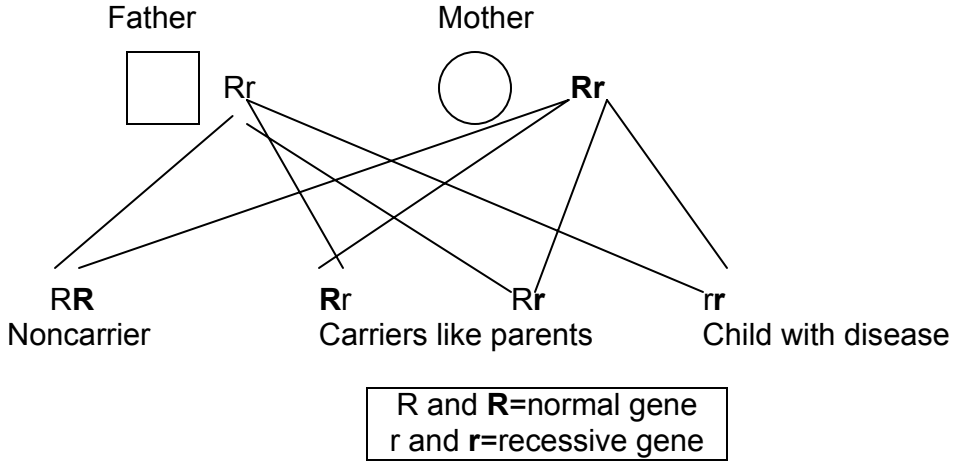
EMORY UNIVERSITY SCHOOL OF MEDICINE
Department of Human Genetics
Division of Medical Genetics
www.genetics.emory.edu

Carrier Screening for Recessive Diseases ©2001

Single gene Defects: To review, all of us have pairs of genes or 'instructions' which tell our bodies how to grow and develop. For each pair, we inherit one gene from our mother and one from our father. When both of the genes in a pair do not work properly, causing improper development, the gene pair is called "recessive". When disease or improper development is caused by having **two** copies of a gene that are not working properly, the disease is called recessive. (When disease or improper development is caused by having **one** copy of a gene that is not working properly, the disease is called "dominant".)

Autosomal recessive: In an autosomal recessive condition we must inherit two non-working genes, one from our mother and one from our father, in order to be affected. The parents are called 'carriers' of the condition. Carrier parents are normal and therefore not affected. However, when carrier parents have children, each child has a 1 in 4, or 25% chance to be affected.



All humans are thought to carry 10-20 genes which do not work properly. It is impossible to screen an individual for every recessive condition. It is estimated that humans have 30,000 genes and at the present, testing is available for a small percentage. Therefore, carrier screening for recessive genes is based on a person's ethnic background or the presence of a disease in their family history. For example, the gene which causes Tay Sachs disease is most common in the Ashkenazi Jewish population, while the gene for Sickle Cell Anemia is most common in the African American population.

The patients' ethnic background is:
Caucasian____ African-American____ Hispanic____ Asian_____ Native American____
Mediterranean_____Jewish_____French Canadian_____Other_____

The father of the pregnancy's ethnic background is:
Caucasian____ African-American____ Hispanic____ Asian_____ Native American____
Mediterranean_____Jewish_____French Canadian_____Other_____

Based on this information, the following carrier testing is recommended for the patient:
_____Hemoglobin electrophoresis to exclude a hemoglobinopathy gene
_____DNA testing for cystic fibrosis
_____Enzyme testing for Tay Sachs
_____DNA testing for Canavan disease
_____Other:_____

Based on this information, the following carrier testing is recommended for the father of the pregnancy:
_____Hemoglobin electrophoresis to exclude a hemoglobinopathy gene
_____DNA testing for cystic fibrosis
_____Enzyme testing for Tay Sachs
_____DNA testing for Canavan disease
_____Other:_____

At the time of your visit, you:
____elected screening for_____
____declined screening for_____
____were undecided_____
____were planning to coordinate screening through your doctor's office for:
_____