

MANIPAL
UNIVERSITY

Diagnostic Tests

Molecular and Cellular Biology Unit

School of Life Sciences

Manipal University

Manipal - 576 104

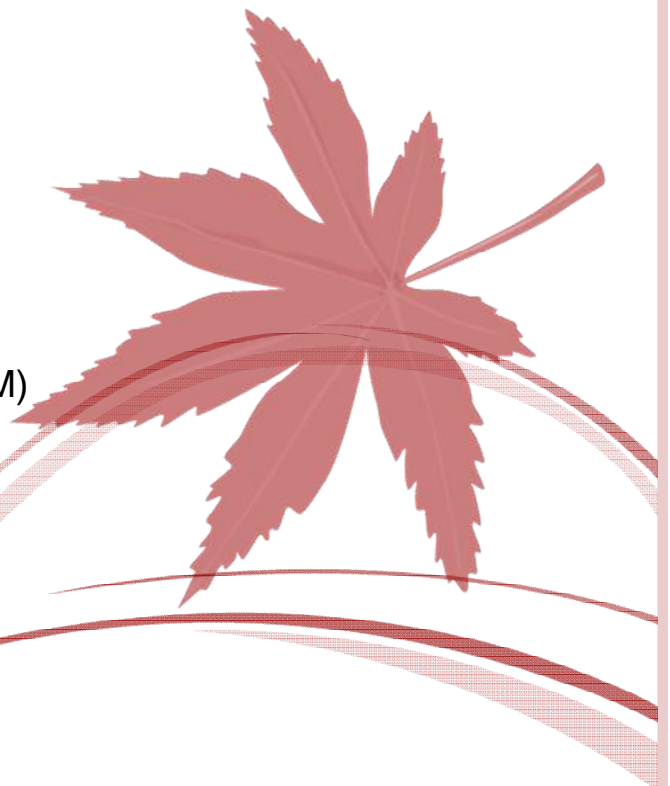
Contact

Tel: 0820 2922058; 0820 2923501

Email: dbt@manipal.edu

Working Hours

Monday to Saturday (9:00 AM–5:00 PM)



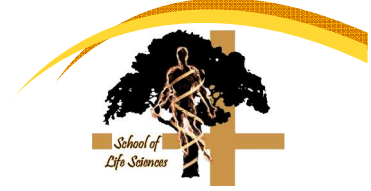


Molecular and Genetic Tests

Test Code	Name of the Condition	Name of Gene	Exons/Mutations	Type of Test	Sample Required
CYS026	Cystic Fibrosis	<i>CFTR</i>	Δf508	QF-PCR	Blood *
PCR006	Hemochromatosis	<i>HFE</i>	H63D and C282Y	PCR and RFLP	Blood *
AMY008	Amyloidosis	<i>TTE Transthyretin</i>	Val30Met	PCR-DNA sequencing	Blood
PCR010	Hepatitis B Virus (HBV)	'S' gene	(Semi-Quantitative)	PCR	Blood
HEP013	Hepatitis B Virus (HBV)	'S' gene	Quantitative	Real Time-PCR	Blood
WIL006	Wilson's Disease	<i>ATP7B</i>	Any single exon	PCR-DNA sequencing	Blood*
PCR011	Wilson's Disease	<i>ATP7B</i>	Exons 8, 12 & 13	PCR-DNA sequencing	Blood *
6ME002	6-Mercaptopurine/ azathioprine toxicity	<i>TPMT</i>	TPMT*2, TPMT*3A, TPMT*3B,TPMT*3C	PCR-RFLP	Blood
PCR016	Hyperhomocysteinuria Venous thromboembolic disease Cardiovascular disease	<i>MTHFR</i>	C677T polymorphism A1298C polymorphism	PCR-RFLP	Blood
PCR027	Beta-Thalassemia	<i>HBB</i>	Exons 1, 2 & 3, intron 1 and parts of intron 2 & the 3'- UTR	PCR and DNA sequencing	Blood *
PCR026	Sickle Cell Anemia	<i>HBB</i>	Exons 1,2 & flanking sequences	PCR- DNA Sequencing	Blood
PCR018	Fragile X	<i>FMR 1 and FMR 2</i>	Triplet expansion	PCR	Blood*
HUN002	Huntington's disease	<i>HTT</i>	Trinucleotide repeat	Capillary gel electrophoresis	Blood

Note:

1. Sample Required: Blood – indicates 5 ml anticoagulated (EDTA/Heparin) blood, unless otherwise mentioned.
2. * - 5 ml blood samples from parents required
3. ** - 5 ml blood samples from parents (required) and other siblings whenever possible
4. Immediately transport the blood sample at room temperature. If delayed, please store samples at 4°C to prevent RBC lysis.
5. Samples not meeting these conditions will be rejected.



