virtusGENETICS
A MEMBER OF VIRTUS HEALTH

virtus duo carrier screen

361 genes in males; 390 genes in females

361 Autosomal Recessive (AR) genes

CLICK HERE TO VIEW X-LINKED (XL) GENES

GENE	CONDITION	INHERITANCE
AAAS	Achalasia-Addisonianism-Alacrima Syndrome	AR
ABCA12	Harlequin ichthyosis	AR
ABCB11	Progressive Familial Intrahepatic Cholestasis, Type 2	AR
ABCB4	Progressive Familial Intrahepatic Cholestasis, Type 3	AR
ABCC6	Pseudoxanthoma elasticum	AR
ABCC8	Familial Hyperinsulinism, ABCC8-Related	AR
ACAD9	Mitochondrial Complex I Deficiency, ACAD9-Related	AR
ACADM	Medium Chain Acyl-CoA Dehydrogenase Deficiency	AR
ACADVL	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	AR
ACAT1	Beta-Ketothiolase Deficiency	AR
ACOX1	Acyl-CoA Oxidase I Deficiency	AR
ACSF3	Combined Malonic and Methylmalonic Aciduria	AR
ADA	Severe Combined Immunodeficiency, ADA-Related	AR
ADAMTS2	Ehlers-Danlos Syndrome, Type VIIC	AR
ADGRG1	Bilateral Frontoparietal Polymicrogyria	AR
AGA	Aspartylglucosaminuria	AR
AGL	Glycogen Storage Disease, Type III (Cori/Forbes)	AR
AGPS	Rhizomelic Chondrodysplasia Punctata, Type 3	AR
AGXT	Hyperoxaluria, Primary, Type 1	AR
AIRE	Autoimmune polyendocrinopathy syndrome, type I	AR
ALDH3A2	Sjogren-Larsson Syndrome	AR
ALDH7A1	Pyridoxine-dependent epilepsy	AR
ALDOB	Hereditary Fructose Intolerance	AR
ALG6	Congenital Disorder of Glycosylation, Type 1C	AR
ALMS1	Alstrom Syndrome	AR
ALPL	Hypophosphatasia, ALPL-Related	AR
AMT	Glycine Encephalopathy, AMT-Related	AR
AP1S1	Mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis, and keratoderma (MEDNIK syndrome)	AR

GENE	CONDITION	INHERITANCE
AQP2	Familial Nephrogenic Diabetes Insipidus, AQP2-Related	AR
ARG1	Argininemia	AR
ARSA	Metachromatic Leukodystrophy, ARSA-Related	AR
ARSB	Mucopolysaccharidosis, Type VI (Maroteaux-Lamy)	AR
ASL	Argininosuccinate Lyase Deficiency	AR
ASNS	Asparagine Synthetase Deficiency	AR
ASPA	Canavan Disease	AR
ASS1	Citrullinemia, Type 1	AR
ATM	Ataxia-Telangiectasia	AR
ATP6V1B1	Renal Tubular Acidosis and Deafness, ATP6V1B1-Related	AR
ATP7B	Wilson Disease	AR
ATP8B1	Progressive Familial Intrahepatic Cholestasis, Type 1	AR
BBS1	Bardet-Biedl Syndrome 1	AR
BBS10	Bardet-Biedl Syndrome 10	AR
BBS12	Bardet-Biedl Syndrome 12	AR
BBS2	Bardet-Biedl Syndrome 2	AR
BBS4	Bardet-Biedl Syndrome 4	AR
BBS9	Bardet-Biedl Syndrome 9	AR
BCKDHA	Maple Syrup Urine Disease, Type 1A	AR
BCKDHB	Maple Syrup Urine Disease, Type 1B	AR
BCS1L	GRACILE Syndrome	AR
BLM	Bloom Syndrome	AR
BSND	Bartter syndrome, Type 4a	AR
BTD	Biotinidase Deficiency	AR
CANT1	Desbuquois dysplasia 1	AR
CAPN3	Limb-Girdle Muscular Dystrophy, Type 2A	AR
CASQ2	Catecholaminergic polymorphic ventricular tachycardia	AR
CBS	Homocystinuria, CBS-Related	AR
CC2D1A	Mental retardation, autosomal recessive 3	AR
CCN6	Progressive Pseudorheumatoid Dysplasia (WISP3 or CCN6)	AR
CDH23	Usher Syndrome, Type 1D	AR
CEP290	Leber Congenital Amaurosis, Type CEP290	AR
CERKL	Retinitis Pigmentosa 26	AR
CFTR	Cystic Fibrosis	AR
CHRNE	Congenital Myasthenic Syndrome, CHRNE-Related	AR
CHRNG	Escobar Syndrome	AR

