

Način nasleđivanja	Naziv bolesti	Gen
AR	Progressive familial intrahepatic cholestasis type 2 (AR) NM_003742.2	ABCB11
AR	ABCC8-related disorders (AR) NM_000352.4	ABCC8
AR	ACAD9 deficiency (AR) NM_014049.4	ACAD9
AR	Medium chain acyl-CoA dehydrogenase deficiency (AR) NM_000016.5	ACADM
AR	Very long-chain acyl-CoA dehydrogenase deficiency (AR) NM_000018.3	ACADVL
AR	Beta-ketothiolase deficiency (AR) NM_000019.3	ACAT1
AR	Peroxisomal acyl-CoA oxidase deficiency (AR) NM_004035.6	ACOX1
AR	Combined malonic and methylmalonic aciduria (ACSF3-related) (AR) NM_174917.4	ACSF3
AR	Adenosine deaminase deficiency (AR) NM_000022.2	ADA
AR	Polymicrogyria (ADGRG1-related) (AR) NM_005682.6	ADGRG1
AR	Aspartylglucosaminuria (AR) NM_000027.3	AGA
AR	Glycogen storage disease type III (AR) NM_000642.2	AGL
AR	Rhizomelic chondrodysplasia punctata type 3 (AR) NM_003659.3	AGPS
AR	Primary hyperoxaluria type 1 (AR) NM_000030.2	AGXT
AR	Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia (AR) NM_000383.3	AIRE
AR	Sjögren-Larsson syndrome (AR) NM_000382.2	ALDH3A2
AR	Hereditary fructose intolerance (AR) NM_000035.3	ALDOB
AR	Congenital disorder of glycosylation (ALG6-related) (AR) NM_013339.3	ALG6
AR	Hypophosphatasia (AR) NM_000478.5	ALPL
AR	Glycine encephalopathy (AMT-related) (AR) NM_000481.3	AMT
AR	Nephrogenic diabetes insipidus (AQP2-related) (AR) NM_000486.5	AQP2
AR	Arginase deficiency (AR) NM_000045.3	ARG1
AR	Metachromatic leukodystrophy (ARSA-related) (AR) NM_000487.5	ARSA
AR	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome) (AR) NM_000046.3	ARSB
AR	Argininosuccinic aciduria (AR) NM_000048.3	ASL
AR	Canavan disease (AR) NM_000049.2	ASPA
AR	Citrullinemia type 1 (AR) NM_000050.4	ASS1
AR	Ataxia telangiectasia (AR) NM_000051.3	ATM
AR	Ataxia with vitamin E deficiency (AR) NM_000370.3	ATP6V1B1
XL	Menkes disease/ATP7A-related disorders (XL) NM_000052.6	ATP7A
AR	Wilson Disease (AR) NM_000053.3	ATP7B
AR	BBS1-related disorders (AR) NM_024649.4	BBS1
AR	Bardet-Biedl syndrome (BBS10-related) (AR) NM_024685.3	BBS10
AR	Bardet-Biedl syndrome (BBS12-related)(AR) NM_152618.2	BBS12
AR	BBS2-related disorders (AR) NM_031885.3	BBS2
AR	Maple syrup urine disease type 1A (AR) NM_000709.3	BCKDHA
AR	Maple syrup urine disease type 1B (AR) NM_183050.2	BCKDHB
AR	GRACILE syndrome/BCS1L-related disorders (AR) NM_004328.4	BCS1L
AR	Bloom syndrome (AR) NM_000057.3	BLM
AR	Bartter syndrome type 4A (AR) NM_057176.2	BSND
AR	Biotinidase deficiency (AR) NM_000060.3	BTD
AR	Limb-girdle muscular dystrophy type 2A (calpainopathy) (AR) NM_000070.2s	CAPN3
AR	Homocystinuria due to CBS deficiency (AR) NM_000071.2	CBS
AR	Usher syndrome type ID (AR) NM_022124.5	CDH23
AR	Retinitis pigmentosa 26 (AR) NM_001030311.2	CERKL
AR	CFTR-related disorders (AR) NM_000492.3	CFTR
AR	Congenital myasthenic syndrome (CHRNE-related) (AR) NM_000080.3	CHRNE
AR	Major histocompatibility complex class II deficiency (CIITA-related) (AR) NM_000246.3	CIITA