Molecular and Genetic Tests

Test Code	Name of the Condition	Name of Gene	Exons/Mutations	Type of Test	Sample Required
PCR020	Thrombosis Association	PAI-I	4G/5G polymorphism	PCR	Blood
PCR021	Thrombosis (Leiden) Association	Factor V	G1691A polymorphism	PCR-RFLP	Blood
PCR022	Thrombosis Association	Prothrombin	G20210A polymorphism	PCR-RFLP	Blood
PCR025	Resistance to drugs association	MDR 1/ ABCB1	C3435T/G2677A/ G26677T polymorphism	PCR-DNA sequencing	Blood
PCR028	Beta2 adrenoreceptor	B2-AR	Complete coding sequence	PCR-DNA sequencing	Blood
JAK002	JAK 2	Jak2	Jak2 (V617F)	PCR-RFLP	Blood
FLT002	Acute Myeloid Leukemia	FLT3	ITD	PCR-Fragment analysis	Blood
GIL001& GIL002	Gilbert's syndrome	UGT1A	UGT1A*28 polymorphism	PCR-DNA sequencing	Blood
SPI012	Spinal muscular atrophy	SMN1	Deletion in exon 7 & 8	PCR-RFLP	Blood
DUC001	Duchenne Muscular Dystrophy	Dystrophin	Deletions in 20 exons	PCR	Blood
HEM026	Hemophilia A Carrier analysis	Factor VIII	Linkage analysis	PCR-RFLP	Blood**
HEM027	Hemophilia B Carrier analysis	Factor IX	Linkage analysis	PCR-RFLP	Blood**
5FU004	5-FU, Methotrexate toxicity	TYMS, DPYD	Polymorphisms	PCR/PCR-RFLP	Blood

Note:

1. In the Table, Sample Required: Blood – indicates 5 ml anticoagulated (EDTA/Heparin) blood, unless otherwise mentioned.
2. * - 5 ml blood samples from parents required.
3. ** - 5 ml blood samples from parents (required) and other siblings whenever possible
4. Immediately transport the blood sample at room temperature. If delayed, please store samples at 4°C to prevent RBC lysis.
5. Samples not meeting these conditions will be rejected.



Molecular and Genetic Tests

Test Code	Name of the Condition	Name of Gene	Exons/Mutations	Type of Test	Sample Required
MY005	Myotonic Dystrophy	<i>DM1</i>	Repeat expansion	PCR	Blood
ACH001	Achondroplasia	<i>FGF-R3</i>	Mutation analysis	PCR-RFLP	Blood
HYP010	Hypochondroplasia	<i>FGF-R3</i>	Mutation analysis	PCR-RFLP	Blood
PRA002	Prader-Willi/Angleman Syndrome	<i>SNRPN</i>	Methylation analysis	MSP-PCR	Blood
ALP011	Alpha-1, Antitrypsin deficiency	<i>AA1</i>	Mutational analysis	PCR-RFLP	Blood
PAP001&103	Papillon Lefvre Syndrome (PLS)	<i>Cathepsin C</i>	Mutation analysis Exon 7	PCR-Sequencing	Blood
EPS002	Epstein Barr Virus A & B	<i>EBV</i>	EBNA	PCR	Blood
HUM005	Human Papilloma virus	<i>HPV</i>	L1	PCR	Blood
HUM006	Human Papilloma virus	<i>HPV</i>	L1 – Subtype	PCR-Sequencing	Blood
RAS001	K-Ras mutation screen	<i>KRAS</i>	Codon 12 and 13	PCR-Sequencing	Tissue
P53001	P53 mutations screen	<i>p53</i>	Exon 4,5,6,7&8	PCR-Sequencing	Tissue/ blood
SPI015	Spinocerebellar ataxia – (SCA 1/ 2/ 3/ 6/ 7)	<i>Any one marker SCA1/ SCA2/ MJD/ CACNA1A/ SCA7</i>	Triplet repeat analysis	QF-PCR	Blood
QFP004	Spinocerebellar ataxia – (SCA 1, 2, 3, 6, and 7)	<i>SCA1, SCA2, MJD, CACNA1A, SCA7</i>	Triplet repeat analysis	QF-PCR	Blood

