Adventia®
carrier screening

ADVENTIA CARRIER SCREENING PANELS

GENES TESTED

Adventia Core 22 genes	Alpha Thalassemia; *HBA1*, *HBA2* • Becker Muscular Dystrophy; X-linked, *DMD* • Beta Thalassemia; *HBB* • Bloom Syndrome; *BLM* • Canavan Disease; *ASPA* • Cystic Fibrosis; *CFTR* • Duchenne Muscular Dystrophy; X-linked, *DMD* • Familial Dysautonomia; *ELP1* • Fanconi Anemia, Type C; *FANCC* • Fragile X Syndrome; X-linked, *FMR1* • Galactosemia; *GALT* • Gaucher Disease; *GBA* • Medium Chain Acyl-CoA Dehydrogenase Deficiency; *ACADM* • Mucolipidosis Type IV; *MCOLN1* • Niemann-Pick Disease, Types A/B; *SMPD1* • Non-Syndromic Hearing Loss (GJB2-Related and GJB6-Related); *GJB2*, *GJB6* • Phenylalanine Hydroxylase Deficiency; *PAH* • Sickle-Cell Disease; *HBB* • Spinal Muscular Atrophy; *SMN1*, *SMN2* • Smith-Lemli-Opitz syndrome; *DHCR7* • Tay-Sachs Disease; *HEXA*
Adventia Comprehensive 231 genes	3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency; *HMGCL* • 3-Methylcrotonyl-CoA Carboxylase Deficiency 1; *MCCC1* • 3-Methylcrotonyl-CoA Carboxylase Deficiency 2; *MCCC2* • 3-Methylglutaconic Aciduria, Type 3 [Costeff Syndrome]; *OPA3* • 3-Phosphoglycerate Dehydrogenase Deficiency; *PHGDH* • 6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency; *PTS* • Abetalipoproteinemia; *MTTP* • Achondrogenesis, Type 1B; *SLC26A2* • Achromatopsia (CNGB3-related); *CNGB3* • Acute Infantile Liver Failure; (TRMU-related), *TRMU* • Acyl-CoA Oxidase I Deficiency; *ACOX1* • Adrenoleukodystrophy, X-linked; *ABCD1* • Aicardi-Goutières Syndrome; *SAMHD1* • Alpha Thalassemia; *HBA1*, *HBA2* • Alport Syndrome (COL4A3-related); *COL4A3* • Alport Syndrome, X-linked; *COL4A5* • Alstrom Syndrome; *ALMS1* • Andermann Syndrome; *SLC12A6* • Argininosuccinate Lyase Deficiency; *ASL* • Aromatase Deficiency; *CYP19A1* • Arthrogryposis Mental Retardation Seizures; *SLC35A3* • Asparagine Synthetase Deficiency; *ASNS* • Aspartylglycosaminuria; *AGA* • Ataxia with Vitamin E Deficiency; *TTPA* • Ataxia-Telangiectasia; *ATM* • Autoimmune Polyglandular Syndrome, Type 1; *AIRE* • Autosomal Recessive Polycystic Kidney Disease; *PKHD1* • Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay; *SACS* • Bardet-Biedl Syndrome (BBS1-related); *BBS1* • Bardet Biedl syndrome (BBS12-related); *BBS12* • Bare Lymphocyte Syndrome (CIITA-related); *CIITA* • Bartter Syndrome (BSND-related); *BSND* • Batten Disease (CLN3-related); *CLN3* • Becker Muscular Dystrophy; *DMD* • Beta Thalassemia; *HBB* • Biotinidase Deficiency; *BTD* • Bloom Syndrome; *BLM* • Canavan Disease; *ASPA* • Carnitine Palmitoyltransferase IA Deficiency; *CPT1A* • Carnitine Palmitoyltransferase II Deficiency; *CPT2* • Carpenter Syndrome, *RAB23* • Cartilage-Hair Hypoplasia; *RMRP* • Cerebrotendinous Xanthomatosis; *CYP27A1* • Choreacanthocytosis; *VPS13A* • Choroideremia, X-linked; *CHM* • Chronic Granulomatous Disease, X-linked; *CYBB* • Citrin Deficiency; *SLC25A13* • Citrullinemia, Type 1; *ASS1* • Combined Malonic and Methylmalonic Acidemia; *ACSF3* • Combined Oxidative Phosphorylation Deficiency 1; *GFM1* • Combined Oxidative Phosphorylation Deficiency 3; *TSFM* • Combined Pituitary Hormone Deficiency 2; *PROP1* • Congenital Disorder of Glycosylation, Type 1A (PMM2- related); *PMM2* • Congenital Disorder of Glycosylation, Type 1B; *MPI* • Congenital Disorder of Glycosylation, Type 1C; *ALG6* • Congenital Finnish Nephrosis; *NPHS1* • Congenital Insensitivity to Pain with Anhidrosis; *NTRK1* • Congenital Myasthenic Syndrome (CHRNE-related); *CHRNE* • Congenital Myasthenic Syndrome (RAPSN-related); *RAPSN* • Congenital Neutropenia (HAX1-related); *HAX1* • Congenital Neutropenia (VPS45-related); *VPS45* • Corneal Dystrophy and Perceptive Deafness; *SLC4A11* • Corticosterone Methyloxidase Deficiency; *CYP11B2* • CRB1-related Retinal Dystrophies; *CRB1* • Creatine Transporter Defect [Cerebral Creatine Deficiency syndrome 1], X-linked; *SLC6A8* • Crigler Najjar syndrome, Type I; *UGT1A1* • Cystic Fibrosis; *CFTR* • Cystinosis; *CTNS* • D-Bifunctional Protein Deficiency; *HSD17B4* • Deafness; Autosomal Recessive 77; *LOXHD1* • Duchenne Muscular Dystrophy, X-linked; *DMD* • Dystrophic Epidermolysis Bullosa (COL7A1-related); *COL7A1* • Ehlers-Danlos Syndrome, Type VIIC; *ADAMTS* • Emery-Dreifuss Muscular Dystrophy 1, X-linked; *EMD* • Enhanced S-Cone Syndrome; *NR2E3* • Ethylmalonic Encephalopathy; *ETHE1* • Fabry Disease, X-linked; *GLA* • Factor IX Deficiency, X-linked; *F9* • Factor V Leiden Thrombophilia; *F5* • Factor XI Deficiency; *F11* • Familial Dysautonomia; *ELP1* • Familial Hypercholesterolemia (LDLR-related); *LDLR* •Familial Mediterranean Fever; *MEFV* • Familial Nephrogenic Diabetes Insipidus (AQP2- related); *AQP2* • Fanconi Anemia, Type C; *FANCC* • Fanconi Anemia, Type G; *FANCG* • Fragile X syndrome, X-linked; *FMR1* • Galactokinase Deficiency [Galactosemia, Type II]; *GALK1* • Galactosemia; *GALT* • Gaucher Disease; *GBA* • Glutaric Acidemia; Type 1; *GCDH* • Glutaric Acidemia, Type 2A; *ETFA* • Glycine Encephalopathy (GLDC-related); *GLDC* • Glycine Encephalopathy (AMT-related); *AMT* • Glycogen Storage Disease, Type 1A; *G6PC* • Glycogen Storage Disease, Type 1B; *SLC37A4* • Glycogen Storage Disease, Type 2 [Pompe Disease]; *GAA* • Glycogen Storage Disease, Type 3; *AGL* • Glycogen Storage Disease, Type 4; *GBE1* • Glycogen Storage Disease, Type 5 [McArdle Disease]; *PYGM* • Glycogen Storage Disease, Type 7; *PFKM* • GRACILE Syndrome; *BCS1L* • Hemochromatosis, Type 2A; *HJV* • Hemochromatosis, Type 3 (TFR2 related); *TFR2* • Hereditary Fructose Intolerance; *ALDOB* • Hermansky-Pudlak Syndrome (HPS1-related); *HPS1* • Hermansky-Pudlak Syndrome (HPS3-related); *HPS3* • Holocarboxylase Synthetase Deficiency; *HLCS* • Homocystinuria due to Cystathionine Beta-synthase Deficiency; *CBS* • Homocystinuria-Megaloblastic Anemia, cblE Type; *MTRR* • Hydrolethalus Syndrome; *HYLS1* • Hypohidrotic Ectodermal Dysplasia, X-linked; *EDA* • Hypophosphatasia (ALPL-related); *ALPL* • Inclusion Body Myopathy Type 2; *GNE* • Isovaleric Acidemia; *IVD* • Joubert Syndrome, Type 2; *TMEM216* • Junctional Epidermolysis Bullosa, Herlitz type; *LAMC2* • Juvenile Retinoschisis, X-linked; *RS1* • Krabbe Disease; *GALC* • Lamellar Ichthyosis, Type 1; *TGM1* • Leber Congenital Amaurosis (LCA5-related); *LCA5* • Leber Congenital Amaurosis, Type *CEP290*; *CEP290* • Leigh Syndrome, French-Canadian Type; *LRPPRC* • Leukoencephalopathy with Vanishing White Matter; *EIF2B5* • Leydig Cell Hypoplasia [Luteinizing Hormone Resistance]; *LHCGR* • Limb-Girdle Muscular Dystrophy, Type 2A; *CAPN3* • Limb-Girdle Muscular Dystrophy, Type 2B; *DYSF* • Limb-Girdle Muscular Dystrophy, Type 2C; *SGCG* • Limb-Girdle Muscular Dystrophy, Type 2D; *SGCA* • Limb-Girdle Muscular Dystrophy, Type 2E; *SGCB* • Lipoamide Dehydrogenase Deficiency [Maple Syrup Urine Disease, Type 3]; *DLD* • Lipoid Adrenal Hyperplasia; *STAR* • Lipoprotein Lipase Deficiency; *LPL* • Long Chain 3-Hydroxyacyl-

