



# Quick Facts



GeneAware screens for diseasecausing variants in over 400 genes by full gene sequencing

# Five Panel Options

## ACMG & ACOG

DISORDERS RECOMMENDED BY THE ACMG & ACOG

#### **ASHKENAZI JEWISH**

DISORDERS SPECIFIC FOR INDIVIDUALS OF ASHKENAZI JEWISH DESCENT

#### BASIC

THE MOST COMMONLY REQUESTED DISORDERS

### COMPLETE

A SELECTION OF COMMON DISORDERS

#### **EXPANDED**

THE MOST COMPREHENSIVE SCREENING

# Empower your patients with the knowledge to understand important family planning options.

GeneAware is a reproductive carrier screen that analyzes DNA from your patients' blood, saliva, or buccal swab sample to help them understand the risk of having a child with an inherited disease. Individuals and couples of reproductive age can have GeneAware testing to identify potential risks of having a child with a genetic condition.

Both the American Congress of Obstetrics and Gynecology (ACOG) and the American College of Medical Genetics and Genomics (ACMG) recommend all individuals who are pregnant or planning a pregnancy be offered carrier screening.<sup>1, 2, 3</sup>

## Goals of Carrier Screening

- Identify individuals at risk to have a child with a genetic disease
- Provide information to individuals to help understand reproductive risks and family planning options
- Identify resources to plan for a child born with extra needs
- Provide peace of mind for individuals that are not at an increased risk based on test results
- > 80% of couples who have babies with autosomal recessive disorders have no history of the disease.<sup>4</sup>
- $1.\,American\,College\,of\,Obstetricians\,and\,Gynecologists, Committee\,Opinion\,\#690, March\,2017.$
- 2. American College of Obstetricians and Gynecologists, Committee Opinion #691, March 2017.
- Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource from the American College of Medical Genetics and Genomics. Genetics in Medicine, 2021.
- Archibald, Alison Dalton et al. "Reproductive genetic carrier screening for cystic fibrosis, fragile X syndrome, and spinal muscular atrophy
  in Australia: outcomes of 12,000 tests." Genetics in medicine: official journal of the American College of Medical Genetics vol. 20,5 (2018):
  513-523. doi:10.1038/gim.2017.134



# Scientists can study a person's genes and identify inherited gene changes.

Inherited gene changes, known as pathogenic variants, are severe and cause genetic disease. Most diseases screened by the GeneAware test are inherited as autosomal recessive diseases, meaning both copies of the gene need to have a change to cause disease. Individuals who have one gene change are carriers of the disease and rarely have symptoms but are at risk to pass the gene change on to their children.

When a parent is a carrier, they can pass on the gene change to their child. If both parents are carriers of the same genetic disease, the chances of passing on the gene change to their children are:



1 IN 4 CHANCE THAT A CHILD WILL INHERIT THE GENE CHANGE, FROM BOTH MOM AND DAD, AND WILL BE AFFECTED BY THE DISEASE.



¥ 25%



1 IN 2 CHANCE THAT A CHILD WILL INHERIT A NORMAL GENE FROM ONE PARENT AND THE GENE CHANGE FROM THE OTHER PARENT AND WILL BE A CARRIER OF THE GENETIC DISEASE.



50%



1 IN 4 CHANCE THAT A CHILD WILL INHERIT THE NORMAL GENE FROM BOTH MOM AND DAD AND WILL NEITHER BE A CARRIER OF THE GENETIC CONDITION NOR BE AFFECTED BY THE DISEASE.



¥ 25%

If a woman is a carrier of an X-linked condition, each child is at increased risk for this condition (50% risk of inheriting the gene change).

# Understanding the Results



#### **Positive (Carrier)**

One or more disease-causing gene change(s) was detected in the genes included in GeneAware. The partner should be tested to determine carrier status for autosomal recessive diseases.



#### **Negative**

No known disease-causing gene change was detected in the genes included in GeneAware.

# **GeneAware Tests**

Female Complete

Male Complete

Female Ashkenazi Jewish

Male Ashkenazi Jewish

Female ACMG & ACOG

Male ACMG & ACOG

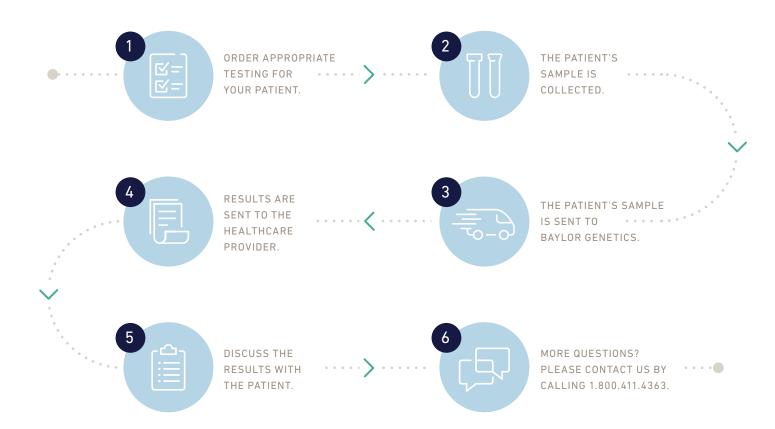
Female Basic

Male Basic

Female Expanded

Male Expanded

# How it Works





# 40+ YEARS OF INNOVATION



4 MILLION+ CLINICAL TESTS PERFORMED



1 MILLION+ FAMILIES HELPED



3 THOUSAND+ TESTS OFFERED



1 MISSION IMPROVE HEALTHCARE THROUGH GENETICS

Baylor Genetics pioneered the history of genetic testing. Now, we're leading the way in precision medicine.

Baylor Genetics is a joint venture of H.U. Group Holdings, Inc. and Baylor College of Medicine, including the #1 NIH funded Department of Molecular and Human Genetics. A pioneer of precision medicine for nearly 40 years, Baylor Genetics now offers a full spectrum of clinically relevant genetic testing, access to world-renowned experts, and the confidence to provide patients with the best care.

**BAYLOR GENETICS** 

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