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TEST PATIENT

Sample Test Name
Sex : F
Date Collected : 00-00-0000
111 TEST ROAD TEST SUBURB
LAB ID: 00000000 UR#:0000000

TEST PHYSICIAN

DR JOHN DOE
111 CLINIC STREET
CLINIC SUBURB VIC 3000

GENOMIC TESTING

Summary of Results

Propionic Acidemia

Risk to Child: Any child of this patient has a 50% chance of inheriting the patient's mutation associated with this disease and being a carrier. If the patient's partner also carries a mutation for this disease, there is a 25% chance that each child of the patient will inherit both parents' mutations and may develop the disease.

Risk to Patient: This patient is a carrier of a genetic mutation for this disease but is not likely to be affected. Since there are many rare mutations, it is possible to carry an untested mutation in addition to the one found in the patient's DNA.

Recommendation: Genetic counseling is recommended for the patient and his or her partner to discuss the potential clinical and/or reproductive implications of this result and to discuss genetic testing of the patient's partner and close relatives.

Result:
Carrier, Heterozygote

Mutations:
PCCB [c.1228C>T (p.R410W)]

PATIENT IS NOT A CARRIER FOR THE FOLLOWING:			
21-Hydroxylase-deficient congenital adrenal hyperplasia	Costeff optic atrophy syndrome	Hereditary fructose intolerance	Pendred syndrome
3-Methylcrotonyl-CoA carboxylase deficiency	Crigler-Najjar syndrome	Herlitz junctional epidermolysis bullosa, LAMA3-related	Phenylketonuria
Achromatopsia	Cystic fibrosis	Herlitz junctional epidermolysis bullosa, LAMB3-related	Polycystic kidney disease
Acrodermatitis enteropathica	Cystinosis	Herlitz junctional epidermolysis bullosa, LAMC2-related	Pompe disease
Alkaptonuria	Diabetes, permanent neonatal	HMG-CoA lyase deficiency	Prekallikrein deficiency
Alpha-1 antitrypsin deficiency	Dihydropyrimidine dehydrogenase deficiency	Homocystinuria, cblE type	Primary hyperoxaluria, type 1
Alpha-mannosidosis	Dubin-Johnson syndrome	Homocystinuria, classic	Primary hyperoxaluria, type 2
Amyotrophic lateral sclerosis	Ehlers-Danlos syndrome, dermatosparaxis	Hurler syndrome	Primary hyperoxaluria, type 3
Andermann syndrome	Ehlers-Danlos syndrome, hypermobility	Hypophosphatasia, autosomal recessive	Prothrombin deficiency
Argininosuccinate lyase deficiency	Ehlers-Danlos syndrome, kyphoscoliotic	Inclusion body myopathy 2	Rh-null syndrome
ARSACS	Factor V Leiden thrombophilia	Juvenile retinoschisis, X-linked	Rhizomelic chondrodysplasia punctate type 1
Aspartylglucosaminuria	Factor XI deficiency	Krabbe disease	Rickets, pseudovitamin D-deficiency
Ataxia with vitamin E deficiency	Familial dysautonomia	Lipoamide dehydrogenase deficiency	Salla disease
Ataxia-telangiectasia	Familial Mediterranean fever	Lipoprotein lipase deficiency, familial	Sandhoff disease
Autoimmune polyglandular syndrome, type I	Fanconi anemia	Maple syrup urine disease	Short-chain acyl-CoA dehydrogenase deficiency
Bardet-Biedl syndrome, BBS1-related	Galactokinase deficiency	Medium-chain acyl-CoA dehydrogenase deficiency	Sick sinus syndrome
Bartter syndrome, type 4a	Galactosemia	Megalencephalic leukoencephalopathy with subcortical cysts	Sickle cell disease
Beta-ketothiolase deficiency	Gaucher disease	Metachromatic leukodystrophy	Smith-Lemli-Opitz syndrome
Beta-thalassemia	Glutaric acidemia, type 1	Methylmalonic acidemia	Spherocytosis, hereditary
Biotinidase deficiency	Glycogen storage disease, type 1a	Mucolipidosis II	Tay-Sachs disease
Bloom syndrome	Glycogen storage disease, type Ib	Mucolipidosis III	Tay-Sachs pseudodeficiency
Canavan disease	Glycogen storage disease, type III	Mucolipidosis IV	Thrombocytopenia, congenital amegakaryocytic

PATIENT IS NOT A CARRIER FOR THE FOLLOWING:			
Carnitine deficiency, primary systemic	Glycogen storage disease, type V	Multiple carboxylase deficiency	Tyrosine hydroxylase deficiency
Carnitine palmitoyltransferase II deficiency	GM1-gangliosidosis	Nephrotic syndrome, steroid-resistant	Tyrosinemia
Cartilage-hair hypoplasia	Hearing loss, DFNB1 and DFNB9 nonsyndromic	Neuronal ceroid lipofuscinosis, CLN3-related	Usher syndrome, type 1F
Cerebrotendinous xanthomatosis	Hearing loss, DFNB59 nonsyndromic	Neuronal ceroid lipofuscinosis, CLN5-related	Very long-chain acyl-CoA dehydrogenase deficiency
Choroideremia	Hemochromatosis	Neuronal ceroid lipofuscinosis, CLN8-related	Von Willebrand disease, type 2 Normandy
Citrullinemia, type I	Hemoglobin C	Neuronal ceroid lipofuscinosis, PPT1-related	Von Willebrand disease, type 3
Cohen syndrome	Hemoglobin D	Neuronal ceroid lipofuscinosis, TPP1-related	Wilson disease
Combined pituitary hormone deficiency, PROP1-related	Hemoglobin E	Niemann-Pick disease	Zellweger syndrome spectrum, PEX1-related
Congenital disorder of glycosylation type Ia	Hemoglobin O	Nijmegen breakage syndrome	

Propionic Acidemia

About: Propionic acidemia is an inherited disorder that causes brain damage in infants and young children due to a defect in protein and fat metabolism. Symptoms may include poor appetite, nausea, vomiting, extreme sleepiness, irritability, low muscle tone and muscle weakness. If not treated, breathing problems, seizures, swelling of the brain, stroke, coma and sometimes even death can occur. With prompt and lifelong treatment, children with propionic academia can often live normal lives. A small number of people with propionic acidemia never show symptoms.[1]

Genetics: Propionic acidemia is caused by mutations in the PCCA or the PCCB genes, which encode the alpha and beta protein subunits, respectively, of propionyl-CoA carboxylase. This enzyme is necessary for breaking down certain amino acids (valine, isoleucine, methionine, and threonine) and fats (odd-chain fatty acids, cholesterol).[2,3] Defects in the protein lead to toxic levels of propionic acid; by-products accumulate in body fluids and can cause brain damage.

The incidence of propionic acidemia is very low worldwide (about 1 in 50,000) but highly variable: 1 in 1,000 in the Inuit people of Greenland. The incidence of propionic acidemia is 1 in 27,264 in Saudi Arabia, and 1 in 250,000 in Germany.[4]

Mutations Tested: The test involves one mutation in the PCCA gene and four mutations in PCCB gene.

PCCA [R3999Q]

PCCB [R410W, T428I, 1218del14ins12, 1172_1173insT]

References

1. Yang X, Sakamoto O, Matsubara Y, et al. Mutation spectrum of the PCCA and PCCB genes in Japanese patients with propionic acidemia. Molecular genetics and metabolism. 2004;81:335-42.
2. Ugarte M, Pérez-Cerdá C, Rodríguez-Pombo P, et al. Overview of mutations in the PCCA and PCCB genes causing propionic acidemia. Human mutation. 1999;14:275-82.
3. Lamhonwah AM, Troxel CE, Schuster S, Gravel RA. Two distinct mutations at the same site in the PCCB gene in propionic acidemia. Genomics. 1990;8:249-54.
4. Desviat LR, Pérez B, Pérez-Cerdá C, et al. Propionic acidemia: mutation update and functional and structural effects of the variant alleles. Molecular genetics and metabolism. 2004;83:28-37.

GENOTYPE/HAPLOTYPE DETAIL

CARRIER STATUS

This section lists the individual mutations that were tested for Carrier Status. Tested mutations are organized by disease and contained in brackets next to their respective genes.

- If the patient carries a tested mutation, it will be highlighted in red in the “Carrier of” section.
- If the patient does not carry a tested mutation, it will be listed in black in the “Not a Carrier of” section.
- If a result could not be obtained for a mutation, it is listed in the “No Data for” section.
- “Pending” indicates that the patient’s test for this disease is still in progress.
- “Unable To Report” indicates that no result can be provided.

Residual risk: since there are many rare mutations, it is possible to carry a mutation that is not included in our test.