	CoA Dehydrogenase Deficiency; *HADHA* • Lysinuric Protein Intolerance; *SLC7A7* • Maple Syrup Urine Disease, Type 1B; *BCKDHB* • Meckel-Gruber Syndrome, Type 1; *MKS1* • Medium Chain Acyl-CoA Dehydrogenase Deficiency; *ACADM* • Megalencephalic Leukoencephalopathy with Subcortical Cysts; *MLC1* • Metachromatic Leukodystrophy (ARSA-related); *ARSA* • Metachromatic Leukodystrophy (PSAP-related); *PSAP* • Methylmalonic Acidemia, cblA Type; *MMAA* • Methlymalonic Acidemia, cblB Type; *MMAB* • Methylmalonic Acidemia and Homocystinuria, Type cblC; *MMACHC* • Methylmalonic Acidemia and Homocystinuria, cblD Type; *MMADHC* • Methylmalonic Acidemia due to Methylmalonyl-CoA Mutase Deficiency; *MMUT* • Microphthalmia/Anophthalmia (VSX2-related); *VSX2* • Mitochondrial Complex 1 Deficiency (ACAD9-related); *ACAD9* • Mitochondrial Complex 1 Deficiency (NDUFAF5- related); *NDUFAF5* • Mitochondrial Complex 1 Deficiency (NDUFS6-related); *NDUFS6* • Mitochondrial Myopathy and Sideroblastic Anemia (MLASA1); *PUS1* • Mucolipidosis II/III; *GNPTAB* • Mucolipidosis III Gamma; *GNPTG* • Mucolipidosis, Type IV; *MCOLN1* • Mucopolysaccharidosis, Type II [Hunter Syndrome], X-liked; *IDS* • Mucopolysaccharidosis, Type IIIB [Sanfilippo B]; *NAGLU* • Mucopolysaccharidosis, Type IIIC [Sanfilippo C]; *HGSNAT* • Mucopolysaccharidosis IIID [Sanfilippo D]; *GNS* • Mucopolysaccharidosis, Type IX; *HYAL1* • Multiple Sulfatase Deficiency; *SUMF1* • Myoneurogastrointestinal Encephalopathy (MNGIE); *TYMP* • Myotubular Myopathy, X-linked; *MTM1* • N-acetylglutamate Synthase Deficiency; *NAGS* • Navajo Neurohepatopathy [MPV17-related Hepatocerebral Mitochondrial DNA Depletion Syndrome]; *MPV17* • Neurological Ceroid Lipofuscinosis (TPP1-related); *TPP1* • Neuronal Ceroid Lipofuscinosis (MFSD8-related); *MFSD8* • Neuronal Ceroid Lipofuscinosis (CLN5-related); *CLN5* • Neuronal Ceroid Lipofuscinosis (CLN6-related); *CLN6* • Neuronal Ceroid Lipofuscinosis (CLN8-related); *CLN8* • Neuronal Ceroid Lipofuscinosis (PPT1-related); *PPT1* • Niemann-Pick Disease, Types A/B; *SMPD1* • Niemann-Pick Disease, Type C1/D; *NPC1* • Niemann-Pick Disease, Type C2; *NPC2* • Nijmegen Breakage Syndrome; *NBN* • Non-Syndromic Hearing Loss (GJB2-related and GJB6-related); *GJB2*, *GJB6* • Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome; *WNT10A* • Omenn Syndrome (RAG2-related); *RAG2* • Ornithine Aminotransferase Deficiency; *OAT* • Ornithine Transcarbamylase Deficiency; *OTC* • Ornithine Translocase Deficiency [Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome]; *SLC25A15* • Pendred Syndrome; *SLC26A4* • Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX1-related); *PEX1* • Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX2-related); *PEX2* • Phenylalanine Hydroxylase Deficiency; *PAH* • Pituitary Hormone Deficiency, Combined 3; *LHX3* • Pontocerebellar Hypoplasia (RARS2-related); *RARS2* • Pontocerebellar Hypoplasia, Type 1A; *VRK1* • Pontocerebellar Hypoplasia, Type 2D; *SEPSECS* • Pontocerebellar Hypoplasia, Type 2E; *VPS53* • Primary Ciliary Dyskinesia (DNAH5-related); *DNAH5* • Primary Ciliary Dyskinesia (DNAI1-related); *DNAI1* • Primary Ciliary Dyskinesia (DNAI2-related); *DNAI2* • Primary Hyperoxaluria; Type 1; *AGXT* • Primary Hyperoxaluria, Type 2; *GRHPR* • Primary Hyperoxaluria, Type 3; *HOGA1* • Pycnodysostosis; *CTSK* • Pyruvate Dehydrogenase Deficiency (PDHB-related); *PDHB* • Pyruvate Dehydrogenase Deficiency, X-linked; *PDHA1* • Renal Tubular Acidosis and Deafness (ATP6V1B1-related); *ATP6V1B* • Retinal Dystrophy (RLBP1-related) [Bothnia Retinal Dystrophy]; *RLBP1* • Retinitis Pigmentosa 59 (DHDDS-related); *DHDDS* • Retinitis Pigmentosa 25 (EYS-related); *EYS* • Retinitis Pigmentosa 26; *CERKL* • Retinitis Pigmentosa 28; *FAM161A* • Retinitis Pigmentosa; *RPGR* • Rhizomelic Chondrodysplasia Punctata, Type 1; *PEX7* • Rhizomelic Chondrodysplasia Punctata, Type 3; *AGPS* • Roberts Syndrome; *ESCO2* • Salla Disease; *SLC17A5* • Sandhoff Disease; *HEXB* • Schimke Immunoosseous Dysplasia; *SMARCAL1* • Segawa Syndrome, (TH-related); *TH* • Severe Combined Immunodeficiency, Type Athabaskan; *DCLRE1C* • Severe Combined Immunodeficiency, X-linked; *IL2RG* • Sickle-Cell Disease; *HBB* • Sjögren-Larsson Syndrome; *ALDH3A2* • Smith-Lemli-Opitz Syndrome; *DHCR7* • Spinal Muscular Atrophy; *SMN1; SMN2* • Steroid-Resistant Nephrotic Syndrome; *NPHS2* • Stuve-Wiedemann Syndrome; *LIFR* • Tay-Sachs Disease; *HEXA* • Tyrosinemia, Type 1; *FAH* • Usher Syndrome, Type 1C; *USH1C* • Usher Syndrome, Type 1F; *PCDH15* • Usher Syndrome, Type 2A; *USH2A* • Usher Syndrome, Type 3; *CLRN1* • Wilson Disease; *ATP7B* • Wolman Disease; *LIPA* • Zellweger Spectrum Disorders (PEX6-related); *PEX6* • Zellweger Spectrum Disorders (PEX10-related); *PEX10*

The following table shows the detection rate and carrier residual risk after a negative result per disorder and gene tested, depending on the panel selected:

Condition	Gene	Population	Carrier frequency before screening (1 in)	Detection rate	Carrier residual risk after negative result (1 in)
3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	*HMGCL*	General population	*500	99%	Reduced
		Portugese	160	99%	15844
		Saudi Arabia	50	99%	4951
3-Methylcrotonyl-CoA Carboxylase Deficiency 1	*MCCC1*	European	147	99%	14556
3-Methylcrotonyl-CoA Carboxylase Deficiency 2	*MCCC2*	General Population	120	99%	11883
3-Methylglutaconic Aciduria, Type 3 (Costeff Syndrome)	*OPA3*	General Population	*500	99%	Reduced
3-Phosphoglycerate Dehydrogenase Deficiency	*PHGDH*	Ashkenazi Jewish	400	99%	39608
		General Population	*500	99%	Reduced
6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency	*PTS*	East Asian	122	99%	12081
		General Population	*500	99%	Reduced
Abetalipoproteinemia	*MTTP*	Ashkenazi Jewish	131	99%	12972
Achondrogenesis, Type 1B	*SLC26A2*	Finnish	75	99%	7427
		General Population	158	99%	15646
Achromatopsia (CNGB3-related)	*CNGB3*	General Population	146	98%	7301

Condition	Gene	Population	Carrier frequency before screening (1 in)	Detection rate	Carrier residual risk after negative result (1 in)
Acute Infantile Liver Failure (TRMU-related)	*TRMU*	General Population	*500	99%	Reduced
		Sephardic Jewish - Yemenite	34	99%	3367
Acyl-CoA Oxidase I Deficiency	*ACOX1*	General Population	*500	99%	Reduced
Adrenoleukodystrophy, X-Linked	*ABCD1*	General Population	16800	95%	338035
		Sephardic Jewish	*500	99%	Reduced
Aicardi-Goutières Syndrome	*SAMHD1*	General Population	*500	95%	Reduced
Alpha Thalassemia	*HBA1, HBA2*	African American	30	98%	276
		Asian	20	98%	184
		Caucasian	*500	98%	Reduced
		General Population	25	98%	230
Alport Syndrome (COL4A3-related)	*COL4A3*	Ashkenazi Jewish	183	99%	18121
		General Population	354	>98%	17701
Alport Syndrome, X-Linked	*COL4A5*	General Population	*500	>90%	Reduced
Alstrom Syndrome	*ALMS1*	General Population	*500	>90%	Reduced
Andermann Syndrome	*SLC12A6*	French Canadian	23	99%	2278
		General Population	*500	99%	Reduced
Argininosuccinate Lyase Deficiency	*ASL*	General Population	132	99%	13071
Aromatase Deficiency	*CYP19A1*	General Population	*500	99%	Reduced
Arthrogryposis Mental Retardation Seizures	*SLC35A3*	Ashkenazi Jewish	453	99%	44856
Asparagine Synthetase Deficiency	*ASNS*	General Population	*500	99%	Reduced
		Sephardic Jewish - Iranian	80	99%	7922
Aspartylglycosaminuria	*AGA*	Finnish	63	99%	6239
		General Population	*500	99%	Reduced
Ataxia with Vitamin E Deficiency	*TTPA*	General Population	*500	99%	Reduced
Ataxia-Telangiectasia	*ATM*	General Population	100	>91%	1120
		Romani Population	36	99%	3565
Autoimmune Polyglandular Syndrome, Type 1	*AIRE*	Finnish	79	>87%	613
		Sardinian	60	>87%	466
		Sephardic Jewish - Iranian	27	>87%	210
Autosomal Recessive Polycystic Kidney Disease	*PKHD1*	Ashkenazi Jewish	105	99%	10398
		Caucasian	100	99%	9902
		General Population	70	99%	6932
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	*SACS*	French Canadian - Charlevoix-Saguenay	21	99%	2080
		General Population	*500	99%	Reduced
Bardet-Biedl syndrome (BBS1-related)	*BBS1*	General Population	330	99%	32677
Bardet Biedl syndrome (BBS12-related)	*BBS12*	General Population	*500	99%	Reduced
Bare Lymphocyte Syndrome (CIITA-related)	*CIITA*	General Population	*500	99%	Reduced
Bartter Syndrome (BSND-related)	*BSND*	General Population	*500	99%	Reduced
Batten Disease (CLN3-related)	*CLN3*	General Population	230	99%	22775
Beta Thalassemia	*HBB*	African-American	8	99%	793
		Chinese	29	99%	2872
		Mediterranean	28	99%	2773
		Middle-Eastern	30	99%	2971
		Thai	20	99%	1981
Biotinidase Deficiency	*BTD*	General Population	120	99%	11883
Bloom Syndrome	*BLM*	Ashkenazi Jewish	100	99%	9902
Canavan Disease	*ASPA*	Ashkenazi Jewish	57	99%	5645
		European	3392	99%	Reduced
Carnitine Palmitoyltransferase IA Deficiency	*CPT1A*	General Population	*500	99%	Reduced
		Hutterite	16	99%	1585

Condition	Gene	Population	Carrier frequency before screening (1 in)	Detection rate	Carrier residual risk after negative result (1 in)
Carnitine Palmitoyltransferase II Deficiency	*CPT2*	Ashkenazi Jewish	45	99%	4456
		General Population	182	99%	18022
Carpenter Syndrome	*RAB23*	General Population	*500	99%	Reduced
Cartilage-Hair Hypoplasia	*RMRP*	Amish	19	99%	1882
		Finnish	76	99%	7526
		General Population	*500	99%	Reduced
Cerebrotendinous Xanthomatosis	*CYP27A1*	General Population	112	99%	11091
		Sephardic Jewish	76	99%	7526
Choreacanthocytosis	*VPS13A*	Ashkenazi Jewish	NA	99%	Reduced
Choroideremia, X-Linked	*CHM*	General Population	*500	>75%	Reduced
Chronic Granulomatous Disease, X-Linked	*CYBB*	General Population	*500	>90%	Reduced
Citrin Deficiency	*SLC25A13*	General Population	*500	>94%	8390
Citrullinemia, Type 1	*ASS1*	General Population	119	>96%	2991
Combined Malonic and Methylmalonic Acidemia	*ACSF3*	General Population	87	99%	8615
Combined Oxidative Phosphorylation Deficiency 1	*GFM1*	General Population	*500	99%	Reduced
Combined Oxidative Phosphorylation Deficiency 3	*TSFM*	Finnish	80	99%	7922
Combined Pituitary Hormone Deficiency 2	*PROP1*	General Population	45	>98%	2251
Congenital Disorder of Glycosylation, Type 1A (PMM2-related)	*PMM2*	Ashkenazi Jewish	61	99%	6041
		Caucasian	60	99%	5942
		General Population	190	99%	18814
Congenital Disorder of Glycosylation, Type 1B	*MPI*	General Population	*500	99%	Reduced
Congenital Disorder of Glycosylation, Type 1C	*ALG6*	General Population	*500	98%	Reduced
Congenital Finnish Nephrosis	*NPHS1*	Finnish	46	99%	4555
		General Population	*500	99%	Reduced
		Groffdale Conference Mennonite	12	99%	1189
Congenital Insensitivity to Pain with Anhidrosis	*NTRK1*	General Population	*500	99%	Reduced
Congenital Myasthenic Syndrome (CHRNE-related)	*CHRNE*	General Population	200	99%	19804
		Roma - Southeastern European	25	99%	2476
Congenital Myasthenic Syndrome (RAPSN-related)	*RAPSN*	General Population	283	99%	28023
Congenital Neutropenia (HAX1-related)	*HAX1*	General Population	*500	99%	Reduced
Congenital Neutropenia (VPS45-related)	*VPS45*	General Population	*500	99%	Reduced
Corneal Dystrophy and Perceptive Deafness	*SLC4A11*	General Population	*500	99%	Reduced
Corticosterone Methyloxidase Deficiency	*CYP11B2*	General Population	*500	99%	Reduced
		Sephardic Jewish - Iranian	30	99%	2971
CRB1-Related Retinal Dystrophies	*CRB1*	General Population	112	99%	11091
Creatine Transporter Defect (Cerebral Creatine Deficiency Syndrome 1), X-Linked	*SLC6A8*	General Population	*500	>98%	Reduced
Crigler Najjar Syndrome, Type I	*UGT1A1*	Amish	61	99%	Reduced
Cystic Fibrosis	*CFTR*	General Population	45	97%	1506
Cystinosis	*CTNS*	French Canadian - Saguenay Lac-St. Jean	39	99%	3862
		General Population	224	99%	22181
		Sephardic Jewish - Moroccan	100	99%	9902
D-Bifunctional Protein Deficiency	*HSD17B4*	General Population	158	96%	3970
Deafness, Autosomal Recessive 77	*LOXHD1*	Ashkenazi Jewish	180	99%	17824
		General Population	*500	99%	Reduced
Dystrophic Epidermolysis Bullosa (COL7A1-related)	*COL7A1*	General Population	370	99%	36638
Dystrophinopathies (Duchenne/Becker Muscular Dystrophy)	*DMD*	General Population	*500	90%	Reduced
Ehlers-Danlos Syndrome, Type VIIC	*ADAMTS2*	Ashkenazi Jewish	187	93%	2691
		General Population	*500	93%	Reduced
Emery-Dreifuss Muscular Dystrophy 1, X-Linked	*EMD*	General Population	200	99%	19804