Note:

1. In the Table, Sample Required: Blood – indicates 5 ml anticoagulated (EDTA/Heparin) blood, unless otherwise mentioned.
2. * - 5 ml blood samples from parents required.
3. ** - 5 ml blood samples from parents (required) and other siblings whenever possible
4. Immediately transport the blood sample at room temperature. If delayed, please store samples at 4°C to prevent RBC lysis.
5. Samples not meeting these conditions will be rejected.



Molecular and Genetic Tests

Test Code	Name of the Condition	Name of Gene	Exons/Mutations	Type of Test	Sample Required
CON101	Non-syndromic hearing loss	<i>Connexin-26</i>	Mutations	PCR/DNA sequencing	Blood
PFA001	Malaria	<i>P. falciparum</i>	Quantitative Detection	Real Time PCR	Blood
PVI001	Malaria	<i>P. vivax</i>	Quantitative Detection	Real Time PCR	Blood
APE001	Apert syndrome	<i>FGF-R2</i>	Mutation	PCR/RFLP	Blood
RES009	Resistance to Warfarin	<i>VKORC1, CYP2C9, PON1</i>	VKORC1-1639G>A CYP2C9*2 PON1Gln192Arg PON1Leu55Met	PCR-RFLP	Blood
MOR004	Morquio A Syndrome	<i>GALNS</i>	All 16 exons	PCR-sequencing	Blood
MOR005	Morquio B Syndrome and GM1 Gangliosidosis	<i>GLB1</i>	All 14 exons	PCR-sequencing	Blood
SET001	Setleis syndrome	<i>TWIST2</i>	Single exon	PCR-Sequencing	Blood
FUH001	Fuhrmann syndrome	<i>WNT7A</i>	All 4 exons	PCR-Sequencing	Blood
PRO058	Progressive Pseudorheumatoid Dysplasia	<i>WISP3</i>	All 5 coding exons	PCR–Sequencing	Blood
GST001	Glutathione -S-Transferase	<i>GST – T1, M1, P1</i>	Polymorphisms/ mutations	PCR/RFLP	Blood
SEQ002 SEQ003 SEQ004	Sequencing of: i) any single exon ii) any five exons iii) any ten exons	<i>Name(s) of the gene and the exon(s) number(s) to be provided</i>	Synthesis of primers and sequencing		

Note:

1. Sample Required: Blood – indicates 5 ml anticoagulated (EDTA/Heparin) blood, unless otherwise mentioned.
2. * - 5 ml blood samples from parents required
3. ** - 5 ml blood samples from parents (required) and other siblings whenever possible
4. Immediately transport the blood sample at room temperature. If delayed, please store samples at 4°C to prevent RBC lysis.
5. Samples not meeting these conditions will be rejected.

Molecular and Genetic Tests: QF-PCR for Prenatal Diagnosis

(Aneuploidy chromosomes 13, 18, 21 and X)

Test Code	Type of Test	Sample	Report Duration
ANE003	Aneuploidy of Chromosome 13	*AF/CVS/FB/POC along with parental blood samples	5 days
ANE004	Aneuploidy of Chromosome 18		5 days
ANE005	Aneuploidy of Chromosome 21		5 days
ANE006	Aneuploidy of sex chromosomes (X & Y)		5 days
ANE007	Aneuploidy of any two chromosomes 13/18/21		5 days
ANE008	Aneuploidy of Chromosome 13, 18, & 21		5 days
ANE009	Aneuploidy of Chromosome 13, 18, 21 and the sex chromosomes (X and Y)		5 days

Note:

All samples for QF-PCR should be transported at room temperature **IMMEDIATELY** (unless otherwise mentioned) under aseptic precautions. Tubes with samples should be protected from damage by padding with soft material. Samples not meeting these requirements will NOT be accepted.

