Beacon Carrier Screening
Information for Patients

What is carrier screening?

Carrier screening is a genetic test used to identify carriers of inherited conditions. Carriers have genetic changes within their DNA that increase their risk to have a child with a genetic disorder. Research has shown that most people are a carrier for at least one condition; however, most people are not aware of their carrier status because they do not have symptoms or a family history of disease.

Who should have carrier screening?

According to the American College of Obstetricians and Gynecologists (ACOG), expanded carrier screening should be offered to pregnant women and women considering pregnancy, regardless of ethnicity and family history. Expanded carrier screening tests for hundreds of genetic conditions and is considered an appropriate approach for carrier screening during pregnancy. If a woman is found to be a carrier for a specific condition, carrier screening for her partner should be considered.

Which genetic conditions are included in Beacon Carrier Screening tests?

Fulgent offers 5 Beacon Carrier Screening Test options, from a small 6-gene panel to a large 335-gene panel. All 5 panels screen for cystic fibrosis and hemoglobinopathies. Other conditions offered on our panels include:

- **Congenital adrenal hyperplasia** (CAH) - a group of disorders in which individuals may experience abnormal hormone production and/or salt-wasting, which can be life-threatening in infancy.
- **Duchenne and Becker muscular dystrophies** (DMD/BMD) - a condition which mainly affect males, causing progressive muscle weakness, delayed motor skills, and heart and respiratory problems. Few males with DMD survive beyond their early thirties, while those with BMD may live into their forties or beyond.
- **Fragile X syndrome -** the most common inherited cause of intellectual disability and autism spectrum disorder.
- **Spinal muscular atrophy** (SMA) - a condition characterized by muscle wasting. In severe cases, individuals may not live past the age of two.

At www.fulgentgenetics.com you can find detailed descriptions for each panel and condition. Please consult with your doctor to determine which test is most appropriate for you.

What causes recessive genetic conditions and how are they inherited?

Genes are the instructions in our DNA that tell our bodies how to grow and develop. For most genes we inherit two copies: one from our mother and one from our father. We all have millions of genetic changes; many are benign and do not cause disease or impact our health. However, some changes may be harmful and can cause disease in ourselves or when passed down to our children. These harmful changes, or "mutations", prevent the gene product from working properly.

An autosomal recessive (AR) conditions occur when a child inherits two mutated copies of a gene, one from each carrier parent. Because the parent carriers have just one abnormal copy, they usually do not display symptoms.

X-linked (XL) recessive conditions occur due to a mutated gene located on the X chromosome. Because males only have one X chromosome, a male will be affected if he inherits a mutated gene from his mother. Because females have two X chromosomes, carrier mothers and daughters usually do not display symptoms.

What will the Beacon Carrier Screening test tell me?

Through testing a sample of your blood or saliva, Beacon Carrier Screening tests look for harmful changes (mutations) in your genes that increase your risk to have a child affected with a recessive genetic condition. Your results will tell you if you are a carrier for the tested conditions (See Screening Panels). If you are identified as a carrier, your partner should consider screening to determine if you are both carriers for the same condition.

What are the benefits of carrier screening?

Carrier screening enables couples to become aware of their reproductive options, prenatal testing options, and the opportunity to prepare for a child with a genetic disorder. It is important to discuss these options with your doctor or clinical genetic counselor to determine which is most appropriate for you and your family.

If I am a carrier, what are the chances I could have an affected child?

If you and your partner are carriers for the same autosomal recessive condition, there is a 1 in 4 (25%) chance with each pregnancy, your child will be affected. For X-linked recessive conditions, only the mother has to be a carrier for her children to be at risk. If the mother is a carrier of an X-linked recessive condition, there is a 1 in 2 (50%) chance her son will be affected.

Can my partner and I receive screening at the same time?

Yes. Typically, women are offered carrier screening first because if they are not found to be a carrier, then follow up testing of their partner will not be necessary. However, it may save time if a couple receives screening concurrently. If you and your partner would like to receive screening at the same time, inform your doctor(s) so that they may coordinate testing for you.

How do I request a Beacon Carrier Screening test from my doctor?

Your doctor can request the proper test kit from Fulgent prior to your next appointment and order the test during your next visit.

When will my results become available?

Once your sample is received at the lab, your results will be available in approximately 2 weeks.

After testing, who can I speak with about my results?

A genetic counselor is a professional who provides information and support to patients as they make decisions about their genetic health. Once the carrier screening test is completed, Fulgent offers genetic counseling to those who have questions about their results. For detailed information about this service, please visit www.fulgentgenetics.com.

Is the Beacon Carrier Screening test covered by insurance?

Fulgent accepts all commercial and private healthcare insurance plans. Most insurance companies have specific criteria they use to determine coverage of genetic testing. Prior to testing, Fulgent's insurance specialists will contact you if there is an anticipated out-of-pocket cost that exceeds $100.

If I have genetic testing, can my employer or health insurance company discriminate against me?

No. The Genetic Information Nondiscrimination Act (GINA) was signed into law in 2008. It protects individuals from discrimination by an employer or a health insurance company based on genetic testing results and genetic information. Importantly, GINA does not offer protections for disability, long term care, or life insurance. It also does not apply to members of the U.S. military or employees of the Federal government, Indian Health Service, or Veterans Health Administration. For detailed information about GINA, please visit :
www.eeoc.gov/laws/types/genetic.cfm

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