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AR	CLN3Neuronal ceroid-lipofuscinosis (CLN3-related) (AR) NM_001042432.1	CLN3
AR	Neuronal ceroid-lipofuscinosis (CLN5-related) (AR) NM_006493.2	CLN5
AR	Neuronal ceroid-lipofuscinosis (CLN6-related) (AR) NM_017882.2	CLN6
AR	Neuronal ceroid-lipofuscinosis/Northern epilepsy (CLN8-related) (AR) NM_018941.3	CLN8
AR	Usher syndrome type IIIA (AR) NM_174878.2	CLRN1
AR	Achromatopsia (CNGB3-related) (AR) NM_019098.4	CNGB3
AR	Alport syndrome (COL4A3-related) (AR) NM_000091.4	COL4A3
AR	Alport syndrome (COL4A4-related) (AR) NM_000092.4	COL4A4
XL	Alport syndrome, X-linked (COL4A5-related) (XL) NM_000495.4	COL4A5
AR	Dystrophic epidermolysis bullosa (AR) NM_000094.3	COL7A1
AR	Carbamoylphosphate synthetase I deficiency (AR) NM_001875.4	CPS1
AR	Carnitine palmitoyltransferase I deficiency (AR) NM_001876.3	CPT1A
AR	Carnitine palmitoyltransferase II deficiency (AR) NM_000098.2	CPT2
AR	Leber congenital amaurosis 8/CRB1-related disorders (AR) NM_201253.2	CRB1
AR	Cystinosis (AR) NM_004937.2	CTNS
AR	Pycnodysostosis (AR) NM_000396.3	CTSK
AR	Chronic granulomatous disease (CYBA-related) (AR) NM_000101.3	CYBA
XL	Chronic granulomatous disease (CYBB-related) (XL) NM_000397.3	CYBB
AR	CYP17A1-related conditions (AR) NM_000102.3	CYP17A1
AR	Aromatase deficiency (AR) NM_031226.2	CYP19A1
AR	Cerebrotendinous xanthomatosis (AR) NM_000784.3	CYP27A1
AR	Maple syrup urine disease type 2 (AR) NM_001918.3	DBT
AR	Smith-Lemli-Opitz syndrome (AR) NM_001360.2	DHCR7
AR	DHDDS-related disorders (AR) NM_024887.3	DHDDS
AR	Dihydroipoamide dehydrogenase deficiency (AR) NM_000108.4	DLD
AR	Primary ciliary dyskinesia (DNAH5-related)(AR) NM_001369.2	DNAH5
XL	DMD-related dystrophinopathy (XL) NM_004006.2	DMD
AR	Primary ciliary dyskinesia (DNAI1-related) (AR) NM_012144.3	DNAI1
AR	Primary ciliary dyskinesia (DNAI2-related) (AR) NM_023036.4	DNAI2
AR	Dysferlinopathy (AR) NM_003494.3	DYSF
XL	Hypohidrotic ectodermal dysplasia (EDA-related) (XL) NM_001399.4	EDA
AR	Leukoencephalopathy with vanishing white matter (EIF2B5-related)(AR) NM_003907.2	EIF2B5
AR	Familial dysautonomia (AR) NM_003640.3	ELP1
XL	Emery-Dreifuss muscular dystrophy (EMD-related) (XL) NM_000117.2	EMD
AR	Cockayne syndrome type A (AR) NM_000082.3	ERCC8
AR	Roberts syndrome (AR) NM_001017420.2	ESCO2
AR	Glutaric acidemia type IIA (AR) NM_000126.3	ETFA
AR	Glutaric acidemia type IIC (AR) NM_004453.3	ETFDH
AR	Ethylmalonic encephalopathy (AR) NM_014297.3	ETHE1
AR	Ellis-Van Creveld syndrome (EVC-related) (AR) NM_153717.2	EVC
AR	Ellis-Van Creveld syndrome (EVC2-related)(AR) NM_147127.4	EVC2
AR	Factor XI deficiency (hemophilia C) (AR) NM_000128.3	F11
XL	Factor IX deficiency (hemophilia B) (XL) NM_000133.3	F9
AR	Tyrosinemia type I (AR) NM_000137.2	FAH
AR	Retinitis pigmentosa 28 (AR) NM_001201543.1	FAM161A
AR	Fanconi anemia type A (AR) NM_000135.2	FANCA