GENE	CONDITION	INHERITANCE
CIITA	Bare Lymphocyte Syndrome, CIITA-Related	AR
CLN3	Ceroid Lipofuscinosis, Neuronal, 3	AR
CLN5	Ceroid Lipofuscinosis, Neuronal, 5	AR
CLN6	Ceroid Lipofuscinosis, Neuronal, 6	AR
CLN8	Ceroid Lipofuscinosis, Neuronal, 8 (Northern Epilepsy)	AR
CLRN1	Usher Syndrome, Type 3	AR
CNGA3	Achromatopsia, CNGA3-Related	AR
CNGB3	Achromatopsia, CNGB3-Related	AR
COL11A2	Fibrochondrogenesis type 2	AR
COL4A3	Alport Syndrome, COL4A3-Related	AR
COL4A4	Alport Syndrome, COL4A4-Related	AR
COL7A1	Dystrophic Epidermolysis Bullosa, COL7A1-Related	AR
CPS1	Carbamoyl Phosphate Synthetase I Deficiency	AR
CPT1A	Carnitine Palmitoyltransferase IA Deficiency	AR
CPT2	Carnitine Palmitoyltransferase II Deficiency	AR
CRB1	Leber congenital amaurosis 8	AR
CTNS	Cystinosis	AR
CTSC	Papillon-Lefevre Syndrome	AR
CTSD	Ceroid Lipofuscinosis, Neuronal, 10 (CLN10 Disease)	AR
CTSK	Pycnodysostosis	AR
CYBA	Chronic Granulomatous Disease, CYBA-Related	AR
CYP11B1	Congenital Adrenal Hyperplasia, 11-beta-hydroxylase-deficient	AR
CYP11B2	Corticosterone Methyloxidase Deficiency	AR
CYP17A1	Congenital Adrenal Hyperplasia, 17-Alpha-Hydroxylase Deficiency	AR
CYP19A1	Aromatase Deficiency	AR
CYP1B1	Primary Congenital Glaucoma	AR
CYP21A2	Congenital Adrenal Hyperplasia, 21-hydroxylase-deficient	AR
CYP27A1	Cerebrotendinous Xanthomatosis	AR
CYP27B1	Vitamin D-dependent rickets type 1A	AR
DBT	Maple Syrup Urine Disease, Type 2	AR
DCLRE1C	Severe Combined Immunodeficiency, Type Athabaskan	AR
DDB2	Xeroderma Pigmentosum Group E	AR
DHCR7	Smith-Lemli-Opitz Syndrome	AR
DHDDS	Retinitis Pigmentosa 59	AR
DLD	Dihydrolipoamide Dehydrogenase Deficiency	AR
DNAH5	Ciliary Dyskinesia, Primary 3	AR

