Beacon Carrier Screening Detecting 400+ Conditions

COVERAGE ~99% at 20x **DELETION / DUPLICATION** ≥ 2 exons **TURNAROUND TIME** 2 weeks*

Condition	Gene	Inheritance	ACOG/ ACMG (6 genes†)	Focus (30 genes†)	Ashkenzi Jewish (61 genes†)	ACMG Tier 3 (113 genes†)	Expanded (427 genes†)	Expanded Plus (436 genes†)
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	*HMGCL*	AR	-	-	-	-	✓	✓
3-Ketothiolase deficiency	*ACAT1*	AR	-	-	-	✓	✓	✓
3-Methylcrotonyl-CoA carboxylase 1 deficiency (3-MCC deficiency)	*MCCC1*	AR	-	-	-	-	✓	✓
3-Methylcrotonyl-CoA carboxylase 2 deficiency (3-MCC deficiency)	*MCCC2*	AR	-	-	-	✓	✓	✓
Abetalipoproteinemia	*MTTP*	AR	-	-	✓	-	✓	✓
Achondrogenesis, type IB; Atelosteogenesis II; Diastrophic dysplasia; Multiple epiphyseal dysplasia	*SLC26A2*	AR	-	-	-	✓	✓	✓
Achromatopsia	*CNGB3*	AR	-	-	-	✓	✓	✓
Acrodermatitis enteropathica	*SLC39A4*	AR	-	-	-	-	✓	✓
Acyl-CoA dehydrogenase-9 (ACAD9) Deficiency	*ACAD9*	AR	-	-	-	-	✓	✓
Adenosine deaminase deficiency	*ADA*	AR	-	-	-	-	✓	✓
Adrenal insufficiency, congenital, with 46,XY sex reversal, partial or complete	*CYP11A1*	AR	-	-	-	✓	✓	✓
Adrenoleukodystrophy, X-linked	*ABCD1*	XL	-	-	-	✓	✓	✓
Aicardi-Goutieres syndrome	*SAMHD1*	AR	-	-	-	-	✓	✓
Aicardi-Goutieres syndrome 2	*RNASEH2B*	AR	-	-	-	✓	✓	✓
Albinism, oculocutaneous, type IA; Albinism, oculocutaneous, type IB	*TYR*	AR	-	-	-	✓	✓	✓
Albinism, oculocutaneous, type II; Albinism, brown oculocutaneous; Skin/hair/eye pigmentation 1	*OCA2*	AR	-	-	-	✓	✓	✓
Alkaptonuria	*HGD*	AR	-	-	-	-	✓	✓
Allan-Herndon-Dudley syndrome	*SLC16A2*	XL	-	-	-	-	✓	✓
Alpha thalassemia	*HBA1/HBA2*	AR	✓	✓	✓	✓	✓	✓
Alpha thalassemia X-linked intellectual disability syndrome	*ATRX*	XL	-	-	-	-	✓	✓
Alpha-1 antitrypsin deficiency	*SERPINA1*	AR	-	-	-	-	-	✓
Alpha-mannosidosis	*MAN2B1*	AR	-	-	-	-	✓	✓
Alport syndrome, COL4A3-related	*COL4A3*	AR	-	-	✓	-	✓	✓
Alport syndrome, COL4A4-related	*COL4A4*	AR	-	-	-	-	✓	✓

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Alport syndrome, COL4A5-related	*COL4A5*	XL	-	-	-	-	✓	✓
Alstrom syndrome	*ALMS1*	AR	-	-	-	-	✓	✓
Anauxetic dysplasia; Cartilage-hair hypoplasia; Metaphyseal dysplasia without hypotrichosis	*RMRP*	AR	-	-	-	-	✓	✓
Andermann syndrome	*SLC12A6*	AR	-	-	-	-	✓	✓
Arginase deficiency	*ARG1*	AR	-	-	-	-	✓	✓
Argininosuccinate lyase deficiency	*ASL*	AR	-	-	-	✓	✓	✓
Aromatase deficiency	*CYP19A1*	AR	-	-	-	-	✓	✓
Arthrogryposis, mental retardation, and seizures	*SLC35A3*	AR	-	-	✓	-	✓	✓
Arts syndrome; Rosenberg-Chutorian syndrome; Phosphoribosylpyrophosphate synthetase superactivity; Non-syndromic hearing loss, PRPS1-related	*PRPS1*	XL	-	-	-	-	✓	✓
Asparagine synthetase deficiency	*ASNS*	AR	-	-	-	-	✓	✓
Aspartylglucosaminuria	*AGA*	AR	-	-	-	✓	✓	✓
Ataxia with isolated vitamin E deficiency	*TTPA*	AR	-	-	-	-	✓	✓
Ataxia-telangiectasia	*ATM*	AR	-	-	-	-	✓	✓
Atransferrinemia	*TF*	AR	-		-	✓	✓	✓
Autoimmune polyendocrinopathy syndrome type I	*AIRE*	AR	-	-	-	✓	✓	✓
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	*SACS*	AR	-	-	-	-	✓	✓
Bardet-Biedl syndrome 14; Joubert syndrome 5; Leber congenital amaurosis 10; Meckel syndrome 4; Senior-Løken syndrome 6	*CEP290*	AR	-	-	-	✓	✓	✓
Bardet-Biedl syndrome 2; Retinitis Pigmentosa 74	*BBS2*	AR	-	-	✓	✓	✓	✓
Bardet-Biedl syndrome type 1	*BBS1*	AR	-	-	-	✓	✓	✓
Bardet-Biedl syndrome type 10	*BBS10*	AR	-	-	-	-	✓	✓
Bardet-Biedl syndrome type 12	*BBS12*	AR	-	-	-	-	✓	✓
Bare lymphocyte syndrome, type II	*CIITA*	AR	-	-	-	-	✓	✓
Bartter syndrome	*BSND*	AR	-	-	-	-	✓	✓
Bernard-Soulier syndrome type A1	*GP1BA*	AR	-	-	-	-	✓	✓
Bernard-Soulier syndrome type C	*GP9*	AR	-	-	-	-	✓	✓
Bilateral frontoparietal polymicrogyria	*ADGRG1*	AR	-	-	-	-	✓	✓
Biotinidase deficiency	*BTD*	AR	-	-	-	✓	✓	✓
Björnstad syndrome; GRACILE syndrome; Mitochondrial complex III deficiency	*BCS1L*	AR	-	-	-	-	✓	✓
Bloom syndrome	*BLM*	AR	-	✓	✓	✓	✓	✓
Butyrlcholinesterase deficiency	*BCHE*	AR	-	-	-	-	-	✓
Canavan disease	*ASPA*	AR	-	✓	✓	✓	✓	✓

