GÉNÉTHON
INNOVER POUR GUÉRIR

CODE	PATHOLOGIE	Omim	Individus	Familles	Atteints
ADDM	MYOPATHY DISTAL	160500	19	15	16
AMC	ARTHROGRYPOSIS	108110	235	74	125
AMD	GLYCOGEN STORAGE DISEASE, POMPE DISEASE	232300	23	16	21
ASP	AMYOTROPHY SCAPULOPERONEAL	181400	33	19	22
BET	BETHLEM MYOPATHY	158810	319	74	210
BMD	MUSCULAR DYSTROPHY BECKER TYPE	300376	77	49	61
BRO	BRODY MYOPATHY	601003	12	9	11
BVVLS1	BROWN-VIALETTO-VAN LAERE SYNDROME 1	211530	3	1	1
CBM	DESMIN RELATED MYOPATHY	601419	19	6	7
CCD	CENTRAL CORE DISEASE	117000	805	140	365
CHAC	CHOREOACANTHOCYTOSIS	200150	1	1	1
CMT	CHARCOT MARIE TOOTH DISEASE	145900	1071	570	689
CMT1	CHARCOT-MARIE-TOOTH DISEASE TYPE 1	118200	93	35	52
CMT1A	CHARCOT-MARIE-TOOTH DISEASE, TYPE 1A	118220	21	11	15
CMT1C	CHARCOT-MARIE-TOOTH DISEASE, TYPE 1C	601098	6	4	4
CMT2	CHARCOT-MARIE-TOOTH DISEASE, AXONAL TYPE 2	118210	709	254	369
CMT2A2	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, AUTOSOMAL DOMINANT, TYPE 2A2A	609260	16	10	12
CMT2C	TRANSIENT RECEPTOR POTENTIAL CHANNEL	605427	6	2	2
CMT4	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4, AR	214400	66	26	44
CMT4B1	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4B1	601382	3	1	1
CMTX	CHARCOT-MARIE-TOOTH DISEASE, X-LINKED DOMINANT	302800	10	4	8
CPT2	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, MYOPATHIC	255110	1	1	1
DBU	MUSCULAR DYSTROPHY SCLEROATONIC	254090	316	85	181
DD	DANON DISEASE	300257	19	11	15
DM1	DYSTROPHIA MYOTONICA, STEINERT DISEASE	160900	584	354	510
DM2	DYSTROPHIA MYOTONICA 2	602668	152	90	131

DMD	MUSCULAR DYSTROPHY DUCHENNE TYPE	310200	418	241	369
EDMD	EMERY-DREIFUSS MUSCULAR DYSTROPHY	181350	394	121	205
FCMD	FUKUYAMA CONGENITAL MUSCULAR DYSTROPHY	253800	6	4	6
FOP	FIBRODYSPLASIA OSSIFICANS PROGRESSIVA	135100	23	11	12
FSHD	FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY	158900	988	375	566
GAN	GIANT AXONAL NEUROPATHY	256850	65	20	26
GSD	GLYCOGEN STORAGE DISEASE 2B		14	9	14
GSDIII	GLYCOGEN STORAGE DISEASE III	232400	41	40	41
HARD	WALKER-WARBURG SYNDROME	236670	6	3	3
HOKPP	HYPOKALEMIC PERIODIC PARALYSIS	170400	45	25	28
HSAN1A	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE IA	162400	16	5	7
HSAN1B	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE 1B	608088	21	8	12
HSAN2B	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE 2B	613115	1	1	1
HSN4	HEREDITARY SENSORY NEUROPATHY	256800	25	16	18
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	173	83	118
IIM	MYOPATHY, FAMILIAL IDIOPATHIC INFLAMMATORY	160750	3	2	2
LGMD	LIMB-GIRDLE MUSCULAR DYSTROPHY		1732	654	922
LGMD2A	MUSCULAR DYSTROPHY, LIMB-GIRDLE,TYPE 2A	253600	727	134	276
LGMD2B	MUSCULAR DYSTROPHY, LIMB-GIRDLE, AUTOSOMAL RECESSIVE 2	253601	38	17	26
LGMD2C	MUSCULAR DYSTROPHY, LIMB-GIRDLE AUTOSOMAL RECESSIVE TYPE 2C	253700	301	68	135
LGMD2D	MUSCULAR DYSTROPHY, LIMB-GIRDLE AUTOSOMAL RECESSIVE TYPE 2D	600119	9	5	9
LGMD2I	MUSCULAR DYSTROPHY, LIMB-GIRDLE AUTOSOMAL RECESSIVE TYPE 2I	607155	67	19	34
LGMD2L	MUSCULAR DYSTROPHY, LIMB-GIRDLE AUTOSOMAL RECESSIVE TYPE 2L	611307	9	5	7
LGMD2N	MUSCULAR DYSTROPHY, LIMB-GIRDLE AUTOSOMAL RECESSIVE TYPE 2N	613158	5	2	3
MAC	MCARDLE DISEASE	232600	184	135	162
MCF	MUSCLE CRAMPS, FAMILIAL	158400	6	5	6
MCMU	MYOPATHY CENTRONUCLEAR	255200	391	85	123
MDC	MUSCULAR DYSTROPHY CONGENITAL	253800	2410	739	1195
MDD	MYOADENYLATE DEAMINASE DEFICIENCY	615511	3	3	3

