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Myeloproliferative neoplasms (MPN) Prognosis

The **"MPN - Prognosis"** NGS panel includes the analysis of 23 genes: ASXL1/CALR/CBL/CSF3R/DNMT3A/EZH2/FLT3/IDH1/IDH2/JAK2/KIT/KRAS/MPL/NPM1/NRAS/RUNX1/SETBP1/SF3B1/SRSF2/TET2/TP53/U2AF1/ZRSR2.

It can be prescribed for **diagnostic** purposes (JAK2/CALR/MPL/CSF3R/SETBP1/SRSF2), however its main use is in **prognosis**:

- For Primary Myelofibrosis, this NGS panel can accurately assist with treatment decisions (e.g. 'high-risk' patient as defined in WHO 2017: CALR unmutated / ASXL1 mutated).
- For Polycythemia Vera or Essential Thrombocythemia, testing for poor prognostic molecular factors may help the clinician in patient follow-up (example of mutations associated with a poor prognosis: ASXL1, DNMT3A, EZH2, IDH1/2, SRSF2, TP53, U2AF1, …).

Note: Testing for a BCR-ABL fusion transcript, rearrangements of PDGFRA, PDGFRB, FGFR1, or PCM1-JAK2 fusion transcript cannot be performed using this NGS analysis (gDNA analysis). Additional techniques are offered by the Eurofins Biomnis laboratory for these five gene rearrangements.

Please note that the morphology (peripheral blood, bone marrow aspiration, bone marrow trephine biopsy), the immunophenotypic analysis and genetic data must be compared for the diagnosis and/or prognosis of a malignant haemopathy.

