Complete Investigation list

Gene	Disease
AAAS	Triple-A syndrome (achalasia-addisonianism-alacrimia)
AARS1	Epileptic encephalopathy, early infantile, type 29
AARS2	Combined oxidative phosphorylation deficiency 8; Leukoencephalopathy, progressive, with ovarian failure
AASS	Hyperlysinemia, type 1 and type 2
ABAT	GABA-transaminase deficiency
ABCA1	Tangier disease
ABCA12	Ichthyosis congenital autosomal recessive type 4A and 4B (harlequin)
ABCA3	Surfactant metabolism dysfunction, pulmonary, type 3
ABCA4	Stargardt disease type 1; Cone-rod dystrophy type 3
ABCB11	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2
ABCB4	Cholestasis, progressive familial intrahepatic, type 3
ABCB7	Anemia, sideroblastic, with ataxia
ABCC2	Dubin-Johnson syndrome
ABCC6	Pseudoxanthoma elasticum; Generalized arterial calcification of infancy, type 2
ABCC8	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)
ABCD1	Adrenoleukodystrophy
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type
ABCG5	Sitosterolemia
ABCG8	Sitosterolemia
ABHD12	PHARC syndrome (polyneuropathy, hearing loss, ataxia, retinitis pigmentosa and cataract)
ABHD5	Chanarin-Dorfman syndrome
ACAD8	Isobutyryl-CoA dehydrogenase deficiency
ACAD9	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency
ACADS	Short-chain acyl-CoA dehydrogenase deficiency
ACADSB	Short/branched-chain acyl-CoA dehydrogenase deficiency (2-methylbuyrylglycinuria)
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
ACAT1	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)
ACE	Renal tubular dysgenesis
ACO2	Infantile cerebellar-retinal degeneration

Complete Investigation list

Gene	Disease
ACOX1	Peroxisomal acyl-CoA oxidase deficiency
ACOX2	Bile acid synthesis defect, congenital, type 6
ACP5	Spondyloenchondrodysplasia with immune dysregulation
ACSF3	Combined malonic and methylmalonic aciduria
ACSL4	Intellectual developmental disorder, X-linked 63
ACTA1	Nemaline myopathy 3; Congenital fiber-type disproportion myopathy 1
ACTN4	Glomerulosclerosis, focal segmental, 1
ACY1	Aminoacylase 1 deficiency
ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)
ADA2	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome
ADAM9	Cone-rod dystrophy 9
ADAMTS10	Weill-Marchesani syndrome, type 1 recessive
ADAMTS13	Thrombotic thrombocytopenic purpura, familial (Schulman-Upshaw syndrome)
ADAMTS17	Weill-Marchesani syndrome, type 4 recessive
ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type
ADAMTSL2	Geleophysic dysplasia type 1
ADAMTSL4	Ectopia lentis et pupillae; Ectopia lentis, isolated, type 2
ADAR	Aicardi-Goutieres syndrome, type 6
ADAT3	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies
ADCK3	Coenzyme Q10 deficiency, primary, 4
ADGRG1	Polymicrogyria, bilateral frontoparietal
ADGRG6	Lethal congenital contracture syndrome 9
ADGRV1	Usher syndrome, type 2C
ADK	Hypermethioninemia due to adenosine kinase deficiency
ADSL	Adenylosuccinase deficiency
ADSS1	Myopathy, distal, 5
AFF2	Intellectual developmental disorder, X-linked 109
AFG3L2	Spastic ataxia, type 5 autosomal recessive
AFP	Alpha-fetoprotein deficiency
AGA	Aspartylglucosaminuria (glycosylasparaginase deficiency)
AGBL5	Retinitis pigmentosa 75
AGK	Cataract 38; Sengers syndrome

Complete Investigation list

Gene	Disease
AGL	Glycogen storage disease, type 3
AGPAT2	Congenital generalized lipodystrophy (Berardinelli-Seip syndrome)
AGPS	Rhizomelic chondrodysplasia punctata, type 3
AGRN	Myasthenic syndrome, congenital, type 8
AGT	Renal tubular dysgenesis
AGTR1	Renal tubular dysgenesis
AGXT	Hyperoxaluria, primary, type 1
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
AHI1	Joubert syndrome, type 3
AICDA	Immunodeficiency with hyper-IgM, type 2
AIMP1	Leukodystrophy, hypomyelinating, type 3
AIMP2	Leukodystrophy, hypomyelinating, type 17
AIPL1	Leber congenital amaurosis, type 4
AIRE	Autoimmune polyendocrinopathy syndrome, type 1
AK1	Hemolytic anemia due to adenylate kinase deficiency
AK2	Reticular dysgenesis
AKR1C2	46,XY disorder of sex development due to testicular 17,20-desmolase deficiency
AKR1D1	Bile acid synthesis defect, congenital, type 2
ALAD	Porphyria, acute hepatic
ALAS2	Anemia, sideroblastic, 1
ALB	Analbuminemia
ALDH18A1	Spastic paraplegia, type 9B, autosomal recessive; Cutis laxa, type 3A (De Barsy syndrome)
ALDH1A3	Microphthalmia, isolated 8
ALDH3A2	Sjogren-Larsson syndrome
ALDH4A1	Hyperprolinemia, type 2
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency
ALDH7A1	Epilepsy, pyridoxine-dependent
ALDOA	Glycogen storage disease type 12
ALDOB	Fructose intolerance, hereditary
ALG1	Congenital disorder of glycosylation, type 1K
ALG11	Congenital disorder of glycosylation, type 1P
ALG12	Congenital disorder of glycosylation, type 1G
ALG2	Myasthenic syndrome, congenital, type 14 with tubular aggregates

Complete Investigation list

Gene	Disease
ALG3	Congenital disorder of glycosylation, type 1D
ALG6	Congenital disorder of glycosylation, type 1C
ALG8	Congenital disorder of glycosylation, type 1H
ALG9	Congenital disorder of glycosylation, type 1L; Gillessen-Kaesbach-Nishimura syndrome
ALMS1	Alström syndrome
ALOX12B	Ichthyosis, congenital, autosomal recessive, type 2
ALOXE3	Ichthyosis, congenital, autosomal recessive, type 3
ALPK3	Cardiomyopathy, familial hypertrophic, type 27
ALPL	Hypophosphatasia, infantile/childhood
ALS2	Amyotrophic lateral sclerosis, type 2 juvenile; Primary lateral sclerosis, juvenile; Spastic paralysis, infantile onset ascending
ALX1	Frontonasal dysplasia, type 3
ALX3	Frontonasal dysplasia, type 1
ALX4	Frontonasal dysplasia, type 2
AMACR	Bile acid synthesis defect, congenital, type 4; Alpha-methylacyl-CoA racemase deficiency
AMBN	Amelogenesis imperfecta, type IF
AMH	Persistent Mullerian duct syndrome, type 1
AMHR2	Persistent Mullerian duct syndrome, type II
AMN	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)
AMPD1	Myopathy due to myoadenylate deaminase deficiency
AMPD2	Pontocerebellar hypoplasia, type 9
AMT	Glycine encephalopathy
ANGPTL3	Hypobetalipoproteinemia, familial, type 2
ANKS6	Nephronophthisis 16
ANO10	Spinocerebellar ataxia, autosomal recessive, type 10
ANO5	Limb-girdle muscular dystrophy, type 12 (LGMD R12)
ANTXR1	GAPO syndrome
ANTXR2	Hyaline fibromatosis syndrome
AP1S1	MEDNIK syndrome
AP1S2	Pettigrew syndrome
AP3B1	Hermansky-Pudlak syndrome, type 2
AP3B2	Epileptic encephalopathy, early infantile, type 48
AP3D1	Hermansky-Pudlak syndrome, type 10
AP4B1	Spastic paraplegia, type 47 autosomal recessive
AP4E1	Spastic paraplegia, type 51 autosomal recessive

Complete Investigation list

Gene	Disease
AP4M1	Spastic paraplegia, type 50 autosomal recessive
AP4S1	Spastic paraplegia, type 52 autosomal recessive
AP5Z1	Spastic paraplegia, type 48 autosomal recessive
APOC2	Hyperlipoproteinemia, type 1B
APOE	Sea-blue histiocyte disease
APRT	Adenine phosphoribosyltransferase deficiency
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia
AQP2	Diabetes insipidus, nephrogenic, type 2
AR	Androgen insensitivity syndrome, complete
ARFGEF2	Periventricular heterotopia with microcephaly
ARG1	Argininemia (arginase deficiency)
ARHGDIA	Nephrotic syndrome, type 8
ARHGEF18	Retinitis pigmentosa 78
ARHGEF9	Developmental and epileptic encephalopathy 8
ARL13B	Joubert syndrome type 8
ARL2BP	Retinitis pigmentosa with or without situs inversus
ARL6	Bardet-Biedl syndrome, type 3
ARMC9	Joubert syndrome 30
ARPC1B	Immunodeficiency, type 71 with inflammatory disease and congenital thrombocytopenia
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)
ARSE	Chondrodysplasia punctata, X-linked recessive
ARSL	Chondrodysplasia punctata, brachytelephalangic
ARV1	Epileptic encephalopathy, early infantile, 38
ARX	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders
ASAH1	Farber lipogranulomatosis; Spinal muscular atrophy with progressive myoclonic epilepsy
ASL	Argininosuccinic aciduria
ASNS	Asparagine synthetase deficiency
ASPA	Canavan disease
ASPH	Traboulsi syndrome
ASPM	Primary microcephaly type 5 autosomal recessive
ASS1	Citrullinemia, type 1
ATAD1	Hyperekplexia 4
ATF6	Achromatopsia, type 7

Complete Investigation list

Gene	Disease
ATIC	AICA-ribosiduria due to ATIC deficiency
ATM	Ataxia-telangiectasia
ATOH7	Persistent hyperplastic primary vitreous, autosomal recessive
ATP13A2	Kufor-Rakeb syndrome; Spastic paraplegia, type 78 autosomal recessive
ATP2A1	Brody myopathy
ATP6AP2	Parkinsonism with spasticity, X-linked
ATP6V0A2	Cutis laxa, autosomal recessive, type 2A; Wrinkly skin syndrome
ATP6V0A4	Renal tubular acidosis, distal, autosomal recessive
ATP6V1A	Cutis laxa, autosomal recessive, type 2D
ATP6V1B1	Renal tubular acidosis with deafness
ATP6V1E1	Cutis laxa, autosomal recessive, type 2C
ATP7A	Menkes disease; Occipital horn syndrome
ATP7B	Wilson disease
ATP8B1	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1
ATR	Seckel syndrome, type 1
ATRX	Intellectual disability-hypotonic facies syndrome, X-linked; Alpha-thalassemia/intellectual developmental disorder syndrome
AUH	3-methylglutaconic aciduria, type 1
AURKC	Spermatogenic failure, type 5
AVIL	Nephrotic syndrome, type 21
AVPR2	Diabetes insipidus, nephrogenic, 1; Nephrogenic syndrome of inappropriate antidiuresis
B2M	Immunodeficiency, type 43
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11
B3GALT6	Ehlers-Danlos syndrome, spondylodysplastic type, 2
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects
B3GLCT	Peters-plus syndrome
B4GALNT1	Spastic paraplegia, type 26 autosomal recessive
B4GALT1	Congenital disorder of glycosylation, type 2D
B4GALT7	Ehlers-Danlos syndrome, spondylodysplastic, type 1
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13
B9D1	Joubert syndrome, type 27; ?Meckel syndrome 9
B9D2	Joubert syndrome, type 34; ?Meckel syndrome, type 10

Complete Investigation list

Gene	Disease
BBS1	Bardet-Biedl syndrome, type 1
BBS10	Bardet-Biedl syndrome, type 10
BBS12	Bardet-Biedl syndrome, type 12
BBS2	Bardet-Biedl syndrome, type 2
BBS4	Bardet-Biedl syndrome, type 4
BBS5	Bardet-Biedl syndrome, type 5
BBS7	Bardet-Biedl syndrome, type 7
BBS9	Bardet-Biedl syndrome, type 9
BCAT2	Hypervalinemia or hyperleucine-isoleucinemia
BCHE	Butyrylcholinesterase deficiency
BCKDHA	Maple syrup urine disease, type 1A
BCKDHB	Maple syrup urine disease, type 1B
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency
BCL10	Immunodeficiency, type 37
BCOR	Microphthalmia, syndromic 2
BCS1L	Mitochondrial complex III deficiency nuclear type 1; GRACILE syndrome; Bjornstad syndrome
BEST1	Bestrophinopathy, AR
BFSP1	Cataract 33 multiple types
BHLHA9	Syndactyly, mesoaxial synostotic, with phalangeal reduction
BIN1	Centronuclear myopathy, type 2
BLM	Bloom syndrome
BLNK	Agammaglobulinemia 4
BLOC1S3	Hermansky-Pudlak syndrome, type 8
BLOC1S6	Hermansky-Pudlak syndrome, type 9
BLVRA	Hyperbiliverdinemia
BMP1	Osteogenesis imperfecta, type 13
BMPER	Diaphanospondylodysostosis
BMPR1B	Acromesomelic dysplasia, Demirhan type
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia
BPGM	Erythrocytosis due to bisphosphoglycerate mutase deficiency
BPNT2	Chondrodysplasia with joint dislocations, GPAPP type
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal; Neurodevelopmental disorder with cerebellar atrophy and with or without seizures
BRCA2	Fanconi anemia, complementation group D1
BRF1	Cerebellofaciodental syndrome

Complete Investigation list

Gene	Disease
BRIP1	Fanconi anemia, complementation group J
BRWD3	Intellectual developmental disorder, X-linked 93
BSCL2	Congenital generalized lipodystrophy, type 2; Encephalopathy, progressive, with or without lipodystrophy
BSND	Bartter syndrome, type 4A
BTD	Biotinidase deficiency
BTK	Agammaglobulinemia X-linked, type 1
BUB1B	Mosaic variegated aneuploidy syndrome 1
C10orf2	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)
C12orf57	Temtamy syndrome
C12orf65	Combined oxidative phosphorylation deficiency, type 7; Spastic paraplegia, type 55, autosomal recessive
C19orf12	Neurodegeneration with brain iron accumulation, type 4
C1QA	C1q deficiency
C1QB	C1q deficiency
C1QBP	Combined oxidative phosphorylation deficiency 33
C1QC	C1q deficiency
C1S	C1s deficiency
C2	C2 deficiency
C2CD3	Orofaciodigital syndrome, type 14
C3	Complement component 3 deficiency
C5	Complement component 5 deficiency
C6	Complement component 6 deficiency
C7	Complement component 7 deficiency
C8B	Complement component 8 deficiency, type 2
C8orf37	Bardet-Biedl syndrome, type21
CA12	Hyperchlorhidrosis, isolated
CA2	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency
CA8	Cerebellar ataxia, impaired intellectual development and dysequilibrium syndrome 3
CABP2	Deafness, autosomal recessive, type 93
CABP4	Congenital stationary night blindness, type 2B
CACNA1D	Sinoatrial node dysfunction and deafness
CACNA2D4	Retinal cone dystrophy 4
CAD	Epileptic encephalopathy, early infantile, 50