PROPIONIC ACIDEMIA

Heterozygous for: PCCB [c.1228C>T (p.R410W)]
Not a Carrier of: PCCA [c.1196G>A (p.R399Q)]; **PCCB** [c.1173dupT (p.V392CfsX2), c.1283C>T (p.T428I), c.1218_1231delinsTAGAGCACAGGA (p.G407RfsX14)]

21-HYDROXYLASE-DEFICIENT CONGENITAL ADRENAL HYPERPLASIA

Not a Carrier of: CYP21A2 [c.293-2A>G, c.1360C>T (p.P454S), c.293-13C>G, c.844G>T (p.V282L)/c.844G>C (p.V282L), c.518T>A (p.I173N), c.719T>A (p.M240K), c.955C>T (p.Q319X), c.1069C>T (p.R357W), c.92C>T (p.P31L), c.713T>A (p.V238E), c.923dupT (p.L308FfsX6), c.332_339del (p.G111VfsX21)]

3-METHYLCROTONYL-COA CARBOXYLASE DEFICIENCY

Not a Carrier of: MCCC1 [c.866C>T (p.A289V), c.1155A>C (p.R385S), c.1594G>C (p.D532H), c.1310T>C (p.L437P)]; **MCCC2** [c.295G>C (p.E99Q), c.1309A>G (p.I437V), c.1015G>A (p.V339M), c.577C>T (p.R193C), c.518C>T (p.S173L)]

ACHROMATOPSIA

Not a Carrier of: CNGA3 [c.847C>T (p.R283W), c.1641C>A (p.F547L), c.829C>T (p.R277C), c.1306C>T (p.R436W)]; **CNGB3** [c.819_826del (p.R274VfsX13), c.991-3T>G, c.886_896delinsT (p.T296YfsX9), c.1304C>T (p.S435F), c.1578+1G>A, c.1006G>T (p.E336X), c.1148delC (p.T383IfsX13)]

ACHROMATOPSIA

Not a Carrier of: SLC39A4 [c.1224_1228del (p.G409LfsX7), c.143T>G (p.L48X)]

ALKAPTONURIA

Not a Carrier of: HGD [c.360T>G (p.C120W), c.481G>A (p.G161R), c.1102A>G (p.M368V), c.342+1G>A]

ALPHA-1 ANTITRYPSIN DEFICIENCY

Not a Carrier of: SERPINA1 [c.863A>T (p.E288V, S allele), c.1096G>A (p.E366K, Z allele)]

ALPHA-MANNOSIDOSIS

Not a Carrier of: MAN2B1 [c.1830+1G>C, c.2248C>T (p.R750W), c.2426T>C (p.L809P)]

AMYOTROPHIC LATERAL SCLEROSIS

Not a Carrier of: ALS2 [c.1867_1868del (p.L623Vfs)]

ANDERMANN SYNDROME

Not a Carrier of: SLC12A6 [c.2032dupT (p.Y678LfsX41), c.3031C>T (p.R1011X), c.2023C>T (p.R675X), c.2436delG (p.T813PfsX2), c.1584_1585delinsG (p.F529LX4), c.1478_1485del (p.F493CfsX48)]

ARGININOSUCCINATE LYASE DEFICIENCY

Not a Carrier of: ASL [c.1153C>T (p.R385C), c.532G>A (p.V178M), c.446+1G>A (IVS5+1G>A), c.1060C>T (p.Q354X), c.346C>T (p.Q116X), c.578G>A (p.R193Q), c.260A>G (p.D87G)]

ARSACS

Not a Carrier of: SACS [c.8844delT (p.I2949FfsX4), c.7504C>T (p.R2502X), c.10907G>A (p.R3636Q), c.12160C>T (p.Q4054X)]

ASPARTYLGLUCOSAMINURIA

Not a Carrier of: AGA [c.488G>C (p.C163S)]

ATAXIA WITH VITAMIN E DEFICIENCY

Not a Carrier of: TTPA [c.303T>G (p.H101Q), c.744delA (p.E249NfsX15)]

ATAXIA-TELANGIECTASIA

Not a Carrier of: ATM [c.1564_1565del (p.E522IfsX43), c.7010_7011del (p.C2337SfsX35), c.7517_7520del (p.R2506RfsX3), c.7638_7646del (p.R2547_S2549del), c.7886_7890del (p.I2629SfsX25), c.8264_8268del (p.Y2755CfsX12), c.397_398insT (p.N133IfsX8), c.2806_2809dupCTAG (p.E937AfsX33), c.7926A>C (p.R2642S), c.1120C>T (p.Q374X), c.4507C>T (p.Q1503X), c.5908C>T (p.Q1980X), c.5932G>T (p.E1978X), c.7449G>A (p.W2483X), c.8494C>T (p.R2832C), c.4852C>T (p.R1618X), c.8011-2A>C, c.5319+2T>C, c.3576G>A (p.K1192K), c.2251-10T>G, c.4612-12A>G, c.4909+1G>A, c.8201_8211delinsGACCTG (p.M2734RfsX11), c.3245_3247delinsTGAT (p.H1082LfsX14), c.8786+1G>A, c.103C>T (p.R35X), c.7327C>T (p.R2443X), c.6095G>A (p.R2032K)]

AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE I

Not a Carrier of: AIRE [c.931delT (p.C311fsX376), c.1189delC (p.L397fsX478), c.64_69del (p.V22_D23del), c.653-6_653-4del (p.G218fsX284), c.402delC (p.S135fsX147), c.1249delC (p.L417fsX478), c.966_969dupCCTG (p.L323fsX372), c.1295_1296insAC (p.C434VfsX47), c.1242_1243insA (p.H415fsX422), c.1072C>T (p.Q358X), c.908G>C (p.R303P), c.290T>C (p.L97P), c.1336T>G (p.C446G), c.1400+1G>A (IVS11+1G>A), c.879+1G>A (IVS7+1G>A), c.462A>T (p.P154P, IVS3-2A>T), c.1344delinsTT (p.C449fsX502), c.755C>T (p.P252L), c.769C>T (p.R257X), c.247A>G (p.K83E), c.415C>T (p.R139X), c.682G>T (p.G228W), c.1A>T (p.M1L), c.43C>T (p.R15C), c.47C>T (p.T16M), c.83T>C (p.L28P), c.86T>C (p.L29P), c.232T>C (p.W78R), c.238G>T (p.V80L), c.254A>G (p.Y85C), c.269A>G (p.Y90C), c.278T>G (p.L93R), c.230T>C (p.F77S), c.1616C>T (p.P539L), c.995+5G>T (IVS8+5G>T), c.1163_1164insA (p.M388fsX422), c.1638A>T (p.X546C), c.932G>A (p.C311Y), c.967_979del (p.L323SfsX51), c.1103dupC (p.P370fsX370), c.1513delG (p.A502fsX519), c.607C>T (p.R203X), c.892G>A (p.E298K), c.463+2T>C (IVS3+2T>C)]

BARDET-BIEDL SYNDROME, BBS1-RELATED

Not a Carrier of: BBS1 [c.1169T>G (p.M390R)]

BARTTER SYNDROME, TYPE 4A

Not a Carrier of: BSND [c.139G>A (p.G47R)]

BETA-KETOTHIOLASE DEFICIENCY

Not a Carrier of: ACAT1 [c.149delC (p.T50NfsX7), c.890C>T (p.T297M), c.1163+2T>C (IVS11+2T>C), c.547G>A (p.G183R), c.814C>T (p.Q272X), c.622C>T (p.R208X), c.826+1G>T (IVS8+1G>T), c.455G>C (p.G152A)]

BETA-THALASSEMIA

Not a Carrier of: HBB [c.118C>T (p.Q40X, cd39C>T), c.316-2A>C (IVS2+849A>C)/c.316-2A>G (IVS2+849A>G), c.92+5G>T (IVS1+5G>T), c.-78A>G (-28A>G), c.-137C>G (-87C>G), c.-138C>T (-88C>T), c.315+1G>A (IVS2+1G>A), c.75T>A (p.G25G, cd24T>A), c.92+1G>A (IVS1+1G>A), c.59A>G (p.N20S, Hb Malay), c.52A>T (p.K18X, 17A>T), c.316-197C>T (IVS2+654C>T), c.-79A>G (-29A>G), c.316-106C>G (IVS2+745C>G), c.93-21G>A (IVS1+110G>A), c.25_26del (p.K9VfsX14, cd8-AA), c.27dupG (p.S10VfsX14, cd8/9+G), c.92+6T>C (IVS1+6T>C), c.135delC (p.F46LfsX16, cd44-C), c.126_129del (p.F42LfsX19, 41/42-TTCT)]

BIOTINIDASE DEFICIENCY

Not a Carrier of: BTD [c.511G>A (p.A171T), c.1330G>C (p.D444H), c.98_104delinsTCC (p.C33FfsX36), c.1368A>C (p.Q456H), c.1612C>T (p.R538C)]

BLOOM SYNDROME

Not a Carrier of: BLM [c.1284G>A (p.W428X), c.1701G>A (p.W567X), c.2207_2212delinsTAGATTC (p.Y736LfsX5, blmAsh), c.2407dupT (p.W803fsX), c.2923delC (p.Q975fsX), c.2506_2507del (p.R836fsX),

BLOOM SYNDROME

c.557_559del (p.S186X), c.1933C>T (p.Q645X), c.2695C>T (p.R899X)]