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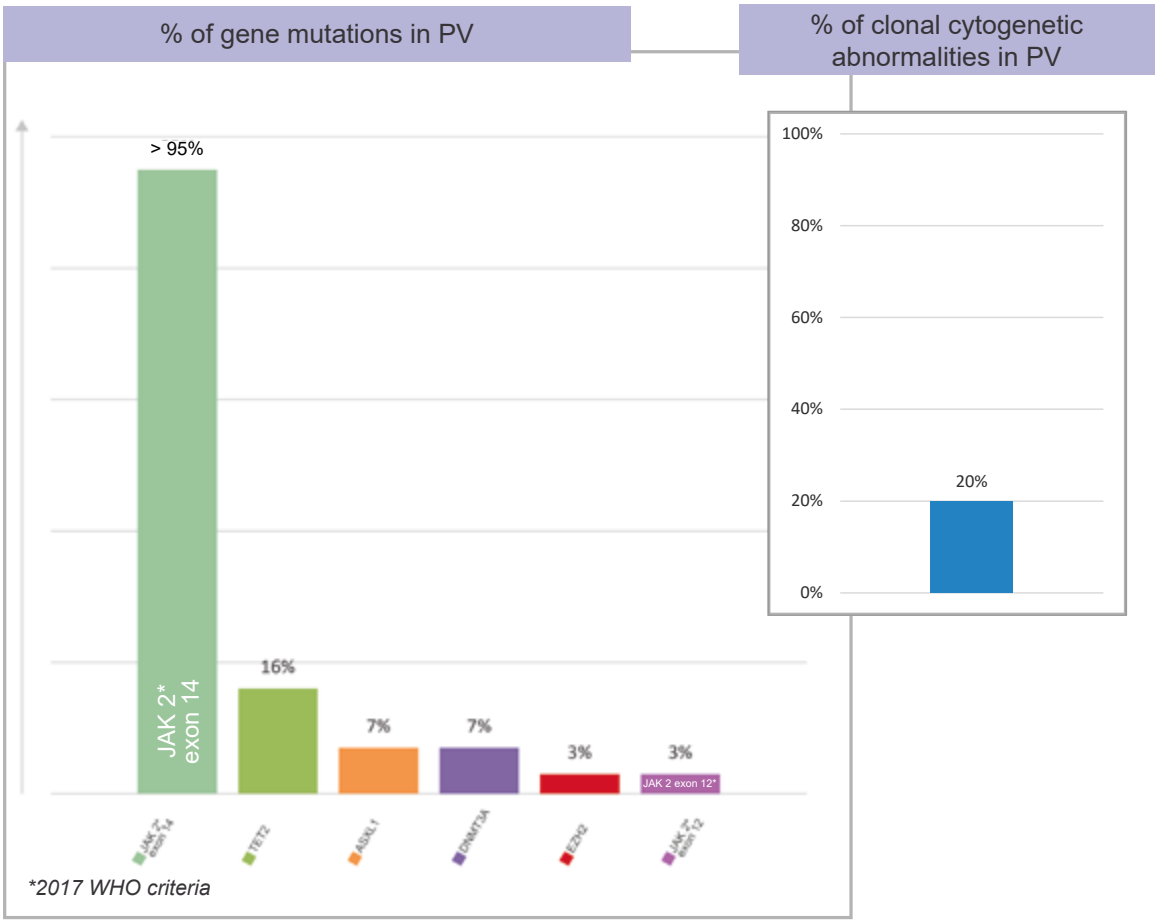
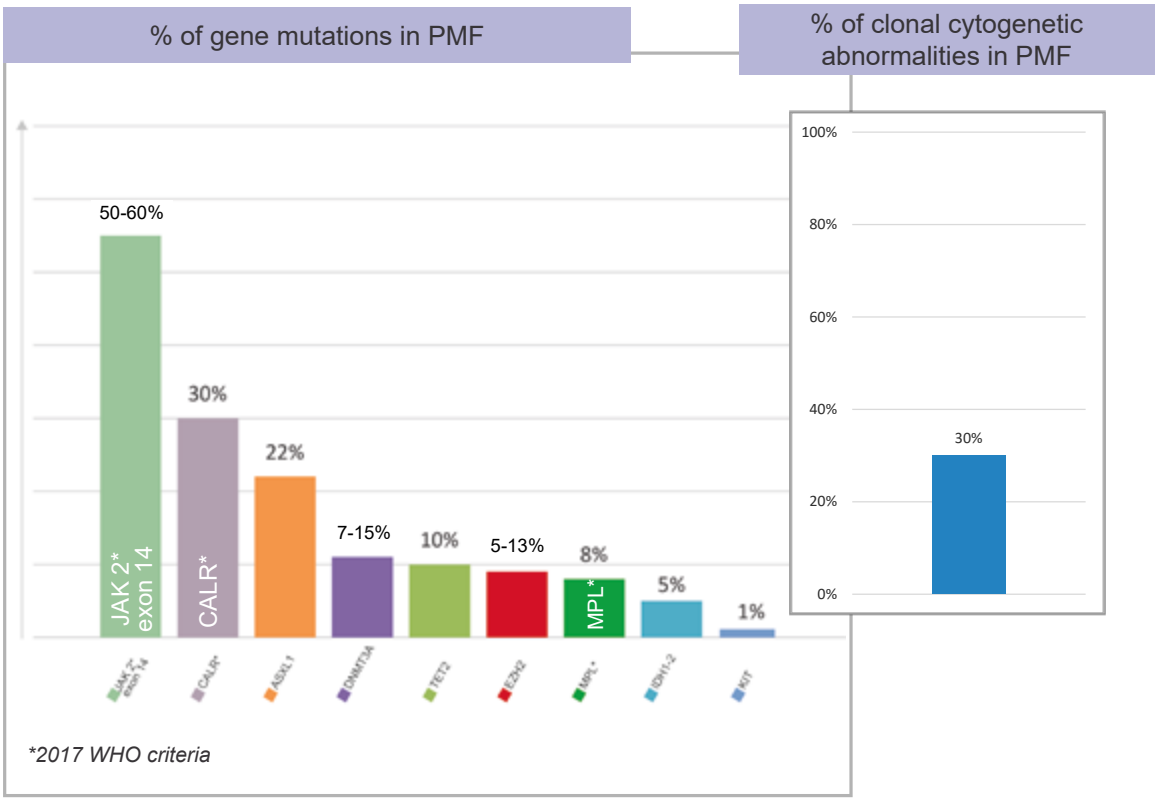
Gene mutations and clonal cytogenetic abnormalities in MPN

