Disorders Tested

Exon Panel

preparent®
CARRIER TEST

CLINICAL IMPACT AREAS

Life Expectancy	Decreased life expectancy and increased childhood mortality
Quality of Life	Severe impact on quality of life; typically no effect on life expectancy
Treatment Benefits	Early medical intervention can increase life expectancy or reduce symptoms
Intellectual Ability	Associated with varying degrees of intellectual disability

	Life Expectancy	Quality of Life	Treatment Benefits	Intellectual Ability
3-hydroxy-3-methylglutaryl CoA lyase deficiency	●		●	●
3-phosphoglycerate dehydrogenase deficiency	●		●	●
6-pyruvoyl-tetrahydropterin synthase (PTS) deficiency	●		●	●
Abetalipoproteinemia			●	
Adenosine deaminase deficiency	●			
Adrenoleukodystrophy (X-linked)*	●		●	●
Agammaglobulinemia (X-linked)*		●	●	
Alpha-1 antitrypsin deficiency	●			
Alpha-mannosidosis	●			●
Alpha-thalassemia†	●		●	
Alport syndrome, COL4A3-related	●			
Andermann syndrome	●			●
Angelman syndrome		●		●
Argininosuccinic aciduria	●		●	●
Arthrogryposis, mental retardation and seizures	●			●
Aspartylglycosaminuria	●			●
Ataxia with vitamin E deficiency	●		●	
Ataxia-telangiectasia	●			
Autosomal recessive polycystic kidney disease	●			
Bardet-Biedl syndrome, BBS1-related	●			●
Bardet-Biedl syndrome, BBS10-related	●			●
Bardet-Biedl syndrome, BBS2-related	●			●
Beta-hemoglobinopathies†		●	●	
Biotinidase deficiency	●		●	●
Bloom syndrome	●			●
Canavan disease	●			●
Carnitine palmitoyltransferase IA deficiency	●		●	
Carnitine palmitoyltransferase II deficiency	●		●	
Cartilage-hair hypoplasia	●			
Cerebrotendinous xanthomatosis		●	●	
Chronic granulomatous disease (X-linked)*		●	●	
Citrin deficiency		●	●	
Citrullinemia, type I	●		●	
Combined pituitary hormone deficiency, PROP1-related		●	●	
Congenital amegakaryocytic thrombocytopenia	●			
Congenital disorder of glycosylation, type IA	●			●
Congenital disorder of glycosylation, type IB	●		●	
Congenital myasthenic syndrome, CHAT-related		●	●	
Congenital myasthenic syndrome, CHRNE-related		●	●	
Congenital myasthenic syndrome, RAPSN-related		●	●	
Crigler-Najjar syndrome	●		●	●
Cystic fibrosis†	●			
Cystinosis†		●		
D-bifunctional protein deficiency	●			
Dihydrolipoamide dehydrogenase deficiency	●		●	●
Dihydropyrimidine dehydrogenase deficiency	●		●	●
Duchenne/Becker muscular dystrophy (X-linked)*†	●			
Dyskeratosis congenita, RTEL1-related	●			
Ehlers Danlos syndrome, type VIIC		●		
Ethlymalonic encephalopathy	●		●	
Familial dysautonomia	●			
Familial hyperinsulinism, ABCC8-related		●		
Fanconi anemia, type C	●		●	●
Fetal akinesia deformation sequence, DOK7-related		●		
Fragile X syndrome (X-linked)*		●		●
Fumarase deficiency	●			●
Galactosemia†	●		●	●
Gaucher disease	●		●	
Glucose-6-phosphate dehydrogenase deficiency (X-linked)*		●	●	
Glutaric acidemia, type I	●		●	●
Glycine encephalopathy, AMT-related	●			●
Glycine encephalopathy, GLDC-related	●			●
Glycogen storage disease, type IA	●		●	
Glycogen storage disease, type IB	●		●	
Glycogen storage disease, type II	●		●	
Glycogen storage disease, type III	●		●	
GM1-gangliosidosis	●			●
GRACILE syndrome	●			
Hereditary fructose intolerance		●	●	
Hermansky-Pudlak syndrome, type III	●			

