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Genetic Conditions

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Genetic Conditions

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Arylsulfatase B deficiency, see Mucopolysaccharidosis type VI

Arylsulfatase E deficiency, see X-linked chondrodysplasia punctata 1

AS, see Angelman syndrome

ASA, see Argininosuccinic aciduria

Asadollahi-Rauch syndrome, see MED13L syndrome

ASAuria, see Argininosuccinic aciduria

ASD, see Autism spectrum disorder

Asidan ataxia, see Spinocerebellar ataxia type 36

ASL deficiency, see Argininosuccinic aciduria

ASNS deficiency, see Asparagine synthetase deficiency

ASNSD, see Asparagine synthetase deficiency

Aspa deficiency, see Canavan disease

Asparagine synthetase deficiency

Aspartoacylase deficiency, see Canavan disease

Aspartyl-tRNA synthetase deficiency, see Hypomyelination with brainstem and spinal cord involvement and leg spasticity

Aspartylglucosamidase deficiency, see Aspartylglucosaminuria

Aspartylglucosaminidase deficiency, see Aspartylglucosaminuria

Aspartylglucosaminuria

Aspartylglycosaminuria, see Aspartylglucosaminuria

Asphyxiating thoracic chondrodystrophy, see Asphyxiating thoracic dystrophy

Asphyxiating thoracic dysplasia, see Asphyxiating thoracic dystrophy

Asphyxiating thoracic dystrophy

Asplenia, familial, see Isolated congenital asplenia

Asplenia, isolated congenital, see Isolated congenital asplenia

ASRAS, see MED13L syndrome

Asymbolia for pain, see Channelopathy-associated congenital insensitivity to pain

Asymmetric hypoplasia of facial structures, see Craniofacial microsomia

Ataxia neuropathy spectrum

Ataxia telangiectasia syndrome, see Ataxia-telangiectasia

Ataxia with isolated vitamin E deficiency, see Ataxia with vitamin E deficiency

Ataxia with lactic acidosis, see Pyruvate dehydrogenase deficiency

Ataxia with lactic acidosis, type II, see Pyruvate carboxylase deficiency

Ataxia with oculomotor apraxia

Ataxia with vitamin E deficiency

Ataxia, delayed dentition, and hypomyelination, see Pol III-related leukodystrophy

Ataxia, fatal X-linked, with deafness and loss of vision, see Arts syndrome

Ataxia-deafness-optic atrophy, lethal, see Arts syndrome

Ataxia-hypogonadism-choroidal dystrophy syndrome, see Boucher-NeuhÃ¤user syndrome

Ataxia-pancytopenia syndrome

Ataxia-telangiectasia

Ataxia-telangiectasia variant 1, see Nijmegen breakage syndrome

ATD, see Asphyxiating thoracic dystrophy

Atelosteogenesis de la Chapelle type, see Atelosteogenesis type 2

Atelosteogenesis type 1

Atelosteogenesis type 2

Atelosteogenesis type 3

Atelosteogenesis type I, see Atelosteogenesis type 1

Atelosteogenesis type III, see Atelosteogenesis type 3

Atelosteogenesis, type 2, see Atelosteogenesis type 2

ATM, see Ataxia-telangiectasia

Atopic dermatitis

Atopic eczema, see Atopic dermatitis

ATP synthase deficiency, see Mitochondrial complex V deficiency

ATP8B1-related intrahepatic cholestasis, see Progressive familial intrahepatic cholestasis

ATP8B1-related intrahepatic cholestasis, see Benign recurrent intrahepatic cholestasis

ATR-X syndrome, see Alpha thalassemia X-linked intellectual disability syndrome

Atrial fibrillation, familial, see Familial atrial fibrillation

Atrio-digital syndrome, see Holt-Oram syndrome

Atriodigital dysplasia, see Holt-Oram syndrome

Atrophia bulborum hereditaria, see Norrie disease

ATRX syndrome, see Alpha thalassemia X-linked intellectual disability syndrome

ATS, see Arterial tortuosity syndrome

ATS, see Andersen-Tawil syndrome

Attention deficit, see Attention-deficit/hyperactivity disorder

Attention deficit disorder, see Attention-deficit/hyperactivity disorder

Attention deficit disorder of childhood with hyperactivity, see Attention-deficit/hyperactivity disorder

Attention deficit disorder with hyperactivity, see Attention-deficit/hyperactivity disorder

Attention deficit disorder with hyperactivity syndrome, see Attention-deficit/hyperactivity disorder

Attention deficit hyperactivity disorder, see Attention-deficit/hyperactivity disorder

