

Exome

Gene panel Bone Marrow Failure (172 genes)

version 2, 09-12-2019

| HGNC approved gene symbol | transcript | median depth | % covered >10x | % covered >20x | % covered >30x | % covered >50x |
|------------------------------|----------------|-----------------|-------------------|-------------------|-------------------|-------------------|
| <i>ABCB6</i> | NM_005689.4 | 111.00 | 100.00 | 99.99 | 99.59 | 95.88 |
| <i>ABCB7</i> | NM_001271696.3 | 91.00 | 100.00 | 99.92 | 99.59 | 97.93 |
| <i>ABL1</i> | NM_005157.6 | 102.00 | 99.77 | 98.16 | 97.49 | 96.43 |
| <i>ACTB</i> | NM_001101.5 | 154.00 | 100.00 | 100.00 | 99.80 | 99.50 |
| <i>ACTN1</i> | NM_001130004.1 | 81.00 | 99.86 | 99.11 | 97.26 | 90.24 |
| <i>ALAS2</i> | NM_000032.5 | 71.00 | 99.96 | 98.57 | 93.16 | 76.45 |
| <i>ANKRD26</i> | NM_014915.2 | 104.00 | 97.43 | 93.90 | 89.45 | 81.40 |
| <i>AP3B1</i> | NM_003664.4 | 99.00 | 98.98 | 97.16 | 95.65 | 90.58 |
| <i>ASXL1</i> | NM_015338.6 | 167.00 | 98.78 | 98.71 | 98.42 | 96.97 |
| <i>ATRX</i> | NM_000489.5 | 99.00 | 100.00 | 99.76 | 98.36 | 91.13 |
| <i>BCOR</i> | NM_001123385.2 | 98.00 | 99.97 | 99.70 | 98.65 | 94.11 |
| <i>BCORL1</i> | NM_001184772.2 | 76.00 | 99.81 | 98.14 | 92.41 | 81.99 |
| <i>BRAF</i> | NM_004333.6 | 105.00 | 99.99 | 99.70 | 99.25 | 95.30 |
| <i>BRCA1</i> | NM_007294.4 | 275.00 | 100.00 | 100.00 | 100.00 | 99.90 |
| <i>BRCA2</i> | NM_000059.3 | 191.00 | 100.00 | 100.00 | 99.99 | 99.76 |
| <i>BRIP1</i> | NM_032043.3 | 135.00 | 100.00 | 100.00 | 100.00 | 99.86 |
| <i>C15orf41</i> | NM_001321759.2 | 110.00 | 100.00 | 100.00 | 100.00 | 99.52 |
| <i>CALR</i> | NM_004343.3 | 91.00 | 100.00 | 98.41 | 94.67 | 87.37 |
| <i>CBL</i> | NM_005188.4 | 121.00 | 100.00 | 100.00 | 99.98 | 99.80 |
| <i>CBLB</i> | NM_170662.5 | 117.00 | 99.78 | 99.27 | 98.32 | 95.48 |
| <i>CDAN1</i> | NM_138477.4 | 77.00 | 100.00 | 99.11 | 94.46 | 80.10 |
| <i>CDC42</i> | NM_001791.4 | 239.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>CDKN2A</i> | NM_000077.4 | 170.00 | 100.00 | 99.50 | 97.48 | 92.36 |
| <i>CEBPA</i> | NM_004364.4 | 75.00 | 88.28 | 69.96 | 63.82 | 61.51 |
| <i>CECR1/ADA2</i> | NM_001282225 | 125.00 | 100.00 | 100.00 | 99.99 | 99.93 |
| <i>CSF3R</i> | NM_156039.3 | 75.00 | 99.67 | 97.21 | 93.58 | 85.38 |
| <i>CTC1</i> | NM_025099.6 | 107.00 | 100.00 | 99.95 | 98.70 | 93.26 |
| <i>CTLA4</i> | NM_005214.5 | 133.00 | 100.00 | 100.00 | 100.00 | 98.97 |
| <i>CUX1</i> | NM_181552.4 | 82.00 | 98.16 | 95.51 | 91.62 | 85.53 |
| <i>CXCR4</i> | NM_003467.3 | 172.00 | 100.00 | 100.00 | 100.00 | 99.21 |
| <i>CYCS</i> | NM_018947.6 | 246.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>DKC1</i> | NM_001363.5 | 76.00 | 99.70 | 97.99 | 94.81 | 86.99 |
