ALTAMEDICA

ARTEMISIA

ELENCO GENI E PATOLOGIE

Test del portatore

2025

ALTAMEDICA
Artemisia SpA - Viale Liegi, 45 – Roma
P.I.00929551000 - C.F.00768340580
Certificazione Qualità UNI EN ISO 9001:2015 Reg. 7662

Servizio di Genetica Medica
Responsabile Dott. Alvaro Mesoraca
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Carrier Screening – Test del portatore

GENI	PATOLOGIE
CFTR	Fibrosi cistica
GJB2	Sordità congenita
SMA	Atrofia Muscolare Spinale
DMD	Distrofia muscolare di Duchenne
FMR1	X-Fragile

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Carrier Screening – Test del portatore 2

GENI	PATOLOGIE
AAAS	Triple-A syndrome (achalasia- addisonianism-alacrimia)
ABCA12	Ichthyosis, congenital, autosomal recessive, type 4A; ICAr, type 4B (harlequin)
ABCA3	Surfactant metabolism dysfunction, pulmonary, type 3
ABCB11	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2
ABCB4	Cholestasis, progressive familial intrahepatic, type 3
ABCC8	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)
ABCD1	Adrenoleukodystrophy
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
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
ACAT1	Alpha-methylacetoacetic aciduria (3- ketothiolase deficiency)
ACOX1	Peroxisomal acyl-CoA oxidase deficiency
ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type
ADGRV1	Usher syndrome, type 2C
AGA	Aspartylglucosaminuria (glycosylasparaginase deficiency)
AGL	Glycogen storage disease, type 3
AGPS	Rhizomelic chondrodysplasia punctata, type 3
AGXT	Hyperoxaluria, primary, type 1
AHI1	Joubert syndrome, type 3
AIRE	Autoimmune polyendocrinopathy syndrome, type 1

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GENI	PATOLOGIE
ALDH3A2	Sjogren-Larsson syndrome
ALDH7A1	Epilepsy, pyridoxine-dependent
ALDOB	Fructose intolerance, hereditary
ALG1	Congenital disorder of glycosylation, type 1K
ALG6	Congenital disorder of glycosylation, type 1C
ALMS1	Alstrom syndrome
ALPL	Hypophosphatasia, infantile/ childhood
AMT	Glycine encephalopathy
AP1S1	MEDNIK syndrome
AP3B1	Hermansky-Pudlak syndrome, type 2
AR	Androgen insensitivity syndrome, complete
ARG1	Argininemia (arginase deficiency)
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)
ARSL	Chondrodysplasia punctata, brachytelephalangic (ARSL o ARSE)
ARX	Epileptic encephalopathy, early infantile, type 1; ArX-related developmental disorders
ASL	Argininosuccinic aciduria
ASNS	Asparagine synthetase deficiency
ASPA	Canavan disease
ASS1	Citrullinemia, type 1
ATM	Ataxia-telangiectasia
ATP6V1B1	Renal tubular acidosis with deafness
ATP7A	Menkes disease; Occipital horn syndrome
ATP7B	Wilson disease
ATP8B1	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1

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ATRX	Mental retardation-hypotonic facies syndrome, X-linked; Alpha- thalassemia/mental retardation syndrome
AUH	3-methylglutaconic aciduria, type 1
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
BBS7	Bardet-Biedl syndrome, type 7
BBS9	Bardet-Biedl syndrome, type 9
BCKDHA	Maple syrup urine disease, type 1A
BCKDHB	Maple syrup urine disease, type 1B
BCS1L	BCS1L-related disorders, including Leigh syndrome
BLM	Bloom syndrome
BSND	Bartter syndrome, type 4A
BTD	Biotinidase deficiency
BTK	Agammaglobulinemia X-linked, type 1
CANT1	Desbuquois dysplasia, type 1; Epiphyseal dysplasia, multiple, type 7
CAPN3	Limb-girdle muscular dystrophy, type 1 (LGMD r1)
CBS	Homocystinuria due to cystathionine beta-synthase
CC2D2A	Joubert syndrome, type 9; Meckel syndrome, type 6
CD3D	Immunodeficiency, type 19
CD3E	Immunodeficiency, type 18
CD40LG	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)
CDH23	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D
CEP290	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10

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CERKL	Retinitis pigmentosa, type 26
CFTR	Cystic fibrosis
CHAT	Myasthenic syndrome, congenital, type 6, presynaptic
CHM	Choroideremia
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
CLN3	Ceroid lipofuscinosis, neuronal, type 3
CLN5	Ceroid lipofuscinosis, neuronal, type 5
CLN6	Ceroid lipofuscinosis, neuronal, type 6
CLN8	Ceroid lipofuscinosis, neuronal, type 8
CLRN1	Usher syndrome, type 3A
CNGB3	Achromatopsia, type 3
COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type
COL4A3	Alport syndrome, autosomal recessive, type 2
COL4A4	Alport syndrome, autosomal recessive, type 2
COL4A5	Alport syndrome, X-linked
COL7A1	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 2; Leigh syndrome due to cytochrome c oxidase deficiency
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
CRB1	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8
CRTAP	Osteogenesis imperfecta, type 7
CSTB	Epilepsy, progressive myoclonic type 1A (Unverricht and Lundborg)
CTNS	Nephropathic cystinosis

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CTSD	Ceroid lipofuscinosis, neuronal, type 10
CTSK	Pycnodysostosis
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
CYP27A1	Cerebrotendinous xanthomatosis
CYP27B1	Vitamin D-dependent rickets, type 1
DBT	Maple syrup urine disease, type 2
DCLRE1C	Omenn syndrome; Severe combined immunodeficiency, Athabascan type
DGUOK	DGUOK-related mitochondrial DNA depletion syndrome
DHCR7	Smith-Lemli-Opitz syndrome
DHDDS	Retinitis pigmentosa, type 59
DKC1	Dyskeratosis congenita, X-linked
DLD	Dihydrolipoamide dehydrogenase deficiency
DLL3	Spondylocostal dysostosis type 1
DMD	Duchenne/Becker muscular dystrophy
DNAH5	Ciliary dyskinesia, primary, type 3, with or without situs inversus
DNAI1	Ciliary dyskinesia, primary, type 1, with or without situs inversus
DNAI2	Ciliary dyskinesia, primary, type 9, with or without situs inversus
DNAJC19	3-methylglutaconic aciduria, type 5
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 1

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DOK7	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10
DPYD	Dihydropyrimidine dehydrogenase deficiency
DSP	Cardiomyopathy, dilated, with woolly hair and keratoderma; Epidermolysis bullosa, lethal acantholytic
DUOX2	Thyroid dyshormonogenesis, type 6
DYNC2H1	Short-rib thoracic dysplasia, type 3, with or without polydactyly
DYSF	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD r2)
EDA	Ectodermal dysplasia, type 1, hypohidrotic, X-linked
EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type
EIF2AK3	Wolcott-rallison syndrome
EIF2B1	Leukoencephalopathy with vanishing white matter
EIF2B2	Leukoencephalopathy with vanishing white matter
EIF2B3	Leukoencephalopathy with vanishing white matter
EIF2B4	Leukoencephalopathy with vanishing white matter
EIF2B5	Leukoencephalopathy with vanishing white matter
ELP1	Familial dysautonomia
EMD	Emery-Dreifuss muscular dystrophy, type 1, X-linked
ERCC2	Trichothiodystrophy, type 1
ERCC6	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1
ERCC8	Cockayne syndrome, type A
ESCO2	Roberts syndrome
ETFA	Glutaric acidemia, type 2A
ETFB	Glutaric acidemia, type 2B
ETFDH	Glutaric acidemia, type 2C
ETHE1	Ethylmalonic encephalopathy
EVC	Ellis-van Creveld syndrome

