

New INSIGHTS



Neuberg
DIAGNOSTICS

CENTER FOR
GENOMIC
MEDICINE



OncoCEPT HAEM

OncoCEPT - HAEM

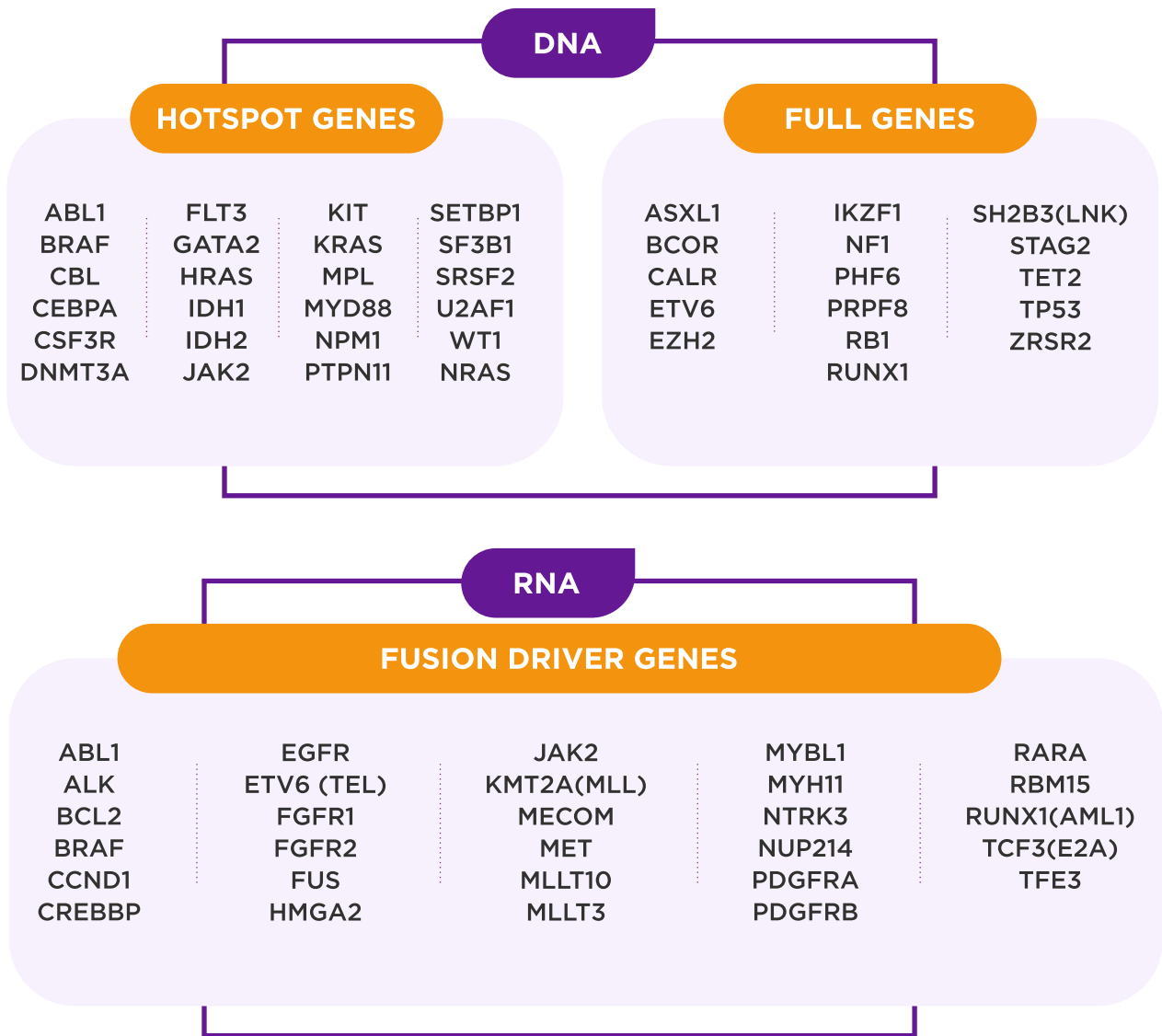
- ▶ Sample Type:- Blood or Bone Marrow
- ▶ Next Generation Sequencing
- ▶ TAT:- 10 working days*

GENES COVERED

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

*for >90% samples

List of Genes in Panel



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

1. Polycythemia
2. Cytopenia(Anaemia, leucopenia, thrombocytopenia)
3. Leukocytosis(Neutrophilia/eosinophilia/monocytosis/basophilia)
4. Thrombocytosis
5. Acute leukaemia (blasts/atypical cells)

6. Dyspoiesis(Dyserythropoiesis/dysgranulopoiesis/dysmegakaryopoiesis)
7. Ring sideroblasts
8. Myelofibrosis
9. Atypical lymphoid cells with hairy cytoplasmic projections
10. Rouleaux formation with lymphoplasmacytic cells

Diseases Covered

Disorder wise:

1. Myeloproliferative Neoplasms

- Polycythaemia Vera (PV)
- Primary Myelofibrosis (PMF)
- Essential thrombocythaemia (ET)
- Chronic myeloid leukemia, BCR-ABL1 positive
- Chronic neutrophilic leukaemia
- Chronic eosinophilic leukaemia, NOS
- Myeloproliferative Neoplasm, unclassifiable

2. Mastocytosis

3. Myeloid/lymphoid neoplasms with eosinophilia & gene rearrangement

- Myeloid/lymphoid neoplasms with PDGFRA rearrangement
- Myeloid/lymphoid neoplasms with PDGFRB rearrangement
- Myeloid/lymphoid neoplasms with FGFR1 rearrangement
- Myeloid/lymphoid neoplasms with PCM1-JAK2

4. Myelodysplastic/myeloproliferative neoplasms

- Chronic myelomonocytic leukaemia (CMML)
- Atypical chronic myeloid leukaemia, BCR-ABL1 negative (aCML)
- Juvenile myelomonocytic leukaemia (JMML)
- Myelodysplastic/myeloproliferative neoplasm with ring sideroblasts & thrombocytosis
- Myelodysplastic/myeloproliferative neoplasm, unclassifiable (MDS/MPN-U)

5. Myelodysplastic syndromes (MDS)

- MDS with single lineage dysplasia
- MDS with ring sideroblasts
- MDS with multilineage dysplasia
- MDS with excess blasts
- MDS with isolated del(5q)
- MDS, unclassifiable
- Childhood myelodysplastic syndrome