Complete Investigation list

Gene	Disease
CALCRL	Lymphatic malformation 8
CANT1	Desbuquois dysplasia, type 1; Epiphyseal dysplasia, multiple, type 7
CAPN1	Spastic paraplegia, type 76 autosomal recessive
CAPN3	Limb-girdle muscular dystrophy, type 1 (LGMD R1)
CARD11	Immunodeficiency, type 11A
CARD9	Candidiasis, familial, type 2 autosomal recessive
CARS2	Combined oxidative phosphorylation deficiency 27
CASK	FG syndrome 4
CASP14	Ichthyosis, congenital, autosomal recessive 12
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, type 2
CASR	Hyperparathyroidism, neonatal
CAST	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads
CAT	Acatalasemia
CATSPER1	Spermatogenic failure, type 7
CAVIN1	Lipodystrophy, congenital generalized, type 4
CBLIF	Intrinsic factor deficiency
CBS	Homocystinuria due to cystathionine beta-synthase
CC2D1A	Intellectual developmental disorder, autosomal recessive 3
CC2D2A	Joubert syndrome, type 9; Meckel syndrome, type 6; COACH syndrome, 2
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome, type 1
CCDC103	Ciliary dyskinesia, primary, type 17
CCDC115	Congenital disorder of glycosylation, type IIo
CCDC174	Hypotonia, infantile, with psychomotor retardation
CCDC39	Ciliary dyskinesia, primary, type 14
CCDC40	Ciliary dyskinesia, primary, type 15
CCDC65	Ciliary dyskinesia, primary, type 27
CCDC8	3M syndrome 3
CCDC88C	Hydrocephalus, congenital, type 1
CCN6	Arthropathy, progressive pseudorheumatoid, of childhood
CCNO	Ciliary dyskinesia, primary, type 29
CD19	Immunodeficiency, common variable, type 3
CD247	Immunodeficiency, type 25
CD27	Lymphoproliferative syndrome 2
CD2AP	Glomerulosclerosis, focal segmental, type 3 susceptibility to
CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect

Complete Investigation list

Gene	Disease
CD36	Platelet glycoprotein 4 deficiency
CD3D	Immunodeficiency, type 19
CD3E	Immunodeficiency, type 18
CD3G	Immunodeficiency, type 17 CD3 gamma deficient
CD40	Immunodeficiency with hyper-IgM, type 3
CD40LG	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)
CD55	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy (CHAPLE)
CD59	CD59 deficiency
CD79A	Agammaglobulinemia 3
CD79B	Agammaglobulinemia 6
CD81	Immunodeficiency, common variable, type 6
CD8A	CD8 deficiency, familial
CDAN1	Dyserythropoietic anemia, congenital, type 1A
CDC14A	Deafness, autosomal recessive, type 105
CDC45	Meier-Gorlin syndrome 7
CDCA7	Immunodeficiency-centromeric instability-facial anomalies syndrome 3
CDH11	Elsahy-Waters syndrome
CDH23	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy
CDHR1	Cone-rod dystrophy, type 15
CDIN1	Dyserythropoietic anemia, congenital, type Ib
CDK10	Al Kaissi syndrome
CDK5RAP2	Primary microcephaly type 3 autosomal recessive
CDKL5	Developmental and epileptic encephalopathy 2
CDSN	Peeling skin syndrome 1
CDT1	Meier-Gorlin syndrome, type 4
CEBPE	Specific granule deficiency
CENPF	Stromme syndrome
CENPJ	Primary microcephaly type 6 autosomal recessive
CEP104	Joubert syndrome 25
CEP120	Short-rib thoracic dysplasia 13 with or without polydactyly
CEP135	Microcephaly 8 primary, autosomal recessive
CEP152	Primary microcephaly type 9 autosomal recessive
CEP164	Nephronophthisis 15
CEP19	Morbid obesity and spermatogenic failure

Complete Investigation list

Gene	Disease
CEP290	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10
CEP41	Joubert syndrome, type 15
CEP55	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly
CEP57	Mosaic variegated aneuploidy syndrome 2
CEP78	Cone-rod dystrophy and hearing loss
CEP83	Nephronophthisis 18
CERKL	Retinitis pigmentosa, type 26
CERS3	Ichthyosis, congenital, autosomal recessive 9
CFAP43	Spermatogenic failure, type 19
CFAP53	Heterotaxy, visceral, 6 autosomal recessive
CFD	Complement factor D deficiency
CFH	Complement factor H deficiency
CFI	Complement factor I deficiency
CFL2	Nemaline myopathy, type 7 autosomal recessive
CFP	Properdin deficiency, X-linked
CFTR	Cystic fibrosis
CHAT	Myasthenic syndrome, congenital, type 6 presynaptic
CHKB	Muscular dystrophy, congenital, megaconial type
CHM	Choroideremia
CHMP1A	Pontocerebellar hypoplasia, type 8
CHRNA1	Multiple pterygium syndrome, lethal type
CHRNB1	Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency
CHRND	Myasthenic syndrome, congenital, type 3B, fast-channel; Multiple pterygium syndrome, lethal type
CHRNE	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency
CHRNG	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type
CHST14	Ehlers-Danlos syndrome, musculocontractural, type 1
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations
CHST6	Macular corneal dystrophy
CHSY1	Temtamy preaxial brachydactyly syndrome
CHUK	Cocoon syndrome
CIB2	Deafness, autosomal recessive, type 48; Usher syndrome, type 1J

Complete Investigation list

Gene	Disease
CIITA	Bare lymphocyte syndrome, type 2 complementation group A
CILK1	Endocrine-cerebroosteodysplasia
CISD2	Wolfram syndrome 2
CIT	Microcephaly 17 primary, autosomal recessive
CKAP2L	Filippi syndrome
CLCF1	Cold-induced sweating syndrome 2
CLCN1	Myotonia congenita, recessive
CLCN2	Leukoencephalopathy with ataxia
CLCN5	Dent disease 1
CLCN7	Osteopetrosis, autosomal recessive type 4
CLCNKA	Bartter syndrome, type 4B, digenic
CLCNKB	Bartter syndrome, type 3; Bartter syndrome, type 4B, digenic
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis
CLDN10	HELIX syndrome
CLDN14	Deafness type 29 autosomal recessive
CLDN16	Hypomagnesemia, type 3 renal
CLDN19	Rena hypomagnesemia type 5 with ocular involvement
CLMP	Congenital short bowel syndrome
CLN3	Ceroid lipofuscinosis, neuronal, type 3
CLN5	Ceroid lipofuscinosis, neuronal, type 5
CLN6	Ceroid lipofuscinosis, neuronal, type 6
CLN8	Ceroid lipofuscinosis, neuronal, type 8
CLP1	Pontocerebellar hypoplasia, type 10
CLPB	3-methylglutaconic aciduria, type 7 with cataracts, neurologic involvement and neutropenia
CLPP	Perrault syndrome 3
CLRN1	Usher syndrome, type 3A
CNGA1	Retinitis pigmentosa type 49
CNGA3	Achromatopsia, type 2
CNGB1	Retinitis pigmentosa type 45
CNGB3	Achromatopsia, type 3
CNNM2	Hypomagnesemia, seizures, and intellectual developmental disorder
CNNM4	Jalili syndrome
CNPY3	Epileptic encephalopathy, early infantile, type 60
CNTNAP1	Lethal congenital contracture syndrome 7
CNTNAP2	Pitt-Hopkins like syndrome 1

Complete Investigation list

Gene	Disease
COA6	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4
COA8	Mitochondrial complex IV deficiency, nuclear type 17
COASY	Neurodegeneration with brain iron accumulation 6
COG1	Congenital disorder of glycosylation, type IIg
COG4	Congenital disorder of glycosylation, type 2J
COG5	Congenital disorder of glycosylation, type 2I
COG6	Congenital disorder of glycosylation, type 2L; Shaheen syndrome
COG7	Congenital disorder of glycosylation, type 2E
COG8	Congenital disorder of glycosylation, type 2H
COL11A1	Fibrochondrogenesis type 1
COL11A2	Otospondylomegaepiphyseal dysplasia, autosomal recessive
COL13A1	Myasthenic syndrome, congenital, 19
COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type
COL18A1	Knobloch syndrome, type 1
COL25A1	Fibrosis of extraocular muscles, congenital, type 5
COL27A1	Steel syndrome
COL4A3	Alport syndrome, autosomal recessive, type 2
COL4A4	Alport syndrome, autosomal recessive, type 2
COL4A5	Alport syndrome, X-linked
COL6A1	Ullrich congenital muscular dystrophy, type 1 (Limb-girdle muscular dystrophy, type 22 [LGMD R22])
COL6A2	Ullrich congenital muscular dystrophy, type 1 (Limb-girdle muscular dystrophy, type 22 [LGMD R22])
COL6A3	Ullrich congenital muscular dystrophy, type 1 (Limb-girdle muscular dystrophy, type 22 [LGMD R22])
COL7A1	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial
COL9A1	Stickler syndrome, type 4
COL9A2	Stickler syndrome, type V
COLEC10	3MC syndrome 3
COLEC11	3MC syndrome 2
COLQ	Myasthenic syndrome, congenital, type 5
COQ2	Primary coenzyme Q10 deficiency, type 1
COQ4	Coenzyme Q10 deficiency, primary, type 7
COQ6	Coenzyme Q10 deficiency, primary, type 6
COQ8A	Primary coenzyme Q10 deficiency, type 4

Complete Investigation list

Gene	Disease
COQ8B	Nephrotic syndrome, type 9
COQ9	Coenzyme Q10 deficiency, primary, type 5
CORO1A	Immunodeficiency, type 8
COX10	Mitochondrial complex IV deficiency, nuclear type 3
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 2; Leigh syndrome due to cytochrome c oxidase deficiency
COX20	Mitochondrial complex IV deficiency, nuclear type 11
COX6B1	Mitochondrial complex IV deficiency, nuclear type 7
CP	Aceruloplasminemia
CPA6	Febrile seizures, familial, type 11
CPAMD8	Anterior segment dysgenesis, type 8
CPLANE1	Joubert syndrome 17
CPLX1	Epileptic encephalopathy, early infantile, 63
CPS1	Carbamoylphosphate synthetase 1 deficiency
CPT1A	Carnitine palmitoyltransferase type 1A deficiency, hepatic
CPT2	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile
CR2	Immunodeficiency, common variable, type 7
CRADD	intellectual developmental disorder, autosomal recessive, type 34 with variant lissencephaly
CRB1	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8
CRB2	Ventriculomegaly with cystic kidney disease
CRBN	intellectual developmental disorder, autosomal recessive, type 2
CRIPT	Short stature with microcephaly and distinctive facies
CRLF1	Cold-induced sweating syndrome type 1
CRPPA	Muscular dystrophy-dystroglycanopathy, type A7; Muscular dystrophy-dystroglycanopathy, type C7
CRTAP	Osteogenesis imperfecta, type 7
CRYAA	Cataract 9 multiple types
CRYAB	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related; Cataract 16 multiple types
CRYBB1	Cataract 17
CRYBB3	Cataract 22
CSF2RB	Surfactant metabolism dysfunction, pulmonary, type 5
CSF3R	Neutropenia, severe congenital, type 7 autosomal recessive
CSPP1	Joubert syndrome 21

Complete Investigation list

Gene	Disease
CSTA	Peeling skin syndrome, type 4
CSTB	Epilepsy, progressive myoclonic type 1A (Unverricht and Lundborg)
CTC1	Cerebroretinal microangiopathy with calcifications and cysts
CTH	Cystathioninuria
CTNS	Nephropathic cystinosis
CTPS1	Immunodeficiency, type 24
CTSA	Galactosialidosis
CTSC	Haim-Munk syndrome; Papillon-Lefevre syndrome
CTSD	Ceroid lipofuscinosis, neuronal, type 10
CTSF	Ceroid lipofuscinosis, neuronal, type 13 (Kufs type)
CTSK	Pycnodysostosis
CUBN	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)
CUL4B	Intellectual developmental disorder, X-linked syndromic, Cabezas type
CUL7	3M syndrome 1
CWC27	Retinitis pigmentosa with or without skeletal anomalies
CWF19L1	Spinocerebellar ataxia, autosomal recessive, type 17
CYB5A	46,XY disorder of sex development due to isolated 17,20-lyase deficiency
CYB5R3	Methemoglobinemia, type 1; Methemoglobinemia, type 2
CYBA	Chronic granulomatous disease, type 4
CYBB	Chronic granulomatous disease, X-linked
CYC1	Mitochondrial complex III deficiency, nuclear type 6
CYP11A1	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
CYP11B2	Hypoaldosteronism, congenital, due to CMO I deficiency
CYP17A1	17 alpha-hydroxylase/17,20-lyase deficiency
CYP19A1	Aromatase deficiency
CYP1B1	Glaucoma, primary congenital, type 3A
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
CYP24A1	Hypercalcemia, infantile, type 1
CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies
CYP26C1	Focal facial dermal dysplasia 4
CYP27A1	Cerebrotendinous xanthomatosis
CYP27B1	Vitamin D-dependent rickets, type 1
CYP2R1	Rickets due to defect in vitamin D 25-hydroxylation
CYP2U1	Spastic paraplegia, type 56 autosomal recessive

Complete Investigation list

Gene	Disease
CYP4F22	Ichthyosis, congenital, autosomal recessive, type 5
CYP4V2	Bietti crystalline corneoretinal dystrophy
CYP7B1	Spastic paraplegia, type 5A, autosomal recessive
D2HGDH	D-2-hydroxyglutaric aciduria
DAG1	Muscular dystrophy-dystroglycanopathy type A9; Muscular dystrophy-dystroglycanopathy type C9
DARS1	Hypomyelination with brainstem and spinal cord involvement and leg spasticity
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation
DBH	Dopamine beta-hydroxylase deficiency
DBT	Maple syrup urine disease, type 2
DCAF17	Woodhouse-Sakati syndrome
DCC	Gaze palsy, familial horizontal, with progressive scoliosis, type 2
DCDC2	Sclerosing cholangitis, neonatal; Nephronophthisis 19
DCHS1	Van Maldergem syndrome 1
DCLRE1C	Omenn syndrome; Severe combined immunodeficiency, Athabascan type
DCPS	Al-Raqad syndrome
DCX	Lissencephaly, X-linked, type 1
DDB2	Xeroderma pigmentosum, complementation group E
DDC	Aromatic L-amino acid decarboxylase deficiency
DDHD1	Spastic paraplegia, type 28 autosomal recessive
DDHD2	Spastic paraplegia, type 54 autosomal recessive
DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type
DDRGK1	Spondyloepimetaphyseal dysplasia, Shohat type
DDX11	Warsaw breakage syndrome
DDX59	Orofaciodigital syndrome V
DENND5A	Epileptic encephalopathy, early infantile, 49
DES	Myopathy, myofibrillar, type 1
DGAT1	Diarrhea 7 protein-losing enteropathy type
DGKE	Nephrotic syndrome, type 7
DGUOK	DGUOK-related mitochondrial DNA depletion syndrome
DHCR24	Desmosterolosis
DHCR7	Smith-Lemli-Opitz syndrome
DHDDS	Retinitis pigmentosa, type 59
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency
DHH	46,XY complete gonadal dysgenesis