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EVC2	Ellis-van Creveld syndrome
EXOSC3	Pontocerebellar hypoplasia, type 1B
EYS	Retinitis pigmentosa, type 25
F11	Factor XI deficiency
F8	Hemophilia A
F9	Hemophilia B
FAH	Tyrosinemia, type 1
FANCA	Fanconi anemia, complementation group A
FANCC	Fanconi anemia, complementation group C
FANCG	Fanconi anemia, complementation group G
FH	Fumarase deficiency
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])
FMR1	Fragile X syndrome
FOXN1	T-cell immunodeficiency, congenital alopecia and nail dystrophy
FRAS1	Fraser syndrome, type 1
FREM2	Fraser syndrome, type 2
FUCA1	Fucosidosis
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

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GENI	PATOLOGIE
GALNS	Mucopolysaccharidosis, type 4A
GALT	Galactosemia
GAMT	Cerebral creatine deficiency syndrome, type 2
GBA	Gaucher disease
GBE1	Glycogen storage disease, type 4
GCDH	Glutaricaciduria, type 1
GCH1	Hyperphenylalaninemia, BH4- deficient, type B
GCSH	Glycine encephalopathy
GDF5	Chondrodysplasia, Grebe type
GFM1	Combined oxidative phosphorylation deficiency, type 1
GJB1	Charcot-Marie-Tooth neuropathy, X- linked dominant, type 1
GJB2	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6
GJB3	hearing loss
GJB6	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6
GLA	Fabry disease
GLB1	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)
GLDC	Glycine encephalopathy
GLE1	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease
GNE	Inclusion body myopathy, type 2 (Nonaka myopathy)
GNPAT	Rhizomelic chondrodysplasia punctata, type 2
GNPTAB	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta
GNPTG	Mucolipidosis III gamma
GNS	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)
GORAB	Geroderma osteodysplasticum
GRHPR	Hyperoxaluria, primary, type 2

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GENI	PATOLOGIE
GSS	Glutathione synthetase deficiency
GUSB	Mucopolysaccharidosis, type 7
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
HAX1	Neutropenia, severe congenital, type 3, autosomal recessive
HBB	HBB-related hemoglobinopathy
HCFC1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)
HEXA	Tay-Sachs disease
HEXB	Sandhoff disease, infantile, juvenile, and adult forms
HFE	Hemochromatosis type 1
HFE2	Hereditary hemochromatosis type 2A, HFE2-related
HGD	Alkaptonuria
HGSNAT	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)
HLCS	Holocarboxylase synthetase deficiency
HMGCL	HMG-CoA lyase deficiency
HOGA1	Hyperoxaluria, primary, type 3
HPD	Tyrosinemia, type 3
HPRT1	Lesch-Nyhan syndrome
HPS1	Hermansky-Pudlak syndrome, type 1
HPS3	Hermansky-Pudlak syndrome, type 3
HSD17B10	HSD10 mitochondrial disease
HSD17B3	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
HSD17B4	D-bifunctional protein deficiency

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HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency
HYLS1	Hydrolethalus syndrome
IDS	Mucopolysaccharidosis, type 2
IDUA	Mucopolysaccharidosis type 1
IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S
IKBKAP	Familial Dysautonomia
IL2RG	Severe combined immunodeficiency, X-linked
ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis
ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia
IVD	Isovaleric acidemia
JAK3	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B- positive type
KCNJ1	Bartter syndrome, type 2
KCNJ11	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)
L1CAM	L1 Syndrome
LAMA2	LAMA2-related muscular dystrophy
LAMA3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LAMB3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LAMC2	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LARGE1	Muscular dystrophy- dystroglycanopathy, type 6A and 6B
LCA5	Leber congenital amaurosis, type 5
LHCGR	Leydig cell hypoplasia
LHX3	Pituitary hormone deficiency, combined, type 3
LIFR	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome
LIG4	LIG4 syndrome
LIPA	Lysosomal acid lipase deficiency

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LOXHD1	Deafness, autosomal recessive, type 77
LPL	Lipoprotein lipase deficiency
LRP2	Donnai-Barrow syndrome
LRPPRC	Leigh syndrome, French-Canadian type
LYST	Chediak-Higashi syndrome
MAN2B1	Alpha-mannosidosis
MCCC1	3-Methylcrotonyl-CoA carboxylase deficiency, type 1
MCCC2	3-Methylcrotonyl-CoA carboxylase deficiency, type 2
MCEE	Methylmalonyl-CoA epimerase deficiency
MCOLN1	Mucolipidosis type 4
MECP2	Encephalopathy, neonatal severe; rett syndrome
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy
MEFV	Familial Mediterranean fever
MESP2	Spondylocostal dysostosis, type 2, autosomal recessive
MFSD8	Ceroid lipofuscinosis, neuronal, type 7
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
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
MOCS1	Molybdenum cofactor deficiency A
MOCS2	Molybdenum cofactor deficiency B

GENI	PATOLOGIE
MPI	Congenital disorder of glycosylation, type 1B
MPL	Thrombocytopenia, congenital amegakaryocytic
MPV17	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE
MTHFR	Homocystinuria due to MTHFr deficiency
MTM1	Myotubular myopathy, X-linked
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
MUT	Methylmalonic aciduria mut(0) type, MUT-related
MVK	Mevalonic aciduria
MYO7A	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2
NAGLU	Mucopolysaccharidosis, type 3B (Sanfilippo B)
NAGS	N-acetylglutamate synthase deficiency
NBN	Nijmegen breakage syndrome
NDUFAF2	Mitochondrial complex I deficiency, nuclear type 10
NDUFS4	Mitochondrial complex I deficiency, nuclear type 1
NDUFS6	Mitochondrial complex I deficiency, nuclear type 9
NDUFS7	Mitochondrial complex I deficiency, nuclear type 3
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4
NEB	Nemaline myopathy type 2
NEU1	Sialidosis, type 1 and type 2
NPC1	Niemann-Pick disease, type C1
NPC2	Niemann-pick disease, type C2
NPHP1	Joubert syndrome type 4

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NPHS1	Nephrotic syndrome, type 1
NPHS2	Nephrotic syndrome, type 2
NR0B1	Adrenal hypoplasia, congenital
NTRK1	Insensitivity to pain, congenital, with anhidrosis
OCRL	Lowe Syndrome; Dent disease type 2
OPA3	3-methylglutaconic aciduria, type 3
OSTM1	Osteopetrosis, autosomal recessive type 5
OTC	Ornithine transcarbamylase deficiency
P3H1	Osteogenesis imperfecta, type 8
PAH	Phenylketonuria
PANK2	Neurodegeneration with brain iron accumulation type 1
PC	Pyruvate carboxylase deficiency
PCBD1	Hyperphenylalaninemia, BH4- deficient, type D
PCCA	Propionic acidemia
PCCB	Propionic acidemia
PCDH15	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency
PDHB	Pyruvate dehydrogenase E1-beta deficiency
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
PEX12	Peroxisome biogenesis disorder type 3A (Zellweger)
PEX13	Peroxisome biogenesis disorder, type 11A (Zellweger syndrome); Peroxisome biogenesis disorder, type 11B
PEX2	Peroxisome biogenesis disorder type 5A (Zellweger)

