

Exome

Gene panel Muscle disorders (195 genes)

version 5, 28-03-2024

HGNC approved gene symbol	transcript	median depth	% covered >10x	% covered >20x	% covered >30x	% covered >50x
ACADVL	NM_000018.4	108.00	99.99	98.99	94.62	87.21
ACTA1	NM_001100.4	126.00	100.00	99.94	99.48	99.06
ACTN2	NM_001103.3	94.00	100.00	99.89	98.97	92.26
ACVR1	NM_001111067.4	116.00	100.00	100.00	99.95	99.63
ADSSL1	NM_152328	87.00	91.26	86.21	86.01	83.65
AGL	NM_000642.3	126.00	100.00	99.99	99.64	97.93
AGRN	NM_198576.4	90.00	98.68	95.39	90.02	80.73
ANO5	NM_213599.2	112.00	99.99	99.62	97.81	92.48
ATP2A1	NM_004320.5	96.00	99.97	99.17	97.51	92.53
B3GALNT2	NM_152490.5	111.00	100.00	99.98	99.27	95.13
B4GAT1	NM_006577	205.50	100.00	100.00	100.00	100.00
BAG3	NM_004281.3	93.00	100.00	99.51	98.01	96.56
BICD2	NM_001003800.2	104.00	100.00	99.99	99.68	98.42
BIN1	NM_139343.3	63.00	99.78	97.12	90.70	69.99
BVES	NM_001199563.2	104.00	100.00	100.00	99.87	99.62
CACNA1H	NM_021098.3	88.00	99.52	97.18	92.96	82.36
CACNA1S	NM_000069.3	92.00	100.00	99.98	99.32	93.08
CAPN3	NM_000070.3	94.00	100.00	100.00	100.00	99.03
CASQ1	NM_001231.5	79.00	100.00	99.94	99.03	96.41
CAV3	NM_033337.3	114.00	100.00	100.00	100.00	99.87
CAVIN1	NM_012232	114.00	100.00	100.00	99.73	98.21
CCDC78	NM_001031737.3	83.00	99.47	96.38	91.09	79.10
CFL2	NM_138638.5	154.00	100.00	99.60	96.57	88.79
CHAT	NM_001142929.1	93.00	100.00	99.29	96.67	87.67
CHCHD10	NM_213720.3	55.00	95.97	86.50	76.91	57.35
CHKB	NM_005198.4	143.00	100.00	99.99	99.44	96.39
CHRNA1	NM_000079.4	90.00	99.95	99.27	95.68	89.22
CHRNA1	NM_000747.3	88.00	99.95	99.20	96.58	88.53
CHRND	NM_000751.3	80.00	99.54	97.05	92.78	82.50
CHRNE	NM_000080.4	105.00	100.00	99.99	99.87	98.46
CLCN1	NM_000083.3	86.00	99.98	98.68	94.27	85.81
CLN3	NM_001042432.1	91.00	99.67	96.29	91.03	87.06
CNTN1	NM_001843.4	101.00	100.00	100.00	99.97	99.62
COL12A1	NM_004370.6	113.00	100.00	99.94	99.38	96.72
COL13A1	NM_001320951.2	71.00	99.99	99.25	96.17	81.37
COL6A1	NM_001848.3	103.00	99.35	97.65	95.02	84.09
COL6A2	NM_001849.4	115.00	99.99	99.56	97.95	90.28
COL6A3	NM_004369.4	169.00	100.00	99.92	99.66	99.00
COLQ	NM_005677.4	76.00	100.00	99.48	95.78	79.55

