



PGPC-0005	Epilepsy, myoclonic juvenile; Epilepsy,
PGPC-0005	Leukoencephalopathy, cystic, without
PGPC-0005	Shwachman-Diamond syndrome; Apl
PGPC-0005	Pendred syndrome; Deafness, autosol
PGPC-0005	Short -rib thoracic dysplasia 11 with o
PGPC-0005	Hyperoxaluria, primary, type III
PGPC-0005	Myoclonic dystonia
PGPC-0005	Leber congenital amaurosis 10; Mecke
PGPC-0005	Niemann-pick disease, type C2
PGPC-0005	Amyotrophic lateral sclerosis 5, juveni
PGPC-0005	Progressive external ophthalmoplegia
PGPC-0005	Tuberous sclerosis 2; Lymphangioleior
PGPC-0005	Familial juvenile hyperuricemic nephri
PGPC-0005	Dyskeratosis congenita, autosomal do
PGPC-0005	Macular dystrophy, corneal, 1; Macula
PGPC-0005	Ichthyosiform erythroderma, congenit
PGPC-0005	Cardiomyopathy, dilated, 1N; Cardiorr
PGPC-0005	Breast-ovarian cancer, familial, suscep
PGPC-0005	Paramyotonia congenita; Hyperkalem
PGPC-0005	Hemophagocytic lymphohistiocytosis,
PGPC-0005	Camurati-Engelmann disease
PGPC-0005	Amyotrophic lateral sclerosis 8; Spinal
PGPC-0005	Epiphyseal dysplasia, multiple, 3
PGPC-0005	Li-Fraumeni syndrome 2
PGPC-0005	Glucose/galactose malabsorption
PGPC-0005	Acyl-CoA dehydrogenase family, mem
PGPC-0006	Nephronophthisis 4; Senior-Loken syn
PGPC-0006	Amyotrophic lateral sclerosis 10, with
PGPC-0006	Homocystinuria due to MTHFR deficie
PGPC-0006	Cone-rod dystrophy 3; Retinitis pigme
PGPC-0006	Coenzyme Q10 deficiency; Progressiv
PGPC-0006	Ehlers-Danlos syndrome, type IV
PGPC-0006	Ichthyosis, harlequin; Ichthyosis, lame
PGPC-0006	Hemolytic uremic syndrome, atypical;
PGPC-0006	Growth hormone insensitivity syndror
PGPC-0006	Glucocorticoid resistance
PGPC-0006	Leukoencephalopathy, cystic, without
PGPC-0006	Ciliary dyskinesia, primary, 6
PGPC-0006	Exostoses, multiple, type 1
PGPC-0006	Refsum disease
PGPC-0006	Niemann-Pick disease, type A; Niemar
PGPC-0006	T cell-negative, B cell-negative, natura
PGPC-0006	Vitreoretinopathy, neovascular inflam
PGPC-0006	Deafness, autosomal recessive 2; Ush

PGPC-0006	Hydroxymethylbilane synthase deficie
PGPC-0006	Retinal cone dystrophy 4
PGPC-0006	Lethal congenital contracture syndron
PGPC-0006	Deafness, autosomal recessive 1A; De
PGPC-0006	Spastic ataxia, Charlevoix-Saguenay ty
PGPC-0006	Breast-ovarian cancer, familial, suscep
PGPC-0006	Bile acid malabsorption, primary
PGPC-0006	Hyperphenylalaninemia, BH4-deficien
PGPC-0006	Glaucoma 3, primary congenital, D; M
PGPC-0006	Albinism, oculocutaneous, type II; Alb
PGPC-0006	Progressive external ophthalmoplegia
PGPC-0006	Surfactant metabolism dysfunction, p
PGPC-0006	Familial Mediterranean fever
PGPC-0006	Familial juvenile hyperuricemic nephro
PGPC-0006	Leber congenital amaurosis 4; Retiniti
PGPC-0006	Ichthyosiform erythroderma, congenit
PGPC-0006	Frontotemporal lobar degeneration w
PGPC-0006	Hemophagocytic lymphohistiocytosis,
PGPC-0006	Liopdystrophy, partial, acquired; Epile
PGPC-0006	Hypercholesterolemia, familial
PGPC-0006	Anemia, dyserythropoietic congenital,
PGPC-0006	Homocystinuria due to cystathionine l
PGPC-0006	Klippel-Feil syndrome 4, autosomal re
PGPC-0006	Immunodysregulation, polyendocrino
PGPC-0050	Microcephaly, primary autosomal rec
PGPC-0050	Congenital disorder of glycosylation, t
PGPC-0050	Ventricular tachycardia, catecholamin
PGPC-0050	Miyoshi muscular dystrophy 1; Muscu
PGPC-0050	Paroxysmal extreme pain disorder
PGPC-0050	Hyperoxaluria, primary, type 1
PGPC-0050	Schizophrenia
PGPC-0050	Infantile liver failure syndrome 1
PGPC-0050	Hemochromatosis
PGPC-0050	Polycystic kidney disease, autosomal r
PGPC-0050	Parkinson disease 2, autosomal recess
PGPC-0050	Platelet glycoprotein IV deficiency
PGPC-0050	Pendred syndrome; Deafness, autoso
PGPC-0050	Cortical dysplasia-focal epilepsy syndr
PGPC-0050	Exostoses, multiple, type 1
PGPC-0050	Candidiasis, familial, 2
PGPC-0050	Refsum disease
PGPC-0050	Refsum disease
PGPC-0050	Multiple endocrine neoplasia type I; H
PGPC-0050	Tyrosinemia, type III; Hawksinuria

