

LIST OF INVESTIGATIONS

Karyotyping:

Test Code	Test	SAMPLE	CONTAINER TYPE	TAT
K001	Peripheral blood cells For Karyotyping [Single]	Peripheral blood	Sodium heparin Vacutainer (2ml)	7-8 days
K002	Peripheral blood cells For Karyotyping [Couple]	Peripheral blood	Sodium heparin Vacutainer (2ml)	7-8 days
K003	Fetal Blood Sample For Karyotyping	Peripheral blood	Sodium heparin Vacutainer (2ml)	7-8 days
K004	Products Of Conception [POC]	Placental tissue	Bits of Placental Tissue in sterile tissue culture container with normal saline or culture media	15-20 days
K006	Amniotic Fluid [AF]	Amniotic fluid	2 leak proof Sterile Tubes (15 ml each)	15-20 days
K007	Fragile X Analysis	Peripheral blood	Sodium heparin Vacutainer (2ml)	7-8 days
K008	Fragile X Analysis (Prenatal)	Chorionic villi/ Amniotic fluid	Bits of chorionic tissue in Sterile culture container with normal saline or culture media / 2 leak proof Sterile Tubes (15 ml each)	15-20 days
K009	High Resolution Chromosome Study	Peripheral blood	Sodium heparin Vacutainer (2ml)	7-8 days
K011	Chorionic Villus Sample [CVS]	Chorionic villi	Bits of chorionic Tissue Sterile tissue culture container with normal saline or culture media	15-20 days
K012	Cord blood sample (Abortus and prenatal)	Cord Blood	Sodium Heparin Vacutainer (2ml)	7-8 days

FISH Investigation:

Test Code	Test	SAMPLE	CONTAINER TYPE	TAT
F012	Del(20q) [MDS]	Bone marrow / Peripheral blood	Sodium heparin Vacutainer (2ml)	5-6 days
F013	Del(5q) [MDS]	Bone marrow / Peripheral blood	Sodium heparin Vacutainer (2ml)	5-6 days
F014	Del(7q) [MDS]	Bone marrow / Peripheral blood	Sodium heparin Vacutainer (2ml)	5-6 days
F016	17p(p53)	Bone marrow / Peripheral blood	Sodium heparin Vacutainer (2ml)	5-6 days
F017	11q(ATM)	Bone marrow / Peripheral blood	Sodium heparin Vacutainer (2ml)	5-6 days
F018	Prader-Willi Syndrome	Peripheral blood	Sodium heparin Vacutainer (2ml)	5-6 days
F019	William's Syndrome	Peripheral blood	Sodium heparin Vacutainer (2ml)	5-6 days
F020	Di-George Syndrome	Peripheral blood	Sodium heparin Vacutainer (2ml)	5-6 days
F021	CHROMOSOMES [13, 18, 21, X & Y]	Amniotic fluid	Sterile container (20ml)	5-6 days
		Chorionic villi	Sterile Container	
		Blood	Heparin vacutainer (2ml)	
F022	Single Chromosome FISH (13/18/21/X/Y)	Amniotic fluid	Sterile container (20ml)	5-6 days
		Chorionic villi	Sterile Container	
		Blood	Heparin vacutainer (2ml)	

Male Infertility

Test Code	Test	SAMPLE	CONTAINER TYPE	TAT
MI001	Y Chromosome Micro-deletion	Peripheral blood	EDTA vacutainer (2ml)	4-5days
MI002	Sperm DNA fragmentation	Semen Sample	Sterile Container	4-5days

REAL Time PCR:

Test Code	Test	SAMPLE	CONTAINER TYPE	TAT
C006	TORCH PCR	Bone marrow / Peripheral blood	EDTA vacutainer (2ml)	5-6 days

Conventional PCR Study and Gel and Sequencing Based Detection:

