GENETHON
CURE THROUGH INNOVATION

Neuromuscular

CODE	PATHOLOGY	Omim	Individuals	Family	Affecteds
ADDM	MYOPATHY DISTAL	160500	9	5	6
AMC	ARTHROGRYPOSIS	108110	146	58	63
AMD	GLYCOGEN STORAGE DISEASE, POMPE DISEASE	232300	10	9	10
ASP	AMYOTROPHY SCAPULOPERONEAL	181400	26	15	16
BET	BETHLEM MYOPATHY	158810	175	63	96
BMD	MUSCULAR DYSTROPHY BECKER TYPE	300376	50	37	39
BRO	BRODY MYOPATHY	601003	11	8	10
BVVLS1	BROWN VIALETTO VAN LAERE SYNDROME	211530	3	1	1
CBM	DESMIN RELATED MYOPATHY	601419	19	6	7
CCD	CENTRAL CORE DISEASE	117000	581	135	230
CHAC	CHOREOACANTHOCYTOSIS	200150	1	1	1
CMT	CHARCOT MARIE TOOTH DISEASE	145900	816	451	520
CMT1	CHARCOT-MARIE-TOOTH DISEASE TYPE 1	118200	59	23	30
CMT1A	CHARCOT-MARIE-TOOTH, TYPE 1A	118220	18	10	12
CMT1C	CHARCOT-MARIE-TOOTH TYPE 1C	601098	6	4	4
CMT2	CHARCOT-MARIE-TOOTH DISEASE, AXONAL TYPE 2	118210	159	91	107
CMT2A2	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, AUTOSOMAL DOMINANT, TYPE 2A2A	609260	15	10	10
TRPV4	TRANSIENT RECEPTOR POTENTIAL CATION CHANNEL, SUBFAMILY V, MEMBER 4	605427	4	1	2
CMT4	CHARCOT-MARIE-TOOTH , TYPE 4	214400	59	27	37
CMT4B1	CHARCOT-MARIE-TOOTH TYPE 4B1	601382	3	1	1
CMTX	CHARCOT-MARIE-TOOTH , X-LINKED	302800	10	4	8
DBU	MUSCULAR DYSTROPHY SCLEROATONIC	254090	204	83	93
DD	DANON DISEASE	300257	9	7	7
DEJ	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS	145900	12	4	
DM1	DYSTROPHIA MYOTONICA, STEINERT DISEASE	160900	363	268	307
DM2	MYOTONIC DYSTROPHY 2	602668	94	69	77
DMD	MUSCULAR DYSTROPHY DUCHENNE TYPE	310200	251	212	211
DMNET	UNDEFINED MUSCULAR DYSTROPHY		183	115	127
EDMD	EMERY-DREIFUSS MUSCULAR DYSTROPHY	181350	297	107	141
FCMD	FUKUYAMA CONGENITAL MUSCULAR DYSTROPHY	253800	6	4	6
FOP	FIBRODYSPLASIA OSSIFICANS PROGRESSIVA	135100	21	10	11
FSHD	FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY	158900	763	307	402
GAN	GIANT AXONAL NEUROPATHY	256850	53	19	26
GSD	GLYCOGEN STORAGE DISEASE 2B	232400	8	8	8
GSDIII	GLYCOGEN STORAGE DISEASE III		42	41	42
GSDNET	UNDEFINED GLYCOGEN STORAGE DISEASE		12	7	7
HARD	WALKER-WARBURG SYNDROME	236670	6	3	3
HOKPP	HYPOKALEMIC PERIODIC PARALYSIS	170400	42	23	26
HSAN1A	NEUROPATHIE SENSITIVE AUTOSOMIQUE HEREDITAIRE	162400	13	3	5
HSAN1B	JROPATHIE SENSITIVE AUTOSOMIQUE HEREDITAIRE, TYPE	608088	38	20	29
HSAN2B	JROPATHIE SENSITIVE AUTOSOMIQUE HEREDITAIRE, TYPE	613119	1	1	1
HSN4	HEREDITARY SENSORY NEUROPATHY		19	14	14
HTM1	MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 1	145600	132	56	113
HTM2	MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 2	154275	133	36	4
HTM3	MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 3	154276	7	5	0
HTM4	MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 4	600467	9	7	0
HTM5	MALIGNANT HYPERTHERMIA SISCEPTIBILITY 5	601887	9	3	0
HTM6	MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 6	601888	610	202	127
IBM	INCLUSION BODY MYOSITIS	147421	148	82	92

