

Comprehensive carrier screening
providing valuable information to
you and your patients



ASPIRA is focused on providing solutions to support you and your patients

Ethnic-specific, pan-ethnic, and expanded carrier screening are acceptable strategies for prepregnancy and prenatal carrier screening. Each health care provider or practice should establish a standard approach **that is consistently offered to and discussed with each patient**, ideally before pregnancy.

- ACOG Committee Opinion Number 690 March 2017

Why Choose **ASPIRA GENETIX**?



Analyzes **more than 300 genes**, in which mutations may cause over 330 different recessive and X-linked disorders.



Provides **full coverage of each gene** via full gene sequencing, a specialized method to detect different types of mutations by using one technology, providing highly accurate results while remaining cost effective.



Can be customized: ASPIRA LABS® understands that not every couple has the same needs, and each patient's circumstances will differ. We have designed five different carrier screening panels to accommodate such needs. Talk with your Sales Representative about additional customization options.

What is **Carrier Screening**?



Carrier screening uses genetic testing to **identify carrier couples and individuals at risk** for passing genetic disorders on to their children. These genetic disorders can include physical disabilities, cognitive impairment, and other severe health problems.



Generally, everyone inherits two copies of each gene: one from their mother and one from their father. A carrier is an individual who has **one mutated copy and one normal copy of the same gene**. Carriers typically do not have signs or symptoms of a genetic disorder.



Traditionally, carrier screening has targeted couples of certain ethnic groups with a high risk of carrying specific genetic disorders. This approach presents difficulties for patients who are multiracial, adopted, or unsure of their ethnic backgrounds. To address this concern, **expanded carrier screening (ECS)** was developed to look for mutations that cause a wide variety of genetic disorders regardless of a patient's ethnicity.



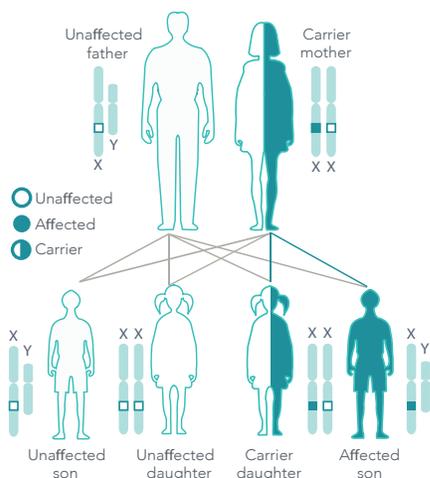
The American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG) have published **guidelines on expanded carrier screening** and its importance in reproductive care.

1 in every 31 Americans is a symptomless carrier of one copy of the mutated gene that causes Cystic Fibrosis

How Are **Genetic Disorders Inherited**?

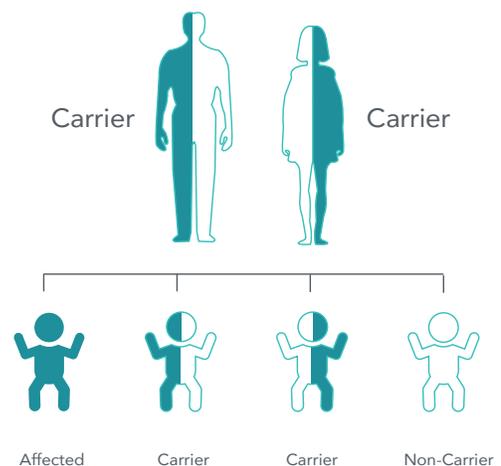
ASPIRA GenetiX Carrier Screening scans genes for mutations that cause autosomal recessive and X-linked recessive disorders.

X-Linked Recessive Inheritance, Carrier Mother (Example: Fragile X Syndrome)



If a woman carries a disease-causing variant on the X-chromosome, each of their children has a **1 in 2 (50%) chance of inheriting the variant**. Sons will be affected and daughters will be carriers.

Autosomal Recessive Conditions (Example: Cystic Fibrosis)



If both parents carry a disease-causing mutation in the same gene, each time they are pregnant there is a **1 in 4 (25%) chance to have an affected child**.

Carrier Screening Panels

Core 6

Focuses on **6 of the most common genetic disorders** seen within the general population. Carrier screening for these disorders has been recommended by the American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG). (Cystic Fibrosis, Spinal Muscular Atrophy, Fragile X Syndrome, Hemoglobinopathies (HBB and HBA), Sickle Cell Disease).

