Beacon Carrier Screening Detecting 320+ Conditions

Coverage: 99% at 20x **Deletion/Duplication:** ≥ 2 exons **Turnaround Time**: 2 weeks*

Condition	Gene	Inheritance	Analytical Detection Rate‡	ACOG/ ACMG (6 genes†)	Focus (30 genes†)	Ashkenazi Jewish (43 genes†)	Expanded (326 genes†)	Expanded Plus (335 genes†)
Alpha thalassemia	HBA1/HBA2	AR	90%	✓	✓	✓	✓	✓
Cystic fibrosis	CFTR	AR	99%	✓	✓	✓	✓	✓
Fragile X syndrome	FMR1	XL	99%	✓	✓	✓	✓	✓
Sickle cell disease; Beta thalassemia	HBB	AR	95%	✓	✓	✓	✓	✓
Spinal muscular atrophy	SMN1	AR	91%	✓	✓	✓	✓	✓
Bloom syndrome	BLM	AR	87%	-	✓	✓	✓	✓
Canavan disease	ASPA	AR	97%	-	✓	✓	✓	✓
Congenital disorder of glycosylation type 1a	PMM2	AR	99%	-	✓	✓	✓	✓
Familial dysautonomia	ELP1 (IKBKAP)	AR	99%	-	✓	✓	✓	✓
Fanconi anemia group C	FANCC	AR	99%	-	✓	✓	✓	✓
Galactosemia	GALT	AR	95%	-	✓	✓	✓	✓
Gaucher disease	GBA	AR	99%	-	✓	✓	✓	✓
Glycogen storage disease, type 1a	G6PC	AR	95%	-	✓	✓	✓	✓
Niemann-Pick disease, type A/B	SMPD1	AR	95%	-	✓	✓	✓	✓
Polycystic kidney disease, PKHD1-related	PKHD1	AR	98%	-	✓	✓	✓	✓
Smith-Lemli-Opitz syndrome	DHCR7	AR	96%	-	✓	✓	✓	✓
Tay-Sachs disease	HEXA	AR	99%	-	✓	✓	✓	✓
Tyrosinemia, type 1	FAH	AR	95%	-	✓	✓	✓	✓
Citrullinemia	ASS1	AR	96%	-	✓	-	✓	✓
Duchenne muscular dystrophy	DMD	XL	93%	-	✓	-	✓	✓
Isovaleric acidemia	IVD	AR	90%	-	✓	-	✓	✓
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	ACADM	AR	98%	-	✓	-	✓	✓
Methylmalonic aciduria and homocystinuria, cblC type	MMACHC	AR	90%	-	✓	-	✓	✓
Mucopolysaccharidosis, type I (Hurler syndrome)	IDUA	AR	95%	-	✓	-	✓	✓
Neuronal ceroid lipofuscinosis, CLN3-related	CLN3	AR	98%	-	✓	-	✓	✓
Phenylalanine hydroxylase deficiency (Phenylketonuria)	PAH	AR	99%	-	✓	-	✓	✓
Pompe disease	GAA	AR	98%	-	✓	-	✓	✓
Rhizomelic chondrodysplasia punctata, type 1	PEX7	AR	99%	-	✓	-	✓	✓
Zellweger syndrome, PEX1-related	PEX1	AR	95%	-	✓	-	✓	✓