GENE	CONDITION	INHERITANCE
DNAI1	Ciliary Dyskinesia, Primary 1	AR
DNAI2	Ciliary Dyskinesia, Primary 9	AR
DNAL1	Ciliary Dyskinesia, Primary, 16	AR
DOK7	Congenital Myasthenic Syndrome, DOK7-Related	AR
DPYD	Dihydropyrimidine Dehydrogenase Deficiency	AR
DYSF	Limb-Girdle Muscular Dystrophy, Type 2B	AR
EDAR	Hypohidrotic Ectodermal Dysplasia	AR
EIF2AK3	Wolcott-Rallison Syndrome	AR
EIF2B5	Leukoencephalopathy with Vanishing White Matter	AR
ELP1	Dysautonomia, familial (IKBKAP or ELP1)	AR
ERCC2	Xeroderma Pigmentosum Group D	AR
ERCC3	Xeroderma Pigmentosum Group B	AR
ERCC4	Xeroderma Pigmentosum Group F	AR
ERCC5	Xeroderma pigmentosum Group G	AR
ERCC6	Cockayne syndrome, type B	AR
ERCC8	Cockayne syndrome, type A	AR
ESCO2	Roberts Syndrome	AR
ETFA	Glutaric Acidemia, Type 2A	AR
ETFB	Glutaric Acidemia, Type 2B	AR
ETFDH	Glutaric Acidemia, Type 2C	AR
ETHE1	Ethylmalonic Encephalopathy	AR
EVC	Ellis-van Creveld Syndrome, EVC-Related	AR
EVC2	Ellis-van Creveld Syndrome, EVC2-related	AR
EXOSC3	Pontocerebellar Hypoplasia, Type 1B	AR
EYS	Retinitis Pigmentosa 25	AR
FAH	Tyrosinemia, Type I	AR
FAM161A	Retinitis Pigmentosa 28	AR
FANCA	Fanconi Anemia, Group A	AR
FANCC	Fanconi Anemia, Group C	AR
FANCG	Fanconi Anemia, Group G	AR
FH	Fumarase Deficiency	AR
FKRP	Limb-Girdle Muscular Dystrophy, Type 2I	AR
FKTN	Walker-Warburg Syndrome, FKTN-Related	AR
G6PC	Glycogen Storage Disease, Type IA	AR
GAA	Glycogen Storage Disease, Type II (Pompe Disease)	AR
GALC	Krabbe Disease	AR

GENE	CONDITION	INHERITANCE
GALE	Galactose epimerase deficiency	AR
GALK1	Galactokinase Deficiency (Galactosemia, Type II)	AR
GALNS	Mucopolysaccharidosis, Type IVA	AR
GALNT3	Hyperphosphatemic familial tumoral calcinosis	AR
GALT	Galactosemia	AR
GAMT	Guanidinoacetate Methyltransferase Deficiency	AR
GBA1	Gaucher Disease	AR
GBE1	Glycogen Storage Disease, Type IV	AR
GCDH	Glutaric Acidemia, Type 1	AR
GCH1	Dopa-responsive dystonia	AR
GDF5	Grebe syndrome	AR
GFM1	Combined Oxidative Phosphorylation Deficiency 1	AR
GH1	Isolated growth hormone deficiency, Type IA/II	AR
GHRHR	Isolated growth hormone deficiency, Type IB	AR
GJB2	Deafness, autosomal recessive 1A	AR
GJB6	Non-Syndromic Hearing Loss (Connexin 30)	AR
GLB1	Mucopolysaccharidosis, Type IVB / GM1 Gangliosidosis	AR
GLDC	Glycine Encephalopathy, GLDC-Related	AR
GLE1	Lethal Congenital Contracture Syndrome 1	AR
GNE	Inclusion Body Myopathy 2	AR
GNPTAB	Mucolipidosis II/IIIA	AR
GNPTG	Mucolipidosis III gamma	AR
GNS	Mucopolysaccharidosis, Type IIID (Sanfilippo D)	AR
GORAB	Geroderma osteodysplastica	AR
GUCY2D	Leber congenital amaurosis 1	AR
GUSB	Mucopolysaccharidosis, Type VII	AR
HADHA	Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	AR
HADHB	Trifunctional protein deficiency	AR
HAX1	Congenital Neutropenia, HAX1-Related	AR
HBA1	Alpha Thalassemia	AR
HBA2	Alpha Thalassemia	AR
HBB	Beta Hemoglobinopathies	AR
HEXA	Tay-Sachs Disease	AR
HEXB	Sandhoff Disease	AR
HJV	Hemochromatosis, Type 2A (HFE2 or HJV)	AR
HGSNAT	Mucopolysaccharidosis, Type IIIC (Sanfilippo C)	AR