Attention-deficit/hyperactivity disorder

ATTR, see Transthyretin amyloidosis

ATXPC, see Ataxia-pancytopenia syndrome

Atypical hemolytic-uremic syndrome

Au-Kline syndrome

Auditory vertigo, see MÃ©niÃ¨re disease

AUH defect, see 3-methylglutaconyl-CoA hydratase deficiency

Aural vertigo, see MÃ©niÃ¨re disease

Auricular fibrillation, see Familial atrial fibrillation

Auriculo-condylar syndrome

Auriculobranchiogenic dysplasia, see Craniofacial microsomia

Auriculocondylar syndrome, see Auriculo-condylar syndrome

Austin syndrome, see Multiple sulfatase deficiency

Autism spectrum disorder

Autism, susceptibility to, 14A, see 16p11.2 deletion syndrome

Autism, susceptibility to, 14B, see 16p11.2 duplication

Autism-dementia-ataxia-loss of purposeful hand use syndrome, see Rett syndrome

Autistic continuum, see Autism spectrum disorder

Autoimmune Addison disease

Autoimmune Addison's disease, see Autoimmune Addison disease

Autoimmune adrenalitis, see Autoimmune Addison disease

Autoimmune chronic lymphocytic thyroiditis, see Hashimoto's disease

Autoimmune diabetes, see Type 1 diabetes

Autoimmune hyperthyroidism, see Graves' disease

Autoimmune lymphoproliferative syndrome

Autoimmune polyendocrinopathy syndrome type 1, see Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy

Autoimmune polyendocrinopathy with candidiasis and ectodermal dystrophy, see Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy

Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy

Autoimmune polyglandular syndrome, type 1, see Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy

Autoimmune thrombocytopenia, see Immune thrombocytopenia

Autoimmune thrombocytopenic purpura, see Immune thrombocytopenia

Autoimmune thyroiditis, see Hashimoto's disease

Autoimmunity-immunodeficiency syndrome, X-linked, see Immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome

Autoinflammation, lipodystrophy, and dermatosis syndrome, see Nakajo-Nishimura syndrome

Autoinflammation, panniculitis, and dermatosis syndrome, see Otulipenia

Autosomal dominant acute necrotizing encephalopathy, see Acute necrotizing encephalopathy type 1

Autosomal dominant adult-onset demyelinating leukodystrophy, see Autosomal dominant leukodystrophy with autonomic disease

Autosomal dominant cerebellar ataxia, deafness, and narcolepsy

Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome, see Autosomal dominant cerebellar ataxia, deafness, and narcolepsy

Autosomal dominant cerebrovascular amyloidosis, see Hereditary cerebral amyloid angiopathy

Autosomal dominant childhood-onset proximal spinal muscular atrophy with contractures, see Spinal muscular atrophy with lower extremity predominance

Autosomal dominant congenital stationary night blindness

Autosomal dominant craniometaphyseal dysplasia, see Craniometaphyseal dysplasia

Autosomal dominant epilepsy with auditory features

Autosomal dominant familial hematuria, retinal arteriolar tortuosity, contractures, see Hereditary angiopathy with nephropathy, aneurysms, and muscle cramps syndrome

Autosomal dominant familial periodic fever, see Tumor necrosis factor receptor-associated periodic syndrome

Autosomal dominant hereditary pancreatitis, see Hereditary pancreatitis

Autosomal dominant hereditary sensory radicular neuropathy, type 1A, see Hereditary sensory

neuropathy type IA

Autosomal dominant HIES, see Autosomal dominant hyper-IgE syndrome

Autosomal dominant hyaline body myopathy, see Myosin storage myopathy

Autosomal dominant hyper-IgE recurrent infection syndrome, see Autosomal dominant hyper-IgE syndrome

Autosomal dominant hyper-IgE syndrome

Autosomal dominant hyperimmunoglobulin E recurrent infection syndrome, see Autosomal dominant hyper-IgE syndrome

Autosomal dominant hypocalcemia

Autosomal dominant hypoparathyroidism, see Autosomal dominant hypocalcemia

Autosomal dominant intellectual disability 25, see Xia-Gibbs syndrome

Autosomal dominant intellectual disability-17, see PACS1 syndrome

Autosomal dominant interstitial kidney disease, see Medullary cystic kidney disease type 1

Autosomal dominant Job syndrome, see Autosomal dominant hyper-IgE syndrome

Autosomal dominant lateral temporal lobe epilepsy, see Autosomal dominant epilepsy with auditory features

Autosomal dominant leukodystrophy with autonomic disease

Autosomal dominant medullary cystic kidney disease, see Medullary cystic kidney disease type 1

Autosomal dominant mental retardation 35, see PPP2R5D-related intellectual disability

Autosomal dominant MYH9 spectrum disorders, see MYH9-related disorder

Autosomal dominant nocturnal frontal lobe epilepsy

Autosomal dominant Opitz G/BBB syndrome, see 22q11.2 deletion syndrome

Autosomal dominant optic atrophy, see Optic atrophy type 1

Autosomal dominant optic atrophy and cataract

Autosomal dominant optic atrophy Kjer type, see Optic atrophy type 1

Autosomal dominant optic atrophy type 3, see Autosomal dominant optic atrophy and cataract

