Tests Available
Cytogenetics Laboratory

Cincinnati Children's
Human Genetics

Microarray - SNP

Prenatal
- Amniocentesis
- Chorionic Villi Sampling

Constitutional
- Peripheral Blood
- Products of Conception

Oncology
- Blood
- Bone Marrow

Chromosome Analysis

Prenatal
- Amniocentesis
- Chorionic Villi Sampling

Constitutional
- Peripheral Blood
  - Routine
  - High Resolution
- Products of Conception

Oncology
- Blood
- Bone Marrow
- Lymph Node
- Tumor

Fanconi Breakage Study
- Blood
- Bone Marrow
- Fibroblast

Sister Chromatid Exchange (SCE) Bloom Syndrome
- Blood

FISH: Constitutional/ Prenatal

1p36 Microdeletion Syndrome
Alagille Syndrome: JAG1 (20p12)
Aneuploidy 13 [Trisomy 13]
Aneuploidy 18 [Trisomy 18]
Cri-Du-Chat Syndrome: D5S23, D5S721 (5p15.2)
DiGeorge/VCF Syndrome: TBX1 (22q11.2)
Down Syndrome [Trisomy 21]
Kallmann Syndrome: KAL (Xp22.3)
Miller-Dieker Syndrome: LIS1 (17p13.3)
Prader-Willi/Angelman Syndrome: SNRPN (15q11-q13)
Retinoblastoma: RB1 (13q14)
SHOX (Xp22/Yp11.3)
Smith-Magenis Syndrome: RAI1 (17p11.2)
Sotos Syndrome: NSD1 (5q35)
SRY (Yp11.3)
Steroid Sulfatase Deficiency: STS (Xp22.3)
Williams Syndrome: ELN (7q11.23)
Wolf-Hirschorn Syndrome: WHS (4p16.3)
X/Y Centromeres [Sex Determination]
X-Inactivation: XIST (Xq13)

Aneuploidy FISH Panel: 13, 18, 21, X, Y
[Prenatal/Constitutional]

Visit our website for test specifics: **www.cincinnatichildrens.org/cytogenetics**
Questions? Ask our genetic counselors!
Phone: (513) 636-4474 • Fax: (513) 636-4373 • Email: cytogenetics@cchmc.org

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Oncology FISH: Panels

ALL Hyperdiploid: Trisomy 4, 10, 17
ALL Risk Stratification: Trisomy 4, 10, 17; t(12;21); t(9;22); KMT2A (11q23)
AML: t(8;21); KMT2A (11q23); CBFB (16q22)
APL: t(15;17); RARA (17q21)
Burkitt Lymphoma: t(8;14); MYC (8q24)
CLL: D13S319 (13q14.3)/LAMP1 (13q34)/CEP 12; ATM (11q22.3); TP53(17p13)
Small B-Cell NHL: CLL panel; t(11;14); t(14;18); MALT1 (18q21)
Large Cell NHL: ALK (2p23.2); t(11;14); t(14;18); TP53 (17p13); BCL6 (3q27)
Combined NHL: Large and Small Cell NHL Panels
Eosinophilia: 4q12 (SCFD2,FIP1L1/LNX,PDGFRA/KIT); PDGFRB (5q32); FGFR1 (8p11); CBFB (16q22)
Fanconi Anemia: 1p36/1q25 (TP73/ABL2); BCL6 (3q27); mono 7/del 7q
Multiple Myeloma (CD138+ sorted cells): 1p32.3/1q21; t(4;14); t(11;14); mono 13/del 13q14.3; t(14;16); t(14;20); TP53 (17p13)
Myeloid Disorder (MDS): mono 5/del 5q; mono 7/del 7q; tri 8; del 20q
Shwachman-Diamond Syndrome (SDS): mono 7/del 7q; tri 8; del 20q
Myeloproliferative Disorder (MPD): 4q12 (SCFD2,LNX, PDGFRA/KIT); PDGFRB (5q32); FGFR1 (8p11), t(9;22)
T-Cell: TRAD (14q11.2); TRB (7q34); TRG (7p14); t(9;22), KMT2A (11q23)

