Name of Disease	ICD10_Identifier	ICD10_Name	MIM_Number	ORPHA_Number	N. samples Muscle tissue	N. samples DNA	N. samples Myoblasts	N. samples Fibroblasts
Duchenne dystrophy, DMD	G71.0	Muscular Dystrophy	310200	98896	182	214	68	41
Becker dystrophy, BMD	G71.0	Muscular Dystrophy	300376	98895	84	225	44	50
DMD/BMD carrier	G71.0	Muscular Dystrophy	310200	262	32	29	17	20
Relatives of distrophinopathic pts		Muscular Dystrophy			0	381	0	0
LGMD2A, calpainopathy	G71.0	Muscular Dystrophy	253600	267	0	0	2	2
LGMD2B, dysferlinopathy	G71.0	Muscular Dystrophy	253601	268	14	46	2	2
LGMD2C, gamma-sarcoglycanopathy	G71.0	Muscular Dystrophy	253700	353	14	69	2	2
LGMD2D, alpha-sarcoglycanopathy	G71.0	Muscular Dystrophy	608099	62	4	4	1	1
LGMD2E, beta-sarcoglycanopathy	G71.0	Muscular Dystrophy	604286	119	6	12	3	3
LGMD2F, delta-sarcoglycanopathy	G71.0	Muscular Dystrophy	601287	219	6	9	2	2
LGMD2I, FKRP	G71.0	Muscular Dystrophy	607155	34515	2	0	1	1
LGMD1B, LMNA	G71.0	Muscular Dystrophy	159001	264	4	8	2	2
LGMD1C, caveolinopathy	G71.0	Muscular Dystrophy	607801	265	37	63	4	4
Other LGMD	G71.0	Muscular Dystrophy			58	54	1	1
Facio-scapulo-humeral, FSHD1	G71.0	Muscular Dystrophy	158900	269	59	6	11	9
Merosinopathy, LAMA2	G71.2	Congenital Myopathies	607855	258	4	1	0	0
Congenital dystrophies	G71.2	Congenital Myopathies	606612	97242	26	17	9	9
Myotonic dystrophy, Steinert, DM1	G71.1	Myotonic Disorders	160900	273	56	73	7	6
Thomsen Becker disease CLCN1 channel	G71.1	Myotonic Disorders	160800	614	2	2	0	0
Selenoproteine (SEPN1)	G71.2	Congenital Myopathies	606210	209193	0	3	0	0
HypoKaliemic Periodic paralysis	G72.3	Periodic paralysis	170400	681	6	0	0	0
Oculo-pharyngeal	G71.0	Muscular Dystrophy	164300	270	13	4	5	4
Nemaline myopathy	G71.2	Congenital Myopathies	256030	607	3	1	2	2
Centralcore myopathy	G71.2	Congenital Myopathies	117000	597	7	1	3	3
Minicore myopathy	G71.2	Congenital Myopathies	255320	598	4	2	3	3
Centronuclear myopathy	G71.2	Congenital Myopathies	602378	595	2	1	0	0
Myotubular myopathy, MTMX	G71.2	Congenital Myopathies	310400	596	5	0	1	1
Fiber type disproportion	G71.2	Congenital Myopathies	255310	2020	5	2	4	5
Tubular aggregates	G71.2	Congenital Myopathies	160565	2593	13	1	1	1
Morbo di Basedow	E05.5	Hypethyroidism		181399	2	0	0	0
Myofibrillar myopathies	G72.8	Congenital Myopathies	601419	593	10	2	4	5
Glycogenosis type II, including Pompe	E74.0	Glycogen Storage Disease	232300	365	109	11	18	28
Glycogenosis type V, Mc Ardle	E74.0	Glycogen Storage Disease	232600	368	19	0	11	11
Glycogenosis type III	E74.0	Glycogen Storage Disease	232400	366	9	45	20	20

Name of Disease	ICD10_Identifier	ICD10_Name	MIM_Number	ORPHA_Number	N. samples Muscle tissue	N. samples DNA	N. samples Myoblasts	N. samples Fibroblasts
Glycogenosis type VII	E74.0	Glycogen Storage Disease	232800	371	1	0	5	5