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<i>CPT2</i>	NM_000098.3	200.00	100.00	99.94	99.55	99.14
<i>CRPPA</i>	NM_001101426	98.00	99.98	98.81	95.92	90.12
<i>CRYAB</i>	NM_001289808.2	120.00	100.00	100.00	100.00	99.88
<i>DAG1</i>	NM_004393.6	182.00	100.00	99.75	98.79	96.99
<i>DES</i>	NM_001927.4	79.00	100.00	99.78	98.41	91.32
<i>DMD</i>	NM_004006.2	93.00	99.91	99.43	98.09	93.54
<i>DNA2</i>	NM_001080449.3	123.00	100.00	99.99	99.55	97.89
<i>DNAJB4</i>	NM_007034.5	146.00	100.00	100.00	100.00	100.00
<i>DNAJB6</i>	NM_058246.4	100.00	100.00	99.96	96.03	76.92
<i>DNM2</i>	NM_001005361.2	85.00	99.95	99.65	97.29	87.62
<i>DNMT3B</i>	NM_006892.4	98.00	100.00	99.64	97.73	91.43
<i>DOK7</i>	NM_173660.5	83.00	93.64	93.28	92.79	86.82
<i>DPAGT1</i>	NM_001382.4	98.00	100.00	100.00	99.61	95.29
<i>DPM1</i>	NM_003859.2	103.00	100.00	100.00	100.00	99.91
<i>DPM2</i>	NM_003863.3	75.00	100.00	99.92	95.28	83.45
<i>DPM3</i>	NM_153741.2	70.00	100.00	100.00	99.97	98.95
<i>DUX4</i>	NM_001306068	0.00	0.00	0.00	0.00	0.00
<i>DYNC1H1</i>	NM_001376.5	136.00	99.99	99.79	98.99	96.91
<i>DYSF</i>	NM_001130987.2	90.00	99.97	99.28	97.22	90.22
<i>ECEL1</i>	NM_004826.4	92.00	100.00	99.88	98.02	87.73
<i>EMD</i>	NM_000117.3	91.00	97.73	89.90	79.49	65.13
<i>ENO3</i>	NM_053013.4	138.00	100.00	100.00	99.69	93.15
<i>ERBB3</i>	NM_001982.3	99.00	99.99	99.28	98.19	96.08
<i>EXOSC8</i>	NM_181503.3	76.00	100.00	99.47	95.94	77.68
<i>FAM111B</i>	NM_198947.4	295.00	100.00	100.00	100.00	100.00
<i>FHL1</i>	NM_001159699.2	95.00	100.00	99.84	98.85	96.37
<i>FKRP</i>	NM_024301.5	104.00	100.00	100.00	99.52	93.81
<i>FKTN</i>	NM_001079802.1	101.00	100.00	100.00	99.91	97.99
<i>FLNC</i>	NM_001458.4	103.00	100.00	99.72	98.18	93.55
<i>FXR1</i>	NM_005087.4	89.00	100.00	99.87	98.89	95.14
<i>GAA</i>	NM_000152.5	82.00	99.99	99.75	97.91	89.63
<i>GBE1</i>	NM_000158.4	110.00	100.00	99.99	99.84	98.06
<i>GFPT1</i>	NM_001244710.1	110.00	100.00	100.00	99.99	99.79
<i>GMPPB</i>	NM_021971.4	147.00	100.00	100.00	99.97	99.90
<i>GNE</i>	NM_005476.7	117.00	100.00	100.00	99.98	99.95
<i>GOLGA2</i>	NM_001366244.1	92.00	99.86	97.37	93.63	88.25
<i>GOSR2</i>	NM_004287.4	124.00	100.00	99.32	97.19	95.42
<i>GYG1</i>	NM_001184720.1	174.00	100.00	100.00	99.98	99.67
<i>GYS1</i>	NM_002103.5	82.00	100.00	100.00	99.80	93.92
<i>HACD1</i>	NM_014241	82.00	94.44	81.65	72.30	70.47
<i>HMGCS1</i>	NM_001098272.3					
<i>HNRNPA1</i>	NM_031157.4	249.00	100.00	100.00	100.00	100.00
<i>HNRNPA2B1</i>	NM_002137.4	116.00	100.00	100.00	100.00	99.84

