Roadmap 2 Rare

Full table of genes and disorders - 122-gene Muscle Disorders panel

GENE	DISORDER
ACTA1	Myopathy, actin, congenital, with cores; myopathy, actin, congenital, with excess of thin myofilaments; myopathy, congenital, with fiber-type disproportion 1; nemaline myopathy 3
ACTN2	Multiple structured core disease; progressive early-onset muscle weakness
AGRN	Congenital myasthenic syndrome; myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
ANO5	Miyoshi muscular dystrophy 3613319; muscular dystrophy, limb-girdle, type 2L611307
ATP2A1	Brody myopathy 601003
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies typeA 11; congenital muscular dystrophies
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAG3	Myopathy, myofibrillar, 6, 612954
BIN1	Centronuclear myopathy, recessive; myopathy, centronuclear, autosomal recessive, 255200
BVES	Muscular dystrophy, limb-girdle, type 2X, 616812

GENE	DISORDER
CACNA1S	{Malignant hyperthermia susceptibility 5}, 601887
CAPN3	Muscular dystrophy, limb-girdle, type 2A, 253600; limb-girdle muscular dystrophy, recessive; limb-girdle muscular dystrophy
CASQ1	Myopathy, vacuolar, with CASQ1 aggregates
CAV3	Muscular dystrophy, limb-girdle, type IC607801; myopathy, distal, Tateyama type 614321; rippling muscle disease 606072
CCDC78	Myopathy, centronuclear, 4, 614807
CFL2	Nemaline myopathy 7, autosomal recessive, 610687; Nemaline myopathy, recessive
CHAT	Myasthenic syndrome, congenital, 6, presynaptic, 254210; congenital myasthenics syndrome associated with episodic apnea; CMS-EA
CHKB	Congenital muscular dystrophy, CKHB-related; muscular dystrophy, congenital, megaconial type, 602541

sanofi

Roadmap 2 Rare

Full table of genes and disorders - 122-gene Muscle Disorders panel

GENE	DISORDER
CHRNA1	Congenital myasthenic syndrome, dominant/recessive; myasthenic syndrome, congenital, 1A, slow-channel, 601462; myasthenic syndrome, congenital, 1B, fast-channel, 608930; slow channel myasthenic syndrome; fast-channel myasthenic syndrome; acetylcholine receptor deficiency syndrome
CHRNB1	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314; myasthenic syndrome, congenital, 2A, slow-channel, 616313; slow channel myasthenic syndrome; fast channel myasthenic syndrome; acetylcholine receptor deficiency syndrome; myasthenic syndrome, slow-channel congenital, 601462; congenital myasthenic syndrome, dominant/recessive

GENE	DISORDER
CHRND	Congenital myasthenic syndrome, dominant/recessive; myasthenic syndrome, slow-channel congenital, 601462; slow channel myasthenic syndrome; fast-channel myasthenic syndrome; acetylcholine receptor deficiency syndrome; ?myasthenic syndrome, congenital, 3A, slow-channel, 616321; ?myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323; myasthenic syndrome, congenital, 3B, fast-channel, 616322

sanofi

Roadmap 2 Rare

Full table of genes and disorders - 122-gene Muscle Disorders panel

GENE	DISORDER
CHRNE	Congenital myasthenic syndrome, dominant/recessive; myasthenic syndrome, slow-channel congenital, 601462; myasthenic syndrome, congenital, 4A, slow-channel, 605809; myasthenic syndrome, congenital, 4B, fast-channel, 616324; myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931; slow channel myasthenic syndrome; fast channel myasthenic syndrome; acetylcholine receptor deficiency syndrome; reduced channel conductance syndrome
COL12A1	EDS/myopathy overlap syndrome
COL13A1	Congenital myasthenic syndrome type 19; myasthenic syndrome, congenital, 19, 616720
COL6A1	Bethlem myopathy 1158810
COL6A2	Bethlem myopathy 1158810
COL6A3	Bethlem myopathy 1158810
COLQ	Congenital myasthenic syndrome, recessive; congenital myasthenic syndrome with endplate acetylcholinesterase deficiency; myasthenic syndrome, congenital, 5, 603034
CPT2	Carnitine palmitoyltransferase deficiency
CRYAB	Cataract 16, multiple types, 613763; myofibrillar myopathy
DAG1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818; limb-girdle muscular dystrophy; congenital muscular dystrophy; limb-girdle muscular dystrophy
DES	Muscular dystrophy, limb-girdle, type 2R, 615325; myofibrillar myopathy; cardiomyopathy; limb girdle muscular dystrophy
DMD	Becker muscular dystrophy300376
DNAJB6	Limb-girdle muscular dystrophy, dominant; muscular dystrophy, limb-girdle, type 1E, 603511
DNM2	Myopathy, centronuclear, 160150; Charcot-Marie-Tooth disease, axonal, type 2M, 606482
DOK7	Congenital myasthenic syndrome; limb-girdle muscular dystrophy

