

I.

Einzelgenliste  
(Sangersequenzierung; alphabetisch)

| <b>GEN</b>      | <b>OMIM</b> | <b>NM</b>      | <b>ENST</b>       |
|-----------------|-------------|----------------|-------------------|
| <b>ACD</b>      | OMIM 609377 | NM_001082486.1 | ENST00000620338.4 |
| <b>ANKRD26</b>  | OMIM 610855 | NM_014915.2    | ENST00000376087.4 |
| <b>ASXL1</b>    | OMIM 605039 | NM_015338.5    | ENST00000375687   |
| <b>C16orf57</b> | OMIM 613276 | NM_024598.3    | ENST00000219281   |
| <b>CBL</b>      | OMIM 165360 | NM_005188.3    | ENST00000264033   |
| <b>CEBPA</b>    | OMIM 116897 | NM_004364.4    | ENST00000498907   |
| <b>CECR1</b>    | OMIM 607575 | NM_001282225.1 | ENST00000399837   |
| <b>c-Kit</b>    | OMIM 164920 | NM_000222.2    | ENST00000288135   |
| <b>c-MPL</b>    | OMIM 159530 | NM_005373.2    | ENST00000372470   |
| <b>CTC1</b>     | OMIM 613129 | NM_025099.5    | ENST00000315684   |
| <b>CXCR4</b>    | OMIM 162643 | NM_003467.2    | ENST00000241393   |
| <b>DKC1</b>     | OMIM 300126 | NM_001363.4    | ENST00000369550   |
| <b>ETV6</b>     | OMIM 600618 | NM_001987.4    | ENST00000396373   |
| <b>GATA1</b>    | OMIM 305371 | NM_002049.3    | ENST00000376670   |
| <b>Gata2</b>    | OMIM 137295 | NM_032638.4    | ENST00000341105   |
| <b>HOXA11</b>   | OMIM 142958 | NM_005523.5    | ENST00000006015   |
| <b>JAK2</b>     | OMIM 147796 | NM_004972.3    | ENST00000381652   |
| <b>KLF1</b>     | OMIM 600599 | NM_006563.3    | ENST00000264834   |
| <b>KRAS</b>     | OMIM 190070 | NM_004985.4    | ENST00000311936   |
| <b>NHP2</b>     | OMIM 606470 | NM_017838.3    | ENST00000274606   |
| <b>NOP10</b>    | OMIM 606471 | NM_018648.3    | ENST00000328848   |
| <b>NRAS</b>     | OMIM 164790 | NM_002524.4    | ENST00000369535   |
| <b>PARN</b>     | OMIM 604212 | NM_002582.3    | ENST00000437198   |
| <b>PTPN11</b>   | OMIM 176876 | NM_002834.3    | ENST00000351677   |
| <b>RPL11</b>    | OMIM 604175 | NM_000975.5    | ENST00000643754.2 |
| <b>RPL15</b>    | OMIM 604174 | NM_002948.3    | ENST00000307839   |
| <b>RPL26</b>    | OMIM 603704 | NM_000987.3    | ENST00000293842   |
| <b>RPL35A</b>   | OMIM 180468 | NM_000996.2    | ENST00000464167   |
| <b>RPL5</b>     | OMIM 603634 | NM_000969.3    | ENST00000370321   |
| <b>RPL9</b>     | OMIM 603686 | NM_000661.4    | ENST00000295955   |
| <b>RPS10</b>    | OMIM 603632 | NM_001014.4    | ENST00000326199   |
| <b>RPS17</b>    | OMIM 180472 | NM_001021.4    | ENST00000330244   |

