Tabella 1. Lista dei geni investigati dal test GENEADVANCE Omnia e delle patologie genetiche a trasmissione ereditaria associate.

Gene	Malattia Genetica
AAAS	Achalasia-addisonianism-alacrima syndrome
ABCA12	Harlequin ichthyosis
ABCA4	Stargardt disease, type 1
ABCB11	Progressive familial intrahepatic cholestasis, type 2
ABCB4	Progressive familial intrahepatic cholestasis, type 3
ABCC6	Pseudoxanthoma elasticum
ABCC8	Familial hyperinsulinism, *ABCC8*-related
ABCD1	Adrenoleukodystrophy, X-linked
ACAD9	Mitochondrial complex I deficiency, *ACAD9*-related
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 deficiency
ACAT1	Beta-ketothiolase deficiency
ACOX1	Acyl-CoA oxidase I deficiency
ACSF3	Combined malonic and methylmalonic aciduria
ADA	Severe combined immunodeficiency, ADA-related
ADAMTS2	Ehlers-Danlos syndrome, type VIIC
ADGRG1	Bilateral frontoparietal polymicrogyria
AGA	Aspartylglucosaminuria
AGL	Glycogen storage disease, type III (Cori/Forbes)
AGPS	Rhizomelic chondrodysplasia punctata, type 3
AGXT	Hyperoxaluria, primary, type 1
AIRE	Autoimmune polyendocrinopathy syndrome, type I
ALDH3A2	Sjögren-Larsson syndrome
ALDH7A1	Pyridoxine-dependent epilepsy
ALDOB	Hereditary fructose intolerance
ALG6	Congenital disorder of glycosylation, type 1C
ALMS1	Alström syndrome
ALPL	Hypophosphatasia, *ALPL*-related
AMH	Persistent Müllerian duct syndrome, type 1
AMHR2	Persistent Müllerian duct syndrome, type 2

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Pec: info@pec.genomicalab.it
P.Iva e C.F: 14554101007
REA: RM-1530210

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Milano: Viale L. Bodio 29-37 (Bodio 3) - 20158 - Tel.: 02.21115330
E-mail: info@genomicalab.it
Website: www.genomicalab.it

AMT	Glycine encephalopathy, *AMT*-related
AP1S1	Mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis, and keratoderma (MEDNIK)
AQP2	Familial nephrogenic diabetes insipidus, *AQP2*-related
AR	Androgen insensitivity syndrome, X-linked
ARG1	Argininemia
ARSA	Metachromatic leukodystrophy, *ARSA*-related
ARSB	Mucopolysaccharidosis, type VI (Maroteaux-Lamy)
ASL	Argininosuccinate lyase deficiency
ASNS	Asparagine synthetase deficiency
ASPA	Canavan disease
ASS1	Citrullinemia, type 1
ATM	Ataxia-telangiectasia
ATP6V1B1	Renal tubular acidosis and deafness, *ATP6V1B1*-related
ATP7A	Menkes syndrome, X-linked
ATP7B	Wilson disease
ATP8B1	Progressive familial intrahepatic cholestasis, type 1
ATRX	Alpha-thalassemia intellectual disability syndrome, X-linked
BBS1	Bardet-Biedl syndrome 1
BBS10	Bardet-Biedl syndrome 10
BBS12	Bardet-Biedl syndrome 12
BBS2	Bardet-Biedl syndrome 2
BBS4	Bardet-Biedl syndrome 4
BBS9	Bardet-Biedl syndrome 9
BCHE	Pseudocholinesterase deficiency
BCKDHA	Maple syrup urine disease, type 1A
BCKDHB	Maple syrup urine disease, type 1B
BCS1L	GRACILE syndrome
BLM	Bloom syndrome
BRIP1	Fanconi anemia, group J
BSND	Bartter syndrome, type 4a
BTD	Biotinidase deficiency
BTK	Isolated growth hormone deficiency, type III, X-linked
CANT1	Desbuquois dysplasia 1
CAPN3	Limb-girdle muscular dystrophy, type 2A
CASQ2	Catecholaminergic polymorphic ventricular tachycardia
CBS	Homocystinuria, *CBS*-related
CC2D1A	Mental retardation, autosomal recessive 3
CDH23	Usher syndrome, type 1D
CEP290	Leber congenital amaurosis, type *CEP290*
CERKL	Retinitis pigmentosa 26
CFTR	Cystic fibrosis
CHM	Choroideremia, X-linked
CHRNE	Congenital myasthenic syndrome, *CHRNE*-related
CHRNG	Escobar syndrome
CIITA	Bare lymphocyte syndrome, *CIITA*-related
CLN3	Ceroid lipofuscinosis, neuronal, 3
CLN5	Ceroid lipofuscinosis, neuronal, *5*
CLN6	Ceroid lipofuscinosis, neuronal, 6