6. Myeloid neoplasms with germline predisposition (CEBPA, RUNX1, ETV6, GATA2)

7. Acute myeloid leukaemia (AML) & related precursor neoplasms

- AML with t(8;21)/RUNX1-RUNX1T1
- AML with inv(16)/CBFB-MYH11
- AML with PMLRARA/variant RARA
- AML with t(9;11)/KMT2A-MLLT3 as well as other KMT2A rearrangements
- AML with t(6;9)/DEK-NUP214
- AML with t(1;22)/RBM15-MKL1
- AML with t(9;22)/BCR-ABL1
- AML with mutated NPM1
- AML with biallelic mutation of CEBPA
- AML with mutated RUNX1
- AML with myelodysplasia related changes
- Therapy related myeloid neoplasms
- AML, not otherwise specified
- Myeloid sarcoma

8. Acute leukaemias of ambiguous lineage

- Acute undifferentiated leukaemia
- Mixed phenotype acute leukaemia with t(9;22)/BCR-ABL1
- Mixed phenotype acute leukaemia with t(v;11q23.3)/KMT2A rearranged
- Mixed phenotype acute leukaemia, B/myeloid
- Mixed phenotype acute leukaemia, T/myeloid

9. Precursor lymphoid neoplasms : B-lymphoblastic leukaemia/lymphoma (B-ALL/LBL)

- B-ALL/LBL, not otherwise specified
- B-ALL/LBL with t(9;22)/BCR-ABL1
- B-2ALL/LBL with t(v;11q3.3)/KMT2A rearranged
- B-ALL/LBL with t(12;21) / ETV6-RUNX1
- B-ALL/LBL with t(1;19)/TCF3-PBX1
- B-ALL/LBL with ZNF384 related fusion

10. Mature B-neoplasms

- Chronic lymphocytic leukaemia (CLL)
- Hairy cell leukaemia (HCL) (BM sample preferred)
- Lymphoplasmacytic lymphoma/Waldenstrom macroglobulinemia (LPL/WM) (BM sample preferred)

A stylized illustration of a DNA double helix structure. The helix is composed of two intertwined strands, one rendered in a vibrant blue and the other in a bright red. The strands are connected by horizontal rungs representing the base pairs. The entire structure is set against a dark, almost black background, which makes the glowing colors of the DNA stand out. The image has a slightly grainy, digital texture, giving it a high-tech or scientific feel.

- BPDCN

Fusions (RNA)

Acute myeloid leukaemia(AML) and related precursor neoplasms

RUNX1-RUNX1T1 t(8;21)	CBFB-MYH11 inv(16)	
PML-RARA t(15;17)	DEK-NUP214 t(6;9)	
RBM15-MKL1 t(1;22)	AML with BCR-ABL1	
KMT2A REARRANGEMENTS		
MLLT3 t(9;11)	TET1	CASP8AP2
AFF1/AF4	PICALM	CBL
MLLT1-ENL	ABI1	CREBBP
MLLT10/AF10	CASC5	DCP1A
ELL	MYO1F	DCPS
PTD	SEPT5	FNBP1
MLLT4/AF6	ACTN4	GAS7
EPS15	FLNA	KIA1524
MLLT11/AF1Q	FOXO3	BTBD18
SEPT9	CEP170B	MYH11
EPS15	MAML2	NEBL
SEPT6	SEPT11	NRIP3
MLLT6/AF17	ABI2	PDS5A
AFF3/LAF4	ACACA	SEPT2
ARHGEF12	AFF4/AF5	SMAP1
GMPS	ARHGEF17	TOP3A

Acute promyelocytic leukaemia

RARA REARRANGEMENTS: 3 types bcr1,bcr2,bcr3	
IRF2BP2-RARA	
NABP1-RARA t(2;17)	
TBL1XR1-RARA t(4;17)	
FIP1L1-RARA t(5;17)	
NPM1-RARA t(11;17)	
NUMA1-RARA	
ZBTB16-RARA t (11;17)	
ADAMTS17-RARA	
STAT5B-RARA	
PRKAR1A-RARA t(17;17)	
BCOR-RARA t(X;17)	
PML-RARA	

Chronic eosinophilic leukaemia

ETV6 REARRANGEMENTS
FLT3
NTRK
LYN
SYK
ABL1
ETV6-JAK2
BCR-JAK2

Pediatric -AML

RBM15-MKL1 t(1;22)
KAT6A-CREBBP
DEK-NUP214 t(6;9)
KMT2A Rearrangements
MLLT3
MLLT10
MLLT11

B-lymphoblastic leukaemia/lymphoma

t(9;22)/BCR-ABL1	t(1;19)/TCF3-PBX1
t(v;11q23.3)/KMT2A	ZNF384 related Fusions
t(12;21) /ETV6-RUNX1	Others