	Life Expectancy	Quality of Life	Treatment Benefits	Intellectual Ability
Homocystinuria, CBS-related	●		●	●
Hypophosphatasia, autosomal recessive	●			
Inclusion body myopathy, type II		●		
Infantile neuroaxonal dystrophy, type 1	●			●
Isovaleric acidemia	●		●	
Joubert syndrome 2	●			●
Junctional epidermolysis bullosa, LAMA3-related	●			
Junctional epidermolysis bullosa, LAMB3-related	●			
Junctional epidermolysis bullosa, LAMC2-related	●			
Juvenile nephronophthisis, type 1†		●		●
Krabbe disease†	●		●	
Lamellar ichthylosis, type I		●		
Leigh syndrome, French Canadian	●			●
Leukoencephalopathy with vanishing white matter	●			●
Limb-girdle muscular dystrophy, type 2A	●	●		
Limb-girdle muscular dystrophy, type 2C		●		
Limb-girdle muscular dystrophy, type 2D		●		
Limb-girdle muscular dystrophy, type 2E		●		
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	●		●	●
Lowe syndrome (X-linked)*	●			●
Lysinuric protein intolerance			●	●
Maple syrup urine disease, type IA	●		●	●
Maple syrup urine disease, type IB	●		●	●
Maple syrup urine disease, type II	●		●	●
MECP2 duplication syndrome (X-linked)*†	●			●
Medium-chain acyl-CoA dehydrogenase deficiency	●		●	●
Megalencephalic leukoencephalopathy with subcortical cysts, type I		●		●
Metachromatic leukodystrophy	●			●
Methylmalonic aciduria, cblC type	●		●	●
Mucolipidosis type IV†		●		●
Mucolipidosis, type II/III alpha/beta	●			●
Mucopolysaccharidosis type IIIA (Sanfilippo A)	●			●
Mucopolysachharidosis, type I-Hurler syndrome	●		●	●
Multiple sulfatase deficiency	●			●
Muscle-eye-brain disease	●			●
Nemaline myopathy 2†	●			
Nephrotic syndrome, type I	●			
Neuronal ceroid lipofuscinosis, CLN3-related†	●			●
Neuronal ceroid lipofuscinosis, CLN5-related	●			●
Neuronal ceroid lipofuscinosis, CLN6-related	●			●
Neuronal ceroid lipofuscinosis, CLN8-related	●			●
Neuronal ceroid lipofuscinosis, PPT1-related	●			●
Neuronal ceroid lipofuscinosis, TPP1-related	●			●
Niemann-Pick disease, type A and B	●			●

	Life Expectancy	Quality of Life	Treatment Benefits	Intellectual Ability
Niemann-Pick disease, type C1	●			●
Nijmegen breakage syndrome	●			●
Nonsyndromic hearing loss, GJB2-related		●	●	
Nonsyndromic hearing loss, GJB6-related†		●	●	
Omenn syndrome	●		●	
Ornithine transcarbamylase deficiency (X-linked)*	●		●	●
Ornithine translocase deficiency	●		●	●
Pendred syndrome			●	
Phenylalanine hydroxylase deficiency			●	●
POLG-related disorders	●			●
Primary carnitine deficiency		●	●	
Primary congenital glaucoma			●	
Primary hyperoxaluria, type I	●		●	
Primary hyperoxaluria, type II	●		●	
Propionic acidemia, PCCA-related	●		●	●
Propionic acidemia, PCCB-related	●		●	●
Pycnodysostosis		●		
Pyruvate carboxylase deficiency	●			●
Retinitis pigmentosa 59		●		
Rhizomelic chondrodysplasia punctata, type I	●			●
Salla disease	●			●
Sandhoff disease	●			●
Severe combined immunodeficiency, IL2RG-related (X-linked)*	●		●	
Sjögren-Larsson syndrome	●			●
Smith-Lemli-Opitz syndrome	●		●	●
Spastic ataxia of Charlevoix-Saguenay (ARSACS)	●			
Spinal muscular atrophy†	●		●	
Steroid-resistant nephrotic syndrome	●			
Sulfate transporter-related osteochondrodysplasia	●			
Tay-Sachs disease†	●			●
Tyrosine hydroxylase deficiency		●	●	●
Tyrosinemia, type I	●		●	
Usher syndrome, type IB		●	●	
Usher syndrome, type IC		●	●	
Usher syndrome, type ID		●	●	
Usher syndrome, type IF		●	●	
Usher syndrome, type IIA		●	●	
Usher syndrome, type III		●	●	
Very long-chain acyl-CoA dehydrogenase deficiency	●		●	
Walker-Warburg syndrome	●			●
Wilson disease	●		●	
Wiskott-Aldrich syndrome (X-linked)*	●		●	
Zellweger spectrum disorders, PEX1-related	●			●
Zellweger spectrum disorders, PEX2-related	●			●

For more information, visit **progenity.com/preparent**

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* Carrier testing for X-linked disorders is not performed in males. † CNV analysis is clinically relevant and performed for select disorders.

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