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PEX26	Peroxisome biogenesis disorder type 7A (Zellweger)
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
PHGDH	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency
PJVK	Hearing loss, autosomal recessive
PKHD1	Polycystic kidney disease, autosomal recessive
PLA2G6	Infantile neuroaxonal dystrophy type 1
PLEKHG5	Charcot-Marie-Tooth disease, recessive intermediate, type C
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1
PLP1	Pelizaeus-Merzbacher disease
PMM2	Congenital disorder of glycosylation, type 1A
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency
POLG	POLG-related disorders
POMGNT1	Muscular dystrophy- dystroglycanopathy, type 3A (Walker- Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDr15])
POMT1	Muscular dystrophy- dystroglycanopathy, type 1A (Walker- Warburg syndrome); Type 1B; Type 1C (limb-girdle 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])
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
POU1F1	Pituitary hormone deficiency, combined, type 1
PPT1	Ceroid lipofuscinosis, neuronal, type 1
PRF1	Hemophagocytic lymphohistiocytosis, familial, type 2
PROP1	Pituitary hormone deficiency, combined, type 2
PRPS1	PrPS1-related disoders
PSAP	Combined SAP deficiency

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GENI	PATOLOGIE
PTS	Hyperphenylalaninemia, BH4- deficient, type A
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia, type 1
PYGM	McArdle disease
QDPR	Hyperphenylalaninemia, BH4- deficient, type C
RAB23	Carpenter syndrome
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
RARS2	Pontocerebellar hypoplasia, type 6
RDH12	Leber congenital amaurosis, type 13
RMRP	Anauxetic dysplasia, type 1
RNASEH2A	Aicardi-Goutieres syndrome, type 4
RNASEH2B	Aicardi-Goutieres syndrome, type 2
RNASEH2C	Aicardi-Goutieres syndrome, type 3
RPE65	rPE65-related Leber congenital amaurosis/early- onset severe retinal dystrophy
RPGRIP1L	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome
RS1	Retinoschisis
RTEL1	Dyskeratosis congenita, autosomal recessive type 5
RYR1	Minicore myopathy with external ophthalmoplegia
SACS	Spastic ataxia, Charlevoix-Saguenay, type
SAMD9	Tumoral calcinosis, familial, normophosphatemic
SAMHD1	Aicardi-Goutieres syndrome, type 5
SBDS	Shwachman-Diamond syndrome
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1
SEPSECS	Pontocerebellar hypoplasia, type 2D

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GENI	PATOLOGIE
SERPINA1	Alpha-1 antitrypsin deficiency
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)
SGSH	Mucopolysaccharidosis, type 3A (Sanfilippo A)
SLC12A1	Bartter syndrome, type 1
SLC12A3	Gitelman syndrome
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy
SLC17A5	Salla disease
SLC22A5	Carnitine deficiency, systemic primary
SLC25A13	Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset
SLC25A15	Hyperornithinemia- hyperammonemia-homocitrullinemia syndrome
SLC25A20	Carnitine-acylcarnitine translocase deficiency
SLC26A2	Achondrogenesis, type 1B (diastrophic dysplasia)
SLC26A4	Deafness, autosomal recessive, type 4; Pendred syndrome
SLC35A3	Arthrogryposis, mental retardation and seizures
SLC37A4	Glycogen storage disease, type 1B
SLC39A4	Acrodermatitis enteropathica
SLC4A11	Corneal endothelial dystrophy, autosomal recessive
SLC5A5	Thyroid dyshormonogenesis, type 1
SLC6A8	Cerebral creatine deficiency syndrome, type 1
SMN1	Spinal muscular atrophy
SMPD1	Niemann-Pick disease, type A; Niemann-Pick disease, type B
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome

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GENI	PATOLOGIE
SRD5A2	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)
ST3GAL5	Salt and pepper developmental regression syndrome
STAR	Lipoid adrenal hyperplasia
STX11	Hemophagocytic lymphohistiocytosis, familial, type 4
STXBP2	Hemophagocytic lymphohistiocytosis, familial, type 5
SUMF1	Multiple sulfatase deficiency
SUOX	Sulfite oxidase deficiency
SURF1	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency
TAT	Tyrosinemia, type 2
TAZ	3-methylglutaconic aciduria (3MGA) Type I
TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy; Hypoparathyroidism-retardation- dysmorphism syndrome; Kenny- Caffey syndrome, type 1
TCIRG1	Osteopetrosis, autosomal recessive, type 1
TCN2	Transcobalamin II deficiency
TECPR2	Spastic paraplegia, type 49, autosomal recessive
TFR2	Hemochromatosis, type 3
TGM1	Ichthyosis, congenital, autosomal recessive, type 1
TH	Segawa syndrome, recessive
TK2	Mitochondrial DNA depletion syndrome, type 2 (myopathic type)
TMEM216	Joubert syndrome, type 2; Meckel syndrome, type 2
TMEM67	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome
TPP1	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7
TREX1	Aicardi-Goutieres syndrome, type 1
TRIM37	Mulibrey nanism
TSEN2	Pontocerebellar hypoplasia, type 2B
TSEN54	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4

GENI	PATOLOGIE
TSFM	Combined oxidative phosphorylation deficiency, type 3
TSHB	Hypothyroidism, congenital, nongoitrous, type 4
TSHR	Hypothyroidism, congenital, nongoitrous, type 1
TTPA	Ataxia with isolated vitamin E deficiency
TYMP	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)
TYR	Oculocutaneous albinism (OCA) type 1A; OCA type 1B
UBR1	Johanson-Blizzard syndrome
UGT1A1	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2
UNC13D	Hemophagocytic lymphohistiocytosis, familial, type 3
USH1C	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A
USH2A	Usher syndrome, type 2A
VDR	Rickets, vitamin D-resistant, type 2A
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion, type 1
VPS13A	Choreoacanthocytosis
VPS13B	Cohen syndrome
VRK1	Pontocerebellar hypoplasia, type 1A
WAS	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked
WNT10A	Odontoonychodermal dysplasia
XPA	Xeroderma pigmentosum, group A
XPC	Xeroderma pigmentosum, group C
ZFYVE26	Spastic paraplegia, type 15, autosomal recessive

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Carrier Screening – Test del portatore Extended

GENI	PATOLOGIE
AAAS	Triple-A syndrome (achalasia- addisonianism-alacrimia)
AARS2	Combined oxidative phosphorylation deficiency 8; Leukoencephalopathy, progressive, with ovarian failure
ABAT	GABA-transaminase deficiency
ABCA12	Ichthyosis, congenital, autosomal recessive, type 4A; ICAr, type 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
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
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
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
ACOX1	Peroxisomal acyl-CoA oxidase deficiency
ACP5	Spondyloenchondrodysplasia with immune dysregulation

