



GENETIC DISEASE CARRIER SCREENING

FOR INFERTILITY SPECIALISTS



A COMPREHENSIVE SERVICE FOR GENETIC DISEASE CARRIER SCREENING

LabCorp and Integrated Genetics offer a comprehensive service to make it easier for infertility specialists to provide patients and donors with genetic disease carrier screening. This cost-effective approach combines easy-to-order, ethnicity-based testing profiles with a convenient process for patients to complete their genetic assessments. LabCorp provides services in accordance with the genetic testing guidelines established by the American College of Obstetricians and Gynecologists (ACOG), the American Society for Reproductive Medicine (ASRM), and the American College of Molecular Genetics (ACMG).

Benefits of this comprehensive service include:

- Patient flyers with step-by-step instructions for completing the genetic disease carrier screening process.
- Ethnicity-based test groups that provide targeted screening for at-risk patients as defined by established guidelines for ACOG, ASRM, and ACMG.
- Access to LabCorp's large service network, including blood collection labs and genetic counselors.
- Multiple connectivity options to ensure efficient and accurate flow of test ordering and result reporting.
- Availability of LabCorp's wide managed care network.

Offering patient flyers can help patients and donors complete the genetic disease carrier screening process.

- Two versions of the patient flyer are available: one that provides information about the availability of telephone genetic counseling services and one that does not.
- Both versions of the patient flyer provide a brief summary of the genetic diseases included in the ethnicity-based test groups with basic questions to help patients understand which category may be best suited for them.

Ethnicity-based Carrier Screening

Ordering ethnicity-based test groups provides genetic testing options that are in accordance with clinical guidelines to optimize patient care.

- Targets specific at-risk groups based on established clinical guidelines.
- Reduces potential for patient anxiety about mutations that have mild clinical implications or that do not consistently result in severe disease.
- Facilitates efficient completion of testing to assist with moving patients and donors to the next step in the IVF process.

Using LabCorp's service network and connectivity options can help provide practice efficiencies, saving staff time on various tasks including blood collection, counseling patients, and managing patient records.

- LabCorp operates more than 1800 blood collection labs nationwide, with online options to find a lab and/or schedule appointments.
- Through Integrated Genetics, a member of the LabCorp Specialty Testing Group, genetic counselors are available to support physicians and patients. Our comprehensive genetic counseling services include a three-generation pedigree, personalized genetic risk assessment, and discussion of relevant test results. Please ask your LabCorp or Integrated Genetics sales representative for additional details.
- LabCorp has flexible connectivity options for test ordering and result delivery, including the ability to interface with many EMR systems.

LabCorp's broad participation in managed care and insurance plans can help reduce patient out-of-pocket expenses.

- Genetic carrier screening is covered by many insurance plans.*
- LabCorp is an in-network provider for many managed care plans; listings of insurance plans with which LabCorp files claims are available online at www.LabCorp.com.

All Ethnicities, Including Caucasian Descent

CFplus® (Cystic Fibrosis 97-mutation Profile);
Spinal Muscular Atrophy (SMA) Carrier Testing;
Fragile X Syndrome, PCR With Reflex to Southern Blot
OR

InheritestSM Carrier Screen (Expanded Testing)
• Analysis of gene mutations associated with more than 90 inherited disorders

African, Asian, Hispanic, Mediterranean, Mixed Descent

CFplus® (Cystic Fibrosis 97-mutation Profile);
Spinal Muscular Atrophy (SMA) Carrier Testing;
Fragile X Syndrome, PCR With Reflex to Southern Blot



Hemoglobinopathy Profile With Reflex to Alpha-thalassemia

Ashkenazi Jewish Descent*

CFplus® (Cystic Fibrosis 97-mutation Profile);
Spinal Muscular Atrophy (SMA) Carrier Testing;
Fragile X Syndrome, PCR With Reflex to Southern Blot



Jewish Descent ACOG (3) Add-on Profile
• Tay-Sachs Disease, Biochemical, Leukocytes
• Canavan Disease, DNA Analysis
• Familial Dysautonomia, DNA Analysis
OR

InheritestSM Select Carrier Screen
• 18 disorders indicated for patients of Ashkenazi Jewish descent

Expanded Testing Option

*For genetic testing ordered by infertility specialists and submitted to LabCorp with insurance billing, 91% of patients were covered by managed care plans. Coverage includes claims paid in full by insurance, as well as claims with patient copays and deductibles when applicable. Based on internal LabCorp billing data for test orders including cystic fibrosis, SMA, fragile X, Tay-Sachs disease, and/or Canavan disease. Analysis includes 20,559 test accessions generated from fertility clinics from August 2010 through October 2011.

*Additional testing may be indicated for patients of Ashkenazi Jewish descent interested in a broader panel of testing.

LabCorp Genetic Disease Carrier Screening Test Information

The American Society for Reproductive Medicine (ASRM) has published guidelines for prospective gamete donors that generally refer to the American College of Obstetricians and Gynecologists (ACOG) guidelines as shown below.

