Igenomix®

PGT-M Disease List

3-methylglutaconic aciduria type 1
6-deficiency pyruvyl-tetrahydropterin synthase
ABO isoimmunization
Achondroplasia
Achromatopsia
Aciduria Argininosuccinic
Acyl-CoA dehydrogenase deficiency
Adenosine deaminase deficiency
Adrenoleukodystrophy
Adult syndrome
Aicardi Goutieres Syndrome
Aicardi-Goutières syndrome
Alagille syndrome
Albright syndrome
Alpha thalassemia
Alpha-1 antitrypsin deficiency
Alpha-thalassemia syndrome X-linked intellectual disability
Alport syndrome
Alström syndrome
Amyotrophic Lateral Sclerosis
Amyotrophic lateral sclerosis familiar
Androgen insensitivity syndrome
Aniridia
APECED syndrome
Apparent mineralocorticoid excess
Arterial tortuosity syndrome
Ataxia-oculomotor apraxia type 1
ATIC deficiency
Autism spectrum disorder
Autosomal dominant limb-girdle muscular dystrophy type 1B
Autosomal recessive limb-girdle muscular dystrophy
Autosomal recessive polycystic kidney disease

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Band like calcification polymicrogyria
Bardet-Biedl syndrome
Bartter syndrome
Best Macular dystrophy
Beta Thalassemia
Biotinidase deficiency
Biotin-responsive basal ganglia disease
Blackfan-Diamond disease
Blepharophimosis-epicanthus inversus-ptosis syndrome
Bruck syndrome
Brugada syndrome
Bruton type agammaglobulinemia
CADASIL
Canavan Disease
Carbamoyl-phosphate synthase deficiency
Carbonic anhydrase deficiency type 2
Cartilage-hair hypoplasia
Catecholaminergic polymorphic ventricular tachycar
Caveolinopathy
CDK9 SYNDROME
Central core myopathy
Cerebellar hypoplasia
Charcot Marie Tooh 2A
Charcot Marie Tooh X-linked
Charcot Marie Tooth 1A
Charcot Marie Tooth 1B
Charcot Marie Tooth 2K
Charcot Marie Tooth 4C
Charcot Marie Tooth 4D
Chondrodysplasia punctata
Chondrodysplasia punctata type 1 Rhizomelic
Chondrodysplasia type Grebe
Choroideremia
Chronic granulomatosis

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Citrullinemia
Cleidocranial dysostosis
Clouston syndrome
COFS syndrome
Cohen syndrome
Combined oxidative phosphorylation deficiency
Congenital adrenal hyperplasia
Congenital cataracts
Congenital cerebellar ataxia
congenital heart disease
Congenital ichthyosis
Congenital lamellar ichthyosis
Congenital muscular dystrophy
Congenital muscular dystrophy by merosin Deficit
Congenital muscular dystrophy megaconial
Congenital myasthenic syndromes with glycosylation defect
Congenital nephrotic syndrome
Congenital tufting enteropathy
Corneal dystrophy
Cornelia de Lange syndrome
Creutzfeldt-Jakob disease
Cystic fibrosis
Cystoid macular dystrophy
Cytomegalic congenital adrenal hypoplasia
D,L-2-hydroxyglutaric aciduria
Darier disease
Deficiency of glucose 6-phosphate dehydrogenase
Dehydrated hereditary stomatocytosis
Diastrophic dysplasia
Diffuse gastric cancer
Dihydropyrimidine dehydrogenase deficiency
Dihydropyrimidine dehydrogenase deficiency
Dominant polycystic kidney disease
Duchenne muscular dystrophy / Becker
Duffy isoinmunization
Dyskeratosis congenita, X-linked
Dystrophic epidermolysis bullosa

Ectodermic dysplasia
EIF2B3-related leukodstrophy
Ellis-van Creveld syndrome
Emery-Dreifuss muscular dystrophy
Epileptic encephalopathy
Fabry disease
Facio-scapulo-humeral dystrophy
Factor XIII deficiency
Familial adenomatous polyposis
Familial amyloidotic polyneuropathy
Familial dysautonomia
Familial hyperaldosteronism
Familial hypertrophic cardiomyopathy
Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis
Familial isolated arrhythmogenic right ventricular dysplasia
Familial isolated dilated cardiomyopathy
Familial mediterranian fever
Familial spastic paraplegia
Family thoracic aortic aneurysm
Fanconi Anemia
Fatal familial insomnia
Fatal mutiple mitochondrial dysfunctions syndrome
FGFR1 related disorder
Focal dermal hypoplasia
Fragile X syndrome
Fraser syndrome
Galactosemy
Gangliosidosis
Gaucher's Disease
Geleophysic dysplasia
Gerstmann-Straussler-Scheinker syndrome
Glanzmann's thrombasthenia
Glycine encephalopathy
Glycogen Storage Disease
Glycogenosis due to glucose-6-phosphatase deficiency
GNE-related disorders
Gorlin syndrome
GSS syndrome