Complete Investigation list

Gene	Disease
DHODH	Miller syndrome
DHPS	Neurodevelopmental disorder with seizures and speech and walking impairment
DHTKD1	2-aminoadipic 2-oxoadipic aciduria
DIAPH1	Seizures, cortical blindness, microcephaly syndrome
DIS3L2	Perlman syndrome
DKC1	Dyskeratosis congenita, X-linked
DLAT	Pyruvate dehydrogenase E2 deficiency
DLD	Dihydrolipoamide dehydrogenase deficiency
DLG3	Intellectual developmental disorder, X-linked 90
DLL3	Spondylocostal dysostosis type 1
DMD	Duchenne/Becker muscular dystrophy
DMGDH	Dimethylglycine dehydrogenase deficiency
DMP1	Hypophosphatemic rickets, autosomal recessive
DMXL2	Developmental and epileptic encephalopathy, type 81
DNAAF1	Ciliary dyskinesia, primary, type 13
DNAAF2	Ciliary dyskinesia, primary, type 10
DNAAF3	Ciliary dyskinesia, primary, type 2
DNAAF4	Ciliary dyskinesia, primary, type 25
DNAAF5	Ciliary dyskinesia, primary, type 18
DNAH1	Spermatogenic failure, type 18
DNAH11	Ciliary dyskinesia, primary, type 7 with or without situs inversus
DNAH5	Ciliary dyskinesia, primary, type 3 with or without situs inversus
DNAH9	Ciliary dyskinesia, primary, type 40
DNAI1	Ciliary dyskinesia, primary, type 1 with or without situs inversus
DNAI2	Ciliary dyskinesia, primary, type 9 with or without situs inversus
DNAJB13	Ciliary dyskinesia, primary, type 34
DNAJB2	Spinal muscular atrophy, distal, autosomal recessive, type 5
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient
DNAJC19	3-methylglutaconic aciduria, type 5
DNAJC21	Bone marrow failure syndrome, type 3
DNAJC6	Parkinson disease, type 19A, juvenile-onset; Parkinson disease, type 19B, early-onset
DNAL1	Ciliary dyskinesia, primary, type 16
DNASE1L3	Systemic lupus erythematosus 16
DNM1L	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 1

Complete Investigation list

Gene	Disease
DNM2	Lethal congenital contracture syndrome, type 5
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 1
DOCK2	Immunodeficiency, type 40
DOCK6	Adams-Oliver syndrome 2
DOCK7	Epileptic encephalopathy, early infantile, 23
DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive
DOK7	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10
DOLK	Congenital disorder of glycosylation, type 1M
DONSON	Microcephaly, short stature, and limb abnormalities
DPAGT1	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13
DPH1	Developmental delay with short stature, dysmorphic features, and sparse hair
DPM1	Congenital disorder of glycosylation, type 1E
DPM2	Congenital disorder of glycosylation, type Iu
DPM3	Congenital disorder of glycosylation, type Io
DPY19L2	Spermatogenic failure, type 9
DPYD	Dihydropyrimidine dehydrogenase deficiency
DPYS	Dihydropyrimidinuria
DRAM2	Cone-rod dystrophy 21
DRC1	Ciliary dyskinesia, primary, type 21
DSG1	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE
DSG4	Hypotrichosis, type 6
DSP	Cardiomyopathy, dilated, with woolly hair and keratoderma; Epidermolysis bullosa, lethal acantholytic
DST	Epidermolysis bullosa simplex, autosomal recessive, type 2
DSTYK	Spastic paraplegia, type 23 autosomal recessive
DTNBP1	Hermansky-Pudlak syndrome, type 7
DUOX2	Thyroid dyshormonogenesis, type 6
DUOXA2	Thyroid dyshormonogenesis, type 5
DYM	Smith-McCort dysplasia; Dyggve-Melchior-Clausen disease
DYNC2H1	Short-rib thoracic dysplasia, type 3 with or without polydactyly
DYNC2I1	Short-rib thoracic dysplasia 8 with or without polydactyly
DYNC2I2	Short-rib thoracic dysplasia 11 with or without polydactyly

Complete Investigation list

Gene	Disease
DYNC2LI1	Short-rib thoracic dysplasia 15 with polydactyly
DYNLT2B	Short-rib thoracic dysplasia 17 with or without polydactyly
DYSF	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)
DZIP1L	Polycystic kidney disease 5
EARS2	Combined oxidative phosphorylation deficiency 12
ECEL1	Arthrogryposis, distal, type 5D
ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency
ECM1	Urbach-Wiethe disease
EDA	Ectodermal dysplasia, type 1 hypohidrotic, X-linked
EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type
EDARADD	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type
EDN1	Auriculocondylar syndrome, type 3
EDN3	Waardenburg syndrome, type 4B
EDNRB	ABCD syndrome
EFEMP2	Cutis laxa, autosomal recessive, type 1B
EFL1	Shwachman-Diamond syndrome 2
EGFR	Inflammatory skin and bowel disease, neonatal, 2
EGR2	Dejerine-Sottas disease
EIF2AK3	Wolcott-Rallison syndrome
EIF2AK4	Pulmonary venoocclusive disease 2
EIF2B1	Leukoencephalopathy with vanishing white matter (VWM)
EIF2B2	Leukoencephalopathy with vanishing white matter (VWM)
EIF2B3	Leukoencephalopathy with vanishing white matter (VWM)
EIF2B4	Leukoencephalopathy with vanishing white matter (VWM)
EIF2B5	Leukoencephalopathy with vanishing white matter (VWM)
EIF4A3	Robin sequence with cleft mandible and limb anomalies
ELAC2	Combined oxidative phosphorylation deficiency 17
ELMO2	Vascular malformation, primary intraosseous
ELOVL4	Ichthyosis, spastic quadriplegia, and intellectual developmental disorder
ELP2	intellectual developmental disorder, autosomal recessive, type 58
EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation
EMD	Emery-Dreifuss muscular dystrophy, type 1 X-linked
EML1	Band heterotopia
EMP2	Nephrotic syndrome, type 10
ENAM	Amelogenesis imperfecta, type 1C
ENO3	Glycogen storage disease XIII

Complete Investigation list

Gene	Disease
ENPP1	Arterial calcification, generalized, of infancy, type 1
ENTPD1	Spastic paraplegia, type 64 autosomal recessive
EOGT	Adams-Oliver syndrome 4
EPB41	Elliptocytosis, type 1
EPB42	Spherocytosis, type 5
EPCAM	Diarrhea 5 with tufting enteropathy, congenital
EPG5	Vici syndrome
EPM2A	Epilepsy, progressive myoclonic, type 2A (Lafora)
EPRS1	Leukodystrophy, hypomyelinating, type 15
EPS8L2	Deafness autosomal recessive, type 106
ERAL1	Perrault syndrome 6
ERBB3	Lethal congenital contractural syndrome, type 2
ERCC1	Cerebrooculofacioskeletal syndrome, type 4
ERCC2	Trichothiodystrophy, type 1
ERCC3	Trichothiodystrophy, type 2
ERCC4	Fanconi anemia, complementation group Q
ERCC5	Cerebrooculofacioskeletal syndrome, type 3
ERCC6	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1
ERCC6L2	Bone marrow failure syndrome, type 2
ERCC8	Cockayne syndrome, type A
ERLIN1	Spastic paraplegia, type 62 autosomal recessive
ERLIN2	Spastic paraplegia, type 18 autosomal recessive
ESCO2	Roberts syndrome; Juberg-Hayward syndrome
ESPN	Deafness, autosomal recessive, type 36
ESR1	Estrogen resistance
ESRRB	Deafness, autosomal recessive, type 35
ETFA	Glutaric acidemia, type 2A
ETFB	Glutaric acidemia, type 2B
ETFDH	Glutaric acidemia, type 2C
ETHE1	Ethylmalonic encephalopathy
EVC	Ellis-van Creveld syndrome
EVC2	Ellis-van Creveld syndrome
EXOSC3	Pontocerebellar hypoplasia, type 1B
EXPH5	Epidermolysis bullosa, nonspecific, autosomal recessive
EXT1	Chondrosarcoma
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities
EYS	Retinitis pigmentosa, type 25

Complete Investigation list

Gene	Disease
F10	Factor X deficiency
F11	Factor XI deficiency
F13A1	Factor XIIIA deficiency
F13B	Factor XIIIB deficiency
F2	Prothrombin deficiency
F5	Factor V deficiency
F7	Factor VII deficiency
F8	Hemophilia A
F9	Hemophilia B
FA2H	Spastic paraplegia, type 35 autosomal recessive
FADD	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations
FAH	Tyrosinemia, type 1
FAM126A	Leukodystrophy, hypomyelinating, type 5
FAM161A	Retinitis pigmentosa, type 28
FAM20A	Amelogenesis imperfecta, type 1G (Enamel-renal syndrome)
FAM20C	Raine syndrome
FAN1	Interstitial nephritis, karyomegalic
FANCA	Fanconi anemia, complementation group A
FANCB	Fanconi anemia, complementation group B
FANCC	Fanconi anemia, complementation group C
FANCD2	Fanconi anemia, complementation group D2
FANCE	Fanconi anemia, complementation group E
FANCF	Fanconi anemia, complementation group F
FANCG	Fanconi anemia, complementation group G
FANCI	Fanconi anemia, complementation group I
FANCL	Fanconi anemia, complementation group L
FANCM	Spermatogenic failure, type 28
FAR1	Peroxisomal fatty acyl-CoA reductase 1 disorder
FARS2	Combined oxidative phosphorylation deficiency 14; Spastic paraplegia, type 77 autosomal recessive
FASTKD2	Combined oxidative phosphorylation deficiency 44
FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2
FBLN5	Cutis laxa, autosomal recessive, type 1A
FBP1	Fructose-1,6-bisphosphatase deficiency
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)
FBXO7	Parkinson disease, type 15 autosomal recessive

Complete Investigation list

Gene	Disease
FDXR	Auditory neuropathy and optic atrophy
FECH	Protoporphyria, erythropoietic, autosomal recessive
FERMT1	Kindler syndrome
FERMT3	Leukocyte adhesion deficiency, type 3
FEZF1	Hypogonadotropic hypogonadism type 22 with or without anosmia
FGA	Afibrinogenemia, congenital
FGB	Congenital afibrinogenemia
FGD1	Aarskog-Scott syndrome; intellectual developmental disorder, X-linked syndromic, type 16
FGD4	Charcot-Marie-Tooth disease, type 4H
FGF23	Tumoral calcinosis, hyperphosphatemic, familial, type 2
FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia
FGG	Afibrinogenemia, congenital; Hypofibrinogenemia, congenital
FH	Fumarase deficiency
FHL1	Emery-Dreifuss muscular dystrophy 6, X-linked
FIBP	Thauvin-Robinet-Faivre syndrome
FIG4	Charcot-Marie-Tooth disease, type 4J; Yunis-Varon syndrome
FKBP10	Bruck syndrome 1
FKBP14	Ehlers-Danlos syndrome, kyphoscoliotic type, 2
FKRP	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])
FKTN	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])
FLAD1	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency
FLG	Ichthyosis vulgaris
FLI1	Bleeding disorder, platelet-type, type 21
FLNA	X-linked otopalatodigital (X-OPD) spectrum disorders;
FLNB	Spondylocarpotarsal synostosis syndrome
FLVCR1	Posterior column ataxia-retinitis pigmentosa syndrome
FLVCR2	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome
FMN2	intellectual developmental disorder, autosomal recessive, type 47
FMO3	Trimethylaminuria
FMR1	Fragile X syndrome
FOLR1	Neurodegeneration due to cerebral folate transport deficiency

Complete Investigation list

Gene	Disease
FOXE1	Bamforth-Lazarus syndrome
FOXE3	Anterior segment dysgenesis, type 2 multiple subtypes
FOXN1	T-cell immunodeficiency, congenital alopecia and nail dystrophy
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked
FOXRED1	Mitochondrial complex I deficiency, nuclear type 19
FRAS1	Fraser syndrome, type 1
FREM1	Manitoba oculotrichoanal syndrome
FREM2	Fraser syndrome, type 2
FRRS1L	Epileptic encephalopathy, early infantile, 37
FSHB	Hypogonadotropic hypogonadism, type 24 without anosmia
FSHR	Ovarian dysgenesis 1
FTCD	Glutamate formiminotransferase deficiency
FTL	L-ferritin deficiency
FTO	Growth retardation, developmental delay, facial dysmorphism
FTSJ1	Intellectual developmental disorder, X-linked 9
FUCA1	Fucosidosis
FUT8	Congenital disorder of glycosylation with defective fucosylation, type 1
FXN	Friedreich ataxia
FYCO1	Cataract 18
FZD6	Nail disorder, nonsyndromic congenital, type 10 (claw-shaped nails)
G6PC	Glycogen storage disease, type 1A
G6PC3	Dursun syndrome
G6PD	Hemolytic anemia, G6PD deficient (favism)
GAA	Glycogen storage disease, type 2
GALC	Krabbe disease
GALE	Galactose epimerase deficiency
GALK1	Galactokinase deficiency with cataracts
GALNS	Mucopolysaccharidosis, type 4A
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, type 1
GALT	Galactosemia
GAMT	Cerebral creatine deficiency syndrome, type 2
GAN	Giant axonal neuropathy, type 1
GAS8	Ciliary dyskinesia, primary, type 33
GATM	Cerebral creatine deficiency syndrome, type 3
GBA	Gaucher Disease
GBA2	Spastic paraplegia, type 46 autosomal recessive
GBE1	Glycogen storage disease, type 4

Complete Investigation list

Gene	Disease
GCDH	Glutaricaciduria, type 1
GCH1	Hyperphenylalaninemia, BH4-deficient, type B
GCK	Permanent neonatal diabetes mellitus (PNDM)
GCM2	Hypoparathyroidism, familial isolated (FIH) 2
GCNT2	Cataract 13 with adult i phenotype
GCSH	?Glycine encephalopathy
GDAP1	Charcot-Marie-Tooth disease, recessive intermediate, type A
GDF1	Right atrial isomerism (Ivemark syndrome)
GDF5	Chondrodysplasia, Grebe type
GDF6	Leber congenital amaurosis, type 17
GDI1	Intellectual developmental disorder, X-linked 41
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay
GFM1	Combined oxidative phosphorylation deficiency, type 1
GFPT1	Myasthenia, congenital, type 12 with tubular aggregates
GGCX	Vitamin K-dependent clotting factors, combined deficiency of, type 1
GH1	Growth hormone deficiency, isolated, type 1A; Kowarski syndrome
GHR	Laron dwarfism
GHRHR	Growth hormone deficiency, isolated, type 1B
GHSR	Growth hormone deficiency, isolated partial
GINS1	Immunodeficiency, type 55
GIPC3	Deafness, autosomal recessive, type 15
GJA1	Craniometaphyseal dysplasia, autosomal recessive
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1
GJB2	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6
GJB6	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6
GJC2	Spastic paraplegia, type 44 autosomal recessive
GK	Glycerol kinase deficiency
GLA	Fabry disease
GLB1	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)
GLDC	Glycine encephalopathy
GLDN	Lethal congenital contracture syndrome 11
GLE1	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease
GLIS2	Nephronophthisis, type 7
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism

Complete Investigation list

Gene	Disease
GLRA1	Hyperekplexia, type 1
GLRB	Hyperekplexia, type 2
GLRX5	Anemia, sideroblastic, type 3 pyridoxine-refractory; Spasticity, childhood-onset, with hyperglycinemia
GLUL	Glutamine deficiency, congenital
GLYCTK	D-glyceric aciduria
GM2A	GM2-gangliosidosis, AB variant
GMPPA	Alacrima, achalasia, and intellectual developmental disorder syndrome
GMPPB	Muscular dystrophy-dystroglycanopathy 14
GNAT1	Night blindness, congenital stationary, type 1G
GNAT2	Achromatopsia, type 4
GNB5	Intellectual developmental disorder with cardiac arrhythmia; Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia
GNE	Inclusion body myopathy, type 2 (Nonaka myopathy)
GNMT	Glycine N-methyltransferase deficiency
GNPAT	Rhizomelic chondrodysplasia punctata, type 2
GNPTAB	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta
GNPTG	Mucolipidosis III gamma
GNRHR	Hypogonadotropic hypogonadism, type 7 without anosmia
GNS	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)
GORAB	Geroderma osteodysplasticum
GOSR2	Epilepsy, progressive myoclonic, type 6
GOT2	Epileptic encephalopathy, early infantile, 82
GP1BA	Bernard-Soulier syndrome, type A1
GP1BB	Bernard-Soulier syndrome, type B
GP6	Bleeding disorder, platelet-type, type 11
GP9	Bernard-Soulier syndrome, type C
GPAA1	Glycosylphosphatidylinositol biosynthesis defect 15
GPC3	Simpson-Golabi-Behmel syndrome, type 1
GPC6	Omodysplasia, type 1
GPD1	Hypertriglyceridemia, transient infantile
GPHN	Molybdenum cofactor deficiency C
GPI	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency
GPIHBP1	Hyperlipoproteinemia, type 1D
GPR143	Ocular albinism, type 1 (Nettleship-Falls type)

Complete Investigation list

Gene	Disease
GPR179	Night blindness, congenital stationary (complete), type 1E, autosomal recessive
GPR68	Amelogenesis imperfecta, type 2A6 (hypomaturation type)
GPR98	Usher syndrome, type 2C
GPSM2	Chudley-McCullough syndrome
GPT2	intellectual developmental disorder, autosomal recessive 49
GPX4	Spondylometaphyseal dysplasia, Sedaghatian type
GRHL2	Ectodermal dysplasia/short stature syndrome
GRHPR	Hyperoxaluria, primary, type 2
GRIA3	Intellectual developmental disorder, X-linked syndromic, Wu type
GRID2	Spinocerebellar ataxia, autosomal recessive, type 18
GRIK2	Intellectual developmental disorder, autosomal recessive 6
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive
GRIP1	Fraser syndrome 3
GRK1	Oguchi disease-2
GRM1	Spinocerebellar ataxia, autosomal recessive, type 13
GRM6	Night blindness, congenital stationary (complete), type 1B, autosomal recessive
GRN	Ceroid lipofuscinosis, neuronal, type 11
GRXCR1	Deafness, autosomal recessive, type 25
GSC	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities
GSS	Glutathione synthetase deficiency
GTF2H5	Trichothiodystrophy, type 3 photosensitive
GTPBP2	Jaberi-Elahi syndrome
GTPBP3	Combined oxidative phosphorylation deficiency 23
GUCY2C	Meconium ileus
GUCY2D	Leber congenital amaurosis, type 1
GUF1	Epileptic encephalopathy, early infantile, 40
GUSB	Mucopolysaccharidosis, type 7
GYG1	Polyglucosan body myopathy, type 2
GYS1	Glycogen storage disease, type 0 muscle
GYS2	Glycogen storage disease, type 0 liver
GZF1	Joint laxity, short stature, and myopia
H6PD	Cortisone reductase deficiency 1
HAAO	Vertebral, cardiac, renal, and limb defects syndrome 1

Complete Investigation list

Gene	Disease
HACE1	Spastic paraplegia and psychomotor retardation with or without seizures
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency
HADHA	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency
HADHB	Mitochondrial trifunctional protein deficiency
HAMP	Hemochromatosis, type 2B
HARS1	Usher syndrome, type 3B
HAX1	Neutropenia, severe congenital, type 3 autosomal recessive
HBA1	Alpha-thalassemia
HBA2	Alpha-thalassemia
HBB	Beta-thalassemia, Sickle cell anemia and other HBB-related hemoglobinopathies
HCCS	Linear skin defects with multiple congenital anomalies 1
HCFC1	intellectual developmental disorder, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)
HELLS	Immunodeficiency-centromeric instability-facial anomalies syndrome 4
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A
HERC1	Macrocephaly, dysmorphic facies, and psychomotor retardation
HERC2	intellectual developmental disorder, autosomal recessive, type 38
HES7	Spondylocostal dysostosis, type 4 autosomal recessive
HESX1	Growth hormone deficiency with pituitary anomalies
HEXA	Tay-Sachs disease
HEXB	Sandhoff disease, infantile, juvenile, and adult forms
HFE	Hemochromatosis
HFE2	Hemochromatosis, type 2A
HFM1	Premature ovarian failure 9
HGD	Alkaptonuria
HGF	Deafness, autosomal recessive, type 39
HGSNAT	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency
HIKESHI	Leukodystrophy, hypomyelinating, type 13
HINT1	Neuromyotonia and axonal neuropathy, autosomal recessive
HJV	Hemochromatosis, type 2A
HK1	Charcot-Marie-Tooth disease, type 4G
HLCS	Holocarboxylase synthetase deficiency
HMGCL	HMG-CoA lyase deficiency
HMGCS2	HMG-CoA synthase-2 deficiency

Complete Investigation list

Gene	Disease
HMOX1	Heme oxygenase-1 deficiency
HMX1	Oculoauricular syndrome
HNMT	intellectual developmental disorder, autosomal recessive, type 51
HOGA1	Hyperoxaluria, primary, type 3
HOXA1	Athabaskan brainstem dysgenesis syndrome
HOXB1	Facial paresis, hereditary congenital, 3
HOXC13	Ectodermal dysplasia 9 hair/nail type
HPCA	Dystonia 2 torsion, autosomal recessive
HPD	Tyrosinemia, type 3
HPGD	Hypertrophic osteoarthropathy, primary, type 1 (pachydermoperiostosis)
HPRT1	Lesch-Nyhan syndrome
HPS1	Hermansky-Pudlak syndrome, type 1
HPS3	Hermansky-Pudlak syndrome, type 3
HPS4	Hermansky-Pudlak syndrome, type 4
HPS5	Hermansky-Pudlak syndrome, type 5
HPS6	Hermansky-Pudlak syndrome, type 6
HPSE2	Urofacial syndrome, type 1
HR	Alopecia universalis; Atrichia with papular lesions
HSD11B2	Apparent mineralocorticoid excess
HSD17B10	HSD10 mitochondrial disease
HSD17B3	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
HSD17B4	D-bifunctional protein deficiency
HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency
HSD3B7	Bile acid synthesis defect, congenital, type 1
HSPA9	Even-plus syndrome
HSPD1	Leukodystrophy, hypomyelinating, type 4
HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type
HTRA1	CARASIL syndrome
HTRA2	3-methylglutaconic aciduria, type 8
HUWE1	Intellectual developmental disorder, X-linked syndromic, Turner type
HYAL1	?Mucopolysaccharidosis, type 9
HYDIN	Ciliary dyskinesia, primary, type 5
HYLS1	Hydrolethalus syndrome
IARS1	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy

Complete Investigation list

Gene	Disease
IBA57	Multiple mitochondrial dysfunctions syndrome 3
ICOS	Immunodeficiency, common variable, 1
IDH3B	Retinitis pigmentosa, type 46
IDS	Mucopolysaccharidosis, type 2
IDUA	Mucopolysaccharidosis type 1
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome
IFNGR1	Immunodeficiency, type 27A, mycobacteriosis
IFNGR2	Immunodeficiency, type 28 mycobacteriosis
IFT122	Cranioectodermal dysplasia 1
IFT140	Retinitis pigmentosa, type 80; Short-rib thoracic dysplasia 9 with or without polydactyly
IFT172	Short-rib thoracic dysplasia 10 with or without polydactyly
IFT43	Short-rib thoracic dysplasia 18 with polydactyly
IFT52	Short-rib thoracic dysplasia 16 with or without polydactyly
IFT80	Short-rib thoracic dysplasia, type 2 with or without polydactyly
IFT81	Short-rib thoracic dysplasia 19 with or without polydactyly
IGBP1	Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia
IGF1	Growth retardation with deafness and intellectual developmental disorder due to IGF1 deficiency
IGF1R	Insulin-like growth factor I, resistance to
IGFALS	Acid-labile subunit deficiency
IGFBP7	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis
IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S
IGLL1	Agammaglobulinemia 2
IGSF1	Hypothyroidism, central, and testicular enlargement
IHH	Acrocapitofemoral dysplasia
IKBKAP	Dysautonomia, familial
IKBKB	Immunodeficiency, type 15
IKBKG	Ectodermal dysplasia and immunodeficiency 1
IL10RA	Inflammatory bowel disease, type 28 early onset, autosomal recessive
IL10RB	Inflammatory bowel disease, type 25 early onset, autosomal recessive
IL11RA	Craniosynostosis and dental anomalies
IL12B	Immunodeficiency, type 29 mycobacteriosis
IL12RB1	Immunodeficiency, type 30
IL17RA	Immunodeficiency, type 51
IL17RC	Candidiasis, familial, 9

Complete Investigation list

Gene	Disease
IL1RAPL1	Intellectual developmental disorder, X-linked 21
IL1RN	Sterile multifocal osteomyelitis with periostitis and pustulosis
IL21R	Immunodeficiency, type 56
IL2RA	Immunodeficiency, type 41 with lymphoproliferation and autoimmunity
IL2RG	Severe combined immunodeficiency, X-linked
IL36RN	Psoriasis, type 14 pustular
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type
ILDR1	Deafness, autosomal recessive, type 42
IMPA1	intellectual developmental disorder, autosomal recessive 59
IMPG2	Retinitis pigmentosa, type 56
INPP5E	Joubert syndrome, type 1
INPP5K	Muscular dystrophy, congenital, with cataracts and intellectual disability
INPPL1	Opsismodysplasia
INS	Permanent neonatal diabetes mellitus (PNDM)
INSR	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, type A
INTS1	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies
INVS	Nephronophthisis, type 2 infantile
IQCB1	Senior-Loken syndrome, type 5
IQCE	Polydactyly, postaxial, type A7
IQSEC2	Intellectual developmental disorder, X-linked 1
IRAK4	Immunodeficiency, type 67 (IRAK4 deficiency)
IRF8	Immunodeficiency, type 32B, monocyte and dendritic cell deficiency
IRX5	Hamamy syndrome
ISCA1	Multiple mitochondrial dysfunctions syndrome 5
ISCA2	Multiple mitochondrial dysfunctions syndrome 4
ISCU	Myopathy with lactic acidosis, hereditary
ISG15	Immunodeficiency, type 38
ITCH	Autoimmune disease, multisystem, with facial dysmorphism
ITGA2B	Glanzmann thrombasthenia
ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital
ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis
ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency
ITGA8	Renal hypodysplasia/aplasia 1
ITGB2	Leukocyte adhesion deficiency

Complete Investigation list

Gene	Disease
ITGB3	Glanzmann thrombasthenia
ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia
ITGB6	Amelogenesis imperfecta, type 1H
ITK	Lymphoproliferative syndrome 1
ITPA	Epileptic encephalopathy, early infantile, type 35
ITPR1	Gillespie syndrome
IVD	Isovaleric acidemia
IYD	Thyroid dyshormonogenesis, type 4
JAGN1	Neutropenia, severe congenital, 6 autosomal recessive
JAK3	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts
JUP	Naxos disease
KANK2	Nephrotic syndrome, type 16
KARS1	Deafness, autosomal recessive, type 89
KATNB1	Lissencephaly 6 with microcephaly
KATNIP	Joubert syndrome 26
KCNE1	Jervell and Lange-Nielsen syndrome 2
KCNJ1	Bartter syndrome, type 2
KCNJ10	SESAME syndrome
KCNJ11	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)
KCNJ13	Leber congenital amaurosis, type 16
KCNV2	Retinal cone dystrophy, type 3B
KCTD7	Epilepsy, progressive myoclonic, type 3 with or without intracellular inclusions
KDM5C	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type
KDSR	Erythrokeratodermia variabilis et progressiva 4
KERA	Cornea plana 2 autosomal recessive
KHDC3L	Hydatidiform mole, recurrent, type 2
KIAA0586	Short-rib thoracic dysplasia 14 with polydactyly
KIAA0753	Orofaciodigital syndrome, type 15
KIAA1109	Alkuraya-Kucinskas syndrome
KIAA1549	Retinitis pigmentosa, type 86
KIAA2022	Intellectual developmental disorder, X-linked 98

Complete Investigation list

Gene	Disease
KIF14	Microcephaly 20 primary, autosomal recessive; ?Meckel syndrome 12
KIF1A	Neuropathy, hereditary sensory, type 2C; Spastic paraplegia, type 30 autosomal recessive
KIF1C	Spastic ataxia 2 autosomal recessive
KIF7	Acrocallosal syndrome; Joubert syndrome, type 12
KIFBP	Goldberg-Shprintzen megacolon syndrome
KISS1R	Hypogonadotropic hypogonadism, type 8 with or without anosmia
KIZ	Retinitis pigmentosa 69
KLHL3	Pseudohypoaldosteronism, type 2D
KLHL40	Nemaline myopathy 8 autosomal recessive
KLHL41	Nemaline myopathy 9
KLHL7	Cold-induced sweating syndrome 3
KLK4	Amelogenesis imperfecta, type 2A1 (hypomaturation type)
KLKB1	Fletcher factor (prekallikrein) deficiency
KNL1	Microcephaly 4 primary, autosomal recessive
KPTN	intellectual developmental disorder, autosomal recessive 41
KREMEN1	Ectodermal dysplasia 13 hair/tooth type
KRT10	Epidermolytic hyperkeratosis
KRT14	Epidermolysis bullosa simplex, autosomal recessive, type 1
KRT25	Woolly hair, autosomal recessive 3
KRT5	Epidermolysis bullosa simplex, autosomal recessive, type 1
KRT85	Ectodermal dysplasia 4 hair/nail type
KY	Myopathy, myofibrillar, type 7
KYNU	Vertebral, cardiac, renal, and limb defects syndrome, type 2
L1CAM	L1 Syndrome
L2HGDH	L-2-hydroxyglutaric aciduria
LAMA1	Poretti-Boltshauser syndrome
LAMA2	LAMA2-related muscular dystrophy
LAMA3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LAMB1	Lissencephaly, type 5
LAMB2	Pierson syndrome; Nephrotic syndrome, type 5 with or without ocular abnormalities
LAMB3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LAMC2	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LAMC3	Cortical malformations, occipital
LAMP2	Danon disease
LARGE1	Muscular dystrophy-dystroglycanopathy, type 6A and 6B

