Disease	Carrier frequency	Characteristics
Spinal muscular atrophy (SMA)	~1 in 50	SMA results in progressive muscle weakness and paralysis. In severe cases, a person will not be able to sit up independently and their breathing and swallowing may also be impaired. In mild cases, symptoms begin in adulthood.
Fragile X syndrome	~1 in 250	This is the most common cause of inherited intellectual disability. Only the mother must be a carrier for a pregnancy to be at risk.
Cystic fibrosis	Caucasian: ~1 in 28 Ashk. Jewish: ~1 in 28 Hispanic: ~1 in 46 Af-Am: ~ 1in 66 Asian: ~1 in 87	A disorder of excess mucus production, primarily affecting the lungs, digestive and reproductive systems. Although there is some variability of clinical expression, most individuals require lifelong medical care and experience reduced life expectancy. Intelligence is not affected.
Tay-Sachs disease	Ashk Jewish: ~1 in 27 Fench Canadian: ~1 in 73 General pop.: ~1 in 300	An enzyme deficiency results in damage to nerves and deterioration of mental and physical abilities. Most affected individuals die in childhood.
Canavan disease	General pop.: rare Ashk. Jewish: `1 in 40	Destruction of the white matter that insulates nerve cells in the brain causes overall muscle weakness and developmental delay leading to severe intellectual disability. Most affected individuals die in childhood.
Familial dysautonomia	General pop.: rare Ashk. Jewish: ~1 in 31	Progressive nervous system disorder that causes vomiting, sweating, unstable blood pressure and temperature. May have some learning disabilities. Only 50% will survive to 30 years of age.
Sickle cell disease	Mediterranean: ~1 in 50 Hispanic: up to 1 in 10 Af-Am: ~1 in 12	Affects the ability of red blood cells to carry oxygen. Symptoms include anemia, repeated infections, shortness of breath, fatigue, jaundice, and bone pain starting in childhood.
Beta Thalassemia	Mediterranean: ~1 in 20 Hispanic: up to 1 in 13 Af Am: ~1 in 10 SE Asian: ~1 in 30	Affects the ability of red blood cells to carry oxygen. In the most severe form, thalassemia major, a child will show symptoms of severe anemia in the first year of life. Without frequent blood transfusions, the condition is life-threatening.

Reproductive Genetic Services

UNC Department of OB-GYN

Division of Maternal-Fetal Medicine

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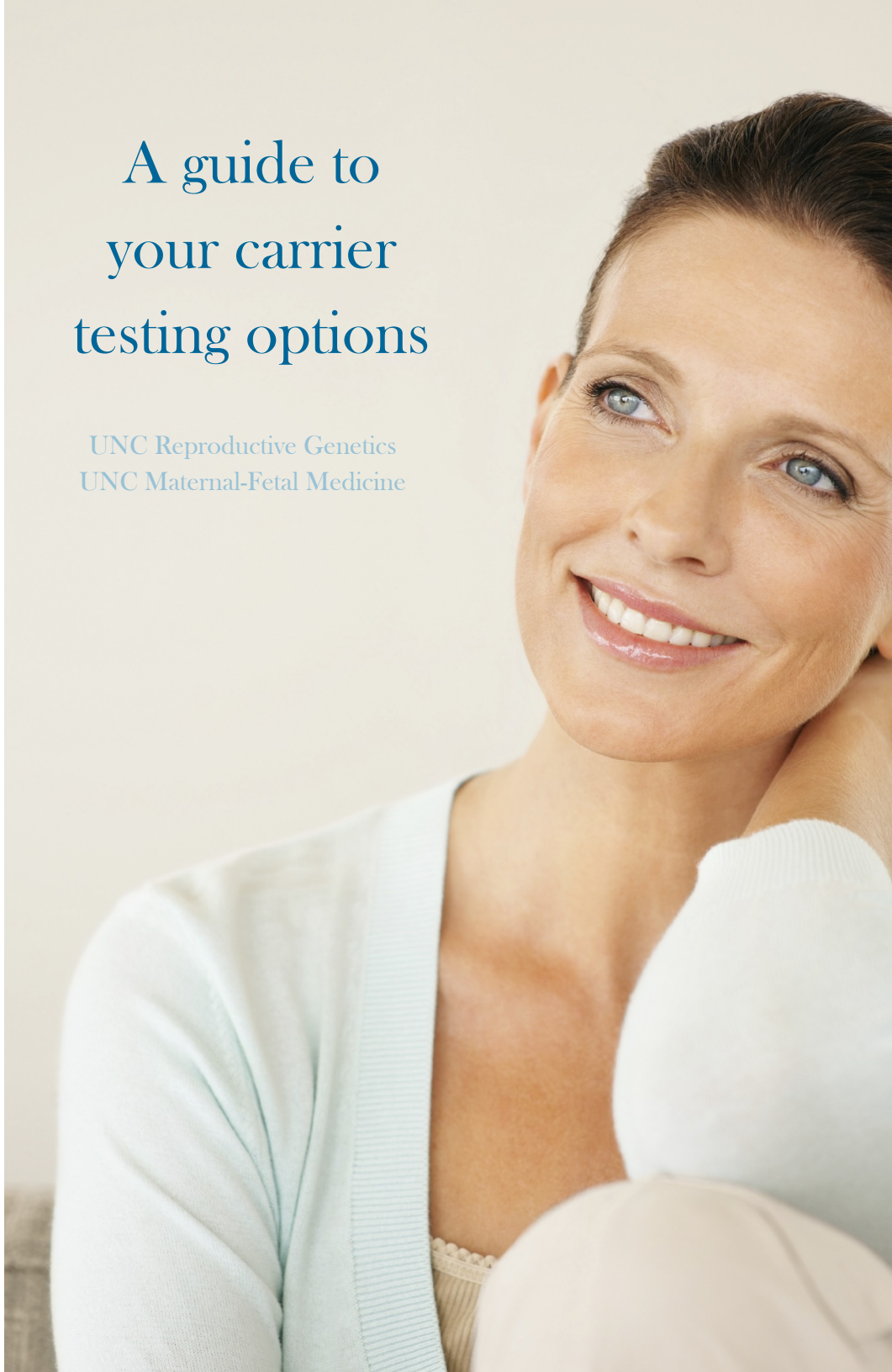
A guide to your carrier testing options

UNC Reproductive Genetics
UNC Maternal-Fetal Medicine



Carrier screening options

UNC Reproductive Genetics

Carrier screening can inform a couple if they are at risk for having a child with certain genetic conditions. There are multiple options available for genetic carrier screening. The current guidelines from the American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) use the patient's ethnicity to make recommendations regarding carrier screening. However, more expanded options are becoming increasingly available. This is intended to be a basic overview of your options. Please speak to your healthcare provider or genetic counselor for more information.

A couple identified at increased risk for a genetic condition can consider all of their reproductive options including preimplantation genetic diagnosis (PGD), use of donor egg or donor sperm, prenatal diagnosis, or no further action.

What if you test positive?

In general, both parents must be carriers for the same condition for a pregnancy to be **at risk** for being affected. Fragile X syndrome is the exception. Only the mother must be a carrier for Fragile X syndrome for the 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 the implications of these results and options for further testing.

What if you test negative?

A negative result reduces the likelihood of being a carrier, but does not eliminate the possibility. 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 undetectable though any known testing.

Basic carrier screening*

All ethnicities	All ethnicities can consider carrier screening for **cystic fibrosis**, **spinal muscular atrophy**, and **Fragile X syndrome**.
African ancestry	Carrier screening is recommended for **thalassemia** and **sickle cell disease**
Southeast Asian or Mediterranean ancestry	Carrier screening is recommended for **thalassemia**
French Canadian/ Cajun/Creole ancestry	Carrier screening is recommended for **Tay-Sachs disease**
Jewish ancestry	Carrier screening is recommended for: **cystic fibrosis, Tay-Sachs disease**, **Canavan disease**, and **familial dysautonomia**

*Recommended by ACOG or ACMG

Expanded carrier screening

Jewish ancestry only¥	A panel of 18 genetic conditions (including the four recommended by ACOG) seen more frequently in individuals of Eastern European (Ashkenazi) Jewish ancestry.
Broad-based screening, not ethnicity based¥	Screening for carrier status for more than ninety genetic conditions (including the 18 genetic conditions included in the expanded Jewish ancestry panel). Approximately 1 in 5 people will test positive for *at least* one condition.

¥See separate brochure for more detailed information regarding these options.
Screening for some of the conditions discussed above may need be ordered separately.

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.

For more information:

If you would like to schedule an appointment with a reproductive genetic counselor to discuss these options, ask your referring provider to refer you to UNC Reproductive Genetics Services. Our schedulers can be reached at 919-843-6094.