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Abetalipoproteinemia	MTTP	AR	98%	-	-	✓	✓	✓
Alport syndrome, COL4A3-related	COL4A3	AR	98%	-	-	✓	✓	✓
Arthrogryposis, mental retardation, and seizures	SLC35A3	AR	98%	-	-	✓	✓	✓
Bardet-Biedl syndrome 2; Retinitis Pigmentosa 74	BBS2	AR	99%	-	-	✓	✓	✓
Carnitine palmitoyltransferase II deficiency	CPT2	AR	95%	-	-	✓	✓	✓
Congenital amegakaryocytic thrombocytopenia	MPL	AR	98%	-	-	✓	✓	✓
Dihydrolipoamide dehydrogenase deficiency	DLD	AR	98%	-	-	✓	✓	✓
Dyskeratosis congenita type 5	RTEL1	AR	99%	-	-	✓	✓	✓
Ehlers-Danlos syndrome, Dermatosparaxis type VIIC	ADAMTS2	AR	98%	-	-	✓	✓	✓
Factor XI deficiency	F11	AR	98%	-	-	✓	✓	✓
Familial hyperinsulinism, ABCC8-related	ABCC8	AR	98%	-	-	✓	✓	✓
Joubert syndrome 2; Meckel syndrome 2	TMEM216	AR	98%	-	-	✓	✓	✓
Maple syrup urine disease type Ia	BCKDHA	AR	98%	-	-	✓	✓	✓
Maple syrup urine disease type Ib	BCKDHB	AR	98%	-	-	✓	✓	✓
Mucolipidosis IV	MCOLN1	AR	99%	-	-	✓	✓	✓
Multiple sulfatase deficiency	SUMF1	AR	98%	-	-	✓	✓	✓
Muscular dystrophy-dystroglycanopathy, FKTN-related; Fukuyama congenital muscular dystrophy	FKTN	AR	99%	-	-	✓	✓	✓
Nemaline myopathy	NEB	AR	98%	-	-	✓	✓	✓
Non-syndromic hearing loss, PCDH15-related; Usher syndrome, type 1F	PCDH15	AR/Digenic	98%	-	-	✓	✓	✓
Phosphoglycerate dehydrogenase deficiency	PHGDH	AR	98%	-	-	✓	✓	✓
Retinitis pigmentosa 59	DHDDS	AR	98%	-	-	✓	✓	✓
Usher syndrome, type 3A	CLRN1	AR	98%	-	-	✓	✓	✓
Wilson disease	ATP7B	AR	98%	-	-	✓	✓	✓
Zellweger syndrome, PEX2-related	PEX2	AR	95%	-	-	✓	✓	✓
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	HMGCL	AR	98%	-	-	-	✓	✓
3-Ketothiolase deficiency	ACAT1	AR	98%	-	-	-	✓	✓
3-Methylcrotonyl-CoA carboxylase 1 deficiency (3-MCC deficiency)	MCCC1	AR	98%	-	-	-	✓	✓
3-Methylcrotonyl-CoA carboxylase 2 deficiency (3-MCC deficiency)	MCCC2	AR	98%	-	-	-	✓	✓
Achondrogenesis, type IB; Atelosteogenesis II; Diastrophic dysplasia; Multiple epiphyseal dysplasia	SLC26A2	AR	90%	-	-	-	✓	✓
Achromatopsia	CNGB3	AR	99%	-	-	-	✓	✓
Acrodermatitis enteropathica	SLC39A4	AR	98%	-	-	-	✓	✓
Acyl-CoA dehydrogenase-9 (ACAD9) Deficiency	ACAD9	AR	98%	-	-	-	✓	✓
Adenosine deaminase deficiency	ADA	AR	93%	-	-	-	✓	✓
Adrenoleukodystrophy, X-linked	ABCD1	XL	99%	-	-	-	✓	✓
Aicardi-Goutieres syndrome	SAMHD1	AR	95%	-	-	-	✓	✓