Chronic myeloid leukaemia

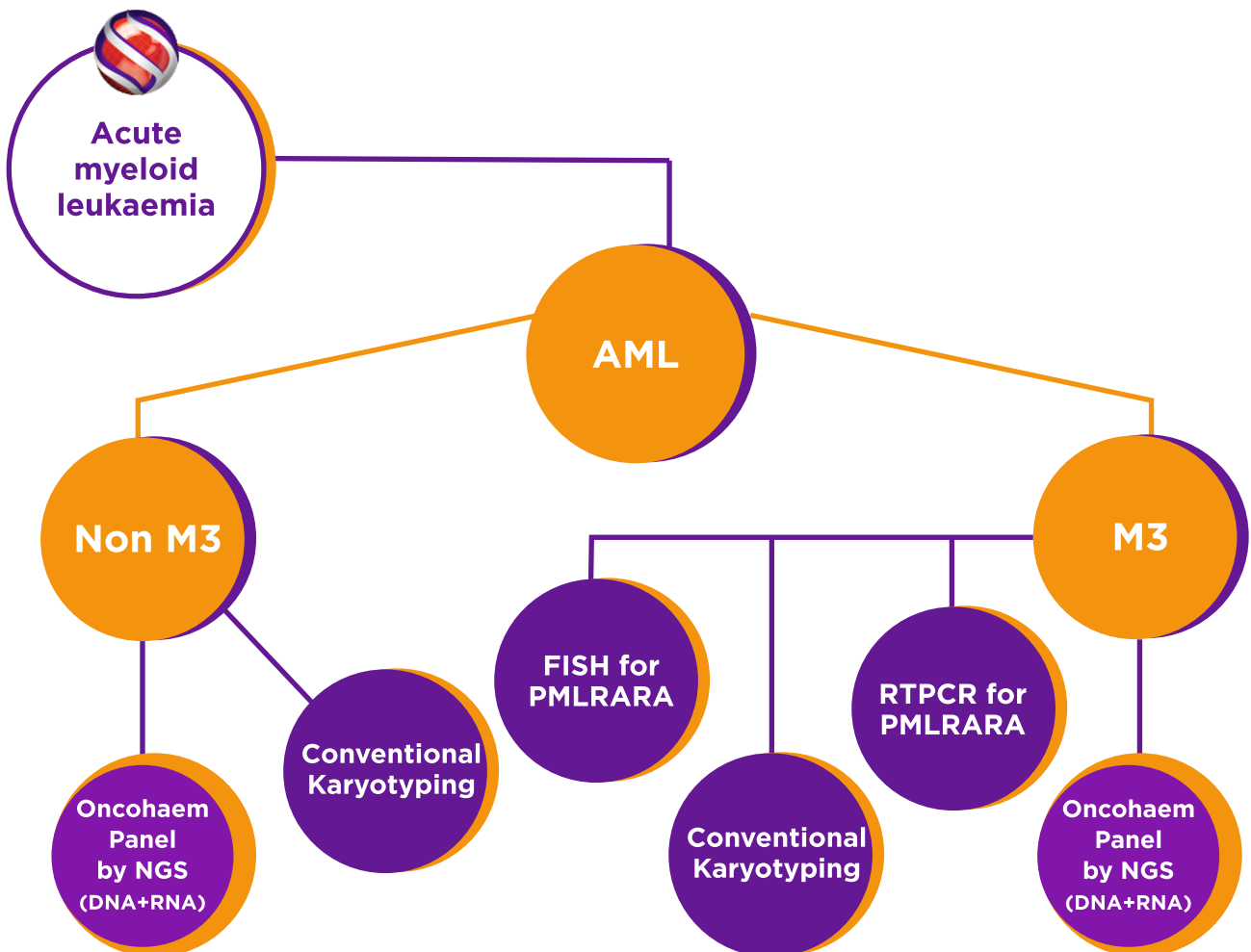
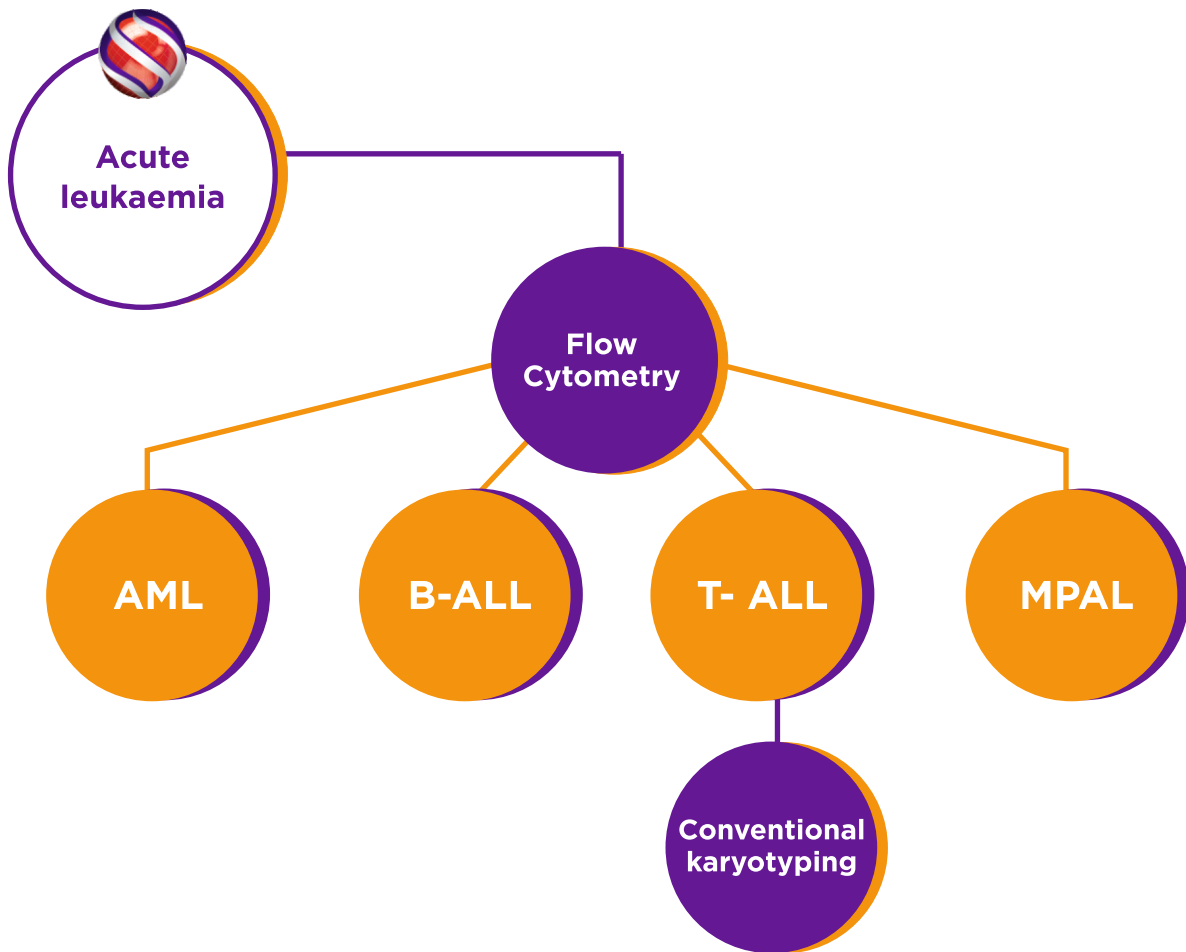
BCR-ABL1

