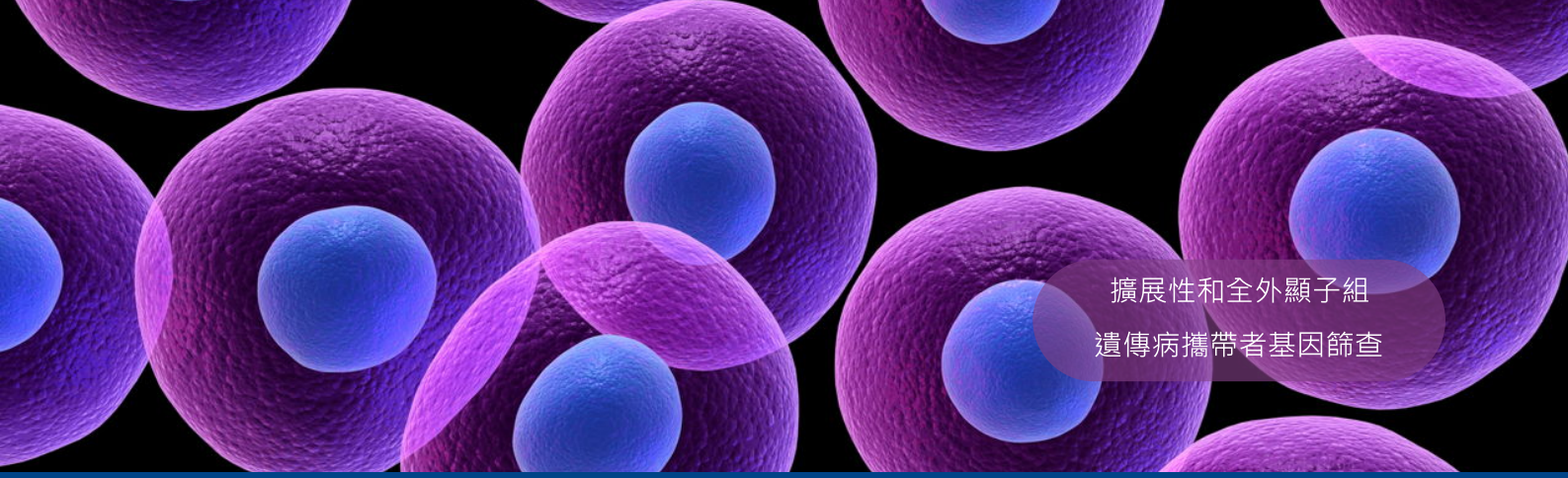

List of Disorders Screened by the Expanded Carrier Test

11-beta-hydroxylase-deficient congenital adrenal hyperplasia

17-alpha-hydroxylase-deficient congenital adrenal hyperplasia

3-beta-hydroxysteroid dehydrogenase type II deficiency (Congenital adrenal hyperplasia)

3-hydroxy-3-methylglutarayl-CoA (HMG-CoA) lyase deficiency

3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC1-related)

3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC2-related)

3-methylglutaconic aciduria type III (Costeff optic atrophy)

Abetalipoproteinemia

ACAD9 deficiency

Achromatopsia (CNGB3-related)

Acrodermatitis enteropathica

Adenosine deaminase deficiency

Aicardi-Goutieres syndrome (SAMHD1-related)

Aldosterone synthase deficiency

Alkaptonuria

Alpha-1 antitrypsin deficiency

Alpha-mannosidosis

Alpha-thalassemia

Alpha-thalassemia X-linked intellectual disability syndrome

Alport Syndrome (COL4A3-related)

Alport Syndrome (COL4A4-related)

Alport Syndrome, X-linked (COL4A5-related)

Alström syndrome

Andermann syndrome

Arginase deficiency

Argininosuccinic aciduria

Aromatase deficiency

Asparagine synthetase deficiency

Aspartylglucosaminuria

Ataxia with vitamin E deficiency

Ataxia-telangiectasia

Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia

Autosomal recessive deafness 77 (DFNB77)

Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)

Bardet-Biedl syndrome (BBS10-related)

Bardet-Biedl syndrome (BBS12-related)

Bardet-Biedl syndrome (BBS1-related)

Bardet-Biedl syndrome (BBS2-related)

Bartter syndrome type IV (BSND-related)

Bernard-Soulier syndrome (GP1BA-related)

Bernard-Soulier syndrome (GP9-related)

