Disorders Tested

Global and Global+ 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
17-alpha-hydroxylase deficiency	●		●	
17-beta-hydroxysteroid dehydrogenase deficiency, type III			●	
3-beta-hydroxysteroid dehydrogenase deficiency, type II	●		●	
3-hydroxy-3-methylglutaryl CoA lyase deficiency	●		●	●
3-methylglutaconic aciduria, type III	●			●
3-phosphoglycerate dehydrogenase deficiency	●		●	●
Abetalipoproteinemia			●	
Achalasia-addisonianism-alacrima syndrome	●		●	
Achromatopsia, *CNGA3*-related		●	●	
Achromatopsia, *CNGB3*-related		●	●	
Acrodermatitis enteropathica			●	
Adenosine deaminase deficiency	●			
Adrenoleukodystrophy **(X-linked)***	●		●	●
Aicardi-Goutieres syndrome, *RNASEH2C*-related	●		●	●
Aicardi-Goutieres syndrome, *SAMHD1*-related	●		●	●
Aicardi-Goutieres syndrome, *TREX1*-related	●		●	●
Alkaptonuria		●		
Alpha-mannosidosis	●			●
Alpha-thalassemia	●		●	
Alport syndrome, *COL4A3*-related	●			
Alport syndrome, *COL4A5*-related **(X-linked)***	●			
Amish infantile epilepsy syndrome	●			●
Andermann syndrome	●			●
Argininosuccinic aciduria	●		●	●
Aromatase deficiency			●	
Arthrogryposis, mental retardation and seizures	●			●
Arts syndrome **(X-linked)***	●		●	●
Aspartylglycosaminuria	●			●
Ataxia-telangiectasia	●			

	Life Expectancy	Quality of Life	Treatment Benefits	Intellectual Ability
Ataxia-telangiectasia-like disorder	●			
Ataxia with vitamin E deficiency	●		●	
Autoimmune polyglandular syndrome, type I		●		
Autosomal recessive polycystic kidney disease	●			
Bardet-Biedl syndrome, *BBS1*-related	●			●
Bardet-Biedl syndrome, *BBS10*-related	●			●
Bardet-Biedl syndrome, *BBS12*-related	●			●
Bartter syndrome, type IV	●		●	
Beta-ketothiolase deficiency	●		●	
Beta-thalassemia		●	●	
Bilateral frontoparietal polymicrogyria	●		●	●
Biotinidase deficiency	●		●	●
Bloom syndrome	●			●
Canavan disease	●			●
Carnitine palmitoyltransferase I deficiency	●		●	
Carnitine palmitoyltransferase II deficiency	●		●	
Carpenter syndrome	●			●
Cartilage-hair hypoplasia	●			
Charcot-Marie-Tooth disease, *GJB1*-related **(X-linked)***		●		
Charcot-Marie-Tooth disease, *PRPS1*-related **(X-linked)***		●		
Chediak-Higashi syndrome	●			●
Cholesteryl ester storage disease	●		●	
Choroideremia **(X-linked)***		●		
Citrullinemia, type I	●		●	
Congenital amegakaryocytic thrombocytopenia	●			
Congenital disorder of glycosylation, type IA	●			●
Congenital disorder of glycosylation, type IB	●		●	
Congenital lipoid adrenal hyperplasia	●			
Congenital neutropenia, *HAX1*-related	●			