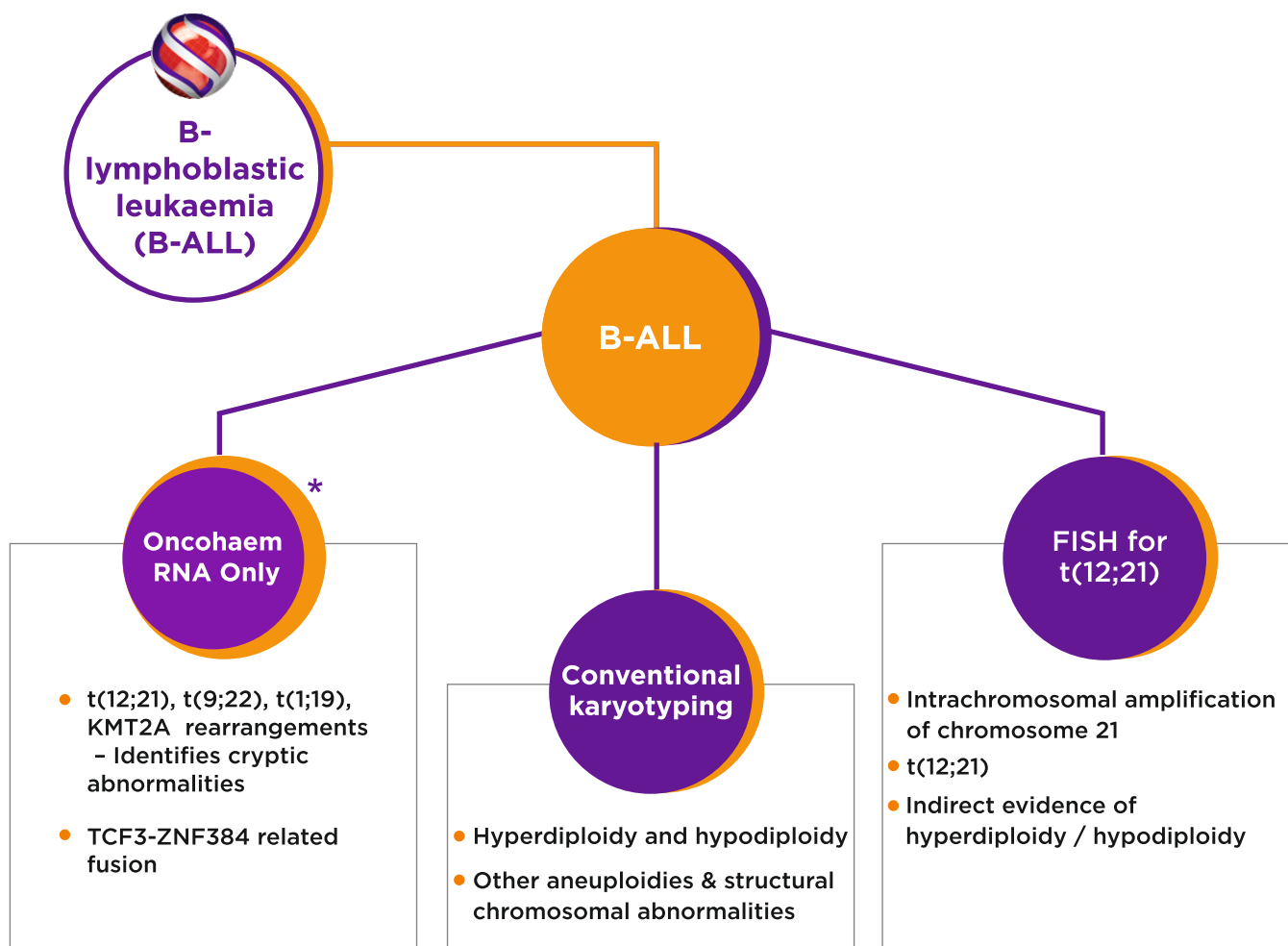
Myeloid/lymphoid neoplasms with eosinophilia & gene arrangement

PDGFRB REARRANGEMENTS	GOLGA4	FGFR1 REARRANGEMENTS	JAK2 REARRANGEMENTS
CCDC6	TNIP1	ZMYM2	PCM1
SART3	HIP1	CNTRL	ETV6
GIT2	KANK1	FGFR1OP	BCR
ERC1	MYO18A	BCR	PAXC
BIN2	COL1A1	MYO18A	PDGFRA REARRANGEMENTS
NIN	DTD1	TRIM24	ETV6
CCDC88C	GOLGB1	FGFR1OP2	FIP1L1
TP53BP1	CEP85L	TPR	KIF5B
NDE1	TRIP11	RANBP2	CDK5RAP2
RABEP1	MPRIIP	LRRFIP1	STRN
SPECC1	CPSF6	CUX1	FOXPI
ETV6	TPM3	CPSF6	TNKS2
WDR48	PDE4DIP	SQSTM1	BCR
CAPRIN1	PRKG2		