CANAVAN DISEASE

Not a Carrier of: ASPA [c.827_828del (p.C276YfsX9), c.244dupA (p.M82NfsX8, 245insA), c.884T>C (p.F295S), c.327T>G (p.Y109X), c.584T>G (p.M195R), c.854A>C (p.E285A), c.693C>A (p.Y231X), c.914C>A (p.A305E), c.433-2A>G (IVS2-2A>G), c.654C>A (p.C218X), c.838C>T (p.P280S), c.820G>A (p.G274R)]

CARNITINE DEFICIENCY, PRIMARY SYSTEMIC

Not a Carrier of: SLC22A5 [c.653_654insTATGGCCATCAGGTTGGAG (p.T219fsX284), c.12C>G (p.Y4X), c.95A>G (p.N32S), c.1319C>T (p.T440M), c.632A>G (p.Y211C), c.505C>T (p.R169W), c.760C>T (p.R254X), c.136C>T (p.P46S), c.849G>T (p.W283C), c.1403C>G (p.T468R)]

CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY

Not a Carrier of: CPT2 [c.149C>A (p.P50H), c.338C>T (p.S113L), c.1507C>T (p.R503C), c.1646G>A (p.G549D), c.1238_1239del (p.K414TfsX7), c.641T>C (p.M214T)]

CARTILAGE-HAIR HYPOPLASIA

Not a Carrier of: RMRP [g.70A>G]

CEREBROTENDINOUS XANTHOMATOSIS

Not a Carrier of: CYP27A1 [c.1321C>T (p.P441S), c.1151C>T (p.P384L), c.409C>T (p.R137W), c.475C>T (p.Q159X), c.691C>T (p.R231X), c.808C>T (p.R270X), c.850A>T (p.K284X), c.1061A>G (p.D354G), c.1183C>T (p.R395C), c.1420C>T (p.R474W), c.1214G>A (p.R405Q), c.1016C>T (p.T339M), c.1415G>C (p.G472A), c.379C>T (p.R127W), c.646G>C (p.A216P), c.380G>A

CEREBROTENDINOUS XANTHOMATOSIS

(p.R127Q), c.844+1G>A (IVS4+1G>A), c.1263+1G>A (IVS7+1G>A), c.1213C>T (p.R405W), c.1184+1G>A (IVS6+1G>A), c.1185-1G>T (IVS6-1G>T), c.1202C>G (p.P401R), c.1222G>T (p.E408X), c.1263+5G>T (IVS7+5G>T), c.435G>T (p.G145G), c.446+1G>A (IVS2+1G>A), c.583G>T (p.E195X), c.779G>A (p.W260X), c.1381C>T (p.Q461X)]

CHOROIDEREMIA

Not a Carrier of: CHM [c.1609+2dupT]

CITRULLINEMIA, TYPE I

Not a Carrier of: ASS1 [c.285G>T (p.R95S), c.952delG (p.A318LfsX58), c.568T>G (p.Y190D), c.1034T>G (p.V345G), c.539G>A (p.S180N), c.1168G>A (p.G390R), c.910C>T (p.R304W), c.928A>C (p.K310Q), c.919C>T (p.R307C), c.805G>A (p.V269M), c.257G>A (p.R86H), c.421-2A>G (IVS6-2A>G), c.814C>T (p.R272C), c.571G>A (p.E191K), c.1138C>T (p.Q380X)]

COHEN SYNDROME

Not a Carrier of: VPS13B [c.8459T>C (p.I2820T), c.3348_3349del (p.C1117FfsX8), c.9259_9260insT (p.L3087FfsX20)]

COMBINED PITUITARY HORMONE DEFICIENCY, PROP1-RELATED

Not a Carrier of: PROP1 [c.301_302del (p.L102CfsX8)]

CONGENITAL DISORDER OF GLYCOSYLATION TYPE IA

Not a Carrier of: PMM2 [c.422G>A (p.R141H), c.357C>A (p.F119L)]

COSTEFF OPTIC ATROPHY SYNDROME

Not a Carrier of: OPA3 [c.415C>T (p.Q139X), c.143-1G>C, c.320_337del (p.Q108_E113del)]

CRIGLER-NAJJAR SYNDROME

Not a Carrier of: UGT1A1 [c.722_723delAG (p.Q239fsX256), c.517delC (p.H173MfsX32), c.1043delA (p.N348TfsX18), c.1186delG (p.D396IfsX16), c.801delC (p.I268SfsX98), c.396_401del (p.H132_K134delinsQ), c.973delG (p.A325LfsX41), c.652dupT (p.S218FfsX40), c.1223dupG (p.A409SfsX13), c.1127A>G (p.H376R), c.1130G>T (p.G377V), c.1448G>A (p.W483X, TAG), c.1449G>A (p.W483X, TGA), c.101C>A (p.P34Q), c.576C>G (p.Y192X), c.1433C>A (p.A478D), c.554A>C (p.Q185P), c.118T>C (p.W40R), c.1477G>C (p.G493R), c.610A>G (p.M204V), c.847C>T (p.Q283X), c.392T>C (p.L131P), c.875C>T (p.A292V), c.1005G>A (p.W335X), c.1305-1G>A (IVS4-1G>A), c.1304+1G>T (IVS4+1G>T), c.864+1G>C (IVS1+1G>C), c.1085-2A>G (IVS3-2A>G), c.877_890delinsA (p.Y293MfsX69), c.1160_1161delinsGT (p.P387R), c.1198A>G (p.N400D), c.1456T>G (p.Y486D), c.674T>G (p.V225G), c.115C>G (p.H39D), c.222C>A (p.Y74X), c.524T>A (p.L175Q), c.529T>C (p.C177R), c.625C>T (p.R209W), c.698T>G (p.L233R), c.881T>C (p.I294T), c.992A>G (p.Q331R), c.1021C>T (p.R341X), c.1069C>T (p.Q357X), c.1070A>G (p.Q357R), c.1102G>A (p.A368T), c.1124C>T (p.S375F), c.1143C>G (p.S381R), c.1201G>C (p.A401P), c.1282A>G (p.K428E), c.1309A>T (p.K437X), c.1388A>C (p.E463A), c.1463C>T (p.S488F), c.991C>T (p.Q331X), c.1006C>T (p.R336W), c.840C>A (p.C280X), c.513_515del (p.F170del), c.835A>T (p.N279Y), c.1220delA (p.K407RfsX5), c.479T>A (p.V160E), c.1108A>G (p.I370V), c.1328T>C (p.L443P), c.1207C>T (p.R403C)]

CYSTIC FIBROSIS

Not a Carrier of: CFTR [c.3659delC (p.T1220KfsX8, 3791delC), c.3773dupT (p.L1258FfsX7, 3905insT), c.3302T>A (p.M1101K), c.1210-11T>G (5T), c.273+3A>C (405+3A>C), c.3752G>A (p.S1251N), c.1364C>A (p.A455E), c.1657C>T (p.R553X), c.3484C>T (p.R1162X), c.3718-2477C>T (3849+10kbC>T), c.2988+1G>A (3120+1G>A), c.2128A>T (p.K710X), c.1652G>A (p.G551D), c.3454G>C (p.D1152H), c.254G>A (p.G85E), c.3140-26A>G (3272-26A>G), c.1585-1G>A (1717-1G>A), c.3846G>A (p.W1282X), c.1477C>T (p.Q493X), c.579+1G>T (711+1G>T), c.1558G>T (p.V520F), c.1040G>C (p.R347P), c.350G>A (p.R117H), c.489+1G>T (621+1G>T), c.3266G>A (p.W1089X), c.1090T>C (p.S364P), c.988G>T (p.G330X), c.3472C>T (p.R1158X), c.3909C>G (p.N1303K), c.1679G>C (p.R560T), c.2657+5G>A (2789+5G>A), c.532G>A (p.G178R), c.1624G>T (p.G542X), c.1521_1523delCTT (p.F508del), c.948delT (p.F316LfsX12, 1078delT), c.1519_1521delATC (p.I507del), c.2052delA (p.K684NfsX38, 2184delA), c.3528delC (p.K1177SfsX15, 3659delC), c.1766+1G>A (1898+1G>A), c.617T>G (p.L206W), c.1055G>A (p.R352Q), c.1572C>A (p.C524X), c.1646G>A (p.S549N), c.1645A>C (p.S549R), c.1721C>A (p.P574H), c.1865G>A (p.G622D), c.2125C>T (p.R709X), c.3587C>G (p.S1196X), c.3612G>A (p.W1204X), c.3712C>T (p.Q1238X), c.935_937del (p.F312del, deltaF311), c.262_263del (p.L88IfsX22, 394delTT), c.442delA (p.I184LfsX5, 574delA), c.531delT (p.I177MfsX12, 663delT), c.803delA (p.N268IfsX17, 935delA), c.805_806del (p.I269PfsX4, 936delTA), c.1545_1546del (p.Y515X, 1677delTA), c.1817_1900del (p.M607_Q634del, 1949del84), c.1911delG (p.Q637HfsX26, 2043delG), c.1923_1931delinsA (p.S641RfsX5, 2055del9>A), c.1973_1985delinsAGAAA (p.R658KfsX4, 2105del13ins5), c.3039delC (p.Y1014TfsX9, 3171delC), c.3744delA (p.K1250RfsX9, 3876delA), c.2175dupA (p.E726RfsX4, 2307insA), c.2737_2738insG (p.Y913X, 2869insG), c.273+1G>A (405+1G>A), c.580-1G>T (712-1G>T), c.1680-1G>A (1812-1G>A), c.2988G>A (p.Q996Q, 3120G>A), c.313delA (p.I105SfsX2, 444delA), c.613C>T (p.P205S), c.1976delA (p.N659IfsX4), c.1647T>G (p.S549R), c.1000C>T (p.R334W), c.1682C>A (p.A561E), c.2249C>T (p.P750L), c.1673T>C (p.L558S), c.4046G>A (p.G1349D), c.3532_3535dupTCAA (p.T1179IfsX17, 3667ins4), c.1075_1079delinsAAAAA (p.Q359_T360delinsKK, Q359K/T360K), c.3299A>C (p.Q1100P), c.695T>A (p.V232D), c.714delT (p.L240X)]

