Suppl 5. Secondary genetic mutations among 193 cases with CBF – AML from Mitelman Database

| **Variables** | **Overall**  N = 193  n (%) | **inv(16**)  N = 76  n (%) | **t(8;21)**  N = 117  n (%) | **p-value** |
| --- | --- | --- | --- | --- |
| BCR-ABL1 | 12 (6.2) | 12 (15.8) | 0 (0.0) | <0.0011 |
| BRCC3 | 8 (4.1) | 0 (0.0) | 8 (6.8) | 0.0231 |
| ELN | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| TYK2 | 1 (0.5) | 0 (0.0) | 1 (0.9) | >0.9991 |
| LIMK1 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| PHF6 | 1 (0.5) | 0 (0.0) | 1 (0.9) | >0.9991 |
| D7S613 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| FRA7G | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| CAV2 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| DHX15 | 1 (0.5) | 0 (0.0) | 1 (0.9) | >0.9991 |
| NRAS | 8 (4.1) | 4 (5.3) | 4 (3.4) | 0.7141 |
| KRAS | 2 (1.0) | 2 (2.6) | 0 (0.0) | 0.1541 |
| KIT | 41 (21.2) | 17 (22.4) | 24 (20.5) | 0.7582 |
| BCORL1 | 2 (1.0) | 1 (1.3) | 1 (0.9) | >0.9991 |
| ADAM12 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| ARF3 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| CAND1 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| CCND1 | 3 (1.6) | 0 (0.0) | 3 (2.6) | 0.2801 |
| CCND2 | 10 (5.2) | 0 (0.0) | 10 (8.5) | 0.0071 |
| CMIP | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| DOCK6 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| KIF14 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| MIOX | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| JAK1 | 1 (0.5) | 0 (0.0) | 1 (0.9) | >0.9991 |
| MYOCD | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| EZH2 | 2 (1.0) | 0 (0.0) | 2 (1.7) | 0.5201 |
| RAD21 | 1 (0.5) | 0 (0.0) | 1 (0.9) | >0.9991 |
| NID2 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.394 |
| PRSS16 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| PTPRT | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| PTNP11 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| PTEN | 2 (1.0) | 1 (1.3) | 1 (0.9) | >0.9991 |
| GATA2 | 1 (0.5) | 0 (0.0) | 1 (0.9) | >0.9991 |
| TMEM125 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| ASXL1 | 11 (5.7) | 1 (1.3) | 10 (8.5) | 0.0531 |
| ASXL2 | 11 (5.7) | 0 (0.0) | 11 (9.4) | 0.0041 |
| RUNX1T1 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| FLT3 | 11 (5.7) | 5 (6.6) | 6 (5.1) | 0.7551 |
| CBL | 2 (1.0) | 2 (2.6) | 0 (0.0) | 0.1541 |
| PML-RARa | 1 (0.5) | 0 (0.0) | 1 (0.9) | >0.9991 |
| IDH2R140 | 2 (1.0) | 0 (0.0) | 2 (1.7) | 0.5201 |
| ZRSR2 | 3 (1.6) | 1 (1.3) | 2 (1.7) | >0.9991 |
| SF3B1 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| TET2 | 1 (0.5) | 0 (0.0) | 1 (0.9) | >0.9991 |
| CALR | 1 (0.5) | 0 (0.0) | 1 (0.9) | >0.9991 |
| WT1 | 1 (0.5) | 1 (1.3) | 0 (0.0) | 0.3941 |
| 1 Fisher's exact test, 2 Pearson's Chi-squared test  BCR-ABL1: BCR-ABL1 fusion gene mutation; BRCC3: BRCA1/BRCA2-containing complex 3 gene mutation; ELN: Elastin gene mutation; TYK2: Tyrosine kinase 2 gene mutation; LIMK1: LIM Domain Kinase 1 gene mutation; PHF6: PHD finger protein 6 gene mutation; D7S613: microsatellite markers D7S613; FRA7G: aphidicolin-induced fragile site on human chromosome 7 mutation; CAV2: Caveolin 2 gene mutation; DHX15: DEAH-Box Helicase 15 gene mutation; NRAS: neuroblastoma RAS viral gene mutation; KRAS: Kirsten rat sarcoma virus gene mutation; KIT: Receptor tyrosine kinase gene KIT mutation; BCORL1: BCORL1 gene mutation; ADAM12: Disintegrin and metalloproteinase domain-containing protein 12 gene mutation; ARF3: ADP-ribosylation factor 3 gene mutation; CAND1: Cullin-associated and neddylation-dissociated protein 1 gene mutation; CCND1: Cyclin D1 gene mutation; CCND2: Cyclin D2 gene mutation; CMIP: C-Maf inducing protein gene mutation; DOCK6: Dedicator of cytokinesis 6 gene mutation; KIF14: Kinesin family member 14 gene mutation; MIOX: Myo-Inositol oxigenase gene mutation; JAK1: Janus kinase 1 gene mutation; MYOCD: Myocardin gene mutation; EZH2: Enhancer Of Zeste 2 Polycomb Repressive Complex 2 Subunit gene mutation; RAD21: RAD21 Cohesin Complex Component gene mutation; NID2: Nidogen-2 gene mutation; PRSS16: Serine protease 16 gene mutation; PTPRT: Protein tyrosine phosphatase receptor type T gene mutation; PTNP11: Protein tyrosine phosphatase non-receptor type 11 gene mutation; PTEN: Phosphatase and tensin homolog gene mutation; GATA2: GATA binding protein 2 gene mutation; TMEM125: Transmembrane protein 125 gene mutation; ASXL1: ASXL Transcriptional regulator 1 gene mutation; ASXL2: ASXL Transcriptional regulator 2 gene mutation; RUNX1T1: RUNX1 partner transcriptional co-repressor 1 gene mutation; FLT3: FMS‐like tyrosine kinase 3 gene mutation; CBL: Casitas B-lineage lymphoma gene mutation; PML-RARa: Promyelocytic leukemia/retinoic acid receptor alpha gene mutation; IDH2R140: Isocitrate dehydrogenase 2 R140 gene mutation; ZRSR2: Zinc finger, RNA-binding motif and serine/arginine rich 2 gene mutation; SF3B1: Splicing factor 3b subunit 1 gene mutation; TET2: Tet methylcytosine dioxygenase 2 gene mutation; CALR: Calreticulin gene mutation; WT1: Wilms' tumor 1 gene mutation. | | | | |