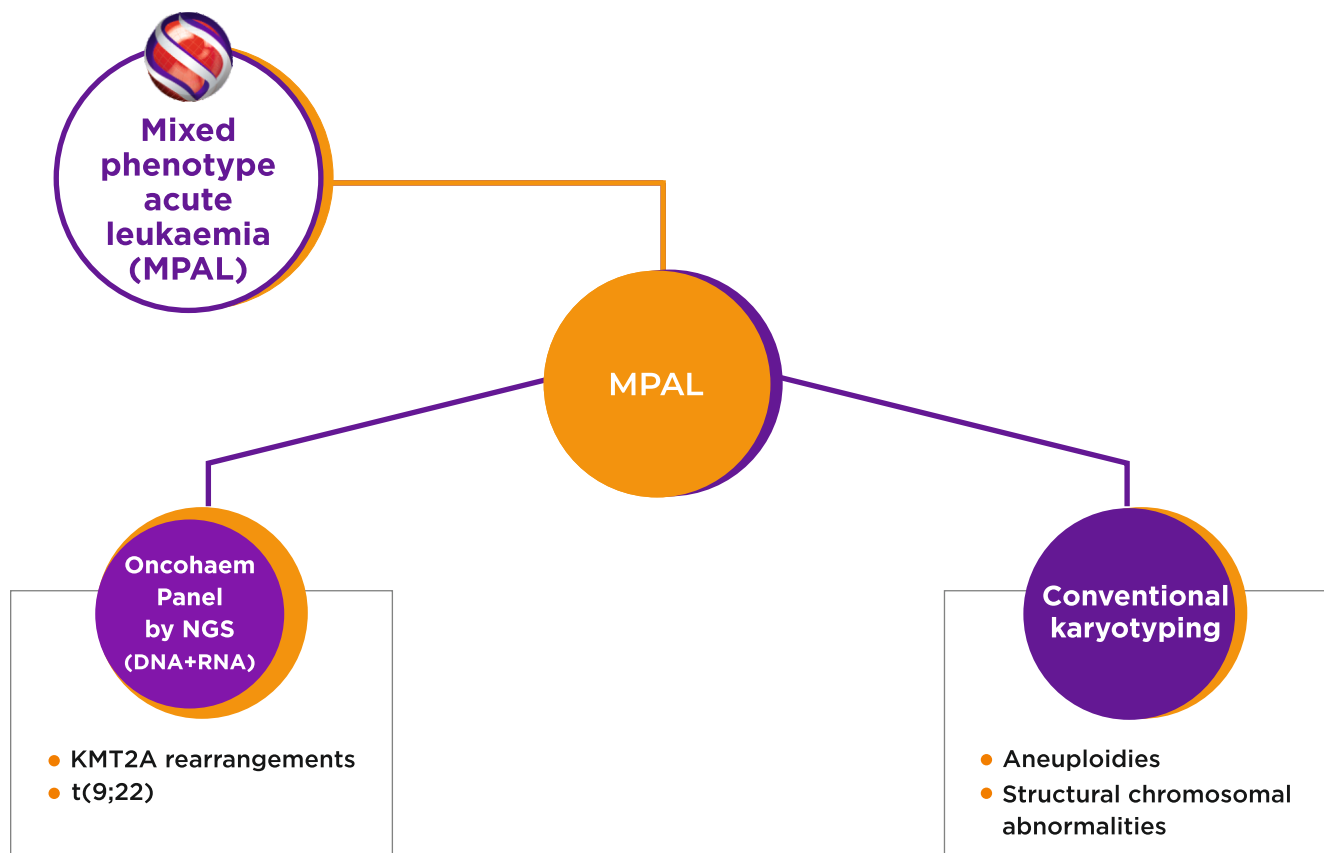
Bone Marrow Examination Is Must For All Suspected Myeloid Neoplasms

(except under special circumstances)

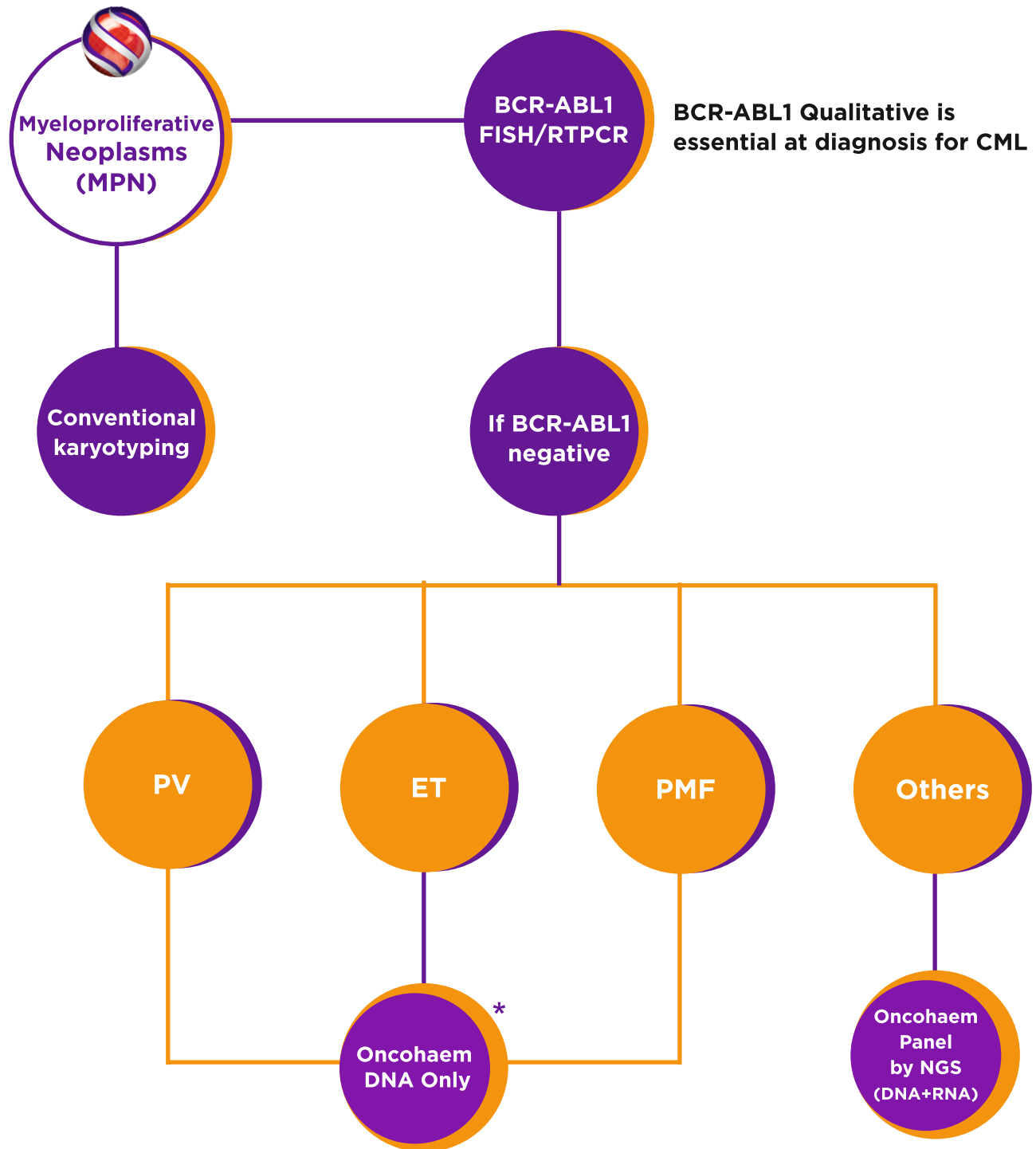




* Ideally Oncohaem panel by NGS (DNA + RNA)



