

Exome

Gene panel Congenital heart defects (139 genes)

version 6, 03-02-2023

HGNC approved gene symbol	transcript	median depth	% covered >10x	% covered >20x	% covered >30x	% covered >50x
<i>ABCC9</i>	NM_005691.3	111.00	100.00	99.88	98.95	96.04
<i>ACTB</i>	NM_001101.5	154.00	100.00	100.00	99.80	99.50
<i>ACTC1</i>	NM_005159.5	163.00	100.00	99.99	99.29	94.86
<i>ACVR2B</i>	NM_001106.4	114.00	99.24	97.09	96.68	96.41
<i>ADNP</i>	NM_001282531.3	337.00	100.00	100.00	100.00	100.00
<i>ALDH1A2</i>	NM_003888	111.00	99.92	97.30	93.12	90.20
<i>AMER1</i>	NM_152424.4	147.00	100.00	100.00	99.97	99.93
<i>ANKRD11</i>	NM_013275.6	157.00	99.87	97.98	96.35	94.10
<i>ARID1A</i>	NM_006015.6	124.00	97.27	93.42	90.87	87.11
<i>ARID1B</i>	NM_001346813.1	112.00	98.71	93.97	88.34	81.37
<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>B3GAT3</i>	NM_012200.4	82.00	100.00	99.77	98.87	83.26
<i>B3GLCT</i>	NM_194318	90.00	99.93	97.95	95.72	94.25
<i>BCOR</i>	NM_001123385.2	98.00	99.97	99.70	98.65	94.11
<i>BRAF</i>	NM_004333.6	105.00	99.99	99.70	99.25	95.30
<i>CBL</i>	NM_005188.4	121.00	100.00	100.00	99.98	99.80
<i>CCBE1</i>	NM_133459.4	103.00	100.00	100.00	99.86	97.09
<i>CCDC103</i>	NM_213607.3	113.00	100.00	99.97	99.82	98.96
<i>CCDC11</i>	NM_032357.4	73.00	100.00	99.64	96.57	89.92
<i>CCDC114</i>	NM_001364171.2	78.00	96.66	96.01	93.26	85.78
<i>CCDC151</i>	NM_145045.5	84.00	99.94	99.45	97.50	89.55
<i>CCDC39</i>	NM_181426.2	103.00	100.00	99.72	98.35	92.32
<i>CCDC40</i>	NM_017950.4	86.00	99.88	97.50	93.74	86.65
<i>CD96</i>	NM_005816.5	124.00	100.00	100.00	100.00	100.00
<i>CDK13</i>	NM_003718.5	119.00	99.94	95.85	88.84	83.43
<i>CFC1</i>	NM_032545.3	2.00	34.48	33.19	31.54	29.08
<i>CHD4</i>	NM_001273.5	121.00	100.00	99.96	99.40	95.61
<i>CHD7</i>	NM_017780.4	143.00	100.00	99.95	99.52	98.44
<i>CITED2</i>	NM_006079.5	67.00	100.00	99.18	95.26	86.46
<i>CREBBP</i>	NM_004380.3	105.00	99.60	98.03	94.76	86.79
<i>CRELD1</i>	NM_015513.5	72.00	99.04	91.56	81.43	67.15
<i>DCHS1</i>	NM_003737.4	119.00	100.00	99.99	99.91	99.16
<i>DDX3X</i>	NM_001356.4	112.00	100.00	100.00	99.55	93.46
<i>DHCR7</i>	NM_001360.2	89.00	99.98	99.76	98.37	94.75
<i>DNAAF3</i>	NM_001256715.2	70.00	99.97	97.88	90.82	71.64
<i>DNAAF4</i>	NM_130810.4	97.00	100.00	98.30	94.17	86.99
<i>DNAH11</i>	NM_001277115	113.00	100.00	100.00	99.81	98.91
<i>DYRK1A</i>	NM_001347721.2	147.00	100.00	99.96	99.87	99.03
<i>EFTUD2</i>	NM_004247.4	91.00	100.00	100.00	99.58	93.95
<i>EHMT1</i>	NM_024757.5	91.00	99.40	98.27	96.69	90.37
<i>ELN</i>	NM_000501.4	52.00	99.53	94.54	82.85	53.41