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GENI	PATOLOGIE
ACSF3	Combined malonic and methylmalonic aciduria
ACSL4	Non-syndromic X-linked intellectual disability
ACTA1	Nemaline myopathy 3; Congenital fiber-type disproportion myopathy 1
ACY1	Aminoacylase 1 deficiency
ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency
ADA2	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome
ADAMTS13	Thrombotic thrombocytopenic purpura, familial (Schulman-Upshaw syndrome)
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
ADGRG1	Polymicrogyria, bilateral frontoparietal
ADGRV1	Usher syndrome, type 2C
ADK	Hypermethioninemia due to adenosine kinase deficiency
ADSL	Adenylosuccinase deficiency
AFF2	FRAXE intellectual disability
AFG3L2	Spastic ataxia, type 5, autosomal recessive
AGA	Aspartylglucosaminuria (glycosylasparaginase deficiency)
AGK	Cataract 38; Sengers syndrome
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

GENI	PATOLOGIE
AIMP1	Leukodystrophy, hypomyelinating, type 3
AIPL1	Leber congenital amaurosis, type 4
AIRE	Autoimmune polyendocrinopathy syndrome, type 1
AK2	Reticular dysgenesis
AKR1D1	Bile acid synthesis defect, congenital, type 2
ALAD	Porphyria, acute hepatic
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
ALG13	Developmental and epileptic encephalopathy, 36
ALG2	Myasthenic syndrome, congenital, type 14, with tubular aggregates
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	Alstrom syndrome
ALOX12B	Ichthyosis, congenital, autosomal recessive, type 2
ALOXE3	Ichthyosis, congenital, autosomal recessive, type 3
ALPL	Hypophosphatasia, infantile/ childhood

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GENI	PATOLOGIE
ALS2	Amyotrophic lateral sclerosis, type 2, juvenile; Primary lateral sclerosis, juvenile; Spastic paralysis, infantile onset ascending
ALX3	Frontonasal dysplasia, type 1
ALX4	Frontonasal dysplasia, type 2
AMACR	Bile acid synthesis defect, congenital, type 4; Alpha-methylacyl- CoA racemase deficiency
AMN	Megaloblastic anemia 1 (Imerslund- Grasbeck syndrome)
AMPD1	Myopathy due to myoadenylate deaminase deficiency
AMPD2	Pontocerebellar hypoplasia, type 9
AMT	Glycine encephalopathy
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	Mental retardation, X-linked, syndromic, type 5 (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
AP4M1	Spastic paraplegia, type 50, autosomal recessive
AP4S1	Spastic paraplegia, type 52, autosomal recessive
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

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GENI	PATOLOGIE
ARHGEF6	Non-syndromic X-linked intellectual disability
ARHGEF9	Developmental and epileptic encephalopathy, 8
ARL13B	Joubert syndrome type 8
ARL6	Bardet-Biedl syndrome, type 3
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)
ARSL	Chondrodysplasia punctata, brachytelephalangic (ARSL o ARSE)
ARV1	Epileptic encephalopathy, early
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
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
ATP6V0A2	Cutis laxa, autosomal recessive, type 2A; Wrinkly skin syndrome
ATP6V0A4	Renal tubular acidosis, distal, autosomal recessive
ATP6V1B1	Renal tubular acidosis with deafness
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	Mental retardation-hypotonic facies syndrome, X-linked; Alpha- thalassemia/mental retardation syndrome

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GENI	PATOLOGIE
AUH	3-methylglutaconic aciduria, type 1
AVPR2	Nephrogenic syndrome of inappropriate antidiuresis
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
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
BCHE	Butyrylcholinesterase deficiency
BCKDHA	Maple syrup urine disease, type 1A
BCKDHB	Maple syrup urine disease, type 1B
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency
BCOR	Microphthalmia, syndromic 2
BCS1L	BCS1L-related disorders, including Leigh syndrome
BHLHA9	Syndactyly, mesoaxial synostotic, with phalangeal reduction
BIN1	Centronuclear myopathy, type 2

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GENI	PATOLOGIE
BLM	Bloom syndrome
BLNK	Agammaglobulinemia 4
BLOC1S3	Hermansky-Pudlak syndrome, type 8
BLOC1S6	Hermansky-Pudlak syndrome, type 9
BMP1	Osteogenesis imperfecta, type 13
BMPER	Diaphanospondylodysostosis
BMPR1B	Acromesomelic dysplasia, Demirhan type
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal; Neurodevelopmental disorder with cerebellar atrophy and with or without seizures
BRF1	Cerebellofaciodental syndrome
BRIP1	Fanconi anemia, complementation group J
BRWD3	Mental retardation, X-linked, type 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
C12ORF57	Temtamy syndrome
C19ORF12	Neurodegeneration with brain iron accumulation, type 4
C2CD3	Orofaciodigital syndrome, type 14
CA2	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3
CABP2	Deafness, autosomal recessive, type 93
CACNA1D	Sinoatrial node dysfunction and deafness
CANT1	Desbuquois dysplasia, type 1; Epiphyseal dysplasia, multiple, type 7
CAPN3	Limb-girdle muscular dystrophy, type 1 (LGMD r1)
CARD11	Immunodeficiency, type 11A

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GENI	PATOLOGIE
CARS2	Combined oxidative phosphorylation deficiency 27
CASK	Syndromic X-linked intellectual disability Najm type
CASP10	Autoimmune lymphoproliferative syndrome
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, type 2
CASR	Hyperparathyroidism, neonatal
CAVIN1	Lipodystrophy, congenital generalized, type 4
CBS	Homocystinuria due to cystathionine beta-synthase
CC2D1A	Mental retardation, autosomal recessive, type 3
CC2D2A	Joubert syndrome, type 9; Meckel syndrome, type 6
CCBE1	Hennekam lymphangiectasia- lymphedema syndrome, type 1
CCDC103	Ciliary dyskinesia, primary, type 17
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
CCNO	Ciliary dyskinesia, primary, type 29
CD19	Immunodeficiency, common variable, type 3
CD247	Immunodeficiency, type 25
CD27	Lymphoproliferative syndrome 2
CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect
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

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GENI	PATOLOGIE
CD79A	Agammaglobulinemia 3
CD79B	Agammaglobulinemia 6
CDH23	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy
CDK5RAP2	Primary microcephaly type 3, autosomal recessive
CDKL5	Developmental and epileptic encephalopathy, 2
CDSN	Peeling skin syndrome 1
CDT1	Meier-Gorlin syndrome, type 4A
CENPJ	Primary microcephaly type 6, autosomal recessive
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
CEP290	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10
CEP41	Joubert syndrome, type 15
CEP57	Mosaic variegated aneuploidy syndrome 2
CEP83	Nephronophthisis 18
CERKL	Retinitis pigmentosa, type 26
CERS3	Ichthyosis, congenital, autosomal recessive 9
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

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GENI	PATOLOGIE
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
CIITA	Bare lymphocyte syndrome, type 2, complementation group A
CLCN1	Myotonia congenita, recessive
CLCN5	Dent disease type 1
CLCN7	Osteopetrosis, autosomal recessive type 4
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis
CLDN19	Renal hypomagnesemia type 5, with ocular involvement
CLN3	Ceroid lipofuscinosis, neuronal, type 3
CLN5	Ceroid lipofuscinosis, neuronal, type 5
CLN6	Ceroid lipofuscinosis, neuronal, type 6
CLN8	Ceroid lipofuscinosis, neuronal, type 8
CLRN1	Usher syndrome, type 3A
CNGA3	Achromatopsia, type 2
CNGB3	Achromatopsia, type 3
COG1	Congenital disorder of glycosylation, type IIg
COG7	Congenital disorder of glycosylation, type 2E
COG8	Congenital disorder of glycosylation, type 2H
COL11A2	Otospondylomegaepiphyseal dysplasia, autosomal recessive
COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type
COL1A2	Ehlers-Danlos syndrome, cardiac valvular type
COL27A1	Steel syndrome
COL3A1	Ehlers-Danlos syndrome, vascular type
COL4A3	Alport syndrome, autosomal recessive, type 2
COL4A4	Alport syndrome, autosomal recessive, type 2
COL4A5	Alport syndrome, X-linked

