INFORMED CONSENT FOR CARRIER TESTING

Before signing this consent form, you should review the information below and discuss prenatal genetic testing with your healthcare provider or genetic counselor. I authorize my physician to obtain a sample of my blood for DNA testing to determine if I am a carrier for any of these diseases as noted on the reverse of this form.

Note: InheriGen Plus includes all below diseases as well as Cystic Fibrosis, Fragile X Syndrome, and Spinal Muscular Atrophy (SMA).

INHERIGEN DISEASE LIST

17_--hydroxylase/17,20-lyase Deficiency
3-Hydroxy-3-Methylglutaryl CoA lyase deficiency
6-pyruvyl-tetrahydropterin synthase (PTPS) deficiency
Abetalipoproteinemia
Achondroplasia, CH đoNGb-associated
Adenosine Deaminase Deficiency
Agammaglobulinemia of the Corpus Callosum with Peripheral Neuropathy (Wiedemann Syndrome)
Aldri-Blader Syndrome
Agranuloctocytic Acardia
Apathy-Glycosaminuria
Ataxia Neuronal injury (ANS)
Ataxia with Vitamin E Deficiency
Ataxia-Telangiectasia
Ataxia with Vitamin E Deficiency
Autoimmune Polyglandular Syndrome, Type 1
Autosomal Recessive Spastic Ataxia of Charcot-Marie-Tooth Disease, Type 4D
Carpenter syndrome
Carnitine Palmitoyltransferase Deficiency, Type 1A
Canavan Disease
Bardet-Biedl syndrome, BBS12-associated
Bardet-Biedl Syndrome, BBS5-associated
Bardet-Biedl Syndrome, BBS1-associated
Bernard-Soulier syndrome (DS), Type A1
Bernard-Soulier syndrome (DS), Type C
Beta-thalassemia
Bilateral Frontaltemporal Polymicrogyria
Blom Syndrome
Canavan Disease
Carbamyltransferease Deficiency, Type 1A
Carntine Palmitoyltransferase Deficiency, Type 2
Carpenter syndrome
Cerebrorenal xanthomatosis
Charcot-Marie-Tooth Disease, Type 4D
Choricentasia
Cirrhosis Deficiency
Colonic Syndrome
Congenital Ameagakaryocytic Thrombocytopenia (CANT)
Congenital Disorder of Glycosylation, Type la
Congenital Disorder of Glycosylation, Type lb
Congenital Finnish Nephrosis
Congenital Myasthenic Syndrome, CHRNA-associated
Congenital Myasthenic Syndrome, RAPSN-associated
CRBP1-associated retinal dystrophies
Crigler-Najjar syndrome
Cytidinosis
Dihydroxyaspartase Deficiency
Dihydropyrimidine dehydrogenase deficiency
Ethylmalonic Encephalopathy
Factor IX Deficiency (hemophilia C)
Familial Dysautonomia
Familial Hypochromatemia, LDLRAP1-associated
Familial Hypochromatemia, LDL-associated
Familial Hyperinsulinism
Familial Mediterranean Fever
Familial Neurohypoglycemia Diabetes Insipidus (FNID), Autosomal Recessive
Fanci Anemia, complementation group C
Fanci Anemia, complementation group G
Galegerma
Gangliosida
Glutaric Acidemia, Type I
Glutaric Acidemia, Type II
Glutaric Acidemia, Type IC
Glycogen Storage Disease, Type la
Glycogen Storage Disease, Type lb
Glycogen Storage Disease, Type II (Pompe Disease)
Glycogen Storage Disease, Type III
Glycogen Storage Disease, Type V (McCrdle Disease)
Gracile Syndrome
Hemmersky-Pudlak syndrome
Holocarbonyl Synthetase Deficiency
Homocystinuria (CBS Deficiency)
Homocystinuria, cbs type
Hurler Syndrome (mucopolysaccharidosis type I)
Hyperglycinemia-Hyperekatomatosis-
Hemocritullina (HHH) Syndrome
Hypophosphatasa
Inclusion body myopathy 2
Joubert syndrome 2
Junctional Epidermolysis Bullosa, Herlit, LMAM-associated
