All diseases on Foresight

Disease	Has a carrier frequency of 1/100 or greater in at least one ethnicity	Has a well-defined phenotype	Detrimental effect on quality of life in most affected individuals	May cause cognitive or physical impairment	Typically requires medical or surgical intervention	Often has childhood onset	Prenatal diagnosis is available
11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia		•	•	•	•	•	•
21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia	•	•	•	•	•	•	•
3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency		•	•	•	•	•	•
3-Methylglutaconic Aciduria Type III (a.k.a. Costeff Optic Atrophy Syndrome)	•	•	•	•	•	•	•
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency	•	•	•	•	•	•	•
Adenosine Deaminase Deficiency		•	•	•	•	•	•
Adrenoleukodystrophy[1]	•	•	•	•	•	•	•
Alpha Thalassemia	•	•	•	•	•	•	•
Alpha-Mannosidosis		•	•	•	•	•	•
Alpha-Sarcoglycanopathies (Including Limb-Girdle Muscular Dystrophy Type 2D)		•	•	•	•	•	•

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Alport Syndrome, X-Linked[1]		•	•	•	•	•	•
Alstrom Syndrome		•	•	•	•	•	•
Andermann Syndrome	•	•	•	•	•	•	•
Argininemia		•	•	•	•	•	•
Argininosuccinic Aciduria		•	•	•	•	•	•
Aspartylglycosaminuria	•	•	•	•	•	•	•
Ataxia With Isolated Vitamin E Deficiency		•	•	•	•	•	•
Ataxia-Telangiectasia		•	•	•	•	•	•
Autoimmune Polyendocrinopathy Syndrome Type I	•	•	•	•	•	•	•
Autosomal Recessive Congenital Ichthyosis 1		•	•	•	•	•	•
Autosomal Recessive Osteopetrosis 1	•	•	•	•	•	•	•
Bardet-Biedl Syndrome 1		•	•	•	•	•	•
Bardet-Biedl Syndrome 10		•	•	•	•	•	•
Bardet-Biedl Syndrome 12		•	•	•	•	•	•

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Bardet-Biedl Syndrome 2		•	•	•	•	•	•
Beta-Sarcoglycanopathies (Including Limb-Girdle Muscular Dystrophy Type 2E)		•	•	•	•	•	•
Biotinidase Deficiency		•	•	•	•	•	•
Bloom Syndrome		•	•	•	•	•	•
Calpainopathies (Including Limb-Girdle Muscular Dystrophy Type 2A)		•	•	•	•	•	•
Canavan Disease	•	•	•	•	•	•	•
Carbamoylphosphate Synthetase I Deficiency		•	•	•	•	•	•
Carnitine Palmitoyltransferase Deficiency Type IA		•	•	•	•	•	•
Carnitine Palmitoyltransferase Deficiency Type II	•	•	•	•	•	•	•
Cartilage-Hair Hypoplasia		•	•	•	•	•	•
Cerebrotendinous Xanthomatosis	•	•	•	•	•	•	•
Citrullinemia		•	•	•	•	•	•
Cohen Syndrome		•	•	•	•	•	•

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COL4A3-related Alport syndrome		•	•	•	•	•	•
COL4A4-related Alport syndrome		•	•	•	•	•	•
Combined Pituitary Hormone Deficiency 2		•	•	•	•	•	•
Congenital Adrenal Hypoplasia with Hypogonadotropic Hypogonadism[1]	•	•	•	•	•	•	•
Congenital Disorder of Glycosylation Type Ia		•	•	•	•	•	•
Congenital Disorder of Glycosylation Type Ib		•	•	•	•	•	•
Congenital Disorder of Glycosylation Type Ic		•	•	•	•	•	•
Cystic Fibrosis	•	•	•	•	•	•	•
Cystinosis		•	•	•	•	•	•
D-Bifunctional Protein Deficiency		•	•	•	•	•	•
Delta-Sarcoglycanopathies (Including Limb-Girdle Muscular Dystrophy Type 2F)		•	•	•	•	•	•

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DFNB1A (a.k.a. *GJB2*-Related Nonsyndromic Hearing Loss and Deafness)	•	•	•	•	•	•	•
Dihydrolipoamide Dehydrogenase Deficiency	•	•	•	•	•	•	•
Dysferlinopathies (Including Limb-Girdle Muscular Dystrophy Type 2B)		•	•	•	•		•
Dyskeratosis Congenita	•	•	•	•	•	•	•
Dystrophinopathies (Including Duchenne Muscular Dystrophy and Becker Muscular Dystrophy)[1]	•	•	•	•	•	•	•
ERCC6-Related Disorders (Including Cockayne Syndrome Type B)		•	•	•	•	•	•
ERCC8-Related Disorders (Including Cockayne Syndrome Type A)		•	•	•	•	•	•
EVC-related Ellis-van Creveld Syndrome and Weyers Acrofacial Dysostosis		•	•	•	•	•	•
EVC2-related Ellis-van Creveld Syndrome and Weyers Acrofacial Dysostosis		•	•	•	•	•	•

