Individual	Disease
PGPC-0002	Myopathy, vacuolar, with CASQ1 aggr
PGPC-0002	Chronic granulomatous disease, autos
PGPC-0002	Usher syndrome, type 2A
PGPC-0002	Hypobetalipoproteinemia, familial; Hy
PGPC-0002	Pulmonary hypertension, primary; Pul
PGPC-0002	Mitochondrial complex IV deficiency
PGPC-0002	Myopathy, myofibrillar 1; Cardiomyop
PGPC-0002	Joubert syndrome 9; Meckel syndrom
PGPC-0002	Gastrointestinal stromal tumor
PGPC-0002	Hypogonadotropic hypogonadism 17,
PGPC-0002	Glucocorticoid resistance
PGPC-0002	Polycystic kidney disease, autosomal r
PGPC-0002	Neuropathy, hereditary sensory and a
PGPC-0002	Pendred syndrome; Deafness, autoso
PGPC-0002	Achromatopsia 3; Macular degenerati
PGPC-0002	Cohen syndrome
PGPC-0002	Deafness, autosomal recessive 12; Usl
PGPC-0002	Fanconia anemia, complementation g
PGPC-0002	Waardenburg syndrome type 4A; ABC
PGPC-0002	Ceroid lipofuscinosis, neuronal, 6
PGPC-0002	Tay-Sachs disease; GM2-gangliosidosi
PGPC-0002	Surfactant metabolism dysfunction, p
PGPC-0002	Floating-Harbor syndrome
PGPC-0002	Meckel syndrome 9
PGPC-0002	Myeloperoxidase deficiency
PGPC-0002	Immunodeficiency 35
PGPC-0002	Glycogen storage disease, type 0, mus
PGPC-0002	Li-Fraumeni syndrome 2
PGPC-0002	Anemia, sideroblastic, and spinocereb
PGPC-0005	Gaucher disease
PGPC-0005	Hypobetalipoproteinemia, familial; Hy
PGPC-0005	Hypobetalipoproteinemia, familial; Hy
PGPC-0005	Cystinuria
PGPC-0005	Singleton-Merten syndrome 1
PGPC-0005	Pulmonary hypertension, primary; Pul
PGPC-0005	Glaucoma, normal tension, susceptibi
PGPC-0005	Polycystic kidney disease 2
PGPC-0005	Aplastic anemia; Dyskeratosis congen
PGPC-0005	Usher syndrome, type IIC
PGPC-0005	Arrhythmogenic right ventricular dysp
PGPC-0005	Hermansky-Pudlak syndrome 7
PGPC-0005	Epilepsy, progressive myoclonic 2B (La
PGPC-0005	Vesicoureteral reflux 8; Ehlers-Danlos

PGPC-0005	Epilepsy, myoclonic juvenile; Epilepsy,
PGPC-0005	Leukoencephalopathy, cystic, without
PGPC-0005	Shwachman-Diamond syndrome; Apla
PGPC-0005	Pendred syndrome; Deafness, autoso
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 nephr
PGPC-0005	Dyskeratosis congenita, autosomal do
PGPC-0005	Macular dystrophy, corneal, 1; Macula
PGPC-0005	Ichthyosiform erythroderma, congenit
PGPC-0005	Cardiomyopathy, dilated, 1N; Cardiom
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; Progressive
PGPC-0006	Ehlers-Danlos syndrome, type IV
PGPC-0006	Ichthyosis, harlequin; Ichthyosis, lame
PGPC-0006	Hemolytic uremic syndrome, atypical;
PGPC-0006	Growth hormone insensitivity syndron
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 nephr
PGPC-0006	Leber congenital amaurosis 4; Retiniti
PGPC-0006	Ichthyosiform erythroderma, congeni
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 adr
PGPC-0050	ACTH-independent macronodular adr
PGPC-0050	Increased analgesia from kappa-opioi
PGPC-0050	Deafness, autosomal recessive 3
PGPC-0050	Hemophagocytic lymphohistiocytosis,
PGPC-0050	Methylmalonic aciduria due to transc
PGPC-0050	Hypercholesterolemia, familial
PGPC-0050	Palmoplantar keratoderma and woolly
PGPC-0050	Cerebral arteriopathy with subcortical
PGPC-0050	Muscular dystrophy-dystroglycanopat

Gene ID (transcript)	Genomic position
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NCF2:NM_000433	chr1:183532364T>A
USH2A:NM_206933	chr1:216496932C>G
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BMPR2:NM_001204	chr2:203420712G>A
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KIT:NM_000222	chr4:55524252G>A
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DST:NM_001723	chr6:56485496C>G
SLC26A4:NM_000441	chr7:107355874C>T
CNGB3:NM_019098	chr8:87645092C>T
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Detected in blood?	Detected in non-enriched saliva?
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Detected in enriched saliva?	Detected in non-enriched buccal?
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Detected in enriched buccal?	Detected in every sample type?
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Notes

Two SNVs

Two SNVs