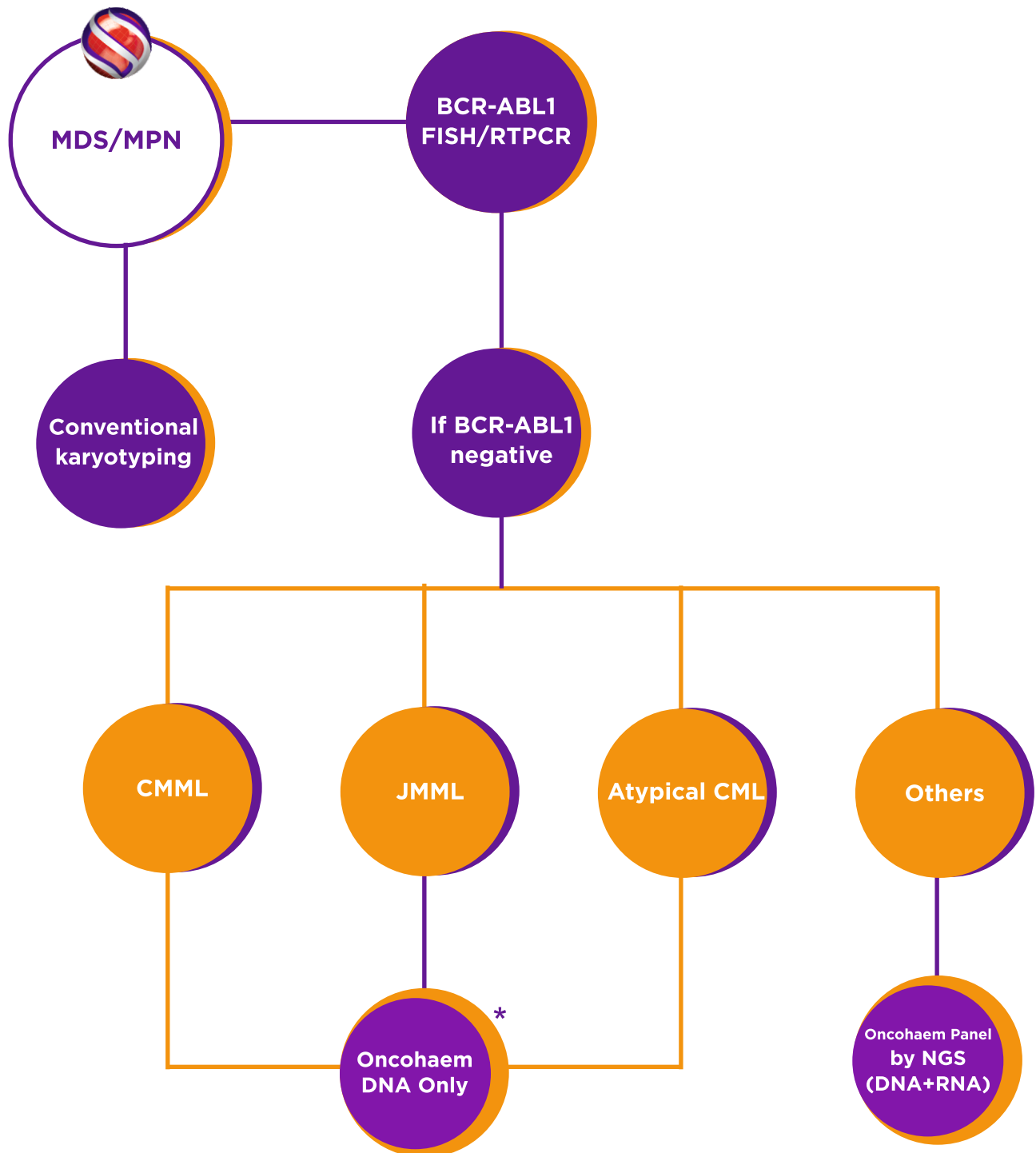
Myeloproliferative Neoplasms (MPN)



*Ideally Oncohaem panel by NGS (DNA + RNA)

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

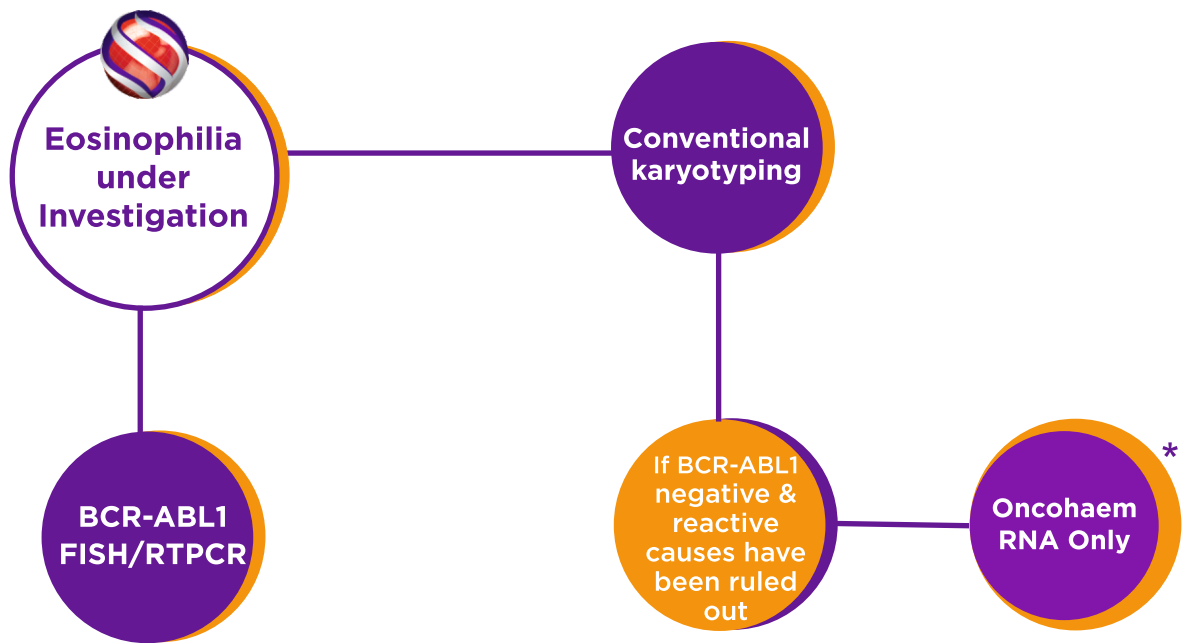
Myelodysplastic syndrome/ Myeloproliferative neoplasm(MDS/MPN)