	Life Expectancy	Quality of Life	Treatment Benefits	Intellectual Ability
Corneal dystrophy and perceptive deafness syndrome		●		
Corticosterone methyloxidase deficiency	●		●	
Creatine transporter defect, *SLC6A8*-related **(X-linked)***	●			●
Crigler-Najjar syndrome	●		●	●
Cystic fibrosis (600 mutations)	●			
Cystinosis		●		
D-bifunctional protein deficiency	●			
Desbuquois dysplasia, type I	●			
Dihydrolipoamide dehydrogenase deficiency	●		●	●
Dihydropyrimidine dehydrogenase deficiency	●		●	●
Du Pan syndrome		●		
Dyskeratosis congenita, *RTEL1*-related	●			
Dyskeratosis congenita, *DKC1*-related **(X-linked)***	●			
Dystrophic epidermolysis bullosa, *COL7A1*-related	●			
Early onset myopathy with fatal cardiomyopathy	●			
Ehlers-Danlos syndrome, type VIIC		●		
Emery-Dreifuss muscular dystrophy **(X-linked)***	●			
Enhanced S-cone syndrome		●	●	
Ethylmalonic encephalopathy	●		●	
Fabry disease **(X-linked)***	●		●	
Familial dysautonomia	●			
Familial hyperinsulinism, *ABCC8*-related		●		
Familial hyperinsulinism, *KCNJ11*-related		●		
Familial Mediterranean fever		●	●	
Fanconi anemia, type A	●		●	●
Fanconi anemia, type C	●		●	●
Fetal akinesia deformation sequence, *DOK7*-related		●		
Fragile X syndrome **(X-linked)***		●		●
Fumarase deficiency	●			●
Galactokinase deficiency		●	●	
Galactosemia	●		●	●
Gaucher disease	●		●	
Geroderma osteodysplastica	●			●
Gitelman syndrome		●	●	
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	●		●	
Glycogen storage disease, type IV	●		●	
Glycogen storage disease, type V		●		

	Life Expectancy	Quality of Life	Treatment Benefits	Intellectual Ability
Glycogen storage disease, type VII		●		
GM1-gangliosidosis	●			●
GRACILE syndrome	●			
Guanidinoacetate methyltransferase deficiency	●		●	●
Hemoglobinopathy, Hb C		●	●	
Hemoglobinopathy, Hb D		●	●	
Hemoglobinopathy, Hb E		●	●	
Hemoglobinopathy, Hb O		●	●	
Hemophilia A **(X-linked)***		●	●	
Hemophilia B **(X-linked)***		●	●	
Hepatocerebral mitochondrial DNA depletion syndrome, *MPV17*-related	●			
Hermansky-Pudlak syndrome, type III	●			
Holocarboxylase synthetase deficiency	●		●	
Homocystinuria, *CBS*-related	●		●	●
Hyperphosphatemic familial tumoral calcinosis		●	●	
Hypohidrotic ectodermal dysplasia		●		
Hypophosphatasia	●			
Isovaleric acidemia	●		●	
Joubert syndrome 2	●			●
Junctional epidermolysis bullosa, *LAMB3*-related	●			
Juvenile retinoschisis **(X-linked)***		●		
Krabbe disease	●		●	
Leber congenital amaurosis, *LCA5*-related		●		
Leber congenital amaurosis, *RDH12*-related		●		
Leigh syndrome, French Canadian	●			●
Limb-girdle muscular dystrophy, type 2A		●		
Limb-girdle muscular dystrophy, type 2D		●		
Limb-girdle muscular dystrophy, type 2E		●		
Limb-girdle muscular dystrophy, type 2I		●		
Lipoprotein lipase deficiency		●	●	
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	●		●	●
Luteinizing hormone resistance		●		
Maple syrup urine disease, type IA	●		●	●
Maple syrup urine disease, type IB	●		●	●
Medium-chain acyl-CoA dehydrogenase deficiency	●		●	●
MEDNIK syndrome	●			●
Metachromatic leukodystrophy	●			●
Methylmalonic aciduria, cblA type	●		●	●
Methylmalonic aciduria, cblB type	●		●	●
Methylmalonic aciduria, cblC type	●		●	●
Methylmalonic aciduria, *MUT*-related	●		●	●
Mitochondrial complex IV deficiency	●			●
Mitochondrial myopathy and sideroblastic anemia	●			