AR	Fanconi anemia type C (AR) NM_000136.2	FANCC
AR	Fanconi anemia type G (AR) NM_004629.1	FANCG

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AR	Fumarate hydratase deficiency (AR) NM_000143.3	FH
AR	FKRP-related disorders (AR) NM_024301.4	FKRP
AR	FKTN-related disorders (AR) NM_001079802.1	FKTN
AR	Glycogen storage disease type Ia (AR) NM_000151.3	G6PC1
XL	Glucose-6-phosphate dehydrogenase deficiency (XL) NM_001042351.2	G6PD
AR	Glycogen storage disease type II (Pompe disease) (AR) NM_000152.3	GAA
AR	Krabbe disease (AR) NM_000153.3	GALC
AR	Galactokinase deficiency galactosemia (AR) NM_000154.1	GALK1
AR	Galactosemia (GALT-related) (AR) NM_000155.3	GALT
AR	Guanidinoacetate methyltransferase deficiency (AR) NM_000156.5	GAMT
AR	Glycogen storage disease type IV/adult polyglucosan body disease (AR) NM_000158.3	GBE1
AR	Glutaric acidemia type I (AR) NM_000159.3	GCDH
AR	Combined oxidative phosphorylation deficiency (GFM1-related) (AR) NM_024996.5	GFM1
XL	Charcot-Marie-Tooth disease, X-linked (GJB1-related) (XL) NM_000166.5	GJB1
AR	GJB2-related DFNB1 nonsyndromic hearing loss and deafness (AR) NM_004004.5	GJB2
XL	Fabry disease (XL) NM_000169.2	GLA
AR	Mucopolysaccharidosis type IVB (Morquio B syndrome)/GM1 gangliosidosis (AR) NM_000404.2	GLB1
AR	Glycine encephalopathy (GLDC-related) (AR) NM_000170.2	GLDC
AR	GLE1-related disorders (AR) NM_001003722.1	GLE1
AR	Inclusion body myopathy 2 (AR) NM_001128227.2	GNE
AR	Mucopolipidosis type II/III (GNPTAB-related) (AR) NM_024312.4	GNPTAB
AR	Mucopolipidosis type III (GNPTG-related) (AR) NM_032520.4	GNPTG
AR	Mucopolysaccharidosis type IIID (Sanfilippo D syndrome) (AR) NM_002076.3	GNS
AR	Bernard-Soulier syndrome (GP1BA-related) (AR) NM_000173.6	GP1BA
AR	Bernard-Soulier syndrome (GP9-related) (AR) NM_000174.4	GP9
AR	Primary hyperoxaluria type 2 (AR) NM_012203.1	GRHPR
AR	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (AR) NM_000182.4	HADHA
AR	Severe congenital neutropenia type 3 (AR) NM_006118.3	HAX1
AR	HBB-related hemoglobinopathies (AR) NM_000518.4	HBB
AR	Tay-Sachs disease/hexosaminidase A deficiency (AR) NM_000520.4	HEXA
AR	Sandhoff disease (AR) NM_000521.3	HEXB
AR	Hereditary hemochromatosis type 1(AR) NM_000410.3	HFE
AR	Alkaptonuria (AR) NM_000187.3	HGD
AR	Mucopolysaccharidosis type IIIC (Sanfilippo C syndrome)/retinitis pigmentosa (AR) NM_152419.2	HGSNAT
AR	Hereditary hemochromatosis type 2 (HJV-related) (AR) NM_213653.3	HJV (HFE)
AR	Holocarboxylase synthetase deficiency (AR) NM_000411.6	HLCS
AR	3-hydroxy-3-methylglutarayl-CoA (HMG-CoA) lyase deficiency (AR) NM_000191.2	HMGCL
AR	Primary hyperoxaluria type 3 (AR) NM_138413.3	HOGA1
AR	Hermansky-Pudlak syndrome type 3 (AR) NM_032383.4	HPS3
AR	HSD17B4-related disorders (AR) NM_000414.3	HSD17B4
AR	Congenital adrenal hyperplasia due to 3-beta	HSD3B2
AR	Mucopolysaccharidosis type IX (AR) NM_153281.1	HYAL1
AR	Hydrolethalus syndrome type 1 (AR) NM_145014.2	HYLS1