| <i>DNAJC21</i> | NM_001012339.3 | 80.00 | 99.99 | 99.57 | 97.95 | 88.57 |
| <i>DNMT3A</i> | NM_022552.4 | 82.00 | 99.86 | 97.74 | 91.61 | 81.41 |
| <i>DTNBP1</i> | NM_032122.4 | 88.00 | 100.00 | 99.97 | 99.65 | 92.80 |
| <i>ELANE</i> | NM_001972.4 | 73.00 | 100.00 | 100.00 | 99.83 | 93.64 |
| <i>ENG</i> | NM_001114753.2 | 70.00 | 99.84 | 98.42 | 94.65 | 83.14 |
| <i>EPG5</i> | NM_020964.3 | 109.00 | 100.00 | 99.81 | 98.80 | 95.03 |
| <i>EPO</i> | NM_000799.4 | 80.00 | 100.00 | 99.17 | 90.78 | 72.64 |
| <i>ERCC4</i> | NM_005236.3 | 123.00 | 100.00 | 99.99 | 99.47 | 94.70 |
| <i>ERCC6L2</i> | NM_020207.5 | 128.00 | 100.00 | 99.99 | 99.90 | 98.74 |
| <i>ETV6</i> | NM_001987.5 | 113.00 | 100.00 | 100.00 | 99.94 | 98.69 |
| <i>EZH2</i> | NM_004456.5 | 100.00 | 100.00 | 99.95 | 99.50 | 98.61 |
| <i>FANCA</i> | NM_000135.4 | 88.00 | 100.00 | 99.40 | 96.04 | 82.95 |

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|--------------------------------------|-------------------|-------------------------|------------------------------|------------------------------|------------------------------|------------------------------|
| <i>FANCB</i> | NM_001018113.3 | 111.00 | 100.00 | 99.84 | 97.74 | 94.77 |
| <i>FANCC</i> | NM_000136.3 | 91.00 | 100.00 | 100.00 | 99.61 | 93.15 |
| <i>FANCD2</i> | NM_001018115.2 | 110.00 | 100.00 | 100.00 | 99.96 | 99.63 |
| <i>FANCE</i> | NM_021922.3 | 87.00 | 95.36 | 89.17 | 86.16 | 80.81 |
| <i>FANCF</i> | NM_022725.4 | 206.00 | 100.00 | 100.00 | 99.86 | 98.80 |
| <i>FANCG</i> | NM_004629.1 | 106.00 | 100.00 | 99.71 | 97.91 | 91.91 |
| <i>FANCI</i> | NM_001113378.1 | 117.00 | 100.00 | 100.00 | 99.99 | 99.61 |
| <i>FANCL</i> | NM_018062.3 | 91.00 | 100.00 | 100.00 | 99.95 | 92.89 |
| <i>FANCM</i> | NM_020937.4 | 138.00 | 100.00 | 99.87 | 99.11 | 96.06 |
| <i>FERMT3</i> | NM_178443.2 | 82.00 | 99.49 | 95.87 | 92.05 | 81.85 |
| <i>FLI1</i> | NM_002017.5 | 115.00 | 100.00 | 99.89 | 99.53 | 97.78 |
| <i>FLT3</i> | NM_004119.3 | 112.00 | 100.00 | 99.49 | 98.67 | 98.06 |
| <i>G6PC3</i> | NM_138387.3 | 83.00 | 99.75 | 94.51 | 85.88 | 79.06 |
| <i>GATA1</i> | NM_002049.4 | 57.00 | 99.23 | 93.53 | 86.02 | 67.31 |
| <i>GATA2</i> | NM_032638.5 | 74.00 | 100.00 | 98.80 | 92.96 | 74.13 |
| <i>GFI1</i> | NM_005263.5 | 69.00 | 100.00 | 99.65 | 98.03 | 88.08 |
| <i>GFI1B</i> | NM_004188.7 | 106.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>GLRX5</i> | NM_016417.3 | 33.00 | 81.54 | 65.97 | 53.73 | 38.54 |
| <i>GNAS</i> | NM_080425.3 | 169.00 | 100.00 | 99.92 | 99.53 | 96.20 |
| <i>GRHL2</i> | NM_024915.4 | 103.00 | 100.00 | 99.87 | 99.02 | 93.15 |
| <i>HAX1</i> | NM_006118.4 | 109.00 | 100.00 | 100.00 | 99.99 | 96.77 |