Beta-ketothiolase deficiency

Biotinidase deficiency

Bloom syndrome

Canavan disease

Carbamoylphosphate synthetase I deficiency

Carnitine palmitoyltransferase I deficiency

Carnitine palmitoyltransferase II deficiency

Carpenter syndrome (RAB23-related)

Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders

Cerebrotendinous xanthomatosis

Charcot-Marie-Tooth disease (NDRG1-related)

Charcot-Marie-Tooth disease, X-linked (GJB1-related)

Chorea-acanthocytosis

Choroideremia

Chronic granulomatous disease (CYBA-related)

Chronic granulomatous disease (CYBB-related)

Citrin deficiency

Citrullinemia type 1

Cockayne syndrome type A

Cockayne syndrome type B

Cohen syndrome

Combined malonic and methylmalonic aciduria (ACSF3-related)

Combined oxidative phosphorylation deficiency (GFM1-related)

Combined oxidative phosphorylation deficiency (TSFM-related)

Combined pituitary hormone deficiency (LHX3-related)

Combined pituitary hormone deficiency (PROP1-related)

Combined SAP Deficiency

Congenital amegakaryocytic thrombocytopenia

Congenital disorder of glycosylation (ALG6-related)

Congenital disorder of glycosylation (MPI-related)

Congenital disorder of glycosylation (PMM2-related)

Congenital ichthyosis (TGM1-related)

Congenital insensitivity to pain with anhidrosis

Congenital myasthenic syndrome (CHRNE-related)

Congenital myasthenic syndrome (RAPSN-related)

Congenital neutropenia (HAX1-related)

Corneal dystrophy and perceptive deafness

Cystic fibrosis/ CFTR-related disorders

Cystinosis

D-bifunctional protein deficiency

DHDDS-related disorders (including Congenital disorder of glycoslylation/ Retinitis pigmentosa 59)

Dihydrolipoamide dehydrogenase deficiency (DLD)

DMD-related dystrophinopathy (Including Duchenne/Becker muscular dystrophy and Dilated cardiomyopathy)

Dystrophic epidermolysis bullosa (COL7A1-related)

Ehlers-Danlos syndrome, dermatosparaxis type

Ellis-van Creveld syndrome (EVC2-related)

Ellis-van Creveld syndrome (EVC-related)

Emery-Dreifuss muscular dystrophy (EMD-related)

Enhanced S-cone syndrome/ Retinitis pigmentosa 37

Ethylmalonic encephalopathy

Fabry disease

Factor IX deficiency (Hemophilia B)

Factor V Leiden thrombophilia

Factor XI deficiency (Hemophilia C)

Familial dysautonomia

Familial hypercholesterolemia (LDLRAP1-related)

Familial hypercholesterolemia (LDLR-related)

Familial hyperinsulinism (ABCC8-related)

Familial hyperinsulinism (KCNJ11-related)

Familial mediterranean fever

Fanconi anemia type A

Fanconi anemia type C

Fanconi anemia type G

Fragile X syndrome

Fumarate hydratase deficiency

Galactokinase deficiency galactosemia

Galactosemia (GALT-related)

Gaucher disease

Gitelman syndrome (SLC12A3-related)

GJB2-related DFNB1 nonsyndromic hearing loss and deafness

Glucose-6-phosphate dehydrogenase (G6PD) deficiency

Glutaric acidemia type I

Glutaric acidemia type II (ETFA-related)

Glutaric acidemia type II (ETFDH-related)

Glycine encephalopathy (AMT-related)

Glycine encephalopathy (GLDC-related)

Glycogen storage disease type Ia

Glycogen storage disease type Ib

Glycogen storage disease type II (Pompe disease)

Glycogen storage disease type IV/ Adult polyglucosan body disease

Glycogen storage disease type V

Glycogen storage disease type III

Glycogen storage disease type VII

GRACILE syndrome/ BCS1L-related disorders (including Mitochondrial complex III deficiency, Bjornstad syndrome, Leigh syndrome)

Guanidinoacetate methyltransferase deficiency

HBB-related hemoglobinopathies (including Beta-thalassemia and Sickle cell disease)

Hereditary fructose intolerance

Hereditary hemochromatosis (HFE-related)

Hereditary hemochromatosis (HJV-related)

Hereditary hemochromatosis (TFR2-related)

Hermansky-Pudlak syndrome (HPS1-related)

Hermansky-Pudlak syndrome (HPS3-related)

Holocarboxylase synthetase deficiency

Homocystinuria (CBS-related)

