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Myeloproliferative neoplasms (MPN) Diagnosis

The **"MPN- Diagnosis 1"** NGS panel includes an analysis of JAK2, CALR and MPL.

According to the WHO 2017, mutations of these three genes are included in the **diagnostic** criteria for the following myeloproliferative neoplasms (MPN):

- Polycythemia Vera (JAK2 exon 14 and 12 mutation – major criteria),
- Primary Myelofibrosis (JAK2, CALR, CPL mutation – major criteria),
- And Essential Thrombocythemia (JAK2, CALR, MPL mutation – major criteria).

The **"MPN- Diagnosis 2"** NGS panel includes an analysis of JAK2, CALR, MPL, CSF3R, SETBP1 and SRSF2.

According to the WHO 2017, mutations of JAK2, CALR, MPL, CSF3R are included in the **diagnostic** criteria for the following myeloproliferative neoplasms (MPN):

- Polycythemia Vera (JAK2 exon 14 and 12 mutation – major criteria),
- Primary Myelofibrosis (JAK2, CALR, CPL mutation – major criteria),
- Essential Thrombocythemia (JAK2, CALR, MPL mutation – major criteria),
- And Chronic Neutrophilia Leukemia (CSF3R mutation).

The presence of a SETBP1 and/or SRSF2 mutation assist in **diagnosis** for the WHO entity 'Atypical CML – BCR-ABL negative'.

NGS analysis enables the detection of any mutation in the three genes ("MPN- Diagnosis 1" panel) or for the six genes ("MPN- Diagnosis 2" panel) above mentioned.

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.

NB: a "MPN-prognosis" NGS panel is also available for an exhaustive analysis of MPN.

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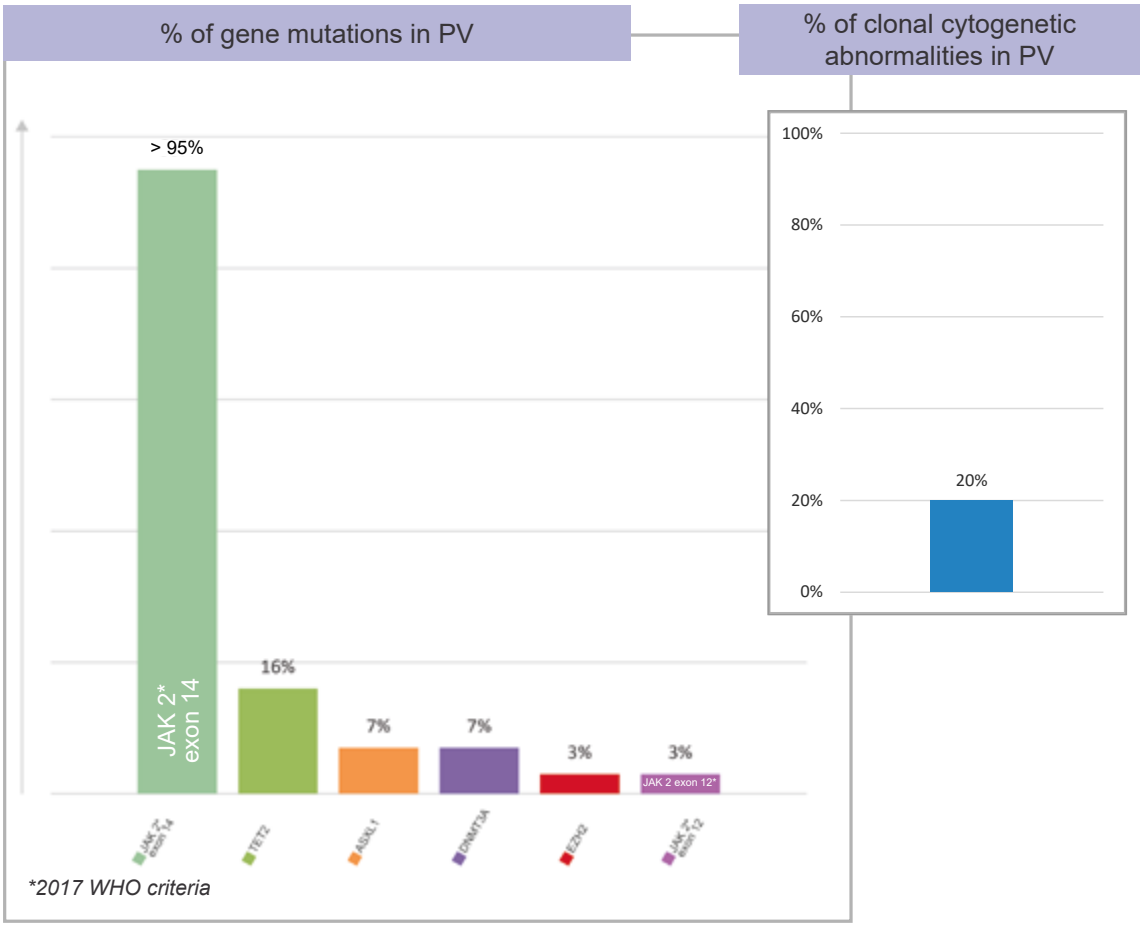
Gene mutations and clonal cytogenetic abnormalities in MPN

