

Exome

Gene panel Congenitale Myastenia Gravis (32 genes)

version 1, 15-01-2020

HGNC approved gene symbol	transcript	median depth	% covered >10x	% covered >20x	% covered >30x	% covered >50x
<i>SYT2</i>	NM_177402.5	102.00	100.00	99.94	99.32	94.22
<i>AGRN</i>	NM_198576.4	90.00	98.68	95.39	90.02	80.73
<i>ALG14</i>	NM_144988.4	118.00	100.00	100.00	100.00	100.00
<i>PREPL</i>	NM_001171613.1	111.00	100.00	100.00	100.00	99.89
<i>GFPT1</i>	NM_001244710.1	110.00	100.00	100.00	99.99	99.79
<i>SLC5A7</i>	NM_021815.5	132.00	100.00	100.00	99.98	99.91
<i>CHRNA1</i>	NM_000079.4	90.00	99.95	99.27	95.68	89.22
<i>CHRND</i>	NM_000751.3	80.00	99.54	97.05	92.78	82.50
<i>CHRNG</i>	NM_005199.5	74.00	99.92	98.46	92.84	76.46
<i>COLQ</i>	NM_005677.4	76.00	100.00	99.48	95.78	79.55
<i>LAMB2</i>	NM_002292.4	140.00	100.00	99.91	99.26	97.86
<i>GMPPB</i>	NM_021971.4	147.00	100.00	100.00	99.97	99.90
<i>DOK7</i>	NM_173660.5	83.00	93.64	93.28	92.79	86.82
<i>PLEC</i>	NM_201380.4	110.00	99.98	99.61	98.00	92.75
<i>MUSK</i>	NM_005592.4	107.00	100.00	100.00	99.90	99.05
<i>ALG2</i>	NM_033087.4	212.00	100.00	100.00	99.77	98.67
<i>CHAT</i>	NM_001142929.1	93.00	100.00	99.29	96.67	87.67
<i>COL13A1</i>	NM_001320951.2	71.00	99.99	99.25	96.17	81.37
<i>SLC18A3</i>	NM_003055.3	223.00	100.00	100.00	100.00	99.99
<i>LRP4</i>	NM_002334.4	111.00	100.00	99.97	99.16	94.95
<i>RAPSN</i>	NM_005055.5	122.00	100.00	100.00	99.69	94.80
<i>DPAGT1</i>	NM_001382.4	98.00	100.00	100.00	99.61	95.29
<i>RPH3A</i>	NM_001143854.2	74.00	99.98	99.12	96.42	85.72
<i>VAMP1</i>	NM_199245.3	104.00	100.00	100.00	100.00	100.00
<i>MYO9A</i>	NM_006901.4	127.00	100.00	100.00	99.99	99.73
<i>CHRNE</i>	NM_000080.4	105.00	100.00	99.99	99.87	98.46
<i>CHRNB1</i>	NM_000747.3	88.00	99.95	99.20	96.58	88.53
<i>SCN4A</i>	NM_000334.4	118.00	99.99	99.72	98.64	96.40
<i>UNC13A</i>	NM_001080421.2	90.00	99.95	99.47	97.10	90.35
<i>SNAP25</i>	NM_130811.3	64.00	100.00	100.00	99.40	85.09
<i>LAMA5</i>	NM_005560.6	73.00	99.35	95.84	90.23	77.43
<i>SLC25A1</i>	NM_005984.5	71.00	91.69	89.40	87.32	77.31