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CLN8	Ceroid lipofuscinosis, neuronal, 8 (a.k.a. Northern epilepsy)
CLRN1	Usher syndrome, type 3
CNGA3	Achromatopsia, *CNGA3*-related
CNGB3	Achromatopsia, *CNGB3*-related
COL11A2	Fibrochondrogenesis, type 2
COL4A3	Alport syndrome, *COL4A3*-related
COL4A4	Alport syndrome, *COL4A4*-related
COL4A5	Alport syndrome, X-linked
COL7A1	Dystrophic epidermolysis bullosa, *COL7A1*-related
CPS1	Carbamoyl phosphate synthetase I deficiency
CPT1A	Carnitine palmitoyltransferase IA deficiency
CPT2	Carnitine palmitoyltransferase II deficiency
CRB1	Leber congenital amaurosis 8
CTNS	Cystinosis
CTSC	Papillon-Lefevre syndrome
CTSD	Ceroid lipofuscinosis, neuronal, 10 (CLN10 disease)
CTSK	Pycnodysostosis
CYBA	Chronic granulomatous disease, *CYBA*-related
CYBB	Chronic granulomatous disease, X-linked
CYP11B1	Congenital adrenal hyperplasia, 11-beta-hydroxylase-deficient
CYP11B2	Corticosterone methyloxidase deficiency
CYP17A1	Congenital adrenal hyperplasia, 17-alpha-hydroxylase deficiency
CYP19A1	Aromatase deficiency
CYP1B1	Primary congenital glaucoma
CYP21A2	Congenital adrenal hyperplasia, 21-hydroxylase-deficient
CYP27A1	Cerebrotendinous xanthomatosis
CYP27B1	Vitamin D–dependent rickets, type 1A
DBT	Maple syrup urine disease, type 2
DCLRE1C	Severe combined immunodeficiency, type athabaskan
DDB2	Xeroderma pigmentosum group E
DHCR7	Smith-Lemli-Opitz syndrome
DHDDS	Retinitis pigmentosa 59
DKC1	Dyskeratosis congenita, X-linked
DLD	Dihydrolipoamide dehydrogenase deficiency
DMD	Duchenne/Becker muscular dystrophy
DNAH5	Ciliary dyskinesia, primary 3
DNAI1	Ciliary dyskinesia, primary 1
DNAI2	Ciliary dyskinesia, primary 9
DNAL1	Ciliary dyskinesia, primary, 16
DOK7	Congenital myasthenic syndrome, *DOK7*-related
DPYD	Dihydropyrimidine dehydrogenase deficiency
DYSF	Limb-girdle muscular dystrophy, type 2B
EDA	Hypohidrotic ectodermal dysplasia, X-linked
EDAR	Hypohidrotic ectodermal dysplasia
EIF2AK3	Wolcott-Rallison syndrome
EIF2B5	Leukoencephalopathy with vanishing white matter
EMD	Emery-Dreifuss muscular dystrophy 1, X-linked
ERCC2	Xeroderma pigmentosum, group D

