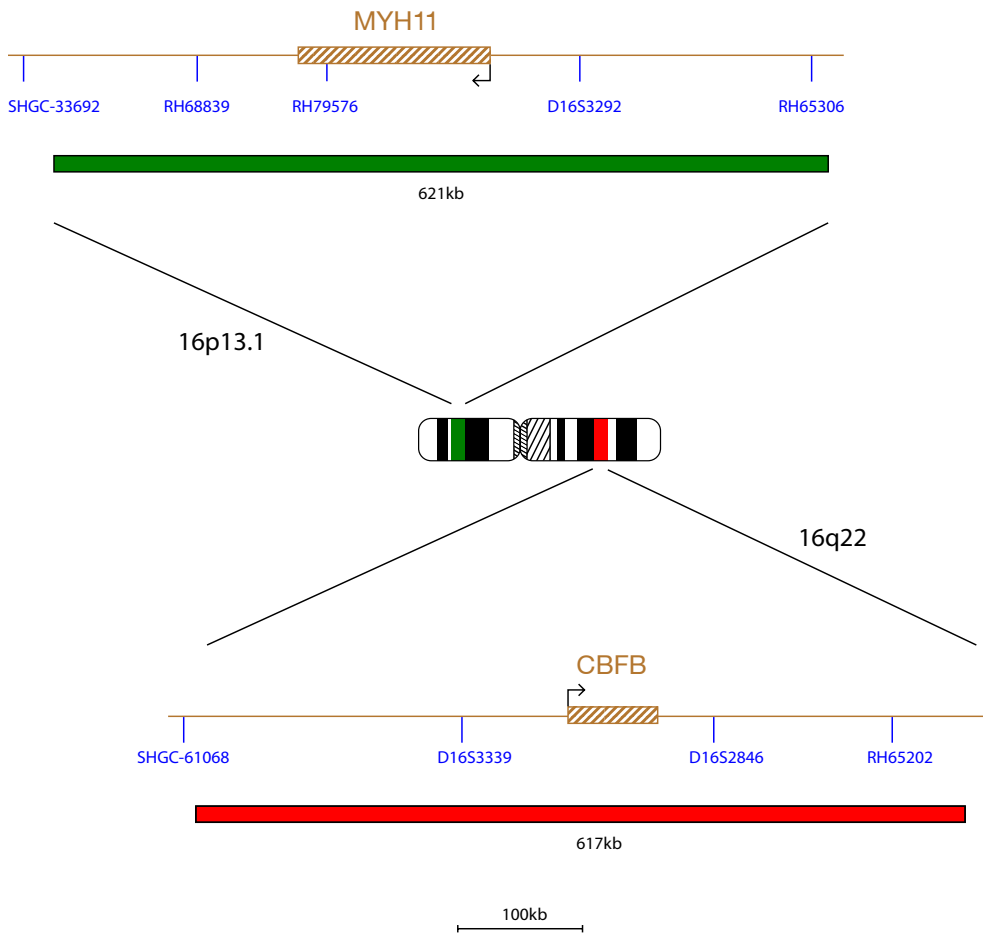
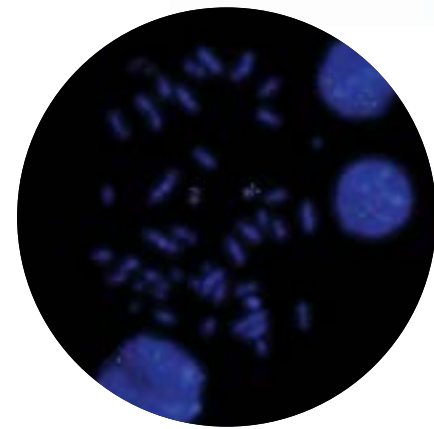
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



CBFβ/MYH11 Translocation, Dual Fusion**

The CBFβ probe, labeled in red, covers a 617kb region, within 16q22 including the CBFB gene. The MYH11 probe, labeled in green, covers a 621kb region within 16p13.1 including the MYH11 gene.

Probe Specification CBFβ, 16q22, Red
 MYH11, 16p13.1, Green

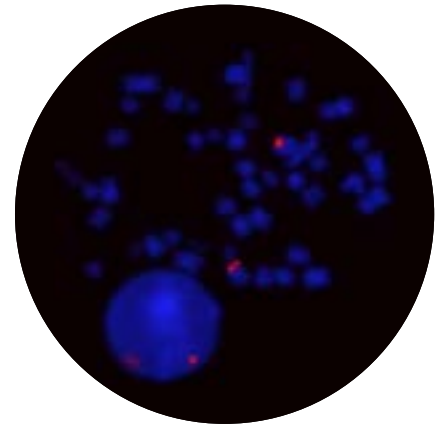


CMP-H077 v005.00

** For sale in Canada only.

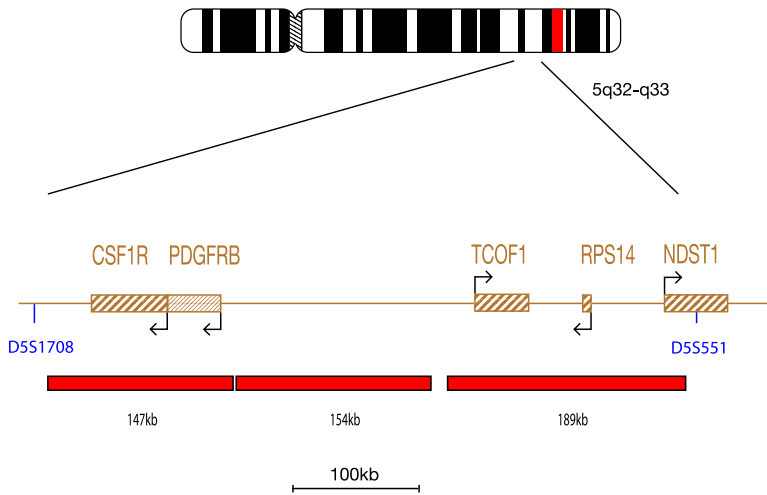
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

CSF1R/RPS14 (5q32-q33) Probe Red



The CSF1R/RPS14 (5q32-q33) probe mix, labeled in red, consists of a 147kb probe covering the CSF1R and PDGFRB genes and two probes (154kb and 189kb) covering the TCOF1 and RPS14 genes including the centromeric end of the NDST1 gene.

Probe Specification CSF1R/RPS14, 5q32-q33, Red



CMP-H133 v001.00

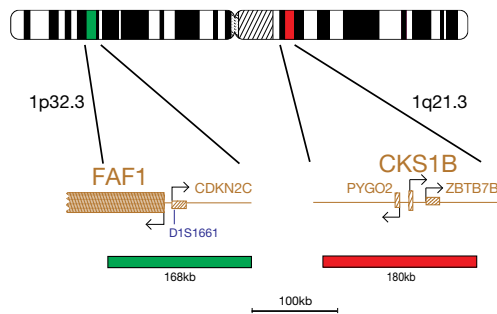
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



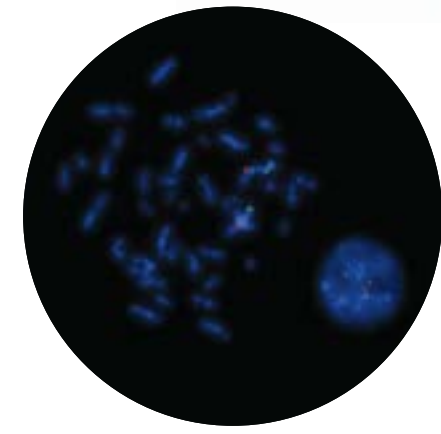
CKS1B/CDKN2C (P18) Amplification/Deletion

The CKS1B/CDKN2C (P18) product consists of a 180kb probe, labeled in red, covering the entire CKS1B gene and flanking regions, including the PYGO2 and ZBTB7B genes, and a green probe covering a 168kb region, including the entire CDKN2C gene, the D1S1661 marker and the centromeric end of the FAF1 gene.

Probe Specification CKS1B, 1q21.3, Red
CDKN2C (P18), 1p32.3, Green



CMP-H013 v002.00

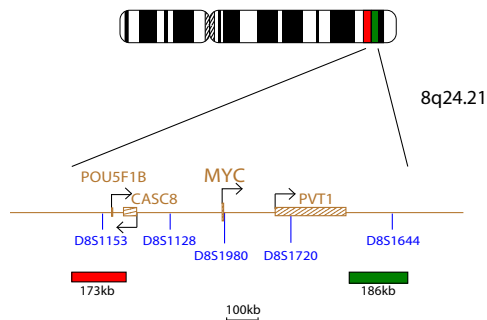


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

cMYC (MYC) Breakapart

The cMYC probe mix consists of a 173kb probe labeled in red, centromeric to the MYC gene that includes the marker D8S1153 and a 186kb probe, labeled in green telomeric to the MYC gene that includes the D8S1644 marker.

Probe Specification cMYC, 8q24.21, Red
cMYC, 8q24.21, Green



CMP-H014 v003.00

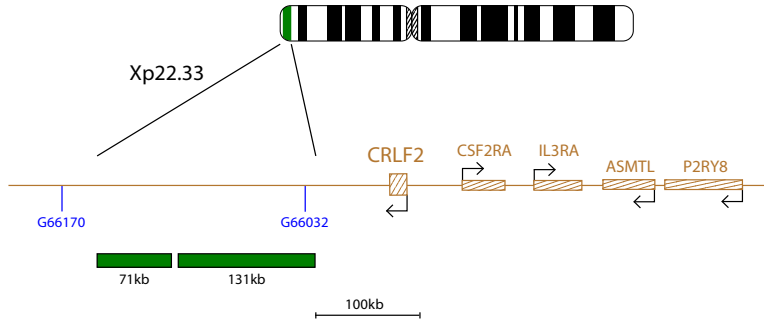


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

CRLF2 Distal

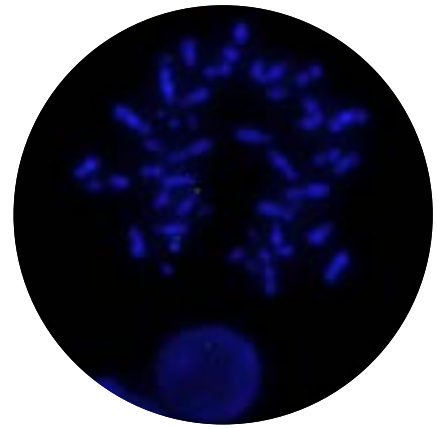
The CRLF2 Distal probe, labeled in green, consists of two probes (71kb and 131kb) that are situated distal (telomeric) to the CRLF2 gene.

Probe Specification CRLF2 Distal, Xp22.33/Yp11.32, Green



CMP-H091 v001.00

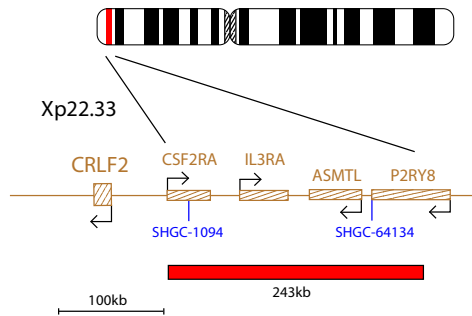
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



CRLF2 Proximal

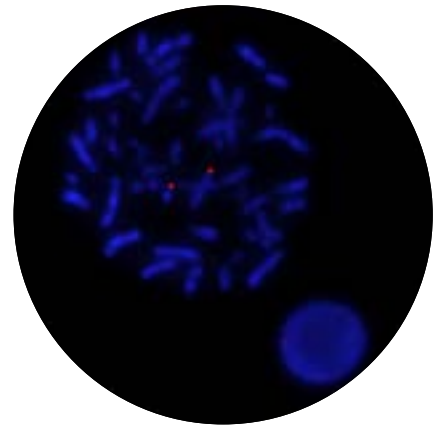
The CRLF2 Proximal probe, labeled in red, consists of a 243kb probe situated proximal (centromeric) to the CRLF2 gene.

Probe Specification CRLF2 Proximal, Xp22.33/Yp11.32, Red



CMP-H090 v002.00

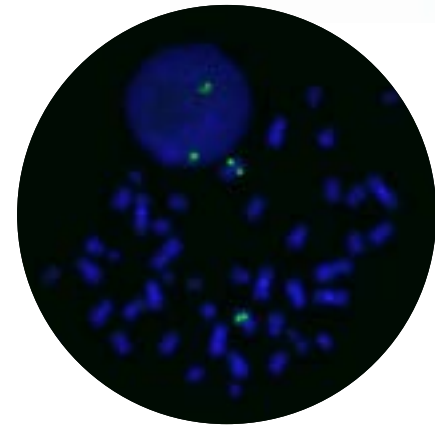
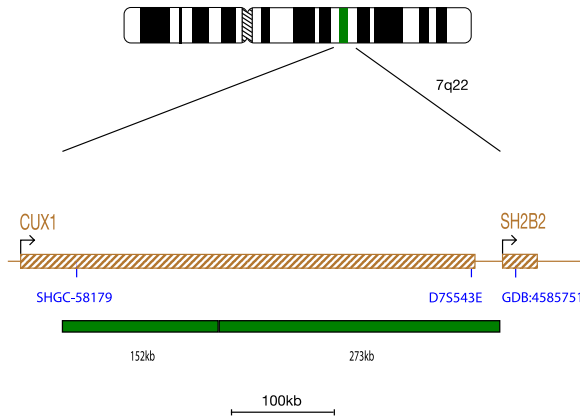
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



CUX1 (7q22) Probe Green

The CUX1 (7q22) probe mix, labeled in green, consists of two probes (152kb and 273kb) covering the telomeric end of the CUX1 gene, including markers SHGC-58179 and D7S543E.

Probe Specification CUX1, 7q22, Green



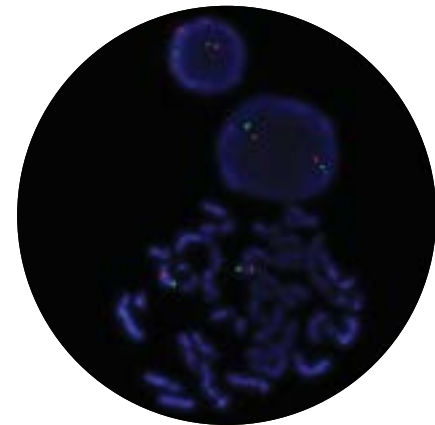
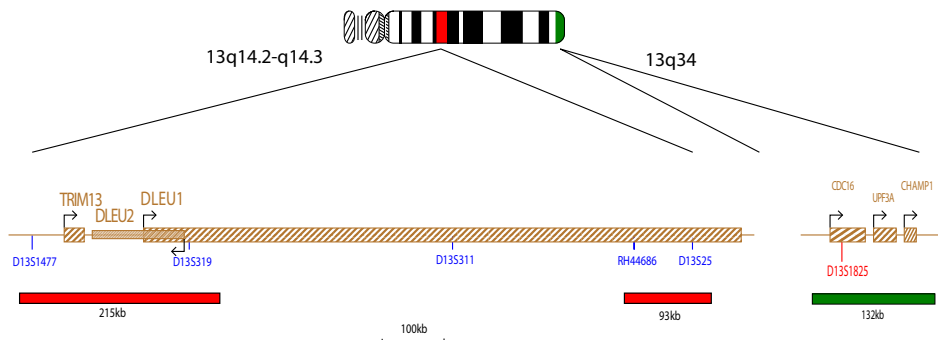
CMP-H130 v001.00

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

13q14.3 Deletion

The 13q14.3 probe, labeled in red, covers the D13S319 and D13S25 markers. The 13qter subtelomere specific probe (clone 163C9), labeled in green, allows identification of chromosome 13 and acts as a control probe.

Probe Specification 13q14.2-q14.3, Red
13qter, 13q34, Green

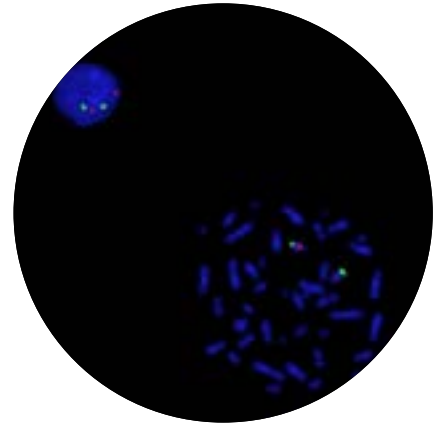


CMP-H001 v003.00

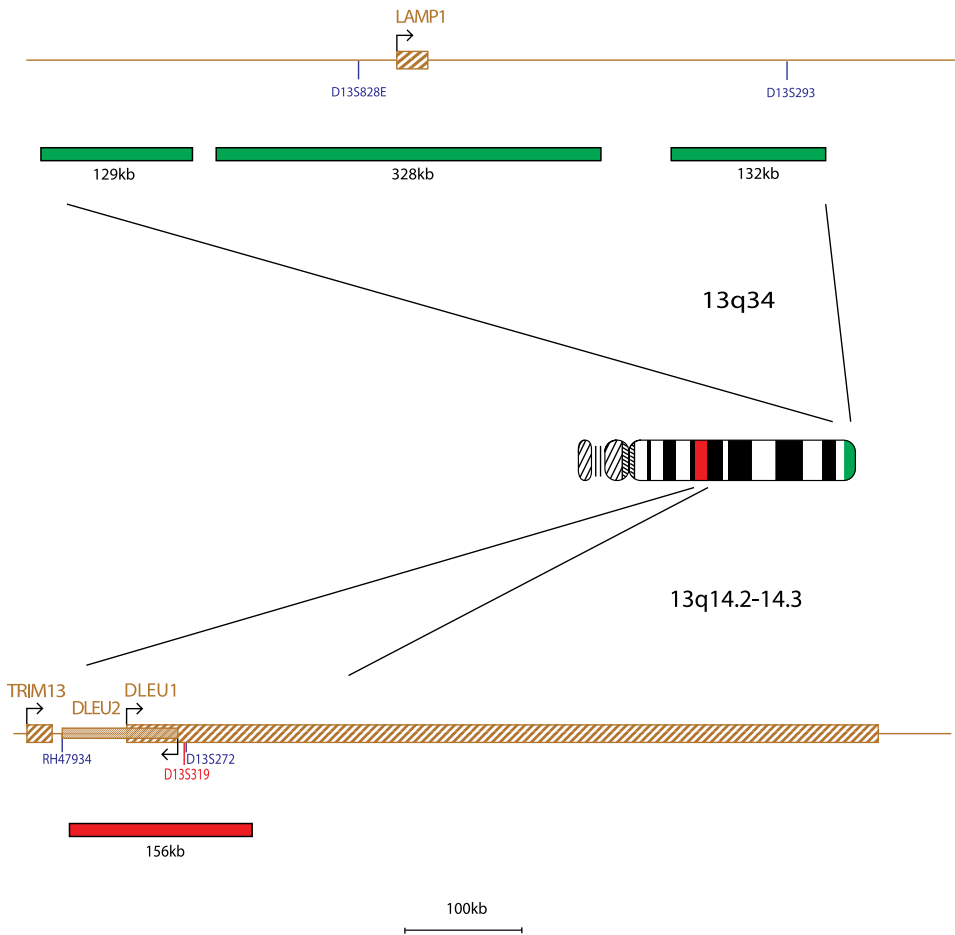
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

D13S319 *Plus* Deletion

The D13S319 probe, labeled in red, covers a 156kb region including most of the DLEU2 gene, part of the DLEU1 gene and the D13S319 and D13S272 markers. The 13qter subtetromere specific probe, labeled in green, allows identification of chromosome 13 and acts as a control probe.



Probe Specification D13S319, 13q14.2-14.3, Red
 13qter, 13q34, Green



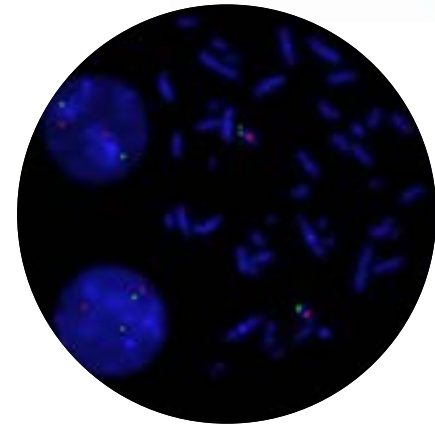
CMP-H079 v004

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

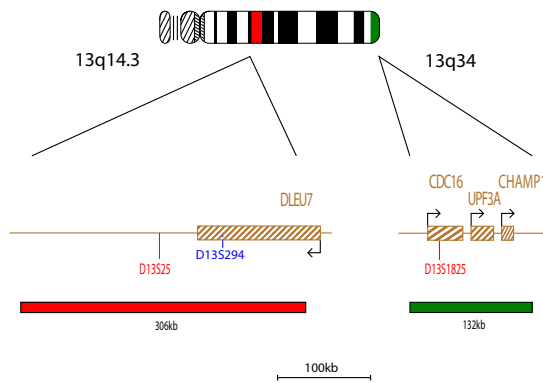


D13S25 Deletion

The D13S25 probe, labeled in red, covers a 306kb region including most of the DLEU7 gene and the D13S25 marker. The 13qter subtelomere specific probe (clone 163C9), labeled in green, allows identification of chromosome 13 and acts as a control probe.



Probe Specification D13S25, 13q14.3, Red
13qter, 13q34, Green

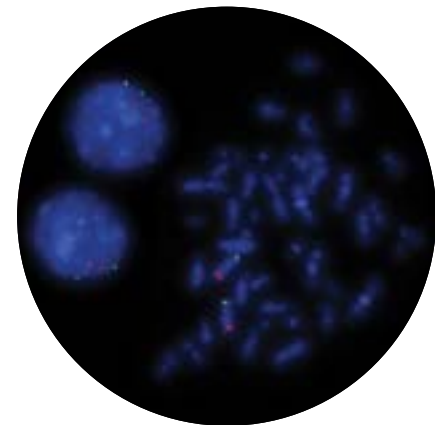


CMP-H016 v004

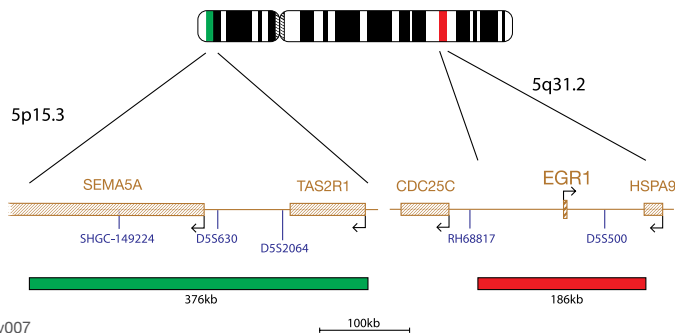
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

Del(5q) Deletion**

The EGR1 probe, labeled in red, covers a 186kb region within 5q31.2 that includes the D5S500 marker. The probe mix also contains a control probe, labeled in green for chromosome 5 at 5p15.3 that includes the marker D5S630.



Probe Specification EGR1, 5q31.2, Red
5p15.3, Green



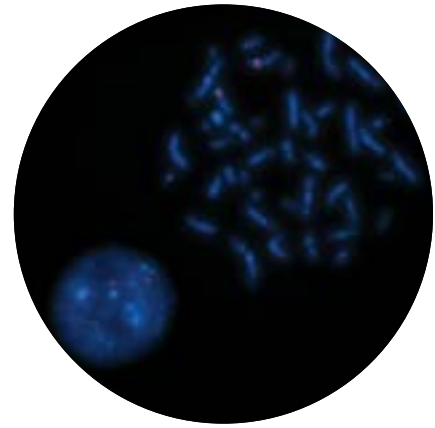
CMP-H017 v007

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

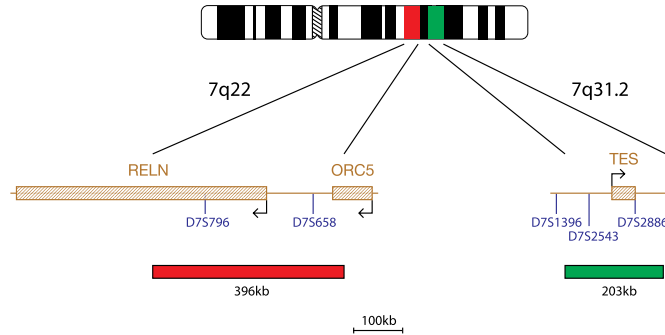
** For sale in Canada only.

Del(7q) Deletion**

The Del (7q) probe mix consists of a 7q22 probe, labeled in red that covers a 396kb region including the telomeric end of the RELN gene to beyond the marker D7S658. In addition a 7q31.2 probe, labeled in green covers a 203kb region that spans the TES gene.



Probe Specification 7q22, Red
7q31.2, Green



CMP-H018 v006

** For sale in Canada only.

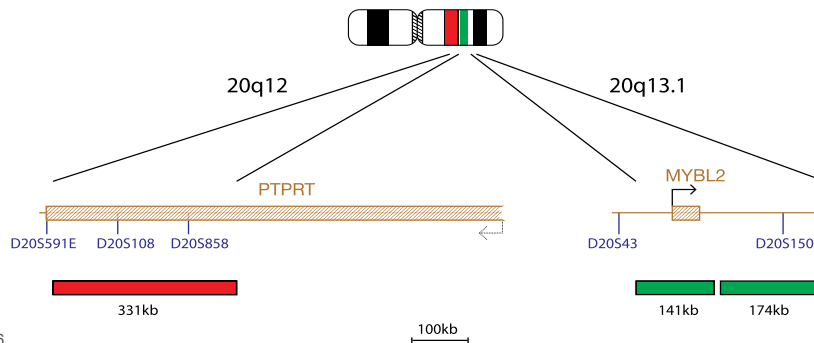
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

Del(20q) Deletion**

The 20q12 probe, labeled in red, covers a 331kb region within the PTPRT gene and includes the D20S108 marker. The 20q13.1 probes, labeled in green (141kb and 174kb), cover the MYBL2 gene and includes the D20S150 marker.



Probe Specification 20q12, Red
20q13.1, Green



CMP-H019 v006

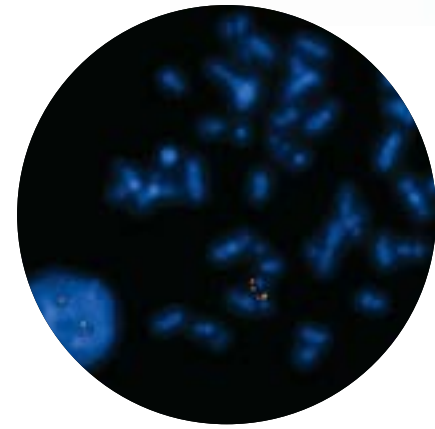
** For sale in Canada only.

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

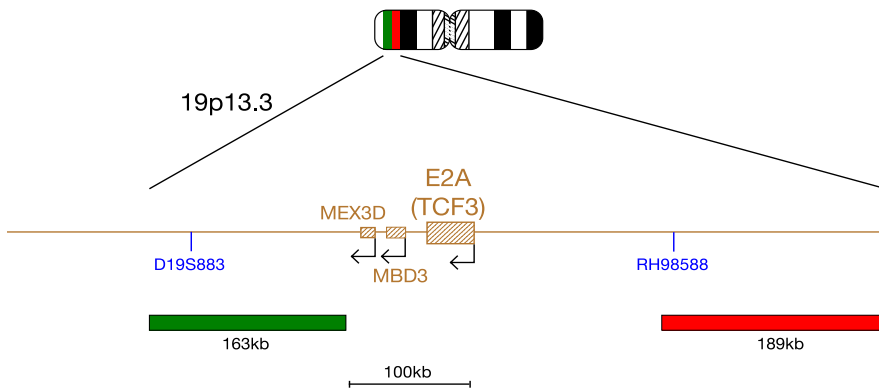


E2A (TCF3) Breakapart

The E2A (TCF3) product consists of a 189kb probe, labeled in red, located centromeric to the TCF3 gene, including the RH98588 marker, and a green probe covering a 163kb region located telomeric to the TCF3 gene, including the D19S883 marker.



Probe Specification E2A, 19p13.3, Red
E2A, 19p13.3, Green



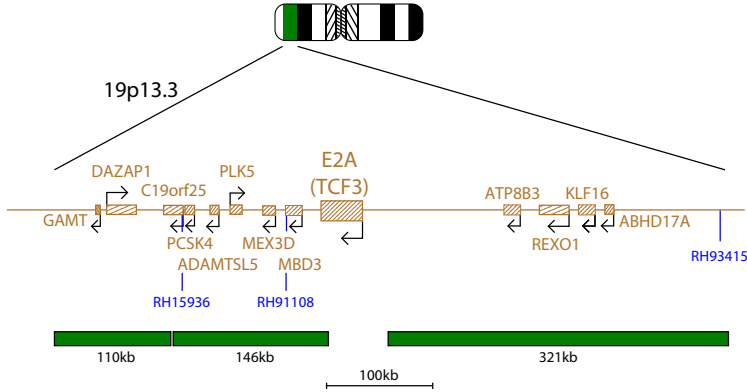
CMP-H020 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

E2A (TCF3)

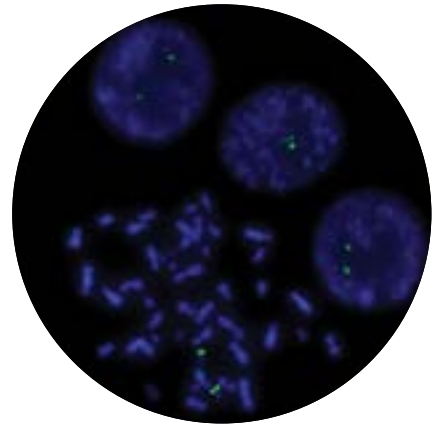
The E2A (TCF3) probe, labeled in green, contains two probes (110kb and 146kb) that cover the 3' end of the TCF3 gene and flanking region and a 321kb probe that covers a region 5' (centromeric) to the gene.

Probe Specification E2A, 19p13.3, Green



CMP-H080 v001

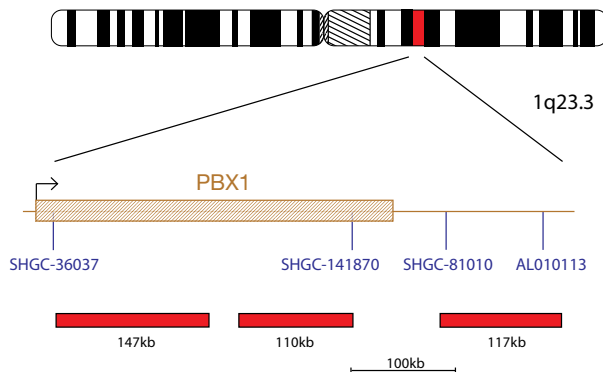
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



PBX1

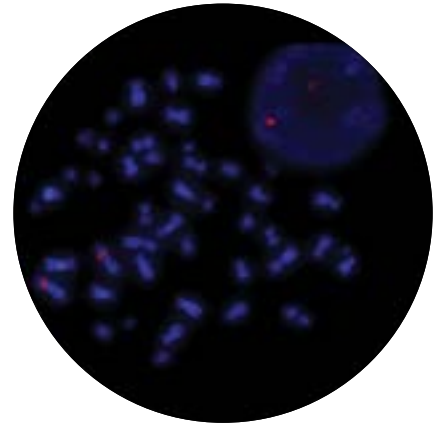
The PBX1 probe, labeled in red, contains two probes (147kb and 110kb) that map within the PBX1 gene and a 117kb probe that maps 3' (telomeric) to the gene.

Probe Specification PBX1, 1q23.3, Red



CMP-H081 v003

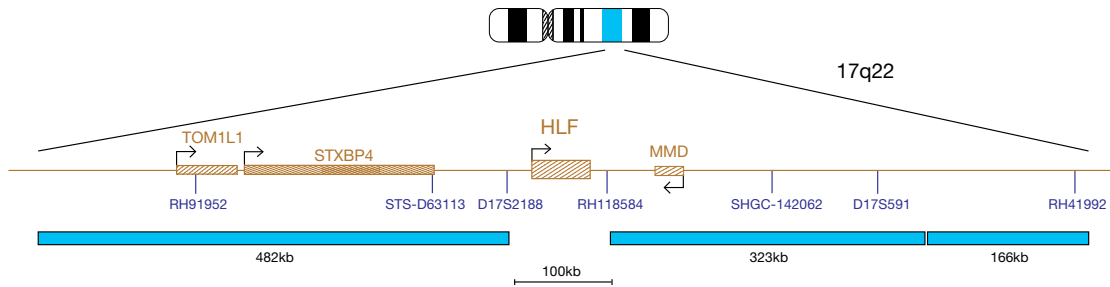
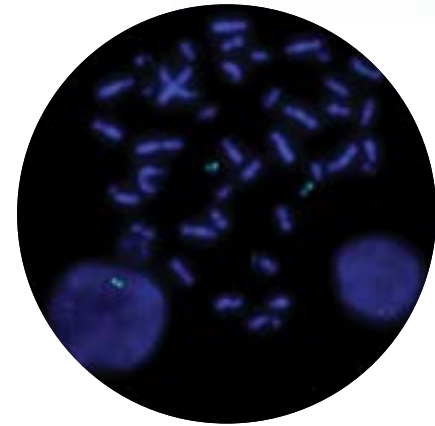
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



HLF

The HLF probe, labeled in blue, consists of a 482kb clone 5' (centromeric) to HLF and two clones (323kb and 166kb) that are 3' of the gene.

Probe Specification HLF, 17q22, Blue



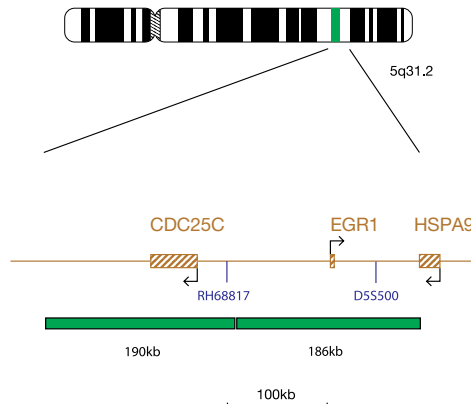
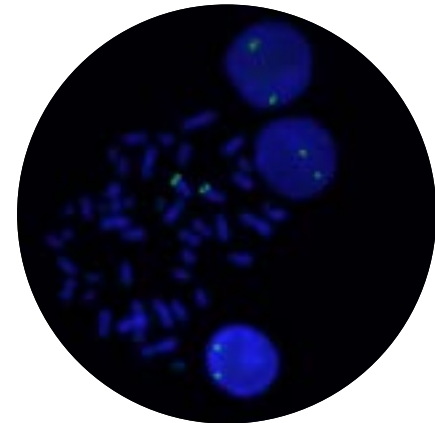
CMP-H082 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

EGR1/CDC25C (5q31) Probe Green

The EGR1/CDC25C (5q31) probe mix, labeled in green, consists of a 190kb probe covering the CDC25C gene and a 186kb probe covering the EGR1 gene, including marker D5S500.

Probe Specification EGR1/CDC25C, 5q31, Green



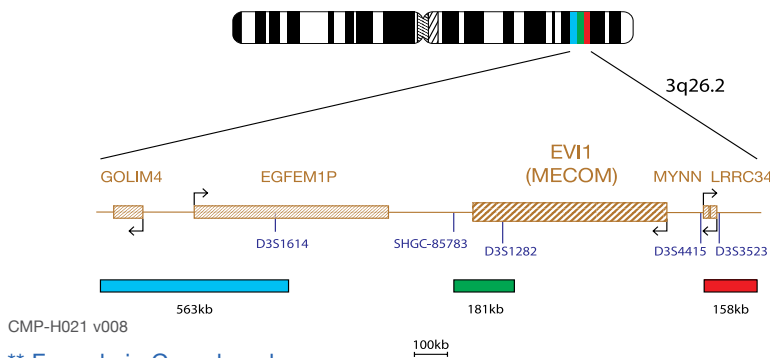
CMP-H132 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

EVI1 (MECOM) Breakapart**

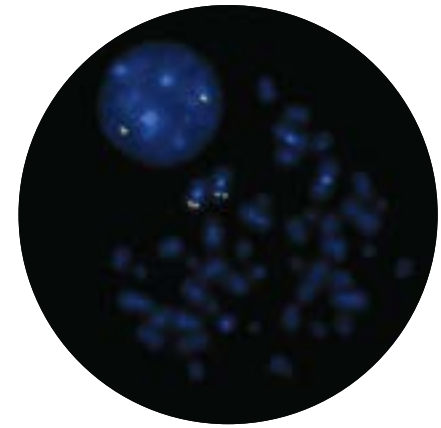
The red component of the EVI1 probe mix consists of a 158kb probe telomeric to the D3S4415 marker and includes the LRRC34 gene. The green component covers a 181kb region that includes the centromeric part of the EVI1 (MECOM) gene and beyond marker D3S1282. The blue component covers a 563kb region centromeric to the EVI1 gene, that includes the D3S1614 marker.

Probe Specification
 EVI1, 3q26.2, Red
 EVI1, 3q26.2, Green
 EVI1, 3q26.2, Blue



CMP-H021 v008

** For sale in Canada only.

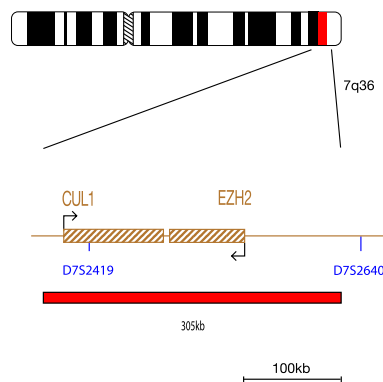


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

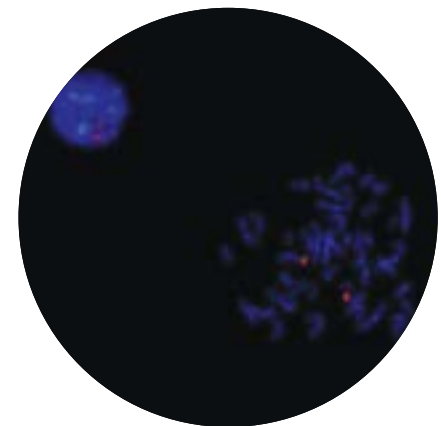
EZH2 (7q36) Probe Red

The EZH2 (7q36) probe, labeled in red, covers a 305kb region, including the EZH2 and CUL1 genes and the D7S2419 marker.

Probe Specification
 EZH2, 7q36, Red



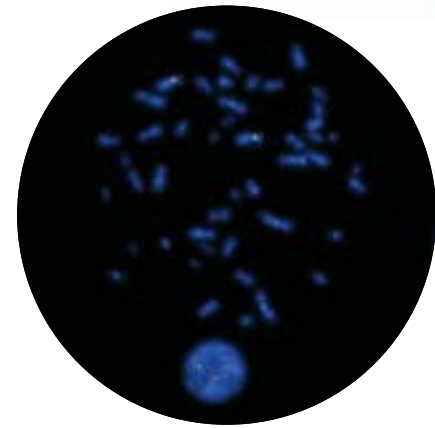
CMP-H129 v001



ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

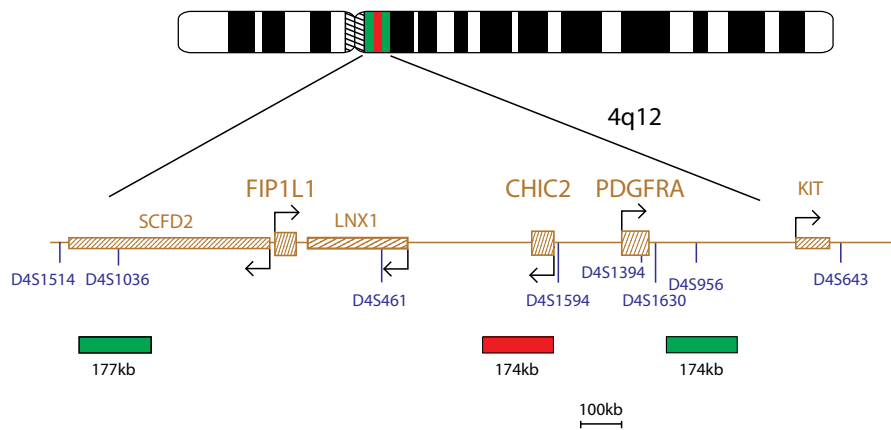


FIP1L1/CHIC2/PDGFR A Deletion/Fusion



The FIP1L1/CHIC2/PDGFR A product consists of a 177kb probe, labeled in green, located centromeric to the FIP1L1 gene, including the D4S1036 marker, a 174kb probe, labeled in red covering the CHIC2 gene and a 174kb probe, labeled in green, located telomeric to the PDGFR A gene, including the D4S956 marker.

Probe Specification	FIP1L1, 4q12, Green
	CHIC2, 4q12, Red
	PDGFR A, 4q12, Green



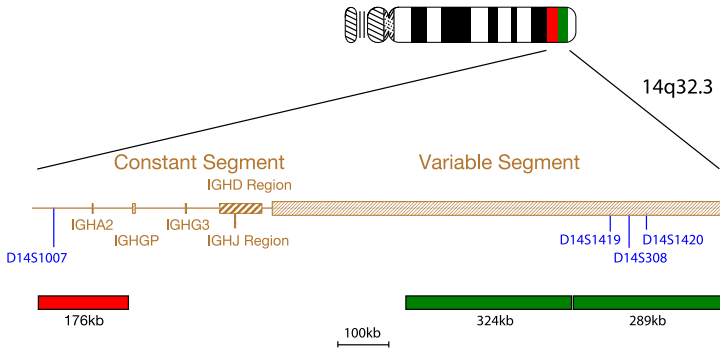
CMP-H022 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

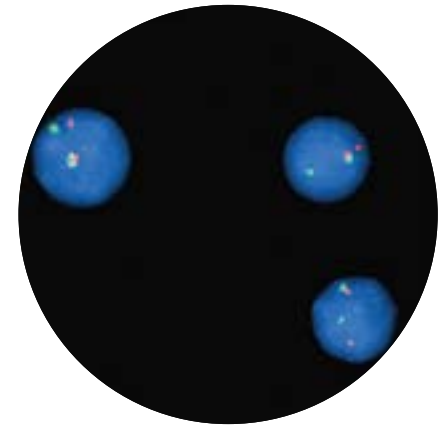
IGH Breakapart

The IGH probe mix consists of a 176kb probe, labeled in red, covering part of the Constant region of the gene and two green probes (324kb and 289kb), covering part of the Variable segment of the gene.

Probe Specification IGHC, 14q32.3, Red
IGHV, 14q32.3, Green



CMP-H023 v003

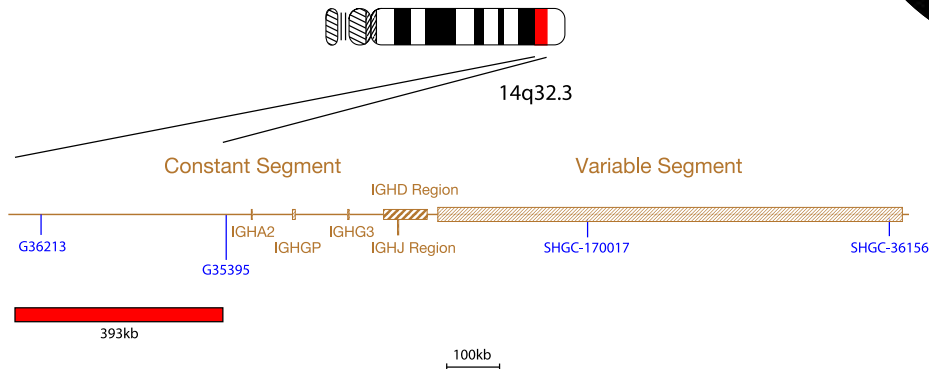


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

IGH Proximal Plus

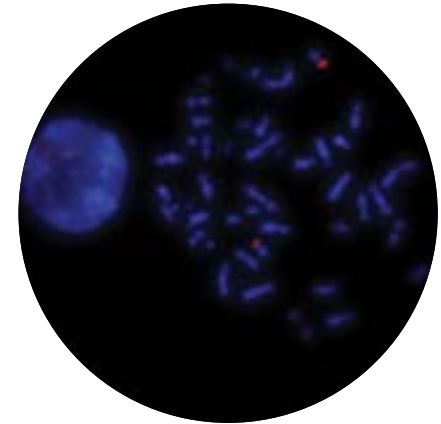
The IGH Proximal Plus product consists of a 393kb probe, labeled in red, proximal to the IGH Constant region.

Probe Specification IGHC, 14q32.33, Red



CMP-H095 v002

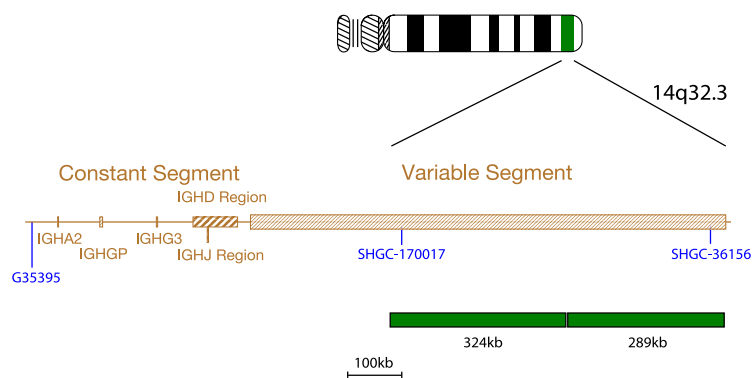
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



IGH Distal Plus

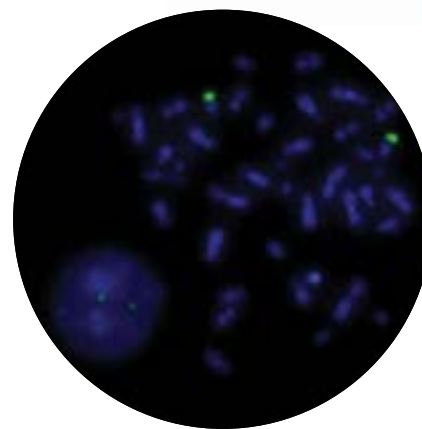
The IGH Distal Plus product consists of a 324kb probe and a 289kb probe within the Variable segment of the IGH region.

Probe Specification IGHV, 14q32.33, Green



CMP-H094 v002

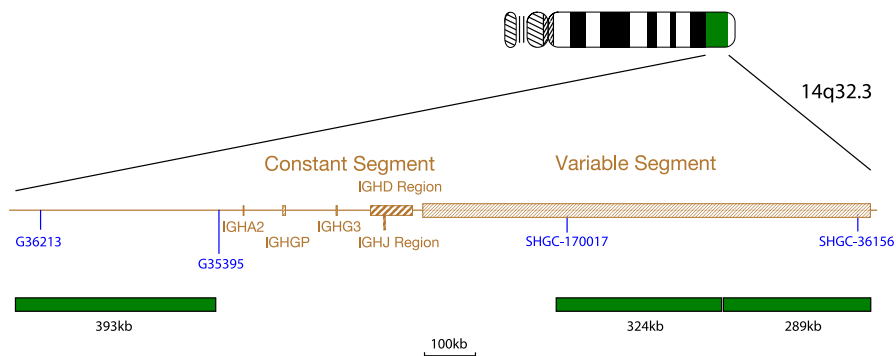
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



IGH Plus

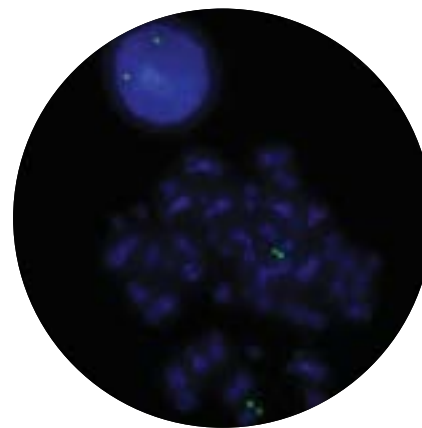
The IGH Plus product consists of probes, labeled in green, proximal to the Constant, and within the Variable segment of the IGH region.

Probe Specification IGH, 14q32.33, Green



CMP-H078 v002

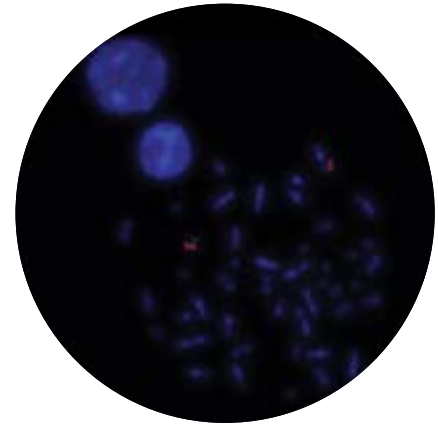
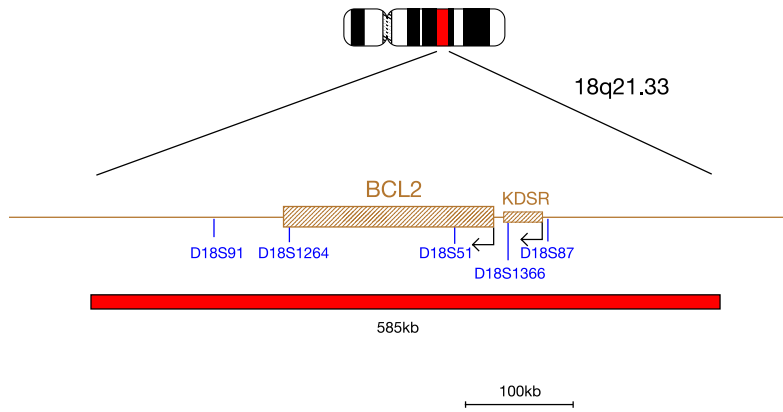
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



BCL2 Plus

The BCL2 Plus product consists of a 585kb probe, labeled in red, covering the BCL2 and KDSR genes.

Probe Specification BCL2, 18q21.33, Red



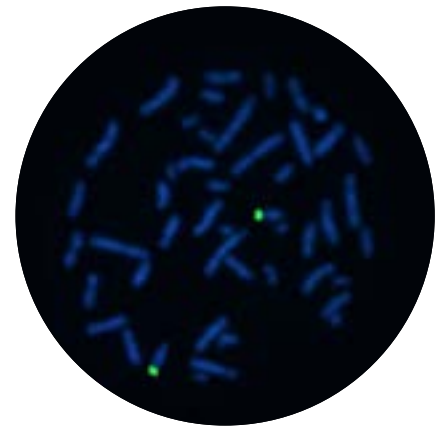
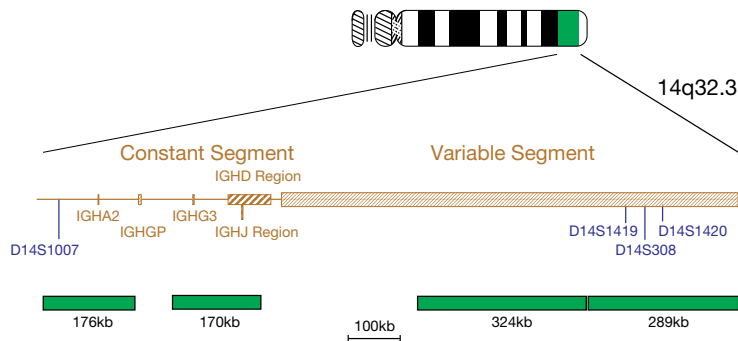
CMP-H026 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

IGH Probe Green

The IGH probe mix, labeled in green, has probes that cover parts of the Constant, J, D and Variable segments of the IGH gene.

Probe Specification Green, 14q32.3, Green



CMP-H024 v003

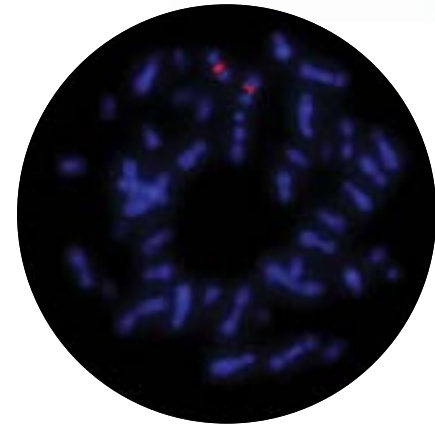
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



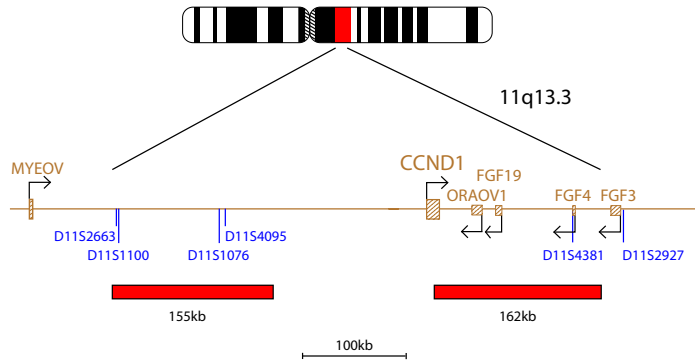


CCND1 Plus

The CCND1 *Plus* product consists of a 155kb probe centromeric to the CCND1 gene, covering a region between the D11S2663 and D11S4095 markers, and a second probe (162kb) covering the telomeric end of CCND1 gene and the region up to the FGF3 gene.



Probe Specification CCND1, 11q13.3, Red

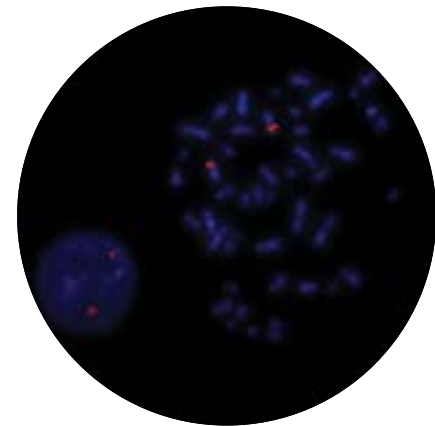


CMP-H027 v003

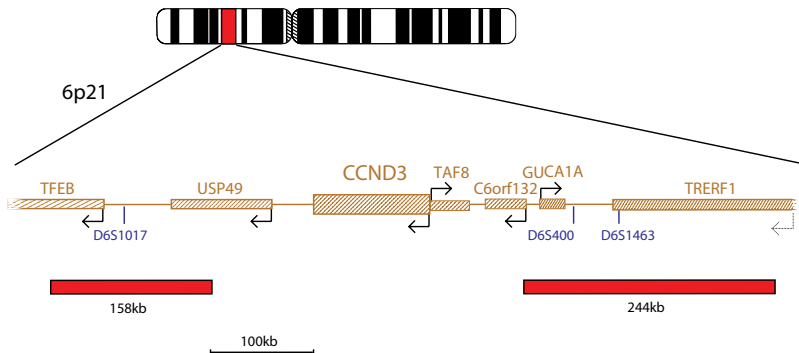
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

CCND3 Plus

The CCND3 *Plus* product consists of a 158kb probe telomeric to the CCND3 gene, including the D6S1017 marker, and a second probe covering the 244kb region centromeric to the TAF8 gene, including the D6S400 and D6S1463 markers.



Probe Specification CCND3, 6p21, Red



CMP-H028 v002

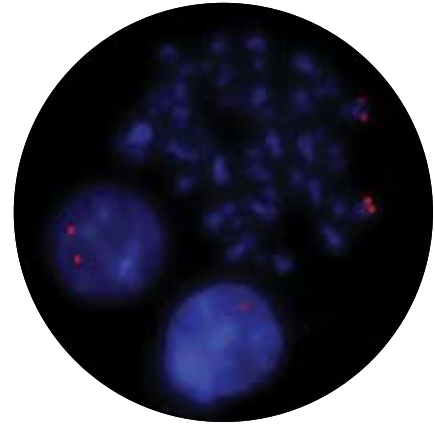
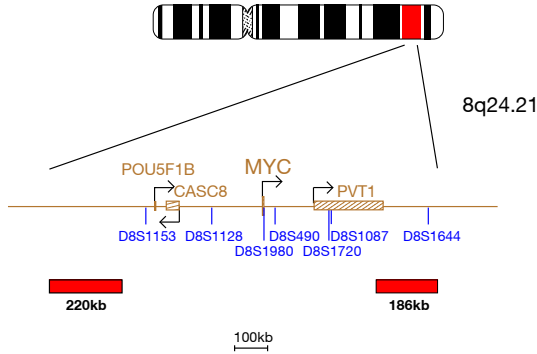
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



cMYC (MYC) Plus

The cMYC (MYC) Plus product consists of a 220kb probe centromeric to the cMYC (MYC) gene and a second probe covering the 186kb region telomeric to the cMYC (MYC) gene, including the D8S1644 marker.

Probe Specification cMYC (MYC), 8q24.21, Red



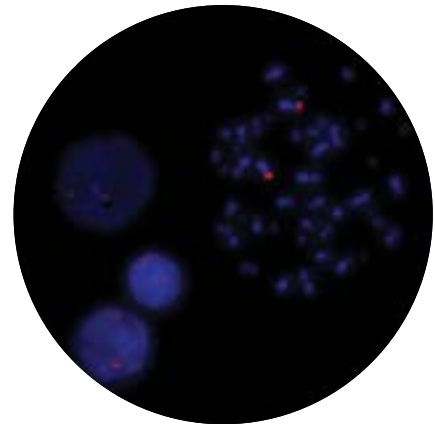
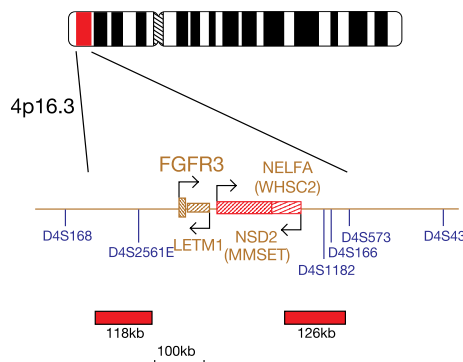
CMP-H029 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

FGFR3 Plus

The FGFR3 Plus product consists of a 118kb probe telomeric to FGFR3, including the D4S2561E marker and a second probe covering the 126kb region centromeric to MMSET, including the D4S1182 marker.

Probe Specification FGFR3, 4p16.3, Red



CMP-H030 v004

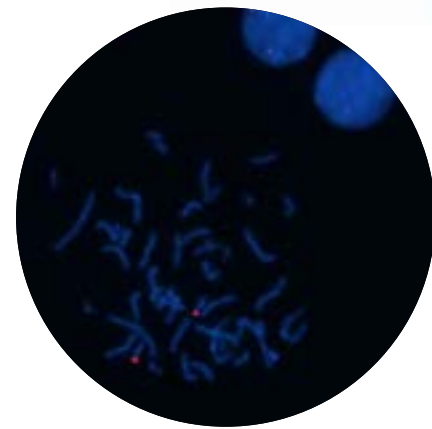
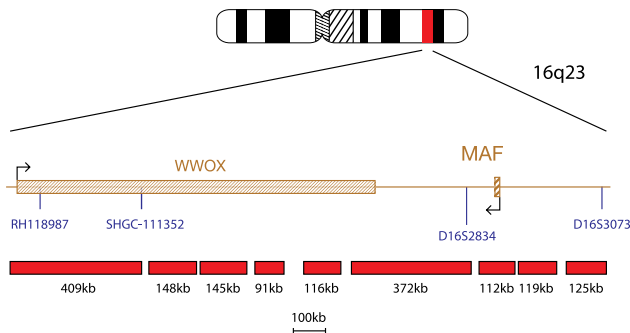
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



MAF v2 Probe Red

The MAF probe mix, labeled in red, encompasses the MAF gene and flanking regions as well as the WWOX gene.

Probe Specification MAF, 16q23, Red



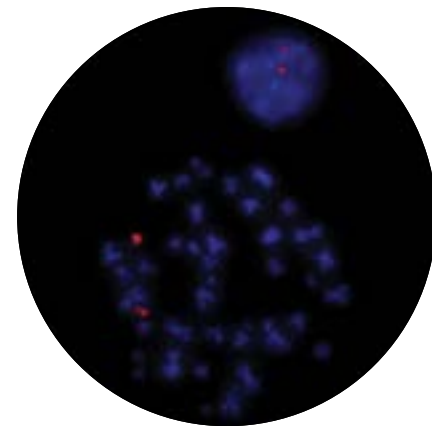
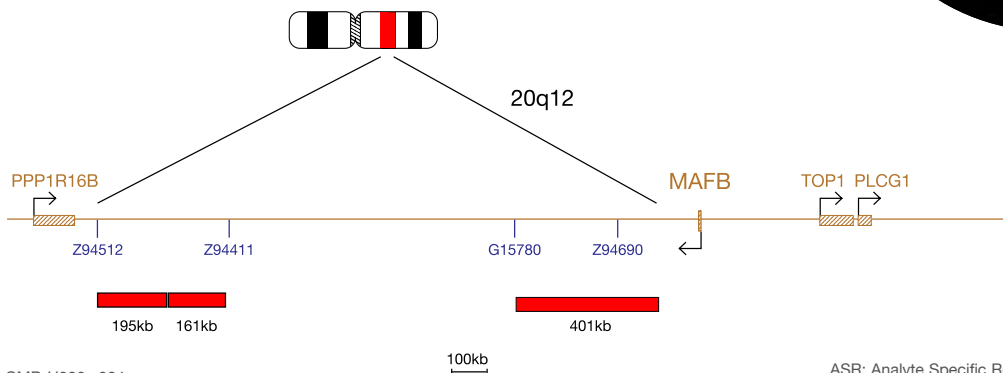
CMP-H139 v001

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

MAFB Plus

The MAFB Plus product consists of three probes (195kb, 161kb and 401kb), labeled in red. The MAFB probes are located on either side of the breakpoint region (between MAFB and PPP1R16B (WWC1)).

Probe Specification MAFB, 20q12, Red

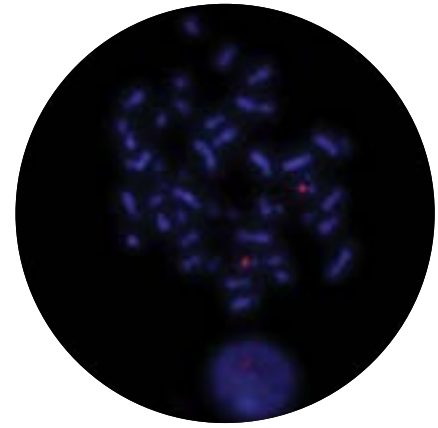


CMP-H032 v004

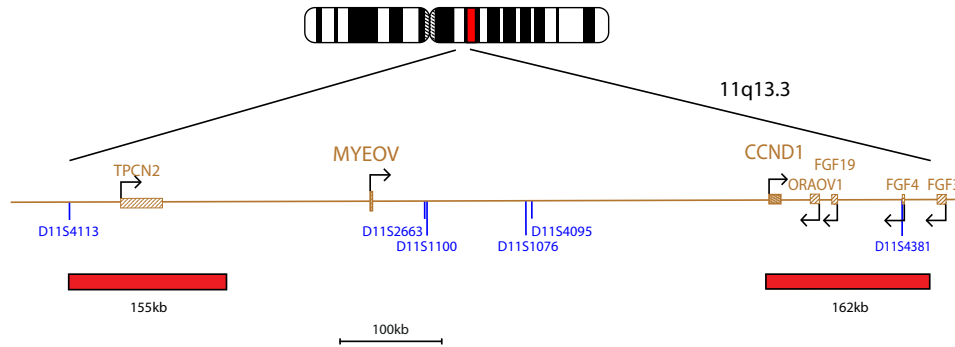
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

MYEOV Plus

The MYEOV *Plus* product consists of a 155kb probe centromeric to the MYEOV gene, which includes the TPCN2 gene, and a second probe, telomeric to the MYEOV gene, covering the 162kb region including the CCND1 and ORAOV1 genes.



Probe Specification MYEOV, 11q13.3, Red

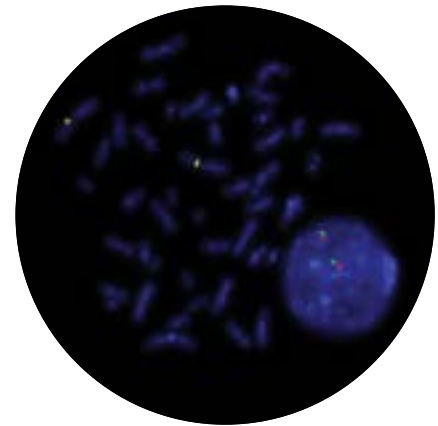


CMP-H033 v003

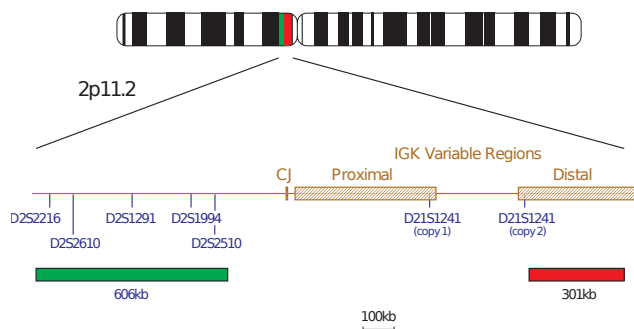
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

IGK Breakapart

The IGK product consists of a 301kb probe, labeled in red, covering a part of the distal IGK Variable region and a green probe, covering a 606kb region telomeric to the Joining segments and the Constant segment of IGK. The green probe extends from a position that is telomeric to the D2S2216 marker and continues to a position that is centromeric to the D2S2510 marker.



Probe Specification IGK, 2p11.2, Red
IGK, 2p11.2, Green



CMP-H034 v005

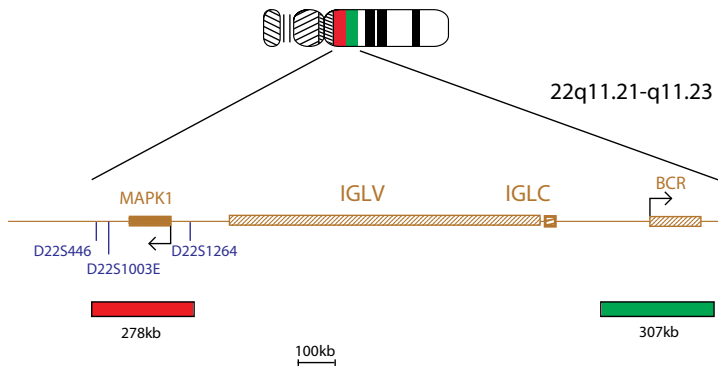
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



IGL Breakapart

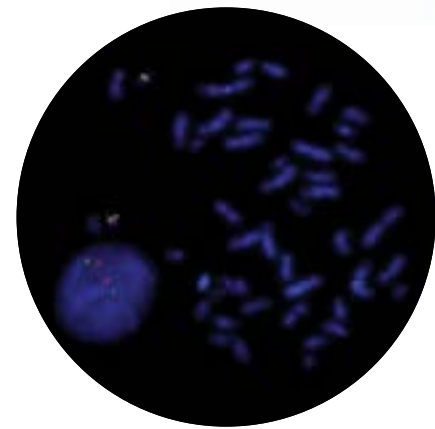
The IGL product consists of a 278kb probe, labeled in red, centromeric to the IGL Variable region and covering the MAPK1 gene, and a green probe, covering a 307kb region telomeric to the IGL Constant segment, including the BCR gene.

Probe Specification
 IGL, 22q11.21-q11.23, Red
 IGL, 22q11.21-q11.23, Green



CMP-H035 v004

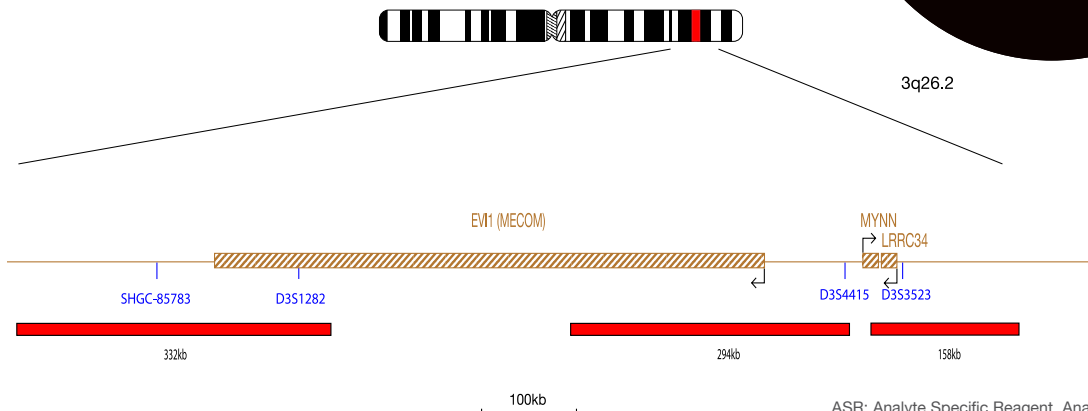
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



MECOM Probe Red

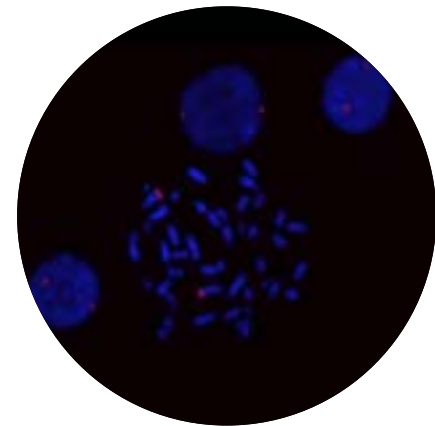
The MECOM probe mix, labeled in red, contains a 332kb probe covering the MECOM gene, including markers SHGC-85783 and D3S1282 and two probes (294kb and 158kb) telomeric to the MECOM gene.

Probe Specification
 3q26.2, Red



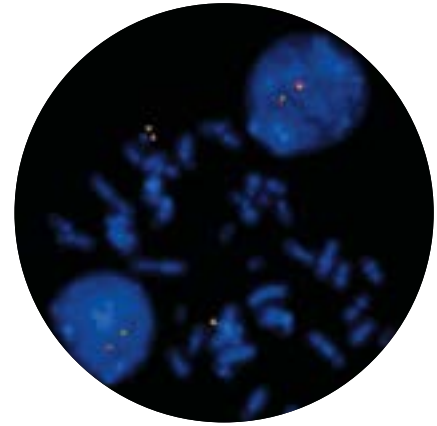
CMP-H099 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

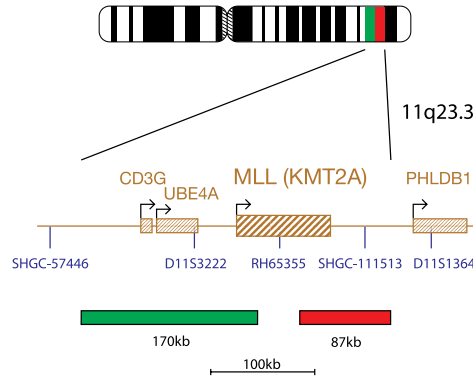


MLL (KMT2A) Breakapart**

The MLL (KMT2A) probe mix consists of an 87kb probe, labeled in red that covers the telomeric end of the MLL (KMT2A) gene and includes the marker SHGC-111513. In addition, a 170kb probe labeled in green covers the centromeric end of the MLL (KMT2A) gene and spans the CD3G and UBE4A genes.



Probe Specification
MLL, 11q23.3, Red
MLL, 11q23.3, Green

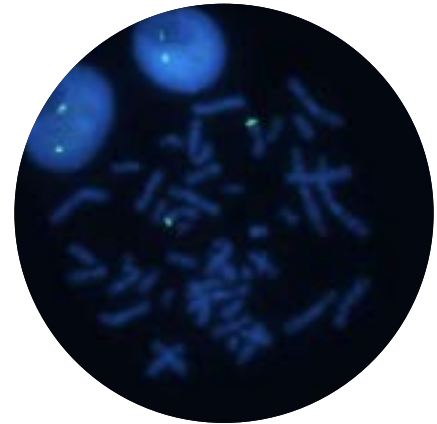


CMP-H036 v006

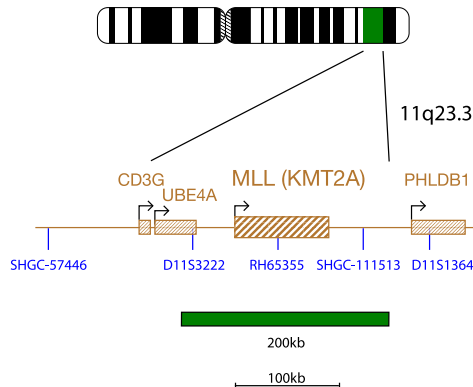
** For sale in Canada only.

MLL (KMT2A)

The MLL (KMT2A) probe, labeled in green, covers a 200kb region including the MLL (KMT2A) gene.