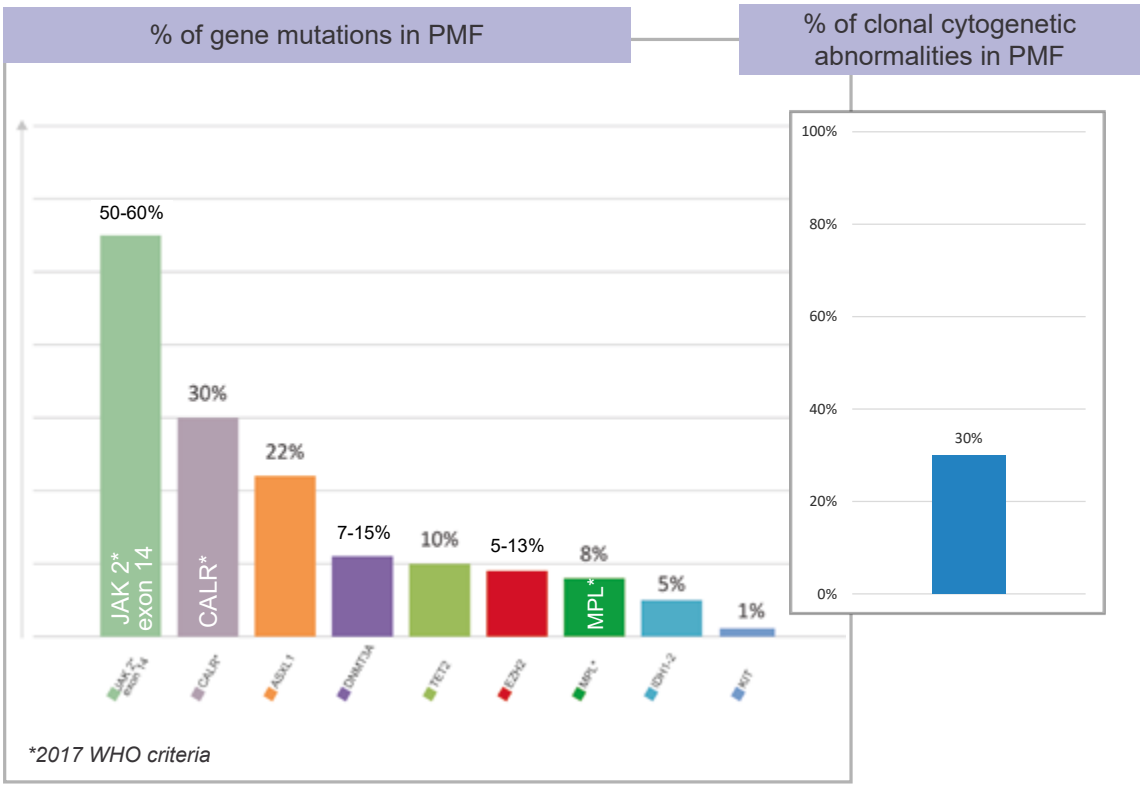
Polycythemia Vera (PV)

Panel 1 and 2



Primary Myelofibrosis (PMF)