Condition	Gene	Population	Carrier frequency before screening (1 in)	Detection rate	Carrier residual risk after negative result (1 in)
Enhanced S-Cone Syndrome	*NR2E3*	General Population	*500	99%	Reduced
Ethylmalonic Encephalopathy	*ETHE1*	General Population	*500	>84%	Reduced
Fabry Disease	*GLA*	General Population	*500	99%	Reduced
Factor IX Deficiency	*F9*	General Population	*500	98%	Reduced
Factor V Leiden Thrombophilia	*F5*	European	NA	99%	Reduced
		General Population	12-33	99%	1189
		US (African American)	NA	99%	Reduced
		US (Caucasian)	NA	99%	Reduced
		US (Hispanic)	NA	99%	Reduced
Factor XI Deficiency	*F11*	Ashkenazi Jewish	11	99%	1090
		European	123	99%	12180
Familial Dysautonomia	*ELP1*	Ashkenazi Jewish	31	99%	3070
Familial Hypercholesterolemia (LDLR-related)	*LDLR*	Ashkenazi Jewish	69	92%	870
		French Canadian	270	92%	3401
		General Population	250	92%	3149
		South African Afrikaner	72	92%	907
Familial Mediterranean Fever	*MEFV*	Armenian	17	99%	1684
		Cypriot	25	99%	2476
		Jewish	58	99%	5744
		Turkish	125	99%	12378
Familial Nephrogenic Diabetes Insipidus (AQP2-related)	*AQP2*	General Population	*500	99%	Reduced
Fanconi Anemia, Type C	*FANCC*	Ashkenazi Jewish	89	99%	8813
		General Population	417	99%	41292
Fanconi Anemia, Type G	*FANCG*	French-Canadian	NA	99%	Reduced
		Korean/Japanese	NA	99%	Reduced
		Portuguese-Brazilian	NA	99%	Reduced
Fragile X Syndrome, X-linked	*FMR1*	African American	267	99%	26439
		Ashkenazi Jewish	102	99%	10101
		Asian	419	99%	41490
		Caucasian	170	99%	16834
		General Population	201	99%	19903
		Hispanic	253	99%	25052
Galactokinase Deficiency (Galactosemia, Type II)	*GALK1*	General Population	122	99%	12081
		Roma	47	99%	4654
Galactosemia	*GALT*	African American	87	99%	8615
		Ashkenazi Jewish	156	99%	15448
		General Population	100	99%	9902
		Irish Travellers	11	99%	1090
Gaucher Disease	*GBA*	Ashkenazi Jewish	15	99%	1486
		General Population	158	99%	15646
Glutaric Acidemia, Type 1	*GCDH*	Amish - Pennsylvania	10	99%	991
		General Population	158	99%	15646
		Oji-Cree First Nations - N. Manitoba	10	99%	991
Glutaric Acidemia, Type 2A	*ETFA*	General Population	*500	99%	Reduced
Glycine Encephalopathy (AMT-related)	*AMT*	General Population	325	99%	32182
Glycine Encephalopathy (GLDC-related)	*GLDC*	Finnish	NA	>96%	Reduced
		General Population	NA	>96%	Reduced
Glycogen Storage Disease, Type 1A	*G6PC*	Ashkenazi Jewish	71	99%	7031
		General Population	177	99%	17527
Glycogen Storage Disease, Type 1B	*SLC37A4*	General Population	354	99%	35053
Glycogen Storage Disease, Type 2 (Pompe Disease)	*GAA*	General Population	100	>90%	1009

Condition	Gene	Population	Carrier frequency before screening (1 in)	Detection rate	Carrier residual risk after negative result (1 in)
Glycogen Storage Disease, Type 3	*AGL*	Faroese	30	99%	2971
		Finnish	1580	99%	Reduced
		Sephardic Jewish - Moroccan	35	99%	3466
Glycogen Storage Disease, Type 4	*GBE1*	Ashkenazi Jewish	68	99%	6734
		General Population	387	>90%	3902
Glycogen Storage Disease, Type 5 (McArdle Disease)	*PYGM*	General Population	*500	99%	49510
		Sephardic Jewish - Kurdish	84	99%	8318
Glycogen Storage Disease, Type 7	*PFKM*	General Population	*500	99%	49510
GRACILE Syndrome	*BCS1L*	Finnish	NA	99%	Reduced
Hemochromatosis, Type 2A	*HJV*	General Population	*500	99%	Reduced
Hemochromatosis, Type 3 (TFR2-related)	*TFR2*	General Population	*500	99%	Reduced
Hereditary Fructose Intolerance	*ALDOB*	Central European	95	99%	9407
		Northern India	NA	99%	Reduced
		Spanish	NA	99%	Reduced
		US	55-120	99%	5447
Hermansky-Pudlak Syndrome (HPS1-related)	*HPS1*	General Population	*500	99%	Reduced
		Puerto Rican (Northwestern)	21	99%	2080
Hermansky-Pudlak Syndrome (HPS3-related)	*HPS3*	Ashkenazi Jewish	235	99%	23270
		General Population	*500	99%	Reduced
Holocarboxylase Synthetase Deficiency	*HLCS*	Faroese	50	99%	4951
		General Population	224	99%	22181
		Japanese	158	99%	15646
Homocystinuria due to Cystathionine Beta-synthase Deficiency	*CBS*	General Population	224	98%	11201
		Norwegian	40	98%	2001
		Qatari	21	98%	1051
Homocystinuria-Megaloblastic Anemia, cbIE Type	*MTRR*	Caucasian	*500	>96%	Reduced
Hydrolethalus Syndrome	*HYLS1*	Finnish	50	99%	4951
Hypohidrotic Ectodermal Dysplasia, X-Linked	*EDA*	General Population	112	>90%	1130
Hypophosphatasia (ALPL-related)	*ALPL*	General Population	150	98%	7501
		Mennonite	25	99%	2476
Inclusion Body Myopathy, Type 2	*GNE*	Asian	58	99%	5744
		Iranian Jewish	11-100	99%	1090
Isovaleric Acidemia	*IVD*	General Population	150	98%	7501
Joubert Syndrome, Type 2	*TMEM216*	Ashkenazi Jewish	110	99%	10893
Junctional Epidermolysis Bullosa, Herlitz Type	*LAMC2*	Italian	858	99%	Reduced
Juvenile Retinoschisis, X-Linked	*RS1*	General Population	*500	>90%	Reduced
Krabbe Disease	*GALC*	Druze Northern Israel	6	99%	595
		General Population	158	99%	15646
Lamellar Ichthyosis, Type 1	*TGM1*	General Population	301	99%	29805
		Norwegian	151	99%	14952
Leber Congenital Amaurosis (LCA5-related)	*LCA5*	Ashkenazi Jewish	100	99%	Reduced
Leber Congenital Amaurosis, Type CEP290	*CEP290*	General Population	185	97%	6188
Leigh Syndrome, French-Canadian Type	*LRPPRC*	French Canadian - Saguenay Lac-St. Jean	23	99%	2278
		General Population	*500	99%	Reduced
Leukoencephalopathy with Vanishing White Matter	*EIF2B5*	General Population	*500	99%	Reduced
Leydig Cell Hypoplasia (Luteinizing Hormone Resistance)	*LHCGR*	Brazilian	NA	99%	Reduced
Limb-Girdle Muscular Dystrophy, Type 2A	*CAPN3*	General Population	158	>90%	1593
Limb-Girdle Muscular Dystrophy, Type 2B	*DYSF*	General Population	311	99%	30796
		Sephardic Jewish - Libyan	10	99%	991

Condition	Gene	Population	Carrier frequency before screening (1 in)	Detection rate	Carrier residual risk after negative result (1 in)
Limb-Girdle Muscular Dystrophy, Type 2C	*SGCG*	General Population	*500	99%	Reduced
		Moroccan	250	99%	24755
		Roma	59	99%	5843
Limb-Girdle Muscular Dystrophy, Type 2D	*SGCA*	General Population	*500	>95%	Reduced
Limb-Girdle Muscular Dystrophy, Type 2E	*SGCB*	General Population	*500	>90%	Reduced
Lipoamide Dehydrogenase Deficiency (Maple Syrup Urine Disease, Type 3)	*DLD*	Ashkenazi Jewish	110	99%	10893
Lipoid Adrenal Hyperplasia	*STAR*	General Population	*500	99%	Reduced
		Korean	170	99%	16834
Lipoprotein Lipase Deficiency	*LPL*	African population	308	98%	15401
		French Canadian	46	98%	2301
		General Population	*500	98%	Reduced
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	*HADHA*	European	210	99%	20795
Lysinuric Protein Intolerance	*SLC7A7*	Finnish	122	99%	12081
		General Population	*500	99%	Reduced
		Italians	NA	99%	Reduced
		Japanese	119	99%	11784
Maple Syrup Urine Disease, Type 1B	*BCKDHB*	Ashkenazi Jewish	97	99%	9605
Meckel-Gruber Syndrome, Type 1	*MKS1*	Finnish	47	>95%	946
		General Population	260	>95%	5232
Medium Chain Acyl-CoA Dehydrogenase Deficiency	*ACADM*	General Population	66	98%	3301
Megalencephalic Leukoencephalopathy with Subcortical Cysts	*MLC1*	General Population	*500	99%	Reduced
		Sephardic Jewish - Libyan	40	99%	3961
Metachromatic Leukodystrophy (ARSA-related)	*ARSA*	General Population	100	99%	9902
		Navajo	40	99%	3961
		Sephardic Jewish - Yemenite	46	99%	4555
Metachromatic Leukodystrophy (PSAP-related)	*PSAP*	General Population	*500	99%	Reduced
Methylmalonic Acidemia, cblA Type	*MMAA*	General Population	316	99%	31291
Methylmalonic Acidemia, cblB Type	*MMAB*	General Population	456	99%	45153
Methylmalonic Acidemia and Homocystinuria, Type cblC	*MMACHC*	General Population	123	99%	12180
Methylmalonic Acidemia and Homocystinuria, Type cblD	*MMADHC*	General Population	*500	99%	Reduced
Methylmalonic acidemia due to Methylmalonyl-CoA Mutase Deficiency	*MMUT*	Asian	53	99%	5249
		General Population	383	99%	37925
Microphthalmia/Anophthalmia (VSX2-related)	*VSX2*	General Population	*500	99%	Reduced
Mitochondrial Complex 1 Deficiency (ACAD9-related)	*ACAD9*	General Population	*500	99%	Reduced
Mitochondrial Complex 1 Deficiency (NDUFAF5-related)	*NDUFAF5*	Ashkenazi Jewish	290	99%	28716
		General Population	*500	99%	Reduced
Mitochondrial Complex 1 Deficiency (NDUFS6-related)	*NDUFS6*	Caucasus Jewish	24	99%	2377
		General Population	*500	99%	Reduced
Mitochondrial Myopathy and Sideroblastic Anemia (MLASA1)	*PUS1*	General Population	*500	99%	Reduced
Mucolipidosis II/III	*GNPTAB*	General Population	200	99%	19804
		Irish Traveller	15	99%	1486
Mucolipidosis III Gamma	*GNPTG*	General Population	*500	97%	Reduced
Mucolipidosis, Type IV	*MCOLN1*	Ashkenazi Jewish	100	97%	3345
		General Population	*500	97%	Reduced
Mucopolysaccharidosis, Type II (Hunter Syndrome), X-Linked	*IDS*	General Population	*500	>82%	Reduced
Mucopolysaccharidosis, Type IIIB (Sanfilippo B)	*NAGLU*	General Population	224	99%	22181

Condition	Gene	Population	Carrier frequency before screening (1 in)	Detection rate	Carrier residual risk after negative result (1 in)
Mucopolysaccharidosis, Type IIIC (Sanfilippo C)	*HGSNAT*	Caucasians	259	97%	8663
Mucopolysaccharidosis IIID (Sanfilippo Syndrome, Type D)	*GNS*	General Population	*500	92%	Reduced
Mucopolysaccharidosis, Type IX	*HYAL1*	General Population	*500	99%	Reduced
Multiple Sulfatase Deficiency	*SUMF1*	Ashkenazi Jewish	279	99%	27627
Myoneurogastrointestinal Encephalopathy (MNGIE)	*TYMP*	General Population	*500	99%	Reduced
		Sephardic Jewish - Iranian	158	99%	15646
Myotubular Myopathy, X-Linked	*MTM1*	General Population	*500	95%	Reduced
N-acetylglutamate Synthase Deficiency	*NAGS*	General Population	*500	99%	Reduced
Navajo Neurohepatopathy (MPV17-related Hepatocerebral Mitochondrial DNA Depletion Syndrome)	*MPV17*	Navajo	20	99%	1981
Neurological Ceroid Lipofuscinosis (TPP1-related)	*TPP1*	General Population	250	99%	24755
		Newfoundland	53	99%	5249
Neuronal Ceroid Lipofuscinosis (CLN5-related)	*CLN5*	Finnish	115	>98%	5751
		General Population	*500	>98%	Reduced
Neuronal Ceroid Lipofuscinosis (CLN6-related)	*CLN6*	General Population	*500	99%	Reduced
Neuronal Ceroid Lipofuscinosis (CLN8-related)	*CLN8*	Finnish	135	99%	13368
		General Population	*500	99%	Reduced
Neuronal Ceroid Lipofuscinosis (MFSD8-related)	*MFSD8*	General Population	*500	99%	Reduced
		Turkish	NA	99%	Reduced
Neuronal Ceroid Lipofuscinosis (PPT1-related)	*PPT1*	Finnish	70	99%	6932
		General Population	199	99%	19705
Niemann-Pick Disease, Types A/B	*SMPD1*	Ashkenazi Jewish	115	99%	11388
		General Population	250	99%	24755
Niemann-Pick Disease, Type C1/D	NPC1	General Population	183	98%	9151
Niemann-Pick Disease, Type C2	NPC2	General Population	*500	99%	Reduced
Nijmegen Breakage Syndrome	NBN	Eastern European	115	99%	15349
		North American	NA	99%	Reduced
Non-Syndromic Hearing Loss (GJB2-related)	GJB2	Ashkenazi Jewish	13	99%	1288
		General Population	50	99%	4951
Non-Syndromic Hearing Loss (GJB6-related)	GJB6	General Population	423	99%	41886
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome	*WNT10A*	General Population	305	99%	30202
Omenn Syndrome (RAG2-related)	*RAG2*	General Population	*500	99%	Reduced
Ornithine Aminotransferase Deficiency	*OAT*	General Population	*500	>96%	12564
Ornithine Transcarbamylase Deficiency	*OTC*	General Population	*500	99%	Reduced
Ornithine Translocase Deficiency [Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome]	*SLC25A15*	French-Canadian	20	99%	1981
		Japanese	NA	99%	Reduced
Pendred Syndrome	*SLC26A4*	Asian	74	99%	7328
		General Population	80	99%	7922
Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX1-related)	*PEX1*	General Population	134	>98%	6701
Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX2-related)	*PEX2*	Ashkenazi Jewish	227	99%	22478
Phenylalanine Hydroxylase Deficiency	*PAH*	Ashkenazi Jewish	250	99%	24755
		Caucasian	50	99%	4951
		East Asian	50	99%	4951
		European	50	99%	4951
		Finland	200	99%	19804
		General Population	140	99%	13863
		Ireland	33	99%	3268
		Japanese	200	99%	19804
		Turkish	26	99%	2575