sanofi

Roadmap 2 Rare

Full table of genes and disorders - 122-gene Muscle Disorders panel

GENE	DISORDER
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093; myasthenic syndrome, congenital, 13, with tubular aggregates, 614750; limb-girdle congenital myasthenic; tubular aggregates; congenital disorder of glycosylation type Ij (CDG-IJ)
DPM3	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15
DYSF	Miyoshi muscular dystrophy 1254130; muscular dystrophy, limb-girdle, type 2B253601; myopathy, distal, with anterior tibial onset 606768
EMD	Emery-Dreifuss muscular dystrophy 1, X-linked310300
FHL1	Emery-Dreifuss muscular dystrophy
FKBP14	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss 6, 14557
FKRP	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5607155
FKTN	Fukuyama congenital muscular dystrophy; muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4253800
FLNC	Myopathy, myofibrillar, 5609524

GENE	DISORDER
GAA	Pompe disease; Glycogen storage disease II
GFPT1	Congenital myasthenic syndrome, recessive; myasthenia, congenital, 12, with tubular aggregates, 610542; limb-girdle congenital myasthenic syndrome; tubular aggregates
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type
GNE	Nonaka myopathy, 605820; distal myopathy; limb-girdle muscular dystrophy; limb-girdle muscular dystrophy; quadriceps sparing myopathy; distal myopathy; Nonaka myopathy, HIBM
HNRNPDL	Muscular dystrophy, limb-girdle, type 1G 609115; limb-girdle muscular dystrophy
INPP5K	Congenital muscular dystrophy overlapping Marinesco-Sjogren syndrome and dystroglycanopathy; congenital muscular dystrophy with cataracts and mild cognitive impairment
ISPD/CRPPA	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052; congenital muscular dystrophy; limb-girdle muscular dystrophy

sanofi

Roadmap 2 Rare

Full table of genes and disorders - 122-gene Muscle Disorders panel

GENE	DISORDER
ITGA7	Congenital muscular dystrophy, ITGA7-related; muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
KBTBD13	Nemaline myopathy, dominant; nemaline myopathy 6, autosomal dominant, 609273
KLHL40	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	Nemaline myopathy 9, 615731
KY	Myopathy, myofibrillar, 7
LAMA2	Muscular dystrophy, congenital, merosin-deficient or partially-deficient, 607855; congenital muscular dystroph
LAMP2	Danon disease 300257
LARGE1	Congenital muscular dystrophy, alpha-dystroglycan related; muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6 613154; muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6 608840
LDB3	Myopathy, myofibrillar, 4
LMNA	Limb-girdle muscular dystrophy, recessive; Emery-Dreifuss muscular dystrophy 2, AD, 181350; limb-girdle muscular dystrophy
LMOD3	Nemaline myopathy 10 616165

GENE	DISORDER
LRP4	Congenital myasthenic syndrome; myasthenic syndrome, congenital, 17, 616304
MEGF10	Myopathy, early-onset, areflexia, respiratory distress, and dysphagia; myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MTM1	Myotubular myopathy, X-linked, 310400
MUSK	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325; congenital myasthenic syndrome, recessive; congenital myasthenic syndrome
MYBPC1	Arthrogryposis, distal, type 1B, 614335; lethal congenital contracture syndrome 4, 614915
MYF6	Myopathy, centronuclear, 3
MYH2	Proximal myopathy and ophthalmoplegia 605637
MYH7	Laing distal myopathy, 160500; cardiomyopathy; distal myopathy
MYO18B	Klippel-Feil syndrome 4, autosomal recessive, with nemaline myopathy and facial dysmorphis

sanofi

Roadmap 2 Rare

Full table of genes and disorders - 122-gene Muscle Disorders panel

GENE	DISORDER
MYOT	Limb-girdle muscular dystrophy, dominant; muscular dystrophy, limb-girdle, type 1A, 159000; limb-girdle muscular dystrophy
MYPN	Congenital cap myopathy; nemaline myopathy, 617336
NEB	Nemaline myopathy; nemaline myopathy, recessive; nemaline myopathy 2, autosomal recessive, 256030
ORAI1	Myopathy, tubular aggregate, 2, 615883
PABPN1	Oculopharyngeal muscular dystrophy
PAX7	Hypotonia; axial hypotonia; ptosis; scoliosis; delayed motor milestones; myopathy, congenital, progressive, with scoliosis, 618578
PFKM	Glycogen storage disease VII232800
PHKA1	Muscle glycogenosis300559
PLEC	Muscular dystrophy with epidermolysis bullosa simplex, 226670; limb-girdle muscular dystrophy
PNPLA2	Neutral lipid storage disease with myopathy; neutral lipid storage disease with myopathy, 610717
POGLUT1	Muscular dystrophy, limb-girdle, autosomal recessive 21