| <i>HOXA11</i> | NM_005523.6 | 100.00 | 100.00 | 99.41 | 94.74 | 80.77 |
| <i>IDH1</i> | NM_005896.3 | 121.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>IDH2</i> | NM_002168.3 | 119.00 | 98.97 | 94.44 | 91.84 | 91.21 |
| <i>IKZF1</i> | NM_006060.6 | 135.00 | 100.00 | 99.82 | 98.86 | 97.74 |
| <i>JAK2</i> | NM_004972.3 | 107.00 | 99.99 | 99.91 | 99.37 | 97.86 |
| <i>JAK3</i> | NM_000215.3 | 91.00 | 99.97 | 99.32 | 97.45 | 91.76 |
| <i>KDM6A</i> | NM_001291415.1 | 93.00 | 100.00 | 99.60 | 97.93 | 88.13 |
| <i>KIF23</i> | NM_138555.4 | 109.00 | 100.00 | 99.97 | 99.31 | 94.44 |
| <i>KIT</i> | NM_000222.2 | 113.00 | 100.00 | 100.00 | 100.00 | 99.91 |
| <i>KLF1</i> | NM_006563.5 | 77.00 | 99.93 | 98.37 | 92.07 | 84.38 |
| <i>KMT2A</i> | NM_001197104.1 | 159.00 | 99.73 | 99.18 | 98.38 | 96.31 |
| <i>KRAS</i> | NM_004985.5 | 120.00 | 100.00 | 99.32 | 98.31 | 88.38 |
| <i>LIG4</i> | NM_002312.3 | 286.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>LPIN2</i> | NM_014646.2 | 106.00 | 100.00 | 99.99 | 99.87 | 99.17 |
| <i>LYST</i> | NM_000081.4 | 130.00 | 100.00 | 99.98 | 99.80 | 98.24 |
| <i>MAD2L2</i> | NM_006341.4 | 87.00 | 100.00 | 100.00 | 98.26 | 79.72 |
| <i>MECOM</i> | NM_004991.4 | 158.00 | 100.00 | 99.95 | 99.34 | 95.32 |
| <i>MLPH</i> | NM_024101.7 | 79.00 | 99.38 | 93.33 | 90.32 | 84.76 |
| <i>MPIG6B</i> | NM_025260 | 77.00 | 99.28 | 93.06 | 87.40 | 84.12 |
| <i>MPL</i> | NM_005373.3 | 100.00 | 100.00 | 99.99 | 99.91 | 97.99 |
| <i>MYD88</i> | NM_001172567.2 | 102.00 | 100.00 | 100.00 | 99.74 | 92.16 |
| <i>MYH9</i> | NM_002473.5 | 82.00 | 99.88 | 97.73 | 91.58 | 77.76 |
| <i>MYSM1</i> | NM_001085487.3 | 131.00 | 99.93 | 98.75 | 96.25 | 93.05 |
| <i>NHP2</i> | NM_017838.3 | 101.00 | 100.00 | 100.00 | 100.00 | 99.83 |
| <i>NOP10</i> | NM_018648.3 | 103.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>NOTCH1</i> | NM_017617.5 | 96.00 | 98.99 | 97.62 | 94.26 | 85.40 |

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| <i>NPM1</i> | NM_002520.6 | 141.00 | 94.61 | 93.42 | 93.23 | 90.43 |
| <i>NRAS</i> | NM_002524.5 | 132.00 | 100.00 | 100.00 | 100.00 | 99.96 |
| <i>PALB2</i> | NM_024675.4 | 239.00 | 100.00 | 100.00 | 99.94 | 99.29 |
| <i>PARN</i> | NM_002582.4 | 101.00 | 100.00 | 100.00 | 99.92 | 97.88 |
| <i>PDGFRA</i> | NM_006206.6 | 114.00 | 100.00 | 100.00 | 99.99 | 99.83 |
| <i>PHF6</i> | NM_001015877.2 | 59.00 | 99.99 | 98.08 | 89.74 | 63.33 |
| <i>PIEZO1</i> | NM_001142864.4 | 89.00 | 99.90 | 98.50 | 93.66 | 80.76 |
| <i>PRKACG</i> | NM_002732.3 | 174.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>PTEN</i> | NM_000314.8 | 152.00 | 100.00 | 100.00 | 100.00 | 99.55 |
| <i>PTPN11</i> | NM_002834.4 | 137.00 | 99.19 | 99.17 | 99.17 | 99.17 |
