



|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| *Gen* | *Chromosoom regio* | *Type afwijking* | *Startpositie* | *Eindpositie* | *Bron\** | *Evidence\** |
| TAL1 | 1p33 | rearrangements | 47681961 | 47698007 | 3,5 | 1 |
| STIL | 1p33 | Loss, translocation | 47715810 | 47779819 | 5 | 1 |
| PBX1 | 1q23.3 | rearrangements | 164528801 | 164821045 | 3,4 | 1 |
| FHIT | 3p14.2 | loss | 59735035 | 61237133 | 1 | 2 |
| IL3 | 5q31.1 | rearrangements | 131396346 | 131398896 | 4 | 1 |
| PDGFRB | 5q32 | loss/rearrangements | 149493401 | 149535422 | 5 | 1 |
| EBF1 | 5q33.3 | loss | 158122924 | 158526788 | 1 | 2 |
| TLX3 | 5q35.1 | rearrangements | 170736287 | 170739138 | 3 | 1 |
| MYB | 6q23.3 | gain | 135502452 | 135540311 | 1 | 2 |
| IKZF1 | 7p12.2 | loss | 50344377 | 50472798 | 1 | 1 |
| TRB@ | 7q34 | rearrangements | 141872083 | 142007136 | 3 | 1 |
| CDKN2A | 9p21.3 | loss | 21967750 | 21994490 | 1 | 2 |
| CDKN2B | 9p21.3 | loss, CNLOH | 22002902 | 22009312 | 1,2 | 2 |
| PAX5 | 9p13.2 | loss, translocatie | 36833271 | 37034476 | 1 | 2 |
| ABL1 | 9q34.12 | rearrangements | 133589267 | 133763062 | 3,4 | 1 |
| NUP214 | 9q34.13 | gain, transl(NUP214-ABL1) | 134000980 | 134109091 | 3 | 1 |
| PTEN | 10q23.31 | loss | 89623194 | 89731687 | 1 | 2 |
| BLNK | 10q24.1 | loss | 97951454 | 98031333 | 1 | 2 |
| TLX1 | 10q24.31 | rearrangements | 102891060 | 102897546 | 3 | 1 |
| ADD3 | 10q25.1 | loss | 111756107 | 111895323 | 1 | 2 |
| LMO2 | 11p13 | rearrangements | 33880122 | 33913836 | 3 | 1 |
| *Gen* | *Chromosoom Regio* | *Type afwijking* | *Startpositie* | *Eindpositie* | *Bron\** | *Level of Evidence\** |
| RAG1 | 11p12 | loss | 36589563 | 36601310 | 1 | 2 |
| RAG2 | 11p12 | loss | 36613492 | 36619829 | 1 | 2 |
| KMT2A (MLL) | 11q23.3 | loss, rearrangement | 118307204 | 118397539 | 1,3,4 | 1 |
| ETV6 | 12p13.2 | loss/rearrangement | 11802787 | 12048325 | 1,3,4 | 1 |
| BTG1 | 12q21.33 | loss | 92534053 | 92539673 | 1 | 1 |
| RB1 | 13q14.2 | loss | 48877882 | 49056026 | 1 | 1 |
| TRAenD@ | 14q11.2 | rearrangements | 22615948 | 23016719 | 3 | 1 |
| IGH@ | 14q32.33 | rearrangements | 106053225 | 106330470 | 3 | 1 |
| NF1 | 17q11.2 | loss | 29421944 | 29704695 | 1 | 2 |
| IKZF3 | 17q12 | loss | 37913967 | 38020441 | 1 | 2 |
| HLF | 17q22 | rearrangements | 53342320 | 53402426 | 3 | 1 |
| TCF4 | 18q21.2 | loss | 52889562 | 53303224 | 1 | 2 |
| TCF3 | 19p13.3 | rearrangements | 1609291 | 1652326 | 3,4 | 1 |
| RUNX1 | 21q22.12 | gain/loss/ rearrangements | 36160097 | 36421595 | 1,3,4 | 2 |
| ERG | 21q22.2 | loss | 39739182 | 40033704 | 1 | 2 |
| TMPRSS2 | 21q22.3 | gain | 42836477 | 42880085 | 1 | 2 |
| BCR | 22q11.23 | rearrangements | 23522551 | 23660224 | 3,4 | 1 |
| CRLF2-P2RY8, CSF2RA, IL3RA | Xp22.33,  Yp11 | loss | 1314887 | 1656037 | 1 | 1 |

Bron:

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

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

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

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

5= Cancer Cytogentics, Heim et al., third edition

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