Polycythemia Vera (PV)



Primary Myelofibrosis (PMF)



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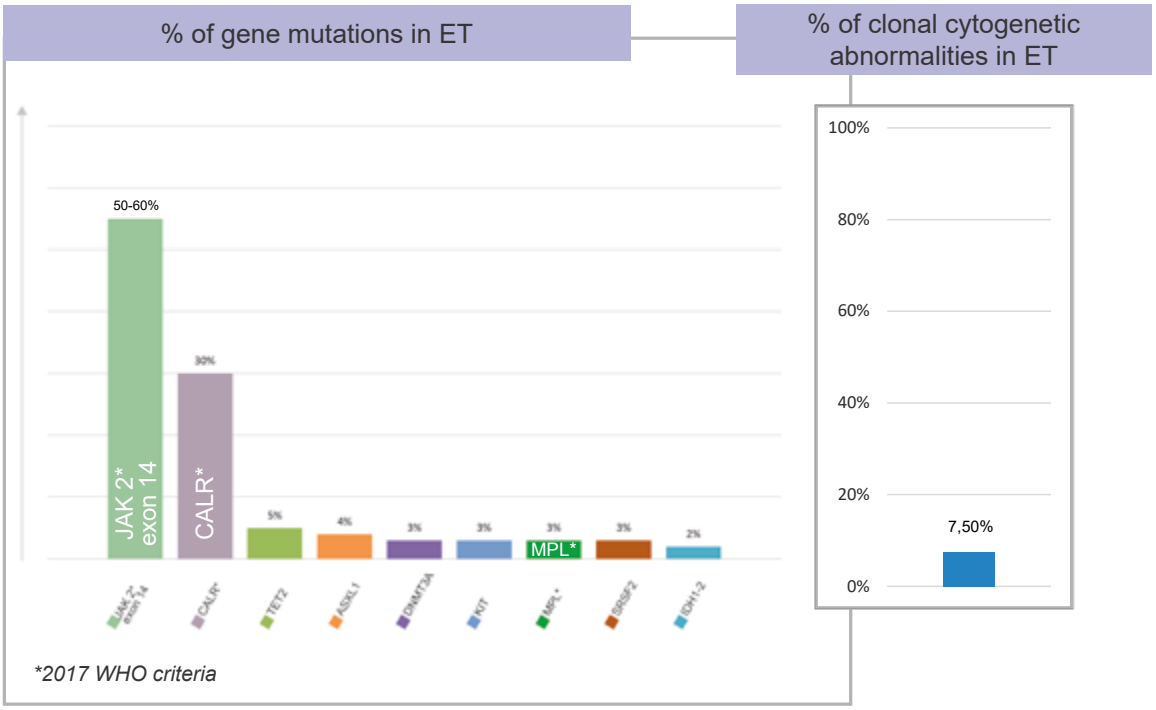
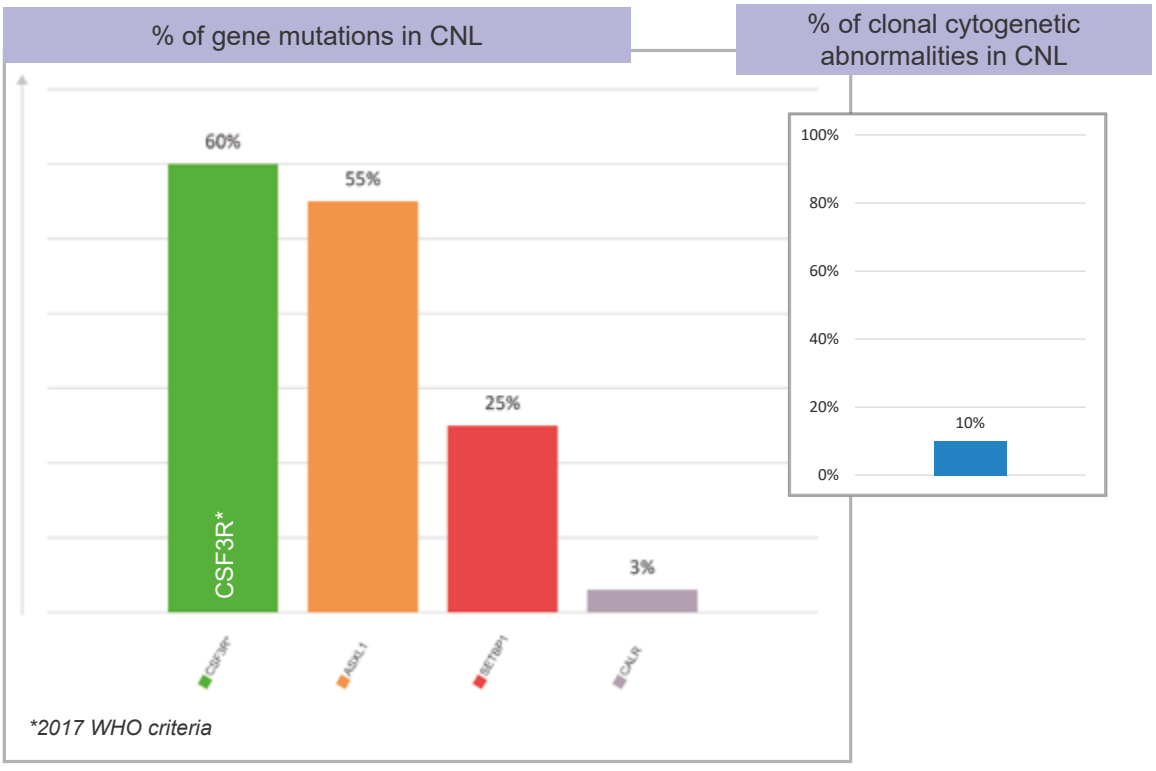
Essential Thrombocythemia (ET)