GENE	CONDITION	INHERITANCE
HLCS	Holocarboxylase Synthetase Deficiency	AR
HMGCL	3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	AR
HMOX1	Heme Oxygenase-1 Deficiency	AR
HPD	Tyrosinemia, Type III	AR
HPS1	Hermansky-Pudlak Syndrome 1	AR
HPS3	Hermansky-Pudlak Syndrome 3	AR
HPS4	Hermansky-Pudlak syndrome 4	AR
HSD17B3	17-beta hydroxysteroid dehydrogenase 3 deficiency	AR
HSD17B4	D-Bifunctional Protein Deficiency	AR
HSD3B2	3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency	AR
HYLS1	Hydrolethalus Syndrome	AR
IDUA	Mucopolysaccharidosis, Type I (Hurler Syndrome)	AR
IVD	Isovaleric Acidemia	AR
KCNJ11	Congenital Hyperinsulinism, KCNJ11-Related	AR
LAMA2	LAMA2-related Muscular Dystrophy	AR
LAMA3	Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related	AR
LAMB3	Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related	AR
LAMC2	Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related	AR
LCA5	Leber Congenital Amaurosis, Type LCA5	AR
LDLR	Familial Hypercholesterolemia, LDLR-Related	AR
LDLRAP1	Familial Hypercholesterolemia, LDLRAP1-Related	AR
LHCGR	Leydig cell hypoplasia	AR
LIFR	Stuve-Wiedemann Syndrome	AR
LIPA	Lysosomal Acid Lipase Deficiency	AR
LOXHD1	Deafness, Autosomal Recessive 77	AR
LPL	Lipoprotein Lipase Deficiency	AR
LRPPRC	Leigh Syndrome, French-Canadian Type	AR
LYST	Chediak-Higashi syndrome	AR
MAN2B1	Alpha Mannosidosis	AR
MCOLN1	Mucolipidosis, Type IV	AR
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy	AR
MESP2	Spondylothoracic Dysostosis, MESP2-Related	AR
MFSD8	Ceroid Lipofuscinosis, Neuronal, 7	AR
MKKS	Bardet-Biedl Syndrome 6	AR
MKS1	Meckel-Gruber Syndrome, Type 1	AR
MLC1	Megalencephalic Leukoencephalopathy with Subcortical Cysts	AR