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IIM	MYOPATHY, FAMILIAL IDIOPATHIC INFLAMMATORY	160750	2	1	1
LGMD	LIMB-GIRDLE MUSCULAR DYSTROPHY		1361	547	653
LGMD1C	MUSCULAR DYSTROPHY , LIMB-GIRDLE,TYPE 1C	607801	20	10	15
LGMD2A	MUSCULAR DYSTROPHY, LIMB-GIRDLE,TYPE 2A	253600	441	109	172
LGMD2B	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2B	253601	16	5	7
LGMD2C	MUSCULAR DYSTROPHY, LIMB-GIRDLE TYPE 2C	253700	170	51	91
LGMD2D	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2D	600119	3	3	3
LGMD2I	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2I	607155	40	18	19
LGMD2L	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2L	611307	6	3	4
LGMD2N	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2N	613158	4	2	2
MAC	MCARDLE DISEASE	232600	147	123	128
MCF	MUSCLE CRAMPS, FAMILIAL	158400	2	2	2
MCMU	MYOPATHY CENTRONUCLEAR	255200	264	81	118
MDC	MUSCULAR DYSTROPHY CONGENITAL	253800	1644	604	702
MDD	MYOADENYLATE DEAMINASE DEFICIENCY	615511	3	3	3
MEAX	MYOPATHY, X-LINKED, WITH EXCESSIVE AUTOPHAGY	310440	5	4	4
MEB	MUSCLE-EYE-BRAIN-DISEASE	253280	9	4	3
MERRF	MERRF SYNDROME	545000	1	1	1
MG	MYASTHENIA GRAVIS	254200	1467	664	663
MITO	MITOCHONDRIAL MYOPATHY	251900	285	227	239
MMF	MYOPATHY MYOFIBRILAR	609524	68	15	30
MNET	UNDEFINED MYOPATHY		2891	2022	1989
MSD	DESMIN-RELATED MYOPATHY	601419	118	37	55
MTD	MYOTONIA CONGENITA, THOMSEN DISEASE	160800	113	75	86
MTSD	MYOPATHY AND TRIGLYCERIDE STORAGE DISEASE		95	84	85
MTU	MYOTUBULAR MYOPATHY	310400	137	56	60
MULT	MINICORE MYOPATHY WITH EXTERNAL OPHTHALMOPLEGI/	255320	306	96	109
MYD	MYOPATHY DISTAL WITH ONSET IN INFANCY	160300	434	226	261
NAPB	NEURITIS WITH BRANCHIAL PREDILECTION	162100	2	2	2
NEM	NEMALINE MYOPATHY	161800	397	137	160
NM	NONAKA MYOPATHY	605820	4	4	4
NSM	NEUROPATHY MOTOR AND SENSORY UNDEFINED		70	52	55
NTO	TOMACULOUS NEUROPATHY	162500	11	5	3
OPMD	OCULOPHARYNGEAL MUSCULAR DYSTROPHY	164300	322	156	207
PFKM	PHOSPHOFRUCTOKINASE, MUSCLE TYPE	610681	4	4	4
PMDM	POLYMYOSITIS		2	2	2
RMD	RIPPLING MUSCLE DISEASE	606072	5	5	5
RSMD1	RIGID SPINE MUSCULAR DYSTROPHY	602771	57	24	24
SCN4	HYPERKALEMIC PERIODIC PARALYSIS	170500	4	4	4
SCN4A	MYOTONIA PERMANENS	608390	2	2	2
SFNP	NEUROPATHY, SMALL FIBER	133020	1	1	1
SGE	SECONDARY GENERALIZED EPILEPSY		6	3	4
SMA	SPINAL MUSCULAR ATROPHY	253300	681	334	387
SMAD	SPINAL MUSCULAR ATROPHY, JUVENILE, PROXIMAL, AUTOSOMAL DOMINANT	158600	20	10	11
SMC	MYASTHENIC SYNDROME CONGENITAL	601462	610	233	302
VCPDM	VOCAL CORD AND PHARYNGEAL DYSFUNCTION WITH DISTAL MYOPATHY, FORMERLY	606070	4	2	3
VLCAD	DEFICIENCY OF ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN,	201475	2	2	2