

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
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

FISH Investigation:

Test Code	Test	SAMPLE	CONTAINER TYPE	TAT
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)	

Conventional PCR Study and Gel and Sequencing Based Detection:

Test Code	Test	SAMPLE	CONTAINER TYPE	TAT
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
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 & 29 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
P012	Huntington's Disease	Peripheral blood	EDTA vacutainer (2ml)	5-6 Days
P013	Spinocerebellar Ataxias (SCA) (Single form)	Peripheral blood	EDTA vacutainer (2ml)	5-6 Days
P014	Spinocerebellar Ataxias SCA Panel (1, 2, 3, 6, 7, 10 & 12)	Peripheral blood	EDTA vacutainer (2ml)	5-6 Days
P015	RETT Syndrome	Peripheral blood	EDTA vacutainer (2ml)	10-15 days

Infectious Disease PCR:

Test Code	Test	TAT	Sample Type	CONTAINER TYPE
ID001	CMV DNA PCR	2-3 Days	Peripheral blood	EDTA vacutainer (2ml)
ID002	CMV Quantitative Real Time PCR	3-4 Days	Peripheral blood	EDTA vacutainer (2ml)
ID003	HBV DNA PCR (Qualitative)	2-3 Days	Peripheral blood	EDTA vacutainer (2ml)
ID004	HBV Genotyping (Sequencing Method)	3-4 Days	Peripheral blood	EDTA vacutainer (2ml)
ID005	HBV Quantitative Real Time PCR	3-4 Days	Peripheral blood	EDTA vacutainer (2ml)
ID006	HCV Genotyping (Sequencing Method)	3-4 Days	Peripheral blood	EDTA vacutainer (2ml)
ID007	HCV Quantitative Real Time PCR	3-4 Days	Peripheral blood	EDTA vacutainer (2ml)
ID008	HCV RNA PCR (Qualitative)	2-3 Days	Peripheral blood	EDTA vacutainer (2ml)
ID009	Hepatitis A Virus RNA RT-PCR(HAV)	3-4 Days	Peripheral blood	EDTA vacutainer (2ml)
ID010	Herpes Simplex	2-3 Days	Peripheral blood	EDTA vacutainer (2ml)
ID011	Human Papilloma Virus (HPV) with LBC	2-3 Days	Peripheral blood	EDTA vacutainer (2ml)
ID012	TB DNA PCR Qualitative (Real Time PCR)	1-2 Days	Peripheral blood	EDTA vacutainer (2ml)
ID013	HIV Proviral DNA PCR	2-3 Days	Peripheral blood	EDTA vacutainer (2ml)
ID014	HIV-1 Quantitative Real Time PCR	3-4 Days	Peripheral blood	EDTA vacutainer (2ml)
ID015	HIV Genotyping	10-15 days`	Peripheral blood	EDTA vacutainer (2ml)

Microarray Analysis:

Test Code	Test	Sample type	TAT
MC001	Chromosomal Microarray Analysis (array CGH) 8X60k	Amniotic Fluid (20ml)	15days
MC003	Chromosomal Microarray Analysis (array CGH) 8X60k	Peripheral Blood (4ml in EDTA vacutainer)	15days
MC006	Array CGH + SNP analysis	All type of Biological samples	30days

NEXT GENERATION SEQUENCING BASED Analysis:

Test Code	Test	Sample type	TAT
NEUROLOGY			
NG013	Neurology Panel	EDTA Blood/Tissue	4-5 weeks
NG014	Muscular Dystrophy and Myopathy Panel	EDTA Blood/Tissue	4-5 weeks
NG015	Mitochondrial Panel	EDTA Blood/Tissue	4-5 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-09	Neuronal/ASD/Delayed Development Karyotyping (K001) aCGH- Microarray (MC001)