Probe Specification
MLL, 11q23.3, Green



CMP-H083 v003

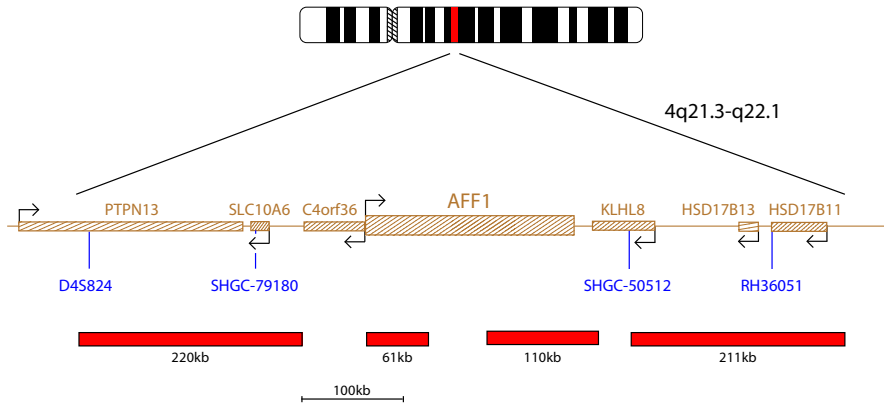
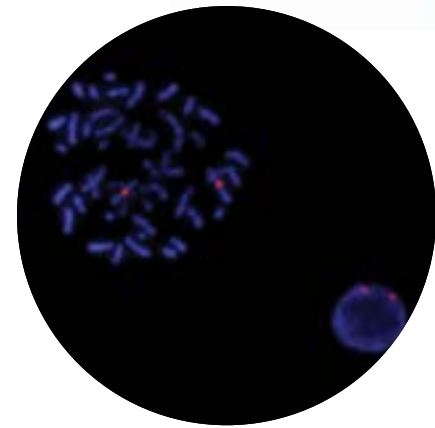
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



AFF1

The AFF1 probe, labeled in red, consists of four clones (220kb, 61kb, 110kb and 211kb) covering the AFF1 gene and surrounding regions.

Probe Specification AFF1, 4q21.3-q22.1, Red



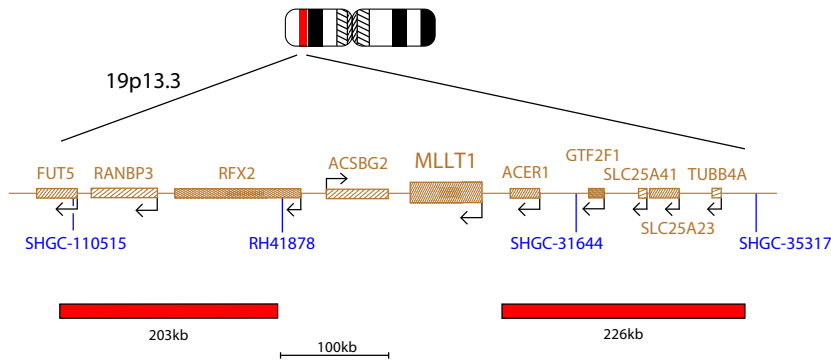
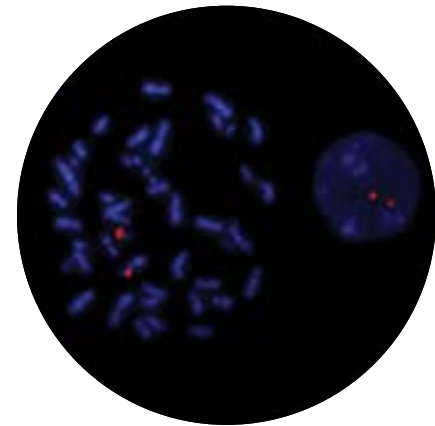
CMP-H087 v004

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

MLLT1

The MLLT1 probe, labeled in red, consists of two clones (203kb and 226kb) that flank the MLLT1 gene.

Probe Specification MLLT1, 19p13.3, Red



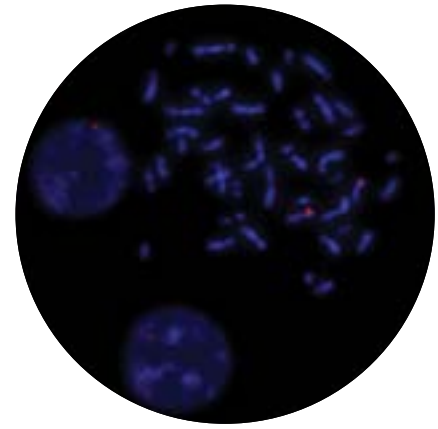
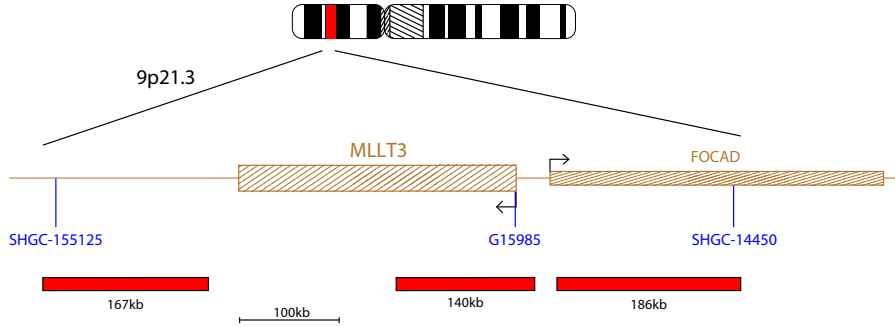
CMP-H084 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

MLLT3

The MLLT3 probe, labeled in red, consists of a 140kb clone that covers the 5' end of the MLLT3 gene and two clones (167kb and 186kb) that flank this.

Probe Specification MLLT3, 9p21.3, Red



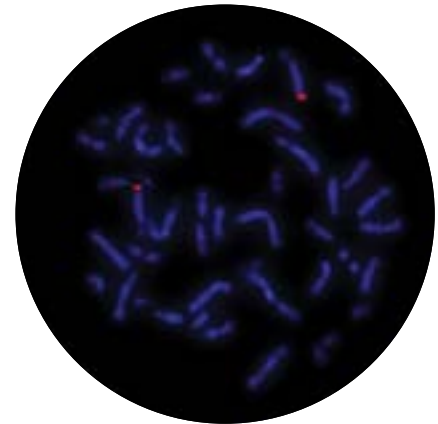
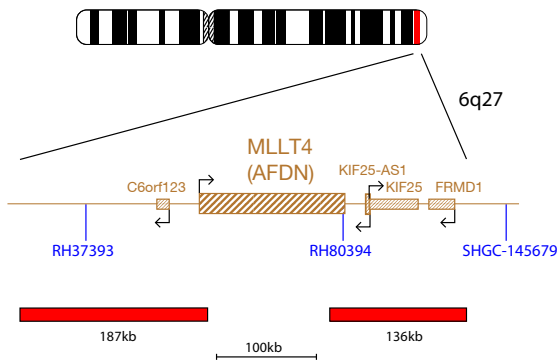
CMP-H085 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

MLLT4 (AFDN)

The MLLT4 (AFDN) probe, labeled in red, consists of two clones (187kb and 136kb) that flank the MLLT4 gene.

Probe Specification MLLT4, 6q27, Red



CMP-H086 v003

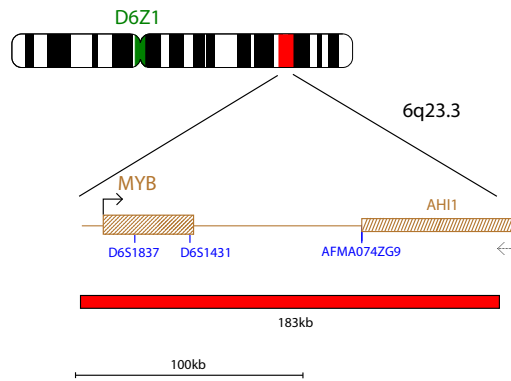
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



MYB Deletion

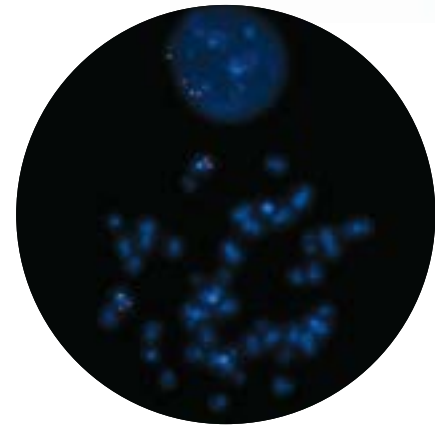
The MYB probe mix consists of a 183kb probe, labeled in red that covers the entire MYB gene and a region telomeric to the gene that includes a centromeric part of the AHI1 gene. This probe mix also contains a control probe for the 6 centromere (D6Z1) labeled in green.

Probe Specification MYB, 6q23.3, Red
D6Z1, 6p11.1-q11.1, Green



CMP-H037 v002

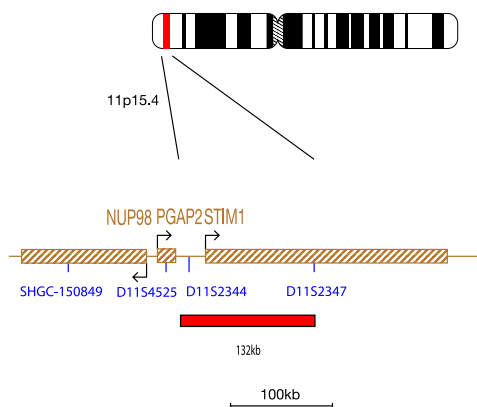
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



NUP98 Proximal Red

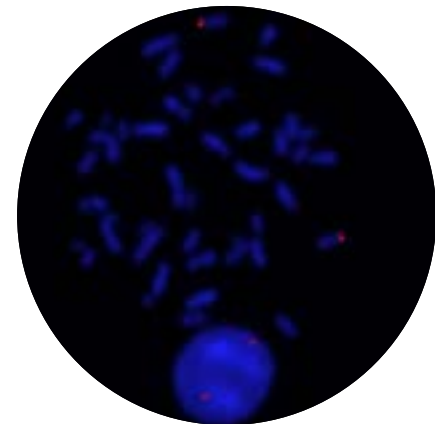
The NUP98 Proximal probe, labeled in red, consists of a 132kb probe proximal to the NUP98 gene, covering the telomeric end of the STIM1 gene including markers D11S2344 and D11S2347.

Probe Specification NUP98 Proximal, 11p15.4, Red



CMP-H118 v001

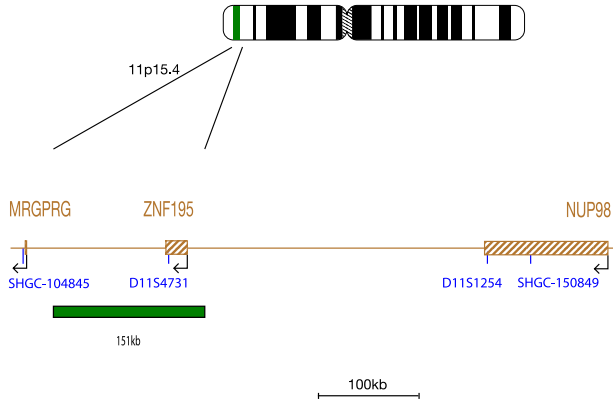
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



NUP98 Distal Green

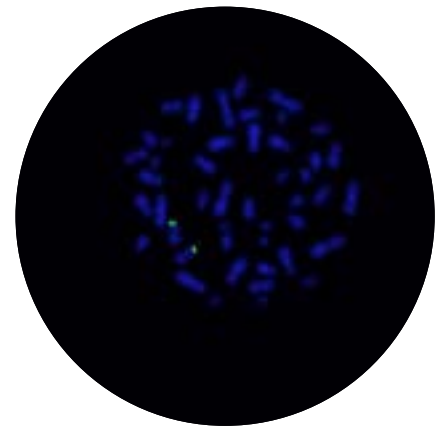
The NUP98 Distal probe, labeled in green, consists of a 151kb probe distal to the NUP98 gene, covering the ZNF195 gene and the D11S4731 marker.

Probe Specification NUP98 Distal, 11p15.4, Green



CMP-H119 v001

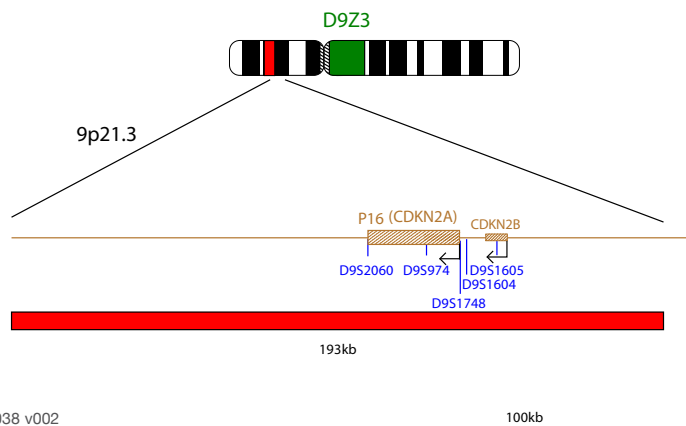
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



P16 (CDKN2A) Deletion

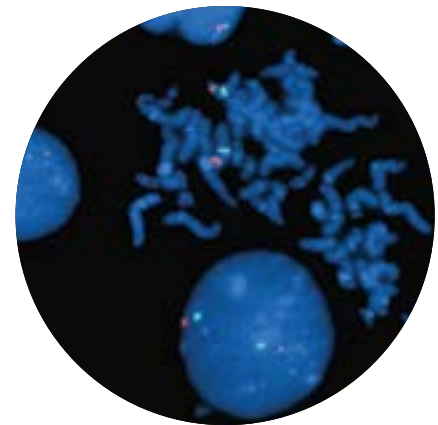
The P16 probe, labeled in red, covers a 193kb region of 9p21.3, including the CDKN2A gene and flanking regions. The probe mix also contains a control probe for chromosome 9 (D9Z3, the heterochromatic block at 9q12) labeled in green.

Probe Specification P16, 9p21.3, Red
D9Z3, 9q12, Green



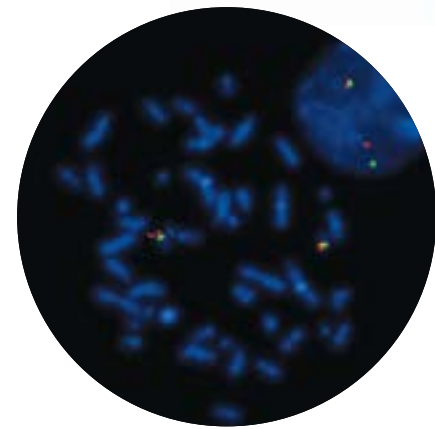
CMP-H038 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

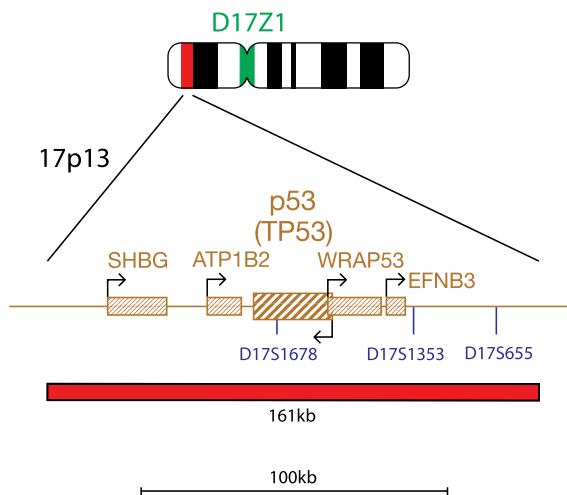


P53 (TP53) Deletion**

The P53 probe mix consists of a 161kb probe, labeled in red that covers the whole P53 (TP53) gene and the flanking regions. The probe mix also contains a control probe for the 17 centromere (D17Z1) that is labeled in green.



Probe Specification P53, 17p13, Red
 D17Z1, 17p11.1-q11.1, Green



CMP-H039 v006

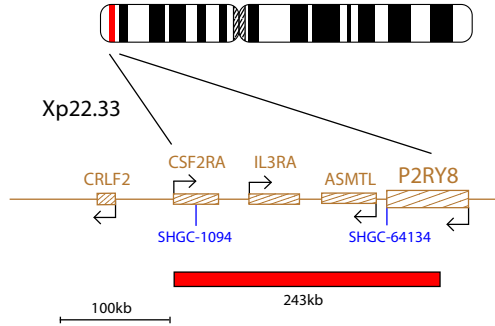
** For sale in Canada only.

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

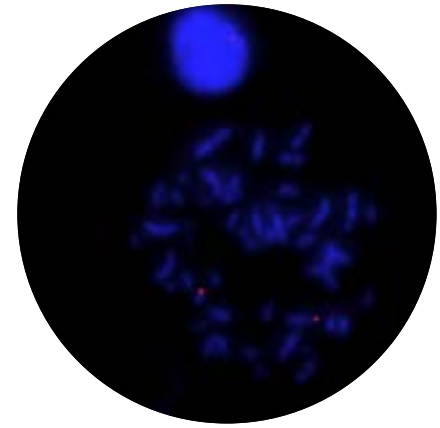
P2RY8 Distal

The P2RY8 Distal probe, labeled in red, consists of a 243kb probe covering the 3' end of, and a region distal (telomeric) to, the P2RY8 gene.

Probe Specification P2RY8 Distal, Xp22.33/Yp11.32, Red



CMP-H093 v002

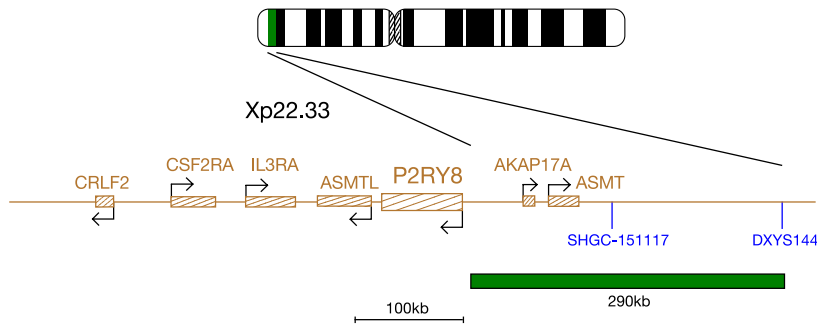


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

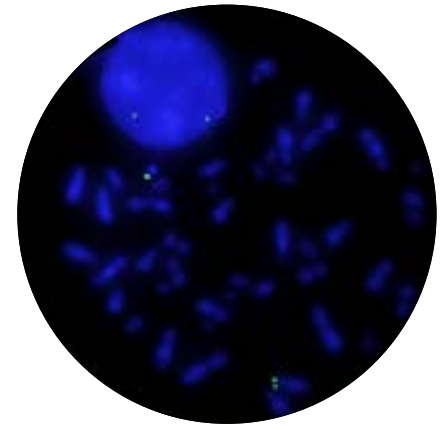
P2RY8 Proximal

The P2RY8 Proximal probe, labeled in green, consists of a 290kb probe that is situated proximal (centromeric) to the P2RY8 gene.

Probe Specification P2RY8 Proximal, Xp22.33/Yp11.32, Green



CMP-H092 v002

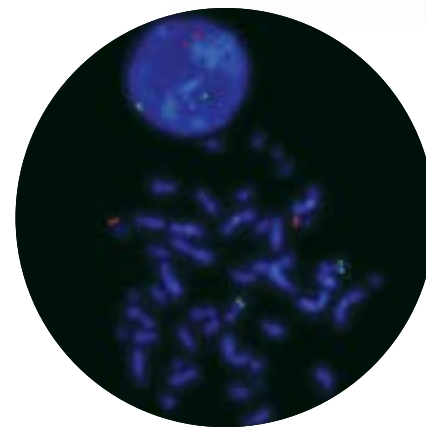


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



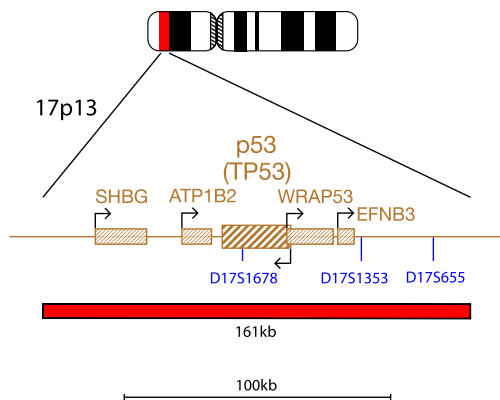


P53 (TP53)/ATM Probe Combination

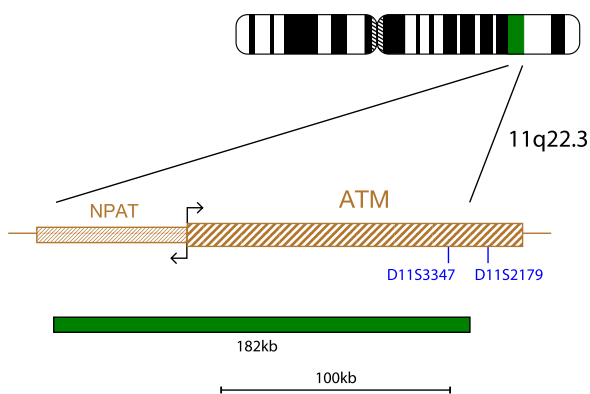


The P53 component consists of a 161kb probe, labeled in red that covers the whole P53 (TP53) gene and flanking regions. The ATM component consists of a 182kb probe, labeled in green that covers the telomeric end of the NPAT gene and the centromeric end of the ATM gene beyond the D11S3347 marker.

Probe Specification P53, 17p13, Red
 ATM, 11q22.3, Green



CMP-H040 v005



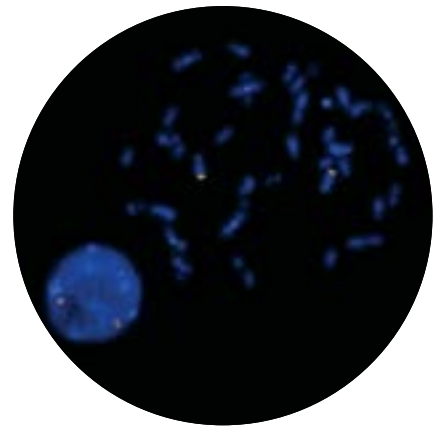
CMP-H041 v005

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

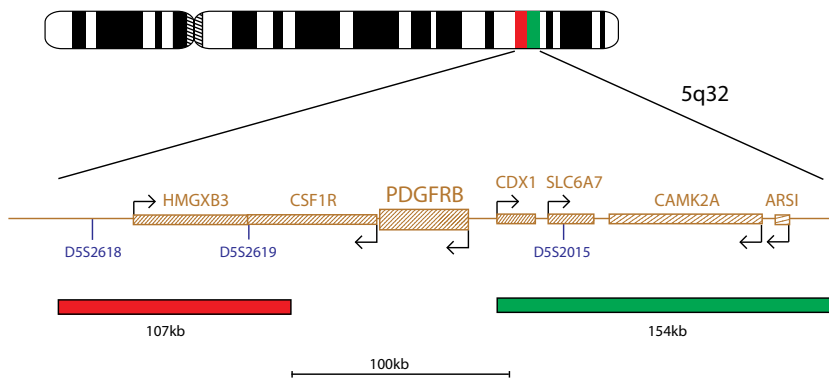


PDGFRB Breakapart

The PDGFRB product consists of a 107kb probe, labeled in red, located centromeric to the PDGFRB gene, including the D5S2618 and D5S2619 markers and a green probe, covering a 154kb region telomeric to the PDGFRB gene, including the D5S2015 marker.



Probe Specification PDGFRB, 5q32, Red
PDGFRB, 5q32, Green



CMP-H042 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

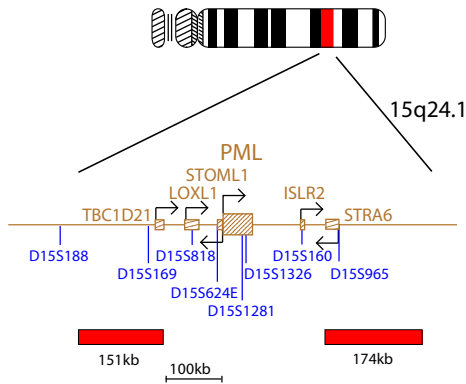




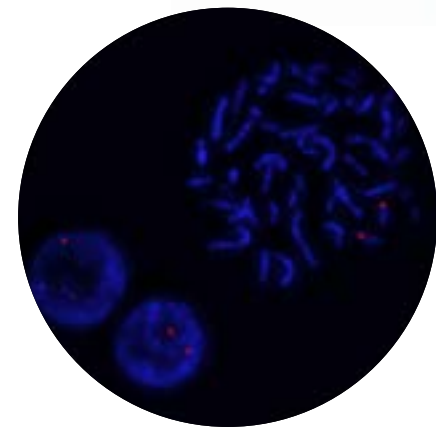
FAST PML

The PML probe mix, labeled in red, consists of a 151kb probe centromeric to the PML gene and a 174kb probe telomeric to the PML gene.

Probe Specification PML, 15q24.1, Red



CMP-H043 v002

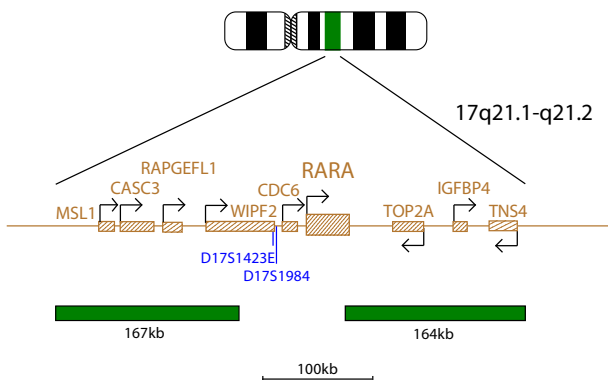


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

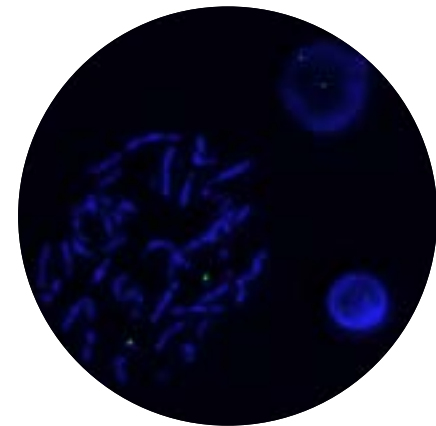
FAST RAR α (RARA)

The RAR α (RARA) probe mix, labeled in green, consists of a 167kb probe centromeric to the RARA gene, including the CASC3 gene, and a 164kb probe, including the telomeric end of the RAR α gene as well as the TOP2A and IGFBP4 genes.

Probe Specification RAR α , 17q21.1-q21.2, Green



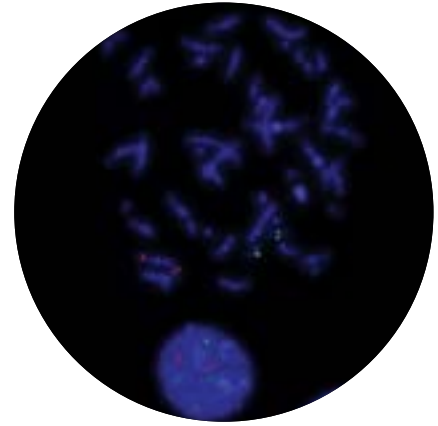
CMP-H044 v002



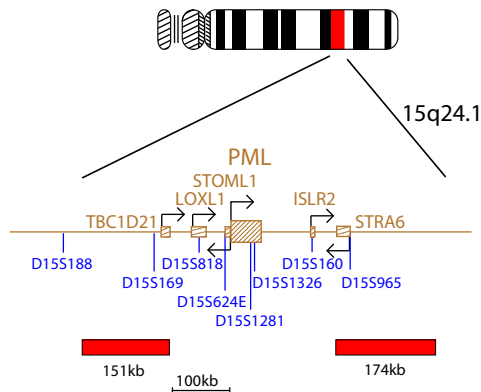
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

PML/RAR α (RARA) Translocation, Dual Fusion

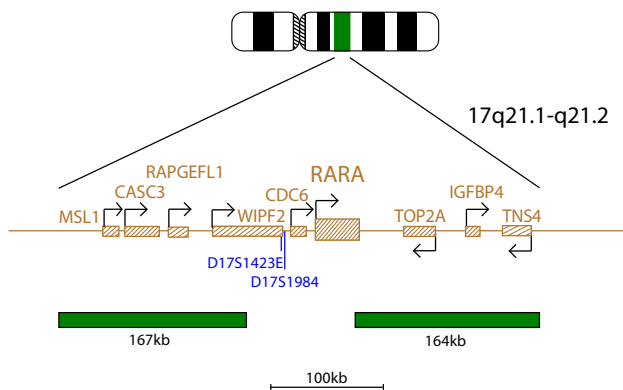
The PML probe mix, labeled in red, consists of a 151kb probe centromeric to the PML gene and a 174kb probe telomeric to the PML gene. The RAR α (RARA) probe mix, labeled in green, consists of a 167kb probe centromeric to the RARA gene, including the CASC3 gene, and a 164kb probe, including the telomeric end of the RARA gene as well as the TOP2A and IGFBP4 genes.



Probe Specification PML, 15q24.1, Red
RAR α , 17q21.1-q21.2, Green



CMP-H043 v002



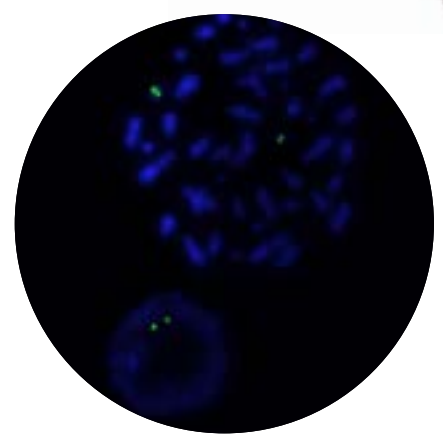
CMP-H044 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

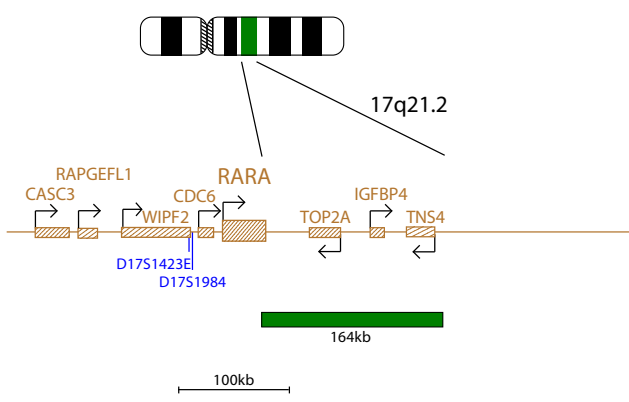


RARα (RARA) Distal

The RARα (RARA) Distal probe, labeled in green, covers a 164kb region, including the telomeric end of the RARA gene as well as the TOP2A and IGFBP4 genes.



Probe Specification RARα Distal, 17q21.2, Green

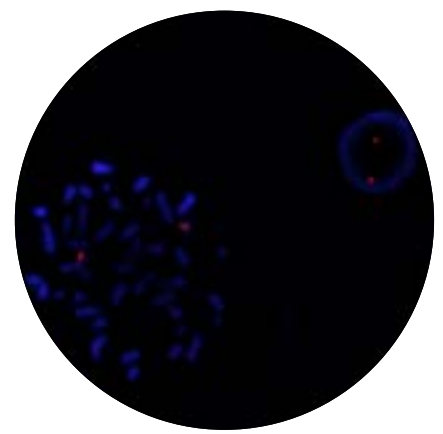


CMP-H069 v002

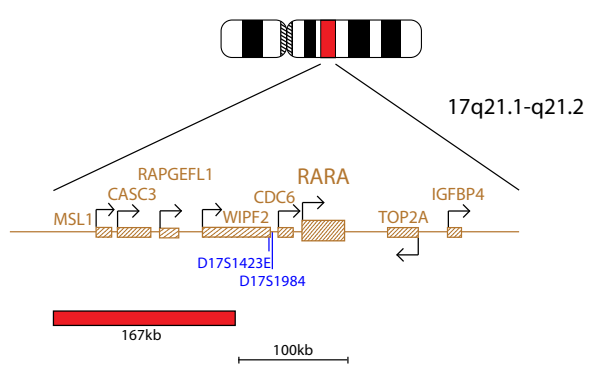
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

RARα (RARA) Proximal

The RARα (RARA) Proximal probe, labeled in red, covers a 167kb region centromeric to the RARA gene, including the CASC3 gene.



Probe Specification RARα Proximal, 17q21.1-q21.2, Red



CMP-H068 v002

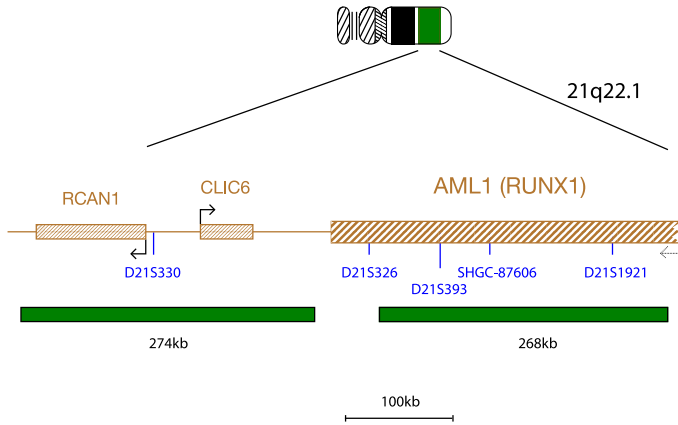
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



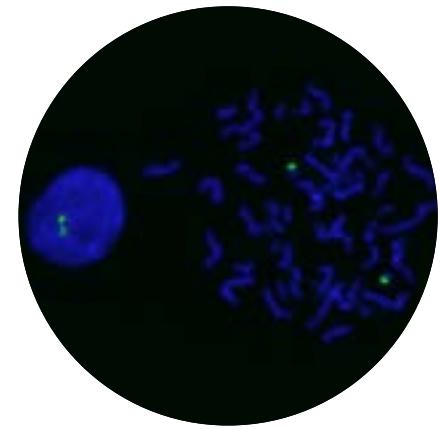
RUNX1 Probe Green

The RUNX1 probe mix, labeled in green, consists of a 274kb probe centromeric to the RUNX1 gene, covering the RCAN1 and CLIC6 genes and a 268kb probe, covering the telomeric end of the RUNX1 gene, including markers D21S393 and D21S1921.

Probe Specification RUNX1, 21q22.12, Green



CMP-H100 v002

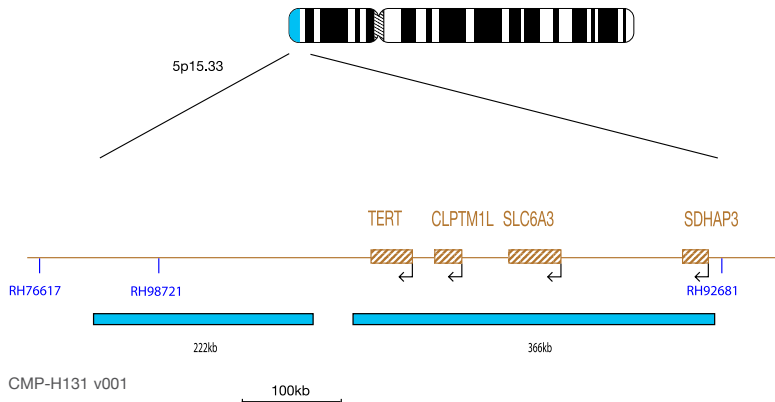


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

TERT (5p15.33) Probe Aqua

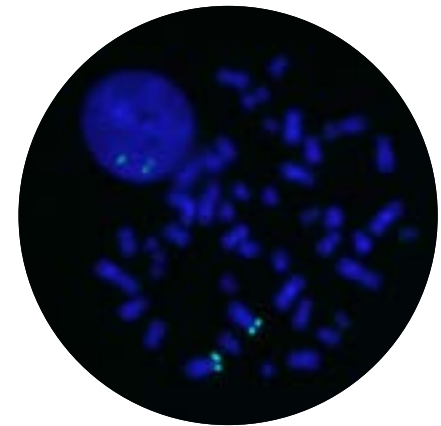
The TERT (5p15.33) probe mix, labeled in aqua, consists of a 222kb probe covering the RH98721 marker and a 366kb probe covering the TERT, CLPTM1L, SLC6A3 and SDHAP3 genes.

Probe Specification TERT, 5p15.33, Aqua



CMP-H131 v001

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

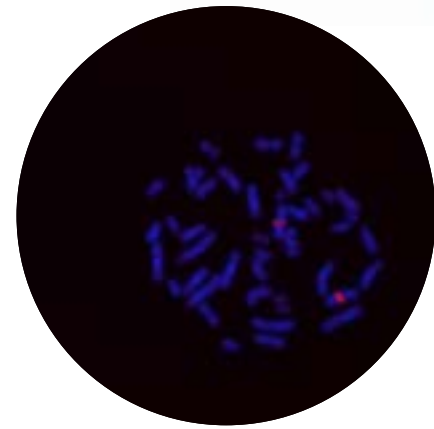
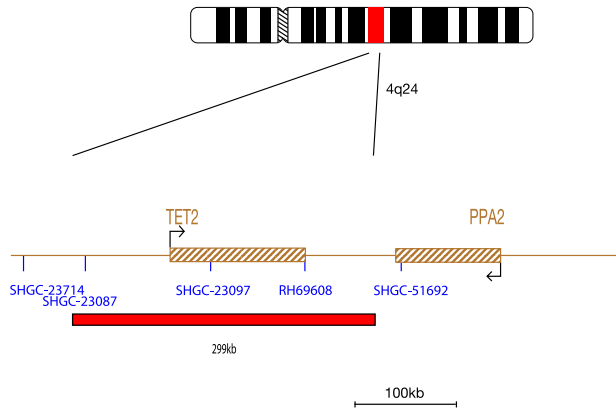




TET2 Probe Red

The TET2 probe, labeled in red, covers a 299kb region including the TET2 gene and markers SHGC-23087, SHGC-23097 and RH69608.

Probe Specification 4q24, Red



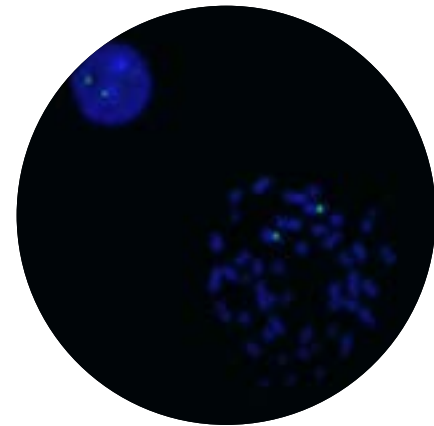
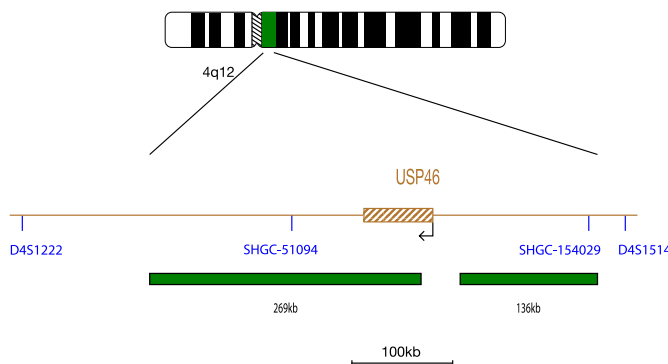
CMP-H106 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

USP46 (4q12) Probe Green

The USP46 (4q12) probe mix, labeled in green, consists of a 269kb probe covering the centromeric end of the USP46 gene and marker SHGC-51094 and a 136kb probe covering marker SHGC-154029.

Probe Specification USP46, 4q12, Green



CMP-H107 v001

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



Chromosome 7 Alpha Satellite Probe Aqua

The chromosome 7 alpha satellite probe, labeled in aqua, covers the highly repeated α -satellite sequences at 7p11.1-7q11.1.

Probe Specification D7Z1: 7p11.1-7q11.1, Aqua

D7Z1



CMP-H128 v001



ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

Chromosome 9 Satellite III Probe Aqua

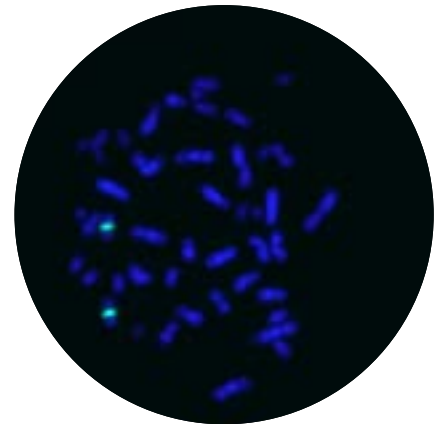
The chromosome 9 satellite III probe, labeled in aqua, covers the highly repeated satellite III sequences at 9q12.

Probe Specification D9Z3, 9q12, Aqua

D9Z3



CMP-H114 v001



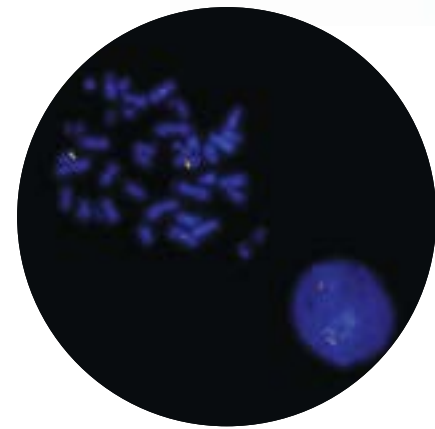
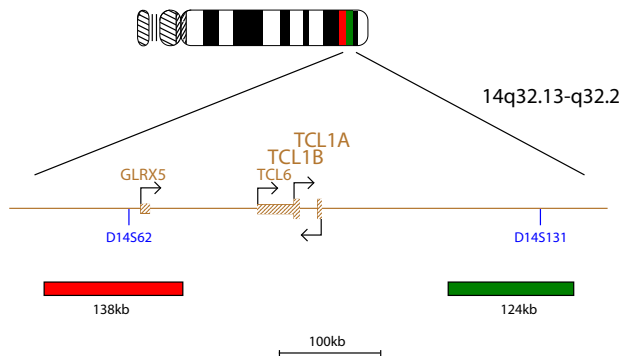
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



TCL1 Breakapart

The TCL1 product consists of a 138kb probe, labeled in red, located centromeric to the TCL1A and TCL1B genes, including the GLRX5 gene and the D14S62 marker, and a green probe covering a 124kb region located telomeric to these genes, including the D14S131 marker.

Probe Specification TCL1, 14q32.13, Red
 TCL1, 14q32.2, Green



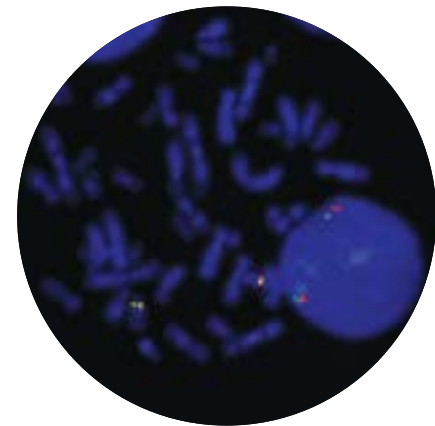
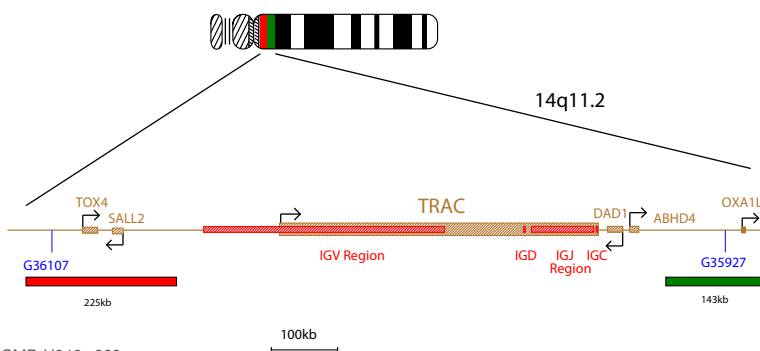
CMP-H045 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

TCRAD Breakapart

The TCRAD product consists of a 225kb probe, labeled in red, located at the centromeric end of the Variable Region of the immunoglobulin gene cluster, including the G36107 marker and a green probe covering a 143kb region located telomeric to the TRAC gene, including the entire OXA1L gene and the G35927 marker.

Probe Specification TCRAD, 14q11.2, Red
 TCRAD, 14q11.2, Green

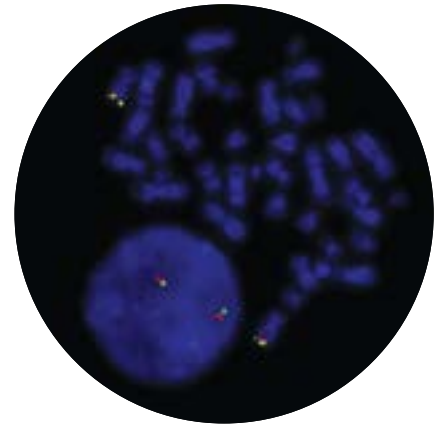


CMP-H046 v002

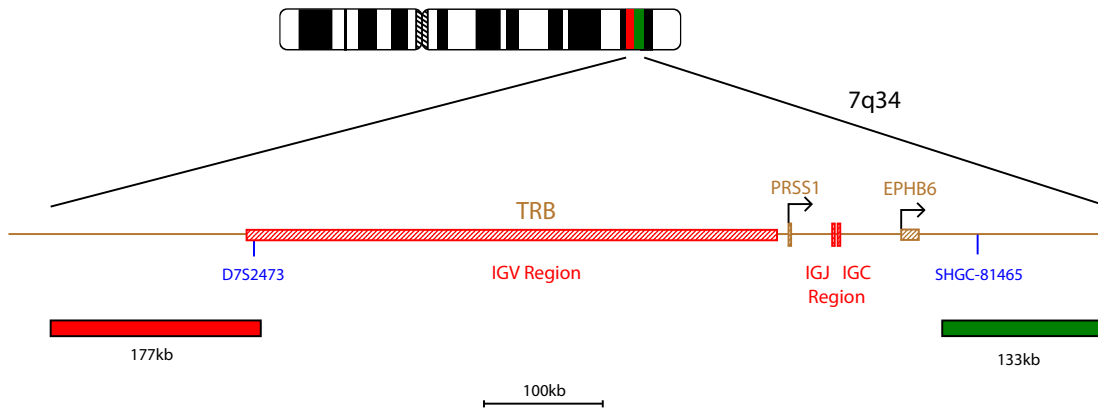
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

TCRB (TRB) Breakapart

The TCRB product consists of a 177kb probe, labeled in red, covering the centromeric end of the TRB gene, including the D7S2473 marker and a green probe covering a 133kb region located telomeric to the TRB gene, including the SHGC-81465 marker.



Probe Specification
TCRB, 7q34, Red
TCRB, 7q34, Green

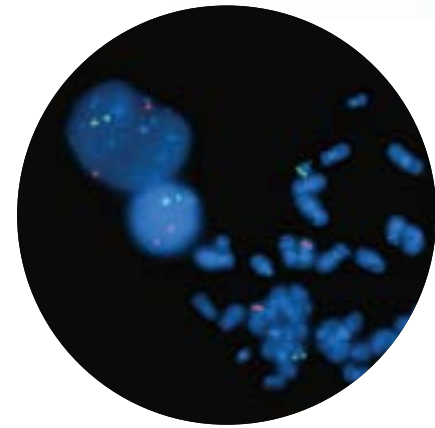


CMP-H047 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

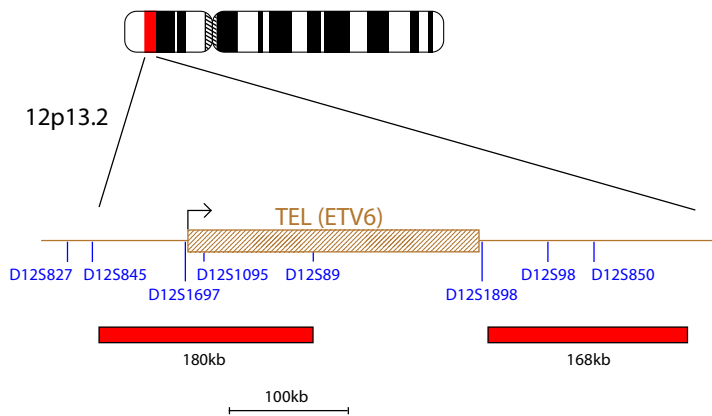


TEL/AML1 (ETV6/RUNX1) Translocation, Dual Fusion

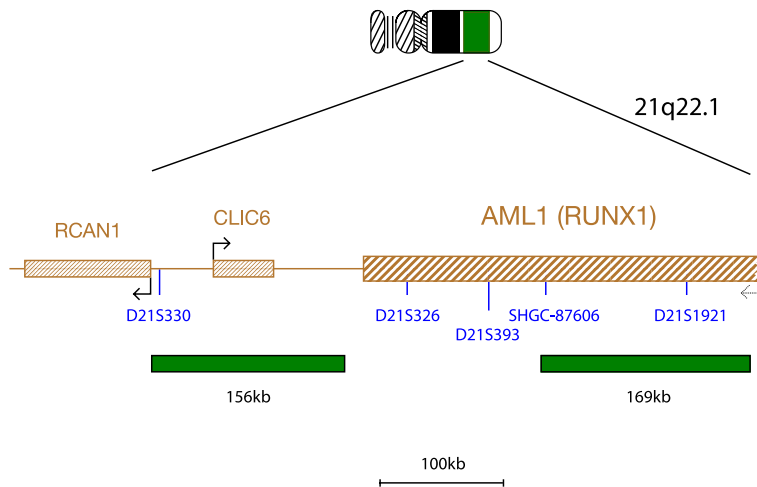


The TEL (ETV6) probe mix, labeled in red, contains a probe covering a 180kb region between the markers D12S845 and D12S89 and a second probe centromeric to the ETV6 gene, extending 168kb from the marker D12S1898. For RUNX1, there are two probes, labeled in green, one covering a 156kb region centromeric to the RUNX1 gene, including the CLIC6 gene and a second probe covering a 169kb region, including the marker D21S1921.

Probe Specification TEL1 (ETV6), 12p13.2, Red
AML1 (RUNX1), 21q22.12, Green



CMP-H048 v002



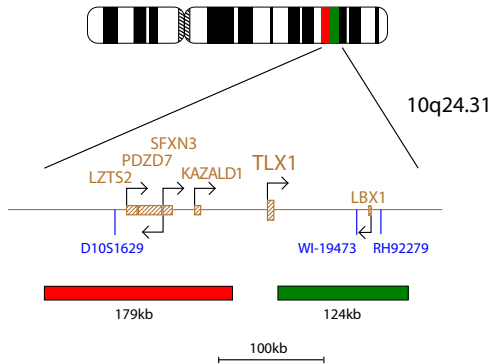
CMP-H049 v004

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

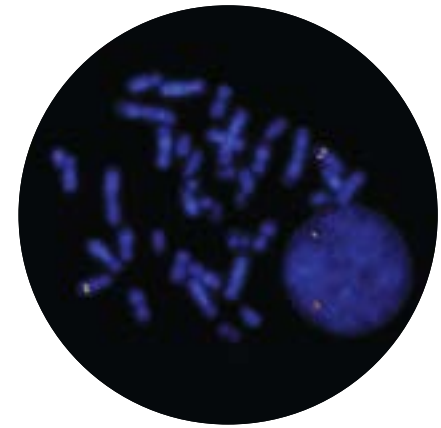
TLX1 Breakapart

The TLX1 product consists of a 179kb probe, labeled in red, located centromeric to the TLX1 gene, including the KAZALD1 gene and the D10S1629 marker and a green probe covering a 124kb region located telomeric to the gene, including the LBX1 gene and the RH92279 marker.

Probe Specification
 TLX1, 10q24.31, Red
 TLX1, 10q24.31, Green



CMP-H050 v002

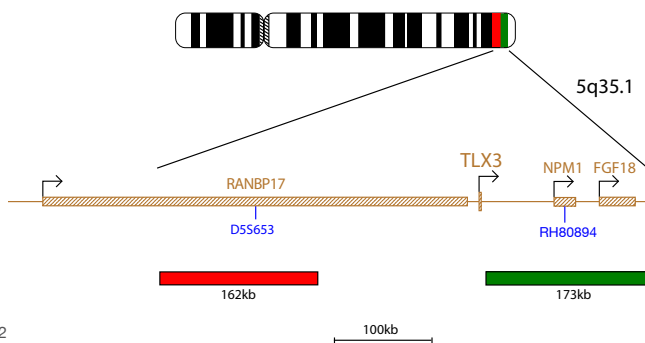


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

TLX3 Breakapart

The TLX3 product consists of a 162kb probe, labeled in red, located centromeric to the TLX3 gene, including the middle part of the RANBP17 gene and the D5S653 marker and a green probe covering a 173kb region located telomeric to the gene, including the NPM1 and FGF18 genes and the RH80894 marker.

Probe Specification
 TLX3, 5q35.1, Red
 TLX3, 5q35.1, Green



CMP-H051 v002



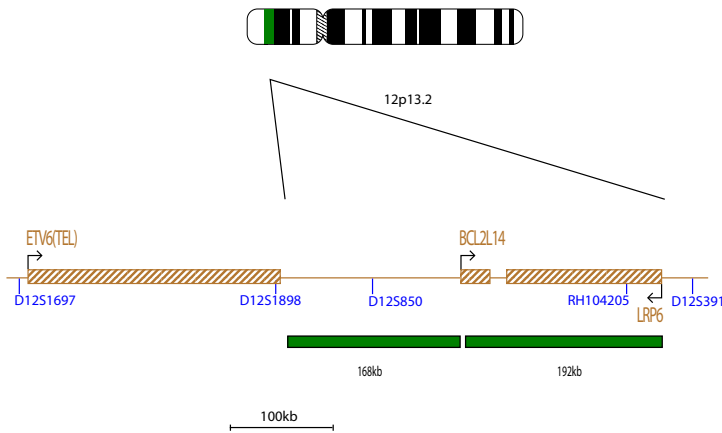
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



ETV6 Proximal Probe Green

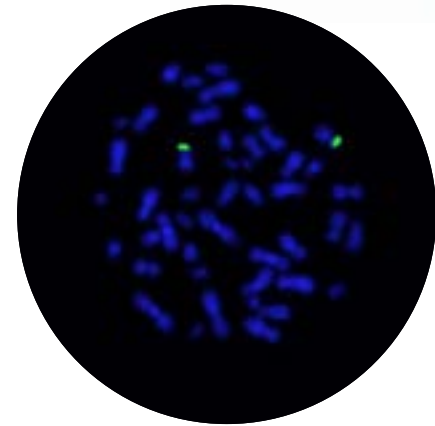
The ETV6 Proximal probe, labeled in green, contains two probes (168kb and 192kb) that map proximal to the ETV6 gene, covering D12S850 and RH104205 markers.

Probe Specification ETV6 Proximal, 12p13.2, Green



CMP-H120 v001

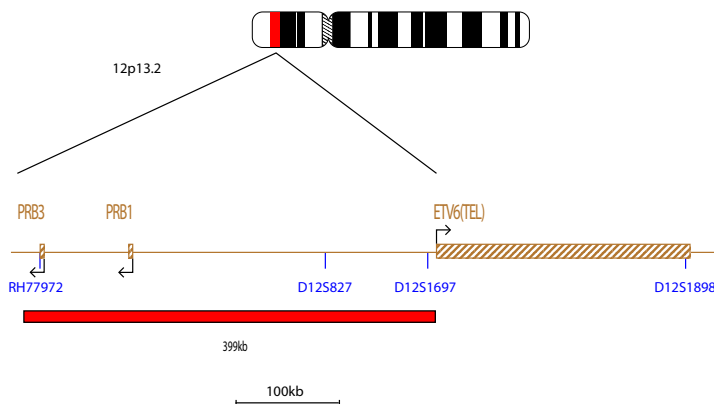
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



ETV6 Distal Probe Red

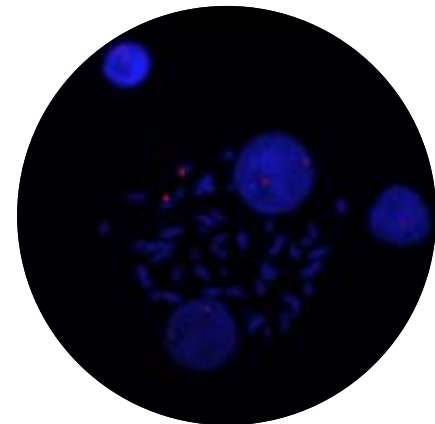
The ETV6 Distal probe, labeled in red, consists of a 399kb probe distal to the ETV6 gene, covering RH77972 and D12S1697 markers.

Probe Specification ETV6 Distal, 12p13.2, Red



CMP-H121 v001

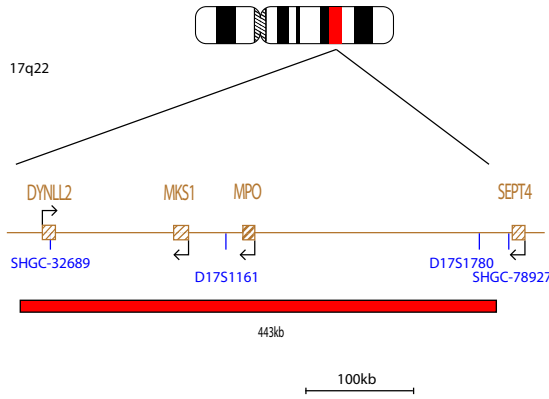
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



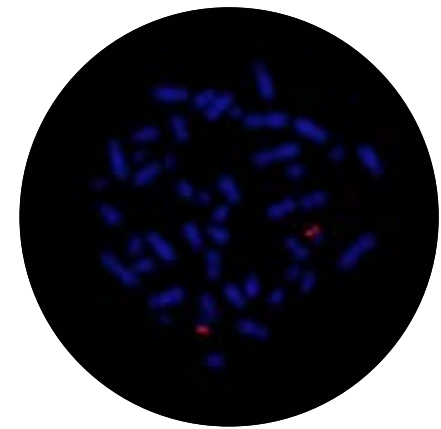
MPO Probe Red

The MPO probe, labeled in red, covers a 443kb region including the MPO gene.

Probe Specification MPO, 17q22, Red



CMP-H109 v001

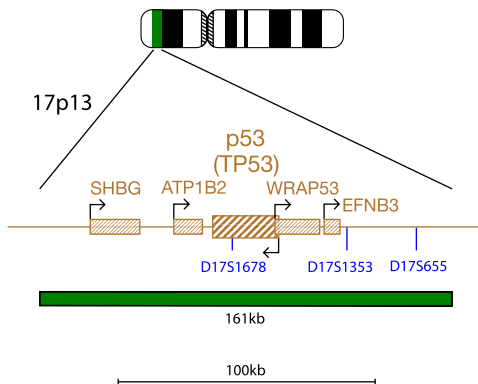


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

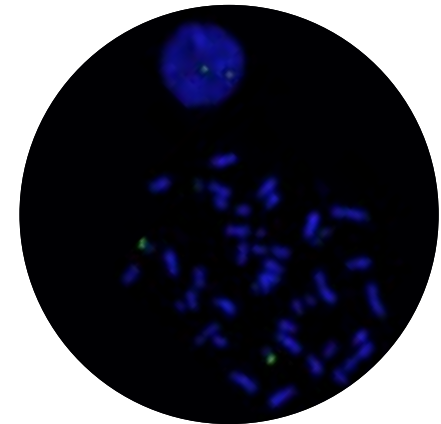
TP53 Probe Green

The TP53 probe, labeled in green, covers a 161kb region including the TP53 gene.

Probe Specification TP53, 17p13, Green



CMP-H110 v002



ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

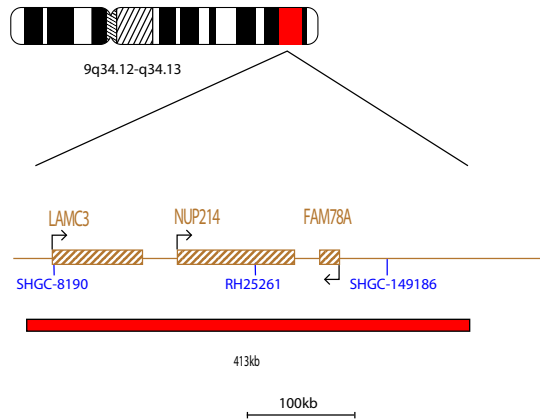




NUP214 Probe Red

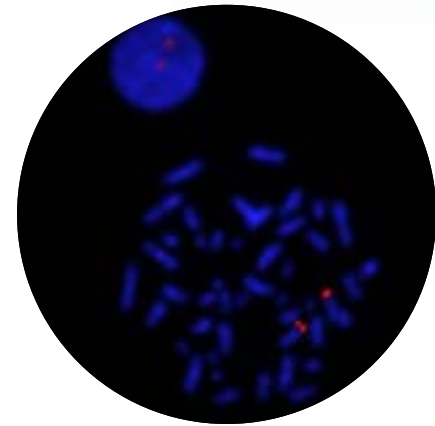
The NUP214 Probe, labeled in red, covers a 413kb region including the NUP214, LAMC3 and FAM78A genes.

Probe Specification NUP214, 9q34.12-q34.13, Red



CMP-H101 v001

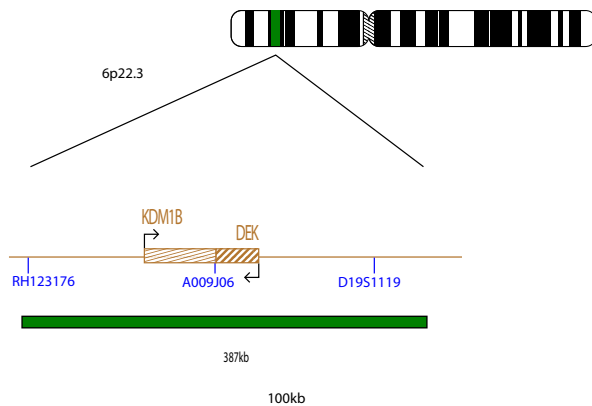
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



DEK Probe Green

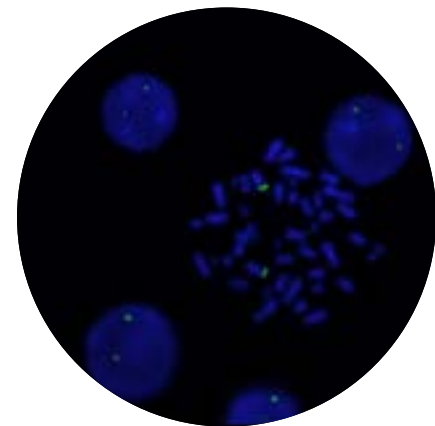
The DEK Probe, labeled in green, covers a 387kb region including the DEK and KDM1B genes.

Probe Specification DEK, 6p22.3, Green



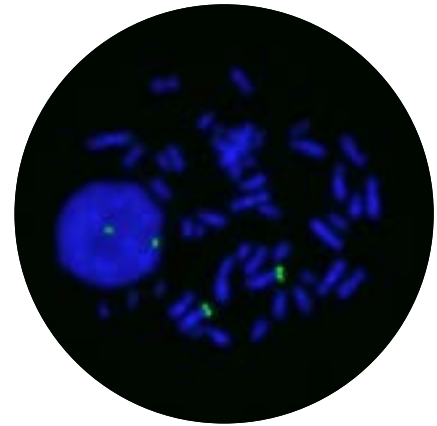
CMP-H102 v001

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

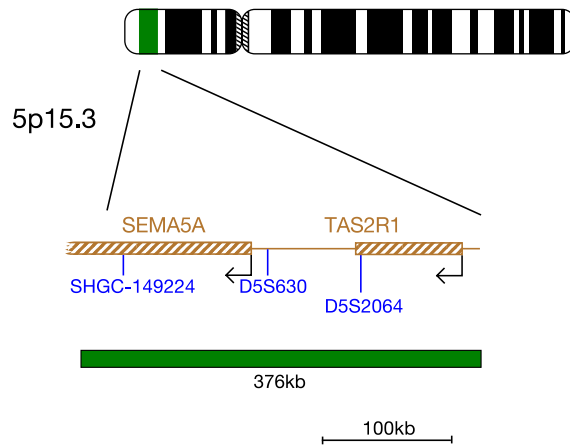


TAS2R1 (5p15.31) Probe Green

The TAS2R1 probe, labeled in green, covers a 376kb region including the centromeric end of SEMA5A gene and TAS2R1 gene.



Probe Specification TAS2R1, 5p15.31, Green



CMP-H113 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.





Alpha Satellite 12 *Plus*

The Alpha Satellite 12 *Plus* Probe is a repeat sequence probe, labeled in red, which recognizes the centromeric repeat sequence D12Z3.

Probe Specification D12Z3, 12p11.1-q11.1, Red



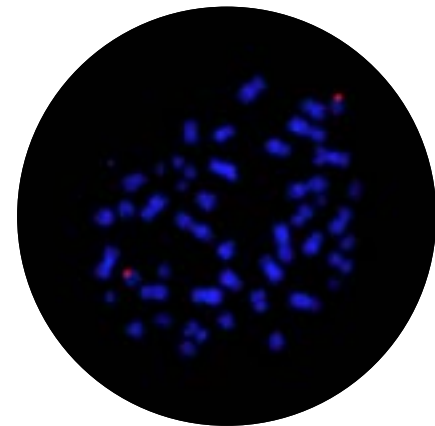
CMP-H002 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

Chromosome 15 Alpha Satellite Probe Red

The chromosome 15 alpha satellite probe, labeled in red, covers the highly repeated α -satellite sequences at 15p11.1-15q11.1.

Probe Specification D15Z4, 15p11.1-15q11.1, Red



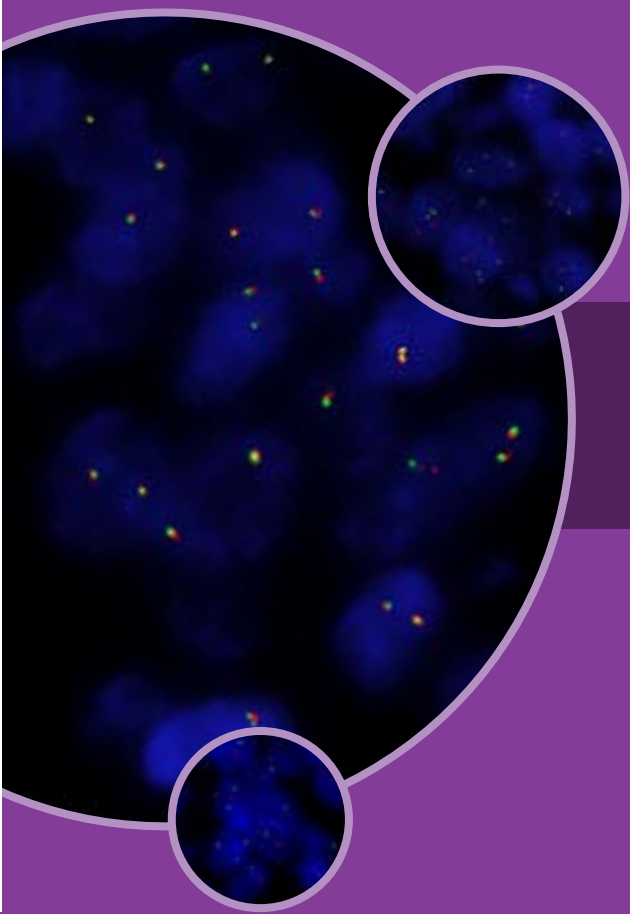
CMP-H115 v001

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



CytoCell
aquarius

Hematopathology



Contents

75	BCL2 Breakapart
75	BCL6 Breakapart
76	CCND1 Breakapart
76	IGH Breakapart
77	IGH/BCL2 Translocation, Dual Fusion
78	IGH/CCND1 Translocation, Dual Fusion
79	IGH/MALT1 Translocation, Dual Fusion
80	IGH/MYC Translocation, Dual Fusion
81	IGK Breakapart
81	IGL Breakapart
82	MALT1 Breakapart
82	MYC Breakapart
83	P16 (CDKN2A) Deletion
83	P53 (TP53) Deletion
84	RB1 Deletion

Hematopathology

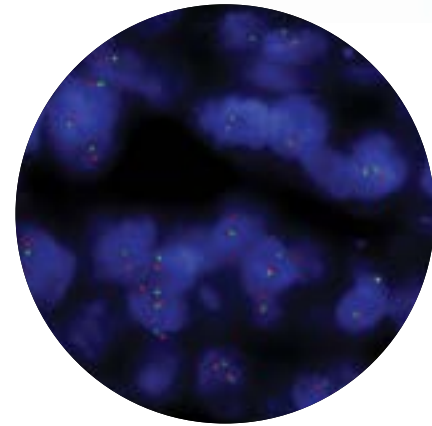
Cytocell offers a range of Hematopathology probes available in the Aquarius® liquid format. These ASR (Analyte Specific Reagent)* probes are directly labeled and ready to use in hybridization buffer.

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

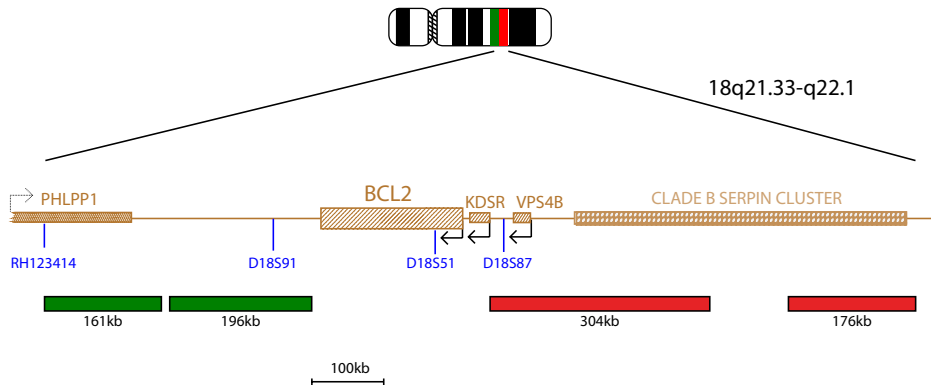


BCL2 Breakapart

The BCL2 product consists of two green 161kb, 196kb probes and two red 304kb, 176kb probes, which are positioned on each side of the BCL2 gene.



Probe Specification
 BCL2, 18q21.33-q22.1, Red
 BCL2, 18q21.33-q22.1, Green

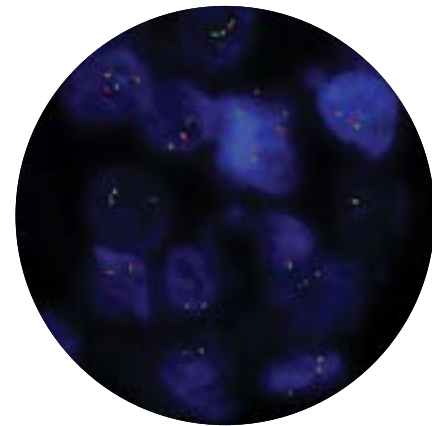


CMP-S052 v002

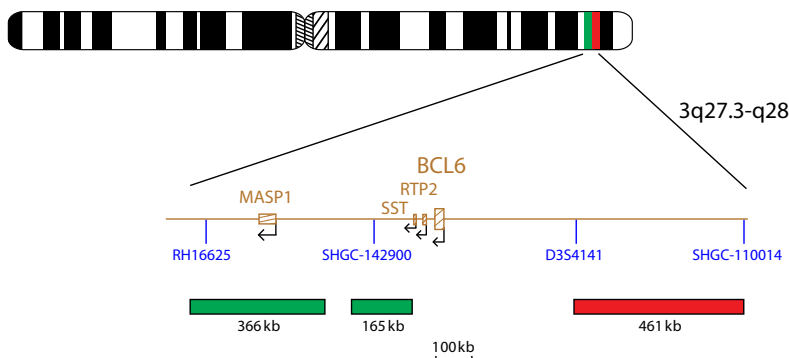
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

BCL6 Breakapart

The BCL6 product consists of a 461kb probe, labeled in red, telomeric to the BCL6 gene, and two green probes, 366kb and 165kb, centromeric to the BCL6 gene.



Probe Specification
 BCL6, 3q27.3-q28, Red
 BCL6, 3q27.3-q28, Green



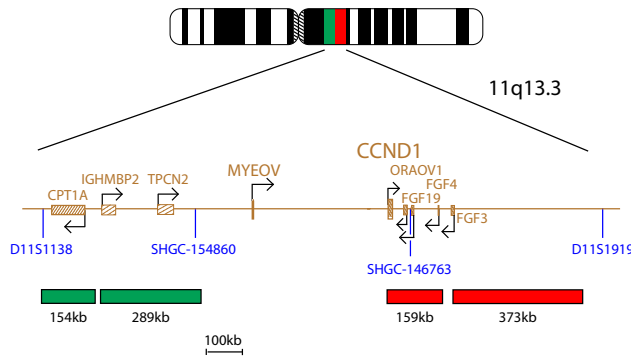
CMP-S047 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

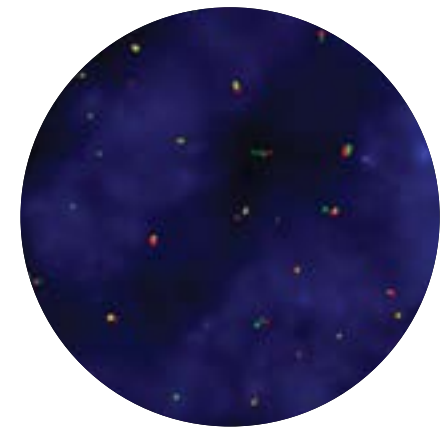
CCND1 Breakapart

The CCND1 product consists of two green 154kb, 289kb probes that are positioned centromeric to the CCND1 gene and two red 159kb, 373kb probes, covering the CCND1 gene and surrounding telomeric regions up to the D11S1919 marker.