	Life Expectancy	Quality of Life	Treatment Benefits	Intellectual Ability
Mucolipidosis, type II/III alpha/beta	●			●
Mucolipidosis, type IV		●		●
Mucopolysaccharidosis, type I-Hurler syndrome	●		●	●
Mucopolysaccharidosis, type II-Hunter syndrome **(X-linked)***	●		●	●
Mucopolysaccharidosis, type IIIC	●		●	●
Mucopolysaccharidosis, type VI	●		●	
Mulibrey nanism		●		
Multiple sulfatase deficiency	●			●
Muscle-eye-brain disease	●			●
Myotubular myopathy, *MTM1*-related **(X-linked)***	●			
Nemaline myopathy 2	●			
Nephrotic syndrome, type I	●			
Neuronal ceroid lipofuscinosis, *CLN5*-related	●			●
Neuronal ceroid lipofuscinosis, *CLN6*-related	●			●
Neuronal ceroid lipofuscinosis, *CLN8*-related	●			●
Neuronal ceroid lipofuscinosis, *MFSD8*-related	●			●
Neuronal ceroid lipofuscinosis, *PPT1*-related	●			●
Neuronal ceroid lipofuscinosis, *TPP1*-related	●			●
Niemann-Pick disease, type A and B	●			●
Niemann-Pick disease, type CI	●			●
Niemann-Pick disease, type CII	●			●
Nijmegen breakage syndrome	●			●
Nonsyndromic hearing loss, *GJB2*-related		●	●	
Omenn syndrome	●		●	
Ornithine transcarbamylase deficiency **(X-linked)***	●		●	●
Ornithine translocase deficiency	●		●	●
Pendred syndrome			●	
Phenylalanine hydroxylase deficiency			●	●
Pontocerebellar hypoplasia, type IA	●			●
Primary congenital glaucoma			●	
Primary hyperoxaluria, type I	●		●	
Primary hyperoxaluria, type II	●		●	
Progressive familial intrahepatic cholestasis, type II	●			
Prolidase deficiency	●			●
Propionic acidemia, *PCCA*-related	●		●	●
Propionic acidemia, *PCCB*-related	●		●	●
Pseudoxanthoma elasticum		●		
Pycnodysostosis		●		
Pyruvate dehydrogenase deficiency, *PDHB*-related	●			●
Pyruvate dehydrogenase deficiency, *PDHA1*-related **(X-linked)***	●			●
Retinitis pigmentosa 59		●		
Rhizomelic chondrodysplasia punctata, type I	●			●
Salla disease	●			●
Sandhoff disease	●			●

	Life Expectancy	Quality of Life	Treatment Benefits	Intellectual Ability
Severe combined immunodeficiency, *RAG1*-related	●		●	
Severe combined immunodeficiency, *IL2RG*-related **(X-linked)***	●		●	
Shwachman-Diamond syndrome	●			●
Sickle cell anemia (Hb S)	●		●	
Sjögren-Larsson syndrome	●			●
Smith-Lemli-Opitz syndrome	●		●	●
Spastic ataxia of Charlevoix-Saguenay (ARSACS)	●			
Spinal muscular atrophy	●		●	
Steroid-resistant nephrotic syndrome	●			
Stuve-Wiedemann syndrome	●			
Sulfate transporter-related osteochondrodysplasia	●			
Tay-Sachs disease	●			●
Tumoral calcinosis, normophosphatemic	●		●	
Tyrosinemia, type I	●		●	
Usher syndrome, type IB		●	●	
Usher syndrome, type IC		●	●	
Usher syndrome, type ID		●	●	
Usher syndrome, type IF		●	●	
Usher syndrome, type III		●	●	
Very long-chain acyl-CoA dehydrogenase deficiency	●		●	
Vitamin D-dependent rickets, type I			●	
Walker-Warburg syndrome	●			●
Wilson disease	●		●	
Zellweger syndrome spectrum, *PEX6*-related	●			●

Global+ Panel includes these additional disorders

Alpha-1 antitrypsin deficiency	●			
Autosomal recessive woolly hair/hypotrichosis		●		
Hereditary fructose intolerance		●	●	
Hereditary hemochromatosis, *HFE*-related		●	●	
Hereditary hemochromatosis, *HFE2*-related	●		●	
Hereditary hemochromatosis, *TFR2*-related		●	●	
Inclusion body myopathy, type II		●		
Lamellar ichthyosis, type I		●		
Limb-girdle muscular dystrophy, type 2B		●		
Pseudocholinesterase deficiency		●	●	
Spastic paraplegia, *ZFYVE26*-related	●			●
Xeroderma pigmentosum	●			●

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

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*X-linked disorders are not tested in males.

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