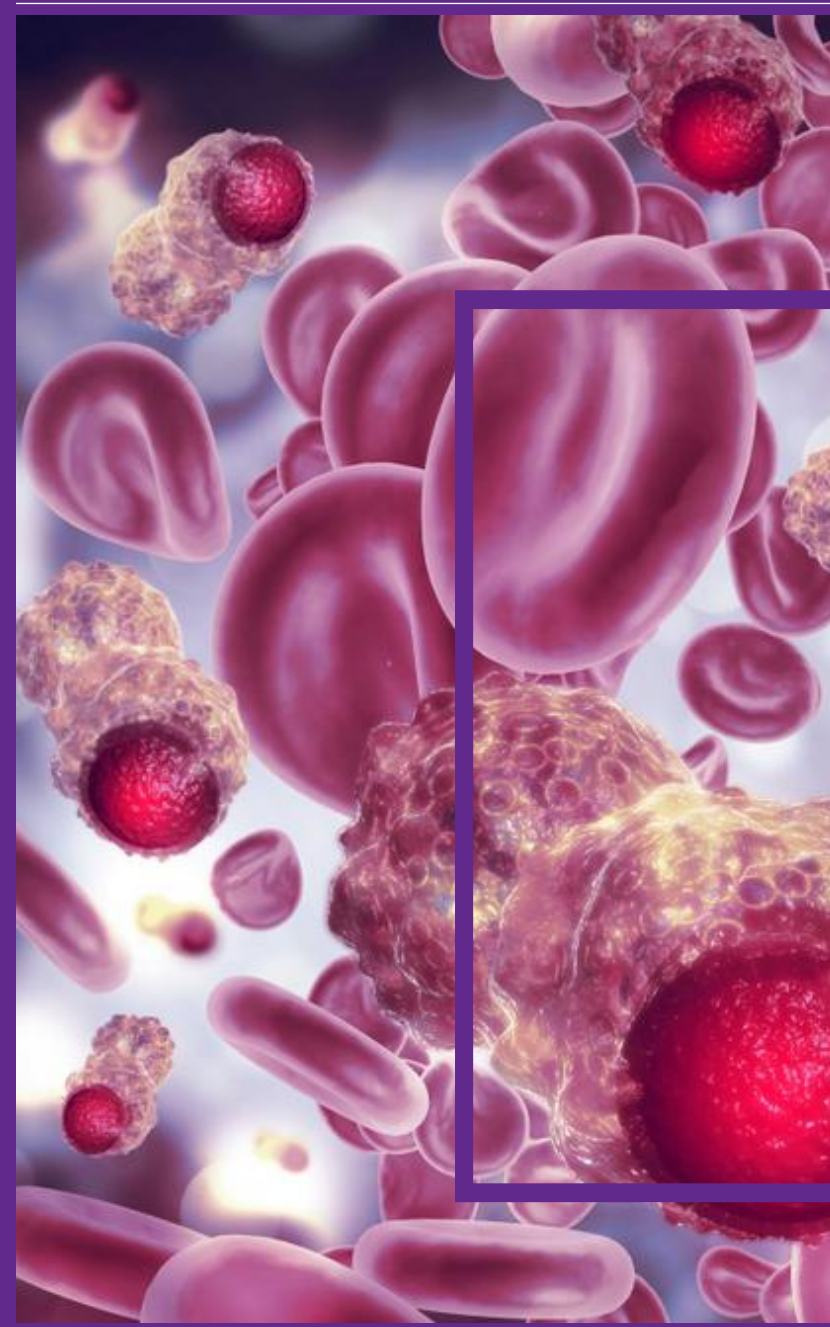


Powered by



Phone : +91-6357244307, 079-61618111  
Email : [contact@ncgmglobal.com](mailto:contact@ncgmglobal.com)  
Website : [www.ncgmglobal.com](http://www.ncgmglobal.com)



# OncoCEPT HAEM

# OncoCEPT - HAEM

- Sample Type:- Blood or Bone Marrow
- Next Generation Sequencing
- TAT:-10 days\*

## Genes Covered

- 40 key DNA target genes
- 29 driver genes
- (a broad fusion panel)

## Diseases Covered

### Disorder wise:

- 1. Myeloproliferative Neoplasms**
  - a. Polycythaemia Vera (PV)
  - b. Primary Myelofibrosis (PMF)
  - c. Essential thrombocythaemia (ET)
  - d. Chronic myeloid leukemia, BCR-ABL1 positive
  - e. Chronic neutrophilic leukaemia
  - f. Chronic eosinophilic leukaemia, NOS
  - g. Myeloproliferative Neoplasm, unclassifiable
- 2. Mastocytosis**
- 3. Myeloid/lymphoid neoplasms with eosinophilia & gene rearrangement**
  - a. Myeloid/lymphoid neoplasms with PDGFRA rearrangement
  - b. Myeloid/lymphoid neoplasms with PDGFRB rearrangement
  - c. Myeloid/lymphoid neoplasms with FGFR1 rearrangement
  - d. Myeloid/lymphoid neoplasms with PCM1-JAK2

