Inheritest®
CARRIER SCREEN

Integrated GENETICS
LabCorp Specialty Testing Group

Expanded panels using next generation sequencing now available

BROADENING YOUR PATIENT'S TESTING OPTIONS FOR GENETIC CARRIER SCREENING

Because Knowledge is a Powerful Tool.

The Inheritest® Carrier Screen provides relevant genetic screening for many inherited diseases found throughout the pan-ethnic US population.

Empower your patients with Inheritest® Carrier Screen

Given the pan-ethnic population in the United States and the difficulty in assigning a single ethnicity to many patients, it has become difficult for many patients to know their family origin. In addition, many patients may not be aware of their genetic family history or disease carrier status. Providing broader testing options to your patients can empower them with information about current and/or future pregnancies.

Utilizing next generation sequencing technology, the Inheritest Carrier Screen provides genetic risk information related to inherited diseases found throughout the US population:

- Analysis of over 7,400 mutations found in 142 different genes, associated with more than 114 different inherited diseases, providing you with carrier status information for these disease-causing mutations
- Succinct and informative summary reports, providing details about any positive result up front for easy review
- Choice to select the panel most appropriate for your patient

Expect more
from your Genetics Testing Partner.

Integrated Genetics offers carrier screening options for diseases found in the general population and for which ACOG and/or ACMG have provided guidelines regarding appropriate screening:[1-4]

- Full hemoglobinopathy screening, utilizing a CBC and hemoglobin electrophoresis assay.[4]
- Full gene sequencing for the partner when the patient has a positive result for any gene on the Inheritest panel.
- Preimplantation genetic diagnosis (PGD) to test the embryos prior to transfer into the uterus.
- Prenatal screening, including cell-free DNA for both common trisomies and sex aneuploidies, and for biochemical screening.
- Diagnostic testing (amniocentesis or CVS) to determine whether the fetus has inherited disease-causing mutations found in one or both parents. Integrated Genetics can perform prenatal testing on amniotic fluid and CVS for any of the diseases included in our Inheritest Carrier Screen. This type of fetal testing will be performed as targeted sequencing of only the identified mutations carried by the parents.

Comprehensive Genetic Counseling Services

You and your patients can choose from two genetic counseling solutions: a focused results discussion or a comprehensive genetic counseling discussion. All sessions are conducted by our trusted team of certified genetic counselors.

Focused Results Counseling through our Telegenetic Counseling to You Program

Our services are available remotely to patients with positive Inheritest® Carrier Screen results through the **Telegenetic Counseling to You** program. Available to patients through tele-video or telephone, the **Telegenetic Counseling to You** session includes:

- An explanation of the Inheritest Carrier Screen results
- A review of appropriate follow-up testing options, if applicable, based on the Inheritest result
- An Inheritest result summary report for the referring physician

Exclusive Comprehensive Counseling Session

Comprehensive genetic counseling is available remotely though our **Telegenetic Counseling to You** program when both the patient and her partner are identified as carriers for the same disease-causing gene. Available to patients though tele-video or telephone, the comprehensive genetic counseling session will include:

- Comprehensive genetic risk assessment
  - Evaluation of the reproductive and family history
  - Review of genetic testing results and medical records
  - Identification of risk factors that can affect the patient or the patient's offspring
- Discussion of the disorder in question, including etiology, heritability, and clinical implications
- Explanation of appropriate follow-up testing and procedure options to support informed decision making
- Summary report to the referring physician

Patients referred by their physicians for genetic counseling can call 855-GC-CALLS (855-422-2557) to schedule an appointment.

All genetic counseling reports are reviewed by a board-certified medical geneticist, providing you with added confidence.