Complete Investigation list

Gene	Disease
LARP7	Alazami syndrome
LARS1	Infantile liver failure syndrome 1 (ILFS1)
LARS2	Perrault syndrome, type 4
LAT	Immunodeficiency, type 52
LBR	Greenberg skeletal dysplasia
LCA5	Leber congenital amaurosis, type 5
LCAT	Familial LCAT deficiency; Fish-eye disease
LCK	Immunodeficiency, type 22
LCT	Lactase deficiency, congenital
LDHA	Glycogen storage disease type 11
LDLRAP1	Hypercholesterolemia, familial, autosomal recessive
LEMD2	Cataract 46 juvenile-onset
LEP	Obesity, morbid, due to leptin deficiency
LEPR	Obesity, morbid, due to leptin receptor deficiency
LGI4	Arthrogryposis multiplex congenita, neurogenic, with myelin defect
LHB	Hypogonadotropic hypogonadism, type 23 with or without anosmia
LHCGR	Leydig cell hypoplasia
LHFPL5	Deafness, autosomal recessive, type 67
LHX3	Pituitary hormone deficiency, combined, type 3
LIAS	Hyperglycinemia, lactic acidosis, and seizures
LIFR	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome
LIG4	LIG4 syndrome
LIM2	Cataract 19 multiple types
LINS1	intellectual developmental disorder, autosomal recessive, type 27
LIPA	Lysosomal Acid Lipase Deficiency
LIPE	Lipodystrophy, familial partial, type 6
LIPH	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2 with or without hypotrichosis
LIPN	Ichthyosis, congenital, autosomal recessive 8
LIPT1	Lipoyltransferase 1 deficiency
LIPT2	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities
LMAN1	Combined deficiency of factor V and factor VIII, type 1
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type
LMF1	Lipase deficiency, combined
LMNA	Cardiomyopathy, dilated, 1A
LMOD3	Nemaline myopathy 10

Complete Investigation list

Gene	Disease
LONP1	CODAS syndrome
LOXHD1	Deafness, autosomal recessive, type 77
LPAR6	Hypotrichosis, type 8 or woolly hair, autosomal recessive, type 1 with or without hypotrichosis
LPIN1	Myoglobinuria, acute recurrent, autosomal recessive
LPIN2	Majeed syndrome
LPL	Lipoprotein lipase deficiency
LRAT	Leber congenital amaurosis type 14
LRBA	Immunodeficiency, common variable, 8 with autoimmunity
LRIG2	Urofacial syndrome 2
LRIT3	Night blindness, congenital stationary (complete), 1F, autosomal recessive
LRMDA	Albinism, oculocutaneous, type 7
LRP2	Donnai-Barrow syndrome
LRP4	Cenani-Lenz syndactyly syndrome
LRP5	Osteoporosis-pseudoglioma syndrome
LRPAP1	Myopia, type 23 autosomal recessive
LRPPRC	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian)
LRRC6	Ciliary dyskinesia, primary, 19
LRSAM1	Charcot-Marie-Tooth disease, axonal, type 2P
LRTOMT	Deafness, autosomal recessive, type 63
LSS	Cataract 44
LTBP2	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma
LTBP3	Dental anomalies and short stature
LTBP4	Cutis laxa, autosomal recessive, type 1C
LYRM7	Mitochondrial complex III deficiency, nuclear type 8
LYST	Chediak-Higashi syndrome
LZTFL1	Bardet-Biedl syndrome, type 17
LZTR1	Noonan syndrome, type 2
MAG	Spastic paraplegia, type 75 autosomal recessive
MAGI2	Nephrotic syndrome, type 15
MAGT1	Congenital disorder of glycosylation, type Icc
MAK	Retinitis pigmentosa type 62
MALT1	Immunodeficiency, type 12
MAN1B1	intellectual developmental disorder, autosomal recessive, type 15
MAN2B1	Alpha-mannosidosis

Complete Investigation list

Gene	Disease
MANBA	Mannosidosis, beta
MAP3K20	Centronuclear myopathy, type 6 with fiber-type disproportion
MAPKBP1	Nephronophthisis 20
MAPT	Supranuclear palsy, progressive atypical (parkinsonism syndrome)
MARS1	Interstitial lung and liver disease
MARS2	Spastic ataxia, type 3 autosomal recessive
MARVELD2	Deafness, autosomal recessive, type 49
MASP1	3MC syndrome 1
MAT1A	Methionine adenosyltransferase deficiency, autosomal recessive
MATN3	Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type
MBOAT7	intellectual developmental disorder, autosomal recessive 57
MBTPS2	Olmsted syndrome, X-linked
MC2R	Glucocorticoid deficiency, due to ACTH unresponsiveness
MCCC1	3-Methylcrotonyl-CoA carboxylase deficiency, type 1
MCCC2	3-Methylcrotonyl-CoA carboxylase deficiency, type 2
MCEE	Methylmalonyl-CoA epimerase deficiency
MCFD2	Combined deficiency of factor V and factor VIII, type 2
MCIDAS	Ciliary dyskinesia, primary, type 42
MCM3AP	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development
MCM4	Immunodeficiency, type 54
MCM9	Ovarian dysgenesis 4
MCOLN1	Mucolipidosis type 4
MCPH1	Microcephaly type 1 primary, autosomal recessive
MDH2	Epileptic encephalopathy, early infantile, 51
MECP2	Encephalopathy, neonatal severe; Rett syndrome
MECR	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy
MED23	intellectual developmental disorder, autosomal recessive, type 18
MED25	Basel-Vanagait-Smirin-Yosef syndrome
MEFV	Familial Mediterranean fever
MEGF10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset
MEGF8	Carpenter syndrome, type 2
MEOX1	Klippel-Feil syndrome 2
MERTK	Retinitis pigmentosa type 38
MESP2	Spondylocostal dysostosis, type 2 autosomal recessive

Complete Investigation list

Gene	Disease
METTL23	intellectual developmental disorder, autosomal recessive 44
MFF	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 2
MFN2	Charcot-Marie-Tooth disease, axonal, type 2A2B
MFRP	Microphthalmia, isolated type 5
MFSD2A	Microcephaly 15 primary, autosomal recessive
MFSD8	Ceroid lipofuscinosis, neuronal, type 7
MGAT2	Congenital disorder of glycosylation, type 2a
MGME1	Mitochondrial DNA depletion syndrome 11
MGP	Keutel syndrome
MICU1	Myopathy with extrapyramidal signs
MID1	Opitz GBBB syndrome, type 1
MIPEP	Combined oxidative phosphorylation deficiency 31
MITF	COMMAD syndrome
MKKS	Bardet-Biedl syndrome type 6
MKS1	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts
MLPH	Griscelli syndrome, type 3
MLYCD	Malonyl-CoA decarboxylase deficiency
MMAA	Methylmalonic aciduria, vitamin B12-responsive
MMAB	Methylmalonic aciduria, vitamin B12-responsive, type cblB
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
MMADHC	Homocystinuria, cblD type, variant 1
MME	Charcot-Marie-Tooth disease, axonal, type 2T
MMP13	Metaphyseal dysplasia, Spahr type
MMP2	Multicentric osteolysis, nodulosis, and arthropathy (MONA)
MMP20	Amelogenesis imperfecta, type 2A2 (hypomaturation type)
MMP21	Heterotaxy, visceral, 7 autosomal
MMUT	Methylmalonic aciduria, mut(0) type
MOCOS	Xanthinuria, type 2
MOCS1	Molybdenum cofactor deficiency A
MOCS2	Molybdenum cofactor deficiency B
MOGS	Congenital disorder of glycosylation, type 2B
MPC1	Mitochondrial pyruvate carrier deficiency
MPDU1	Congenital disorder of glycosylation, type 1F
MPDZ	Hydrocephalus, congenital, type 2 with or without brain or eye anomalies

Complete Investigation list

Gene	Disease
MPI	Congenital disorder of glycosylation, type 1B
MPIG6B	Thrombocytopenia, anemia, and myelofibrosis
MPL	Thrombocytopenia, congenital amegakaryocytic
MPLKIP	Trichothiodystrophy, type 4 nonphotosensitive
MPO	Myeloperoxidase deficiency
MPV17	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE
MPZ	Dejerine-Sottas disease
MRAP	Glucocorticoid deficiency, type 2
MRE11	Ataxia-telangiectasia-like disorder 1
MRPS16	Combined oxidative phosphorylation deficiency 2
MRPS22	Combined oxidative phosphorylation deficiency type 5
MRPS34	Combined oxidative phosphorylation deficiency 32
MSH3	Familial adenomatous polyposis, type 4
MSMO1	Microcephaly, congenital cataract, and psoriasiform dermatitis
MSRB3	Deafness, autosomal recessive, type 74
MSTO1	Myopathy, mitochondrial, and ataxia
MTFMT	Combined oxidative phosphorylation deficiency 15
MTHFD1	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia
MTHFR	Homocystinuria due to MTHFR deficiency
MTM1	Myotubular myopathy, X-linked
MTMR2	Charcot-Marie-Tooth disease, type 4B1
MTO1	Combined oxidative phosphorylation deficiency 10
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type
MTRR	Homocystinuria-megaloblastic anemia, cbl E type
MTTP	Abetalipoproteinemia
MUSK	Fetal akinesia deformation sequence, type 1; Myasthenic syndrome, congenital, type 9 associated with acetylcholine receptor deficiency
MUTYH	Adenomas, multiple colorectal
MVK	Mevalonic aciduria
MYBPC1	Lethal congenital contracture syndrome, type 4
MYD88	Immunodeficiency, type 68
MYH2	Proximal myopathy and ophthalmoplegia
MYMK	Carey-Fineman-Ziter syndrome
MYO15A	Deafness, autosomal recessive, type 3

Complete Investigation list

Gene	Disease
MYO18B	Klippel-Feil syndrome, type 4 autosomal recessive, with myopathy and facial dysmorphism
MYO1E	Glomerulosclerosis, focal segmental, 6
MYO3A	Deafness, autosomal recessive, type 30
MYO5A	Griscelli syndrome, type 1
MYO5B	Microvillus inclusion disease
MYO6	Deafness, autosomal recessive, type 37
MYO7A	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2
MYPN	Nemaline myopathy, type 11 autosomal recessive
NADK2	2,4-dienoyl-CoA reductase deficiency
NAGA	Schindler disease, type I
NAGLU	Mucopolysaccharidosis, type 3B (Sanfilippo B)
NAGS	N-acetylglutamate synthase deficiency
NALCN	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1
NANS	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type
NARS2	Combined oxidative phosphorylation deficiency 24
NAXE	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy
NBAS	Infantile liver failure syndrome, type 2; Short stature, optic nerve atrophy, and Pelger-Huet anomaly
NBEAL2	Gray platelet syndrome
NBN	Nijmegen breakage syndrome
NCAPD3	Microcephaly 22 primary, autosomal recessive
NCF1	Chronic granulomatous disease, type 1
NCF2	Chronic granulomatous disease, type 2
NCF4	Chronic granulomatous disease 3 autosomal recessive
NDE1	Lissencephaly, type 4 (with microcephaly)
NDP	Norrie disease
NDRG1	Charcot-Marie-Tooth disease, type 4D
NDST1	intellectual developmental disorder, autosomal recessive, type 46
NDUFA1	Mitochondrial complex I deficiency, nuclear type 12
NDUFA10	Mitochondrial complex I deficiency, nuclear type 22
NDUFA11	Mitochondrial complex I deficiency, nuclear type 14
NDUFA12	Mitochondrial complex I deficiency, nuclear type 23
NDUFA2	Mitochondrial complex I deficiency, nuclear type 13
NDUFA9	Mitochondrial complex I deficiency, nuclear type 26

Complete Investigation list

Gene	Disease
NDUFAF1	Mitochondrial complex I deficiency, nuclear type 11
NDUFAF2	Mitochondrial complex I deficiency, nuclear type 10
NDUFAF3	Mitochondrial complex I deficiency, nuclear type 18
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16
NDUFAF6	Mitochondrial complex I deficiency, nuclear type 17
NDUFB3	Mitochondrial complex I deficiency, nuclear type 25
NDUFB9	Mitochondrial complex I deficiency, nuclear type 24
NDUFS1	Mitochondrial complex I deficiency, nuclear type 5
NDUFS2	Mitochondrial complex I deficiency, nuclear type 6
NDUFS3	Mitochondrial complex I deficiency, nuclear type 8
NDUFS4	Mitochondrial complex I deficiency, nuclear type 1
NDUFS6	Mitochondrial complex I deficiency, nuclear type 9
NDUFS7	Mitochondrial complex I deficiency, nuclear type 3
NDUFS8	Mitochondrial complex I deficiency, nuclear type 2
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4
NDUFV2	Mitochondrial complex I deficiency, nuclear type 7
NEB	Nemaline myopathy type 2
NECTIN1	Cleft lip/palate-ectodermal dysplasia syndrome; Orofacial cleft 7
NECTIN4	Ectodermal dysplasia-syndactyly syndrome, type 1
NEFL	Charcot-Marie-Tooth disease, type 1F
NEK1	Short-rib thoracic dysplasia, type 6 with or without polydactyly
NEK8	Renal-hepatic-pancreatic dysplasia, type 2
NEK9	Lethal congenital contracture syndrome 10
NEU1	Sialidosis, type 1 and type 2
NEUROG3	Diarrhea 4 malabsorptive, congenital
NFU1	Multiple mitochondrial dysfunctions syndrome 1
NGF	Neuropathy, hereditary sensory and autonomic, type 5
NGLY1	Congenital disorder of deglycosylation
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation
NHLRC1	Epilepsy, progressive myoclonic, type 2B (Lafora)
NHP2	Dyskeratosis congenita, autosomal recessive type 2
NHS	Nance-Horan syndrome; Cataract 40, X-Linked
NIN	Seckel syndrome, type 7
NIPAL4	Ichthyosis, congenital, autosomal recessive, type 6
NKX2-6	Conotruncal heart malformations
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia

Complete Investigation list

Gene	Disease
NKX6-2	Spastic ataxia 8 autosomal recessive, with hypomyelinating leukodystrophy
NLRP1	Autoinflammation with arthritis and dyskeratosis
NLRP7	Hydatidiform mole, recurrent, type 1
NME8	Ciliary dyskinesia, primary, type 6
NMNAT1	Leber congenital amaurosis type 9
NNT	Glucocorticoid deficiency 4 with or without mineralocorticoid deficiency
NONO	Intellectual developmental disorder, X-linked syndromic 34
NOP10	Dyskeratosis congenita, autosomal recessive type 1
NPC1	Niemann-Pick disease, type C1
NPC2	Niemann-pick disease, type C2
NPHP1	Joubert syndrome type 4
NPHP3	Meckel syndrome type 7
NPHP4	Nephronophthisis type 4
NPHS1	Nephrotic syndrome, type 1
NPHS2	Nephrotic syndrome, type 2
NPR2	Acromesomelic dysplasia, Maroteaux type
NR0B1	Adrenal hypoplasia, congenital
NR1H4	Cholestasis, progressive familial intrahepatic, type 5
NR2E3	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37
NRL	Retinal degeneration, autosomal recessive, clumped pigment type
NRXN1	Pitt-Hopkins-like syndrome, type 2
NSDHL	CHILD syndrome; CK syndrome
NSMCE2	Seckel syndrome, type 10
NSUN2	Intellectual developmental disorder, autosomal recessive 5
NT5C2	Spastic paraplegia, type 45 autosomal recessive
NT5C3A	Anemia, hemolytic, due to UMPH1 deficiency
NT5E	Calcification of joints and arteries
NTHL1	Familial adenomatous polyposis, type 3
NTRK1	Insensitivity to pain, congenital, with anhidrosis
NUBPL	Mitochondrial complex I deficiency, nuclear type 21
NUP107	Nephrotic syndrome, type 11
NUP62	Striatonigral degeneration, infantile
NUP93	Nephrotic syndrome, type 12
NYX	Night blindness, congenital stationary (complete), 1A, X-linked
OAT	Gyrate atrophy of choroid and retina