GENE	CONDITION	INHERITANCE
MLYCD	Malonyl-CoA decarboxylase deficiency	AR
MMAA	Methylmalonic Aciduria, MMAA-Related	AR
MMAB	Methylmalonic Aciduria, MMAB-Related	AR
MMACHC	Methylmalonic Aciduria and Homocystinuria, Type cblC	AR
MMADHC	Methylmalonic Aciduria and Homocystinuria, Type cblD	AR
MOCS1	Molybdenum cofactor deficiency	AR
MPI	Congenital Disorder of Glycosylation, Type 1B	AR
MPL	Congenital Amegakaryocytic Thrombocytopenia	AR
MPV17	Hepatocerebral Mitochondrial DNA Depletion Syndrome, MPV17-Related	AR
MRE11	Ataxia-telangiectasia-like disorder 1	AR
MTHFR	Homocystinuria due to Deficiency of MTHFR	AR
MTRR	Homocystinuria, Type cblE	AR
MTTP	Abetalipoproteinemia	AR
MMUT	Methylmalonic Aciduria, Type mut(0) (MUT or MMUT)	AR
MYO15A	Deafness, autosomal recessive, 3	AR
MYO7A	Usher Syndrome, Type 1B	AR
NAGLU	Mucopolysaccharidosis, Type IIIB (Sanfilippo B)	AR
NAGS	N-acetylglutamate Synthase Deficiency	AR
NBN	Nijmegen Breakage Syndrome	AR
NDRG1	Charcot-Marie-Tooth Disease type 4D	AR
NDUFAF5	Mitochondrial Complex I Deficiency, NDUFAF5-Related	AR
NDUFS4	Mitochondrial complex I deficiency	AR
NDUFS6	Mitochondrial Complex I Deficiency, NDUFS6-Related	AR
NEB	Nemaline Myopathy, NEB-Related	AR
NEU1	Sialidosis	AR
NPC1	Niemann-Pick Disease, Type C1/D	AR
NPC2	Niemann-Pick Disease, Type C2	AR
NPHP1	Juvenile Nephronophthisis	AR
NPHS1	Congenital Finnish Nephrosis	AR
NPHS2	Steroid-Resistant Nephrotic Syndrome	AR
NR2E3	Enhanced S-Cone Syndrome	AR
NTRK1	Congenital Insensitivity to Pain with Anhidrosis (CIPA)	AR
OAT	Ornithine Aminotransferase Deficiency	AR
OPA3	Costeff Syndrome (3-Methylglutaconic Aciduria, Type 3)	AR
PAH	Phenylketonuria	AR
PANK2	Pantothenate Kinase-Associated Neurodegeneration	AR

GENE	CONDITION	INHERITANCE
PC	Pyruvate Carboxylase Deficiency	AR
PCCA	Propionic Acidemia, PCCA-Related	AR
PCCB	Propionic Acidemia, PCCB-Related	AR
PCDH15	Usher Syndrome, Type 1F	AR
PDHB	Pyruvate Dehydrogenase Deficiency, PDHB-Related	AR
PEPD	Prolidase deficiency	AR
PET100	Cytochrome-c oxidase deficiency	AR
PEX1	Peroxisome Biogenesis Disorder 1A (Zellweger)	AR
PEX10	Peroxisome Biogenesis Disorder 6A (Zellweger)	AR
PEX12	Peroxisome Biogenesis Disorder 3A (Zellweger)	AR
PEX2	Peroxisome Biogenesis Disorder 5A (Zellweger)	AR
PEX6	Peroxisome Biogenesis Disorder 4A (Zellweger)	AR
PEX7	Rhizomelic Chondrodysplasia Punctata, Type 1	AR
PFKM	Glycogen Storage Disease, Type VII	AR
PHGDH	Phosphoglycerate Dehydrogenase Deficiency	AR
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1	AR
PKHD1	Polycystic Kidney Disease, Autosomal Recessive	AR
PLA2G6	Infantile neuroaxonal dystrophy 1	AR
PMM2	Congenital Disorder of Glycosylation, Type 1A, PMM2-Related	AR
PNPO	Pyridoxal 5'-phosphate-dependent epilepsy	AR
POLG	POLG-Related Disorders	AR
POLH	Xeroderma pigmentosum Variant	AR
POMGNT1	Muscle-Eye-Brain Disease, POMGNT1-Related	AR
POR	Cytochrome P450 oxidoreductase deficiency	AR
PPT1	Ceroid Lipofuscinosis, Neuronal, 1	AR
PREPL	Myasthenic syndrome, congenital, 22	AR
PROP1	Combined Pituitary Hormone Deficiency 2	AR
PSAP	Metachromatic Leukodystrophy, PSAP-Related	AR
PTS	6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency	AR
PUS1	Mitochondrial Myopathy and Sideroblastic Anaemia (MLASA1)	AR
RAB23	Carpenter Syndrome	AR
RAG1	Omenn Syndrome, RAG1-Related	AR
RAG2	Omenn Syndrome, RAG2-Related	AR
RAPSN	Congenital Myasthenic Syndrome, RAPSN-Related	AR
RARS2	Pontocerebellar Hypoplasia, Type 1 and 6, RARS2-Related	AR
RDH12	Leber Congenital Amaurosis, Type RDH12	AR