AR	Mucopolysaccharidosis type I (AR) NM_000203.4	IDUA
XL	X-linked severe combined immunodeficiency (XL) NM_000206.2	IL2RG
AR	Isovaleric acidemia (AR) NM_002225.3	IVD
AR	KCNJ11-related disorders (AR) NM_000525.3	KCNJ11
AR	LAMA2-related muscular dystrophy (AR) NM_000426.3	LAMA2

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AR	LAMA3-related disorders (AR) NM_000227.4	LAMA3
AR	Junctional epidermolysis bullosa (LAMB3-related) (AR) NM_000228.2	LAMB3
AR	Junctional epidermolysis bullosa (LAMC2-related) (AR) NM_005562.2	LAMC2
AR	Leber congenital amaurosis 5 (AR) NM_181714.3	LCA5
AR	Familial hypercholesterolemia (LDLRAP1-related) (AR) NM_015627.2	LDLRAP1
AR	Combined pituitary hormone deficiency (LHX3- related) (AR) NM_014564.4	LHX3
AR	Stüve-Wiedemann syndrome (AR) NM_002310.5	LIFR
AR	Lysosomal acid lipase deficiency (AR) NM_000235.3	LIPA
AR	Autosomal recessive deafness 77 (AR) NM_144612.6	LOXHD1
AR	Lipoprotein lipase deficiency (AR) NM_000237.2	LPL
AR	Leigh syndrome, French Canadian type (AR) NM_133259.3	LRPPRC
AR	Alpha-mannosidosis (AR) NM_000528.3	MAN2B1
AR	3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC1-related) (AR) NM_020166.4	MCCC1
AR	3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC2-related) (AR) NM_022132.4	MCCC2
AR	Mucopolidosis type IV (AR) NM_020533.2	MCOLN1
AR	Postnatal progressive microcephaly with seizures and brain atrophy/infantile cerebral and cerebellar atrophy (AR) NM_004268.4	MED17
AR	Familial Mediterranean fever (AR) NM_000243.2	MEFV
AR	Spondylothoracic dysostosis (AR) NM_001039958.1	MESP2
AR	Neuronal ceroid-lipofuscinosis (MFSD8-related) (AR) NM_152778.2	MFSD8
AR	MKS1-related disorders (AR) NM_017777.3	MKS1
AR	Megalencephalic leukoencephalopathy with subcortical cysts type 1 (AR) NM_015166.3	MLC1
AR	Methylmalonic acidemia (MMAA-related) (AR) NM_172250.2	MMAA
AR	Methylmalonic acidemia (MMAB-related) (AR) NM_052845.3	MMAB
AR	Methylmalonic acidemia with homocystinuria, cobalamin C type (AR) NM_015506.2	MMACHC
AR	Methylmalonic acidemia with homocystinuria, cobalamin D type (AR) NM_015702.2	MMADHC
AR	Congenital disorder of glycosylation (MPI-related) (AR) NM_002435.2	MPI
AR	Congenital amegakaryocytic thrombocytopenia (AR) NM_005373.2	MPL
AR	Mitochondrial DNA depletion syndrome (MPV17- related) (AR) NM_002437.4	MPV17
AR	Homocystinuria due to MTHFR deficiency (AR) NM_005957.4	MTHFR
XL	X-linked myotubular myopathy (XL) NM_000252.2	MTM1
AR	Homocystinuria, cobalamin E type (AR) NM_002454.2	MTRR
AR	Abetalipoproteinemia (AR) NM_000253.3	MTTP
AR	Methylmalonic acidemia (MUT-related) (AR) NM_000255.3	MMUT
AR	Usher syndrome type IB/MYO7A-related disorders (AR) NM_000260.3	MYO7A
AR	Mucopolysaccharidosis type IIIB (Sanfilippo B syndrome) (AR) NM_000263.3	NAGLU
AR	-acetylglutamate synthase deficiency (AR) NM_153006.2	NAGS
AR	Nijmegen breakage syndrome (AR) NM_002485.4	NBN