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GENI	PATOLOGIE
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
COLQ	Myasthenic syndrome, congenital, type 5
COQ2	Primary coenzyme Q10 deficiency, type 1
COQ8A	Primary coenzyme Q10 deficiency, type 4
COQ9	Coenzyme Q10 deficiency, primary, type 5
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
COX6B1	Mitochondrial complex IV deficiency, nuclear type 7
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
CRB1	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8
CRLF1	Cold-induced sweating syndrome type 1
CRTAP	Osteogenesis imperfecta, type 7
CSTB	Epilepsy, progressive myoclonic type 1A (Unverricht and Lundborg)
CTNS	Nephropathic cystinosis
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
CUL4B	Mental retardation, X-linked, syndromic, type 15 (Cabezas type)
CYBA	Chronic granulomatous disease, type 4
CYBB	Chronic granulomatous disease, X- linked

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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
CYP27A1	Cerebrotendinous xanthomatosis
CYP27B1	Vitamin D-dependent rickets, type 1
CYP7B1	Spastic paraplegia, type 5A, autosomal recessive
DBT	Maple syrup urine disease, type 2
DCAF17	Woodhouse-Sakati syndrome
DCLRE1C	Omenn syndrome; Severe combined immunodeficiency, Athabascan type
DCX	Lissencephaly, X-linked, type 1
DDB2	Xeroderma pigmentosum, complementation group E
DDC	Aromatic L-amino acid decarboxylase deficiency
DDX11	Warsaw breakage syndrome
DGAT1	Diarrhea 7, protein-losing enteropathy type
DGUOK	DGUOK-related mitochondrial DNA depletion syndrome
DHCR24	Desmosterolosis
DHCR7	Smith-Lemli-Opitz syndrome
DHDDS	Retinitis pigmentosa, type 59
DKC1	Dyskeratosis congenita, X-linked
DLD	Dihydrolipoamide dehydrogenase deficiency
DLG3	Mental retardation, X-linked, type 90
DLL3	Spondylocostal dysostosis type 1
DMD	Duchenne/Becker muscular dystrophy
DMP1	Hypophosphatemic rickets, autosomal recessive

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GENI	PATOLOGIE
DNAH11	Ciliary dyskinesia, primary, type 7, with or without situs inversus
DNAH5	Ciliary dyskinesia, primary, type 3, with or without situs inversus
DNAI1	Ciliary dyskinesia, primary, type 1, with or without situs inversus
DNAI2	Ciliary dyskinesia, primary, type 9, with or without situs inversus
DNAJC19	3-methylglutaconic aciduria, type 5
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 1
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
DPAGT1	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13
DPM1	Congenital disorder of glycosylation, type 1E
DPYD	Dihydropyrimidine dehydrogenase deficiency
DSP	Cardiomyopathy, dilated, with woolly hair and keratoderma; Epidermolysis bullosa, lethal acantholytic
DUOX2	Thyroid dyshormonogenesis, type 6
DYNC2H1	Short-rib thoracic dysplasia, type 3, with or without polydactyly
DYSF	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD r2)
EDA	Ectodermal dysplasia, type 1, hypohidrotic, X-linked
EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type
EDN3	Waardenburg syndrome, type 4B
EDNRB	ABCD syndrome
EFEMP2	Cutis laxa, autosomal recessive, type 1B
EFNB1	craniofrontonasal syndrome
EGR2	Dejerine-Sottas disease
EIF2AK3	Wolcott-rallison syndrome
EIF2B1	Leukoencephalopathy with vanishing white matter
EIF2B2	Leukoencephalopathy with vanishing white matter
EIF2B3	Leukoencephalopathy with vanishing white matter
EIF2B4	Leukoencephalopathy with vanishing white matter

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GENI	PATOLOGIE
EIF2B5	Leukoencephalopathy with vanishing white matter
ELP1	Familial dysautonomia
EMD	Emery-Dreifuss muscular dystrophy, type 1, X-linked
ENPP1	Arterial calcification, generalized, of infancy, type 1
EPG5	Vici syndrome
EPM2A	Epilepsy, progressive myoclonic, type 2A (Lafora)
ERBB3	Lethal congenital contractural syndrome, type 2
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
ERCC8	Cockayne syndrome, type A
ESCO2	Roberts syndrome
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
EYS	Retinitis pigmentosa, type 25
F11	Factor XI deficiency
F2	Prothrombin deficiency
F5	Factor V deficiency
F8	Hemophilia A
F9	Hemophilia B
FAH	Tyrosinemia, type 1

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GENI	PATOLOGIE
FAM126A	Leukodystrophy, hypomyelinating, type 5
FAM161A	Retinitis pigmentosa, type 28
FAM20C	Raine syndrome
FANCA	Fanconi anemia, complementation group A
FANCB	VACTERL association, X-linked, with or without hydrocephalus
FANCC	Fanconi anemia, complementation group C
FANCD2	Fanconi anemia, complementation group D2
FANCE	Fanconi anemia, complementation group E
FANCG	Fanconi anemia, complementation group G
FANCI	Fanconi anemia, complementation group I
FANCL	Fanconi anemia, complementation group L
FASTKD2	Combined oxidative phosphorylation deficiency 44
FBLN5	Cutis laxa, autosomal recessive, type 1A
FBP1	Fructose-1,6-bisphosphatase deficiency
FBXO7	Parkinson disease, type 15, autosomal recessive
FERMT3	Leukocyte adhesion deficiency, type 3
FGA	Afibrinogenemia, congenital
FGD1	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic, type 16
FGD4	Charcot-Marie-Tooth disease, type 4H
FH	Fumarase deficiency
FHL1	X-linked myopathy with postural muscle atrophy
FKBP10	Bruck syndrome 1
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])
FMO3	Trimethylaminuria
FMR1	Fragile X syndrome
FOLR1	Neurodegeneration due to cerebral folate transport deficiency
FOXN1	T-cell immunodeficiency, congenital alopecia and nail dystrophy

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FOXP3	Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome
FOXRED1	Mitochondrial complex I deficiency, nuclear type 19
FRAS1	Fraser syndrome, type 1
FREM2	Fraser syndrome, type 2
FTSJ1	Mental retardation, X-linked 44
FUCA1	Fucosidosis
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
GATM	Cerebral creatine deficiency syndrome, type 3
GBA	Gaucher disease
GBE1	Glycogen storage disease, type 4
GCDH	Glutaricaciduria, type 1
GCH1	Hyperphenylalaninemia, BH4- deficient, type B
GCSH	Glycine encephalopathy
GDAP1	Charcot-Marie-Tooth disease, recessive intermediate, type A
GDF5	Chondrodysplasia, Grebe type
GDI1	Intellectual disability, X-linked 41
GFM1	Combined oxidative phosphorylation deficiency, type 1
GFPT1	Myasthenia, congenital, type 12, with tubular aggregates

