Carrier Screening in Pregnancy

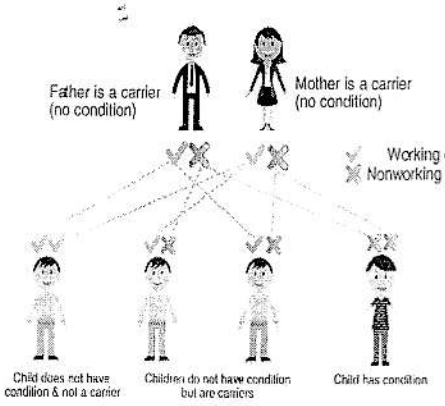
Office of Earle Oki, MD PC

WHAT IS CARRIER SCREENING?

Carrier screening is an optional genetic test that is available to all pregnant women and their partners. This test reads through our genes, which are recipes for how the body grows and develops. Most of the time, we have two copies of every gene, one from mom and one from dad. However, sometimes one copy of a gene has a typo, called a mutation, which means the body can no longer use that important recipe. Having only ONE working copy of a gene is called being a carrier.

WHAT IF I AM HEALTHY AND HAVE NO SIGNS OF ANY DISORDER?

Usually, carriers have absolutely no signs of the conditions they carry and are completely healthy people. Most of the time, there is no family history of the conditions we carry.



HOW CAN THIS TESTING BENEFIT MY PREGNANCY?

Carrier testing can give you more information about potential risks to the baby. If you elect to have carrier testing, we may discover that you are a carrier of a specific condition. Again, it is rare that this will indicate any health problems for you. If you are a carrier, we will likely recommend testing your partner as well. If both mom and dad are carriers of the SAME condition, there is usually a 25% chance to have a child with that disorder. If only one parent is a carrier, the risk of having a child with that condition is usually much less than 1%. For some conditions, however, there could be up to a 50% chance to have an affected child if only mom is a carrier.

Please indicate if you are interested in carrier screening:

☐ **YES**, I would like more information.

☐ **NO,** I would not like any additional information. I am not interested in carrier screening.

☐ I would like to check with my OB to see if this testing has already been done.

Print Name

Signature

Date

WHAT KIND OF CONDITIONS COULD I CARRY?

You have the option to test only for the 2 conditions below (and we may recommend others based on your ethnicity) or to test for over 150 conditions. Keep in mind that while testing more conditions will give you the most information, it will also be more likely to come back positive. For some parents, any positive result can cause added anxiety during pregnancy.

Cystic Fibrosis (CF)- CF mostly affects the lungs, but can also affect digestion. The effect on the lungs makes it difficult to breathe, so people with cystic fibrosis often pass away in their 20s or 30s. As many as 1 in 25 people care carriers of cystic fibrosis, depending on ethnicity.

Spinal muscular atrophy (SMA)- SMA causes muscle weakness, which makes it difficult for people to sit up straight or swallow. There are many different types of SMA; in the most severe form, children with this condition pass away before 3 years of age. About 1 in every 54 people carry this condition.

WHAT ARE THE NEXT STEPS IF I DISCOVER MY PREGNANCY IS AT RISK FOR ONE OF THESE CONDITIONS?

Unfortunately, most genetic conditions do not have a cure. Some people want to know if their child will have a condition because it could affect their pregnancy decisions and/or help them prepare to have a child with special needs. It is completely up to you to decide if you want this information.