Carrier Screening

We want women and their families to have the healthiest outcome possible for their pregnancy. The good news is that most babies are born healthy. However, everyone has a small chance to have a baby who could be born with a problem. Carrier screening is one way to get information about the health of your baby. This testing is optional: it is your choice to have carrier screening or not. This information sheet may be helpful to you in making this choice.

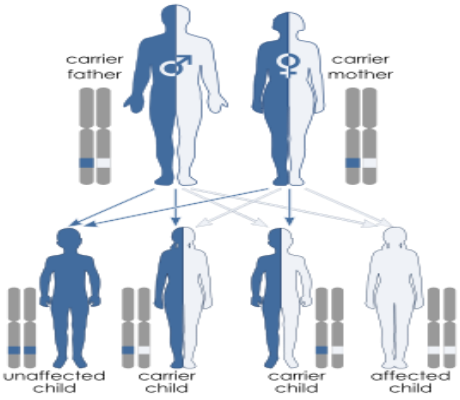
What is carrier screening?

Carrier screening can sometimes help families learn about genetic conditions that could affect their children's health. These genetic conditions happen because of changes in our DNA. DNA is what people inherit from their parents (in the egg and sperm) that tell the body how to grow and develop. Some changes in our DNA can cause health concerns, learning difficulties, and/or birth defects. Carrier screening should be considered even if you do not have a family history of any of these issues. However, less than 1% of families who have carrier screening will find they are at high risk to have a baby with a genetic condition.

There are two main ways these conditions can be inherited: autosomal recessive inheritance and X-linked inheritance (see below).

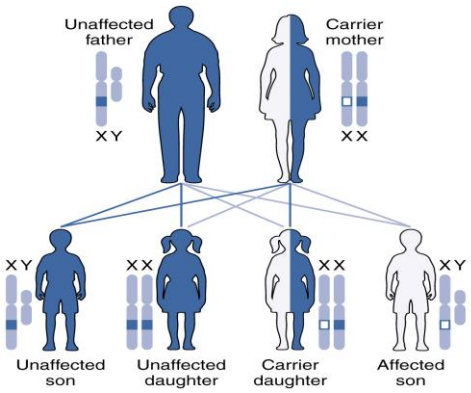
What is autosomal recessive inheritance?

- Both parents have to be carriers of the same condition
- Being a carrier typically does not cause a person to have health concerns
- When both parents are carriers, there is a 1 out of 4 (25%) chance any child could be born with the condition
- Both boys and girls can be affected



What is X-linked inheritance?

- Only the mother is a carrier of the condition
- 1 out of 2 of her sons (50%) would be born with the condition
- 1 out of 2 of her daughters (50%) would be a carrier of the condition
- Typically only boys are affected, although sometimes females are affected too



LEGACY HEALTH

Two important things to remember:

- We cannot screen for all birth defects or genetic conditions
- Carrier screening will not find all carriers of these conditions

Am I a carrier?

Most people who are carriers are healthy themselves and have a healthy family. Some experts believe that we are all carriers of a few of these conditions. One way to find out is through carrier screening.

Ask your doctor or a genetic counselor about your chance to be a carrier of a genetic condition(s).

Why should I consider carrier screening?

- You are worried about having a child with a genetic condition
- You are adopted and/or do not know much about your family history
- You want more information to prepare for the birth of your child

How accurate is carrier screening?

How well the test works depends on many factors including your race. Ask your doctor for more information.

- Genetic counseling is strongly recommended when you have a family history of a genetic condition or birth defect

Who wouldn't want carrier screening?

Carrier screening can:

- Cause you to worry
- Take 2-3 weeks for results
- Be expensive – you may have out-of-pocket costs
- Not be as helpful if the father of the baby is not able to be tested
- Test for some, but not all, genetic conditions and birth defects
- Not guarantee you will have a healthy baby

Some families may feel that the chance for their child to be born with a problem is small and not something they are worried about. Less than 1% of families who choose to have carrier screening learn that their pregnancy has an increased chance for a genetic condition.

What are other options?
The newborn screen is a screening test that babies have within days after birth. This test looks for some, but not all, of these same types of conditions. There are some people who may prefer to wait until after the baby is born to learn this type of information.

How is carrier screening done?
A blood sample is needed, possibly from both parents. Results are generally available in 2-3 weeks.

What type of results could I get from carrier screening?

- Your carrier screen shows you are a carrier or "positive" for one (or more) condition(s):
    - Genetic counseling and carrier screening for your partner are recommended
    - There are rare cases where finding out you are a carrier may impact your own health
    - Your carrier screen results may also be important for your family members

- Your carrier screen is negative for all conditions tested:
    - In most cases no more testing is needed
    - A negative carrier screen does not guarantee that your baby will be born healthy, it just makes it more likely
    - These tests can not find all carriers of these conditions

What are my options for carrier screening?
There are many options:

1. If you choose to have carrier screening, our recommended approach is a small panel which includes all ethnicity based recommendations
2. Some people may choose to screen for less or more conditions
3. Some people decline carrier screening

Ethnicity	Genetic Conditions Recommended for Carrier Screening
African American	Sickle cell anemia, cystic fibrosis, spinal muscular atrophy
Ashkenazi Jewish	Panel of conditions including Tay-Sachs, cystic fibrosis, and spinal muscular atrophy
Asian American	Blood disorders (hemoglobinopathies), cystic fibrosis, and spinal muscular atrophy
European Caucasian	Cystic fibrosis and spinal muscular atrophy
Hispanic American	Cystic fibrosis and spinal muscular atrophy

Experts currently recommend carrier screening for a handful of specific conditions. It is not known what benefit there is to screening for hundreds of conditions. However, the more conditions you screen for the more likely you will:

- Have a positive result
- Be a carrier for a mild condition that may not concern you
- Be a carrier for a condition that can cause problems some of the time but not all of the time – we won't be able to tell whether it is a risk for your pregnancy
- Be a carrier of a condition that does not cause problems in childhood, but typically only when a person becomes an adult

What happens if both of us are found to be carriers of a recessive condition or I am found to be a carrier of an X-linked condition?

There is a 25% (1 in 4) chance your pregnancy could have the condition. You can have prenatal diagnosis if you want to learn more about the health of the pregnancy. In most cases, additional testing is invasive. Please ask your doctor about the risks with prenatal diagnosis.

Will my insurance cover carrier screening?

Carrier screening may not be covered. If it is important to know before testing, please contact your doctor's office.

If you want carrier screening or have more questions, talk to your doctor or a genetic counselor.