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Carbamoylphosphate synthetase I deficiency	*CPS1*	AR	-	-	-	-	✓	✓
Carnitine palmitoyltransferase IA deficiency	*CPT1A*	AR	-	-	-	-	✓	✓
Carnitine palmitoyltransferase II deficiency	*CPT2*	AR	-	-	✓	✓	✓	✓
Carnitine-acylcarnitine translocase deficiency	*SLC25A20*	AR	-	-	-	-	✓	✓
Carpenter syndrome	*RAB23*	AR	-	-	-	-	✓	✓
Catecholaminergic polymorphic ventricular tachycardia, CASQ2-related	*CASQ2*	AR	-	-	-	-	✓	✓
Catecholaminergic polymorphic ventricular tachycardia, TRDN-related	*TRDN*	AR	-	-	-	-	✓	✓
Cerebrotendinous xanthomatosis	*CYP27A1*	AR	-	-	-	✓	✓	✓
Charcot-Marie-Tooth disease, GDAP1-related	*GDAP1*	AR	-	-	-	-	✓	✓
Charcot-Marie-Tooth disease, SH3TC2-related	*SH3TC2*	AR	-	-	-	-	✓	✓
Charcot-Marie-Tooth disease, SURF1-related	*SURF1*	AR	-	-	-	-	✓	✓
Charcot-Marie-Tooth disease, type 4B1	*MTMR2*	AR	-	-	-	-	✓	✓
Charcot-Marie-Tooth disease, type 4D	*NDRG1*	AR	-	-	-	-	✓	✓
Charcot-Marie-Tooth disease, X-linked type 1	*GJB1*	XL	-	-	-	-	✓	✓
Chediak-Higashi syndrome	*LYST*	AR	-	-	-	-	✓	✓
Childhood-onset severe retinal dystrophy, AIPL1-related	*AIPL1*	AR	-	-	-	-	✓	✓
Chondrodysplasia punctata type 1, X-linked	*ARSE*	XL	-	-	-	-	✓	✓
Choreoacanthocytosis	*VPS13A*	AR	-	-	✓	-	✓	✓
Choroideremia	*CHM*	XL	-	-	-	-	✓	✓
Chronic granulomatous disease	*CYBA*	AR	-	-	-	-	✓	✓
Chronic granulomatous disease, X-linked	*CYBB*	XL	-	-	-	-	✓	✓
Citrin deficiency	*SLC25A13*	AR	-	-	-	-	✓	✓
Citrullinemia	*ASS1*	AR	-	✓	-	-	✓	✓
Cockayne syndrome type A	*ERCC8*	AR	-	-	-	-	✓	✓
Cockayne syndrome type B; De Sanctis-Cacchione syndrome	*ERCC6*	AR	-	-	-	-	✓	✓
Cohen syndrome	*VPS13B*	AR	-	-	-	-	✓	✓
Combined malonic and methylmalonic aciduria	*ACSF3*	AR	-	-	-	-	✓	✓
Combined oxidative phosphorylation deficiency, GFM1-related	*GFM1*	AR	-	-	-	-	✓	✓
Combined oxidative phosphorylation deficiency, TSFM-related	*TSFM*	AR	-	-	-	-	✓	✓

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Combined pituitary hormone deficiency 2	*PROP1*	AR	-	-	-	-	✓	✓
Combined pituitary hormone deficiency 3	*LHX3*	AR	-	-	-	-	✓	✓
Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	*CYP11B1*	AR	-	-	-	-	✓	✓
Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	*CYP17A1*	AR	-	-	-	-	✓	✓
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	*CYP21A2*	AR	-	-	-	✓	✓	✓
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	*HSD3B2*	AR	-	-	-	-	✓	✓
Congenital adrenal hypoplasia, X-linked	*NR0B1*	XL	-	-	-	✓	✓	✓
Congenital amegakaryocytic thrombocytopenia	*MPL*	AR	-	-	✓	-	✓	✓
Congenital disorder of glycosylation type 1a	*PMM2*	AR	-	✓	✓	✓	✓	✓
Congenital disorder of glycosylation type Ib	*MPI*	AR	-	-	-	-	✓	✓
Congenital disorder of glycosylation type Ic	*ALG6*	AR	-	-	-	-	✓	✓
Congenital hydrocephalus 1	*CCDC88C*	AR	-	-	-	✓	✓	✓
Congenital hyperinsulinism; Permanent neonatal diabetes mellitus	*KCNJ11*	AR	-	-	-	-	✓	✓
Congenital hypothyroidism, TSHB-related	*TSHB*	AR	-	-	-	-	✓	✓
Congenital ichthyosis	*TGM1*	AR	-	-	-	-	✓	✓
Congenital insensitivity to pain with anhidrosis	*NTRK1*	AR	-	-	-	-	✓	✓
Congenital myasthenic syndrome, CHRNE-related	*CHRNE*	AR	-	-	-	✓	✓	✓
Congenital myasthenic syndrome, RAPSN-related; Fetal akinesia deformation sequence	*RAPSN*	AR	-	-	-	-	✓	✓
Congenital nephrotic syndrome, type 1	*NPHS1*	AR	-	-	-	✓	✓	✓
Congenital nephrotic syndrome, type 2	*NPHS2*	AR	-	-	-	-	✓	✓
Congenital secretory chloride diarrhea	*SLC26A3*	AR	-	-	-	-	✓	✓
Congenital hypothyroidism, DUOX2-related	*DUOX2*	AR	-	-	-	-	✓	✓
Congenital hypothyroidism, DUOXA2-related	*DUOXA2*	AR	-	-	-	-	✓	✓
Corneal endothelial dystrophy	*SLC4A11*	AR	-	-	-	-	✓	✓
Corticosterone methyloxidase deficiency	*CYP11B2*	AR	-	-	-	-	✓	✓
Costeff syndrome	*OPA3*	AR	-	-	-	-	✓	✓
Creatine deficiency syndrome	*SLC6A8*	XL	-	-	-	✓	✓	✓