AR	Charcot-Marie-Tooth disease, type 4D (AR)	NDRG1
AR	Mitochondrial complex I deficiency/Leigh syndrome (NDUFAF5-related) (AR) NM_024120.4	NDUFAF5
AR	Mitochondrial complex I deficiency/Leigh syndrome (NDUFS6-related) (AR) NM_004553.4	NDUFS6
AR	Niemann-Pick disease type C (NPC1-related) (AR) NM_000271.4	NPC1
AR	Niemann-Pick disease type C (NPC2-related)(AR) NM_006432.3	NPC2
AR	Nephrotic syndrome/congenital Finnish nephrosis (NPHS1-related) (AR) NM_004646.3	NPHS1
AR	Nephrotic syndrome/steroid-resistant nephrotic syndrome (NPHS2-related) (AR) NM_014625.3	NPHS2
AR	Enhanced S-cone syndrome/retinitis pigmentosa (AR) NM_014249.3	NR2E3
AR	Congenital insensitivity to pain with anhidrosis (AR) NM_001012331.1	NTRK1
AR	Ornithine aminotransferase deficiency (AR) NM_000274.3	OAT
AR	OPA3-related conditions (AR) NM_025136.3	OPA3

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XL	Ornithine transcarbamylase deficiency (XL) NM_000531.5	OTC
AR	Phenylalanine hydroxylase deficiency (AR) NM_000277.1	PAH
AR	Pyruvate carboxylase deficiency (AR) NM_000920.3	PC
AR	Propionic acidemia (PCCA-related) (AR) NM_000282.3	PCCA
AR	Usher syndrome type IF/PCDH15-related disorders (AR) NM_033056.3	PCDH15
XL	Pyruvate dehydrogenase complex deficiency (PDHA1-related) (XL) NM_000284.3	PDHA1
AR	Pyruvate dehydrogenase complex deficiency (PDHB)	PDHB
AR	Zellweger spectrum disorder (PEX1-related) (AR) NM_000466.2	PEX1
AR	Zellweger spectrum disorder (PEX10-related) (AR) NM_153818.1	PEX10
AR	Zellweger spectrum disorder (PEX12-related) (AR) NM_000286.2	PEX12
AR	Zellweger spectrum disorder (PEX2-related) (AR) NM_000318.2	PEX2
AR	Zellweger spectrum disorder (PEX6-related) (AR) NM_000287.3	PEX6
AR	Rhizomelic chondrodysplasia punctata type 1/Refsum disease (PEX7-related) (AR) NM_000288.3	PEX7
AR	Glycogen storage disease type VII (AR) NM_000289.5	PFKM
AR	Phosphoglycerate dehydrogenase deficiency/Neu Laxova syndrome type 1 (AR) NM_006623.3	PHGDH
AR	Polycystic kidney disease (PKHD1-related) (AR) NM_138694.3	PKHD1
AR	Congenital disorder of glycosylation (PMM2-related) (AR) NM_000303.2	PMM2
AR	POMGNT1-related disorders (AR) NM_017739.3	POMGNT1
AR	Neuronal ceroid-lipofuscinosis (PPT1-related) (AR) NM_000310.3	PPT1
AR	Combined pituitary hormone deficiency (PROP1-related)(AR) NM_006261.4	PROP1
AR	PSAP-related disorders (AR) NM_002778.3	PSAP
AR	Tetrahydrobiopterin deficiency (PTS-related) (AR) NM_000317.2	PTS
AR	Mitochondrial myopathy and sideroblastic anemia 1 (AR) NM_025215.5	PUS1
AR	Glycogen storage disease type V (AR) NM_005609.3	PYGM
AR	Carpenter syndrome (RAB23-related) (AR) NM_183227.2	RAB23
AR	Severe combined immunodeficiency (RAG2-related) (AR) NM_000536.3	RAG2
AR	RAPSN-related disorders (AR) NM_005055.4	RAPSN
AR	Pontocerebellar hypoplasia (RARS2-related) (AR) NM_020320.3	RARS2
AR	Leber congenital amaurosis 13 (AR) NM_152443.2	RDH12