Probe Specification
 CCND1, 11q13.3, Red
 CCND1, 11q13.3, Green



CMP-S048 v002

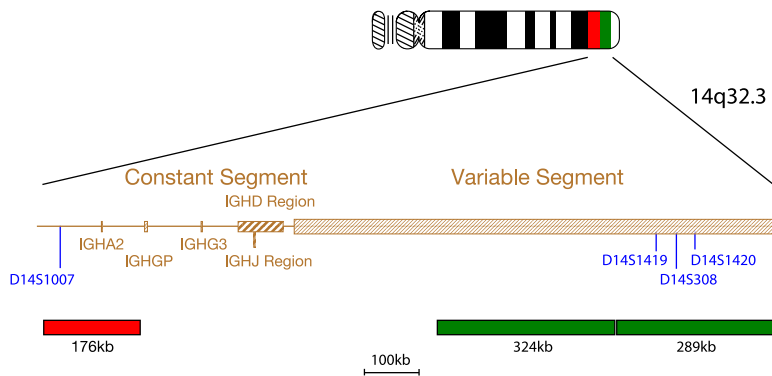


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

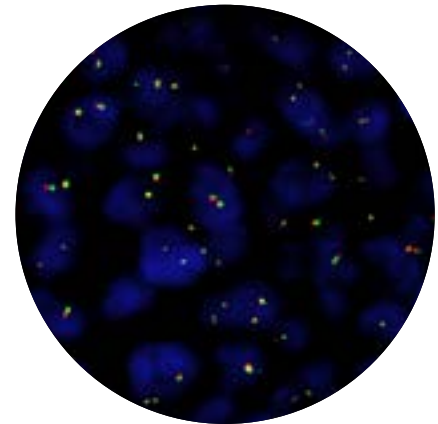
IGH Breakapart

The IGH probe mix consists of a 176kb probe, labeled in red, covering part of the Constant region of the gene and two green probes (324kb and 289kb), covering part of the Variable segment of the gene.

Probe Specification
 IGHC, 14q32.3, Red
 IGHV, 14q32.3, Green



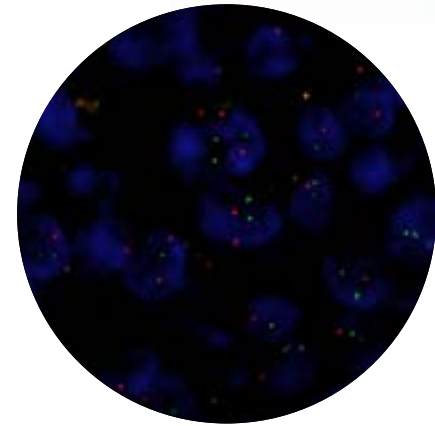
CMP-H023 v003



ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

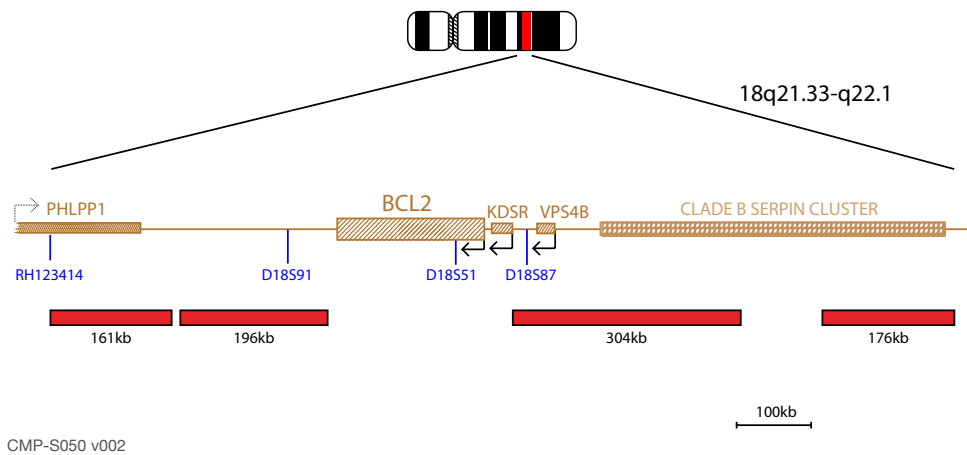
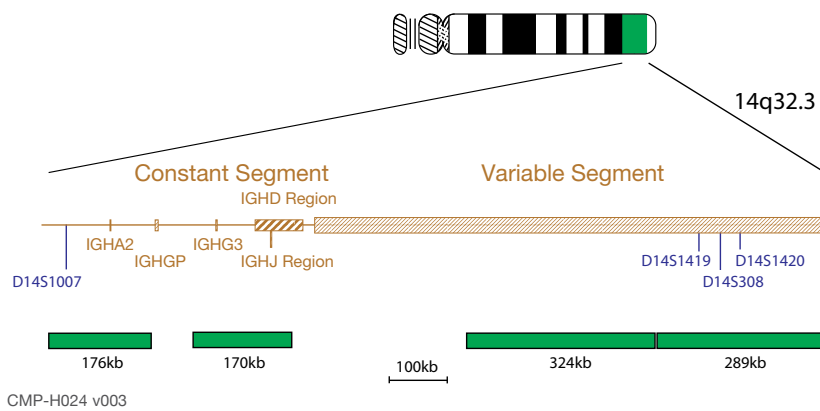


IGH/BCL2 Translocation, Dual Fusion



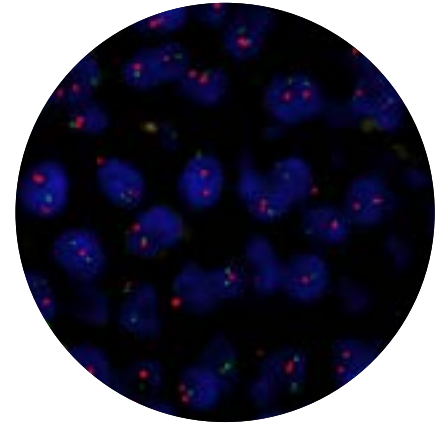
The IGH/BCL2 product consists of probes, labeled in green, covering the Constant, J, D and Variable segments of the IGH gene, and four probes labeled in red, positioned centromeric and telomeric to the BCL2 gene.

Probe Specification BCL2, 18q21.33-q22.1, Red
 IGH, 14q32.33, Green



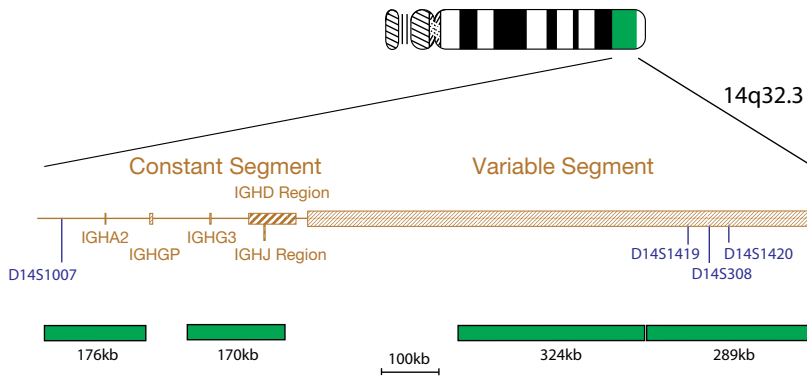
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

IGH/CCND1 Translocation, Dual Fusion

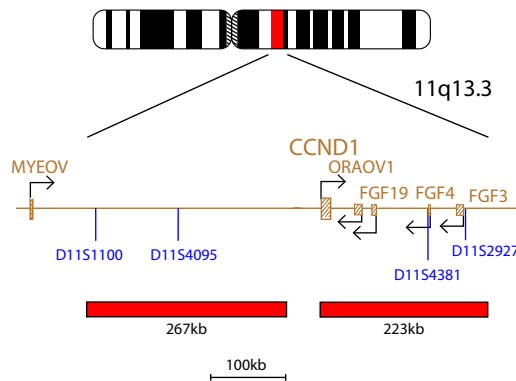


The IGH/CCND1 product consists of probes, labeled in green, covering the Constant, J, D and Variable segments of the IGH gene, and CCND1 probes labeled in red. The CCND1 probe mix contains a 267kb probe centromeric to CCND1 gene, covering the region between the D11S1100 and the D11S4095 markers, and a second 223kb probe covering the telomeric end of CCND1 gene.

Probe Specification CCND1 (BCL1), 11q13.3, Red
IGH, 14q32.33, Green



CMP-H024 v003



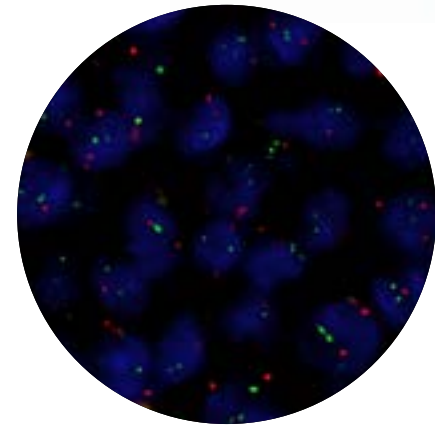
CMP-S053 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

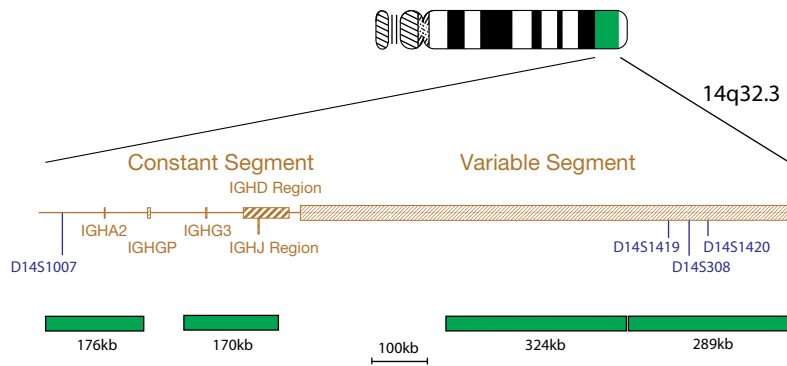


IGH/MALT1 Translocation, Dual Fusion

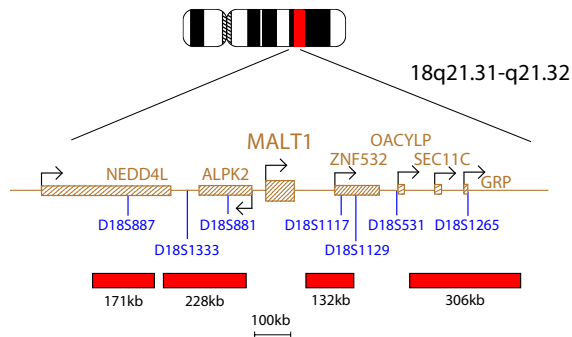
The IGH/MALT1 product consists of probes, labeled in green, covering the Constant, J, D and Variable segments of the IGH gene, and MALT1 probes, labeled in red. The MALT1 probe mix contains four probes, two 171kb, 228kb probes positioned centromeric to the MALT1 gene and two 132kb, 306kb probes positioned telomeric to the MALT1 gene.



Probe Specification MALT1, 18q21.31-q21.32, Red
IGH, 14q32.33, Green



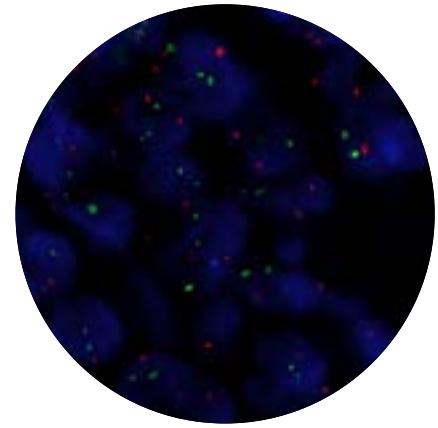
CMP-H024 v003



CMP-S054 v002

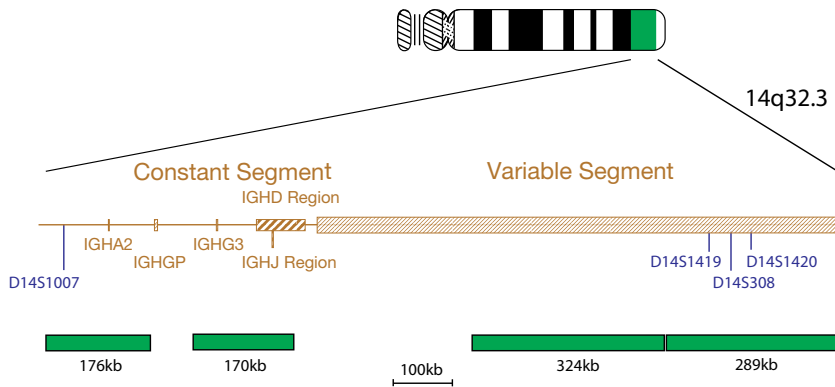
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

IGH/MYC Translocation, Dual Fusion

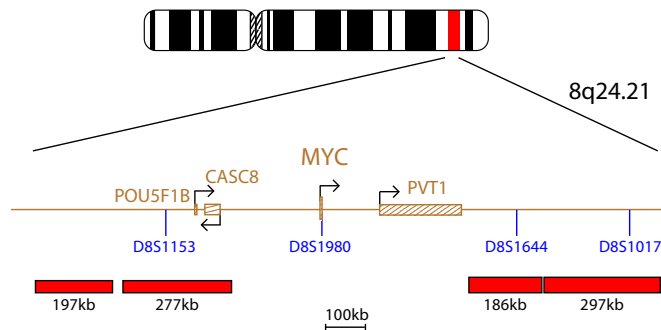


The IGH/MYC product consists of probes, labeled in green, covering the Constant, J, D and Variable segments of the IGH gene, and MYC probes, labeled in red. The MYC probe mix contains four probes, two 197kb, 277kb probes positioned centromeric to the MYC gene and two 186kb, 297kb probes positioned telomeric to the MYC gene, including the D8S1644 marker.

Probe Specification MYC, 8q24.21, Red
 IGH, 14q32.33, Green



CMP-H024 v003



CMP-S051 v002

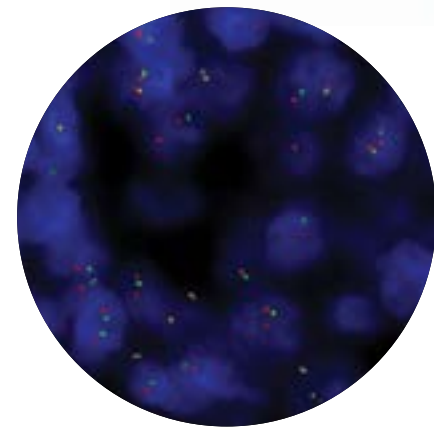
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



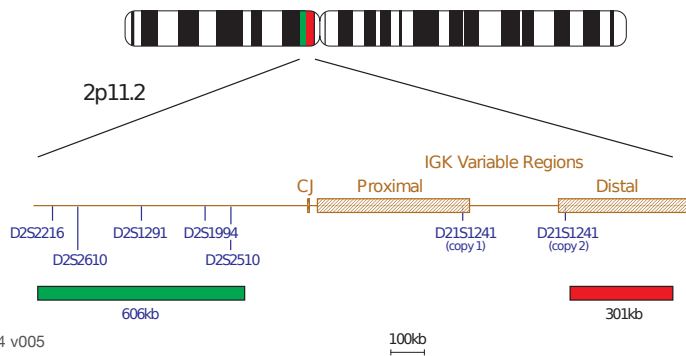


IGK Breakapart

The IGK product consists of a 301kb probe, labeled in red, covering a part of the distal IGK Variable region and a green probe, covering a 606kb region telomeric to the Joining segments and the Constant segment of IGK. The green probe extends from a position that is telomeric to the D2S2216 marker and continues to a position that is centromeric to the D2S2510 marker.



Probe Specification
IGK, 2p11.2, Red
IGK, 2p11.2, Green

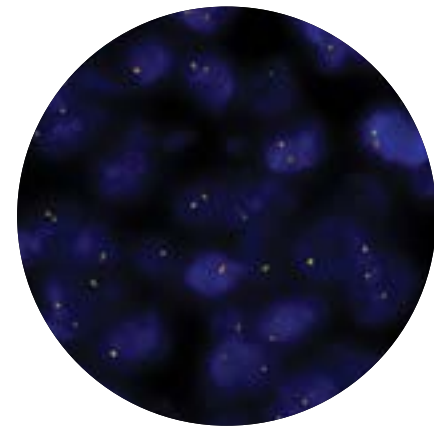


CMP-H034 v005

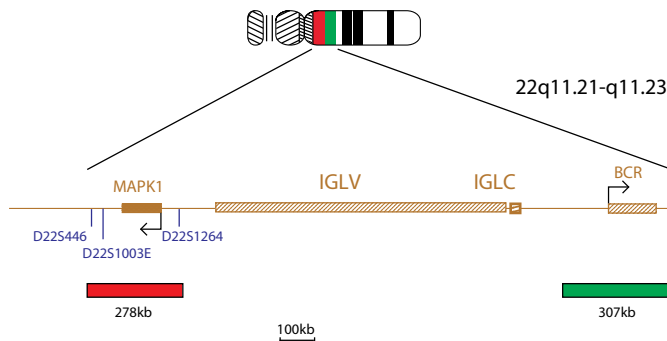
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

IGL Breakapart

The IGL product consists of a 278kb probe, labeled in red, centromeric to the IGL Variable region and covering the MAPK1 gene, and a green probe, covering a 307kb region telomeric to the IGL Constant segment, including the BCR gene.



Probe Specification
IGL, 22q11.21-q11.23, Red
IGL, 22q11.21-q11.23, Green



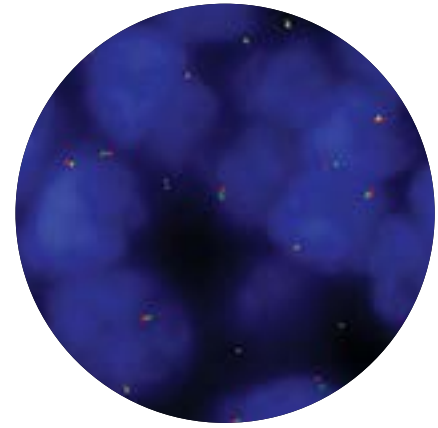
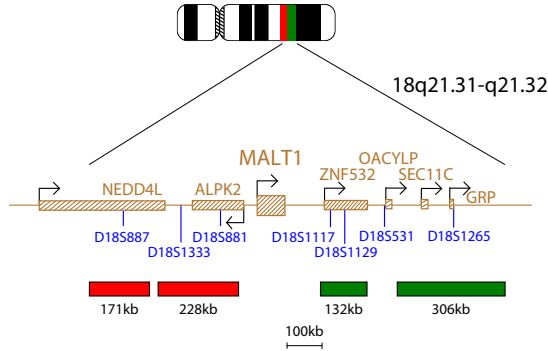
CMP-H035 v004

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

MALT1 Breakapart

The MALT1 Breakapart probe consists of two green clones (132kb and 306kb) and two red clones (171kb and 228kb), which are positioned on each side of the MALT1 gene.

Probe Specification
 MALT1, 18q21.31-q21.32, Red
 MALT1, 18q21.31-q21.32, Green



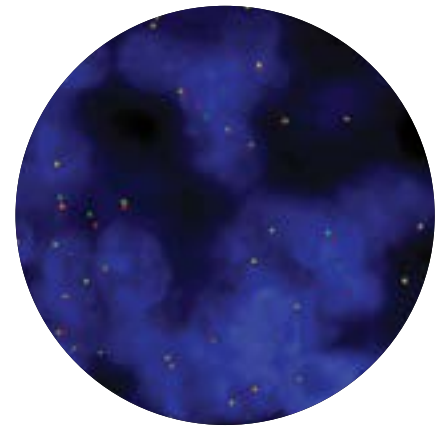
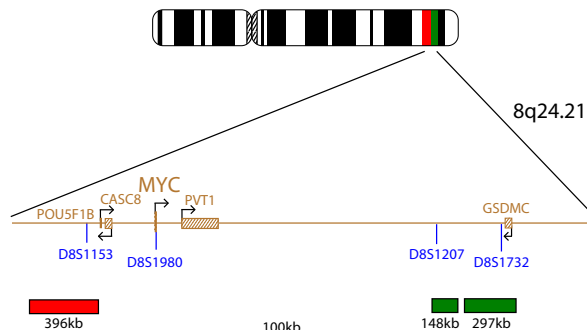
CMP-S011 v005

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

MYC Breakapart

The MYC Breakapart probe consists of a 396kb probe, labeled in red, centromeric to MYC gene, including the marker D8S1153 and two green probes (148kb and 297kb) telomeric to the MYC gene, covering the GSDMC gene.

Probe Specification
 MYC, 8q24.21, Red
 MYC, 8q24.21, Green



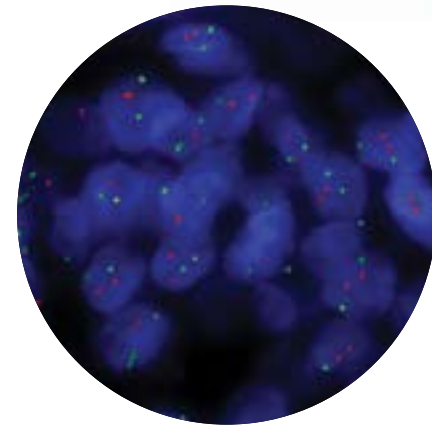
CMP-S049 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

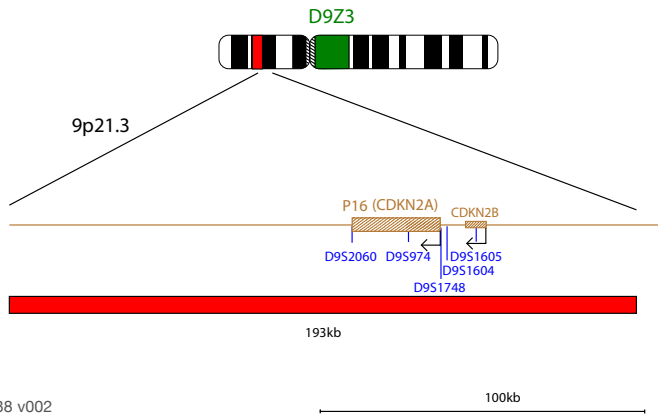


P16 (CDKN2A) Deletion

The P16 probe, labeled in red, covers a 193kb region of 9p21.3, extending from 105kb telomeric of P16 (CDKN2A) gene to 46kb centromeric of CDKN2B. The probe mix also contains a control probe for chromosome 9 (D9Z3, the heterochromatic block at 9q12) labeled in green.



Probe Specification P16, 9p21.3, Red
D9Z3, 9q12, Green



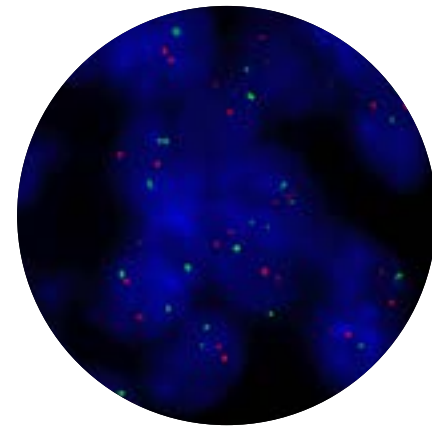
CMP-H038 v002

100kb

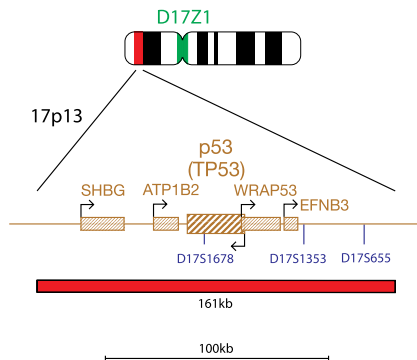
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

P53 (TP53) Deletion

The P53 probe mix consists of a 161kb probe, labeled in red that covers the whole P53 (TP53) gene and the flanking regions. The probe mix also contains a control probe for the 17 centromere (D17Z1) that is labeled in green.



Probe Specification P53, 17p13.1, Red
D17Z1, 17p11.1-q11.1, Green



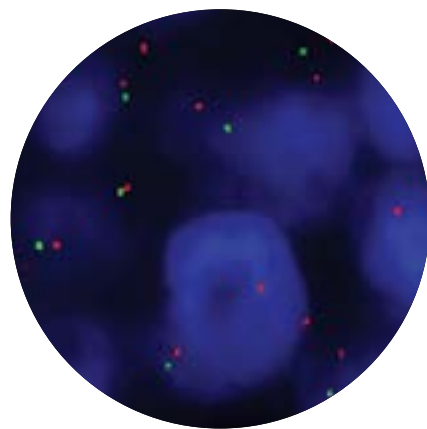
CMP-H039 v006

100kb

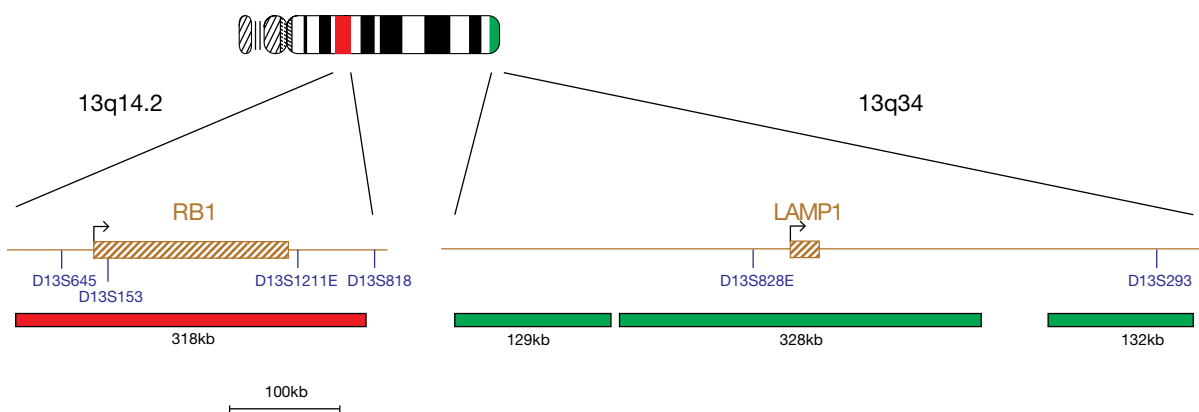
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

RB1 Deletion

The RB1 Deletion probe consists of a 318kb red probe spanning the RB1 gene region. The 13qter probe in green, which includes the LAMP1 gene, acts as a control for chromosome 13.*



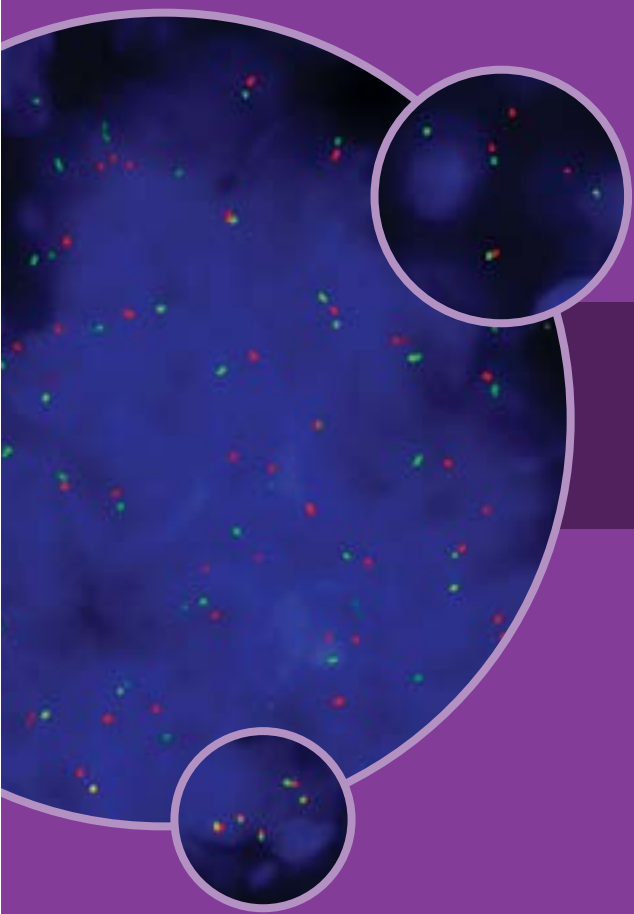
Probe Specification RB1, 13q14.2, Red
 13qter, 13q34, Green



CMP-S016 v006

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.





Pathology



Contents

87	Tissue Pretreatment Kit
88	1p36
88	1q25
89	19p13
89	19q13
90	ALK Breakapart
90	CDKN2A Probe Gold
91	CHOP (DDIT3) Breakapart
91	Centromere 17 Probe Aqua
92	Centromere 7 Probe Green
92	Centromere 3 Probe Red
93	C-MET (MET) Amplification
93	EGFR Amplification
94	EML4 Breakapart
94	ERG Control
95	ETV6 Proximal Probe Green
95	ETV6 Distal Probe Red
96	EWSR1 Breakapart
97	EWSR1/ERG Translocation, Dual Fusion
98	FOXO1 Proximal Probe Green
98	FOXO1 Distal Probe Red
99	FUS Proximal Probe Red
99	FUS Distal Probe Green
100	FLI1/EWSR1 Translocation, Dual Fusion
101	FGFR1 Breakapart/Amplification
101	HER2 (ERBB2) Amplification
102	MDM2 Amplification
102	MPO Probe Red
103	N-MYC (MYCN) Amplification
103	PAX3 Breakapart
104	PAX7 Breakapart
104	RET Proximal
105	RET Distal
105	ROS1 Breakapart
106	ROS1-GOPC (FIG) Proximal
106	ROS1-GOPC (FIG) Distal
107	SRD (CHD5) Deletion
107	SYT (SS18) Breakapart
108	TMPRSS2 Breakapart
108	TFE3 Proximal Probe Red
109	TFE3 Distal Probe Green
109	TOP2A Amplification/Deletion
110	ZNF217 Amplification
110	TP53 Green

Pathology

Cytocell offers a range of Pathology probes available in the Aquarius® liquid format. These probes are directly labeled and ready to use in hybridization buffer.

Tissue Pretreatment Kit

Introducing the first Pretreatment kit capable of preparing slides for FISH and/or CISH analysis on formalin-fixed, paraffin-embedded (FFPE) tissue.

Our ready-to-use Tissue Pretreatment Kit has been optimized to produce excellent visual results with our extensive Aquarius® Pathology FISH range.

To further extend the utility of the kit we have also validated its use with other commercially available FISH (fluorescence *in situ* hybridization) DNA probes.*



Product Information

Aquarius® Tissue Pretreatment Kit**

Kit Components

Reagent 1 (1x1L)
Reagent 2 (1x10mL)

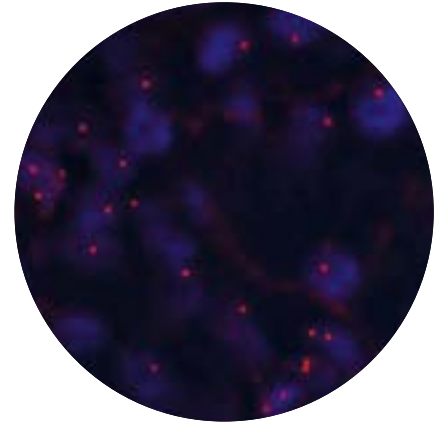
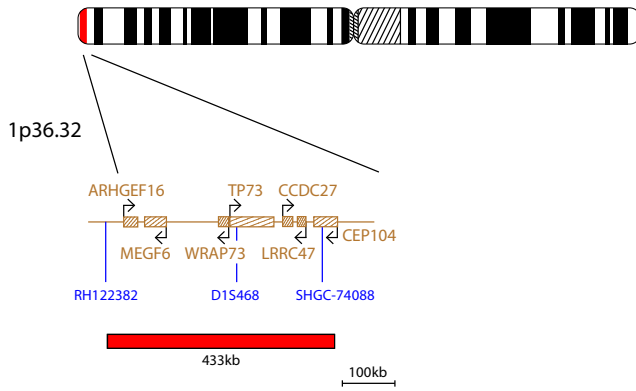
* A list of manufacturers is available upon request.

** This product is provided under an agreement between Life Technologies Corporation and Cytocell Ltd and is available for human diagnostics or life science use only.

1p36

The 1p36 probe, labeled in red, is 433kb in size and covers the region between markers RH122382 and SHGC-74088.

Probe Specification 1p36 Probe, 1p36.32, Red



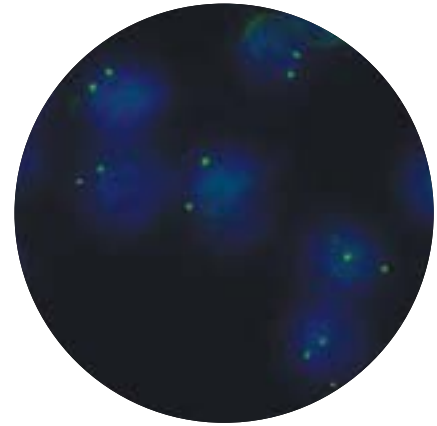
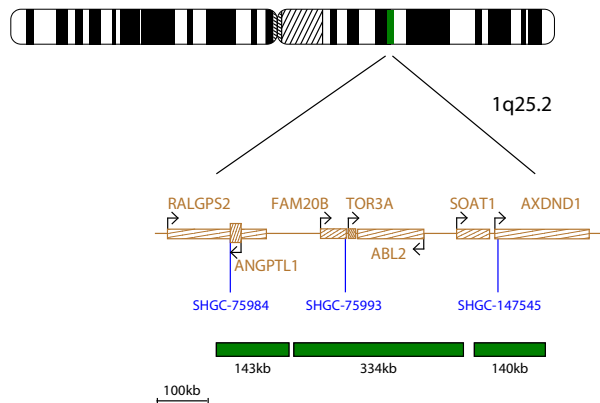
CMP-S062 v001

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

1q25

The 1q25 probe, labeled in green, consists of three probes (143kb, 334kb and 140kb) that cover regions including markers SHGC-75984 and SHGC-147545.

Probe Specification 1q25 Probe, 1q25.2 Green



CMP-S063 v001

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

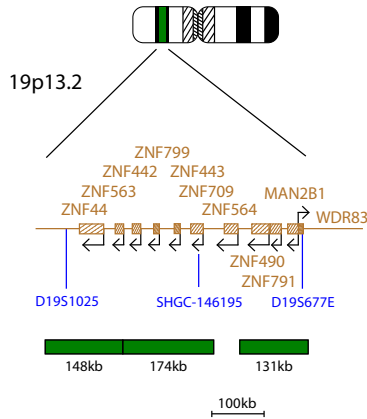




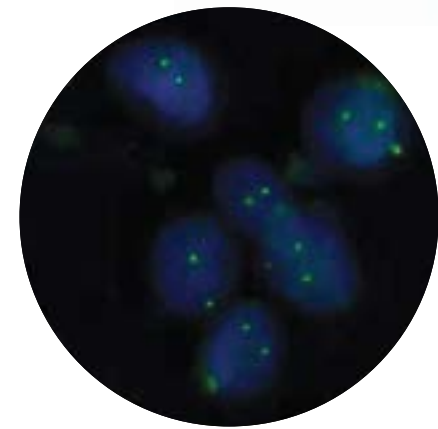
19p13

The 19p13.2 probe, labeled in green, consists of three probes (148kb, 174kb and 131kb) that cover regions including markers D19S1025 and D19S677E.

Probe Specification 19p13 Probe, 19p13.2, Green



CMP-S066 v001

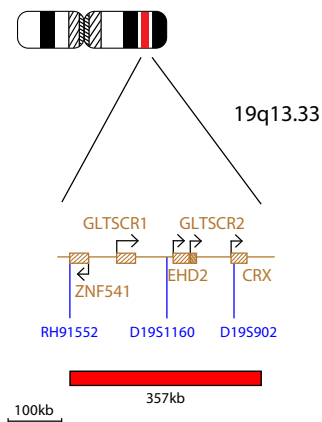


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

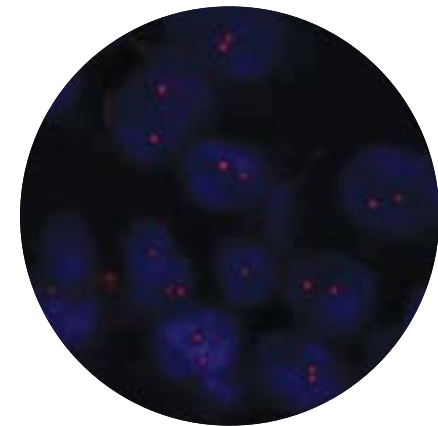
19q13

The 19q13 probe, labeled in red, is 357kb in size and covers the region between markers RH91552 and D19S902.

Probe Specification 19q13 Probe, 19q13.33, Red



CMP-S068 v001

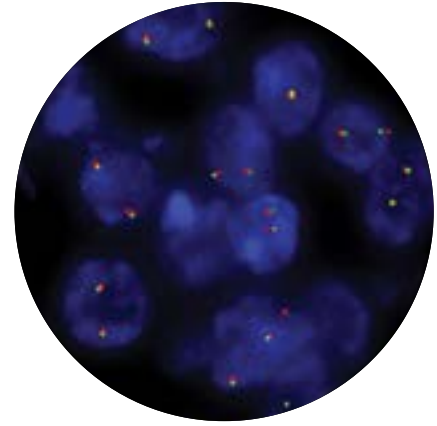
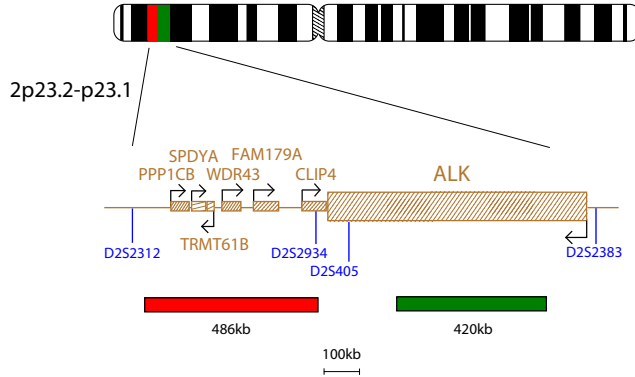


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

ALK Breakapart

The ALK Breakapart probe consists of a green 420kb probe, covering part of the ALK gene and a red 486kb probe, which is telomeric to the ALK gene.

Probe Specification ALK, 2p23.2-p23.1, Red
ALK, 2p23.2-p23.1, Green



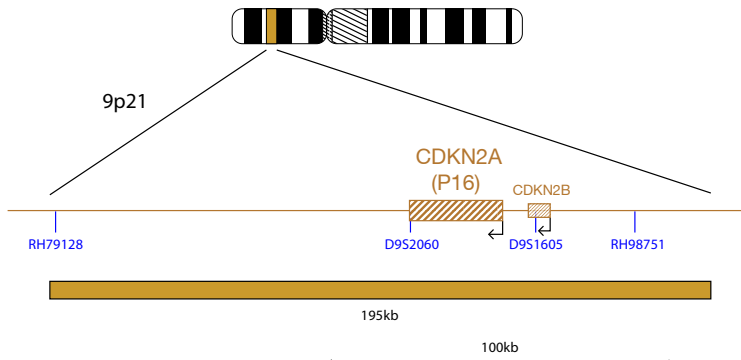
CMP-S023 v004

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

CDKN2A Probe Gold*

The Cytocell Aquarius® CDKN2A Probe covers CDKN2A (P16) gene and flanking regions, and is labeled in gold.

Probe Specification CDKN2A, 9p21, Gold



CMP-S032 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

* For sale in the US only.

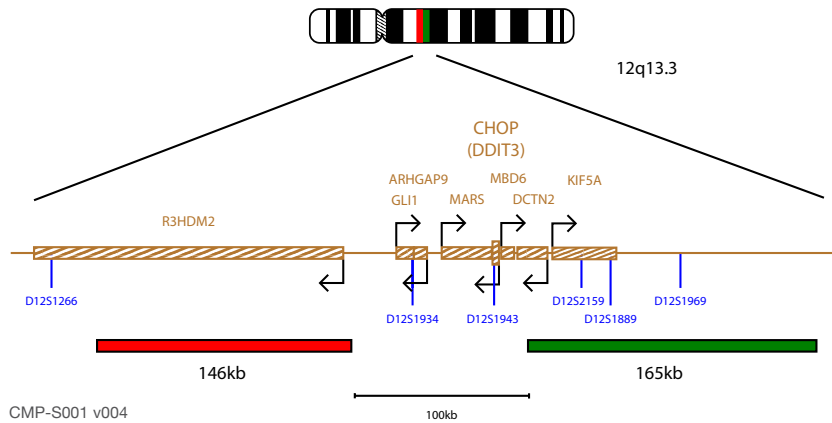
This product has not been licensed in accordance with Canadian law.



CHOP (DDIT3) Breakapart

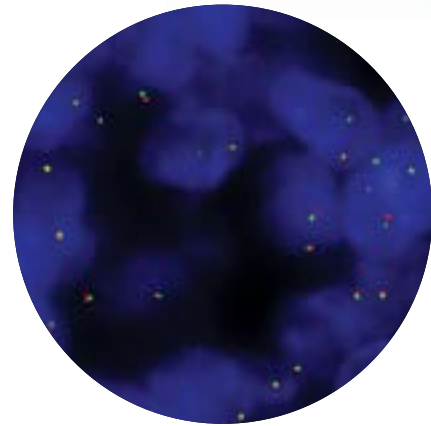
The CHOP Breakapart probe consists of a green 165kb probe and a red 146kb probe, which are positioned on each side of the CHOP (DDIT3) gene.

Probe Specification
CHOP, 12q13.3, Red
CHOP, 12q13.3, Green



CMP-S001 v004

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



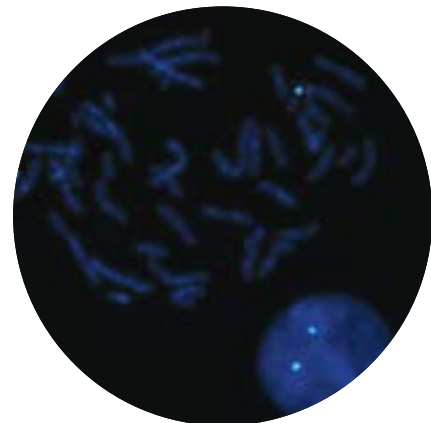
Centromere 17 Probe Aqua*

The Cytocell Aquarius® Centromere 17 Probe covers the chromosome 17 centromere (D17Z1) region and is labeled in aqua.

Probe Specification
17cen, D17Z1, Aqua



CMP-S031 v003



* For sale in the US only.
This product has not been licensed in accordance with Canadian law.

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

Centromere 7 Probe Green*

The Cytocell Centromere 7 Probe covers the chromosome 7 centromere (D7Z1) region and is labeled in green.

Probe Specification 7cen, D7Z1, Green



CMP-S030 v003



* For sale in the US only.
This product has not been licensed in accordance with Canadian law.

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

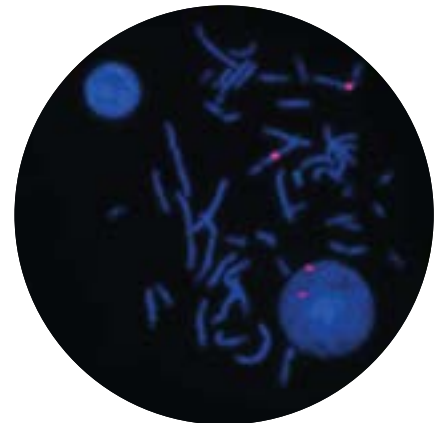
Centromere 3 Probe Red*

The Cytocell Centromere 3 Probe covers the chromosome 3 centromere (D3Z1) region and is labeled in red.

Probe Specification 3cen, D3Z1, Red



CMP-S029 v003



* For sale in the US only.
This product has not been licensed in accordance with Canadian law.

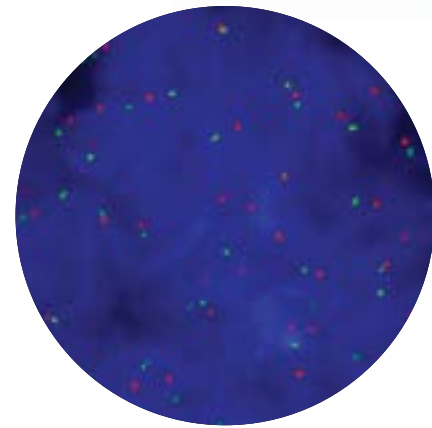
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



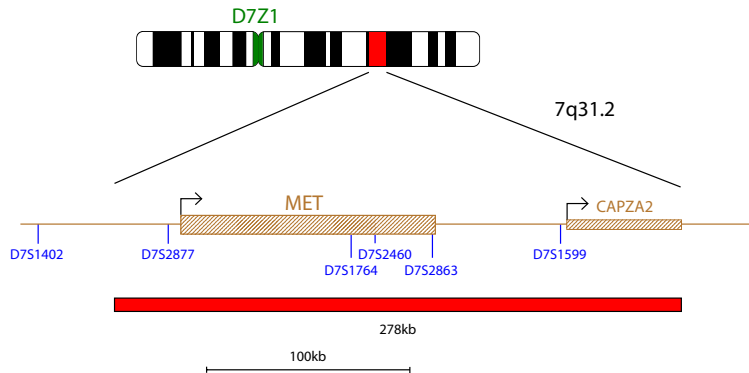


C-MET (MET) Amplification

The C-MET Amplification probe consists of a 278kb red probe spanning the C-MET (MET) gene. The centromeric probe in green acts as a control for chromosome 7.



Probe Specification
C-MET, 7q31.2, Red
D7Z1, 7p11.1-q11.1, Green

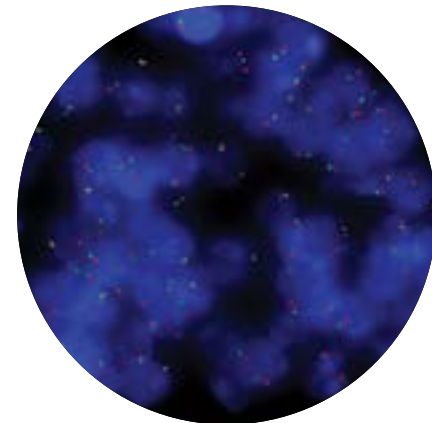


CMP-S002 v003

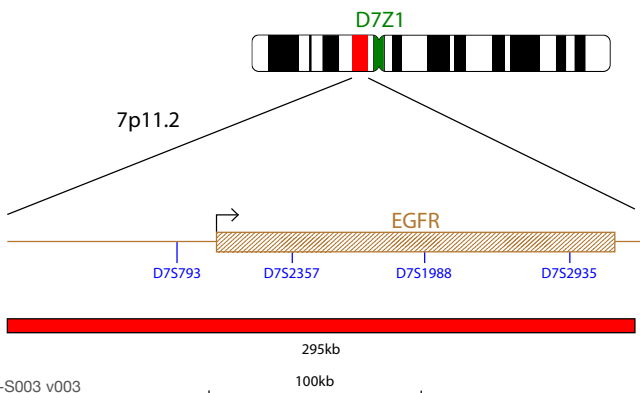
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

EGFR Amplification

The EGFR Amplification probe consists of a red 295kb probe spanning the gene region 7p11.2. The centromeric probe in green acts as a control for chromosome 7.



Probe Specification
EGFR, 7p11.2, Red
D7Z1, 7p11.1-q11.1, Green



CMP-S003 v003

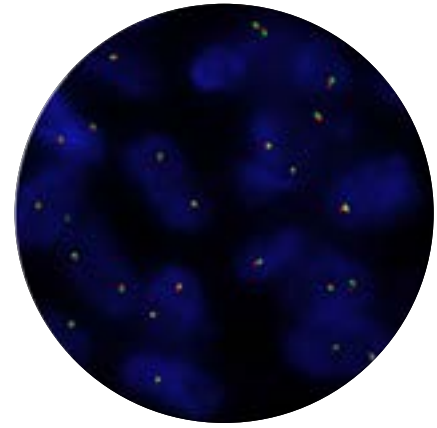
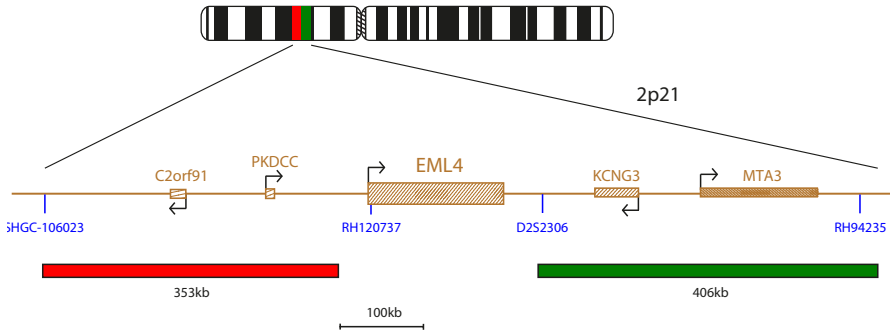
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



EML4 Breakapart

The EML4 Breakapart probe consists of a green 406kb probe and a red 353kb probe, which are positioned on each side of the EML4 gene.

Probe Specification EML4, 2p21, Red
EML4, 2p21, Green



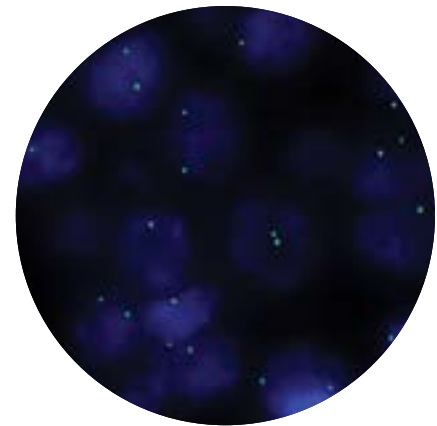
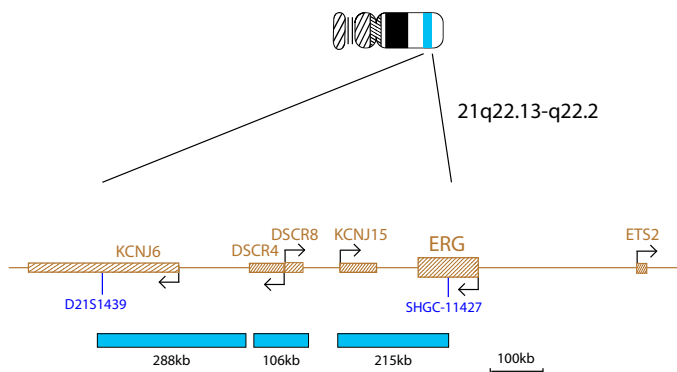
CMP-S024 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

ERG Control

The ERG Control probe consists of three 288kb, 106kb and 215kb blue probes that cover the centromeric (3') region of the ERG gene up to the D21S1439 marker.

Probe Specification 21q22.13-q22.2, Blue



CMP-S026 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

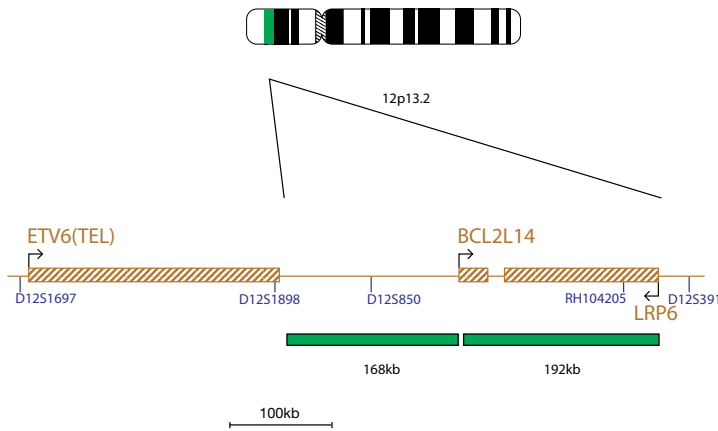




ETV6 Proximal Probe Green

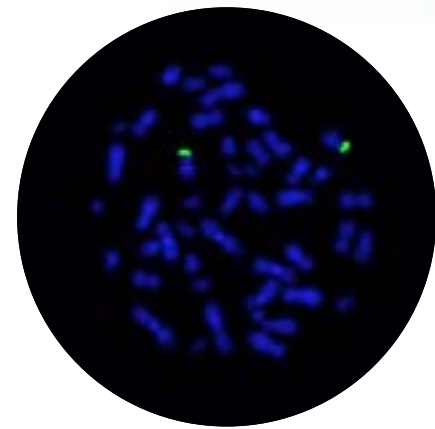
The ETV6 Proximal probe, labeled in green, contains two probes (168kb and 192kb) that map proximal to the ETV6 gene, covering D12S850 and RH104205 markers.

Probe Specification ETV6 Proximal, 12p13.2, Green



CMP-H120 v001

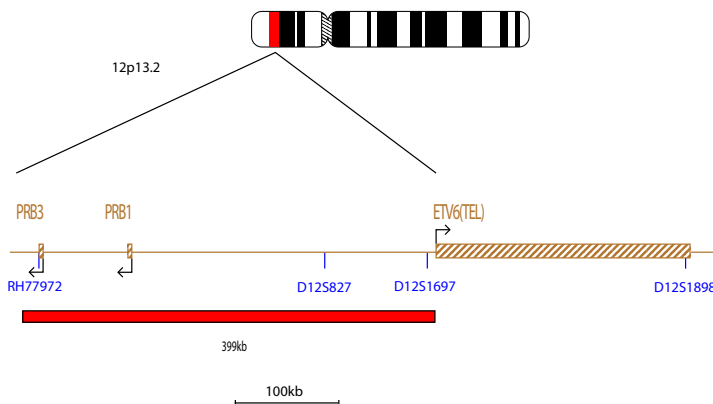
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



ETV6 Distal Probe Red

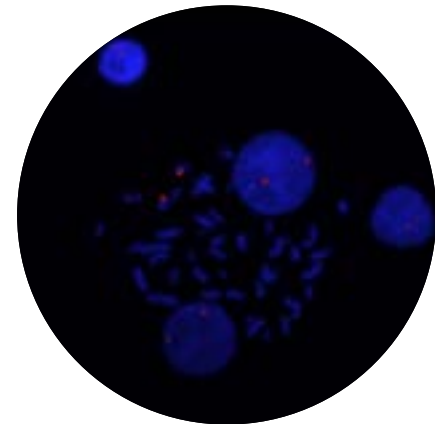
The ETV6 Distal probe, labeled in red, consists of a 399kb probe distal to the ETV6 gene, covering RH77972 and D12S1697 markers.

Probe Specification ETV6 Distal, 12p13.2, Red



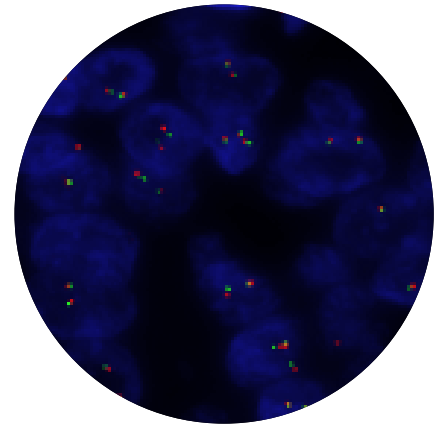
CMP-H121 v001

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

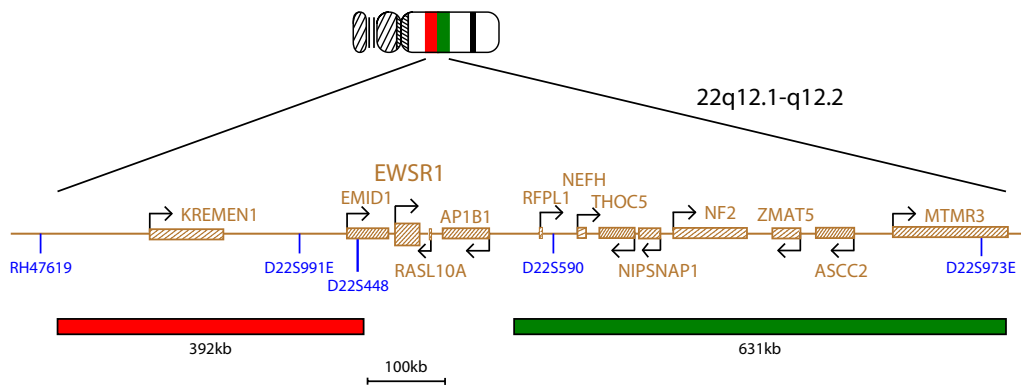


EWSR1 Breakapart

The EWSR1 Breakapart probe contains a red 392kb probe and a green 631kb probe, which are positioned on each side of the EWSR1 gene.



Probe Specification
EWSR1, 22q12.1-q12.2, Red
EWSR1, 22q12.1-q12.2, Green

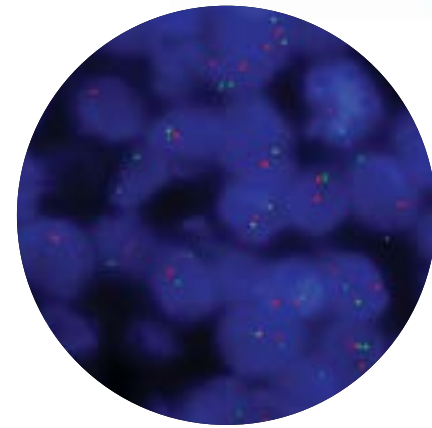


CMP-S004 v004

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

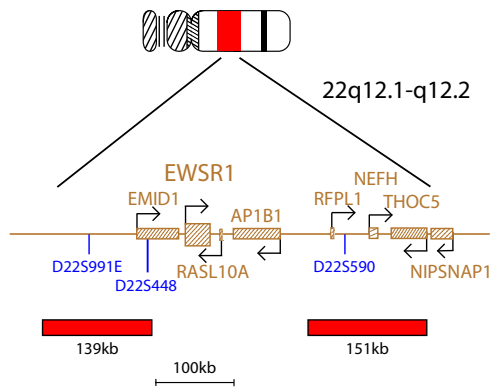


EWSR1/ERG Translocation, Dual Fusion

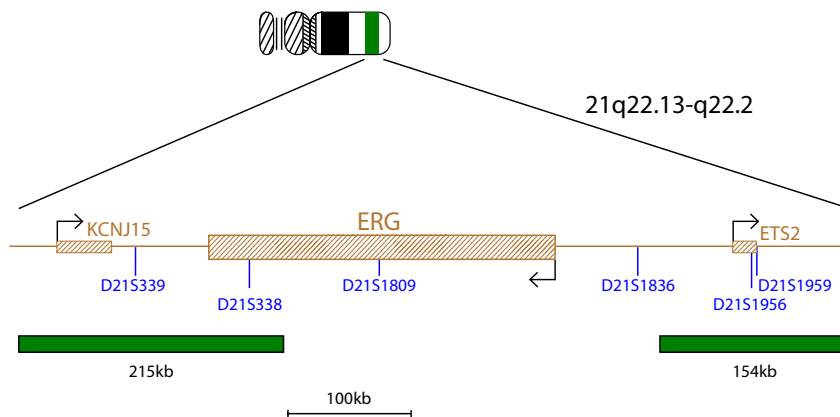


The EWSR1/ERG Translocation probe consists of green probes (215kb, 154kb) flanking the breakpoint region at the ERG gene locus and red probes (139kb, 151kb) flanking the breakpoint region at the EWSR1 locus.

Probe Specification EWSR1, 22q12.1-q12.2 Red
 ERG, 21q22.13-q22.2 Green



CMP-S005 v003



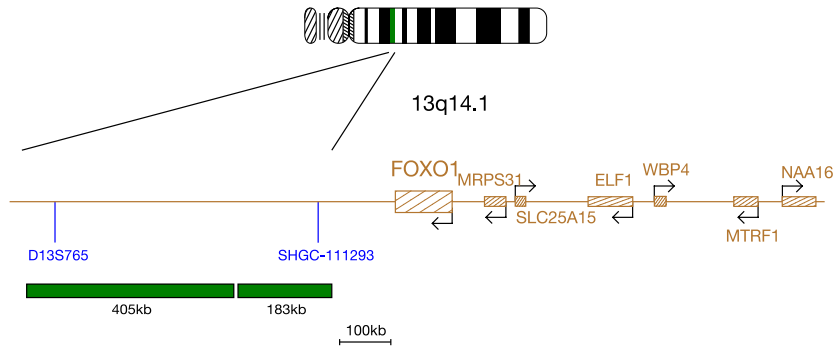
CMP-S006 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

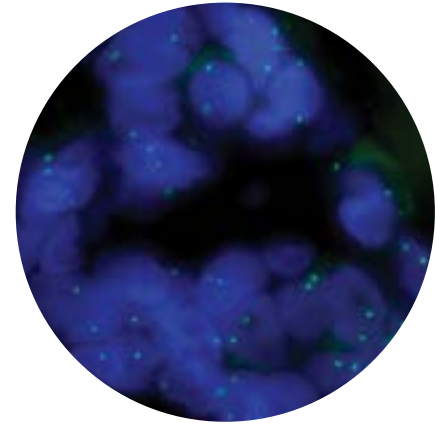
FOXO1 Proximal Green

The FOXO1 Proximal Probe, labeled in green, consists of two probes (405kb, and 183kb) that cover regions including markers D13S765 and SHGC-111293.

Probe Specification 13q14.1 Green



CMP-S074 v003

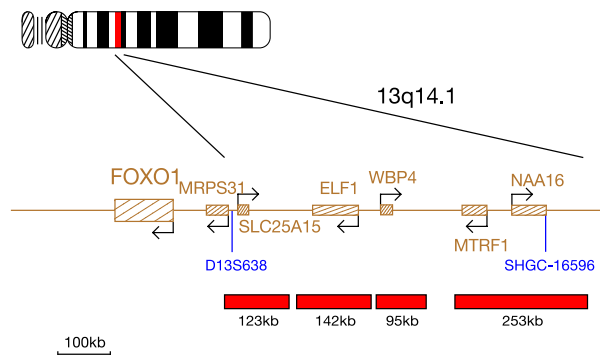


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

FOXO1 Distal Red

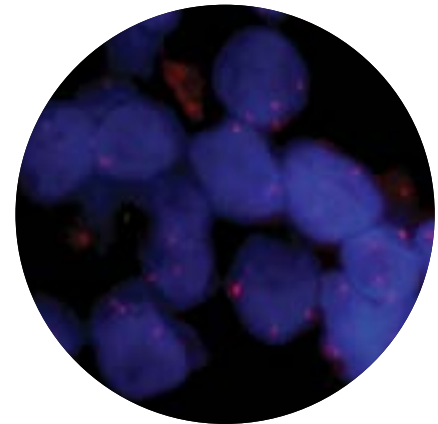
The FOXO1 Distal Probe, labeled in red, consists of four probes (123kb, 142kb, 95kb and 253kb) that cover regions including markers D13S638 and SHGC-16596.

Probe Specification 13q14.1, Red



CMP-S075 v003

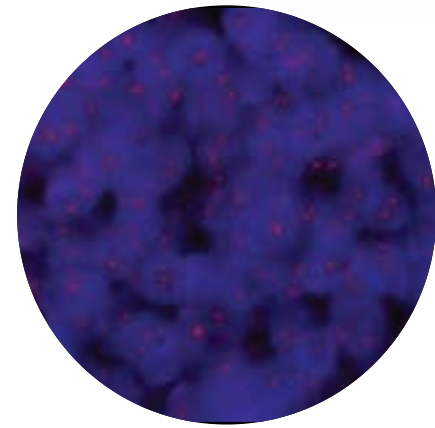
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



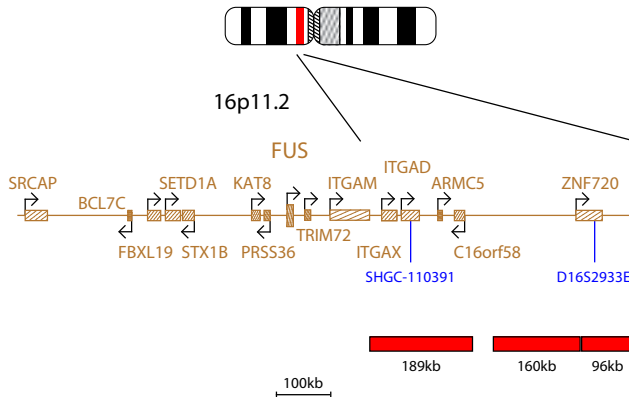


FUS Proximal Red

The FUS Proximal Probe, labeled in red, consists of three probes (189kb, 160kb and 96kb) that cover regions including the markers SHGC-110391 and D16S2933E.



Probe Specification 16p11.2 Red

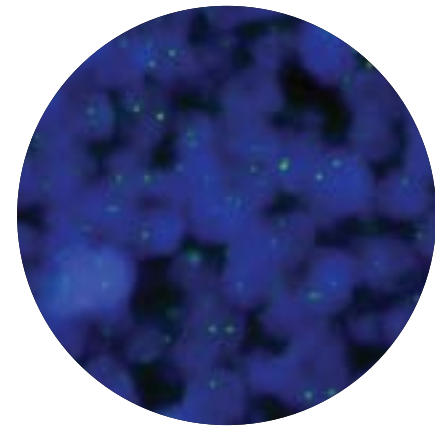


CMP-S076 v001

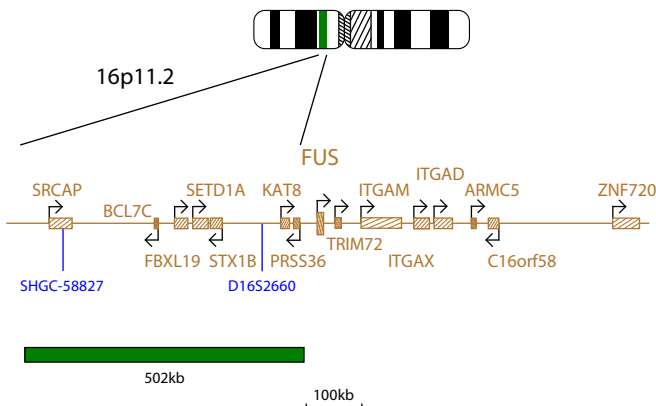
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

FUS Distal Green

The FUS Distal Probe, labeled in green, is 502kb in size and covers the region including markers SHGC-58827 and D16S2660.



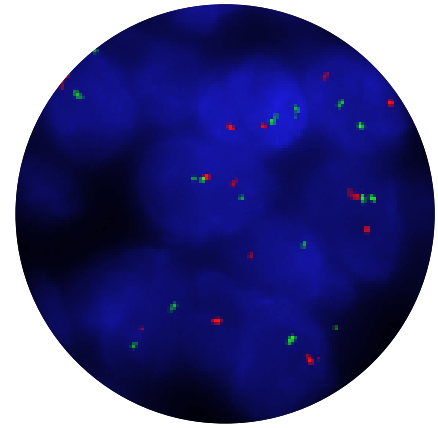
Probe Specification 16p11.2 Green



CMP-S077 v002

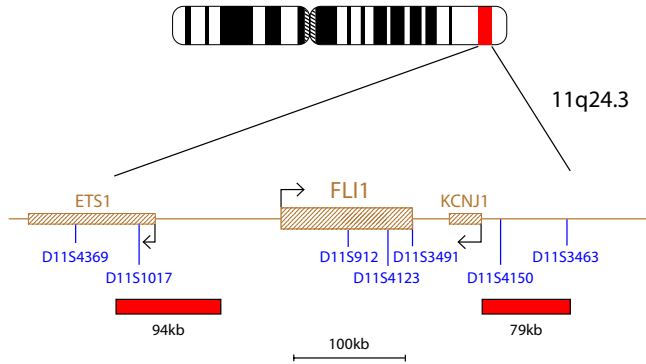
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

FLI1/EWSR1 Translocation, Dual Fusion

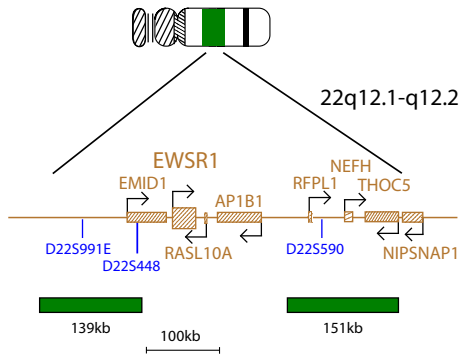


The FLI1/EWSR1 Translocation probe consists of green probes (139kb, 151kb) flanking the breakpoint region at the EWSR1 gene locus and red probes (94kb, 79kb) flanking the breakpoint region at the FLI1 locus.

Probe Specification FLI1, 11q24.3, Red
 EWSR1, 22q12.1-q12.2 Green



CMP-S007 v003



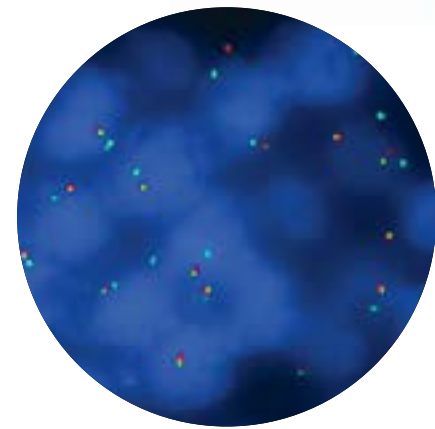
CMP-S008 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

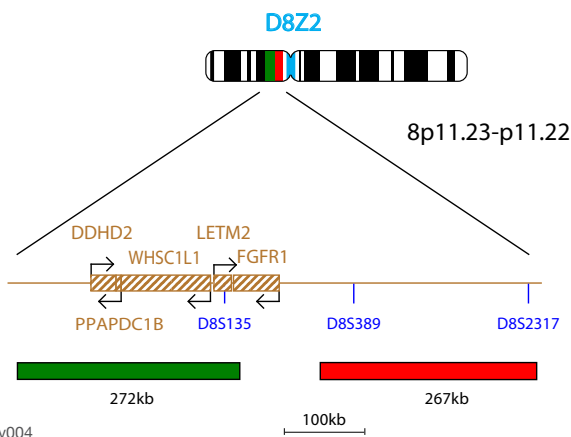


FGFR1 Breakapart/Amplification

The FGFR1 Breakapart/Amplification probe consists of a green 272kb probe and a red 267kb probe which are positioned on each side of the FGFR1 gene. The 8-centromere probe in blue acts as a control for chromosome 8.



Probe Specification
 FGFR1, 8p11.23-p11.22, Red
 FGFR1, 8p11.23-p11.22, Green
 D8Z2, 8p11.1-q11.1, Blue

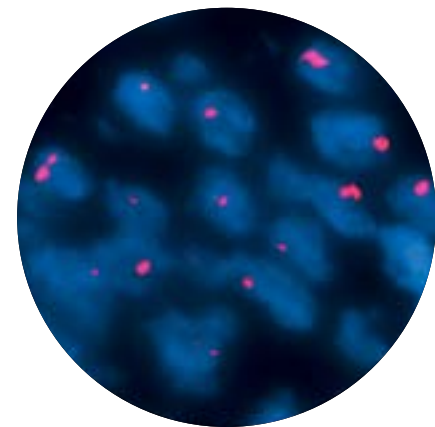


CMP-S009 v004

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

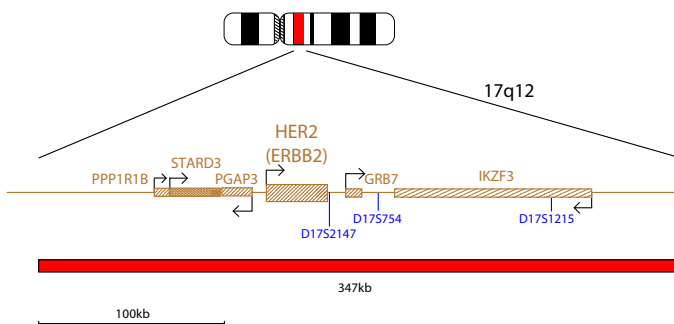
HER2 (ERBB2) Amplification

The HER2 amplification probe consists of a 347kb probe labeled in red, spanning the ERBB2 gene and neighboring regions.



(Amplified HER2)

Probe Specification
 HER2 (ERBB2), 17q12, Red



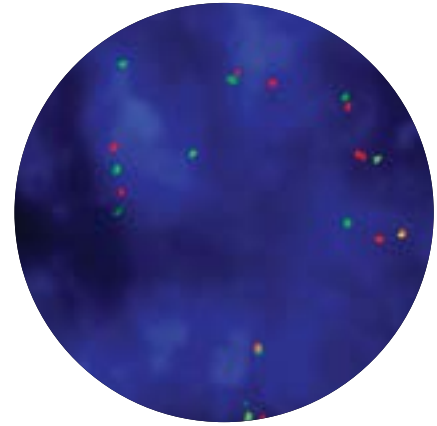
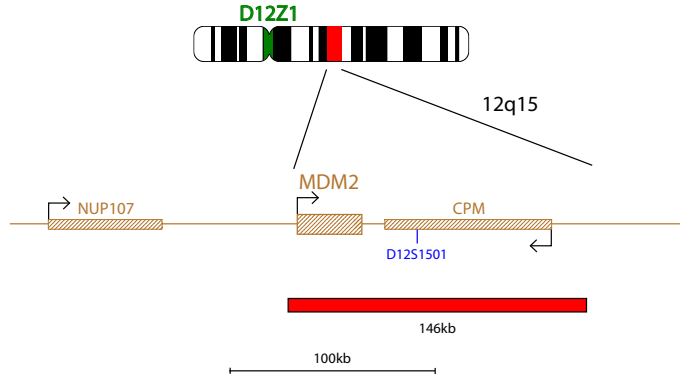
CMP-S021 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

MDM2 Amplification

The MDM2 amplification probe consists of a 146kb red probe spanning the MDM2 gene and neighboring CPM gene, with a green centromeric probe for chromosome 12 provided as a control.

