# 2 MYELOID GENEN PANEL

Releasedate 05-08-2021

MDS, AML, MPN, CMML, HES, aCML, JMML, MDS/MPN, MASTOCYTOSE

| *Gen* | *Chromosoom regio* | *Type afwijking* | *Startpositie* | *Eindpositie* | *Bron** | *Ziekte* | *Evidence** |
|---|---|---|---|---|---|---|---|
| CSF3R | 1p34.3 | mutatie | 36931643 | 36948915 | 1 | aCML,CNL | 1 |
| MPL | 1p34.2 | CNLOH | 43803474 | 43820135 | 1,2,3,4 | MPN,MDS RARSt,MDS/MPN | 1 |
| RBM15 | 1p13.3 | t(1;22) | 110881944 | 110889303 | 1 | AML | 1 |
| NRAS | 1p13.2 | CNLOH | 115247077 | 115259515 | 1,2 | JMML,CMML,MDS | 1 |
| DNMT3A | 2p23.3 | loss,CNLOH | 25455844 | 25565459 | 1,3,5 | MDS | 1 |
| SF3B1 | 2q33.1 | loss, CNLOH | 198256700 | 198299771 | 1 | MDS/MPN,MDS | 1 |
| IDH1 | 2q34 | loss, CNLOH | 209100952 | 209119806 | 1 | MDS,AML,MPN | 1 |
| RASSF1 | 3p21.31 | loss | 50367216 | 50378367 | 5 | MDS | 2 |
| GATA2 | 3q21.3 | Inv(3)/t(3;3) | 128198264 | 128212030 | 1 | AML | 1 |
| MECOM FUSIE | 3q26.2 | CNLOH,<br>Inv(3)/t(3;3) | 168801286 | 169381563 | 1,3 | MDS,AML | 3 (CNLOH)<br>1 (t/inv) |
| FIP1L1 | 4q12 | FIP1L1-PDGFRA<br>loss | 54243819 | 54326103 | 1,4,5 | HES<br>MDS | 1<br>2 |
| CHIC2 | 4q12 | loss | 54875957 | 54930788 | 1 | HES | 1 |
| PDGFRA | 4q12 | FIP1L1-PDGFRA<br>loss | 55095263 | 55164412 | 1,4,5 | HES<br>MDS | 1<br>2 |
| KIT | 4q12 | mutatie,translocatie | 55524094 | 55606881 | 1 | Mastocytose,AML | 1 |
| TET2 | 4q24 | CNLOH, loss | 106067942 | 106200958 | 1,2,3,5 | CMML,sAML,MPN,MDS/MPN,MDS,AML | 1 |
| PDGFRB | 5q32 | translocatie | 149493401 | 149535422 | 1,4 | HES | 1 |
| RPS14 | 5q33.1 | loss | 149823791 | 149829319 | 3 | CMML,MPN,MDS/MPN,MDS,AML | 1 |
| NPM1 | 5q35.1 | loss,translocatie | 170814707 | 170837888 | 1,4,5 | MDS,AML | 1 |
| DEK | 6p22.3 | t(6;9) | 18224399 | 18264799 | 1 | AML | 1 |
| CUX1 | 7q22.1 | loss,CNLOH | 101459183 | 101927250 | 3,6 | CMML,MPN,MDS/MPN,MDS,AML | 1 |

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|---|---|---|---|---|---|---|---|
| EZH2 | 7q36.1 | loss,CNLOH | 148504463 | 148580601 | 1,3,4,5,6 | CMML,MPN,MDS/MPN,MDS,AML | 1 |
| PCM1 | 8p22 | t(8;9) | 17780365 | 17887457 | 1,4 | HES | 1 |
| FGFR1 | 8p11.23p11.22 | translocatie | 38268655 | 38326352 | 1,4 | HES | 1 |
| RUNX1T1 | 8q21.3 | trans,CNLOH | 92967194 | 93115454 | 1,4 | AML | 1 |
| JAK2 | 9p24.1 | CNLOH,gain,t(8;9) | 4985244 | 5128183 | 1,2,3,4 | MPN,HES,PV,ET, IMF,MDS RARSt,MDS/MPN | 1 |
| MLLT3 | 9p21.3 | t(9;11) | 20341662 | 20622514 | 1 | AML | 1 |
| ABL1 | 9q34.12 | gain,t(9;22) | 133589267 | 133763062 | 3 | CML,AML | 1 |
| NUP214 | 9q34.13 | t(6;9) | 134000980 | 134110057 | 1 | AML | 1 |
| HRAS | 11p15.5 | loss | 532241 | 535550 | 5 | MDS | 2 |
| WT1 | 11p13 | CNLOH | 32409321 | 32457081 | 1,2 | AML | 2 |
| CCND1 | 11q13 | loss | 69455872 | 69469242 | 5 | MDS | 2 |
| KMT2A FUSIE | 11q23.3 | PTD-KMT2A*, translocation | 118307204 | 118397539 | 1 | AML | 1 |
| CBL | 11q23.3 | loss,CNLOH | 119076989 | 119178859 | 1,2,3 | JMML,CMML,MPN,MDS/MPN,MDS | 1 |
| ETV6 | 12p13.2 | loss,CNLOH, translocation | 11802787 | 12048325 | 1,3,5 | HES,CMML,MPN,MDS/MPN,MDS,AML | 1 |
| ETNK1 | 12p12.1 | mutatie | 22778075 | 22843608 | 1 | aCML | 1 |
| KRAS | 12p12.1 | mutatie | 25357722 | 25403854 | 1,5 | JMML,MDS | 1 |
| PTPN11 | 12q24.13 | mutatie | 112856535 | 112947717 | 1 | JMML | 1 |
| NCOR2 | 12q24.31 | loss | 124808956 | 125052010 | 5 | MDS | 2 |
| FLT3 | 13q12.2 | CNLOH | 28577410 | 28674729 | 2,4 | AML,MDS | 2 |
| RB1 | 13q14.2 | loss | 48877882 | 49056026 | 3 | CMML,MPN,MDS/MPN,MDS | 1 |
| PML | 15q24.1 | t(15;17) | 74287013 | 74340155 | 1,4 | AML | 1 |

