



**GENETICS PROGRAM
CYTOGENETICS LABORATORY**

TESTING CURRENTLY AVAILABLE

Chromosome Analysis (Karyotype)

- Constitutional chromosome analysis
- Acquired chromosome abnormalities in oncology (e.g. leukemia)

Constitutional FISH Test

- DiGeorge/VCFS TUPLE1 and 22q13.3 deletion syndrome probe (22q11.2/22q13.3)
- Kallmann (KAL1) and Steroid Sulfatase deficiency (STS) probe (Xp22.3)
- Molar pregnancy (chromosomes X, Y, 13, 16, 18, 21)
- Saethre-Chotzen/Williams-Beuren probe (7p21.1/7q11.23)
- Smith-Magenis/Miller-Dieker probe (17p13.3/17p11.2)
- SRY (Sex determining region Y) probe (Yp11.31)
- Wolf Hirschhorn probe (4p16.3)
- XIST probe (Xq13.2)

Microarray Test

- Postnatal SNP array analysis
- Prenatal SNP array analysis
- Oncology SNP array CLL analysis

Oncology FISH Test

- BCR/ABL dual color dual fusion translocation probe (t(9;22)(q34;q11.2))
- CFBF break apart rearrangement probe (16q22)
- CEP8 probe (8p11.1-q11.1)
- EGR1/D5S23, D5S721 dual color probe (5q31/5p15.2)
- IGH/BCL2 translocation dual fusion probe (t(14;18)(q32.33;q21.33))
- IGH/CCND1 dual color dual fusion translocation probe (t(11;14)(q13.3;32.3))
- MALT1 dual color break apart rearrangement probe (18q21.3)
- MLL break apart probe (11q23)
- MYC dual color break apart rearrangement probe (8q24.21)
- PML/RARA dual color dual fusion translocation probe (t(15;17)(q24;q21.1q21.2))
- TEL/AML1 (ETV/RUNX1) translocation dual fusion probe t(12;21)(p13.2;q22.12)
- D7S522/CEP7 probe (7q31/7p11.1-q11.1)
- D20S108 probe (20q12)
- Lymphoma Panel (BCL6, MYC and BCL2)
- Multiple Myeloma Panel (CKS1B/CDKN2C(P18), 13q14, IGH and TP53 / Reflex IGH/FGFR3, IGH/MYEOV and IGH/MAF)