| <i>PUS1</i> | NM_025215.6 | 98.00 | 100.00 | 99.76 | 97.98 | 93.40 |
| <i>RAB27A</i> | NM_183235.3 | 120.00 | 100.00 | 100.00 | 100.00 | 99.59 |
| <i>RAD21</i> | NM_006265.3 | 100.00 | 100.00 | 100.00 | 99.92 | 98.46 |
| <i>RAD51</i> | NM_002875.5 | 98.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>RAD51C</i> | NM_058216.3 | 116.00 | 100.00 | 100.00 | 99.92 | 98.06 |
| <i>RASGRP2</i> | NM_001098671.2 | 91.00 | 100.00 | 99.62 | 96.87 | 83.68 |
| <i>RBM8A</i> | NM_005105.4 | 102.00 | 100.00 | 100.00 | 99.84 | 99.08 |
| <i>RGS2</i> | NM_002923.4 | 91.00 | 100.00 | 100.00 | 99.84 | 98.84 |
| <i>RMRP</i> | NR_003051 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| <i>RPL11</i> | NM_000975.5 | 103.00 | 100.00 | 100.00 | 100.00 | 96.50 |
| <i>RPL15</i> | NM_001253383.3 | 101.00 | 99.89 | 98.96 | 95.97 | 93.10 |
| <i>RPL17</i> | NM_001199343.2 | 149.00 | 100.00 | 100.00 | 100.00 | 99.73 |
| <i>RPL18</i> | NM_000979.4 | 92.00 | 100.00 | 100.00 | 99.58 | 96.96 |
| <i>RPL26</i> | NM_000987.5 | 113.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>RPL27</i> | NM_000988.5 | 171.00 | 100.00 | 100.00 | 100.00 | 99.79 |
| <i>RPL31</i> | NM_000993.5 | 151.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>RPL35A</i> | NM_000996.4 | 170.00 | 99.88 | 97.95 | 93.28 | 92.58 |
| <i>RPL36</i> | NM_033643.3 | 75.00 | 99.87 | 95.34 | 85.89 | 71.59 |
| <i>RPL5</i> | NM_000969.5 | 115.00 | 99.76 | 97.47 | 94.63 | 91.69 |
| <i>RPL9</i> | NM_001024921.3 | 122.00 | 100.00 | 100.00 | 100.00 | 99.56 |
| <i>RPS10</i> | NM_001014.5 | 129.00 | 100.00 | 100.00 | 99.97 | 99.49 |
| <i>RPS14</i> | NM_005617.4 | 136.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>RPS15</i> | NM_001018.4 | 162.00 | 84.04 | 80.32 | 80.32 | 80.26 |
| <i>RPS15A</i> | NM_001019.5 | 101.00 | 100.00 | 100.00 | 100.00 | 99.96 |
| <i>RPS17</i> | NM_001021.6 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| <i>RPS19</i> | NM_001022.4 | 84.00 | 100.00 | 100.00 | 99.57 | 97.74 |
| <i>RPS24</i> | NM_033022.4 | 218.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>RPS26</i> | NM_001029.5 | 98.00 | 100.00 | 100.00 | 100.00 | 99.93 |
| <i>RPS27</i> | NM_001030.6 | 110.00 | 100.00 | 100.00 | 99.69 | 97.33 |
| <i>RPS27A</i> | NM_001177413.1 | 147.00 | 100.00 | 100.00 | 100.00 | 99.36 |
| <i>RPS28</i> | NM_001031.5 | 33.00 | 99.93 | 89.83 | 59.83 | 26.50 |
| <i>RPS29</i> | NM_001032.4 | 177.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>RPS7</i> | NM_001011.4 | 153.00 | 92.30 | 87.48 | 85.87 | 81.52 |
| <i>RPS8</i> | NM_001012.2 | 105.00 | 100.00 | 100.00 | 100.00 | 99.33 |
| <i>RTKL1</i> | NM_001283009.2 | 90.00 | 100.00 | 99.89 | 98.36 | 86.89 |
| <i>RTL1</i> | NM_001134888.2 | 180.00 | 100.00 | 99.94 | 99.60 | 98.43 |