|               |             |                |                 |
|---------------|-------------|----------------|-----------------|
| <b>RPS19</b>  | OMIM 603474 | NM_001022.3    | ENST00000598742 |
| <b>RPS24</b>  | OMIM 602412 | NM_001026.4    | ENST00000372360 |
| <b>RPS26</b>  | OMIM 603701 | NM_001029.3    | ENST00000356464 |
| <b>RPS28</b>  | OMIM 603685 | NM_001031.4    | ENST00000600659 |
| <b>RPS29</b>  | OMIM 603633 | NM_001032.4    | ENST00000245458 |
| <b>RPS7</b>   | OMIM 603658 | NM_001011.3    | ENST00000304921 |
| <b>RPSA</b>   | OMIM150370  | NM_002295.5    | ENST00000301821 |
| <b>RRAS2</b>  | OMIM 600098 | NM_012250      |                 |
| <b>RTEL1</b>  | OMIM 608833 | NM_032957.4    | ENST00000508582 |
| <b>RUNX1</b>  | OMIM 151385 | NM_001754.4    | ENST00000437180 |
| <b>SBDS</b>   | OMIM 607444 | NM_016038.2    | ENST00000246868 |
| <b>SETBP1</b> | OMIM 611060 | NM_015559.2    | ENST00000282030 |
| <b>TERC</b>   | OMIM 602322 | NR_001566.1    |                 |
| <b>TERT</b>   | OMIM 187270 | NM_198253.2    | ENST00000310581 |
| <b>TINF2</b>  | OMIM 604319 | NM_001099274.1 | ENST00000267415 |
| <b>TSR2</b>   | OMIM 300945 | NM_058163.1    | ENST00000375151 |
| <b>WRAP53</b> | OMIM 612661 | NM_018081.2    | ENST00000316024 |

|            |                 |
|------------|-----------------|
| <b>II.</b> | Krankheitsliste |
|------------|-----------------|

### 1. JMML-NGS panel

| <b>Gen</b> | <b>Exon</b> |
|------------|-------------|
| ASXL1      | 12, 13      |
| CBL        | 7-10        |
| GATA2      | 2-6         |
| GATA2_ebox | Intron 4    |
| JAK2       | 14          |
| JAK3       | 11-19, 21   |
| KRAS       | 2-5         |
| NF1        | 1-58        |
| NRAS       | 2-5         |
| PTPN11     | 2-15        |
| RAC2       | 1-6         |
| RRAS       | 1-6         |
| RUNX1      | 2-9         |
| SETBP1     | 4           |
| SH2B3      | 3-7         |

## 2. DBA-NGS panel

| <b>Gen</b> | <b>Exon</b> |
|------------|-------------|
| CECR1      | 1-10        |
| EPO        | 1-5         |
| GATA1      | 2-6         |
| RPL10      | 1-6         |
| RPL10A     | 1-6         |
| RPL11      | 1-6         |
| RPL15      | 2-5         |
| RPL18      | 1-7         |
| RPL19      | 1-6         |
| RPL26      | 2-4         |
| RPL27      | 2-5         |
| RPL3       | 1-10        |
| RPL31      | 2-5         |
| RPL34      | 2-5         |
| RPL35      | 1-4         |
| RPL35A     | 2-5         |
| RPL37      | 1-4         |
| RPL5       | 1-8         |
| RPL9       | 1-6         |
| RPLP0      | 2-8         |
| RPS10      | 2-6         |
| RPS11      | 1-5         |
| RPS15A     | 2-5         |
| RPS17      | 1-5         |
| RPS19      | 2-6         |
| RPS20      | 1-5         |
| RPS24      | 1-5         |
| RPS26      | 1-4         |
| RPS27      | 1-4         |
| RPS28      | 1-3         |
| RPS29      | 1-3         |
| RPS7       | 2-7         |
| TP53       | 2-11        |
| TSR2       | 1-5         |

### 3. RCC/SAA-NGS panel

| <b>Gen</b> | <b>Exon</b> |
|------------|-------------|
| RUNX1      | 2-9         |
| GATA2_ebox | Intron 4    |
| SAMD9L     | 5           |
| SAMD9      | 3           |
| GATA2      | 2-6         |

### 4. DC-NGS panel

| <b>Gen</b> | <b>Exon</b> |
|------------|-------------|
| ACD        | 1-12        |
| CTC1       | 1-23        |
| DKC1       | 1-15        |
| NAF1       | 1-8         |
| NHP2       | 1-4         |
| NOP10      | 1-2         |
| PARN       | 1-23        |
| POT1       | 5-19        |
| RPA1       | 1-17        |
| RTEL1      | 2-35        |
| SAMD9      | 3           |
| SAMD9L     | 5           |
| TERC       | 1           |
| TERT       | 1-16        |
| TERT-Prom  |             |
| TINF2      | 1-6         |
| USB1       | 1-7         |
| WRAP53     | 1-10        |

