PATIENT INFORMATION

Inheritest® Carrier Screen

Prepare today for your baby's tomorrow

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The Inheritest® Carrier Screen can help determine if you are at risk for having a baby with an inherited disorder. Early insight can help you better prepare for your baby.

What is carrier screening?

Carrier screening is a genetic test that can help you learn if you (and if indicated, your partner) are a carrier of variants in certain genes associated with disorders like cystic fibrosis or spinal muscular atrophy.

Anyone can carry a genetic disorder

You could be a carrier of a genetic disorder regardless of your ethnicity and even when there is no family history of the disorder. More than 80% of infants with cystic fibrosis (CF) are born to families with no prior family history.[1]

It is estimated that every person carries three to five genetic mutations in their DNA.[2] Being a carrier does not typically cause any related symptoms for that person, but it can mean that their children are at risk of inheriting the disorder.

Who should consider carrier screening?

According to the American College of Obstetricians and Gynecologists (ACOG), screening for CF and SMA should be offered to all women who are considering pregnancy or are currently pregnant.[3] Carrier screening for additional disorders is also available, based upon each family's needs and preferences.

When is the best time to have carrier screening?

Carrier screening can be done at any time but is most useful if it is done before the pregnancy or as early as possible during pregnancy. Early knowledge can empower your family planning choices and ensure you have early access to available treatments.

Choice in carrier screening

Inheritest is a helpful step in family planning, with panels that screen for 2 disorders to more than 500. Your doctor may help you decide which panel is right for you. Visit our website and watch our patient education videos to learn more.

A simple process with speedy results

1. A blood sample is collected from you (and/or your partner)

2. The sample is sent to our laboratory for analysis
3. In approximately two weeks, your results are returned to your doctor

What if the test shows you are a carrier of a genetic disorder?

If your test shows you're a carrier of an autosomal recessive disorder, the next step is for your partner to have carrier screening performed to assess a true risk for your child. Your doctor and our genetic counselors can help you better understand your test results and help you prepare for the future.

If your test result is negative, could you still be a carrier?

A negative result reduces, but does not completely eliminate, the risk of being a carrier of the genetic disorders included in a carrier screening panel. Screening cannot test for all mutations or all disorders.

The chance you'll have an affected child varies

- **Autosomal recessive inheritance**
  Many genetic disorders are inherited through "autosomal recessive inheritance". With autosomal recessive inheritance, both parents must carry a variant in the same gene in order to have an increased chance (25%) of having a child affected with a disorder, such as cystic fibrosis.

- **X-linked inheritance**
  With X-linked disorders such as fragile X syndrome, the most common cause of inherited intellectual disability,[4] only the mother needs to be a carrier in order to be at risk for having a child with the disorder.

  When the mother is a carrier of an X-linked condition, there is a 50% chance of passing the mutation on to a child. While these disorders are seen most commonly in boys, in some cases girls can also show symptoms.

"Prior testing would have helped us prepare emotionally for a baby with cystic fibrosis, and we could have avoided that first week of Hayden being really sick while we searched for a diagnosis"

– Elizabeth B., Hayden's mom

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References
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