Homocystinuria due to MTHFR deficiency

Homocystinuria, cobalamin E type

Hydrolethalus syndrome type 1

Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome

Hypohidrotic ectodermal dysplasia (EDA-related)

Hypophosphatasia

Inclusion body myopathy 2

Isovaleric acidemia

Joubert syndrome 2/ TMEM216-related disorders

Junctional epidermolysis bullosa (LAMA3-related)

Junctional epidermolysis bullosa (LAMB3-related)

Junctional epidermolysis bullosa (LAMC2-related)

Krabbe disease

LAMA2-related muscular dystrophy

Leber congenital amaurosis 10/ CEP290-related disorders

Leber congenital amaurosis 13

Leber congenital amaurosis 2

Leber congenital amaurosis 5

Leber congenital amaurosis 8/ CRB1-related disorders

Leigh syndrome, French Canadian type

Lethal congenital contracture syndrome 1 / Lethal arthrogryposis with anterior horn cell disease

Leukoencephalopathy with vanishing white matter (EIF2B5-related)

Limb-girdle muscular dystrophy type 2A (calpainopathy)

Limb-girdle muscular dystrophy type 2B (dysferlinopathy)

Limb-girdle muscular dystrophy type 2C

Limb-girdle muscular dystrophy type 2D

Limb-girdle muscular dystrophy type 2E

Lipoid congenital adrenal hyperplasia (STAR-related)

Lipoprotein lipase deficiency

Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency

Lysinuric protein intolerance

Lysosomal acid lipase deficiency (includes Wolman disease and Cholesterol ester storage disease)

Major histocompatibility complex class II deficiency (CIITA-related)

Maple syrup urine disease (MSUD) type 1A

Maple syrup urine disease (MSUD) type 1B

Maple syrup urine disease (MSUD) type 2

Medium chain acyl-CoA dehydrogenase (MCAD) deficiency

Megalencephalic leukoencephalopathy with subcortical cysts type 1

Menkes disease/ ATP7A-related disorders (including Occipital horn syndrome and Distal hereditary motor neuropathy)

Metachromatic leukodystrophy (ARSA-related)

Methylmalonic acidemia (MMAA-related)

Methylmalonic acidemia (MMAB-related)

Methylmalonic acidemia (MUT-related)

Methylmalonic acidemia with homocystinuria, cobalamin C type

Methylmalonic acidemia with homocystinuria, cobalamin D type

Microphthalmia / clinical anophthalmia (VSX2-related)

Mitochondrial complex I deficiency/ Leigh syndrome (NDUFAF5-related)

Mitochondrial complex I deficiency/ Leigh syndrome (NDUFS6- related)

Mitochondrial myopathy and sideroblastic anemia 1

Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease

Mitochondrial DNA depletion syndrome (MPV17-related)

MKS1-related disorders

Mucolipidosis type II/III (GNPTAB-related)

Mucolipidosis type IV

Mucolipidosis type III (GNPTG-related)

Mucopolysaccharidosis type I (includes Hurler, Hurler-Scheie, and Scheie syndromes)

Mucopolysaccharidosis type II (Hunter syndrome)

Mucopolysaccharidosis type IX

Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)

Mucopolysaccharidosis type IIIA (Sanfilippo A syndrome)

Mucopolysaccharidosis type IIIB

Mucopolysaccharidosis type IIIC (Sanfilippo syndrome)/ Retinitis pigmentosa 73

Mucopolysaccharidosis type IIID (Sanfilippo syndrome)

Mucopolysaccharidosis type IVB (Morquio B syndrome)/ GM1 gangliosidosis

Multiple sulfatase deficiency

N-Acetylglutamate synthase deficiency

Nemaline myopathy 2

Nephrogenic diabetes insipidus (AQP2-related)

Nephrotic syndrome/ Congenital Finnish nephrosis (NPHS1-related)

Nephrotic syndrome/Steroid-resistant nephrotic syndrome (NPHS2-related)

Neuronal ceroid lipofuscinosis (TPP1-related)

Neuronal ceroid-lipofuscinosis (CLN3-related)

Neuronal ceroid-lipofuscinosis (CLN5-related)

Neuronal ceroid-lipofuscinosis (CLN6-related)

Neuronal ceroid-lipofuscinosis (MFSD8-related)

Neuronal ceroid-lipofuscinosis (PPT1-related)

Neuronal ceroid-lipofuscinosis/ Northern epilepsy (CLN8-related)

Niemann-Pick disease type A/B

Niemann-Pick disease type C (NPC1-related)

