

MOLECULAR HAEMATOPATHOLOGY

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 Haemochromatosis Gene Mutation Screening	
C282Y & H63D gene mutation	EDTA PB
Thalassaemia & Haemoglobinopathy Investigation	
α-thalassaemia genotyping	
α^0 -thalassaemia test 1 - [5 common deletions: -- ^{SEA} , - (α) ^{20.5} , -- ^{MED} , -- ^{FIL} , and -- ^{THAI1}]	EDTA PB
α^+ -thalassaemia test 2 - [$-\alpha^{3.7}$ and $\alpha\alpha^{\text{anti}3.7}$]	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/or Hb Quong Sze mutation]	EDTA PB
α -thalassaemia tests panel (Tests 1-5)	
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 BLK
PCR for BCL-2 translocation t(14;18)(q32;q21) - Major breakpoint region	EDTA PB/BM/ Tissue BLK
IgH gene rearrangement (clonality assessment)	EDTA PB/BM/ Tissue BLK
T-cell receptor gamma / beta gene rearrangement (clonality assessment)	EDTA PB/BM/ Tissue BLK

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Myeloproliferative Diseases (MPDs)	
JAK2 tyrosine kinase gene Val617Phe Mutation Screening	EDTA PB/BM
ZNF198/FGFR1 derived from Ph ^{NEG} 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 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
Chronic Myeloid 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

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TEST NAME	SPECIMEN / REQUIREMENT
Tyrosine Kinase Mutation Screening	
FLT3 D835 (Qualitative)	Fresh EDTA BM (preferable) / PB
FLT3 D835 (Characterized)	Fresh EDTA BM (preferable) / PB
FLT3 internal tandem duplication (Qualitative)	Fresh EDTA BM (preferable) / PB
FLT3 internal tandem duplication (Characterized)	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-RAR α	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

Remarks:

1. Please send either 3 ml Bone Marrow (BM), or 10 ml Periperal Blood (PB) in EDTA for any molecular testings, and send it before 3pm within 4 hours collected.
2. Please call 2632-3949 for advance booking for all the above tests.