

DMCH MOLECULAR GENETICS LAB TESTS		
S.No	TEST NAME	SAMPLE TYPE
1	Apo-B Mutation Analysis	3ml EDTA Blood
2	Acute Pancreatitis (PRESS1, SPINK1, CFTR)	3ml EDTA Blood
3	Acute Myeloid Leukemia(AML)	3ml EDTA Blood /Bone Marrow
4	Acute Lymphoblastic Leukemia(ALL)	3ml EDTA Blood /Bone Marrow
5	Alpha Thalassemia	3ml EDTA Blood
6	BCR-ABL FISH	3ml Heparin Bone Marrow
7	Beta Thalassemia -Individual	3ml EDTA Blood
8	Beta Thalassemia Panel - Family	3ml EDTA Blood
9	BCR - ABL –Quantitative -RNA	3ml EDTA Blood
10	Clopirogrel Sensitivity	3ml EDTA Blood
11	CFTR delta 508 Genotyping	3ml EDTA Peripheral Blood
12	Chromosome Analysis (Aneuploidy)	3ml Heparin Blood
13	Chromosome Analysis (HM) (for Haem. Malignancies)	3ml Heparin Bone Marrow
14	CLL-Panel FISH	3ml Heparin Bone Marrow
15	Duchenne Muscular Dystrophy(DMD)	3ml EDTA Blood
16	DYPD Genotyping	3ml EDTA Blood
17	Factor v Leiden (R506Q) Genotyping	3ml EDTA Blood
18	FSHR (S680N) Genotype	3ml EDTA Blood
19	HFE Mutational Analysis	3ml EDTA Blood
20	HLA-B27 Genotyping	3ml EDTA Blood
21	HPV 16/18 DNA Testing	Cervical Brushing
22	HER2 FISH	Block/Slides
23	JAK-2 (V617F) Genotype	3ml EDTA Blood
24	MTHFR (A1298C and C677T) Genotyping	3ml EDTA Blood
25	Myeloproliferative Neoplasm(MPN)I	3ml EDTA Blood /Bone Marrow
26	Myeloproliferative Neoplasm(MPN)II	3ml EDTA Blood /Bone Marrow

27	Multiple Myeloma FISH	3ml Heparin Bone Marrow
28	MDS Panel FISH	3ml Heparin Bone Marrow
29	NUDT15 genotyping	3ml EDTA Blood
30	PML –RARA –Qualitative –RNA	3ml EDTA Blood
31	PML RARA FISH	3ml Heparin Bone Marrow
32	Prothrombin(G20210A) Genotyping	3ml EDTA Blood
33	Prenatal Beta Thalassemia –Carrier screening	CVS or 10ml amniotic fluid
34	Qualitative BCR-ABL -RNA	3ml EDTA Blood
35	Statin Sensitivity	3ml EDTA Blood
36	Thrombophilia panel (MTHFR, Factor V Leiden & Prothrombin)	3ml EDTA Blood
37	TPMT genotyping	3ml EDTA Blood
38	Warfarin Sensitivity	3ml EDTA Blood
39	XMN1 Polymorphism (Thalassemia)	3ml EDTA Blood
40	Y- Chromosome micro-deletion	3ml EDTA Blood
41	Breast Cancer BRCA1/2(NGS)	3ml EDTA Blood
42	BRCA1/2 Variant Screening(NGS)	3ml EDTA Peripheral Blood
43	BRAF Testing(NGS)	3ml bone Marrow (EDTA), (FFPE block)
44	EGFR Testing (NGS)	Formalin fix paraffin embed tissue (FFPE block)
45	GIST Testing (KIT, PDGFRA) (NGS)	FFPE Block
46	IDH1/IDH2 Testing(NGS)	3ml Bone Marrow/ Peripheral Blood (EDTA)
47	JAK2 reflex Panel(NGS)	3ml EDTA Blood /Bone Marrow
48	PIK3CA Testing (NGS)	Formalin fix paraffin embed tissue (FFPE block)
49	Somatic BRCA 1/2 Testing (NGS)	Formalin fix paraffin embed tissue (FFPE block)