

GENETICS

- **Cytogenetics/Karyotyping:**
 - Chromosomal Study- Blood (Karyotyping)
 - Chromosomal Study–Special (Karyotyping) (AF/BM/CVS/D)
 - FISH (Fluorescence In Situ Hybridization) Panels (X,Y, 13, 18, 21, Digeorge, Miller dieker, Prader – Willi, combinations)
- **Next-Generation Sequencing (NGS):**
 - Whole Exome Sequencing (WES)
 - Whole Genome Sequencing (WGS)
- **Molecular Genetics / DNA Testing:**
 - Alpha Thalassaemia by PCR
 - Alpha Thalassemia Gene sequencing
 - Alzheimer Gene screen
 - Androgen Receptor
 - β -Thalassaemia by PCR
 - BCL I&II
 - BCR-ABL (PCR)
 - BRCA1 & BRCA2
 - Cardiovascular Disease Mutations-CVD-12 by PCR
 - Celiac disease gene
 - CLL Panel / GLI:(12q13), ATM: (11q22), p53: (17p13), DLEU: 12q15
 - Congenital Adrenal Hyperplasia (PCR)
 - Congenital absence of vas deference
 - Congenital Deafness screen
 - Colon cancer
 - Cystic Fibrosis-17 by PCR
 - Cystic Fibrosis (Full CFTR Gene)
 - Familial Mediterranean Fever (FMF) by PCR (12 Mutations)
 - FII Prothrombin by PCR
 - FLT3
 - FV Cambridge Gene (R307T) by PCR
 - FV Hong Kong Gene by PCR
 - FV Leiden – 1691 (Activated Protein C) by PCR
 - FV (Codon 1702) by PCR
 - FV HR2 Aplotype (Codon 1299) by PCR
 - FXIII by PCR
 - Fragile X Syndrome by Sequencing
 - FRIEDRICH's ATAXIA
 - GAD Abs (Anti Glutamic Acid Decarboxylase)
 - Genetic screen full sequence (up to 2, 1-10, more than 10 genes)
 - Hearing Loss Panel
 - HLA (A,B,C, DR) by PCR
 - HLA B27 by PCR
 - HLA B8 by PCR
 - HLA DQ by PCR
 - HLA DR by PCR
 - Homocysteine Gene (MTHFR – C677T & A1298c) by PCR
 - Huntington Disease
 - IGR (idiopathic growth retardation)

- Intersex
- Lactose intolerance by PCR
- MTHFR – C^{677T} by PCR (Homocystein Gene)
- MTHFR – A^{1294C} by PCR
- Myotonic Dystrophy
- Neonatal Haemoglobinopathies
- NIPT (Non-invasive Prenatal Testing: 13,18,21,X,Y) by NGS
- ONCOblot
- ONCOsure
- Osteogenesis Imperfecta screen
- Parkinson screen
- Paternity Testing (STR's)
- Peripheral Neuropathy Panel
- PML-PARa
- SRY (Sex-determining Region Y protein)
- Sibling Testing (STR's)
- Celiac disease gene
- **Flow Cytometry/Immunophenotyping:**
 - ALL Panel, Flow Cytometry
 - AML Panel, Flow Cytometry
 - CD Markers (CD 1a to CD 104 - extensive list for various cell types)
 - CLL Panel, Flow Cytometry
 - Immunophenotyping (surface markers by FLOWCYTOMETRY for: Leukemia and Lymphoma (each marker))
 - Leukaemia Panel
 - Lymphoma Panel
- **Tissue Pathology:**
 - Immunofluorescence study on Tissue
 - Culture- Tissue
- **Pregnancy & Prenatal Screening:**
 - Pregnancy Test
 - β-HCG – Free
 - β-HCG
 - 1st Trimester Prenatal Interpretive Screening
 - Down Syndrome Dual Test 1st Trimester (PAPP-a, Free β-HCG)
 - Down Syndrome (AFP, β- HCG, FE^α)(Triple Test)
 - Down Syndrome (AFP, β-HCG, FE^α, Inhibin A) (Quadruple Test)
 - Integrated Test (wk 12 + wk 16)
 - Quadruple Test
 - PAPP-a
 - Placental Growth Factor (PIGF)
 - Soluble Fms-like Tyrosine Kinase-1 (sFlt-1)
 - sFlt-1/PIGF Ratio
- **Genetic / Inherited Metabolic Diseases:**
 - Alpha-Fucosidase
 - Alpha-Glucosidase I
 - Alpha-Galactosidase
 - Alpha-Iduronidase

- Arylsulfatase A
- Gal- γ P-Uridyl Transferase (Quantitative)
- Galactokinase
- Galactose
- Hexosaminidase – Total
- Hexosaminidase A
- Iduronat- γ -Sulfatase
- Neonatal Galactose- Total
- Coproporphyrins (Quantitative)
- Porphyrins – Total

Remarks Key (as provided by you):

- **Dy/A:** Daily (or As Directed/Available)
- **S:** Serum
- **ABS:** Serum (presumably Acute Blood Sample)
- **U:** Urine
- **24U:** 24-hour Urine
- **EWB:** EDTA Whole Blood
- **HWB:** Heparin Whole Blood
- **CP:** Citrated Plasma
- **EP:** EDTA Plasma
- **HP:** Heparin Plasma
- **AF:** Amniotic Fluid
- **BF:** Bronchial Fluid / Body Fluid
- **BM:** Bone Marrow
- **CSF:** Cerebrospinal Fluid
- **D:** Direct Sample (e.g., discharge)
- **DB:** Dried Blood Spot
- **EnS:** Endocervical Swab
- **ES:** Eye Swab
- **FP:** Fluoride Plasma
- **GnS:** Genital Swab
- **HVS:** High Vaginal Swab
- **LBC:** Liquid Based Cytology
- **NS:** Nasal Swab
- **SF:** Synovial Fluid / Seminal Fluid
- **SP:** Sputum
- **ST:** Stool
- **US:** Random Urine Sample (or unspecified urine)
- **CWB:** Cord Whole Blood
- **LI-HEP:** Lithium Heparin
- **ASP:** Aspirate
- **Vacutec:** Vacutainer tube for collection
- **Call:** Call for details/appointment/specific instructions.

