

Myeloproliferative neoplasms (MPN)

- Chronic myeloid leukemia (CML)
- Chronic neutrophilic leukemia (CNL)
- Polycythemia vera (PV)
- Primary myelofibrosis (PMF)
- Essential thrombocythemia (ET)

Mastocytosis

Myeloid/lymphoid neoplasms with eosinophilia and rearrangement of PDGFRA, PDGFRB or FGFR1, or with PCM1-JAK2

Myelodysplastic / myeloproliferative neoplasms (MDS/MPN)

- Chronic myelomonocytic leukemia (CMML)
- Atypical chronic myeloid leukemia (aCML)
- Juvenile myelomonocytic leukemia (JMML)

Myelodysplastic syndromes (MDS)

- MDS with single lineage dysplasia (MDS-SLD)
- MDS with ring sideroblasts (MDS-RS)
- MDS with multilineage dysplasia (MDS-MLD)
- MDS with excess blasts (MDS-EB)
- MDS with isolated del(5q)

1 STAGE OF ANALYSIS	2	3	4	5
Cytology Cytochemistry	Immunophenotyping	Conventional cytogenetics: hematological karyotyping	Molecular cytogenetics: Fluorescent in situ Hybridization (FISH)	Molecular Biology including NGS panel
Peripheral blood and/or bone marrow aspirate smears analysis			BCR-ABL	Qualitative BCR-ABL Quantitative BCR-ABL (MRD (follow up)) BCR-ABL mutation (Resistance to imatinib) MPN - Diagnosis panel MPN - Prognosis panel
Bone marrow aspirate smears analysis				CKIT
Peripheral blood and/or bone marrow aspirate smears analysis		Bone marrow karyotyping in first line diagnosis and/or blood karyotyping if immature cells present (for MF: blood in first intention)	FIP1L1/PDGFR α PDGFR β (5q33) FGFR1 PCM1-JAK2	
Peripheral blood and/or bone marrow aspirate smears analysis + cytochemistry (Perl's stain)	Immunophenotyping (blasts evaluation and characterisation) <i>(See our "Acute leukemia" leaflet ref. DS3-INTGB)</i>		BCR-ABL Chromosomes 5, 7, 8 status EVI1 (MECOM) TP53 TEL (ETV6) 20q MLL (KMT2A)	Qualitative BCR-ABL CMML panel
			Chromosomes 5, 7, 8 status EVI1 (MECOM) TP53 TEL (ETV6) 20q MLL (KMT2A)	MDS panel

Sample requirements

Sample must reach laboratory within 48 hours.

	Preanalytical			Temperature	Required informations	Test request form
	Blood	Bone marrow	Other			
Cytology / Cytochemistry	2 slides unfixed and unstained	≥4 slides unfixed and unstained		Room temperature Do not freeze	- Clinical data - FBC	
Immunophenotyping	1 EDTA tube (or lithium heparin tube) 2 slides unfixed and unstained	2 ml in EDTA tube (or lithium heparin tube)		Room temperature Do not freeze	- Clinical data - FBC	
Conventional cytogenetics : Haematological aryotyping	5ml 1 lithium heparin tube	2-5ml 1 lithium heparin tube	Biological fluid	Room temperature Do not freeze		
Molecular cytogenetics : FISH	5ml 1 lithium heparin tube	2-5ml 1 lithium heparin tube	FISH also available on: - unstained slides (peripheral blood or bone marrow smear) - paraffin embedded lymph nodes No analysis from bone marrow biopsy	Room temperature Do not freeze		B8-INTGB Malignant blood disorders + BW12-INTGB Fish request form for Malignant Hemopathies and Solid Tumors
Molecular biology Qualitative BCR-ABL	Whole blood 1 EDTA tube and 1 PaxGen tube	Bone marrow 1 EDTA tube		EDTA tube : Room temperature PaxGen tube : Freeze -20°C	- Clinical data - FBC - Bone marrow report - ± Immuno-phenotyping report	
Molecular Biology Quantitative BCR-ABL	Whole blood 1 EDTA tube and 1 PaxGen tube			EDTA tube : Room temperature PaxGen tube : Freeze -20°C		
All others molecular biology tests	Whole blood 2 EDTA tubes	Bone marrow 1 EDTA tube		Room temperature Do not freeze		
Additional testing Plasma			Plasma IMATINIB (Glivec®) assay			

All the preanalytical information and the gene list by NGS panel can be found on www.eurofins-biomnis.ie > Test Guide

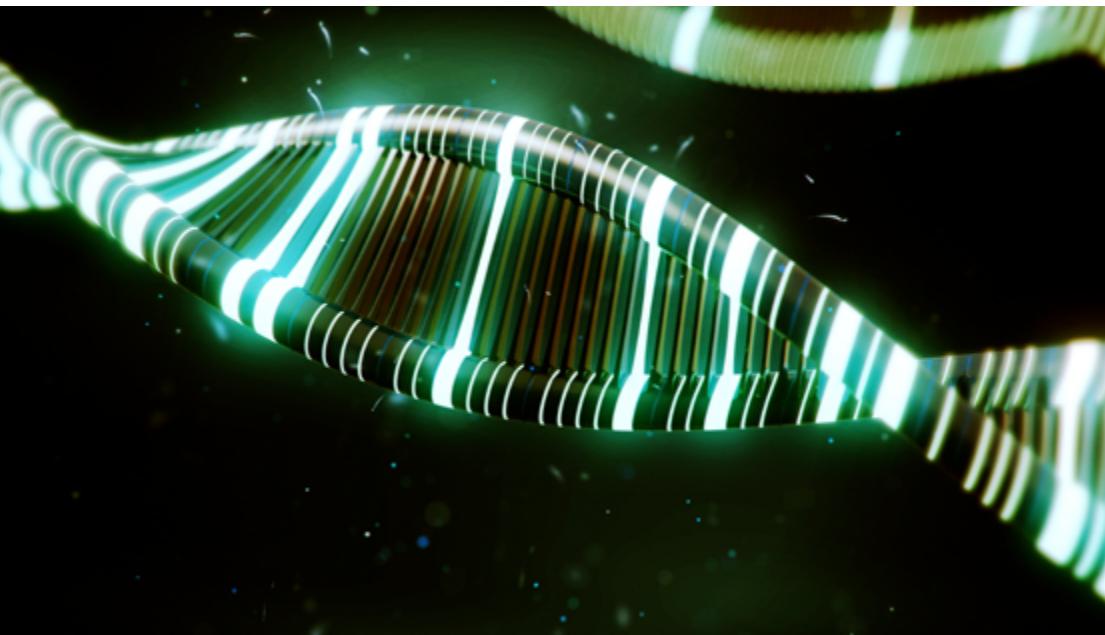
 ABL1, ASXL1, BRAF, CALR, CBL, CEBPA, CSF3R, DNMT3A, ETV6, EZH2, FLT3, HRAS, IDH1, IDH2, JAK2, KIT, KRAS, MPL, NPM1, NRAS, PTPN11, RUNX1, SETBP1, SF3B1, SRSF2, TET2, TP53, U2AF1, WT1, ZRSR2.



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References

*Swerdlow SH, Campo E, Harris NL, Jaffe ES, Pileri SA, Stein H, Thiele J. WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th edition) IARC Lyon 2017



**Pathological diagnosis
and follow up of patients with**

**Chronic Myeloid
Neoplasms**

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