Glycogenosis type XIII	E74.0	Glycogen Storage Disease	131370	99849	1	0	0	0
MAD deficiency	E71.3		102770	26791	37	8	3	3
MtDNA macrodeletions		Mitochondrial Myopathy		35697	88	59	25	24
MtDNA multiple deletions		Mitochondrial Myopathy		35697	82	130	40	30
LEBER	H47	Mitochondrial Myopathy	535000	104	4	37	0	0
MERRF	G40.3	Mitochondrial Myopathy not elsewhere classified	545000	551	26	39	6	6
MELAS	G71.3	Mitochondrial Myopathy not elsewhere classified	540000	550	24	38	5	5
NARP	G31.8	Mitochondrial Myopathy	551500	644	2	3	0	0
PEO 3243 point mutations	G71.3	Mitochondrial Myopathy not elsewhere classified	540000	550	9	9	1	1
Other mtDNA point mutations		Mitochondrial Myopathy	540000	35697	23	45	4	12
MtDNA depletion	Q99.8	Mitochondrial Myopathy	609560	35698	9	15	0	0
Other mitochondrial disorders	G71.3	Mitochondrial Myopathy	540000	35698	306	248	54	35
Lipid storage myopathy	G73	Lipid storage disorder unspecified	255100	206953	51	28	2	2
LCAD deficiency	E71.3	Mitochondrial Myopathy	201460	99900	6	0	0	0
MCAD deficiency	E71.3	Lipid storage disorder unspecified	201450	42	1	0	0	0
CPT deficiency	E71.3	Disorders of fatty-acid metabolism	255110	157	16	36	1	1
Inflammatory Myopathies	M60.8	Inflammatory Myopathies	160750	48918	739	47	109	82
IBM	M60.8	Inflammatory Myopathies not elsewhere classified	147421	611	57	18	15	14
Critical Illness Myopathy					12	0	0	0
Myasthenia Gravis	G70.0	Myasthenia Gravis	254200	589	17	0	0	0
Malignant hyperthermia	T88.3	Malignant hyperthermia	145600	423	12	192	1	0
Corea di Huntington	G10	Huntington's disease	143100	399	2	0	0	0
Essential hyperckemia					138	62	39	32
Spinal muscular atrophy, SMA-1,2,3	G12.0	Spinal muscular atrophy and related syndroms	253550	70	55	54	3	13

Name of Disease	ICD10_Identifier	ICD10_Name	MIM_Number	ORPHA_Number	N. samples Muscle tissue	N. samples DNA	N. samples Myoblasts	N. samples Fibroblasts
Spinal bulbar muscular atrophy, Kennedy	G12.2	Other spinal muscular atrophy and related syndroms	313200	481	3	8	0	0
Amyotrophic lateral sclerosis	G12.2	Motor neuron disease	105400	803	115	101	21	28
Mild non specific myopathic signs					1250	944	192	192
Mild non specific neurogenic signs					783	24	32	33
Normal muscle biopsy		Controls			1223	56	175	153
Dilatative idiopathic cardiomyopathy	I42.0	Dilated cardiomyopathy	115200	83618	5	0	0	0
Secondary dilatative cardiomyopathy	I42.0	Dilated cardiomyopathy		217629	3	0	0	0
Myocarditis		Cardiomyopathy			3	0	0	0
Ischemic cardiopathy		Cardiopathy			4	0	0	0
Degenerative neuropathy (hereditary and acquired)	G60.8	Degenerative neuropathies	162400	36386	504	1	1	1
Inflammatory neuropathy	G72.4	Inflammatory neuropathies			60	2	0	0
Normal nerve biopsy		Controls			25	0	0	0
Spastic Paraparesis (Paraplegin. Def.)	G11.4	Hereditary spastic paraplegia	602783	685	1	7	1	1
S. of Down	Q90	Genetic diseases	190685	870	1	186	0	26