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Alkaptonuria	HGD	AR	90%	-	-	-	✓	✓
Alpha thalassemia X-linked intellectual disability syndrome	ATRX	XL	99%	-	-	-	✓	✓
Alpha-mannosidosis	MAN2B1	AR	99%	-	-	-	✓	✓
Alport syndrome, COL4A4-related	COL4A4	AR	98%	-	-	-	✓	✓
Alport syndrome, COL4A5-related	COL4A5	XL	98%	-	-	-	✓	✓
Alstrom syndrome	ALMS1	AR	98%	-	-	-	✓	✓
Anauxetic dysplasia; Cartilage-hair hypoplasia; Metaphyseal dysplasia without hypotrichosis	RMRP	AR	99%	-	-	-	✓	✓
Andermann syndrome	SLC12A6	AR	98%	-	-	-	✓	✓
Arginase deficiency	ARG1	AR	98%	-	-	-	✓	✓
Argininosuccinate lyase deficiency	ASL	AR	90%	-	-	-	✓	✓
Aromatase deficiency	CYP19A1	AR	98%	-	-	-	✓	✓
Arts syndrome; Rosenberg-Chutorian syndrome; Phosphoribosylpyrophosphate synthetase superactivity; Non-syndromic hearing loss, PRPS1-related	PRPS1	XL	98%	-	-	-	✓	✓
Asparagine synthetase deficiency	ASNS	AR	98%	-	-	-	✓	✓
Aspartylglucosaminuria	AGA	AR	98%	-	-	-	✓	✓
Ataxia with isolated vitamin E deficiency	TTPA	AR	98%	-	-	-	✓	✓
Ataxia-telangiectasia	ATM	AR	92%	-	-	-	✓	✓
Autoimmune polyendocrinopathy syndrome type I	AIRE	AR	98%	-	-	-	✓	✓
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	SACS	AR	95%	-	-	-	✓	✓
Bardet-Biedl syndrome 14; Joubert syndrome 5; Leber congenital amaurosis 10; Meckel syndrome 4; Senior-Løken syndrome 6	CEP290	AR	98%	-	-	-	✓	✓
Bardet-Biedl syndrome type 1	BBS1	AR	99%	-	-	-	✓	✓
Bardet-Biedl syndrome type 10	BBS10	AR	99%	-	-	-	✓	✓
Bardet-Biedl syndrome type 12	BBS12	AR	99%	-	-	-	✓	✓
Bare lymphocyte syndrome, type II	CIITA	AR	98%	-	-	-	✓	✓
Bartter syndrome	BSND	AR	98%	-	-	-	✓	✓
Bernard-Soulier syndrome type A1	GP1BA	AR	98%	-	-	-	✓	✓
Bernard-Soulier syndrome type C	GP9	AR	98%	-	-	-	✓	✓
Bilateral frontoparietal polymicrogyria	ADGRG1	AR	98%	-	-	-	✓	✓
Biotinidase deficiency	BTD	AR	99%	-	-	-	✓	✓
Björnstad syndrome; GRACILE syndrome; Mitochondrial complex III deficiency	BCS1L	AR	98%	-	-	-	✓	✓
Carbamoylphosphate synthetase I deficiency	CPS1	AR	98%	-	-	-	✓	✓
Carnitine palmitoyltransferase IA deficiency	CPT1A	AR	90%	-	-	-	✓	✓
Carnitine-acylcarnitine translocase deficiency	SLC25A20	AR	98%	-	-	-	✓	✓
Carpenter syndrome	RAB23	AR	98%	-	-	-	✓	✓
Cerebrotendinous xanthomatosis	CYP27A1	AR	98%	-	-	-	✓	✓
Charcot-Marie-Tooth disease, type 4D	NDRG1	AR	98%	-	-	-	✓	✓

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Charcot-Marie-Tooth disease, X-linked type 1	GJB1	XL	90%	-	-	-	✓	✓
Chediak-Higashi syndrome	LYST	AR	90%	-	-	-	✓	✓
Chondrodysplasia punctata type 1, X-linked	ARSE	XL	98%	-	-	-	✓	✓
Choreoacanthocytosis	VPS13A	AR	98%	-	-	-	✓	✓
Choroideremia	CHM	XL	95%	-	-	-	✓	✓
Chronic granulomatous disease	CYBA	AR	99%	-	-	-	✓	✓
Chronic granulomatous disease, X-linked	CYBB	XL	99%	-	-	-	✓	✓
Citrin deficiency	SLC25A13	AR	95%	-	-	-	✓	✓
Cockayne syndrome type A	ERCC8	AR	98%	-	-	-	✓	✓
Cockayne syndrome type B; De Sanctis-Cacchione syndrome	ERCC6	AR	99%	-	-	-	✓	✓
Cohen syndrome	VPS13B	AR	98%	-	-	-	✓	✓
Combined malonic and methylmalonic aciduria	ACSF3	AR	98%	-	-	-	✓	✓
Combined oxidative phosphorylation deficiency, GFM1-related	GFM1	AR	98%	-	-	-	✓	✓
Combined oxidative phosphorylation deficiency, TSFM-related	TSFM	AR	98%	-	-	-	✓	✓
Combined pituitary hormone deficiency 2	PROP1	AR	98%	-	-	-	✓	✓
Combined pituitary hormone deficiency 3	LHX3	AR	98%	-	-	-	✓	✓
Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	CYP11B1	AR	98%	-	-	-	✓	✓
Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	CYP17A1	AR	98%	-	-	-	✓	✓
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2	AR	99%	-	-	-	✓	✓
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	HSD3B2	AR	98%	-	-	-	✓	✓
Congenital adrenal hypoplasia, X-linked	NR0B1	XL	99%	-	-	-	✓	✓
Congenital disorder of glycosylation type Ib	MPI	AR	98%	-	-	-	✓	✓
Congenital disorder of glycosylation type Ic	ALG6	AR	98%	-	-	-	✓	✓
Congenital hyperinsulinism; Permanent neonatal diabetes mellitus	KCNJ11	AR	99%	-	-	-	✓	✓
Congenital ichthyosis	TGM1	AR	95%	-	-	-	✓	✓
Congenital insensitivity to pain with anhidrosis	NTRK1	AR	99%	-	-	-	✓	✓
Congenital myasthenic syndrome, CHRNE-related	CHRNE	AR	99%	-	-	-	✓	✓
Congenital myasthenic syndrome, RAPSN-related; Fetal akinesia deformation sequence	RAPSN	AR	99%	-	-	-	✓	✓
Congenital nephrotic syndrome, type 1	NPHS1	AR	98%	-	-	-	✓	✓
Congenital nephrotic syndrome, type 2	NPHS2	AR	98%	-	-	-	✓	✓
Congenital secretory chloride diarrhea	SLC26A3	AR	98%	-	-	-	✓	✓
Corneal endothelial dystrophy	SLC4A11	AR	98%	-	-	-	✓	✓
Corticosterone methyloxidase deficiency	CYP11B2	AR	98%	-	-	-	✓	✓
Costeff syndrome	OPA3	AR	98%	-	-	-	✓	✓

