Amby Lab

Carrier status testing

Amby Lab is making high-quality genetic Carrier Status testing applying SNP microarray genotyping to identify alterations in human genome.

Carrier Status testing detects genetic variants that may cause inherited conditions. Being a carrier means you have one variant for the condition. Carriers typically don't have the condition but can pass the variant to their children.

If **both parents** are carriers, there is a 25% chance their child will have the condition and a 50% chance that their child will be the carrier of the same condition. If **a woman** is the carrier of **X-linked mutation,** there are a 50% chance her son will have the condition and a 50% chance her daughter will be a carrier of the same condition.

Understanding your carrier status helps you work with your doctor to prepare for the health of your future family.

Important note: If you do not have frequent mutations we tested, that does not mean the absence of a chance that a child will have rare mutations, not covered by this test, as well as emergence of new (*de novo*) mutations not observed in his parents.

Amby Lab identifies alterations in the SNPs associated with diseases listed below. This list contains monogenic inherited diseases, for which all detectable mutations are responsible for more than 50% of the cases.

DISEASE LIST

DISEASE NAME	OMIM ID	COVERAGE OF CASES
Abetalipoproteinemia	200100	100%
Aceruloplasminemia	604290	100%
Achondrogenesis, type ib	600972	100%
Achromatopsia 3	262300	100%
Acth deficiency, isolated	201400	100%
Acyl-coa dehydrogenase, short-chain, deficiency of	201470	100%
Albinism, oculocutaneous, type 1	203100	75%
Albinism, oculocutaneous, type ii	203200	67%
Albinism, oculocutaneous, type iii	203290	100%
Alkaptonuria	203500	100%
Alpha-thalassemia	604131	77%
Amegakaryocytic thrombocytopenia, congenital	604498	86%
Amelogenesis imperfecta, hypomaturation type, iia1	204700	100%

Amelogenesis imperfecta, type ig	614253	100%
Amyloidosis, familial visceral	105200	100%
Amyotrophic lateral sclerosis 9	611895	100%
Argininemia	207800	80%
Arterial calcification, generalized, of infancy, 2	614473	65%
Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	208920	100%
Bardet-biedl syndrome 1	209900	100%
Bardet-biedl syndrome 2	615981	100%
Bardet-biedl syndrome 3	600151	100%
Bietti crystalline corneoretinal dystrophy	210370	100%
Bile acid synthesis defect, congenital, 3	613812	100%
Blood group, junior system	614490	100%
Breast-ovarian cancer, familial, susceptibility to, 2	612555	83%
C1q deficiency	613652	100%
Calcification of joints and arteries	211800	100%
Carbamoyl phosphate synthetase i deficiency, hyperammonemia due to	237300	100%
Carnitine deficiency, systemic primary	212140	64%
Carpenter syndrome 1	201000	100%
Ceroid lipofuscinosis, neuronal, 1	256730	97%
Ceroid lipofuscinosis, neuronal, 2	204500	100%
Charcot-marie-tooth disease, 4c	609260	100%
Charcot-marie-tooth disease, axonal, type 2a2	615376	100%
Charcot-marie-tooth disease, recessive intermediate c	601596	92%
Charcot-marie-tooth disease, x-linked dominant, 1	302800	80%
Cholestasis, benign recurrent intrahepatic, 2	605479	100%
Cholestasis, intrahepatic, of pregnancy 3	614972	100%
Cholestasis, progressive familial intrahepatic, 2	601847	100%
Ciliary dyskinesia, primary, 19	614935	100%
Citrullinemia, type ii, adult-onset	603471	100%
Citrullinemia, type ii, neonatal-onset	605814	100%
Coach syndrome	216360	100%
Cockayne syndrome b	133540	100%
Combined oxidative phosphorylation deficiency 1	609060	100%
Combined oxidative phosphorylation deficiency 15	614947	100%
Complement component 5 deficiency	609536	100%
Complement component 9 deficiency	613825	75%
Cone-rod dystrophy 3	604116	100%
Cystinosis, nephropathic	219800	100%
Cystinuria	220100	100%
De sanctis-cacchione syndrome	278800	100%
Deafness, autosomal dominant 6	600965	100%
Deafness, autosomal recessive 12	601386	100%
Deafness, autosomal recessive 18a	602092	100%
Deafness, autosomal recessive 2	600060	100%
Deafness, autosomal recessive 31	607084	100%
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	600791	85%
Deafness, autosomal recessive 59	610220	100%
Deafness, autosomal recessive 7	600974	100%
Deafness, onychodystrophy, osteodystrophy, mental retardation, and seizures syndrome	222760	100%