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ERCC3	Xeroderma pigmentosum, group B
ERCC4	Xeroderma pigmentosum, group F
ERCC5	Xeroderma pigmentosum, group G
ERCC6	Cockayne syndrome, type B
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, *EVC*-related
EVC2	Ellis-van Creveld syndrome, *EVC2*-related
EXOSC3	Pontocerebellar hypoplasia, type 1B
EYS	Retinitis pigmentosa 25
F11	Factor XI deficiency
F2	Prothrombin deficiency
F8	Hemophilia A
F9	Hemophilia B
FAH	Tyrosinemia, type I
FAM161A	Retinitis pigmentosa 28
FANCA	Fanconi anemia, group A
FANCC	Fanconi anemia, group C
FANCG	Fanconi anemia, group G
FH	Fumarase deficiency
FKRP	Limb-girdle muscular dystrophy, type 2I
FKTN	Walker-Warburg syndrome, *FKTN*-related
G6PC	Glycogen storage disease, type IA
G6PD	Glucose-6-phosphate dehydrogenase deficiency
GAA	Glycogen storage disease, type II (Pompe disease)
GALC	Krabbe disease
GALE	Galactose epimerase deficiency
GALK1	Galactokinase deficiency (galactosemia, type II)
GALNS	Mucopolysaccharidosis, type IVA
GALNT3	Hyperphosphatemic familial tumoral calcinosis
GALT	Galactosemia
GAMT	Guanidinoacetate methyltransferase deficiency
GBA	Gaucher disease
GBE1	Glycogen storage disease, type IV
GCDH	Glutaric acidemia, type 1
GCH1	Dopa-responsive dystonia
GDF5	Grebe syndrome
GFM1	Combined oxidative phosphorylation deficiency 1
GH1	Isolated growth hormone deficiency, type IA/II
GHRHR	Isolated growth hormone deficiency, type IB
GJB1	Charcot-Marie-Tooth disease with deafness, X-linked
GJB2	Non-syndromic hearing loss (a.k.a. connexin 26)
GJB3	Erythrokeratodermia variabilis et progressiva

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GJB6	Non-syndromic hearing loss (a.k.a. connexin 30)
GLA	Fabry disease
GLB1	Mucopolysaccharidosis, type IVB / GM1 gangliosidosis
GLDC	Glycine encephalopathy, *GLDC*-related
GLE1	Lethal congenital contracture syndrome 1
GNE	Inclusion body myopathy 2
GNPTAB	Mucolipidosis II/IIIA
GNPTG	Mucolipidosis III gamma
GNS	Mucopolysaccharidosis, type IIID (Sanfilippo D)
GORAB	Geroderma osteodysplastica
GP1BA	Bernard-Soulier syndrome, type A2
GP1BB	Bernard-Soulier syndrome, type B
GP9	Bernard-Soulier syndrome, type C
GRHPR	Primary hyperoxaluria, type 2
GUCY2D	Leber congenital amaurosis 1
GUSB	Mucopolysaccharidosis, type VII
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
HADHB	Trifunctional protein deficiency
HAX1	Congenital neutropenia, *HAX1*-related
HBA1	Alpha-thalassemia
HBA2	Alpha-thalassemia
HBB	Beta-hemoglobinopathies
HEXA	Tay-Sachs disease
HEXB	Sandhoff disease
HFE	Hemochromatosis, type 1
HFE2	Hemochromatosis, type 2A
HGD	Alkaptonuria
HGSNAT	Mucopolysaccharidosis, type IIIC (Sanfilippo C)
HLCS	Holocarboxylase synthetase deficiency
HMGCL	3-hydroxy-3-methylglutaryl-coenzyme A lyase deficiency
HMOX1	Heme oxygenase-1 deficiency
HOGA1	Primary hyperoxaluria, type 3
HPD	Tyrosinemia, type 3
HPS1	Hermansky-Pudlak syndrome 1
HPS3	Hermansky-Pudlak syndrome 3
HPS4	Hermansky-Pudlak syndrome 4
HSD17B3	17-beta hydroxysteroid dehydrogenase 3 deficiency
HSD17B4	D-bifunctional protein deficiency
HSD3B2	3-beta-hydroxysteroid dehydrogenase type II deficiency
HYLS1	Hydrolethalus syndrome
IDS	Mucopolysaccharidosis, type II (Hunter syndrome)
IDUA	Mucopolysaccharidosis, type I (Hurler syndrome)
IKBKAP	Dysautonomia, familial (*IKBKAP* or *ELP1*)
IL2RG	Severe combined immunodeficiency, X-linked
ITGB3	Glanzmann thrombasthenia
IVD	Isovaleric acidemia
KCNJ11	Congenital hyperinsulinism, *KCNJ11*-related
LAMA2	*LAMA2*-related muscular dystrophy