PGPC-0050	Cardiomyopathy, dilated 1U
PGPC-0050	ACTH-independent macronodular adrenocortical dysplasia
PGPC-0050	ACTH-independent macronodular adrenocortical dysplasia
PGPC-0050	Increased analgesia from kappa-opioid receptor
PGPC-0050	Deafness, autosomal recessive 3
PGPC-0050	Hemophagocytic lymphohistiocytosis, familial
PGPC-0050	Methylmalonic aciduria due to transcobalamin II deficiency
PGPC-0050	Hypercholesterolemia, familial
PGPC-0050	Palmoplantar keratoderma and woolly hair
PGPC-0050	Cerebral arteriopathy with subcortical white matter changes
PGPC-0050	Muscular dystrophy-dystroglycanopathy

<b>Gene ID (transcript)</b>	<b>Genomic position</b>
CASQ1:NM_001231	chr1:160160801T>C
NCF2:NM_000433	chr1:183532364T>A
USH2A:NM_206933	chr1:216496932C>G
APOB:NM_000384	chr2:21231278G>A
BMPR2:NM_001204	chr2:203420712G>A
FASTKD2:NM_014929	chr2:207631566A>G
DES:NM_001927	chr2:220284876C>T
CC2D2A:NM_001080522	chr4:15589552delG
KIT:NM_000222	chr4:55524252G>A
SPRY4:NM_001293289	chr5:141694021G>T
NR3C1:NM_001024094	chr5:142780337delCTCinsTTT
PKHD1:NM_170724	chr6:51637536G>T
DST:NM_001723	chr6:56485496C>G
SLC26A4:NM_000441	chr7:107355874C>T
CNGB3:NM_019098	chr8:87645092C>T
VPS13B:NM_017890	chr8:100887731delCCAGCTGTTCTinsC
CDH23:NM_022124	chr10:73377112G>A
FANCF:NM_022725	chr11:22646532C>T
EDNRB:NM_001122659	chr13:78477665G>A
CLN6:NM_017882	chr15:68504005G>A
HEXA:NM_000520	chr15:72638921insGATA
ABCA3:NM_001089	chr16:2367764T>A
SRCAP:NM_006662	chr16:30740333A>G
B9D1:NM_001243475	chr17:19241062C>G
MPO:NM_000250	chr17:56355397G>A
TYK2:NM_003331	chr19:10463118G>C
GYS1:NM_002103	chr19:49472545T>A
CHEK2:NM_145862	chr22:29121087A>G
ABCB7:NM_001271696	chrX:74334588C>T
GBA:NM_001171811	chr1:155205634T>C
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IFIH1:NM_022168	chr2:163124637T>C
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PKD2:NM_000297	chr4:88989089A>C
TERT:NM_198253	chr5:1293767G>A
ADGRV1:NM_032119	chr5:89979568G>A
DSP:NM_001008844	chr6:7565727A>T
DTNBP1:NM_183040	chr6:15524715G>A
NHLRC1:NM_198586	chr6:18122402C>T
TNXB:NM_019105	chr6:32052313C>T