- 4. Myelodysplastic/myeloproliferative neoplasms**
  - a. Chronic myelomonocytic leukaemia (CMML)
  - b. Atypical chronic myeloid leukaemia, BCR-ABL1 negative (aCML)
  - c. Juvenile myelomonocytic leukaemia (JMML)
  - d. Myelodysplastic/myeloproliferative neoplasm with ring sideroblasts & thrombocytosis
  - e. Myelodysplastic/myeloproliferative neoplasm, unclassifiable (MDS/MPN-U)
- 5. Myelodysplastic syndromes(MDS)**
  - a. MDS with single lineage dysplasia
  - b. MDS with ring sideroblasts
  - c. MDS with multilineage dysplasia
  - d. MDS with excess blasts
  - e. MDS with isolated del(5q)
  - f. MDS, unclassifiable
  - g. Childhood myelodysplastic syndrome

## Diseases Covered

### Disorder wise:

- 6. Myeloid neoplasms with germline predisposition (CEBPA, RUNX1, ETV6, GATA2)**
- 7. Acute myeloid leukaemia(AML) & related precursor neoplasms**
  - a. AML with t(8;21)/RUNX1-RUNX1T1
  - b. AML with inv(16)/CBFB-MYH11
  - c. AML with PMLRARA/variant RARA
  - d. AML with t(9;11)/KMT2A-MLL3 as well as other KMT2A rearrangements
  - e. AML with t(6;9)/DEK-NUP214
  - f. AML with t(1;22)/RBM15-MKL1
  - g. AML with t(9;22)/BCR-ABL1
  - h. AML with mutated NPM1
  - i. AML with biallelic mutation of CEBPA
  - j. AML with mutated RUNX1
  - k. AML with myelodysplasia related changes
  - l. Therapy related myeloid neoplasms
  - m. AML, not otherwise specified
  - n. Myeloid sarcoma
- 8. Acute leukaemias of ambiguous lineage**
  - a. Acute undifferentiated leukaemia
  - b. Mixed phenotype acute leukaemia with t(9;22)/BCR-ABL1
  - c. Mixed phenotype acute leukaemia with t(v;11q23.3)/KMT2A rearranged
  - d. Mixed phenotype acute leukaemia, B/myeloid
  - e. Mixed phenotype acute leukaemia, T/myeloid
- 9. Precursor lymphoid neoplasms : B-lymphoblastic leukaemia/lymphoma (B-ALL/LBL)**
  - a. B-ALL/LBL, not otherwise specified
  - b. B-ALL/LBL with t(9;22)/BCR-ABL1
  - c. B-ALL/LBL with t(v;11q23.3)/KMT2A rearranged
  - d. B-ALL/LBL with t (12;21) / ETV6-RUNX1
  - e. B-ALL/LBL with t(1;19)/TCF3-PBX1
  - f. B-ALL/LBL with ZNF384 related fusion
- 10. Mature B-neoplasms**
  - a. Chronic lymphocytic leukaemia (CLL)
  - b. Hairy cell leukaemia (HCL) (BM sample preferred)
  - c. Lymphoplasmacytic lymphoma/ Waldenstrom macroglobulinemia (LPL/WM) (BM sample preferred)

\*TAT mentioned is for working days for >90% samples

**CBC/BM findings under evaluation whenever a myeloid neoplasm is suspected(after ruling out other reactive/secondary causes):**

- |   |  |
|---|--|
| 1. Polycythemia   | 6. Dyspoiesis(Dyserythropoiesis/<br>dysgranulopoiesis/dysmegakaryopoiesis) |
| 2. Cytopenia(Anaemia, leucopenia,<br>thrombocytopenia)                | 7. Ring sideroblasts   |
| 3. Leukocytosis(Neutrophilia/eosinophilia/<br>monocytosis/basophilia) | 8. Myelofibrosis   |
| 4. Thrombocytosis   | 9. Atypical lymphoid cells with hairy<br>cytoplasmic projections           |
| 5. Acute leukaemia(blasts/atypical cells)                             | 10. Rouleaux formation with<br>lymphoplasmacytic cells                     |

**List of Genes in Panel**

DNA						
HOTSPOT GENES				FULL GENES		
ABL1	FLT3	KIT	SETBP1	ASXL1	IKZF1	SH2B3(LNK)
BRAF	GATA2	KRAS	SF3B1	BCOR	NF1	STAG2
CBL	HRAS	MPL	SRSF2	CALR	PHF6	TET2
CEBPA	IDH1	MYD88	U2AF1	ETV6	PRPF8	TP53
CSF3R	IDH2	NPM1	WT1	EZH2	RB1	ZRSR2
DNMT3A	JAK2	PTPN11			RUNX1	

RNA				
FUSION DRIVER GENES				
ABL1	EGFR	JAK2	MYBL1	RARA
ALK	ETV6 (TEL)	KMT2A(MLL)	MYH11	RBM15
BCL2	FGFR1	MECOM	NTRK3	RUNX1(AML1)
BRAF	FGFR2	MET	NUP214	TCF3(E2A)
CCND1	FUS	MLLT10	PDGFRA	TFE3
CREBBP	HMGA2	MLLT3	PDGFRB	

**Mutations (DNA)**

AML			MDS/MPN				PMF		CNL
NPM1	FLT3-TKD	ASXL1	SF3B1	SRSF2	NRAS	ZRSR2	JAK2	IDH1	CSF3R
CEBPA	TP53	DNMT3A	JAK2	DNMT3A	TP53	STAG2	MPL	IDH2	SETBP1
RUNX1	WT1	IDH1	MPL	U2AF1	SETBP1	IDH1	CALR	SRSF2	ASXL1
KIT	TET2	IDH2	CALR	TP53	Others	IDH2	ASXL1	SF3B1	JAK2
FLT3-ITD			TET2	EZH2		BCOR	EZH2	TET2	

