

WES CONGENITAL HEARTDISEASE ¹ DG 2.16

| <i>Gene</i> | <i>Median coverage</i> | <i>% covered >10x</i> | <i>% covered >20x</i> | <i>OMIM disease ID</i> |
|-------------|------------------------|--------------------------|--------------------------|------------------------|
| ACTC1 | 111.1 | 100.0 | 98.9 | 102540 |
| ACVR2B | 115.6 | 99.7 | 97.0 | 602730 |
| ALDH1A2 | 105.1 | 99.9 | 98.6 | 603687 |
| ANKRD1 | 98.1 | 99.9 | 98.6 | 609599 |
| BRAF | 72.5 | 92.4 | 80.2 | 164757 |
| CFAP53 | 131.8 | 99.1 | 97.0 | 614759 |
| CFC1 | 125.8 | 91.0 | 80.1 | 605194 |
| CHD7 | 137.0 | 99.9 | 99.4 | 608892 |
| CITED2 | 149.7 | 99.2 | 99.0 | 602937 |
| CRELD1 | 98.9 | 99.8 | 95.9 | 607170 |
| EHMT1 | 127.7 | 94.6 | 94.2 | 607001 |
| ELN | 103.1 | 100.0 | 98.9 | 130160 |
| FBN1 | 137.1 | 100.0 | 99.8 | 134797 |
| FLT4 | 160.3 | 99.2 | 99.1 | 136352 |
| FOXC2 | 122.3 | 100.0 | 100.0 | 602402 |
| FOXH1 | 84.5 | 100.0 | 99.5 | 603621 |
| FOXL1 | 144.0 | 100.0 | 99.4 | 603252 |
| GATA4 | 87.6 | 95.9 | 86.7 | 600476 |
| GATA5 | 74.0 | 100.0 | 99.2 | 611496 |
| GATA6 | 110.2 | 98.3 | 92.5 | 601656 |
| GDF1 | 50.7 | 97.8 | 84.7 | 602880 |
| GJA5 | 207.8 | 100.0 | 100.0 | 121013 |
| HAND1 | 162.9 | 100.0 | 100.0 | 602406 |
| HAND2 | 85.1 | 100.0 | 99.2 | 602407 |
| HEY2 | 162.7 | 99.4 | 96.6 | 604674 |

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|---------|-------|-------|-------|--------|
| JAG1 | 133.7 | 99.2 | 97.1 | 601920 |
| KMT2D | 136.2 | 100.0 | 99.7 | 602113 |
| KRAS | 67.2 | 99.4 | 97.3 | 190070 |
| LEFTY2 | 69.3 | 99.5 | 91.7 | 601877 |
| MCTP2 | 120.6 | 99.5 | 97.4 | 616297 |
| MED13L | 108.5 | 99.9 | 99.6 | 608771 |
| MMP21 | 94.9 | 100.0 | 98.0 | 608416 |
| MYH11 | 122.1 | 100.0 | 99.5 | 160745 |
| MYH6 | 95.5 | 99.0 | 95.3 | 160710 |
| MYH7 | 92.2 | 99.5 | 96.4 | 160760 |
| NKX2-5 | 120.8 | 100.0 | 99.9 | 600584 |
| NKX2-6 | 139.9 | 100.0 | 100.0 | 611770 |
| NODAL | 144.8 | 100.0 | 100.0 | 601265 |
| NOTCH1 | 141.8 | 99.8 | 98.9 | 190198 |
| NOTCH2 | 123.7 | 100.0 | 99.6 | 610205 |
| NR2F2 | 236.6 | 100.0 | 100.0 | 107773 |
| PKD1L1 | 108.7 | 100.0 | 99.3 | 609721 |
| PLD1 | 116.4 | 99.9 | 99.3 | 602382 |
| PTPN11 | 78.3 | 98.6 | 90.7 | 176876 |
| RAF1 | 108.3 | 100.0 | 99.9 | 164760 |
| SHROOM3 | 151.4 | 99.9 | 99.1 | 604570 |
| SMAD6 | 180.5 | 98.8 | 89.1 | 602931 |
| SOS1 | 102.0 | 99.6 | 97.4 | 182530 |
| TAB2 | 170.6 | 99.9 | 99.5 | 605101 |
| TBX1 | 101.2 | 93.0 | 86.9 | 602054 |
| TBX20 | 108.2 | 100.0 | 99.9 | 606061 |
| TBX5 | 135.3 | 100.0 | 100.0 | 601620 |
| TDGF1 | 120.9 | 99.7 | 94.8 | 187395 |
| TFAP2B | 168.3 | 99.2 | 96.8 | 601601 |
| TLL1 | 129.8 | 100.0 | 99.9 | 606742 |

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|--------|-------|-------|------|--------|
| TNNI3K | 105.8 | 99.9 | 99.3 | 613932 |
| ZFPM2 | 155.6 | 100.0 | 99.8 | 603693 |
| ZIC3 | 140.9 | 100.0 | 99.8 | 300265 |

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.

Median Coverage describes the average number of reads seen across 50 exomes.

% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.

% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors