fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
AAAS	Achalasia-addisonianism-alacrimia syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ABCA12	Congenital ichthyosis, ABCA12-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ABCA3	Surfactant metabolism dysfunction, pulmonary 3	AR	General Population	1 in 116	99%	1 in 11,501	1 in 5,336,464
ABCA4	Stargardt disease	AR	General Population	1 in 51	98%	1 in 2,501	1 in 510,204
ABCB11	Progressive familial intrahepatic cholestasis	AR	General Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
ABCB4	Progressive familial intrahepatic cholestasis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ABCC8	Familial hyperinsulinism	AR	General Population Ashkenazi Jewish Population Finnish Population Middle-Eastern Population	1 in 112 1 in 44 1 in 25 1 in 25	98% 98% 98% 98%	1 in 5,551 1 in 2,151 1 in 1,201 1 in 1,201	1 in 2,486,848 1 in 378,576 1 in 120,100 1 in 120,100
ABCD1	Adrenoleukodystrophy, X-linked	XL	General Population	1 in 21,000	99%	1 in 2,099,901	1 in 8,399,804
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ACAD9	Acyl-CoA dehydrogenase-9 (ACAD9) deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	AR	General Population Caucasian / European Population East Asian Population Native American Population	1 in 69 1 in 52 1 in 198 1 in 43	98% 99% 99% 96%	1 in 3,401 1 in 5,101 1 in 19,701 1 in 1,051	1 in 938,676 1 in 1,061,008 <1 in 10 million 1 in 180,772
ACADS	Short-chain acyl-coA dehydrogenase (SCAD) deficiency	AR	General Population African/African American Population Caucasian / European Population Middle-Eastern Population South Asian/Indian Population	1 in 85 1 in 52 1 in 76 1 in 52 1 in 51	99% 99% 99% 99% 99%	1 in 8,401 1 in 5,101 1 in 7,501 1 in 5,101 1 in 5,001	1 in 2,856,340 1 in 1,061,008 1 in 2,280,304 1 in 1,061,008 1 in 1,020,204
ACADSB	Short branched chain acyl-CoA dehydrogenase (SBCAD) deficiency	AR	General Population Hmong Population	1 in 368 1 in 6	99% 99%	1 in 36,701 1 in 501	<1 in 10 million <1 in 10 million
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR	General Population Middle-Eastern Population Native American Population South Asian/Indian Population	1 in 118 1 in 74 1 in 61 1 in 73	93% 93% 93% 93%	1 in 1,672 1 in 1,044 1 in 858 1 in 1,030	1 in 789,184 1 in 309,024 1 in 209,352 1 in 300,760
ACAT1	3-ketothiolase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ACSF3	Combined malonic and methylmalonic aciduria	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ADA	Adenosine deaminase deficiency	AR	General Population	1 in 224	93%	1 in 3,187	1 in 2,855,552
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 248	98% 98%	1 in 24,951 1 in 12,351	<1 in 10 million <1 in 10 million
ADGRG1	Bilateral frontoparietal polymicrogyria	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ADGRV1	Usher syndrome, type IIC	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ADK	Hypermethioninemia due to adenosine kinase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
AFF2	Fragile XE syndrome	XL	General Population	<1 in 50,000	98%	1 in 2,499,951	1 in 9,999,804
AGA	Aspartylglucosaminuria	AR	General Population Finnish Population	<1 in 500 1 in 71	98% 98%	1 in 24,951 1 in 3,501	<1 in 10 million 1 in 994,284
AGL	Glycogen storage disease type III	AR	General Population Faroese Population Inuit Population North African Jewish Population	1 in 158 1 in 28 1 in 25 1 in 37	95% 95% 95% 95%	1 in 3,141 1 in 541 1 in 481 1 in 721	1 in 1,985,112 1 in 60,592 1 in 48,100 1 in 106,708
AGPAT2	Congenital generalized lipodystrophy, type 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
AGPS	Rhizomelic chondrodysplasia punctata, type 3	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
AGXT	Primary hyperoxaluria type 1	AR	General Population Caucasian / European Population	1 in 120 1 in 173	99% 99%	1 in 11,901 1 in 17,201	1 in 5,712,480 <1 in 10 million
AHCY	Hypermethioninemia due to deficiency of S-adenosylhomocysteine hydrolase	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
AHI1	Joubert syndrome, AHI1-related	AR	General Population	1 in 448	99%	1 in 44,701	<1 in 10 million
AIMP1	Hypomyelinating leukodystrophy 3	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
AIPL1	Childhood-onset severe retinal dystrophy, AIPL1-related	AR	General Population	1 in 409	99%	1 in 40,801	<1 in 10 million
AIRE	Autoimmune polyendocrinopathy syndrome type I	AR	General Population Finnish Population	1 in 150 1 in 79	98% 98%	1 in 7,451 1 in 3,901	1 in 4,470,600 1 in 1,232,716
AK2	Reticular dysgenesis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
AKR1D1	Congenital Bile Acid Synthesis Defect	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ALDH3A2	Sjögren-Larsson syndrome	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
ALDH4A1	Hyperprolinemia type II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ALDH7A1	Pyridoxine-dependent epilepsy	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ALDOB	Hereditary fructose intolerance	AR	General Population African/African American Population Caucasian / European Population Middle-Eastern Population	1 in 122 1 in 250 1 in 67 1 in 97	99% 99% 99% 99%	1 in 12,101 1 in 24,901 1 in 6,601 1 in 9,601	1 in 5,905,288 <1 in 10 million 1 in 1,769,068 1 in 3,725,188
ALG6	Congenital disorder of glycosylation type Ic	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ALMS1	Alstrom syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
ALOX12B	Autosomal recessive, congenital, ichthyosis 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ALOXE3	Congenital ichthyosiform erythroderma	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ALPL	Hypophosphatasia	AR	General Population Caucasian / European Population Mennonite Population	1 in 158 1 in 274 1 in 25	95% 95% 95%	1 in 3,141 1 in 5,461 1 in 481	1 in 1,985,112 1 in 5,985,256 1 in 48,100
AMH	Persistent mullerian duct syndrome, type I	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
AMHR2	Persistent mullerian duct syndrome, type II	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
AMN	Megaloblastic anemia 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
AMPD2	Pontocerebellar hypoplasia type 9	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
AMT	Glycine encephalopathy	AR	General Population Finnish Population	1 in 373 1 in 117	98% 98%	1 in 18,601 1 in 5,801	<1 in 10 million 1 in 2,714,868
ANO10	Spinocerebellar ataxia 10	AR	General Population	1 in 93	99%	1 in 9,201	1 in 3,422,772
ANO5	Gnathodiaphyseal dysplasia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ANTXR2	Hyaline fibromatosis syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
AP1S1	MEDNIK syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
AP1S2	X-linked Intellectual disability, AP1S2-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
AP3B1	Hermansky-Pudlak syndrome 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
AP3D1	Hermansky-Pudlak syndrome 10	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
APOPT1	Mitochondrial complex IV deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
AQP2	Nephrogenic diabetes insipidus	AR	General Population Finnish Population	<1 in 500 1 in 169	95% 95%	1 in 9,981 1 in 3,361	<1 in 10 million 1 in 2,272,036
AR	Androgen insensitivity syndrome	XL	General Population	1 in 14,286	98%	1 in 714,251	1 in 1,428,571
ARG1	Arginase deficiency	AR	General Population	1 in 296	98%	1 in 14,751	<1 in 10 million
ARL13B	Joubert syndrome, ARL13B-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ARSA	Metachromatic leukodystrophy	AR	General Population Caucasian / European Population Yemenite Jewish Population	1 in 100 1 in 78 1 in 75	99% 99% 99%	1 in 9,901 1 in 7,701 1 in 7,401	1 in 3,960,400 1 in 2,402,712 1 in 2,220,300
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	AR	General Population Western Australian Population	1 in 250 1 in 283	98% 98%	1 in 12,451 1 in 14,101	<1 in 10 million <1 in 10 million
ARSE	Chondrodysplasia punctata type 1, X-linked	XL	General Population	1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
ARX	X-linked intellectual disability, ARX-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
ASL	Argininosuccinate lyase deficiency	AR	General Population	1 in 132	90%	1 in 1,311	1 in 692,208
ASNS	Asparagine synthetase deficiency	AR	General Population Iranian Jewish Population	<1 in 500 1 in 80	99% 99%	1 in 49,901 1 in 7,901	<1 in 10 million 1 in 2,528,320
ASPA	Canavan disease	AR	General Population Ashkenazi Jewish Population	1 in 300 1 in 55	97% 96%	1 in 9,968 1 in 1,351	<1 in 10 million 1 in 297,220
ASS1	Citrullinemia	AR	General Population East Asian Population	1 in 119 1 in 132	96% 96%	1 in 2,951 1 in 3,276	1 in 1,404,676 1 in 1,729,728
ATM	Ataxia-telangiectasia	AR	General Population	1 in 100	92%	1 in 1,239	1 in 495,600
ATP13A2	Kufor-Rakeb syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ATP6V0A2	Cutis laxa, type IIA	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ATP6V0A4	Renal tubular acidosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ATP6V1B1	Renal tubular acidosis with deafness	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ATP6V1E1	Cutis laxa, type IIC	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ATP7A	Menkes disease	XL	General Population	1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
ATP7B	Wilson disease	AR	General Population Caucasian / European Population Ashkenazi Jewish Population	1 in 87 1 in 42 1 in 70	98% 98% 98%	1 in 4,301 1 in 2,051 1 in 3,451	1 in 1,496,748 1 in 344,568 1 in 966,280
ATP8B1	Progressive familial intrahepatic cholestasis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ATRX	Alpha thalassemia X-linked intellectual disability syndrome	XL	General Population	<1 in 250,000	99%	1 in 24,999,901	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
AVPR2	Nephrogenic diabetes insipidus	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
B9D1	Joubert syndrome 27	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
B9D2	Meckel syndrome 10	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
BBS1	Bardet-Biedl syndrome type 1	AR	General Population	1 in 367	99%	1 in 36,601	<1 in 10 million
BBS10	Bardet-Biedl syndrome type 10	AR	General Population	1 in 395	99%	1 in 39,401	<1 in 10 million
BBS12	Bardet-Biedl syndrome type 12	AR	General Population	1 in 791	99%	1 in 79,001	<1 in 10 million
BBS2	Bardet-Biedl syndrome 2	AR	General Population Ashkenazi Jewish Population	1 in 621 1 in 107	99% 99%	1 in 62,001 1 in 10,601	<1 in 10 million 1 in 4,537,228
BBS2	Retinitis Pigmentosa 74	AR	General Population Ashkenazi Jewish Population	1 in 621 1 in 107	99% 99%	1 in 62,001 1 in 10,601	<1 in 10 million 1 in 4,537,228
BBS5	Bardet-Biedl syndrome 5	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
BBS7	Bardet-Biedl syndrome 7	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
BBS9	Bardet-Biedl syndrome 9	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
BCHE	Butyrylcholinesterase deficiency	AR	General Population	1 in 28	99%	1 in 2,701	1 in 302,512
BCKDHA	Maple syrup urine disease type Ia	AR	General Population Mennonite Population	1 in 321 1 in 10	98% 98%	1 in 16,001 1 in 451	<1 in 10 million 1 in 18,040
BCKDHB	Maple syrup urine disease type Ib	AR	General Population Ashkenazi Jewish Population	1 in 364 1 in 97	98% 98%	1 in 18,151 1 in 4,801	<1 in 10 million 1 in 1,862,788
BCS1L	Björnstad syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
BCS1L	GRACILE syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
BCS1L	Mitochondrial complex III deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
BLM	Bloom syndrome	AR	General Population Ashkenazi Jewish Population	1 in 800 1 in 134	87% 99%	1 in 6,147 1 in 13,301	<1 in 10 million 1 in 7,129,336
BLOC1S3	Hermansky-Pudlak syndrome 8	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
BLOC1S6	Hermansky-Pudlak syndrome 9	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
BMP1	Osteogenesis imperfecta, type XIII	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
BMPER	Diaphanospondylodysostosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
BRIP1	Fanconi anemia group J	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
BRWD3	X-linked intellectual disability, BRWD3-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
BSND	Bartter syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
BTD	Biotinidase deficiency	AR	General Population Caucasian / European Population Latino Population Middle-Eastern Population	1 in 124 1 in 71 1 in 136 1 in 55	99% 99% 99% 99%	1 in 12,301 1 in 7,001 1 in 13,501 1 in 5,401	1 in 6,101,296 1 in 1,988,284 1 in 7,344,544 1 in 1,188,220
BTK	X-linked agammaglobulinemia	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
C19orf12	Mitochondrial membrane protein-associated neurodegeneration	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
C8orf37	Bardet-Biedl Syndrome 21	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CAD	Early Infantile Epileptic Encephalopathy 50	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CANT1	Desbuquois dysplasia 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CAPN3	Limb-girdle muscular dystrophy type 2A	AR	General Population Caucasian / European Population	<1 in 500 1 in 103	98% 98%	1 in 24,951 1 in 5,101	<1 in 10 million 1 in 2,101,612
CASP14	Congenital Ichthyosis 12	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CASQ2	Catecholaminergic polymorphic ventricular tachycardia	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million
CAVIN1	Congenital Generalized Lipodystrophy 4	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	AR	General Population Caucasian / European Population Middle-Eastern Population	1 in 224 1 in 86 1 in 21	99% 99% 99%	1 in 22,301 1 in 8,501 1 in 2,001	<1 in 10 million 1 in 2,924,344 1 in 168,084
CC2D1A	Autosomal recessive intellectual developmental disorder 3	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CC2D2A	Joubert syndrome 9	AR	General Population	1 in 201	99%	1 in 20,001	1 in 16,080,804
CCDC103	Primary ciliary dyskinesia, type 17	AR	General Population	1 in 316	98%	1 in 15,751	<1 in 10 million
CCDC151	Primary ciliary dyskinesia, type 30	AR	General Population	1 in 365	98%	1 in 18,201	<1 in 10 million
CCDC39	Primary ciliary dyskinesia, type 14	AR	General Population	1 in 211	98%	1 in 10,501	1 in 8,862,844
CCDC8	3-M Syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CCDC88C	Congenital hydrocephalus 1	AR	General Population	1 in 137	99%	1 in 13,601	1 in 7,453,348
CD247	Severe Combined Immunodeficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CD3D	Severe Combined Immunodeficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CD3E	Severe Combined Immunodeficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
CD3G	Severe Combined Immunodeficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CD40LG	Hyper IgM syndrome, X-linked	XL	General Population	1 in 50,000	98%	1 in 2,499,951	1 in 9,999,904
CD8A	Familial CD8 Deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CDAN1	Dyserythropoietic congenital anemia, type Ia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CDCA7	Immunodeficiency-centromeric instability-facial anomalies syndrome 3	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CDH23	Usher syndrome, type 1D	AR	General Population	1 in 285	90%	1 in 2,841	1 in 11,364
CEP104	Joubert syndrome 25	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CEP152	Seckel syndrome 5	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CEP152	Microcephaly 9	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CEP290	Joubert syndrome 5	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Leber congenital amaurosis 10	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Bardet-Biedl syndrome 14	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	CEP290-related disorders	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Senior-Løken syndrome 6	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Meckel syndrome 4	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CERKL	Retinitis pigmentosa 26	AR	General Population	1 in 148	98%	1 in 7,351	1 in 4,351,792
CERS3	Congenital ichthyosis 9	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CFTR	Cystic Fibrosis	AR	General Population African/African American Population Ashkenazi Jewish Population Caucasian / European Population East Asian Population Latino Population	1 in 32 1 in 61 1 in 24 1 in 25 1 in 94 1 in 58	99% 99% 99% 99% 99% 99%	1 in 3,101 1 in 6,001 1 in 2,301 1 in 2,401 1 in 9,301 1 in 5,701	1 in 396,928 1 in 1,464,244 1 in 220,896 1 in 240,100 1 in 3,497,176 1 in 1,322,632
CHAT	Congenital myasthenic syndrome 6	AR	General Population	1 in 197	99%	1 in 19,601	<1 in 10 million
CHM	Choroideremia	XL	General Population	1 in 25,000	95%	1 in 499,981	1 in 1,999,964
CHRNE	Congenital myasthenic syndrome	AR	General Population	1 in 408	99%	1 in 40,701	<1 in 10 million
CHRNG	Multiple pterygium syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CHST6	Macular corneal dystrophy, CHST6-related	AR	General Population	1 in 79	99%	1 in 7,801	1 in 2,465,116
CIITA	Bare lymphocyte syndrome, type II	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CLCF1	Crisponi cold-induced sweating syndrome 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CLCN1	Autosomal recessive congenital myotonia	AR	General Population	1 in 176	99%	1 in 17,501	1 in 12,320,704
CLCNKB	Bartter syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
CLCNKB	Bartter syndrome type 3	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CLN3	Neuronal ceroid lipofuscinosis	AR	General Population Finnish Population	1 in 230 1 in 72	98% 98%	1 in 11,451 1 in 3,551	<1 in 10 million 1 in 1,022,688
CLN5	Neuronal ceroid lipofuscinosis 5	AR	General Population Finnish Population	<1 in 500 1 in 115	95% 95%	1 in 9,981 1 in 2,281	<1 in 10 million 1 in 1,049,260
CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	AR	General Population	<1 in 500	92%	1 in 6,239	<1 in 10 million
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related	AR	General Population Finnish Population	<1 in 500 1 in 135	95% 95%	1 in 9,981 1 in 2,681	<1 in 10 million 1 in 1,447,740
CLP1	Pontocerebellar hypoplasia type 10	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CLRN1	Usher syndrome, type 3A	AR	General Population Ashkenazi Jewish Population Finnish Population	1 in 500 1 in 120 1 in 70	98% 98% 98%	1 in 24,951 1 in 5,951 1 in 3,451	<1 in 10 million 1 in 2,856,480 1 in 966,280
CNGA1	Retinitis Pigmentosa, CNGA1-related	AR	General Population	1 in 210	99%	1 in 20,901	<1 in 10 million
CNGA3	Leber congenital amaurosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CNGB1	Retinitis Pigmentosa, CNGB1-related	AR	General Population	1 in 296	99%	1 in 29,501	<1 in 10 million
CNGB3	Achromatopsia	AR	General Population Micronesian Population	1 in 87 1 in 2	99% 99%	1 in 8,601 1 in 101	1 in 2,993,148 1 in 808
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
COASY	Pontocerebellar hypoplasia type 12	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
COL11A2	Otospondylomegaepiphyseal dysplasia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
COL11A2	Fibrochondrogenesis 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
COL11A2	Nonsyndromic hearing loss 53	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
COL17A1	Junctional epidermolysis bullosa	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
COL27A1	Steel syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
COL4A3	Alport syndrome, COL4A3-related	AR	General Population Ashkenazi Jewish Population	1 in 267 1 in 188	98% 98%	1 in 13,301 1 in 9,351	<1 in 10 million 1 in 7,031,952
COL4A4	Alport syndrome, COL4A4-related	AR	General Population	1 in 267	98%	1 in 13,301	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
COL4A5	Alport syndrome, COL4A5-related	XL	General Population	1 in 139	98%	1 in 6,901	1 in 27,604
COL7A1	Dystrophic epidermolysis bullosa	AR	General Population	1 in 196	97%	1 in 6,501	1 in 5,096,784
COLQ	Congenital myasthenic syndrome 5	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
COQ4	Primary Coenzyme Q10 deficiency 7	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CORO1A	Immunodeficiency 8	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
COX10	Mitochondrial complex IV deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
COX15	Mitochondrial complex IV deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
COX20	Mitochondrial complex IV deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
COX6B1	Mitochondrial complex IV deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CP	Aceruloplasminemia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CPLANE1	Joubert syndrome 17	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CPS1	Carbamoylphosphate synthetase I deficiency	AR	General Population	1 in 570	98%	1 in 28,451	<1 in 10 million
CPT1A	Carnitine palmitoyltransferase IA deficiency	AR	General Population Hutterite Population	1 in 354 1 in 16	90% 90%	1 in 3,531 1 in 151	1 in 4,999,896 1 in 9,664
CPT2	Carnitine palmitoyltransferase II deficiency	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 51	95% 95%	1 in 9,981 1 in 1,001	<1 in 10 million 1 in 204,204
CRADD	Intellectual developmental disorder with variant lissencephaly	AR	General Population Bukharian Jewish Population	1 in 500 1 in 30	99% 99%	1 in 49,901 1 in 2,901	<1 in 10 million 1 in 348,120
CRB1	Leber congenital amaurosis 8	AR	General Population	1 in 104	98%	1 in 5,151	1 in 2,142,816
CRB1	Retinitis pigmentosa 12	AR	General Population	1 in 104	98%	1 in 5,151	1 in 2,142,816
CRLF1	Crisponi cold-induced sweating syndrome 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CRTAP	Osteogenesis imperfecta, type VII	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CRYL1	GJB6-CRYL1 related nonsyndromic hearing loss	UK	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
CTC1	Cerebroretinal microangiopathy with calcifications and cysts 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CTNS	Cystinosis	AR	General Population British Population Moroccan Jewish Population	1 in 158 1 in 81 1 in 100	99% 99% 99%	1 in 15,701 1 in 8,001 1 in 9,901	1 in 9,923,032 1 in 2,592,324 1 in 3,960,400
CTSA	Galactosialidosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CTSC	Papillon-Lefevre syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CTSD	Neuronal ceroid lipofuscinosis, CTSD-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CTSF	Neuronal ceroid lipofuscinosis 13	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CTSK	Pycnodysostosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CUL4B	X-linked intellectual disability, CUL4B-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
CUL7	Three M syndrome 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CWC27	Retinitis pigmentosa with or without skeletal anomalies	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CYBA	Chronic granulomatous disease	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million
CYBB	Chronic granulomatous disease, X-linked	XL	General Population	1 in 149,254	99%	1 in 14,925,301	<1 in 10 million
CYP11A1	Congenital adrenal insufficiency	AR	General Population	1 in 114	99%	1 in 11,301	1 in 5,153,256
CYP11B1	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	AR	General Population Morrocan Jewish Population	1 in 158 1 in 35	98% 98%	1 in 7,851 1 in 1,701	1 in 4,961,832 1 in 238,140
CYP11B2	Corticosterone methyloxidase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
CYP19A1	Aromatase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CYP1B1	Primary congenital glaucoma	AR	General Population	1 in 50	99%	1 in 4,901	1 in 980,200
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR	General Population Inuit Population Middle-Eastern Population	1 in 61 1 in 9 1 in 35	99% 99% 99%	1 in 6,001 1 in 801 1 in 3,401	1 in 1,464,244 1 in 28,836 1 in 476,140
CYP27A1	Cerebrotendinous xanthomatosis	AR	General Population Morrocan Jewish Population	1 in 500 1 in 5	98% 98%	1 in 24,951 1 in 201	<1 in 10 million 1 in 4,020
CYP27B1	Vitamin D–dependent rickets, type 1	AR	General Population	1 in 181	99%	1 in 18,001	1 in 13,032,724
CYP4F22	Congenital ichthyosis 5	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CYP7B1	Congenital bile acid synthesis defect 3	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DBT	Maple syrup urine disease, type II	AR	General Population	1 in 481	98%	1 in 24,001	<1 in 10 million
DCAF17	Woodhouse-Sakati syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DCLRE1C	Severe combined immunodeficiency with sensitivity to ionizing radiation	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
DCX	Lissencephaly, X-linked	XL	General Population	1 in 42,500	98%	1 in 2,124,951	1 in 8,499,904
DDB2	Xeroderma pigmentosum, group E	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DDC	Aromatic l-amino acid decarboxylase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DDR2	Spondylometaepiphyseal dysplasia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DDX11	Warsaw breakage syndrome	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 68	99% 99%	1 in 49,901 1 in 6,701	<1 in 10 million 1 in 1,822,672