Junctional Epidermolysis Bullosa, Herlit, LAMC2-associated
Junctional Epidermolysis Bullosa, Herlit, LAMC2-associated
Krabbe Disease
Lamellar ichthyosis, Type 1
Leber Congenital Amaurosis, CEP290-associated
Leber congenital Amaurosis, RHOH-associated
Leigh Syndrome, French-Canadian Type
Lekuonecephalopathy with Vanshing White Matter (WWW)
Limb-Girdle Muscular Dystrophy, Type 2A
Limb-Girdle Muscular Dystrophy, Type 2C
Limb-Girdle Muscular Dystrophy, Type 2D
Limb-Girdle Muscular Dystrophy, Type 2E
Lipoprotein Lipoarity Deficiency
Long-Chain 3-Hydroxyacyl-Coenzyme A Defhydrase (LCHAD) Deficiency
Lysozyme Protein Intolerance
Maple Syrup Urine Disease, Type 1A
Maple Syrup Urine Disease, Type 1B
Meckel-Gruber Syndrome
Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency
Megalencephalic Leukoecephalopathy with Subcortical Cysts
Metachromatic Leukodystrophy
Mitochondrial Myopathy, MAMM-associated
Mitochondrial Myocardia, MIT-associated
Mitochondrial Myocardiopathy and Homocystinuria, CblAm (cblC) Type
Mucolipidosis, Type IV
Navaz Neurohepatopathy
Nemaline Myopathy, NEM-associated
Nephrotic Syndrome, Steroid-Resistant Type 2
Neuroan Cereol Lipofuscinosis, CLN3-associated
Neuroan Cereol Lipofuscinosis, CLN4-associated
Neuroan Cereol Lipofuscinosis, CLN5-associated
Neuroan Cereol Lipofuscinosis, PPT1-associated
Neuroan Cereol Lipofuscinosis, TP1-associated
Neuermann-Pick Disease, Type A/B
Neuermann-Pick Disease, Type C
Niemann-Pick Disease
Ockolostenatae Albinism, Type 1
Ockolostenatae Albinism, Type 4
Oxalato-ornitho-dermal dystrophy/ Scott-Shultz-Passarge Syndrome
Pendred Syndrome
Phenylketonuria (PKU)
Primary Ciliary Dyskinesia, DNAS-associated
Primary Ciliary Dyskinesia, DNA1-associated
Primary Congenital Glaucoma
Primary Hypersusis, Type 1
Primary Hypersusis, Type 2
Progressive Pseudosarcomatous Dysplasia
Proline Deficiency
Propionic Acidemia, PCG-associated
Propionic Acidemia, PCGA-associated
Pseudoxanthoma Elasticum
Pycnodysostosis
Pyridoxine-Dependent Epilepsy
Pyruvate Carboxylase Deficiency
Reticilin Pigmentosus, RLP1-associated
Rettlin Pigmentosus, EYS-associated
Rhizomelic Chondrodysplasia Punctata, Type 1
Salta Disease
Sardhosa Disease
Sanfilippo, Type A
Sanfilippo, Type B
Sanfilippo, Type C
Segawa Syndrome
Severe Combined Immunodeficiency, Abahsane associated (SCID)
Shof/Braachied Chain Vacil-CoA Dehydratase (SCID) Deficiency
Sicklecell, Type 2
Sjogren-Larsson syndrome
Smith-Leim-Opi per Syndrome Starbarg Disease
Stuve-Wiedemann syndrome
(Schwarz-Jampel Syndrome Type 2)
Sulfate Transporer-Related Osteochondrodysplasia
Say-Sachc Syndrome
Tay-Sachs Disease
Tay-Sachs Syndrome (alligrow syndrome; Achalasia-Addisoninom-Ataxiaism)
Tyrosinemia
Usher Syndrome, Type IB
Usher Syndrome, Type IC
Usher Syndrome, Type ID
Usher Syndrome, Type IF
Usher Syndrome, Type II
Usher Syndrome, Type III
Very Long-Chain Aco-CoA Dehydratase (MCAD) Deficiency
Vitamin D-dependent Rickets, Type I
Walker-Warburg Syndrome
Weiner Syndrome
Wilson Disease
X-Linked Juvenile Retinoschisis
X-Linked Severe Combined Immunodeficiency
Zelweger Syndrome Spectrum

I understand that a sample of blood may be drawn by venipuncture, a procedure which carries a negligible risk.