GENE	CONDITION	INHERITANCE
RLBP1	Retinal Dystrophies, RLBP1-Associated	AR
RMRP	Cartilage-Hair Hypoplasia	AR
RNASEH2C	Aicardi-Goutieres syndrome, RNASEH2C-related	AR
RPE65	Leber Congenital Amaurosis 2	AR
RPGRIP1L	Ciliopathies, RPGRIP1L-Related	AR
RTEL1	Dyskeratosis Congenita, RTEL1-Related	AR
SACS	Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	AR
SAMD9	MIRAGE syndrome	AR
SAMHD1	Aicardi-Goutieres Syndrome	AR
SBDS	Shwachman-Diamond syndrome	AR
SEPSECS	Pontocerebellar Hypoplasia, Type 2D	AR
SERPINA1	Alpha-1-Antitrypsin Deficiency	AR
SGCA	Limb-Girdle Muscular Dystrophy, Type 2D	AR
SGCB	Limb-Girdle Muscular Dystrophy, Type 2E	AR
SGCD	Limb-Girdle Muscular Dystrophy, Type 2F	AR
SGCG	Limb-Girdle Muscular Dystrophy, Type 2C	AR
SGSH	Mucopolysaccharidosis, Type IIIA (Sanfilippo A)	AR
SLC12A3	Gitelman Syndrome	AR
SLC12A6	Agenesis of the Corpus Callosum with Peripheral Neuropathy (Andermann Syndrome)	AR
SLC17A5	Salla Disease	AR
SLC19A2	Megaloblastic Anaemia Syndrome	AR
SLC22A5	Carnitine Deficiency	AR
SLC25A13	Citrullinemia, Type II	AR
SLC25A15	Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome	AR
SLC25A20	Carnitine-acylcarnitine translocase deficiency	AR
SLC26A2	Achondrogenesis, Type 1B	AR
SLC26A3	Congenital Chloride Diarrhea	AR
SLC26A4	Pendred Syndrome	AR
SLC35A3	Autism Spectrum, Epilepsy and Arthrogryposis	AR
SLC37A4	Glycogen Storage Disease, Type IB	AR
SLC39A4	Acrodermatitis Enteropathica	AR
SLC45A2	Oculocutaneous albinism, Type 4	AR
SLC7A7	Lysinuric Protein Intolerance	AR
SMARCAL1	Schimke Immunoosseous Dysplasia	AR
SMN1	Spinal Muscular Atrophy	AR

GENE	CONDITION	INHERITANCE
SMPD1	Niemann-Pick Disease, Types A/B	AR
SRD5A2	5-alpha reductase deficiency	AR
ST3GAL5	GM3 synthase deficiency	AR
STAR	Lipoid Congenital Adrenal Hyperplasia	AR
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	AR
SUMF1	Multiple Sulfatase Deficiency	AR
SURF1	Leigh Syndrome	AR
TAT	Tyrosinemia, Type II	AR
TCIRG1	Osteopetrosis, Infantile Malignant, TCIRG1-Related	AR
TECPR2	Hereditary Spastic Paraparesis, Type 49	AR
TFR2	Hemochromatosis, Type 3, TFR2-Related	AR
TGM1	Lamellar Ichthyosis, Type 1	AR
TH	Segawa Syndrome, TH-Related	AR
TMC1	Deafness, autosomal dominant 36, autosomal recessive 7	AR
TMEM216	Joubert Syndrome 2 / Meckel Syndrome 2	AR
TPO	Congenital hypothyroidism	AR
TPP1	Ceroid Lipofuscinosis, Neuronal, 2	AR
TREX1	Aicardi-Goutieres syndrome, TREX1-related	AR
TRIM32	Bardet-Biedl syndrome 11	AR
TRIM37	Mulibrey nanism syndrome	AR
TRMU	Acute Infantile Liver Failure, TRMU-Related	AR
TSEN54	Pontocerebellar hypoplasia	AR
TSFM	Combined Oxidative Phosphorylation Deficiency 3	AR
TSHB	Congenital hypothyroidism	AR
TSHR	Hypothyroidism, congenital, nongoitrous, 1	AR
SKIC3	Tricho-Hepato-Enteric Syndrome (TTC37 or SKIC3)	AR
TTN	Familial dilated cardiomyopathy	AR
TTPA	Ataxia with Vitamin E Deficiency	AR
TYMP	Myoneurogastrointestinal Encephalopathy (MNGIE)	AR
TYR	Oculocutaneous Albinism, Type 1	AR
TYRP1	Oculocutaneous albinism, Type 3	AR
UGT1A1	Crigler-Najjar Syndrome	AR
USH1C	Usher Syndrome, Type 1C	AR
USH2A	Usher Syndrome, Type 2A	AR
VPS13A	Choreo-acanthocytosis	AR