Probe Specification MDM2, 12q15, Red
D12Z1, 12p11.1-q11.1, Green



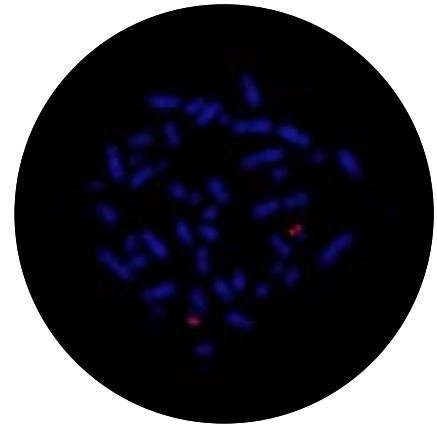
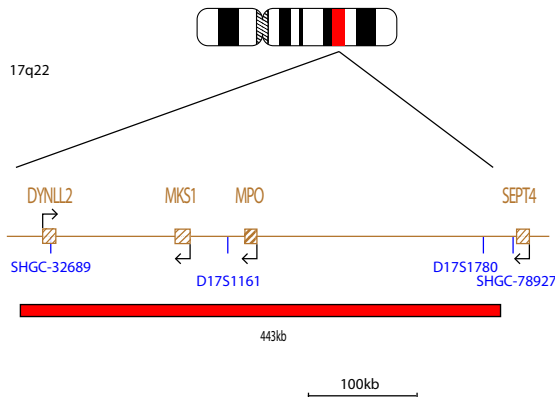
CMP-S012 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

MPO Probe Red

The MPO probe, labeled in red, covers a 443kb region including the MPO gene.

Probe Specification MPO, 17q22, Red



CMP-H109 v001

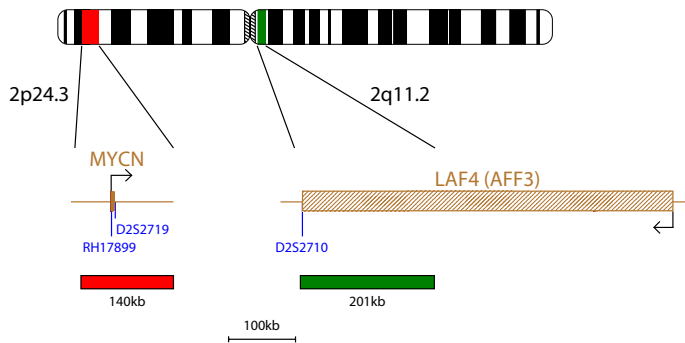
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



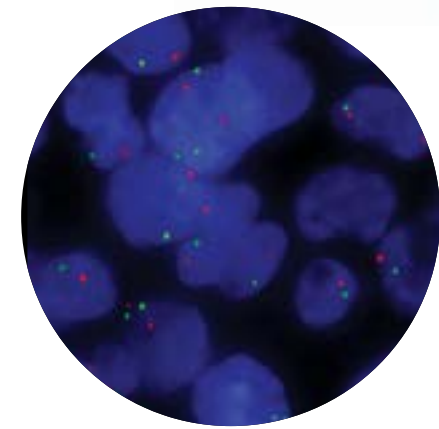
N-MYC (MYCN) Amplification

The N-MYC (MYCN) Amplification probe consists of a red 140kb probe spanning the MYCN gene region at 2p24.3. The LAF4 (AFF3) gene probe in green at 2q11 acts as a control for chromosome 2.

Probe Specification N-MYC, 2p24.3, Red
LAF4, 2q11.2, Green



CMP-S013 v004

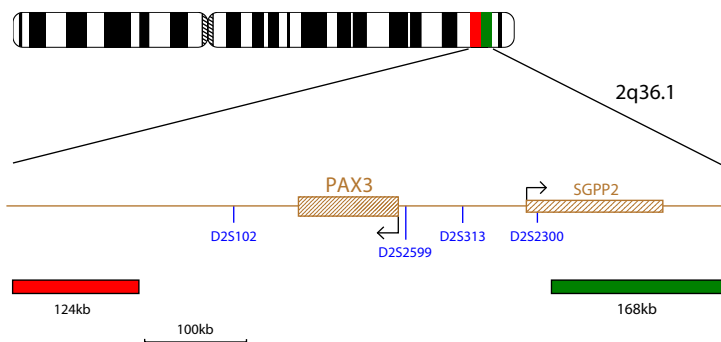


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

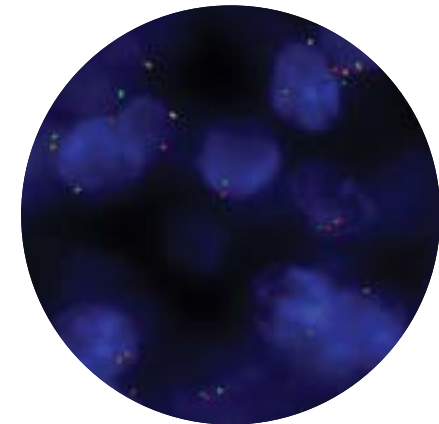
PAX3 Breakapart

The PAX3 Breakapart probe consists of a green 168kb probe and a red 124kb probe, which are positioned on each side of the PAX3 gene.

Probe Specification PAX3, 2q36.1, Red
PAX3, 2q36.1, Green



CMP-S014 v003

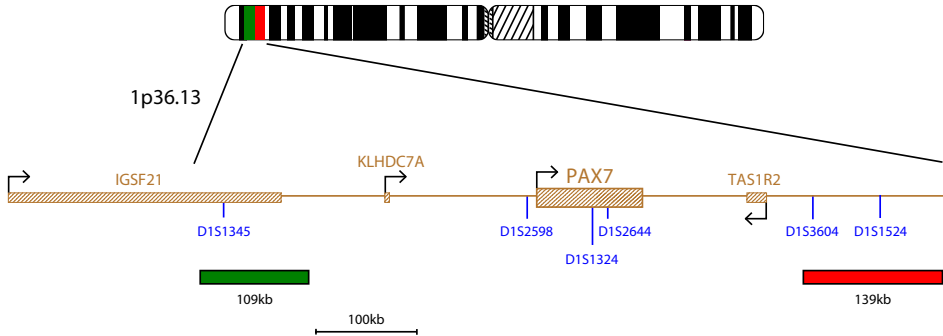
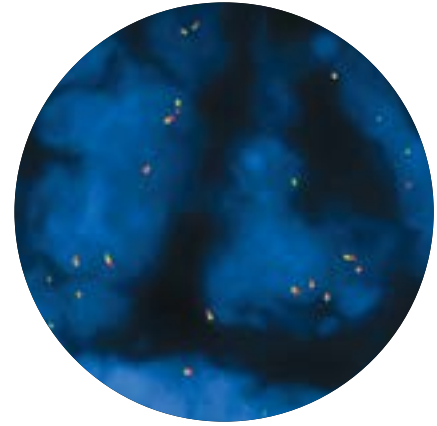


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

PAX7 Breakapart

The PAX7 Breakapart probe consists of a green 109kb probe and a red 139kb probe, which are positioned on each side of the PAX7 gene.

Probe Specification
PAX7, 1p36.13, Red
PAX7, 1p36.13, Green



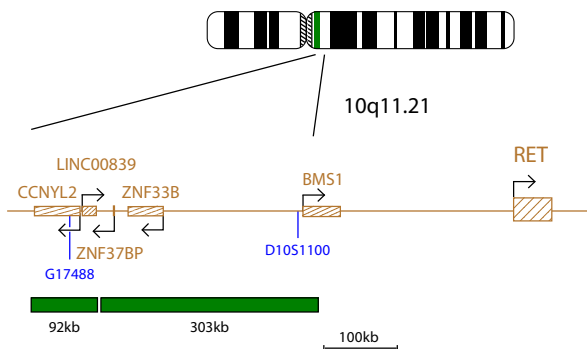
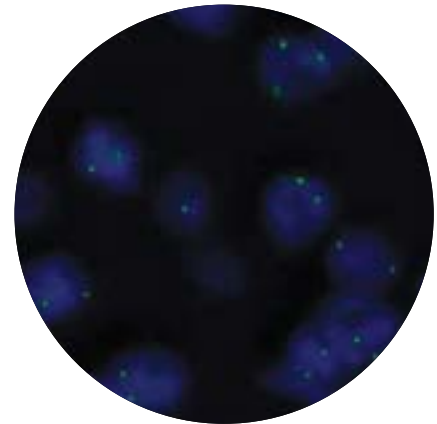
CMP-S015 v004

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

RET Proximal

The RET Proximal probe consists of two probes (92kb and 303kb), labeled in green, situated proximal to the RET gene and covering markers G17488 and D10S1100.

Probe Specification
RET Proximal, 10q11.21, Green



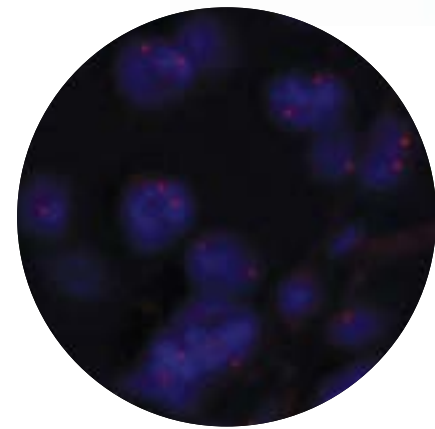
CMP-S057 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

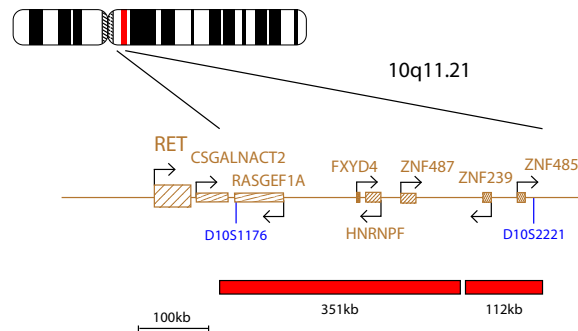


RET Distal

The RET Distal probe consists of two probes (351kb and 112kb), labeled in red, situated distal to the RET gene and covering markers D10S1176 and D10S2221.



Probe Specification RET Distal, 10q11.21, Red

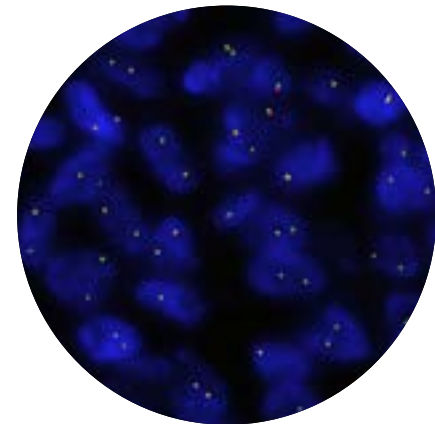


CMP-S056 v002

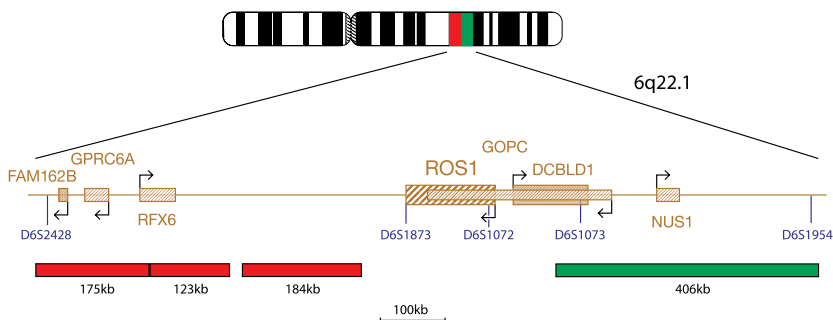
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

ROS1 Breakapart

The ROS1 Breakapart probe consists of a green 406kb probe and three red 175kb, 123kb and 184kb probes, which are positioned on each side of the ROS1 gene.



Probe Specification ROS1, 6q22.1, Red
ROS1, 6q22.1, Green



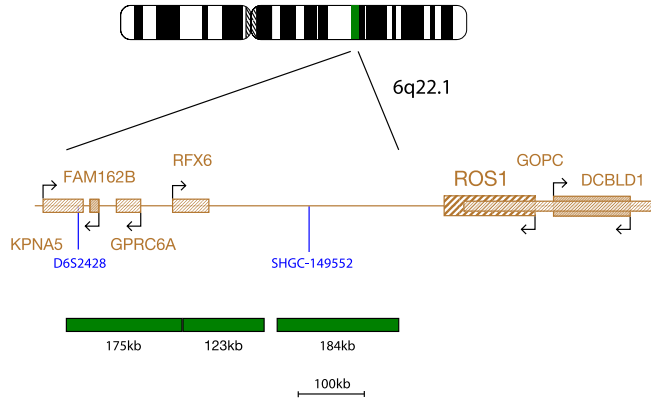
CMP-S045 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

ROS1-GOPC (FIG) Proximal

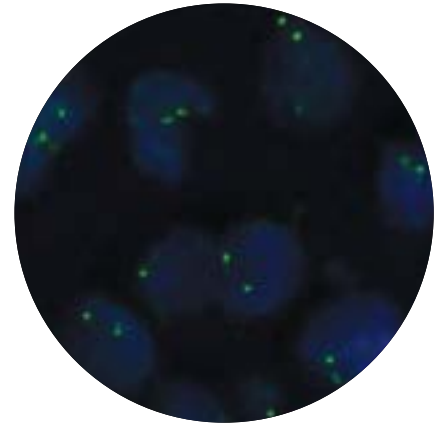
The ROS1-GOPC(FIG) Proximal probe consists of 3 probes (175kb, 123kb, 184kb), labeled in green, situated proximal to the ROS1 gene and including markers D6S2428 and SHGC-149552.

Probe Specification ROS1-GOPC (FIG) Proximal, 6q22.1, Green



CMP-S059 v003

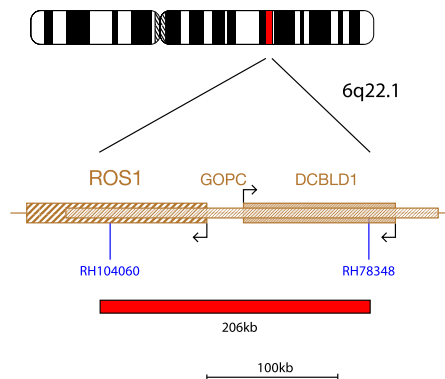
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



ROS1-GOPC (FIG) Distal

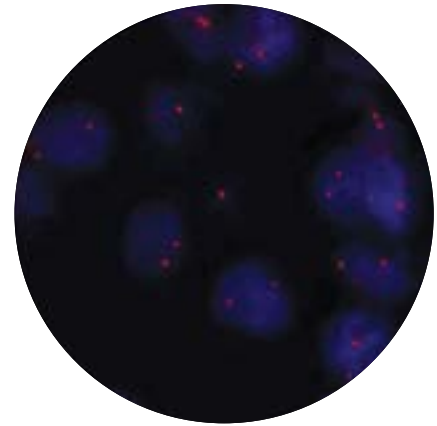
The ROS1-GOPC (FIG) Distal probe consists of a red probe (206kb) covering the distal part of the ROS1 gene and a region up to RH78348.

Probe Specification ROS1-GOPC (FIG) Distal, 6q22.1, Red



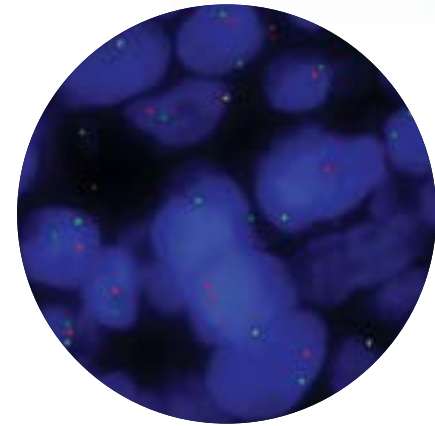
CMP-S058 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

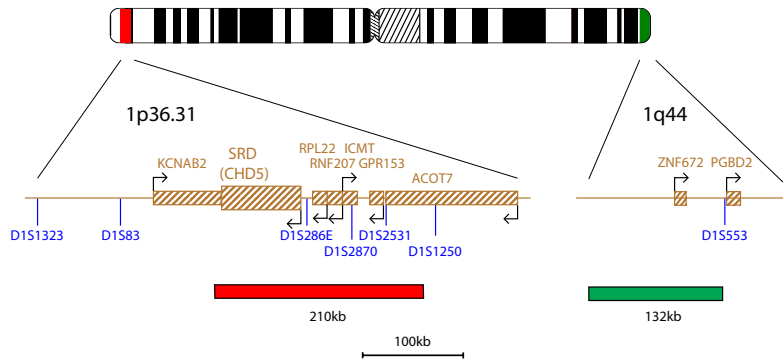


SRD (CHD5) Deletion

The SRD (CHD5) Deletion probe consists of a 210kb red probe spanning the SRD (CHD5) gene region. The 1qter probe in green acts as a control for chromosome 1.



Probe Specification SRD (CHD5), 1p36.31, Red
1qter, 1q44, Green

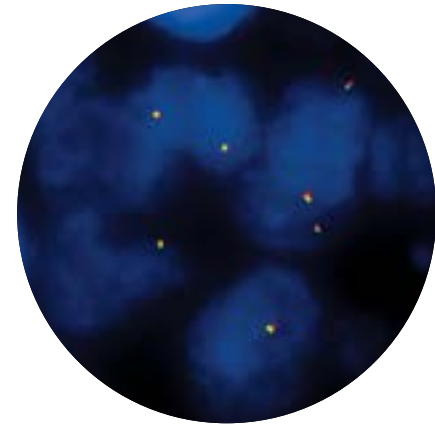


CMP-S017 v004

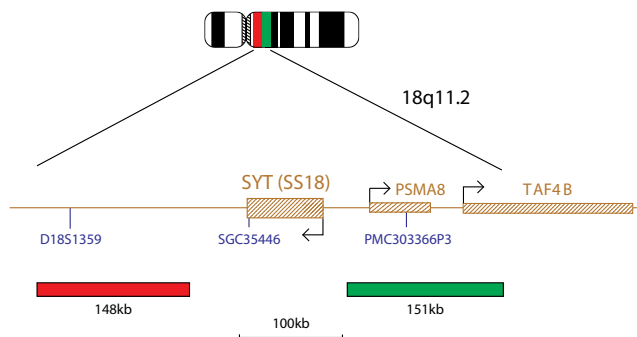
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

SYT (SS18) Breakapart

The SYT (SS18) Breakapart probe consists of a green 151kb probe and a red 148kb probe, which are positioned on each side of the the SYT (SS18) gene.



Probe Specification SYT, 18q11.2, Red
SYT, 18q11.2, Green

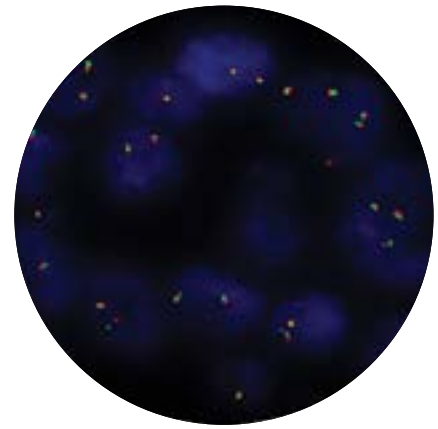


CMP-S018 v003

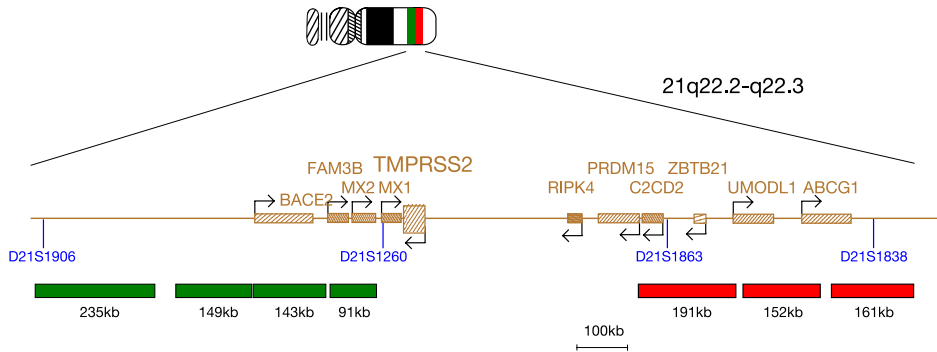
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

TMPRSS2 Breakapart

The TMPRSS2 Breakapart Probe consists of three red probes (191kb, 152kb and 161kb), which are telomeric to the TMPRSS2 gene and begin centromeric to the D21S1863 marker. Four green probes (235kb, 149kb, 143kb and 91kb) are also included that are centromeric to the TMPRSS2 gene and begin centromeric to the D21S1906 marker.



Probe Specification TMPRSS2, 21q22.2-q22.3, Red
 TMPRSS2, 21q22.2-q22.3, Green

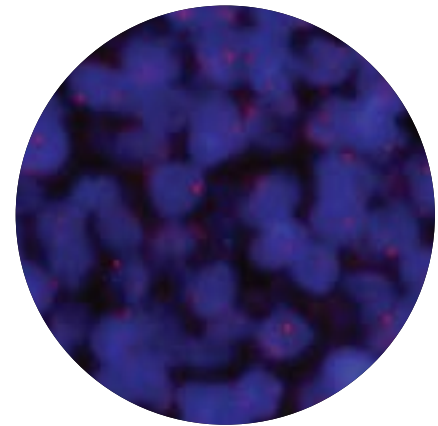


CMP-S025 v004

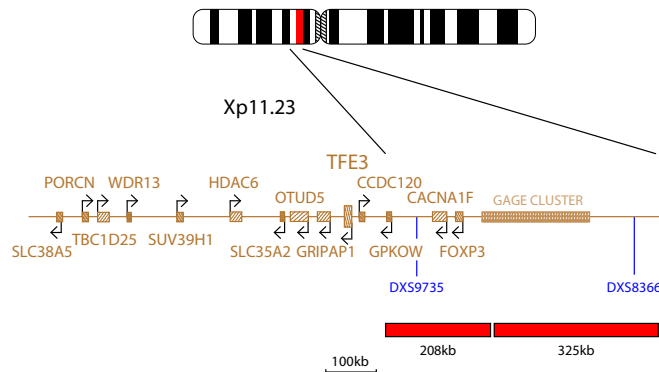
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

TFE3 Proximal Probe Red

The TFE3 Proximal Probe, labeled in red, consists of two probes (208kb and 325kb) that cover regions including the markers DXS9735 and DXS8366.



Probe Specification TFE3 Proximal Probe, Xp11.23, Red



CMP-S078 v001

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

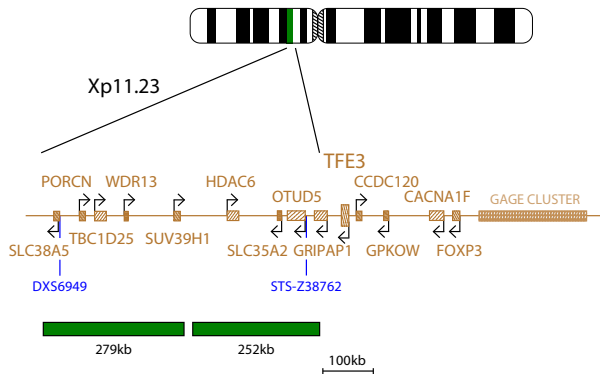




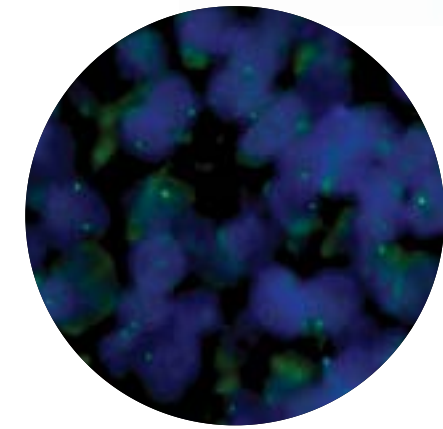
TFE3 Distal Probe Green

The TFE3 Distal Probe, labeled in green, consists of two probes (279kb and 252kb) that cover the regions including markers DXS6949 and STS-Z38762.

Probe Specification TFE3 Distal Probe, Xp11.23, Green



CMP-S079 v001

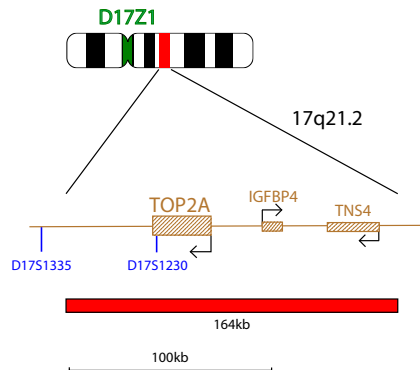


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

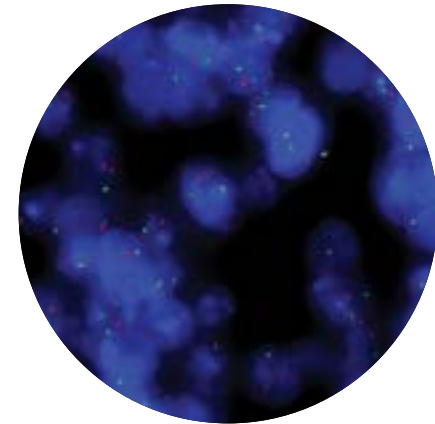
TOP2A Amplification/Deletion

The TOP2A Amplification/Deletion probe consists of a red 164kb probe spanning the TOP2A gene region. The centromeric probe in green acts as a control for chromosome 17.

Probe Specification TOP2A, 17q21.2, Red
D17Z1, 17p11.1-q11.1, Green



CMP-S019 v003

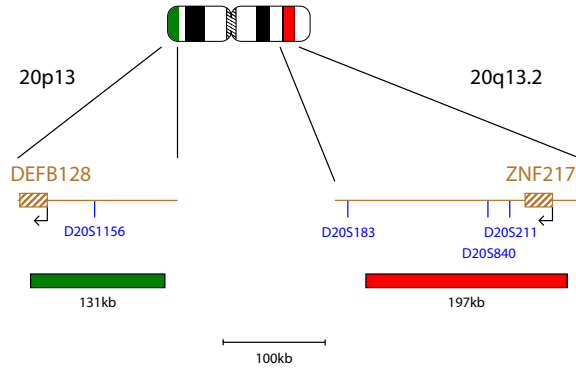


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

ZNF217 Amplification

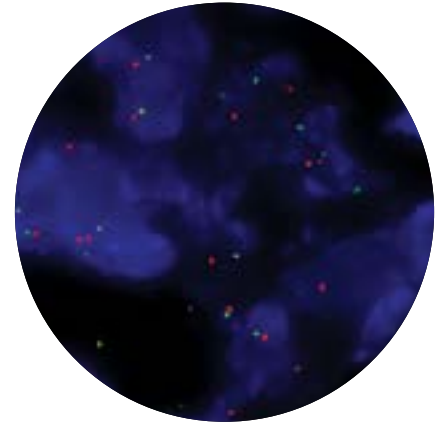
The ZNF217 Amplification probe consists of a 197kb red probe spanning the ZNF217 gene and neighboring regions. The accompanying 20pter probe in green acts as a control for chromosome 20.

Probe Specification ZNF217, 20q13.2, Red
20pter, 20p13, Green



CMP-S020 v004

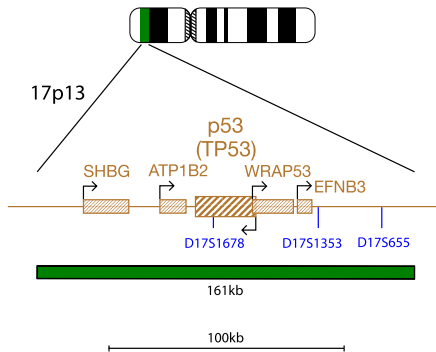
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



TP53 Probe Green

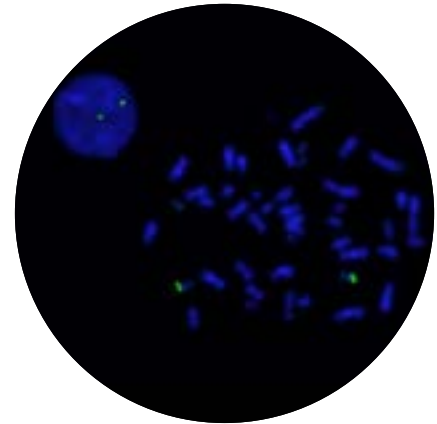
The TP53 probe, labeled in green, covers a 161kb region including the TP53 gene.

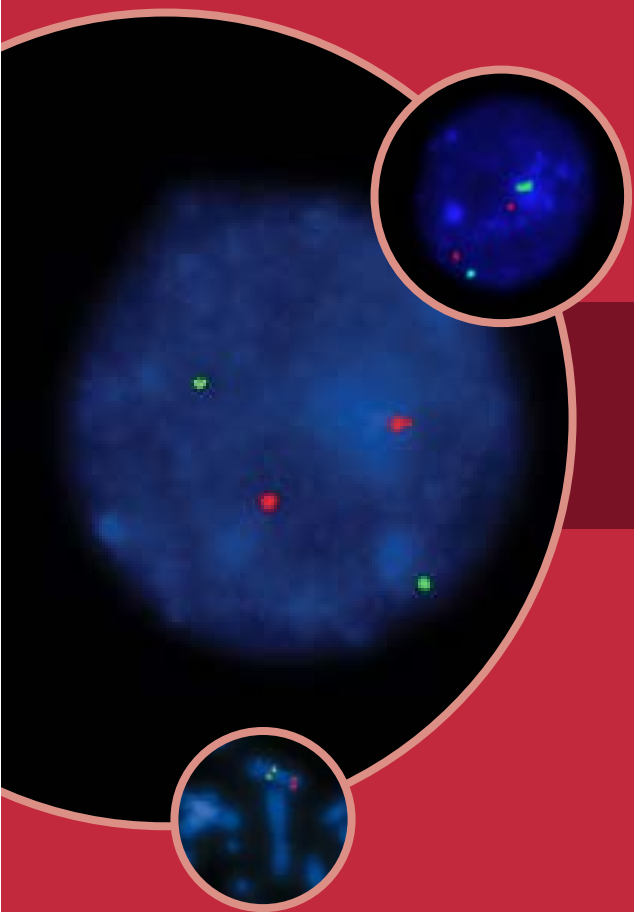
Probe Specification TP53, 17p13, Green



CMP-H110 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.





Microdeletion



Contents

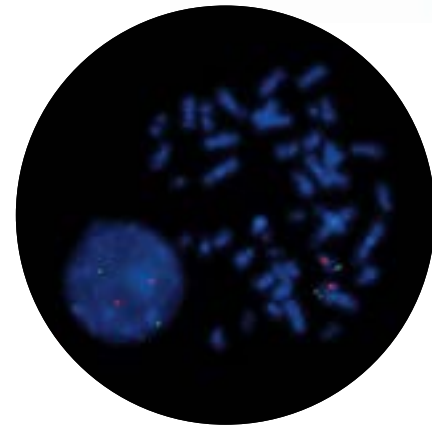
- 113 Angelman (UBE3A/D15S10) Region
- 114 Cri-du-chat Region/Sotos Region
- 115 DiGeorge II (10p14)
- 116 DiGeorge/VCFS TUPLE1 Region and 22q13.3 Region
- 117 DiGeorge/VCFS N25 Region and 22q13.3 Region
- 118 DiGeorge TBX1 Region and 22q13.3 Region
- 119 Kallmann (KAL1) Region/STS Region
- 120 Prader-Willi/Angelman (SNRPN) Region
- 121 Saethre-Chotzen Region/Williams-Beuren Region
- 122 SHOX
- 123 Smith-Magenis (RAI1) Region/Miller-Dieker Region
- 124 SRY
- 125 Williams-Beuren Region
- 126 Wolf-Hirschhorn Region

Microdeletion

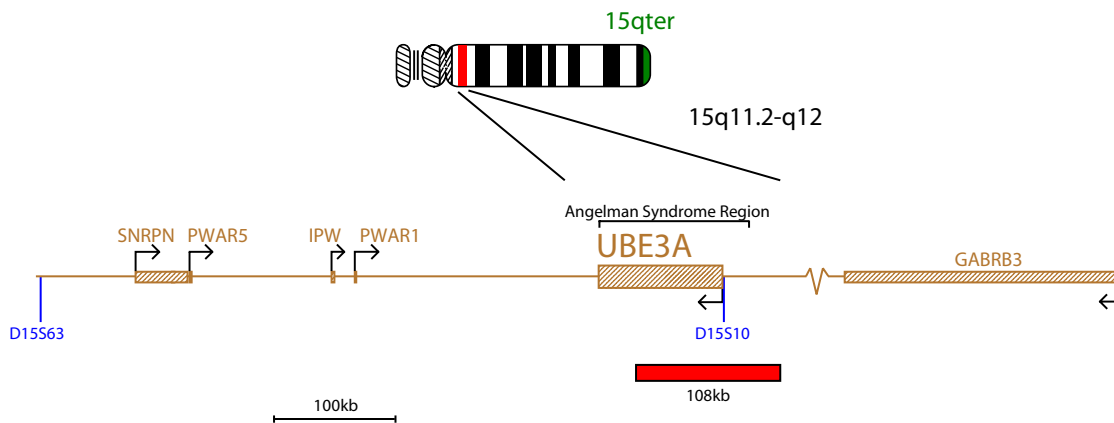
Cytocell offers a range of Microdeletion probes available in the Aquarius® liquid format. These probes are directly labeled and ready to use in hybridization buffer.

Angelman (UBE3A/D15S10) Region

The Angelman (UBE3A/D15S10) probe is 108kb, labeled in red, and covers most of the UBE3A gene and includes the D15S10 marker. The 15qter subtelomere specific probe (clone 154P1), labeled in green, allows identification of chromosome 15 and acts as a control probe.



Probe Specification UBE3A, 15q11.2-q12, Red
 15qter, 15q26.3, Green

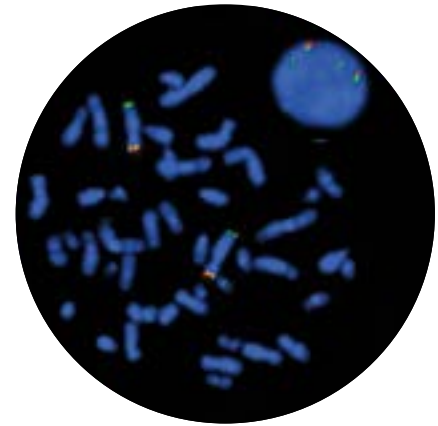


CMP-U002 v002

* For sale in the US only. This product has not been licensed in accordance with Canadian law.

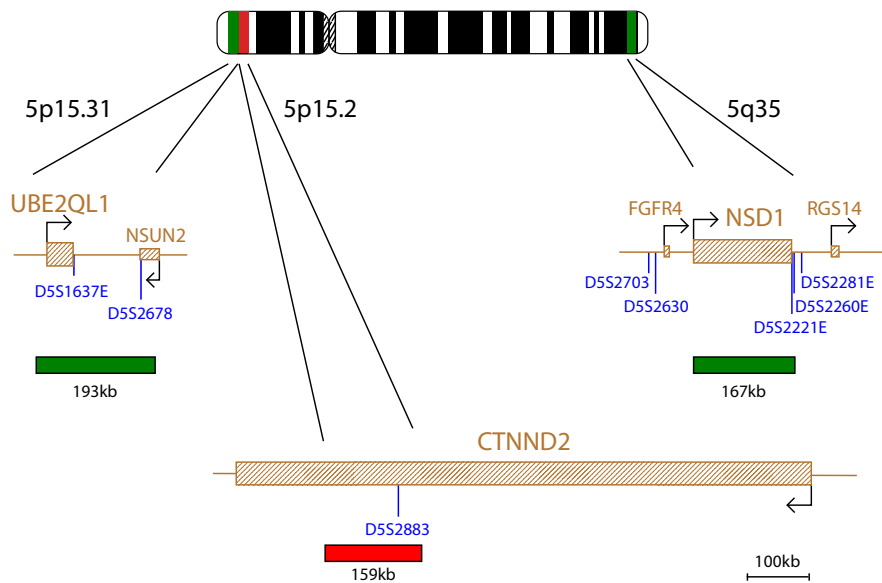
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

Cri-du-Chat Region/ Sotos Region



The CTNND2 probe is 159kb, labeled in red and covers a region including the D5S2883 marker. The UBE2QL1 probe is 193kb, labeled in green and covers a region including the D5S1637E and D5S2678 markers, as well as the entire UBE2QL1 gene. The Sotos probe is 167kb, labeled in green and covers the NSD1 gene. The three unique sequences act as control probes for each other and allow identification of chromosome 5.

Probe Specification
 Cri-du-chat (CTNND2), 5p15.2, Red
 Cri-du-chat (UBE2QL1), 5p15.31, Green
 Sotos, 5q35, Green



CMP-
U004 v002

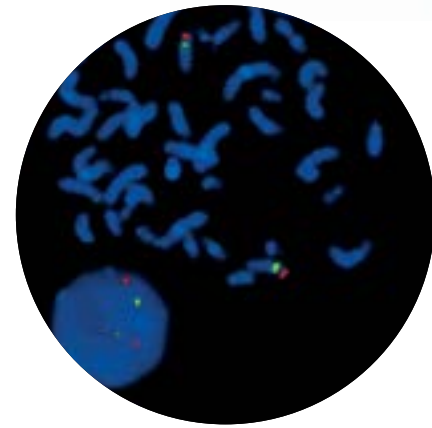
* For sale in the US only. This product has not been licensed in accordance with Canadian law.

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

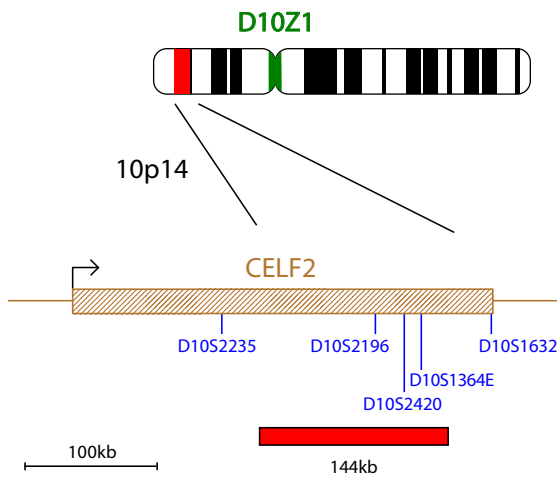


DiGeorge II (10p14)*

The CELF2 probe is 144kb, labeled in red and covers a region including the D10S2196, D10S2420 and D10S1364E markers. The probe mix also contains a control probe for the chromosome 10 centromere (D10Z1), labeled in green.



Probe Specification CELF2, 10p14, Red
D10Z1, 10p11.1-q11.1, Green



CMP-U005 v002

* For sale in the US only. This product has not been licensed in accordance with Canadian law.

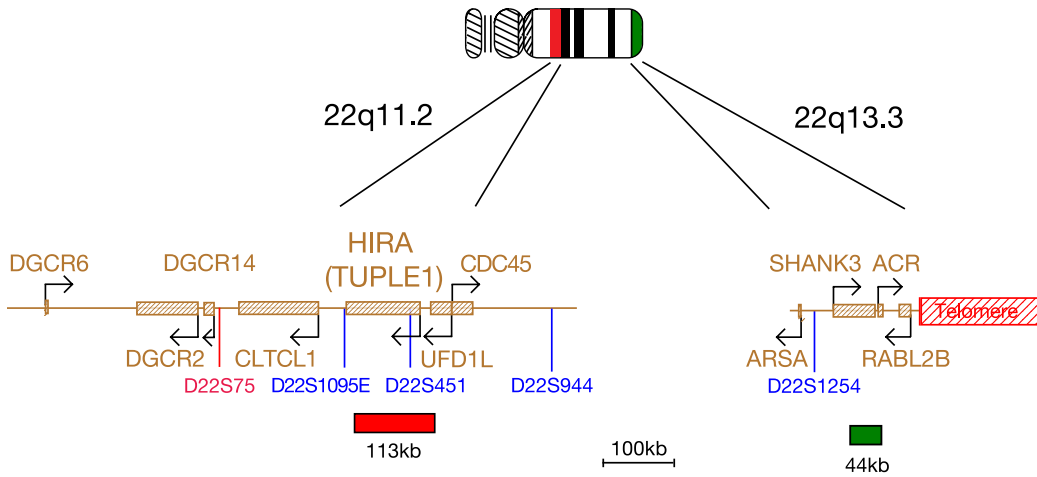
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

DiGeorge/VCFS TUPLE1 Region and 22q13.3 Region



The TUPLE1 probe is 113kb, labeled in red, and covers most of the TUPLE1 (HIRA) gene. The N85A3 (44kb) probe, labeled in green, is located within 22q13.3 and covers the telomeric end of the SHANK3 gene, allowing for identification of the most distal 22q13.3 deletions. The two unique sequences provide control probes for each other and allow identification of chromosome 22.

Probe Specification
 TUPLE1, 22q11.2, Red
 N85A3, 22q13.3, Green



CMP-U008 v005

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

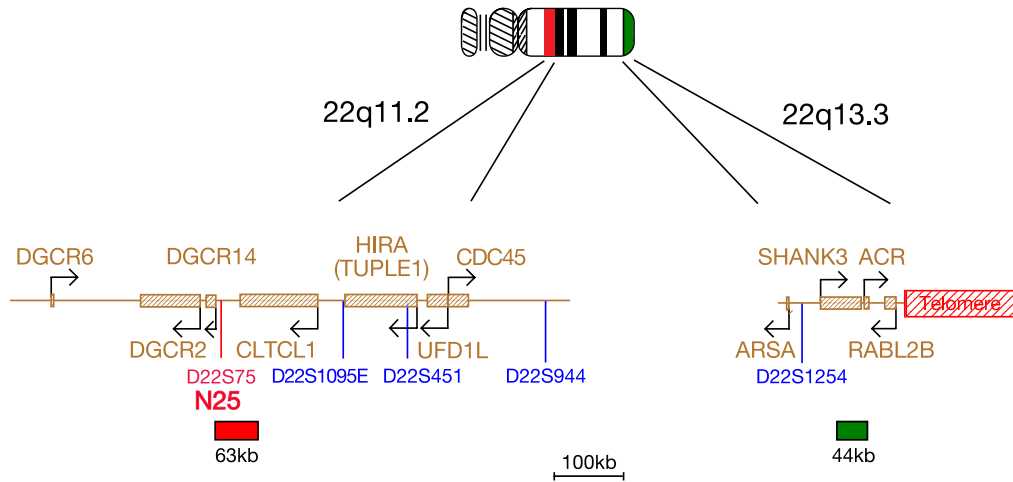


DiGeorge/VCFS N25 Region and 22q13.3 Region*



The N25 probe is 63kb, labeled in red and covers a region including the D22S75 marker and the centromeric end of the CLTCL1 gene. The N85A3 (44kb), labeled in green, is located within 22q13.3 band and covers the telomeric end of the SHANK3 gene allowing for identification of the most distal 22q13.3 deletions. The two unique sequences provide control probes for each other and allow identification of chromosome 22.

Probe Specification N25, 22q11.21, Red
 N85A3, 22q13.33, Green

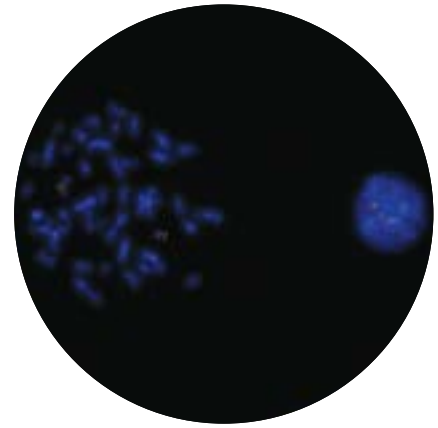


CMP-U007 v003

* For sale in the US only. This product has not been licensed in accordance with Canadian law.

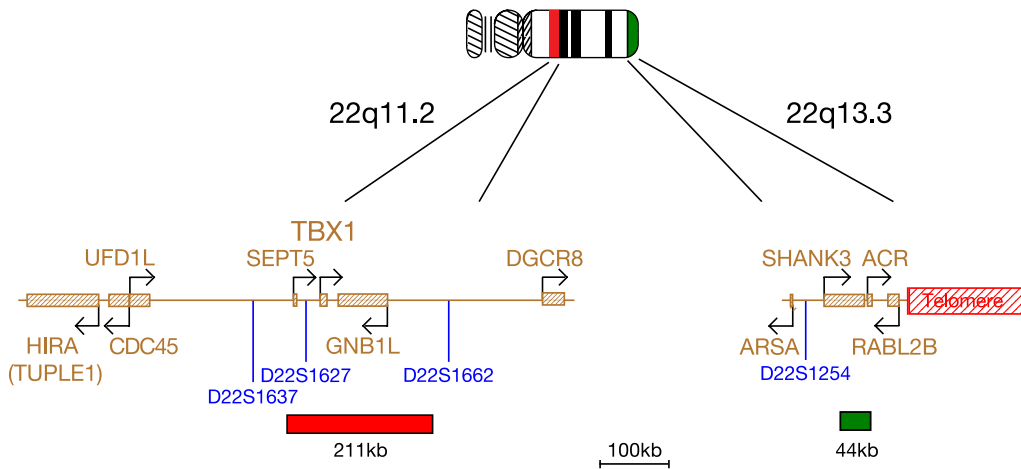
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

DiGeorge TBX1 Region and 22q13 Region*



The TBX1 probe is 211kb, labeled in red, covers the entire TBX1 gene and includes the D22S1627 marker. The N85A3 (44kb), labeled in green, is located within 22q13.33 and covers the telomeric end of the SHANK3 gene, allowing for identification of the most distal 22q13.3 deletions. The two unique sequences act as control probes for each other and allow identification of chromosome 22.

Probe Specification TBX1, 22q11.2, Red
 N85A3, 22q13.3, Green



CMP-U006 v003

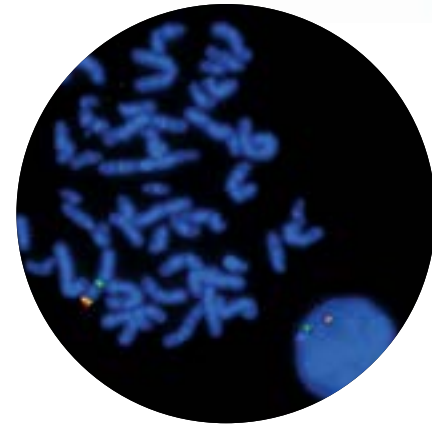
* For sale in the US only. This product has not been licensed in accordance with Canadian law.

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

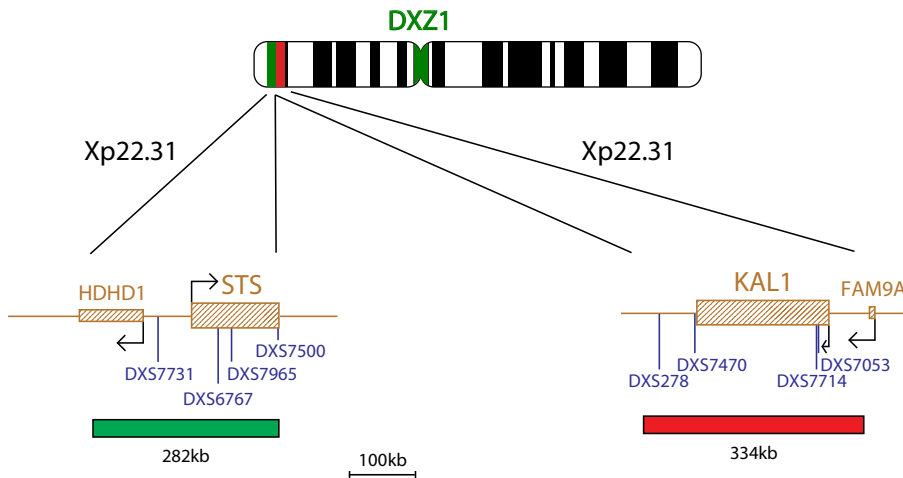


Kallmann (KAL1) Region/ STS Region

The KAL1 probe is 334kb, labeled in red that spans the entire KAL1 gene and the DXS278 and DXS7053 markers. The STS probe is 282kb, labeled in green and covers most of the HDHD1 and STS genes. The probe mix also contains a control probe for the X centromere (DXZ1), labeled in green.



Probe Specification	KAL1, Xp22.31, Red
	STS, Xp22.31, Green
	DXZ1, Xp11.1-q11.1, Green



CMP-U009 v002

* For sale in the US only. This product has not been licensed in accordance with Canadian law.

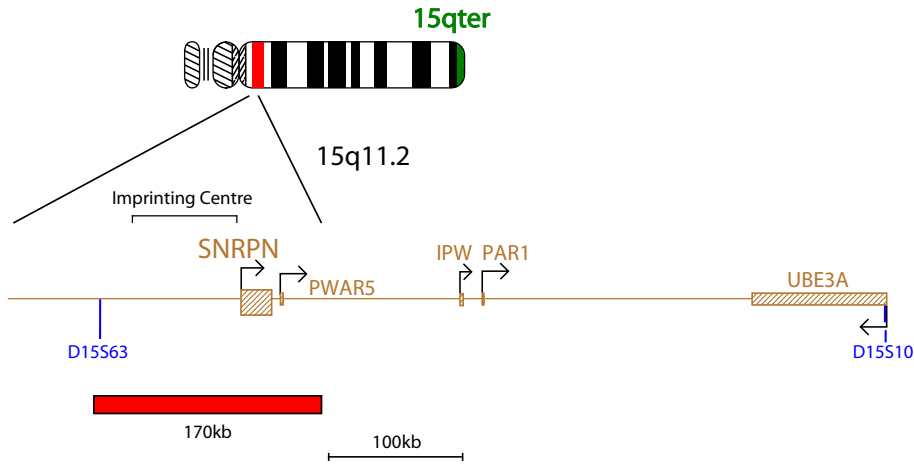
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

Prader-Willi/Angelman (SNRPN) Region

The Prader-Willi/Angelman (SNRPN) probe is 170kb, labeled in red and covers the whole SNRPN gene as well as the entire imprinting center. The 15qter subtelomere specific probe (clone 154P1), labeled in green, allows identification of chromosome 15 and acts as a control probe.



Probe Specification SNRPN, 15q11.2, Red
15qter, 15q26.3, Green



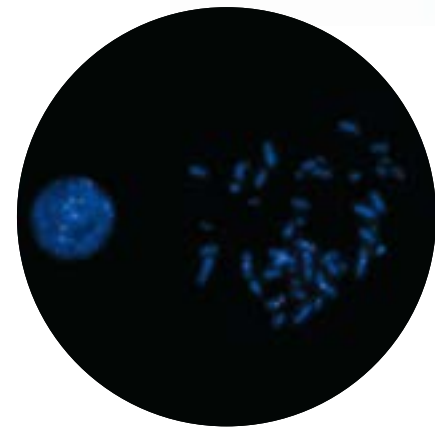
CMP-U013 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

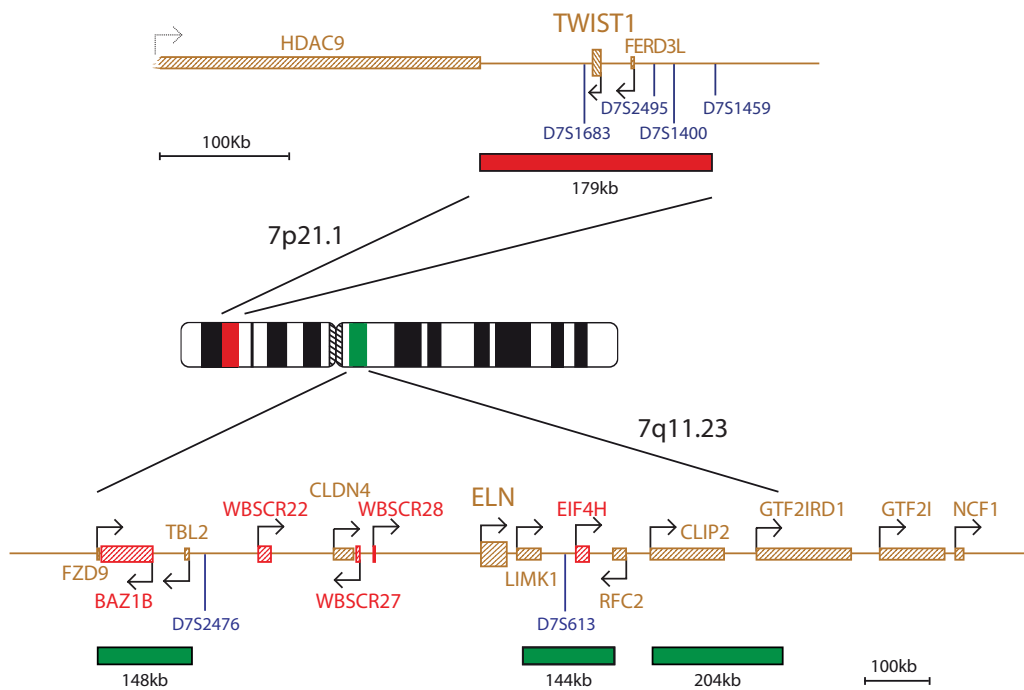


Saethre-Chotzen Region/ Williams-Beuren Region

The TWIST1 probe is 179kb, labeled in red and covers a region including the entire TWIST1 gene and flanking DNA. The Williams-Beuren region probe, labeled in green, consists of three non-overlapping clones (148kb, 144kb and 204kb), which cover much of the deletion region.



Probe Specification TWIST1, 7p21.1, Red
 WBSCR, 7q11.23, Green

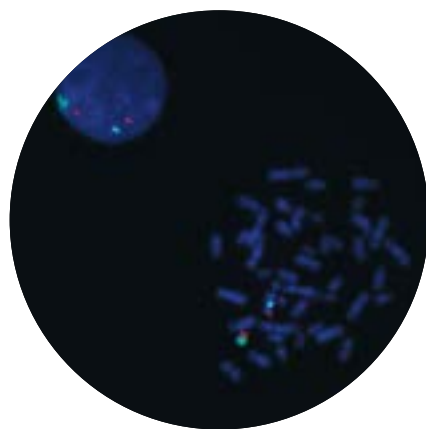


CMP-U015 v002

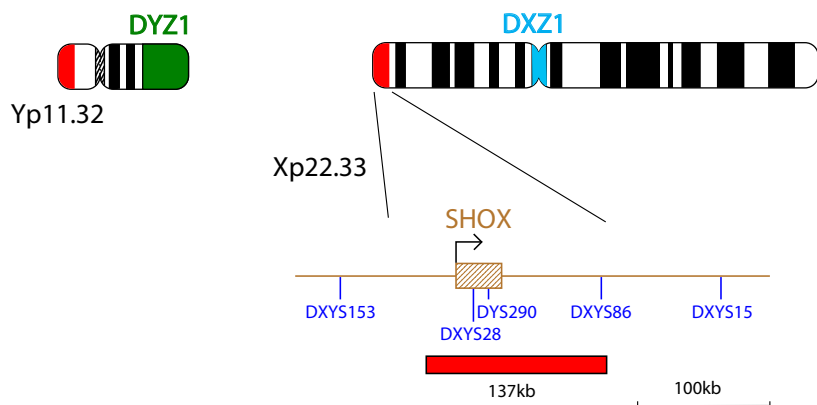
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

SHOX

The SHOX probe is 137kb, labeled in red and covers a region including the entire SHOX gene and flanking DNA on chromosomes Y and X. The probe mix also contains control probes for the X centromere (DXZ1) labeled in blue, and for chromosome Y (DYZ1, the heterochromatic block at Yq12) labeled in green.



Probe Specification SHOX, Xp22.33/Yp11.32, Red
 DYZ1, Yq12, Green
 DXZ1, Xp11.1-q11.1, Blue



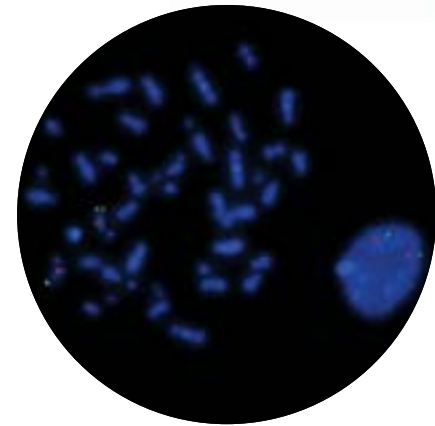
CMP-U016 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

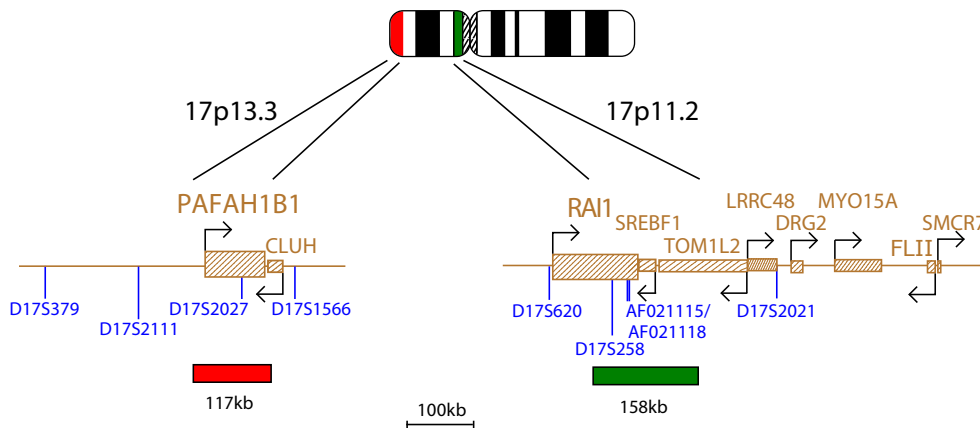


Smith-Magenis (RAI1) Region/Miller-Dieker Region

The Miller-Dieker (LIS1) probe is 117kb, labeled in red and covers the entire LIS1 (PAFAH1B1) gene. The Smith-Magenis (RAI1) probe is 158kb, labeled in green and covers the centromeric end of the RAI1 gene and includes the D17S258 marker. The two unique sequences act as control probes for each other and allow identification of chromosome 17.



Probe Specification LIS1 (PAFAH1B1), 17p13.3, Red
RAI1, 17p11.2, Green

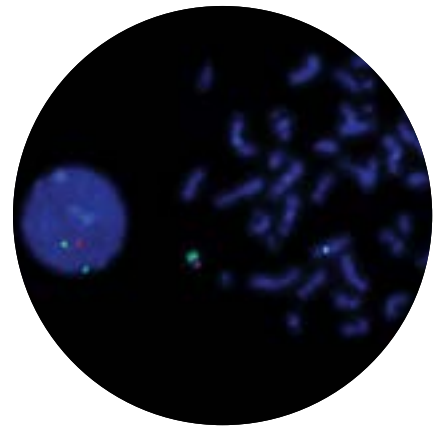


CMP-U017 v002

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

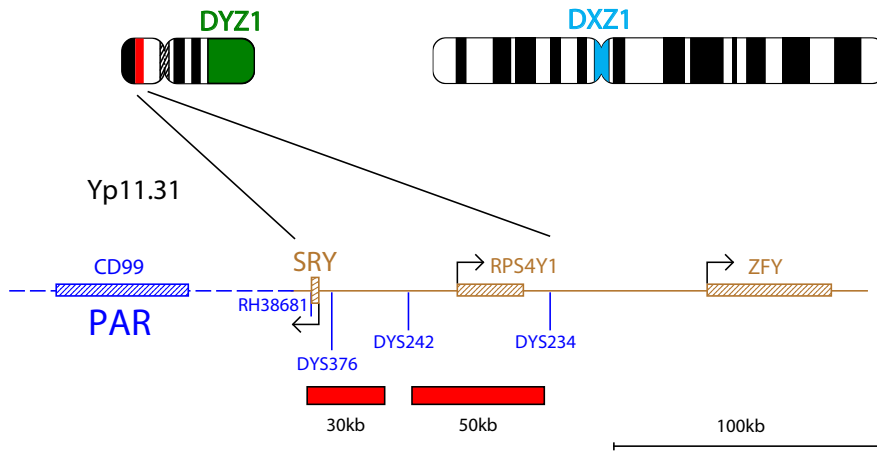
SRY

The SRY probe, labeled in red, consists of two non-overlapping probes, 30kb and 50kb. The probes cover the entire SRY gene and flanking DNA, including the RPS4Y1 gene. The probe mix also contains control probes for the X centromere (DXZ1), labeled in blue, and for chromosome Y (DYZ1, the heterochromatic block at Yq12), labeled in green.



Probe Specification

- SRY, Yp11.31, Red
- DYZ1, Yq12, Green
- DXZ1, Xp11.1-q11.1, Blue



CMP-U019 v003

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

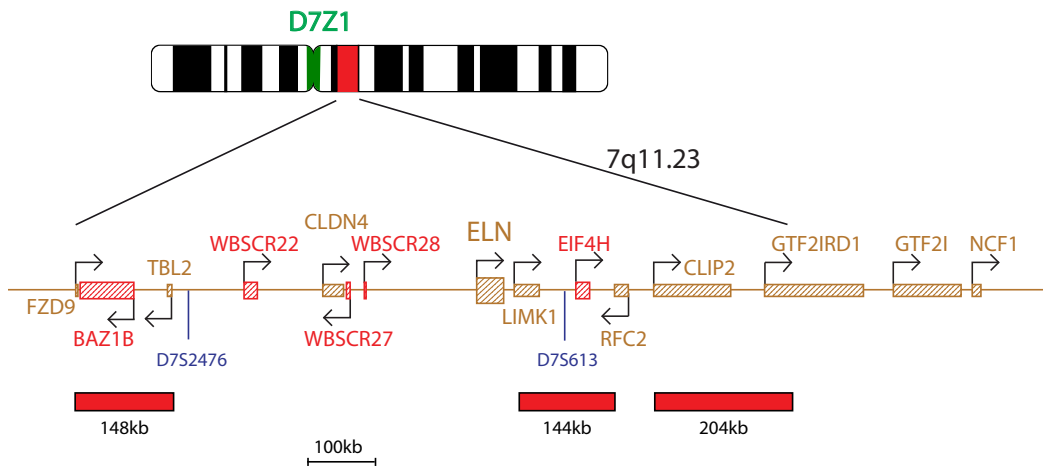


Williams-Beuren Region

The Williams-Beuren probe, labeled in red, consists of three non-overlapping clones (148kb, 144kb and 204kb), which cover much of the deletion region. The probe mix also contains a control probe for the 7 centromere (D7Z1), labeled in green.



Probe Specification Williams-Beuren, 7q11.23, Red
D7Z1, 7p11.1-q11.1, Green



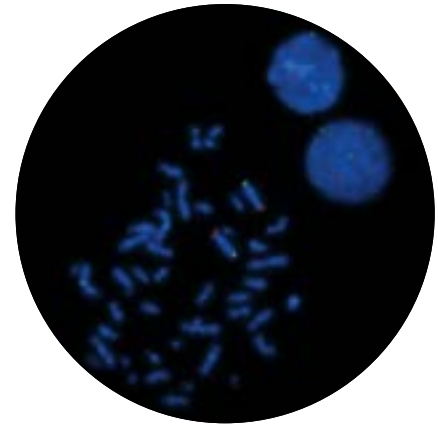
CMP-U020 v002

* For sale in the US only. This product has not been licensed in accordance with Canadian law.

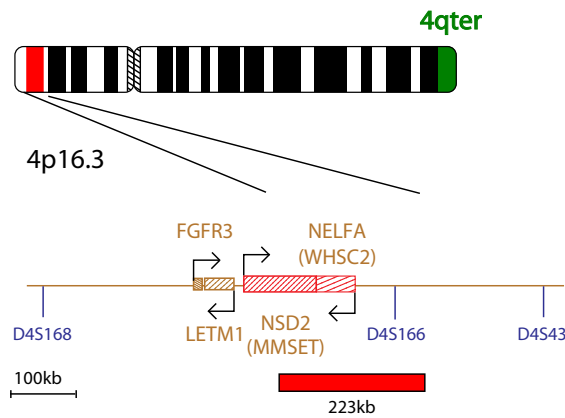
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

Wolf-Hirschhorn Region

The Wolf-Hirschhorn probe is 223kb, labeled in red and covers a region centromeric to the FGFR3 and LETM1 genes, including the D4S166 marker. The 4qter subtetromere specific probe (clone CTC-963K6), labeled in green, allows identification of chromosome 4 and acts as a control probe.



Probe Specification Wolf-Hirschhorn, 4p16.3, Red
4qter, 4q35.2, Green

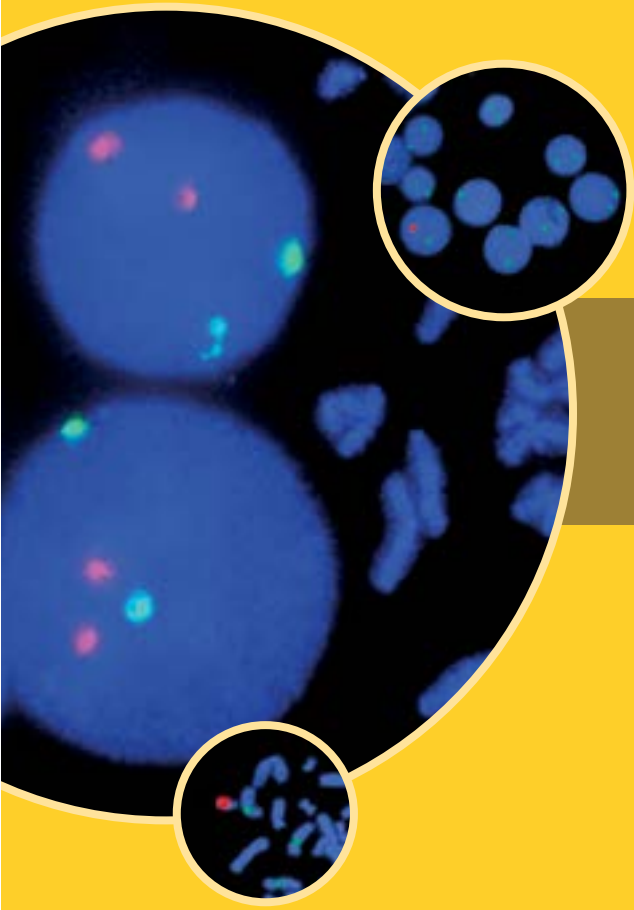


CMP-U021 v004

* For sale in the US only. This product has not been licensed in accordance with Canadian law.

ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.





Satellites



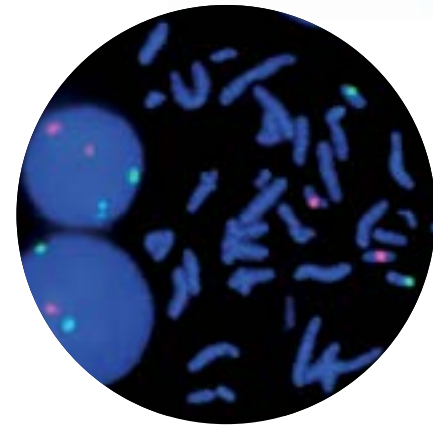
Contents

- 129 Satellite Enumeration Probes
- 130 Blue Labeled Satellite Enumeration Probes
- 130 Dual Labeled Satellite Probe Sets
- 130 Acro-P-Arm Probe





Satellite Enumeration Probes



Cytocell's Satellite Enumeration probes are chromosome specific sequences generated from highly repeated human satellite DNA located in the centromeric, pericentromeric or heterochromatic regions of each chromosome.

Cytocell offers a complete range of satellite probes available in the Aquarius® liquid format. The probes are available independently and directly labeled in either red or green fluorophore (Texas Red® or FITC spectra respectively). The probes are supplied concentrated and packaged in an economical 15µl format.

Green Satellite Enumeration Probes

	Cat. No. LPE 001G-A
	Cat. No. LPE 002G-A
	Cat. No. LPE 003G-A
	Cat. No. LPE 004G-A
	Cat. No. LPE 005G-A (Chromosome 1,5,19)
	Cat. No. LPE 006G-A
	Cat. No. LPE 007G-A
	Cat. No. LPE 008G-A
	Cat. No. LPE 009G-A
	Cat. No. LPE 010G-A
	Cat. No. LPE 011G-A
	Cat. No. LPE 012G-A
	Cat. No. LPE 013G-A (Chromosome 13,21)
	Cat. No. LPE 014G-A (Chromosome 14,22)
	Cat. No. LPE 015G-A
	Cat. No. LPE 016G-A
	Cat. No. LPE 017G-A
	Cat. No. LPE 018G-A
	Cat. No. LPE 020G-A
	Cat. No. LPE 0XG-A
	Cat. No. LPE 0YcG-A
	Cat. No. LPE 0YqG-A

Red Satellite Enumeration Probes

	Cat. No. LPE 001R-A
	Cat. No. LPE 002R-A
	Cat. No. LPE 003R-A
	Cat. No. LPE 004R-A
	Cat. No. LPE 005R-A (Chromosome 1,5,19)
	Cat. No. LPE 006R-A
	Cat. No. LPE 007R-A
	Cat. No. LPE 008R-A
	Cat. No. LPE 009R-A
	Cat. No. LPE 010R-A
	Cat. No. LPE 011R-A
	Cat. No. LPE 012R-A
	Cat. No. LPE 013R-A (Chromosome 13,21)
	Cat. No. LPE 014R-A (Chromosome 14,22)
	Cat. No. LPE 015R-A
	Cat. No. LPE 016R-A
	Cat. No. LPE 017R-A
	Cat. No. LPE 018R-A
	Cat. No. LPE 020R-A
	Cat. No. LPE 0XR-A
	Cat. No. LPE 0YcR-A
	Cat. No. LPE 0YqR-A

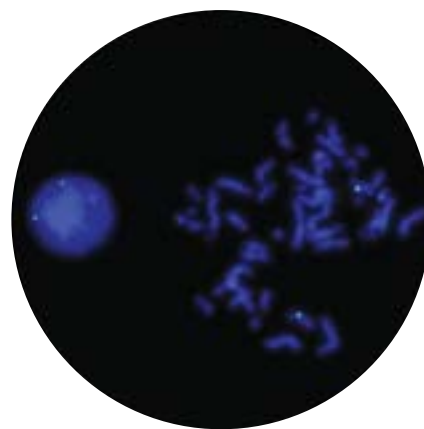
ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



Blue Labeled Satellite Enumeration Probes

Cytocell offers a limited range of satellite probes directly labeled in a blue fluorophore. The probes are supplied concentrated and packaged in an economical 30µl format.

- Chromosome 8
- Chromosome 12
- Chromosome 17
- Other chromosomes are available through the myProbes® custom FISH probes program

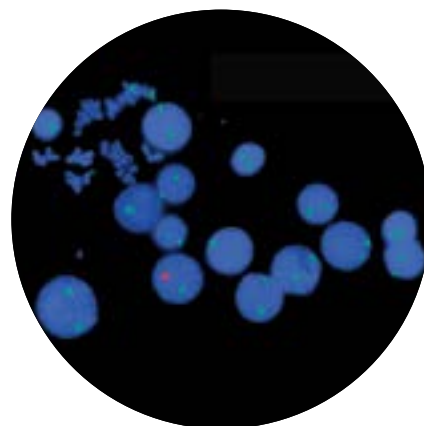


ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

Dual Labeled Satellite Probe Sets

We also offer two dual labeled X&Y probe sets are available in the Aquarius® liquid range in a 100µl format.

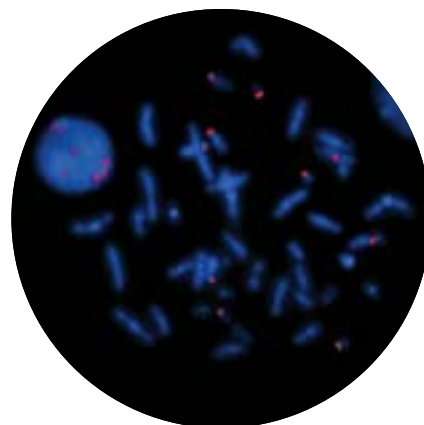
1. XYc: Xp11.1-q11.1 directly labeled with a green fluorophore and Yp11.1-q11.1 with a red fluorophore.
2. XYq: Xp11.1-q11.1 directly labeled with a green fluorophore and Yq12 with a red fluorophore.



ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.