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GENI	PATOLOGIE
GHR	Laron dwarfism
GJB1	Charcot-Marie-Tooth neuropathy, X- linked dominant, type 1
GJB2	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6
GJB3	Hearing loss
GJB6	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6
GJC2	Spastic paraplegia, type 44, autosomal recessive
GLA	Fabry disease
GLB1	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)
GLDC	Glycine encephalopathy
GLE1	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease
GNE	Inclusion body myopathy, type 2 (Nonaka myopathy)
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
GP1BA	Bernard-Soulier syndrome, type A1
GP9	Bernard-Soulier syndrome, type C
GPC3	Simpson-Golabi-Behmel syndrome type 1
GRHPR	Hyperoxaluria, primary, type 2
GRIK2	Mental retardation, autosomal recessive, type, 6
GRIP1	Fraser syndrome 3
GRN	Ceroid lipofuscinosis, neuronal, type 11
GSS	Glutathione synthetase deficiency
GTF2H5	Trichothiodystrophy, type 3, photosensitive
GUCY2D	Leber congenital amaurosis, type 1
GUSB	Mucopolysaccharidosis, type 7

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GENI	PATOLOGIE
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
HAX1	Neutropenia, severe congenital, type 3, autosomal recessive
HBB	HBB-related hemoglobinopathy
HCFC1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A
HESX1	Growth hormone deficiency with pituitary anomalies
HEXA	Tay-Sachs disease
HEXB	Sandhoff disease, infantile, juvenile, and adult forms
HFE	hemochromatosis type 1
HFE2	Hereditary hemochromatosis type 2A, HFE2-related
HGD	Alkaptonuria
HGSNAT	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency
HLCS	Holocarboxylase synthetase deficiency
HMGCL	HMG-CoA lyase deficiency
HMOX1	Heme oxygenase-1 deficiency
HOGA1	Hyperoxaluria, primary, type 3
HPD	Tyrosinemia, type 3
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
HSD11B2	Apparent mineralocorticoid excess

GENI	PATOLOGIE
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
HSPG2	Dyssegmental dysplasia, Silverman- Handmaker type
HUWE1	intellectual disability, X-linked syndromic, Turner type
HYAL1	Mucopolysaccharidosis, type 9
HYLS1	Hydrolethalus syndrome
ICOS	Immunodeficiency, common variable, 1
IDS	Mucopolysaccharidosis, type 2
IDUA	Mucopolysaccharidosis type 1
IFNGR1	Immunodeficiency, type 27A, mycobacteriosis
IFNGR2	Immunodeficiency, type 28, mycobacteriosis
IFT80	Short-rib thoracic dysplasia, type 2, with or without polydactyly
IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S
IKBKAP	Familial Dysautonomia
IKBKB	Immunodeficiency, type 15
IKBKG	immunodeficiency without anhidrotic ectodermal dysplasia
IL12B	Immunodeficiency, type 29, mycobacteriosis
IL12RB1	Immunodeficiency, type 30
IL1RAPL1	Mental retardation, X-linked, type 21/34
IL1RN	Sterile multifocal osteomyelitis with periostitis and pustulosis
IL2RG	Severe combined immunodeficiency, X-linked
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type
INSR	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, type A
INVS	Nephronophthisis, type 2, infantile
IQCB1	Senior-Loken syndrome, type 5
ISPD	Walker-Warburg (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7

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GENI	PATOLOGIE
ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis
ITGB3	Glanzmann thrombasthenia
ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia
IVD	Isovaleric acidemia
JAK3	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B- positive type
KCNJ1	Bartter syndrome, type 2
KCNJ11	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)
KCTD7	Epilepsy, progressive myoclonic, type 3, with or without intracellular inclusions
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type
L1CAM	L1 Syndrome
LAMA2	LAMA2-related muscular dystrophy
LAMA3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
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
LARGE1	Muscular dystrophy- dystroglycanopathy, type 6A and 6B
LBR	Greenberg skeletal dysplasia
LCA5	Leber congenital amaurosis, type 5
LDLR	Hypercholesterolemia, familial, 1
LDLRAP1	Hypercholesterolemia, familial, autosomal recessive
LHCGR	Leydig cell hypoplasia
LHX3	Pituitary hormone deficiency, combined, type 3
LIFR	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome
LIG4	LIG4 syndrome
LIPA	Lysosomal acid lipase deficiency
LIPH	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type
LMNA	LMNA-related disorders, autosomal recessive

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GENI	PATOLOGIE
LOXHD1	Deafness, autosomal recessive, type 77
LPL	Lipoprotein lipase deficiency
LRAT	Leber congenital amaurosis type 14
LRP2	Donnai-Barrow syndrome
LRPPRC	Leigh syndrome, French-Canadian type
LYST	Chediak-Higashi syndrome
MAK	retinitis pigmentosa type 62
MAN2B1	Alpha-mannosidosis
MANBA	Mannosidosis, beta
MAT1A	Methionine adenosyltransferase deficiency, autosomal recessive
MBTPS2	osteogenesis imperfecta
MCCC1	3-Methylcrotonyl-CoA carboxylase deficiency, type 1
MCCC2	3-Methylcrotonyl-CoA carboxylase deficiency, type 2
MCEE	Methylmalonyl-CoA epimerase deficiency
MCOLN1	Mucolipidosis type 4
MCPH1	Microcephaly type 1, primary, autosomal recessive
MECP2	Encephalopathy, neonatal severe; rett syndrome
MED12	FG syndrome 1
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy
MEFV	Familial Mediterranean fever
MESP2	Spondylocostal dysostosis, type 2, autosomal recessive
MFSD8	Ceroid lipofuscinosis, neuronal, type 7
MGAT2	Congenital disorder of glycosylation, type 2a
MID1	X-linked Opitz G/BBB 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
MLYCD	Malonyl-CoA decarboxylase deficiency

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GENI	PATOLOGIE
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
MOCS1	Molybdenum cofactor deficiency A
MOCS2	Molybdenum cofactor deficiency B
MOGS	Congenital disorder of glycosylation, type 2B
MPDU1	Congenital disorder of glycosylation, type 1F
MPI	Congenital disorder of glycosylation, type 1B
MPL	Thrombocytopenia, congenital amegakaryocytic
MPV17	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE
MPZ	Dejerine-Sottas disease
MRE11A	Ataxia-telangiectasia-like disorder 1
MRPS16	Combined oxidative phosphorylation deficiency 2
MRPS22	Combined oxidative phosphorylation deficiency type 5
MTHFR	Homocystinuria due to MTHFr deficiency
MTM1	Myotubular myopathy, X-linked
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
MUT	Methylmalonic aciduria mut(0) type, MUT-related
MVK	Mevalonic aciduria
MYD88	Immunodeficiency, type 68
MYO15A	Deafness, autosomal recessive, type 3
MYO5A	Griscelli syndrome, type 1
MYO7A	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2
NAGA	Schindler disease, type I

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GENI	PATOLOGIE
NAGLU	Mucopolysaccharidosis, type 3B (Sanfilippo B)
NAGS	N-acetylglutamate synthase deficiency
NBN	Nijmegen breakage syndrome
NCF2	Chronic granulomatous disease, type 2
NDP	Norrie disease
NDRG1	Charcot-Marie-Tooth disease, type 4D
NDUFA1	Mitochondrial complex I deficiency, nuclear type
NDUFAF2	Mitochondrial complex I deficiency, nuclear type 10
NDUFAF4	Mitochondrial complex 1 deficiency, nuclear type 15
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16
NDUFAF6	Mitochondrial complex I deficiency, nuclear type 17
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
NEB	Nemaline myopathy type 2
NEU1	Sialidosis, type 1 and type 2
NEUROG3	Diarrhea 4, malabsorptive, congenital
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)
NHS	Nance-Horan syndrome
NPC1	Niemann-Pick disease, type C1
NPC2	Niemann-pick disease, type C2
NPHP1	Joubert syndrome type 4
NPHP3	Meckel syndrome type 7