GENE	CONDITION	INHERITANCE
VPS13B	Cohen Syndrome	AR
VPS45	Congenital Neutropenia, VPS45-Related	AR
VPS53	Pontocerebellar Hypoplasia, Type 2E	AR
VRK1	Pontocerebellar Hypoplasia, Type 1A	AR
VSX2	Microphthalmia/Anophthalmia, VSX2-Related	AR
VWF	Von Willebrand disease	AR
WNT10A	Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome	AR
WRN	Werner Syndrome	AR
XPA	Xeroderma pigmentosum Group A	AR
XPC	Xeroderma Pigmentosum Group C	AR
ZFYVE26	Spastic Paraplegia Type 15	AR

29 X-linked genes (XL)

GENE	CONDITION	INHERITANCE
ABCD1	Adrenoleukodystrophy, X-Linked	XL
AR	Androgen insensitivity syndrome, X-Linked	XL
ATP7A	Menkes Syndrome, X-Linked	XL
ATRX	Alpha-Thalassemia Intellectual Disability Syndrome, X-Linked	XL
BTK	Isolated growth hormone deficiency, Type III, X-linked	XL
CHM	Choroideremia, X-Linked	XL
COL4A5	Alport Syndrome, X-Linked	XL
CYBB	Chronic Granulomatous Disease, X-Linked	XL
DKC1	Dyskeratosis congenita, X-Linked	XL
DMD	Duchenne/Becker Muscular Dystrophy	XL
EDA	Hypohidrotic Ectodermal Dysplasia, X-Linked	XL
EMD	Emery-Dreifuss Muscular Dystrophy 1, X-Linked	XL
F8	Haemophilia A	XL
F9	Haemophilia B	XL
FMR1	Fragile X syndrome	XL
GJB1	Charcot-Marie-Tooth Disease with Deafness, X-Linked	XL
GLA	Fabry Disease	XL
IDS	Mucopolysaccharidosis, Type II (Hunter Syndrome)	XL
IL2RG	Severe Combined Immunodeficiency, X-Linked	XL
MECP2	Intellectual developmental disorder, X-linked syndromic	XL
MTM1	Myotubular Myopathy, X-Linked	XL
NR0B1	Congenital Adrenal Hypoplasia, X-linked	XL

GENE	CONDITION	INHERITANCE
OCRL	Lowe syndrome, X-Linked	XL
OTC	Ornithine Transcarbamylase Deficiency	XL
PDHA1	Pyruvate Dehydrogenase Deficiency, X-Linked	XL
PRPS1	Arts syndrome, X-Linked	XL
RS1	Juvenile Retinoschisis, X-Linked	XL
SLC6A8	Creatine Transporter Defect (Cerebral Creatine Deficiency Syndrome 1, X-Linked)	XL
WAS	Wiskott-Aldrich syndrome, X-Linked	XL