Chronic Neutrophilia Leukemia (CNL)



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“MPN - Prognosis” NGS Panel – Targeted genes

Gene	Transcript	Exon rank
CSF3R	NM_000760	Full coding region
DNMT3A	NM_022552	Full coding region
EZH2	NM_001203247	Full coding region
JAK2	NM_004972	Full coding region
RUNX1	NM_001754	Full coding region
TET2	NM_001127208	Full coding region
TP53	LRG_TP53 (LRG-specific mixed numbering)	Full coding region
ZRSR2	NM_005089	Full coding region
ASXL1	NM_015338	9, 11, 12, 14
CALR	NM_004343	9
CBL	NM_005188	8, 9
FLT3	NM_004119	13, 14, 15, 20

Gene	Transcript	Exon rank
IDH1	NM_005896	4
IDH2	NM_002168	4
KIT	NM_000222	2, 8, 9, 10, 11, 13, 17, 18
KRAS	NM_033360	2, 3
MPL	NM_005373	10
NPM1	NM_002520	10, 11
NRAS	NM_002524	2, 3
SETBP1	NM_015559	4
SF3B1	NM_012433	10, 11, 12, 13, 14, 15, 16
SRSF2	NM_003016	1
U2AF1	NM_006758	2, 6

Test code: MYSPR

Pre-analytical: 2 x 5 ml EDTA whole blood or 2 ml EDTA bone marrow

Turnaround time: 10 days (Results may require an extended turnaround time of one week, depending on the confirmation tests required by Sanger sequencing)

Contact

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Reference

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

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