Universal Carrier Screening

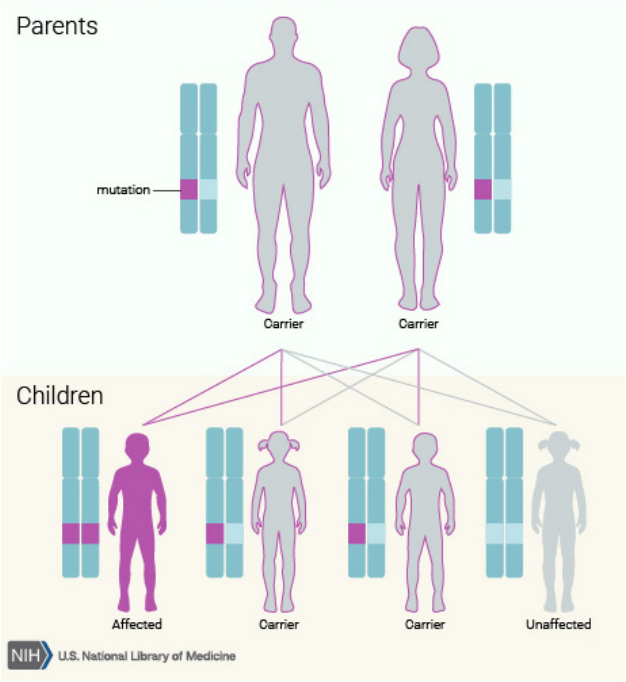
Most babies are born healthy. However, there is a chance that any parent might have a child with a genetic disease. Most babies born with a genetic disease do not have a family history of that disease.

You are being offered universal genetic carrier screening. This is a blood test to learn if you and your partner are at risk to have a child with one of many different genetic diseases. These diseases are passed down from healthy parents who are "carriers" of the genetic disease but do not show signs of the disease.

What is universal carrier screening?

Universal carrier screening looks at many instructions in our bodies, called "genes," for the most common ways these instructions might be misspelled. Misspellings in a gene can cause them not to work correctly. If a person has one working copy of a gene and one misspelled gene, that person will be a carrier of a genetic disease, but will not show signs of the disease. Some of the diseases affect only males because they have only one copy of each gene, while females have two copies.

Autosomal Recessive



In this example, two unaffected parents each carry one copy of a gene mutation for an autosomal recessive disorder. They have one affected child and three unaffected children, two of which carry one copy of the gene mutation.

What if both parents are carriers?

Parents who are carriers of the same genetic condition each have one non-working copy of a gene and one working copy. These parents have a 1 in 4 chance (25 percent) with each pregnancy to have a child with the condition. The child has the condition if they have inherited two non-working genes — one form each parent.

When both parents are carriers for the same condition the chances for the baby are:

- 25 percent chance that the baby is healthy, with two working copies of the gene
- 50 percent chance that the baby would be a carrier, with one working copy and one non-working copy of the gene
- 25 percent chance that the baby would have the genetic condition, with two non-working genes.

These couples have the option to elect testing during the pregnancy to learn whether the baby has the disease.

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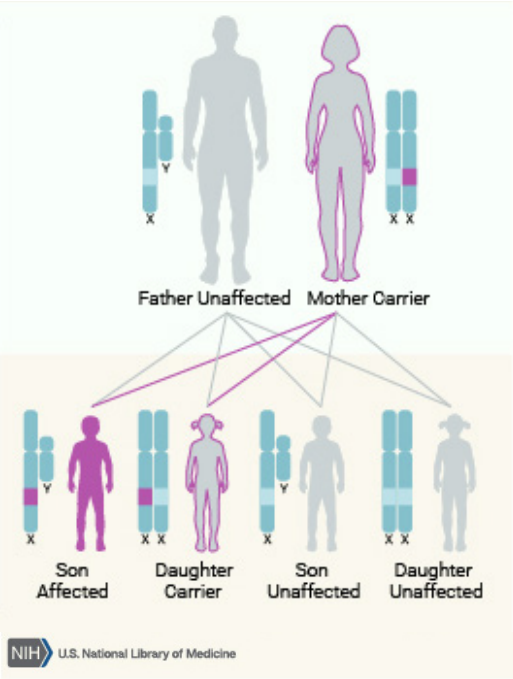
X-linked Recessive



X & Y chromosomes determine sex of a person.

- Males have both an X and a Y chromosome.
- Females have two X chromosomes.

If a gene is misspelled on the X chromosome, a male would not have a second working copy (because he only has one X). Therefore, the male would be affected. Females can be carriers, but not affected by the genetic disease, because they have a second, working copy of the gene.

What happens if my test results show that I am a carrier?

All patients will be notified by their provider about their results. Patients who are found to be a carrier will be referred to a genetic counselor to review the results. You and your partner would be offered other tests to find out more about risks for a pregnancy.

What do the results mean?

A "normal" result is good news. Normal indicates a very low chance you are a carrier of the genetic diseases that were tested. However, the test is not perfect and may not identify all carriers.

How is this genetic screening done?

Universal carrier screening is a blood test.

Why should I have this testing?

Results from a universal carrier screening test can give you information about your risks of having a baby with certain genetic diseases. Some couples find this information helpful for family planning.

Why should I **not** have this testing?

Not all couples value the information that is provided through universal carrier screening. Thus, the results can cause additional and unwanted anxiety in the pregnancy.

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Do my partner and I get the screening done at the same time?

In most cases, both parents do not need to be tested at the same time. It does not matter if the mom or the dad is tested first. Screening both mom and dad provides the most accurate risk assesst. Conditions which affect mostly males but females can be carriers will only be screened for in females.

Anyone who has a family history of genetic disease should be referred to a genetic counselor before testing. This will ensure the correct test is ordered.

Does insurance cover the cost of the test?

The cost depends on the type of health insurance and benefits you have. Ask your provider for more information.

Is this testing required?

No. The test is optional.

Where do I get more information about universal carrier screening?

Learn more about universal carrier screening by talking to your health care provider or genetic counselor.

www.VanderbiltHealth.com 615.322.5000