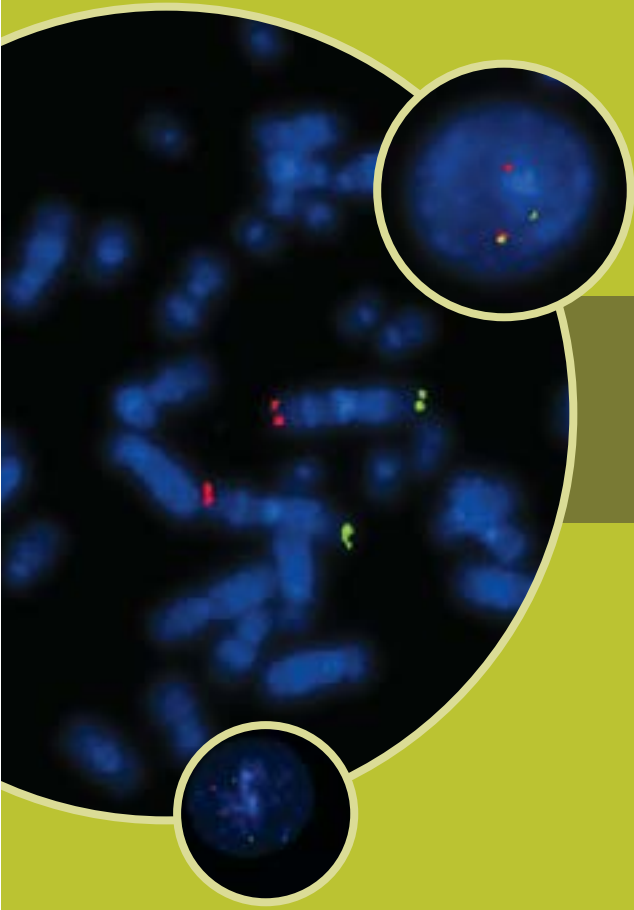
Acro-P-Arm Probe

NOR (Nucleolar Organizer Regions) probe is specific for rRNA genes located in the short arms of the acrocentric chromosomes (13, 14, 15, 21 and 22), labeled in red.



ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.



A large, circular fluorescence microscopy image of a chromosome spread, showing blue-stained chromosomes with several red and green fluorescent spots. Two smaller circular inset images show magnified views of individual chromosomes with these spots.

Subtelomere Specific Probes



Contents

133 Aquarius® Subtelomere Specific Probes

Subtelomere Specific Probes

Cytocell's subtelomere specific probes are located in the most distal region of chromosome specific DNA on each chromosome. Beyond this unique sequence material is the 100 to 300kb region of telomere associated repeat followed by the cap of between 3 to 20kb of tandemly repeated (TTAGGG)_n sequence.

The original second-generation set of probes is derived from PAC clones and was established in conjunction with the Institute of Molecular Medicine, part of Oxford University, in the UK. Continuing product improvements have led to some substitutions with alternative cosmid (35–40kb) or BAC (150kb) clones to give improved signal strength or chromosome specificity.

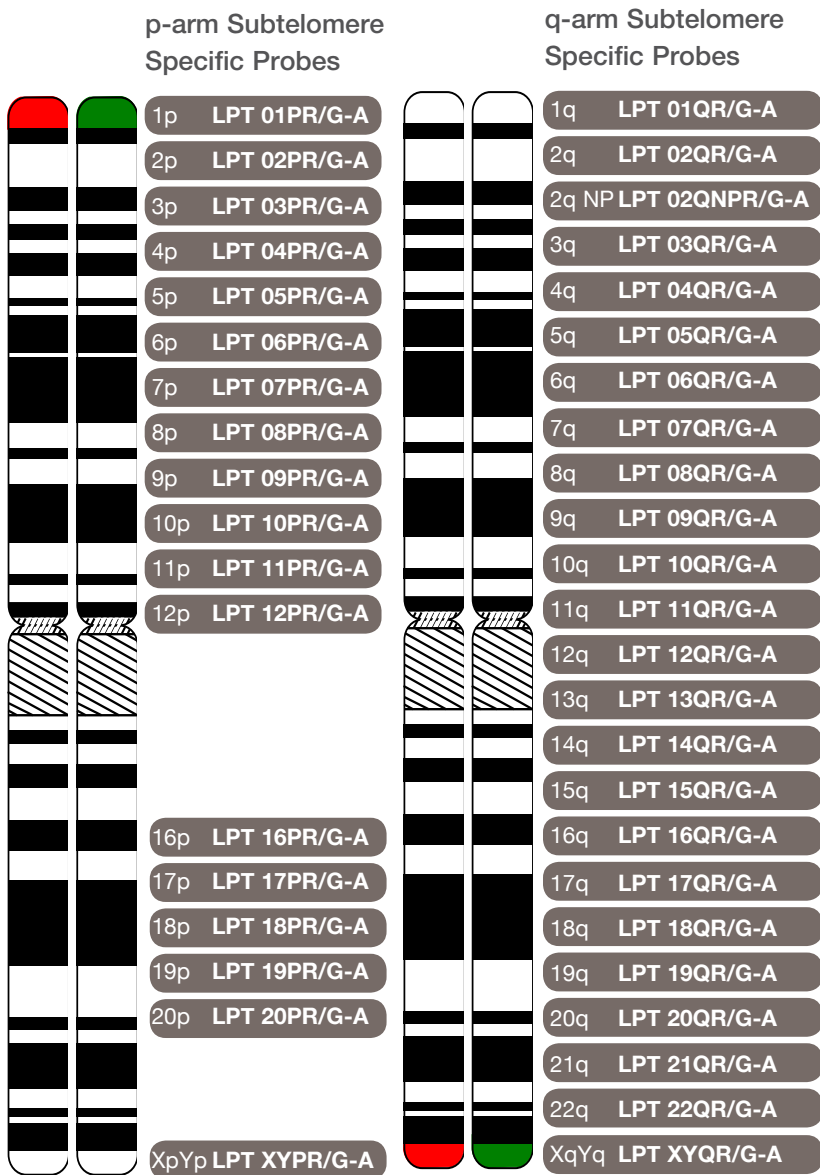
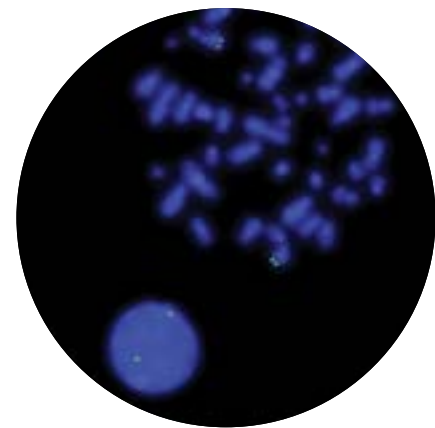
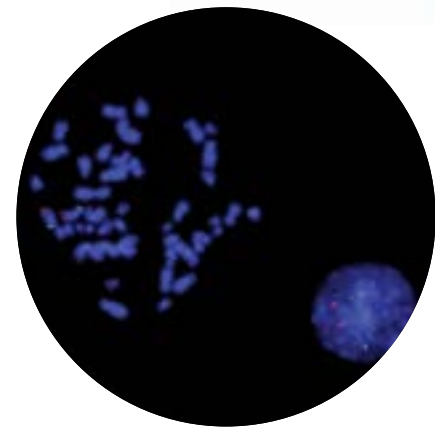




Subtelomere Specific Probes

Cytocell offers a complete set of Subtelomere Specific Probes available in the Aquarius® liquid format. The set identifies 41 of the 46 human subtelomeres with the exclusion of the p-arm telomeres of the acrocentric chromosomes. The probes are available independently and directly labeled in either a red or a green fluorophore (Texas Red® or FITC spectra respectively).

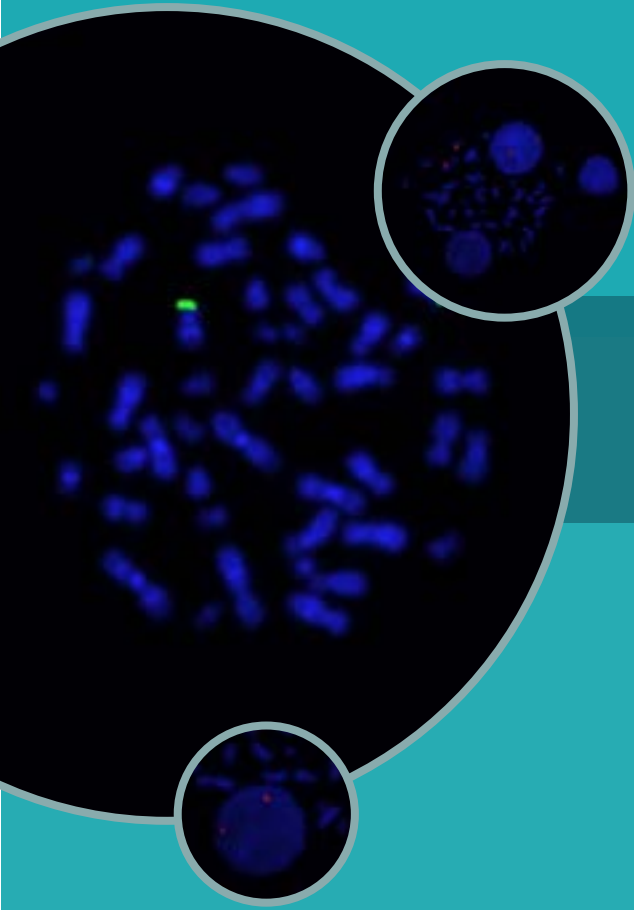
The probes are supplied concentrated and packaged in an economical 15µl format.



ASR: Analyte Specific Reagent. Analytical and performance characteristics are not established.







Custom FISH Probes



myProbes[®] Custom FISH Probes

Custom FISH Probes designed to your specifications

myProbes[®] is a custom design and manufacture service that provides unique fluorescence *in situ* hybridization (FISH) probes using the BAC-2-FISH[™] process. This process utilizes CytoCell's proprietary BAC clone collection containing >220,000 clones to produce fully quality-assured custom FISH probes for virtually any sequence in the entire human genome. Over 2,000 myProbes projects have been completed since 2010.

Based on your specific interests and research, custom FISH probes may range from a simple catalog probe modification to a truly unique product.

CytoCell offers expert consultation from start to finish on your project. Contact us to learn more.

Step 1. Select the gene/target of interest

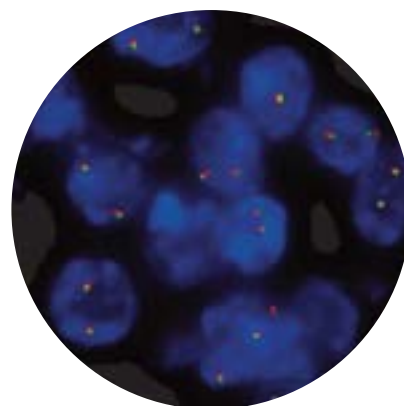
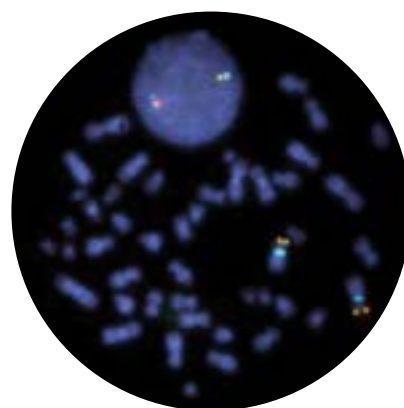
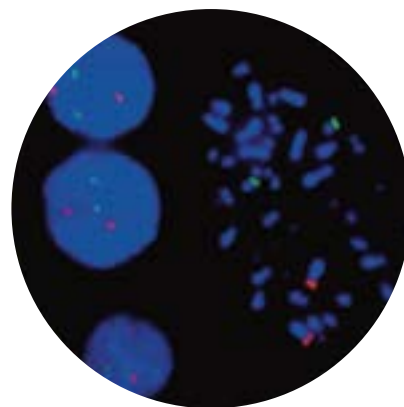
Step 2. Specify the sample type(s)

Step 3. Specify the probe strategy required

Step 4. Choose the color

All custom myProbes are tested on your specific sample type (when available) to ensure reproducibility. Our process and quality assurances are designed to produce high-quality probes and accurate results.

A selection of previously manufactured custom probes are searchable online: www.cytoCell-us.com/custom-search

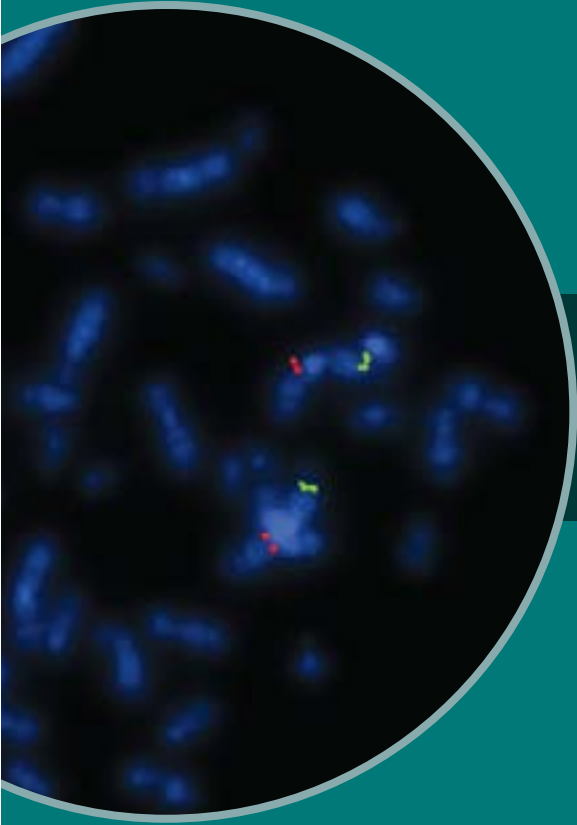


For USA: Analyte Specific Reagent. Analytical and performance characteristics are not established. Laboratories must undertake all appropriate validation of any LDT, as per the CLIA Regulations. Custom probes are specifically developed for individual customers' ANALYTE SPECIFIC REAGENT (ASR) requirements. Therefore, prior to any future ordering of these probes, users should review the design of such probes to confirm they are suitable for their requirements.

For Canada: For Research Use Only. Not for use in diagnostic procedures. Custom probes are specifically developed for individual customers' RESEARCH USE ONLY (RUO) requirements and not with the intention of being used for *in vitro* diagnostic examination. Therefore, prior to any future ordering of these probes, users should review the design of such probes to confirm they are suitable for their requirements.




Cytocell
aquarius



FISH Accessories



FISH Accessories and Ancillary Items

From filters, slides and counterstains to hybridization solutions and chambers, OGT offers a comprehensive range of accessories and ancillary items to support FISH.

Accessories

Cat. No.	Description	Unit Size
PCN009	Porcelain Wash Jars - 12 slide capacity	2
PCN004	Hybridization Chamber	1
PCN007	24 Square Template Slides	100
PCN008	8 Square Template Slides	100
PCN002	Slide Surface Thermometer	1

Ancillary Reagents

Cat. No.	Description	Unit Size
DES500L	0.125µg/ml DAPI	500µl
DES1000L	0.125µg/ml DAPI	1000µl
DFS500L	1.0µg/ml DAPI	500µl
DSS500L	0.0625µg/ml DAPI	500µl
HB500L	Hybridization Solution B	500µl
HB1000L	Hybridization Solution B	1000µl
LPS100	Aquarius® Tissue Pretreatment Kit*	Reagent 1 (1x1L) Reagent 2 (1x10ml)
PCA003	20x SSC	100ml
PCA005	Rubber Solution Glue	15g
PCN003	Mounting Medium	10ml

* LPS100 is provided under agreement between Life Technologies Corporation and Cytocell Ltd and is available for human diagnostics or life science use only





Microscope Filters*

Cat. No.	Description	Unit Size
CF69008	Chroma® Filter: 69008 ET-Aqua/FITC/Texas Red Triple Filter Set	1
CF69011	Chroma® Filter: 69011 ET-Aqua/Green/Orange Triple Filter Set	1
CF49000	Chroma® Filter: 49000 ET-DAPI Single Filter	1
CF49302	Chroma® Filter: 49302 ET-Aqua Single Filter	1
CF49303	Chroma® Filter: 49303 ET-Green Single Filter	1
CF49306	Chroma® Filter: 49306 ET-Red Single Filter	1
CF59010	Chroma® Filter: 59010 ET-Green/Red Dual Filter	1
CF59011	Chroma® Filter: 59011 ET-Green/Orange Dual Filter	1
CF59022	Chroma® Filter: 59022 ET-FITC/Texas Red Dual Filter	1

Blocks






Cat. No.	Description	Unit Size
CBZ0001	Chroma® Block: Zeiss Microscope	1
CBBX051	Chroma® Block: Olympus BX51	1
CBBX061	Chroma® Block: Olympus BX61	1
CBNK050	Chroma® Block: Nikon 50i	1
CBDM550	Chroma® Block: Leica DM5500	1

* Microscope filters are available on request. These filters can be ordered with or without a filter cube.

Chroma is a registered trademark of Chroma Technology Corporation. Zeiss is a trademark of Carl Zeiss AG. Olympus is a trademark of Olympus Corporation. NIKON is a trademark of Nikon Corporation. LEICA is a trademark of LEICA MICROSYSYSTEMS IR GMBH.

Ordering Guide

Recommended products are available from OGT.

	Sample and slide preparation	<ul style="list-style-type: none"> Tissue Pretreatment Kit * (Cat. No. LPS 100) Template slides (Cat. No. PCN 007/008)
	Pre-denaturation	<ul style="list-style-type: none"> Slide surface thermometer (Cat. No. PCN 002) Rubber solution glue (Cat. No. PCA 005)
	Denaturation	<ul style="list-style-type: none"> Slide surface thermometer (Cat. No. PCN 002)
	Hybridization	<ul style="list-style-type: none"> Hybridization solution (Cat. No. HB 500L/1000L) Hybridization chamber (Cat. No. PCN 004)
	Post-hybridization washes	<ul style="list-style-type: none"> Porcelain Wash Jars (Cat. No. PCN 009) SCC buffer (Cat. No. PCA 003) DAPI (Cat. No. DES 500L/100L, DFS 500L or DSS 500L) Mounting medium (Cat. No. PCN 003)
	Analyze	<ul style="list-style-type: none"> Filter block (Cat. No. CBZ 001, CBBX 051, CBBX 061, CBNK 050 or CBDM 550) Filter (Cat. No. CF 69008/69011/49000, 49302, 49303/49306/59010/59011/59022)

*This product is provided under agreement between Life Technologies Corporation and Cytocell Ltd and is available for human diagnostics or life science use only. See page 87 for product information.



SureSeq™ NGS Products for Hematology and Solid Tumor Cancer Research



Contents

RUO

143 SureSeq myPanel™ NGS Custom Cancer Panels

- 145 SureSeq myPanel NGS Custom AML Panel
- 147 SureSeq myPanel NGS Custom Breast Cancer Panel
- 149 SureSeq myPanel NGS Custom Colorectal Cancer Panel
- 151 SureSeq myPanel NGS Custom Melanoma Cancer Panel
- 153 SureSeq myPanel NGS Custom Prostate Cancer Panel

SureSeq

- 155 SureSeq CLL + CNV Panel
- 157 SureSeq Core MPN Panel
- 159 SureSeq Ovarian Cancer Panel
- 161 SureSeq Myeloid Panel
- 163 Interpret Software
- 165 SureSeq NGS Library Preparation Kit
- 167 SureSeq FFPE DNA Repair Mix

Overview

The application of next generation sequencing (NGS) technologies to cancer research in recent years has provided novel insights into disease initiation, progression and response to therapy. This powerful technique allows for accurate analysis of nucleotide-level aberrations such as single nucleotide variants (SNVs), insertions and deletions (indels) and structural aberrations including copy-number variations (CNVs) and translocations.

SureSeq™ NGS products include targeted panels and library preparation products for the accurate detection of a wide range of somatic variants and structural aberrations, using a streamlined NGS workflow.

Utilizing hybridization-based enrichment, SureSeq NGS cancer panels deliver unparalleled coverage uniformity, excellent run-to-run consistency and ensure highly reproducible data. These factors are particularly important when studying heterogeneous cancer samples, where the ability to detect mutations with low minor allele frequency (MAF) at high accuracy is required. More so, facilitated by OGT's excellent bait design and software, structural aberrations including CNVs, loss-of-heterozygosity (LOH) and translocations can easily be identified.

SureSeq NGS products include pre-designed panels for specific disease groups, as well as an expanded library of pre-optimized custom cancer panel content that allows researchers to create NGS cancer panels meeting their exact requirements.

For more information, visit www.ogt.com/ngs_products

SureSeq myPanel™ NGS Custom Cancer Panels

Simply mix and match the genes, exonic or intronic content you need to create an NGS cancer panel that meets your exact requirements.

SureSeq myPanel offers:

- Hybridization-based panel delivering unparalleled coverage uniformity — Detect low frequency variants consistently with confidence and minimize the requirement for supplementary fill-in with Sanger sequencing
- Pre-optimized panels that meet your technical requirements and work with your samples (including FFPE tissue) — No more laborious in-house optimization, decreasing assay development time
- Bespoke panel content — Sequence only what's relevant for your cancer research, increase throughput and save on sequencing reagents
- Panel content designed with experts and from current literature to target all relevant regions including intronic and splice sites — Get the most comprehensive insight into disease-driving mutations

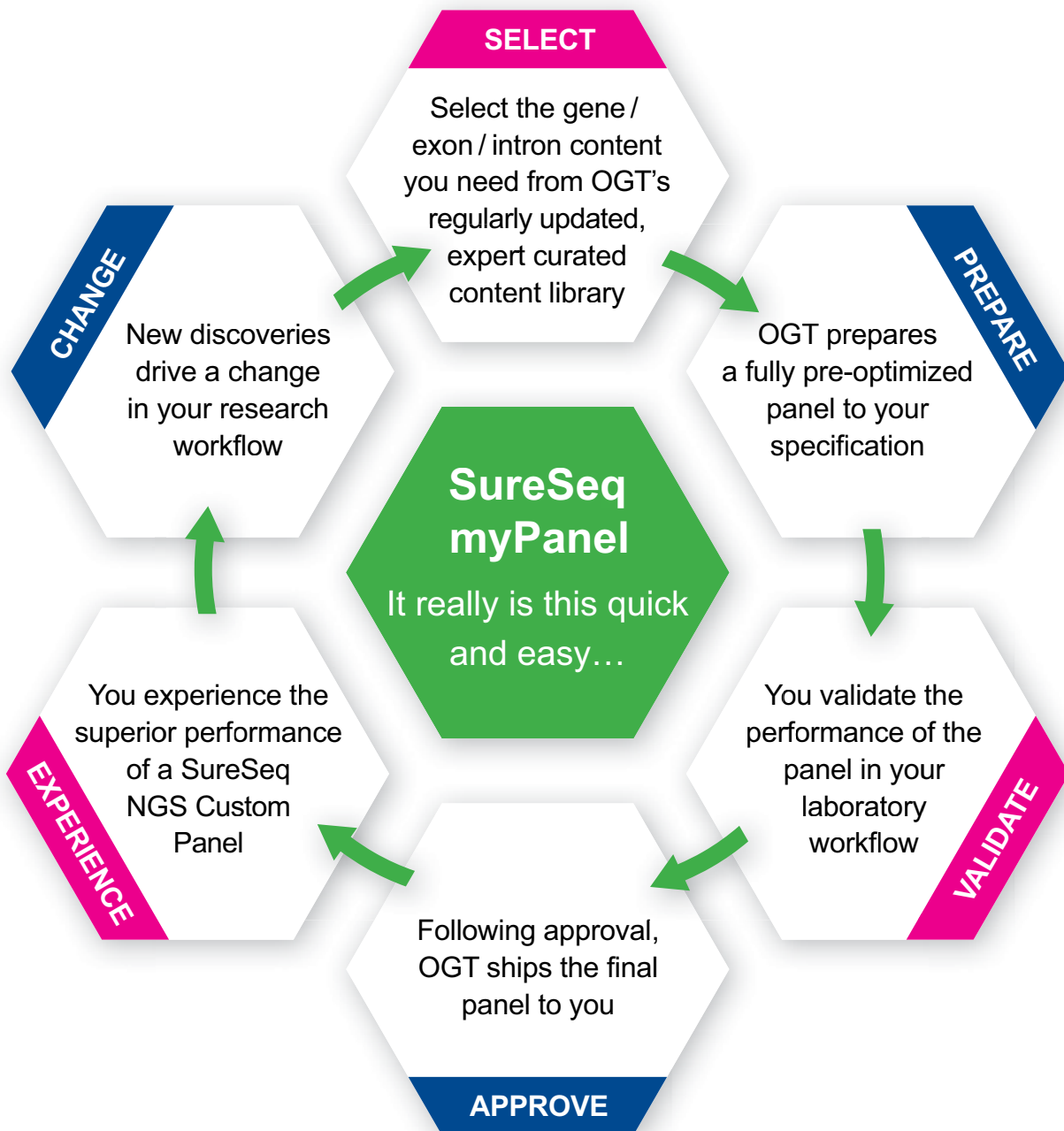
SureSeq myPanel pre-optimized NGS custom panel content is available for research into a wide range of conditions, including:

Hematology Panels	Solid Tumor Panels
Acute Myeloid Leukemia	Bladder Cancer
Chronic Lymphocytic Leukemia	Breast Cancer
Chronic Myeloid Leukemia	Colorectal Cancer
Multiple Myeloma	Glioma
Myelodysplastic Syndromes (MDS)	Lung Cancer
Myeloid Disorders	Melanoma
Myeloproliferative neoplasms (MPN)/ Myelodysplastic syndromes (MDS) Overlap	Ovarian Cancer
	Prostate Cancer
	Sarcoma

Put our expertise to work for you

Go to www.ogt.com/ngs_products to find out more about SureSeq NGS Custom Cancer Panels, library preparation kits, reagents and Interpret, our complimentary, easy-to-use next generation sequencing analysis solution.

Getting started with your next SureSeq myPanel NGS Custom Cancer panel could not be simpler



SureSeq myPanel NGS Custom AML Panel

Acute myeloid leukemia (AML) is the most common type of acute leukemia in adults. Our understanding of AML has been transformed in recent years to a disease classified largely based on genetic, genomic and molecular characteristics. Key genes implicated in AML progression include *CEBPA*, *NPM1*, *FLT3* and *KMT2A (MLL)* with mutations in multiple additional genes identified in recent research¹.

Choose your perfect AML NGS panel from our range of fully tested and optimized panel content. Simply mix and match the genes or individual exons you require for your research and get the most out of your sequencing runs.

SureSeq myPanel Custom AML Panels offer:

- Unparalleled coverage uniformity across all content including *CEBPA* — confidently detect AML variants and remove the requirement for supplementary fill-in approaches
- Bespoke panels with pre-optimized content — create your ideal AML panel and sequence only what’s relevant for your AML research
- Robust detection of *FLT3*-ITDs and *KMT2A*-PTDs — streamline your laboratory workflow with a single NGS assay for comprehensive aberration detection in AML
- Complimentary Interpret data analysis software — easy-to-use analysis solution for accurate detection of all variants

Select from any of the following myPanel AML gene or exonic content:

<i>ASXL1</i>	<i>BCOR</i>	<i>BCORL1</i>	<i>CBLB</i>	<i>CBLC</i>	<i>CEBPA</i>
<i>CUX1</i>	<i>DDX41</i>	<i>DNMT3A</i>	<i>ETV6</i>	<i>FLT3</i>	<i>GATA1</i>
<i>IDH1</i>	<i>IDH2</i>	<i>IKZF1</i>	<i>IRF1</i>	<i>JAK3</i>	<i>KIT</i>
<i>KMT2A</i>	<i>KRAS</i>	<i>NPM1</i>	<i>NRAS</i>	<i>PHF6</i>	<i>RUNX1</i>
<i>SMC1A</i>	<i>TET2</i>	<i>TP53</i>	<i>U2AF1</i>	<i>WT1</i>	

REFERENCES

1. Döhner *et al.*, Blood 2017; 129(4):424–447
2. Steudel *et al.*, Genes Chromosomes Cancer 2003; 37(3):237-51

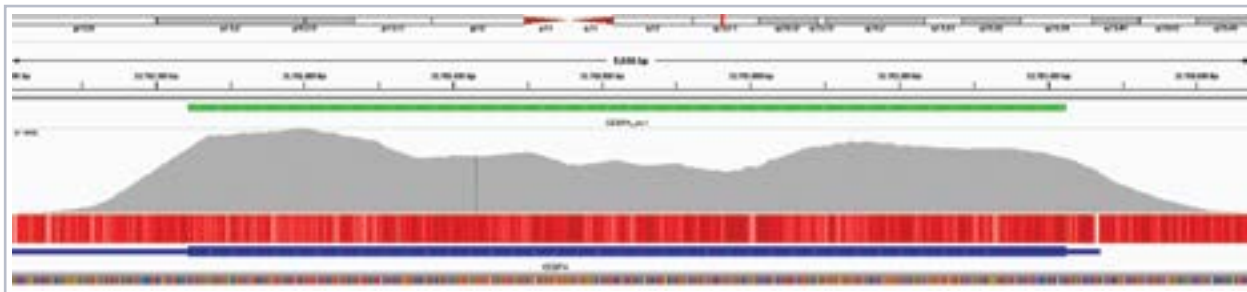


Figure 1: Illustration of the excellent coverage uniformity of the *CEBPA* gene. Depth of coverage per base (grey). Targeted region (green). Gene coding region as defined by RefSeq (blue). GC percentage (red).



Figure 2: Facilitated by OGT's expert bait design, *FLT3*-ITDs of various sizes and even regions containing multiple ITDs can be confidently detected. ITD sizes are **A** 174 bp, **B** 225 bp, **C** 195 bp with additional 6 bp, **D** 120 bp and **E** 168 bp with additional 69 bp.



Figure 3: PTB detected spanning exons 2-8 of *KMT2A* by OGT's Interpret NGS analysis software.

SureSeq myPanel NGS Custom Breast Cancer Panel

Breast cancer is the most common cancer in women after skin cancer. Approximately one out of eight women will be diagnosed in their lifetime with some form of breast cancer. Next generation sequencing (NGS) has enabled the simultaneous study of mutations in high-penetrance breast cancer predisposition genes. These include *BRCA1*, *BRCA2* and other high-risk breast cancer susceptibility genes such as *TP53* (Li-Fraumeni syndrome), *PTEN* (Cowden's syndrome) and *PIK3CA*, as well as more moderate-risk genes such as *PALB2*, *BRIP1*, *RAD51C* and *RAD51D*.

Superior Coverage Uniformity

Mutations in *BRCA1* and *BRCA2* genes lead to an increased susceptibility to breast, ovarian, and other cancers. Figure 1a, illustrates the superior uniformity of coverage of key exons of *BRCA1*, and Figure 1b, *BRCA2* from an FFPE sample with SureSeq compared to an amplicon-based panel.

Key Genomic Regions Covered

The *PI3K* pathway is the most frequently enhanced oncogenic pathway in breast cancer. Among mechanisms of *PI3K* enhancement, *PIK3CA* mutations are most frequently (~30%) observed, with the majority of *PIK3CA* somatic mutations located in two "hot spots": E542K or E545K in exon 9, and H1047R or H1047L in exon 20¹, figures 2a and 2b.

GC-rich regions: handled with ease

Sequencing of another frequently mutated breast cancer gene, *TP53*, where point mutations are predominantly located in exons 5-8², is often hampered by the GC-rich content, which can lead to technical challenges in assay design and analysis. OGT's innovative bait design overcomes this issue, offering a high level of uniform coverage for these difficult genes to sequence in FFPE samples (Figure 3).

Choose your ideal breast cancer NGS panel from our range of fully tested and optimized NGS panel content. Simply mix and match the genes or individual exons you require and get the most out of your sequencing runs. Use in conjunction with the SureSeq FFPE DNA Repair Mix* for improved NGS library yields, %OTR and mean target coverage from challenging FFPE derived samples.

Select from any of the following myPanel breast cancer whole gene or exonic content below:

<i>APC</i>	<i>BRCA2</i>	<i>CHEK2</i>	<i>GATA3</i>	<i>PIK3CA</i>
<i>RB1</i>	<i>ATM</i>	<i>BRIP1</i>	<i>EGFR</i>	<i>MSH6</i>
<i>PTEN</i>	<i>SF3B1</i>	<i>BARD1</i>	<i>CDH1</i>	<i>ERBB2</i>
<i>NBN</i>	<i>RAD51C</i>	<i>STK11</i>	<i>BRCA1</i>	<i>CDK12</i>
<i>ESR1</i>	<i>PALB2</i>	<i>RAD51D</i>	<i>TP53</i>	

REFERENCES

1. Mukohara, Breast Cancer (Dove Med Press). 2015; 7: 111-123.
2. Langerød et al, Clin Cancer Res; 2013; 3569-80

*The SureSeq™ FFPE DNA Repair Mix can only be purchased in conjunction with SureSeq NGS panels, not as a standalone product.

For research use only. Not for use in diagnostic procedures.

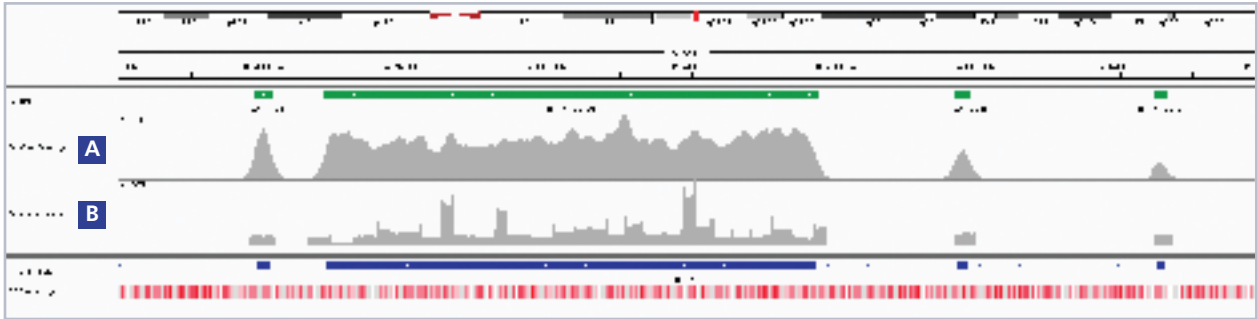


Figure 1: *BRCA1* exons 8, 9, 10 and 11 coverage, Figure 1b: *BRCA2* exons 11, 12 and 13. **A** SureSeq, **B** Amplicon panel. Depth of coverage per base (grey). Targeted region (green). Gene coding region as defined by RefSeq (blue). GC percentage (red).

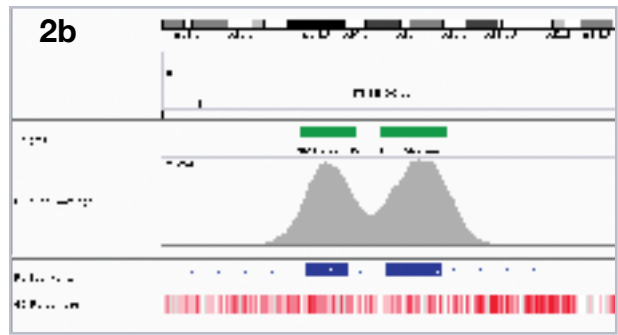
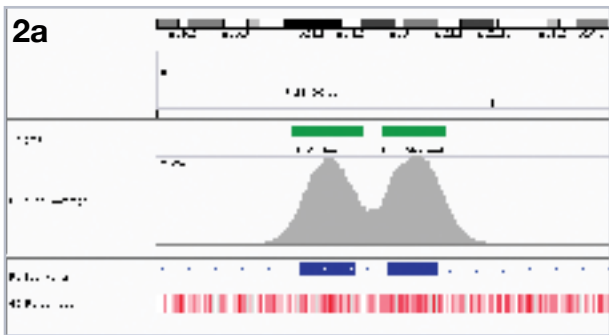


Figure 2a and 2b: Illustration of the excellent uniformity of coverage of *PIK3CA* exons 9 (2a) and 20 (2b). Depth of coverage per base (grey). Targeted region (green). Gene coding region as defined by RefSeq (blue). GC percentage (red).

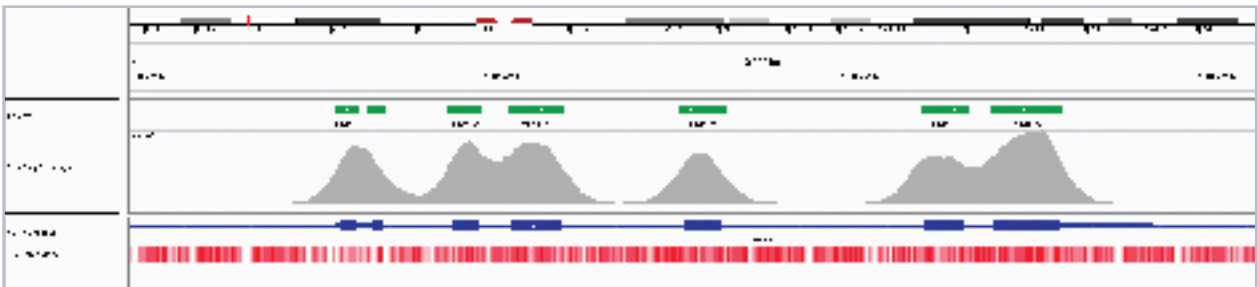


Figure 3: *TP53* exons 3 – 9, exceptional uniformity of coverage in spite of the high GC content of the region. Depth of coverage per base (grey). Targeted region (green). Gene coding region as defined by RefSeq (blue). GC percentage (red).

For information on the SureSeq FFPE DNA Repair Mix, see page 167.*

SureSeq myPanel NGS Custom Colorectal Cancer Panel

Colorectal cancer (CRC) is the third most common cancer in men (746,000 cases, 10.0% of the total) and the second in women (614,000 cases, 9.2% of the total) worldwide¹. Next generation sequencing (NGS) has enabled the simultaneous study of mutations in high-penetrance colorectal cancer genes. These include *KRAS*, *APC* and *TP53* as well as more moderate-risk genes such as *ERBB2*, *PTEN* and *BRAF*.²

Superior Coverage Uniformity

KRAS mutations are found in approximately 35-45% of colorectal cancers with around 80% occurring in codon 12 and 15% in codon 13 of exon 2; other commonly reported mutations are found in exons 3 and 4³. The tumor suppressor gene *APC* plays an important role in CRC development. Absence of the *APC* protein leads to accumulation of beta-catenin in the cytoplasm, which may contribute to tumor progression. 60% of all somatic mutations in *APC* occur within the mutation cluster region between codons 1286 and 1513 on exon 15⁴. Figures 1a, b, c and 2 illustrate the superior uniformity of coverage of these key genomic regions.

Approximately 8-15% of colorectal cancers involve mutations in the *BRAF* gene, with up to 90% of these a result of a mutation at V600E, located on exon 15⁵. In *TP53*, another frequently mutated cancer gene, point mutations are predominantly located in exons 5-8², however sequencing is often hampered by the GC-rich content, which can lead to technical challenges in assay design and analysis. OGT's innovative bait design overcomes this issue, offering a high level of uniform coverage for these difficult genes to sequence in FFPE samples, Figure 4.

Choose your ideal colorectal cancer NGS panel from our range of fully tested and optimized NGS panel content. Simply mix and match the genes or individual exons you require and get the most out of your sequencing runs. Use in conjunction with the SureSeq FFPE DNA Repair Mix* for improved NGS library yields, %OTR and mean target coverage from challenging FFPE derived samples.

Select from any of the following myPanel colorectal whole gene or exonic content below:

<i>APC</i>	<i>CDH1</i>	<i>ERBB2</i>	<i>KRAS</i>	<i>MSH6</i>	<i>PIK3CA</i>	<i>STK11</i>
<i>BRAF</i>	<i>CHEK2</i>	<i>HRAS</i>	<i>MET</i>	<i>NRAS</i>	<i>PTEN</i>	<i>TP53</i>

REFERENCES

1. http://globocan.iarc.fr/Pages/fact_sheets_cancer.aspx
2. Han et al, PLoS One. 2013; 8(5): e64271
3. Tan et al, World J Gastroenterol. 2012 Oct 7; 18(37): 5171-51804
4. More, et al, Hum Mol Genet (1992) 1 (4): 229-233
5. <https://www.mycancergenome.org/content/disease/colorectal-cancer/braf/54/>

*The SureSeq™ FFPE DNA Repair Mix can only be purchased in conjunction with SureSeq NGS panels, not as a standalone product.

For research use only. Not for use in diagnostic procedures.

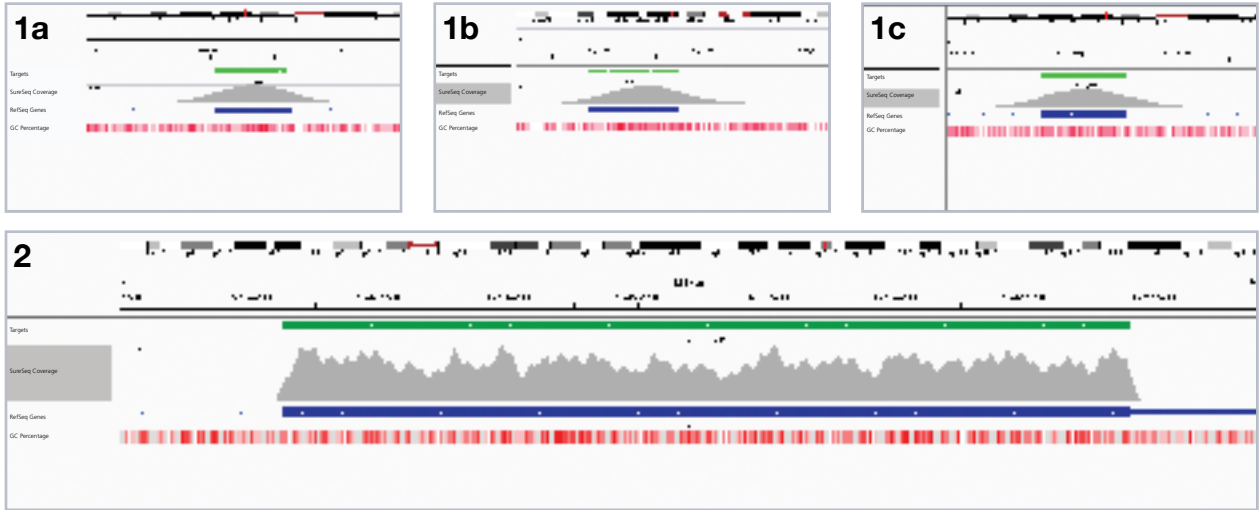
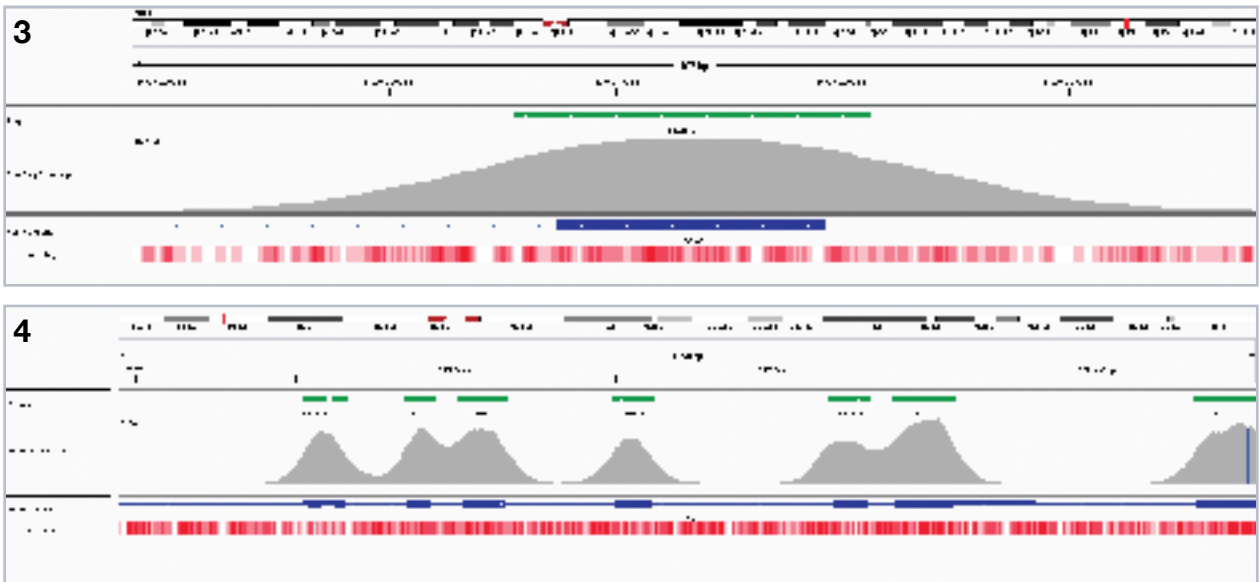


Figure 1a, b, c: *KRAS* exons 2, 3 and 4 coverage. **Figure 2:** *APC* exon 15 coverage. Depth of coverage per base (grey). Targeted region (green). Gene coding region as defined by RefSeq (blue). GC percentage (red).



Figures 3 and 4: Illustration of the excellent uniformity of coverage of (Figure 3) *BRAF* exon 15 and *TP53* exons 3 - 9 (Figure 4). Depth of coverage per base (grey). Targeted region (green). Gene coding region as defined by RefSeq (blue). GC percentage (red).

For information on the SureSeq FFPE DNA Repair Mix, see page 167.*

SureSeq myPanel NGS Custom Melanoma Cancer Panel

Cutaneous melanoma (CM) is the most dangerous form of skin tumor and causes 90% of skin cancer mortality¹. With recurrent somatic mutations in *BRAF*, *NRAS*, *KIT* and *NF1* among the most common genetic aberrations underlying pathogenesis of melanoma, next generation sequencing (NGS) has been an invaluable tool in helping to characterize the overall genomic landscape of melanomas.

Superior Coverage Uniformity

The most frequently activated pathway in melanoma is the mitogen-activated protein kinase (MAPK) pathway, often activated through mutations in the V600 codon of *BRAF* (in 35–50% of melanomas) and the Q61 codon of *NRAS* (10–25%)², with mutations being mutually exclusive.

Mutations of *KIT* are found in particular subsets of melanoma, where the mutations activate signal-transduction pathways (MAPK and PI3K) that ultimately lead to cell proliferation. Approximately 70% of *KIT* mutations identified in melanoma are found in exon 11, most commonly L576P (Figure 1a and 1b).

Neurofibromatosis type 1 (*NF1*) is a relatively common tumor predisposition syndrome related to germline aberrations of *NF1*, a tumor suppressor gene. Recent studies have additionally shown *NF1* to play a critical role in somatic events in a wide range of tumors, including melanoma. The tumor suppressor function of neurofibromin is largely attributed to a small central region which comprises 360 amino acids encoded by exons 20-27a³. OGT's expert bait design offers excellent uniformity for all of these key genes associated with melanoma (Figure 1).

Choose your ideal melanoma NGS panel from our range of fully tested and optimized NGS panel content. Simply mix and match the genes or individual exons you require and get the most out of your sequencing runs. Use in conjunction with the SureSeq FFPE DNA Repair Mix* for improved NGS library yields, %OTR and mean target coverage from challenging FFPE derived samples.

Select from any of the following myPanel melanoma whole gene or exonic content below:

<i>BRAF</i>	<i>KIT</i>	<i>NF1</i> **	<i>NRAS</i>
-------------	------------	---------------	-------------

REFERENCES

1. Garbe *et al*, European Journal of Cancer 63 (2016) 201-217
2. Tsao *et al*, Genes & Dev. 2012. 26: 1131-1155
3. Yap *et al*, Oncotarget, 2014, Vol. 5, No. 15

*The SureSeq™ FFPE DNA Repair Mix can only be purchased in conjunction with SureSeq NGS panels, not as a standalone product.

**Due to the presence of pseudogenes in *NF1*, it is recommended that an orthogonal technique is used to verify any mutations detected.

For research use only. Not for use in diagnostic procedures.

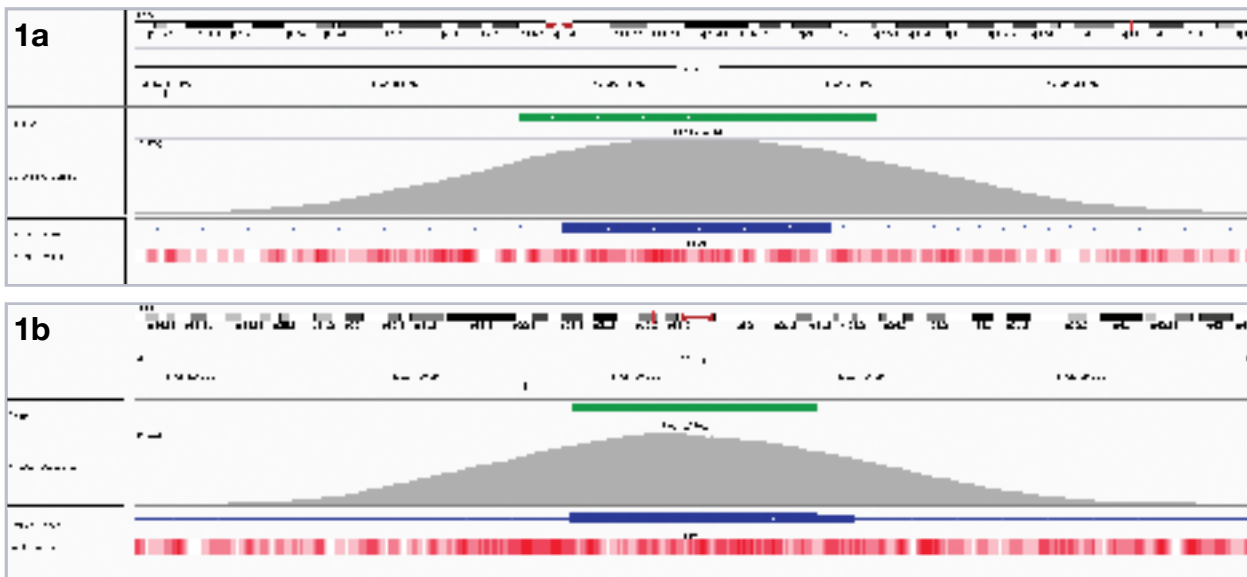


Figure 1: Illustration of the exceptional uniformity of coverage of *BRAF* exon 15 (1a), *NRAS* exon 2 (1b) with a SureSeq melanoma panel. Depth of coverage per base (grey). Targeted region (green). Gene coding region as defined by RefSeq (blue). GC percentage (red).

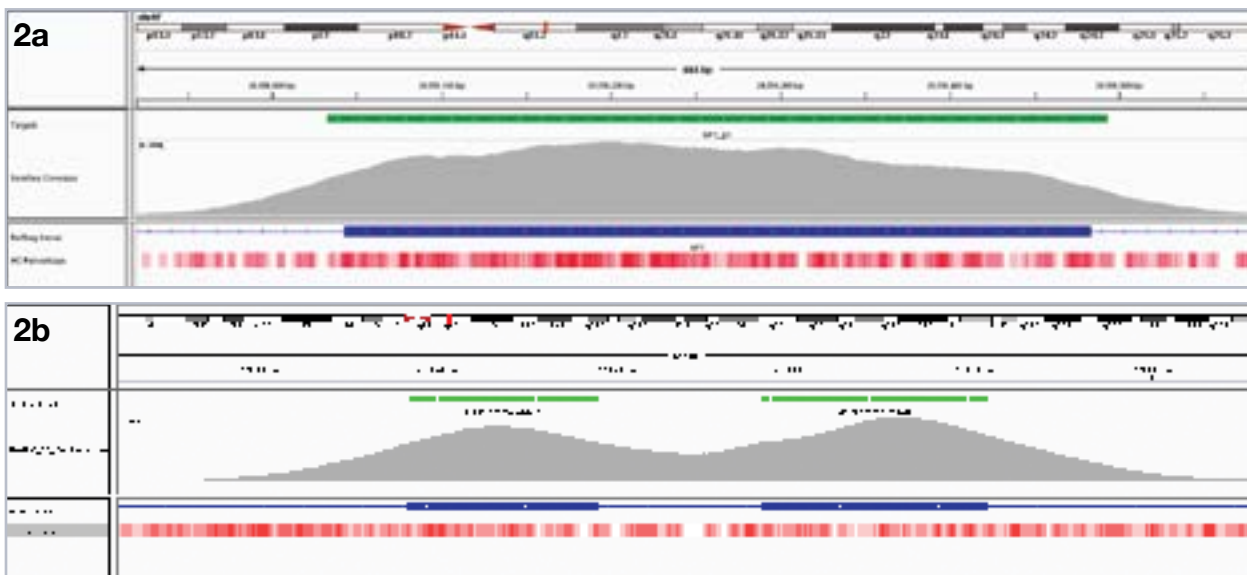


Figure 2: Even coverage of *NF1* exon 21 (2a) and *KIT* exons 10 and 11 (2b) with a SureSeq melanoma panel. Depth of coverage per base (grey). Targeted region (green). Gene coding region as defined by RefSeq (blue). GC percentage (red).

For information on the SureSeq FFPE DNA Repair Mix, see page 167.*

SureSeq myPanel NGS Custom Prostate Cancer Panel

Prostate cancer is now the second leading cause of cancer in men, with recent genome-wide studies helping to clarify the genetic basis of this common but complex disease¹. Many of these studies have reinforced the importance of homologous end repair genes including: *ATM*, *BRCA1*, *BRCA2* and *PALB2*, in the mechanism of prostate cancer development. Mutations in these genes result in cells having to repair lesions through other non-conservative mutagenic mechanisms.

Superior Coverage Uniformity

A number of genetic factors have been found that increase prostate cancer risk, including heritable mutations in the genes *BRCA1* and *BRCA2*. *BRCA1* is a key player in cellular control systems, having been linked to DNA damage response and repair, transcriptional regulation and chromatin modelling², while *BRCA2* function is linked to DNA recombination and repair processes, being of particular importance in the regulation of *RAD51* activity. Figure 1a, illustrates the superior uniformity of coverage of key exons of *BRCA1*, and Figure 1b, *BRCA2* from an FFPE sample.

PALB2 is a *BRCA2* binding protein and the *BRCA2-PALB2* interaction is essential for *BRCA2*-mediated DNA repair. Recently it has been shown that correct *PALB2* function is necessary for the homologous recombination repair via interaction with *BRCA1*, revealing that *PALB2* is actually a linker between *BRCA1* and *BRCA2*³. Figures 2 illustrate the excellent uniformity of coverage of key exons of *PALB2*.

Choose your ideal prostate cancer NGS panel from our range of fully optimized NGS panel content. Simply mix and match the genes or individual exons you require and get the most out of your sequencing runs. Use in conjunction with the SureSeq FFPE DNA Repair Mix* for improved NGS library yields, %OTR (on target rate) and mean target coverage from challenging FFPE derived samples.

Select from any of the following myPanel Prostate Cancer whole gene or exonic content below:

<i>ATM</i>	<i>BRCA1</i>	<i>BRCA2</i>	<i>PALB2</i>
------------	--------------	--------------	--------------

REFERENCES

1. Thoma, C. (2015) The complex relationships of malignant cells in lethal metastatic castration-resistant disease, *Nature Reviews Urology* 12, 237
2. Castro, E. *et al.*, (2012) The role of *BRCA1* and *BRCA2* in prostate cancer. *Asian Journal of Andrology*, 14 (3):409-414.
3. Pakkanen, S. *et al.*, (2009) *PALB2* variants in hereditary and unselected Finnish Prostate cancer cases. *Journal of Negative Results in BioMedicine*, 8 (1).

*The SureSeq™ FFPE DNA Repair Mix can only be purchased in conjunction with SureSeq NGS panels, not as a standalone product.

For research use only. Not for use in diagnostic procedures.

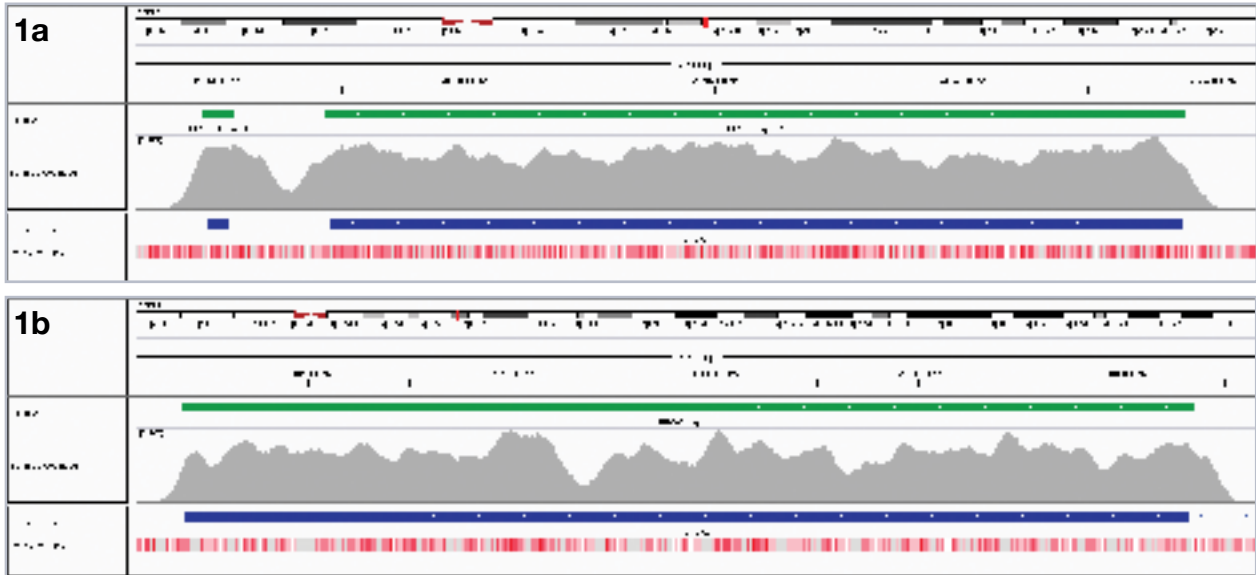
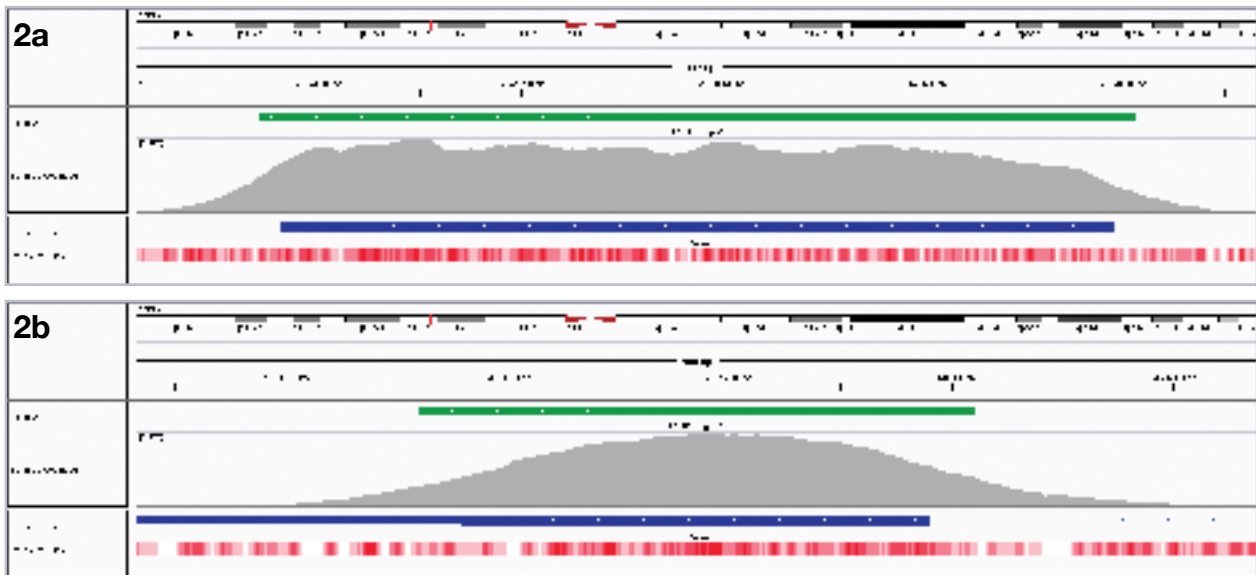


Figure 1a: *BRCA1* exon 9 and 10 coverage, Figure 1b: *BRCA2* exon 11 coverage. Depth of coverage per base (grey). Targeted region (green). Gene coding region as defined by RefSeq (blue). GC percentage (red).



Figures 2a and 2b: Illustration of the excellent uniformity of coverage of *PALB2* exons 5 (2a) and 13 (2b). Depth of coverage per base (grey). Targeted region (green). Gene coding region as defined by RefSeq (blue). GC percentage (red).

For information on the SureSeq FFPE DNA Repair Mix, see page 167.*

SureSeq CLL + CNV Panel

Chronic lymphocytic leukemia (CLL) is the most common type of leukemia in adults. A wide variety of chromosomal abnormalities are associated with CLL, ranging from single nucleotide variants (SNVs) and insertions/deletions (indels) up to large copy-number variations (CNVs), including trisomies.

The SureSeq CLL + CNV Panel has been designed in collaboration with recognized cancer experts to detect 12 key genes and 5 chromosomal regions implicated in CLL progression (Table 1). The SureSeq CLL + CNV Panel alleviates the burden of running multiple assays and streamlines your CLL research to deliver a comprehensive genomic profile for each CLL sample using a single workflow.

The SureSeq CLL + CNV Panel offers:

- Unparalleled uniformity and high depth of coverage — detect low-frequency SNVs and indels with confidence
- CNV detection ranging from loss of single exons to full chromosome arms and trisomy 12 — profile your samples for CNVs in the 5 most commonly aberrant regions in CLL
- Time savings - replace multiple assays with a single NGS panel, increasing throughput and reducing turnaround time
- Complimentary data analysis software — analyse your data with Interpret data analysis software, OGT's powerful and easy-to-use analysis solution for accurate identification of all variants and CNVs

OGT's expert bait design delivers outstanding uniformity and depth of coverage, offering confident detection of low frequency SNVs and indels down to 1% minor allele frequency (MAF) in 14 genes, including 2 genes and 24 SNPs to allow for easy sample tracking². The SureSeq CLL + CNV Panel covers the 5 most common CNVs in CLL and enables detection down to 10% MAF, corresponding to 20% tumor content. Compared to array data, often considered the gold standard for CNV detection, the events reported with the SureSeq CLL + CNV Panel were 100% concordant, even in genomic regions containing multiple aberrations (Figures 1-2). More so, facilitated by OGT's excellent bait design, loss-of-heterozygosity (LOH) can be identified. With CNV detection ranging from loss of single exons to full chromosome arms and trisomy 12, your data provides a more comprehensive genetic picture for each CLL sample from a single assay.

The SureSeq CLL + CNV Panel comes with OGT's complimentary data analysis software, Interpret. Designed to work seamlessly with all SureSeq panels, Interpret delivers fast and accurate detection of all SNVs, indels, LOH and CNVs covered by the panel, for an effortless translation of all your CLL data into meaningful results.



REFERENCES

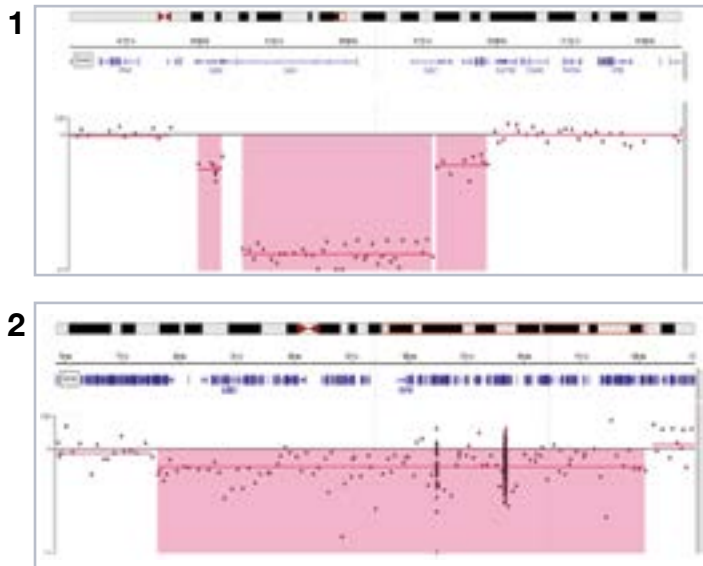
1. Döhner *et al.*, N Engl J Med 2000;343:1910-1916
2. Pengelly *et al.*, Genome Med 2013;5:89

The SureSeq CLL + CNV Panel in numbers

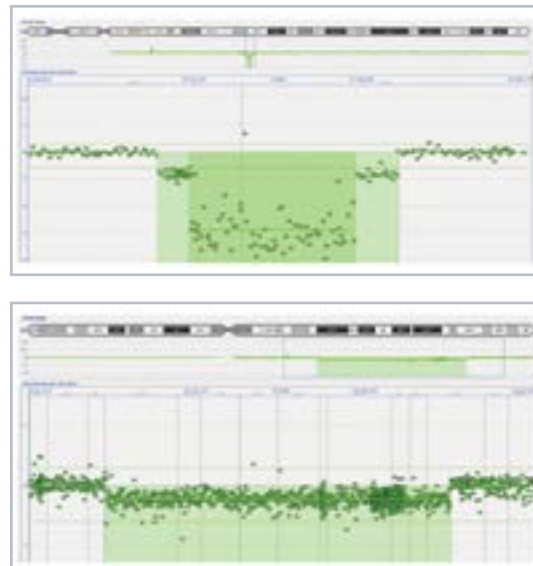
Feature	Specification
Number of genes	14
Uniformity Coverage	>99% of bases at >20% of the mean target coverage
Mean target coverage	>1000x
DNA input recommended	>500ng high quality DNA
Gene list	<i>ATM, PLAG2, BIRC3, BRAF, TP53, XPO1, SF3B1, KRAS, MYD88, SAMHD1, NOTCH1</i> and <i>BTK</i>
CNV list	17p (covering <i>TP53</i>), 11q (covering <i>ATM</i>), 13q (covering <i>RB1/DLEU2/DLEU7</i>), 6q (6q23.2-6q23.3 covering <i>MYB</i>) and Trisomy 12
Sample tracking	<i>CXCR4</i> and <i>SRY</i> + 24 SNP profiling panel ²
Limit of Detection	SNVs/indels: MAF of 1% within the 14 genes CNVs: MAF of 10% within the 5 chromosomal regions
CNV detection size	11q: - single exon to whole gene of <i>ATM</i> - > 5-10 Mb for the rest of the 11q arm 17p: - single exon to whole gene of <i>TP53</i> - > 5-10 Mb for the rest of the 17p arm 13q: - del(13)(q14) type I (short) and del(13)(q14) type II (larger) events covering <i>RB1/DLEU2/DLEU7</i> - > 10-20 Mb for the rest of the 13q arm 6q (6q23.2-6q23.3): - single exon to whole gene of <i>MYB</i> plus 1MB flanking sequence on either side Trisomy 12: - whole chromosome
LOH detection size	11q and 17p: 5-10 Mb 13q: 10-20 Mb
Samples per MiSeq® v2 run	16 samples/ run

Table 1: The SureSeq CLL + CNV Panel targets the 5 most common chromosomal regions implicated in CLL and 14 genes, including 2 genes and 24 SNPs for easy sample tracking².

NGS



ARRAY



2

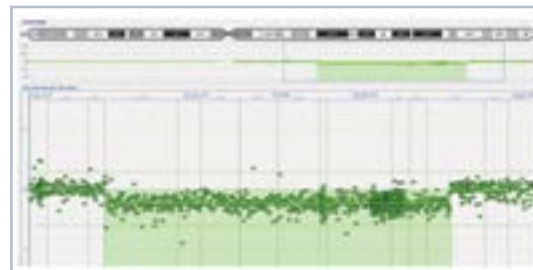
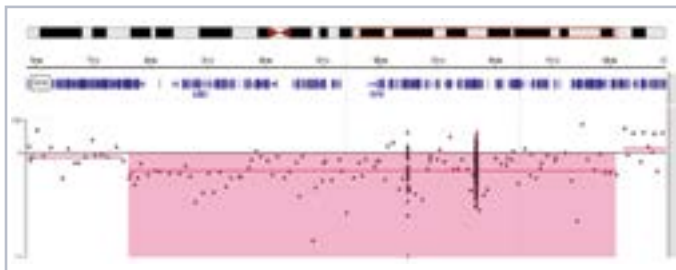


Figure 1: 0.6Mb biallelic loss called within a larger ~1Mb single allele deletion in the region covering *DLEU2/DLEU1/DLEU7* on chromosome 13q.
Figure 2: 42.7Mb deletion of 11q covering *ATM*.

SureSeq Core MPN Panel

Myeloproliferative neoplasms (MPNs) are a heterogeneous group of diseases characterized by the overproduction of one or more types of blood cells. The SureSeq Core MPN Panel has been designed in collaboration with recognized cancer experts to detect somatic variants in 3 clinically relevant MPN-associated genes; *JAK2*, *MPL* and *CALR* (Table 1). The SureSeq Core MPN Panel provides researchers with a single, 1-day NGS workflow for studies into the diagnosis, aetiology and prognosis of MPNs.

The SureSeq Core MPN Panel offers:

- Unparalleled uniformity and high depth of coverage — detect low frequency SNVs and indels with confidence
- Time and cost savings — replace multiple single gene assays with a focused NGS panel
- 1 day from sample to sequencer — streamlined library preparation and rapid 30-minute hybridization
- Additional *BCR-ABL* fusion gene detection — customize your panel by adding *BCR-ABL* translocation content
- Complimentary Interpret NGS data analysis software — easy-to-use analysis solution for accurate identification of all variants and translocations

The SureSeq Core MPN Panel is able to consistently detect SNVs and indels down to 1% minor allele frequency (MAF), using a streamlined 1-day workflow. Facilitated by OGT's expert bait design, the panel delivers the turn-around time of an amplicon-based protocol with the superior coverage uniformity of a hybridization-based panel, enabling confident detection of key MPN variants including a 52 bp deletion in *CALR* exon 9 and a 6 bp deletion in *JAK2* exon 12 (Figures 1 and 2).

The Core MPN Panel in numbers

Feature	Specification
Target regions	<i>JAK2</i> exons 12 and 14 <i>CALR</i> exon 9 <i>MPL</i> exon 10
Panel size	1kb
Mean target coverage	>1000x
Coverage uniformity	V617F
DNA input recommended	>500ng high quality DNA
Limit of detection	SNVs / indels: 1% MAF
Workflow	30 minutes hybridization, 1-day sample-to-sequencer
Samples per MiSeq® v2 run	48 samples / run

Table 1: The SureSeq Core MPN Panel targets 4 exons in 3 genes implicated in MPNs, covering various key MPN driver mutations.

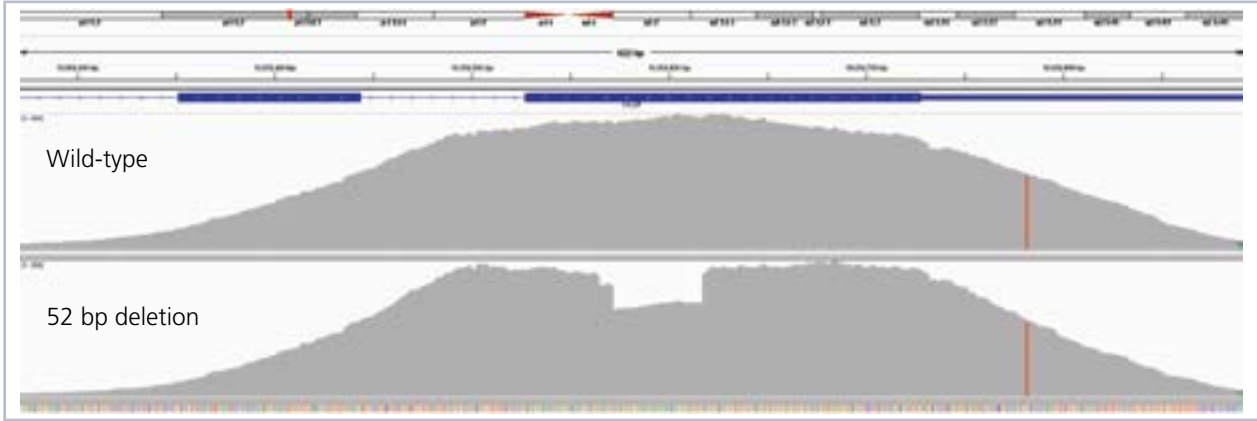


Figure 1: Detection of a 52 bp deletion (type 1) in exon 9 of *CALR* (bottom panel), compared to a wild-type sample (top panel).

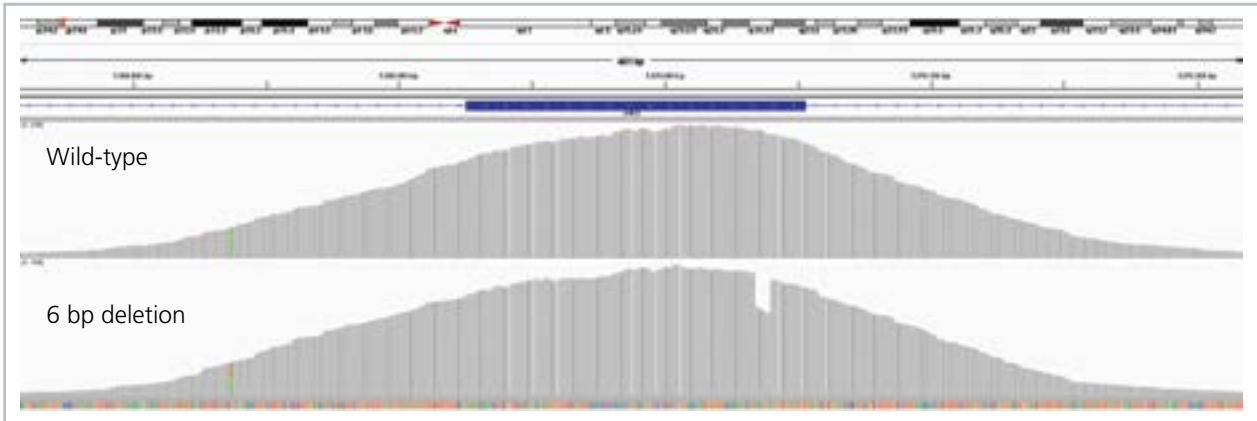


Figure 2: Detection of a 6 bp deletion in exon 12 of *JAK2* (bottom panel), compared to a wild-type sample (top panel).