Autosomal dominant partial epilepsy with auditory features, see Autosomal dominant epilepsy with auditory features

Autosomal dominant porencephaly type 1, see Familial porencephaly

Autosomal dominant spastic paraplegia 31, see Spastic paraplegia type 31

Autosomal dominant spastic paraplegia 8, see Spastic paraplegia type 8

Autosomal dominant vitreoretinochoroidopathy

Autosomal recessive axonal neuropathy with neuromyotonia

Autosomal recessive cerebellar ataxia type 1

Autosomal recessive cerebellar ataxia with mental retardation, see VLDLR-associated cerebellar hypoplasia

Autosomal recessive cerebellar hypoplasia with cerebral gyral simplification, see VLDLR-associated cerebellar hypoplasia

Autosomal recessive Charcot-Marie-Tooth disease type 2 with neuromyotonia, see Autosomal recessive axonal neuropathy with neuromyotonia

Autosomal recessive chronic granulomatous disease, see Chronic granulomatous disease

Autosomal recessive complete congenital stationary night blindness, see Autosomal recessive congenital stationary night blindness

Autosomal recessive congenital ichthyosis 4B, see Harlequin ichthyosis

Autosomal recessive congenital methemoglobinemia

Autosomal recessive congenital stationary night blindness

Autosomal recessive craniometaphyseal dysplasia, see Craniometaphyseal dysplasia

Autosomal recessive deafness-onychodystrophy syndrome, see DOORS syndrome

Autosomal recessive distal renal tubular acidosis with deafness, see Renal tubular acidosis with deafness

Autosomal recessive distal spinal muscular atrophy 1, see Spinal muscular atrophy with respiratory distress type 1

Autosomal recessive hereditary spastic paraplegia, see Troyer syndrome

Autosomal recessive HIES, see DOCK8 immunodeficiency syndrome

Autosomal recessive hyper-IgE syndrome, see DOCK8 immunodeficiency syndrome

Autosomal recessive hypotrichosis

Autosomal recessive incomplete congenital stationary night blindness, see Autosomal recessive congenital stationary night blindness

Autosomal recessive infantile hypercalcemia, see Idiopathic infantile hypercalcemia

Autosomal recessive infantile parkinsonism, see Tyrosine hydroxylase deficiency

Autosomal recessive Larsen syndrome, see CHST3-related skeletal dysplasia

Autosomal recessive localized hypotrichosis, see Autosomal recessive hypotrichosis

Autosomal recessive long QT syndrome (LQTS), see Jervell and Lange-Nielsen syndrome

Autosomal recessive neuromyotonia and axonal neuropathy, see Autosomal recessive axonal neuropathy with neuromyotonia

Autosomal recessive OPA3, see Costeff syndrome

Autosomal recessive optic atrophy 3, see Costeff syndrome

Autosomal recessive primary microcephaly

Autosomal recessive sensorineural hearing impairment, enlarged vestibular aqueduct, and goiter, see Pendred syndrome

Autosomal recessive spastic ataxia of Charlevoix-Saguenay

Autosomal recessive spastic paraplegia 15, see Spastic paraplegia type 15

Autosomal recessive spastic paraplegia 5A, see Spastic paraplegia type 5A

Autosomal recessive spastic paraplegia complicated with thin corpus callosum, see Spastic paraplegia type 11

Autosomal recessive spastic paraplegia type 49, see Spastic paraplegia type 49

Autosomal recessive spastic paraplegia with mental impairment and thin corpus callosum, see Spastic paraplegia type 11

Autosomal recessive spinocerebellar ataxia 8, see Autosomal recessive cerebellar ataxia type 1

Autosomal recessive T cell-negative, B cell-positive, NK cell-negative severe combined immunodeficiency, see JAK3-deficient severe combined immunodeficiency

Autosomal recessive T-B+NK- SCID, see JAK3-deficient severe combined immunodeficiency

Autosomal recessive woolly hair with or without hypotrichosis, see Autosomal recessive hypotrichosis

AUTS14A, see 16p11.2 deletion syndrome

AUTS14B, see 16p11.2 duplication

AVED, see Ataxia with vitamin E deficiency

AxD, see Alexander disease

Axenfeld and Rieger anomaly, see Axenfeld-Rieger syndrome

Axenfeld anomaly, see Axenfeld-Rieger syndrome

Axenfeld syndrome, see Axenfeld-Rieger syndrome

Axenfeld-Rieger syndrome

axial spondylarthritis, see Ankylosing spondylitis

AXRA, see Axenfeld-Rieger syndrome

AXRS, see Axenfeld-Rieger syndrome

Ayerza syndrome, see Pulmonary arterial hypertension

Azorean ataxia, see Spinocerebellar ataxia type 3

Azorean disease, see Spinocerebellar ataxia type 3