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Fabry Disease[1]	•	•	•	•	•	•	•
Familial Dysautonomia	•	•	•	•	•	•	•
Familial Hyperinsulinemic Hypoglycemia Type 1	•	•	•	•	•	•	•
Familial Hyperinsulinemic Hypoglycemia Type 2		•	•	•	•	•	•
Familial Mediterranean Fever	•	•	•	•	•		•
Fanconi Anemia Complementation Group A		•	•	•	•	•	•
Fanconi Anemia Complementation Group C	•	•	•	•	•	•	•
Fragile X Syndrome[1]	•	•	•	•	•	•	•
Galactokinase Deficiency with Cataracts	•	•	•	•	•	•	•
Galactosemia	•	•	•	•	•	•	•
Gamma-Sarcoglycanopathies (Including Limb-Girdle Muscular Dystrophy Type 2C)		•	•	•	•	•	•
Gaucher Disease	•	•	•	•	•	•	•
Glutaricaciduria Type I	•	•	•	•	•	•	•

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Glycine Encephalopathy		•	•	•	•	•	•
Glycine Encephalopathy		•	•	•	•	•	•
Glycogen Storage Disease Ia	•	•	•	•	•	•	•
Glycogen Storage Disease Ib/Ic		•	•	•	•	•	•
Glycogen Storage Disease II (a.k.a. Pompe Disease)	•	•	•	•	•	•	•
Glycogen Storage Disease III		•	•	•	•	•	•
GM1-Gangliosidoses (Including Mucopolysaccharidosis Type IVB, Morquio Syndrome B)		•	•	•	•	•	•
GRACILE Syndrome		•	•	•	•	•	•
HADHA-Related disorders (Including Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency and Trifunctional Protein Deficiency)		•	•	•	•	•	•
Hb Beta Chain-Related Hemoglobinopathy	•	•	•	•	•	•	•
Hereditary Fructose Intolerance	•	•	•	•	•	•	•

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Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	•	•	•	•	•	•	•
HGSNAT-Related diseases (Including Mucopolysaccharidosis Type IIIC, a.k.a. Sanfilippo C)		•	•	•	•	•	•
Holocarboxylase Synthetase Deficiency		•	•	•	•	•	•
Homocystinuria due to Cystathionine Beta-Synthase Deficiency		•	•	•	•	•	•
Hurler Syndrome	•	•	•	•	•	•	•
Hydrolethalus Syndrome	•	•	•	•	•	•	•
Hypophosphatasia		•	•	•	•	•	•
Inclusion Body Myopathy~2		•	•	•	•	•	•
Isovaleric Acidemia		•	•	•	•	•	•
Junctional Epidermolysis Bullosa, *LAMA3*-Related		•	•	•	•	•	•
Junctional Epidermolysis Bullosa, *LAMB3*-Related		•	•	•	•	•	•
Junctional Epidermolysis Bullosa, *LAMC2*-Related		•	•	•	•	•	•

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Krabbe Disease		•	•	•	•	•	•
Leigh Syndrome, French-Canadian Type	•	•	•	•	•	•	•
Lipoid Adrenal Hyperplasia		•	•	•	•	•	•
Lysosomal Acid Lipase Deficiency (Including Cholesteryl Ester Storage Disease and Wolman Disease)		•	•	•		•	•
Maple Syrup Urine Disease Type 1B	•	•	•	•	•	•	•
Maple Syrup Urine Disease Type IA	•	•	•	•	•	•	•
Maple Syrup Urine Disease Type II		•	•	•	•	•	•
Medium Chain Acyl-CoA Dehydrogenase Deficiency	•	•	•	•	•	•	•
Megalencephalic Leukoencephalopathy With Subcortical Cysts	•	•	•	•	•	•	•
Menkes Syndrome; Occiptal Horn Syndrome; Distal Spinal Muscular Atrophy X-Linked[1]		•	•	•	•	•	•
Merosin-Deficient Congenital Muscular Dystrophy		•	•	•	•	•	•

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Metachromatic Leukodystrophy		•	•	•	•	•	•
Methylmalonic Acidemia and Homocystinuria, cblC Type		•	•	•	•	•	•
Methylmalonic Acidemia, cblA Type		•	•	•	•	•	•
Methylmalonic Acidemia, cblB Type		•	•	•	•	•	•
Methylmalonic Acidemia, Mut Type	•	•	•	•	•	•	•
MKS1-Related diseases (Including Meckel Syndrome 1 and Bardet-Biedl Syndrome 13)	•	•	•	•	•	•	•
Mucolipidosis II/III		•	•	•	•	•	•
Mucolipidosis III Gamma		•	•	•	•	•	•
Mucolipidosis IV		•	•	•	•	•	•
Mucopolysaccharidosis II (a.k.a. Hunter Syndrome)[1]	•	•	•	•	•	•	•
Mucopolysaccharidosis Type IIIB (Sanfilippo B)		•	•	•	•	•	•
Muscular Dystrophy-Dystroglycanopathy 5 (Including Limb-Girdle Muscular Dystrophy Type 2I)		•	•	•	•	•	•