AR	Cartilage-hair hypoplasia – anauxetic dysplasia spectrum disorders (AR) NR_003051.3	RMRP(NME1)
AR	RPE65-related disorders (AR) NM_000329.2	RPE65
XL	X-linked juvenile retinoschisis (XL) NM_000330.3	RS1
AR	Autosomal recessive spastic ataxia of Charlevoix Saguenay (ARSACS) (AR) NM_014363.5	SACS
AR	Aicardi-Goutières syndrome (SAMHD1-related) (AR) NM_015474.3	SAMHD1
AR	Pontocerebellar hypoplasia (SEPSECS-related) (AR) NM_016955.3	SEPSECS
AR	Alpha-1 antitrypsin deficiency (AR) NM_000295.4	SERPINA1
AR	Limb-girdle muscular dystrophy type 2D (AR) NM_000023.2	SGCA
AR	Limb-girdle muscular dystrophy type 2E (AR) NM_000232.4	SGCB
AR	Limb-girdle muscular dystrophy type 2C (AR) NM_000231.2	SGCG
AR	Mucopolysaccharidosis type IIIA (Sanfilippo A syndrome) (AR) NM_000199.3	SGSH
AR	Gitelman syndrome (SLC12A3-related) (AR) NM_000339.2	SLC12A3
AR	Andermann syndrome (AR) NM_133647.1	SLC12A6
AR	Sialic acid storage disorders (AR) NM_012434.4	SLC17A5
AR	Primary carnitine deficiency (AR) NM_003060.3	SLC22A5
AR	Citrin deficiency (AR) NM_014251.2	SLC25A13
AR	SLC26A2-related disorders (AR) NM_000112.3	SLC26A2
AR	Pendred syndrome (AR) NM_000441.1	SLC26A4
AR	Glycogen storage disease type Ib (AR) NM_001164277.1	SLC37A4

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AR	Acrodermatitis enteropathica (AR) NM_130849.3	SLC39A4
AR	Corneal dystrophy and perceptive deafness (AR) NM_032034.3	SLC4A11
AR	Lysinuric protein intolerance (AR) NM_001126106.2	SLC7A7
AR	Schimke immuno-osseous dysplasia (AR) NM_014140.3	SMARCAL1
AR	Niemann-Pick disease type A/B (AR) NM_000543.4	SMPD1
AR	Lipoid congenital adrenal hyperplasia (AR) NM_000349.2	STAR
AR	Multiple sulfatase deficiency (AR) NM_182760.3	SUMF1
AR	Tyrosinemia type II (AR) NM_000353.2	TAT
AR	Osteopetrosis (TCIRG1-related) (AR) NM_006019.3	TCIRG1
AR	Hereditary hemochromatosis type 3 (AR) NM_003227.3	TFR2
AR	Congenital ichthyosis (TGM1-related) (AR) NM_000359.2	TGM1
AR	Tyrosine hydroxylase deficiency (AR) NM_199292.	TH
AR	Joubert syndrome 2/TMEM216-related disorders (AR) NM_001173990.2	TMEM216
AR	Neuronal ceroid-lipofuscinosis (TPP1-related) (AR) NM_000391.3	TPP1
AR	Transient infantile liver failure (AR) NM_018006.4	TRMU
AR	Ataxia with vitamin E deficiency (AR) NM_000370.3	TTPA
AR	Mitochondrial neurogastrointestinal encephalopathy disease (AR) NM_001953.4	TYMP
AR	Usher syndrome type IC/USH1C-related disorders (AR) NM_005709.3	USH1C
AR	Usher syndrome type IIA/USH2A-related disorders (AR) NM_206933.2	USH2A
AR	Chorea-acanthocytosis (AR) NM_033305.2	VPS13A
AR	Cohen syndrome (AR) NM_017890.4	VPS13B
AR	VRK1-related disorders (AR) NM_003384.2	VRK1
AR	Microphthalmia/clinical anophthalmia (VSX2- related) (AR) NM_182894.2	VSX2
AR	WNT10A-related disorders (AR) NM_025216.2	WNT10A
AR	Xeroderma pigmentosum complementation group A (AR) NM_000380.3	XPA
AR	Xeroderma pigmentosum complementation group C (AR) NM_004628.4	XPC
AR	Spastic paraplegia type 15 (AR) NM_015346.3	ZFYVE26