Figure 3: *BCR-ABL* translocation reported in Interpret. Split-reads covering both *BCR* (left panel) and *ABL1* (right panel) are detected, indicative of the *BCR-ABL* gene fusion.

We would like to thank Professor Nick Cross (National Genetics Reference Laboratory - Wessex, UK) for providing the validated research samples and West Midlands Regional Genetic Laboratory, Birmingham, for providing the *BCR-ABL* samples.

SureSeq Ovarian Cancer Panel

Hybridization-based NGS panel validated on FFPE samples and whole blood; it allows the analysis of variants associated with ovarian cancer and research into therapeutic response



The SureSeq Ovarian Cancer Panel:

- Contains the latest evidence-based genes involved in ovarian cancer research — gain insight into homologous repair deficiencies and cell cycle dysregulation
- Is validated for research use on FFPE and whole blood — detect germline mutations in DNA derived from blood as well as both germline and somatic mutations in DNA derived from FFPE tissue
- Utilizes hybridization-based enrichment — sensitive and reproducible detection of low-frequency variants, even in heterogeneous cancer samples
- Fast and easy workflow — streamlined library preparation, short 4-hour hybridization and intuitive software allowing easy variant analysis
- Delivers excellent uniformity of coverage across the whole panel — over 99% of targeted regions are covered to at least 20% of mean target coverage

Ovarian cancer is the leading cause of death from gynecological cancers in the Western world¹. Next generation sequencing (NGS) is quickly becoming a commonly used tool for analysis of mutations — both single nucleotide variants (SNVs) and insertion/deletions (indels) — in genes associated with ovarian cancer. The SureSeq Ovarian Cancer Panel has been developed with leading cancer experts and covers all coding exons of seven genes (Table 1). The panel allows detection of known and novel variants in tumor suppressor genes as well as genes involved in homologous repair to advance research into ovarian cancer treatment. It has been validated on DNA derived from FFPE tissue and whole blood to allow investigation of both germline and somatic mutations.

Utilizing hybridization-based enrichment, the SureSeq Ovarian Cancer Panel delivers excellent run-to-run consistency and extremely uniform coverage across the whole region of interest (Figure 1) to allow sensitive detection of variants present, even at low minor allele frequency (MAF) (Table 2).

The SureSeq Ovarian Cancer panel is optimized to work with the SureSeq NGS Library Preparation Kit*. For more information, see page 165.

REFERENCES

1. Helleman J. *et al.* (2006) Molecular profiling of platinum resistant ovarian cancer. *Int J Cancer* 118(8):1963-71.

*The SureSeq™ FFPE DNA Repair Mix can only be purchased in conjunction with SureSeq NGS panels, not as a standalone product.

For research use only. Not for use in diagnostic procedures.



BRCA1	BRCA2	TP53	PTEN	ATM	ATR	NF1
-------	-------	------	------	-----	-----	-----

Table 1: The SureSeq Ovarian Cancer Panel targets seven genes implicated in ovarian cancer.

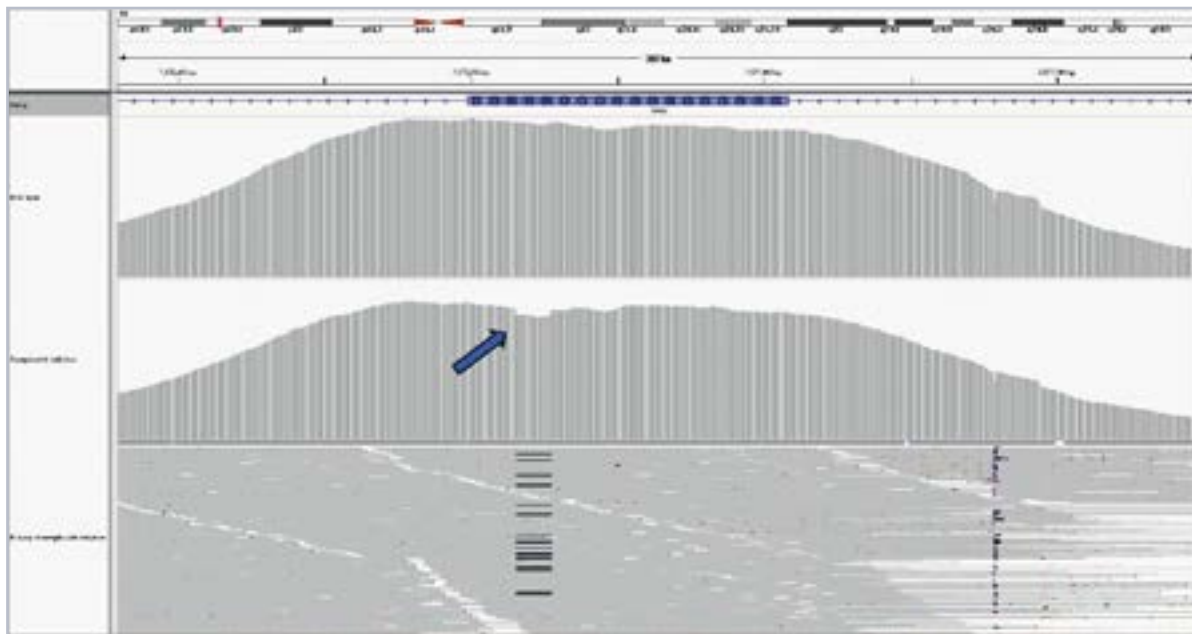


Figure 1: OGT's expert bait design delivers excellent uniformity of coverage. Shown here is FFPE sample, exon 7 of *TP53* (NM_000546). The top panel shows a normal control, the bottom panel shows a deletion (c.754_765delCTCACCATCATC) at 6% frequency. Mean target coverage >1400x, 12 samples per MiSeq lane.

Gene	Variant detected	Type of variant	Mean target coverage	% MAF detected
<i>BRCA1</i>	c.3424G>C (p.Ala1142Pro)	SNV	881	11.92%
<i>BRCA2</i>	c.556G>C (p.Ala186Pro)	SNV	728	1.92%
<i>TP53</i>	c.1129A>C (p.Thr377Pro)	SNV	1024	3.03%
<i>ATR</i>	c.7274G>A (p.Arg2425Gln)	SNV	1579	38.63%
<i>NF1</i>	c.8137_8138insG (p.Phe2714ValfsTer16)	Insertion	683	1.45%
<i>NF1</i>	c.3354delT (p.Ser1118ArgfsTer24)	Deletion	621	1.13%
<i>ATR</i>	c.4154delC (p.Thr1385MetfsTer3)	Deletion	506	1.61%

Table 2: Example mutations detected in FFPE clinical research samples using the SureSeq Ovarian Cancer Panel. The ability to detect MAFs as low as 1.13% gives added confidence in the variants being called and facilitates the exploration of tumour heterogeneity. Rows 1–4: low-frequency SNVs; rows 5–7: low-frequency indels. Samples kindly provided by Biopathology Department of Gustave Roussy, Villejuif, France.

For information on the SureSeq FFPE DNA Repair Mix, see page 167.*

SureSeq Myeloid Panel

25-gene myeloid disorder hybridization-based NGS enrichment panel that delivers accurate and easy identification of variants

The SureSeq Myeloid Panel delivers:

- Most up-to-date content designed in collaboration with recognized cancer experts — detect SNVs and indels in 25 genes implicated in a variety of MPNs
- Time and cost saving solution — replace multiple single gene assays with one comprehensive panel
- Sensitive and reproducible variant detection even in heterogeneous samples — detect low-frequency alleles down to 1% MAF with confidence
- Fast and easy workflow — streamlined library preparation, rapid hybridization and intuitive software allowing easy variant analysis
- Excellent coverage uniformity — 99% of targeted regions are covered to at least 20% of mean target coverage

Myeloproliferative neoplasms (MPNs) are a group of diseases that affect normal blood cell production in the bone marrow resulting in overproduction of one or more cell types (i.e. red cells, white cells or platelets). There are numerous different sub-types of MPNs that are distinguished from each other by the type of cell which is most affected and the genetic profile. The SureSeq Myeloid Panel targets selected key genes known to contain driver mutations for a range of MPNs including polycythaemia vera (PV), essential thrombocythaemia (ET) and myelofibrosis (MF) (Table 1). To obtain the optimal sensitivity whilst maximizing throughput, hot exons where clinically relevant mutations are known, and every exon for tumor suppressor, hereditary and highly implicated research-related genes, are targeted. This allows detection of previously characterized as well as novel variants in myeloid samples.

Instead of assaying for single genes in a sequential manner, the mutational status of twenty-five genes can be rapidly and simultaneously determined with the use of the SureSeq Myeloid Panel.

The SureSeq Myeloid Panel has been validated with samples from the National Institute for Biological Standards and Control (NIBSC) and has been shown to accurately detect alleles down to 1% minor allele frequency (MAF) at a read depth of >1000x.

OGT's expert bait design ensures efficient and more uniform capture of all targeted regions than amplicon-based technologies, so that all variants present can be called with maximum confidence. This has been demonstrated on the *CALR* gene, which is commonly mutated in various MPNs. It is critical to identify key *CALR* indels (types 1 & 2 causing a frameshift) as well as increasingly recognized point mutations in this gene. The SureSeq Myeloid Panel delivers superior performance to panels designed using standard algorithms, by ensuring uniform coverage over the regions of interest (Figure 1).





ASXL1	EGLN1	IDH2	NRAS	SRSF2
CBL	EPAS1	JAK2	RUNX1	TET2
CALR	EPOR	KIT	SETBP1	TP53
CSF3R	EZH2	KRAS	SF3B1	U2AF1
DNMT3A	IDH1	MPL	SH2B3	VHL

Table 1: The SureSeq Myeloid Panel targets 25 genes implicated in a variety of MPNs. The gene content has been defined with input from recognized cancer experts including Professor Mike Griffiths (West Midlands Regional Genetics Laboratory, UK) and Professor Nick Cross (National Genetics Reference Laboratory – Wessex, UK).

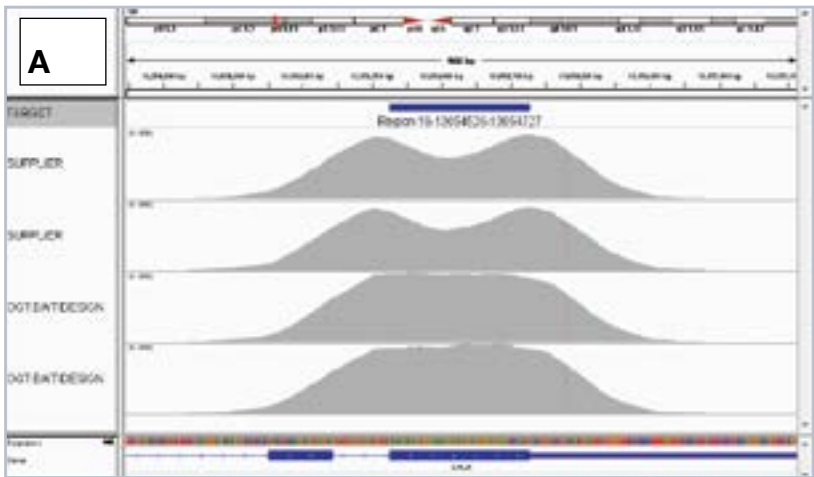
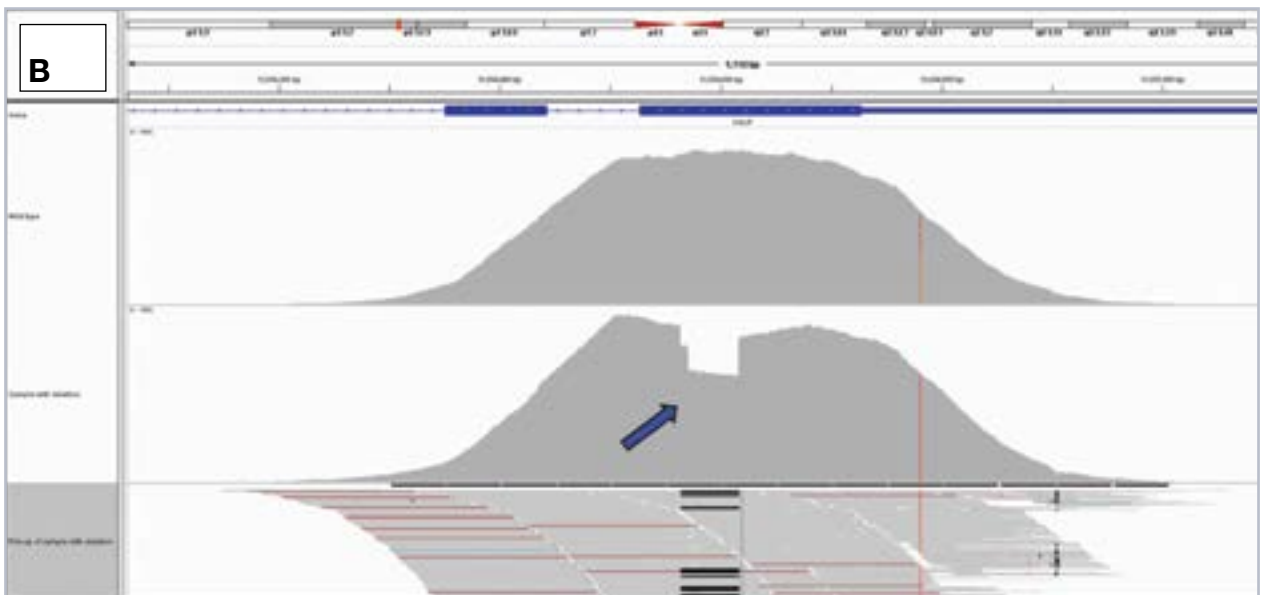


Figure 1: A) OGT's expert bait design delivers improved uniformity of coverage. Shown here is exon 9 of *CALR*. The top two captures have been completed using baits designed with standard commercially available software. They have a considerable dip in coverage in the middle of the exon due to the fact it presents a low complexity region with low nucleotide diversity. Most algorithms would want to avoid such regions in the design. However, OGT's superior bait design can increase the evenness of coverage of such regions. B) The top panel shows a normal control sample and the bottom panel shows 23% deletion (c.1092_1143del_52bp) in *CALR* exon 9. Mean target coverage >1000x, 24 samples per MiSeq lane.



Interpret Software

Interpret is a powerful and easy-to-use next generation sequencing analysis solution. Sequencing data can be quickly processed to deliver accurate identification of single nucleotide variants (SNVs) and indels as well as structural variants such as internal and partial tandem duplications (ITDs and PTDs), and copy-number variations (CNVs) including trisomies, loss-of-heterozygosity (LOH) and translocations. Coupled with a comprehensive and powerful filtering framework, the software delivers accurate mutation calling with 100% sensitivity and 99.9% specificity at >1% variant allele frequency (VAF)*.

Interpret is designed to work seamlessly with OGT's extensive range of SureSeq and CytoSure NGS panels and offers flexible accessibility to analyse your data whether through a stand-alone computer, laboratory server or another web enabled device.

Interpret Software offers:

- Extensive customization options — easily customize variant and batch reports and database links to meet the exact needs of your laboratory
- Comprehensive range of filtering options — standardize your analysis workflow and overlay bespoke variant filtering to meet your analytical criteria
- Security and control — log and track user activity and standardize analysis protocols through multiple access permission levels
- Powerful analysis capability — optimized for use with OGT's NGS panels for confident annotation and reporting of low frequency variants

Used in conjunction with OGT's NGS panels, Interpret compliments the expert panel design and hybridization-based approach of SureSeq and CytoSure NGS to enable the effortless translation of NGS data into meaningful results.

For more information on Interpret, visit www.ogt.com/InterpretNGS



*Sensitivity and specificity determined using Horizon Discovery OncoSpan and TruQ7 and HapMap (NA12878) standards.

For research use only. Not for use in diagnostic procedures.

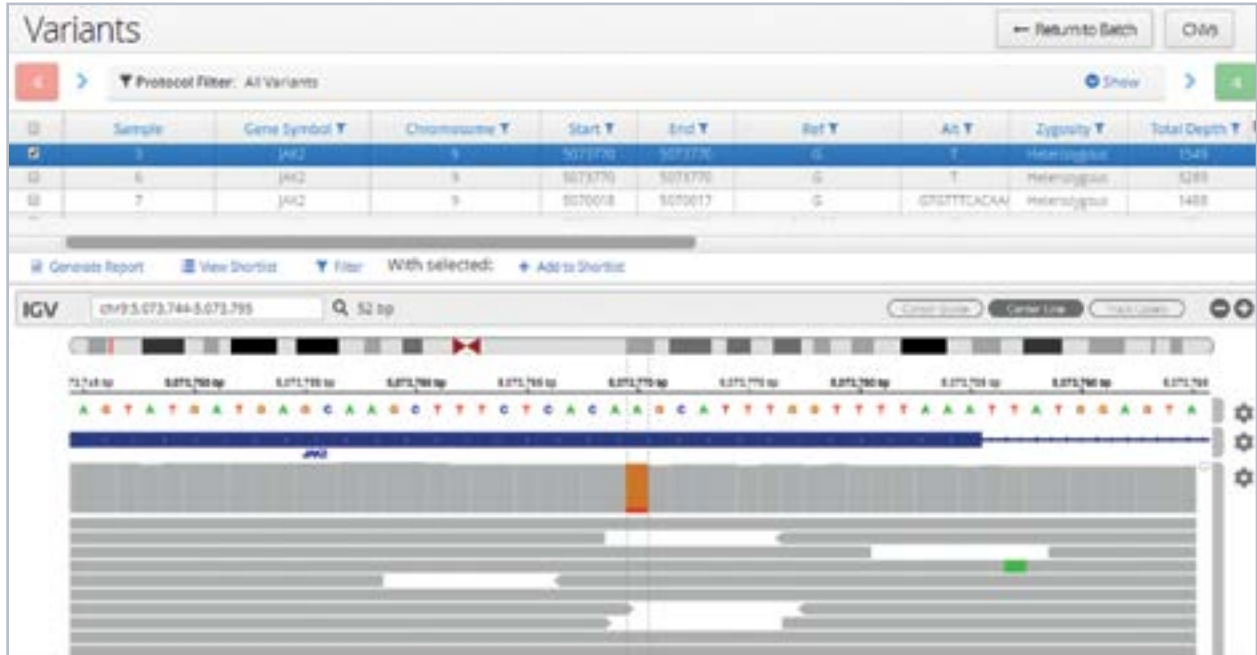


Figure 1: Following analysis, all variants are displayed in a table, below which is an IGV window allowing a more detailed review of the data and additional verification. In this example a low frequency *JAK2* V617F SNV has been selected and the user is able to view the aligned reads generated by the pipeline.

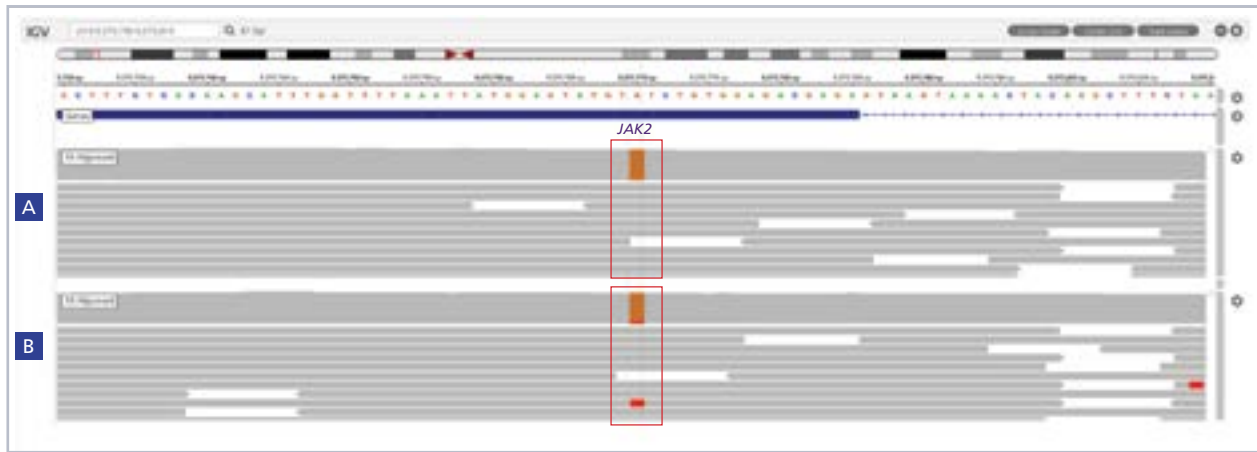


Figure 2: Detection of low frequency somatic variants, as **A** 1% and **B** 9% *JAK2* V617F mutations using a SureSeq Myeloid Panel.

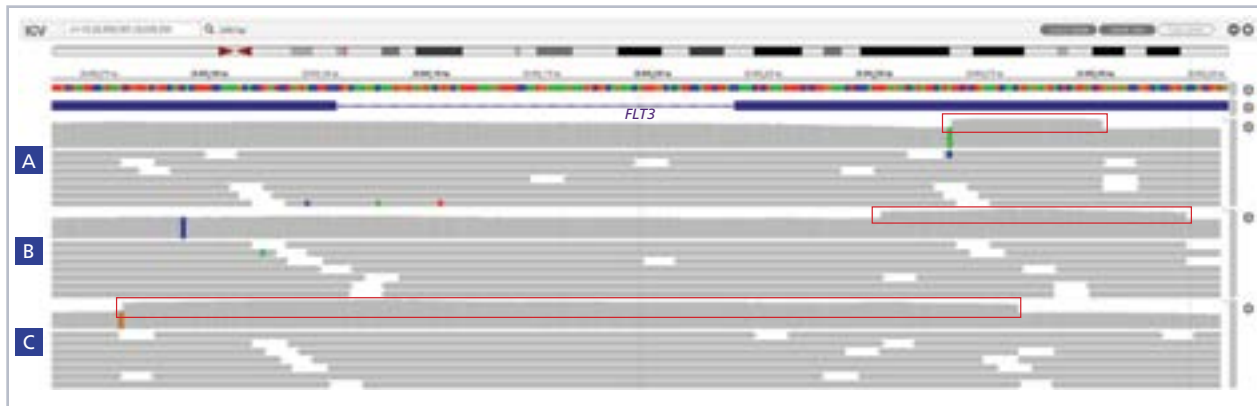


Figure 3: Detection of *FLT3* ITDs of difference sizes, **A** 33 bp, **B** 69 bp, **C** 201 bp, using a SureSeq myPanel Custom AML Panel. Note how OGT's innovative panel design in conjunction with Interpret is able to identify and call ITDs much longer than the sequencing read length of 150 bp.

SureSeq NGS Library Preparation Kit

The complete library preparation solution for unparalleled next generation sequencing (NGS) results.

The SureSeq NGS Library Preparation Kit generates NGS libraries suitable for the capture of targeted genomic regions using hybridization. With a streamlined workflow, significantly reduced hands-on steps and hybridization times as low as 30 minutes, SureSeq offers all of the benefits of hybridization in as little as a 1-day workflow. The SureSeq NGS Library Preparation Kit delivers high performance in the quality metrics that really matter, giving more reliable, more trustworthy data. Exceptionally low levels of duplication (Figure 2) ensures more accurate calling, more even coverage and higher levels of confidence in the data produced.

The inclusion of the SureSeq Hyb & Wash buffer, optimized for use with SureSeq NGS panels, simplifies this key step while offering excellent coverage uniformity (Figure 3) and reproducibility. It contains all the components ready-to-use to perform the hybridization and wash steps in SureSeq sequence capture protocols, eliminating the requirement to dilute buffers and the possibility of cross-contamination during buffer preparation.

SureSeq myPanel offers:

- Greater trust in your data — high performance with low duplication rates, high sequence quality and high percentage of on-target bases
- Simpler hybridization — all components of the SureSeq Hyb & Wash buffer are ready-to-use with no requirement for multiple wash buffers
- Rapid process — streamlined protocol, minimal manual handling, automation and a rapid hybridization step offers increased reliability as well as throughput
- Reliable results — NGS targeted panel, complete NGS library preparation solution and powerful



Rapid process with streamlined protocol

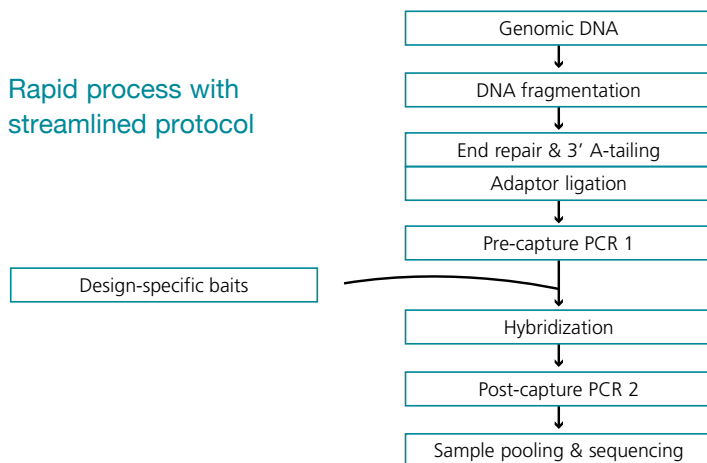


Figure 1: A streamlined protocol, including enrichment by hybridization. The complete procedure can be completed in 1.5 days with minimal handling time.

For research use only. Not for use in diagnostic procedures.

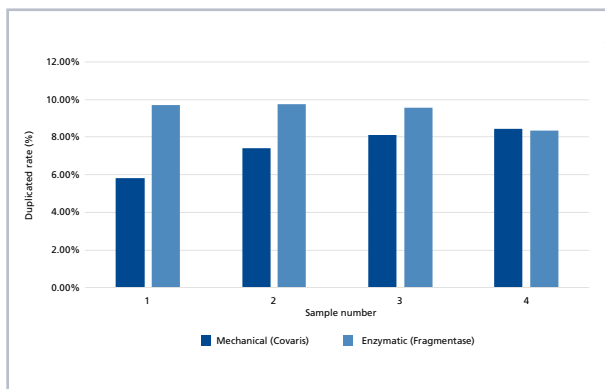


Figure 2: The SureSeq NGS Library Preparation Kit delivers low levels of sequence duplication. The duplication rates are shown for samples fragmented by mechanical or enzymatic methods. Samples were prepared using the SureSeq NGS Library Preparation kit and hybridized with a SureSeq myPanel Custom Myeloid 49 Gene Plus panel.

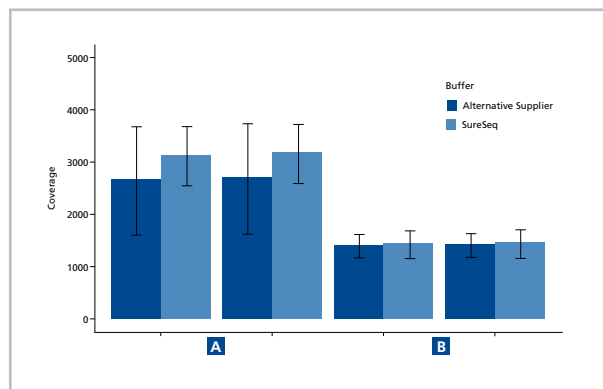


Figure 3: Superior performance. Comparison of the mean target coverage generated using the SureSeq Hyb & Wash Buffer Kit (light blue) compared to an alternative supplier kit (dark blue). **A** SureSeq myPanel custom CLL panel and **B** SureSeq myPanel custom CLL plus CNV panel.

Ordering information

Product	Contents	Cat. No.
SureSeq NGS Library Preparation Complete Solution (16)	Bundle of 1x SureSeq library preparation kit (16), containing adaptors, PCR primers and enzymes, 1x SureSeq Solution (16) NGS Index Kit – Collection A, 1x SureSeq Hyb & Wash Kit (16), 1x Dynabeads M270 Streptavidin (2ml) and 1x AMPure XP beads (10ml). Sufficient for 16 samples	500084
SureSeq NGS Library Preparation Complete Solution (48)	Bundle of 3x SureSeq NGS Library Preparation Kit (16), containing adaptors, PCR primers and enzymes, 1x SureSeq Solution (48) NGS Index Kit – Collection B, 3x SureSeq NGS Hyb & Wash Kit (16), 3x Dynabeads M270 Streptavidin (2ml) and 3x AMPure XP beads (10ml). Sufficient for 48 samples	500085
SureSeq NGS Library Preparation and Hyb & Wash Kit (16)	Bundle of 1x SureSeq NGS Library Preparation Kit (16), containing adaptors, PCR primers and enzymes, 1x SureSeq Hyb & Wash Kit (16) NGS Index Kit – Collection A and 1x SureSeq Hyb & Wash Kit (16). Sufficient for 16 samples	500082
SureSeq NGS Library Preparation and Hyb & Wash (48)	Bundle of 3x SureSeq NGS Library Preparation Kit (16), containing adaptors, PCR primers and enzymes, 1x SureSeq NGS Index Kit - Collection B and 3x SureSeq Hyb & Wash kit (16). Sufficient for 48 samples	500083
SureSeq NGS Library Preparation Kit (16)	Bundle of 1 x library preparation kit (16), containing adaptors, PCR primers and enzymes sufficient for 16 samples and 1 x SureSeq NGS Index Kit – Collection A	500070
SureSeq NGS Library Preparation Kit (48)	Bundle of 3 x library preparation kit (16), containing adaptors, PCR primers and enzymes sufficient for 48 samples and 1 x SureSeq NGS Index Kit – Collection B	500073
SureSeq NGS Hyb & Wash Kit (16)	Hybridization buffer, Wash buffer, Cot and blocking oligos. Sufficient for 16 samples	500075
SureSeq NGS Hyb & Wash Kit (48)	Bundle of 3x SureSeq NGS Hyb & Wash Kit (16), containing Hybridization buffer, Wash buffer, Cot and blocking oligos. Sufficient for 48 samples	500086

The SureSeq NGS Library Preparation Kit was jointly developed between Oxford Gene Technology and Bionline Reagents Limited.

SureSeq FFPE DNA Repair Mix

SureSeq FFPE DNA Repair Mix:

- Optimized to repair a broad range of damage in FFPE-derived DNA — remove artefacts caused by fixation and long-term storage
- Improves NGS library yields, %OTR and mean target coverage — get excellent sequencing data for confident variant calling from FFPE DNA
- Allows decreased amount of input DNA — preserve your precious samples and get meaningful results from as little as 100 ng of FFPE DNA

Tissue biopsies are typically archived as formalin-fixed, paraffin-embedded (FFPE) blocks, which preserve tissue morphology and allow long-term storage at room temperature. However, the methods used for fixation significantly damage and compromise the quality of nucleic acids from these samples. Consequently, it may be difficult to distinguish between true and damage-induced low-frequency mutations in such samples. The SureSeq FFPE DNA Repair Mix is a mixture of enzymes that has been optimized to remove a broad range of damage that can cause artefacts in sequencing data (Table 1).

The SureSeq FFPE DNA Repair Mix has been shown to significantly improve NGS library yields, preserving original complexity and delivering high-quality sequencing data for confident calling of variants with low minor allele fractions (MAFs). It also increases depth of coverage and %OTR improving sensitivity of your test (Figure 1).

Pathology labs often have to work with very limited amounts of material. Additionally, FFPE samples are usually irreplaceable. This leads to the need to reduce DNA input in downstream applications including NGS. Often amplicon-based approaches are chosen as they require very little input material. Unfortunately, due to PCR bias and lower complexity from smaller input amounts, these methods are not well suited to detect low-frequency mutations in heterogeneous tumor samples. Hybridization-based approaches eliminate the problem of PCR bias providing much more reliable data but they typically require higher DNA inputs of 500 ng – 1 µg. Using the SureSeq FFPE DNA Repair Mix a reduction in the amount of starting material down to 100 ng depending on required depth of coverage is possible.



Damage	Repaired?
Deamination of cytosine to uracil	✓
Nicks and gaps	✓
Oxidised bases	✓
Blocked 3' ends	✓
DNA fragmentation	✗
DNA-protein crosslinks	✗

Table 1: The SureSeq FFPE DNA Repair Mix is capable of removing a variety of DNA damage caused by fixation and long-term storage.



Figure 1: The SureSeq FFPE DNA Repair Mix significantly improves mean target coverage resulting in more confident calls. Data obtained using 500ng of FFPE DNA from ovarian and colon cancer samples; 16 samples per MiSeq lane.

*The SureSeq™ FFPE DNA Repair Mix can only be purchased in conjunction with SureSeq NGS panels, not as a standalone product.

For research use only. Not for use in diagnostic procedures.

CytoSure™

CytoSure NGS and Array Products



**NGS & Array Products
for Cytogenetics and
Rare Disease Research**

Contents

RUO

CytoSure Next Generation Sequencing (NGS)

- 171 CytoSure Constitutional NGS
- 173 CytoSure NGS Comprehensive FH Panel

Constitutional Arrays

- 175 CytoSure Constitutional v3 and
Constitutional v3 + LOH Arrays

CytoSure Disease-Focused Arrays

- 177 CytoSure Medical Research Exome Array
- 179 CytoSure Disease-Focused Arrays
- 180 CytoSure Autism Research Array
- 180 CytoSure Cardiomyopathy Research Array
- 181 CytoSure Epilepsy Research Array
- 181 CytoSure Eye Disease Research Array
- 182 CytoSure NMD Research Array

Cancer Arrays

- 183 CytoSure Cancer +SNP Arrays

- 186 CytoSure Custom Designed aCGH Arrays
- 187 CytoSure Interpret Software
- 189 CytoSure Genomic DNA Labelling kits
- 191 CytoSure Sample Tracking Spike-ins
- 192 Accessories

Overview

The CytoSure™ brand offers comprehensive microarray and Next Generation Sequencing (NGS) solutions for research into a wide range of genetic aberrations, including constitutional and molecular disorders.

For a number of years, array comparative genomic hybridization (aCGH) has been considered the primary technique for research into copy number variation (CNV) and loss of heterozygosity (LOH) analysis in cytogenetic and rare disease samples. CytoSure microarrays have been designed and optimized to provide the best analysis possible, offering genome-wide coverage and single-exon resolution, facilitating accurate detection of microdeletions and duplications. Probe design and development is critical and OGT's probe design algorithms have allowed us to generate an Oligome™ database of ~26.5 million optimized probes to select from, ensuring optimal probe selection and array performance. All our array products include our class-leading CytoSure Interpret software for fast and simple interpretation of your data.

Building on our experience with CNV and LOH calling from our array offering, OGT has developed the CytoSure NGS platform, which includes the CytoSure Constitutional NGS panel and the Familial Hypercholesterolemia (FH) panels, adding the benefit of SNV and indel analysis capability. The CNV calling performance has been made possible by excellent panel design, in tandem with state-of-the-art analytical software. The CytoSure Constitutional NGS panel has been designed to offer a comprehensive targeted intellectual disability (ID) and developmental delay (DD) genetic assay — with over 700 ID/DD genes targeted at the exon level and a backbone of additional targets spread throughout the genome. It enables the seamless transition from microarrays to NGS, delivering a significant increase in information obtained from a single assay without extensive analysis time and costly data generation and storage.

For more information, visit www.ogt.com/CytoSure

RUO: For research use only. Not for use in diagnostic procedures.

CytoSure™ Constitutional NGS

Unparalleled CNV calling in a targeted Constitutional NGS panel.

The CytoSure Constitutional NGS solution delivers:

- The ability to detect CNV, SNV, indel, LOH, and mosaicism
- Advanced panel design and software
- Robust single-exon CNV calling unlike other large targeted panels or exomes
- The most up-to-date content for ID and DD
- A targeted >700-gene panel, minimising variants of uncertain significance (VUS) detection
- Regions across the genome are targeted to create a backbone of coverage
- Cost-effective analysis



Comprehensive aberration detection

The NGS panel is designed to cover important genes for ID/DD and also contains a backbone of baits covering common single nucleotide polymorphisms (SNPs), this allows detection of a comprehensive range of aberration types including CNVs, SNVs, indels and LOH in a single assay (Figure 1). This also includes detection within mosaic samples (Figure 2). The Interpret software (see page 163) user interface is conveniently arranged and also has the ability to switch between CNV/LOH calls and SNV/indel analysis, enabling a step-by-step approach to the interpretation process.

Advanced panel design and software for robust single-exon CNV calling

A key requirement in enabling the transition from a microarray-based technology to NGS for CNV detection is the ability to ensure that CNV data from the NGS panel is concordant with that from microarrays, particularly for small, sub-gene duplications and deletions. OGT's expertise in bait design ensures uniform sequencing coverage of the desired regions. This, coupled with Interpret's proprietary CNV calling algorithm, allows robust detection of even the smallest CNVs.

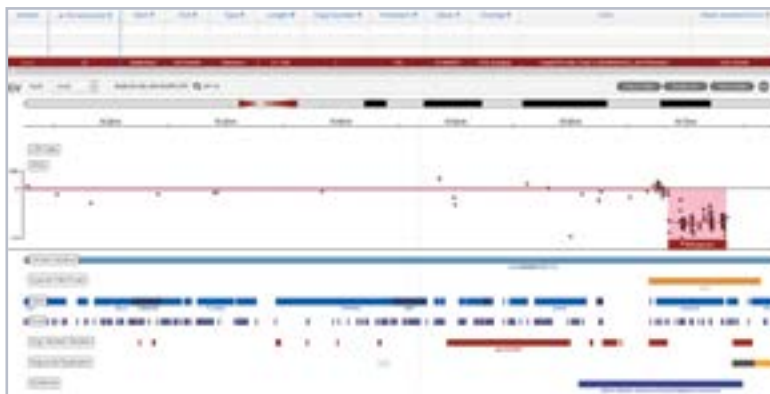


Figure 1: Detection of a 51kb deletion within the *SHANK3* gene on chromosome 22.*

For research use only. Not for use in diagnostic procedures.

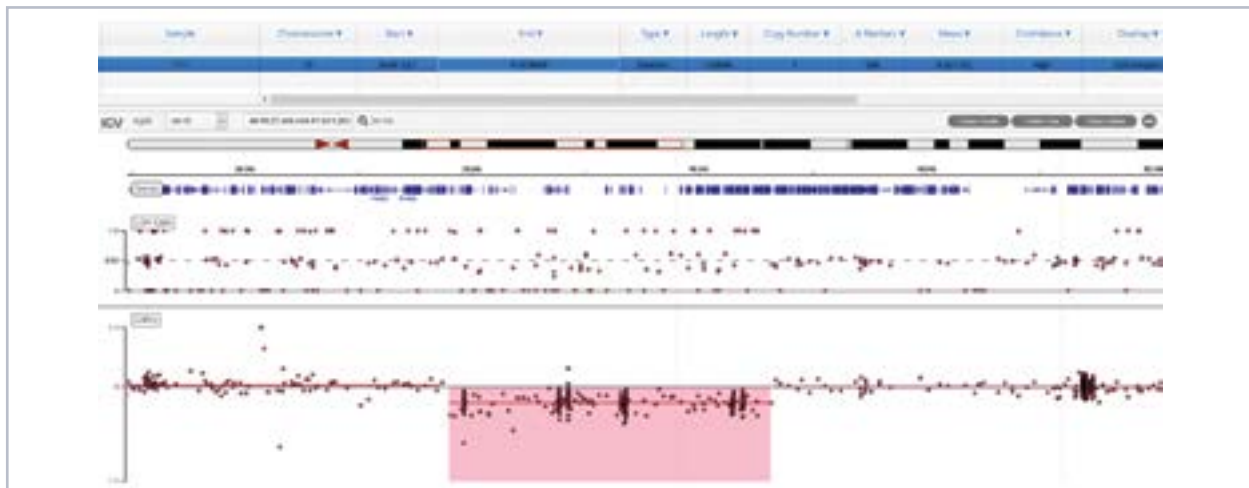


Figure 2: Sample with a 7Mb deletion at 15q14q15.1 within a mosaic sample.*

Ordering details and related products

For information on complimentary Interpret software, please see page 163.

Product	Contents	Cat. No.
CytoSure Constitutional NGS Solution (24)	Bundle of 1x CytoSure Constitutional NGS Panel (24), 1x CytoSure NGS Library Preparation Kit (24) and 1x CytoSure NGS Hybridization & Wash Kit (24)	502005-B24
CytoSure Constitutional NGS Panel (24)	Enrichment baits sufficient for 24 samples	502003-24
CytoSure NGS Library Preparation Kit (24)	Library Preparation Kit containing adaptors, PCR primers and enzymes sufficient for 24 samples	502001-24
CytoSure NGS Hybridization & Wash Kit (24)	Hybridization buffer, Wash buffer, Cot and blocking oligos. Sufficient for 24 samples	502002-24
CytoSure Constitutional NGS Solution (96)	Bundle of 1x CytoSure Constitutional NGS Panel (96), 1x CytoSure NGS Library Preparation Kit (96) and 1x CytoSure NGS Hybridization & Wash Kit (96)	502005-B96
CytoSure Constitutional NGS Panel (96)	Enrichment baits sufficient for 96 samples	502003-96
CytoSure NGS Library Preparation Kit (96)	Library Preparation Kit containing adaptors, PCR primers and enzymes sufficient for 96 samples	502001-96
CytoSure NGS Hybridization & Wash Kit (96)	Hybridization buffer, Wash buffer, Cot and blocking oligos. Sufficient for 96 samples	502002-96
CytoSure NGS Index Kit	48 Indexes sufficient for 4 reactions each	502004

* Clinical research sample provided courtesy of Centre hospitalier universitaire de Sherbrooke (CIUSSSE-CHUS)

CytoSure™ Comprehensive FH Panel

The CytoSure Comprehensive FH Panel facilitates the detection of exonic CNVs involved in Familial Hypercholesterolemia (FH) when used in conjunction with OGT’s Interpret bioinformatics solution.

To date, most NGS CNV analysis approaches have been designed for whole genome/exome sequencing and besides being less robust than standard aCGH, they are not suitable for small targeted NGS panels. This is illustrated with the *LDLR* gene, thought to have a prevalence between 1/500 and 1/200¹ in FH. When intragenic CNVs detected using the panel were confirmed with aCGH (custom CytoSure arrays are available for confirmation of CNV detection), the concordance was 100% over the targeted exons of the NGS panel, offering confident and reliable CNV and SNV determination.

The CytoSure Comprehensive FH Panel offers:

- Confident variant detection, minimising the requirement for supplementary fill-in with Sanger sequencing
- Detection of CNVs as well as SNVs in a single assay — no need to run MLPA alongside your NGS panel
- Pre-optimized panels that meet your technical requirements — no more laborious in-house optimization, decreasing assay development time

The hybridization methodology, combined with our bait design expertise, allows generation of panels with outstanding completeness and coverage uniformity. Together, this allows the areas of CNV to be easily identified within each sample using our proprietary algorithm — delivering an increased understanding of the sample without an increase in cost or time.

All exons

<i>LDLR</i>	<i>LDLRAP1</i>	<i>APOB</i>	<i>PCSK9</i>
<i>APOE</i>	<i>LIPA</i>	<i>ABCG5</i>	<i>ABCG8</i>

REFERENCES

1. Brice, P; Burton, H; Edwards, CW; Humphries, SE; Aitman, TJ; (2013) Familial hypercholesterolaemia: A pressing issue for European health care. *Atherosclerosis*, 231(2), pp. 223-226.

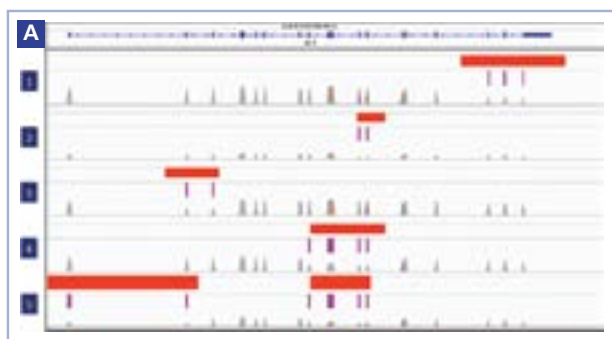
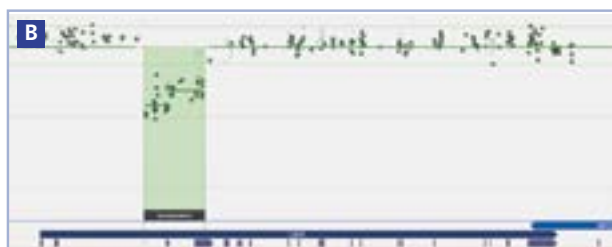


Figure 1: CNV in *LDLR* gene shown using IGV from the Broad Institute **A**: Red bars indicate areas of CNV (data from aCGH), purple bars represent deleted exons (data from NGS): 5 samples are shown, each with at least one area of CNV. There is complete concordance between the aCGH and NGS data. Note the evenness of the NGS coverage (even peak height) across each exon, allowing the areas of CNV to be easily identified. The data from the custom CytoSure aCGH array, confirms the deletions in *LDLR*. **B** A 2 exon deletion and **C**, a deletion of 2 exons and 4 exons, corresponding to samples 3 and 5 in **A** respectively.



Ordering details and related products

For information on complimentary Interpret software, please see page 163.

Product	Contents	Cat. No.
CytoSure Comprehensive FH Panel (16 reactions)	Enrichment baits; Interpret Software	601004-16
CytoSure Comprehensive FH Panel (96)	Enrichment baits; Interpret Software	601004-96
SureSeq NGS Library Preparation Kit (16)	Bundle of 1x library preparation kit (16) containing adaptors, PCR primers and enzymes sufficient for 16 samples and 1x SureSeq NGS Index Kit – Collection A	500070
SureSeq NGS Library Preparation Kit (48)	Bundle of 3x library preparation kit (16), containing adaptors, PCR primers and enzymes sufficient for 48 samples and 1x SureSeq NGS Index Kit – Collection B	500073

CytoSure Constitutional v3 and v3 + LOH arrays

Enhanced exon-level coverage of developmental disorder genes and the latest ClinGen* and DDD content

The CytoSure Constitutional v3 arrays deliver:

- Up-to-date developmental disorder content — all the latest research-validated genes and regions
- Single exonic CNV detection in the genes that matter — enabling high resolution CNV detection in up to 502 genes of interest
- Integrated sample tracking probes and optimized labeling kits — the complete solution for reliable analysis and reporting
- Streamlined data analysis and interpretation — straightforward and fast analysis of CNVs and LOH

Enhanced exon-level coverage of all developmental disorders and the latest ClinGen and DDD content

CytoSure Constitutional v3 arrays have been developed in collaboration with experts at the Wellcome Trust Sanger Institute. These unique arrays combine the most up-to-date and relevant developmental delay content from the recent Deciphering Developmental Disorders (DDD) study with the latest updates from ClinGen the Clinical Genome Resource¹.

Higher probe density across the exons and introns of important developmental delay genes allows improved detection of small (<500bp) deletions and duplications that might otherwise be missed or require manual calling on other constitutional cytogenetics array designs (Figures 1 and 2). An informed, sophisticated approach to array design has been used, with more probes being located in regions of the genome that are most likely to detect a biologically relevant aberration (Table 1). The addition of a research-validated collection of single nucleotide polymorphism (SNP) probes on the CytoSure Constitutional v3 +LOH array facilitates the precise identification of loss of heterozygosity (LOH) and uniparental disomy (UPD) in addition to accurate copy number (CN) detection.

Streamlined data analysis and interpretation

CytoSure Interpret Software, provided free of charge with all CytoSure arrays, is a powerful, easy-to-use package for the analysis of CNV and SNP data which includes a host of innovative features to enable the automation of data analysis workflows.

CytoSure Constitutional v3 arrays are available in a range of formats to match your resolution and throughput requirements. All CytoSure arrays have been research-validated using CytoSure Genomic DNA Labelling Kits.



REFERENCES

1. NCBI (2015) ClinGen Dosage Sensitivity Map [online] Available from: <http://ncbi.nlm.nih.gov/projects/dbvar/clingen> [Accessed 28 May 2015]

* Formerly known as ISCA/ICCG.

	Format	Cat. no.	Top priority genes	Medium priority genes	Lower priority genes	Decipher Syndrome regions	ClinGen regions	High priority backbone resolution	Medium priority backbone resolution	Low priority backbone resolution	LOH resolution
CytoSure Constitutional v3	8x60k	020045	Exon targeted	Whole-gene targeted	Whole-gene targeted	Whole-gene targeted	Whole-gene targeted	189kb	375kb	663kb	-
CytoSure Constitutional v3	4x180k	020046	Exon targeted	Exon targeted	Exon targeted	Whole-gene targeted	Whole-gene targeted	68kb	74kb	162kb	-
CytoSure Constitutional v3 +LOH	4x180k	020047	Exon targeted	Whole-gene targeted	Whole-gene targeted	Whole-gene targeted	Whole-gene targeted	68kb	74kb	162kb	7Mb and above

Table 1: Selection guide for CytoSure Constitutional v3 arrays. For a complete list of genes covered, please email: products@ogt.com

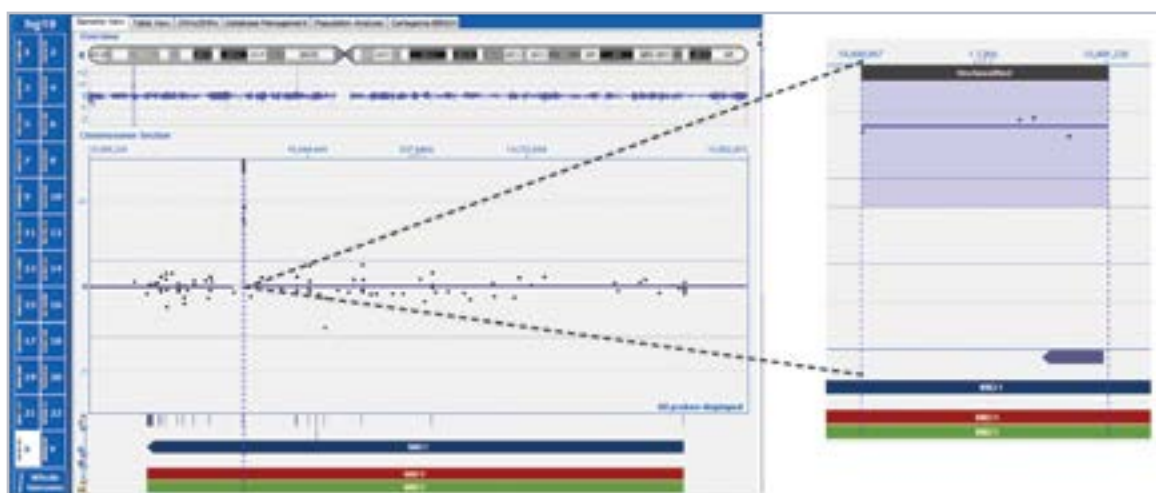


Figure 1: Accurate detection of a small, single-exon (<500bp; 4 probes) duplication in *MID1* associated with Opitz-G syndrome.*

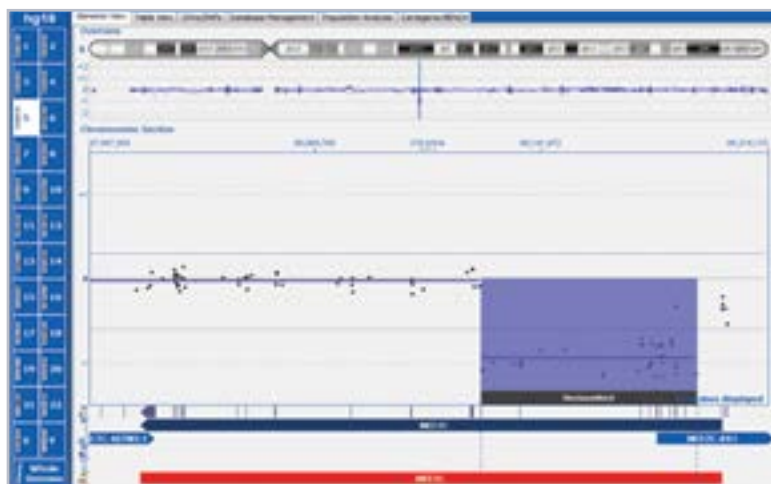


Figure 2: Enhanced probe coverage. A 68kb deletion covering *MEF2C* gene. The CytoSure Constitutional v3 8x60k array contains 36 probes in this region. This deletion was previously called manually on the CytoSure ISCA v2 8x60k array which has only 2 probes in this deletion.*

* Data kindly provided by West Midlands Regional Genetics Laboratory (WMRGL) Birmingham UK. Find out more at www.ogt.com/cytosure

CytoSure Medical Research Exome Array

Complete coverage of all medically relevant genes; ultra-high resolution, exon-focused CNV calling in inherited molecular disease

CytoSure Medical Research Exome array delivers:

- Highly targeted optimized probes — detect single or multiple exonic CNVs
- Medical research relevant content — over 4600 hand-curated, research-validated genes
- Optimized labeling kits and integrated sample tracking probes — for confident analysis and reporting
- Combine with NGS exome analysis — comprehensive mutation spectrum analysis in rare disease

The CytoSure Medical Research Exome Array is a highly targeted exon-focused array capable of detecting medically relevant microdeletions and microduplications. Developed in collaboration with leading molecular genetics experts at Emory University, this array covers the most medically relevant regions of the genome gathered from their research into molecular disorders. The Medical Research Exome Array makes an ideal complement to an exome sequencing approach to provide a comprehensive mutation spectrum analysis in rare disease.

Highly targeted optimized probes

All probes were tested *in silico* and scored on quality before the highest scoring probes were printed on an array and tested in the laboratory. Only the most accurate, best performing probes were used in the final design. Probes have been selected to target the exonic regions of 4,645 genes. For the majority of genes there are a minimum of 4 probes per exon. For very large exons, probes are distributed evenly along the exon with one probe every 125bp. For any array design, good backbone coverage is important to ensure accurate normalization. In the untargeted backbone, the CytoSure Medical Research Exome array has one probe every 42kb. Industry-leading coverage levels have been achieved with this design process — 88% of genes have 100% coverage, with 98% of genes having >75% coverage.

All probes in the Medical Research Exome Array can be used to create customized, high value, disease-specific arrays in multiplex formats (see page 186). Emory University has used this approach to create disease-specific arrays, each of which is also available as a catalog product (see pages 179-182).



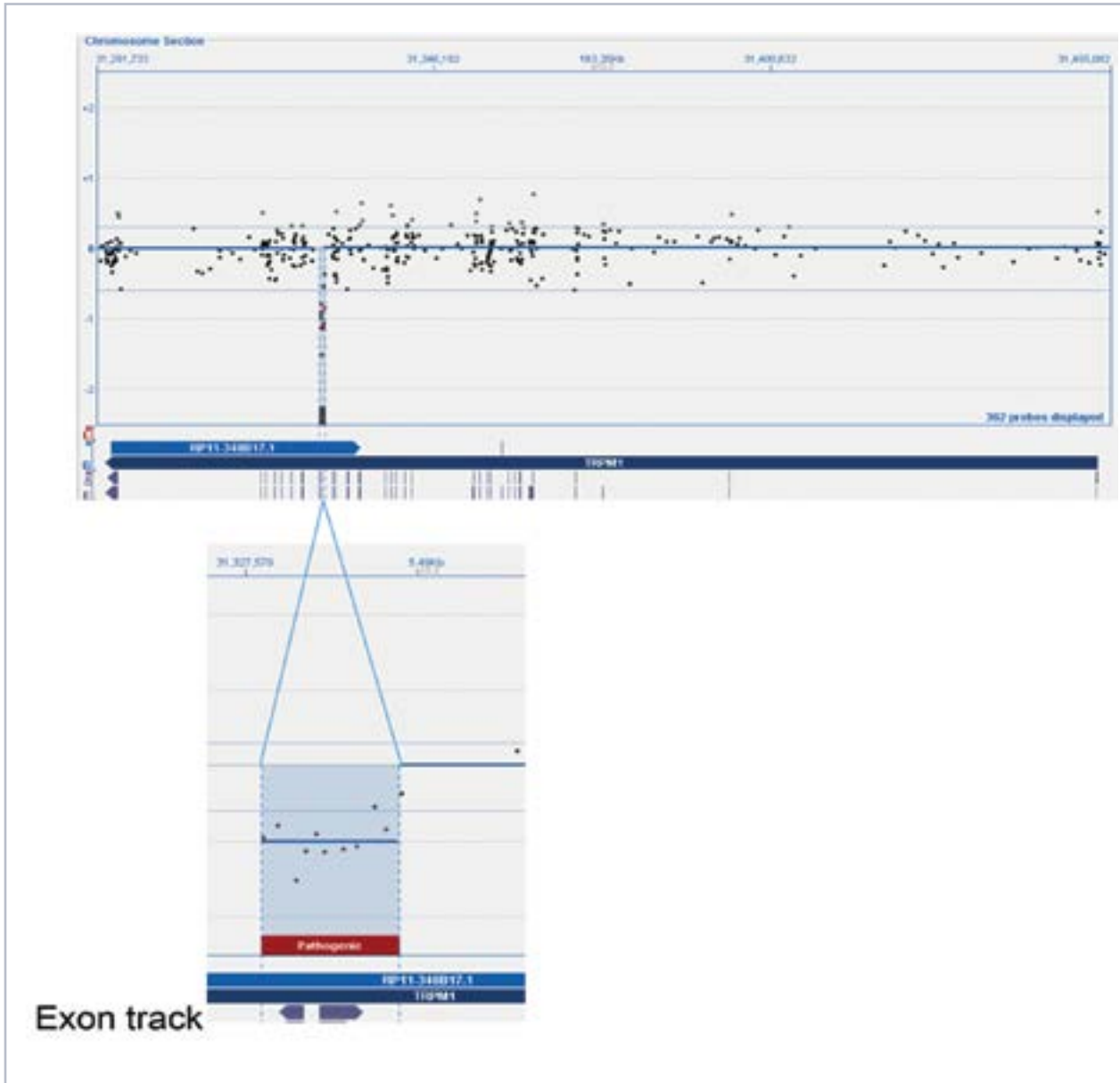


Figure 1: CytoSure™ Interpret Software clearly displays small aberrations and enables easy identification of genes and exons. Mutations in *TRPM1* may be associated with congenital stationary night blindness. Shown here in the top panel is an overview of the whole gene. In the bottom panel, the close-up view shows a very small 684bp deletion which contains 10 probes, and also spans a single exon*.

* Data kindly provided by Madhuri Hegde, Ph.D., FACMG, Emory University.

CytoSure

Disease-focused Arrays

Ultra-high resolution, exon-focused CNV calling for specific inherited molecular disease

CytoSure Disease-Focused arrays deliver:

- Accurate detection of copy number variation at the exon-level — a perfect complement to sequencing analysis
- Array content taken from the Medical Research Exome Array — fully optimized and research-validated by Emory University
- Multiplex (4x180k) format is cost-effective and allows for higher sample throughput
- Easy data interpretation using optimized protocols for high signal-to-noise ratios and industry-leading CytoSure Interpret Software

Array content fully optimized and research-validated

CytoSure disease-focused research arrays are designed to accurately identify small intragenic copy number variations (CNVs). They are exon-focused, high-resolution, 4x180k aCGH (array comparative genomic hybridization) array designs covering medically-relevant genes for research into specific disorders. The content for the disease-focused research arrays has been designed and optimized in collaboration with leading molecular genetics experts at Emory University.

For the best results, combine the CytoSure disease-focused arrays with the CytoSure Genomic DNA Labelling kits (page 189) and CytoSure Interpret Software (page 187).

CytoSure Autism Research Array

Autism spectrum disorders (ASD) affect 21.7 million people globally¹. CNV linked to ASD have been described at 11 loci across 8 chromosomes², hence understanding CNV status is critical for research into the genetic basis of this disease.

- 227 genes covered
- Diseases covered by the array:
 - Autism
 - Hearing loss
 - XLID (X-linked intellectual disability)



REFERENCES

1. Global Burden of Disease Study 2013 Collaborators (2015). Global, regional, and national incidence, prevalence, and years lived with disability for 301 acute and chronic diseases and injuries in 188 countries, 1990-2013: a systematic analysis for the Global Burden of Disease Study 2013. *Lancet* 386 (9995), 743-800
2. Menasha, I. *et al* (2013) Prioritization of Copy Number Variation Loci Associated with Autism from AutDB—An Integrative Multi-Study Genetic Database. *PLOS one* 8 (6), e66707

For research use only. Not for use in diagnostic procedures.

CytoSure Cardiomyopathy Research Array

CNVs are associated with a number of cardiomyopathies, including Long QT syndrome (LQTS)^{1,2} and dilated cardiomyopathy (DCM)³, so it is important to include CNV into any research. The array includes genes that cause genetic syndromes with cardiomyopathy as a feature (e.g. Duchenne/ Becker MD, Emery-Dreifuss MD).

- 223 genes covered
- Examples of diseases covered by the array:
 - Cardiomyopathies including LQTS (Long QT syndrome), DCM (dilated cardiomyopathy), LVNC (left ventricular non-compaction)
 - Hereditary neuropathies
 - Connective tissue disorders



REFERENCES

1. Eddy, C.A. *et al*. (2008) Identification of large gene deletions and duplications in KCNQ1 and KCNH2 in patients with long QT syndrome. *Heart Rhythm* 5, 1275-1281
2. Tester, D.J. *et al*. (2010) Prevalence and spectrum of large deletions or duplications in the major long QT syndrome-susceptibility genes and implications for long QT syndrome genetic testing. *Am J Cardiol* 106, 1124-1128
3. Norton, N. *et al*. (2011) Genome-wide studies of copy number variation and exome sequencing identify rare variants in bag 3 as a cause of dilated cardiomyopathy. *Am J Hum Genet.* 88, 273-82

For research use only. Not for use in diagnostic procedures.

CytoSure Epilepsy Research Array

While over 200 single-gene defects have been described in epilepsy¹, CNVs also play a key role in this disease. An important study identified 437 CNVs in 323/805 (40%) individuals with epilepsy (1–4 per patient) ranging from 18kb to 142Mb in size², many of which were associated with the disease.

- 212 genes covered
- Examples of diseases covered by the array:
 - Epilepsy
 - Brain malformations
 - SCID (severe combined immune deficiency)



REFERENCES

1. Kumar, D. ed. (2008) Genomics and clinical medicine. Oxford: Oxford University Press. p. 279.
2. Olson, H. *et al* (2014) Copy number variation plays an important role in clinical epilepsy. *Annals of Neurology* 75(6), 943–958

For research use only. Not for use in diagnostic procedures.

CytoSure Eye Disease Research Array

The CytoSure Eye Disease Research Array includes genes important for syndromic and non-syndromic inherited retinal and choroidal dystrophies, as well as ocular developmental disorders.

- 221 genes covered
- Examples of diseases covered by the array:
 - Retinitis pigmentosa
 - Stargardt disease
 - Congenital stationary night blindness
 - Usher syndrome



For research use only. Not for use in diagnostic procedures.

CytoSure NMD Research Array

It is estimated that around 16/10,000 of the population are affected by some form of neuromuscular disease (NMD)¹. The CytoSure NMD Research array is focused primarily on the muscular dystrophies. In the most common form of muscular dystrophy, Duchenne muscular dystrophy, between 60% and 75% of disease relevant mutations are CNVs².

- 205 genes covered
- Examples of diseases covered by the array:
 - DMD (Duchenne muscular dystrophy)
 - Limb girdle MD
 - CMD (Congenital muscular dystrophy)
 - Emery-Dreifuss MD
 - Congenital disorders of glycosylation
 - MODY (Maturity onset diabetes of the young)



REFERENCES

1. Deenen, J.C.W. *et al* (2015) The Epidemiology of Neuromuscular Disorders: A Comprehensive Overview of the Literature. *Journal of Neuromuscular Disease* 2(1) 73-85
2. Prior, T.W. and Bridgeman, S.J. (2005) Experience and Strategy for the Molecular Testing of Duchenne Muscular Dystrophy. *J Mol Diagn.* 7(3) 317-26

For research use only. Not for use in diagnostic procedures.

Other disease-focused research arrays

Other disease-focused arrays are also available including arrays focused on:

- Ciliopathies 4x180k
- Metabolic disorders 4x180k
- Skeletal dysplasia 4x180k
- Hereditary cancer 4x180k
- Duchenne muscular dystrophy (DMD) 8x60k

See www.ogt.com/CytoSure for more information.

For research use only. Not for use in diagnostic procedures.

CytoSure Cancer +SNP Arrays

Reliable detection of copy number changes and loss of heterozygosity (LOH) on a single array for hematological malignancies and solid tumors

CytoSure Cancer +SNP arrays deliver:

- Unique SNP probe technology allowing the use of any reference sample with no restriction digest
- Unparalleled performance through design optimization
- Fast and easy analysis using CytoSure Interpret™ software
- Versatile array designs across a choice of formats

Unique SNP Probe technology allows the use of any reference sample with no restriction digest

CytoSure Cancer +SNP arrays combine aCGH (array comparative genomic hybridization)-based CNV detection with fully research-validated SNP content allowing confident and cost-effective CNV and LOH identification using a single array (Figure 1). Due to the unique design of the SNP probes where an intensity-based comparison is made between the two SNP alleles there are no changes to the standard aCGH protocol, no restriction digest is required and any reference sample can be used. The ability to use matched reference samples (e.g. buccal swab tissue from the same individual) is particularly important when investigating aberrations in cancer as it enables constitutional abnormalities to be filtered out.

Improved results over other technologies

Array comparative genomic hybridization (aCGH) using 60-mer oligonucleotide probes has been shown to offer higher signal-to-noise ratios, increased sensitivity and increased specificity compared to other technologies¹.

With other platforms, the use of 60-mer technology for LOH analysis typically requires a restriction digest, which can compromise sample quality, limits the target SNPs to those overlapping restriction sites, and requires a genotyped reference for comparison. However, due to OGT's unique SNP technology, there is no restriction digest required, the most informative SNPs can be targeted and any reference sample can be used (e.g. normal tissue from the same individual to enable constitutional abnormalities to be filtered out).

Combined with the *in silico* and empirical optimization carried out across all OGT catalog arrays as well as easy customization to include any additional regions of interest, OGT's Cancer +SNP arrays deliver flexible and robust analysis of CNV and LOH combined in a single assay.



REFERENCES

1. Curtis, C. *et al* (2009) The pitfalls of platform comparison: DNA copy number array technologies assessed. *BMC Genomics* 10, 588-610
2. Hurles, M. *et al* (2010) Characterising and predicting haploinsufficiency in the human genome. *PLoS Genetics* 6, 10, e1001154 1-11.

Fast and easy analysis using CytoSure Interpret Software

OGT's CytoSure Interpret Software, which accompanies all CytoSure arrays, is a powerful and easy-to-use package for straightforward analysis of CNV and SNP data (Figure 1), delivering:

- Feature-rich, highly-customisable analysis workflows to meet any lab's requirements
- Automation of the data analysis processes, including batch upload of LIMS information to the database
- Extensive cancer-specific annotation tracks including regions from the Mitelman Database, the Cancer Gene Consensus Genes, the Atlas of Genetics and Cytogenetics in Oncology and Haematology and the Hurles Haploinsufficiency data².

For more information on complimentary CytoSure Interpret Software, see page 187.

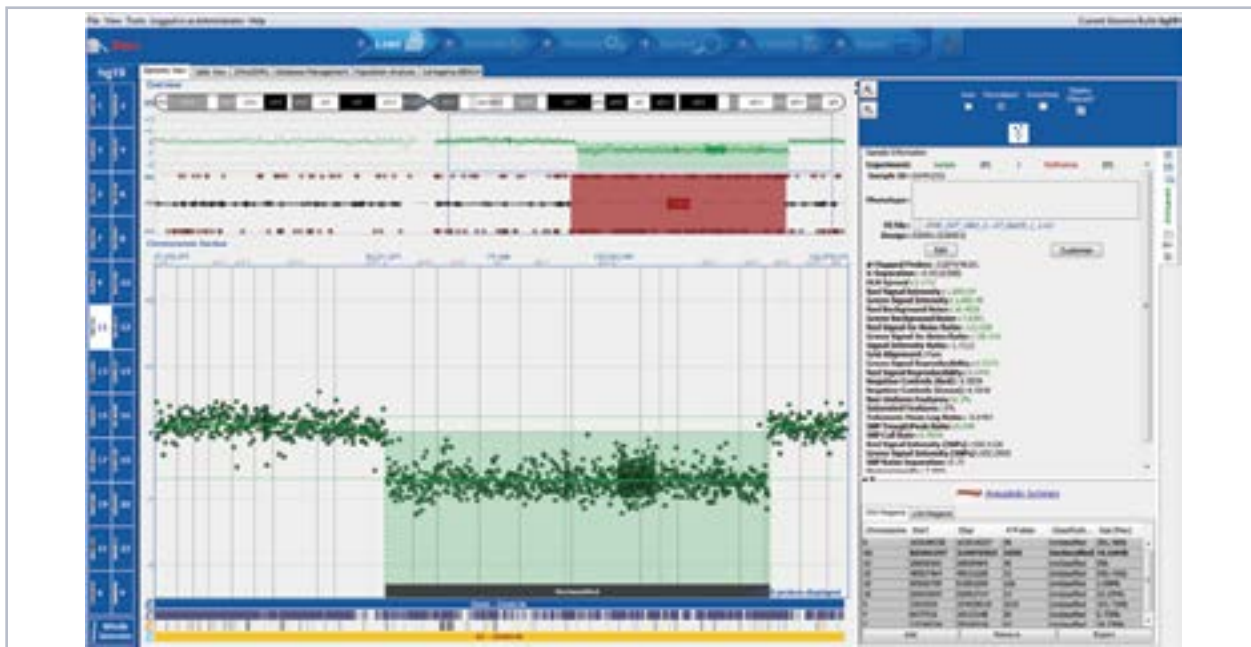


Figure 1: Shown here is a CLL research sample run on the CytoSure Consortium Cancer +SNP array (8x60k) with a deletion and corresponding LOH. CytoSure Interpret offers an intuitive user interface for easy interpretation of genetic findings. Samples kindly provided by Dr Jon Strefford, University of Southampton.

Versatile array designs across a choice of formats

Three fully customisable Cancer +SNP designs are available, designed using different formats to suit any analysis and throughput requirement.

Array	Copy number resolution		LOH resolution
	Backbone	Average gene resolution (Hg19)	
CytoSure Haematological Cancer +SNP (8x60k)	1 probe every 117kb	1 probe every 68kb	30Mb
CytoSure Cancer +SNP (4x180k)	1 probe every 44kb	1 probe every 25kb	20Mb
CytoSure Consortium Cancer +SNP (4x180k)	1 probe every 36kb	1 probe every 23kb	10Mb

Table 1: CytoSure Cancer +SNP arrays selection guide. For a complete list of genes covered by each array, email support@ogt.com

* Samples kindly provided by Dr Jon Strefford, University of Southampton.

CytoSure Hematological Cancer +SNP array (8x60k)

This design offers a balance between throughput and resolution, allowing investigation of large CNV and LOH in a cost-effective manner.

This array delivers:

- Whole-genome coverage for CNV and LOH analysis
- Enhanced resolution across regions relevant for research into CLL, MM, MPN and MDS (Chronic Lymphocytic leukemia, Multiple Myeloma, Myeloproliferative Neoplasms, Myelodysplastic Syndromes)

CytoSure Cancer +SNP array (4x180k)

This design, developed in collaboration with Prof. Jacqueline Schoumans (Head of the Cancer Cytogenetic Unit at Lausanne University Hospital), focusses on CNV detection across the target regions.

This array delivers:

- Whole-genome coverage for CNV and LOH analysis
- Whole-gene CNV resolution across more than 1500 cancer-associated genes
- Exon resolution across 18 genes

<i>CDKN2A</i>	<i>IKZF1</i>	<i>NRAS</i>
<i>CDKN2B</i>	<i>IK2F2</i>	<i>PAX5</i>
<i>CEBPA</i>	<i>JAK2</i>	<i>RB1</i>
<i>EBF1</i>	<i>KIT</i>	<i>RUNX1</i>
<i>ETV6</i>	<i>MPL</i>	<i>TET2</i>
<i>FLT3</i>	<i>NF1</i>	<i>WT1</i>

Table 2: Genes covered at single-exon resolution on the CytoSure Cancer +SNP array

CytoSure Consortium Cancer +SNP array (4x180k)

This design focuses on the content recommended by the Cancer Cytogenetics Microarray Consortium (CCMC) now known as the Cancer Genomics Consortium (CGC), with more probes dedicated to SNP analysis than the other arrays. The recommended content is intended to help standardize research across cancer genomics, similar to the successful model introduced by ISCA/ICCG, now known as ClinGen.