Harlequin type ichthyosis congenital
Hemolytic anemia due to red cell pyruvate kinase deficiency
Hemophagocytic lymphohistiocytosis
Hemophilia A
Hemophilia B
Hereditary angioedema
Hereditary breast cancer
Hereditary chronic pancreatitis
Hereditary fructose intolerance
Hereditary hearing loss
Hereditary hemorrhagic telangiectasia
Hereditary multiple exostosis
Hereditary Parkinson
Hereditary spherocytosis
Hermansky-Pudlak syndrome
Hipomielinizante leukodystrophy
HLA typing
Holoprosencephaly
Homocystinuria
Hunters Syndrome
Huntington
Hurler Syndrome
Hyper IgD syndrome
Hyperinsulinism
Hyperkalemic periodic paralysis
Hypochondroplasia
Hypohidrotic ectodermal dysplasia
Hypoparathyroidism-retardation-dysmorphism syndrom
Hypophosphatemic rickets X-linked
Idiopathic dystonia
Incontinentia pigmenti
Infantile liver failure syndrome 1
intestinal atresia
IPEX syndrome
Isolated sulfite oxidase deficiency
isovaleric acidemia
Jeune asphyxiating thoracic dystrophy
Joubert syndrome
Junctional epidermolysis bullosa

Kell isoimmunization
Kennedy disease
Krabbe disease
L1 syndrome
L-aromatic amino acid decarboxylase deficiency
Larsen syndrome
LCHADD
Leber congenital amaurosis
Leigh disease with leukodystrophy
Leigh syndrome
Leigh-like syndrome
Leopard syndrome
Leprechaunism
Leri Weill Dyschondrosteosis
Leucodystrophy
leukoencephalopathy with vanishing white matter
Li-Fraumeni syndrome
Limb girdle muscular dystrophy
Lipofuscinosis
Lissencephaly type 1 (X-chromosome)
Long QT syndrome
Lowe syndrome
Lymphedema-Distichiasis
Lynch syndrome
Macular dystrophy
Marfan syndrome
McArdle disease
Menkes disease
Mental Retardation
Mental retardation syndrome - strabismus
Merkel-Gruber syndrome
Metachromatic leucodystrophy
Methyl malonic acidemia
Microcephaly
Microphthalmia
Microvillus inclusion disease
Mitochondrial complex IV deficiency
Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency
Mitochrondrial DNA depletion syndrome

Molybdenum cofactor deficiency
Morbus-Crouzon syndrome
Mucin-1 kidney disease
Mucolipidosis II
Mucopolysaccharidosis I
Mucopolysaccharidosis II
Mucopolysaccharidosis IIIA
Mucopolysaccharidosis IV-A
Muenke syndrome
Multiple Endocrine Neoplasia 1
Multiple Endocrine Neoplasia 2A
Multiple Endocrine Neoplasia 2B
Multiple intestinal atresia
Myoclonic dystonia
N-acetyl-alpha-D-galactosaminidase deficiency
Nail-patella syndrome
Nanoftalmia
Nemaline myopathy
Neurofibromatosis 1
Neurofibromatosis 2
Niemann Pick A/B syndrome
Niemann Pick C syndrome
Nonketotic hyperglycinemia
Nonspecific X-linked intellectual deficit
Noonan Syndrome
Norrie disease
nuclear cataract
Oculocutaneous albinism
Oculo-dental-digital dysplasia
Ohtahara Syndrome
Ornithine transcarbamylase deficiency
Osteochondrodysplasia
Osteogenesis imperfecta
Osteopetrosis
Oxoglutaricaciduria

Papillo-renal syndrome
Paraganglioma/Pheochromocytoma
Partial STAT1 deficiency
Pelizaeus Merzbacher
Pelizaeus Merzbacher-like
Peraganglioma / pheochromocytoma
Periventricular Heterotopia
Peters plus syndrome
Peutz Jeghers syndrome
Pfeiffer syndrome
Phenylketonuria
Pompe disease
Pressure sensitive neuropathy
Primary ciliary dyskinesia
Progressive external ophthalmoplegia
Progressive familial intrahepatic cholestasis
Propionic acidemia
Proximal myopathy with extrapyramidal signs
Pseudoachondroplasia
Pseudohermaphroditism
Pyridoxal phosphate-responsive seizures
Pyruvate carboxylase deficiency
Rendu-Osler-Weber disease
Retinitis Pigmentosa
Retinoblastoma
RhCE isoimmunization
RhD isoimmunization
Sandhoff disease
Schaaf-Yang syndrome
Schwannomatosis
Sensory ataxic neuropathy - dysarthria - ophthalmo
Severe combined immunodeficiency T-B + X-linked
Shwachman-Diamond syndrome

Shwachman-Diamond syndrome
Sickle cell anemia
Simple epidermolysis bullosa
Simpson-Golabi-Behmel syndrome
Smith Lemli Opitz syndrome
Spinal Muscular Atrophy
Spinocerebellar ataxia
spinocerebellar ataxia type 36
Spondylometaphyseal dysplasia
Stargardt's Disease
Steinert myotonic dystrophy
Steroid resistant nephrotic syndrome
Stickler syndrome type 1
Stuve-Wiedemann syndrome
Synaptic congenital myasthenic syndromes
Syndrome type 2 lethal congenital contracture
Syndrome Van der Knapp
Tay Sachs
Temtamy's syndrome
Thymine-uraciluria
Tibial muscular dystrophy
Treacher Collins syndrome
Tuberous sclerosis
Tyrosinemia type I
Urine Disease smelling maple syrup
Usher syndrome
Van den Ende-Gupta syndrome
Van der Woude syndrome
Variable PenetranceHypertrophic Cardiomyopthy
Vitelliform macular dystrophy
Von Hippel Lindau syndrome
Walker-Warburg syndrome
Warburg micro syndrome
Wiskott-Aldrich syndrome
Wolman syndrome
Woodhouse-Sakati syndrome
X-linked adrenoleukodystrophy
X-linked Agammaglobulinemia
Zellweger syndrome