Niemann-Pick disease type C (NPC2-related)

Nijmegen breakage syndrome

Ornithine aminotransferase deficiency

Ornithine transcarbamylase (OTC) deficiency

Osteopetrosis (TCIRG1-related)

Pendred syndrome

Peroxisomal acyl-CoA oxidase deficiency

Phenylalanine hydroxylase deficiency (including Phenylketonuria (PKU))

Phosphoglycerate dehydrogenase deficiency/ Neu-Laxova syndrome

Polycystic kidney disease (PKHD1-related)

Polymicrogyria (ADGRG1-related)

POMGNT1-related disorders (including Muscle eye brain disease)

Pontocerebellar hypoplasia (RARS2-related)

Pontocerebellar hypoplasia (SEPSECS-related)

Pontocerebellar hypoplasia (VRK1-related)

Postnatal progressive microcephaly with seizures and brain atrophy/ Infantile cerebral and cerebellar atrophy (MED17-related)

Primary carnitine deficiency

Primary Ciliary Dyskinesia (DNAH5-related)

Primary Ciliary Dyskinesia (DNAI1-related)

Primary Ciliary Dyskinesia (DNAI2-related)

Primary hyperoxaluria type 1

Primary hyperoxaluria type 2

Primary hyperoxaluria type 3

Progressive familial intrahepatic cholestasis type 2

Propionic acidemia (PCCA-related)

Propionic acidemia (PCCB-related)

Propionic acidemia (PCCB-related)

PRPS1-related disorders (including Charcot-Marie-Tooth disease type 5 and Arts syndrome)

Pycnodysostosis

Pyruvate carboxylase deficiency

Pyruvate dehydrogenase deficiency (PDHA1-related)

Pyruvate dehydrogenase deficiency (PDHB-related)

Renal tubular acidosis with deafness (ATP6V1B1-related)

Retinitis pigmentosa 25

Retinitis pigmentosa 26

Retinitis Pigmentosa 28

Rhizomelic chondrodysplasia punctata type 1/ Refsum disease (PEX7-related)

Rhizomelic chondrodysplasia punctata type 3

Roberts syndrome

RPGRIP1L-related disorders (including Joubert syndrome 7, COACH syndrome and Meckel syndrome 5)

RTEL-1-related disorders (including Dyskeratosis congenita)

Sandhoff disease

Schimke immuno-osseous dysplasia

Severe combined immune deficiency (DCLRE1C-related)

Severe combined immunodeficiency/ Omenn syndrome (RAG2-related)

Severe congenital neutropenia (VPS45-related)

Sialic acid storage disorders

Sjögren-Larsson syndrome

SLC26A2-related disorders (including Diatrophic dysplasia, Atelosteogenesis type 2, Achondrogenesis type 1B/ Multiple metaphyseal dysplasia)

SLC35A3-related disorder

Smith-Lemli-Opitz syndrome

Spastic paraplegia type 15

Spastic paraplegia type 49

Spinal muscular atrophy

Spondylothoracic dysostosis

Steel Syndrome

Stüve-Wiedemann syndrome

Tay-Sachs disease/ Hexosaminidase A deficiency

Tetrahydrobiopterin deficiency (PTS-related)

Transient infantile liver failure (TRMU-related)

Tyrosine hydroxylase deficiency

Tyrosinemia type I

Tyrosinemia type II

Usher syndrome type IB/ MYO7A-related disorders

Usher syndrome type IC/ USH1C-related disorders

Usher syndrome type ID

Usher syndrome type IF/ PCDH15-related disorders

Usher syndrome type IIA/ USH2A-related disorders

Usher syndrome type IIIA

Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency

Walker-Warburg syndrome/ FKRP-related disorders

Walker-Warburg syndrome/ FKTN-related disorders

Wilson disease

WNT10A-related disorders (including Odonto-onycho-dermal dysplasia and Schopf-Schulz-Passarge syndrome)

Xeroderma pigmentosum complementation group A

Xeroderma pigmentosum complementation group C

X-linked adrenoleukodystrophy

X-linked creatine transporter deficiency

X-linked juvenile retinoschisis

X-linked myotubular myopathy

X-linked severe combined immunodeficiency (X-SCID)

Zellweger spectrum disorder (PEX10-related)

Zellweger spectrum disorder (PEX12-related)

Zellweger spectrum disorder (PEX1-related)

Zellweger spectrum disorder (PEX2-related)

Zellweger spectrum disorder (PEX6-related)

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