CMML	t-MNs	Mastocytosis	JMML
ASXL1	TP53	KIT	PTPN11
TET2	IDH2	CBL	CBL
SRSF2	TET2	RUNX1	KRAS
RUNX1	PTPN11	RAS family	NRAS
	IDH1	ASXL1	NF1
			SH2B3
			ASXL1

MNGP	CEL	TAM	PV	ET
CEBPA	GATA2	EZH2	JAK2V617F	JAK2
RUNX1	Same as JMML	SH2B3	JAK2 exon 12	CALR
ETV6	CSF3R	RAS pathway genes	SH2B3/LNK	MPL

MDS			BPDCN	
SF3B1	RUNX1	STAG2	TET2	IDH2
TET2	U2AF1	IDH1	NPM1	KIT
ASXL1	TP53	IDH2	ASXL1	RB1
SRSF2	EZH2	CBL	RAS family	BRAF
DNMT3A	ZRSR2	NRAS	NRAS	TP53
	Others	BCOR	KRAS	

aCML	APL
SETBP1	FLT3-ITD
CSF3R	FLT3-TKD

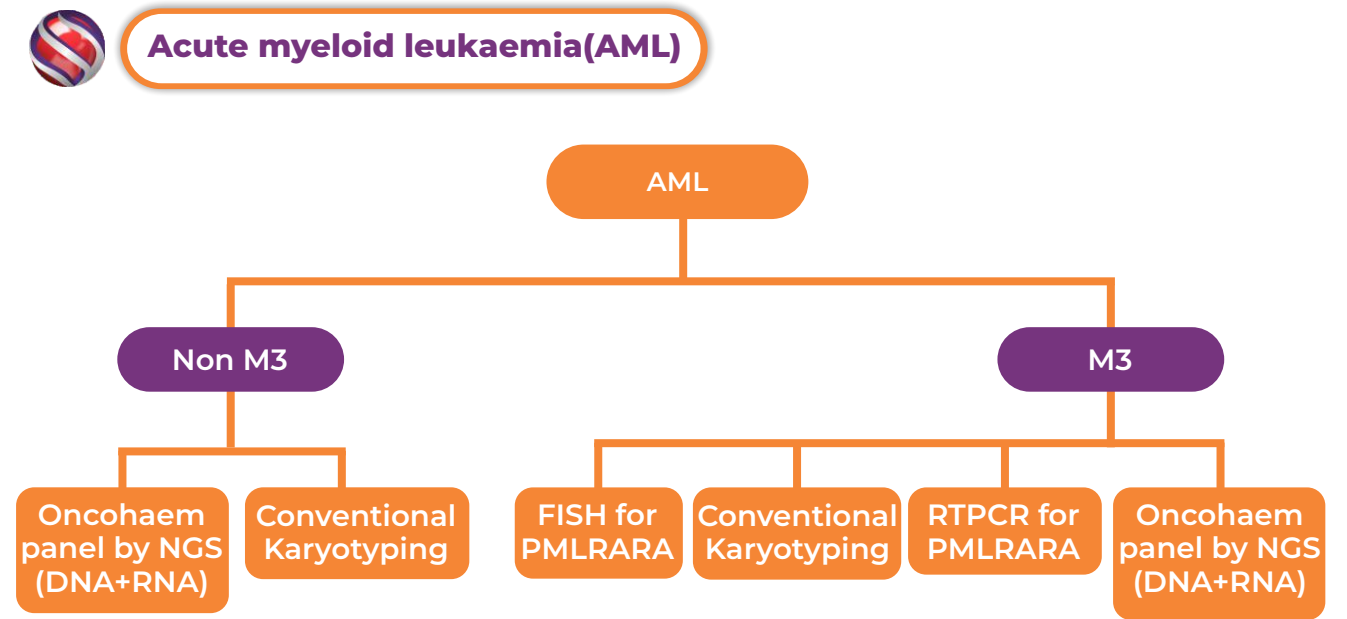
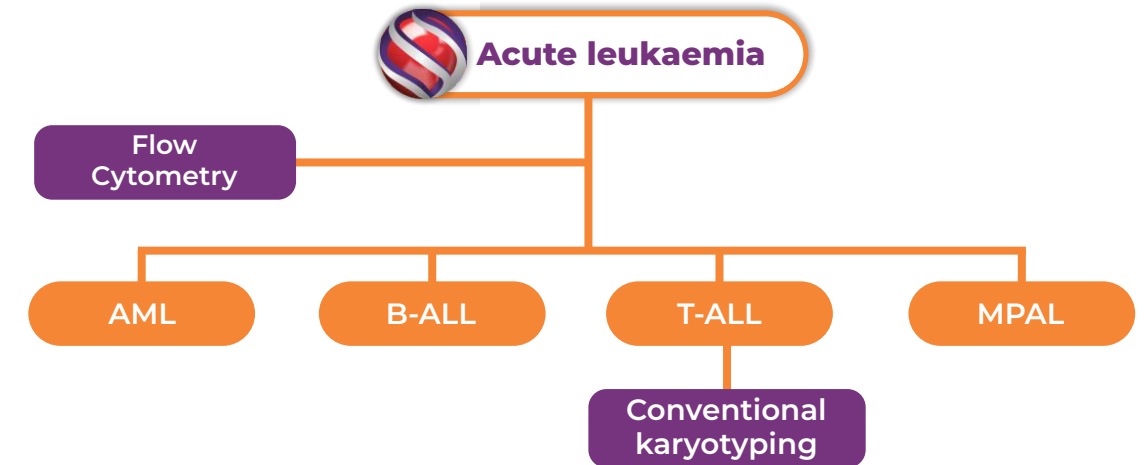
- Acute myeloid leukaemia (AML)
- Primary Myelofibrosis (PMF)
- Therapy related myeloid neoplasms (t-MNs)
- Myeloid proliferations ass. with Down's (TAM)
- Chronic eosinophilic leukaemia (CEL)
- Acute promyelocytic leukaemia (APL)
- Chronic neutrophilic leukaemia (CNL)
- Mastocytosis
- Chronic myelomonocytic leukaemia (CMML)
- Juvenile myelomonocytic leukaemia (JMML)
- Myelodysplastic Syndrome / myeloproliferative neoplasms (MDS/MPN)
- Myelodysplastic Syndrome(MDS)
- Myeloid neoplasms with germline predisposition(MNGP)
- Atypical chronic myeloid leukaemia (aCML)
- Essential Thrombocythemia (ET)
- Polycythemia (PV)
- Blastic plasmacytoid dendritic cell neoplasm(BPDCN)