*Ideally Oncohaem panel by NGS (DNA + RNA)

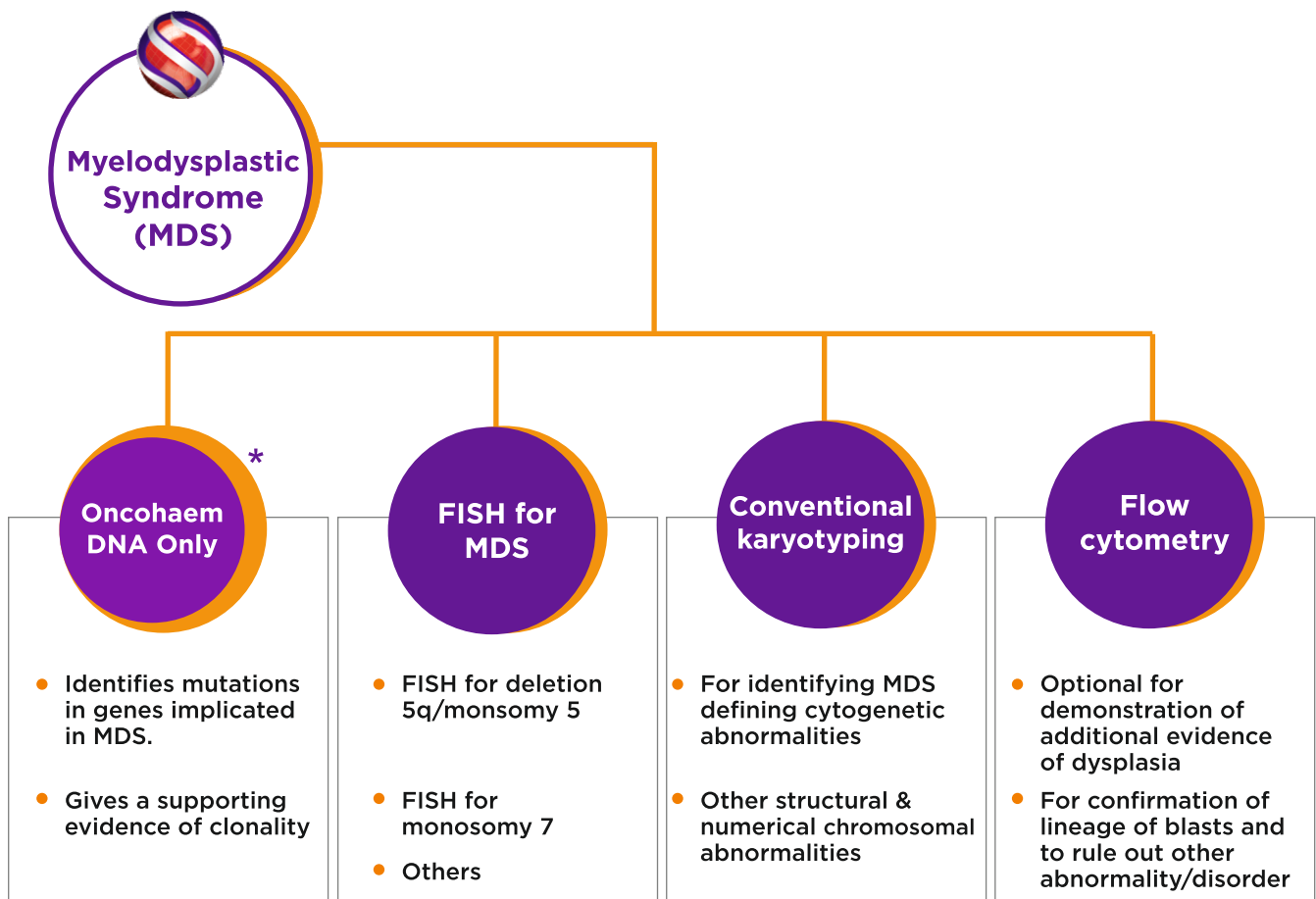
- Chronic myelomonocytic leukaemia(CMML)
- Atypical CML(aCML)
- Juvenile myelomonocytic leukaemia(JMML)
- MDS/MPN-RS-T
- MDS/MPN-U

Eosinophilia under Investigation



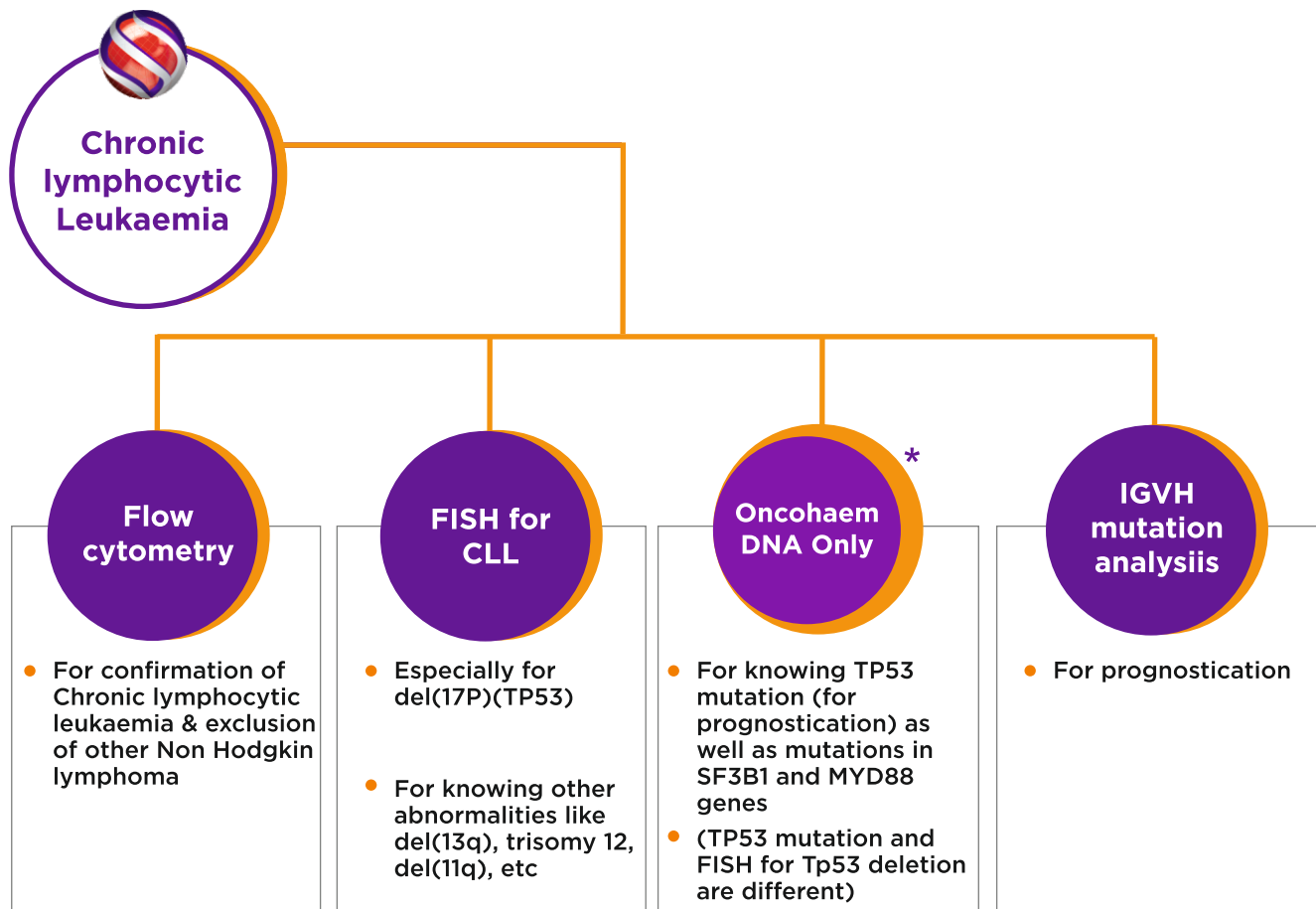
*Ideally Oncohaem panel by NGS (DNA + RNA)