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LAMA3	Herlitz junctional epidermolysis bullosa, *LAMA3*-related
LAMB3	Herlitz junctional epidermolysis bullosa, *LAMB3*-related
LAMC2	Herlitz junctional epidermolysis bullosa, *LAMC2*-related
LCA5	Leber congenital amaurosis, type *LCA5*
LDLR	Familial hypercholesterolemia, *LDLR*-related
LDLRAP1	Familial hypercholesterolemia, *LDLRAP1*-related
LHCGR	Leydig cell hypoplasia
LIFR	Stuve-Wiedemann syndrome
LIPA	Lysosomal acid lipase deficiency
LIPH	Woolly hair/hypotrichosis syndrome
LOXHD1	Deafness, autosomal recessive 77
LPL	Lipoprotein lipase deficiency
LRPPRC	Leigh syndrome, French-Canadian type
LYST	Chediak-Higashi syndrome
MAN2B1	Alpha-mannosidosis
MAT1A	Hypermethioninemia
MCCC1	3-methylcrotonyl-CoA carboxylase 1 deficiency
MCCC2	3-methylcrotonyl-CoA carboxylase 2 deficiency
MCOLN1	Mucolipidosis, type IV
MECP2	RETT syndrome
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy
MEFV	Familial Mediterranean fever
MESP2	Spondylothoracic dysostosis, *MESP2*-related
MFSD8	Ceroid lipofuscinosis, neuronal, 7
MKKS	Bardet-Biedl syndrome 6
MKS1	Meckel-Gruber syndrome, type 1
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts
MLYCD	Malonyl-CoA decarboxylase deficiency
MMAA	Methylmalonic aciduria, *MMAA*-related
MMAB	Methylmalonic aciduria, *MMAB*-related
MMACHC	Methylmalonic aciduria and homocystinuria, type cblC
MMADHC	Methylmalonic aciduria and homocystinuria, type cblD
MOCS1	Molybdenum cofactor deficiency
MPI	Congenital disorder of glycosylation, type 1B
MPL	Congenital amegakaryocytic thrombocytopenia
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome, *MPV17*-related
MRE11	Ataxia-telangiectasia-like disorder 1
MTHFR	Homocystinuria due to deficiency of *MTHFR*
MTM1	Myotubular myopathy, X-linked
MTRR	Homocystinuria, type cblE
MTTP	Abetalipoproteinemia
MUT	Methylmalonic aciduria, type mut(0)
MYO15A	Deafness, autosomal recessive, 3
MYO7A	Usher syndrome, type 1B
NAGLU	Mucopolysaccharidosis, type IIIB (Sanfilippo B)
NAGS	N-acetylglutamate synthase deficiency
NBN	Nijmegen breakage syndrome
NDRG1	Charcot-Marie-Tooth disease type 4D