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|---|---|---|---|---|---|---|---|
| IDH2 | 15q26.1 | loss | 90627211 | 90645708 | 1 | MDS,AML,MPN | 1 |
| MYH11 | 16p13.11 | inv/t(16;16) | 15796991 | 15950887 | 1 | AML | 1 |
| CBFB | 16q22.1 | inv/t(16;16),loss | 67063049 | 67134958 | 1 | AML | 1 |
| PRPF8 | 17p13.3 | loss | 1553922 | 1588176 | 5 | MDS | 2 |
| TP53 | 17p13.1 | loss,CNLOH | 7571719 | 7590863 | 1,2,3,5,6 | CML,CMML,MPN,MDS/MPN MDS,AML | 1 |
| NF1 | 17q11.2 | loss,CNLOH | 29421944 | 29704695 | 1,2,3,5 | JMML,MPN,MDS | 1 |
| RARA | 17q21.2 | t(15;17) | 38465422 | 38513895 | 1,4 | AML | 1 |
| SRSF2 | 17q25.1 | loss,CNLOH | 74730197 | 74733493 | 1,3 | CMML,MDS | 1 |
| SETBP1 | 18q12.3 | mutatie | 42260137 | 42648475 | 1 | CMML/atypCML | 1 |
| CALR | 19p13.2 | mutatie | 13049413 | 13055304 | 1,4 | MPN | 1 |
| CEBPA | 19q13.11 | CNLOH | 33790839 | 33793470 | 1,2,4 | AML | 1 |
| ASXL1 | 20q11.21 | loss,CNLOH | 30946146 | 31027122 | 1,3,4,5 | CMML,MPN,MDS/MPN,MDS,AML | 1 |
| RUNX1 | 21q22.12 | loss,CNLOH,translocatie | 36160097 | 36421595 | 1,3,4,5 | CMML,MPN,MDS/MPNAML,MDS | 1 |
| ERG | 21q22.2 | amplificatie | 39739182 | 40033704 | 6 | AML | 3 |
| U2AF1 | 21q22.3 | loss,CNLOH | 44513073 | 44527688 | 1,3 | MDS,MPN | 1 |
| BCR | 22q11.23 | gain,translocatie | 23522551 | 23660224 | 3 | CML,AML | 1 |
| MKL1 | 22q13.1 | t(1;22) | 40806291 | 41032690 | 1 | AML | 1 |
| ZRSR2 | Xp22.2 | loss | 15808574 | 15841382 | 1 | MDS | 1 |
| BCOR | Xp11.4 | loss | 39910498 | 40036582 | 1 | MDS | 1 |
| STAG2 | Xq25 | loss | 123094409 | 123236505 | 1 | MDS | 1 |

Bron:

1= WHO Classification of tumours of haematopoietic and lymphoid tissues, Swerdlow et al., revised 4th edition 2017

2= artikel **O'Keefe** et al., Copy neutral loss of heterozygosity: a novel chromosomal lesion in myeloid malignancies, **Blood 2010**

3= artikel **Kanagal-Shamanna** et al., Assessing copy number aberrations and copy neutral loss of heterozygosity across the genome as best practice: An evidence based review of clinical utility from the cancer genomics consortium (CGC) working group for myelodysplastic syndrome, myelodysplastic/myeloproliferative and myeloproliferative neoplasms, **cancer genetics 2018**

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4= artikel **Rack** et al., European recommandations and quality assurance for cytogenomic analysis of haematological neoplasm, **Leukemia 2019**
5= artikel **Schoumans** et al., Guidelines for genomic array analysis in acquired haematological neoplastic disorders, **Genes, Chromosomes&Cancer 2016**
6= artikel **Xu** et al., Assessing copy number abnormalities and copy-neutral loss-of-heterozygosity across the genome as best practice in diagnostic evaluation of acute myeloid leukemia: An evidence-based review from the cancer genomics consortium (CGC) myeloid neoplasms working group, **Cancer Genetics 2018**
*PTD-KMT2A are duplications which are variable in size and most commonly involve exons 2 or 3, spanning through exon 6 or exons 8–11
Evidence: 1)present in WHO classification 2)recurrent (>5 cases for CNV en >2 for CNLOH in well-powered studies with expert consensus) 3)other recurrent abnormalities