# Fusions (RNA)

Acute myeloid leukaemia(AML) and related precursor neoplasms			Acute promyelocytic leukaemia			Chronic eosinophilic leukaemia			Pediatric -AML		
RUNX1-RUNX1T1 t(8;21) CBFB-MYH11 inv(16)			RARA REARRANGEMENTS: 3 types bcr1,bcr2,bcr3			ETV6 REARRANGEMENTS			RBM15-MKL1 t(1;22) KAT6A-CREBBP		
PML-RARA t(15;17) DEK-NUP214 t(6;9)			IRF2BP2-RARA			FLT3			DEK-NUP214 t(6;9)		
RBM15-MKL1 t(1;22) AML with BCR-ABL1			NABP1-RARA t(2;17)			NTRK			KMT2A Rearrangements		
KMT2A REARRANGEMENTS			TBLX1R1-RARA t(4;17)			LYN			MLLT3		
MLLT3 t(9;11)	TET1	CASP8AP2	FIP1L1-RARA t(5;17)			SYK			MLLT10		
AFF1/AF4	PICALM	CBL	NPM1-RARA t(11;17)			ABL1			MLLT1		
MLLT1-ENL	ABI1	CREBBP	NUMA1-RARA			ETV6-JAK2			Chronic myeloid leukaemia		
MLLT10/AF10	CASC5	DCPIA	ZBTB16-RARA t(11;17)			BCR-JAK2			BCR-ABL1		
ELL	MYO1F	DCPS	ADAMTS17-RARA			B-lymphoblastic leukaemia/lymphoma					
PTD	SEPT5	FNBP1	STAT5B-RARA			t(9;22)/BCR-ABL1			t(1;19)/TCF3-PBX1		
MLLT4/AF6	ACTN4	GAS7	PRKARIA-RARA t(17;17)			t(v;11q23.3)/KMT2A			ZNF384 related Fusions		
EPS15	FLNA	KIA1524	BCOR-RARA t(X;17)			t(12;21)/ETV6-RUNX1					
MLLT11/AF10	FOXO3	BTBD18	PML-RARA								
SEPT9	CEP170B	MYH11									
EPS15	MAML2	NEBL									
SEPT6	SEPT11	NRIP3									
MLLT6/AF17	ABI2	PDS5A									
AFF3/LAF4	ACACA	SEPT2									
ARHGEF12	AFF4/AF5	SMAP1									
GMPS	ARHGEF17	TOP3A									

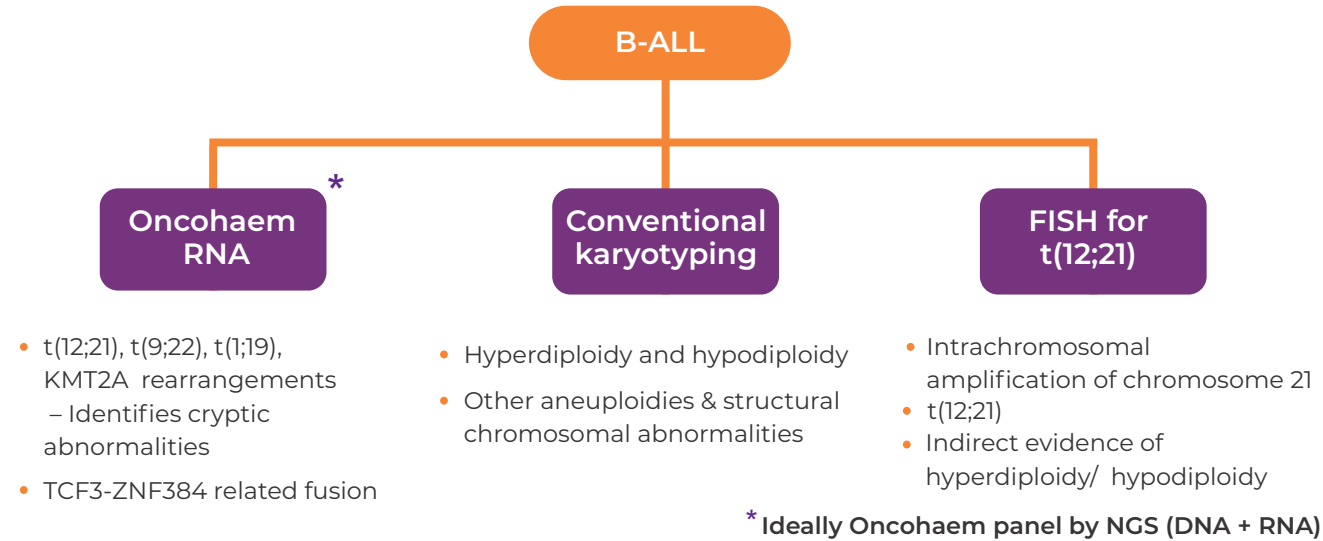
Myeloid/lymphoid neoplasms with eosinophilia & gene arrangement			
PDGFRB REARRANGEMENTS		FGFR1 REARRANGEMENTS	
CCDC6	GOLGA4	ZMYM2	JAK2 REARRANGEMENTS
SART3	TNIP1	CNTRL	PCM1
GIT2	HIP1	FGFR1OP	ETV6
ERC1	KANK1	BCR	BCR
BIN2	MYO18A	MYO18A	PDGFRA REARRANGEMENTS
NIN	COL1A1	TRIM24	ETV6
CCDC88C	DTD1	FGFR1OP2	FIP1L1
TP53BP1	GOLGB1	TPR	KIF5B
NDE1	CEP85L	RANBP2	CDK5RAP2
RABEP1	TRIP11	LRRFIP1	STRN
SPECC1	MPPRIP	CUX1	FOXP1
ETV6	CPSF6	CPSF6	TNKS2
WDR48	TPM3	SQSTM1	BCR
CAPRIN1	PDE4DIP		
	PRKG2		

