



# A guide to your carrier testing options

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# Carrier screening options

## UNC Reproductive Genetics

Carrier screening is a blood test that can find out if you are at risk for having a child with certain genetic conditions. The newest guidelines from the American College of Obstetricians and Gynecologists (ACOG) recommend carrier screening be offered to all women either before or during pregnancy. This is a basic overview of your options. Please speak to your healthcare provider or genetic counselor for more information.

### Basic carrier screening

Cystic fibrosis	Cystic fibrosis (CF) is a lung and digestive disease. CF usually requires lifelong medical care and shortens lifespan. The risk of being a carrier is different among different races and ethnicities.
Spinal muscular atrophy	Spinal muscular atrophy (SMA) is a condition that results in progressive muscle weakness and paralysis. In severe cases, children may not survive past 2 years of age. The risk of being a carrier is approximately 2% regardless of race or ethnicity.
Sickle cell and other inherited anemias	Sickle cell disease and other inherited anemias are caused by problems with the red blood cells. People with these conditions may require hospitalizations and blood transfusions.

### Expanded carrier screening

Expanded carrier screening	Expanded carrier screening is available that can test for over 100 conditions at the same time. Approximately 1 in 5 people are found to be a carrier for <i>at least</i> one condition. Some test results may also indicate a risk for health problems in a carrier parent. This testing is available to anyone.
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### Does insurance cover this testing?

Insurance may cover the costs of these tests, but we recommend that you contact your insurance company to confirm your individual coverage.

### Who should consider expanded carrier screening?

Anyone can choose to have expanded carrier screening. However, a genetic counseling visit is recommended to discuss further carrier screening for these groups:

- If either you or your partner has Jewish ancestry
- If either you or your partner has Cajun/Creole or French Canadian ancestry
- If either you or your partner has a family history of intellectual disability or a family history of a specific genetic condition
- If you and your partner are related to each other by blood

### What if you are found to be a carrier?

For most conditions, both parents must be carriers for the same condition for a pregnancy to be **at risk** for being affected. However, for some conditions included in the expanded carrier screening, only the woman needs to be a carrier for a pregnancy to be **at risk**.

If either you or your partner is found to be a carrier for one of these genetic conditions, the next step would be an appointment with a genetic counselor to discuss what these results mean and what other testing is recommended.

### What if your test is “negative”?

A “negative” result means you are at **low risk** to be a carrier, but does not rule-out the possibility of being a carrier. There are no tests that can detect all genetic changes. Three to five percent of all newborns have some significant physical or mental disability, many of which are not included in any known testing.

### For more information:

If you would like to schedule an appointment with a reproductive genetic counselor to discuss these options, ask your healthcare provider to refer you to UNC Reproductive Genetics Services. Our schedulers can be reached at 984-974-6094 (UNC) or 919-784-6425 (REX).