CYSTINOSIS

Not a Carrier of: CTNS [c.18_21del (p.T7FfsX7), c.614_616del (p.D205del), 57-kb deletion,

CYSTINOSIS

c.382C>T (p.Q128X), c.544T>C (p.W182R), c.922G>A (p.G308R), c.397A>T (p.I133F), c.414G>A (p.W138X), c.473T>C (p.L158P)]

DIABETES, PERMANENT NEONATAL

Not a Carrier of: ABCC8 [c.215A>G (p.N72S), c.1144G>A (p.E382K), c.134C>T (p.P45L)]; **GCK** [c.1190G>T (p.R397L), c.1019+2T>G (IVS8+2T>G)]

DIHYDROPYRIMIDINE DEHYDROGENASE DEFICIENCY

Not a Carrier of: DPYD [c.1339+1G>T (IVS11+1G>T), c.703C>T (p.R235W), c.2657G>A (p.R886H), c.1905+1G>A (IVS14+1G>A), c.1679T>G (p.I560S), c.2933A>G (p.H978R), c.1003G>T (p.V335L), c.1156G>T (p.E386X), c.257C>T (p.P86L), c.2329G>T (p.A777S), c.545T>A (p.M182K)]

DUBIN-JOHNSON SYNDROME

Not a Carrier of: ABCC2 [c.3449G>A (p.R1150H), c.3517A>T (p.I1173F)]

EHLERS-DANLOS SYNDROME, DERMATOSPARAXIS

Not a Carrier of: ADAMTS2 [c.673C>T (p.Q225X), c.2384G>A (p.W795X)]

EHLERS-DANLOS SYNDROME, HYPERMOBILITY

Not a Carrier of: TNXB [c.2116_2117dupGT (p.E707X), c.3290_3291del (p.K1097RfsX48, 3551_3552delAA)]

EHLERS-DANLOS SYNDROME, KYPHOSCOLIOTIC

Not a Carrier of: PLOD1 [c.1362delC (p.Y455TfsX2), c.467-2delA, c.1677dupC (p.I560HfsX8, 1702insC), c.975+2_975+3insTT, c.153dupC (p.N52QfsX52), c.426T>A (p.Y142X), c.979C>T (p.Q327X), c.145C>T (p.Q49X), c.1336T>G (p.W446G), c.2117A>G (p.H706R), c.955C>T (p.R319X), c.2032G>A (p.G678R),

EHLERS-DANLOS SYNDROME, KYPHOSCOLIOTIC

c.1533C>G (p.Y511X), c.1836G>C (p.W612C), c.2008C>T (p.R670X), c.1999G>A (p.A667T)]

FACTOR V LEIDEN THROMBOPHILIA

Not a Carrier of: F5 [c.1601G>A (p.R534Q, Factor V Leiden)]

FACTOR XI DEFICIENCY

Not a Carrier of: F11 [c.403G>T (p.E135X), c.901T>C (p.F301L), c.438C>A (p.C146X), c.1716+1G>A (IVS14+1G>A)]

FAMILIAL DYSAUTONOMIA

Not a Carrier of: IKBKAP [c.2204+6T>C (IVS20+6T>C), c.2087G>C (p.R696P)]

FAMILIAL MEDITERRANEAN FEVER

Not a Carrier of: MEFV [c.2082G>A (p.M694I), c.2177T>C (p.V726A), c.2040G>C (p.M680I), c.2230G>T (p.A744S), c.2080A>G (p.M694V), c.1958G>A (p.R653H), c.2084A>G (p.K695R), c.2282G>A (p.R761H)]

FANCONI ANEMIA

Not a Carrier of: FANCC [c.456+4A>T (IVS4+4A>T), c.1642C>T (p.R548X), c.1661T>C (p.L554P), c.67delG (p.D23IfsX23, 322delG), c.553C>T (p.R185X), c.37C>T (p.Q13X)]

GALACTOKINASE DEFICIENCY

Not a Carrier of: GALK1 [c.1144C>T (p.Q382X), c.1031C>T (p.T344M), c.766C>T (p.R256W), c.1045G>A (p.G349S)]

GALACTOSEMIA

Not a Carrier of: GALT [c.652C>G (p.L218V), c.940A>G (p.N314D), c.563A>G (p.Q188R), c.-119_-116del, c.253-2A>G (IVS2-2A>G), c.404C>T (p.S135L), c.512T>C (p.F171S),

GALACTOSEMIA

c.584T>C (p.L195P), c.607G>A (p.E203K), c.626A>G (p.Y209C), c.855G>T (p.K285N)]

GAUCHER DISEASE

Not a Carrier of: GBA [c.1488T>C (p.L483P, L444P), c.1342G>C (p.D448H, D409H), c.1604G>A (p.R535H), c.1226A>G (p.N409S, N370S), c.1297G>T (p.V433L, V394L), c.1504C>T (p.R502C, R463C), c.115+1G>A (IVS2+1G>A), c.84dupG (p.L29AfsX18)]

GLUTARIC ACIDEMIA, TYPE 1

Not a Carrier of: GCDH [c.1262C>T (p.A421V), c.1204C>T (p.R402W), c.877G>A (p.A293T), c.1198G>A (p.V400M), c.680G>C (p.R227P)]

GLYCOGEN STORAGE DISEASE, TYPE 1A

Not a Carrier of: G6PC [c.247C>T (p.R83C), c.248G>A (p.R83H), c.79delC (p.Q27RfsX9), c.562G>C (p.G188R), c.809G>T (p.G270V), c.724C>T (p.Q242X), c.980_982del (p.F327del), c.1039C>T (p.Q347X), c.379_380dupTA (p.Y128TfsX3)]

GLYCOGEN STORAGE DISEASE, TYPE IB

Not a Carrier of: SLC37A4 [c.1042_1043del (p.L348VfsX53), c.1015G>T (p.G339C), c.352T>C (p.W118R)]

GLYCOGEN STORAGE DISEASE, TYPE III

Not a Carrier of: AGL [c.2590C>T (p.R864X), c.3682C>T (p.R1228X), c.3965delT (p.V1322AfsX27), c.4260-12A>G (IVS32-12A>G)]

GLYCOGEN STORAGE DISEASE, TYPE V

Not a Carrier of: PYGM [c.148C>T (p.R50X), c.613G>A (p.G205S)]

GM1-GANGLIOSIDOSIS

Not a Carrier of: GLB1 [c.176G>A (p.R59H)]

HEARING LOSS, DFNB1 AND DFNB9 NONSYNDROMIC

Not a Carrier of: GJB2 [c.377_378insATGCGGA (p.R127CfsX85), c.439G>A (p.E147K), c.109G>A (p.V37I), c.35delG (p.G12VfsX2), c.-23+1G>A, c.167delT (p.L56RfsX26), c.235delC (p.L79CfsX3), c.231G>A (p.W77X), c.269T>C (p.L90P), c.71G>A (p.W24X), c.299_300del (p.H100RfsX14), c.1A>G (p.M1V), c.283G>A (p.V95M), c.370C>T (p.Q124X)]; **OTOF** [c.2485C>T (p.Q829X)]

HEARING LOSS, DFNB59 NONSYNDROMIC

Not a Carrier of: DFNB59 [c.509_512del (p.S170CfsX37), c.726delT (p.F242LfsX7), c.988delG (p.V330LfsX7), c.113dupT (p.K41EfsX8), c.499C>T (p.R167X), c.731T>G (p.L244R)]

HEMOCHROMATOSIS

Not a Carrier of: HFE [c.845G>A (p.C282Y), c.187C>G (p.H63D), c.193A>T (p.S65C)]; **HFE2** [c.959G>T (p.G320V)]; **TFR2** [c.515T>A (p.M172K), c.750C>G (p.Y250X)]

HEMOGLOBIN C

Not a Carrier of: HBB [c.19G>A (p.E7K, Hemoglobin C)]

HEMOGLOBIN D

Not a Carrier of: HBB [c.364G>C (p.E122Q, Hemoglobin D-Punjab)]

HEMOGLOBIN E

Not a Carrier of: HBB [c.79G>A (p.E27K, Hemoglobin E)]