BONE MARROW EXAMINATION IS MUST FOR ALL SUSPECTED MYELOID NEOPLASMS (except under special circumstances)

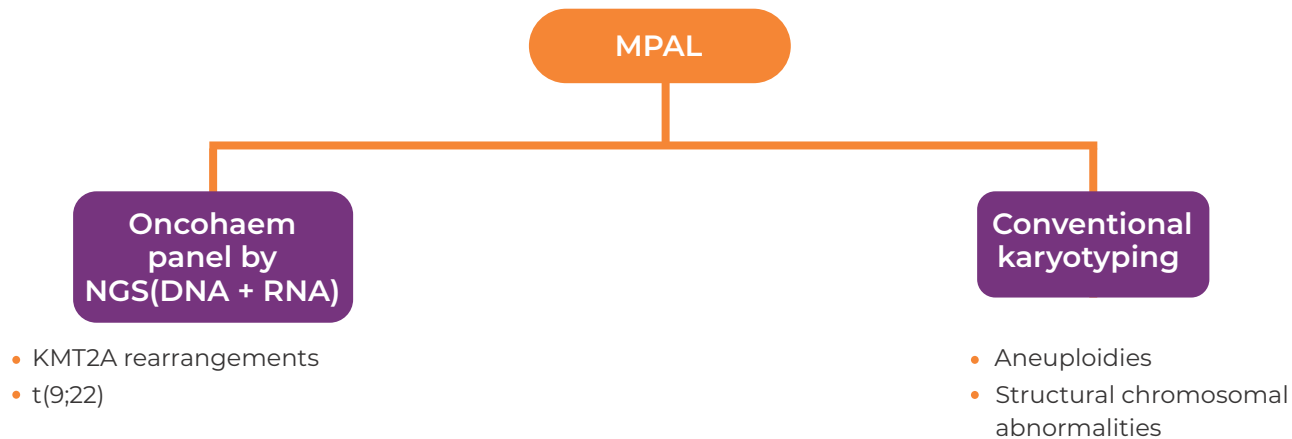




## B-lymphoblastic leukaemia(B-ALL)



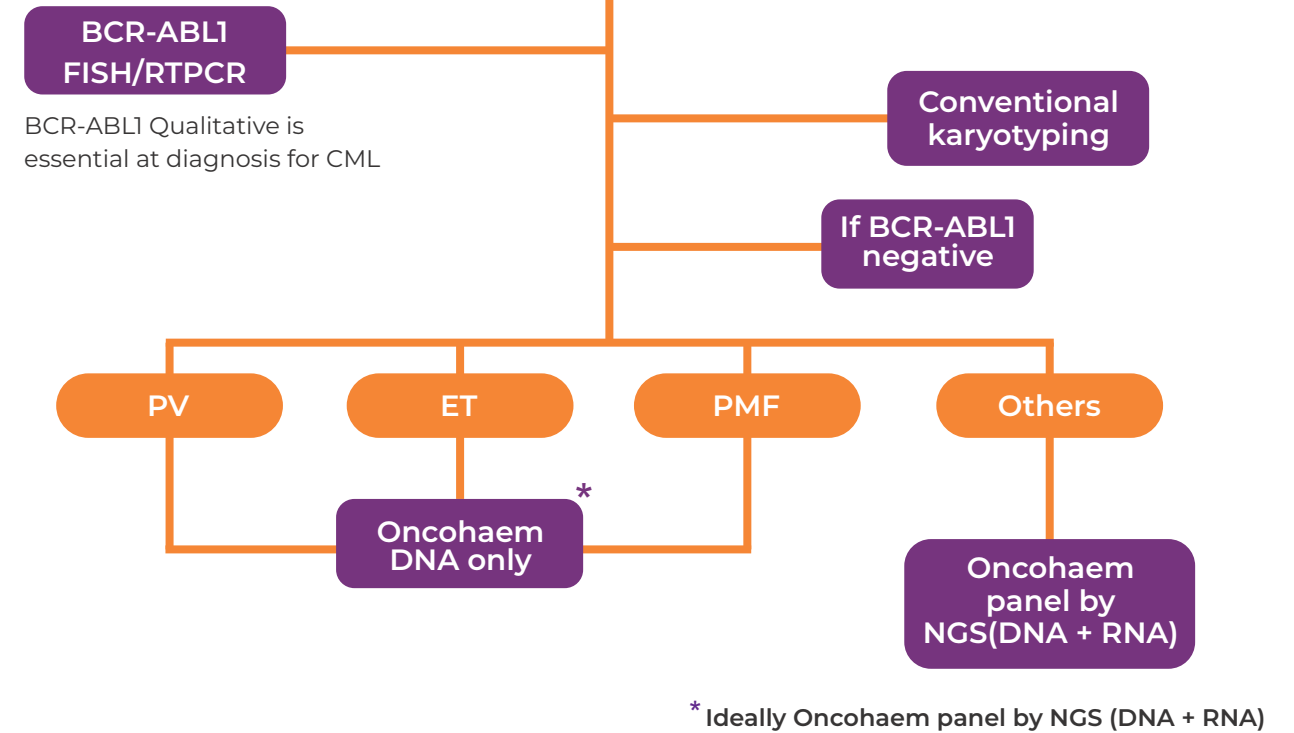
## Mixed phenotype acute leukaemia (MPAL)