DGUOK	Mitochondrial DNA depletion syndrome 3	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DHCR24	Desmosterolosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DHCR7	Smith-Lemli-Opitz syndrome	AR	General Population African/African American Population Ashkenazi Jewish Population	1 in 30 1 in 138 1 in 36	96% 96% 96%	1 in 726 1 in 3,426 1 in 876	1 in 87,120 1 in 1,891,152 1 in 126,144
DHDDS	Retinitis pigmentosa 59	AR	General Population Ashkenazi Jewish Population	1 in 296 1 in 118	98% 98%	1 in 14,751 1 in 5,851	<1 in 10 million 1 in 2,761,672
DKC1	X-linked dyskeratosis congenita	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
DLAT	Pyruvate dehydrogenase E2 deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DLD	Dihydrolipoamide dehydrogenase deficiency	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 107	98% 98%	1 in 24,951 1 in 5,301	<1 in 10 million 1 in 2,268,828
DLG3	X-linked intellectual disability, DLG3-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
DLL3	Spondylocostal dysostosis 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DMD	Duchenne Muscular Dystrophy	XL	General Population	1 in 2,350	93%	1 in 33,558	1 in 134,260
DMD	Becker Muscular Dystrophy	XL	General Population	1 in 2,350	93%	1 in 33,558	1 in 134,260
DNAH5	Primary ciliary dyskinesia, DNAH5-related	AR	General Population Ashkenazi Jewish Population	1 in 142 1 in 113	98% 99%	1 in 7,051 1 in 11,201	1 in 4,004,968 1 in 5,062,852
DNAI1	Primary ciliary dyskinesia, DNAI1-related	AR	General Population	1 in 230	98%	1 in 11,451	<1 in 10 million
DNAI2	Primary ciliary dyskinesia, DNAI2-related	AR	General Population	1 in 447	98%	1 in 22,301	<1 in 10 million
DNAL1	Primary ciliary dyskinesia, DNAL1-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
DNMT3B	ICF Syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DOCK8	Hyper-IgE syndrome due to DOCK8 defiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DOK7	Congenital myasthenic syndrome, DOK7-related	AR	General Population	1 in 472	98%	1 in 23,551	<1 in 10 million
DPYD	Dihydropyrimidine dehydrogenase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
DTNBP1	Hermansky-Pudlak syndrome 7	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DUOX2	Congenital hypothyroidism, DUOX2-related	AR	General Population	1 in 366	91%	1 in 4,057	1 in 5,938,797
DUOXA2	Congenital hypothyroidism, DUOXA2-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly	AR	General Population	1 in 68	98%	1 in 3,351	1 in 924,876
DYSF	Limb-girdle muscular dystrophy type 2B	AR	General Population Japanese Population Libyan Jewish Population	<1 in 500 1 in 332 1 in 18	95% 95% 95%	1 in 9,981 1 in 6,621 1 in 341	<1 in 10 million 1 in 8,792,688 1 in 24,552
EDA	Hypohidrotic ectodermal dysplasia	XL	General Population	1 in 14,167	99%	1 in 1,416,601	1 in 5,666,472
EIF2AK3	Wolcott-Rallison Syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
EIF2B1	Leukoencephalopathy with vanishing white matter	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
EIF2B2	Leukoencephalopathy with vanishing white matter	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
EIF2B3	Leukoencephalopathy with vanishing white matter	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
EIF2B4	Leukoencephalopathy with vanishing white matter	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
EIF2B5	Leukoencephalopathy with vanishing white matter	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ELP1	Familial Dysautonomia	AR	General Population Ashkenazi Jewish Population	1 in 300 1 in 31	99% 99%	1 in 29,901 1 in 3,001	<1 in 10 million 1 in 372,124
EMD	Emery-Dreifuss muscular dystrophy	XL	General Population	1 in 81,967	99%	1 in 8,196,601	<1 in 10 million
EPB42	Spherocytosis, type 5	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ERBB3	Lethal congenital contractural syndrome 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ERCC2	Xeroderma pigmentosum, group D	AR	General Population	1 in 65	99%	1 in 6,401	1 in 1,664,260
ERCC2	Photosensitive trichothiodystrophy 1	AR	General Population	1 in 65	99%	1 in 6,401	1 in 1,664,260
ERCC2	Cerebrooculofacioskeletal syndrome 2	AR	General Population	1 in 65	99%	1 in 6,401	1 in 1,664,260

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
ERCC3	Xeroderma pigmentosum, complementation group B	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ERCC3	Photosensitive Trichothiodystrophy 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ERCC4	Fanconi anemia, complementation group Q	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ERCC4	Xeroderma pigmentosum, complementation group F	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ERCC5	Xeroderma Pigmentosa, group G	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ERCC6	De Sanctis-Cacchione syndrome	AR	General Population Japanese Population	1 in 500 1 in 74	99% 99%	1 in 49,901 1 in 7,301	<1 in 10 million 1 in 2,161,096
ERCC6	Cockayne syndrome type B	AR	General Population Japanese Population	1 in 500 1 in 74	99% 99%	1 in 49,901 1 in 7,301	<1 in 10 million 1 in 2,161,096
ERCC8	Cockayne syndrome type A	AR	General Population	1 in 822	98%	1 in 41,051	<1 in 10 million
ESCO2	Roberts syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ETFA	Glutaric aciduria IIA	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
ETFB	Glutaric aciduria IIB	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
ETFDH	Glutaric aciduria IIC	AR	General Population East Asian Population	1 in 250 1 in 74	98% 98%	1 in 12,451 1 in 3,651	<1 in 10 million 1 in 1,080,696
ETHE1	Ethylmalonic encephalopathy	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
EVC	Weyers acrofacial dysostosis, EVC-related	AR	General Population Amish Population	1 in 142 1 in 7	98% 98%	1 in 7,051 1 in 301	1 in 4,004,968 1 in 8,428
EVC	Ellis-van Creveld syndrome, EVC-related	AR	General Population Amish Population	1 in 142 1 in 7	98% 98%	1 in 7,051 1 in 301	1 in 4,004,968 1 in 8,428
EVC2	Weyers acrodental dysostosis, EVC2-related	AR	General Population Amish Population	1 in 240 1 in 7	98% 98%	1 in 11,951 1 in 301	<1 in 10 million 1 in 8,428
EVC2	Ellis-van Creveld syndrome, EVC2-related	AR	General Population Amish Population	1 in 240 1 in 7	98% 98%	1 in 11,951 1 in 301	<1 in 10 million 1 in 8,428
EXOSC3	Pontocerebellar hypoplasia type 1B	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
EYS	Retinitis pigmentosa 25	AR	General Population	1 in 66	98%	1 in 3,251	1 in 858,264
F11	Factor XI deficiency	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 11	98% 98%	1 in 24,951 1 in 501	<1 in 10 million 1 in 22,044
F2	Prothrombin-related conditions	AR	General Population Caucasian / European Population	1 in 33 1 in 4	99% 99%	1 in 3,201 1 in 301	1 in 422,532 1 in 4,816
F5	Factor V deficiency	AR	General Population Caucasian / European Population Latino Population African/African American Population East Asian Population Native American Population	1 in 36 1 in 19 1 in 45 1 in 83 1 in 222 1 in 80	99% 99% 99% 99% 99% 99%	1 in 3,501 1 in 1,801 1 in 4,401 1 in 8,201 1 in 22,101 1 in 7,901	1 in 504,144 1 in 136,876 1 in 792,180 1 in 2,722,732 <1 in 10 million 1 in 2,528,320
F7	Factor VII deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
F8	Hemophilia A	XL	General Population	1 in 3,250	48%	1 in 6,249	1 in 25,000
F9	Hemophilia B	XL	General Population	1 in 15,000	99%	1 in 1,499,901	1 in 5,999,804
FA2H	Spastic paraplegia type 35	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FAH	Tyrosinemia, type 1	AR	General Population Ashkenazi Jewish Population Finnish Population French Canadian Population South Asian/Indian Population	1 in 99 1 in 150 1 in 122 1 in 66 1 in 172	95% 95% 95% 95% 95%	1 in 1,961 1 in 2,981 1 in 2,421 1 in 1,301 1 in 3,421	1 in 776,556 1 in 1,788,600 1 in 1,181,448 1 in 343,464 1 in 2,353,648
FAM126A	Hypomyelinating leukodystropy type 5	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FAM126A	Hypomyelinating leukodystropy type 5	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FAM161A	Retinitis pigmentosa 28	AR	General Population	1 in 296	98%	1 in 14,751	<1 in 10 million
FANCA	Fanconi anemia group A	AR	General Population Moroccan Jewish Indian Jewish Population	1 in 239 1 in 100 1 in 27	99% 99% 99%	1 in 23,801 1 in 9,901 1 in 2,601	<1 in 10 million 1 in 3,960,400 1 in 280,908
FANCB	Fanconi anemia group B	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
FANCC	Fanconi anemia group C	AR	General Population Ashkenazi Jewish Population	1 in 535 1 in 99	99% 99%	1 in 53,401 1 in 9,801	<1 in 10 million 1 in 3,881,196
FANCD2	Fanconi anemia, group D2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FANCE	Fanconi anemia, group E	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FANCF	Fanconi anemia, group F	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FANCG	Fanconi anemia group G	AR	General Population	1 in 632	90%	1 in 6,311	<1 in 10 million
FANCI	Fanconi anemia, group I	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FANCL	Fanconi anemia, group L	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
FBP1	Fructose-1,6-bisphosphatase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FBXL4	Mitochondrial DNA depletion syndrome 13	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FGD1	X-linked Aarskog-Scott syndrome	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
FH	Fumarase deficiency	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 99	99% 99%	1 in 49,901 1 in 9,801	<1 in 10 million 1 in 3,881,196
FHL1	Emery-Dreifuss muscular dystrophy 6	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
FHL1	X-linked myopathy with postural muscle atrophy	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
FKRP	Muscular dystrophy-dystroglycanopathy, FKRP-related	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
FKTN	Muscular dystrophy-dystroglycanopathy, FKTN-related	AR	General Population Ashkenazi Jewish Population Japanese Population	<1 in 500 1 in 150 1 in 82	99% 99% 99%	1 in 49,901 1 in 14,901 1 in 8,101	<1 in 10 million 1 in 8,940,600 1 in 2,657,128
FKTN	Fukuyama congenital muscular dystrophy	AR	General Population Ashkenazi Jewish Population Japanese Population	<1 in 500 1 in 150 1 in 82	99% 99% 99%	1 in 49,901 1 in 14,901 1 in 8,101	<1 in 10 million 1 in 8,940,600 1 in 2,657,128
FMO3	Trimethylaminuria	AR	General Population	1 in 139	99%	1 in 13,801	1 in 7,763,356
FMR1	Fragile X Syndrome Intermediate Allele	XL	General Population Ashkenazi Jewish Population	1 in 259 1 in 115	99% 99%	1 in 25,801 1 in 11,401	1 in 103,204 1 in 45,604
FMR1	Fragile X Syndrome Premutation	XL	General Population Ashkenazi Jewish Population	1 in 259 1 in 115	99% 99%	1 in 25,801 1 in 11,401	1 in 103,204 1 in 45,604
FMR1	Fragile X Syndrome Full Mutation	XL	General Population	1 in 11,111	99%	1 in 1,111,001	1 in 4,444,004
FOLR1	Cerebral folate deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FOXN1	T-cell immunodeficiency with congenital alopecia and nail dystrophy	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FOXN1	T-cell immunodeficiency with thymic aplasia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FOXP3	IPEX syndrome	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
FOXRED1	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FRAS1	Fraser syndrome	AR	General Population	1 in 250	99%	1 in 24,901	<1 in 10 million
FREM2	Fraser syndrome	AR	General Population	1 in 354	99%	1 in 35,301	<1 in 10 million
FTCD	Glutamate formiminotransferase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FTSJ1	X-linked intellectual disability, FTSJ1-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
FUCA1	Fucosidosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FXN	Friedreich ataxia	AR	General Population Caucasian / European Population	1 in 80 1 in 80	4% 98%	1 in 83 1 in 3951	1 in 26,653 1 in 1,264,320
G6PC	Glycogen storage disease, type 1a	AR	General Population Ashkenazi Jewish Population	1 in 177 1 in 64	95% 95%	1 in 3,521 1 in 1,261	1 in 2,492,868 1 in 322,816
G6PC3	Severe congenital neutropenia 4	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
G6PD	Glucose-6-phosphate dehydrogenase deficiency	XL	General Population	1 in 7	98%	1 in 301	1 in 1,204
GAA	Pompe disease	AR	General Population African/African American Population East Asian Population Ashkenazi Jewish Population	1 in 100 1 in 60 1 in 112 1 in 76	98% 98% 98% 99%	1 in 4,951 1 in 2,951 1 in 5,551 1 in 7,501	1 in 1,980,400 1 in 708,240 1 in 2,486,848 1 in 2,280,304
GALC	Krabbe disease	AR	General Population Israeli Druze Population	1 in 158 1 in 6	99% 99%	1 in 15,701 1 in 501	1 in 9,923,032 1 in 12,024
GALE	Galactose epimerase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
GALK1	Galactokinase deficiency	AR	General Population Irish Population	1 in 110 1 in 64	95% 95%	1 in 2,181 1 in 1,261	1 in 959,640 1 in 322,816
GALNS	Mucopolysaccharidosis IVA (Morquio syndrome A)	AR	General Population	1 in 224	97%	1 in 7,434	1 in 6,660,864
GALNT3	Familial hyperphosphatemic tumoral calcinosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
GALT	Galactosemia	AR	General Population African/African American Population Ashkenazi Jewish Population	1 in 110 1 in 94 1 in 127	99% 99% 99%	1 in 10,901 1 in 9,301 1 in 12,601	1 in 4,796,440 1 in 3,497,176 1 in 6,401,308
GAMT	Guanidinoacetate methyltransferase deficiency	AR	General Population	1 in 371	99%	1 in 37,001	<1 in 10 million
GBA	Gaucher disease	AR	General Population African/African American Population Ashkenazi Jewish Population	1 in 77 1 in 35 1 in 15	99% 99% 99%	1 in 7,601 1 in 3,401 1 in 1,401	1 in 2,341,108 1 in 476,140 1 in 84,060
GBE1	Glycogen storage disease IV	AR	General Population	1 in 387	99%	1 in 38,601	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
GCDH	Glutaric aciduria, type I	AR	General Population	1 in 87	98%	1 in 4,301	1 in 1,496,748
			Amish Population	1 in 9	98%	1 in 401	1 in 14,436
GDAP1	Charcot-Marie-Tooth disease, GDAP1-related	AR	General Population	1 in 152	99%	1 in 15,101	1 in 9,181,408
GDF5	Du Pan Syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
GFM1	Combined oxidative phosphorylation deficiency, GFM1-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
GFPT1	Congenital myasthenic syndrome 12	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
GHR	Growth hormone insensitivity syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
GHRHR	Isolated growth hormone deficiency, type 1B	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
GJB1	Charcot-Marie-Tooth disease, X-linked type 1	XL	General Population	1 in 667	90%	1 in 6,661	1 in 26,644
GJB2	Nonsyndromic hearing loss 1A	AR	General Population	1 in 42	99%	1 in 4,101	1 in 688,968
			African/African American Population	1 in 25	99%	1 in 2,401	1 in 240,100
			Ashkenazi Jewish Population	1 in 21	99%	1 in 2,001	1 in 168,084
			Caucasian / European Population	1 in 33	99%	1 in 3,201	1 in 422,532
			Latino Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
			Middle-Eastern Population	1 in 83	99%	1 in 8,201	1 in 2,722,732
			South Asian/Indian Population	1 in 148	99%	1 in 14,701	1 in 8,702,992
GJB6	GJB6-CRYL1 related nonsyndromic hearing loss	AR	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
GLA	Fabry disease	XL	General Population	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
GLB1	GM1-gangliosidosis	AR	General Population	1 in 134	99%	1 in 13,301	1 in 7,129,336
			Maltese Population	1 in 30	99%	1 in 2,901	1 in 348,120
			Roma Population	1 in 50	99%	1 in 4,901	1 in 980,200
GLB1	Mucopolysaccharidosis type IVB (Morquio syndrome B)	AR	General Population	1 in 134	99%	1 in 13,301	1 in 7,129,336
			Maltese Population	1 in 30	99%	1 in 2,901	1 in 348,120
			Roma Population	1 in 50	99%	1 in 4,901	1 in 980,200
GLDC	Glycine encephalopathy, GLDC-related	AR	General Population	1 in 193	98%	1 in 9,601	1 in 7,411,972
			British Columbia Canadian Population	1 in 125	99%	1 in 12,401	1 in 6,200,500
			Finnish Population	1 in 117	99%	1 in 11,601	1 in 5,429,268
GLE1	Lethal congenital contracture syndrome 1	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 80	98%	1 in 3,951	1 in 1,264,320
GNE	Inclusion body myopathy type 2 (Nonaka myopathy)	AR	General Population	<1 in 500	99%	1 in 49,901	1 in 99,802,000
			Iranian Jewish Population	1 in 11	99%	1 in 1,001	1 in 44,044
GNPAT	Rhizomelic chondrodysplasia punctata, type 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
GNPTAB	Mucolipidosis II alpha/beta	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNPTAB	Mucolipidosis III alpha/beta	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNPTG	Mucolipidosis III gamma	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNRHR	Hypogonadotropic hypogonadism, GNRHR-related	AR	General Population	1 in 347	99%	1 in 34,601	<1 in 10 million
GNS	Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
GORAB	Geroderma osteodysplasticum	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
GP1BA	Bernard-Soulier syndrome type A1	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
GP9	Bernard-Soulier syndrome type C	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
GPR143	X-linked Ocular albinism, GPR143-related	XL	General Population	1 in 25,000	99%	1 in 2,499,901	<1 in 10 million
GRHPR	Primary hyperoxaluria type II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
GRIP1	Fraser syndrome	AR	General Population	1 in 84	99%	1 in 8,301	1 in 2,789,136
GSS	Glutathione synthetase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
GUCY2D	Leber congenital amaurosis 1	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
GUSB	Mucopolysaccharidosis type VII	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
GYS2	Glycogen storage disease, type 0, liver	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
HADH	Familial hyperinsulinemic hypoglycemia 4	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
HADHA	Trifunctional protein deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
HADHB	Trifunctional protein deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
HAMP	Hemochromatosis, type 2B	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
HAX1	Severe congenital neutropenia, HAX1-related	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
HBA1	Alpha thalassemia	AR	General Population	1 in 1000	98%	1 in 860	1 in 3,440,364
			General Population†	1 in 18	98%	1 in 860	1 in 3,440,364
			Southeast Asian Population	≤1 in 7	98%	≤1 in 305	≤1 in 17,228
			Southeast Asian Population†	≤1 in 14	98%	≤1 in 305	≤1 in 17,228
			Mediterranean Population	≤1 in 6	98%	≤1 in 229	≤1 in 457,556
			Mediterranean Population†	1 in 500	98%	≤1 in 229	≤1 in 457,556
			African/African American Population	1 in 30	98%	1 in 1,451	1 in 5,804,000
HBA2	Alpha thalassemia	AR	General Population	1 in 1000	98%	1 in 860	1 in 3,440,364
			General Population†	1 in 18	98%	1 in 860	1 in 3,440,364
			Southeast Asian Population	≤1 in 7	98%	≤1 in 305	≤1 in 17,228
			Southeast Asian Population†	≤1 in 14	98%	≤1 in 305	≤1 in 17,228
			Mediterranean Population	≤1 in 6	98%	≤1 in 229	≤1 in 457,556
			Mediterranean Population†	1 in 500	98%	≤1 in 229	≤1 in 457,556
			African/African American Population	1 in 30	98%	1 in 1,451	1 in 5,804,000
HBB	Sickle cell disease	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			African/African American Population	1 in 10	95%	1 in 181	1 in 7,240
			East Asian Population	1 in 50	95%	1 in 981	1 in 196,200
			Latino Population	1 in 128	95%	1 in 2,541	1 in 1,300,992
			Mediterranean Population	1 in 3	95%	1 in 41	1 in 492
			South Asian/Indian Population	1 in 25	95%	1 in 481	1 in 48,100
HBB	Hemoglobin C disease	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			African/African American Population	1 in 10	95%	1 in 181	1 in 7,240
			East Asian Population	1 in 50	95%	1 in 981	1 in 196,200
			Latino Population	1 in 128	95%	1 in 2,541	1 in 1,300,992
			Mediterranean Population	1 in 3	95%	1 in 41	1 in 492
			South Asian/Indian Population	1 in 25	95%	1 in 481	1 in 48,100
HBB	Beta thalassemia	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			African/African American Population	1 in 10	95%	1 in 181	1 in 7,240
			East Asian Population	1 in 50	95%	1 in 981	1 in 196,200
			Latino Population	1 in 128	95%	1 in 2,541	1 in 1,300,992
			Mediterranean Population	1 in 3	95%	1 in 41	1 in 492
			South Asian/Indian Population	1 in 25	95%	1 in 481	1 in 48,100
HCFC1	Methylmalonic acidemia with homocystinuria, type cblX	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
HELLS	Immunodeficiency, Centromeric region instability, Facial anomalies syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
HEXA	Tay-Sachs disease	AR	General Population	1 in 300	99%	1 in 29,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 27	99%	1 in 2,601	1 in 280,908
			Moroccan Jewish Population	1 in 110	99%	1 in 10,901	1 in 4,796,440
HEXB	Sandhoff disease	AR	General Population	1 in 600	98%	1 in 29,951	<1 in 10 million
HFE	Hereditary Hemochromatosis	AR	General Population	1 in 10	99%	1 in 901	1 in 36,040
			African/African American Population	1 in 17	99%	1 in 1,601	1 in 108,868
			Caucasian / European Population	1 in 3	99%	1 in 201	1 in 2,412
			East Asian Population	1 in 12	99%	1 in 1,101	1 in 52,848
			Latino Population	1 in 6	99%	1 in 501	1 in 12,024
HGD	Alkaptonuria	AR	General Population	1 in 250	90%	1 in 2,491	1 in 2,491,000
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	AR	General Population	1 in 434	98%	1 in 21,651	<1 in 10 million
			Caucasian / European Population	1 in 345	98%	1 in 17,201	<1 in 10 million
HINT1	Neuromyotonia and axonal neuropathy	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
HJV	Hemochromatosis, type 2A	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
HLCS	Holocarboxylase synthetase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
HMGCS2	3-hydroxy-3-methylglutaryl-CoA synthase 2 deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
HOGA1	Primary hyperoxaluria type III	AR	General Population	1 in 184	99%	1 in 18,301	<1 in 10 million
HPD	Tyrosinemia type III	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
HPS1	Hermansky-Pudlak syndrome 1	AR	General Population	1 in 354	98%	1 in 17,651	<1 in 10 million
			Puerto Rican Population	1 in 21	98%	1 in 1,001	1 in 84,084
HPS3	Hermansky-Pudlak syndrome 3	AR	General Population	1 in 354	98%	1 in 17,651	<1 in 10 million
HPS4	Hermansky-Pudlak syndrome 4	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
HPS5	Hermansky-Pudlak syndrome 5	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
HPS6	Hermansky-Pudlak syndrome 6	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
HSD17B10	HSD10 mitochondrial disease	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
HSD17B3	17-Beta-Hydroxysteroid Dehydrogenase Deficiency	AR	General Population Palestinian Population	1 in 192 1 in 8	98% 98%	1 in 9,551 1 in 351	1 in 7,335,168 1 in 11,232
HSD17B4	D-bifunctional protein deficiency	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
HSD3B2	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
HSD3B7	Congenital bile acid synthesis defect	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
HYAL1	Mucopolysaccharidosis type IX	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
HYLS1	Hydrolethalus syndrome	AR	General Population Finnish Population	<1 in 500 1 in 50	98% 98%	1 in 24,951 1 in 2,451	<1 in 10 million 1 in 490,200
IDH3B	Retinitis pigmentosa, IDH3B-related	AR	General Population	1 in 296	99%	1 in 29,501	<1 in 10 million
IDS	Mucopolysaccharidosis type II (Hunter syndrome)	XL	General Population	1 in 50,000	91%	1 in 555,545	1 in 2,222,204
IDUA	Mucopolysaccharidosis, type I (Hurler syndrome)	AR	General Population Caucasian / European Population	<1 in 500 1 in 153	95% 95%	1 in 9,981 1 in 3,041	<1 in 10 million 1 in 1,861,092
IFT140	Mainzer-Saldino syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
IFT140	Asphyxiating thoracic dystrophy	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
IGHMBP2	Charcot-Marie-Tooth disease type 2S	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
IGHMBP2	Spinal muscular atrophy with respiratory distress 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
IGSF1	X-linked central hypothyroidism and testicular enlargement	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
IKBKB	Immunodeficiency 15B	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
IL1RAPL1	X-linked intellectual disability, IL1RAPL1-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
IL2RA	Immunodeficiency due to CD25 deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
IL2RG	Severe combined immunodeficiency, X-linked	XL	General Population	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
IL7R	Severe Combined Immunodeficiency 104	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
INPP5E	Joubert syndrome 1	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
INVS	Nephronophthisis 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ITGA2B	Glanzmann thrombasthenia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ITGA6	Junctional epidermolysis bullosa	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ITGB3	Glanzmann thrombasthenia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ITGB4	Junctional epidermolysis bullosa	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ITPA	Developmental and epileptic encephalopathy 35	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
IVD	Isovaleric Acidemia	AR	General Population African/African American Population Caucasian / European Population East Asian Population	1 in 167 1 in 100 1 in 115 1 in 407	90% 90% 90% 90%	1 in 1,661 1 in 991 1 in 1,141 1 in 4,061	1 in 1,109,548 1 in 396,400 1 in 524,860 1 in 6,611,308
IYD	Thyroid dyshormonogenesis, IYD-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
JAK3	Severe combined immunodeficiency, JAK3-related	AR	General Population	1 in 299	99%	1 in 29,801	<1 in 10 million
KCNJ1	Bartter syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
KCNJ1	Bartter syndrome type 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
KCNJ11	Congenital hyperinsulinism	AR	General Population Caucasian / European Population	1 in 423 1 in 232	99% 99%	1 in 42,201 1 in 23,101	<1 in 10 million <1 in 10 million
KCNJ11	Permanent neonatal diabetes mellitus	AR	General Population Caucasian / European Population	1 in 423 1 in 232	99% 99%	1 in 42,201 1 in 23,101	<1 in 10 million <1 in 10 million
KCTD7	Progressive myoclonic epilepsy type 3	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
KDM5C	X-linked intellectual disability, KDM5C-related	XL	General Population	<1 in 50,000	98%	1 in 2,499,951	<1 in 10 million
KIF14	Primary Autosomal Recessive Microcephaly 20	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
L1CAM	L1 syndrome	XL	General Population	1 in 15,000	99%	1 in 1,499,901	1 in 5,999,804
LAMA2	Muscular dystrophy, LAMA2-related	AR	General Population Caucasian / European Population	<1 in 500 1 in 125	99% 99%	1 in 49,901 1 in 12,401	<1 in 10 million 1 in 6,200,500
LAMA3	Junctional epidermolysis bullosa, LAMA3-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
LAMA3	Laryngo-onycho-cutaneous syndrome	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
LAMB3	Junctional epidermolysis bullosa, LAMB3-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
LAMC2	Junctional epidermolysis bullosa, LAMC2-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
LARS	Infantile liver failure syndrome 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
LCA5	Leber congenital amaurosis 5	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
LCK	Immunodeficiency 22	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
LDLR	Familial Hypercholesterolemia	AD	General Population Amish Population Caucasian / European Population French Canadian Population	1 in 8 1 in 2 1 in 7 1 in 8	99% 99% 99% 99%	1 in 701 1 in 101 1 in 601 1 in 701	1 in 22,432 1 in 808 1 in 16,828 1 in 22,432
LDLR	Familial hypercholesterolemia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
LDLRAP1	Familial Hypercholesterolemia	AR	General Population Amish Population Caucasian / European Population French Canadian Population	1 in 8 1 in 2 1 in 7 1 in 8	99% 99% 99% 99%	1 in 701 1 in 101 1 in 601 1 in 701	1 in 22,432 1 in 808 1 in 16,828 1 in 22,432
LHCGR	Leydig cell hypoplasia	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
LHX3	Combined pituitary hormone deficiency 3	AR	General Population	1 in 45	98%	1 in 2,201	1 in 396,180
LIFR	Stuve-Wiedemann syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
LIG4	LIG4 syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
LIG4	Severe combined immunodeficiency with sensitivity to ionizing radiation	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
LIPA	Lysosomal acid lipase deficiency	AR	General Population Caucasian / European Population Iranian Jewish Population	<1 in 500 1 in 112 1 in 26	99% 99% 99%	1 in 49,901 1 in 11,101 1 in 2,501	<1 in 10 million 1 in 4,973,248 1 in 260,104
LIPN	Congenital Ichthyosis 8	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
LMAN1	Combined factor V and VIII deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
LOXHD1	Nonsyndromic hearing loss 77	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 180	98% 98%	1 in 24,951 1 in 8,951	<1 in 10 million 1 in 6,444,720
LPAR6	Hypotrichosis 8	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
LPL	Familial lipoprotein lipase deficiency	AR	General Population French Canadian Population	1 in 500 1 in 46	99% 99%	1 in 49,901 1 in 4,501	<1 in 10 million 1 in 828,184
LRAT	Leber congenital amaurosis 14	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
LRP2	Donnai–Barrow syndrome	AR	General Population	1 in 214	99%	1 in 10,651	1 in 9,117,256
LRPPRC	Leigh syndrome with Complex IV deficiency	AR	General Population Faroese Population French Canadian Population	1 in 447 1 in 21 1 in 22	98% 98% 98%	1 in 22,301 1 in 1,001 1 in 1,051	<1 in 10 million 1 in 84,084 1 in 92,488
LTBP4	Cutis laxa with severe pulmonary, gastrointestinal, and urinary abnormalities	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
LYST	Chediak-Higashi syndrome	AR	General Population	<1 in 500	90%	1 in 4,991	1 in 9,982,000
MAK	Retinitis Pigmentosa 62	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MALT1	Immunodeficiency 12	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MAN2B1	Alpha-Mannosidosis	AR	General Population Caucasian / European Population	1 in 354 1 in 274	99% 99%	1 in 35,301 1 in 27,301	<1 in 10 million <1 in 10 million
MANBA	Beta-Mannosidosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MAT1A	Methionine adenosyltransferase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency (3-MCC deficiency)	AR	General Population	1 in 95	98%	1 in 4,701	1 in 1,786,380
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency (3-MCC deficiency)	AR	General Population	1 in 95	98%	1 in 4,701	1 in 1,786,380
MCEE	Methylmalonyl-CoA epimerase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MCOLN1	Mucolipidosis IV	AR	General Population Ashkenazi Jewish Population	1 in 300 1 in 100	99% 99%	1 in 29,901 1 in 9,901	<1 in 10 million 1 in 3,960,400
MCPH1	Primary microcephaly 1, recessive	AR	General Population	1 in 147	99%	1 in 14,601	1 in 8,585,388
MED17	Postnatal Progressive Microcephaly with Seizures and Brain Atrophy	AR	General Population Bukharan/Kurdish Jewish Population	<1 in 500 1 in 20	99% 99%	1 in 49,901 1 in 1,901	<1 in 10 million 1 in 152,080
MEFV	Familial Mediterranean fever	AR	General Population Mediterranean Population	1 in 20 1 in 7	99% 90%	1 in 1,901 1 in 61	1 in 152,080 1 in 1,708
MEGF8	Carpenter syndrome 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MESP2	Spondylocostal dysostosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
MFSD8	Neuronal ceroid lipofuscinosis, MFSD8-related	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
MID1	Opitz GBBB syndrome, type I	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
MKKS	Bardet-Biedl syndrome 6	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MKS1	Bardet-Biedl syndrome 13	AR	General Population Finnish Population	1 in 260 1 in 47	98% 98%	1 in 12,951 1 in 2,301	<1 in 10 million 1 in 432,588
MKS1	Joubert syndrome 28	AR	General Population Finnish Population	1 in 260 1 in 47	98% 98%	1 in 12,951 1 in 2,301	<1 in 10 million 1 in 432,588
MKS1	Meckel syndrome 1	AR	General Population Finnish Population	1 in 260 1 in 47	98% 98%	1 in 12,951 1 in 2,301	<1 in 10 million 1 in 432,588
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	AR	General Population Libyan Jewish Population	<1 in 500 1 in 40	99% 99%	1 in 49,901 1 in 3,901	<1 in 10 million 1 in 624,160
MLYCD	Malonyl-CoA decarboxylase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
MMAA	Methylmalonic aciduria, cblA type	AR	General Population	1 in 301	97%	1 in 10,001	<1 in 10 million
MMAB	Methylmalonic aciduria, cblB type	AR	General Population	1 in 435	98%	1 in 21,701	<1 in 10 million
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	AR	General Population	1 in 134	90%	1 in 1,331	1 in 713,416
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
MPI	Congenital disorder of glycosylation type Ib	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
MPL	Congenital amegakaryocytic thrombocytopenia	AR	General Population Ashkenazi Jewish Population	1 in 102 1 in 55	98% 98%	1 in 5,051 1 in 2,701	1 in 2,060,808 1 in 594,220
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	AR	General Population Native American Population	<1 in 500 1 in 20	96% 96%	1 in 12,476 1 in 476	<1 in 10 million 1 in 38,080
MRE11	Ataxia-Telangiectasia-Like Disorder 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MTHFD1	Combined immunodeficiency and megaloblastic anemia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MTHFR	Homocystinuria, MTHFR-related	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
MTM1	Myotubular myopathy, X-linked	XL	General Population	1 in 25,000	98%	1 in 1,249,951	1 in 4,999,904
MTMR2	Charcot-Marie-Tooth disease, type 4B1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MTR	Methylmalonic acidemia, cblG type	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MTRR	Homocystinuria-megaloblastic anemia, cobalamin E type	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
MTTP	Abetalipoproteinemia	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 180	98% 98%	1 in 24,951 1 in 8,951	<1 in 10 million 1 in 6,444,720
MUT	Methylmalonic aciduria–methylmalonyl–CoA mutase deficiency	AR	General Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
MVK	Hyperimmunoglobulinemia D syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MVK	Mevalonate kinase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MYO15A	Nonsyndromic hearing loss, MYO15A-related	AR	General Population Balinese Population Pakistani Population	1 in 500 1 in 6 1 in 77	98% 98% 98%	1 in 24,951 1 in 251 1 in 3,801	<1 in 10 million 1 in 6,024 1 in 1,170,708
MYO7A	Usher syndrome, type 1B	AR	General Population East Asian Population	1 in 206 1 in 62	98% 98%	1 in 10,251 1 in 3,051	1 in 8,446,824 1 in 756,648
MYO7A	Non-syndromic hearing loss, MYO7A-related	AR	General Population East Asian Population	1 in 206 1 in 62	98% 98%	1 in 10,251 1 in 3,051	1 in 8,446,824 1 in 756,648
NAGA	Schindler disease types 1 and 3	AR	General Population	1 in 94	99%	1 in 9,301	1 in 3,497,176
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	AR	General Population Caucasian / European Population East Asian Population	<1 in 500 1 in 346 1 in 298	99% 99% 99%	1 in 49,901 1 in 34,501 1 in 29,701	<1 in 10 million <1 in 10 million <1 in 10 million
NAGS	N-acetylglutamate synthase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
NBAS	SOPH syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NBEAL2	Gray platelet syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NBN	Nijmegen breakage syndrome	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
NCF2	Chronic granulomatous disease 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NCF4	Chronic granulomatous disease 4	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NDP	Norrie disease	XL	General Population	<1 in 50,000	98%	1 in 2,499,951	<1 in 10 million
NDRG1	Charcot-Marie-Tooth disease, type 4D	AR	General Population	1 in 22	98%	1 in 1,051	1 in 92,488
NDUFA11	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NDUFAF2	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NDUFAF5	Mitochondrial complex I deficiency (Leigh syndrome)	AR	General Population Ashkenazi Jewish Population	1 in 447 1 in 290	98% 98%	1 in 22,301 1 in 14,451	<1 in 10 million <1 in 10 million
NDUFS4	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
NDUFS4	Mitochondrial complex I deficiency	AR	General Population Hutterite Population	<1 in 500 1 in 27	99% 99%	1 in 49,901 1 in 2,601	<1 in 10 million 1 in 280,908
NDUFS6	Mitochondrial complex I deficiency (Leigh syndrome)	AR	General Population Bukharan/Kurdish Jewish Population	<1 in 500 1 in 24	99% 99%	1 in 49,901 1 in 2,301	<1 in 10 million 1 in 220,896
NDUFS7	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NEB	Nemaline myopathy	AR	General Population Amish Population Ashkenazi Jewish Population Finnish Population	1 in 112 1 in 11 1 in 108 1 in 112	98% 98% 98% 98%	1 in 5,551 1 in 501 1 in 5,351 1 in 5,551	1 in 2,486,848 1 in 22,044 1 in 2,311,632 1 in 2,486,848
NEU1	Sialidosis, type I and II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NGLY1	Congenital disorder of deglycosylation	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NIPAL4	Autosomal Recessive Congenital Ichthyosis 6	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NONO	X-linked intellectual disability syndrome 34	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
NPC1	Niemann-Pick disease, type C1	AR	General Population	1 in 194	90%	1 in 1,931	1 in 1,498,456
NPC2	Niemann-Pick disease, type C2	AR	General Population	1 in 194	99%	1 in 19,301	<1 in 10 million
NPHP1	Joubert syndrome 4	AR	General Population Finnish Population	1 in 480 1 in 124	98% 98%	1 in 23,951 1 in 6,151	<1 in 10 million 1 in 3,050,896
NPHP1	Nephronophthisis	AR	General Population Finnish Population	1 in 480 1 in 124	98% 98%	1 in 23,951 1 in 6,151	<1 in 10 million 1 in 3,050,896
NPHP1	NPHP1-related disorders	AR	General Population Finnish Population	1 in 480 1 in 124	98% 98%	1 in 23,951 1 in 6,151	<1 in 10 million 1 in 3,050,896
NPHP1	Senior-Løken syndrome 1	AR	General Population Finnish Population	1 in 480 1 in 124	98% 98%	1 in 23,951 1 in 6,151	<1 in 10 million 1 in 3,050,896
NPHP3	Nephronophthisis 3	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NPHP3	Meckel syndrome 7	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NPHS1	Congenital nephrotic syndrome, type 1	AR	General Population Finnish Population	1 in 289 1 in 50	98% 98%	1 in 14,401 1 in 2,451	<1 in 10 million 1 in 490,200
NPHS2	Congenital nephrotic syndrome, type 2	AR	General Population Finnish Population	1 in 289 1 in 50	98% 98%	1 in 14,401 1 in 2,451	<1 in 10 million 1 in 490,200
NR0B1	Congenital adrenal hypoplasia, X-linked	XL	General Population	1 in 6,250	99%	1 in 624,901	1 in 2,499,804
NR2E3	Retinitis pigmentosa 37	AR	General Population	1 in 209	98%	1 in 10,401	1 in 8,695,236
NR2E3	Enhanced S-cone syndrome	AR	General Population	1 in 209	98%	1 in 10,401	1 in 8,695,236
NTRK1	Congenital insensitivity to pain with anhidrosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
OAT	Gyrate atrophy of choroid and retina	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
OBSL1	3M syndrome 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
OCA2	Oculocutaneous albinism type II	AR	General Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
OCRL	Lowe syndrome	XL	General Population	1 in 250,000	95%	1 in 4,999,981	<1 in 10 million
OCRL	Dent disease 2	XL	General Population	1 in 250,000	95%	1 in 4,999,981	<1 in 10 million
OPA3	Costeff syndrome	AR	General Population Iraqi Jewish Population	<1 in 500 1 in 50	98% 98%	1 in 24,951 1 in 2,451	<1 in 10 million 1 in 490,200
OPHN1	X-linked intellectual disability with cerebellar hypoplasia and distinctive facial appearance	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
OTC	Ornithine transcarbamylase deficiency	XL	General Population	1 in 7,000	90%	1 in 69,991	1 in 279,984
OTOF	Nonsyndromic hearing loss, OTOF-related	AR	General Population Spanish Population	<1 in 500 1 in 106	99% 99%	1 in 49,901 1 in 10,501	<1 in 10 million 1 in 4,452,424
P3H1	Osteogenesis imperfecta, type VIII	AR	General Population West African Population African American Population	<1 in 500 1 in 67 1 in 250	99% 99% 99%	1 in 49,901 1 in 6,601 1 in 24,901	<1 in 10 million 1 in 1,769,068 <1 in 10,000,000
PAH	Phenylalanine Hydroxylase deficiency (Phenylketonuria)	AR	General Population Caucasian / European Population Middle-Eastern Population South East Asian	1 in 93 1 in 63 1 in 74 1 in 59	99% 99% 99% 99%	1 in 9,201 1 in 6,201 1 in 7,301 1 in 5,801	1 in 3,422,772 1 in 1,562,652 1 in 2,161,096 1 in 1,369,036
PAK3	X-linked intellectual disability, PAK3-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PANK2	Pantothenate kinase-associated neurodegeneration	AR	General Population	1 in 289	99%	1 in 28,801	<1 in 10 million
PC	Pyruvate carboxylase deficiency	AR	General Population	1 in 250	95%	1 in 4,981	1 in 4,981,000

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
PCBD1	Tetrahydrobiopterin deficiency, PCBD1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PCCA	Propionic acidemia, PCCA-related	AR	General Population Native American Population	1 in 224 1 in 85	96% 96%	1 in 5,576 1 in 2,101	1 in 4,996,096 1 in 714,340
PCCB	Propionic acidemia, PCCB-related	AR	General Population Native American Population	1 in 224 1 in 85	99% 99%	1 in 22,301 1 in 8,401	<1 in 10 million 1 in 2,856,340
PCDH15	Non-syndromic hearing loss, PCDH15-related	AR	General Population Ashkenazi Jewish Population	1 in 395 1 in 72	98% 98%	1 in 19,701 1 in 3,551	1 in 78,804 1 in 14,204
PCDH15	Usher syndrome, type 1F	AR	General Population Ashkenazi Jewish Population	1 in 395 1 in 72	98% 98%	1 in 19,701 1 in 3,551	1 in 78,804 1 in 14,204
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
PDE6A	Retinitis pigmentosa, PDE6A-related	AR	General Population	1 in 133	99%	1 in 13,201	1 in 7,022,932
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	XL	General Population	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PDHB	Pyruvate dehydrogenase E1-beta deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
PDHX	Pyruvate dehydrogenase E3-binding protein deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PDP1	Pyruvate dehydrogenase phosphatase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PEPD	Prolidase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PET100	Mitochondrial complex IV deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PEX1	Zellweger syndrome, PEX1-related	AR	General Population	1 in 147	95%	1 in 2,921	1 in 1,717,548
PEX10	Zellweger syndrome, PEX10-related	AR	General Population Japanese Population	1 in 500 1 in 354	95% 95%	1 in 9,981 1 in 7,061	<1 in 10 million 1 in 9,998,376
PEX11B	Zellweger spectrum disorder	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PEX12	Zellweger syndrome, PEX12-related	AR	General Population	1 in 373	95%	1 in 7,441	<1 in 10 million
PEX13	Zellweger spectrum disorder	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PEX14	Zellweger spectrum disorder	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PEX16	Zellweger spectrum disorder	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PEX19	Zellweger spectrum disorder	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PEX2	Zellweger syndrome, PEX2-related	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 123	95% 95%	1 in 9,981 1 in 2,441	<1 in 10 million 1 in 1,200,972
PEX26	Zellweger syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PEX3	Zellweger spectrum disorder	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PEX5	Zellweger spectrum disorder	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PEX6	Zellweger syndrome, PEX6-related	AR	General Population Yemenite Jewish Population	1 in 280 1 in 18	99% 99%	1 in 27,901 1 in 1,701	<1 in 10 million 1 in 122,472
PEX7	Rhizomelic chondrodysplasia punctata, type 1	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
PFKM	Glycogen storage disease VII	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 120	99% 99%	1 in 49,901 1 in 11,901	<1 in 10 million 1 in 5,712,480
PGK1	Phosphoglycerate kinase 1 deficiency	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PGM3	Immunodeficiency 23	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PHF8	X-linked intellectual disability, Siderius type	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 280	98% 98%	1 in 24,951 1 in 13,951	<1 in 10 million <1 in 10 million
PHKA1	Glycogen storage disease type IXd	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PHKA2	Glycogen storage disease type IXa	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PHKB	Glycogen storage disease type IXb	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PHKG2	Glycogen storage disease type IXc	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PHYH	Refsum disease	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PIGN	Multiple congenital anomalies hypotonia seizures syndrome 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PIP5K1C	Lethal congenital contractural syndrome 3	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PJVK	Nonsyndromic hearing loss 59	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PKHD1	Polycystic kidney disease, PKHD1-related	AR	General Population Ashkenazi Jewish Population	1 in 70 1 in 107	98% 98%	1 in 3,451 1 in 5,301	1 in 966,280 1 in 2,268,828
PLA2G6	Infantile neuroaxonal dystrophy	AR	General Population	1 in 500	97%	1 in 16,634	<1 in 10 million
PLEKHG5	Charcot-Marie-Tooth disease type C	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PLEKHG5	Distal spinal muscular atrophy type 4	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
PLOD1	Ehlers-Danlos syndrome with kyphoscoliosis, PLOD1-related	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
PLOD2	Bruck syndrome 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PLP1	Spastic paraplegia type 2	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PLP1	Pelizaeus-Merzbacher disease	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PMM2	Congenital disorder of glycosylation type 1a	AR	General Population Ashkenazi Jewish Population Caucasian / European Population	1 in 63 1 in 57 1 in 71	99% 99% 99%	1 in 6,201 1 in 5,601 1 in 7,001	1 in 1,562,652 1 in 1,277,028 1 in 1,988,284
PNP	Purine nucleoside phosphorylase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PNPLA1	Autosomal recessive congenital ichthyosis 10	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POLG	Ataxia neuropathy spectrum	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	Progressive external ophthalmoplegia	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	Myocerebrohepatopathy syndrome	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	POLG-related disorders	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	Alpers-Huttenlocher syndrome	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLH	Xeroderma pigmentosum	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POLR1C	Hypomyelinating Leukodystrophy, POLR1C-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POLR1C	Treacher Collins syndrome, POLR1C-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POMGNT1	Muscular dystrophy-dystroglycanopathy	AR	General Population Finnish Population	1 in 462 1 in 111	98% 98%	1 in 23,051 1 in 5,501	<1 in 10 million 1 in 2,442,444
POMGNT1	Retinitis pigmentosa 76	AR	General Population Finnish Population	1 in 462 1 in 111	98% 98%	1 in 23,051 1 in 5,501	<1 in 10 million 1 in 2,442,444
POMT1	Muscular dystrophy-dystroglycanopathy, POMT1-related	AR	General Population	1 in 290	99%	1 in 28,901	<1 in 10 million
POMT2	Muscular dystrophy-dystroglycanopathy, POMT2-related	AR	General Population	1 in 371	99%	1 in 37,001	<1 in 10 million
POR	Antley-Bixler syndrome	AR	General Population	1 in 159	98%	1 in 7,901	1 in 5,025,036
POU1F1	Combined pituitary hormone deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POU3F4	X-linked hearing loss, POU3F4-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PPIB	Osteogenesis imperfecta, type IX	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	AR	General Population Caucasian / European Population Finnish Population	1 in 368 1 in 488 1 in 75	98% 98% 98%	1 in 18,351 1 in 24,351 1 in 3,701	<1 in 10 million <1 in 10 million 1 in 1,110,300
PQBP1	Renpenning syndrome	XL	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PRCD	Retinitis pigmentosa 36	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PRDM5	Brittle cornea syndrome 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	AR	General Population	1 in 149	99%	1 in 14,801	1 in 8,821,396
PRICKLE1	Progressive myoclonic epilepsy, type 1B	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PRKDC	PRKDC-related immunodeficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PROP1	Combined pituitary hormone deficiency 2	AR	General Population	1 in 45	98%	1 in 2,201	1 in 396,180
PRPS1	Rosenberg-Chutorian syndrome	XL	General Population	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PRPS1	Arts syndrome	XL	General Population	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PRPS1	Non-syndromic hearing loss, PRPS1-related	XL	General Population	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PRPS1	Phosphoribosylpyrophosphate synthetase superactivity	XL	General Population	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PSAP	Metachromatic leukodystrophy due to saposin-b deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