This array delivers:

- Whole-genome coverage for CNV and LOH analysis
- Enhanced coverage of 130 cancer-associated genomic regions
- Whole-gene CNV resolution of more than 500 cancer-associated genes

CytoSure Custom Designed aCGH Arrays

Focused custom aCGH arrays designed to your specification by the microarray experts

Custom arrays deliver:

- Complete confidence in the design of your array
- Flexible array content and format
- Customization of any existing catalog array
- Full custom designs including probes from our existing designs (e.g. Medical Research Exome Array), from our proprietary Oligome™ database, or new designs using our superior in-house design pipeline

High-quality aCGH arrays, perfectly matched to your exact specifications

CytoSure Custom arrays allow you to benefit from OGT's extensive array design expertise to produce an array matching your precise specifications. These arrays are ideal if you want to know the precise coordinates of an aberration by analysing specific areas of the genome at high resolution.

Complete confidence in the design of your array

OGT have designed hundreds of custom arrays for some of the world's leading researchers. The array content is selected from OGT's proprietary Oligome database — a database of more than 26.5 million oligonucleotide probes, or can be *de novo* designed using the proprietary OGT probe design pipeline. All *de novo* probes are in-silico optimized and optional empirical validation of the array content ensures optimal performance. A dedicated project manager from our experienced team of bioinformaticians is assigned to each new custom-design project. This gives a single point of contact throughout the process and ensures a close collaboration with our experts from initial consultation to delivery of the final design.

Flexible array content and format

CytoSure Custom arrays can be designed against any fully or partially sequenced genome as well as against sequencing data. In addition, OGT has extensively research-validated SNP content for detection of loss of heterozygosity (LOH) and uniparental disomy, which can be incorporated into the array design. CytoSure Custom arrays can be designed in a variety of formats depending on your desired level of focus, with 1, 2, 4, or 8 arrays available per slide to provide the most cost-effective solution for your research (Figure 1).

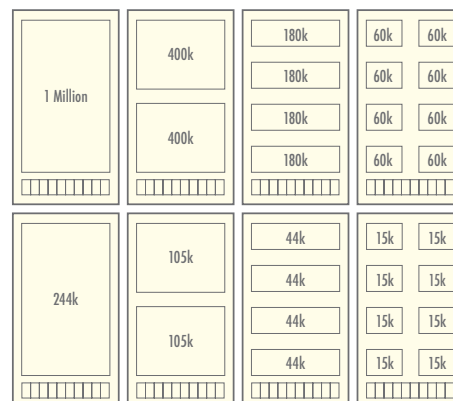


Figure 1: Multiple arrays on a single slide can reduce costs and improve efficiency.

CytoSure™ Interpret Software

A powerful and easy-to-use package for analysis of aCGH (array comparative genomic hybridization) data with multiple features allowing standardized data analysis (using Accelerate Workflow) or customized, user-defined analysis.



CytoSure Interpret Software delivers:

- Fast, accurate and simple analysis of aCGH data
- Comprehensive data annotation with direct links to external databases and online resources
- A robust relational database allowing sophisticated data querying and filtering
- Extensive customization options
- Fully integrated, automatic analysis of array image files

Effortless translation of oligo aCGH into meaningful results

The “Accelerate Workflow” provides automated data analysis based on predefined settings. This unique feature minimize user intervention and maximizes the consistency and speed of analysis. Batch processing allows an unlimited number of samples to be analysed simultaneously with the Circular Binary Segmentation (CBS) algorithm (Figure 1). Regions of LOH are analysed with our proprietary SNP calling algorithm (Figure 2).

Direct links to external databases and online resources

CytoSure Interpret Software includes extensive annotation tracks covering syndromes, genes, exons, CNVs and segmental duplication — linked to publicly available databases such as ISCA, Decipher, Database of Genomic Variants and the Cancer Gene Census (Figure 3) providing results in context. Each track can reference hg18, hg19 or hg38 information.

Sophisticated data querying and filtering

The powerful relational database enables storage of sample data according to its relationship with other data and back-ups are straightforward with the choice of full, partial or mini back up. The database is customisable with a choice of management systems designed to integrate with your current IT infrastructure. The unique “Family Tree” viewer allows probands to be linked to other family members to view aberrations across three generations.

Designed to meet the needs of your laboratory

Complete flexibility to optimize data analysis settings and customize data reports. The permission-based log-on structure enables greater flexibility for management of user accounts.

Compatible with a variety of microarray scanners

The CytoSure Interpret Feature Extraction Module allows analysis of TIFF images from a variety of microarray scanners. The module comes pre-loaded with template files enabling images to be feature-extracted and seamlessly loaded into the Accelerate Workflow without the need for user intervention.

For research use only. Not for use in diagnostic procedures.

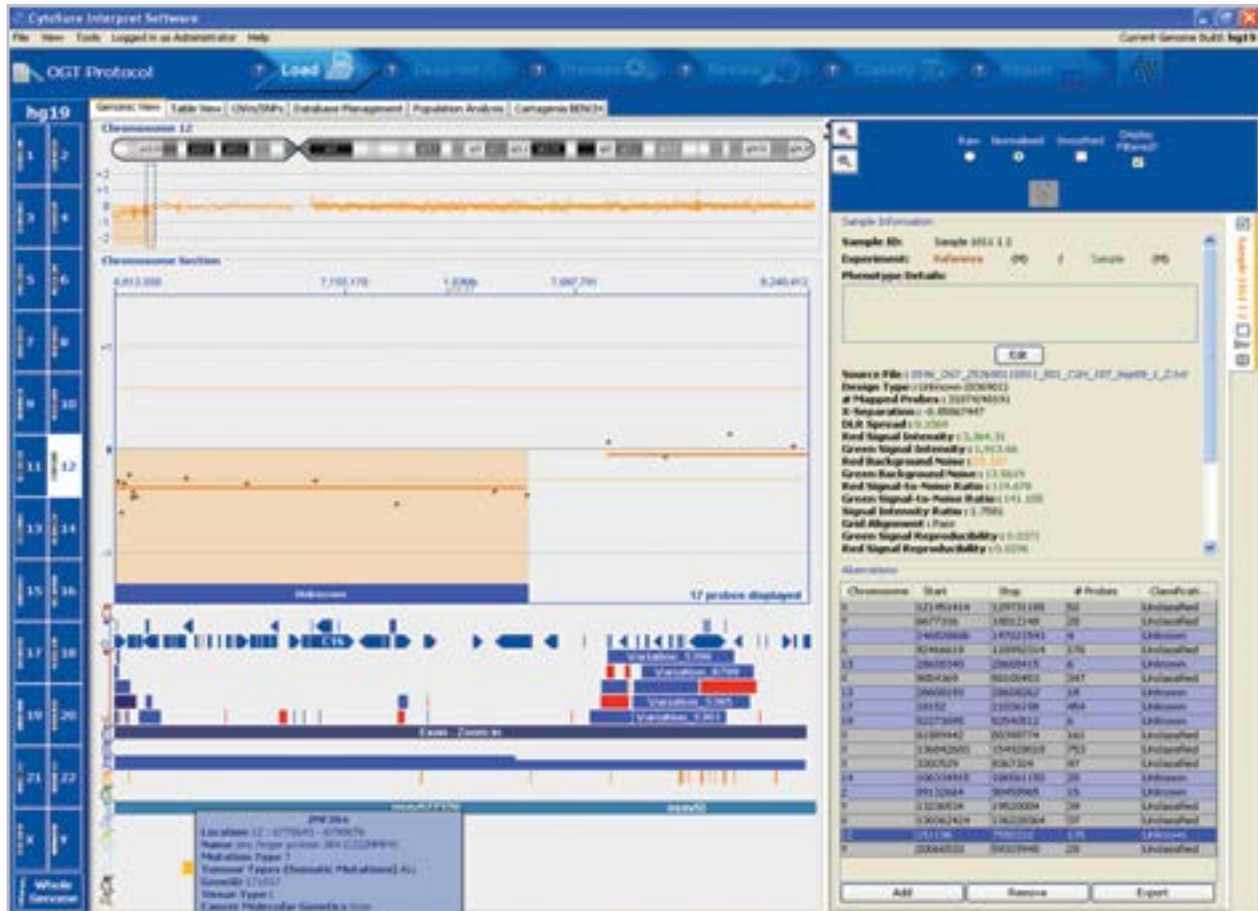


Figure 1: Automated aberration detection with CytoSure Interpret Software, showing clear detection of chromosomal abnormalities. The gain on chromosome 12 for this chronic lymphocytic leukaemia (CLL) sample contains the zinc finger protein gene ZP384, easily identified in the Cancer Gene Census genes track.



Figure 2: Automated SNP detection and LOH calling with CytoSure Interpret Software. The dark red rectangles indicate regions of LOH. The green and the bright red rectangles indicate amplifications and deletions respectively. This is the same CLL sample as displayed in Figure 1 and clearly illustrates the gain in the telomere region of the p arm and the region of LOH in p13.31-p12.3

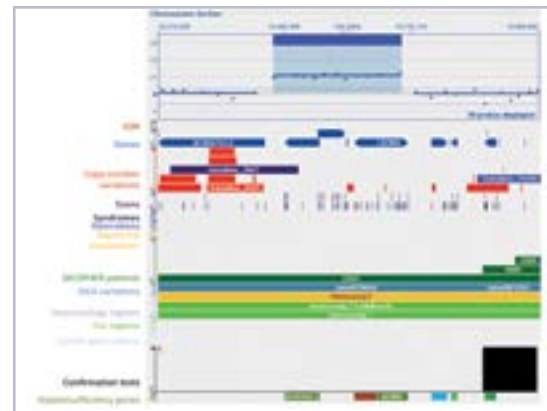


Figure 3: Fully customisable tracks simplify interpretation of aberrations.

CytoSure Genomic DNA Labelling Kits

Efficient and reproducible labeling of DNA samples for use in aCGH

CytoSure Genomic DNA Labelling Kits deliver:

- Optimized formats to suit your throughput requirements
- Reliable high-quality results through higher signal intensity
- A fast and simple procedure

Everything you need, from reagents to plasticware

The labeling of DNA samples used in array comparative genomic hybridization (aCGH) is a critical step in the experimental process as poor labeling can result in inaccurate data. As part of a complete labeling solution — protocols, reagents, clean-up plates or columns and collection tubes — OGT's CytoSure Genomic DNA Labelling Kits have been uniquely developed and optimized to enable rapid delivery of high-quality results with high signal-to-noise ratios.

Tested with a wide range of sample types to ensure optimal performance

Offering reliable, high-quality results, the CytoSure Genomic DNA Labelling Kits ensure superior signal-to-noise ratios for confident detection of copy number variation. This high signal-to-noise ratio means that even small aberrations can be reliably detected (Figure 1).

CytoSure Genomic DNA Labelling Kits offer much faster DNA labeling and clean up than traditional enzymatic labeling procedures. Labeling reactions using both the 24 and 96 reaction kits can easily be completed in a single day (Figure 2). The procedure can also be automated for implementation in high-throughput workflows.



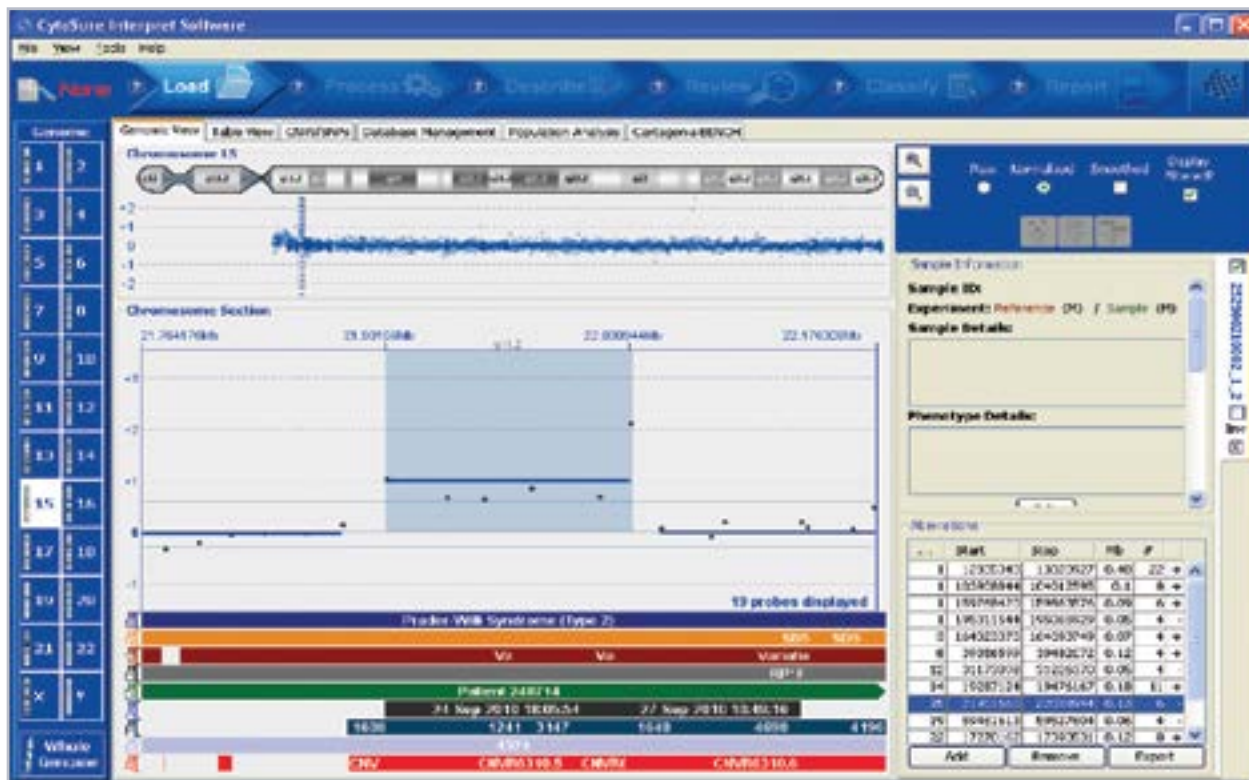


Figure 1: Reliable detection of small aberrations. DNA labelled using the CytoSure Genomic DNA Labelling Kit was run on a CytoSure ISCA 8x60K array. CytoSure Interpret Software combined with high DNA signal intensity allowed detection of a small (130 kb) DNA amplification.

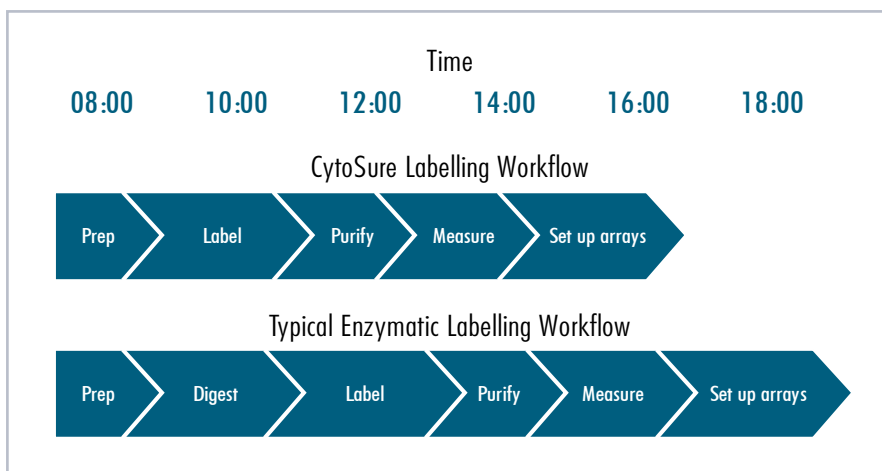


Figure 2: Two typical labeling workflows: With no need to digest, CytoSure Genomic DNA Labelling Kits save you at least 2 hours.

Product	Contents	Cat. No.
CytoSure Genomic DNA Labelling Kit	24 reactions: clean-up columns, dyes, nucleotide mix, random primers, enzyme, collection tubes	020020
CytoSure HT Genomic DNA Labelling Kit	96 reactions: 2 purification plates, nucleotide mix, random primers, enzyme	500040

CytoSure Sample Tracking Spike-ins

Reliable sample identity tracking for use with CytoSure arrays

CytoSure Sample Tracking Spike-ins deliver:

- Confidence in results
- Simple one-step procedure with no alteration to existing workflows
- Easy identification of sample mix-up

Complete confidence in results

Increasing numbers of aCGH (array comparative genomic hybridization) samples combined with higher-throughput array formats means that it is imperative to track samples throughout the labeling, hybridization and analysis process to maintain sample identity. CytoSure Sample Tracking Spike-ins are uniquely designed to enable reliable sample tracking and easy identification of sample mix-up using OGT's class-leading CytoSure Arrays and CytoSure Interpret Software.

Each CytoSure Sample Tracking Spike-in is designed to a specific, unique region of the genome. Oligonucleotide probes complementary to the sample tracking spike-ins are included on all of the arrays supplied and optimized by OGT. Eight different CytoSure Sample Tracking Spike-ins are available. Each spike-in has been carefully prepared to ensure that there is no cross-hybridization with other probes on the array or with any other region on the genome. In addition, colour-coded caps are used for ease of identification, aiding correct usage.



Accessories

Product	Description	Cat. No.
Oligo aCGH/ChIP-on-Chip Hybridization Kit	Hybridization reagents for 100 samples	500013
	Hybridization reagents for 25 samples	500014
DNA clean up plate	96 well plate for the clean up of DNA	500041
DNA clean up columns	24 columns for the clean up of DNA	500020
Wash Buffer 1 & 2 set	Buffers for post hybridization washing of arrays – 3x 4L	500015
Backing plate (gaskets)	Backing for 8x arrays	500010
	Backing for 4x arrays	500011
	Backing for 2x arrays	500012
	Backing for 1x arrays	500017
COT Human DNA (250µl)	Blocking reagent to prevent non-specific hybridization	500025
Human Genomic DNA, Male (100µg)	Reference DNA	500026
Human Genomic DNA, Female (100µg)	Reference DNA	500027



Educational Resources

Contents

FISH Probes

- 195 FISH glossary: Chromosome basics, guide to chromomaps and common terms explained
- 200 Recommended FISH protocol for AML/MDS FDA-cleared IVD Cytocell FISH probes
- 202 AML/MDS FDA-cleared IVD Cytocell FISH Probes: Guide to expected abnormal clinical results

Next Generation Sequencing

- 206 Choosing the best enrichment assay
- 208 One-day hybridization protocol
- 211 Comprehensive Results from a single NGS assay: Panel and Software highlights

Arrays

- 212 Basics of array aCGH
- 214 Troubleshooting for arrays

Overview

Oxford Gene Technology is committed to providing comprehensive support services and resources for our products.

This new section of the catalog is a small selection of technical resources to provide introductions, guidance for product selection, protocol overviews, and troubleshooting for your workflows.

Please read on for handy references and proven tips for FISH, NGS and arrays. Visit www.ogt.com/support for the full offering of content from our in-house experts and customers. We have application notes, posters, protocols, instructional videos and regularly updated feature pages to share the most recent advancements in the field.

www.ogt.com/support

FISH Glossary

Chromosome basics, guide to chromomaps and common terms explained

Regulatory Abbreviations

ASR	Analyte Specific Reagent. ASRs are “building blocks” of LDTs. [21 CFR 864.4020]
CGMP	Current Good Manufacturing Practice. See also QSR. [21 CFR 820]
CLIA	Clinical Laboratory Improvement Amendments of 1988 (CLIA '88, 42 USC 263a). Clinical laboratory testing (except research) in the U.S. is regulated by CMS via the CLIA Regulations. [42 CFR 493]
CMS	Centers for Medicare and Medicaid Services.
FDA	Food and Drug Administration.
GPR	General Purpose Reagent. [21 CFR 864.4010]
IFU	Instructions For Use. May also be referred to as a package insert.
IVD	<i>In Vitro</i> Diagnostic Medical Device. [21 CFR 809.3]
LDT	Laboratory Developed Test. Sometimes called a “home brew” test or an “in-house” test. Most LDTs in the U.S. are regulated under the CLIA Regulations.
QSR	Quality System Regulation. See also CGMP. [21 CFR 820]
RUO	For Research Use Only. Not for use in diagnostic procedures.
UDI	Unique Device Identification/Identifier. [78 FR 58785, 21 CFR 830]

Chromosome Basics: Glossary of terms

Nucleus	An organelle found in eukaryotic cells that contains genetic material, deoxyribonucleic acid (DNA).
DNA (Deoxyribonucleic acid)	A self-replicating material which encodes hereditary information present in nearly all living organisms as the main constituent of chromosomes. DNA encodes hereditary genetic information using four nucleotides; adenine, thymine, cytosine and guanine.
Chromosome	A condensed form of the highly organised structure of nucleic acids and proteins found in the nucleus of most living cells, carrying genetic information in the form of genes. Humans have 23 pairs of chromosomes.
Chromatid	One of the two threadlike strands (sister chromatids) into which a chromosome divides longitudinally during cell division.
p arm	The short arm of the chromosome (with the exception of a metacentric chromosome which has chromosome arms of equal length).
q arm	The long arm of the chromosome (with the exception of a metacentric chromosome which has chromosome arms of equal length).
Centromere	A primary constriction which separates the p arm from the q arm and holds the pair of sister chromatids together.
Telomere	A compound structure at the end(s) of a chromosome, consisting of repetitive nucleotide sequences.
Gene	A unit of heredity which is transferred from a parent to offspring and is known to determine some characteristics of the offspring. A gene consists of a sequence of nucleotides in DNA that encode the synthesis of a gene product, such as a protein or RNA molecule.

Anatomy of a chromosome

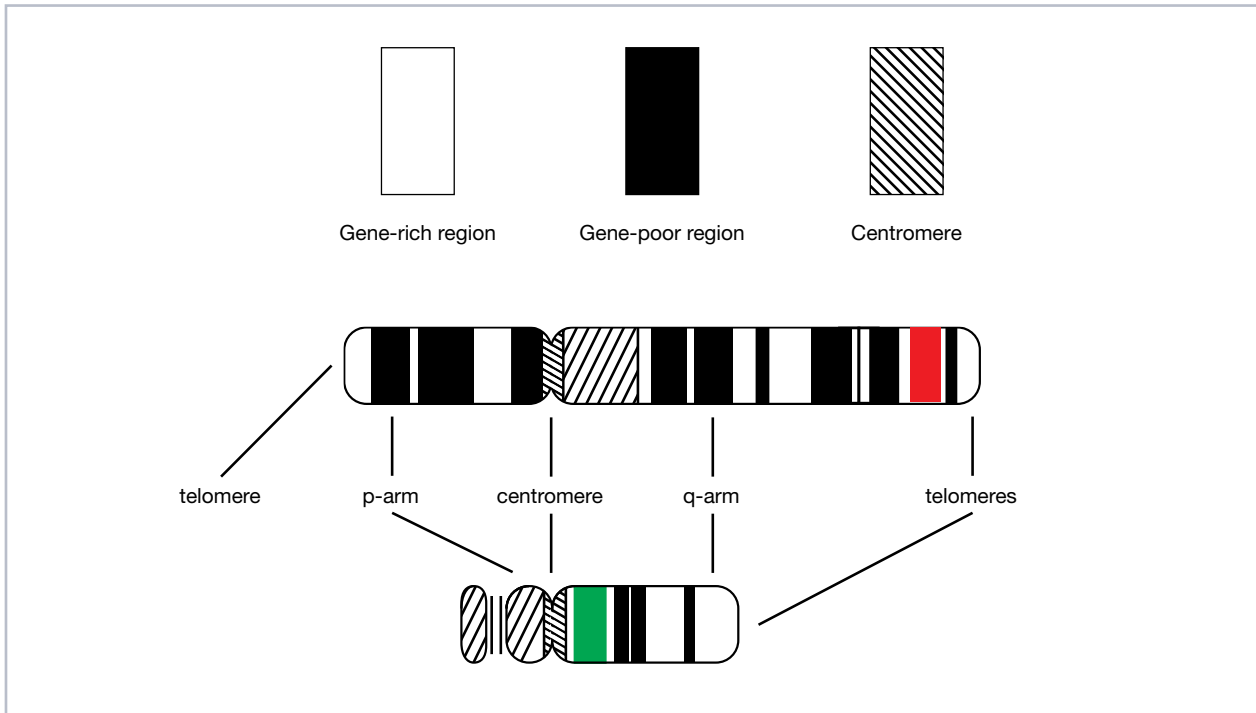


Figure 1: Characteristic banding pattern produced by exposure to trypsin during G banding. G band light regions tend to be gene rich. G band dark regions tend to be gene poor. Giemsa banding is a technique used in cytogenetics to produce a visible karyotype by staining condensed chromosomes

Types of chromosome

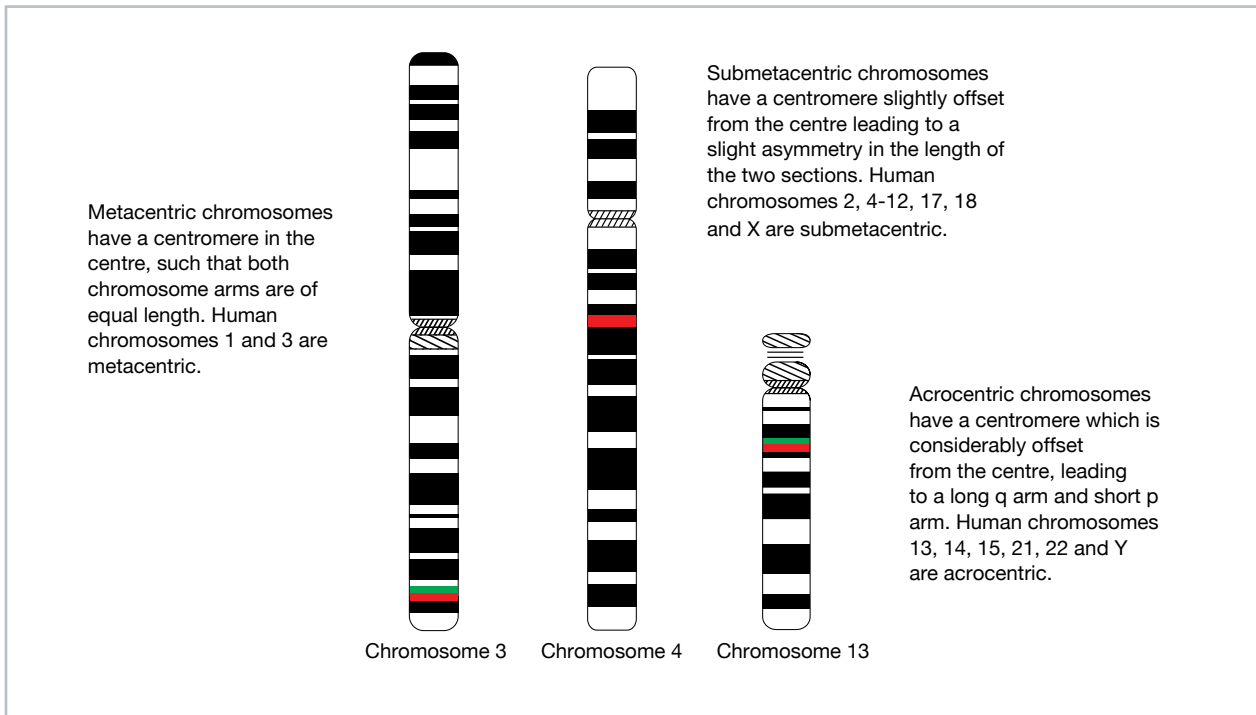


Figure 2: Metacentric (left), submetacentric (center) and acrocentric (right) chromosomes

Probe maps and gene orientation

Probe maps in the OGT Product Catalog, Second Edition, have been updated to include information on gene orientation to allow the 3' or 5' positions of the probes to be seen in relation to the gene, or region, of interest. The arrows on the genes indicate the direction of transcription. Those with an arrow above, pointing to the right, are located on the plus, or sense, strand of the DNA. Those with an arrow below the gene, pointing to the left, are on the minus, antisense strand. In both cases, the 5' end is that with the arrow as genes are transcribed in a 5' to 3' direction. Examples can be seen below.

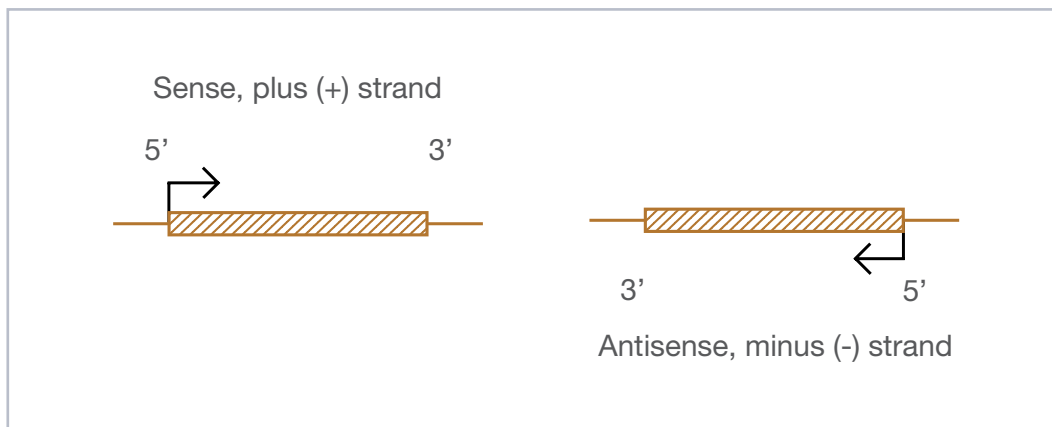


Figure 3: Arrows on genes in chromomaps indicate the direction of transcription.

HGNC Nomenclature

Gene names have also been updated to reflect current HUGO Gene Nomenclature Committee (HGNC) approved symbols. Where this affects existing product names, the approved HGNC symbol is placed into brackets. All gene names were checked and updated according to the HGNC database¹ as of April 2019.

1. HGNC Database, HUGO Gene Nomenclature Committee (HGNC), EMBL Outstation - Hinxton, European Bioinformatics Institute, Wellcome Trust Genome Campus, Hinxton, Cambridgeshire, CB10 1SD, UK. www.genenames.org

Types of probes

In theory, any region of a chromosome can be a target for a FISH probe. Cytocell probes may label chromosomes anywhere along the p or q arm: the subtelomere, the centromere, or any specific gene region in between.

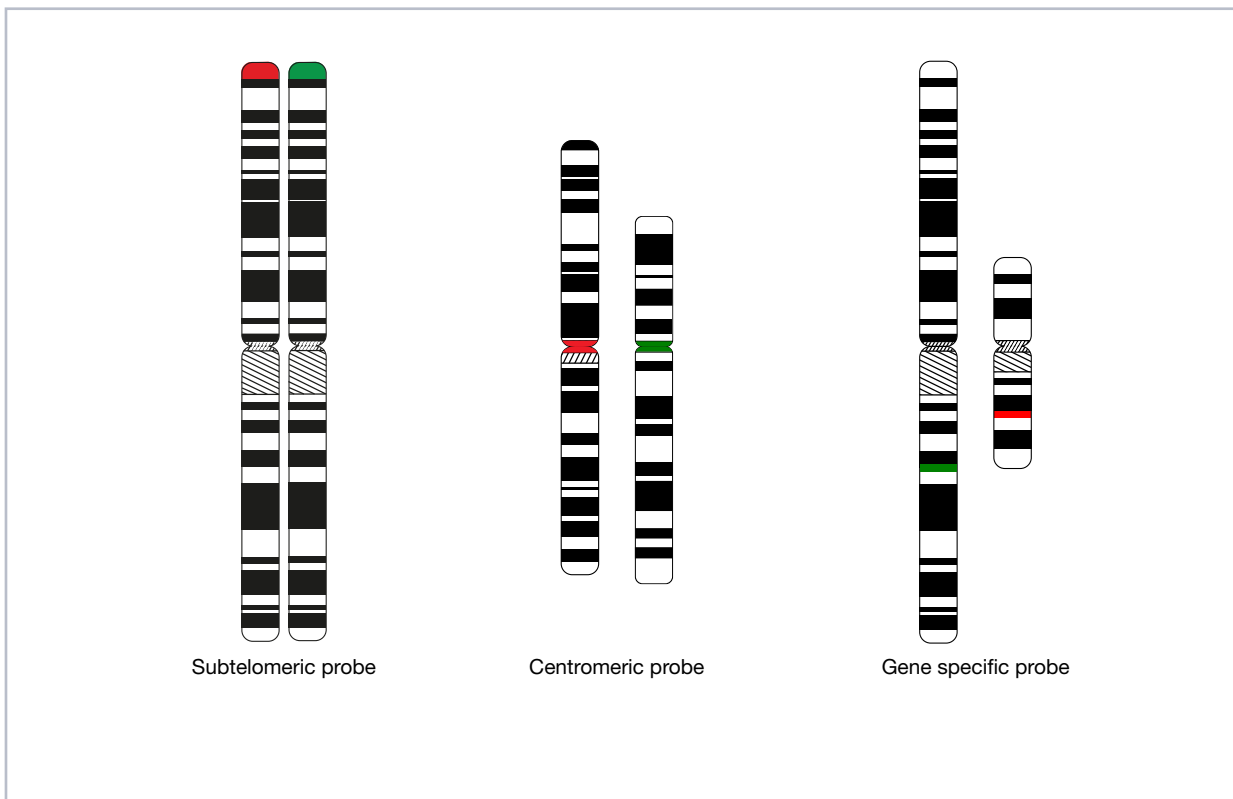
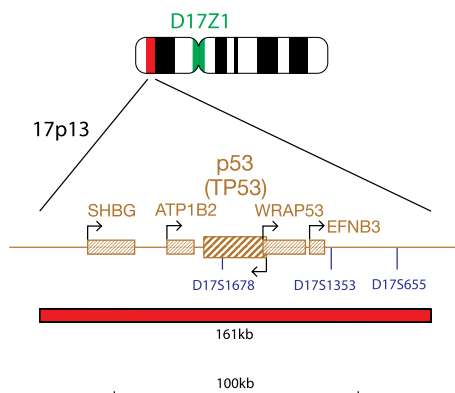


Figure 4: Illustration of Cytocell subtelomeric, centromeric and gene specific FISH probes.

Probe design

An example of the convention at OGT for assignment of a fluorophore can be viewed with the FDA-cleared probe for AML/MDS: P53 (TP53) Deletion (USA-LPH 017):*

- Green = control
- Red = target region of interest



* There are some exceptions to this convention, please consult the IFU for probe map and full details.

Probe nomenclature explained

Using the MLL (KMT2A) Breakapart probe (USA-LPH 013) probe as an example:

MLL (*KMT2A*) Breakapart

Breakapart = Probe design / signal pattern, one fluorophore for 3' end and a different color fluorophore for 5' end

Guide to Chromosome Region

11q23.3 =

Chromosome 11 / q arm / Band 23 / Sub-band 3

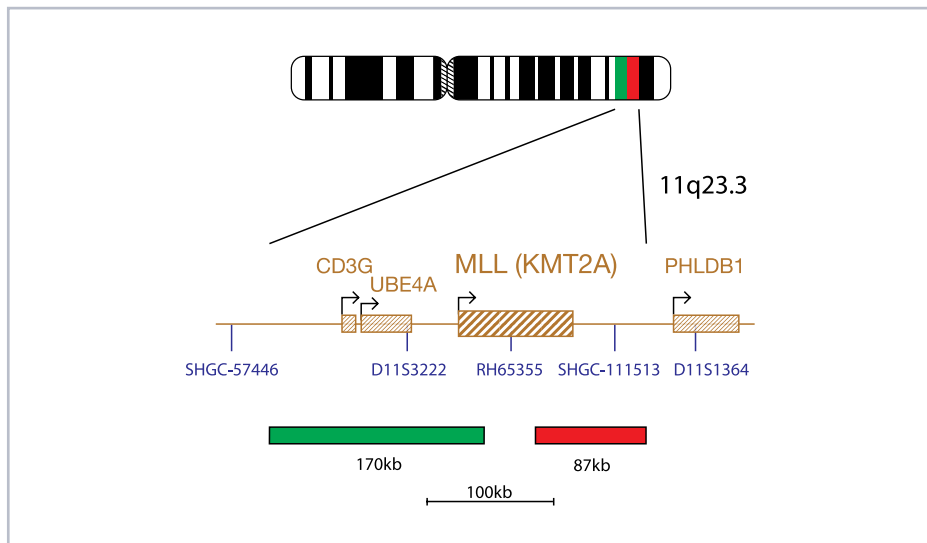


Figure 5: Cytocell chromomap for MLL (KMT2A) Breakapart FISH Probe Kit (US only).

Cytocell catalog product numbers explained:

- LPH** Hematology probes.
- LPS** Solid tumour and hematopathology probes.
- LPD** Dual use probes.
- LPE** Chromosome enumeration probes.
- LPU** Microdeletion probes.
- LPT** Subtelomeric probes.



Recommended FISH protocol for Cytocell Aquarius® AML and MDS FISH Probe Kits



This protocol is provided for use with the FDA-cleared range of Cytocell Aquarius FISH probe kits for acute myeloid leukemia (AML) or myelodysplastic syndrome (MDS). Reference the Package Insert (Instructions for Use (IFU)) for warnings, precautions, storage, and handling. Study the Package Insert carefully before using this quick reference guide.

This can be found in the product packaging and by using the Resources section of the Cytocell website (www.cytocell-us.com/resources).

Note: This quick reference guide does not replace the content from the Package Insert. For more information on FDA-cleared FISH probe test kits for AML and MDS, see the hematology section.

Overview







The Cytocell Aquarius AML/MDS range of FISH probe test kits are used to detect common chromosomal rearrangements in fixed bone marrow specimens from patients with AML or MDS. The tests are indicated for the characterization of patient specimens consistent with World Health Organization guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are to be interpreted by a qualified pathologist or cytogeneticist. The tests are not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic.

*Refer to individual test kit Package Insert for the specific intended use and limitations.
For In Vitro Diagnostic Use. Rx only.*

Materials Provided

The FISH probes are provided in a 100µl (10 test) per vial, ready-to-use format and premixed with hybridization solution (formamide; dextran sulfate; saline-sodium citrate (SSC)). In addition a 150µl vial of ready-to-use DAPI counterstain with antifade (0.125µg/ml DAPI (4,6-diamidino-2-phenylindole)) is provided. See Package Insert for additional details.

Recommended FISH protocol for Cytocell AML and MDS FISH Probe Kits

Step 1		Sample and slide preparation	<ul style="list-style-type: none"> • The FISH probes for AML/MDS are designed for use on bone marrow cells fixed in Carnoy's solution (3:1 methanol/ acetic acid) that are prepared according to the laboratory or institution guidelines. • Spot the cell sample onto a glass microscope slide. Allow to dry. • Immerse the slide in 2x Saline Sodium Citrate (SSC) for 2 minutes at room temperature (RT) without agitation. • Dehydrate in an ethanol series (70%, 85% and 100%), each for 2 minutes at RT. • Allow to dry.
Step 2		Pre-denaturation	<ul style="list-style-type: none"> • Remove the probe from the freezer and allow it to warm to RT. Briefly centrifuge tubes before use. • Ensure that the probe solution is sufficiently mixed with a pipette or a vortex mixer. • Remove 10µl of probe per test, and transfer it to a microcentrifuge tube. Quickly return the remaining probe to -20°C. • Place the probe and the sample slide to prewarm on a 37°C (+/- 1°C) hotplate for 5 minutes. • Spot 10µl of probe mixture onto the cell sample and carefully apply a 24x24mm coverslip. Seal with rubber solution glue and allow the glue to dry completely.
Step 3		Denaturation	<ul style="list-style-type: none"> • Denature the sample and probe simultaneously by heating the slide on a hotplate at 75°C (+/- 1°C) for 2 minutes.
Step 4		Hybridization	<ul style="list-style-type: none"> • Place the slide in a humid, lightproof container at 37°C (+/- 1°C) overnight.
Step 5		Post-hybridization washes	<ul style="list-style-type: none"> • Remove the DAPI from the freezer and allow it to warm to RT. • Remove the coverslip and all traces of glue carefully. • Immerse the slide in 0.4x Saline Sodium Citrate (SSC) (pH 7.0) at 72°C (+/- 1°C) for 2 minutes without agitation. • Drain the slide and immerse it in 2xSSC + 0.05% Tween-20 at RT (pH 7.0) for 30 seconds without agitation. • Drain the slide and apply 10µl of DAPI antifade onto each sample. • Cover with a 24x24mm coverslip, remove any bubbles. • Edge the slide with clear nail varnish to seal. • Allow the color to develop in the dark for 10 minutes.
Step 6		Analyze	<ul style="list-style-type: none"> • View with a fluorescence microscope. • For optimal visualization of the probes, a 100-Watt mercury lamp (or equivalent) is recommended with plan apochromat objectives 63x or 100x. • Filters designed specifically for detection of DAPI, FITC, Texas Red®, and Aqua or DEAC fluorophores individually or in combination (e.g. dual or triple filters) are optimal for best results. • The final hybridized slides are analyzable for up to 1 month when stored in darkness and at 2-8°C.

Please refer to the Cytocell FDA cleared test kit IFU for more detailed information. This can be found in the product packaging and using the Resources section of the Cytocell website (<https://www.cytocell-us.com/resources>).

For *In Vitro* Diagnostic Use. Rx only. Product availability may vary from country to country and is subject to varying regulatory requirements.

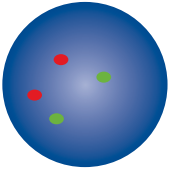
Cytocell® FISH Probes for AML and MDS

Guide to expected abnormal clinical results

AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual Fusion FISH Probe Kit AML

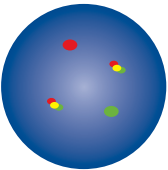
Cat. No. USA LPH-026

Expected normal signal pattern



In a normal cell, two red and two green signals (2R, 2G) are expected.

Expected abnormal signal pattern



In a cell with a t(8;21)(q22;q22) translocation the expected signal pattern will be one red, one green and two fusions (1R, 1G, 2F).

Other signal patterns are possible in aneuploid/unbalanced specimens.

Characterization of Normal Cut-off Values*

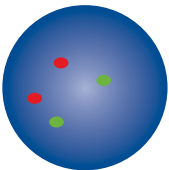
Abnormal signal pattern	Number of samples analyzed to generate the cut-off	Number of nuclei evaluated per sample	Maximum number of false positive signal pattern	Normal cut-off value (per 200 nuclei)	Normal cut-off value (%)
1R, 1G, 2F	1290	200	1	5	2.3

*The device has not been specifically validated in patients with <20% blast count.

CBFB (CBFB)/MYH11 Translocation, Dual Fusion FISH Probe Kit AML

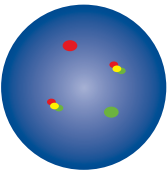
Cat. No. USA LPH-022

Expected normal signal pattern



In a normal cell, two red and two green signals (2R, 2G) are expected.

Expected abnormal signal pattern



In a cell with an inv(16)(p13q22) or a t(16;16)(p13;q22) the expected signal pattern will be one red, one green and two fusions (1R, 1G, 2F).

Other signal patterns are possible in aneuploid/unbalanced specimens.

Characterization of Normal Cut-off Values*

Abnormal signal pattern	Number of samples analyzed to generate the cut-off	Number of nuclei evaluated per sample	Maximum number of false positive signal pattern	Normal cut-off value (per 200 nuclei)	Normal cut-off value (%)
1R, 1G, 2F	1300	200	1	5	2.3

*The device has not been specifically validated in patients with <20% blast count.

The Cytocell Aquarius AML/MDS range of FISH probe test kits are fluorescence *in situ* hybridization (FISH) tests used to detect common chromosomal rearrangements in fixed bone marrow specimens from patients with acute myeloid leukemia (AML) or myelodysplastic syndrome (MDS). The tests are indicated for the characterization of patient specimens consistent with World Health Organization guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are to be interpreted by a qualified pathologist or cytogeneticist. The tests are not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic. Reporting and interpretation of FISH results should be consistent with professional standards of practice and should take into consideration other clinical and diagnostic information. This kit is intended as an adjunct to other diagnostic laboratory tests and therapeutic action should not be initiated on the basis of the FISH result alone. Failure to adhere to the protocol may affect the performance and lead to false results. Each lab is responsible for establishing their own cut-off values. Each laboratory should test sufficiently large number of samples to establish normal population distribution of the signal levels and to assign a cut-off value. The product is for professional use only and is intended to be interpreted by a qualified Pathologist or Cytogeneticist. Refer to individual test kit Package Insert for the specific intended use and limitations. For *In Vitro* Diagnostic Use. Rx only. Product availability may vary from country to country and is subject to varying regulatory requirements. Please contact your local representatives for availability.



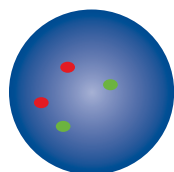
Continued on next page

Del(5q) Deletion FISH Probe Kit

MDS AML

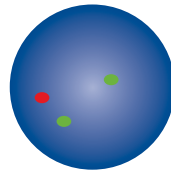
Cat. No. USA LPH-024

Expected normal signal pattern



In a normal cell, two red and two green signals (2R, 2G) are expected.

Expected abnormal signal pattern



A cell with a hemizygous deletion of 5q31.2 will have one red and two green signals (1R, 2G).

Other signal patterns are possible in aneuploid/unbalanced specimens.

Characterization of Normal Cut-off Values

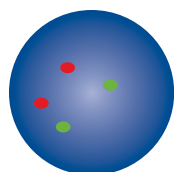
Abnormal signal pattern	Number of samples analyzed to generate the cut-off	Number of nuclei evaluated per sample	Maximum number of false positive signal pattern	Normal cut-off value (per 200 nuclei)	Normal cut-off value (%)
1R, 2G	1300	200	7	13	6.3

Del(7q) Deletion FISH Probe Kit

MDS AML

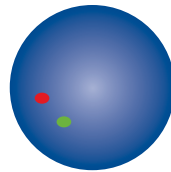
Cat. No. USA LPH-025

Expected normal signal pattern



In a normal cell, two red and two green signals (2R, 2G) are expected.

Expected abnormal signal pattern



One red and one green signal pattern (1R, 1G) will be observed in cells with either monosomy 7 or hemizygous deletion of both CDRs on 7q.

Other signal patterns are possible in aneuploid/unbalanced specimens.

Characterization of Normal Cut-off Values

Abnormal signal pattern	Number of samples analyzed to generate the cut-off	Number of nuclei evaluated per sample	Maximum number of false positive signal pattern	Normal cut-off value (per 200 nuclei)	Normal cut-off value (%)
1R, 1G	1300	200	9	15	7.4

The Cytocel Aquarius AML/MDS range of FISH probe test kits are fluorescence *in situ* hybridization (FISH) tests used to detect common chromosomal rearrangements in fixed bone marrow specimens from patients with acute myeloid leukemia (AML) or myelodysplastic syndrome (MDS). The tests are indicated for the characterization of patient specimens consistent with World Health Organization guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are to be interpreted by a qualified pathologist or cytogeneticist. The tests are not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic. Reporting and interpretation of FISH results should be consistent with professional standards of practice and should take into consideration other clinical and diagnostic information. This kit is intended as an adjunct to other diagnostic laboratory tests and therapeutic action should not be initiated on the basis of the FISH result alone. Failure to adhere to the protocol may affect the performance and lead to false results. Each lab is responsible for establishing their own cut-off values. Each laboratory should test sufficiently large number of samples to establish normal population distribution of the signal levels and to assign a cut-off value. The product is for professional use only and is intended to be interpreted by a qualified Pathologist or Cytogeneticist. Refer to individual test kit Package Insert for the specific intended use and limitations. For *In Vitro* Diagnostic Use. Rx only. Product availability may vary from country to country and is subject to varying regulatory requirements. Please contact your local representatives for availability.

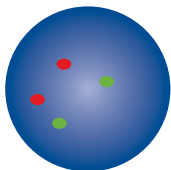


Del(20q) Deletion FISH Probe Kit

MDS

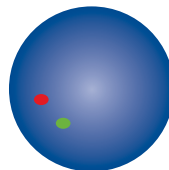
Cat. No. USA LPH-020

Expected normal signal pattern



In a normal cell, two red and two green signals (2R, 2G) are expected.

Expected abnormal signal pattern



One red and one green signal pattern (1R, 1G) will be observed in cells with either monosomy or hemizygous deletion of both bands on 20q.

Other signal patterns are possible in aneuploid/unbalanced specimens.

Characterization of Normal Cut-off Values

Abnormal signal pattern	Number of samples analyzed to generate the cut-off	Number of nuclei evaluated per sample	Maximum number of false positive signal pattern	Normal cut-off value (per 200 nuclei)	Normal cut-off value (%)
1R, 1G	1300	200	6	12	5.7

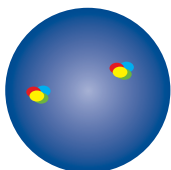
EVI1 (MECOM) Breakapart FISH Probe Kit

MDS AML

Cat. No. USA LPH-036

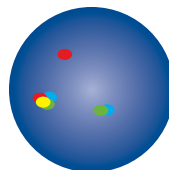
The three-color strategy shows the presence of either a translocation or an inversion and allows for each different type of rearrangement to be distinguished.

Expected normal signal pattern

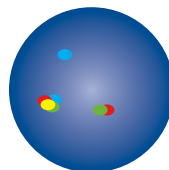


In a normal cell, two red/green/blue fusion signals (2RGB) are expected.

Expected abnormal signal pattern



In a cell with a t(3;3)(q21;q26.2) translocation, one red signal, one green/blue fusion signal and one red/green/blue fusion signal will be observed (1R, 1GB and 1RGB).



In a cell with an inv(3)(q21q26.2) inversion, one red/green fusion, one separate blue signal and one red/green/blue fusion signal will be observed (1RG, 1B and 1RGB).

Other signal patterns are possible in aneuploid/unbalanced specimens.

Characterization of Normal Cut-off Values

Abnormal signal pattern	Number of samples analyzed to generate the cut-off	Number of nuclei evaluated per sample	Maximum number of false positive signal pattern	Normal cut-off value (per 200 nuclei)	Normal cut-off value (%)
1R, 1GB and 1RGB	25	200	3	8	4
1RG, 1B and 1RGB	25	200	3	8	4

The Cytocell Aquarius AML/MDS range of FISH probe test kits are fluorescence *in situ* hybridization (FISH) tests used to detect common chromosomal rearrangements in fixed bone marrow specimens from patients with acute myeloid leukemia (AML) or myelodysplastic syndrome (MDS). The tests are indicated for the characterization of patient specimens consistent with World Health Organization guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are to be interpreted by a qualified pathologist or cytogeneticist. The tests are not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic. Reporting and interpretation of FISH results should be consistent with professional standards of practice and should take into consideration other clinical and diagnostic information. This kit is intended as an adjunct to other diagnostic laboratory tests and therapeutic action should not be initiated on the basis of the FISH result alone. Failure to adhere to the protocol may affect the performance and lead to false results. Each lab is responsible for establishing their own cut-off values. Each laboratory should test sufficiently large number of samples to establish normal population distribution of the signal levels and to assign a cut-off value. The product is for professional use only and is intended to be interpreted by a qualified Pathologist or Cytogeneticist. Refer to individual test kit Package Insert for the specific intended use and limitations. For *In Vitro* Diagnostic Use. Rx only. Product availability may vary from country to country and is subject to varying regulatory requirements. Please contact your local representatives for availability.

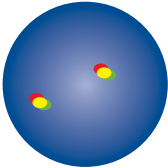


MLL (KMT2A) Breakpart FISH Probe Kit

MDS AML

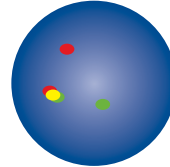
Cat. No. USA LPH-013

Expected normal signal pattern



In a normal cell, two red/green fusion signals are expected (2F).

Expected abnormal signal pattern



In a cell with a balanced *MLL (KMT2A)* rearrangement, the expected signal pattern will be one red, one green and one fusion (1R, 1G, 2F).

Other signal patterns are possible in aneuploid/unbalanced specimens.

Characterization of Normal Cut-off Values

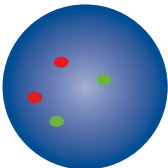
Abnormal signal pattern	Number of samples analyzed to generate the cut-off	Number of nuclei evaluated per sample	Maximum number of false positive signal pattern	Normal cut-off value (per 200 nuclei)	Normal cut-off value (%)
1R, 1G, 1F	1600	200	3	8	3.8

P53 (TP53) Deletion FISH Probe Kit

MDS AML

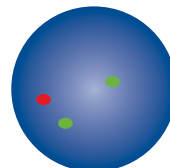
Cat. No. USA LPH-017

Expected normal signal pattern



In a normal cell, two red and two green signals (2R, 2G) are expected.

Expected abnormal signal pattern



A cell with a *TP53* deletion, will have one red and two green signals (1R, 2G).

Other signal patterns are possible in aneuploid/unbalanced specimens.

Characterization of Normal Cut-off Values

Abnormal signal pattern	Number of samples analyzed to generate the cut-off	Number of nuclei evaluated per sample	Maximum number of false positive signal pattern	Normal cut-off value (per 200 nuclei)	Normal cut-off value (%)
1R, 2G	1600	200	8	14	6.8

The Cytocell Aquarius AML/MDS range of FISH probe test kits are fluorescence in situ hybridization (FISH) tests used to detect common chromosomal rearrangements in fixed bone marrow specimens from patients with acute myeloid leukemia (AML) or myelodysplastic syndrome (MDS). The tests are indicated for the characterization of patient specimens consistent with World Health Organization guidelines for Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th Edition) and in conjunction with other clinicopathological criteria. The assay results are to be interpreted by a qualified pathologist or cytogeneticist. The tests are not intended for use as a stand-alone diagnostic, disease screening, or as a companion diagnostic. Reporting and interpretation of FISH results should be consistent with professional standards of practice and should take into consideration other clinical and diagnostic information. This kit is intended as an adjunct to other diagnostic laboratory tests and therapeutic action should not be initiated on the basis of the FISH result alone. Failure to adhere to the protocol may affect the performance and lead to false results. Each lab is responsible for establishing their own cut-off values. Each laboratory should test sufficiently large number of samples to establish normal population distribution of the signal levels and to assign a cut-off value. The product is for professional use only and is intended to be interpreted by a qualified Pathologist or Cytogeneticist. Refer to individual test kit Package Insert for the specific intended use and limitations. For In Vitro Diagnostic Use. Rx only. Product availability may vary from country to country and is subject to varying regulatory requirements. Please contact your local representatives for availability.

Next Generation Sequencing

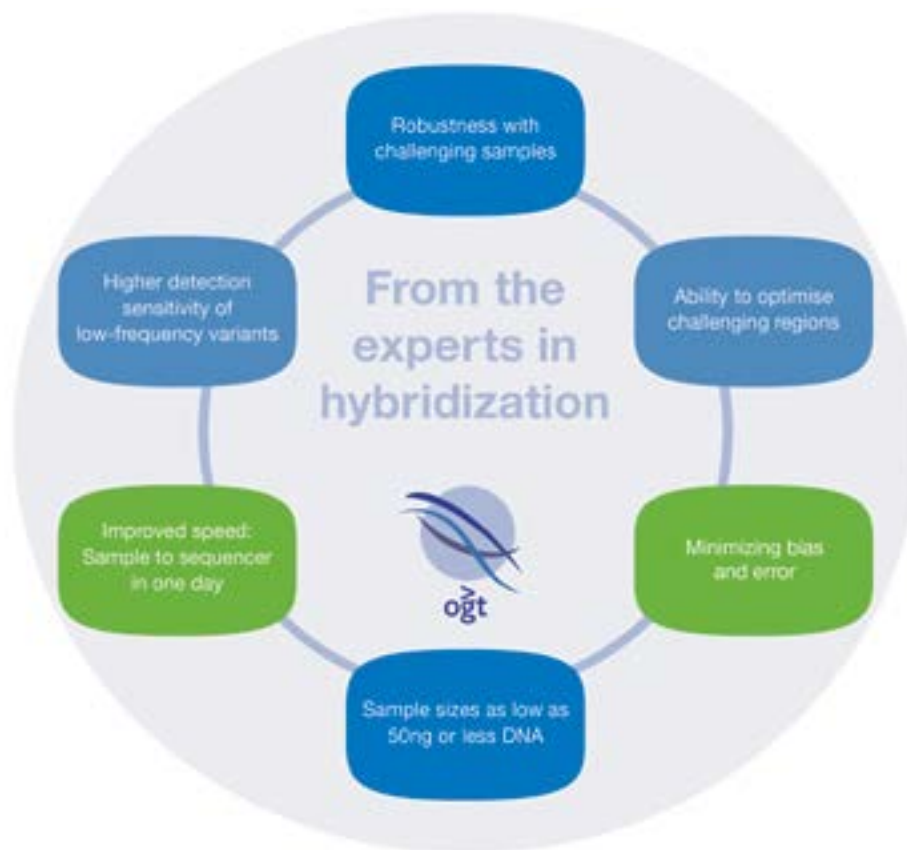
Choosing the best enrichment assay

Next generation sequencing (NGS) is now in routine use for a broad range of research and clinical applications. The rapid rate of adoption has been facilitated by falling reagent costs, benchtop instruments, improved chemistries and improved data analysis solutions. However, the cost and complexity of data analysis still remain significant hurdles — particularly for whole genome sequencing. In the majority of cases, targeted approaches, such as custom NGS panels, are more cost-effective and generate significantly less, but equally meaningful data in a much shorter timescale.

Targeted sequencing requires an initial sequence enrichment step, which, if poorly designed, can be a source of bias and error in the downstream sequencing assay¹.

Which enrichment assay?

Two broad categories of enrichment assays exist: amplicon (PCR) and hybridization. As a very general rule, hybridization-based assays, when designed well, offer superior performance². Please see the following pages for an overview detailing hybridization vs. amplicon. Additional information and a downloadable whitepaper can be found online at www.ogt.com/enrichment.



REFERENCES

1. Aird, D. *et al* (2011) Analyzing and minimizing PCR amplification bias in Illumina sequencing libraries. *Genome Biology* 12:R18 doi:10.1186/gb-2011-12-2-r18
2. Samorodnitsky, E. *et al* (2015) Evaluation of hybridization capture versus amplicon-based methods for whole-exome sequencing. *Hum Mutat* 36(9), 903-915

SureSeq: For Research Use Only; Not for use in diagnostic procedures.

Hybridization vs. Amplicon Enrichment

Higher detection sensitivity of low-frequency variants

Uniformity of enrichment means that all regions are represented more equally, and that variants present in any region will be called. It also allows much lower average sequencing depths to be used, enabling larger numbers of samples to be multiplexed in a run, and significant cost savings.

Reliable detection of low frequency mutations

High uniformity of coverage allows the reliable detection of low frequency somatic events even in FFPE derived DNA. Example is of a 12 bp deletion (c.754_756delCTGAGGATCAATD) in exon 3 of TP53, 6% frequency, using the SureSeq Ovarian Cancer Panel.

Robustness with challenging samples

Unlike amplicon-based assays, hybridization is less susceptible to contaminants found in FFPE-derived DNA. Use of an upstream FFPE repair step can significantly improve mean target coverage.

Improved performance with the SureSeq™ FFPE DNA Repair Mix

The SureSeq FFPE DNA Repair Mix significantly improves mean target coverage resulting in more confident calls.

Improved speed: Sample to sequencer in one day

With a short enzymatic fragmentation step, combined end-repair and adaptor ligation steps, and optimized hybridization in as little as 30 minutes, you can go from sample to sequencer in a single day.

Hybridisation quality – Amplicon speed

The hybridization step has been optimized to take as little as 30 minutes with good quality DNA, without compromising results.

Minimizing bias and error

Hybridization-based assays avoid the tendency towards bias and error seen with amplicon methods as the degree of multiplexing and number of PCR cycles increases.

Minimal variation in amplification efficiency

Comparison of amplicon and hybridization-based enrichment of the GC-rich exons 4 and 5 of the TP53 gene illustrating the superior coverage uniformity.

Sample sizes as low as 50ng or less

Hybridization has moved on. Well-designed hybridization assays can now utilise lower amounts of input DNA, whilst still producing clean, bias-free, high quality data.

Confident detection with low DNA input

Effect of reduced amount of DNA input on mean target coverage and %QTR.

Ability to optimize challenging regions

OGT's innovative bait-design delivers uniform and complete coverage of difficult-to-sequence GC rich regions of the genome.

Outstanding coverage uniformity

Illustration of the excellent uniformity of coverage of the CD38A gene. Depth of coverage per base (grey). Targeted region (green). Gene coding region as defined by RefSeq (blue). GC percentage (red). Repeat regions (pink).



One day hybridization protocol for SureSeq NGS

The application of a one-day hybridization-based enrichment protocol incorporating a rapid (30 minute) hybridization step

Hybridization-based enrichment protocols for next-generation sequencing (NGS) generate higher quality data (e.g. enhanced coverage uniformity, more complete coverage, and more accurate assessment of insertions/deletions (indels) and internal tandem duplications (ITDs)). However, they are generally more time consuming than PCR-based enrichment approaches. OGT has developed a rapid (30 minute) hybridization protocol that enables Illumina sequencer-ready libraries to be generated from purified DNA in 1-day.

This enhanced version of the SureSeq™ library preparation protocol incorporates an enzymatic DNA fragmentation in combination with a rapid hybridization of just 30 minutes. This enhanced protocol reduces the overall processing time by 6 hours, resulting in a streamlined, 1-day workflow. It offers a similar turn-around time to amplicon-based enrichment protocols, without the associated disadvantages, such as PCR bias, allelic bias (indels) and drop-outs, as well as poor uniformity of coverage.

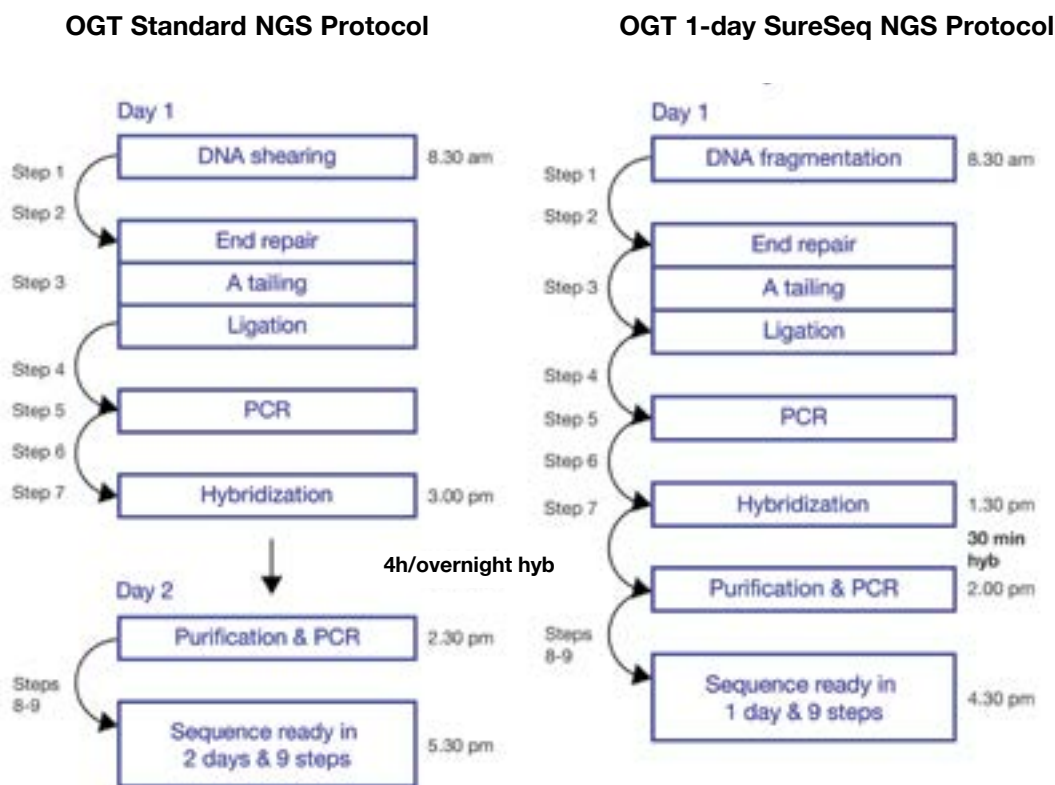


Figure 1: Comparison of workflows

Comparison of the data generated by the 1-day and standard NGS protocols

- Four different hematological panels have been used, with a size range from 0.5 Kb to 138 Kb.
- Data presented here are from 24* samples that were processed using the enhanced LPK in combination with four hematological panels on an Illumina MiSeq.
- The quality of the data generated with the 1-day protocol is comparable to the standard 4-hour hybridization protocol.
- OGT 1-day protocol generated >85% of the % on-target bases generated with the standard protocol. The % change is consistent for all panel sizes.

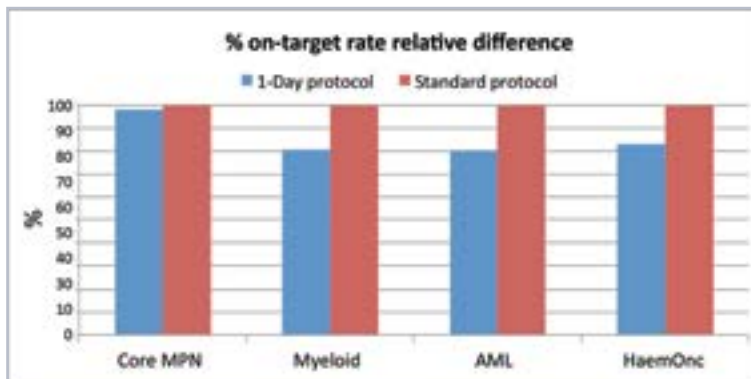


Figure 2: On-target rate comparison between 1-day and standard NGS protocol.

The MTC generated is dependent on the size of each panel. Overall, both workflows generated very good coverage. The MTC generated with the 1-day protocol is >80% of the MTC generated with the standard protocol. The % change is consistent for all panels.

All panels meet the following uniformity specifications: >99% of bases covered at >20% of the mean (after de-duplication). This permits the reliable detection of more complex rearrangements (i.e.) indels and ITDs.

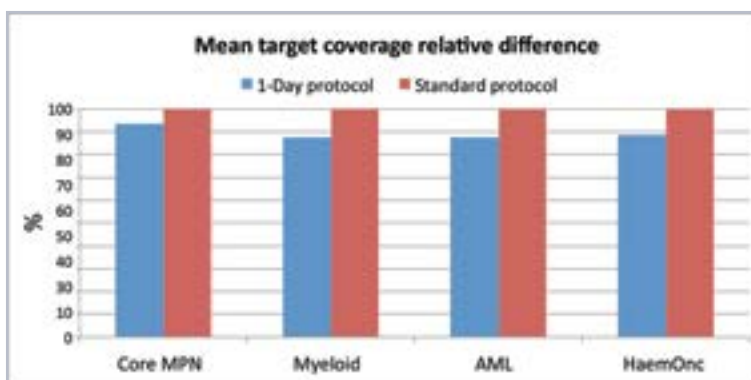


Figure 3: Mean target coverage comparison between 1-day and standard NGS protocol.

Accurate detection of difficult to sequence genes

Mutations in the *CEBPA* and *FLT3* genes are among the most common molecular alterations in *AML*. Sequencing of the *CEBPA* gene is often hampered by a repetitive nucleotide sequence and a very high GC-rich content. Genes such as *FLT3 ITDs* are challenging to target because they are by nature repetitive, can be long and are generally masked in most panel designs.

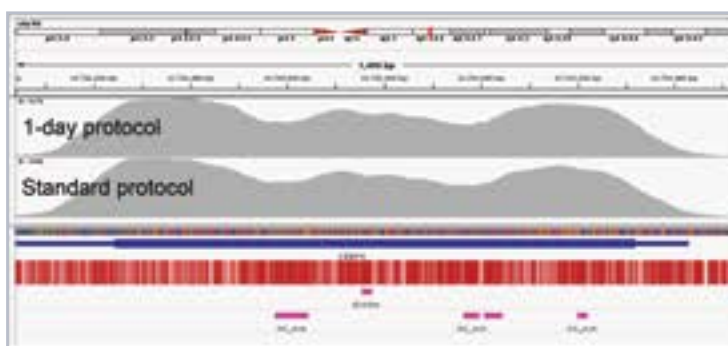


Figure 4: Excellent uniformity of coverage of the *CEBPA* gene averaging ~2000x coverage. Depth of coverage per base (grey). GC percentage (red). Repeat regions and GC-rich regions (pink). Data shown from 1-day protocol.

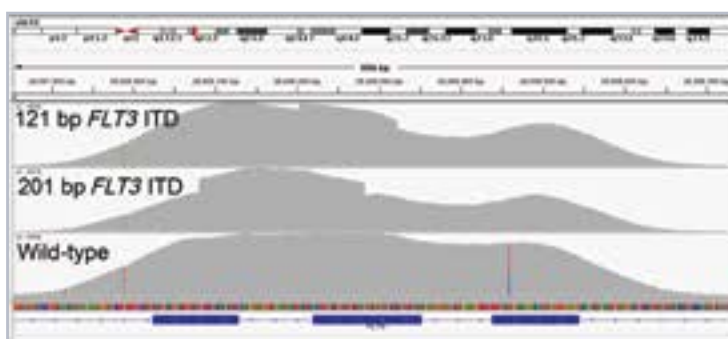


Figure 5: Detection of 121 bp and 201 bp *FLT3* ITD. Wild-type sample (bottom panel).

Conclusions

- We have successfully utilized the OGT 1-day hybridization-based SureSeq LPK protocol in combination with four hematological cancer panels to reliably and routinely detect somatic SNVs by NGS down to a 1% VAF.
- The uniformity of coverage of this approach permitted the detection of key *CALR* and *JAK2* indels (including 52 bp deletions and 5 bp insertions) and *FLT3* ITDs to be identified.
- This enhanced protocol incorporates an enzymatic fragmentation step which permits the high throughput preparation of 24-48 samples (panel size dependent) from genomic DNA to sequencer in a 1-day workflow.
- To achieve >1000x de-duplicated depth (required for confident detection of 1% VAF), 24-48 samples (panel size dependent) can be reliably sequenced in a single MiSeq (V2 300 bp) run. This allows the generation of high quality data in a cost effective and timely manner.

Comprehensive results from a single NGS assay: panel and software highlights

Streamline your research and alleviate the burden of running multiple assays

Investigating both structural aberrations and SNVs/indels is imperative to advance research into progression and treatment of various diseases. For example, copy-number variations (CNVs) are common in Chronic lymphocytic leukemia (CLL) and the BCR-ABL fusion gene is a hallmark of Chronic Myeloid Leukemia (CML). Facilitated by OGT's excellent bait design and Interpret software, SureSeq panels can reliably detect copy-number variations, including trisomies, and translocations; for a more comprehensive understanding of the genetic makeup of each sample – using a single NGS assay.

NGS



Array

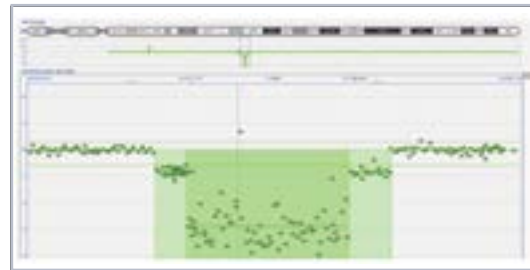


Figure 1: SureSeq CLL + CNV Panel: 0.6Mb biallelic loss called within a larger ~1Mb single allele deletion in the region covering *DLEU2/DLEU1/DLEU7* on chromosome 13q, fully concordant with array data.



Figure 2: *BCR-ABL* translocation reported in Interpret. Split-reads covering both *BCR* (left panel) and *ABL1* (right panel) are detected, indicative of the *BCR-ABL* gene fusion.

Basics of array comparative hybridization (aCGH)

Array comparative genomic hybridization (aCGH) is a powerful tool for analysis of CNV and LOH and is used in a multitude of different applications. CytoSure oligo aCGH products leverage OGT's expert probe design to enable superior CNV resolution to other platforms, detecting microdeletions and microduplications at exon-level resolution across a wide range of disorders.

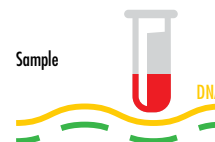
This illustration provides the basics of how aCGH works and the steps involved. For more information, please visit: www.ogt.com/arrays.



Step 1 Labelling



Sample and reference DNA are labelled with different fluorescent dyes



Sample DNA is typically labelled with Cy3 — which looks red under regular light but fluoresces “Green” (575nm) under laser excitation



Reference DNA is typically labelled with Cy5 — which looks blue under regular light but fluoresces “red” (675nm) under laser excitation



Step 2 Preparation



Samples are mixed into a single tube



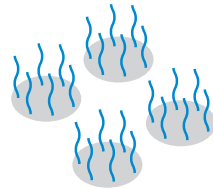
The mixture is pipetted onto a "gasket" slide with chambers which hold the solution



The microarray slide is then held in place against this, ready for hybridization



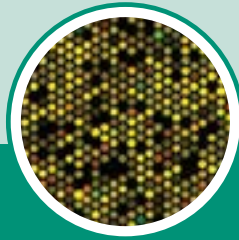
Step 3 Hybridization



Microarrays contain tightly packed "spots" of DNA oligos, also referred to as probes. Each spot is usually designed to target a different region of the genome



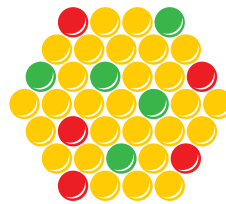
Both the sample and reference labelled DNA compete to bind to the probes, in a process known as "competitive hybridization"



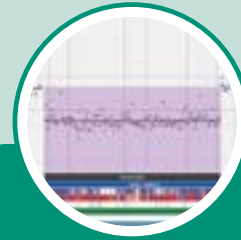
Step 4 Scanning



Slides are scanned — each spot shows the relative amounts of sample vs reference DNA at a particular genomic sequence



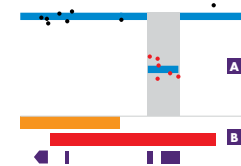
Once scanned, each spot indicates the relative amounts of the sample DNA against the reference DNA at the respective genomic locus. Spots with less sample than reference are indicative of a loss, those with more sample than reference are indicative of a gain



Step 5 Analysis



Purpose-built software allows for analysis and interpretation of aCGH results



A Probe and variant visualisation

B Database tracks

CytoSure Interpret software uses sophisticated algorithms to define areas of CNV or LOH. Track-based analysis allows for interpretation of these regions against external and internal databases

Troubleshooting for arrays

Wet-lab processing is key to achieving the highest quality array data. This quick reference provides an overview of common problems and solutions to improve your data quality. Additional troubleshooting guidelines can be found on www.ogt.com/arrays.