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Crigler-Najjar syndrome	*UGT1A1*	AR	-	-	-	-	✓	✓
Cystic fibrosis	*CFTR*	AR	✓	✓	✓	✓	✓	✓
Cystinosis	*CTNS*	AR	-	-	-	-	✓	✓
D-bifunctional protein deficiency	*HSD17B4*	AR	-	-	-	-	✓	✓
Dent disease 2; Lowe syndrome	*OCRL*	XL	-	-	-	-	✓	✓
Dihydrolipoamide dehydrogenase deficiency	*DLD*	AR	-	-	-	✓	✓	✓
Dihydropyrimidine dehydrogenase deficiency	*DPYD*	AR	-	-	✓	-	✓	✓
Donnai-Barrow syndrome; Faciooculoacousticorenal syndrome	*LRP2*	AR	-	-	-	✓	✓	✓
Duchenne muscular dystrophy	*DMD*	XL	-	✓	-	✓	✓	✓
Dyskeratosis congenita type 5	*RTEL1*	AR	-	-	✓	-	✓	✓
Dystrophic epidermolysis bullosa	*COL7A1*	AR	-	-	-	✓	✓	✓
Ehlers-Danlos syndrome with kyphoscoliosis, PLOD1-related	*PLOD1*	AR	-	-	-	-	✓	✓
Ehlers-Danlos syndrome, autosomal recessive, due to tenascin X deficiency	*TNXB*	AR	-	-	-	✓	✓	✓
Ehlers-Danlos syndrome, Dermatosparaxis type VIIC	*ADAMTS2*	AR	-	-	✓	-	✓	✓
Ellis-van Creveld syndrome, EVC-related; Weyers acrofacial dysostosis, EVC-related	*EVC*	AR	-	-	-	-	✓	✓
Ellis-van Creveld syndrome, EVC2-related; Weyers acrodental dysostosis, EVC2-related	*EVC2*	AR	-	-	-	✓	✓	✓
Emery-Dreifuss muscular dystrophy	*EMD*	XL	-	-	-	-	✓	✓
Enhanced S-cone syndrome; Retinitis pigmentosa 37	*NR2E3*	AR	-	-	✓	-	✓	✓
Ethylmalonic encephalopathy	*ETHE1*	AR	-	-	-	-	✓	✓
Fabry disease	*GLA*	XL	-	-	-	✓	✓	✓
Factor V deficiency	*F5*	AR	-	-	-	-	-	✓
Factor XI deficiency	*F11*	AR	-	-	✓	-	✓	✓
Familial dysautonomia	*ELP1 (IKBKAP)*	AR	-	✓	✓	✓	✓	✓
Familial hypercholesterolemia	*LDLRAP1*	AR	-	-	-	-	-	✓
Familial hyperinsulinism, ABCC8-related	*ABCC8*	AR	-	-	✓	✓	✓	✓
Familial lipoprotein lipase deficiency	*LPL*	AR	-	-	-	-	-	✓
Familial Mediterranean fever	*MEFV*	AR	-	-	✓	-	✓	✓
Fanconi anemia group A	*FANCA*	AR	-	-	-	-	✓	✓
Fanconi anemia group C	*FANCC*	AR	-	✓	✓	✓	✓	✓
Fanconi anemia group G	*FANCG*	AR	-	-	-	-	✓	✓
Fragile X syndrome	*FMR1*	XL	✓	✓	✓	✓	✓	✓
Fraser syndrome	*GRIP1*	AR	-	-	-	✓	✓	✓

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Friedreich ataxia	*FXN*	AR	-	-	-	✓	✓	✓
Fumarase deficiency	*FH*	AR	-	-	-	-	✓	✓
Galactokinase deficiency	*GALK1*	AR	-	-	-	-	✓	✓
Galactose epimerase deficiency	*GALE*	AR	-	-	-	-	✓	✓
Galactosemia	*GALT*	AR	-	✓	✓	✓	✓	✓
Gaucher disease	*GBA*	AR	-	✓	✓	✓	✓	✓
Gitelman syndrome	*SLC12A3*	AR	-	-	-	-	✓	✓
Glucose-6-phosphate dehydrogenase deficiency	*G6PD*	XL	-	-	-	-	-	✓
Glutamate formiminotransferase deficiency	*FTCD*	AR	-	-	-	-	✓	✓
Glutaric aciduria IIA	*ETFA*	AR	-	-	-	-	✓	✓
Glutaric aciduria IIB	*ETFB*	AR	-	-	-	-	✓	✓
Glutaric aciduria IIC	*ETFDH*	AR	-	-	-	-	✓	✓
Glutaric aciduria, type I	*GCDH*	AR	-	-	-	-	✓	✓
Glycine encephalopathy, AMT-related	*AMT*	AR	-	-	-	-	✓	✓
Glycine encephalopathy, GLDC-related	*GLDC*	AR	-	-	-	-	✓	✓
Glycogen storage disease IV	*GBE1*	AR	-	-	✓	✓	✓	✓
Glycogen storage disease type III	*AGL*	AR	-	-	-	-	✓	✓
Glycogen storage disease type V	*PYGM*	AR	-	-	-	-	✓	✓
Glycogen storage disease VII	*PFKM*	AR	-	-	✓	-	✓	✓
Glycogen Storage disease, type 1a	*G6PC*	AR	-	✓	✓	✓	✓	✓
Glycogen storage disease, type Ib	*SLC37A4*	AR	-	-	-	✓	✓	✓
Guanidinoacetate methyltransferase deficiency	*GAMT*	AR	-	-	-	-	✓	✓
Gyrate atrophy of choroid and retina	*OAT*	AR	-	-	-	-	✓	✓
Hartnup disorder	*SLC6A19*	AR	-	-	-	-	✓	✓
Hemochromatosis, HFE-related	*HFE*	AR	-	-	-	-	-	✓
Hemochromatosis, type 2A	*HJV (HFE2)*	AR	-	-	-	-	✓	✓
Hemochromatosis, type 3	*TFR2*	AR	-	-	-	-	✓	✓
Hemophagocytic lymphohistiocytosis, familial, 2	*PRF1*	AR	-	-	-	✓	✓	✓
Hemophilia A	*F8*	XL	-	-	-	✓	✓	✓
Hemophilia B	*F9*	XL	-	-	-	✓	✓	✓
Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	*MPV17*	AR	-	-	-	-	✓	✓
Hereditary folate malabsorption	*SLC46A1*	AR	-	-	-	-	✓	✓
Hereditary fructose intolerance	*ALDOB*	AR	-	-	-	✓	✓	✓
Hermansky-Pudlak syndrome 1	*HPS1*	AR	-	-	-	✓	✓	✓
Hermansky-Pudlak syndrome 3	*HPS3*	AR	-	-	✓	✓	✓	✓