PTPRC	PTPRC related-severe combined immunodeficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PTS	Tetrahydrobiopterin deficiency	AR	General Population	1 in 354	96%	1 in 8,826	<1 in 10 million
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
PYGL	Glycogen storage disease VI	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PYGM	Glycogen storage disease type V	AR	General Population Caucasian / European Population	<1 in 500 1 in 206	99% 99%	1 in 49,901 1 in 20,501	<1 in 10 million <1 in 10 million
QDPR	Tetrahydrobiopterin deficiency, QDPR-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
RAB23	Carpenter syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
RAG1	Omenn syndrome, RAG1-related	AR	General Population	1 in 290	98%	1 in 14,451	1 in 16,763,160
RAG2	Omenn syndrome, RAG2-related	AR	General Population	1 in 137	98%	1 in 6,801	1 in 3,726,948
RAPSN	Congenital myasthenic syndrome, RAPSN-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RAPSN	Fetal akinesia deformation sequence	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RARS2	Pontocerebellar hypoplasia type 6	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
RAX	Microphthalmia, isolated 3	AR	General Population	1 in 289	99%	1 in 28,801	<1 in 10 million
RD3	Leber congenital amaurosis 12	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RDH12	Leber congenital amaurosis type 13	AR	General Population Caucasian / European Population	<1 in 500 1 in 456	98% 98%	1 in 24,951 1 in 22,751	<1 in 10 million <1 in 10 million
RDH5	Fundus albipunctatus	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RFX5	Bare lymphocyte syndrome type II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RFXANK	MHC class II deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RFXAP	Bare lymphocyte syndrome type II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RHAG	Rh Deficiency syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RLBP1	Retinal dystrophy, RLBP1-related	AR	General Population Caucasian / European Population	1 in 296 1 in 84	98% 98%	1 in 14,751 1 in 4,151	<1 in 10 million 1 in 1,394,736
RMRP	Metaphyseal dysplasia without hypotrichosis	AR	General Population Amish Population Finnish Population	<1 in 500 1 in 16 1 in 76	99% 99% 99%	1 in 49,901 1 in 1,501 1 in 7,501	<1 in 10 million 1 in 96,064 1 in 2,280,304
RMRP	Cartilage-Hair Hypoplasia Anauxetic Dysplasia Spectrum Disorder	AR	General Population Amish Population Finnish Population	<1 in 500 <1 in 500 <1 in 500	99% 99% 99%	1 in 49,901 1 in 49,901 1 in 49,901	<1 in 10 million <1 in 10 million <1 in 10 million
RMRP	Anauxetic dysplasia	AR	General Population Amish Population Finnish Population	<1 in 500 1 in 16 1 in 76	99% 99% 99%	1 in 49,901 1 in 1,501 1 in 7,501	<1 in 10 million 1 in 96,064 1 in 2,280,304
RMRP	Cartilage-hair hypoplasia	AR	General Population Amish Population Finnish Population	<1 in 500 1 in 16 1 in 76	99% 99% 99%	1 in 49,901 1 in 1,501 1 in 7,501	<1 in 10 million 1 in 96,064 1 in 2,280,304
RNASEH2A	Aicardi-Goutieres syndrome 4	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RNASEH2B	Aicardi Goutieres syndrome 2	AR	General Population	1 in 217	99%	1 in 10,801	1 in 9,375,268
RNASEH2C	Aicardi-Goutieres syndrome 3	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ROGDI	Kohlschutter-Tonz syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RP2	X-linked Retinitis pigmentosa, RP2-related	XL	General Population	1 in 4,000	99%	1 in 399,901	1 in 1,600,000
RPE65	Retinitis pigmentosa 20	AR	General Population	1 in 228	98%	1 in 11,351	<1 in 10 million
RPE65	Leber congenital amaurosis 2	AR	General Population	1 in 228	98%	1 in 11,351	<1 in 10 million
RPGR	X-linked Retinitis pigmentosa, RPGR-related	XL	General Population	1 in 3,000	75%	1 in 11,997	1 in 48,000
RPGRIP1	Leber congenital amaurosis and Cone-rod dystrophy	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RPGRIP1L	COACH syndrome	AR	General Population	1 in 259	98%	1 in 12,901	<1 in 10 million
RPGRIP1L	Joubert syndrome 7	AR	General Population	1 in 259	98%	1 in 12,901	<1 in 10 million
RPGRIP1L	Meckel syndrome 5	AR	General Population	1 in 259	98%	1 in 12,901	<1 in 10 million
RS1	Juvenile retinoschisis, X-linked	XL	General Population	1 in 2,500	96%	1 in 62,476	1 in 249,956
RSPH9	Primary ciliary dyskinesia 12	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RTEL1	Dyskeratosis congenita type 5	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 203	99% 99%	1 in 49,901 1 in 20,201	<1 in 10 million <1 in 10 million
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR	General Population French Canadian Population	<1 in 500 1 in 19	95% 95%	1 in 9,981 1 in 361	<1 in 10 million 1 in 27,436
SAG	Retinitis pigmentosa 47	AR	General Population	1 in 228	98%	1 in 11,351	<1 in 10 million
SAMD9	Normophosphatemic Familial Tumoral Calcinosis	AR	General Population Yemeni Jewish Population	<1 in 500 1 in 25	99% 99%	1 in 49,901 1 in 2,401	<1 in 10 million 1 in 240,100
SAMHD1	Aicardi-Goutieres syndrome	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SBDS	Shwachman-Diamond syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SCO1	Mitochondrial complex IV deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SCO2	Mitochondrial complex IV deficiency	AR	General Population	1 in 150	99%	1 in 14,901	1 in 8,940,600
SDCCAG8	Bardet-Biedl syndrome and Senior-Loken syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SDR9C7	Autosomal recessive congenital ichthyosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
SEC23B	Congenital dyserythropoietic anemia, type II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SELENON	Rigid spine muscular dystrophy	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SEPSECS	Pontocerebellar hypoplasia type 2D	AR	General Population Moroccan/Iraqi Jewish Population	<1 in 500 1 in 44	99% 99%	1 in 49,901 1 in 4,301	<1 in 10 million 1 in 756,976
SERPINA1	Alpha-1 antitrypsin deficiency	AR	General Population Caucasian / European Population	1 in 33 1 in 19	95% 95%	1 in 641 1 in 361	1 in 84,612 1 in 27,436
SERPINF1	Osteogenesis imperfecta, type VI	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SGCA	Limb-girdle muscular dystrophy, type 2D	AR	General Population Caucasian / European Population Finnish Population	<1 in 500 1 in 288 1 in 150	98% 98% 98%	1 in 24,951 1 in 14,351 1 in 7,451	<1 in 10 million <1 in 10 million 1 in 4,470,600
SGCB	Limb-girdle muscular dystrophy, type 2E	AR	General Population Caucasian / European Population	1 in 500 1 in 406	98% 98%	1 in 24,951 1 in 20,251	<1 in 10 million <1 in 10 million
SGCD	Limb-girdle muscular dystrophy, type 2F	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SGCG	Limb-girdle muscular dystrophy, type 2C	AR	General Population Moroccan Population Roma / Gypsy Population	1 in 381 1 in 250 1 in 96	98% 98% 98%	1 in 19,001 1 in 12,451 1 in 4,751	<1 in 10 million <1 in 10 million 1 in 1,824,384
SGSH	Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	AR	General Population Caucasian / European Population	1 in 454 1 in 253	98% 98%	1 in 22,651 1 in 12,601	<1 in 10 million <1 in 10 million
SH3TC2	Charcot-Marie-Tooth disease, SH3TC2-related	AR	General Population	1 in 69	99%	1 in 6,801	1 in 1,877,076
SKIV2L	Trichohepatoenteric syndrome 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC12A1	Bartter syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
SLC12A1	Bartter syndrome, type 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC12A3	Gitelman syndrome	AR	General Population	1 in 100	98%	1 in 4,951	1 in 1,980,400
SLC12A6	Andermann syndrome	AR	General Population French Canadian Population	<1 in 500 1 in 23	98% 99%	1 in 24,951 1 in 2,201	<1 in 10 million 1 in 202,492
SLC16A2	Allan-Herndon-Dudley syndrome	XL	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC17A5	Sialic acid storage disorder	AR	General Population Finnish Population	<1 in 500 1 in 100	91% 91%	1 in 5,545 1 in 1,101	<1 in 10 million 1 in 440,400
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC19A3	Biotin-responsive basal ganglia disease	AR	General Population	1 in 109	99%	1 in 5,401	1 in 2,354,836
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly syndrome	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 106	99% 99%	1 in 49,901 1 in 10,501	<1 in 10 million 1 in 4,452,424
SLC22A5	Systemic primary carnitine deficiency	AR	General Population African/African American Population East Asian Population Faroese Population Pacific Islander Population South Asian/Indian Population	1 in 129 1 in 86 1 in 77 1 in 9 1 in 37 1 in 51	99% 99% 99% 99% 99% 99%	1 in 12,801 1 in 8,501 1 in 7,601 1 in 801 1 in 3,601 1 in 5,001	1 in 6,605,316 1 in 2,924,344 1 in 2,341,108 1 in 28,836 1 in 532,948 1 in 1,020,204
SLC25A13	Citrin deficiency	AR	General Population East Asian Population	<1 in 500 1 in 65	95% 95%	1 in 9,981 1 in 1,281	<1 in 10 million 1 in 333,060
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	AR	General Population French Canadian Population	<1 in 500 1 in 37	99% 99%	1 in 49,901 1 in 3,601	<1 in 10 million 1 in 532,948
SLC25A20	Carnitine-acylcarnitine translocase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC26A2	Diastrophic dysplasia	AR	General Population Finnish Population	1 in 158 1 in 50	90% 90%	1 in 1,571 1 in 491	1 in 992,872 1 in 98,200
SLC26A2	Achondrogenesis, type IB	AR	General Population Finnish Population	1 in 158 1 in 50	90% 90%	1 in 1,571 1 in 491	1 in 992,872 1 in 98,200
SLC26A2	Multiple epiphyseal dysplasia	AR	General Population Finnish Population	1 in 158 1 in 50	90% 90%	1 in 1,571 1 in 491	1 in 992,872 1 in 98,200
SLC26A2	Atelosteogenesis II	AR	General Population Finnish Population	1 in 158 1 in 50	90% 90%	1 in 1,571 1 in 491	1 in 992,872 1 in 98,200
SLC26A3	Congenital secretory chloride diarrhea	AR	General Population Middle-Eastern Population	<1 in 500 1 in 57	98% 98%	1 in 24,951 1 in 2,801	<1 in 10 million 1 in 638,628
SLC26A4	Pendred syndrome	AR	General Population African/African American Population Caucasian / European Population East Asian Population	1 in 80 1 in 76 1 in 88 1 in 74	98% 98% 98% 98%	1 in 3,951 1 in 3,751 1 in 4,351 1 in 3,651	1 in 1,264,320 1 in 1,140,304 1 in 1,531,552 1 in 1,080,696
SLC27A4	Ichthyosis prematurity syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC2A10	Arterial tortuosity syndrome	AR	General Population	1 in 300	99%	1 in 29,901	<1 in 10 million
SLC2A2	Fanconi-Bickel syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
SLC34A3	Hereditary hypophosphatemic rickets with hypercalciuria	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC35A3	Arthrogryposis, intellectual disability, and seizures	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 453	98% 98%	1 in 24,951 1 in 22,601	<1 in 10 million <1 in 10 million
SLC37A4	Glycogen storage disease, type Ib	AR	General Population Ashkenazi Jewish Population	1 in 158 1 in 71	95% 95%	1 in 3,141 1 in 1,401	1 in 1,985,112 1 in 397,884
SLC39A4	Acrodermatitis enteropathica	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC3A1	Cystinuria, type I	AR	General Population Caucasian / European Population	1 in 50 1 in 42	98% 98%	1 in 2,451 1 in 2,051	1 in 490,200 1 in 344,568
SLC45A2	Oculocutaneous albinism, type IV	AR	General Population Japanese Population	1 in 159 1 in 146	98% 98%	1 in 7,901 1 in 7,251	1 in 5,025,036 1 in 4,234,584
SLC46A1	Hereditary folate malabsorption	AR	General Population Puerto Rican Population	<1 in 500 1 in 500	99% 99%	1 in 49,901 1 in 49,901	<1 in 10 million <1 in 10 million
SLC4A1	Distal Renal Tubular Acidosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC4A11	Corneal endothelial dystrophy	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC5A5	Thyroid dyshormonogenesis, SLC5A5-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC6A19	Hartnup disorder	AR	General Population	1 in 87	99%	1 in 8,601	1 in 2,993,148
SLC6A8	Creatine deficiency syndrome	XL	General Population	1 in 3,434	98%	1 in 171,651	1 in 686,716
SLC7A7	Lysinuric protein intolerance	AR	General Population Finnish Population Japanese Population	<1 in 500 1 in 122 1 in 119	95% 95% 95%	1 in 9,981 1 in 2,421 1 in 2,361	<1 in 10 million 1 in 1,181,448 1 in 1,123,836
SLC7A9	Cystinuria, non-type I	AR	General Population	1 in 42	98%	1 in 2,051	1 in 344,568
SMARCAL1	Schimke immunoosseous dysplasia	AR	General Population	1 in 500	90%	1 in 4,991	1 in 9,982,000