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<i>EMG1</i>	NM_006331.8	147.00	100.00	100.00	99.95	99.45
<i>EP300</i>	NM_001429.4	137.00	100.00	99.79	98.41	94.86
<i>EVC</i>	NM_153717.3	87.00	94.33	93.34	90.84	81.90
<i>EVC2</i>	NM_147127.5	107.00	100.00	99.82	98.45	95.67
<i>FGFR2</i>	NM_022970.3	103.00	100.00	100.00	99.86	99.06
<i>FLNA</i>	NM_001110556.2	83.00	99.51	98.02	94.58	84.51
<i>FLNB</i>	NM_001457.4	111.00	100.00	99.95	99.52	96.92
<i>FLT4</i>	NM_182925.5	89.00	99.71	98.36	95.86	88.96
<i>FOXC1</i>	NM_001453.3	82.00	94.51	89.16	83.11	74.83
<i>FOXC2</i>	NM_005251.3	102.00	99.92	98.40	93.34	83.10
<i>FOXF1</i>	NM_001451.3	139.00	100.00	99.99	99.12	92.83
<i>FOXH1</i>	NM_003923.3	104.00	100.00	100.00	99.97	97.03
<i>FTO</i>	NM_001080432.3	163.00	100.00	100.00	100.00	98.69
<i>GALNS</i>	NM_000512.5	73.00	100.00	98.54	91.29	76.06
<i>GATA4</i>	NM_002052.5	66.00	98.10	85.73	74.64	63.70
<i>GATA5</i>	NM_080473.5	59.00	99.78	97.78	90.19	66.82
<i>GATA6</i>	NM_005257.5	103.00	96.82	92.37	88.25	83.10
<i>GDF1</i>	NM_001492.6	52.00	90.97	75.21	65.18	53.13
<i>GJA1</i>	NM_000165.5	161.00	100.00	100.00	100.00	100.00
<i>GLI3</i>	NM_000168.6	132.00	100.00	100.00	99.80	96.33
<i>GPC3</i>	NM_004484.4	109.00	99.86	98.68	95.69	90.65
<i>HAND1</i>	NM_004821.3	109.00	99.55	92.76	82.72	75.47
<i>HAND2</i>	NM_021973.3	65.00	100.00	99.79	98.39	75.57
<i>HCCS</i>	NM_005333.5	79.00	100.00	100.00	99.26	93.21
<i>HEY2</i>	NM_012259.3	159.00	100.00	100.00	100.00	99.72
<i>HRAS</i>	NM_005343.4	135.00	100.00	100.00	100.00	100.00
<i>JAG1</i>	NM_000214.3	130.00	99.51	98.00	97.73	96.77
<i>KANSL1</i>	NM_015443.3	122.00	99.97	99.61	98.08	92.98
<i>KAT6B</i>	NM_012330.4	162.00	99.99	99.71	98.51	92.94
<i>KDM6A</i>	NM_001291415.1	93.00	100.00	99.60	97.93	88.13
<i>KMT2A</i>	NM_001197104.1	159.00	99.73	99.18	98.38	96.31
<i>KMT2D</i>	NM_003482.3	128.00	100.00	99.76	98.28	92.89
<i>KRAS</i>	NM_004985.5	120.00	100.00	99.32	98.31	88.38
<i>LEFTY2</i>	NM_003240.5	144.00	100.00	99.84	99.07	97.41
<i>MAP2K1</i>	NM_002755.3	94.00	100.00	99.91	99.14	97.26
<i>MAP2K2</i>	NM_030662.3	94.00	100.00	99.09	93.83	79.82
<i>MATR3</i>	NM_199189.3	107.00	100.00	99.96	99.87	97.55
<i>MED12</i>	NM_005120.3	82.00	99.94	98.45	94.27	86.12
<i>MED13L</i>	NM_015335.4	116.00	99.97	99.79	98.57	95.67
<i>MEGF8</i>	NM_001271938.2	79.00	99.78	96.97	90.98	78.59
<i>MGP</i>	NM_000900.5	101.00	100.00	98.90	92.83	84.96
<i>MID1</i>	NM_000381.4	98.00	100.00	99.82	97.87	92.69
<i>MKKS</i>	NM_170784.2	581.00	100.00	100.00	100.00	100.00
<i>MMP21</i>	NM_147191.1	118.00	99.66	95.92	90.46	80.47
<i>MYH11</i>	NM_002474.3	101.00	100.00	99.66	98.19	92.66
<i>MYH6</i>	NM_002471.3	117.00	100.00	99.73	98.13	94.16