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Holocarboxylase synthetase deficiency	*HLCS*	AR	-	-	-	-	✓	✓
Homocystinuria due to cystathionine beta-synthase deficiency	*CBS*	AR	-	-	-	-	✓	✓
Homocystinuria-megaloblastic anemia, cobalamin E type	*MTRR*	AR	-	-	-	✓	✓	✓
Homocystinuria, MTHFR-related	*MTHFR*	AR	-	-	-	-	-	✓
Hydrolethalus syndrome	*HYLS1*	AR	-	-	-	-	✓	✓
Hyper IgM syndrome, X-linked	*CD40LG*	XL	-	-	-	-	✓	✓
Hyperimmunoglobulinemia D syndrome	*MVK*	AR	-	-	-	✓	✓	✓
Hypermethioninemia due to adenosine kinase deficiency	*ADK*	AR	-	-	-	-	✓	✓
Hypermethioninemia due to deficiency of S-adenosylhomocysteine hydrolase	*AHCY*	AR	-	-	-	-	✓	✓
Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	*SLC25A15*	AR	-	-	-	-	✓	✓
Hyperprolinemia type II	*ALDH4A1*	AR	-	-	-	-	✓	✓
Hypogonadotropic hypogonadism, GNRHR-related	*GNRHR*	AR	-	-	-	-	✓	✓
Hypohidrotic ectodermal dysplasia	*EDA*	XL	-	-	-	-	✓	✓
Hypophosphatasia	*ALPL*	AR	-	-	-	✓	✓	✓
Inclusion body myopathy type 2 (Nonaka myopathy)	*GNE*	AR	-	-	-	-	✓	✓
Infantile neuroaxonal dystrophy	*PLA2G6*	AR	-	-	-	-	✓	✓
Isovaleric acidemia	*IVD*	AR	-	✓	-	-	✓	✓
Joubert syndrome 2; Meckel syndrome 2	*TMEM216*	AR	-	-	✓	✓	✓	✓
Joubert syndrome 28; Meckel syndrome 1; Bardet-Biedl syndrome 13	*MKS1*	AR	-	-	-	-	✓	✓
Joubert syndrome 4; Senior-Løken syndrome 1; Nephronophthisis	*NPHP1*	AR	-	-	-	-	✓	✓
Joubert syndrome 9; Meckel syndrome 6; COACH syndrome	*CC2D2A*	AR	-	-	-	✓	✓	✓
Joubert syndrome, AHI1-related	*AHI1*	AR	-	-	-	✓	✓	✓
Joubert syndrome, ARL13B-related	*ARL13B*	AR	-	-	-	-	✓	✓
Junctional epidermolysis bullosa, LAMA3-related; Laryngo-onycho-cutaneous syndrome	*LAMA3*	AR	-	-	-	-	✓	✓
Junctional epidermolysis bullosa, LAMB3-related	*LAMB3*	AR	-	-	-	-	✓	✓
Junctional epidermolysis bullosa, LAMC2-related	*LAMC2*	AR	-	-	-	-	✓	✓
Juvenile retinoschisis, X-linked	*RS1*	XL	-	-	-	✓	✓	✓
Krabbe disease	*GALC*	AR	-	-	-	-	✓	✓