SMN1	Spinal muscular atrophy	AR	General Population African/African American Population Ashkenazi Jewish Population Caucasian / European Population East Asian Population Latino Population Sephardic Jewish Population	1 in 54 1 in 72 1 in 67 1 in 47 1 in 59 1 in 68 1 in 34	91% 71% 91% 95% 93% 90% 96%	1 in 590 1 in 246 1 in 734 1 in 921 1 in 830 1 in 671 1 in 826	1 in 127,440 1 in 70,848 1 in 196,712 1 in 173,148 1 in 195,880 1 in 182,512 1 in 112,336
SMN1	Spinal muscular atrophy silent carrier	AR	General Population	1 in 54	91%	1 in 590	1 in 127,440
SMPD1	Niemann-Pick disease, type A/B	AR	General Population Ashkenazi Jewish Population Latino Population	1 in 250 1 in 115 1 in 106	95% 95% 95%	1 in 4,981 1 in 2,281 1 in 2,101	1 in 4,981,000 1 in 1,049,260 1 in 890,824
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SNX10	Osteopetrosis 8	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SP110	Hepatic venoocclusive disease with immunodeficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SPATA7	Leber congenital amaurosis (LCA) and juvenile retinitis pigmentosa (RP)	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SPG11	SPG11-related Neuromuscular Disorders	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
SPG21	Mast syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SPG7	Spastic paraplegia type 7	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
SPINK5	Netherton syndrome	AR	General Population Ashkenazi Jewish Population	1 in 224 1 in 17	99% 99%	1 in 23,301 1 in 1,601	<1 in 10 million 1 in 108,868
SPR	Sepiapterin Reductase Deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SRD5A2	5-alpha reductase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ST3GAL5	Salt and pepper developmental regression syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
STAR	Lipoid congenital adrenal hyperplasia	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
STK4	Combined immunodeficiency due to STK4 deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
STX11	Familial hemophagocytic lymphohistiocytosis	AR	General Population	1 in 112	99%	1 in 11,101	1 in 4,973,248
STXBP2	Familial hemophagocytic lymphohistiocytosis	AR	General Population	1 in 112	99%	1 in 11,101	1 in 4,973,248
SUCLA2	Mitochondrial DNA depletion syndrome 5	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SUMF1	Multiple sulfatase deficiency	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 320	98% 98%	1 in 24,951 1 in 15,951	<1 in 10 million <1 in 10 million
SUOX	Sulfite oxidase deficiency	AR	General Population	1 in 300	99%	1 in 29,901	1 in 13,395,648
SURF1	Charcot-Marie-Tooth disease, SURF1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SURF1	Leigh syndrome, SURF1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
SYN1	X-linked epilepsy with variable learning disabilities	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
SYNE4	Autosomal recessive deafness 76	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TAT	Tyrosinemia, type II	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
TAZ	Barth syndrome	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
TBCE	Hypoparathyroidism-retardation-dysmorphism syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TBX19	Adrenocorticotropic hormone deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TCIRG1	Osteopetrosis 1	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
TCTN1	Joubert syndrome 13	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TCTN2	Meckel syndrome 8	AR	General Population Ethiopian Jewish Population Yemenite Jewish Population	<1 in 500 1 in 42 1 in 78	99% 99% 99%	1 in 49,901 1 in 4,101 1 in 7,701	<1 in 10 million 1 in 688,968 1 in 2,402,712
TCTN2	Joubert syndrome 24	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TCTN3	Joubert syndrome 18	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TECPR2	Spastic paraplegia 49	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
TERT	Dyskeratosis congenita type 4	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TF	Atransferrinemia	AR	General Population	1 in 116	99%	1 in 11,501	1 in 5,336,464
TFR2	Hemochromatosis, type 3	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
TG	Thyroid dyshormonogenesis, TG-related	AR	General Population	1 in 241	99%	1 in 24,001	<1 in 10 million
TGM1	Congenital ichthyosis	AR	General Population	1 in 224	95%	1 in 4,461	1 in 3,997,056
TH	Segawa syndrome	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
THOC2	X-linked Intellectual disability, THOC2-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
TK2	Mitochondrial DNA depletion syndrome 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TMC1	Nonsyndromic hearing loss 7	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TMEM138	Joubert syndrome 16	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TMEM216	Joubert syndrome 2	AR	General Population Ashkenazi Jewish Population	1 in 141 1 in 92	98% 98%	1 in 7,001 1 in 4,551	1 in 3,948,564 1 in 1,674,768
TMEM216	Meckel syndrome 2	AR	General Population Ashkenazi Jewish Population	1 in 141 1 in 92	98% 98%	1 in 7,001 1 in 4,551	1 in 3,948,564 1 in 1,674,768
TMEM231	Joubert syndrome 20	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TMEM237	Joubert syndrome 14	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TMEM38B	Osteogenesis imperfecta, type XIV	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TMEM67	COACH syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TMEM70	Mitochondrial complex V deficiency type 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TMPRSS3	Nonsyndromic hearing loss, TMPRSS3-related	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
TNFSF11	Osteopetrosis 2	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
TNXB	Ehlers–Danlos-like syndrome due to tenascin-X deficiency	AR	General Population	1 in 28	99%	1 in 2,701	1 in 302,512
TPO	Thyroid dyshormonogenesis, TPO-related	AR	General Population	1 in 373	99%	1 in 37,201	<1 in 10 million
TPP1	Neuronal ceroid lipofuscinosis, TPP1-related	AR	General Population French Canadian Population	1 in 252 1 in 53	97% 97%	1 in 8,368 1 in 1,734	1 in 8,434,944 1 in 367,608
TRAPPC11	Limb-girdle muscular dystrophy 18	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TRDN	Catecholaminergic polymorphic ventricular tachycardia	AR	General Population	1 in 354	98%	1 in 17,651	<1 in 10 million
TREX1	Aicardi-Goutieres syndrome 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TRHR	Generalized thyrotropin-releasing hormone resistance	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TRIM32	Limb-girdle muscular dystrophy, type 2H	AR	General Population Hutterite Population	<1 in 500 1 in 12	98% 98%	1 in 24,951 1 in 551	<1 in 10 million 1 in 26,448
TRIM32	Bardet-Biedl syndrome 11	AR	General Population Hutterite Population	<1 in 500 1 in 12	98% 98%	1 in 24,951 1 in 551	<1 in 10 million 1 in 26,448
TRIM37	Mulibrey nanism	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TRMU	Liver failure, acute infantile	AR	General Population Yemeni Jewish Population	<1 in 500 1 in 34	98% 98%	1 in 24,951 1 in 1,651	<1 in 10 million 1 in 224,536
TRPM6	Hypomagnesemia 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TSEN54	Pontocerebellar hypoplasia type 2A	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
TSFM	Combined oxidative phosphorylation deficiency, TSFM-related	AR	General Population Finnish Population	<1 in 500 1 in 80	98% 98%	1 in 24,951 1 in 3,951	<1 in 10 million 1 in 1,264,320

fulgent | Beacon™ Carrier Screening

Beacon Carrier Screening Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
TSHB	Congenital hypothyroidism, TSHB-related	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
TSHR	Congenital hypothyroidism, TSHR-related	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
TTC37	Trichohepatoenteric syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
TTC7A	Gastrointestinal defects and immunodeficiency syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TTC8	Bardet-Biedl syndrome 8	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TTPA	Ataxia with isolated vitamin E deficiency	AR	General Population Caucasian / European Population	<1 in 500 1 in 267	98% 90%	1 in 24,951 1 in 2,661	<1 in 10 million 1 in 2,841,948
TULP1	Leber congenital amaurosis 15	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TULP1	Retinitis pigmentosa 14	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TYMP	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
TYR	Oculocutaneous albinism types 1A and 1B	AR	General Population	1 in 20	99%	1 in 1,901	1 in 152,080
TYRP1	Oculocutaneous albinism, type III	AR	General Population African Population	<1 in 500 1 in 47	98% 98%	1 in 24,951 1 in 2,301	<1 in 10 million 1 in 432,588
UGT1A1	Crigler-Najjar syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
UNC13D	Familial hemophagocytic lymphohistiocytosis type 3	AR	General Population	1 in 149	99%	1 in 14,801	1 in 8,821,396
UPF3B	Lujan-Fryns syndrome, UPF3B-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
USH1C	Usher syndrome, type IC	AR	General Population French Canadian Population	1 in 353 1 in 227	90% 90%	1 in 3,521 1 in 2,261	1 in 4,971,652 1 in 2,052,988
USH1C	Non-syndromic hearing loss, USH1C-related	AR	General Population French Canadian Population	1 in 353 1 in 227	90% 90%	1 in 3,521 1 in 2,261	1 in 4,971,652 1 in 2,052,988
USH1G	Usher syndrome type IG	AR	General Population	1 in 434	99%	1 in 43,301	<1 in 10 million
USH2A	Usher syndrome, type 2A	AR	General Population Caucasian / European Population Ashkenazi Jewish Population Iranian Jewish Population	1 in 126 1 in 73 1 in 35 1 in 60	96% 96% 99% 99%	1 in 3,126 1 in 1,801 1 in 3,401 1 in 5,901	1 in 1,575,504 1 in 525,892 1 in 476,140 1 in 1,416,240
VDR	Vitamin D-dependent rickets, type 2A	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
VLDLR	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
VPS13A	Choreoacanthocytosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
VPS13B	Cohen syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
VPS45	Severe congenital neutropenia, VPS45-related	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
VPS53	Pontocerebellar hypoplasia type 2E	AR	General Population Moroccan Jewish Population	<1 in 500 1 in 37	98% 98%	1 in 24,951 1 in 1,801	<1 in 10 million 1 in 266,548
VRK1	Pontocerebellar hypoplasia type 1A	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
VSX2	Microphthalmia with or without coloboma	AR	General Population	1 in 91	98%	1 in 4,501	1 in 1,638,364
WAS	Thrombocytopenia, X-linked	XL	General Population	1 in 125,000	99%	1 in 12,499,901	<1 in 10 million
WAS	Severe congenital neutropenia, WAS-related	XL	General Population	1 in 125,000	99%	1 in 12,499,901	<1 in 10 million
WAS	Wiskott-Aldrich syndrome	XL	General Population	1 in 125,000	99%	1 in 12,499,901	<1 in 10 million
WHRN	Usher syndrome type 2D	AR	General Population	1 in 282	99%	1 in 28,101	<1 in 10 million
WISP3	Progressive pseudorheumatoid dysplasia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
WNT1	Osteogenesis imperfecta type 15	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
WNT10A	Schopf-Schulz-Passarge syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
WNT10A	Odontoonychodermal dysplasia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
WRN	Werner syndrome	AR	General Population Caucasian / European Population Japanese Population	1 in 308 1 in 112 1 in 71	98% 98% 98%	1 in 15,351 1 in 5,551 1 in 3,501	<1 in 10 million 1 in 2,486,848 1 in 994,284
XPA	Xeroderma pigmentosum, group A	AR	General Population Japanese Population	1 in 500 1 in 74	99% 99%	1 in 49,901 1 in 7,301	<1 in 10 million 1 in 2,161,096
XPC	Xeroderma pigmentosum, group C	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
ZAP70	Infantile-onset multisystem autoimmune disease type 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ZAP70	Immunodeficiency 48	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome 2	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ZDHHC9	X-linked intellectual disability, ZDHHC9-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
ZFYVE26	Spastic paraplegia 15	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ZNF469	Brittle cornea syndrome 1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ZNF711	X-linked intellectual disability, ZNF711-related	XL	General Population	<1 in 50,000	93%	1 in 714,272	1 in 2,857,143

* For genes that have tested negative
† The carrier frequency for heterozygous alpha thalassemia carriers (αα) is described in rows marked with a dagger symbol. The carrier frequency for alpha thalassemia trait cis (α- -) is 1 in 1000.
Abbreviations: AR, autosomal recessive; XL, X-linked

