

**GENETICS PROGRAM
CYTOGENETICS LABORATORY TESTS**

Chromosome Analysis (Karyotype)
Constitutional study, peripheral blood
Hematologic disorders, bone marrow

Constitutional FISH Test
DiGeorge/VCFS TUPLE1 and 22q13.3 deletion syndrome probe
Kallmann (KAL1) and Steroid Sulfatase deficiency (STS) probe
Saethre-Chotzen/Williams-Beuren probe
Smith-Magenis/Miller-Dieker probe
SRY (Sex determining region Y) probe
XIST probe

Microarray Test
Postnatal SNP array analysis, peripheral blood
Prenatal SNP array analysis, amniotic fluid
Oncology SNP array analysis, bone marrow and peripheral blood

Oncology FISH Test
Acute myeloid leukemia (AML) (KMT2A, TEL/AML1, PML/RARA, CBFβ)
Chronic myeloid leukemia (CML) (BCR/ABL)
Follicular lymphoma (FL) (IGH/BCL2)
High-grade B-cell lymphoma (BCL6, MYC and BCL2 rearrangements)
Mantle cell lymphoma (MCL) (IGH/CCND1)
Marginal zone lymphoma (MZCL) (BCL6 (3q), D7S522 (7q del), MALT1)
Myelodysplastic syndrome (MDS) (EGR1 (5q del) D7S522 (7q del), CEP8 (+8), D20S108 (20q del))
Multiple myeloma (MM) (CKS1B/CDKN2C(P18), D13S319/LAMP1, IGH and TP53 / reflex IGH/FGFR3, IGH/MYEOV and IGH/MAF)