Complete Investigation list

Gene	Disease
OBSL1	3M syndrome 2
OCA2	Oculocutaneous albinism type 2
OCLN	Pseudo-TORCH syndrome, type 1
OCRL	Lowe Syndrome; Dent disease type 2
ODAD1	Ciliary dyskinesia, primary, type 20
ODAD2	Ciliary dyskinesia, primary, type 23
ODAD3	Ciliary dyskinesia, primary, type 30
OFD1	Joubert syndrome 10; Simpson-Golabi-Behmel syndrome, type 2
OPA1	Behr syndrome
OPA3	3-methylglutaconic aciduria, type 3
OPHN1	Intellectual developmental disorder, X-linked syndromic, Billuart type
ORAI1	Immunodeficiency, type 9
ORC1	Meier-Gorlin syndrome, type 1
ORC4	Meier-Gorlin syndrome, type 2
ORC6	Meier-Gorlin syndrome, type 3
OSGEP	Galloway-Mowat syndrome 3
OSTM1	Osteopetrosis, autosomal recessive type 5
OTC	Ornithine transcarbamylase deficiency
OTOA	Deafness, autosomal recessive, type 22
OTOF	Deafness, autosomal recessive, type 9
OTOG	Deafness, autosomal recessive, type 18B
OTOGL	Deafness, autosomal recessive, type 84B
OTUD6B	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies
OTULIN	Autoinflammation, panniculitis, and dermatosis syndrome
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency
P2RY12	Bleeding disorder, platelet-type, type 8
P3H1	Osteogenesis imperfecta, type 8
P3H2	Myopia, high, with cataract and vitreoretinal degeneration
PADI6	Preimplantation embryonic lethality 2
PAH	Phenylketonuria
PAK3	Intellectual developmental disorder, X-linked 30
PALB2	Fanconi anemia, complementation group N
PAM16	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type
PANK2	Neurodegeneration with brain iron accumulation type 1
PAPSS2	Brachyolmia, type 4 with mild epiphyseal and metaphyseal changes
PARK7	Parkinson disease, type 7 autosomal recessive, early-onset

Complete Investigation list

Gene	Disease
PARN	Dyskeratosis congenita, autosomal recessive 6
PATL2	Oocyte maturation defect 4
PAX7	Rhabdomyosarcoma 2 alveolar
PC	Pyruvate carboxylase deficiency
PCARE	Retinitis pigmentosa, type 54
PCBD1	Hyperphenylalaninemia, BH4-deficient, type D
PCCA	Propionic acidemia
PCCB	Propionic acidemia
PCDH12	Microcephaly, seizures, spasticity, and brain calcification
PCDH15	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic
PCDH19	Developmental and epileptic encephalopathy 9
PCK2	PEPCK deficiency, mitochondrial
PCNT	Microcephalic osteodysplastic primordial dwarfism, type 2
PCSK1	Obesity with impaired prohormone processing
PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy
PDE10A	Dyskinesia, limb and orofacial, infantile-onset
PDE6A	Retinitis pigmentosa type 43
PDE6B	Retinitis pigmentosa type 40
PDE6C	Cone dystrophy type 4
PDE6G	Retinitis pigmentosa type 57
PDE6H	Retinal cone dystrophy 3 and achromatopsia 6
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency
PDHB	Pyruvate dehydrogenase E1-beta deficiency
PDHX	Lacticacidemia due to PDX1 deficiency
PDP1	Pyruvate dehydrogenase phosphatase deficiency
PDSS1	Coenzyme Q10 deficiency, primary, type 2
PDSS2	Coenzyme Q10 deficiency, primary, type 3
PDX1	Pancreatic agenesis type 1
PDXK	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy
PEPD	Prolidase deficiency
PET100	Mitochondrial complex IV deficiency, nuclear type 12
PEX1	Heimler syndrome type 1
PEX10	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B
PEX11B	Peroxisome biogenesis disorder 14B
PEX12	Peroxisome biogenesis disorder type 3A (Zellweger)

Complete Investigation list

Gene	Disease
PEX13	Peroxisome biogenesis disorder, type 11A (Zellweger syndrome); Peroxisome biogenesis disorder, type 11B
PEX14	Peroxisome biogenesis disorder, type 13A (Zellweger syndrome)
PEX16	Peroxisome biogenesis disorder, type 8A (Zellweger syndrome); Peroxisome biogenesis disorder, type 8B
PEX19	Peroxisome biogenesis disorder, type 12A (Zellweger syndrome)
PEX2	Peroxisome biogenesis disorder type 5A (Zellweger)
PEX26	Peroxisome biogenesis disorder type 7A (Zellweger)
PEX3	Peroxisome biogenesis disorder, type 10A (Zellweger syndrome)
PEX5	Peroxisome biogenesis disorder type 2A (Zellweger)
PEX6	Peroxisome biogenesis disorder, type 4A (Zellweger syndrome); Peroxisome biogenesis disorder, type 4B; Heimler syndrome 2
PEX7	Rhizomelic chondrodysplasia punctata, type 1
PFKM	Glycogen storage disease, type 7
PGAM2	Glycogen storage disease X
PGAP1	intellectual developmental disorder, autosomal recessive 42
PGAP2	Hyperphosphatasia with impaired intellectual development syndrome 3
PGAP3	Hyperphosphatasia with impaired intellectual development syndrome 4
PGK1	Phosphoglycerate kinase 1 deficiency
PGM1	Congenital disorder of glycosylation, type 1t
PGM3	Immunodeficiency, type 23
PHF8	Intellectual developmental disorder, X-linked syndromic, Siderius type
PHGDH	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency
PHKA1	Muscle glycogenosis
PHKA2	Glycogen storage disease, type IXa1/2
PHKB	Glycogen storage disease, type 9B
PHKG2	Glycogen storage disease type 9c
PHOX2A	Fibrosis of extraocular muscles, congenital, 2
PHYH	Refsum disease
PI4KA	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis
PIBF1	Joubert syndrome 33
PIEZO1	Lymphedema, hereditary, type 3
PIEZO2	Arthrogryposis, distal, with impaired proprioception and touch
PIGC	Glycosylphosphatidylinositol biosynthesis defect 16
PIGG	intellectual developmental disorder, autosomal recessive 53
PIGL	Zunich neuroectodermal syndrome

Complete Investigation list

Gene	Disease
PIGM	Glycosylphosphatidylinositol deficiency
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome, type 1
PIGO	Hyperphosphatasia with impaired intellectual development syndrome 2
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3
PIGV	Hyperphosphatasia with impaired intellectual development syndrome 1
PIGW	Glycosylphosphatidylinositol biosynthesis defect 11
PIGY	Hyperphosphatasia with impaired intellectual development syndrome 6
PINK1	Parkinson disease, type 6 early onset
PIP5K1C	Lethal congenital contractural syndrome, type 3
PJVK	Deafness, autosomal recessive, type 59
PKD1L1	Heterotaxy, visceral, 8 autosomal
PKHD1	Polycystic kidney disease type 4
PKLR	Pyruvate kinase deficiency
PKP1	Ectodermal dysplasia/skin fragility syndrome
PLA2G6	Infantile neuroaxonal dystrophy type 1
PLAA	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies
PLCB1	Epileptic encephalopathy, early infantile, type 12
PLCB4	Auriculocondylar syndrome, type 2
PLCD1	Nail disorder, nonsyndromic congenital, type 3 (leukonychia)
PLCE1	Nephrotic syndrome, type 3
PLD1	Cardiac valvular defect, developmental
PLEC	Epidermolysis bullosa simplex with muscular dystrophy
PLEKHG5	Charcot-Marie-Tooth disease, recessive intermediate, type C
PLG	Plasminogen deficiency, type I
PLK4	Microcephaly and chorioretinopathy, autosomal recessive, 2
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1
PLOD2	Bruck syndrome 2
PLOD3	Lysyl hydroxylase 3 deficiency
PLP1	Pelizaeus-Merzbacher disease
PLPBP	Epilepsy, early-onset, vitamin B6-dependent
PMM2	Congenital disorder of glycosylation, type 1A
PMP22	Dejerine-Sottas disease
PMPCA	Spinocerebellar ataxia, autosomal recessive, type 2
PMPCB	Multiple mitochondrial dysfunctions syndrome 6
PNKP	Ataxia-oculomotor apraxia, type 4; Microcephaly, seizures, and developmental delay

Complete Investigation list

Gene	Disease
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency
PNPLA1	Ichthyosis, congenital, autosomal recessive, type 10
PNPLA2	Neutral lipid storage disease with myopathy
PNPLA6	Boucher-Neuhauser syndrome; Oliver-McFarlane syndrome; Spastic paraplegia, type 39 autosomal recessive
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency
PNPT1	Combined oxidative phosphorylation deficiency 13
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis
POC1B	Cone-rod dystrophy 20
POLE	FILS syndrome
POLG	POLG-related disorders
POLH	Xeroderma pigmentosum, variant type
POLR1C	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3
POLR1D	Treacher Collins syndrome, type 2
POLR3A	Leukodystrophy, hypomyelinating, type 7
POLR3B	Leukodystrophy, hypomyelinating, type 8
POMC	Obesity, adrenal insufficiency, and red hair due to POMC deficiency
POMGNT1	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])
POMGNT2	Muscular dystrophy-dystroglycanopathy, type 8A (Walker-Warburg syndrome); Type 8C (limb-girdle muscular dystrophy, type 24 [LGMD R24])
POMK	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12
POMP	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma
POMT1	Muscular dystrophy-dystroglycanopathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy, type 11 [LGMD R11]) muscular dystrophy, type 11 [LGMD R11])
POMT2	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14]) muscular dystrophy, type 14 [LGMD R14])
POP1	Anauxetic dysplasia, type 2
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
POU1F1	Pituitary hormone deficiency, combined, type 1
POU3F4	Deafness, X-linked, type 2

Complete Investigation list

Gene	Disease
PPA2	Sudden cardiac failure, infantile
PPIB	Osteogenesis imperfecta, type 9
PPM1K	Maple syrup urine disease, mild variant
PPP1R15B	Microcephaly, short stature, and impaired glucose metabolism 2
PPT1	Ceroid lipofuscinosis, neuronal, type 1
PQBP1	Renpenning syndrome
PRCD	Retinitis pigmentosa, type 36
PRDM12	Neuropathy, hereditary sensory and autonomic, type VIII
PRDM5	Brittle cornea syndrome, type 2
PRDX1	Methylmalonic aciduria and homocystinuria, cblC type, digenic
PREPL	Myasthenic syndrome, congenital, type 22
PRF1	Hemophagocytic lymphohistiocytosis, familial, type 2
PRG4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome
PRICKLE1	Epilepsy, progressive myoclonic, type 1B
PRKCD	Autoimmune lymphoproliferative syndrome, type 3
PRKDC	Immunodeficiency 26, with or without neurologic abnormalities
PRKN	Parkinson disease, type 2 juvenile
PRKRA	Dystonia, type 16
PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures
PROC	Thrombophilia due to protein C deficiency, autosomal recessive
PRODH	Hyperprolinemia, type 1
PROM1	Retinitis pigmentosa, type 41
PROP1	Pituitary hormone deficiency, combined, type 2
PROS1	Thrombophilia due to protein S deficiency, autosomal recessive
PRPH2	Leber congenital amaurosis 18; Retinitis punctata albescens
PRPS1	Phosphoribosylpyrophosphate synthetase (PRS) deficiency
PRRX1	Agnathia-otocephaly complex
PRSS1	Trypsinogen deficiency
PRSS12	Intellectual developmental disorder, autosomal recessive 1
PRSS56	Microphthalmia, isolated, type 6
PRUNE1	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies
PRX	Charcot-Marie-Tooth disease, type 4F
PSAP	Combined SAP deficiency
PSAT1	Neu-Laxova syndrome, type 2
PSMB8	Autoinflammation, lipodystrophy, and dermatosis syndrome

Complete Investigation list

Gene	Disease
PSMC3IP	Ovarian dysgenesis 3
PSPH	Phosphoserine phosphatase deficiency
PTF1A	Pancreatic agenesis 2
PTH	Hypoparathyroidism, familial isolated, type 1
PTH1R	Chondrodysplasia, Blomstrand type; Eiken syndrome
PTPN23	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity
PTPRC	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive
PTPRO	Nephrotic syndrome, type 6
PTPRQ	Deafness, autosomal recessive, type 84A
PTRH2	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease
PTS	Hyperphenylalaninemia, BH4-deficient, type A
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia, type 1
PXDN	Anterior segment dysgenesis, type 7 with sclerocornea
PYCR1	Cutis laxa, autosomal recessive, type 2B
PYCR2	Leukodystrophy, hypomyelinating, type 10
PYGL	Glycogen storage disease, type 6
PYGM	McArdle disease
PYROXD1	Myopathy, myofibrillar, type 8
QARS1	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy
QDPR	Hyperphenylalaninemia, BH4-deficient, type C
RAB18	Warburg micro syndrome, type 3
RAB23	Carpenter syndrome
RAB27A	Griscelli syndrome, type 2
RAB28	Cone-rod dystrophy 18
RAB33B	Smith-McCort dysplasia 2
RAB39B	Intellectual developmental disorder, X-linked 72
RAB3GAP1	Warburg micro syndrome; Martsolf syndrome
RAB3GAP2	Warburg micro syndrome; Martsolf syndrome
RAD50	Nijmegen breakage syndrome-like disorder
RAD51C	Fanconi anemia, complementation group O
RAG1	Omenn syndrome; Severe combined immunodeficiency, B cell-negative
RAG2	Omenn syndrome; Severe combined immunodeficiency, B cell-negative
RAPSN	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11 associated with AChR deficiency
RARB	Microphthalmia, syndromic 12

Complete Investigation list

Gene	Disease
RARS1	Leukodystrophy, hypomyelinating, type 9
RARS2	Pontocerebellar hypoplasia, type 6
RASGRP1	Immunodeficiency, type 64
RAX	Microphthalmia, syndromic 16
RBBP8	Jawad syndrome; Seckel syndrome, type 2
RBCK1	Polyglucosan body myopathy 1 with or without immunodeficiency
RBM8A	Thrombocytopenia-absent radius syndrome
RBP3	Retinitis pigmentosa 66
RBP4	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome
RCBTB1	Retinal dystrophy with or without extraocular anomalies
RD3	Leber congenital amaurosis, type 12
RDH12	Leber congenital amaurosis, type 13
RDH5	Fundus albipunctatus
RDX	Deafness, autosomal recessive, type 24
RECQL4	Baller-Gerold syndrome; RAPADILINO syndrome; Rothmund-Thomson syndrome
REEP6	Retinitis pigmentosa 77
RELN	Lissencephaly 2 (Norman-Roberts type)
REN	Renal tubular dysgenesis
RETREG1	Neuropathy, hereditary sensory and autonomic, type 2B
RFT1	Congenital disorder of glycosylation, type In
RFX5	Bare lymphocyte syndrome, type 2
RFX6	Mitchell-Riley syndrome
RFXANK	Bare lymphocyte syndrome, type 2 complementation group B
RFXAP	Bare lymphocyte syndrome, type 2
RHAG	Anemia, hemolytic, Rh-null, regulator type
RHO	Retinitis pigmentosa, type 4; Retinitis punctata albescens
RIN2	Macs syndrome
RIPK4	Popliteal pterygium syndrome, Bartsocas-Papas type
RIPOR2	Deafness, autosomal recessive, type 104
RLBP1	Bothnia retinal dystrophy; Fundus albipunctatus
RMND1	Combined oxidative phosphorylation deficiency 11
RMRP	Anauxetic dysplasia 1
RNASEH1	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2
RNASEH2A	Aicardi-Goutieres syndrome, type 4
RNASEH2B	Aicardi-Goutieres syndrome, type 2