Diastrophic dysplasia	222600	100%
Dubin-johnson syndrome	237500	67%
Ectopia lentis 2, isolated, autosomal recessive	225100	67%
Enterokinase deficiency	226200	100%
Epidermolysis bullosa, junctional, herlitz type	226700	100%
Epidermolysis bullosa, junctional, non-herlitz type	226650	100%
Epilepsy, pyridoxine-dependent	266100	67%
Fabry disease	301500	59%
Factor v deficiency	227400	100%
Factor xi deficiency	612416	91%
Factor xiii, a subunit, deficiency of	613225	80%
Familial cold autoinflammatory syndrome 2	611762	100%
Fanconi anemia, complementation group c	227645	100%
Fanconi anemia, complementation group i	609053	100%
Fanconi anemia, complementation group j	609054	100%
Fructose intolerance, hereditary	229600	100%
Galactokinase deficiency	230200	100%
Gitelman syndrome	263800	93%
Glanzmann thrombasthenia	273800	100%
Glaucoma 1, open angle, a	137750	100%
Glaucoma 1, open angle, o	613100	100%
Glaucoma 3, primary congenital, a	231300	100%
Glaucoma 3, primary congenital, d	613086	100%
Glutathione synthetase deficiency	266130	100%
Glycogen storage disease 0, liver	240600	100%
Glycogen storage disease 1	232220	92%
Glycogen storage disease ii	232300	96%
Glycogen storage disease ixb	261750	100%
Glycogen storage disease v	232600	94%
Glycogen storage disease x	261670	100%
Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type ii	233710	100%
Hartnup disorder	234500	100%
Hemophagocytic lymphohistiocytosis, familial, 3	608898	100%
Hemophilia b	306900	100%
Hereditary leiomyomatosis and renal cell cancer	605839	83%
Hurler syndrome	607014	93%
Hyperbilirubinemia, rotor type	237450	100%
Hypercholesterolemia, familial	144400	67%
Hyperekplexia, hereditary 1	149400	100%
Hyperinsulinemic hypoglycemia, familial, 2	601820	88%
Hyperinsulinemic hypoglycemia, familial, 4	609975	100%
Hyperphenylalaninemia, bh4-deficient, d	264070	67%
Hyperthyroxinemia, familial dysalbuminemic	615999	100%
Hypogonadotropic hypogonadism 11 with or without anosmia	614840	100%
Hypogonadotropic hypogonadism 4 with or without anosmia	610628	100%
Hypogonadotropic hypogonadism 6 with or without anosmia	612702	100%
Hypomagnesemia 3, renal	248250	100%
Hypophosphatasia, infantile	241500	94%
Hypothyroidism, congenital, nongoitrous, 1	275200	100%

Hypouricemia, renal, 1	220150	100%
Ichthyosis prematurity syndrome	608649	100%
Ichthyosis, congenital, autosomal recessive 3	606545	100%
Immunodeficiency, common variable, 2	240500	100%
Immunoglobulin a deficiency 2	609529	100%
Inclusion body myopathy 2, autosomal recessive	600737	100%
Irak4 deficiency	607676	100%
Iron-refractory iron deficiency anemia	206200	80%
Joubert syndrome 2	608091	100%
Joubert syndrome 3	608629	100%
Joubert syndrome 5	610188	100%
Joubert syndrome 6	610688	100%
Joubert syndrome 7	611560	100%
Kanzaki disease	609242	100%
Lactase deficiency, congenital	223000	100%
Laron syndrome	262500	100%
Leber congenital amaurosis 10	611755	100%
Leber congenital amaurosis 13	612712	100%
Leber congenital amaurosis 2	204100	100%
Leber congenital amaurosis 3	604232	100%
Leber congenital amaurosis 4	604393	100%
Leber congenital amaurosis 5	604537	100%
Leber congenital amaurosis 8	613835	100%
Lecithin:cholesterol acyltransferase deficiency	245900	100%
Left ventricular noncompaction 7	615092	100%
Lesch-nyhan syndrome	300322	100%
Lig4 syndrome	606593	100%
Lipodystrophy, congenital generalized, type 3	612526	100%
Lipoid proteinosis of urbach and wiethe	247100	100%
Long qt syndrome 1	192500	89%
Macular degeneration, juvenile	248200	86%
Maple syrup urine disease	248600	80%
Meckel syndrome, type 1	249000	73%
Meckel syndrome, type 3	607361	100%
Meckel syndrome, type 5	611561	100%
Methylmalonic aciduria and homocystinuria, cblc type	277400	100%
Methylmalonyl-coa epimerase deficiency	251120	100%
Microcephalic osteodysplastic primordial dwarfism, type ii	210720	75%
Microcephaly 6, primary, autosomal recessive	608393	100%
Microcephaly 9, primary, autosomal recessive	614852	100%
Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma	251750	100%
Mitochondrial complex i deficiency	252010	67%
Mitochondrial complex iii deficiency, nuclear type 1	124000	100%
Molybdenum cofactor deficiency, complementation group a	252150	100%
Mononeuropathy of the median nerve, mild	613353	100%
Mucolipidosis ii alpha/beta	252500	75%
Mucopolysaccharidosis, type iiia	252900	100%
Muscular dystrophy, congenital merosin-deficient, 1a	607855	100%