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L1 syndrome	*L1CAM*	XL	-	-	-	✓	✓	✓
Leber congenital amaurosis 2; Retinitis pigmentosa 20	*RPE65*	AR	-	-	-	-	✓	✓
Leber congenital amaurosis 5	*LCA5*	AR	-	-	-	-	✓	✓
Leber congenital amaurosis 8; Retinitis pigmentosa 12	*CRB1*	AR	-	-	-	-	✓	✓
Leber congenital amaurosis type 13	*RDH12*	AR	-	-	-	-	✓	✓
Leigh syndrome with Complex IV deficiency	*LRPPRC*	AR	-	-	-	-	✓	✓
Lethal congenital contracture syndrome 1	*GLE1*	AR	-	-	-	-	✓	✓
Leukoencephalopathy with vanishing white matter	*EIF2B5*	AR	-	-	-	-	✓	✓
Limb-girdle muscular dystrophy type 2A	*CAPN3*	AR	-	-	-	-	✓	✓
Limb-girdle muscular dystrophy type 2B	*DYSF*	AR	-	-	-	-	✓	✓
Limb-girdle muscular dystrophy, type 2C	*SGCG*	AR	-	-	-	-	✓	✓
Limb-girdle muscular dystrophy, type 2D	*SGCA*	AR	-	-	-	-	✓	✓
Limb-girdle muscular dystrophy, type 2E	*SGCB*	AR	-	-	-	-	✓	✓
Limb-girdle muscular dystrophy, type 2F	*SGCD*	AR	-	-	-	-	✓	✓
Limb-girdle muscular dystrophy, type 2H; Bardet-Biedl syndrome 11	*TRIM32*	AR	-	-	-	-	✓	✓
Lipoid congenital adrenal hyperplasia	*STAR*	AR	-	-	-	-	✓	✓
Lissencephaly, X-linked	*DCX*	XL	-	-	-	-	✓	✓
Liver failure, acute infantile	*TRMU*	AR	-	-	-	-	✓	✓
Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency; Trifunctional protein deficiency	*HADHA*	AR	-	-	-	-	✓	✓
Lujan-Fryns syndrome, UPF3B-related	*UPF3B*	XL	-	-	-	-	✓	✓
Lujan-Fryns syndrome, ZDHHC9-related	*ZDHHC9*	XL	-	-	-	-	✓	✓
Lysinuric protein intolerance	*SLC7A7*	AR	-	-	-	-	✓	✓
Lysosomal acid lipase deficiency	*LIPA*	AR	-	-	-	-	✓	✓
Macular corneal dystrophy, CHST6-related	*CHST6*	AR	-	-	-	-	✓	✓
Maple syrup urine disease type Ia	*BCKDHA*	AR	-	-	✓	-	✓	✓
Maple syrup urine disease type Ib	*BCKDHB*	AR	-	-	✓	✓	✓	✓
Maple syrup urine disease, type II	*DBT*	AR	-	-	-	-	✓	✓
Meckel syndrome 5; Joubert syndrome 7; COACH syndrome	*RPGRIP1L*	AR	-	-	-	-	✓	✓
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	*ACADM*	AR	-	✓	-	✓	✓	✓

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Megalencephalic leukoencephalopathy with subcortical cysts	*MLC1*	AR	-	-	-	✓	✓	✓
Menkes disease	*ATP7A*	XL	-	-	-	-	✓	✓
Mental retardation, X-linked, associated with fragile site FRAXE	*AFF2*	XL	-	-	-	✓	✓	✓
Metachromatic leukodystrophy	*ARSA*	AR	-	-	-	✓	✓	✓
Metachromatic leukodystrophy due to saposin-b deficiency	*PSAP*	AR	-	-	-	-	✓	✓
Methylmalonic acidemia, MUT-related	*MUT*	AR	-	-	-	✓	✓	✓
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC*	AR	-	✓	-	✓	✓	✓
Methylmalonic aciduria and homocystinuria, cblD type	*MMADHC*	AR	-	-	-	-	✓	✓
Methylmalonic aciduria and homocystinuria, cblF type	*LMBRD1*	AR	-	-	-	-	✓	✓
Methylmalonic aciduria and homocystinuria, cblJ type	*ABCD4*	AR	-	-	-	-	✓	✓
Methylmalonic aciduria, cblA type	*MMAA*	AR	-	-	-	-	✓	✓
Methylmalonic aciduria, cblB type	*MMAB*	AR	-	-	-	-	✓	✓
Methylmalonyl-CoA epimerase deficiency	*MCEE*	AR	-	-	-	-	✓	✓
Microcephaly, primary autosomal recessive, 1	*MCPH1*	AR	-	-	-	✓	✓	✓
Microphthalmia with or without coloboma	*VSX2*	AR	-	-	-	-	✓	✓
Microphthalmia, isolated 3	*RAX*	AR	-	-	-	-	✓	✓
Mitochondrial complex I deficiency (Leigh syndrome), NDUFAF5-related	*NDUFAF5*	AR	-	-	-	-	✓	✓
Mitochondrial complex I deficiency (Leigh syndrome), NDUFS6-related	*NDUFS6*	AR	-	-	✓	-	✓	✓
Mitochondrial complex IV deficiency, nuclear type 2	*SCO2*	AR	-	-	-	✓	✓	✓
Mitochondrial myopathy and sideroblastic anemia 1	*PUS1*	AR	-	-	-	-	✓	✓
Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	*TYMP*	AR	-	-	-	-	✓	✓
Mucolipidosis III alpha/beta; Mucolipidosis II alpha/beta	*GNPTAB*	AR	-	-	-	✓	✓	✓
Mucolipidosis III gamma	*GNPTG*	AR	-	-	-	-	✓	✓
Mucolipidosis IV	*MCOLN1*	AR	-	-	✓	✓	✓	✓
Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	*SGSH*	AR	-	-	-	-	✓	✓
Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	*GNS*	AR	-	-	-	-	✓	✓
Mucopolysaccharidosis IVA (Morquio syndrome A)	*GALNS*	AR	-	-	-	-	✓	✓
Mucopolysaccharidosis type II (Hunter syndrome)	*IDS*	XL	-	-	-	-	✓	✓