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Creatine deficiency syndrome	SLC6A8	XL	98%	-	-	-	✓	✓
Crigler-Najjar syndrome	UGT1A1	AR	98%	-	-	-	✓	✓
Cystinosis	CTNS	AR	99%	-	-	-	✓	✓
D-bifunctional protein deficiency	HSD17B4	AR	98%	-	-	-	✓	✓
Dent disease 2; Lowe syndrome	OCRL	XL	95%	-	-	-	✓	✓
Dihydropyrimidine dehydrogenase deficiency	DPYD	AR	98%	-	-	-	✓	✓
Dystrophic epidermolysis bullosa	COL7A1	AR	97%	-	-	-	✓	✓
Ellis-van Creveld syndrome, EVC2-related; Weyers acrodental dysostosis, EVC2-related	EVC2	AR	98%	-	-	-	✓	✓
Ellis-van Creveld syndrome, EVC-related; Weyers acrofacial dysostosis, EVC-related	EVC	AR	98%	-	-	-	✓	✓
Emery-Dreifuss muscular dystrophy	EMD	XL	99%	-	-	-	✓	✓
Enhanced S-cone syndrome; Retinitis pigmentosa 37	NR2E3	AR	98%	-	-	-	✓	✓
Ethylmalonic encephalopathy	ETHE1	AR	98%	-	-	-	✓	✓
Fabry disease	GLA	XL	99%	-	-	-	✓	✓
Familial Mediterranean fever	MEFV	AR	99%	-	-	-	✓	✓
Fanconi anemia group A	FANCA	AR	98%	-	-	-	✓	✓
Fanconi anemia group G	FANCG	AR	90%	-	-	-	✓	✓
Fumarase deficiency	FH	AR	90%	-	-	-	✓	✓
Galactokinase deficiency	GALK1	AR	95%	-	-	-	✓	✓
Gitelman syndrome	SLC12A3	AR	98%	-	-	-	✓	✓
Glutaric aciduria IIA	ETFA	AR	98%	-	-	-	✓	✓
Glutaric aciduria IIB	ETFB	AR	98%	-	-	-	✓	✓
Glutaric aciduria IIC	ETFDH	AR	98%	-	-	-	✓	✓
Glutaric aciduria, type I	GCDH	AR	98%	-	-	-	✓	✓
Glycine encephalopathy, AMT-related	AMT	AR	98%	-	-	-	✓	✓
Glycine encephalopathy, GLDC-related	GLDC	AR	98%	-	-	-	✓	✓
Glycogen storage disease IV	GBE1	AR	99%	-	-	-	✓	✓
Glycogen storage disease type III	AGL	AR	95%	-	-	-	✓	✓
Glycogen storage disease type V	PYGM	AR	99%	-	-	-	✓	✓
Glycogen storage disease VII	PFKM	AR	98%	-	-	-	✓	✓
Glycogen storage disease, type Ib	SLC37A4	AR	95%	-	-	-	✓	✓
Guanidinoacetate methyltransferase deficiency	GAMT	AR	99%	-	-	-	✓	✓
Gyrate atrophy of choroid and retina	OAT	AR	98%	-	-	-	✓	✓
Hemochromatosis, type 2A	HJV (HFE2)	AR	99%	-	-	-	✓	✓
Hemochromatosis, type 3	TFR2	AR	98%	-	-	-	✓	✓
Hemophilia A	F8	XL	48%	-	-	-	✓	✓
Hemophilia B	F9	XL	99%	-	-	-	✓	✓
Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	MPV17	AR	96%	-	-	-	✓	✓
Hereditary fructose intolerance	ALDOB	AR	99%	-	-	-	✓	✓
Hermansky-Pudlak syndrome 1	HPS1	AR	98%	-	-	-	✓	✓