HEMOGLOBIN O

Not a Carrier of: HBB [c.364G>A (p.E122K, Hemoglobin O)]

HEREDITARY FRUCTOSE INTOLERANCE

Not a Carrier of: ALDOB [c.448G>C (p.A150P), c.524C>A (p.A175D), c.1005C>G (p.N335K), c.612T>A (p.Y204X)/c.612T>G (p.Y204X), c.360_363del (p.N120KfsX32)]

HERLITZ JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMA3-RELATED

Not a Carrier of: LAMA3 [c.1981C>T (p.R661X)]

HERLITZ JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMB3-RELATED

Not a Carrier of: LAMB3 [c.958_1034dup (p.N345KfsX77, 957ins77), c.124C>T (p.R42X), c.727C>T (p.Q243X), c.1903C>T (p.R635X)]

HERLITZ JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMC2-RELATED

Not a Carrier of: LAMC2 [c.283C>T (p.R95X)]

HMG-COA LYASE DEFICIENCY

Not a Carrier of: HMGCL [c.505_506del (p.S169LfsX8, 504_505delCT), c.122G>A (p.R41Q), c.109G>T (p.E37X)]

HOMOCYSTINURIA, CBLE TYPE

Not a Carrier of: MTRR [c.1953-6_1953-2del, c.1728_1730del (p.L576del, 1726delTTG), c.1622_1623dupTA (p.M542X), c.7A>T (p.R3W), c.1573C>T (p.R525X)]

HOMOCYSTINURIA, CLASSIC

Not a Carrier of: CBS [c.1591_1594del (p.F531GfsX9), c.892dupC (p.Q298PfsX32), c.1619_1622dupTGAA (p.F542EfsX37), c.1046G>A (p.S349N), c.676G>A (p.A226T), c.1126G>A (p.D376N), c.464C>T (p.A155V), c.503T>C

HOMOCYSTINURIA, CLASSIC

(p.V168A), c.694C>G (p.H232D), c.650C>T (p.S217F), c.129G>A (p.W43X), c.141T>A (p.D47E), c.969G>A (p.W323X), c.715G>A (p.E239K), c.262C>T (p.P88S), c.494G>A (p.C165Y), c.526G>A (p.E176K), c.384G>C (p.E128D), c.1063G>C (p.A355P), c.253G>A (p.G85R), c.376A>G (p.M126V), c.796A>G (p.R266G), c.1304T>C (p.I435T), c.1471C>T (p.R491C), c.1039+1G>T (IVS9+1G>T), c.828+1G>A (IVS7+1G>A), c.954+1G>A (IVS8+1G>A), c.833T>C (p.I278T), c.1106G>A (p.R369H), c.1330G>A (p.D444N), c.1105C>T (p.R369C), c.919G>A (p.G307S), c.434C>T (p.P145L), c.341C>T (p.A114V), c.415G>A (p.G139R), c.430G>A (p.E144K), c.1150A>G (p.K384E), c.1616T>C (p.L539S), c.797G>A (p.R266K), c.1397C>T (p.S466L), c.1058C>T (p.T353M), c.572C>T (p.T191M), c.146C>T (p.P49L), c.1060G>A (p.V354M), c.1111G>A (p.V371M), c.451G>A (p.G151R), c.1224-2A>C (IVS11-2A>C), c.1006C>T (p.R336C), c.172C>T (p.R58W), c.442G>A (p.G148R), c.770C>T (p.T257M), c.346G>A (p.G116R), c.1007G>A (p.R336H), c.869C>T (p.P290L), c.1135C>T (p.R379W), c.1039G>A (p.G347S), c.361C>T (p.R121C), c.325T>C (p.C109R), c.904G>A (p.E302K), c.959T>C (p.V320A), c.1566delG (p.K523SfsX18), c.233C>G (p.P78R), c.306G>C (p.K102N), c.1358+1G>A (IVS12+1G>A), c.302T>C (p.L101P)]

HURLER SYNDROME

Not a Carrier of: IDUA [c.1814_1815del (p.F605CfsX53), c.1044_1049del (p.D349_N350del), c.1695_1705del (p.L566GfsX2), c.1205G>A (p.W402X), c.208C>T (p.Q70X)]

HYPOPHOSPHATASIA, AUTOSOMAL RECESSIVE

Not a Carrier of: ALPL [c.571G>A (p.E191K), c.1133A>T (p.D378V), c.1001G>A (p.G334D), c.979T>C (p.F327L), c.1559delT (p.L520RfsX86)]

INCLUSION BODY MYOPATHY 2

Not a Carrier of: GNE [c.2228T>C (p.M743T), c.1807G>C (p.V603L)]

JUVENILE RETINOSCHISIS, X-LINKED

Not a Carrier of: RS1 [c.214G>A (p.E72K), c.221G>T (p.G74V), c.325G>C (p.G109R)]

KRABBE DISEASE

Not a Carrier of: GALC [c.857G>A (p.G286D)]

LIPOAMIDE DEHYDROGENASE DEFICIENCY

Not a Carrier of: DLD [c.685G>T (p.G229C)]

LIPOPROTEIN LIPASE DEFICIENCY, FAMILIAL

Not a Carrier of: LPL [c.644G>A (p.G215E)]

MAPLE SYRUP URINE DISEASE

Not a Carrier of: BCKDHB [c.548G>C (p.R183P), c.832G>A (p.G278S), c.1114G>T (p.E372X)]

MEDIUM-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY

Not a Carrier of: ACADM [c.985A>G (p.K329E), c.199T>C (p.Y67H)]

MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS

Not a Carrier of: HEPACAM [c.275G>A (p.R92Q)]; **MLC1** [c.298_423+108del, c.135dupC (p.C46LfsX34), c.176G>A (p.G59E), c.178-10T>A, c.278C>T (p.S93L)]

METACHROMATIC LEUKODYSTROPHY

Not a Carrier of: ARSA [c.1283C>T (p.P428L), c.293C>T (p.S98F), c.542T>G (p.I181S), c.257G>A (p.R86Q), c.641C>T (p.A214V), c.465+1G>A, c.1210+1G>A, c.1408_1418del (p.A470LfsX99)]

METHYLMALONIC ACIDEMIA

Not a Carrier of: MMAA [c.503delC (p.T168MfsX10), c.433C>T (p.R145X)]; **MUT** [c.2150G>T (p.G717V), c.349G>T (p.E117X), c.655A>T (p.N219Y), c.322C>T (p.R108C), c.1105C>T (p.R369C)]

MUCOLIPIDOSIS II

Not a Carrier of: GNPTAB [c.3503_3504del (p.L1168QfsX5), c.3613C>T (p.R1205X), c.1581delC (p.C528VfsX19), c.310C>T (p.Q104X), c.3565C>T (p.R1189X), c.2533C>T (p.Q845X), c.616_619del (p.T206YfsX6)]

MUCOLIPIDOSIS III

Not a Carrier of: GNPTAB [c.10A>C (p.K4Q), c.3335+6T>G (IVS17+6T>G)]; **GNPTG** [c.499dupC (p.L167PfsX32), c.347_349del (p.N116del)]

MUCOLIPIDOSIS IV

Not a Carrier of: MCOLN1 [del6.4kb, c.406-2A>G (IVS3-2A>G)]

MULTIPLE CARBOXYLASE DEFICIENCY

Not a Carrier of: HLCS [c.710T>C (p.L237P), c.1711G>A (p.D571N), c.1522C>T (p.R508W), c.1741G>A (p.G581S), c.1648G>A (p.V550M), c.1993C>T (p.R665X), c.782delG (p.G261VfsX20, 780delG)]

NEPHROTIC SYNDROME, STEROID-RESISTANT

Not a Carrier of: NPHS2 [c.436delA (p.R146EfsX35), c.1036delC (p.L346YfsX2), c.413G>A (p.R138Q)]

NEURONAL CEROID LIPOFUSCINOSIS, CLN3-RELATED

Not a Carrier of: CLN3 [c.461-280_677+382del]

NEURONAL CEROID LIPOFUSCINOSIS, CLN5-RELATED

Not a Carrier of: CLN5 [c.1175_1176del (p.Y392X)]

NEURONAL CEROID LIPOFUSCINOSIS, CLN8-RELATED

Not a Carrier of: CLN8 [c.70C>G (p.R24G)]

NEURONAL CEROID LIPOFUSCINOSIS, PPT1-RELATED

Not a Carrier of: PPT1 [c.364A>T (p.R122W), c.451C>T (p.R151X)]

NEURONAL CEROID LIPOFUSCINOSIS, TPP1-RELATED

Not a Carrier of: TPP1 [c.509-1G>C, c.622C>T (p.R208X)]

NIEMANN-PICK DISEASE

Not a Carrier of: NPC1 [c.2974G>T (p.G992W), c.3182T>C (p.I1061T)]; **NPC2** [c.58G>T (p.E20X)]; **SMPD1** [c.1493G>T (p.R498L, R496L), c.1829_1831del (p.R610del, deltaR608), c.911T>C (p.L304P, L302P), c.1267C>T (p.H423Y, H421Y), c.996delC (p.P333SfsX52, P330SfsX382)]