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Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	*NAGLU*	AR	-	-	-	-	✓	✓
Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	*HGSNAT*	AR	-	-	-	-	✓	✓
Mucopolysaccharidosis type IVB (Morquio syndrome B); GM1-gangliosidosis	*GLB1*	AR	-	-	-	-	✓	✓
Mucopolysaccharidosis type IX	*HYAL1*	AR	-	-	-	-	✓	✓
Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	*ARSB*	AR	-	-	-	-	✓	✓
Mucopolysaccharidosis type VII	*GUSB*	AR	-	-	-	-	✓	✓
Mucopolysaccharidosis, type I (Hurler syndrome)	*IDUA*	AR	-	✓	-	✓	✓	✓
Multiple pterygium syndrome	*CHRNG*	AR	-	-	-	-	✓	✓
Multiple sulfatase deficiency	*SUMF1*	AR	-	-	✓	-	✓	✓
Muscular dystrophy-dystroglycanopathy, FKRP-related	*FKRP*	AR	-	-	-	✓	✓	✓
Muscular dystrophy-dystroglycanopathy, FKTN-related; Fukuyama congenital muscular dystrophy	*FKTN*	AR	-	-	✓	✓	✓	✓
Muscular dystrophy-dystroglycanopathy, POMT1-related	*POMT1*	AR	-	-	-	-	✓	✓
Muscular dystrophy-dystroglycanopathy, POMT2-related	*POMT2*	AR	-	-	-	-	✓	✓
Muscular dystrophy-dystroglycanopathy; Retinitis pigmentosa 76	*POMGNT1*	AR	-	-	-	-	✓	✓
Muscular dystrophy, LAMA2-related	*LAMA2*	AR	-	-	-	-	✓	✓
Myotonia congenita, autosomal dominant; Myotonia congenita, autosomal recessive, Myotonia levior	*CLCN1*	AR	-	-	-	✓	✓	✓
Myotubular myopathy, X-linked	*MTM1*	XL	-	-	-	-	✓	✓
N-acetylglutamate synthase deficiency	*NAGS*	AR	-	-	-	-	✓	✓
Nemaline myopathy	*NEB*	AR	-	-	✓	✓	✓	✓
Nephrogenic diabetes insipidus	*AQP2*	AR	-	-	-	-	✓	✓
Neuronal ceroid lipofuscinosis, CLN3-related	*CLN3*	AR	-	✓	-	-	✓	✓
Neuronal ceroid lipofuscinosis, CLN5-related	*CLN5*	AR	-	-	-	-	✓	✓
Neuronal ceroid lipofuscinosis, CLN6-related	*CLN6*	AR	-	-	-	-	✓	✓
Neuronal ceroid lipofuscinosis, CLN8-related	*CLN8*	AR	-	-	-	-	✓	✓
Neuronal ceroid lipofuscinosis, MFSD8-related	*MFSD8*	AR	-	-	-	-	✓	✓
Neuronal ceroid lipofuscinosis, PPT1-related	*PPT1*	AR	-	-	-	-	✓	✓

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Neuronal ceroid lipofuscinosis, TPP1-related	*TPP1*	AR	-	-	-	-	✓	✓
Niemann-Pick disease, type A/B	*SMPD1*	AR	-	✓	✓	✓	✓	✓
Niemann-Pick disease, type C1	*NPC1*	AR	-	-	-	-	✓	✓
Niemann-Pick disease, type C2	*NPC2*	AR	-	-	-	-	✓	✓
Nijmegen breakage syndrome	*NBN*	AR	-	-	-	-	✓	✓
Non-syndromic hearing loss, MYO7A-related; Usher syndrome, type 1B	*MYO7A*	AR	-	-	-	-	✓	✓
Non-syndromic hearing loss, PCDH15-related; Usher syndrome, type 1F	*PCDH15*	AR/Digenic	-	-	✓	✓	✓	✓
Non-syndromic hearing loss, USH1C-related; Usher syndrome, type IC	*USH1C*	AR	-	-	-	-	✓	✓
Nonsyndromic hearing loss, GJB2-related	*GJB2*	AR	-	-	✓	✓	✓	✓
Nonsyndromic hearing loss, GJB6-related	*GJB6*	AR	-	-	-	-	✓	✓
Nonsyndromic hearing loss, LOXHD1-related	*LOXHD1*	AR	-	-	✓	-	✓	✓
Nonsyndromic hearing loss, OTOF-related	*OTOF*	AR	-	-	-	-	✓	✓
Norrie disease	*NDP*	XL	-	-	-	-	✓	✓
Omenn syndrome, RAG1-related	*RAG1*	AR	-	-	-	-	✓	✓
Omenn syndrome, RAG2-related	*RAG2*	AR	-	-	-	-	✓	✓
Opitz GBBB syndrome, type I	*MID1*	XL	-	-	-	✓	✓	✓
Ornithine transcarbamylase deficiency	*OTC*	XL	-	-	-	✓	✓	✓
Osteogenesis imperfecta, type VIII	*P3H1*	AR	-	-	-	-	✓	✓
Osteopetrosis, TCIRG1-related	*TCIRG1*	AR	-	-	✓	-	✓	✓
Pantothenate kinase-associated neurodegeneration	*PANK2*	AR	-	-	-	-	✓	✓
Pendred syndrome	*SLC26A4*	AR	-	-	-	✓	✓	✓
Peroxisomal acyl-CoA oxidase deficiency	*ACOX1*	AR	-	-	-	-	✓	✓
Phenylalanine hydroxylase deficiency (Phenylketonuria)	*PAH*	AR	✓	✓	✓	✓	✓	✓
Phosphoglycerate dehydrogenase deficiency	*PHGDH*	AR	-	-	✓	-	✓	✓
Phosphoglycerate kinase 1 deficiency	*PGK1*	AR	-	-	-	-	✓	✓
Polycystic kidney disease, PKHD1-related	*PKHD1*	AR	-	✓	✓	✓	✓	✓
Pompe disease	*GAA*	AR	-	✓	✓	✓	✓	✓
Pontocerebellar hypoplasia type 1A	*VRK1*	AR	-	-	✓	-	✓	✓
Pontocerebellar hypoplasia type 1B	*EXOSC3*	AR	-	-	-	-	✓	✓
Pontocerebellar hypoplasia type 6	*RARS2*	AR	-	-	-	✓	✓	✓