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Muscular Dystrophy-Dystroglycanopathy Type 3 (Including Muscle-Eye-Brain Disease)		•	•	•	•	•	•
Muscular Dystrophy-Dystroglycanopathy Type 4 (Including Walker-Warburg Syndrome and Fukuyama Congenital Muscular Dystrophy)		•	•	•	•	•	•
MYO7A-Related Disorders (Including Usher Syndrome Type 1B, DFNB2, and DFNA11)		•	•	•	•	•	•
Nemaline Myopathy 2		•	•	•	•	•	•
Nephrotic Syndrome Type 2		•	•	•	•	•	•
Nephrotic Syndrome Type~1	•	•	•	•	•	•	•
Neuronal Ceroid Lipofuscinosis 1	•	•	•	•	•	•	•
Neuronal Ceroid Lipofuscinosis 2		•	•	•	•	•	•
Neuronal Ceroid Lipofuscinosis 3	•	•	•	•	•	•	•
Neuronal Ceroid Lipofuscinosis 5	•	•	•	•	•	•	•
Neuronal Ceroid Lipofuscinosis 6		•	•	•	•	•	•
Neuronal Ceroid Lipofuscinosis 8 (Including Northern Epilepsy)		•	•	•	•	•	•

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Niemann-Pick Disease Type A/B	•	•	•	•	•	•	•
Niemann-Pick Disease Type C1/D		•	•	•	•	•	•
Niemann-Pick Disease Type C2		•	•	•	•	•	•
Nijmegen Breakage Syndrome		•	•	•	•	•	•
Ornithine Transcarbamylase Deficiency[1]	•	•	•	•	•	•	•
Pendred Syndrome	•	•	•	•	•	•	•
Peroxisome Biogenesis Disorder 1		•	•	•	•	•	•
Peroxisome Biogenesis Disorder 3		•	•	•	•	•	•
Peroxisome Biogenesis Disorder 4	•	•	•	•	•	•	•
Peroxisome Biogenesis Disorder 5		•	•	•	•	•	•
Peroxisome Biogenesis Disorder 6		•	•	•	•	•	•
Peroxisome Biogenesis Disorder 9 (a.k.a. Rhizomelic Chondrodysplasia Punctata Type 1)		•	•	•	•	•	•
Phenylalanine Hydroxylase Deficiency	•	•	•	•	•	•	•
Polycystic Kidney Disease Type I	•	•	•	•	•	•	•

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Primary Carnitine Deficiency		•	•	•	•	•	•
Primary Hyperoxaluria Type I		•	•	•	•	•	•
Primary Hyperoxaluria Type II		•	•	•	•	•	•
Primary Hyperoxaluria Type III	•	•	•	•	•	•	•
Propionic Acidemia Complementation Group pccA	•	•	•	•	•	•	•
Propionic Acidemia Complementation Group pccBC	•	•	•	•	•	•	•
Pycnodysostosis		•	•	•	•	•	•
Pyruvate Carboxylase Deficiency	•	•	•	•	•	•	•
Salla Disease	•	•	•	•	•	•	•
Sandhoff Disease		•	•	•	•	•	•
Sanfilippo, Type A		•	•	•	•	•	•
Segawa Syndrome		•	•	•	•	•	•
Short Chain Acyl-CoA Dehydrogenase Deficiency	•	•		•			•
Sjogren-Larsson Syndrome		•	•	•	•	•	•
Smith-Lemli-Opitz Syndrome	•	•	•	•	•	•	•

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Spastic Ataxia, Charlevoix-Saguenay Type	•	•	•	•	•	•	•
Spastic Paraplegia 15		•	•	•	•	•	•
Menkes Syndrome; Occiptal Horn Syndrome; Distal Spinal Muscular Atrophy X-Linked[1]		•	•	•	•	•	•
Spondylothoracic Dysostosis 2		•	•	•	•	•	•
Sulfate Transporter-Related Osteochondrodysplasia	•	•	•	•	•	•	•
TMEM216-Related Disorders (Including Joubert Syndrome 2)	•	•	•	•	•	•	•
Tyrosinemia Type II		•	•	•	•	•	•
Tyrosinemia Type II		•	•	•	•	•	•
USH1C-Related Disorders (Including Usher Syndrome Type IC and DFNB18A)		•	•	•	•	•	•
USH2A-Related Disorders (Including Usher Syndrome Type 2A and Retinitis Pigmentosa 39)		•	•	•	•	•	•
Usher Syndrome Type 1F		•	•	•	•	•	•
Usher Syndrome Type 3A		•	•	•	•	•	•

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Very Long Chain Acyl-CoA Dehydrogenase Deficiency	•	•	•	•	•	•	•
Wilson Disease	•	•	•	•	•	•	•
X-Linked Combined Immunodeficiency (Including SCID)*		•	•	•	•	•	•
X-Linked Juvenile Retinoschisis[1]	•	•	•	•	•	•	•
X-Linked Myotubular Myopathy[1]	•	•	•	•	•	•	•
Xeroderma Pigmentosum Group A	•	•	•	•	•	•	•
Xeroderma Pigmentosum Group C		•	•	•	•	•	•

[1] Inherited in an X-linked manner.