

Inherited Disease Panel  
Gene panel Muscle Disorders\*, version 1, 18-2-2018

HGNC approved gene symbol	Transcript	OMIM gene ID	Median depth	% covered >10x	% covered >20x	% covered >30x	% covered >50x
<i>ACTA1</i>	NM_0011100	102610	165	100	100	100	100
<i>ANO5**</i>	NM_001142649	608662	160	100	100	100	100
<i>CAPN3**</i>	NM_173088	114240	118	100	100	100	100
<i>CAV3**</i>	NM_001234	601253	159	100	100	100	100
<i>COL6A1</i>	NM_001848	120220	122	100	100	100	98
<i>COL6A2</i>	NM_058174	120240	114	100	100	100	97
<i>COL6A3</i>	NM_004369	120250	202	100	100	100	100
<i>COLQ</i>	NM_005677	603033	126	100	100	100	100
<i>DAG1</i>	NM_001177643	128239	292	100	100	100	100
<i>DES</i>	NM_001927	125660	143	100	100	100	100
<i>DMD**</i>	NM_004013	300377	119	100	100	100	100
<i>DNAJB6</i>	NM_005494	611332	137	100	100	100	100
<i>DOK7</i>	NM_001256896	610285	245	100	100	100	100
<i>DPM3</i>	NM_018973	605951	190	100	100	100	100
<i>DYSF**</i>	NM_001130976	603009	122	100	100	100	100
<i>EMD**</i>	NM_000117	300384	107	100	100	100	100
<i>FHL1</i>	NM_001159702	300163	97	100	100	100	99
<i>FKRP**</i>	NM_001039885	606596	187	100	100	100	100
<i>FKTN</i>	NM_001079802	607440	164	100	100	100	100
<i>FLNC</i>	NM_001127487	102565	129	100	100	100	100
<i>GAA</i>	NM_000152	606800	136	100	100	100	100
<i>GFPT1</i>	NM_001244710	138292	161	100	100	100	99
<i>GNE</i>	NM_001128227	603824	162	100	100	100	100
<i>ISPD</i>	NM_001101426	614631	148	100	100	100	99
<i>ITGA7</i>	NM_001144996	600536	125	100	100	100	99
<i>LAMA2</i>	NM_000426	156225	162	100	100	100	100
<i>LDB3</i>	NM_001080116	605906	116	100	100	100	100
<i>LMNA**</i>	NM_001282625	150330	105	100	100	100	100
<i>MICU1**</i>	NM_001195518	605084	***	100	100	100	100
<i>MYH7</i>	NM_000257	160760	163	100	100	100	100
<i>MYOT**</i>	NM_001135940	604103	192	100	100	100	100
<i>NEB</i>	NM_004543	161650	178	100	100	100	100
<i>PLEC</i>	NM_201380	601282	180	100	100	100	99
<i>POMGNT1</i>	NM_001243766	606822	135	100	100	100	100
<i>POMT1</i>	NM_001077365	607423	152	100	100	100	100
<i>POMT2</i>	NM_013382	607439	112	100	100	100	98
<i>RAPSN</i>	NM_005055	601592	109	100	100	100	100
<i>RYR1</i>	NM_000540	180901	109	100	99	99	98
<i>SEPN1</i>	NM_206926	NA	132	89	89	88	87
<i>SGCA**</i>	NM_000023	600119	131	100	100	100	100
<i>SGCB**</i>	NM_000232	600900	140	100	100	100	100
<i>SGCD**</i>	NM_000337	601411	132	100	100	100	100

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<i>SGCG</i> **	NM_000231	608896	127	100	100	100	100
<i>SMCHD1</i>	NM_015295	614982	***	100	100	100	100
<i>TCAP</i> **	NM_003673	604488	141	100	100	100	100
<i>TPM3</i>	NM_001043351	191030	175	100	100	100	100
<i>TRIM32</i> **	NM_001099679	602290	256	100	100	100	100
<i>TTN</i>	NM_133437	188840	333	100	100	100	100

\* Muscular dystrophies / myopathies

\*\* Core genes (if necessary additional Sanger sequencing will be performed for 100% coverage)

\*\*\*For these genes Sanger sequencing is performed

MLPA for *DMD* is included in the gene panel Muscle Disorders