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<i>MYH7</i>	NM_000257.4	125.00	100.00	99.78	97.96	91.85
<i>NIPBL</i>	NM_133433.4	115.00	100.00	99.96	99.71	98.33
<i>NKX2-5</i>	NM_004387.4	101.00	100.00	99.00	91.43	70.88
<i>NKX2-6</i>	NM_001136271.2	109.00	100.00	100.00	100.00	99.96
<i>NODAL</i>	NM_018055.5	106.00	100.00	99.98	99.88	98.84
<i>NOTCH1</i>	NM_017617.5	96.00	98.99	97.62	94.26	85.40
<i>NOTCH2</i>	NM_024408.4	113.00	99.79	98.82	97.44	95.01
<i>NPHP3</i>	NM_153240.5	103.00	100.00	99.98	99.66	97.00
<i>NR2F2</i>	NM_021005.4	154.00	100.00	98.97	96.03	93.28
<i>NRAS</i>	NM_002524.5	132.00	100.00	100.00	100.00	99.96
<i>NSD1</i>	NM_022455.4	160.00	100.00	99.96	99.55	98.36
<i>PACS1</i>	NM_018026.4	90.00	98.63	95.32	91.95	86.70
<i>PITX2</i>	NM_000325.6	107.00	99.81	96.63	91.73	85.63
<i>PKD1L1</i>	NM_138295	99.00	99.98	99.60	97.84	91.53
<i>PRDM6</i>	NM_001136239	96.00	99.79	93.49	80.75	62.81
<i>PRKD1</i>	NM_002742.3	117.00	99.62	98.26	97.14	94.31
<i>PTPN11</i>	NM_002834.4	137.00	99.19	99.17	99.17	99.17
<i>RAB23</i>	NM_016277.5	106.00	100.00	100.00	100.00	99.99
<i>RAF1</i>	NM_001354689.3	109.00	100.00	99.97	99.24	95.23
<i>RAI1</i>	NM_030665.4	183.00	100.00	99.98	99.36	97.99
<i>RBM8A</i>	NM_005105.4	102.00	100.00	100.00	99.84	99.08
<i>RIT1</i>	NM_006912.6	208.00	100.00	100.00	100.00	100.00
<i>ROR2</i>	NM_004560.4	105.00	100.00	99.84	98.47	94.86
<i>RRAS</i>	NM_006270.5	81.00	99.92	97.91	90.66	82.00
<i>SALL1</i>	NM_002968.3	159.00	100.00	99.99	99.52	98.13
<i>SALL4</i>	NM_020436.5	136.00	99.85	97.97	96.28	95.83
<i>SF3B4</i>	NM_005850.5	123.00	100.00	97.65	90.47	77.51
<i>SH3PXD2B</i>	NM_001017995.3	109.00	99.93	98.53	95.46	87.65
<i>SHOC2</i>	NM_007373.3	113.00	100.00	100.00	99.92	99.77
<i>SMAD6</i>	NM_005585.5	33.00	93.35	67.37	53.09	43.72
<i>SMARCA2</i>	NM_003070.5	95.00	98.60	97.70	97.20	95.00
<i>SMARCA4</i>	NM_001128849.2	92.00	100.00	99.90	98.89	92.48
<i>SMARCB1</i>	NM_003073.5	102.00	100.00	100.00	99.68	98.57
<i>SMARCE1</i>	NM_003079.5	147.00	100.00	100.00	99.97	99.72
<i>SMC1A</i>	NM_006306.4	90.00	99.96	99.41	98.06	92.98
<i>SOS1</i>	NM_005633.3	114.00	100.00	99.63	98.47	97.25
<i>STRA6</i>	NM_022369.4	73.00	100.00	99.87	97.54	82.20
<i>TAB2</i>	NM_001292034.3	269.00	100.00	100.00	99.98	99.93
<i>TBX1</i>	NM_080647.1	45.00	88.72	77.49	65.14	46.35
<i>TBX20</i>	NM_001077653.2	90.00	100.00	100.00	99.45	94.94
<i>TBX3</i>	NM_005996.4	63.00	99.94	98.12	92.36	69.52
<i>TBX5</i>	NM_000192.3	117.00	100.00	99.94	99.21	94.35
<i>TDGF1</i>	NM_003212	140.00	100.00	100.00	100.00	100.00
<i>TFAP2B</i>	NM_003221.4	110.00	99.98	99.34	97.57	93.97
<i>TLL1</i>	NM_012464.5	103.00	100.00	100.00	99.77	97.00
<i>TWIST1</i>	NM_000474.4	71.00	95.32	84.83	77.64	71.43

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<i>UBR1</i>	NM_174916.3	103.00	100.00	99.98	99.81	97.40
<i>VPS13B</i>	NM_152564.4	119.00	100.00	99.71	98.81	97.33
<i>ZEB2</i>	NM_014795.4	609.00	100.00	100.00	99.99	99.95
<i>ZFPM2</i>	NM_012082.4	222.00	100.00	99.46	98.03	93.62
<i>ZIC3</i>	NM_003413.4	79.00	100.00	99.54	96.37	82.57