1. Amniotic fluid or AF (10ml in sterile container).
2. Chorionic villus sample or CVS (in 10 ml HBSS in sterile container).
3. Fetal blood or FB (heparin/EDTA in sterile container).
4. Products of conception or POC (in sterile normal saline).
5. 5 ml blood samples from parents required.



Molecular and Genetic Tests: Prenatal Diagnosis for mutations

Test Code	Name of the Condition	Name of Gene	Exons/Mutations	Type of Test	Sample Required**
PRE017	Prenatal Diagnosis for SMA	<i>SMN1</i>	Deletion in exon 7 & 8 and maternal cell contamination	PCR/RFLP and QF-PCR with chromosome 21 microsatellites	CVS/AF & **5ml of blood
PRE018	Prenatal Diagnosis for Thalassemia	<i>HBB</i>	Mutational analysis and maternal cell contamination	PCR/DNA sequencing and QF-PCR with chromosome 21 microsatellites	CVS/ amniotic fluid or fetal blood. **5ml blood
PRE019	Prenatal Diagnosis for DMD	<i>DMD</i>	Deletions and maternal cell contamination	Multiplex PCR and QF-PCR with chromosome 21 microsatellites	CVS/AF & **5ml of blood
PRE020	Prenatal Diagnosis for Hemophilia A	<i>Factor VIII</i>	Carrier X chromosome	Linkage analysis with microsatellites/ intragenic SNPs	CVS/AF & **5ml of blood (proband's blood sample)
PRE022	Prenatal Diagnosis for Achondroplasia	<i>FGFR3</i>	Mutation analysis and maternal cell contamination	PCR-RFLP/ QF-PCR	CVS/AF & **5ml of blood
PRE023	Prenatal Diagnosis for Hypochondroplasia	<i>FGFR3</i>	Mutation analysis and maternal cell contamination	PCR-RFLP/ QFPCR	CVS/AF & **5ml of blood
PRE024	Prenatal Diagnosis for Cystic Fibrosis	<i>CTFR</i>	Δf508	QF-PCR	CVS/AF & **5ml of blood
PRO057	Prenatal Diagnosis for Progressive Pseudorheumatoid	<i>WISP3</i>	Mutation analysis and maternal cell contamination	PCR– Sequencing/QF-PCR	CVS/AF & **5ml of blood

Note:

1. In the Table , Sample Source: Blood – indicates 5 ml anticoagulated (EDTA/Heparin) blood, unless otherwise mentioned.
2. Immediately transport the blood sample at room temperature. If delayed, please store samples at 4°C to prevent RBC lysis.
3. ** 5 ml blood - Additionally, 5 ml blood sample from mother is required and other siblings whenever possible.
4. Samples not meeting these conditions will be rejected.



Flow Cytometry Tests

Test Code	Panel	CD markers	Name of the condition	Sample Required
ALL004	ALL	CD3/CD7/CD10/CD19/ CD34/CD33/CD13/CD45	Acute Lymphoid Leukemia	Peripheral blood or Bone Marrow
ALL003	ALL	CD3/CD7/CD10/CD13/ CD19/CD33/CD34/CD45/ CD 79b/TDT	Acute Lymphoid Leukemia (With anti TDT & CD79)	Peripheral blood or Bone Marrow
AML001	AML	CD3/CD7/CD10/CD19/ CD34/CD33/CD13/CD45/ CD14	Acute Myeloid Leukemia	Blood or Bone Marrow
CLP002	CLPD	CD5/CD10/CD19/CD20/ CD23/CD25/CD103/ kappa/lambda	Chronic Lymphoproliferative Disorder (With CD103)	Blood or Bone Marrow
CLP001	CLPD	CD5/CD10/CD19/CD20/ CD23/CD25/kappa/ lambda	Chronic Lymphoproliferative Disorder (Without CD103)	Blood or Bone Marrow
DNA003	DNA ploidy	---	DNA Ploidy – Leukemia	Blood or Bone Marrow
DNA004	DNA ploidy	---	DNA Ploidy in Solid tumors	Paraffin embedded tissue blocks
HIV004	HIV	CD4 / CD8/CD3	HIV Monitoring	Blood or Bone Marrow
HLA008	HLA-B27	HLA-B27	Ankylosing Spondylitis	Blood
PNH002	PNH	CD55/ CD59	Paroxysmal nocturnal hemoglobinuria (PNH) - RBC	Blood or Bone Marrow
PNH003	PNH	CD55/ CD59/CD16	Paroxysmal nocturnal hemoglobinuria (PNH) - WBC	Blood or Bone Marrow

