TEST NAME	SPECIMEN / REQUIREMENT
Thrombophilia Screening	
Factor V Leiden mutation screening	EDTA PB
Prothrombin G20210A mutation screening	EDTA PB
Methylenetetrahydrofolate reductase gene C677T mutation screening	EDTA PB
Hereditary Haemochromatiosis Gene Mutation Screening	
C282Y & H63D mutation of HFE gene	EDTA PB
Thalassaemia & Haemoglobinopathy Investigation	
α-thalassaemia genotyping	
α^0-thalassaemia test 1 – [5 common deletions: $--^{SEA}$, $-(\alpha)^{20.5}$, $--^{MED}$, $--^{FIL}$, and $--^{THAI}$]	EDTA PB
α^+-thalassaemia test 2 – [$-\alpha^{3.7}$]	EDTA PB
α^+-thalassaemia test 3 – [$-\alpha^{4.2}$]	EDTA PB
α-thalassaemia test 4 – [α-globin gene DNA sequencing]	EDTA PB
α-thalassaemia test 5 – [Hb Constant Spring and Hb Quong Sze mutation]	EDTA PB
β-thalassaemia genotyping	
β-thalassaemia test – [β-globin gene DNA sequencing]	EDTA PB
Hereditary Persistence of Fetal Haemoglobin screening [HPFH-1,2,3,6 and SEA] and Chinese $^{G}\gamma\ (^{A}\gamma\delta\beta)^0$ thalassaemia	EDTA PB
Lymphoproliferative Diseases (LPDs)	
PCR for BCL-1 translocation t(11;14)(q13;q32)	EDTA PB / BM / Tissue Block
PCR for BCL-2 translocation t(14;18)(q32;q21) – Major breakpoint region	EDTA PB / BM / Tissue Block
Myeloproliferative Diseases (MPDs)	
JAK2 tyrosine kinase gene Val617Phe Mutation Screening	EDTA PB/BM

ZNF198/FGFR1 derived from PhNEG myeloproliferative disorder with t(8;13)	EDTA PB/BM
Acute Lymphoblastic Leukaemia (ALL)	
MLL/AF4 derived from ALL with t(4;11)(q21;q23)	Fresh EDTA BM (preferable) / PB
E2A/PBX1 derived from ALL with t(1;19)(q23;p13)	Fresh EDTA BM (preferable) / PB
TEL/AML1 derived from ALL with t(12;21)(p13;q22)	Fresh EDTA BM (preferable) / PB
BCR/ABL P190 (e1a2) derived from ALL with t(9;22)(q34;q11)	Fresh EDTA BM (preferable) / PB
Acute Myeloid Leukemia (AML)	
AML1/ETO derived from AML with t(8;21)(q22;q22)	Fresh EDTA BM (preferable) / PB
CBFβ/MYH11 derived from AMLEso with inv(16)(p13;q22)	Fresh EDTA BM (preferable) / PB
MLL/AF9 derived from AML with t(9;11)(p22;q23)	Fresh EDTA BM (preferable) / PB
MLL/AF10 derived from AML with t(10;11)(p12;q23)	Fresh EDTA BM (preferable) / PB
PML/RARα derived from AML with t(15;17)(q22;q21)	Fresh EDTA BM (preferable) / PB
NPM1 mutation test	Fresh EDTA BM (preferable) / PB
Chronic Meyloid Leukemia (CML)	
BCR/ABL (b2a2 & b3a2) derived from CML with t(9;22)(q34;q11)	Fresh EDTA BM (preferable) / PB
Quantitative RT-PCR for BCR/ABL transcript (b2a2 & b3a2)	Fresh EDTA PB (3 x 3 ml)
Chronic Myelomonocytic Leukemia & Myelodysplastic Syndrome (CMML & MDS)	
TEL/PDGFRβ derived from CMML / MDS with t(5;12)(q33;p13)	Fresh EDTA BM (preferable) / PB
Chronic Eosinophilic Leukemia (CEL)	
FIP1L1/PDGFRα derived from CEL with an interstitial deletion on chromosome 4q12	Fresh EDTA BM (preferable) / PB
Tyrosine Kinase Mutation Screening	
FLT3-ITD mutation screening	Fresh EDTA BM (preferable) / PB

c-Kit exon 8 and 17	Fresh EDTA BM (preferable) / PB
BCR/ABL tyrosine kinase domain	Fresh EDTA BM (preferable) / PB
Fluorescence In-situ Hybridization (FISH) Detection of Gene Fusion	
BCR-ABL	Fresh EDTA BM (preferable) / PB
PML-RARα	Fresh EDTA BM (preferable) / PB
AML1-ETO	Fresh EDTA BM (preferable) / PB
TEL-AML1	Fresh EDTA BM (preferable) / PB
Giant Platelet Syndrome	
MYH9 mutation screening (Exon 16, 26, 30, 38, 40)	EDTA PB
Neuroblastoma Monitoring of Residual Disease	
Tyrosine hydroxylase transcripts	Fresh EDTA BM (preferable) / PB
Fluorescence In-situ Hybridization (FISH) Detection of Chromosomal deletion & translocation for Multiple Myeloma	
Tyrosine hydroxylase transcripts	Fresh EDTA BM (preferable) / PB
Neuroblastoma Monitoring of Residual Disease	
HLA-B*15:02 for carbamazepine prescription	EDTA PB (3 mL)
HLA-B*58:01 for allopurinol prescription	EDTA PB (3 mL)
Remarks:	
1. Please send either 3 ml Bone Marrow or 6 ml Peripheral Blood in EDTA for molecular tests. Specimen should be sent before 3pm.	
2. Please call 2632 3949 for advance booking for all the above tests.	