## Myeloproliferative Neoplasms (MPN)



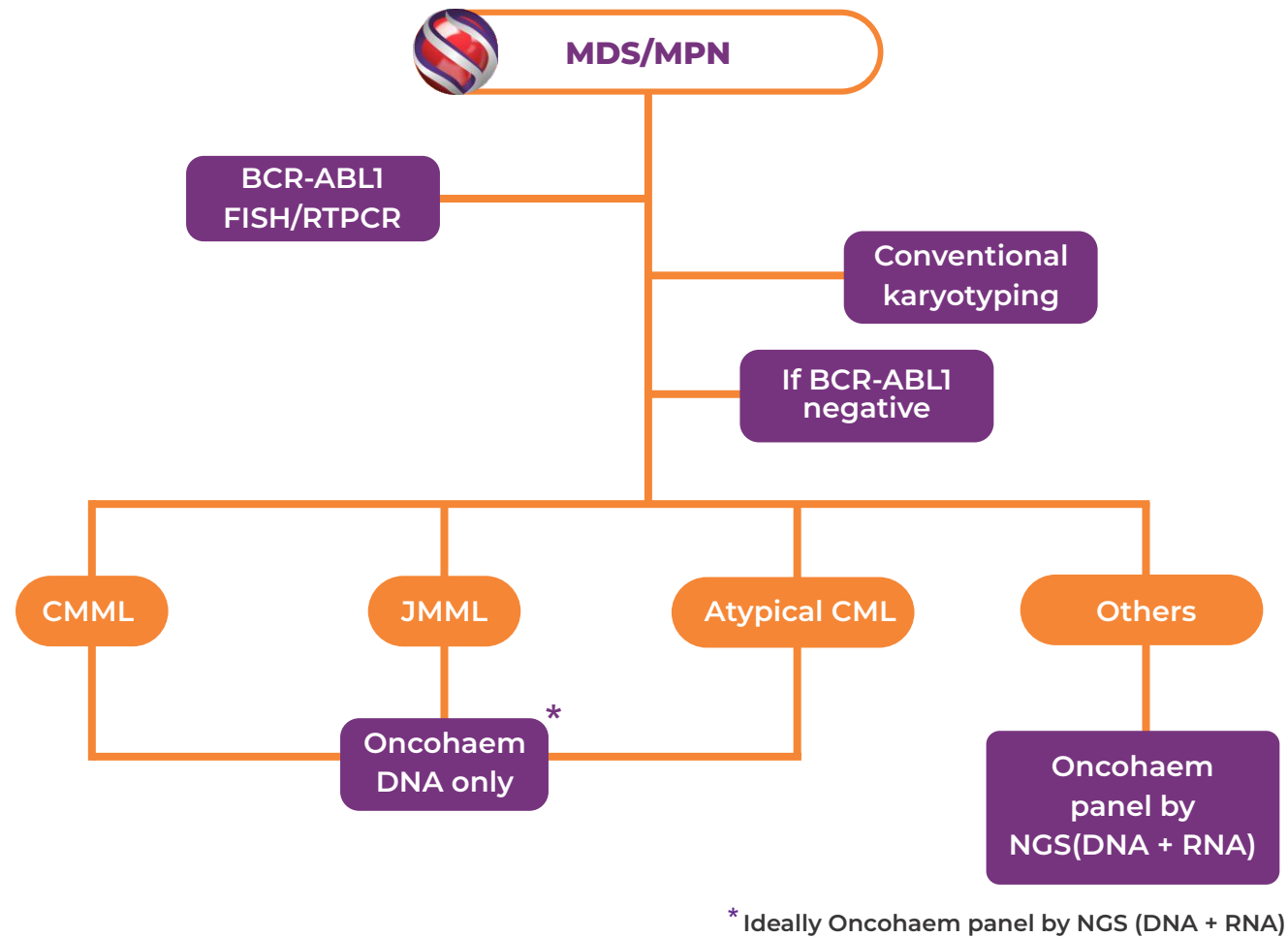
## Myeloproliferative Neoplasms (MPN)