NIJMEGEN BREAKAGE SYNDROME

Not a Carrier of: NBN [c.657_661del (p.K219NfsX16)]

PENDRED SYNDROME

Not a Carrier of: SLC26A4 [c.1246A>C (p.T416P), c.707T>C (p.L236P), c.1001+1G>A, c.1151A>G (p.E384G), c.716T>A (p.V239D), c.919-2A>G, c.2168A>G (p.H723R), c.1540C>A (p.Q514K)]

PHENYLKETONURIA

Not a Carrier of: PAH [c.143T>C (p.L48S), c.473G>A (p.R158Q), c.727C>T (p.R243X), c.782G>A (p.R261Q), c.842C>T (p.P281L), c.1066-11G>A (IVS10-11G>A), c.1208C>T (p.A403V), c.1222C>T (p.R408W), c.1223G>A (p.R408Q), c.1241A>G (p.Y414C), c.728G>A (p.R243Q), c.838G>A (p.E280K), c.442-1G>A (IVS4-1G>A), c.611A>G (IVS6-96A>G), c.1068C>A (p.Y356X)/c.1068C>G (p.Y356X), c.117C>G (p.F39L), c.194T>C (p.I65T), c.734T>C (p.V245A), c.331C>T (p.R111X), c.721C>T (p.R241C), c.1238G>C (p.R413P)]

POLYCYSTIC KIDNEY DISEASE

Not a Carrier of: PKHD1 [c.8829dupC (p.I2944HfsX6), c.107C>T (p.T36M), c.1486C>T (p.R496X), c.10412T>G (p.V3471G), c.10444C>T (p.R3482C), c.2414C>T (p.P805L), c.9530T>C (p.I3177T), c.10174C>T (p.Q3392X), c.664A>G (p.I222V), c.9689delA (p.D3230VfsX34), c.8870T>C (p.I2957T)]

POMPE DISEASE

Not a Carrier of: GAA [c.2741delinsCAG (p.Q914PfsX30), c.1935C>A (p.D645E), c.2560C>T (p.R854X), c.525delT (p.E176RfsX45), c.925G>A (p.G309R)]

PREKALLIKREIN DEFICIENCY

Not a Carrier of: KLKB1 [c.1205G>A (p.W402X), c.1643G>A (p.C548Y)]

PRIMARY HYPEROXALURIA, TYPE 1

Not a Carrier of: AGXT [c.508G>A (p.G170R), c.33dupC (p.K12QfsX156)]

PRIMARY HYPEROXALURIA, TYPE 2

Not a Carrier of: GRHPR [c.103delG (p.D35TfsX11), c.403_404+2del]

PRIMARY HYPEROXALURIA, TYPE 3

Not a Carrier of: HOGA1 [c.700+5G>T, c.944_946del (p.E315del)]

PROTHROMBIN DEFICIENCY

Not a Carrier of: F2 [c.481G>T (p.D161Y, D118Y), c.787C>T (p.R263C, R220C), c.124C>T (p.R42W, R-2W), c.542G>A (p.C181Y, C138Y), c.940C>T (p.R314C, R271C), c.1054G>A (p.E352K, E309K), c.1499G>A (p.R500Q, R457Q), c.1741C>T (p.R581C, R538C)]

RH-NULL SYNDROME

Not a Carrier of: RHAG [c.808G>A (p.V270I)]

RHIZOMELIC CHONDRODYSPLASIA PUNCTATE TYPE 1

Not a Carrier of: PEX7 [c.875T>A (p.L292X), c.653C>T (p.A218V), c.649G>A (p.G217R)]

RICKETS, PSEUDOVITAMIN D-DEFICIENCY

Not a Carrier of: CYP27B1 [c.1166G>A (p.R389H), c.262delG (p.V88WfsX71, 958delG), c.589+1G>A (IVS3+1G>A), c.1319_1325dupCCCACCC (p.F443PfsX24, 3398dupCCCACCC)]

SALLA DISEASE

Not a Carrier of: SLC17A5 [c.115C>T (p.R39C)]

SANDHOFF DISEASE

Not a Carrier of: HEXB [c.76delA (p.M26CfsX5), c.445+1G>A (IVS2+1G>A)]

SHORT-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY

Not a Carrier of: ACADS [c.1170C>G (p.I390M), c.1138C>T (p.R380W), c.1058C>T (p.S353L), c.529T>C (p.W177R), c.319C>T (p.R107C), c.136C>T (p.R46W), c.417G>C (p.W139C), c.1095G>T (p.Q365H), c.1108A>G (p.M370V), c.596C>T (p.A199V), c.505A>C (p.T169P)]

SICK SINUS SYNDROME

Not a Carrier of: SCN5A [c.3893C>T (p.P1298L), c.659C>T (p.T220I), c.4222G>A (p.G1408R), c.4895G>A (p.R1632H)]

SICKLE CELL DISEASE

Not a Carrier of: HBB [c.20A>T (p.E7V, Hemoglobin S)]

SMITH-LEMLI-OPITZ SYNDROME

Not a Carrier of: DHCR7 [c.452G>A (p.W151X), c.1210C>T (p.R404C), c.278C>T (p.T93M), c.506C>T (p.S169L), c.724C>T (p.R242C), c.725G>A (p.R242H), c.906C>G (p.F302L), c.976G>T (p.V326L), c.1054C>T (p.R352W), c.1228G>A (p.G410S), c.1342G>A (p.E448K), c.832-1G>C (IVS8-1G>C)]

SPHEROCYTOSIS, HEREDITARY

Not a Carrier of: ANK1 [c.444+16C>T (5703+16C>T), c.1387G>A (p.V463I)]; **EPB42** [c.357G>A (p.W119X), c.424G>A (p.A142T), c.929G>A (p.R310Q), c.523G>T (p.D175Y), c.922+1G>A (IVS6+1G>A), c.949C>T (p.R317C)]

TAY-SACHS DISEASE

Not a Carrier of: HEXA [c.613delC (p.L205WfsX2), c.986G>A (p.W329X), c.1003A>T (p.I335F), c.1373G>A (p.C458Y), c.1074-1G>T (IVS9-1G>T), c.533G>A(p.R178H)/c.533G>T(p.R178L), c.1510C>T (p.R504C), c.1073+1G>A (IVS9+1G>A), c.805G>A (p.G269S), c.1496G>A (p.R499H), c.509G>A (p.R170Q), c.915_917del (p.F305del, deltaTTC910-912), c.629C>T (p.S210F), c.508C>T (p.R170W), c.1421+1G>C (IVS12+1G>C), c.571-1G>T (IVS5-1G>T), c.1274_1277dupTATC

TAY-SACHS DISEASE

(p.Y427IfsX5, 1278insTATC), c.574G>C (p.V192L), c.346+1G>C (IVS2+1G>C)]

TAY-SACHS PSEUDODEFICIENCY

Not a Carrier of: HEXA [c.739C>T (p.R247W), c.745C>T (p.R249W)]

THROMBOCYTOPENIA, CONGENITAL AMEGAKARYOCYTIC

Not a Carrier of: MPL [c.305G>C (p.R102P), c.127C>T (p.R43X)]

TYROSINE HYDROXYLASE DEFICIENCY

Not a Carrier of: TH [c.698G>A (p.R233H), c.707T>C (p.L236P)]

TYROSINEMIA

Not a Carrier of: FAH [c.192G>T (p.Q64H), c.554-1G>T, c.607-6T>G, c.782C>T (p.P261L), c.786G>A (p.W262X), c.1009G>A (p.G337S), c.1062+5G>A]

USHER SYNDROME, TYPE 1F

Not a Carrier of: PCDH15 [c.733C>T (p.R245X)]

VERY LONG-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY

Not a Carrier of: ACADVL [c.848T>C (p.V283A)]

VON WILLEBRAND DISEASE, TYPE 2 NORMANDY

Not a Carrier of: VWF [c.2311A>G (p.M771V), c.2561G>A (p.R854Q), c.2451T>A (p.H817Q), c.2287A>G (p.R763G), c.2344C>T (p.R782W), c.2354G>A (p.G785E), c.2359G>A (p.E787K), c.2362T>C (p.C788R), c.2363G>A (p.C788Y), c.2372C>T (p.T791M), c.2384A>G (p.Y795C), c.2635G>A (p.D879N), c.3159G>T (p.Q1053H), c.3178T>C (p.C1060R),

VON WILLEBRAND DISEASE, TYPE 2 NORMANDY

c.2411G>T (p.C804F), c.2435C>T (p.P812L), c.2447G>A (p.R816Q), c.2446C>T (p.R816W), c.3232G>A (p.E1078K), c.3673T>G (p.C1225G)]