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Hermansky-Pudlak syndrome 3	HPS3	AR	98%	-	-	-	✓	✓
Holocarboxylase synthetase deficiency	HLCS	AR	98%	-	-	-	✓	✓
Homocystinuria due to cystathionine beta-synthase deficiency	CBS	AR	99%	-	-	-	✓	✓
Homocystinuria-megaloblastic anemia, cobalamin E type	MTRR	AR	98%	-	-	-	✓	✓
Hydrolethalus syndrome	HYLS1	AR	98%	-	-	-	✓	✓
Hyper IgM syndrome, X-linked	CD40LG	XL	98%	-	-	-	✓	✓
Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	SLC25A15	AR	99%	-	-	-	✓	✓
Hypohidrotic ectodermal dysplasia	EDA	XL	99%	-	-	-	✓	✓
Hypophosphatasia	ALPL	AR	95%	-	-	-	✓	✓
Inclusion body myopathy type 2 (Nonaka myopathy)	GNE	AR	80%	-	-	-	✓	✓
Infantile neuroaxonal dystrophy	PLA2G6	AR	97%	-	-	-	✓	✓
Joubert syndrome 28; Meckel syndrome 1; Bardet-Biedl syndrome 13	MKS1	AR	98%	-	-	-	✓	✓
Joubert syndrome 4; Senior-Løken syndrome 1; Nephronophthisis	NPHP1	AR	98%	-	-	-	✓	✓
Junctional epidermolysis bullosa, LAMA3-related; Laryngo-onycho-cutaneous syndrome	LAMA3	AR	98%	-	-	-	✓	✓
Junctional epidermolysis bullosa, LAMB3-related	LAMB3	AR	98%	-	-	-	✓	✓
Junctional epidermolysis bullosa, LAMC2-related	LAMC2	AR	98%	-	-	-	✓	✓
Juvenile retinoschisis, X-linked	RS1	XL	96%	-	-	-	✓	✓
Krabbe disease	GALC	AR	99%	-	-	-	✓	✓
L1 syndrome	L1CAM	XL	99%	-	-	-	✓	✓
Leber congenital amaurosis 2; Retinitis pigmentosa 20	RPE65	AR	98%	-	-	-	✓	✓
Leber congenital amaurosis 5	LCA5	AR	98%	-	-	-	✓	✓
Leber congenital amaurosis 8; Retinitis pigmentosa 12	CRB1	AR	98%	-	-	-	✓	✓
Leber congenital amaurosis type 13	RDH12	AR	98%	-	-	-	✓	✓
Leigh syndrome with Complex IV deficiency	LRPPRC	AR	98%	-	-	-	✓	✓
Lethal congenital contracture syndrome 1	GLE1	AR	98%	-	-	-	✓	✓
Leukoencephalopathy with vanishing white matter	EIF2B5	AR	98%	-	-	-	✓	✓
Limb-girdle muscular dystrophy type 2A	CAPN3	AR	98%	-	-	-	✓	✓
Limb-girdle muscular dystrophy type 2B	DYSF	AR	95%	-	-	-	✓	✓
Limb-girdle muscular dystrophy, type 2C	SGCG	AR	98%	-	-	-	✓	✓
Limb-girdle muscular dystrophy, type 2D	SGCA	AR	98%	-	-	-	✓	✓
Limb-girdle muscular dystrophy, type 2E	SGCB	AR	98%	-	-	-	✓	✓
Limb-girdle muscular dystrophy, type 2F	SGCD	AR	98%	-	-	-	✓	✓
Limb-girdle muscular dystrophy, type 2H; Bardet-Biedl syndrome 11	TRIM32	AR	98%	-	-	-	✓	✓
Lipoid congenital adrenal hyperplasia	STAR	AR	98%	-	-	-	✓	✓
Lissencephaly, X-linked	DCX	XL	98%	-	-	-	✓	✓