- Chronic myeloid leukaemia(CML)
- Polycythaemia Vera(PV)
- Primary Myelofibrosis(PMF)
- Essential Thrombocythaemia(ET)
- Chronic neutrophilic leukaemia(CNL)
- Chronic eosinophilic leukaemia(CEL)
- MPN-U

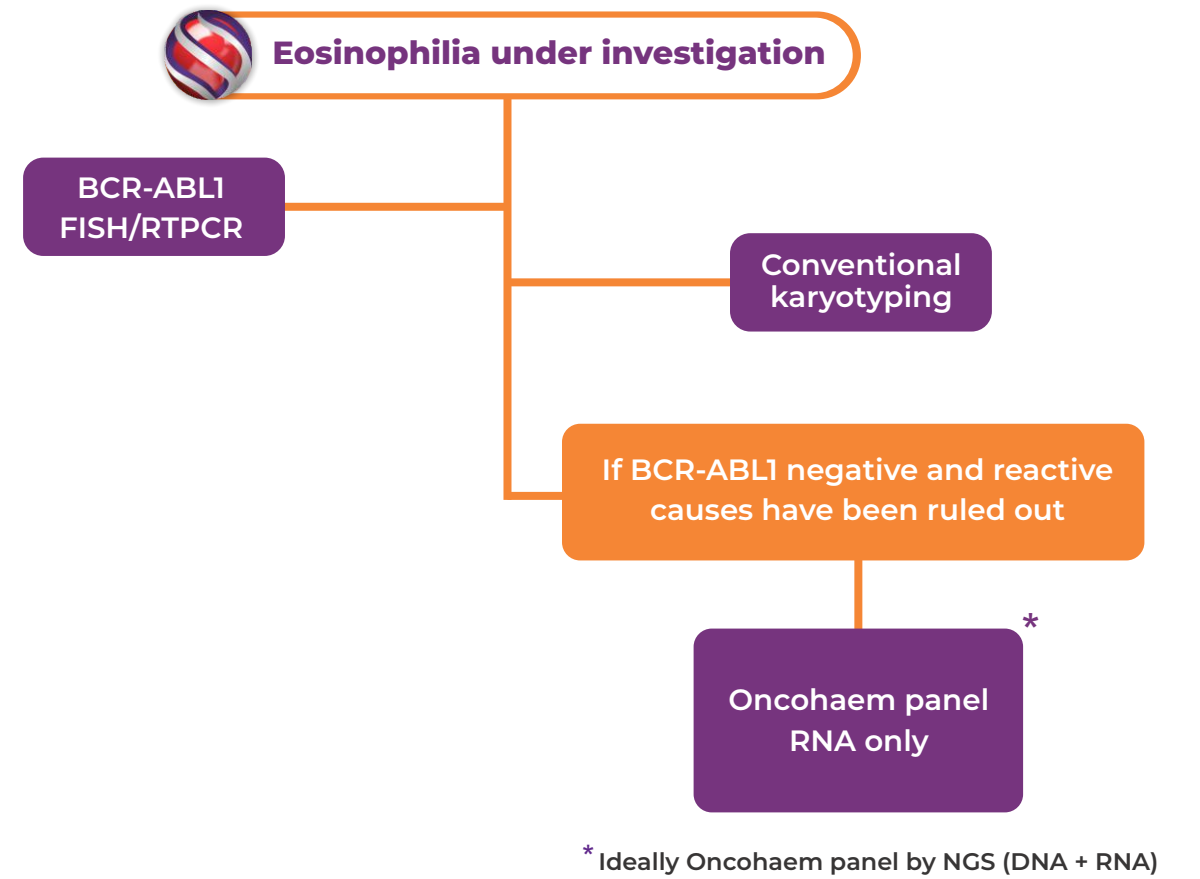


## Myelodysplastic syndrome/ Myeloproliferative neoplasm (MDS/MPN)



- Chronic myelomonocyticleukaemia(CMML)
- Atypical CML(aCML)
- Juvenile myelomonocytic leukaemia(JMML)
- MDS/MPN-RS-T
- MDS/MPN-U

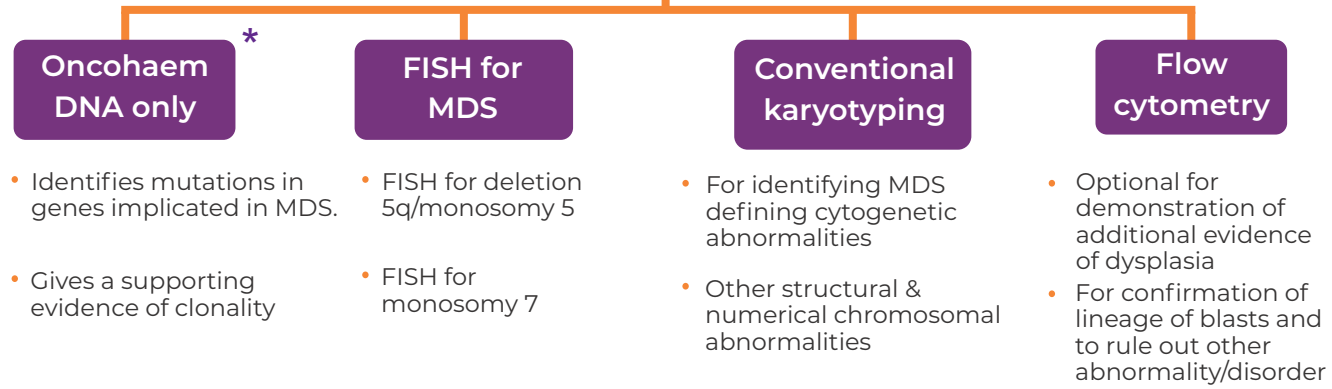
## Eosinophilia under investigation



\* Ideally Oncohaem panel by NGS (DNA + RNA)