Complete Investigation list

Gene	Disease
RNASEH2C	Aicardi-Goutieres syndrome, type 3
RNASET2	Leukoencephalopathy, cystic, without megalencephaly
RNF168	RIDDLE syndrome
RNF216	Gordon Holmes syndrome
ROBO3	Gaze palsy, familial horizontal, with progressive scoliosis, type 1
ROGDI	Kohlschutter-Tonz syndrome
ROM1	Retinitis pigmentosa, type 7 digenic
ROR2	Robinow syndrome, autosomal recessive
RORC	Immunodeficiency, type 42
RP1	Retinitis pigmentosa, type 1
RP2	Retinitis pigmentosa, type 2 X-linked
RPE65	RPE65-related Leber congenital amaurosis/early-onset severe retinal dystrophy
RPGR	Retinitis pigmentosa, type 3 X-linked; Cone-rod dystrophy, X-linked, 1
RPGRIP1	Leber congenital amaurosis, type 6
RPGRIP1L	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome
RPL10	Intellectual developmental disorder, X-linked syndromic 35
RPS6KA3	Coffin-Lowry syndrome
RRM2B	Mitochondrial DNA depletion syndrome, type 8A (encephalomyopathic type with renal tubulopathy) and type 8B (MNGIE type)
RS1	Retinoschisis
RSPH1	Ciliary dyskinesia, primary, type 24
RSPH3	Ciliary dyskinesia, primary, type 32
RSPH4A	Ciliary dyskinesia, primary, type 11
RSPH9	Ciliary dyskinesia, primary, type 12
RSPO4	Anonychia congenita
RSPRY1	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type
RTEL1	Dyskeratosis congenita, autosomal recessive type 5
RTN4IP1	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures
RTTN	Microcephaly, short stature, and polymicrogyria with seizures
RUSC2	intellectual developmental disorder, autosomal recessive 61
RXYLT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10
RYR1	RYR1 related congenitalmyopathy
S1PR2	Deafness, autosomal recessive, type 68
SACS	Spastic ataxia, Charlevoix-Saguenay, type

Complete Investigation list

Gene	Disease
SAG	Oguchi disease, type 1
SAMD9	Tumoral calcinosis, familial, normophosphatemic
SAMHD1	Aicardi-Goutieres syndrome, type 5
SAR1B	Chylomicron retention disease
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis
SBDS	Shwachman-Diamond syndrome
SBF1	Charcot-Marie-Tooth disease, type 4B3
SBF2	Charcot-Marie-Tooth disease, type 4B2
SC5D	Lathosterolosis
SCARB2	Epilepsy, progressive myoclonic, type 4 with or without renal failure
SCARF2	Van den Ende-Gupta syndrome
SCN1B	Epileptic encephalopathy, early infantile, type 52
SCN4A	Myasthenic syndrome, congenital, type 16
SCN9A	Indifference to pain and autosomal recessive hereditary sensory neuropathy type 2D
SCNN1A	Pseudohypoaldosteronism, type 1
SCNN1B	Pseudohypoaldosteronism, type 1
SCNN1G	Pseudohypoaldosteronism, type 1
SCO1	Mitochondrial complex IV deficiency, nuclear type 4
SCO2	Mitochondrial complex IV deficiency, nuclear type 2
SCYL1	Spinocerebellar ataxia, autosomal recessive, type 21
SDCCAG8	Bardet-Biedl syndrome, type 16
SDHA	Mitochondrial respiratory chain complex II deficiency; Leigh syndrome
SDHAF1	Mitochondrial complex II deficiency
SDR9C7	Ichthyosis, congenital, autosomal recessive 13
SEC23A	Craniolenticulosutural dysplasia
SEC23B	Dyserythropoietic anemia, congenital, type 2
SEC24D	Cole-Carpenter syndrome 2
SECISBP2	Thyroid hormone metabolism, abnormal
SELENON	Multiminicore disease (rigid spine syndrome)
SEMA4A	Cone-rod dystrophy, type 10; Retinitis pigmentosa, type 35
SEPSECS	Pontocerebellar hypoplasia, type 2D
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome (MEGDEL)
SERPINA1	Alpha-1 antitrypsin deficiency
SERPINB7	Palmoplantar keratoderma, Nagashima type
SERPINB8	Peeling skin syndrome 5

Complete Investigation list

Gene	Disease
SERPINC1	Thrombophilia due to antithrombin III deficiency
SERPINE1	Plasminogen activator inhibitor-1 deficiency
SERPINF1	Osteogenesis imperfecta, type 6
SERPINF2	Alpha-2-plasmin inhibitor deficiency
SERPING1	Angioedema, hereditary, types 1 and 2
SERPINH1	Osteogenesis imperfecta, type 10
SETX	Spinocerebellar ataxia, autosomal recessive, type 1
SFRP4	Pyle disease
SFTPB	Surfactant metabolism dysfunction, pulmonary, type 1
SFXN4	Combined oxidative phosphorylation deficiency 18
SGCA	Limb-girdle muscular dystrophy, type 3 (LGMD R3)
SGCB	Limb-girdle muscular dystrophy, type 4 (LGMD R4)
SGCD	Limb-girdle muscular dystrophy, type 6 (LGMD R6)
SGCG	Limb-girdle muscular dystrophy, type 5 (LGMD R5)
SGPL1	Nephrotic syndrome, type 14
SGSH	Mucopolysaccharidosis, type 3A (Sanfilippo A)
SH2D1A	Lymphoproliferative syndrome, X-linked, type 1
SH3PXD2B	Frank-ter Haar syndrome
SH3TC2	Charcot-Marie-Tooth disease, type 4C
SI	Sucrase-isomaltase deficiency, congenital
SIL1	Marinesco-Sjogren syndrome
SIX6	Optic disc anomalies with retinal and/or macular dystrophy
SKIV2L	Trichohepatoenteric syndrome 2
SLC10A2	Bile acid malabsorption, primary
SLC11A2	Anemia, hypochromic microcytic, with iron overload 1
SLC12A1	Bartter syndrome, type 1
SLC12A3	Gitelman syndrome
SLC12A5	Epileptic encephalopathy, early infantile, 34
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy
SLC13A5	Epileptic encephalopathy, early infantile, 25
SLC16A1	Monocarboxylate transporter 1 deficiency
SLC16A2	Allan-Herndon-Dudley syndrome
SLC17A5	Salla disease
SLC18A3	Myasthenic syndrome, congenital, 21 presynaptic
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome
SLC19A3	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)

Complete Investigation list

Gene	Disease
SLC1A1	Dicarboxylic aminoaciduria
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly
SLC22A12	Hypouricemia, renal
SLC22A5	Carnitine deficiency, systemic primary
SLC24A1	Night blindness, congenital stationary (complete), type 1D, autosomal recessive
SLC24A4	Amelogenesis imperfecta, type IIA5
SLC24A5	Albinism, oculocutaneous, type 6
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria
SLC25A12	Epileptic encephalopathy, early infantile, type 39
SLC25A13	Citrullinemia, type 2 neonatal-onset; Citrullinemia, type 2 adult-onset
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome
SLC25A19	Microcephaly, Amish type; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)
SLC25A20	Carnitine-acylcarnitine translocase deficiency
SLC25A22	Developmental and epileptic encephalopathy 3
SLC25A26	Combined oxidative phosphorylation deficiency 28
SLC25A3	Mitochondrial phosphate carrier deficiency
SLC25A38	Anemia, sideroblastic, type 2 pyridoxine-refractory
SLC25A4	Mitochondrial DNA depletion syndrome, type 12B (cardiomyopathic type) AR
SLC25A46	Neuropathy, hereditary motor and sensory, type VIB
SLC26A2	Achondrogenesis, type 1B (diastrophic dysplasia)
SLC26A3	Diarrhea 1 secretory chloride, congenital
SLC26A4	Deafness, autosomal recessive, type 4; Pendred syndrome
SLC26A5	Deafness, autosomal recessive, type 61
SLC27A4	Ichthyosis prematurity syndrome
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome
SLC2A1	GLUT1 deficiency syndrome 1, infantile onset, severe
SLC2A10	Arterial tortuosity syndrome
SLC2A2	Fanconi-Bickel syndrome
SLC2A9	Hypouricemia, renal, type 2
SLC30A10	Hypermanganesemia with dystonia, type 1
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration
SLC34A1	Hypercalcemia, infantile, type 2
SLC34A2	Pulmonary alveolar microlithiasis
SLC34A3	Hypophosphatemic rickets with hypercalciuria

Complete Investigation list

Gene	Disease
SLC35A1	Congenital disorder of glycosylation, type 2F
SLC35A3	Arthrogryposis, impaired intellectual development, and seizures
SLC35C1	Congenital disorder of glycosylation, type 2C
SLC35D1	Schneckenbecken dysplasia
SLC37A4	Glycogen storage disease, type 1B
SLC38A8	Foveal hypoplasia 2 with or without optic nerve misrouting and/or anterior segment dysgenesis
SLC39A13	Ehlers-Danlos syndrome, spondylodysplastic type, 3
SLC39A14	Hypermanganesemia with dystonia 2
SLC39A4	Acrodermatitis enteropathica
SLC39A8	Congenital disorder of glycosylation, type IIn
SLC3A1	Cystinuria
SLC45A1	Intellectual developmental disorder with neuropsychiatric features
SLC45A2	Albinism, oculocutaneous, type 4
SLC46A1	Folate malabsorption, hereditary
SLC4A1	Distal renal tubular acidosis
SLC4A11	Corneal endothelial dystrophy, autosomal recessive
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities
SLC52A2	Brown-Vialetto-Van Laere syndrome, type 2
SLC52A3	Brown-Vialetto-Van Laere syndrome, type 1
SLC5A1	Glucose/galactose malabsorption
SLC5A2	Renal glucosuria
SLC5A5	Thyroid dyshormonogenesis, type 1
SLC5A7	Myasthenic syndrome, congenital, type 20 presynaptic
SLC6A17	intellectual developmental disorder, autosomal recessive 48
SLC6A19	Hartnup disorder
SLC6A3	Parkinsonism-dystonia, infantile
SLC6A5	Hyperekplexia, type 3
SLC6A8	Cerebral creatine deficiency syndrome, type 1
SLC6A9	Glycine encephalopathy with normal serum glycine
SLC7A14	Retinitis pigmentosa 68
SLC7A7	Lysinuric protein intolerance
SLC7A9	Cystinuria
SLC9A3	Diarrhea 8 secretory sodium, congenital
SLC9A6	Intellectual developmental disorder, X-linked syndromic, Christianson type
SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal recessive, type 2

Complete Investigation list

Gene	Disease
SLITRK6	Deafness and myopia
SLURP1	Meleda disease
SLX4	Fanconi anemia, complementation group P
SMARCAL1	Schimke immunoosseous dysplasia
SMARCD2	Specific granule deficiency 2
SMG9	Heart and brain malformation syndrome
SMN1	Spinal muscular atrophy
SMN2	{Spinal muscular atrophy, type III, modifier of}
SMOC1	Microphthalmia. with limb anomalies
SMOC2	Dentin dysplasia, type 1 with microdontia and misshapen teeth
SMPD1	Niemann-Pick disease, type A; Niemann-Pick disease, type B
SMS	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome
SNX10	Osteopetrosis, autosomal recessive, type 8
SNX14	Spinocerebellar ataxia, autosomal recessive, type 20
SOBP	Impaired intellectual development, anterior maxillary protrusion, and strabismus
SOD1	Spastic tetraplegia and axial hypotonia, progressive; Amyotrophic lateral sclerosis, type 1
SOHLH1	Ovarian dysgenesis 5
SOST	Sclerosteosis, type 1; Van Buchem disease
SOX18	Hypotrichosis-lymphedema-telangiectasia syndrome
SOX3	Panhypopituitarism, X-linked
SP110	Hepatic venoocclusive disease with immunodeficiency
SP7	Osteogenesis imperfecta, type XII
SPAG1	Ciliary dyskinesia, primary, type 28
SPARC	Osteogenesis imperfecta, type XVII
SPART	Spactic paraplegia, type 20 autosomal recessive
SPATA5	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities
SPATA7	Leber congenital amaurosis, type 3
SPEG	Centronuclear myopathy, type 5
SPG11	Amyotrophic lateral sclerosis, type 5 juvenile
SPG21	Mast syndrome
SPG7	Spastic paraplegia, type 7 autosomal recessive

Complete Investigation list

Gene	Disease
SPINK1	Tropical calcific pancreatitis
SPINK5	Netherton syndrome
SPINT2	Diarrhea 3 secretory sodium, congenital, syndromic
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency
SPRTN	Ruijs-Aalfs syndrome
SPTA1	Pyropoikilocytosis; Apherocytosis, type 3
SPTBN2	Spinocerebellar ataxia, autosomal recessive, type 14
SPTBN4	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness
SQSTM1	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset
SRD5A2	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)
SRD5A3	Congenital disorder of glycosylation, type 1Q; Kahrizi syndrome
SRPX2	Rolandic epilepsy, impaired intellectual development, and speech dyspraxia
ST14	Ichthyosis, congenital, autosomal recessive, type 11
ST3GAL3	Intellectual developmental disorder, autosomal recessive 12 ; Developmental and epileptic encephalopathy 15
ST3GAL5	Salt and pepper developmental regression syndrome
STAC3	Native American myopathy
STAG3	Premature ovarian failure 8
STAMBP	Microcephaly-capillary malformation syndrome
STAR	Lipoid adrenal hyperplasia
STAT1	Immunodeficiency, type 31B, mycobacterial and viral infections
STAT2	Immunodeficiency, type 44
STAT5B	Laron syndrome with immunodeficiency
STIL	Microcephaly, type 7 primary, autosomal recessive
STIM1	Immunodeficiency, type 10
STK4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations
STRA6	Microphthalmia, isolated, with coloboma, type 8
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy
STRC	Deafness, autosomal recessive, type 16
STUB1	Spinocerebellar ataxia, autosomal recessive, type 16
STX11	Hemophagocytic lymphohistiocytosis, familial, type 4
STXBP2	Hemophagocytic lymphohistiocytosis, familial, type 5

