PerkinElmer
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PerkinElmer GENOMICS

OMIM Disease by Gene

Gene	Disease / Phenotype
ACTA1	Myopathy, actin, congenital, with cores, Myopathy, actin, congenital, with excess of thin myofilaments, Myopathy, congenital, with fiber-type disproportion 1, Myopathy, scapulohumeroperoneal, Nemaline myopathy 3, autosomal dominant or recessive
ANO5	Gnathodiaphyseal dysplasia, Miyoshi muscular dystrophy 3, Muscular dystrophy, limb-girdle, autosomal recessive 12
ATP2A1	Brody myopathy
BAG3	Cardiomyopathy, dilated, 1HH, Myopathy, myofibrillar, 6
CAPN3	Muscular dystrophy, limb-girdle, autosomal dominant 4, Muscular dystrophy, limb-girdle, autosomal recessive 1
CAV3*	Cardiomyopathy, familial hypertrophic, Creatine phosphokinase, elevated serum, Long QT syndrome 9, Myopathy, distal, Tateyama type, Rippling muscle disease 2
CLCN1	Myotonia congenita, dominant, Myotonia congenita, recessive, Myotonia levior, recessive
COL12A1	Bethlem myopathy 2, Ullrich congenital muscular dystrophy 2
COL6A1	Bethlem myopathy 1, Ullrich congenital muscular dystrophy 1
COL6A2	Bethlem myopathy 1, Myosclerosis, congenital, Ullrich congenital muscular dystrophy 1
COL6A3	Bethlem myopathy 1, Dystonia 27, Ullrich congenital muscular dystrophy 1
CRPPA	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7
CRYAB*	Cardiomyopathy, dilated, 1II, Cataract 16, multiple types, Myopathy, myofibrillar, 2, Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related
DAG1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9
DES*	Cardiomyopathy, dilated, 1I, Myopathy, myofibrillar, 1, Scapuloperoneal syndrome, neurogenic, Kaeser type
DMD	Becker muscular dystrophy, Cardiomyopathy, dilated, 3B, Duchenne muscular dystrophy
DNAJB6	Muscular dystrophy, limb-girdle, autosomal dominant 1
DNM2	Centronuclear myopathy 1, Charcot-Marie-Tooth disease, axonal type 2M, Charcot-Marie-Tooth disease, dominant intermediate B, Lethal congenital contracture syndrome 5
DYSF	Miyoshi muscular dystrophy 1, Muscular dystrophy, limb-girdle, autosomal recessive 2, Myopathy, distal, with anterior tibial onset
EMD	Emery-Dreifuss muscular dystrophy 1, X-linked
FHL1*	Emery-Dreifuss muscular dystrophy 6, X-linked, Hemophagocytic lymphohistiocytosis, familial, 1, Myopathy, X-linked, with postural muscle atrophy, Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, Reducing body myopathy, X-linked 1b, with late childhood or adult onset, Scapuloperoneal myopathy, X-linked dominant, Uruguay faciocardiomusculoskeletal syndrome
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5
FKTN	Cardiomyopathy, dilated, 1X, Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4
FLNC*	Cardiomyopathy, familial hypertrophic, 26, Cardiomyopathy, familial restrictive 5, Myopathy, distal, 4, Myopathy, myofibrillar, 5
GAA	Glycogen storage disease II
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14
GNE*	Nonaka myopathy, Sialuria
HNRNPA1*	Amyotrophic lateral sclerosis 20, Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3
ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency
LAMA2	Muscular dystrophy, congenital, merosin deficient or partially deficient, Muscular dystrophy, limb-girdle, autosomal recessive 23
LARGE1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6
LDB3*	Cardiomyopathy, dilated, 1C, with or without LVNC, Cardiomyopathy, hypertrophic, 24, Left ventricular noncompaction 3, Myopathy, myofibrillar, 4
LIMS2	Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue

