BAYLOR GENETICS

Outpatient

Reproductive

GeneAware™

Reproductive Carrier Screening



Identify Reproductive Carrier Status to Inform Clinical Decision-Making

Empower Your Patients with the Knowledge to Understand Important Family Planning Options

GeneAware™ is a reproductive carrier screen that analyzes DNA from your patients' samples 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 College 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.[1, 2, 3]

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 expected to have an increased reproductive risk for the conditions tested

> 80% of couples who have children with autosomal recessive disorders have no history of the disease.[4]

References

1. American College of Obstetricians and Gynecologists, Committee Opinion #690, March 2017.
2. American College of Obstetricians and Gynecologists, Committee Opinion #691, March 2017.
3. 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.
4. 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



Quick Facts

600+ Genes

The GeneAware™ testing portfolio has the ability to screen disease causing variants in over 600 genes by full-gene sequencing.

Panel Options

ACMG & ACOG

Disorders recommended by ACMG and ACOG

Ashkenazi Jewish

Disorders specific to individuals of Ashkenazi Jewish descent

Basic

Our most commonly requested disorders

Complete

A selection of common disorders

Up to 157 genes

Expanded or Expanded Plus

Extensive panels offering our broadest carrier screening coverage across all conditions available

Up to 445 genes

Comprehensive or Comprehensive Plus

Our most comprehensive carrier screening panels

Up to 611 genes



Clinical 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 expect to 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 ACMG & ACOG: 24 genes
Male ACMG & ACOG: 22 genes

Female Ashkenazi Jewish: 39 genes
Male Ashkenazi Jewish: 37 genes

Female Basic: 6 genes
Male Basic: 4 genes

Female Complete: 157 genes
Male Complete: 147 genes

Female Expanded: 421 genes
Male Expanded: 381 genes

Female Expanded Plus: 445 genes
Male Expanded Plus: 400 genes

Female Comprehensive: 566 Genes
Male Comprehensive: 523 Genes

Female Comprehensive Plus: 611 genes
Male Comprehensive Plus: 559 genes

Ordering & Reporting Process

1. Order appropriate testing for your patient.
2. The patient's sample is collected.
3. The patient's sample is sent to Baylor Genetics.
4. Results are sent to the healthcare provider.
5. Discuss the results with the patient.
6. More questions? Please contact us by calling 1.800.411.4363.



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It is important to understand that genetic tests, even if negative, cannot rule out every variant. Genetic testing, while highly accurate, might not detect a variant present in the gene(s) tested. This can be due to limitations of the information available about the gene(s) being tested, or limitations of the testing technology. It is not possible to exclude risks for all genetic diseases for patients or their family members. It is possible that even if the test identifies the underlying genetic cause for a disease, this information may not help in predicting the progression of disease or change management or treatment of disease.

The tests described have been developed and their performance characteristics determined by the laboratory performing the testing, which is accredited by the College of American Pathologists (CAP) and certified under the Clinical Laboratory Improvement Amendments (CLIA). These tests are laboratory-developed tests (LDTs) and have not been cleared or approved by the U.S. Food and Drug Administration (FDA). Clinical testing is performed in accordance with CLIA requirements and CAP accreditation standards, helping to ensure high-quality, reliable laboratory practices.

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