Complete Investigation list

Gene	Disease
SUCLA2	Mitochondrial DNA depletion syndrome, type 5 (encephalomyopathic with or without methylmalonic aciduria)
SUCLG1	Mitochondrial DNA depletion syndrome, type 9 (encephalomyopathic, type with methylmalonic aciduria)
SUFU	Joubert syndrome, type 32
SUGCT	Glutaric aciduria, type 3
SULT2B1	Ichthyosis, congenital, autosomal recessive, type 14
SUMF1	Multiple sulfatase deficiency
SUN5	Spermatogenic failure, type 16
SUOX	Sulfite oxidase deficiency
SURF1	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders
SYNE1	Spinocerebellar ataxia, autosomal recessive, type 8
SYNE4	Deafness, autosomal recessive, type 76
SYNJ1	Epileptic encephalopathy, early infantile, 53
SYP	Intellectual developmental disorder, X-linked 96
SYT14	Spinocerebellar ataxia, autosomal recessive, type 11
SZT2	Epileptic encephalopathy, early infantile, 18
TAC3	Hypogonadotropic hypogonadism, type 10 with or without anosmia
TACO1	Mitochondrial complex IV deficiency, nuclear type 8
TACR3	Hypogonadotropic hypogonadism, type 11 with or without anosmia
TACSTD2	Corneal dystrophy, gelatinous drop-like
TAF1	Dystonia-Parkinsonism, X-linked
TAF13	Intellectual developmental disorder, autosomal recessive 60
TAF2	Intellectual developmental disorder, autosomal recessive 40
TAF6	Alazami-Yuan syndrome
TALDO1	Transaldolase deficiency
TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration
TAP1	Bare lymphocyte syndrome, type 1
TAP2	Bare lymphocyte syndrome, type 1 due to TAP2 deficiency
TAPBP	Bare lymphocyte syndrome, type 1
TAPT1	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type
TAT	Tyrosinemia, type 2
TAZ	Barth syndrome

Complete Investigation list

Gene	Disease
TBC1D20	Warburg micro syndrome 4
TBC1D23	Pontocerebellar hypoplasia, type 11
TBC1D24	DOORS (deafness, onychodystrophy, osteodystrophy, impaired intellectual development, and seizures) syndrome; Epileptic encephalopathy, early infantile, type 16; Deafness, autosomal recessive, type 86
TBC1D7	Macrocephaly/megalencephaly syndrome, autosomal recessive
TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum
TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy; Hypoparathyroidism-retardation-dysmorphism syndrome; Kenny-Caffey syndrome, type 1
TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3
TBX15	Cousin syndrome
TBX19	Congenital isolated adrenocorticotropic hormone deficiency
TBXAS1	Ghosal syndrome
TCAP	Limb-girdle muscular dystrophy, type 7 (LGMD R7)
TCIRG1	Osteopetrosis, autosomal recessive, type 1
TCN2	Transcobalamin II deficiency
TCTN1	Joubert syndrome, type 13
TCTN2	Joubert syndrome, type 24; ?Meckel syndrome, type 8
TCTN3	Joubert syndrome 18
TDP1	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy
TDP2	Spinocerebellar ataxia, autosomal recessive, type 23
TDRD7	Cataract 36
TECPR2	Spastic paraplegia, type 49 autosomal recessive
TECR	intellectual developmental disorder, autosomal recessive, type 14
TECRL	Ventricular tachycardia, catecholaminergic polymorphic, 3
TECTA	Deafness, autosomal recessive, type 21
TELO2	You-Hoover-Fong syndrome
TENM3	Microphthalmia, isolated, with coloboma 9
TERT	Dyskeratosis congenita, autosomal recessive, type 4
TEX15	Spermatogenic failure, type 25
TF	Atransferrinemia
TFR2	Hemochromatosis, type 3
TFRC	Immunodeficiency, type 46

Complete Investigation list

Gene	Disease
TG	Thyroid dyshormonogenesis, type 3
TGDS	Catel-Manzke syndrome
TGM1	Ichthyosis, congenital, autosomal recessive, type 1
TGM5	Peeling skin syndrome, type 2
TH	Segawa syndrome, recessive
THOC2	Intellectual developmental disorder, X-linked 12
THOC6	Beaulieu-Boycott-Innes syndrome
THRB	Thyroid hormone resistance, autosomal recessive
TIMM50	3-methylglutaconic aciduria, type 9
TIMM8A	Mohr-Tranebjaerg syndrome
TIMMDC1	Mitochondrial complex I deficiency, nuclear type 31
TJP2	Cholestasis, progressive familial intrahepatic, type 4
TK2	Mitochondrial DNA depletion syndrome , type 2 (myopathic type)
TKT	Short stature, developmental delay, and congenital heart defects
TLE6	Preimplantation embryonic lethality
TMC1	Deafness, autosomal recessive, type 7
TMC6	Epidermodysplasia verruciformis
TMC8	Epidermodysplasia verruciformis
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1
TMEM107	Meckel syndrome, type 13; Orofaciodigital syndrome, type 16
TMEM126A	Optic atrophy 7
TMEM126B	Mitochondrial complex I deficiency, nuclear type 29
TMEM138	Joubert syndrome 16
TMEM165	Congenital disorder of glycosylation, type 2K
TMEM199	Congenital disorder of glycosylation, type 2P
TMEM216	Joubert syndrome, type 2; Meckel syndrome, type 2
TMEM231	Joubert syndrome, type 20; Meckel syndrome,type 11
TMEM237	Joubert syndrome, type 14
TMEM260	Structural heart defects and renal anomalies syndrome
TMEM38B	Osteogenesis imperfecta, type XIV
TMEM67	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2
TMIE	Deafness, autosomal recessive, type 6
TMPRSS15	Enterokinase deficiency
TMPRSS3	Deafness, autosomal recessive, type 45573
TMPRSS6	Iron-refractory iron deficiency anemia

Complete Investigation list

Gene	Disease
TMTC3	Lissencephaly 8
TNFRSF11A	Osteopetrosis, autosomal recessive, type 7
TNFRSF11B	Paget disease of bone, type 5 juvenile-onset
TNFRSF13B	Immunodeficiency, common variable, type 2
TNFSF11	Osteopetrosis, autosomal recessive, type 2
TNIK	Intellectual developmental disorder, autosomal recessive 54
TNNT1	Nemaline myopathy , type 5 Amish type
TNXB	Ehlers-Danlos syndrome, classic-like
TOE1	Pontocerebellar hypoplasia, type 7
TOP3A	Microcephaly, growth restriction, and increased sister chromatid exchange 2
TP53RK	Galloway-Mowat syndrome 4
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency
TPK1	Episodic encephalopathy due to thiamine pyrophosphokinase deficiency
TPM3	Nemaline myopathy, type 1; Congenital fiber-type disproportion myopathy
TPO	Thyroid dyshormonogenesis, type 2A
TPP1	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7
TPRN	Deafness, autosomal recessive, type 79
TRAF3IP1	Senior-Loken syndrome, type 9
TRAIP	Seckel syndrome, type 9
TRAPPC11	Limb-girdle muscular dystrophy, type 18 (LGMD R18)
TRAPPC12	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity
TRAPPC6B	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy
TRAPPC9	Intellectual developmental disorder, autosomal recessive 13
TRDN	Ventricular tachycardia, catecholaminergic polymorphic, type 5 with or without muscle weakness
TREM2	Nasu-Hakola disease
TREX1	Aicardi-Goutieres syndrome, type 1
TRHR	Hypothyroidism, congenital, nongoitrous, type 7
TRIM2	Charcot-Marie-Tooth disease, type 2R
TRIM32	Limb-girdle muscular dystrophy, type 8 (LGMD R8)
TRIM37	Mulibrey nanism
TRIOBP	Deafness, autosomal recessive, type 28
TRIP11	Achondrogenesis, type 1A

Complete Investigation list

Gene	Disease
TRIP13	Mosaic variegated aneuploidy syndrome 3
TRIP4	Spinal muscular atrophy with congenital bone fractures 1
TRIT1	Combined oxidative phosphorylation deficiency 35
TRMT10A	Microcephaly, short stature, and impaired glucose metabolism 1
TRMT10C	Combined oxidative phosphorylation deficiency 30
TRMT5	Combined oxidative phosphorylation deficiency 26
TRMU	Liver failure, transient infantile
TRNT1	Retinitis pigmentosa and erythrocytic microcytosis
TRPM1	Night blindness, congenital stationary (complete), type 1C, autosomal recessive
TRPM6	Familial hypomagnesemia with secondary hypocalcemia
TRPV6	Hyperparathyroidism, transient neonatal
TSEN15	Pontocerebellar hypoplasia, type 2F
TSEN2	Pontocerebellar hypoplasia, type 2B
TSEN34	Pontocerebellar hypoplasia type 2C
TSEN54	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4
TSFM	Combined oxidative phosphorylation deficiency, type 3
TSHB	Hypothyroidism, congenital, nongoitrous, type 4
TSHR	Hypothyroidism, congenital, nongoitrous, type 1
TSPAN7	Intellectual developmental disorder, X-linked 58
TSPYL1	Sudden infant death with dysgenesis of the testes syndrome
TTC19	Mitochondrial complex III deficiency, nuclear type 2
TTC21B	Short-rib thoracic dysplasia, type 4 with or without polydactyly
TTC37	Trichohepatoenteric syndrome 1
TTC7A	Gastrointestinal defects and immunodeficiency syndrome
TTC8	Bardet-Biedl syndrome, type 8
TTI2	Intellectual developmental disorder, autosomal recessive 39
TTLL5	Cone-rod dystrophy 19
TTN	Limb-girdle muscular dystrophy type 10 (LGMDR10); Early-onset myopathy with fatal cardiomyopathy (Salih myopathy)
TTPA	Ataxia with isolated vitamin E deficiency
TUBA8	Cortical dysplasia, complex, with other brain malformations, type 8
TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, type 3
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, type 1
TUFM	Combined oxidative phosphorylation deficiency 4
TULP1	Leber congenital amaurosis, type 15
TUSC3	Intellectual developmental disorder, autosomal recessive 7

Complete Investigation list

Gene	Disease
TWIST2	Focal facial dermal dysplasia, type 3 (Setleis type)
TWNK	Mitochondrial DNA depletion syndrome, type 7 (hepatocerebral type); Perrault syndrome type 5
TXNL4A	Burn-McKeown syndrome
TYK2	Immunodeficiency, type 35
TYMP	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)
TYR	Oculocutaneous albinism (OCA) type 1A; OCA type 1B
TYROBP	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy, type 1 (Nasu-Hakola disease)
TYRP1	Albinism, oculocutaneous, type 3
UBA1	Spinal muscular atrophy, X-linked 2, infantile
UBA5	Epileptic encephalopathy, early infantile, 44
UBE2A	Intellectual developmental disorder, X-linked syndromic, Nascimento type
UBE2T	Fanconi anemia, complementation group T
UBE3A	Angelman syndrome
UBE3B	Kaufman oculocerebrofacial syndrome
UBR1	Johanson-Blizzard syndrome
UCHL1	Spastic paraplegia, type 79 autosomal recessive
UFM1	Leukodystrophy, hypomyelinating, type 14
UGT1A1	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2
UMPS	Orotic aciduria
UNC13D	Hemophagocytic lymphohistiocytosis, familial, type 3
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies, type 2
UNG	Immunodeficiency with hyper IgM, type 5
UPB1	Beta-ureidopropionase deficiency
UPF3B	Intellectual developmental disorder, X-linked syndromic 14
UQCRB	Mitochondrial complex III deficiency, nuclear, type 3
UQCRC2	Mitochondrial complex III deficiency, nuclear type 5
UQCRQ	Mitochondrial complex III deficiency, nuclear, type 4
UROD	Porphyria cutanea tarda
UROS	Porphyria, congenital erythropoietic
USB1	Poikiloderma with neutropenia
USH1C	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A
USH1G	Usher syndrome, type 1G
USH2A	Usher syndrome, type 2A
USP18	Pseudo-TORCH syndrome 2

Complete Investigation list

Gene	Disease
USP9X	Intellectual developmental disorder, X-linked 99
UVSSA	UV-sensitive syndrome, type 3
VAC14	Striatonigral degeneration, childhood-onset
VARS1	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy
VARS2	Combined oxidative phosphorylation deficiency 20
VDR	Rickets, vitamin D-resistant, type 2A
VIPAS39	Arthrogryposis, renal dysfunction and cholestasis, type 2
VKORC1	Vitamin K-dependent clotting factors, combined deficiency of, type 2
VLDLR	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1
VPS13A	Choreoacanthocytosis
VPS13B	Cohen syndrome
VPS13C	Parkinson disease 23 autosomal recessive, early onset
VPS33B	Arthrogryposis, renal dysfunction and cholestasis, type 1
VPS37A	Spastic paraplegia, type 53 autosomal recessive
VPS45	Neutropenia, severe congenital, type 5
VPS53	Pontocerebellar hypoplasia, type 2E
VRK1	Pontocerebellar hypoplasia, type 1A
VSX2	Microphthalmia with coloboma 3; Isolated microphthalmia 2
VWF	von Willibrand disease, type 3
WARS2	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures
WAS	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked
WASHC4	Intellectual developmental disorder, autosomal recessive 43
WASHC5	Ritscher-Schinzel syndrome, type 1
WDR19	Nephronophthisis, type 13; Senior-Loken syndrome, type 8
WDR35	Cranioectodermal dysplasia 2
WDR45B	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures
WDR62	Microcephaly, type 2 primary, autosomal recessive, with or without cortical malformations
WDR72	Amelogenesis imperfecta, type 2A3 (hypomaturation type)
WDR73	Galloway-Mowat syndrome 1
WDR81	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome, type 2; Hydrocephalus, congenital, 3, with brain anomalies
WEE2	Oocyte maturation defect 5

Complete Investigation list

Gene	Disease
WFS1	Wolfram syndrome, type 1
WHRN	Usher syndrome, type 2D; Deafness, autosomal recessive, type 31
WIPF1	Wiskott-Aldrich syndrome 2
WNK1	Neuropathy, hereditary sensory and autonomic, type 2
WNT1	Osteogenesis imperfecta, type XV
WNT10A	Ectodermal dysplasia 16 (odontoonychodermal dysplasia)
WNT10B	Split-hand/foot malformation, type 6
WNT3	Tetra-amelia syndrome
WNT7A	Fuhrmann syndrome
WRAP53	Dyskeratosis congenita, autosomal recessive, type 3
WRN	Werner syndrome
WWOX	Epileptic encephalopathy, early infantile, type 28; Spinocerebellar ataxia, autosomal recessive, type 12
XDH	Xanthinuria, type 1
XIAP	Lymphoproliferative syndrome, X-linked, 2
XPA	Xeroderma pigmentosum, group A
XPC	Xeroderma pigmentosum, group C
XPNPEP3	Nephronophthisis-like nephropathy, type 1
XRCC4	Short stature, microcephaly, and endocrine dysfunction
XYLT1	Desbuquois dysplasia, type 2
XYLT2	Spondyloocular syndrome
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia, type 2
YY1AP1	Grange syndrome
ZAP70	Autoimmune disease, multisystem, infantile-onset, type 2; Immunodeficiency, type 48
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 2
ZC3H14	intellectual developmental disorder, autosomal recessive, type 56
ZDHHC9	Intellectual developmental disorder, X-linked syndromic, Raymond type
ZFYVE26	Spastic paraplegia, type 15 autosomal recessive
ZMPSTE24	Mandibuloacral dysplasia with, type B lipodystrophy
ZMYND10	Ciliary dyskinesia, primary, type 22
ZNF408	Retinitis pigmentosa, type 72
ZNF423	Joubert syndrome, type 19
ZNF469	Brittle cornea syndrome, type 1
ZNF711	Intellectual developmental disorder, X-linked 97
ZNHIT3	PEHO syndrome

Complete Investigation list

Gene	Disease
ZP1	Oocyte maturation defect, type 1