Ashkenazi Jewish Carrier Screening 44 Gene Panel

This panel focuses on **44 of the most impactful genes** commonly known to cause recessive genetic disorders as seen at high frequencies within the Ashkenazi Jewish population. In addition to spinal muscular atrophy (SMA) and fragile X syndrome, ACOG recommends that couples of Ashkenazi Jewish ancestry be offered carrier screening for Tay-Sachs disease, Canavan disease, Cystic Fibrosis, and Familial Dysautonomia. Furthermore, advocacy groups such as the Jewish Genetic Disease Consortium also recommend testing for other disorders such as mucopolidosis IV, Niemann-Pick disease type A, Fanconi anemia group C, Bloom syndrome, and Gaucher disease. Visit the Jewish Genetic Disease Consortium website to view the full list of genetic disorders recommended for carrier screening for Jewish couples.

Carrier Screening 37 Gene Panel

This pan-ethnic panel that analyzes **37 genes for pathogenic mutations** known to cause AR and X-linked prevalent disorders with an elevated carrier frequency across ethnicities.

Carrier Screening 155 Gene Panel

A smaller, expanded pan-ethnic panel appropriate for patients that want to screen for common and severe disorders. Genes included from the smaller panels as well as well-defined disorders that may have a severe impact on the quality of life, and disorders found on the newborn screen. All diseases listed have known reported pathogenic variants and represent a carrier frequency <1:500.

Carrier Screening 327 Gene Panel

This panel screens for more than **320 recessive and X-linked conditions** for patients of all ethnicities. Most of the diseases included in this panel have a significant impact on quality of life, and/or daily activities.

Note: Male patients will not be screened for X-linked conditions.

Testing Highlights

Cystic Fibrosis

Full gene sequencing is performed which is able to identify 1700 known disease-causing variants associated with Cystic Fibrosis.



Fragile X Syndrome

PCR amplification is used to detect the CGG repeat expansion of the FMR1 gene down to a single repeat. We can also detect AGG interruptions, which may decrease the size of the CGG repeat expansion when inherited from the mother.



Spinal Muscular Atrophy

ASPIRA LABS® can detect SMA "silent carriers" by testing for the SMN1 specific haplotype (g.27134T>G). "Silent Carriers" are patients with two copies of the SMN1 gene on one chromosome and zero copies on the other. Standard SMA screening counts the number of SMN1 copies, if a patient has two copies of the SMN1 gene, there is still a chance they are a carrier. ASPIRA LABS® enhanced testing improves detection rates and/or residual risk for SMA carrier screening.

Silent Carrier: Two SMN1 Copies (two on one chromosome)





Specimen & Shipping Requirements

ASPIRA LABS® accepts blood and saliva samples for Carrier Screening panels. Visit aspirawh.com or contact your sales representative to order the appropriate test kit.

Please see below for shipping requirements:



Blood

- + One 4-mL EDTA (lavender top)
- + This is our preferred specimen type for testing. Blood specimens can be sent at ambient temperature if express delivery is arranged such that arrival is within 72 hours of collection. Otherwise, samples will need to be refrigerated.



Saliva

- + DNA Genotek OGD-610 kits are preferred and can be provided upon request.
- + Saliva specimens can be sent at ambient temperature if express delivery is arranged such that arrival is within 72-96 hours of collection.

Detection Rates

A broad range of laboratory and bioinformatic tools are employed to ensure the **highest detection rate of any carrier screening panels** on the market. ASPIRA GenetiX analytical detection rate for all genes is >98%. ASPIRA LABS® uses an in-house algorithm to calculate residual risk for carrier couples.

Note: Residual risk is the chance that the patient being screened is a carrier even after a negative screening test result. Aspira Labs® calculates RR in combination with known population carrier frequency and detection of disease causing alleles by NGS.



Turnaround Time

Once the sample is received at the lab, results will be available in approximately **two weeks**.



Genetic Counseling Services

Pre- and Post-Testing Genetic Counseling is available for your patients.



Cost / Insurance

We accept **all commercial and private healthcare** insurance plans and also offer financial assistance.

Inherited Diseases Covered by ASPIRA GENETIX Carrier Screening

- + Cystic Fibrosis
- + Fragile X
- + Sickle Cell Anemia
- + Spinal Muscular Atrophy
- + Tay-Sachs Disease
- + Alpha & Beta Thalassemia
- + Canavan Disease
- + Bloom Syndrome
- + Wilson Disease
- + Duchenne muscular dystrophy
- + Over 320 hereditary conditions

ASPIRA LABS®

offers Pre and Post Test Genetic Counseling

Personalized Genetic expertise for your patients

PRE-TEST CONSULTATION

Talk with a genetic expert by phone or video once your test is ordered. We review personal and family health history, answer any questions you may have, and determine if you meet eligibility for test authorization.

RESULTS CONSULTATION

Review test results with a genetic counselor or medical geneticist. During this phone or video session, you are provided an opportunity to discuss the results of the test and their implications.

References:

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3. Edwards 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; 125:3.
4. Gross SJ et al. Carrier screening in individuals of Ashkenazi Jewish descent. ACMG Practice Guidelines. *Genet Med* 2008;10(1):64-56.
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