Note:

1. **Required sample:** Heparinized/ EDTA 5 ml of peripheral blood or 3 ml of bone marrow.
2. All samples must be transported immediately at room temperature. Do not freeze the samples
3. Samples not meeting the requirements will not be accepted.
4. Rs. 800 extra will be charged for additional antibody.
5. Reports will be available within a minimum of 3 working days.



Flow Cytometry Tests

Any single Antibody (percentage)

Test Code	CD Marker
CD2001	CD2
CD3001	CD3
CD4005	CD4
CD5001	CD5
CD7001	CD7
CD8001	CD8
CD1010	CD10
CD1101	CD11a
CD1005	CD13
CD1401	CD14
CD1501	CD15
CD1601	CD16
CD1006	CD19
CD2020	CD20
CD2022	CD22
CD2023	CD23
CD2025	CD25
CD3101	CD31
CD3002	CD33
CD3006	CD34

Test Code	CD Marker
CD4041	CD41
CD4401	CD44
CD4003	CD45
CD5055	CD55
CD5056	CD56
CD5059	CD59
CD6101	CD61
CD6069	CD69
CD7101	CD71
CD7901	CD79b
CD1103	CD103
CD1117	CD117
CD1330	CD133
CD2350	CD235a
HLA104	HLADR
KAP001	Kappa
LAM006	Lambda
TCR001	TCR- $\alpha\beta$
TCR002	TCR- $\gamma\delta$
TDT001	TDT

Note:

1. **Required sample:** Heparinized/ EDTA 5 ml of peripheral blood or 3 ml of bone marrow.
2. All samples must be transported immediately at room temperature. Do not freeze the samples
3. Samples not meeting the requirements will not be accepted.
4. Rs. 800 extra will be charged for additional antibody.
5. Reports will be available within a minimum of 3 working days.



Genetic Tests - Karyotyping

Test Code	Sample	Report Duration
KAR010	Blood (2ml, heparinized tube) * Transport within 24 hours	7 days
KAR011	Amniotic fluid (20ml in sterile container) * Transport within 12 hours	20 days
KAR012	Chorionic villus sampling (10ml in HBSS, PBS or Saline in sterile container) * Transport within 12 hours	20 days
KAR013	Bone marrow (1ml in heparinized tube) * Transport immediately	8 days
KAR014	Percutaneous umbilical blood sampling (2ml in heparinized container) * Transport immediately	8 days
KAR015	Products of Conception (aseptically remove a small piece ~1 cm of fetal skin, or other types of fetal tissue, place in sterile normal saline and send it IMMEDIATELY to the laboratory)	21 days
CHR005	Chromosomal breakage syndrome Blood (2ml, heparinized tube) * Transport within 24 hours	8 days

Note:

1. All samples for karyotyping should be transported at room temperature **IMMEDIATELY** (unless otherwise mentioned) under aseptic precautions to enable proper culture of cells.
2. Sample-containing tubes should be protected from damage by padding with soft material.
3. Samples not meeting these requirements will NOT be accepted.

Other Tests

Test Code	Name	Sample	Report Duration
LEA003	Lead analysis (Blood) (Stripping voltametry)	2ml anticoagulated Blood (EDTA or Heparinized) Transport immediately at room temperature. Do Not Freeze	2 days
AAS001	Metal analysis (Atomic Absorption Spectrometry) Mercury/Lead/Zinc/ Copper/Iron/Cobalt/ Magnesium/Manganese/ Chromium/Nickel/ Aluminium/Molybdenum/ Calcium	2ml anticoagulated Blood (EDTA/Heparin) or Tissue/Hair/Soil/Medicinal extract Transport immediately at room temperature. Do Not Freeze	1 week