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<i>HNRNPDL</i>	NM_031372.3	103.00	99.97	97.22	90.21	84.28
<i>HRAS</i>	NM_005343.4	135.00	100.00	100.00	100.00	100.00
<i>HSPB8</i>	NM_014365.2	75.00	100.00	99.99	98.27	88.66
<i>HSPG2</i>	NM_005529.7	80.00	99.42	98.10	94.35	84.23
<i>IGHMBP2</i>	NM_002180.2	108.00	100.00	99.79	98.11	93.08
<i>INPP5K</i>	NM_016532.4	91.00	100.00	100.00	99.44	95.39
<i>ISCU</i>	NM_213595.3	97.00	100.00	100.00	99.61	93.86
<i>ITGA7</i>	NM_001144996.2	90.00	99.98	99.20	95.77	86.69
<i>JAG2</i>	NM_002226.5					
<i>KBTBD13</i>	NM_001101362.2	152.00	100.00	99.96	99.62	98.74
<i>KCNJ2</i>	NM_000891.3	290.00	100.00	100.00	100.00	100.00
<i>KLHL40</i>	NM_152393.4	79.00	100.00	99.94	98.52	93.50
<i>KLHL41</i>	NM_006063.3	220.00	100.00	100.00	100.00	100.00
<i>KLHL9</i>	NM_018847.4	324.00	100.00	100.00	100.00	100.00
<i>KY</i>	NM_178554.6	124.00	100.00	100.00	100.00	99.18
<i>LAMA2</i>	NM_000426.3	108.00	100.00	99.99	99.72	98.00
<i>LAMP2</i>	NM_002294.3	80.00	100.00	99.41	97.44	87.79
<i>LARGE1</i>	NM_133642	92.00	100.00	99.81	98.57	94.49
<i>LDB3</i>	NM_007078.3	92.00	99.92	99.13	95.89	86.14
<i>LDHA</i>	NM_005566.4	162.00	100.00	100.00	100.00	100.00
<i>LIMS2</i>	NM_001161403.2	90.00	99.97	99.29	98.41	93.06
<i>LMNA</i>	NM_170707.4	57.00	100.00	98.85	90.66	64.94
<i>LMOD3</i>	NM_198271.4	208.00	100.00	99.47	97.91	96.00
<i>LPIN1</i>	NM_001349206.2	97.00	99.97	99.26	97.96	95.19
<i>LRIF1</i>	NM_018372.4	278.00	100.00	100.00	100.00	100.00
<i>LRP12</i>	NM_013437.4	249.00	100.00	100.00	100.00	99.99
<i>MAP3K20</i>	NM_016653.3	121.00	100.00	100.00	99.71	97.24
<i>MATR3</i>	NM_018834.6	107.00	100.00	99.96	99.87	97.55
<i>MB</i>	NM_005368.3	114.00	100.00	100.00	100.00	99.36
<i>MEGF10</i>	NM_001256545.2	107.00	100.00	100.00	99.97	99.38
<i>MICU1</i>	NM_001195518.2	108.00	100.00	100.00	99.92	98.89
<i>MLIP</i>	NM_001281747.2					
<i>MSTN</i>	NM_005259.3	179.00	100.00	100.00	100.00	100.00
<i>MSTO1</i>	NM_018116.3	50.50	88.38	74.18	63.27	50.35
<i>MTM1</i>	NM_000252.2	77.00	100.00	99.89	98.80	93.14
<i>MUSK</i>	NM_005592.4	107.00	100.00	100.00	99.90	99.05
<i>MYBPC3</i>	NM_000256.3	102.00	99.75	98.33	96.61	93.05
<i>MYF6</i>	NM_002469.3	100.00	99.97	98.60	95.32	92.34
<i>MYH2</i>	NM_017534.6	212.00	100.00	100.00	99.90	98.41
<i>MYH3</i>	NM_002470.4	140.00	99.98	99.40	98.18	96.10
<i>MYH7</i>	NM_000257.4	125.00	100.00	99.78	97.96	91.85
<i>MYL1</i>	NM_079420.3	99.00	100.00	100.00	100.00	98.89
<i>MYL2</i>	NM_000432.3	86.00	100.00	100.00	100.00	99.69

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<i>MYMK</i>	NM_001080483	95.00	100.00	100.00	98.78	89.69
<i>MYO18B</i>	NM_032608.7	81.00	100.00	99.25	96.49	87.13
<i>MYOT</i>	NM_006790.3	96.00	100.00	100.00	99.93	98.16
<i>MYPN</i>	NM_032578.3	126.00	100.00	99.99	99.75	98.30
<i>NEB</i>	NM_001271208.2	132.00	89.12	87.74	87.02	86.02
<i>ORAI1</i>	NM_032790.3	171.00	98.41	94.73	92.06	88.15
<i>PABPN1</i>	NM_004643.3	68.00	67.92	64.86	60.54	56.37
<i>PAX7</i>	NM_001135254.2	82.00	99.50	95.23	85.32	69.36
<i>PFKM</i>	NM_000289.6	104.00	100.00	100.00	99.88	98.32
<i>PGAM2</i>	NM_000290.4	128.00	100.00	99.29	95.98	83.51
<i>PGK1</i>	NM_000291.4	95.00	100.00	99.65	97.73	90.81
<i>PGM1</i>	NM_002633.3	95.00	100.00	99.66	97.87	92.01
<i>PHKA1</i>	NM_002637.4	84.00	100.00	99.64	97.70	90.39
<i>PIP5K1C</i>	NM_012398.3	77.00	99.14	92.96	85.58	75.89
<i>PLEC</i>	NM_201380.4	110.00	99.98	99.61	98.00	92.75
<i>PNPLA2</i>	NM_020376.4	81.00	99.98	98.19	89.34	76.52
<i>POGLUT1</i>	NM_152305.3	105.00	100.00	100.00	100.00	99.06
<i>POMGNT1</i>	NM_017739.3	97.00	100.00	100.00	99.96	99.66
<i>POMGNT2</i>	NM_032806.6	170.00	100.00	100.00	100.00	99.97
<i>POMK</i>	NM_032237.5	225.00	100.00	100.00	100.00	100.00
<i>POMT1</i>	NM_001077365.2	90.00	100.00	99.59	98.18	91.78
<i>POMT2</i>	NM_013382.5	89.00	100.00	99.99	99.79	96.46
<i>POPDC3</i>	NM_022361.5					
<i>PYGM</i>	NM_005609.4	100.00	100.00	99.71	97.66	92.23
<i>PYROXD1</i>	NM_024854.5	104.00	99.35	95.15	87.72	77.26
<i>RAPSN</i>	NM_005055.5	122.00	100.00	100.00	99.69	94.80
<i>RBCK1</i>	NM_031229.4	66.00	99.65	96.62	91.36	80.06
<i>RXYLT1</i>	NM_014254	113.00	100.00	99.65	97.16	90.13
<i>RYR1</i>	NM_000540.3	92.00	98.83	97.88	95.27	87.63
<i>RYR3</i>	NM_001036.5	106.00	99.92	99.53	98.89	96.14
<i>SCN4A</i>	NM_000334.4	118.00	99.99	99.72	98.64	96.40
<i>SELENON</i>	NM_020451	71.00	89.70	89.21	84.74	67.28
<i>SGCA</i>	NM_000023.4	77.00	100.00	99.72	98.39	93.21
<i>SGCB</i>	NM_000232.4	126.00	99.70	96.92	96.44	96.10
<i>SGCD</i>	NM_000337.5	100.00	100.00	100.00	99.89	98.24
<i>SGCG</i>	NM_000231.2	115.00	100.00	100.00	99.99	98.86
<i>SLC52A2</i>	NM_001363118.2	94.00	100.00	99.81	98.56	93.23
<i>SLC52A3</i>	NM_033409.4	88.00	100.00	99.90	98.79	93.47
<i>SMCHD1</i>	NM_015295.2	103.00	99.95	99.52	98.41	96.83
<i>SMPX</i>	NM_014332.3	66.00	100.00	100.00	99.54	82.71
<i>SPEG</i>	NM_005876.5	78.00	99.51	96.62	90.67	77.67
<i>SPTBN4</i>	NM_020971.3	66.00	97.14	92.54	83.75	69.04
<i>SQSTM1</i>	NM_003900.5	105.00	100.00	99.96	99.51	98.66