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NDUFAF5	Mitochondrial complex I deficiency, *NDUFAF5*-related
NDUFS4	Mitochondrial complex I deficiency
NDUFS6	Mitochondrial complex I deficiency, *NDUFS6*-related
NEB	Nemaline myopathy, *NEB*-related
NEU1	Sialidosis
NLRP7	Hydatidiform mole, recurrent
NPC1	Niemann-Pick disease, type C1/D
NPC2	Niemann-Pick disease, type C2
NPHP1	Juvenile nephronophthisis
NPHS1	Congenital Finnish nephrosis
NPHS2	Steroid-resistant nephrotic syndrome
NR0B1	Congenital adrenal hypoplasia, X-linked
NR2E3	Enhanced S-cone syndrome
NTRK1	Congenital insensitivity to pain with anhidrosis (CIPA)
OAT	Ornithine aminotransferase deficiency
OCRL	Lowe syndrome, X-linked
OPA3	Costeff syndrome (3-methylglutaconic aciduria, type 3)
OTC	Ornithine transcarbamylase deficiency
PAH	Phenylketonuria
PANK2	Pantothenate kinase-associated neurodegeneration
PC	Pyruvate carboxylase deficiency
PCCA	Propionic acidemia, *PCCA*-related
PCCB	Propionic acidemia, *PCCB*-related
PCDH15	Usher syndrome, type 1F
PDHA1	Pyruvate dehydrogenase deficiency, X-linked
PDHB	Pyruvate dehydrogenase deficiency, *PDHB*-related
PEPD	Prolidase deficiency
PET100	Cytochrome-c oxidase deficiency
PEX1	Peroxisome biogenesis disorder 1A (Zellweger)
PEX10	Peroxisome biogenesis disorder 6A (Zellweger)
PEX12	Peroxisome biogenesis disorder 3A (Zellweger)
PEX2	Peroxisome biogenesis disorder 5A (Zellweger)
PEX6	Peroxisome biogenesis disorder 4A (Zellweger)
PEX7	Rhizomelic chondrodysplasia punctata, type 1
PFKM	Glycogen storage disease, type VII
PHGDH	Phosphoglycerate dehydrogenase deficiency
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1
PKHD1	Polycystic kidney disease, autosomal recessive
PLA2G6	Infantile neuroaxonal dystrophy 1
PMM2	Congenital disorder of glycosylation, type 1A, *PMM2*-related
PNPO	Pyridoxal 5´-phosphate-dependent epilepsy
POLG	*POLG*-related disorders
POLH	Xeroderma pigmentosum variant
POMGNT1	Muscle-eye-brain disease, *POMGNT1*-related
POR	Cytochrome P450 oxidoreductase deficiency
PPT1	Ceroid lipofuscinosis, neuronal, 1
PREPL	Myasthenic syndrome, congenital, 22
PROP1	Combined pituitary hormone deficiency 2

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PRPS1	Arts syndrome, X-linked
PSAP	Metachromatic leukodystrophy, *PSAP*-related
PTS	6-pyruvoyl-tetrahydropterin synthase (PTPS) deficiency
PUS1	Mitochondrial myopathy and sideroblastic anemia (MLASA1)
PYGM	Glycogen storage disease, type V (McArdle disease)
RAB23	Carpenter syndrome
RAG1	Omenn syndrome, *RAG1*-related
RAG2	Omenn syndrome, *RAG2*-related
RAPSN	Congenital myasthenic syndrome, *RAPSN*-related
RARS2	Pontocerebellar hypoplasia, type 1 and 6, *RARS2*-related
RDH12	Leber congenital amaurosis, type *RDH12*
RLBP1	Retinal dystrophies, *RLBP1*-associated
RMRP	Cartilage-hair hypoplasia
RNASEH2C	Aicardi-Goutieres syndrome, *RNASEH2C*-related
RPE65	Leber congenital amaurosis 2
RPGRIP1L	Ciliopathies, *RPGRIP1L*-related
RS1	Juvenile retinoschisis, X-linked
RTEL1	Dyskeratosis congenita, *RTEL1*-related
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay
SAMD9	MIRAGE syndrome
SAMHD1	Aicardi-Goutires syndrome
SBDS	Shwachman-Diamond syndrome
SEPSECS	Pontocerebellar hypoplasia, type 2D
SERPINA1	Alpha-1-antitrypsin deficiency
SGCA	Limb-girdle muscular dystrophy, type 2D
SGCB	Limb-girdle muscular dystrophy, type 2E
SGCD	Limb-girdle muscular dystrophy, type 2F
SGCG	Limb-girdle muscular dystrophy, type 2C
SGSH	Mucopolysaccharidosis, type IIIA (Sanfilippo A)
SLC12A3	Gitelman syndrome
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy (Andermann syndrome)
SLC17A5	Salla disease
SLC19A2	Megaloblastic anemia syndrome
SLC22A5	Carnitine deficiency
SLC25A13	Citrullinemia, type II
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome
SLC25A20	Carnitine-acylcarnitine translocase deficiency
SLC26A2	Achondrogenesis, type 1B
SLC26A3	Congenital chloride diarrhea
SLC26A4	Pendred syndrome
SLC35A3	Autism spectrum, epilepsy, and arthrogryposis
SLC37A4	Glycogen storage disease, type IB
SLC39A4	Acrodermatitis enteropathica
SLC3A1	Cystinuria, type A
SLC45A2	Oculocutaneous albinism, type 4
SLC4A11	Corneal dystrophy and perceptive deafness
SLC6A8	Creatine transporter defect (cerebral creatine deficiency syndrome 1, X-linked)
SLC7A7	Lysinuric protein intolerance