VON WILLEBRAND DISEASE, TYPE 3

Not a Carrier of: VWF [c.3940delG (p.V1314SfsX34), c.1384delG (p.A462QfsX15), c.3258_3259insT (p.D1087X), c.3736_3737dupCC (p.P1247LfsX7), c.4324_4331dupAGTGTGGA (p.D1444EfsX84), c.7172_7173insT (p.E2391DfsX3), c.1693C>T (p.Q565X), c.3800T>A (p.L1267X), c.2016_2019del (p.S673TfsX67), c.2269_2270del (p.L757VfsX22), c.3943C>T (p.R1315C), c.4036C>T (p.Q1346X), c.4092_4093del (p.L1365VfsX11), c.4368C>A (p.Y1456X), c.5053+1G>A (IVS28+1G>A), c.5170+10C>T (IVS29+10C>T), c.5557C>T (p.R1853X), c.6182delT (p.F2061SfsX38), c.6520T>G (p.C2174G), c.6977-1G>C (IVS40-1G>C), c.7085G>T (p.C2362F), c.7603C>T (p.R2535X), c.7630C>T (p.Q2544X), c.7683delT (p.Q2562SfsX2), c.7729+7C>T (IVS45+7C>T), c.8012G>A (p.C2671Y), c.8155+3G>T (IVS50+3G>T), c.8216G>A (p.C2739Y), c.8262T>G (p.C2754W), c.139G>C (p.D47H), c.276delT (p.F92LfsX11), c.817C>T (p.R273W), c.970C>T (p.R324X), c.1071C>A (p.Y357X), c.1093C>T (p.R365X), c.1110-1G>A (IVS9-1G>A), c.1830C>A (p.Y610X), c.1858G>T (p.E620X), c.191delG (p.G64AfsX19), c.212C>A (p.S71X), c.652C>T (p.Q218X), c.666G>A (p.W222X), c.1117C>T (p.R373X), c.1131G>T (p.W377C), c.2157delA (p.D720TfsX21), c.7300C>T (p.R2434X), c.374_387del (p.G125VfsX3), c.874+1G>A (IVS7+1G>A), c.893dupG (p.M299YfsX4), c.1657dupT (p.W553LfsX97), c.3212G>T (p.C1071F), c.4626C>G (p.Y1542X), c.7139dupT (p.L2380FfsX11), c.7674dupC (p.S2559LfsX8), c.8411G>A (p.C2804Y)]

WILSON DISEASE

Not a Carrier of: ATP7B [c.2333G>T (p.R778L), c.3207C>A (p.H1069Q)]

ZELLWEGER SYNDROME SPECTRUM, PEX1-RELATED

Not a Carrier of: PEX1 [c.2097dupT (p.I700YfsX42), c.2528G>A (p.G843D)]

RESIDUAL RISK AFTER NEGATIVE TEST RESULTS

21-HYDROXYLASE-DEFICIENT CONGENITAL ADRENAL HYPERPLASIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Yupik Eskimos	1:9	100.0%	Negligible
General	1:60	69.0%	1:191

3-METHYLCROTONYL-COA CARBOXYLASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
German and Turkish	1:146	4.0%	1:151

ACHROMATOPSIA

CNGB3

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pingelapese	1:3	100.0%	Negligible
European	1:91	91.0%	1:1001

CNGA3

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
European	1:181	42.0%	1:311

ACRODERMATITIS ENTEROPATHICA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Tunisian	1:500	78.0%	1:2269

ALKAPTONURIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Czech, Slovak	1:90	50.0%	1:179
European (non-Slovak or Czech)	1:250	11.0%	1:281

ALPHA-1 ANTITRYPSIN DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Southern European	1:7	95.0%	1:121
North American	1:12	95.0%	1:221
African	1:14	95.0%	1:261
Northern European	1:15	95.0%	1:281
Middle East and North African	1:16	95.0%	1:301
Southeast Asian	1:84	95.0%	1:1661
Far East Asian	1:570	95.0%	1:11381

ALPHA-MANNOSIDOSIS

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:354	35.0%	1:544

AMYOTROPHIC LATERAL SCLEROSIS

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

ANDERMANN SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
French-Canadian	1:23	100.0%	Negligible

ARGININOSUCCINATE LYASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:194	50.0%	1:387

ARSACS

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
French-Canadian	1:21	96.0%	1:501

ASPARTYLGLUCOSAMINURIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Finnish	1:68	98.0%	1:3351

ATAXIA WITH VITAMIN E DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Mediterranean, North African	1:274	80.0%	1:1366

ATAXIA-TELANGIECTASIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Amish	1:100	100.0%	Negligible
Costa Rican	1:100	86.0%	1:708
North African Jewish	1:100	100.0%	Negligible
Norwegian	1:100	55.0%	1:221
Polish	1:100	39.0%	1:163
Sardinian	1:100	95.0%	1:1981
Turkish	1:100	33.0%	1:149

AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE I

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Iranian Jewish	1:48	100.0%	Negligible
Finnish	1:80	71.0%	1:273
Slovenian	1:104	67.0%	1:313
Norwegian	1:150	48.0%	1:288
Polish	1:250	71.0%	1:860

BARDET-BIEDL SYNDROME, BBS1-RELATED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
North American, European	1:387	79.0%	1:1839

BARTTER SYNDROME, TYPE 4A

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

BETA-KETOTHIOLASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Vietnamese	1:500	88.0%	1:4159

BETA-THALASSEMIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Mediterranean	1:7	91.0%	1:68
Thai	1:11	91.0%	1:112
West African	1:11	75.0%	1:41
Middle Eastern	1:49	91.0%	1:534
Chinese	1:100	91.0%	1:1101

BIOTINIDASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:120	89.0%	1:1083

BLOOM SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:107	99.0%	1:10601

CANAVAN DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:41	97.0%	1:1540

CARNITINE DEFICIENCY, PRIMARY SYSTEMIC

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Taiwanese	1:1000	35.0%	1:153

CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

CARTILAGE-HAIR HYPOPLASIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Old Order Amish	1:10	100.0%	Negligible
Finnish	1:76	92.0%	1:939

CEREBROTENDINOUS XANTHOMATOSIS

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
North African Jewish	1:5	79.0%	1:20
Dutch	1:111	100.0%	Negligible

CHOROIDEREMIA

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

CITRULLINEMIA, TYPE I

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:119	46.0%	1:220

COHEN SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Old Order Amish	1:11	99.0%	1:1001

COMBINED PITUITARY HORMONE DEFICIENCY, PROP1-RELATED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Worldwide	1:63	55.0%	1:139

CONGENITAL DISORDER OF GLYCOSYLATION TYPE IA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Danish	1:60	88.0%	1:493

COSTEFF OPTIC ATROPHY SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Iraqi Jewish	1:10	100.0%	Negligible

CRIGLER-NAJJAR SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Caucasian	1:500	75.0%	1:1997

CYSTIC FIBROSIS

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:24	94.0%	1:384
Non-Hispanic Caucasian	1:25	88.0%	1:206
Hispanic Caucasian	1:58	72.0%	1:205
African American	1:61	64.0%	1:171
Asian American	1:94	49.0%	1:183

CYSTINOSIS

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
US general, Northern European	1:158	50.0%	1:315

DIABETES, PERMANENT NEONATAL

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

DIHYDROPYRIMIDINE DEHYDROGENASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Caucasian	1:56	52.0%	1:116

DUBIN-JOHNSON SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Iranian Jewish	1:18	100.0%	Negligible
Moroccan Jewish	1:18	100.0%	Negligible

EHLERS-DANLOS SYNDROME, DERMATOSPARAXIS

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

EHLERS-DANLOS SYNDROME, HYPERMOBILITY

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

EHLERS-DANLOS SYNDROME, KYPHOSCOLIOTIC

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

FACTOR V LEIDEN THROMBOPHILIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
European American	1:18	100.0%	0
Hispanic American	1:45	100.0%	0
Native American	1:80	100.0%	0
African American	1:83	100.0%	0
Asian American	1:222	100.0%	0

FACTOR XI DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:11	98.0%	1:501
U.K. Pan-ethnic	1:500	39.0%	1:819

FAMILIAL DYSAUTONOMIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:31	99.0%	1:3001

FAMILIAL MEDITERRANEAN FEVER

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Armenian	1:3	79.0%	1:11
Ashkenazi Jewish	1:4	54.0%	1:8
Non-Ashkenazi Jewish	1:4	69.0%	1:11
Turkish	1:6	76.0%	1:22
Arab	1:7	53.0%	1:14

FANCONI ANEMIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:89	99.0%	1:8801

GALACTOKINASE DEFICIENCY

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

GALACTOSEMIA

Duarte Variant

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Northern European	1:9	100.0%	0
Western European	1:11	100.0%	0
Eastern European	1:12	100.0%	0
Southern European	1:18	100.0%	0
Asian	1:56	100.0%	0

Classic

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Northern European	1:111	80.0%	1:551
Southern European	1:234	80.0%	1:1166
Western European	1:270	80.0%	1:1346
African American	1:1010	80.0%	1:5046
Eastern European	1:1016	80.0%	1:5076

GAUCHER DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:18	90.0%	1:171
Pan-ethnic	1:50	64.0%	1:137

GLUTARIC ACIDEMIA, TYPE 1

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Caucasian	1:159	38.0%	1:256

GLYCOGEN STORAGE DISEASE, TYPE 1A

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:71	93.0%	1:1001
Non-Jewish	1:158	62.0%	1:414