Providing Choice for You and Your Patient

To provide the best service to you and your patients, Inheritest® Carrier Screen using next generation sequencing technology is now available in three distinct offerings:

Comprehensive Panel	142-gene panel that includes >114 diseases for the most complete screening
Ashkenazi Jewish Panel	39-gene panel that includes diseases specific to individuals of Ashkenazi Jewish decent
Society Guided Panel	12-gene panel that includes diseases included in ACMG and ACOG guidelines

The Inheritest NGS Advantage

Facts About Inheritest	Comprehensive Panel	Ashkenazi Jewish Panel	Society Guided Panel
Number of Diseases	>114	>39	>12
Number of Genes	142	39	12
Number of Mutations	>7,400	>2,300	>1,200
Cystic Fibrosis Mutations	609	609	609
Spinal Muscular Atrophy Copy # Mutations	Included	Included	Included
Fragile X Syndrome Expansion Mutations*	Included	Included	Included
Detection of Insertions/Deletions	Yes	Yes	Yes
Capacity to Add New Diseases	Yes	Yes	Yes
Comprehensive Genetic Counseling	Yes	Yes	Yes
Sample Types Accepted	Blood	Blood	Blood

*Females only

Additional Support Services

If your patient's result is positive, the following tests are available:

- Full gene sequencing for partners and non-blood relatives
- Mutation-specific sequencing for blood relatives

Experience You Can Trust

Trust in the experience of our board-certified molecular geneticists. These genetics experts reviewed the inherited diseases found within the pan-ethnic US population to deliver a broad carrier screening option relevant to your population.

Comprehensive Test Offerings

Society Guided Panel
Beta hemoglobinopathy, includes sickle cell disease, hemoglobins C, D, E, and O, and beta thalassemias
Bloom syndrome
Canavan disease
Cystic fibrosis
Familial dysautonomia
Fanconi anemia group C
Fragile X syndrome*
Gaucher disease
Mucolipidosis type IV
Niemann-Pick disease types A and B
Spinal muscular atrophy
Tay-Sachs disease

Ashkenazi Jewish Panel	
Abetalipoproteinemia	Glycogen storage disease type Ia
Alport syndrome, *COL4A3*-related	Joubert syndrome 2
Arthrogryposis, mental retardation, and seizures (AMRS)	Maple syrup urine disease type 1A
Bardet-Biedl syndrome, *BBS2*-related	Maple syrup urine disease type 1B
Bloom syndrome	Mucolipidosis type IV
Canavan disease	Multiple sulphatase deficiency
Carnitine palmitoyltransferase II deficiency	Nemaline myopathy, *NEB*-related
Congenital amegakaryocytic thrombocytopenia	Niemann-Pick disease types A and B
Congenital disorder of glycosylation type 1a	Phosphoglycerate dehydrogenase deficiency, *PHGDH*-related
Cystic fibrosis	Polycystic kidney disease, autosomal recessive
Dihydrolipoamide dehydrogenase deficiency	Retinitis pigmentosa 59
Dyskeratosis congenita, *RTEL1*-related	Smith-Lemli-Opitz syndrome
Ehlers-Danlos syndrome type VIIC	Spinal muscular atrophy
Familial dysautonomia	Tay-Sachs disease
Familial hyperinsulinism, *ABCC8*-related	Tyrosinemia type 1
Fanconi anemia group C	Usher syndrome type IF
Fragile X syndrome*	Usher syndrome type IIIA
Galactosemia, *GALT*-related	Walker-Warburg syndrome, *FKTN*-related
Gaucher disease	Wilson disease
	Zellweger spectrum disorder, *PEX2*-related