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NPHP4	Nephronophthisis type 4
NPHS1	Nephrotic syndrome, type 1
NPHS2	Nephrotic syndrome, type 2
NR0B1	Adrenal hypoplasia, congenital
NR2E3	Enhanced S-cone syndrome (Goldmann-Favre); retinitis pigmentosa, type 37
NSMCE3	Lung disease, immunodeficiency, and chromosome breakage syndrome
NSUN2	Mental retardation, autosomal recessive, type 5
NTRK1	Insensitivity to pain, congenital, with anhidrosis
NUP62	Striatonigral degeneration, infantile
OAT	Gyrate atrophy of choroid and retina
OCA2	Oculocutaneous albinism type 2
OCRL	Lowe Syndrome; Dent disease type 2
OFD1	Oorofaciodigital syndrome I
OPA3	3-methylglutaconic aciduria, type 3
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance
ORAI1	Immunodeficiency, type 9
OSTM1	Osteopetrosis, autosomal recessive type 5
OTC	Ornithine transcarbamylase deficiency
OTOA	Deafness, autosomal recessive, type 22
OTOF	Deafness, autosomal recessive, type 9
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency
P3H1	Osteogenesis imperfecta, type 8
PAH	Phenylketonuria
PAK3	Mental retardation, X-linked, type 30
PANK2	Neurodegeneration with brain iron accumulation type 1
PC	Pyruvate carboxylase deficiency
PCBD1	Hyperphenylalaninemia, BH4- deficient, type D
PCCA	Propionic acidemia

GENI	PATOLOGIE
PCCB	Propionic acidemia
PCDH15	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic
PCDH19	Developmental and epileptic encephalopathy, 9
PCNT	Microcephalic osteodysplastic primordial dwarfism, type 2
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
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
PEX12	Peroxisome biogenesis disorder type 3A (Zellweger)
PEX13	Peroxisome biogenesis disorder, type 11A (Zellweger syndrome); Peroxisome biogenesis disorder, type 11B
PEX16	Peroxisome biogenesis disorder, type 8A (Zellweger syndrome); Peroxisome biogenesis disorder, type 8B
PEX2	Peroxisome biogenesis disorder type 5A (Zellweger)
PEX26	Peroxisome biogenesis disorder type 7A (Zellweger)
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
PGM3	Immunodeficiency, type 23
PHGDH	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency
PHKB	Glycogen storage disease, type 9B
PHKG2	Glycogen storage disease type 9c
PHYH	Refsum disease

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GENI	PATOLOGIE
PIGN	Multiple congenital anomalies- hypotonia-seizures syndrome, type 1
PJVK	Hearing loss, autosomal recessive
PKHD1	Polycystic kidney disease, autosomal recessive
PKLR	Pyruvate kinase deficiency
PLA2G6	Infantile neuroaxonal dystrophy type 1
PLCE1	Nephrotic syndrome, type 3
PLEC	Epidermolysis bullosa simplex with muscular dystrophy
PLEKHG5	Charcot-Marie-Tooth disease, recessive intermediate, type C
PLG	Plasminogen deficiency, type I
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1
PLP1	Pelizaeus-Merzbacher disease
PMM2	Congenital disorder of glycosylation, type 1A
PMP22	Dejerine-Sottas disease
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency
POLG	POLG-related disorders
POLH	Xeroderma pigmentosum, variant type
POMGNT1	Muscular dystrophy- dystroglycanopathy, type 3A (Walker- Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDr15])
POMT1	Muscular dystrophy- dystroglycanopathy, type 1A (Walker- Warburg syndrome); Type 1B; Type 1C (limb-girdle 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])
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
POU1F1	Pituitary hormone deficiency, combined, type 1
PPT1	Ceroid lipofuscinosis, neuronal, type 1
PQBP1	Renpenning syndrome
PRCD	Retinitis pigmentosa, type 36
PRDM5	Brittle cornea syndrome, type 2
PRF1	Hemophagocytic lymphohistiocytosis, familial, type 2
PROP1	Pituitary hormone deficiency, combined, type 2
PRPS1	PrPS1-related disoders

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PRSS12	Mental retardation, autosomal recessive, type 1
PRX	Charcot-Marie-Tooth disease, type 4F
PSAP	Combined SAP deficiency
PTH1R	Chondrodysplasia, Blomstrand type; Eiken syndrome
PTPRC	Severe combined immunodeficiency, Autosomal T cell-negative, B-cell/natural killer-recessive cell positive
PTS	Hyperphenylalaninemia, BH4- deficient, type A
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia, type 1
PYGL	Glycogen storage disease, type 6
PYGM	McArdle disease
QDPR	Hyperphenylalaninemia, BH4- deficient, type C
RAB23	Carpenter syndrome
RAB27A	Griscelli syndrome, type 2
RAB39B	Intellectual disability, X-linked 72
RAB3GAP1	Warburg micro syndrome, type 1
RAB3GAP2	Martsolf syndrome
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
RARS2	Pontocerebellar hypoplasia, type 6
RDH12	Leber congenital amaurosis, type 13
RELN	Lissencephaly 2 (Norman-roberts type)
RFT1	Congenital disorder of glycosylation, type In
RLBP1	Bothnia retinal dystrophy; Fundus albipunctatus
RMRP	Anauxetic dysplasia, type 1
RNASEH2A	Aicardi-Goutieres syndrome, type 4
RNASEH2B	Aicardi-Goutieres syndrome, type 2
RNASEH2C	Aicardi-Goutieres syndrome, type 3
RP2	Retinitis pigmentosa, type 2, X-linked

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GENI	PATOLOGIE
RPE65	rPE65-related Leberÿcongenitalÿamaurosis/early- onset severe retinal dystrophyÿ
RPGRIP1L	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome
RPL10	Intellectual disability, 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
RTEL1	Dyskeratosis congenita, autosomal recessive type 5
RYR1	Minicore myopathy with external ophthalmoplegia
SACS	Spastic ataxia, Charlevoix-Saguenay, type
SAMD9	Tumoral calcinosis, familial, normophosphatemic
SAMHD1	Aicardi-Goutieres syndrome, type 5
SBDS	Shwachman-Diamond syndrome
SC5D	Lathosterolosis
SCNN1A	Pseudohypoaldosteronism, type 1
SCNN1B	Pseudohypoaldosteronism, type 1
SCNN1G	Pseudohypoaldosteronism, type 1
SCO1	Mitochondrial complex IV deficiency, nuclear type 4
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1
SEC23B	Dyserythropoietic anemia, congenital, type 2
SELENON	Congenital myopathy 3 with rigid spine
SEPSECS	Pontocerebellar hypoplasia, type 2D
SERPINA1	Alpha-1 antitrypsin deficiency
SFTPB	Surfactant metabolism dysfunction, pulmonary, type 1
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)
SGSH	Mucopolysaccharidosis, type 3A (Sanfilippo A)