Muscular dystrophy, limb-girdle, type 2a	253600	100%
Muscular dystrophy, limb-girdle, type 2b	253601	100%
Myoclonic epilepsy of unverricht and lundborg	254800	100%
Myopathy, congenital, with fiber-type disproportion	255310	100%
Nanophthalmos 2	609549	100%
Nephronophthisis 2	602088	100%
Neurodegeneration due to cerebral folate transport deficiency	613068	100%
Neurodegeneration with brain iron accumulation 1	234200	100%
Neuropathy, hereditary sensory and autonomic, type iii	223900	100%
Neutropenia, severe congenital, 4, autosomal recessive	612541	100%
Niemann-pick disease, type a	257200	100%
Niemann-pick disease, type c1	257220	100%
Night blindness, congenital stationary, type 1b	257270	100%
Night blindness, congenital stationary, type 1f	615058	100%
Nonaka myopathy	605820	100%
Obesity	601665	100%
Odontoonychodermal dysplasia	257980	100%
Orofaciodigital syndrome iv	258860	100%
Osteogenesis imperfecta, type viii	610854	100%
Osteogenic sarcoma	259500	86%
Osteopetrosis, autosomal recessive 1	259700	100%
Pancreatitis, hereditary	167800	96%
Papillon-lefevre syndrome	245000	100%
Paragangliomas 5	614165	100%
Parkinson disease 2, autosomal recessive juvenile	600116	100%
Pendred syndrome	274600	83%
Peripheral neuropathy, myopathy, hoarseness, and hearing loss	614369	100%
Peroxisome biogenesis disorder 5a (zellweger)	614866	100%
Peroxisome biogenesis disorder 9b	614879	100%
Peters anomaly	604229	100%
Peutz-jeghers syndrome	175200	100%
Phenylketonuria	261600	55%
Pigmented nodular adrenocortical disease, primary, 2	610475	100%
Polycystic kidney disease, autosomal recessive	263200	100%
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	221770	100%
Prekallikrein deficiency	612423	100%
Propionic acidemia	606054	100%
Prostate cancer, hereditary, 1	601518	100%
Pseudohypoaldosteronism, type i, autosomal recessive	264350	100%
Pulmonary hypertension, primary, 1	178600	100%
Pycnodysostosis	265800	100%
Pyridoxamine 5-prime-phosphate oxidase deficiency	610090	100%
Pyruvate kinase deficiency of red cells	266200	100%
Retinitis pigmentosa 14	600132	100%
Retinitis pigmentosa 20	613794	100%
Retinitis pigmentosa 26	608380	100%
Retinitis pigmentosa 28	606068	83%
Rhizomelic chondrodysplasia punctata, type 1	215100	71%
Rigid spine muscular dystrophy 1	602771	100%

Rothmund-thomson syndrome	268400	100%
Rubinstein-taybi syndrome 1	180849	100%
Sarcosinemia	268900	100%
Schopf-schulz-passarge syndrome	224750	100%
Senior-loken syndrome 4	606996	100%
Senior-loken syndrome 5	609254	100%
Severe combined immunodeficiency with sensitivity to ionizing radiation	602450	100%
Sitosterolemia	210250	77%
Spastic paraplegia 54, autosomal recessive	615033	100%
Spastic paraplegia 5a	270800	100%
Spastic paraplegia 7, autosomal recessive	607259	100%
Spondylocostal dysostosis 1, autosomal recessive	277300	100%
Stuve-wiedemann syndrome	601559	100%
Succinic semialdehyde dehydrogenase deficiency	271980	100%
Supranuclear palsy, progressive, 1	601104	100%
Tangier disease	205400	100%
Three m syndrome 1	273750	100%
Thrombophilia due to protein c deficiency	176860	100%
Thrombotic thrombocytopenic purpura, congenital	274150	83%
Thyroid dyshormonogenesis 3	274700	100%
Tooth agenesis, selective, 4	150400	100%
Treacher collins syndrome 3	248390	100%
Trimethylaminuria	602079	71%
Tumoral calcinosis, hyperphosphatemic, familial	610233	100%
Tyrosinemia, type i	276700	100%
Usher syndrome, type i	276900	59%
Usher syndrome, type if	602083	100%
Usher syndrome, type iiia	276902	100%
Van der woude syndrome 1	119300	100%
Vas deferens, congenital bilateral aplasia of	277180	71%
Vitamin d hydroxylation-deficient rickets, type 1a	264700	100%
Von willebrand disease, type 1	193400	100%
Werner syndrome	277700	100%
Wilson disease	277900	100%
Xeroderma pigmentosum, complementation group a	278700	100%