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Liver failure, acute infantile	TRMU	AR	98%	-	-	-	✓	✓
Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency; Trifunctional protein deficiency	HADHA	AR	98%	-	-	-	✓	✓
Lysinuric protein intolerance	SLC7A7	AR	95%	-	-	-	✓	✓
Lysosomal acid lipase deficiency	LIPA	AR	99%	-	-	-	✓	✓
Maple syrup urine disease, type II	DBT	AR	98%	-	-	-	✓	✓
Meckel syndrome 5; Joubert syndrome 7; COACH syndrome	RPGRIP1L	AR	98%	-	-	-	✓	✓
Megalencephalic leukoencephalopathy with subcortical cysts	MLC1	AR	97%	-	-	-	✓	✓
Menkes disease	ATP7A	XL	99%	-	-	-	✓	✓
Metachromatic leukodystrophy	ARSA	AR	95%	-	-	-	✓	✓
Metachromatic leukodystrophy due to saposin-b deficiency	PSAP	AR	98%	-	-	-	✓	✓
Methylmalonic acidemia, MUT-related	MUT	AR	96%	-	-	-	✓	✓
Methylmalonic aciduria and homocystinuria, cblD type	MMADHC	AR	98%	-	-	-	✓	✓
Methylmalonic aciduria, cblA type	MMAA	AR	97%	-	-	-	✓	✓
Methylmalonic aciduria, cblB type	MMAB	AR	98%	-	-	-	✓	✓
Microphthalmia with or without coloboma	VSX2	AR	98%	-	-	-	✓	✓
Mitochondrial complex I deficiency (Leigh syndrome), NDUFAF5-related	NDUFAF5	AR	98%	-	-	-	✓	✓
Mitochondrial complex I deficiency (Leigh syndrome), NDUFS6-related	NDUFS6	AR	98%	-	-	-	✓	✓
Mitochondrial myopathy and sideroblastic anemia 1	PUS1	AR	98%	-	-	-	✓	✓
Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	TYMP	AR	98%	-	-	-	✓	✓
Mucolipidosis III alpha/beta; Mucolipidosis II alpha/beta	GNPTAB	AR	95%	-	-	-	✓	✓
Mucolipidosis III gamma	GNPTG	AR	95%	-	-	-	✓	✓
Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	SGSH	AR	98%	-	-	-	✓	✓
Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	GNS	AR	98%	-	-	-	✓	✓
Mucopolysaccharidosis IVA (Morquio syndrome A)	GALNS	AR	97%	-	-	-	✓	✓
Mucopolysaccharidosis type II (Hunter syndrome)	IDS	XL	91%	-	-	-	✓	✓
Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	NAGLU	AR	99%	-	-	-	✓	✓
Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	HGSNAT	AR	98%	-	-	-	✓	✓
Mucopolysaccharidosis type IVB (Morquio syndrome B); GM1-gangliosidosis	GLB1	AR	99%	-	-	-	✓	✓
Mucopolysaccharidosis type IX	HYAL1	AR	98%	-	-	-	✓	✓
Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	ARSB	AR	98%	-	-	-	✓	✓
Mucopolysaccharidosis type VII	GUSB	AR	98%	-	-	-	✓	✓
Multiple pterygium syndrome	CHRNG	AR	98%	-	-	-	✓	✓
Muscular dystrophy, LAMA2-related	LAMA2	AR	99%	-	-	-	✓	✓