Inheritest®
CARRIER SCREEN

*Females only

Comprehensive Panel			
Abetalipoproteinemia	Familial dysautonomia	Metachromatic leukodystrophy	Primary hyperoxaluria type 1
Adenosine deaminase deficiency	Familial hyperinsulinism, *ABCC8*-related	Methylmalonic acidemia, *MMAA*-related	Primary hyperoxaluria type 2
Alpha-mannosidosis	Familial Mediterranean fever	Methylmalonic acidemia, *MMAB*-related	Propionic acidemia, *PCCA*-related
Alport syndrome, *COL4A3*-related	Fanconi anemia group C	Methylmalonic acidemia, *MUT*-related	Propionic acidemia, *PCCB*-related
Andermann syndrome	Fragile X syndrome*	Mitochondrial acetoacetyl-CoA thiolase deficiency	Pyruvate dehydrogenase deficiency, *PDHA1*-related
Argininosuccinic aciduria	Fucosidosis	Mucolipidosis type II and III, *GNPTAB*-related	Retinitis pigmentosa 59
Arthrogryposis, mental retardation, and seizures (AMRS)	Galactosemia, *GALT*-related	Mucolipidosis type IV	Rhizomelic chondrodysplasia punctata type 1
Aspartylglucosaminuria	Galactosialidosis	Mucopolysaccharidosis type I	Salla disease
Ataxia with vitamin E deficiency	Gaucher disease	Mucopolysaccharidosis type II	Sandhoff disease
Ataxia-telangiectasia	Glutaric acidemia type 1	Mucopolysaccharidosis type IIIA	Sialidosis
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	Glutathione synthetase deficiency	Mucopolysaccharidosis type IIIB	Sjogren-Larsson syndrome
Bardet-Biedl syndrome, *BBS1*-related	Glycine encephalopathy, *AMT*-related	Mucopolysaccharidosis type IIIC	Smith-Lemli-Opitz syndrome
Bardet-Biedl syndrome, *BBS2*-related	Glycine encephalopathy, *GLDC*-related	Mucopolysaccharidosis type IIID	Spinal muscular atrophy
Bardet-Biedl syndrome, *BBS10*-related	Glycogen storage disease type Ia	Mucopolysaccharidosis type IV A	Sulfate transporter-related osteochondrodysplasias, includes achondrogenesis type 1B, atelosteogenesis type 2, diastrophic dysplasia, and recessive multiple epiphyseal dysplasia
Beta hemoglobinopathy, includes sickle cell disease, hemoglobins C, D, E, and O, and beta thalassemias	Glycogen storage disease type Ib	Mucopolysaccharidosis type VI	
Beta-mannosidosis	Glycogen storage disease type III	Mucopolysaccharidosis type VII	
Bloom syndrome	GM1 gangliosidosis and mucopolysaccharidosis type IVB	Multiple sulphatase deficiency	
Canavan disease	GRACILE syndrome	Nemaline myopathy, *NEB*-related	Systemic primary carnitine deficiency
Carbamoyl phosphate synthetase I deficiency	Guanidinoacetate methyltransferase deficiency	Nephrotic syndrome, *NPHS1*-related	Tay-Sachs disease
Carnitine palmitoyltransferase II deficiency	Hereditary fructose intolerance	Nephrotic syndrome, *NPHS2*-related	Tyrosinemia type 1
Carnitine-acylcarnitine translocase deficiency	HMG-CoA lyase deficiency	Neuronal ceroid-lipofuscinosis, *CLN3*-related	Usher syndrome type IF
Cartilage-hair hypoplasia	Holocarboxylase synthetase deficiency	Neuronal ceroid-lipofuscinosis, *CLN5*-related	Usher syndrome type IIIA
Citrullinemia type I	Homocystinuria, *CBS*-related	Neuronal ceroid-lipofuscinosis, *CLN8*-related	Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
Cobalamin C disease	Hypophosphatasia, autosomal recessive	Neuronal ceroid-lipofuscinosis, *PPT1*-related	Walker-Warburg syndrome, *FKTN*-related
Cohen syndrome	Joubert syndrome 2	Neuronal ceroid-lipofuscinosis, *TPP1*-related	Wilson disease
Congenital amegakaryocytic thrombocytopenia	Junctional epidermolysis bullosa, *LAMA3*-related	Niemann-Pick disease types A and B	Xeroderma pigmentosum, *ERCC5*-related
Congenital disorder of glycosylation type 1a	Junctional epidermolysis bullosa, *LAMB3*-related	Niemann-Pick disease type C, *NPC1*-related	Xeroderma pigmentosum, *XPA*-related
Cystic fibrosis	Junctional epidermolysis bullosa, LAMC2-related	Niemann-Pick disease type C, *NPC2*-related	Xeroderma pigmentosum, *XPC*-related
Cystinosis	Krabbe disease	Nijmegen breakage syndrome	X-linked severe combined Immunodeficiency (SCID)
D-bifunctional protein deficiency	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)	Ornithine transcarbamylase deficiency	Zellweger spectrum disorder, *PEX1*-related
Dihydrolipoamide dehydrogenase deficiency	Leigh syndrome, autosomal recessive, includes French Canadian type	Phenylalanine hydroxylase deficiency, includes phenylketonuria (PKU)	Zellweger spectrum disorder, *PEX2*-related
Dihydropyrimidine dehydrogenase deficiency	Leigh syndrome, French Canadian type	Phosphoglycerate dehydrogenase deficiency, *PHGDH*-related	Zellweger spectrum disorder, *PEX6*-related
Dyskeratosis congenita, *RTEL1*-related	Maple syrup urine disease type 1A	Polycystic kidney disease, autosomal recessive	Zellweger spectrum disorder, *PEX10*-related
Ehlers-Danlos syndrome type VIIC	Maple syrup urine disease type 1B	Pompe disease	Zellweger spectrum disorder, *PEX12*-related
Ethylmalonic encephalopathy	Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)		Zellweger spectrum disorder, *PEX26*-related