GLYCOGEN STORAGE DISEASE, TYPE IB

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
European	1:354	47.0%	1:667
Japanese	1:354	50.0%	1:707

GLYCOGEN STORAGE DISEASE, TYPE III

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
General	1:158	20.0%	1:197

GLYCOGEN STORAGE DISEASE, TYPE V

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
US general	1:158	41.0%	1:267
Spanish	1:206	41.0%	1:348

GM1-GANGLIOSIDOSIS

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

HEARING LOSS, DFNB1 AND DFNB9 NONSYNDROMIC

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

HEARING LOSS, DFNB59 NONSYNDROMIC

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

HEMOCHROMATOSIS

HFE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Northern European	1:3	63.0%	1:6

HFE2

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
French-Canadian	1:500	100.0%	Negligible
Greek	1:500	70.0%	1:1664
Italian	1:500	4.0%	1:521

HEMOGLOBIN C

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
African American	1:52	100.0%	0
Native American	1:489	100.0%	0
Hispanic American	1:1517	100.0%	0
Caucasian	1:2754	100.0%	0
Asian Indian	1:4768	100.0%	0
Filipino	1:4775	100.0%	0
Middle Eastern	1:5476	100.0%	0
Asian	1:6607	100.0%	0
Southeast Asian	1:14200	100.0%	0

HEMOGLOBIN D

HbD-Punjab

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
South Asian	1:232	100.0%	0

HEMOGLOBIN E

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Bangladeshi	1:24	100.0%	0
Chinese	1:221	100.0%	0
Pakistani	1:529	100.0%	0
Asian Indian	1:578	100.0%	0
White Irish	1:1961	100.0%	0
White British	1:9091	100.0%	0
African	1:10000	100.0%	0

HEMOGLOBIN O

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
South Asian	1:233	100.0%	0
General	1:1428	100.0%	0
African American	1:30000	100.0%	0

HEREDITARY FRUCTOSE INTOLERANCE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Middle Eastern	1:97	50.0%	1:193
US general	1:122	50.0%	1:243
African American	1:226	50.0%	1:451

HERLITZ JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMA3-RELATED

LAMA3, LAMB3, LAMC2

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
US general	1:781	45.0%	1:1419

HERLITZ JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMB3-RELATED

LAMA3, LAMB3, LAMC2

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
US general	1:781	45.0%	1:1419

HERLITZ JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMC2-RELATED

LAMA3, LAMB3, LAMC2

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
US general	1:781	45.0%	1:1419

HMG-COA LYASE DEFICIENCY

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

HOMOCYSTINURIA, CBLE TYPE

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

HOMOCYSTINURIA, CLASSIC

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
UK	1:500	50.0%	1:999
US general	1:500	26.0%	1:675

HURLER SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Caucasian	1:159	79.0%	1:753

HYPOPHOSPHATASIA, AUTOSOMAL RECESSIVE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Japanese	1:194	41.0%	1:328

INCLUSION BODY MYOPATHY 2

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Middle Eastern Jewish	1:15	100.0%	Negligible
Japanese	Unknown	100.0%	Negligible

JUVENILE RETINOSCHISIS, X-LINKED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Finnish	1:65	95.0%	1:1281

KRABBE DISEASE

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

LIPOAMIDE DEHYDROGENASE DEFICIENCY

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

LIPOPROTEIN LIPASE DEFICIENCY, FAMILIAL

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Caucasian	1:500	46.0%	1:925

MAPLE SYRUP URINE DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:97	99.0%	1:1921

MEDIUM-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Caucasian	1:40	70.0%	1:131

MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

METACHROMATIC LEUKODYSTROPHY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Australian	1:100	46.0%	1:184
Polish	1:100	54.0%	1:216

METHYLMALONIC ACIDEMIA

MUT

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Japanese	1:187	22.0%	1:239
Black	1:237	35.0%	1:364
Caucasian	1:237	19.0%	1:292
Hispanic	1:237	41.0%	1:401

MMAA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Japanese	1:448	64.0%	1:1243
Caucasian	1:568	43.0%	1:996

MUCOLIPIDOSIS II

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Japanese	1:500	60.0%	1:1249
Predominantly white	1:500	56.0%	1:1135

MUCOLIPIDOSIS III

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

MUCOLIPIDOSIS IV

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:127	95.0%	1:2521

MULTIPLE CARBOXYLASE DEFICIENCY

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

NEPHROTIC SYNDROME, STEROID-RESISTANT

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

NEURONAL CEROID LIPOFUSCINOSIS, CLN3-RELATED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Finnish	1:70	85.0%	1:461
West German	1:188	85.0%	1:1248

NEURONAL CEROID LIPOFUSCINOSIS, CLN5-RELATED

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

NEURONAL CEROID LIPOFUSCINOSIS, CLN8-RELATED

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

NEURONAL CEROID LIPOFUSCINOSIS, PPT1-RELATED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Finnish	1:70	98.0%	1:3451

NEURONAL CEROID LIPOFUSCINOSIS, TPP1-RELATED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Newfoundlander	1:53	69.0%	1:169

NIEMANN-PICK DISEASE

Type A

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:90	97.0%	1:2968

NIJMEGEN BREAKAGE SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Eastern European Slavic	1:155	100.0%	Negligible
North American	1:158	70.0%	1:524

PENDRED SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Chinese	1:50	84.0%	1:307
Japanese	1:50	53.0%	1:105
Northern European Caucasian	1:60	50.0%	1:119

PHENYLKETONURIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Irish	1:34	72.0%	1:119
Turkish	1:34	65.0%	1:95
French-Canadian	1:45	56.0%	1:101
Polish	1:45	78.0%	1:201
Spanish	1:51	41.0%	1:86
Chinese	1:53	54.0%	1:114
Danish	1:55	43.0%	1:96
US Caucasian	1:62	51.0%	1:125
Korean	1:102	62.0%	1:267
Japanese	1:174	70.0%	1:578

POLYCYSTIC KIDNEY DISEASE

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

POMPE DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
African American	1:59	60.0%	1:146
Dutch	1:185	40.0%	1:308
Taiwanese, Chinese	1:185	80.0%	1:921

PREKALLIKREIN DEFICIENCY

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

PRIMARY HYPEROXALURIA, TYPE 1

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
European	1:173	44.0%	1:308
Worldwide	1:289	44.0%	1:515

PRIMARY HYPEROXALURIA, TYPE 2

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

PRIMARY HYPEROXALURIA, TYPE 3

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Worldwide	1:913	75.0%	1:3649

PROPIONIC ACIDEMIA

PCCA, PCCB

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Japanese	1:160	35.0%	1:246

PCCB

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Northern European	1:160	30.0%	1:228
Spanish	1:160	50.0%	1:320

PROTHROMBIN DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Iranian, Italian	1:707	54.0%	1:1536

RH-NULL SYNDROME

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

RHIZOMELIC CHONDRODYSPLASIA PUNCTATE TYPE 1

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:158	51.0%	1:321

RICKETS, PSEUDOVITAMIN D-DEFICIENCY

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

SALLA DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Northeastern Finnish	1:100	95.0%	1:1981

SANDHOFF DISEASE

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

SHORT-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:15	65.0%	1:41

SICK SINUS SYNDROME

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

SICKLE CELL DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
African American	1:15	100.0%	0
Native American	1:150	100.0%	0
Hispanic American	1:203	100.0%	0
Middle Eastern	1:478	100.0%	0
Caucasian	1:642	100.0%	0
Asian Indian	1:652	100.0%	0
Filipino	1:879	100.0%	0
Asian	1:1315	100.0%	0
Southeast Asian	1:2365	100.0%	0

SMITH-LEMLI-OPITZ SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Northwestern European	1:50	69.2%	1:150
General	1:68	69.2%	1:219
Southern European	1:83	69.2%	1:267
Middle Eastern	1:129	69.2%	1:417
Hispanic	1:135	69.2%	1:436
African American	1:339	69.2%	1:1098

SPHEROCYTOSIS, HEREDITARY

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

TAY-SACHS DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:31	99.0%	1:3001
Non-Jewish	1:250	46.0%	1:462

TAY-SACHS PSEUDODEFICIENCY

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

THROMBOCYTOPENIA, CONGENITAL AMEGAKARYOCYTIC

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

TYROSINE HYDROXYLASE DEFICIENCY

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

TYROSINEMIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
French-Canadian	1:66	87.0%	1:501
Ashkenazi Jewish	1:100	99.0%	1:9901
US general	1:150	60.0%	1:374

USHER SYNDROME, TYPE 1F

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

VERY LONG-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

VON WILLEBRAND DISEASE, TYPE 2 NORMANDY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:500	75.0%	1:1997

VON WILLEBRAND DISEASE, TYPE 3

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Swedish, Finnish	1:500	10.0%	1:555

WILSON DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Asian	1:90	57.0%	1:208
European	1:90	35.0%	1:138

ZELLWEGER SYNDROME SPECTRUM, PEX1-RELATED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:147	80.0%	1:731

TEST METHODOLOGY

Genotyping by PCR-based enrichment and next-generation sequencing.

RESULT STATUS DEFINITIONS

Final

Test results that are available at the time of report issue or have been revised from pending status to final status.