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Muscular dystrophy-dystroglycanopathy, FKRP-related	FKRP	AR	98%	-	-	-	✓	✓
Muscular dystrophy-dystroglycanopathy; Retinitis pigmentosa 76	POMGNT1	AR	98%	-	-	-	✓	✓
Myotubular myopathy, X-linked	MTM1	XL	98%	-	-	-	✓	✓
N-acetylglutamate synthase deficiency	NAGS	AR	98%	-	-	-	✓	✓
Nephrogenic diabetes insipidus	AQP2	AR	95%	-	-	-	✓	✓
Neuronal ceroid lipofuscinosis, CLN5-related	CLN5	AR	95%	-	-	-	✓	✓
Neuronal ceroid lipofuscinosis, CLN6-related	CLN6	AR	92%	-	-	-	✓	✓
Neuronal ceroid lipofuscinosis, CLN8-related	CLN8	AR	95%	-	-	-	✓	✓
Neuronal ceroid lipofuscinosis, MFSD8-related	MFSD8	AR	95%	-	-	-	✓	✓
Neuronal ceroid lipofuscinosis, PPT1-related	PPT1	AR	98%	-	-	-	✓	✓
Neuronal ceroid lipofuscinosis, TPP1-related	TPP1	AR	97%	-	-	-	✓	✓
Niemann-Pick disease, type C1	NPC1	AR	90%	-	-	-	✓	✓
Niemann-Pick disease, type C2	NPC2	AR	99%	-	-	-	✓	✓
Nijmegen breakage syndrome	NBN	AR	99%	-	-	-	✓	✓
Nonsyndromic hearing loss, GJB2-related	GJB2	AR	99%	-	-	-	✓	✓
Nonsyndromic hearing loss, GJB6-related	GJB6	AR	99%	-	-	-	✓	✓
Nonsyndromic hearing loss, LOXHD1-related	LOXHD1	AR	98%	-	-	-	✓	✓
Non-syndromic hearing loss, MYO7A-related; Usher syndrome, type 1B	MYO7A	AR	98%	-	-	-	✓	✓
Non-syndromic hearing loss, USH1C-related; Usher syndrome, type IC	USH1C	AR	90%	-	-	-	✓	✓
Omenn syndrome, RAG1-related	RAG1	AR	98%	-	-	-	✓	✓
Omenn syndrome, RAG2-related	RAG2	AR	98%	-	-	-	✓	✓
Ornithine transcarbamylase deficiency	OTC	XL	90%	-	-	-	✓	✓
Osteopetrosis, TCIRG1-related	TCIRG1	AR	98%	-	-	-	✓	✓
Pendred syndrome	SLC26A4	AR	98%	-	-	-	✓	✓
Peroxisomal acyl-CoA oxidase deficiency	ACOX1	AR	98%	-	-	-	✓	✓
Pontocerebellar hypoplasia type 1A	VRK1	AR	98%	-	-	-	✓	✓
Pontocerebellar hypoplasia type 1B	EXOSC3	AR	98%	-	-	-	✓	✓
Pontocerebellar hypoplasia type 6	RARS2	AR	98%	-	-	-	✓	✓
Pontocerebellar hypoplasia, type 2D	SEPSECS	AR	98%	-	-	-	✓	✓
Postnatal progressive microcephaly with seizures and brain atrophy	MED17	AR	99%	-	-	-	✓	✓
Primary ciliary dyskinesia, DNAH5-related	DNAH5	AR	98%	-	-	-	✓	✓
Primary ciliary dyskinesia, DNAI1-related	DNAI1	AR	98%	-	-	-	✓	✓
Primary ciliary dyskinesia, DNAI2-related	DNAI2	AR	98%	-	-	-	✓	✓
Primary ciliary dyskinesia, DNAL1-related	DNAL1	AR	98%	-	-	-	✓	✓
Primary ciliary dyskinesia, type 14	CCDC39	AR	98%	-	-	-	✓	✓
Primary ciliary dyskinesia, type 17	CCDC103	AR	98%	-	-	-	✓	✓