All Ethnicities (including Caucasian Descent)

Test Name	Test No.	Methodology	Clinical Guidelines	Disease Incidence Rate
CFplus® (Cystic Fibrosis 97-mutation Profile)	450020	DNA amplification, solution-phase multiplex allele-specific primer extension and hybridization	ACOG ¹ , ASRM ²	About 1 in 2500 in non-Hispanic white people (Caucasians); less common in other ethnic groups ¹
Spinal Muscular Atrophy (SMA) Carrier Testing	450010	Real-time polymerase chain reaction (PCR)	ACOG ³ , with patient history or by request	About 1 in 10,000 live births ³
Fragile X Syndrome, PCR With Reflex to Southern Blot	510234	PCR followed by agarose gel and capillary electrophoresis, and if required, southern blot hybridization	ACOG ⁴ , candidates include patient history or by request	About 1 in 3600 males; about 1 in 4000 to 1 in 6000 females ⁴

All Ethnicities, Expanded

Inheritest SM Carrier Screen	451381	Illumina® iSelect® Infinium® Beadchip Platform		
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Additional test groups available for ordering include:

African, Asian, Hispanic, Mediterranean, and Mixed Descent

Hemoglobinopathy Profile With Reflex to Alpha-Thalassemia (CBC Without Differential; Hemoglobinopathy Fractionation by HPLC; Ferritin)	451363	CBC = Automated cell counter; Hemoglobinopathy fractionation = HPLC and sodium hydrosulfite reduction; Ferritin = ECLIA, and if required, PCR and gel electrophoresis	ACOG ⁵	Sickle cell: About 1 in 300 African Americans; sickle cell disorders also found in people of Greek, Italian, Turkish, Arab, Southern Iranian, and Asian Indian descent. ⁵ Thalassemia: One form (beta-thalassemia) occurs in about 1 in 100,000 people worldwide. ⁶
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Jewish Descent (per ACOG guidelines)

Jewish Descent ACOG (3) Add-on Profile (Tay-Sachs Disease, Biochemical, Leukocytes; Canavan Disease; Familial Dysautonomia)	343649	Tay-Sachs, Biochemical, Leukocytes = determination of enzymatic activity using heat inactivation; Canavan Disease and Familial Dysautonomia = PCR, primer extension, flow-sorted bead array analysis	ACOG ⁷	In people of Eastern European Jewish descent: TSD: About 1 in 3000; also found in people of French-Canadian and Cajun descent ⁷ Canavan: About 1 in 6400 ⁷ FD: About 1 in 3600 ⁷
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All Ethnicities, Expanded

Inheritest SM Select Carrier Screen	451349	Illumina® iSelect® Infinium® Beadchip Platform		
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Patients with Recurrent Miscarriage

Chromosome Analysis, Blood (Constitutional)	052019	Lymphocyte culture with mitogen stimulation, colchicines arrest, methanol/acetic acid fixation, spread preparation, trypsin-Giemsa banding, chromosome analysis of 20 metaphases with preparation and analysis of multiple karyotypes	ACOG ⁹	
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References

- American College of Obstetricians and Gynecologists. Update on carrier screening for cystic fibrosis. ACOG Committee Opinion No 325. *Obstetrics & Gynecology*. 2005;106:1465-1468.
- Practice Committee of American Society for Reproductive Medicine; Practice Committee of Society for Assisted Reproductive Technology. 2008 Guidelines for gamete and embryo donation: a Practice Committee report. *Fertility and Sterility*. 2008;90(suppl 5):S30-S44.
- American College of Obstetricians and Gynecologists. Spinal muscular atrophy. ACOG Committee Opinion No 432. *Obstetrics & Gynecology*. 2009;113:1194-1196.
- American College of Obstetricians and Gynecologists. Carrier screening for fragile X syndrome. ACOG Committee Opinion No 469. *Obstetrics & Gynecology*. 2009;116:1008-1010.
- American College of Obstetricians and Gynecologists. Hemoglobinopathies in pregnancy. ACOG Practice Bulletin No 78. *Obstetrics & Gynecology*. 2007;109:229-237.
- Galanello R, Origa R. Beta-thalassemia. *Orphanet J Rare Dis*. 2010;5:1-15.
- American College of Obstetricians and Gynecologists. Preconception and prenatal carrier screening for genetic disease in individuals of Eastern European Jewish descent. ACOG Committee Opinion No 442. *Obstetrics & Gynecology*. 2009;114:950-953.
- Gross SJ, Pletcher BA, Monaghan KG. ACMG Practice Guidelines. Carrier screening in individuals of Ashkenazi Jewish descent. *Genetics in Medicine*. 2008;10:54-56.
- American College of Obstetricians and Gynecologists. Management of recurrent early pregnancy loss. ACOG Practice Bulletin No 24. *International Journal of Gynecology & Obstetrics*. 2002;78:179-190.



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LabCorp Specialty Testing Group

Please contact your local account representative for more information, or visit www.labcorp.com.