EFHC1:NM_018100	chr6:52317597T>C
RNASET2:NM_003730	chr6:167343204C>T
SBDS:NM_016038	chr7:66453476A>G
SLC26A4:NM_000441	chr7:107341628T>C
WDR34:NM_052844	chr9:131418941G>A
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DRD2:NM_016574	chr11:113283488G>A
CEP290:NM_025114	chr12:88523494C>G
NPC2:NM_006432	chr14:74953134C>T
SPG11:NM_001160227	chr15:44918690C>T
POLG:NM_002693	chr15:89861826T>C
TSC2:NM_001318829	chr16:2110795G>A
UMOD:NM_003361	chr16:20352618C>A
ACD:NM_022914	chr16:67694606A>G
CHST6:NM_021615	chr16:75512734C>A
ALOX12B:NM_001139	chr17:7984479G>A
TCAP:NM_003673	chr17:37822316G>A
BRCA1:NM_007294	chr17:41222975C>T
SCN4A:NM_000334	chr17:62028920C>G
UNC13D:NM_199242	chr17:73839326C>T
TGFB1:NM_000660	chr19:41847860G>A
VAPB:NM_004738	chr20:57014075T>G
COL9A3:NM_001853	chr20:61472082T>C
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ACAD9:NM_014049	chr3:128622922G>A
NPHP4:NM_001291594	chr1:5940243G>A
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CLCN6:NM_001256959	chr1:11866183C>T
ABCA4:NM_000350	chr1:94467548C>G
ADCK3:NM_020247	chr1:227174240delCAC
COL3A1:NM_000090	chr2:189863424C>A
ABCA12:NM_015657	chr2:215876355G>A
CFI:NM_001318057	chr4:110667485T>C
GHR:NM_001242400	chr5:42711376G>A
NR3C1:NM_001024094	chr5:142780337delCTCinsTTT
RNASET2:NM_003730	chr6:167343204C>T
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EXT1:NM_000127	chr8:119124443C>G
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RAG1:NM_000448	chr11:36596200G>A
CAPN5:NM_004055	chr11:76826581delC
MYO7A:NM_000260	chr11:76903189G>A

HMBS:NM_001258209	chr11:118959973A>T
CACNA2D4:NM_172364	chr12:1969369G>A
ADCY6:NM_015270	chr12:49168798C>A
GJB2:NM_004004	chr13:20763554delA
SACS:NM_014363	chr13:23929378G>A
BRCA2:NM_000059	chr13:32910456C>G
SLC10A2:NM_000452	chr13:103701690G>A
GCH1:NM_001024024	chr14:55369314G>A
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OCA2:NM_000275	chr15:28228553C>T
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ABCA3:NM_001089	chr16:2350115G>T
MEFV:NM_000243	chr16:3293310A>G
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UNC13D:NM_199242	chr17:73839326C>T
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ALG6:NM_013339	chr1:63872032T>C
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DYSF:NM_001130979	chr2:71762413G>A
SCN9A:NM_002977	chr2:167141109G>T
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HFE:NM_001300749	chr6:26091185A>T
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PARK2:NM_013987	chr6:162864388C>G
CD36:NM_001127444	chr7:80300431insA
SLC26A4:NM_000441	chr7:107330653G>A
CNTNAP2:NM_014141	chr7:145813093A>G
EXT1:NM_000127	chr8:119124443C>G
CARD9:NM_052813	chr9:139258789delT
PHYH:NM_001323083	chr10:13320339G>A
PHYH:NM_001323083	chr10:13325784C>T
MEN1:NM_130801	chr11:64575505C>T
HPD:NM_002150	chr12:122277904G>C

PSEN1:NM_000021	chr14:73602899C>G
ARMC5:NM_001105247	chr16:31475712C>A
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MC1R:NM_002386	chr16:89986091G>A
MYO15A:NM_016239	chr17:18023066delC
STXBP2:NM_001127396	chr19:7710134C>T
CD320:NM_016579	chr19:8369919delCTC
LDLR:NM_000527	chr19:11231203G>A
KANK2:NM_001329451	chr19:11277236A>G
NOTCH3:NM_000435	chr19:15285063G>T
FKRP:NM_001039885	chr19:47259048C>G









Yes	Yes
Yes	Yes
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Yes	Yes
Yes	Yes
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Yes	Yes
Yes	Yes
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**Notes**

Two SNVs

Two SNVs