



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| --- | --- | --- | --- | --- | --- | --- | --- |
| *Gen* | *Chromosoom regio* | *Type afwijking* | *Startpositie* | *Eindpositie* | *Bron\** | *Ziekte* | *Evidence\** |
| FAF1 | 1p32.3 | loss | 50906935 | 51425936 | 1,2 | MM | 1 |
| CDKN2C | 1p32.3 | loss | 51434366 | 51440309 | 1,2 | MM | 1 |
| MTF2 | 1p22.1 | loss | 93544792 | 93604638 | 1,2 | MM | 2 |
| TMED5 | 1p22.1 | loss | 93615299 | 93646246 | 1,2 | MM | 2 |
| FAM46C | 1p12 | loss | 118148604 | 118171011 | 1,2 | MM | 1 |
| CKS1B | 1q21.3 | gain | 154947118 | 154951725 | 1,2 | MM | 1 |
| MYCN | 2p24.3 | gain | 16080559 | 16087129 | 2,7 | CLL | 1 |
| REL | 2p16.1 | gain | 61108629 | 61155291 | 2,7 | CLL | 2 |
| XPO1 | 2p15 | mutation | 61705070 | 61765418 | 7 | CLL | 2 |
| CXCR4 | 2q22.1 | mutation | 136871918 | 136875725 | 5 | LPL | 1 |
| SF3B1 | 2q33.1 | mutation | 198256700 | 198299771 | 7 | CLL | 1 |
| MYD88 | 3p22.2 | mutation | 38179969 | 38184510 | 7 | CLL,LPL | 1 |
| SETD2 | 3p21.31 | loss, cth | 47057897 | 47205467 | 6,7 | CLL | 1 |
| FGFR3 | 4p16.3 | t(4;14) | 1795039 | 1810599 | 1,3,4 | MM | 1 |
| WHSC1 | 4p16.3 | t(4;14) | 1873123 | 1983934 | 1,5 | MM | 1 |
| CCND3 | 6p21.1 | t(6;14) | 41902671 | 42016610 | 1,3,5 | MM | 1 |
| FOXO3 | 6q21 | loss | 108881025 | 109005971 | 7 | CLL | 2 |
| POT1 | 7q31.33 | mutation | 124462439 | 124570037 | 7 | CLL | 2 |
| BRAF | 7q34 | mutation | 140433812 | 140624564 | 5 | HCL | 1 |
| TRIM35 | 8p21.2 | loss | 27142402 | 27168836 | 7 | CLL | 2 |
| MYC | 8q24.21 | Focal gain, t(8;14) | 128748314 | 128753680 | 1,2,3,4,7 | CLL, MM | 2 |
| *Gen* | *Chromosoom regio* | *Type afwijking* | *Startpositie* | *Eindpositie* | *Bron\** | *Ziekte* | *Evidence\** |
| PVT1 | 8q24.21 | focal gain | 128806778 | 129113499 | 1,2 | MM | 2 |
| MAFA | 8q24.3 | t(8;14) | 144510229 | 144512602 | 5 | MM | 1 |
| NOTCH1 | 9q34.3 | mutation | 139388884 | 139440238 | 7 | CLL | 1 |
| PTEN | 10q23.31 | loss | 89623194 | 89731687 | 3 | MM | 1 |
| PIK3A | 10q24.1 | gain | 98353068 | 98480279 | 2 | CLL | 2 |
| CCND1 | 11q13.3 | t(11;14) | 69455873 | 69469242 | 1,3,4,5 | MM | 1 |
| MRE11A | 11q21 | loss | 94150468 | 94227040 | 7 | CLL | 2 |
| BIRC3 | 11q22.2 | loss, CNLOH | 102188193 | 102210135 | 2,7 | CLL | 1 |
| ATM | 11q22.3 | loss, CNLOH | 108093558 | 108239826 | 2,7 | CLL | 1 |
| H2AFX | 11q23.3 | loss | 118964584 | 118966177 | 7 | CLL | 2 |
| CCND2 | 12p13.32 | t(12;14) | 4382901 | 4414522 | 5 | MM | 1 |
| RB1 | 13q14.2 | loss | 48877883 | 49056026 | 1,2,3,5,7 | MM, CLL | 1 |
| DLEU regio | 13q14.2q14.3 | loss | 50456688 | 51417885 | 2,7 | CLL | 1 |
| TGDS | 13q32.1 | loss | 95226307 | 95248529 | 3 | MM | 1 |
| IGH | 14q32.33 | transl. | 106053225 | 106330470 | 3 | MM | 1 |
| MGA | 15q15.1 | loss | 41952610 | 42062141 | 2,7 | CLL | 2 |
| MAF | 16q23.2 | loss,t(14;16) | 79627744 | 79634622 | 1,3,4,5 | MM | 1 |
| TP53 | 17p13.1 | loss/CNLOH, mutatie | 7571719 | 7590868 | 1,2,4,5,7,8,9 | MM, CLL, HCL,LPL | 1 |
| MAFB | 20q12 | t(14;20) | 39314516 | 39317876 | 1,3,4,5 | MM | 1 |
| PRAME | 22q11.22 | gain | 22890117 | 22901768 | 3 | MM | 1 |

Bron:

1= genenlijst **WHGD** landelijk 2017

2 = artikel **Schoumans** et al., Guidelines for genomic array analysis in acquired haematological neoplastic disorders, **Genes, Chromosomes&Cancer 2016**

3= artikel **Pugh** et al., Assessing genome-wide copy number aberrations and copy-neutral loss-of-heterozygosity as best practice: An evidence-based review from the Cancer Genomics Consortium (CGC) working group for plasma cell disorders, **cancer genetics 2018**

4= artikel **Rack** et al., European recommandations and quality assurance for cytogenomic analysis of haematological neoplasm, **Leukemia 2019**

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

6= artikel **Parker** et al., Genomic disruption of the histone methyltransferase SETD2 in chronic lymphocytic leukaemia, **Leukemia** 2016

7= artikel **Chun** et al., Assessing copy number aberrations and copy-neutral loss-of-heterozygosity across the genome as best practice: An evidence-based review from the Cancer Genomics Consortium (CGC) working group for chronic lymphocytic leukemia, **cancer genetics 2018**

8= Advani et al; doi: 10.1016/j.hemonc.2019.05.002

9= Poulain et al; doi: 10.1158/1078-0432.CCR-17-0007)

Evidence: 1)present in WHO classification 2)recurrent (>5 cases for CNV en >2 for CNLOH in well-powered studies with expert consensus)