#### 4.NGS BMF-MDS-NGS panel

| Gen     | Exon               |
|---------|--------------------|
| ANKRD26 | UTR-5'             |
| ARID2   | 1-21               |
| ASXL1   | 12-13              |
| BCOR    | 1-14               |
| BCORL1  | 1-13               |
| BRAF    | 11, 15             |
| CBL     | 8-9                |
| CEBPA   | 1                  |
| cKIT    | 2,8-13             |
| CSF3R   | 14-17              |
| CSMD1   | 1-70               |
| CTC1    | 1-23               |
| CTCF    | 1-10               |
| CTLA4   | 1-4                |
| CUX1    | 1-33               |
| DDX41   | 1-17               |
| DKC1    | 1-15               |
| DNMT3A  | 16-23              |
| DNAJC21 | 1-13               |
| EFL1    | 1-19               |
| ERCC6L2 | 1-19               |
| ETV6    | 1-8                |
| EZH2    | 8,15-19            |
| FLT3    | 11-12,14-15,20     |
| G6PC3   | 1-6                |
| GATA1   | 1-5                |
| GATA2   | 1-5,Ebox           |
| HLA-A   | 1-8                |
| HLA-B   | 1-7                |
| HLA-C   | 1-8                |
| HLTF    | 1-25               |
| HOXA11  | 1-2                |
| IDH1    | 4                  |
| IDH2    | 4                  |
| IKZF1   | 1-9                |
| JAK2    | 12,14              |
| JAK3    | 13,15,17,19,21     |
| KRAS    | 1-5                |
| LIG4    | 1-2                |
| MECOM   | 5-13               |
| MPL     | 10                 |
| MYSM1   | 1-20               |
| NF1     | 1-58               |
| NHP2    | 1-4                |
| NOP10   | 1-2                |
| NOTCH1  | 1-34               |
| NPM1    | 12                 |
| NRAS    | 2-3                |
| PARN    | 1-24               |
| PHF6    | 1-9                |
| PIGA    | 1-6                |
| PPM1D   | 6                  |
| PTPN11  | 1-15               |
| RAD21   | 1-13               |
| RBM8A   | 1-6, UTR5, Intron1 |
| RPA1    | 1-17               |
| RRAS    | 1-6                |
| RTEL1   | 1-34               |
|         |                    |
|         |                    |

|               |           |
|---------------|-----------|
| RUNX1         | 1-9       |
| SAMD9         | 3         |
| SAMD9L        | 4         |
| SETBP1        | 4 Hotspot |
| SMARCA2       | 1-36      |
| SMARCD2       | 1-14      |
| SRP54         | 1-15      |
| SRP72         | 1-19      |
| SRSF2         | 1         |
| STAG2         | 1-33      |
| STAT3         | 1-23      |
| STAT5         | 15-18     |
| TCAB1/WRAP53  | 1-10      |
| TERC          | 1         |
| TERT          | 1-16      |
| TERT promoter |           |
| TET2          | 3,8-9     |
| THPO          | 1-6       |
| TINF2         | 6         |
| TP53          | 1-12      |
| TPP1          | 1-13      |
| U2AF1         | 2,6       |
| USB1          | 1-9       |
| WT1           | 1-11      |

| III.                         | Bearbeitungsdauer |
|------------------------------|-------------------|
| Methode                      | Dauer             |
| <b>Sanger (JMML)</b>         | 7 Werktage        |
| <b>Sanger (Einzelgene)</b>   | 7 Werktage        |
| <b>NGSPanel JMML</b>         | 4-6 Wochen        |
| <b>NGSPanel DBA</b>          | 4-6 Wochen        |
| <b>NGSPanel DC</b>           | 4-6 Wochen        |
| <b>NGSPanel RCC/SAA</b>      | 4-6 Wochen        |
| <b>NGS BMF-MDS-NGS panel</b> | 4-6 Wochen        |