Integrated Genetics – Your Complete Resource for Genetic Testing

Experience You Can Trust

- 25 years of genetic testing expertise
- 15 years of sequencing experience
- Every result reviewed and interpreted by board-certified clinical molecular geneticists

Broad Test Offerings

- Three carrier screening options
- Follow-up partner testing for identified carriers
- Prenatal or diagnostic testing options, such as amniocentesis and CVS samples, to determine whether the fetus has inherited the identified disease-causing mutations
- Preimplantation genetic diagnosis (PGD) for *in vitro* fertilization

Comprehensive Service Offerings

- Largest commercial genetic counseling network in the country
- Laboratory-based genetic counselors available for consultation and support
- Extensive network of patient service centers, allowing for easy and convenient sample collection
- Multiple online and electronic ordering and resulting capabilities, providing your office with easy solutions
- Knowledgeable client services team to help answer any questions about your testing

To learn more about our **Inheritest® Carrier Screen** offerings or other genetic tests, please visit **www.integratedgenetics.com.**

Test Code	Test Description	Specimen Requirements
451950	Comprehensive Panel (142 genes)	10 mL whole blood, ACD-A (yellow top) or EDTA (lavender top) tube
451920	Ashkenazi Jewish Panel (39 genes)	
451960	Society Guided Panel (12 genes)	

REFERENCES:

1. Edwards JG, Feldman G, Goldberg J, et al., Expanded carrier screening in reproductive medicine-points to consider: a joint statement of the American College of Medical Genetics and Genomics, American College of Obstetricians and Gynecologists, National Society of Genetic Counselors, Perinatal Quality Foundation, and Society for Maternal-Fetal Medicine. *Obstet Gynecol* 2015 Mar; 125(3):653-62. doi:10.1097/AOG.0000000000000666.
2. Prior PW, "Carrier screening for spinal muscular atrophy". ACMG Practice Guidelines. 10.11 (2008).
3. Carrier screening for fragile x syndrome. Committee Opinion No. 469. The American College of Obstetricians and Gynecologists. *Obstet Gynecol* 2010; 116.4:1008-1010.
4. Hemoglobinopathies in pregnancy. ACOG Practice Bulletin No. 78. The American College of Obstetricians and Gynecologists. *Obstet Gynecol* 2007 Jan; 109(1):229-37.

Inheritest® CARRIER SCREEN

Integrated GENETICS
LabCorp Specialty Testing Group

rep-983-v1-0116
L15010-0116-1