I understand that no testing other than those marked on the reverse of this form will be performed on this sample. I understand that I have the option to obtain genetic counseling before signing the informed consent.

I understand that my sample will be stored at the laboratory for not more than 60 days, and then will be destroyed.

I understand that if I am a carrier of any of the above diseases, the probability of detecting my mutation by molecular methods can be dependent on my ethnicity and specific mutations analyzed. Information on the detection rate for each individual disease based on ethnicity is available upon request and will be included in the results report.

If I am positive for any of the molecular genetic tests (i.e. I am a carrier) genetic counseling will be recommended to me and additional recommendations for testing may be provided.

I understand that my result, when negative, only applies to mutations analyzed. There remains a chance that I may be a carrier of a mutation that was not part of this test, as these tests cannot detect all mutations/carriers. The residual risk that I may still be a carrier upon a negative result will be included in the results report.

In rare instances, it may be necessary to obtain another sample in order to determine a result. This will be at no additional cost.

I understand that the methods used by GenPath are highly accurate; however, rare errors can occur due to mislabeling of samples, bone marrow transplantation, blood transfusion, incorrect reporting of family history or relationships, or because some abnormalities are present in such a small fraction of cells that they may not be detectable (mosaicism).

I understand that results will be reported to the indicated healthcare provider and if noted, copied to the additional healthcare provider indicated on the front of this form.

I understand that results may only be disclosed to others by my written consent and/or if demanded by an order of a court of competent jurisdiction.

My signature constitutes my acknowledgement that I have had the opportunity to read supplemental materials regarding the diseases tested and have my questions answered by a healthcare provider or genetic counselor, and that I give my authorization and consent for the testing noted on the reverse of this form.
I hereby authorize the laboratory to furnish my designated insurance carrier the information on this form if necessary for reimbursement. I also authorize payment to the laboratory. I understand that I am responsible for any amount not paid by insurance for reasons including, but not limited to, non-covered and non-authorized services. I permit a copy of this authorization to be used in place of the original.

GenPath provides this consent form for physician use. It is the physician’s responsibility to obtain informed consent from the patient/guardian. If a signed consent form is not forwarded to the laboratory, it is believed that the physician has obtained consent and that the patient’s signature is on file in his/her medical records.

Note: Patient consent is required in the following states: Alaska, Arizona, Florida, Georgia, Massachusetts, Michigan, Nebraska, New Mexico, New York, South Carolina, South Dakota, and Vermont. Patient consent is suggested in all other states.

By my signature below I attest to the following:

1. I have read and understand the information provided on this form.
2. I understand that I may request genetic counseling PRIOR to signing this form and having blood drawn as well as after testing is completed.
3. I understand that I am agreeing to be tested for any or all of the genetic diseases listed above, that can be transmitted to my fetus.

Patient’s signature ___________________________ Date __________

Signature of Parent/Guardian if patient is a minor ___________________________

Print name of Parent/Guardian __________________________

PHYSICIAN AUTHORIZATION:

By my signature below, I indicate that I am the referring physician and that I have explained the purpose of the test(s) described above. The patient has been given the opportunity to ask questions and/or to seek genetic counseling. The patient has voluntarily decided to have the test(s) performed by BioReference Laboratories, Inc.

Physician’s signature ___________________________ Date __________

Physician name (printed) ___________________________ Account number ______

I hereby authorize the laboratory to furnish my designated insurance carrier the information on this form if necessary for reimbursement. I also authorize payment to the laboratory. I understand that I am responsible for any amount not paid by insurance for reasons including, but not limited to, non-covered and non-authorized services. I permit a copy of this authorization to be used in place of the original.

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