Panel 1 and 2



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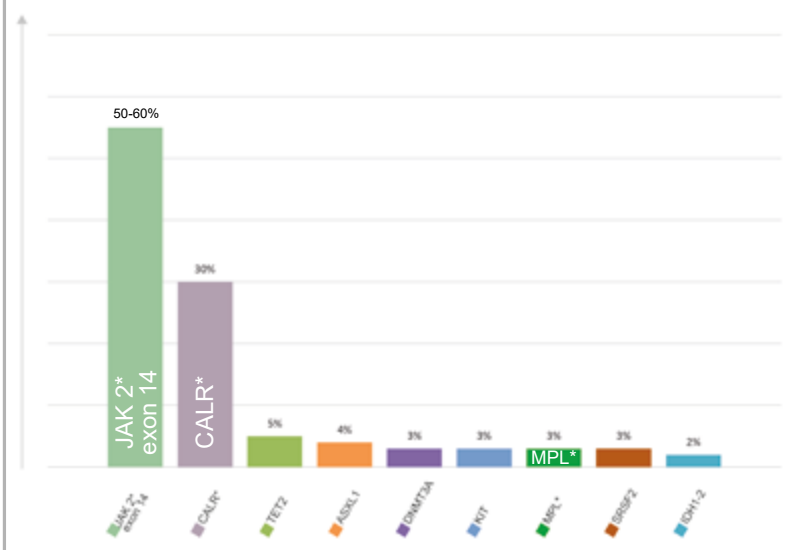
Gene mutations and clonal cytogenetic abnormalities in MPN

Essential Thrombocythemia (ET)

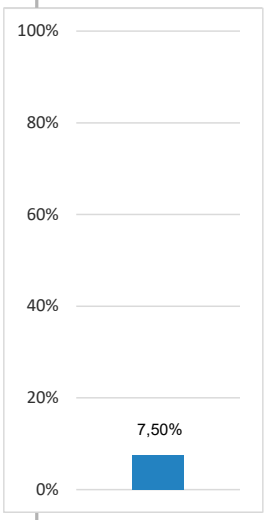
Panel 1 and 2

% of gene mutations in ET



*2017 WHO criteria

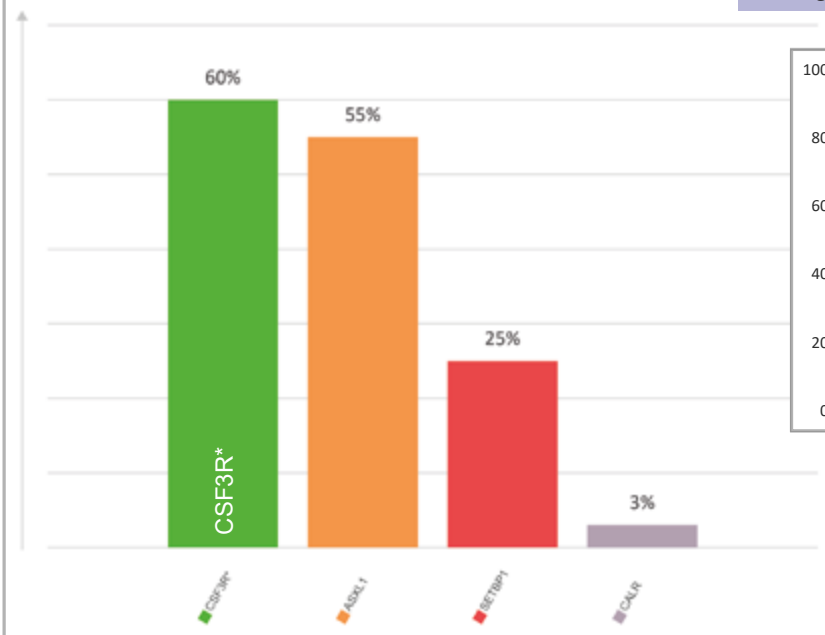
% of clonal cytogenetic abnormalities in ET



Chronic Neutrophilia Leukemia (CNL)

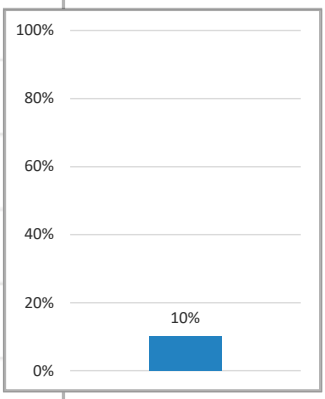
Panel 2

% of gene mutations in CNL



*2017 WHO criteria

% of clonal cytogenetic abnormalities in CNL



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Targeted genes

"MPN - Diagnosis 1" Panel

Gene	Transcript	Exon rank
JAK2	NM_004972	Full coding region
CALR	NM_004343	9
MPL	NM_005373	10

“MPN - Diagnosis 2” Panel

Gene	Transcript	Exon rank
CSF3R	NM_000760	Full coding region
JAK2	NM_004972	Full coding region
CALR	NM_004343	9
MPL	NM_005373	10
SETBP1	NM_015559	4
SRSF2	NM_003016	1

Test code: MYSD1

Test code: MYSD2

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

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

Contact

International division – Eurofins Biomnis
international@eurofins-biomnis.com
Tel.: +33 4 72 80 23 85

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

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