Myelodysplastic Syndrome (MDS)

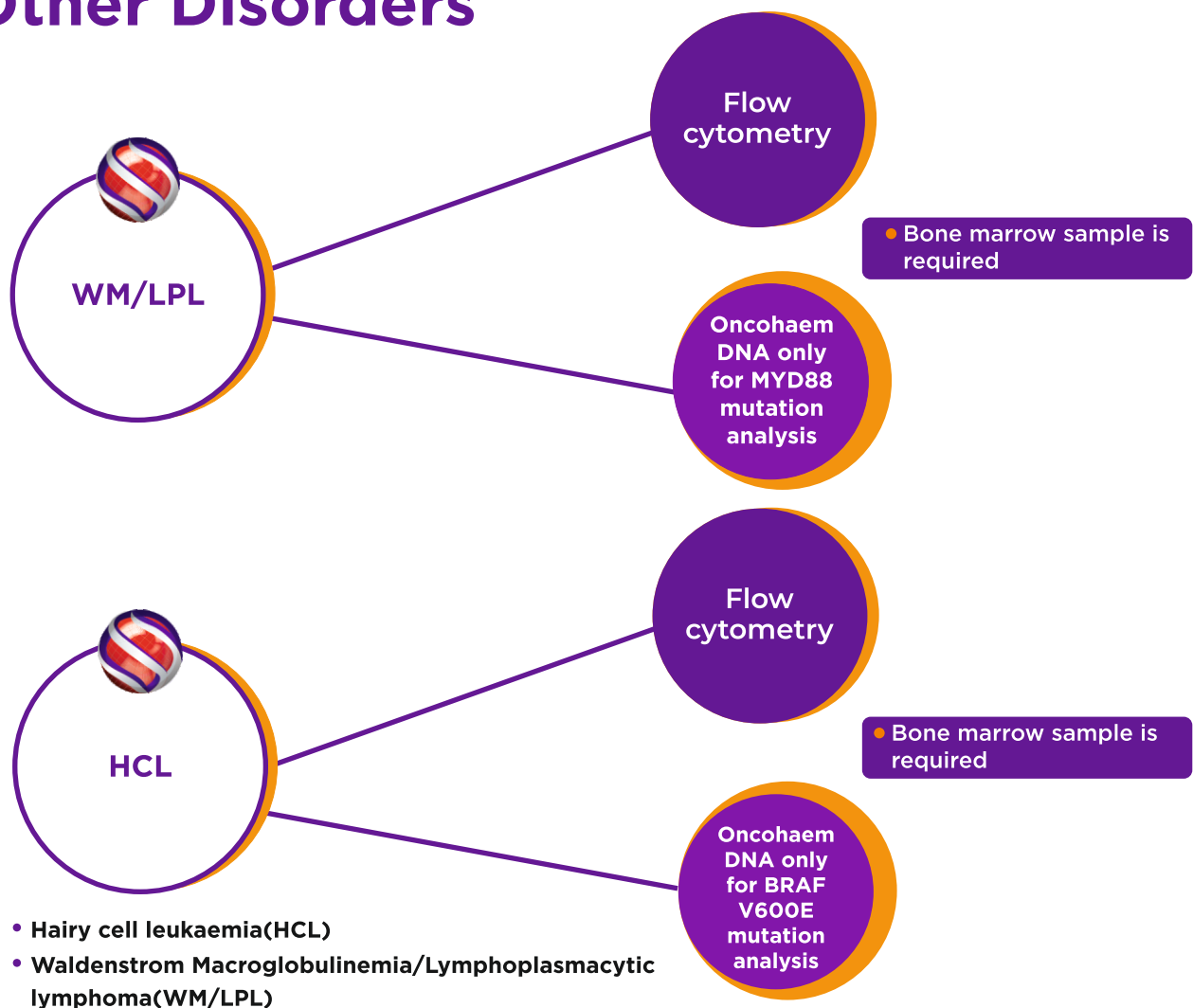


*Ideally Oncohaem panel by NGS (DNA + RNA)

Chronic lymphocytic Leukaemia (CLL)



Other Disorders



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 translocations 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.

Our Services



**Inherited Genetic
Disorder**



**Reproductive
Genetics**



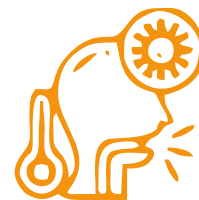
Cancer Genomics



Haemato Oncology



**Transplant
Immunology**



**Infectious
Disorders**



Pharmacogenomics



Research Services

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