| <i>RUNX1</i> | NM_001754.4 | 62.00 | 99.92 | 97.26 | 86.93 | 67.08 |

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| <i>SAMD9</i> | NM_017654.4 | 305.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>SAMD9L</i> | NM_152703.5 | 314.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>SBDS</i> | NM_016038.4 | 131.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>SEC23B</i> | NM_006363.6 | 114.00 | 100.00 | 99.88 | 98.62 | 95.62 |
| <i>SETBP1</i> | NM_015559.3 | 149.00 | 99.37 | 98.06 | 96.13 | 92.51 |
| <i>SF3B1</i> | NM_012433.3 | 116.00 | 99.89 | 98.67 | 96.62 | 93.73 |
| <i>SLC25A38</i> | NM_017875.4 | 157.00 | 100.00 | 100.00 | 100.00 | 99.53 |
| <i>SLC37A4</i> | NM_001164277.1 | 99.00 | 100.00 | 99.72 | 98.33 | 94.76 |
| <i>SLX4</i> | NM_032444.4 | 108.00 | 99.95 | 99.40 | 97.80 | 93.27 |
| <i>SMC1A</i> | NM_006306.4 | 90.00 | 99.96 | 99.41 | 98.06 | 92.98 |
| <i>SMC3</i> | NM_005445.3 | 90.00 | 99.96 | 99.33 | 96.55 | 89.06 |
| <i>SRP72</i> | NM_006947.4 | 112.00 | 100.00 | 100.00 | 100.00 | 99.89 |
| <i>SRSF2</i> | NM_001195427.2 | 195.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>STAG2</i> | NM_001042750.2 | 74.00 | 99.83 | 98.63 | 95.78 | 84.54 |
| <i>STEAP3</i> | NM_182915.3 | 105.00 | 99.91 | 98.04 | 95.30 | 88.53 |
| <i>STIM1</i> | NM_003156.3 | 108.00 | 100.00 | 99.98 | 99.61 | 96.79 |
| <i>STK4</i> | NM_006282.5 | 107.00 | 99.61 | 98.36 | 96.88 | 91.64 |
| <i>TAZ</i> | NM_000116.5 | 59.00 | 99.42 | 96.11 | 88.35 | 67.89 |
| <i>TERC</i> | NR_001566 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| <i>TERT</i> | NM_198253.3 | 84.00 | 99.76 | 97.73 | 95.01 | 87.45 |
| <i>TET2</i> | NM_001127208.2 | 214.00 | 100.00 | 99.97 | 99.60 | 98.45 |
| <i>THPO</i> | NM_000460.4 | 163.00 | 100.00 | 99.87 | 99.02 | 95.94 |
| <i>TINF2</i> | NM_001099274.3 | 157.00 | 100.00 | 100.00 | 100.00 | 99.93 |
| <i>TP53</i> | NM_000546.5 | 130.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>TSR2</i> | NM_058163.3 | 60.00 | 99.92 | 96.32 | 85.84 | 63.00 |
| <i>U2AF1</i> | NM_006758.2 | 188.00 | 100.00 | 100.00 | 100.00 | 96.91 |
| <i>UBE2T</i> | NM_014176.4 | 108.00 | 100.00 | 100.00 | 100.00 | 99.62 |
| <i>USB1</i> | NM_024598.4 | 78.00 | 99.35 | 92.32 | 81.86 | 76.72 |
| <i>WAS</i> | NM_000377.3 | 53.00 | 99.58 | 94.05 | 81.44 | 54.23 |
| <i>WRAP53</i> | NM_018081.2 | 99.00 | 100.00 | 99.93 | 99.30 | 96.51 |
| <i>WT1</i> | NM_024426.6 | 70.00 | 99.88 | 98.27 | 91.39 | 74.72 |
| <i>XRCC2</i> | NM_005431.2 | 226.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>YARS2</i> | NM_001040436.3 | 139.00 | 100.00 | 100.00 | 100.00 | 100.00 |
| <i>ZRSR2</i> | NM_005089.3 | 79.00 | 94.19 | 86.42 | 78.72 | 67.92 |