PGT-M Disease List

BANKER IVF

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

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

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Choroideremia
Chronic granulomatosis
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

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BANKER IVF

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

PGT-M Disease List

BANKER IVF

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

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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