All tests listed in panels are available as individual tests

Oncology FISH: Individual Probes

t(4;14)	LSI IGH (14q32)/FGFR3 (4p16) (Dual Fusion)*
t(8;14)	LSI IGH (14q32)/ MYC (8q24)/CEP 8 (Dual Fusion)*
t(8;21)	LSI RUNX1T1 (8q21.3)/LSI RUNX1 (21q22) (Dual Fusion)*
t(9;22)	LSI BCR (22q11.2)/ABL1 (9q34) (Dual Fusion)*
t(11;14)	LSI IGH (14q32)/CCND1 (11q13) XT (Dual Fusion)*
t(11;18)	LSI BIRC3 (11q21)/MALT1 (18q21) (Dual Fusion)*
t(12;21)	LSI ETV6 (12p13)/LSI RUNX1 (21q22) (ES Fusion)*
t(14;16)	LSI IGH (14q32)/MAF (16q32) (Dual Fusion)*
t(14;18)	LSI IGH (14q32)/LSI BCL2 (18q21) (Dual Fusion)*
t(14;18)	LSI IGH (14q32)/LSI MALT1 (18q21) (Dual Fusion)*
t(14;20)	IGH (14q32.3)/MAFB (20q12) (Dual Fusion)*
t(15;17)	LSI PML (15q22)/RARA (17q21) (Dual Fusion)*

Oncology FISH: Individual Probes, cont.

1	LSI 1p36 (MEGF6, TP73)/LSI 1q25 (ABL2)
2	MYCN (2p24.1)/AFF3 (2q11) (Amplification) ALK (2p23) (Break Apart)*
3	EVI1 (3q26) (TC BA Probe)* LSI BCL6 (3q27) (Break Apart)*
4	LSI 4q12 (SCFD2, FIP1L1/LNX,PDGFRA/KIT) (TC BA Probe)*
5	D5S630 (5p15.31)/EGR1 (5q31.2) PDGFRB (5q32) (Break Apart)*
6	LSI MYB (6q23)/CEP 6
7	LSI D7S522 (7q31)/CEP 7 EGFR (7p11.2)/D7Z1 (7p11.1-q11.1) (Amplification) TRG (7p14) (Break Apart)* TRB (7q34) (Break Apart)*
8	LSI MYC (8q24) (Break Apart)* FGFR1 (8p11) (TC Break Apart/Amplification)*
9	LSI p16 (9p21)/CEP 9 LSI ASS (9q34)
10	LSI PTEN (10q32)/CEP 10
11	LSI KMT2A (11q23) (Break Apart)*
11/17	ATM (11q22.3)/TP53 (17p13)
12	LSI CHOP (DDIT3) (12q13) (Break Apart)* LSI ETV6 (12p13) (Break Apart)* LSI MDM2 (12q15)/CEP 12
12/13	LSI D13S319 (13q14.3)/LAMP1 (13q34)/CEP 12
13	LSI 13 (RB1) (13q14) LSI D13S319 (13q14.3) LSI FOXO1 (13q14) (Break Apart)* LSI LAMP1 (13q34)
14	LSI IGH (14q32) (Break Apart)* LSI TCR alpha/delta (14q11.2) (Break Apart)* TRAD (14q11.2) (Break Apart)*
16	LSI FUS (16p11) (Break Apart)* LSI CBFB (16q22) (Break Apart)* CBFB (16q22)/MYH11 (16p13) (Dual Fusion Translocation Probe)*
17	TP53 (17p13)/D17Z1(17p11.1-q11.1) LSI RARA (17q21) (Break Apart)*
18	LSI MALT1 (18q21) (Break Apart)* LSI BCL2 (18q21.33) (Break Apart)* LSI SYT (18q11.2) (Break Apart)*
19	LSI 19q13/19p13 LSI E2A (19p13) (Break Apart)*
20	D20S108 (20q12)/D20S150 (20q13.12)
21	LSI 21 (21q22.13-q22.2)
22	LSI 22 (BCR) (22q11.2) LSI EWSR1 (22q12) (Break Apart)*
X	Renal Cell Carcinoma: TFE3 (Xp11.23)(Break Apart)* - if positive, reflex to ASPSCR1/PRCC/TFE3 translocation analysis*
XY	Sex Mismatched BMT CEP X (SO) / Y (SG)

**Available for PET samples. Other probes may also be available, please call us to discuss your needs.*

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