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Pontocerebellar hypoplasia, type 2D	*SEPSECS*	AR	-	-	-	-	✓	✓
Postnatal progressive microcephaly with seizures and brain atrophy	*MED17*	AR	-	-	-	-	✓	✓
Primary ciliary dyskinesia, DNAH5-related	*DNAH5*	AR	-	-	✓	-	✓	✓
Primary ciliary dyskinesia, DNAI1-related	*DNAI1*	AR	-	-	✓	-	✓	✓
Primary ciliary dyskinesia, DNAI2-related	*DNAI2*	AR	-	-	✓	-	✓	✓
Primary ciliary dyskinesia, DNAL1-related	*DNAL1*	AR	-	-	-	-	✓	✓
Primary ciliary dyskinesia, type 14	*CCDC39*	AR	-	-	-	-	✓	✓
Primary ciliary dyskinesia, type 17	*CCDC103*	AR	-	-	-	-	✓	✓
Primary ciliary dyskinesia, type 30	*CCDC151*	AR	-	-	-	-	✓	✓
Primary congenital glaucoma	*CYP1B1*	AR	-	-	-	-	✓	✓
Primary hyperoxaluria type 1	*AGXT*	AR	-	-	-	✓	✓	✓
Primary Hyperoxaluria type II	*GRHPR*	AR	-	-	-	-	✓	✓
Primary hyperoxaluria type III	*HOGA1*	AR	-	-	✓	-	✓	✓
Progressive external ophthalmoplegia; Alpers-Huttenlocher syndrome; Ataxia neuropathy spectrum; Myocerebrohepatopathy syndrome	*POLG*	AR	-	-	-	✓	✓	✓
Progressive Familial Intrahepatic Cholestasis	*ABCB11*	AR	-	-	-	-	✓	✓
Propionic acidemia, PCCA-related	*PCCA*	AR	-	-	-	-	✓	✓
Propionic acidemia, PCCB-related	*PCCB*	AR	-	-	-	-	✓	✓
Prothrombin-related conditions	*F2*	AR	-	-	-	-	-	✓
Pycnodysostosis	*CTSK*	AR	-	-	-	-	✓	✓
Pyruvate carboxylase deficiency	*PC*	AR	-	-	-	-	✓	✓
Pyruvate dehydrogenase E1-alpha deficiency	*PDHA1*	XL	-	-	-	-	✓	✓
Pyruvate dehydrogenase E1-beta deficiency	*PDHB*	AR	-	-	-	-	✓	✓
Renal tubular acidosis with deafness	*ATP6V1B1*	AR	-	-	-	-	✓	✓
Renpenning syndrome	*PQBP1*	XL			-	-	✓	✓
Retinitis pigmentosa 25	*EYS*	AR	-	-	-	-	✓	✓
Retinitis pigmentosa 26	*CERKL*	AR	-	-	-	-	✓	✓
Retinitis pigmentosa 28	*FAM161A*	AR	-	-	✓	-	✓	✓
Retinitis pigmentosa 59	*DHDDS*	AR	-	-	✓	✓	✓	✓
Retinitis Pigmentosa, CNGA1-related	*CNGA1*	AR	-	-	-	-	✓	✓
Retinitis Pigmentosa, CNGB1-related	*CNGB1*	AR	-	-	-	-	✓	✓
Retinitis pigmentosa, IDH3B-related	*IDH3B*	AR	-	-	-	-	✓	✓

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Retinitis pigmentosa, PDE6A-related	*PDE6A*	AR	-	-	-	-	✓	✓
Rhizomelic chondrodysplasia punctata, type 1	*PEX7*	AR	✓	✓	-	-	✓	✓
Rhizomelic chondrodysplasia punctata, type 3	*AGPS*	AR	-	-	-	-	✓	✓
Roberts syndrome	*ESCO2*	AR	-	-	-	-	✓	✓
Sandhoff disease	*HEXB*	AR	-	-	-	-	✓	✓
Schimke immunoosseous dysplasia	*SMARCAL1*	AR	-	-	-	-	✓	✓
Schindler disease type I; Schindler disease type III	*NAGA*	AR	-	-	-	✓	✓	✓
Schopf-Schulz-Passarge syndrome; Odontoonychodermal dysplasia	*WNT10A*	AR	-	-	-	-	✓	✓
Segawa syndrome	*TH*	AR	-	-	-	-	✓	✓
Severe combined immunodeficiency with sensitivity to ionizing radiation	*DCLRE1C*	AR	-	-	-	-	✓	✓
Severe combined immunodeficiency, JAK3-related	*JAK3*	AR	-	-	-	-	✓	✓
Severe combined immunodeficiency, X-linked	*IL2RG*	XL	-	-	-	-	✓	✓
Severe Congenital Neutropenia, HAX1-related	*HAX1*	AR	-	-	-	-	✓	✓
Severe congenital neutropenia, VPS45-related	*VPS45*	AR	-	-	-	-	✓	✓
Short branched chain acyl-CoA dehydrogenase (SBCAD) deficiency	*ACADSB*	AR	-	-	-	-	✓	✓
Short-chain acyl-coA dehydrogenase (SCAD) Deficiency	*ACADS*	AR	-	-	-	-	✓	✓
Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*	AR	-	-	-	✓	✓	✓
Sialic acid storage disorder	*SLC17A5*	AR	-	-	-	-	✓	✓
Sickle cell disease; Beta thalassemia	*HBB*	AR	✓	✓	✓	✓	✓	✓
Sjögren-Larsson syndrome	*ALDH3A2*	AR	-	-	-	-	✓	✓
Smith-Lemli-Opitz syndrome	*DHCR7*	AR	-	-	✓	✓	✓	✓
Spastic paraplegia 15	*ZFYVE26*	AR	-	-	-	-	✓	✓
Spastic paraplegia 2, X-linked (SPG2)	*PLP1*	XL	-	-	-	✓	✓	✓
Spastic paraplegia 49	*TECPR2*	AR	-	-	-	-	✓	✓
Spastic paraplegia type 7	*SPG7*	AR	-	-	-	-	✓	✓
SPG11-related Neuromuscular Disorders	*SPG11*	AR	-	-	-	-	✓	✓
Spinal muscular atrophy	*SMN1*	AR	✓	✓	✓	✓	✓	✓
Spinocerebellar ataxia, autosomal recessive 10	*ANO10*	AR	-	-	-	✓	✓	✓
Spondylocostal dysostosis	*MESP2*	AR	-	-	-	-	✓	✓