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Primary ciliary dyskinesia, type 30	CCDC151	AR	98%	-	-	-	✓	✓
Primary congenital glaucoma	CYP1B1	AR	99%	-	-	-	✓	✓
Primary hyperoxaluria type 1	AGXT	AR	99%	-	-	-	✓	✓
Primary Hyperoxaluria type II	GRHPR	AR	99%	-	-	-	✓	✓
Primary hyperoxaluria type III	HOGA1	AR	99%	-	-	-	✓	✓
Progressive external ophthalmoplegia; Alpers-Huttenlocher syndrome; Ataxia neuropathy spectrum; Myocerebrohepatopathy syndrome	POLG	AR	95%	-	-	-	✓	✓
Progressive Familial Intrahepatic Cholestasis	ABCB11	AR	98%	-	-	-	✓	✓
Propionic acidemia, PCCA-related	PCCA	AR	96%	-	-	-	✓	✓
Propionic acidemia, PCCB-related	PCCB	AR	99%	-	-	-	✓	✓
Pycnodysostosis	CTSK	AR	98%	-	-	-	✓	✓
Pyruvate carboxylase deficiency	PC	AR	95%	-	-	-	✓	✓
Pyruvate dehydrogenase E1-alpha deficiency	PDHA1	XL	98%	-	-	-	✓	✓
Pyruvate dehydrogenase E1-beta deficiency	PDHB	AR	98%	-	-	-	✓	✓
Renal tubular acidosis with deafness	ATP6V1B1	AR	98%	-	-	-	✓	✓
Retinitis pigmentosa 25	EYS	AR	98%	-	-	-	✓	✓
Retinitis pigmentosa 26	CERKL	AR	98%	-	-	-	✓	✓
Retinitis pigmentosa 28	FAM161A	AR	98%	-	-	-	✓	✓
Rhizomelic chondrodysplasia punctata, type 3	AGPS	AR	98%	-	-	-	✓	✓
Roberts syndrome	ESCO2	AR	99%	-	-	-	✓	✓
Sandhoff disease	HEXB	AR	98%	-	-	-	✓	✓
Schimke immunoosseous dysplasia	SMARCAL1	AR	90%	-	-	-	✓	✓
Schopf-Schulz-Passarge syndrome; Odontoonychodermal dysplasia	WNT10A	AR	99%	-	-	-	✓	✓
Segawa syndrome	TH	AR	98%	-	-	-	✓	✓
Severe combined immunodeficiency with sensitivity to ionizing radiation	DCLRE1C	AR	98%	-	-	-	✓	✓
Severe combined immunodeficiency, X-linked	IL2RG	XL	99%	-	-	-	✓	✓
Severe Congenital Neutropenia, HAX1-related	HAX1	AR	98%	-	-	-	✓	✓
Severe congenital neutropenia, VPS45-related	VPS45	AR	98%	-	-	-	✓	✓
Short-chain acyl-coA dehydrogenase (SCAD) Deficiency	ACADS	AR	99%	-	-	-	✓	✓
Sialic acid storage disorder	SLC17A5	AR	91%	-	-	-	✓	✓
Sjögren-Larsson syndrome	ALDH3A2	AR	98%	-	-	-	✓	✓
Spastic paraplegia 15	ZFYVE26	AR	98%	-	-	-	✓	✓
Spastic paraplegia 49	TECPR2	AR	98%	-	-	-	✓	✓
Spondylocostal dysostosis	MESP2	AR	98%	-	-	-	✓	✓
Steel syndrome	COL27A1	AR	98%	-	-	-	✓	✓
Stuve-Wiedemann syndrome	LIFR	AR	98%	-	-	-	✓	✓
Systemic primary carnitine deficiency	SLC22A5	AR	76%	-	-	-	✓	✓

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Tetrahydrobiopterin deficiency	PTS	AR	96%	-	-	-	✓	✓
Trichohepatoenteric syndrome	TTC37	AR	98%	-	-	-	✓	✓
Tyrosinemia, type II	TAT	AR	98%	-	-	-	✓	✓
Usher syndrome, type 1D	CDH23	AR/Digenic	90%	-	-	-	✓	✓
Usher syndrome, type 2A	USH2A	AR	96%	-	-	-	✓	✓
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	ACADVL	AR	93%	-	-	-	✓	✓
Wiskott-Aldrich syndrome; Thrombo-cytopenia, X-linked; Severe Congenital Neutropenia, WAS-related	WAS	XL	99%	-	-	-	✓	✓
Wolcott-Rallison syndrome	EIF2AK3	AR	98%	-	-	-	✓	✓
Xeroderma pigmentosum, group A	XPA	AR	99%	-	-	-	✓	✓
Xeroderma pigmentosum, group C	XPC	AR	99%	-	-	-	✓	✓
Zellweger syndrome, PEX10-related	PEX10	AR	95%	-	-	-	✓	✓
Zellweger syndrome, PEX12-related	PEX12	AR	95%	-	-	-	✓	✓
Zellweger syndrome, PEX6-related	PEX6	AR	95%	-	-	-	✓	✓
Alpha-1 antitrypsin deficiency	SERPINA1	AR	95%	-	-	-	-	✓
Butyrlcholinesterase deficiency	BCHE	AR	99%	-	-	-	-	✓
Factor V deficiency	F5	AR	99%	-	-	-	-	✓
Familial hypercholesterolemia	LDLRAP1	AR	99%	-	-	-	-	✓
Familial lipoprotein lipase deficiency	LPL	AR	99%	-	-	-	-	✓
Glucose-6-phosphate dehydrogenase deficiency	G6PD	XL	98%	-	-	-	-	✓
Hemochromatosis, HFE-related	HFE	AR	99%	-	-	-	-	✓
Homocystinuria, MTHFR-related	MTHFR	AR	98%	-	-	-	-	✓
Prothrombin-related conditions	F2	AR	99%	-	-	-	-	✓

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Contact Us

Website
www.FulgentGenetics.com

Phone (USA)
(+1) 626-350-0537

Fax
(+1) 626-454-1667

Email
info@FulgentGenetics.com

Shipping Address
4978 Santa Anita Ave., Suite 205, Temple City, California 91780