GENE	DISORDER
POMGNT1	Limb-girdle muscular dystrophy, recessive; muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type; limb-girdle muscular dystrophy
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830; limb-girdle muscular dystrophy
POMK	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12
POMT1	Limb-girdle muscular dystrophy, recessive; muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type; limb-girdle muscular dystrophy
POMT2	Limb-girdle muscular dystrophy, recessive; muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type; limb-girdle muscular dystrophy
PYGM	Glycogen storage disease V McArdle disease 232600AR

sanofi

Roadmap 2 Rare

Full table of genes and disorders - 122-gene Muscle Disorders panel

GENE	DISORDER
RAPSN	Congenital myasthenic syndrome, recessive; congenital myasthenic syndrome; myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326; acute respiratory crises; late and early onset
RBCK1	Polyglucosan body myopathy 1 with or without immunodeficiency 615895
RXYLT1/ TMEM5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type
RYR1	{Malignant hyperthermia susceptibility 1}, 145600; central core disease, 117000; minicore myopathy with external ophthalmoplegia, 255320; neuromuscular disease, congenital, with uniform type 1 fiber, 117000 King-Denborough syndrome, 145600
SCN4A	Congenital myopathy
SELENON	Muscular dystrophy, rigid spine, 1, 602771; congenital myopathy; muscular dystophy; rigid spine syndrome
SGCA	Muscular dystrophy, limb-girdle, type 2D, 608099; limb-girdle muscular dystrophy
SGCB	Limb-girdle muscular dystrophy, recessive; muscular dystrophy, limb-girdle, type 2E, 604286; limb-girdle muscular dystrophy
SGCD	Limb-girdle muscular dystrophy, recessive; muscular dystrophy, limb-girdle, type 2F, 601287; limb-girdle muscular dystrophy
SGCG	Limb-girdle muscular dystrophy, recessive; muscular dystrophy, limb-girdle, type 2C, 253700; limb-girdle muscular dystrophy
SLC18A3	Congenital myasthenic syndrome; ophthalmopleggia and apnea; myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC25A1	?Myasthenic syndrome, congenital, 23, presynaptic; 618197
SLC5A7	Congenital myasthenic syndrome; hereditory motor neuropathy; myasthenic syndrome, congenital, 20, presynaptic, 617143
SPEG	Centronuclear myopathy 5 615959
STAC3	Myopathy, congenital, Baily-Bloch, 255995
STIM1	Myopathy, tubular aggregate, 1, 160565
SYNE1	Emery-Dreifuss muscular dystrophy 4, autosomal dominant 612998

sanofi

Roadmap 2 Rare

Full table of genes and disorders - 122-gene Muscle Disorders panel

GENE	DISORDER
SYNE2	Emery-Dreifuss muscular dystrophy 5, autosomal dominant 612999; congenital muscular dystrophy
SYT2	Myasthenic syndrome, congenital, 7, presynaptic, 616040
TCAP	Limb-girdle muscular dystrophy, recessive; muscular dystrophy, limb-girdle, type 2G, 601954; limb-girdle muscular dystrophy
TMEM43	Emery-Dreifuss muscular dystrophy 7, AD 614302
TNNT1	Nemaline myopathy; nemaline myopathy, recessive; nemaline myopathy 5, Amish type, 605355
TNPO3	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TOR1AIP1	Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, OMIM:617072
TPM2	CAP myopathy 2 609285; nemaline myopathy 4, autosomal dominant 609285; arthrogryposis multiplex congenita, distal, type1 108120: arthrogryposis, distal, type 2B 601680
TPM3	CAP myopathy 1 609284; myopathy, congenital, with fiber-type disproportion 255310; nemaline myopathy 1, autosomal dominant or recessive 609284
TRAPPC11	Muscular dystrophy, limb-girdle, type 2S, 615356
TRIM32	Limb-girdle muscular dystrophy, recessive; muscular dystrophy, limb-girdle, type 2H, 254110; limb-girdle muscular dystrophy
TRIP4	Severe congenital myopathy with congenital bone fractures, 616866; spinal muscular atrophy with congenital bone fractures 1, 616866
TTN	Muscular dystrophy, limb-girdle, type 2J, 608807; limb-girdle muscular dystrophy; distal myopathy; myofibrillar myopathy; congenital myopathy; dilated cardiomyopathy; HMERF; arthrogryposis
VAMP1	Congenital myasthenic syndrome; presynaptic CMS
VCP	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1167320
VMA21	Myopathy, X-linked, with excessive autophagy, 310440; X-linked myopathy with excessive autophagy

sanofi