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Steel syndrome	*COL27A1*	AR	-	-	-	-	✓	✓
Stuve-Wiedemann syndrome	*LIFR*	AR	-	-	-	-	✓	✓
Surfactant metabolism dysfunction, pulmonary, 3; Interstitial lung disease	*ABCA3*	AR	-	-	-	✓	✓	✓
Systemic primary carnitine deficiency	*SLC22A5*	AR	-	-	-	-	✓	✓
Tay-Sachs disease	*HEXA*	AR	-	✓	✓	✓	✓	✓
Tetrahydrobiopterin deficiency	*PTS*	AR	-	-	-	-	✓	✓
Tetrahydrobiopterin deficiency, PCBD1-related	*PCBD1*	AR	-	-	-	-	✓	✓
Tetrahydrobiopterin deficiency, QDPR-related	*QDPR*	AR	-	-	-	-	✓	✓
Thiamine metabolism dysfunction syndrome 2 (biotin or thiamine responsive)	*SLC19A3*	AR	-	-	-	✓	✓	✓
Thyroid dyshormonogenesis, IYD-related	*IYD*	AR			-	-	✓	✓
Thyroid dyshormonogenesis, SLC5A5-related	*SLC5A5*	AR	-	-	-	-	✓	✓
Thyroid dyshormonogenesis, TG-related	*TG*	AR	-	-	-	-	✓	✓
Thyroid dyshormonogenesis, TPO-related	*TPO*	AR	-	-	-	-	✓	✓
Treacher Collins syndrome, POLR1C-related	*POLR1C*	AR			-	-	✓	✓
Trichohepatoenteric syndrome	*TTC37*	AR	-	-	-	-	✓	✓
Trichothiodystrophy 1, photosensitive; Xeroderma pigmentosum, group D	*ERCC2*	AR	-	-	-	✓	✓	✓
Trimethylaminuria	*FMO3*	AR	-	-	-	✓	✓	✓
Tyrosinemia, type 1	*FAH*	AR	✓	✓	✓	✓	✓	✓
Tyrosinemia, type II	*TAT*	AR	-	-	-	-	✓	✓
Usher syndrome type 2D	*WHRN*	AR	-	-	-	-	✓	✓
Usher syndrome type IG	*USH1G*	AR	-	-	-	-	✓	✓
Usher syndrome, type 1D	*CDH23*	AR/Digenic	-	-	-	-	✓	✓
Usher syndrome, type 2A	*USH2A*	AR	-	-	-	✓	✓	✓
Usher syndrome, type 3A	*CLRN1*	AR	-	-	✓	✓	✓	✓
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	*ACADVL*	AR	-	-	-	✓	✓	✓
Vitamin D-dependent rickets, type 1A	*CYP27B1*	AR	-	-	-	✓	✓	✓
Wilson disease	*ATP7B*	AR	-	-	✓	✓	✓	✓
Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked; Severe Congenital Neutropenia, WAS-related	*WAS*	XL	-	-	-	-	✓	✓
Wolcott-Rallison syndrome	*EIF2AK3*	AR	-	-	-	-	✓	✓
X-linked Aarskog-Scott syndrome	*FGD1*	XL	-	-	-	-	✓	✓

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† This gene count reflects testing for female patients. Male patients will not be screened for X-linked conditions.
‡ Please visit our website for ethnicity-specific analytical detection rates.

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Condition	Gene	Inheritance	ACOG/ ACMG (6 genes†)	Focus (30 genes†)	Ashkenzi Jewish (61 genes†)	ACMG Tier 3 (113 genes†)	Expanded (427 genes†)	Expanded Plus (436 genes†)
X-linked epilepsy with variable learning disabilities	*SYN1*	XL	-	-	-	-	✓	✓
X-linked hearing loss, POU3F4-related	*POU3F4*	XL	-	-	-	-	✓	✓
X-linked intellectual disability with cerebellar hypoplasia and distinctive facial appearance	*OPHN1*	XL	-	-	-	-	✓	✓
X-linked Intellectual disability, AP1S2-related	*AP1S2*	XL	-	-	-	-	✓	✓
X-linked intellectual disability, ARX-related	*ARX*	XL	-	-	-	✓	✓	✓
X-linked intellectual disability, BRWD3-related	*BRWD3*	XL	-	-	-	-	✓	✓
X-linked intellectual disability, CUL4B-related	*CUL4B*	XL	-	-	-	-	✓	✓
X-linked intellectual disability, DLG3-related	*DLG3*	XL	-	-	-	-	✓	✓
X-linked intellectual disability, FTSJ1-related	*FTSJ1*	XL	-	-	-	-	✓	✓
X-linked intellectual disability, IL1RAPL1-related	*IL1RAPL1*	XL	-	-	-	-	✓	✓
X-linked intellectual disability, KDM5C-related	*KDM5C*	XL	-	-	-	-	✓	✓
X-linked intellectual disability, PAK3-related	*PAK3*	XL	-	-	-	-	✓	✓
X-linked intellectual disability, Siderius type	*PHF8*	XL	-	-	-	-	✓	✓
X-linked Intellectual disability, THOC2-related	*THOC2*	XL	-	-	-	-	✓	✓
X-linked intellectual disability, ZNF711-related	*ZNF711*	XL	-	-	-	-	✓	✓
X-linked Ocular albinism, GPR143-related	*GPR143*	XL	-	-	-	-	✓	✓
X-linked Retinitis pigmentosa, RP2-related	*RP2*	XL	-	-	-	-	✓	✓
X-linked Retinitis pigmentosa, RPGR-related	*RPGR*	XL	-	-	-	✓	✓	✓
Xeroderma pigmentosum, group A	*XPA*	AR	-	-	-	-	✓	✓
Xeroderma pigmentosum, group C	*XPC*	AR	-	-	-	✓	✓	✓
Zellweger syndrome, PEX1-related	*PEX1*	AR	-	✓	-	-	✓	✓
Zellweger syndrome, PEX10-related	*PEX10*	AR	-	-	-	-	✓	✓
Zellweger syndrome, PEX12-related	*PEX12*	AR	-	-	-	-	✓	✓
Zellweger syndrome, PEX2-related	*PEX2*	AR	-	-	✓	-	✓	✓
Zellweger syndrome, PEX6-related	*PEX6*	AR	-	-	-	-	✓	✓

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