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# Chronic Myelomonocytic Leukemia (CMML)

The "**CMML**" NGS panel includes the analysis of 19 genes: ASXL1/CBL/DNMT3A/EZH2/FLT3/IDH1/IDH2/JAK2/KRAS/NPM1/NRAS/RUNX1/SETBP1/SF3B1/SRSF2/TET2/TP53/U2AF1/ZRSR2

It can be used in the three areas of **diagnosis, prognosis and theranostics.**

- According to the WHO 2017, the analysis of the TET2, SRSF2, ASXL1 and SETBP1 genes can help in the **diagnosis** of CMML (criterion 5 of the WHO 2017 in combination with criteria 1 to 4), in particular in the absence of contributing cytogenetic data. The combination of mutations TET2/SRSF2 is highly suggestive of CMML.

- The panel can also be used for helping to make **prognosis** and **therapeutic** decisions. According to the new ELN 2018 recommendations, a panel of at least four genes is required (ASXL1, NRAS, RUNX1 and SETBP1) to decide on an allogenic graft and a panel of 20 genes is recommended for any therapeutic decision. This panel also addresses the theranostic impact (IDH1, IDH2, NPM1 and FLT3 therapeutic targets).

**NB:** The genes BCOR and NF1 are not included in the panel offered and the gene TP53 was added as per the ELN 2018 recommendations.

**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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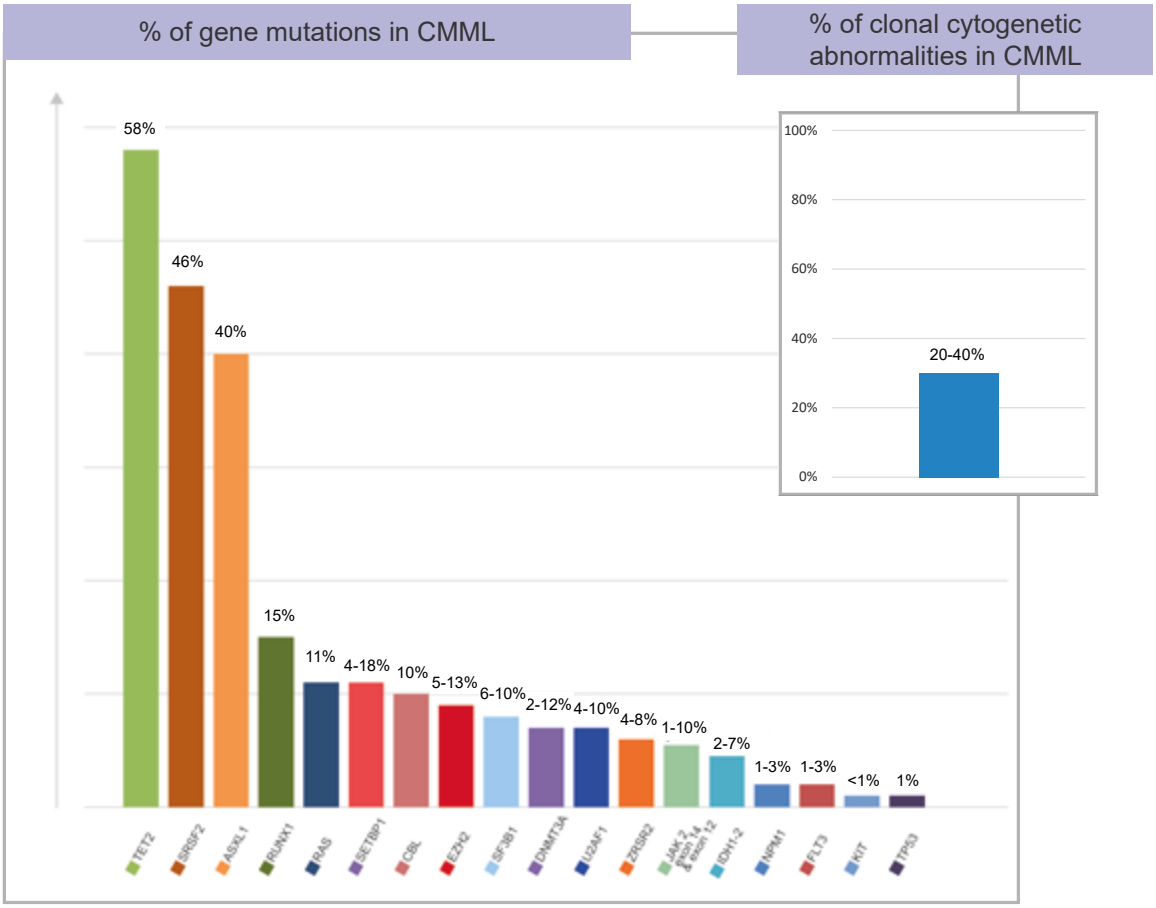
## Gene mutations and clonal cytogenetic abnormalities in CMML



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## “CMML” NGS panel – Targeted genes

| Gene | Transcript | Exon rank |
|---|---|---|
| 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 |
| CBL | NM_005188 | 8, 9 |
| FLT3 | NM_004119 | 13, 14, 15, 20 |

| Gene | Transcript | Exon rank |
|---|---|---|
| IDH1 | NM_005896 | 4 |
| IDH2 | NM_002168 | 4 |
| KRAS | NM_033360 | 2, 3 |
| 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: MYSMO

## 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 of one week, depending on the confirmation tests required by Sanger sequencing)

## Contact

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## References

1. 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

2. Itzykson et al, Diagnosis and Treatment of Chronic Myelomonocytic Leukemias in Adults, Recommendations From the European Hematology Association and the European LeukemiaNet, Hemasphere 2018

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