Important QC metrics

DLRS values

This is perhaps the most important QC metric and calculates the probe-to-probe log ratio noise of an array. A poor Derivative Log Ratio Spread (DLRS) will mean that it is more difficult to accurately call amplifications or deletions. The DLRS value should be <0.3 . Higher values can indicate poor quality DNA. To detect very small aberrations, a DLRS value of <0.2 may be required. An excellent array would have a DLRS value of around 0.15; although for some sample types (e.g., formalin fixed paraffin embedded), this may be difficult to achieve. Check the quality of the DNA on a high percentage agarose gel for degradation. If the DNA is degraded, shown by a smear on the gel, re-extract the sample.

Signal to Noise

This value is calculated by dividing the signal intensity by the background noise and indicates how clearly the spots can be detected above the background level. This metric is dependent on how well the sample labelling and washing steps worked. It is often easier to look at this metric first and then, if it does not pass, identify where the problem occurred by looking at the background noise and the signal intensity. An excellent value for signal to noise would be above 100, between 100 and 30 is good but below 30 is poor. It is difficult to reliably detect aberrations on arrays where the Signal-to-Noise is <30 .

Background Noise

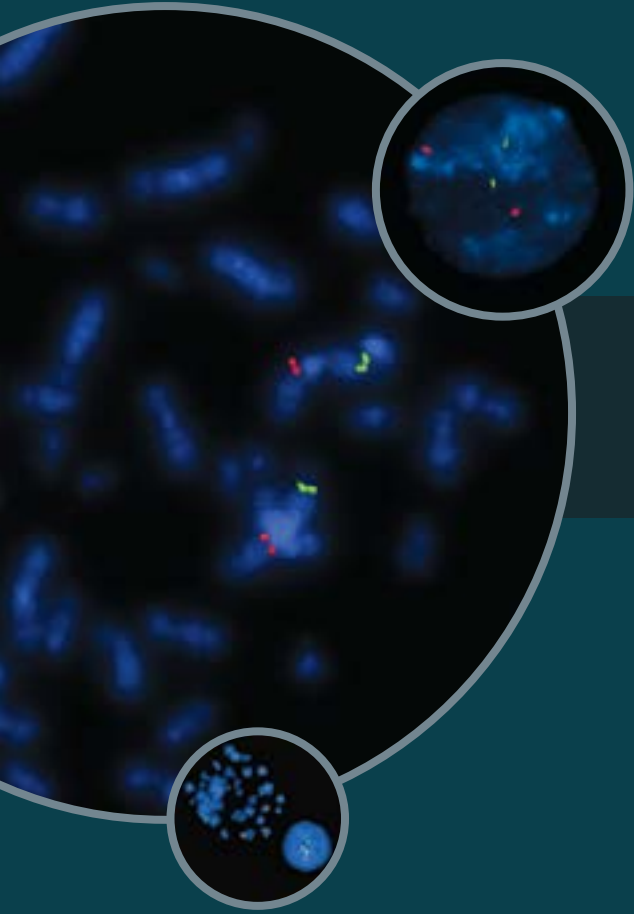
This metric is calculated as the standard deviation of negative control probes on the array. The values are recorded for both the green and red channel and can be classified into Excellent, Good and Poor. The values will depend on the array format being used. A poor background does not necessarily indicate that the array has failed. This is a secondary metric as it is incorporated into the Signal-to-Noise metric.

PROBLEM

IDENTIFIER

SOLUTION

Low A260/280 values	Protein Contamination	Re-purify samples using proteinase steps
High A260/280 values	RNA Contamination	Ensure that your DNA extraction protocol includes RNase
Low A260/230 values	Contamination of salts or solvents (e.g. Phenol)	Re-purified by ethanol precipitation Resuspending the DNA in TE buffer
Inaccurate Sample Concentration	High DNA concentration (> 350ng/μl)	Dilute DNA 1:2 in water or suitable buffer
Low Dye Incorporation— (poor pmol/μl or DNA concentration values)	The wrong temperatures or times are being used	Check temperatures with a calibrated thermometer Check incubation times against protocol
	Incorrect volumes used in mastermix preparation	Check correct volumes are being added Check pipettes are calibrated correctly
	Insufficient mixing of samples, reagents and mastermixes	Gently vortex all reagent tubes (except Klenow) Flick mix Klenow tube Briefly spin to drive contents off tube walls
	Too much exposure to light or air	Use a closed thermal cycler with heated lid
	Loss of solution from evaporation	Use PCR machine with a heated lid If using tubes, make sure lids are tightly closed If using plates, use caps not a plate sealer
Black holes on array	Low volume of Hybridization solution	Ensure the correct volume of hybridization solution has been used Check no leakage of hybridization solution has occurred
Non-uniform signal intensities	Split, deformity or crack in backing slide	Check the backing slide seal is intact and has not cracked Report any gasket slide failures to support@ogt.com
Bubble Scarring/Scotching	Jig assembly untouched for too long after hybridization oven rotation malfunction	Check oven rotators are working Remove jigs from oven one at a time Disassemble under wash buffer rapidly
Fluorescent smears across the slide	Wash-step contamination with fluorescent material Dried-out arrays during the hybridization or wash steps	Ensure dishes are regularly cleaned with appropriate solvent Ensure clean gloves, forceps and dishes Carry out additional acetonitrile wash for 1 min at room temp
High Background Signal	Wash-step contamination with fluorescent material Wash conditions not stringent enough	Ensure dishes are regularly cleaned with appropriate solvent Ensure clean gloves and forceps Check stirrer is producing a vortex prior to adding slides in wash buffer Check temperature of oven and washes
Poor Signal intensity	Overly stringent wash or hybridization conditions Cy5-labelled DNA was exposed to light	Check protocol for correct wash instructions Cover tubes with foil or use amber tubes Check temperature of oven and washes
Low Cy5 signal towards the edges of a feature	Wet ozone: outer edges of features dry quicker than inside, exposing edges to ozone	Ensure slides are scanned immediately after washing Enclose scanners in a box with ozone scrubbers
Low Cy5 signal gradient with more signal loss at one end of the slide	Dry ozone: Degradation during scanning, with exposed end degrading quicker	



Indices of FISH probes



Aquarius® Hematology Probe Range Summary

Probe Name	Chromosome Region	Probe Type	Control Probe	Volume (µl)	Cat. No.	Page
13q14.3	13q14.2-q14.3	Deletion	D13S1825	100	LPH 006-A	31
AFF1	4q21.3-q22.1		–	50	LPH 507-A	49
Alpha Satellite 12 <i>Plus</i>	12p11.1-q11.1	Enumeration	–	100	LPH 069-A	71
AML1/ETO (RUNX1/RUNX1T1) [IVD]	21q22.1 / 8q21.3	Translocation		10 tests	USA-LPH 026*	12
AML1/ETO (RUNX1/RUNX1T1) Dual Fusion	21q22.1 / 8q21.3	Translocation	–	100	LPH 026-A†	21
AML1 (RUNX1)	21q22.1	Breakapart	–	100	LPH 027-A†	20
AML (RUNX1) Distal Probe Green	21q22.1		–	50	LPH 573-A††	–
AML (RUNX1) Proximal Probe Red	21q22.1		–	50	LPH 574-A††	–
ATM	11q22.3	Deletion	D11Z1	100	LPH 011-A	22
BCL2 <i>Plus</i>	18q21.33		–	50	LPH 518-A	42
BCL6	3q27.3	Breakapart	–	100	LPH 035-A	25
BCR/ABL (ABL1) Dual Fusion	22q11.22-q11.23/9q34.11-q34.12	Translocation	–	100	LPH 007-A	23
BCR/ABL (ABL1) <i>Plus</i> Dual Fusion	22q11.22-q11.23/9q34.11-q34.12	Translocation	–	100	LPH 038-A	24
CBFB Proximal Probe Red	16q22		–	50	LPH 526-A	26
CBFB Distal Probe Green	16q22		–	50	LPH 527-A	26
CBFβ (CBFB)/MYH11 Dual Fusion	16p13.1 / 16q22	Translocation	–	10 tests	USA-LPH 022*	13
CBFβ/MYH11 Dual Fusion	16p13.1 / 16q22	Translocation	–	100	LPH 022-A†	27
CCND1 <i>Plus</i>	11q13.3		–	50	LPH 519-A	43
CCND3 <i>Plus</i>	6p21		–	50	LPH 522-A	43
Centromere 17 Probe Green	17p11.1-q11.1		–	50	LPH 572-A††	–
Chromosome 7 Alpha Satellite Probe Aqua	7p11.1-7q11.1		–	33.3	LPH 545-A	62
Chromosome 9 Satellite III Probe Aqua	9q12		–	33.3	LPH 547-A	62
Chromosome 15 Alpha Satellite Probe Red	15p11.1-15q11.1		–	33.3	LPH 548-A	71
CKS1B/CDKN2C(P18)	1p32.3/1q21.3	Amplification/ Deletion	–	100	LPH 039-A	29
cMYC (MYC)	8q24.21	Breakapart	–	100	LPH 010-A	29
cMYC (MYC) <i>Plus</i>	8q24.21		–	50	LPH 523-A	44
CRLF2 Distal	Xp22.33/Yp11.32		–	50	LPH 511-A	30
CRLF2 Proximal	Xp22.33/Yp11.32		–	50	LPH 512-A	30
CSF1R/RPS14 (5q32-q33) Probe Red	5q32-q33		–	33.3	LPH 540-A	28
CUX1 (7q22) Probe Green	7q22		–	33.3	LPH 543-A	31
D13S319 <i>Plus</i>	13q14.2-14.3	Deletion	LAMP1	100	LPH 068-A	32
D13S25	13q14.3	Deletion	D13S1825	100	LPH 043-A	33
DEK Probe Green	6p22.3		–	50	LPH 531-A	69
Del(5q) [IVD]	5p15.3/5q31.2	Deletion	5p15.3	10 tests	USA-LPH 024*	14
Del(5q)	5p15.3/5q31.2	Deletion	5p15.3	100	LPH 024-A†	33
Del(7q) [IVD]	7q22 / 7q31.2	Deletion	–	10 tests	USA-LPH 025*	15
Del(7q)	7q22 / 7q31.2	Deletion	–	100	LPH 025-A†	34
Del(20q) [IVD]	20q12/ 20q13.1	Deletion	–	10 tests	USA-LPH 020*	16
Del(20q)	20q12/ 20q13.1	Deletion	–	100	LPH 020-A†	34
E2A (TCF3)	19p13.3	Breakapart	–	100	LPH 019-A	35
E2A (TCF3)	19p13.3		–	50	LPH 503-A	36

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

† For sale in Canada only.

†† See product page/website for availability in your region.



Aquarius® Hematology Probe Range Summary

Probe Name	Chromosome Region	Probe Type	Control Probe	Volume (µl)	Cat. No.	Page
EGR1/CDC25C Probe Green	5q31		–	33.3	LPH 541-A	37
ETV6 Distal Probe Red	12p13.2		–	50	LPD 502-A	67
ETV6 Proximal Probe Green	12p13.2		–	50	LPD 501-A	67
EVI1 (MECOM) [IVD]	3q26.2	Breakapart	–	10 tests	USA-LPH 036*	17
EVI1 (MECOM)	3q26.2	Breakapart	–	100	LPH 036-A [†]	38
EZH2 (7q36) Probe Red	7q36		–	33.3	LPH 544-A	38
FAST PML	15q24.1		–	50	LPH 501-A	57
FAST RARα (RARA)	17q21.1-q21.2		–	50	LPH 502-A	57
FGFR3 <i>Plus</i>	4p16.3		–	50	LPH 521-A	44
FIP1L1/CHIC2/PDGFRα	4q12	Deletion/Fusion	–	100	LPH 032-A	39
HLF	17q22		–	50	LPH 505-A	37
IGH Breakapart	14q32.3	Breakapart	–	100	LPH 014-A	40
IGH Proximal <i>Plus</i>	14q32.3		–	50	LPH 515-A	40
IGH Distal <i>Plus</i>	14q32.3		–	50	LPH 516-A	41
IGH <i>Plus</i>	14q32.3		–	50	LPH 517-A	41
IGH Probe Green	14q32.3		–	50	LPH 568-A	42
IGK	2p11.2	Breakapart	–	100	LPH 034-A	46
IGL	22q11.21-q11.23	Breakapart	–	100	LPH 033-A	47
MAF v2 Probe Red	16q23		–	50	LPH 567-A	45
MAFB <i>Plus</i>	20q12		–	50	LPH 524-A	45
MECOM Probe Red	3q26.2		–	100	LPH 528-A	47
MLL (KMT2A) [IVD]	11q23.3	Breakapart	–	10 tests	USA-LPH 013*	18
MLL (KMT2A)	11q23.3	Breakapart	–	100	LPH 013-A [†]	48
MLL (KMT2A) Distal Probe Red	11q23.3		–	50	LPH 570-A ^{††}	–
MLL (KMT2A) Proximal Probe Green	11q23.3		–	50	LPH 569-A ^{††}	–
MLL (KMT2A)	11q23.3		–	50	LPH 506-A	48
MLLT1	19p13.3		–	50	LPH 508-A	49
MLLT3	9p21.3		–	50	LPH 509-A	50
MLLT4 (AFDN)	6q27		–	50	LPH 510-A	50
MPO Probe Red	17q22		–	50	LPD 503-A	68
MYB	6q23.3	Deletion	D6Z1	100	LPH 016-A	51
MYEOV <i>Plus</i>	11q13.3		–	50	LPH 525-A	46
NUP98 Proximal Red	11p15.4		–	50	LPH 532-A	51
NUP98 Distal Green	11p15.4		–	50	LPH 533-A	52
NUP214 Probe Red	9q34.12-q34.13		–	50	LPH 530-A	69
P16 (CDKN2A)	9p21.3	Deletion	D9Z3	100	LPH 009-A	52
P2RY8 Distal	Xp22.33/Yp11.32		–	50	LPH 513-A	54
P2RY8 Proximal	Xp22.33/Yp11.32		–	50	LPH 514-A	54
P53 (TP53) [IVD]	17p13	Deletion	D17Z1	10 tests	USA-LPH 017*	19
P53 (TP53)	17p13	Deletion	D17Z1	100	LPH 017-A [†]	53
P53 (TP53) Probe Red	17p13		–	50	LPH 571-A ^{††}	–
P53 (TP53)/ATM Probe Combination	17p13/11q22.3	Deletion	–	100	LPH 052-A	55
PBX1	1q23.3		–	50	LPH 504-A	36
PDGFRB	5q32	Breakapart	–	100	LPH 031-A	56
PML/RARα (RARA) Dual Fusion	15q24.1/17q21.1-q21.2	Translocation	–	100	LPH 023-A	58

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

[†] For sale in Canada only.

^{††} For sale in the US only. These products have not been licensed in accordance with Canadian law.



Aquarius® Hematology Probe Range Summary

Probe Name	Chromosome Region	Probe Type	Control Probe	Volume (µl)	Cat. No.	Page
RARα (RARA) Proximal	17q21.1-q21.2		–	50	LPH 062-A	59
RARα (RARA) Distal	17q21.2		–	50	LPH 063-A	59
RUNX1 Probe Green	21q22.12		–	50	LPH 529-A	60
TAS2R1 (5p15.31) Probe Green	5p15.31		–	33.3	LPH 546-A	70
TCL1	14q32.13-q32.2	Breakapart	–	100	LPH 046-A	63
TCRAD	14q11.2	Breakapart	–	100	LPH 047-A	63
TCRB (TRB)	7q34	Breakapart	–	100	LPH 048-A	64
TEL/AML1 (ETV6/RUNX1) Dual Fusion	12p13.2/21q22.1	Translocation	–	100	LPH 012-A	65
TERT (5p15.33) Probe Aqua	5p15.33		–	33	LPH 542-A	60
TET2 Probe Red	4q24		–	50	LPH 534-A	61
TLX1	10q24.31	Breakapart	–	100	LPH 049-A	66
TLX3	5q35.1	Breakapart	–	100	LPH 050-A	66
TP53 Probe Green	17p13		–	50	LPD 504-A	68
USP46(4q12) Probe Green	4q12		–	50	LPH 535-A	61

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

† For sale in Canada only.

†† For sale in the US only. These products have not been licensed in accordance with Canadian law.





Aquarius® Tissue Pretreatment Kit Summary

Product Description	Kit Format	Cat. No.	Page
Aquarius Tissue Pretreatment Kit*	Reagent 1 (1x1L), Reagent 2 (1x10ml)	LPS 100	87

* This product is provided under an agreement between Life Technologies Corporation and Cytocell Ltd and is available for human diagnostic or life science use only.

Aquarius® Hematopathology Probe Range Summary

Probe Name	Chromosome Region	Probe Type	Control Probe	Volume (µl)	Cat. No.	Page
BCL2	18q21.33-q22.1	Breakapart	–	100	LPS 028-A	75
BCL6	3q27.3-q28	Breakapart	–	100	LPS 029-A	75
CCND1	11q13.3	Breakapart	–	100	LPS 030-A	76
IGH	14q32.3	Breakapart	–	100	LPS 032-A	76
IGH/BCL2 Dual Fusion	14q32.3/18q21.33-q22.1	Translocation	–	100	LPS 033-A	77
IGH/CCND1 Dual Fusion	14q32.3/11q13.3	Translocation	–	100	LPS 031-A	78
IGH/MALT1 Dual Fusion	14q32.3/18q21.31-q21.32	Translocation	–	100	LPS 034-A	79
IGH/MYC Dual Fusion	14q32.3/8q24.21	Translocation	–	100	LPS 035-A	80
IGK	2p11.2	Breakapart	–	100	LPS 038-A	81
IGL	22q11.21-q11.23	Breakapart	–	100	LPS 039-A	81
MALT1	18q21.31-q21.32	Breakapart	–	100	LPS 017-A	82
MYC	8q24.21	Breakapart	–	100	LPS 027-A	82
P16 (CDKN2A)	9p21.3	Deletion	D9Z3	100	LPS 036-A	83
P53 (TP53)	17p13	Deletion	D17Z1	100	LPS 037-A	83
RB1	13q14.2	Deletion	LAMP1	100	LPS 011-A	84

Aquarius® Pathology Probe Range Summary

Probe Name	Chromosome Region	Probe Type	Control Probe	Volume (µl)	Cat. No.	Page
1p36	1p36.32		–	50	LPS 513-A	88
1q25	1q25.2		–	50	LPS 512-A	88
19p13	19p13.2		–	50	LPS 515-A	89
19q13	19q13.33		–	50	LPS 516-A	89
ALK	2p23.2-p23.1	Breakapart	–	100	LPS 019-A	90
CDKN2A Probe Gold	9p21.3		–	15/75	LPS 532-A ^{††}	90
CHOP (DDIT3)	12q13.3	Breakapart	–	100	LPS 015-A	91
Centromere 3 Probe Red	D3Z1		–	15/75	LPS 526-A ^{††}	92
Centromere 7 Probe Green	D7Z1		–	15/75	LPS 527-A ^{††}	92
Centromere 17 Probe Aqua	D17Z1		–	15/75	LPS 528-A ^{††}	91
C-MET (MET)	7q31.2	Amplification	D7Z1	100	LPS 004-A	93
EGFR	7p11.2	Amplification	D7Z1	100	LPS 003-A	93
ERG	21q22.13-q22.2	Control	–	50	LPS 099-A	94
EML4	2p21	Breakapart	–	100	LPS 020-A	94
ETV6 Distal Probe Red	12p13.2		–	50	LPD 502-A	95
ETV6 Proximal Probe Green	12p13.2		–	50	LPD 501-A	95
EWSR1	22q12.1-q12.2	Breakapart	–	100	LPS 006-A	96

^{††} For sale in the US only. These products have not been licensed in accordance with Canadian law.



Aquarius® Pathology Probe Range Summary

Probe Name	Chromosome Region	Probe Type	Control Probe	Volume (µl)	Cat. No.	Page
EWSR1/ERG Dual Fusion	21q22.1-q22.2/22q12.1-q12.2	Translocation	–	100	LPS 008-A	97
FGFR1	8p11.23-p11.22	Breakapart/ Amplification	D8Z2	100	LPS 018-A	101
FLI1/EWSR1 Dual Fusion	11q24.3/22q12.1-q12.2	Translocation	–	100	LPS 007-A	100
FOXO1 Proximal Green	13q14.1		–	50	LPS 518-A	98
FOXO1 Distal Red	13q14.1		–	50	LPS 519-A	98
FUS Proximal Red	16p11.2		–	50	LPS 520-A	99
FUS Distal Green	16p11.2		–	50	LPS 521-A	99
HER2 (ERBB2)	17q12	Amplification	–	100	LPS 001-A	101
MDM2	12q15	Amplification	D12Z1	100	LPS 016-A	102
MPO Probe Red	17q22		–	50	LPD 503-A	102
N-MYC (MYCN)	2p24.3/2q11.2	Amplification	AFF3	100	LPS 009-A	103
PAX3	2q36.1	Breakapart	–	100	LPS 012-A	103
PAX7	1p36.13	Breakapart	–	100	LPS 013-A	104
RET Distal	10q11.21		–	50	LPS 509-A	105
RET Proximal	10q11.21		–	50	LPS 508-A	104
ROS1	6q22.1	Breakapart	–	100	LPS 022-A	105
ROS1-GOPC (FIG) Distal	6q22.1		–	50	LPS 511-A	106
ROS1-GOPC (FIG) Proximal	6q22.1		–	50	LPS 510-A	106
SRD (CHD5)	1p36.31	Deletion	ZNF672	100	LPS 010-A	107
SYT (SS18)	18q11.2	Breakapart	–	100	LPS 014-A	107
TFE3 Proximal Probe Red	Xp11.23		–	50	LPS 522-A	108
TFE3 Distal Probe Green	Xp11.23		–	50	LPS 523-A	109
TMPRSS2	21q22.2-q22.3	Breakapart	–	50	LPS 098-A	108
TOP2A	17q21.2	Amplification/ Deletion	D17Z1	100	LPS 002-A	109
TP53 Probe Green	17p13		–	50	LPD 504-A	110
ZNF217	20q13.2	Amplification	20pter	100	LPS 005-A	110

** For sale in the US only. These products have not been licensed in accordance with Canadian law.





Aquarius® Microdeletion Probe Range Summary

Probe Name	Chromosome Region	Probe Loci	Control Probe	Volume (µl)	Cat. No.*	Page
Angelman (UBE3A/D15S10) Region	15q11.2-q12	UBE3A/D15S10	15qter	50 or 100	LPU 006-A ^{††}	113
Cri-du-chat Region/Sotos Region	5p15.31/5p15.2/5q35	UBE2QL1, CTNND2, NSD1	–	50 or 100	LPU 013-A ^{††}	114
DiGeorge II (10p14)	10p14	CELF2	D10Z1	50 or 100	LPU 015-A ^{††}	115
DiGeorge TBX1 Region and 22q13.3 Region	22q11.2/22q13.3	TBX1, SHANK3	–	50 or 100	LPU 014-A ^{††}	118
DiGeorge/VCFS N25 Region and 22q13.3 Region	22q11.2/22q13.3	N25/D22S75, SHANK3	–	50 or 100	LPU 010-A ^{††}	117
DiGeorge/VCFS TUPLE1 Region and 22q13.3 Region	22q11.2/22q13.3	TUPLE1, SHANK3	–	50 or 100	LPU 004-A	116
Kallmann (KAL1) Region/STS Region	Xp22.31	KAL1, STS	DXZ1	50 or 100	LPU 016-A ^{††}	119
Prader-Willi/Angelman (SNRPN) Region	15q11.2	SNRPN	15qter	50 or 100	LPU 005-A	120
Saethre-Chotzen Region/Williams-Beuren Region	7p21.1/7q11.23	TWIST1, WBSCR/ELN	–	50 or 100	LPU 024-A	121
SHOX	Xp22.33/Yp11.32	SHOX	DXZ1, DYZ1	50 or 100	LPU 025-A	122
Smith-Magenis (RAI1) Region/Miller-Dieker Region	17p11.2/17p13.3	RAI1, PAFAH1B1 (LIS1)	–	50 or 100	LPU 019-A	123
SRY	Yp11.31	SRY	DXZ1, DYZ1	50 or 100	LPU 026-A	124
Williams-Beuren Region	7q11.23	WBSCR/ELN	D7Z1	50 or 100	LPU 011-A ^{††}	125
Wolf-Hirschhorn Region	4p16.3	NSD2 (MMSET), NELFA (WHSC2)	4qter	50 or 100	LPU 009-A ^{††}	126

* For smaller volume probes add -SA to catalog number, e.g: LPU ###-SA

^{††} See product page for availability in your region.



Aquarius® Satellite Enumeration Probe Range Summary

Chromosome	Locus	Chromosome Region	DNA Class	Volume (µl)	Cat. No.*	Page
1	D1Z1	1q12	satellite III	15	LPE 001R/G-A	129
2	D2Z2	2p11.1-q11.1	α-satellite	15	LPE 002R/G-A	129
3	D3Z1	3p11.1-q11.1	α-satellite	15	LPE 003R/G-A	129
4	D4Z1	4p11.1-q11.1	α-satellite	15	LPE 004R/G-A	129
1/5/19	D1Z7	1p11.1-q11.1	α-satellite	15	LPE 005R/G-A	129
	D5Z2	5p11.1-q11.1				129
	D19Z3	19p11.1-q11.1				129
6	D6Z1	6p11.1-q11.1	α-satellite	15	LPE 006R/G-A	129
7	D7Z1	7p11.1-q11.1	α-satellite	15	LPE 007R/G-A	129
8	D8Z2	8p11.1-q11.1	α-satellite	15	LPE 008R/G-A	129
	D8Z2	8p11.1-q11.1	α-satellite	30	LPE 008B-A	130
9	D9Z3	9q12	satellite III	15	LPE 009R/G-A	129
10	D10Z1	10p11.1-q11.1	α-satellite	15	LPE 010R/G-A	129
11	D11Z1	11p11.1-q11.1	α-satellite	15	LPE 011R/G-A	129
12	D12Z3	12p11.1-q11.1	α-satellite	15	LPE 012R/G-A	129
	D12Z3	12p11.1-q11.1	α-satellite	30	LPE 012B-A	130
13/21	D13Z1	13p11.1-q11.1	α-satellite	15	LPE 013R/G-A	129
	D21Z1	21p11.1-q11.1				129
14/22	D14Z1	14p11.1-q11.1	α-satellite	15	LPE 014R/G-A	129
	D22Z1	22p11.1-q11.1				129
15	D15Z4	15p11.1-q11.1	α-satellite	15	LPE 015R/G-A	129
16	D16Z2	16p11.1-q11.1	α-satellite	15	LPE 016R/G-A	129
17	D17Z1	17p11.1-q11.1	α-satellite	15	LPE 017R/G-A	129
	D17Z1	17p11.1-q11.1	α-satellite	30	LPE 017B-A	130
18	D18Z1	18p11.1-q11.1	α-satellite	15	LPE 018R/G-A	129
20	D20Z1	20p11.1-q11.1	α-satellite	15	LPE 020R/G-A	129
X	DXZ1	Xp11.1-q11.1	α-satellite	15	LPE 0XR/G-A	129
Yc	DYZ3	Yp11.1-q11.1	α-satellite	15	LPE 0YcR/G-A	129
Yq	DYZ1	Yq12	satellite III	15	LPE 0YqR/G-A	129
XYc Dual Labeled	DXZ1	Xp11.1-q11.1	α-satellite	100	LPE 0XYc-A	130
	DYZ3	Yp11.1-q11.1				
XYq Dual Labeled	DXZ1	Xp11.1-q11.1	α-satellite	100	LPE 0XYq-A	130
	DYZ1	Yq12	satellite III			

* R specifies a red label and G specifies a green label and B specifies a blue label.

Acro-P-Arm Probe

Chromosome	Color	Volume (µl)	Cat. No.	Page
13, 14, 15, 21, 22	Red	100	LPE NOR-A	130





Aquarius® Subtelomere Specific Probe Range Summary

Probe Specificity	Clone Name	Marker (STS)	Max. physical distance from Telomere (kb)	Cat. No.*	Page
1p	CEB108	RH120573	987	LPT 01PR/G-A	133
1q	160H23	GDB:315525	54	LPT 01QR/G-A	133
2p	dJ892G20	D2S2983	18	LPT 02PR/G-A	133
2q	dJ1011O17	D2S2986	277	LPT 02QR/G-A	133
2q NP	172I13	D2S447	311	LPT 02QNPR/G-A	133
3p	dJ1186B18	D3S4559	213	LPT 03PR/G-A	133
3q	196F4	D3S1272	959	LPT 03QR/G-A	133
4p	36P21	D4S3360	67	LPT 04PR/G-A	133
4q	dJ963K6	D4S139	372	LPT 04QR/G-A	133
5p	189N21	RH120167	2254	LPT 05PR/G-A	133
5q	240G13	D5S2907	222	LPT 05QR/G-A	133
6p	62I11	STS-H99640	147	LPT 06PR/G-A	133
6q	57H24	D6S2522	230	LPT 06QR/G-A	133
7p	109a6	RH104000	118	LPT 07PR/G-A	133
7q	2000a5	RH48601	138	LPT 07QR/G-A	133
8p	dJ580L5	RH40619	150	LPT 08PR/G-A	133
8q	489D14	D8S595	202	LPT 08QR/G-A	133
9p	43N6	RH65569	226	LPT 09PR/G-A	133
9q	112N13	D9S2168	167	LPT 09QR/G-A	133
10p	306F7	STS-N35887	271	LPT 10PR/G-A	133
10q	137E24	RH44494	138	LPT 10QR/G-A	133
11p	dJ908H22	D11S2071	189	LPT 11PR/G-A	133
11q	dJ770G7	D11S4974	3447	LPT 11QR/G-A	133
12p	496A11	D12S200	771	LPT 12PR/G-A	133
12q	221K18	RH81094	90	LPT 12QR/G-A	133
13q	163C9	D13S1825	17	LPT 13QR/G-A	133
14q	dJ820M16	D14S1420	143	LPT 14QR/G-A	133
15q	154P1	D15S936	328	LPT 15QR/G-A	133
16p	121I4	SHGC-16929	147	LPT 16PR/G-A	133
16q	240G10	RH80305	331	LPT 16QR/G-A	133
17p	2111b1	D17S2199	143	LPT 17PR/G-A	133
17q	362K4	-	34	LPT 17QR/G-A	133
18p	74G18	D18S552	141	LPT 18PR/G-A	133
18q	dJ964M9	D18S1390	155	LPT 18QR/G-A	133
19p	dJ546C11	D19S676E	260	LPT 19PR/G-A	133
19q	F21283	RH102404	49	LPT 19QR/G-A	133
20p	dj1061L1	D20S210	165	LPT 20PR/G-A	133
20q	81F12	RH10656	153	LPT 20QR/G-A	133
21q	63H24	D21S1446	29	LPT 21QR/G-A	133
22q	99K24	D22S1726	101	LPT 22QR/G-A	133
XpYp**	839D20	DXYS129	344	LPT XYPR/G-A	133
XqYq***	225F6 C8.2/1	DXYS154 SYBL1	64 131	LPT XYQR/G-A LPT XYQR/G-A	133

* R specifies a red label, G specifies a green label

** This probe is specific for the p-arms of both X and Y

*** This probe is specific for the q-arms of both X and Y

NP Non Polymorphic



Accessories

Cat. No.	Description	Unit Size
PCN009	Porcelain Wash Jars - 12 Slide Capacity	2
PCN004	Hybridization Chamber	1
PCN007	24 Square Template Slides	100
PCN008	8 Square Template Slides	100
PCN002	Slide Surface Thermometer	4

Ancillary Reagents

Cat. No.	Description	Unit Size
DES500L	0.125µg/ml DAPI	500µl
DES1000L	0.125µg/ml DAPI	1000µl
DFS500L	1.0µg/ml DAPI	500µl
DSS500L	0.0625µg/ml DAPI	500µl
HB500L	Hybridization Solution B	500µl
HB1000L	Hybridization Solution B	1000µl
LPS 100	Aquarius Tissue Pretreatment Kit*	Reagent 1 (1x1L), Reagent 2 (1x10ml)
PCA003	20x SSC	100ml
PCA005	Rubber Solution Glue	15g
PCN003	Mounting Medium	10ml

Microscope Filters**

Cat. No.	Description	Unit Size
CF69008	Chroma® Filter: 69008 ET-Aqua/FITC/Texas Red Triple Filter Set	1
CF69011	Chroma® Filter: 69011 ET-Aqua/Green/Orange Triple Filter Set	1
CF49000	Chroma® Filter: 49000 ET-DAPI Single Filter	1
CF49302	Chroma® Filter: 49302 ET-Aqua Single Filter	1
CF49303	Chroma® Filter: 49303 ET-Green Single Filter	1
CF49306	Chroma® Filter: 49306 ET-Red Single Filter	1
CF59010	Chroma® Filter: 59010 ET-Green/Red Dual Filter	1
CF59011	Chroma® Filter: 59011 ET-Green/Orange Dual Filter	1
CF59022	Chroma® Filter: 59022 ET-FITC/Texas Red Dual Filter	1

Blocks

Cat. No.	Description	Unit Size
CBZ0001	Chroma® Block: Zeiss Microscope	1
CBBX051	Chroma® Block: Olympus BX51	1
CBBX061	Chroma® Block: Olympus BX61	1
CBNK050	Chroma® Block: Nikon 50i	1
CBDM550	Chroma® Block: Leica DM5500	1

Chroma is a registered trademark of Chroma Technology Corporation. Zeiss is a trademark of Carl Zeiss AG. Olympus is a trademark of Olympus Corporation. NIKON is a trademark of Nikon Corporation. LEICA is a trademark of LEICA MICROSYSTEMS IR GMBH.

* This product (LPS 100) is provided under an agreement between Life Technologies Corporation and Cytocell Ltd and is available for human diagnostics or life science use only.

** Microscope filters are available on request. These filters can be ordered with or without a filter cube.





Index by Chromosome

Chromosome Region	Probe Name	Regulatory Status	Control Region	Cat. No.	Page
1-22, X, Y	Satellite Enumeration Probes-Aquarius	ASR	–	LPE xxxR/G-A	129
1-22, X, Y	Subtelomere Specific Probes-Aquarius	ASR	–	LPT xxxP/Q, R/G-A	133
1p32.3/1q21.3	CKS1B/CDKN2C (P18) Amplification/Deletion	ASR	–	LPH 039-A	29
1p36.13	PAX7 Breakapart	ASR	–	LPS 013-A	104
1p36.31	SRD (CHD5) Deletion	ASR	ZNF672	LPS 010-A	107
1p36.32	1p36	ASR	–	LPS 513-A	88
1q21.3/1p32.3	CKS1B/CDKN2C (P18) Amplification/Deletion	ASR	–	LPH 039-A	29
1q23.3	PBX1	ASR	–	LPH 504-A	36
1q25.2	1q25	ASR	–	LPS 512-A	88
2p11.2	IGK Breakapart	ASR	–	LPH 034-A	46
2p11.2	IGK Breakapart (Hematopathology)	ASR	–	LPS 038-A	81
2p21	EML4 Breakapart	ASR	–	LPS 020-A	94
2p23.2-p23.1	ALK Breakapart	ASR	–	LPS 019-A	90
2p24.3/2q11.2	N-MYC (MYCN) Amplification	ASR	AFF3 (2q11.2)	LPS 009-A	103
2q11.2/2p24.3	N-MYC (MYCN) Amplification	ASR	AFF3 (2q11.2)	LPS 009-A	103
2q36.1	PAX3 Breakapart	ASR	–	LPS 012-A	103
3cen	Centromere 3 Probe Red	ASR	–	LPS 526-A ^{††}	92
3q26.2	EVI1 (MECOM) Breakapart	ASR	–	LPH 036-A [†]	38
3q26.2	MECOM Probe Red	ASR	–	LPH 528-A	47
3q27.3-q28	BCL6 Breakapart (Hematopathology)	ASR	–	LPS 029-A	75
3q27.3	BCL6 Breakapart	ASR	–	LPH 035-A	25
4p16.3	FGFR3 <i>Plus</i>	ASR	–	LPH 521-A	44
4p16.3	Wolf-Hirschhorn	ASR	4qter	LPU 009-A ^{††}	126
4q12	FIP1L1/CHIC2/PDGFRA Deletion/Fusion	ASR	–	LPH 032-A	39
4q12	USP46 (4q12) Probe Green	ASR	–	LPH 535-A	61
4q21.3-q22.1	AFF1	ASR	–	LPH 507-A	49
4q24	TET2 Probe Red	ASR	–	LPH 534-A	61
5p15.3/5q31.2	Del(5q) Deletion	IVD	5p15.3	USA-LPH 024*	14
5p15.3/5q31.2	Del(5q) Deletion	ASR	5p15.3	LPH 024-A [†]	33
5p15.31/5p15.2/5q35	Cri-du-chat Region/Sotos Region	ASR	–	LPU 013-A ^{††}	114
5p15.31	TAS2R1 (5p15.31) Probe Green	ASR	–	LPH 546-A	70
5p13.33	TERT (5p15.33) Probe Aqua	ASR	–	LPH 542-A	60
5p15.2/5p15.31/5q35	Cri-du-chat Region/Sotos Region	ASR	–	LPU 013-A ^{††}	114
5q31	EGR1/CDC25C Probe Green	ASR	–	LPH 541-A	37
5q31.2/5p15.3	Del(5q) Deletion	IVD	5p15.3	USA-LPH 024*	14
5q31.2/5p15.3	Del(5q) Deletion	ASR	5p15.3	LPH 024-A [†]	33
5q32.33	CSF1R/RPS14 (5q32-q33) Probe Red	ASR	–	LPH 540-A	28
5q32	PDGFRB Breakapart	ASR	–	LPH 031-A	56
5q35/5p15.31/5p15.2	Cri-du-chat Region/Sotos Region	ASR	–	LPU 013-A ^{††}	114
5q35.1	TLX3 Breakapart	ASR	–	LPH 050-A	66
6p21	CCND3 <i>Plus</i>	ASR	–	LPH 522-A	43
6q22.1	ROS1 Breakapart	ASR	–	LPS 022-A	105

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

[†] For sale in Canada only.

^{††} See product page/website for availability in your region.



Index by Chromosome

Chromosome Region	Probe Name	Regulatory Status	Control Region	Cat. No.	Page
6q22.1	ROS1-GOPC (FIG) Proximal	ASR	–	LPS 510-A	106
6q22.1	ROS1-GOPC (FIG) Distal	ASR	–	LPS 511-A	106
6p22.3	DEK Probe Green	ASR	–	LPH 531-A	69
6q23.3	MYB Deletion	ASR	D6Z1	LPH 016-A	51
6q27	MLLT4 (AFDN)	ASR	–	LPH 510-A	50
7cen	Centromere 7 Probe Green	ASR	–	LPS 527-A ^{††}	92
7p11.1	Chromosome 7 Alpha Satellite Probe Aqua	ASR	–	LPH 545-A	62
7p21.1/7q11.23	Saethre-Chotzen Region/Williams-Beuren Region	ASR	–	LPU 024-A	121
7p11.2	EGFR Amplification	ASR	D7Z1	LPS 003-A	93
7q11.23	Williams-Beuren Region	ASR	D7Z1	LPU 011-A ^{††}	125
7q11.23/7p21.1	Saethre-Chotzen Region/Williams-Beuren Region	ASR	–	LPU 024-A	121
7q22	CUX1 (7q22)	ASR	–	LPH 543-A	31
7q22/7q31.2	Del(7q) Deletion	IVD	–	USA-LPH 025*	15
7q22/7q31.2	Del(7q) Deletion	ASR	–	LPH 025-A [†]	34
7q31.2/7q22	Del(7q) Deletion	IVD	–	USA-LPH 025*	15
7q31.2/7q22	Del(7q) Deletion	ASR	–	LPH 025-A [†]	34
7q31.2	C-MET (MET) Amplification	ASR	D7Z1	LPS 004-A	93
7q34	TCRB (TRB) Breakapart	ASR	–	LPH 048-A	64
7q36	EZH2 (7q36) Probe Red	ASR	–	LPH 544-A	38
8p11.23-p11.22	FGFR1 Breakapart/Amplification	ASR	D8Z2	LPS 018-A	101
8q21.3/21q22.1	AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual Fusion	IVD	–	USA-LPH 026*	12
8q21.3/21q22.1	AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual Fusion	ASR	–	LPH 026-A [†]	21
8q24.21	cMYC (MYC) Breakapart	ASR	–	LPH 010-A	29
8q24.21	cMYC (MYC) <i>Plus</i>	ASR	–	LPH 523-A	44
8q24.21	MYC Breakapart (Hematopathology)	ASR	–	LPS 027-A	82
8q24.21/14q32.33	IGH/MYC Translocation, Dual Fusion (Hematopathology)	ASR	–	LPS 035-A	80
9p21	CDKN2A Probe Gold	ASR		LPS 532 - A ^{††}	90
9p21.3	P16 (CDKN2A) Deletion (Hematopathology)	ASR	D9Z3	LPS 036-A	83
9p21.3	P16 (CDKN2A) Deletion	ASR	D9Z3	LPH 009-A	52
9p21.3	MLLT3	ASR	–	LPH 509-A	50
9q12	Chromosome 9 Satellite III probe aqua	ASR	–	LPH 547-A	62
9q34.11-q34.12/22q11.22-q11.23	BCR/ABL (ABL1) Translocation, Dual Fusion	ASR	–	LPH 007-A	23
9q34.11-q34.12/22q11.22-q11.23	BCR/ABL (ABL1) <i>Plus</i> Translocation, Dual Fusion	ASR	–	LPH 038-A	24
9q34.12-q34.13	NUP214 Probe Red	ASR	–	LPH 530-A	69
10p14	DiGeorge II (10p14)	ASR	D10Z1	LPU 015-A ^{††}	115
10q11.21	RET Proximal	ASR	–	LPS 508-A	104
10q11.21	RET Distal	ASR	–	LPS 509-A	105
10q24.31	TLX1 Breakapart	ASR	–	LPH 049-A	66
11p15.4	NUP98 Proximal Red	ASR	–	LPH 532-A	51
11p15.4	NUP98 Distal Green	ASR	–	LPH 533-A	52

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

† For sale in Canada only.

†† See product page/website for availability in your region.



Index by Chromosome

Chromosome Region	Probe Name	Regulatory Status	Control Region	Cat. No.	Page
11q13.3	CCND1 Breakapart (Hematopathology)	ASR	–	LPS 030-A	76
11q13.3	CCND1 <i>Plus</i>	ASR	–	LPH 519-A	43
11q13.3/14q32.3	IGH/CCND1 Translocation, Dual Fusion (Hematopathology)	ASR	–	LPS 031-A	78
11q13.3	MYEOV <i>Plus</i>	ASR	–	LPH 525-A	46
11q22.3	ATM Deletion	ASR	D11Z1	LPH 011-A	22
11q22.3/17p13	P53 (TP53)/ATM Probe Combination	ASR	–	LPH 052-A	55
11q23.3	MLL (KMT2A) Breakapart	ASR	–	LPH 013-A [†]	48
11q23.3	MLL (KMT2A) Breakapart	IVD	–	USA-LPH 013*	18
11q23.3	MLL (KMT2A)	ASR	–	LPH 506-A	48
11q23.3	MLL (KMT2A) Distal Probe Red	ASR	–	LPH 570-A ^{††}	–
11q23.3	MLL (KMT2A) Proximal Probe Green	ASR	–	LPH 569-A ^{††}	–
11q24.3/22q12.1-q12.2	FLI1/EWSR1 Translocation, Dual Fusion	ASR	–	LPS 007-A	100
12p11.1-q11.1	Alpha Satellite 12 <i>Plus</i>	ASR	–	LPH 069-A	71
12p13.2	ETV6 Proximal Probe Green	ASR	–	LPD 501-A	95
12p13.2	ETV6 Distal Probe Red	ASR	–	LPD 502-A	95
12p13.2/21q22.1	TEL/AML1 (ETV6/RUNX1) Translocation, Dual Fusion	ASR	–	LPH 012-A	65
12q13.3	CHOP (DDIT3) Breakapart	ASR	–	LPS 015-A	91
12q15	MDM2 Amplification	ASR	D12Z1	LPS 016-A	102
13q14.1	FOXO1 Proximal Green	ASR	–	LPS 518-A	98
13q14.1	FOXO1 Distal Red	ASR	–	LPS 519-A	98
13q14.2-q14.3	13q14.3 Deletion	ASR	D13S1825	LPH 006-A	31
13q14.2-14.3	D13S319 <i>Plus</i> Deletion	ASR	LAMP1	LPH 068-A	32
13q14.2	RB1 Deletion	ASR	LAMP1	LPS 011-A	84
13q14.3	D13S25 Deletion	ASR	D13S1825	LPH 043-A	33
14q11.2	TCRAD Breakapart	ASR	–	LPH 047-A	63
14q32.13-q32.2	TCL1 Breakapart	ASR	–	LPH 046-A	63
14q32.3	IGH Breakapart	ASR	–	LPH 014-A	40
14q32.3	IGH Breakapart (Hematopathology)	ASR	–	LPS 032-A	76
14q32.3	IGH <i>Plus</i>	ASR	–	LPH 517-A	41
14q32.3	IGH Proximal <i>Plus</i>	ASR	–	LPH 515-A	40
14q32.3	IGH Distal <i>Plus</i>	ASR	–	LPH 516-A	41
14q32.3	IGH Probe Green	ASR	–	LPH 568-A	42
14q32.3/11q13.3	IGH/CCND1 Translocation, Dual Fusion (Hematopathology)	ASR	–	LPS 031-A	78
14q32.3/18q21.31-q21.32	IGH/MALT1 Translocation, Dual Fusion (Hematopathology)	ASR	–	LPS 034-A	79
14q32.3/18q21.33-q22.1	IGH/BCL2 Translocation, Dual Fusion (Hematopathology)	ASR	–	LPS 033-A	77
14q32.3/8q24.21	IGH/MYC Translocation, Dual Fusion (Hematopathology)	ASR	–	LPS 035-A	80
15p11.1-15q11.1	Chromosome 15 Alpha Satellite Probe Red	ASR	–	LPH 548-A	71
15q11.2	Prader-Willi/Angelman (SNRPN) Region	ASR	15qter	LPU 005-A	120
15q11.2-q12	Angelman (UBE3A/D15S10) Region	ASR	15qter	LPU 006-A ^{††}	113
15q24.1	FAST PML	ASR	–	LPH 501-A	57

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

[†] For sale in Canada only.

^{††} See product page/website for availability in your region.

Index by Chromosome

Chromosome Region	Probe Name	Regulatory Status	Control Region	Cat. No.	Page
15q24.1/17q21.1-q21.2	PML/RAR α (RARA) Translocation, Dual Fusion	ASR	–	LPH 023-A	58
16p11.2	FUS Proximal Red	ASR	–	LPS 520-A	99
16p11.2	FUS Distal Green	ASR	–	LPS 521-A	99
16p13.1/16q22	CBF β /MYH11 Translocation, Dual Fusion	ASR	–	LPH 022-A [†]	27
16p13.1/16q22	CBF β /MYH11 Translocation, Dual Fusion	IVD	–	USA-LPH 022*	13
16p13.1/16q22	CBF β /MYH11 Translocation, Dual Fusion	ASR	–	LPH 022-A [†]	27
16q22	CBFB Proximal Probe Red	ASR	–	LPH 526-A	26
16q22	CBFB Distal Probe Green	ASR	–	LPH 527-A	26
16q22/16p13.1	CBF β /MYH11 Translocation, Dual Fusion	ASR	–	LPH 022-A [†]	27
16q22/16p13.1	CBF β /MYH11 Translocation, Dual Fusion	IVD	–	USA-LPH 022*	13
16q23	MAF v2 Probe Red	ASR	–	LPH 567-A	45
17p11.1-q11.1	Centromere 17 Aqua	ASR	–	LPS 528-A ^{††}	91
17p11.1-q11.1	Centromere 17 Probe Green	ASR	–	LPH 572-A ^{††}	–
17p11.2/17p13.3	Smith-Magenis (RAI1) Region/Miller-Dieker Region	ASR	–	LPU 019-A	123
17q12	HER2 (ERBB2) Amplification	ASR	–	LPS 001-A	101
17p13	P53 (TP53) Deletion	IVD	D17Z1	USA-LPH 017*	19
17p13	P53 (TP53) Deletion	ASR	D17Z1	LPH 017-A [†]	53
17p13	P53 (TP53) Deletion	ASR	D17Z1	LPS 037-A	83
17p13	P53 (TP53) Probe Red	ASR	–	LPH 571-A ^{††}	–
17p13	TP53 Probe Green	ASR	–	LPD 504-A	68
17p13/11q22.3	P53(TP53)/ATM Probe Combination	ASR	–	LPH 052-A	55
17p13.3/17p11.2	Smith-Magenis (RAI1) Region/Miller-Dieker Region	ASR	–	LPU 019-A	123
17q21.1-q21.2	FAST RAR α (RARA)	ASR	–	LPH 502-A	57
17q21.1-q21.2	RAR α (RARA) Proximal	ASR	–	LPH 062-A	59
17q21.2	RAR α (RARA) Distal	ASR	–	LPH 063-A	59
17q21.1-q21.2/15q24.1	PML/RAR α (RARA) Translocation, Dual Fusion	ASR	–	LPH 023-A	58
17q21.2	TOP2A Amplification/Deletion	ASR	D17Z1	LPS 002-A	109
17q22	HLF	ASR	–	LPH 505-A	37
17q22	MPO Probe Red	ASR	–	LPD 503-A	68
18q11.2	SYT (SS18) Breakapart	ASR	–	LPS 014-A	107
18q21.31-q21.32	MALT1 Breakapart	ASR	–	LPS 017-A	82
18q21.31-q21.32/14q32.3	IGH/MALT1 Dual Fusion	ASR	–	LPS 034-A	79
18q21.33-q22.1	BCL2 Breakapart (Hematopathology)	ASR	–	LPS 028-A	75
18q21.33	BCL2 <i>Plus</i>	ASR	–	LPH 518-A	42
18q21.33-q22.1/14q32.3	IGH/BCL2 Translocation, Dual Fusion (Hematopathology)	ASR	–	LPS 033-A	77
19p13.2	19p13	ASR	–	LPS 515-A	89
19p13.3	MLLT1	ASR	–	LPH 508-A	49
19p13.3	E2A (TCF3) Breakapart	ASR	–	LPH 019-A	35
19p13.3	E2A (TCF3)	ASR	–	LPH 503-A	36
19q13.33	19q13	ASR	–	LPS 516-A	89
20q12	MAFB <i>Plus</i>	ASR	–	LPH 524-A	45

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

[†] For sale in Canada only.

^{††} See product page/website for availability in your region.



Index by Chromosome

Chromosome Region	Probe Name	Regulatory Status	Control Region	Cat. No.	Page
20q12/20q13.1	Del(20q) Deletion	IVD	–	USA-LPH 020*	16
20q12/20q13.1	Del (20q) Deletion	ASR	–	LPH 020-A [†]	34
20q13.1/20q12	Del(20q) Deletion	IVD	–	USA-LPH 020*	16
20q13.1/20q12	Del(20q) Deletion	ASR	–	LPH 020-A [†]	34
20q13.2/20p13	ZNF217 Amplification	ASR	DEFB128	LPS 005-A	110
21q22.1	AML1 (RUNX1) Breakapart	ASR	–	LPH 027-A [†]	20
21q22.1/12p13.2	TEL/AML1 (ETV6/RUNX1) Translocation, Dual Fusion	ASR	–	LPH 012-A	65
21q22.1	RUNX1 Probe Green	ASR	–	LPH 529-A	60
21q22.1 / 8q21.3	AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual Fusion	ASR	–	LPH 026-A [†]	21
21q22.1 / 8q21.3	AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual Fusion	IVD	–	USA-LPH 026*	12
21q22.1	AML (RUNX1) Distal Probe Green	ASR	–	LPH 573-A ^{††}	–
21q22.1	AML (RUNX1) Proximal Probe Red	ASR	–	LPH 574-A ^{††}	–
21q22.13-q22.2	ERG Control	ASR	–	LPS 099-A	94
21q22.13-q22.2/22q12.1-q12.2	EWSR1/ERG Translocation, Dual Fusion	ASR	–	LPS 008-A	97
21q22.2-q22.3	TMPRSS2 Breakapart	ASR	–	LPS 098-A	108
22q11.21-q11.23	IGL Breakapart	ASR	–	LPH 033-A	47
22q11.21-q11.23	IGL Breakapart (Hematopathology)	ASR	–	LPS 039-A	81
22q11.2/22q13.3	DiGeorge TBX1 Region & 22q13.3 Region	ASR	–	LPU 014-A ^{††}	118
22q11.2/22q13.3	DiGeorge/VCFS N25 Region & 22q13.3 Region	ASR	–	LPU 010-A ^{††}	117
22q11.2/22q13.3	DiGeorge/VCFS TUPLE1 Region & 22q13.3 Region	ASR	–	LPU 004-A	116
22q11.22-q11.23/9q34.11-q34.12	BCR/ABL (ABL1) Translocation, Dual Fusion	ASR	–	LPH 007-A	23
22q11.22-q11.23/9q34.11-q34.12	BCR/ABL (ABL1) Plus Translocation, Dual Fusion	ASR	–	LPH 038-A	24
22q12.1-q12.2	EWSR1 Breakapart	ASR	–	LPS 006-A	96
22q12.1-q12.2/11q24.3	FLI1/EWSR1 Translocation, Dual Fusion	ASR	–	LPS 007-A	100
22q12.1-q12.2/21q22.13-q22.2	EWSR1/ERG Translocation, Dual Fusion	ASR	–	LPS 008-A	97
22q13.3/22q11.2	DiGeorge TBX1 Region & 22q13.3 Region	ASR	–	LPU 014-A ^{††}	118
22q13.3/22q11.2	DiGeorge/VCFS N25 Region & 22q13.3 Region	ASR	–	LPU 010-A ^{††}	117
22q13.3/22q11.2	DiGeorge/VCFS TUPLE1 Region & 22q13.3 Region	ASR	–	LPU 004-A	116
Xp11.1-q11.1/Yp11.1-q11.1	Dual labeled Satellite Probe Set XYc	ASR	–	LPE 0XYc-A	130
Xp11.1-q11.1/Yq12	Dual labeled Satellite Probe Set XYq	ASR	–	LPE 0XYq-A	130
Xp11.23	TFE3 Proximal Probe Red	ASR	–	LPS 522-A	108
Xp11.23	TFE3 Distal Probe Green	ASR	–	LPS 523-A	109
Xp22.31	Kallmann (KAL1) Region/STS Region	ASR	DXZ1	LPU 016-A ^{††}	119
Xp22.33/Yp11.32	CRLF2 Distal	ASR	–	LPH 511-A	30
Xp22.33/Yp11.32	CRLF2 Proximal	ASR	–	LPH 512-A	30
Xp22.33/Yp11.32	SHOX	ASR	DXZ1, DYZ1	LPU 025-A	122
Xp22.33/Yp11.32	P2RY8 Distal	ASR	–	LPH 513-A	54
Xp22.33/Yp11.32	P2RY8 Proximal	ASR	–	LPH 514-A	54
Yp11.1-q11.1/Xp11.1-q11.1	Dual labeled Satellite Probe Set XYc	ASR	–	LPE 0XYc-A	130

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

[†] For sale in Canada only.

^{††} See product page/website for availability in your region.

Index by Chromosome

Chromosome Region	Probe Name	Regulatory Status	Control Region	Cat. No.	Page
Yp11.31	SRY	ASR	DXZ1, DYZ1	LPU 026-A	124
Yp11.32/Xp22.33	CRLF2 Distal	ASR	–	LPH 511-A	30
Yp11.32/Xp22.33	CRLF2 Proximal	ASR	–	LPH 512-A	30
Yp11.32/Xp22.33	SHOX	ASR	DXZ1, DYZ1	LPU 025-A	122
Yp11.32/Xp22.33	P2RY8 Distal	ASR	–	LPH 513-A	54
Yp11.32/Xp22.33	P2RY8 Proximal	ASR	–	LPH 514-A	54
Yq12/Xp11.1-q11.1	Dual labeled Satellite Probe Set XYq	ASR	–	LPE 0XYq-A	130
Various	Acro-P-Arm Probe	ASR	–	LPE NOR-A	130

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

† For sale in Canada only.

†† See product page/website for availability in your region.





Index by Gene Name

HGNC Gene Name	Chromosome Region	Product Name	Cat. No.
ABL1	9q34.11-q34.12	BCR/ABL (ABL1) Translocation, Dual Fusion	LPH 007-A
		BCR/ABL (ABL1) <i>Plus</i> Translocation, Dual Fusion	LPH 038-A
AFDN (MLLT4)	6q27	MLLT4 (AFDN)	LPH 510-A
AFF1	4q21.3-q22.1	AFF1	LPH 507-A
ALK	2p23.2-p23.1	ALK Breakapart	LPS 019-A
ANGPTL1	1q25.2	1q25	LPS 512-A
ANOS1	Xp22.31	Kallmann (KAL1) Region/STS Region	LPU 016-A ^{††}
ASS1	9q34.11	BCR/ABL (ABL1) Translocation, Dual Fusion	LPH 007-A
ATM	11q22.3	ATM Deletion	LPH 011-A
		P53 (TP53)/ATM Probe Combination	LPH 052-A
BCL2	18q21.33-q22.1	BCL2 Breakapart (Hematopathology)	LPS 028-A
		BCL2 <i>Plus</i>	LPH 518-A
		IGH/BCL2 Translocation, Dual Fusion (Hematopathology)	LPS 033-A
BCL6	3q27.3	BCL6 Breakapart	LPH 035-A
	3q27.3-q28	BCL6 Breakapart (Hematopathology)	LPS 029-A
BCR	22q11.22-q11.23	BCR/ABL (ABL1) Translocation, Dual Fusion	LPH 007-A
		BCR/ABL (ABL1) <i>Plus</i> Translocation, Dual Fusion	LPH 038-A
	22q11.21-q11.23	IGL Breakapart (Hematopathology)	LPS 039-A
CBFB	16q22	IGL Breakapart	LPH 039-A
		CBFB/MYH11 Translocation, Dual Fusion [IVD]	USA-LPH 022*
		CBFB/MYH11 Translocation, Dual Fusion	LPH 022-A [†]
		CBFB Proximal Probe Red	LPH 526-A
CCND1	11q13.3	CBFB Distal Probe Green	LPH 527-A
		CCND1 Breakapart (Hematopathology)	LPS 030-A
		CCND1 <i>Plus</i>	LPH 519-A
CCND3	6p21	IGH/CCND1 Translocation, Dual Fusion (Hematopathology)	LPS 031-A
		CCND3 <i>Plus</i>	LPH 522-A
CDKN2A	9p21.3	P16 (CDKN2A) Deletion	LPH 009-A
		P16 (CDKN2A) Deletion (Hematopathology)	LPS 036-A
	9p21	CDKN2A Probe Gold	LPS 532-A ^{††}
CDKN2C	1p32.3	CKS1B/CDKN2C(P18) Amplification/Deletion	LPH 039-A
CELF2	10p14	DiGeorge II (10p14)	LPU 015-A ^{††}
CHD5	1p36.31	SRD (CHD5) Deletion	LPS 010-A
CHIC2	4q12	FIP1L1/CHIC2/PDGFR A Deletion/Fusion	LPH 032-A
CKS1B	1q21.3	CKS1B/CDKN2C(P18) Amplification/Deletion	LPH 039-A
CRLF2	Yp11.32/Xp22.33	CRLF2 Distal	LPH 511-A
		CRLF2 Proximal	LPH 512-A
CUX1	7q22	CUX1 (7q22) Probe Green	LPH 543-A
CTNND2	5p15.2	Cri-du-chat Region/Sotos Region	LPU 013-A ^{††}
DLEU1	13q14.2-14.3	D13S319 <i>Plus</i> Deletion	LPH 068-A
		13q14.3 Deletion	LPH 006-A
DLEU2	13q14.2-14.3	D13S319 <i>Plus</i> Deletion	LPH 068-A
		13q14.3 Deletion	LPH 006-A

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

[†] For sale in Canada only.

^{††} See product page/website for availability in your region.



Index by Gene Name

HGNC Gene Name	Chromosome Region	Product Name	Cat. No.
DLEU7	13q14.3	D13S25 Deletion	LPH 043-A
DDIT3	12q13.3	CHOP (DDIT3) Breakapart	LPS 015-A
DEK	6p22.3	DEK Probe Green	LPH 531-A
EGFR	7p11.2	EGFR Amplification	LPS 003-A
EGR1	5q31.2	Del(5q) Deletion [IVD]	USA-LPH 024*
		Del(5q) Deletion	LPH 024-A†
	5q31	EGR1/CDC25C (5q31) Probe Green	LPH 541-A
ELN	7q11.23	Williams-Beuren Region	LPU 011-A††
	7q11.23	Saethre-Chotzen Region/Williams-Beuren Region	LPU 024-A
EML4	2p21	EML4 Breakapart	LPS 020-A
ERBB2	17q12	HER2 (ERBB2) Amplification	LPS 001-A
ERG	21q22.13-q22.2	ERG Control	LPS 099-A
		EWSR1/ERG Translocation, Dual Fusion	LPS 008-A
ETV6	12p13.2	ETV6 Proximal Probe Green	LPD 501-A
		ETV6 Distal Probe Red	LPD 502-A
		TEL/AML1 (ETV6/RUNX1) Translocation, Dual Fusion	LPH 012-A
EWSR1	22q12.1-q12.2	EWSR1 Breakapart	LPS 006-A
		FLI1/EWSR1 Translocation, Dual Fusion	LPS 007-A
		EWSR1/ERG Translocation, Dual Fusion	LPS 008-A
EZH2	7q36	EZH2 (7q36)	LPH 544-A
FGFR1	8p11.23-p11.22	FGFR1 Breakapart/Amplification	LPS 018-A
FGFR3	4p16.3	FGFR3 <i>Plus</i>	LPH 521-A
		Wolf-Hirschhorn Region	LPU 009-A††
FIP1L1	4q12	FIP1L1/CHIC2/PDGFRA Deletion/Fusion	LPH 032-A
FLI1	11q24.3	FLI1/EWSR1 Translocation, Dual Fusion	LPS 007-A
FOXO1	13q14.1	FOXO1 Proximal Probe Green	LPS 518-A
		FOXO1 Distal Probe Red	LPS 519-A
FUS	16p11.2	FUS Proximal Probe Red	LPS 520-A
		FUS Distal Probe Green	LPS 521-A
GLTSCR1/GLTSCR2	19q13.33	19q13	LPS 516-A
HIRA	22q11.2	DiGeorge/VCFS TUPLE1 Region & 22q13.3 Region	LPU 004-A
HLF	17q22	HLF	LPH 505-A
IGH	14q32.3	IGH Breakapart	LPH 014-A
		IGH Probe Green	LPH 568-A
		IGH Proximal <i>Plus</i>	LPH 515-A
		IGH Distal <i>Plus</i>	LPH 516-A
		IGH Breakapart (Hematopathology)	LPS 032-A
		IGH <i>Plus</i>	LPH 517-A
		IGH/BCL2 Translocation, Dual Fusion (Hematopathology)	LPS 033-A
		IGH/CCND1 Translocation, Dual Fusion (Hematopathology)	LPS 031-A
		IGH/MALT1 Translocation, Dual Fusion (Hematopathology)	LPS 034-A
		IGH/MYC Translocation, Dual Fusion (Hematopathology)	LPS 035-A
IGK	2p11.2	IGK Breakapart	LPH 034-A
		IGK Breakapart (Hematopathology)	LPS 038-A

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

† For sale in Canada only.

†† See product page/website for availability in your region.





Index by Gene Name

HGNC Gene Name	Chromosome Region	Product Name	Cat. No.
IGL	22q11.21-q11.23	IGL Breakapart	LPH 033-A
		IGL Breakapart (Hematopathology)	LPS 039-A
KMT2A	11q23.3	MLL (KMT2A) Breakapart [IVD]	USA-LPH 013*
		MLL (KMT2A) Breakapart	LPH 013-A†
		MLL (KMT2A)	LPH 506-A
		MLL (KMT2A) Distal Probe Red	LPH 570-A††
		MLL (KMT2A) Proximal Probe Green	LPH 569-A††
MAF	16q23	MAF v2 Probe Red	LPH 567-A
MAFB	20q12	MAFB <i>Plus</i>	LPH 524-A
MALT1	18q21.32	MALT1 Breakapart	LPS 017-A
		IGH/MALT1 Translocation, Dual Fusion (Hematopathology)	LPS 034-A
MAPK1	22q11.21-q11.23	IGL Breakapart	LPH 033-A
		IGL Breakapart (Hematopathology)	LPS 039-A
MDM2	12q15	MDM2 Amplification	LPS 016-A
MECOM	3q26.2	EVI1 (MECOM) Breakapart [IVD]	USA- LPH 036*
		EVI1 (MECOM) Breakapart	LPH 036-A†
		MECOM Probe Red	LPH 528-A
MET	7q31.2	C-MET (MET) Amplification	LPS 004-A
MLLT1	19p13.3	MLLT1	LPH 508-A
MLLT3	9p21.3	MLLT3	LPH 509-A
MLLT4 (AFDN)	6q27	MLLT4 (AFDN)	LPH 510-A
MPO	17q22	MPO Probe Red	LPD 503-A
MYB	6q23.3	MYB Deletion	LPH 016-A
MYBL2	20q13.1	Del(20q) Deletion [IVD]	USA-LPH 020*
		Del(20q) Deletion	LPH 020-A†
MYC	8q24.21	cMYC (MYC) Breakapart	LPH 010-A
		MYC Breakapart (Hematopathology)	LPS 027-A
		cMYC (MYC) <i>Plus</i>	LPH 523-A
		IGH/MYC Translocation, Dual Fusion (Hematopathology)	LPS 035-A
MYCN	2p24.3	N-MYC (MYCN) Amplification	LPS 009-A
MYEOV	11q13.3	MYEOV <i>Plus</i>	LPH 525-A
MYH11	16p13.11	CBFβ/MYH11 Translocation, Dual Fusion [IVD]	USA -LPH 022*
		CBFβ/MYH11 Translocation, Dual Fusion	LPH 022-A†
NSD1	5q35.2-q35.3	Cri-Du-Chat Region/ and Sotos Region	LPU 013-A††
NUP98	11p15.4	NUP98 Proximal Probe Red	LPH 532-A
		NUP98 Distal Probe Green	LPH 533-A
NUP214	9q34.13	NUP214 Probe Red	LPH 530-A
P2RY8	Yp11.32/Xp22.33	P2RY8 Distal red	LPH 513-A
		P2RY8 Proximal green	LPH 514-A
PAX3	2q36.1	PAX3 Breakapart	LPS 012-A
PAX7	1p36.13	PAX7 Breakapart	LPS 013-A
PBX1	1q23.3	PBX1	LPH 504-A
PDGFRA	4q12	FIP1L1/CHIC2/PDGFRA Deletion/Fusion	LPH 032-A
PDGFRB	5q32	PDGFRB Breakapart	LPH 031-A

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

† For sale in Canada only.

†† See product page/website for availability in your region.



Index by Gene Name

HGNC Gene Name	Chromosome Region	Product Name	Cat. No.
PML	15q24.1	FAST PML	LPH 501-A
		PML/RARA (RARA) Translocation, Dual Fusion	LPH 023-A
RAI1	17p11.2	Smith-Magenis (RAI1) /Miller-Dieker Region	LPU 019-A
RARA	17q21.1-q21.2	FAST RARα (RARA)	LPH 502-A
		RARα (RARA) Proximal Red	LPH 062-A
		RARα (RARA) Distal Green	LPH 063-A
		PML/RARA (RARA) Translocation, Dual Fusion	LPH 023-A
RB1	13q14.2	RB1 Deletion	LPS 011-A
RELN	7q22	Del(7q) Deletion [IVD]	USA-LPH 025*
		Del(7q) Deletion	LPH 025-A†
RET	10q11.21	RET Proximal Green	LPS 508-A
		RET Distal Red	LPS 509-A
ROS1	6q22.1	ROS1 Breakapart	LPS 022-A
		ROS1-GOPC (FIG) Proximal Green	LPS 510-A
		ROS1-GOPC (FIG) Distal Red	LPS 511-A
RUNX1	21q22.1	AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual Fusion [IVD]	USA-LPH 026*
		AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual Fusion	LPH 026-A†
		AML1 (RUNX1) Breakapart	LPH 027-A†
		AML1 (RUNX1) Distal Probe Green	LPH 573-A††
		AML1 (RUNX1) Proximal Probe Red	LPH 574-A††
		RUNX1 Probe Green	LPH 529-A
		TEL/AML1 (ETV6/RUNX1) Translocation, Dual Fusion	LPH 012-A
RUNX1T1	8q21.3	AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual Fusion [IVD]	USA-LPH 026*
		AML1/ETO (RUNX1/RUNX1T1) Translocation, Dual Fusion	LPH 026-A†
SHOX	Xp22.33/Yp11.32	SHOX	LPU 025-A
SNRPN	15q11.2	Prader-Willi/Angelman (SNRPN) Region	LPU 005-A
SRY	Yp11.31	SRY	LPU 026-A
SS18	18q11.2	SYT (SS18) Breakapart	LPS 014-A
STS	Xp22.31	Kallmann (KAL1) Region/STS Region	LPU 016-A††
TAS2R1	5p15.3	TAS2R1 (5p15.31) Probe Green	LPH 546-A
TBX1	22q11.21/22q13.33	DiGeorge TBX1 Region & 22q13.3 Deletion Region	LPU 014-A††
TCF3	19p13.3	E2A (TCF3) Breakapart	LPH 019-A
		E2A (TCF3)	LPH 503-A
TCL1A/TCL1B	14q32.13-q32.2	TCL1 Breakapart	LPH 046-A
TERT	5p15.33	TERT (5p15.33) Probe Aqua	LPH 542-A
TES	7q22/7q31.2	Del(7q) Deletion [IVD]	USA-LPH 025*
		Del(7q) Deletion	LPH 025-A†
TET2	4q24	TET2 Probe Red	LPH 534-A
TFE3	Xp11.23	TFE3 Proximal Probe Red	LPS 522-A
	Xp11.23	TFE3 Distal Probe Green	LPS 523-A
TLX1	10q24.31	TLX1 Breakapart	LPH 049-A
TLX3	5q35.1	TLX3 Breakapart	LPH 050-A
TMPRSS2	21q22.2-q22.3	TMPRSS2 Breakapart	LPS 098-A
TOP2A	17q21.2	TOP2A Amplification/Deletion	LPS 002-A

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

† For sale in Canada only.

†† See product page/website for availability in your region.





Index by Gene Name

HGNC Gene Name	Chromosome Region	Product Name	Cat. No.
TP53	17p13	P53 (TP53) Deletion [IVD]	USA-LPH 017*
		P53 (TP53) Deletion	LPH 017-A†
		P53 (TP53) Deletion (Hematopathology)	LPS 037-A
		P53 (TP53)/ATM Probe Combination	LPH 052-A
		TP53 Probe Green	LPD 504-A
		P53 (TP53) Probe Red	LPH 571-A††
TP73	1p36.32	1p36	LPS 513-A
TRA (TCRA)	14q11.2	TCRAD Breakapart	LPH 047-A
TRB (TCRB)	7q34	TCRB (TRB) Breakapart	LPH 048-A
TRD (TCRD)	14q11.2	TCRAD Breakapart	LPH 047-A
TWIST1	7p21.1/7q11.23	Saethre-Chotzen Region/Williams-Beuren Region	LPU 024-A
UBE2QL1	5p15.31	Cri-du-Chat and SOTOS Region	LPU 013-A††
UBE3A	15q11.2	Angelman (UBE3A/D15S10) Region	LPU 006-A††
USP46	4q12	USP46 (4q12) Probe Green	LPH 535-A
ZNF217	20p13/ 20q13.2	ZNF217 Amplification	LPS 005-A
ZNF443	19p13.2	19p13	LPS 515-A

* FDA-cleared, Class II IVD FISH Probe Kits for AML/MDS. For sale in the US only. These products have not been licensed in accordance with Canadian law.

† For sale in Canada only.

†† See product page/website for availability in your region.





Notes



Helpful Links

OGT's team of experts share their knowledge and expertise in regularly updated Tips and Tricks for NGS, FISH and arrays. Bookmark these pages for the latest advice.

Fish 'n' Tips:
www.ogt.com/FISH-tips



NGS Success:
www.ogt.com/NGS-tips



Array Advice:
www.ogt.com/array-tips





A Sysmex Group Company

Oxford Gene Technology Inc

520 White Plains Road, Suite 500, Tarrytown, NY 10591, USA

T: 914-467-5285

www.ogt.com

For FISH probe orders:

T: 1-833-208-4OGT (4648)

F: 1-833-824-6514

E: OGTorders@sysmex.com

www.cytocell-us.com