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SLC7A9	Cystinuria, type B
SMARCAL1	Schimke immunoosseous dysplasia
SMN1	Spinal muscular atrophy
SMPD1	Niemann-Pick disease, types A/B
SRD5A2	5-alpha reductase deficiency
ST3GAL5	GM3 synthase deficiency
STAR	Lipoid congenital adrenal hyperplasia
STRC	Deafness, autosomal recessive 16
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
SUMF1	Multiple sulfatase deficiency
SURF1	Leigh syndrome
TAT	Tyrosinemia, type II
TCIRG1	Osteopetrosis, infantile malignant, *TCIRG1*-related
TECPR2	Hereditary spastic paraparesis, type 49
TFR2	Hemochromatosis, type 3, *TFR2*-related
TGM1	Lamellar ichthyosis, type 1
TH	Segawa syndrome, *TH*-related
TMC1	Deafness, autosomal dominant 36, autosomal recessive 7
TMEM216	Joubert syndrome 2/Meckel syndrome 2
TPO	Congenital hypothyroidism
TPP1	Ceroid lipofuscinosis, neuronal, 2
TREX1	Aicardi-Goutieres syndrome, *TREX1*-related
TRIM32	Bardet-Biedl syndrome 11
TRIM37	Mulibrey nanism syndrome
TRMU	Acute infantile liver failure, *TRMU*-related
TSEN54	Pontocerebellar hypoplasia
TSFM	Combined oxidative phosphorylation deficiency 3
TSHB	Congenital hypothyroidism
TSHR	Hypothyroidism, congenital, nongoitrous, 1
TTC37	Tricho-hepato-enteric syndrome
TTN	Familial dilated cardiomyopathy
TTPA	Ataxia with vitamin E deficiency
TYMP	Myoneurogastrointestinal encephalopathy (MNGIE)
TYR	Oculocutaneous albinism, type 1
TYRP1	Oculocutaneous albinism, type 3
UGT1A1	Crigler-Najjar syndrome
UPB1	Beta-ureidopropionase deficiency
USH1C	Usher syndrome, type 1C
USH2A	Usher syndrome, type 2A
VPS13A	Choreo-acanthocytosis
VPS13B	Cohen syndrome
VPS45	Congenital neutropenia, *VPS45*-related
VPS53	Pontocerebellar hypoplasia, type 2E
VRK1	Pontocerebellar hypoplasia, type 1A
VSX2	Microphthalmia/Anophthalmia, *VSX2*-related
VWF	Von Willebrand disease
WAS	Wiskott-Aldrich syndrome, X-linked

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WISP3	Progressive pseudorheumatoid dysplasia
WNT10A	Odonto-onycho-dermal dysplasia/Schopf-Schulz-Passarge syndrome
WRN	Werner syndrome
XPA	Xeroderma pigmentosum group A
XPC	Xeroderma pigmentosum group C
ZFYVE26	Spastic paraplegia type 15

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