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<i>STAC3</i>	NM_145064.3	119.00	99.99	98.86	95.53	88.92
<i>STIM1</i>	NM_003156.3	108.00	100.00	99.98	99.61	96.79
<i>SYNE1</i>	NM_182961.4	122.00	100.00	99.81	99.32	97.98
<i>SYNE2</i>	NM_182914.2	100.00	100.00	99.90	99.21	96.52
<i>TCAP</i>	NM_003673.4	86.00	100.00	99.96	98.49	92.07
<i>TIA1</i>	NM_022173.4	115.00	100.00	100.00	99.90	98.85
<i>TMEM126B</i>	NM_001193538.3	107.00	100.00	100.00	100.00	99.84
<i>TMEM43</i>	NM_024334.2	89.00	100.00	100.00	99.77	97.64
<i>TNNI2</i>	NM_003282.4	104.00	99.98	99.02	97.36	93.41
<i>TNNT1</i>	NM_003283.6	72.00	99.53	95.20	87.19	72.98
<i>TNPO3</i>	NM_012470.3	106.00	100.00	99.95	99.71	98.89
<i>TOR1AIP1</i>	NM_015602.3	182.00	100.00	100.00	99.90	96.38
<i>TPM2</i>	NM_213674.1	70.00	99.97	98.19	94.18	85.00
<i>TPM3</i>	NM_152263.4	119.00	99.98	99.98	99.77	99.28
<i>TRAPPC11</i>	NM_021942.6	113.00	100.00	99.99	99.41	97.12
<i>TRIM32</i>	NM_012210.3	169.00	100.00	100.00	100.00	100.00
<i>TRIM54</i>	NM_187841.3	83.00	100.00	99.84	98.76	94.61
<i>TRIM63</i>	NM_032588.3	96.00	100.00	99.97	99.61	98.66
<i>TRIP4</i>	NM_016213.5	96.00	100.00	99.80	98.37	94.74
<i>TRPV4</i>	NM_021625.5	100.00	100.00	99.90	98.48	92.60
<i>TTC19</i>	NM_017775.4	96.00	97.21	89.48	85.66	82.94
<i>TTN</i>	NM_001267550.2	284.00	99.95	99.76	99.54	99.10
<i>UBA1</i>	NM_003334.4	86.00	99.89	97.47	91.05	77.91
<i>VCP</i>	NM_007126.5	125.00	100.00	99.87	99.56	99.28
<i>VIPAS39</i>	NM_001193315.1	98.00	100.00	100.00	99.99	99.85
<i>VMA21</i>	NM_001017980.3	74.00	100.00	100.00	99.97	91.65
<i>VRK1</i>	NM_003384.3	99.00	100.00	100.00	100.00	97.04