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SH2D1A	Lymphoproliferative syndrome, X- linked, type 1
SHROOM4	X-linked intellectual disability, Stocco dos Santos type
SIL1	Marinesco-Sjogren syndrome
SKIV2L	Trichohepatoenteric syndrome, type 2 (diarrhea, syndromic)
SLC12A1	Bartter syndrome, type 1
SLC12A3	Gitelman syndrome
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy
SLC16A2	Allan-Herndon-Dudley syndrome
SLC17A5	Salla disease
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome
SLC19A3	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)
SLC22A5	Carnitine deficiency, systemic primary
SLC25A13	Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset
SLC25A15	Hyperornithinemia- hyperammonemia-homocitrullinemia syndrome
SLC25A20	Carnitine-acylcarnitine translocase deficiency
SLC25A22	Epileptic encephalopathy, early infantile, type 3
SLC26A2	Achondrogenesis, type 1B (diastrophic dysplasia)
SLC26A3	Diarrhea 1, secretory chloride, congenital
SLC26A4	Deafness, autosomal recessive, type 4; Pendred syndrome
SLC27A4	Ichthyosis prematurity syndrome
SLC35A1	Congenital disorder of glycosylation, type 2F
SLC35A3	Arthrogryposis, mental retardation 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
SLC39A4	Acrodermatitis enteropathica
SLC45A2	Albinism, oculocutaneous, type 4

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SLC4A11	Corneal endothelial dystrophy, autosomal recessive
SLC5A5	Thyroid dyshormonogenesis, type 1
SLC6A8	Cerebral creatine deficiency syndrome, type 1
SLC7A7	Lysinuric protein intolerance
SLC9A6	Christianson syndrome
SMARCAL1	Schimke immunoosseous dysplasia
SMN1	Spinal muscular atrophy
SMPD1	Niemann-Pick disease, type A; Niemann-Pick disease, type B
SMS	syndromic X-linked intellectual disability Snyder type
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome
SOX3	46,XX sex reversal 3
SP110	Hepatic venoocclusive disease with immunodeficiency
SPG11	Amyotrophic lateral sclerosis, type 5, juvenile
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency
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
ST3GAL3	Mental retardation, autosomal recessive 12
ST3GAL5	Salt and pepper developmental regression syndrome
STAR	Lipoid adrenal hyperplasia
STAT1	Immunodeficiency, type 31B, mycobacterial and viral infections
STIM1	Immunodeficiency, type 10
STRA6	Microphthalmia, isolated, with coloboma, type 8
STS	Recessive X-linked ichthyosis
STX11	Hemophagocytic lymphohistiocytosis, familial, type 4
STXBP2	Hemophagocytic lymphohistiocytosis, familial, type 5
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)
SUMF1	Multiple sulfatase deficiency

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GENI	PATOLOGIE
SUOX	Sulfite oxidase deficiency
SURF1	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency
SYNE4	Deafness, autosomal recessive, type 76
SYP	Intellectual disability, X-linked 96
TAT	Tyrosinemia, type 2
TAZ	3-methylglutaconic aciduria (3MGA) Type I
TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy; Hypoparathyroidism-retardation- dysmorphism syndrome; Kenny- Caffey syndrome, type 1
TCIRG1	Osteopetrosis, autosomal recessive, type 1
TCN2	Transcobalamin II deficiency
TECPR2	Spastic paraplegia, type 49, autosomal recessive
TERT	Dyskeratosis congenita, autosomal recessive, type 4
TF	Atransferrinemia
TFR2	Hemochromatosis, type 3
TG	Thyroid dyshormonogenesis, type 3
TGM1	Ichthyosis, congenital, autosomal recessive, type 1
TH	Segawa syndrome, recessive
TIMM8A	Deafness dystonia syndrome
TK2	Mitochondrial DNA depletion syndrome , type 2 (myopathic type)
TMC1	Deafness, autosomal recessive, type 7
TMEM216	Joubert syndrome, type 2; Meckel syndrome, type 2
TMEM67	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome
TMPRSS3	Deafness, autosomal recessive, type 8/10
TNFRSF11B	Paget disease of bone, type 5, juvenile-onset
TPO	Thyroid dyshormonogenesis, type 2A
TPP1	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7
TRAPPC9	Mental retardation, autosomal recessive, type 13
TREX1	Aicardi-Goutieres syndrome, type 1
TRIM32	Limb-girdle muscular dystrophy, type 8 (LGMD r8)

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TRIM37	Mulibrey nanism
TRMU	Liver failure, transient infantile
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
TSPYL1	Sudden infant death-dysgenesis of the testes syndrome
TTC37	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)
TTC8	Bardet-Biedl syndrome, type 8
TTN	Limb-girdle muscular dystrophy type 10 (LGMDr10); Early-onset myopathy with fatal cardiomyopathy (Salih myopathy)
TTPA	Ataxia with isolated vitamin E deficiency
TUFM	Combined oxidative phosphorylation deficiency 4
TULP1	Leber congenital amaurosis, type 15
TUSC3	Mental retardation, autosomal recessive, type 7
TWNK	Mitochondrial DNA depletion syndrome, type 7 (hepatocerebral type); Perrault syndrome type 5
TYK2	Immunodeficiency, type 35
TYMP	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)
TYR	Oculocutaneous albinism (OCA) type 1A; OCA type 1B
TYRP1	Albinism, oculocutaneous, type 3
UBA1	Infantile-onset X-linked spinal muscular atrophy
UBE2A	Syndromic X-linked intellectual disability Nascimento type
UBE3A	Angelman syndrome
UBR1	Johanson-Blizzard syndrome
UGT1A1	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2
UNC13D	Hemophagocytic lymphohistiocytosis, familial, type 3
UPF3B	Mental retardation, X-linked,syndromic, type 14

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UQCRB	Mitochondrial complex III deficiency, nuclear, type 3
UQCRQ	Mitochondrial complex III deficiency, nuclear, type 4
UROS	Porphyria, congenital erythropoietic
USH1C	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A
USH1G	Usher syndrome, type 1G
USH2A	Usher syndrome, type 2A
VDR	Rickets, vitamin D-resistant, type 2A
VIPAS39	Arthrogryposis, renal dysfunction and cholestasis, type 2
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion, type 1
VPS13A	Choreoacanthocytosis
VPS13B	Cohen syndrome
VPS33B	Arthrogryposis, renal dysfunction and cholestasis, type 1
VPS45	Neutropenia, severe congenital, type 5
VPS53	Pontocerebellar hypoplasia, type 2E
VRK1	Pontocerebellar hypoplasia, type 1A
VSX2	Microphthalmia with coloboma 3; Isolated microphthalmia 2
WAS	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked
WNT10A	Odontoonychodermal dysplasia
WNT3	Tetra-amelia syndrome
WNT7A	Fuhrmann syndrome
WRN	Werner syndrome
XIAP	X-linked lymphoproliferative disease due to XIAP deficiency
XPA	Xeroderma pigmentosum, group A
XPC	Xeroderma pigmentosum, group C
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 2
ZDHHC9	Mental retardation, X-linked syndromic, raymond type
ZFYVE26	Spastic paraplegia, type 15, autosomal recessive
ZIC3	VACTERL association, X-linked, with or without hydrocephalus

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GENI	PATOLOGIE
ZMPSTE24	Mandibuloacral dysplasia with, type B lipodystrophy
ZNF469	Brittle cornea syndrome, type 1
ZNF711	Mental retardation, X-linked, type 97

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