Test Code	Test	SAMPLE	CONTAINER TYPE	TAT
P001	β-Thalassemia Five common mutations (IVS 1-5 G>C, IVS 1-1 G>T, 619 bp deletion, FS 8/9 +G, FS 41/42 -CTTT)	Amniotic fluid / Chorionic villi / Blood	Sterile container (20ml)/ Sterile Normal Saline Container / EDTA vacutainer (2ml)	4-5days
P002	Fragile X Syndrome Southern Blot based analysis for FRAXA locus using EcoRI, PstI, and EcoRI+EagI digestion	Peripheral blood	EDTA vacutainer (2ml)	10-15days
P003	Glucose 6 phosphate dehydrogenase Orissa, Mediterranean and keralakalian mutations	Peripheral blood	EDTA vacutainer (2ml)	6-7days
P004	Hemophilia-A Intron-22 inversion by Southern Blot analysis & Carrier analysis @ markers (BclI & XbaI RFLP) Hemophilia-B Carrier analysis @ intragenic markers (HhaI, DdeI, TaqI)	Peripheral blood	EDTA vacutainer (2ml)	10-15 days
P005	Spinal Muscular Atrophy • Deletion analysis (exon7 & 8 only) • Carrier analysis using MLPA	Peripheral blood	EDTA vacutainer (2ml)	6-7days
P006	MTHFR gene polymorphisms 677T>C and 1298A>C mutations	Peripheral blood	EDTA vacutainer (2ml)	10 days
P007	Duchenne and Becker Muscular Dystrophy • Deletion analysis for exons 24 & 79 by MLPA analysis	Peripheral blood	EDTA vacutainer (2ml)	6-7days
P008	Cystic fibrosis Four common mutations (ΔF508, G542X, G551D, R553X) and 5T	Peripheral blood	EDTA vacutainer (2ml)	6-7days
P009	Non-Syndromic Hearing Loss (NSHL) Connexin 26 (GJB2) exon 2 screening	Peripheral blood	EDTA vacutainer (2ml)	10 Days
P011	Mitochondrial encephalopathy Leigh's Disease (LD)	Peripheral blood	EDTA vacutainer (2ml)	10 Days
P012	Mitochondrial encephalopathy LHON panel	Peripheral blood	EDTA vacutainer (2ml)	10 Days

Infectious Disease PCR:

Test Code	Test	Sample type	TAT
ID001	CMV DNA PCR	EDTA vacutainer (2ml)	2-3 Days
ID002	CMV Quantitative Real Time PCR	EDTA vacutainer (2ml)	3-4 Days
ID010	Herpes Simplex	EDTA vacutainer (2ml)	2-3 Days
ID011	Human Papilloma Virus (HPV) with LBC	EDTA vacutainer (2ml)	2-3 Days
ID012	TB DNA PCR Qualitative (Real Time PCR)	EDTA vacutainer (2ml)	1-2 Days
ID013	HIV Proviral DNA PCR	EDTA vacutainer (2ml)	2-3 Days
ID014	HIV-1 Quantitative Real Time PCR	EDTA vacutainer (2ml)	3-4 Days
ID015	HIV Genotyping	EDTA vacutainer (2ml)	10-15 days`

Microarray Analysis:

Test Code	Test	Sample type	TAT
MC001	Chromosomal Micarrray Analysis (array CGH) 8X60k	Amniotic Fluid (20ml)	15days
MC002	Chromosomal Micarrray Analysis (array CGH) 8X60k	Product of Conception (15ml sterile tube)	15days
MC003	Chromosomal Micarrray Analysis (array CGH) 8X60k	Peripheral Blood (4ml in EDTA vaccutainer)	15days
MC004	Pre-implantation Genetic Screening (PGS) 8X60k	Blastomere (ONE or TWO) in Kit provided by Centre (Embryo Biopsy) Minimum four Biopsy per Patient	48hours
MC005	Pre-implantation Genetic Screening (PGS) 8X60k	Blastocyst Biopsy (Trophectoderm) in Kit provided by Centre (Embryo Biopsy) Minimum four Biopsy per Patient	7-12 days
MC006	Array CGH + SNP analysis	All type of Biological samples	30days

NEXT GENERATION SEQUENCING BASED Analysis:

Test Code	Test	Sample type	TAT
CANCER PANELS			
NG002	Breast Carcinoma Panel	Saliva sample/EDTA Blood/Tissue	15days
NG005	Comprehensive Cancer Panel (Germline Panel)	Saliva sample/EDTA Blood/Tissue	4 – 6 weeks
CLINICAL EXOME TEST			
NG016	Clinical Exome Test covers 4800 genes associated with known clinical phenotypes	Saliva sample/EDTA Blood/Tissue	8 - 10 weeks

Package Genetic Diagnosis:

Test Code	Test
PAC-01	Male Infertility Karyotyping (K001) Y chromosome microdeletion (MI001) Sperm DNA fragmentation (MI002)
PAC-02	Female Infertility Karyotyping (K001) FMR1 Gene Mutation Analysis FSH R Gene Mutation Analysis Cyp21A2
PAC-03	Amniotic Fluid Karyotyping (K006) FISH 5 chromosomes (F021) ACGH- Microarray (MC001)
PAC-04	Amniotic Fluid Karyotyping (K006) FISH 5 chromosomes (F021)
PAC-05	Product of Conception Karyotyping (K004) FISH 5 chromosomes (F021) ACGH- Microarray (MC002)
PAC-06	Product of Conception Karyotyping (K004) FISH 5 chromosomes (F021)
PAC-08	Chorionic Villus Sample Karyotyping (K0011) FISH 5 chromosomes (F021) ACGH- Microarray (MC002)
PAC-09	Chorionic Villus Sample Karyotyping (K0011) FISH 5 chromosomes (F021)