Condition	Gene	Population	Carrier frequency before screening (1 in)	Detection rate	Carrier residual risk after negative result (1 in)
Pituitary Hormone Deficiency, Combined 3	*LHX3*	General Population	*500	99%	Reduced
Pontocerebellar Hypoplasia (RARS2-related)	*RARS2*	General Population	*500	99%	Reduced
Pontocerebellar Hypoplasia, Type 1A	*VRK1*	Ashkenazi Jewish	225	99%	22280
		General Population	*500	99%	Reduced
Pontocerebellar Hypoplasia, Type 2D	*SEPSECS*	General Population	*500	99%	Reduced
		Sephardic Jewish - Moroccan, Iraqi	43	99%	4258
Pontocerebellar Hypoplasia, Type 2E	*VPS53*	Moroccan Jewish	37	99%	3664
Primary Ciliary Dyskinesia (DNAH5-related)	*DNAH5*	Amish or Mennonite	NA	99%	Reduced
		General Population	106-164	99%	10497
Primary Ciliary Dyskinesia (DNAI1-related)	*DNAI1*	Ashkenazi Jewish	357	99%	35351
		General Population	150-232	99%	14853
Primary Ciliary Dyskinesia (DNAI2-related)	*DNAI2*	Ashkenazi Jewish	200	99%	19804
		General Population	*500	99%	Reduced
Primary Hyperoxaluria, Type 1	*AGXT*	European	173	97%	5787
Primary Hyperoxaluria, Type 2	*GRHPR*	General Population	*500	99%	Reduced
Primary Hyperoxaluria, Type 3	*HOGA1*	General Population	309	99%	30598
Pycnodysostosis	*CTSK*	Danish	NA	99%	Reduced
Pyruvate Dehydrogenase Deficiency (PDHB-related)	*PDHB*	General Population	*500	99%	Reduced
Pyruvate Dehydrogenase Deficiency, X-Linked	*PDHA1*	General Population	*500	99%	Reduced
Renal Tubular Acidosis and Deafness (ATP6V1B1-related)	*ATP6V1B1*	General Population	*500	99%	Reduced
		Sephardic Jewish - Syrian	140	99%	13863
Retinal Dystrophy (RLBP1-related) (Bothnia Retinal Dystrophy)	*RLBP1*	Swedish	84	99%	8318
Retinitis Pigmentosa 25 (EYS-related)	*EYS*	Ashkenazi Jewish	189	99%	Reduced
Retinitis Pigmentosa 26	*CERKL*	General Population	137	99%	13566
		Sephardic Jewish - Yemenite	24	99%	2377
Retinitis Pigmentosa 28	*FAM161A*	Ashkenazi Jewish	214	99%	21191
		General Population	289	99%	28617
		Sephardic Jewish - Libyan, Moroccan, Tunisian, Bulgarian	41	99%	4060
Retinitis Pigmentosa 59 (DHDDS-related)	*DHDDS*	Ashkenazi Jewish	117	99%	11586
Retinitis Pigmentosa, X-linked	*RPGR*	General Population	259	99%	25647
Rhizomelic Chondrodysplasia Punctata, Type 1	*PEX7*	General Population	158	99%	15646
Rhizomelic Chondrodysplasia Punctata, Type 3	*AGPS*	General Population	*500	99%	Reduced
Roberts Syndrome	*ESCO2*	General Population	*500	99%	Reduced
Salla Disease	*SLC17A5*	Finnish	100	>95%	2013
		General Population	*500	93%	Reduced
Sandhoff Disease	*HEXB*	General Population	180	>90%	1815
Schimke Immunoosseous Dysplasia	*SMARCAL1*	General Population	*500	99%	Reduced
Segawa Syndrome (TH-related)	*TH*	Caucasian	224	99%	22181
		General Population	*500	99%	Reduced
Severe Combined Immunodeficiency, Type Athabaskan	*DCLRE1C*	Navajo and Apache Native Americans	48	99%	4753
Severe Combined Immunodeficiency, X-Linked	*IL2RG*	General Population	*500	99%	Reduced
Sickle-Cell Disease	*HBB*	General Population	NA	99%	Reduced
Sjögren-Larsson Syndrome	*ALDH3A2*	Swedish	100	99%	9902
		Dutch	NA	99%	Reduced

Condition	Gene	Population	Carrier frequency before screening (1 in)	Detection rate	Carrier residual risk after negative result (1 in)
Smith-Lemli-Opitz Syndrome	*DHCR7*	African American	183	99%	18121
		Ashkenazi Jewish	43	99%	4258
		General Population	71	99%	7031
		Hispanic	167	99%	16537
		Northern European	54	99%	5348
		Southern European	66	99%	6536
Spinal Muscular Atrophy	*SMN1, SMN2*	Caucasian	35	95%	705
		Ashkenazi Jewish	41	90%	414
		Hispanic	117	90%	1180
		Asian	53	90%	535
		African American	66	71%	230
Steroid-Resistant Nephrotic Syndrome	*NPHS2*	General Population	*500	99%	Reduced
Stuve-Wiedemann Syndrome	*LIFR*	General Population	*500	99%	Reduced
Tay-Sachs Disease	*HEXA*	Ashkenazi Jewish	25	99%	2476
		General Population	250	99%	24755
Tyrosinemia, Type 1	*FAH*	Finnish	122	99%	12081
		French Canadian	66	99%	6536
		French Canadian - Saguenay Lac-St. Jean	16	99%	1585
		General Population	125	99%	12378
Usher Syndrome, Type 1C	*USH1C*	French Canadian / Acadian	227	99%	22478
		General Population	353	99%	34954
Usher Syndrome, Type 1F	*PCDH15*	Ashkenazi Jewish	40 - 126	99%	3961
Usher Syndrome, Type 2A	*USH2A*	Bukharan Jewish	NA	99%	Reduced
		General Population	158	95%	3180
		Sephardic Jewish - Iraqi, Iranian	36	99%	3565
Usher Syndrome, Type 3	*CLRN1*	Ashkenazi Jewish	120	99%	11883
		Finnish	70	99%	6932
Wilson Disease	*ATP7B*	Ashkenazi Jewish	67	99%	6635
		Canary Islands	25	99%	2476
		General Population	90	99%	8912
		Sardinian	42	99%	4159
		Sephardic Jewish - North African, Iraqi, Yemenite, Iranian, Bukharian	65	99%	6437
Wolman Disease	*LIPA*	General Population	*500	99%	Reduced
		Sephardic Jewish - Iranian	32	99%	3169
Zellweger Spectrum Disorders (PEX6-Related)	*PEX6*	French Canadian	55	99%	5447
		General Population	280	99%	27726
Zellweger Spectrum Disorders (PEX10-related)	*PEX10*	General Population	*500	97%	Reduced

* <1 in 500

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