KHSC Cytogenetics Test List

Chromosome Analysis (Karyotype)

- Constitutional chromosome analysis (from blood, amniotic fluid, solid tissue)
- Acquired chromosome abnormalities in oncology (from bone marrow, tumour tissue)

Constitutional FISH Tests

- Wolf Hirschhorn (4p16.3)
- Cri-du-Chat (5p15.2/5q31)
- NOR Acro-P-Arm
- Prader-Willi region (SNRPN/CEP15/PML)
- Smith-Magenis region (SMS/RARA)
- DiGeorge region (HIRA/ARSA)
- SHOX deficiency disorders (SHOX (Xp22.33,Yp11.32/DYZ1/DXZ1))
- Kallmann (KAL1) syndrome (Xp22.3/CEPX)
- Steroid Sulfatase deficiency (STS) probe (STS (Xp22.3/CEPX)
- SRY (Sex determining region Y) (Yp11.3/CEPX)
- Williams Syndrome Region (ELN) 7q11.23
- CEPX/CEPY
- AneuVysion Multi-colour 5 probe panel (13,21,18,X,Y)
- TelVysion Multi-colour FISH probes

Oncology FISH Tests

- Acute leukemia (AML/ALL):
    - RUNX1/RUNX1T1 – t(8;21)
    - PML/RARA – t(15;17)
    - CBFB – inv(16), t(16;16)
    - EGR1 – del 5q
    - ELN – del 7q
    - ETV6/RUNX1 – t(12;21)
    - BCR/ABL – t(9;22)
    - KMT2A (MLL) (11q23)
    - CEP4/CEP10
    - FIP1L1/CHIC2/PDGFRA (4q12)
    - RPN1/MECOM (3q21.3/3q26.2)
    - ALL Multiprobe panel
- Multiple Myeloma:
    - TP53 – del 17p
    - IGH/FGFR3 – t(4;14)
    - IGH breakapart
    - IGH/MAF – t(14;16)
    - IGH/MAFB – t(14;20)
    - CCND1/IGH – t(11;14)
    - Del 13q (13q14.2-q14.3/13q34)
- Chronic Myelogenous Leukemia (CML):

    - BCR/ABL – t(9;22)
    - BCR/ABL plus ASS1
- Chronic Lyphocytic Leukemia (CLL):
    - TP53 – del 17p
    - ATM – del 11q22.3
    - MYB – del 6q23
    - Del 13q (13q14.2-q14.3/13q34)
    - CEP12
- Myelodysplastic Syndrome (MDS):
    - EGR1 – del 5q
    - ELN – del 7q, monosomy 7
    - MYC – trisomy 8
    - RPN1/MECOM (3q21.3/3q26.2)
    - Del 20q (20q12/20q13.12)
- Solid Tumour
    - MDM2 (12q15)
- Solid tumour – formalin fixed paraffin embedded (FFPE) FISH:
    - Breast cancer - PathVysion HER-2/neu (17q11.2-q12/CEP17)
    - Lung cancer - ALK (2p23)
    - Lymphoma:
        - C-MYC (8q24)
        - IGH/BCL2 – t(14;18)
        - BCL6 (3q27)