## Myelodysplastic Syndrome (MDS)

### Myelodysplastic Syndrome (MDS)



\* Ideally Oncohaem panel by NGS (DNA + RNA)

## Chronic lymphocytic Leukaemia (CLL)

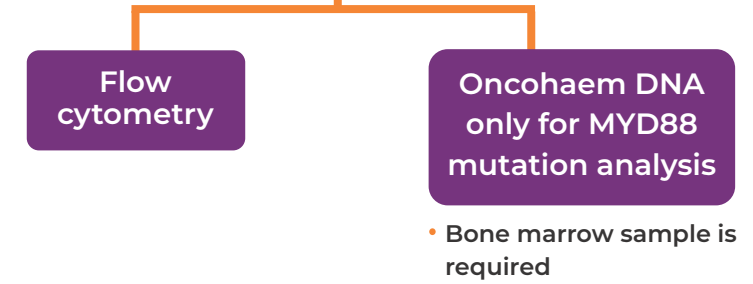
### Chronic lymphocytic leukaemia



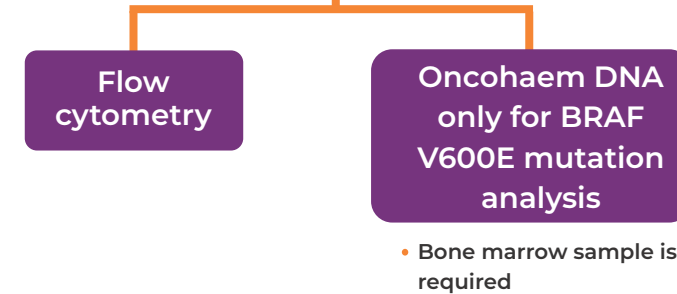
- Flow cytometry**
  - For confirmation of Chronic lymphocytic leukaemia & exclusion of other Non Hodgkin lymphoma
- FISH for CLL**
  - Especially for del17p(TP53)
  - For knowing other abnormalities like del(13q), trisomy 12, del(11q), etc
- Oncohaem DNA only**
  - For knowing TP53 mutation (for prognostication) as well as mutations in SF3B1 and MYD88 genes
  - (TP53 mutation and FISH for TP53 deletion are different)
- IGVH mutation analysis**
  - For prognostication

## Other Disorders

### WM/LPL



### HCL



- Hairy cell leukaemia(HCL)
- Waldenstrom Macroglobulinemia/Lymphoplasmacytic lymphoma(WM/LPL)

Current analysis of hematological malignancies involves multiple sequential tests and laborious workflows. Adoption of next-generation sequencing (NGS) methods into clinical research laboratories has created an unprecedented opportunity to profile the multiple relevant driver genes in myeloid malignancies. Targeted NGS assay is designed to assist in the understanding of myeloid cancers. Specifically, it interrogates all relevant DNA mutations and fusion transcripts associated with myeloid disorders in a single NGS run.

### Why OncoCEPT - HAEM?

- Targetable fusions/rearrangements like PDGFRA, PDGFRB, etc. can be checked
- Rare transcripts, which are not regularly checked or are usually missed, can also be detected. e.g. ETV6-JAK2 and ETV6-FLT3 can be found in CEL which can be detected by this technique.
- Transcripts that are cryptic can also be detected, which karyotyping may fail to identify.
- Fusions resulting from translocations can be confirmed. e.g. not all translocation characterized as t(5;12)(q31-33;p12) lead to ETV6-PDGFRB fusion.
- Fusions suspected but not found by karyotyping can be detected.
- Multiple fusions of a particular gene can be seen, whereas, FISH can only detect a particular translocation depending upon the probe used.
- Testing individually for each fusion by FISH is expensive, however, here, whole panel can be covered in a very reasonable rate.
- Mutations other than FLT3, JAK2, NPM1 & CEBPA are also covered like – IDH2, DNMT3A, etc.
- A suspected case can be confirmed if mutation can be found. e.g. a suspected case of JMML having a mutation KRAS can confirm the diagnosis.
- Drugs are now available that can be used to target the mutations. e.g. IDH1 inhibitor.
- Mutation can help in knowing the prognosis of the disease. e.g. TP53 mutation in AML carries adverse prognosis.

Your  
**PARTNERS**  
in HEALTH 

#### **Dr. Arpan Mehta**

**Laboratory Haematologist &  
Molecular Haemato-oncologist**  
arpan.mehta@supratechlabs.com  
**+91-9978338329**

#### **Dr. Parth Shah**

**Scientific Consultant**  
parth.shah@supratechlabs.com  
**079-40408181**

#### **Dr. Sandip Shah**

**Consultant Pathologist  
M.D (Pathology & Bacteriology)  
Laboratory Director**  
drsandip@neubergdiagnostics.com  
**079-40408181**