MEAX	MYOPATHY, X-LINKED, WITH EXCESSIVE AUTOPHAGY	310440	10	6	6
MEB	MUSCLE-EYE-BRAIN-DISEASE	253280	9	4	3
MELAS	MELAS SYNDROME	540000	2	2	2
MERRF	MERRF SYNDROME	545000	2	1	2
MG	MYASTHENIA GRAVIS	254200	1641	677	747
MMF	MYOPATHY, MYOFIBRILLAR	609524	123	20	61
MSD	DESMIN-RELATED MYOPATHY	601419	165	39	83
MTD	MYOTONIA CONGENITA, THOMSEN DISEASE	160800	128	85	99
MTSD	MYOPATHY AND TRIGLYCERIDE STORAGE DISEASE		151	105	134
MTU	MYOTUBULAR MYOPATHY	310400	142	60	65
MULT	MINICORE MYOPATHY WITH EXTERNAL OPHTHALMOPLEGIA	255320	405	97	175
MYD	MYOPATHY DISTAL WITH ONSET IN INFANCY	160300	600	249	389
NAPB	NEURITIS WITH BRANCHIAL PREDILECTION	162100	2	2	2
NEM	NEMALINE MYOPATHY	161800	526	142	226
NM	NONAKA DISTAL MYOPATHY	605820	5	4	4
NTO	TOMACULOUS NEUROPATHY	162500	11	5	3
OPMD	OCULOPHARYNGEAL MUSCULAR DYSTROPHY	164300	377	162	243
PFKM	PHOSPHOFRUCTOKINASE, MUSCLE TYPE	610681	4	4	4
PMDM	POLYMYOSITIS		2	2	2
RMD	RIPPLING MUSCLE DISEASE	606072	4	4	4
RSMD1	RIGID SPINE MUSCULAR DYSTROPHY	602771	57	24	24
SCN4	HYPERKALEMIC PERIODIC PARALYSIS	170500	7	5	5
SCN4A	MYOTONIA PERMANENS	608390	2	2	2
SFNP	ERYTHROMELALGIA, PRIMARY	133020	1	1	1
SMA	SPINAL MUSCULAR ATROPHY	253300	954	461	617
SMAD	SPINAL MUSCULAR ATROPHY, AUTOSOMAL DOMINANT	158600	26	11	12
SMC	MYASTHENIC SYNDROME	601462	814	251	430
VCPDM	VOCAL CORD AND PHARYNGEAL DYSFUNCTION WITH DISTAL MYOPATHY	606070	4	2	3
VLCAD	DEFICIENCY OF ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN	201475	2	2	2

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