Gene	Disease / Phenotype
LMNA*	Cardiomyopathy, dilated, 1A, Charcot-Marie-Tooth disease, type 2B1, Emery-Dreifuss muscular dystrophy 2, autosomal dominant, Emery-Dreifuss muscular dystrophy 3, autosomal recessive, Heart-hand syndrome, Slovenian type, Hutchinson-Gilford progeria, Lipodystrophy, familial partial, type 2, Malouf syndrome, Mandibuloacral dysplasia, Muscular dystrophy, congenital, Restrictive dermopathy, lethal
MYH7	Cardiomyopathy, dilated, 1S, Cardiomyopathy, hypertrophic, 1, Laing distal myopathy, Left ventricular noncompaction 5, Myopathy, myosin storage, autosomal dominant, Myopathy, myosin storage, autosomal recessive, Scapuloperoneal syndrome, myopathic type
MYOT*	Myopathy, myofibrillar, 3, Myopathy, spheroid body
NEB	Nemaline myopathy 2, autosomal recessive
PABPN1*	Oculopharyngeal muscular dystrophy
PLEC	Epidermolysis bullosa simplex with muscular dystrophy, Epidermolysis bullosa simplex with nail dystrophy, Epidermolysis bullosa simplex with pyloric atresia, Epidermolysis bullosa simplex, Ogna type, Muscular dystrophy, limb-girdle, autosomal recessive 17
PNPLA2*	Neutral lipid storage disease with myopathy
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, Retinitis pigmentosa 76
POMK	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2
PYGM*	McArdle disease
RAPSN	Fetal akinesia deformation sequence 2, Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency
RYR1	Central core disease, King-Denborough syndrome, Malignant hyperthermia susceptibility 1, Minicore myopathy with external ophthalmoplegia, Neuromuscular disease, congenital, with uniform type 1 fiber
SCN4A	Brugada syndrome 6, Hyperkalemic periodic paralysis, type 2, Hypokalemic periodic paralysis, type 2, Myasthenic syndrome, congenital, 16, Myotonia congenita, atypical, acetazolamide-responsive, Paramyotonia congenita
SELENON	Muscular dystrophy, rigid spine, 1, Myopathy, congenital, with fiber-type disproportion
SGCA	Muscular dystrophy, limb-girdle, autosomal recessive 3
SGCB	Muscular dystrophy, limb-girdle, autosomal recessive 4
SGCD	Cardiomyopathy, dilated, 1L, Muscular dystrophy, limb-girdle, autosomal recessive 6
SGCE	Dystonia-11, myoclonic
SGCG	Muscular dystrophy, limb-girdle, autosomal recessive 5
SIL1	Marinesco-Sjogren syndrome
SMCHD1	Bosma arhinia microphthalmia syndrome, Fascioscapulohumeral muscular dystrophy 2, digenic
SYNE1	Arthrogryposis multiplex congenita, myogenic type, Emery-Dreifuss muscular dystrophy 4, autosomal dominant, Spinocerebellar ataxia, autosomal recessive 8
TCAP	Cardiomyopathy, hypertrophic, 25, Muscular dystrophy, limb-girdle, autosomal recessive 7
TIA1*	Welander distal myopathy
TNPO3	Muscular dystrophy, limb-girdle, autosomal dominant 2
TPM2	Arthrogryposis, distal, type 1A, Arthrogryposis, distal, type 2B4, CAP myopathy 2, Nemaline myopathy 4, autosomal dominant
TPM3	CAP myopathy 1, Myopathy, congenital, with fiber-type disproportion, Nemaline myopathy 1, autosomal dominant or recessive
TRAPPC11	Muscular dystrophy, limb-girdle, autosomal recessive 18
TRIM32	Bardet-Biedl syndrome 11, Muscular dystrophy, limb-girdle, autosomal recessive 8
TTN	Cardiomyopathy, dilated, 1G, Cardiomyopathy, familial hypertrophic, 9, Muscular dystrophy, limb-girdle, autosomal recessive 10, Myopathy, myofibrillar, 9, with early respiratory failure, Salih myopathy, Tibial muscular dystrophy, tardive
VCP*	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, Charcot-Marie-Tooth disease, type 2Y, Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1
*Adult- only age of onset	