Prenatal Genetic Testing

The basic building blocks of heredity are called genes. Genes provide the code or set of directions for making us who we are. The genes we inherit from our parents determine everything about us--the color of our hair and eyes, how tall we will grow, and how our bodies will function. Every cell in our bodies contains this inherited genetic code.

All of our genes are grouped on structures called chromosomes. We inherit a set of 23 chromosomes from each of our parents. This gives us one gene from each parent--a pair--for every physical trait and each function our bodies perform. When one or both of the genes in the pair is defective, the trait or function governed by the genes may not be normal. Certain genetic diseases occur when a person inherits one or more genes that don't work properly. These faulty genes are called "mutations." If a piece of a chromosome is missing or if an extra chromosome is present, this causes problems too.

Many genetic conditions or chromosome abnormalities can be discovered before birth. Amniocentesis, chorionic villi sampling, or cordocentesis are tests that can be used to detect these problems. Every expectant mother does not need to undergo prenatal genetic testing. Your doctor may suggest it, however, if you or your partner are from certain ethnic backgrounds or have family members with certain genetic problems.

Genetic tests can diagnose or rule out many conditions, including these:

- **Down Syndrome** results from extra genes along the 21st chromosome. Ninety-five percent of all cases of Down syndrome occur because there are three copies of the 21st chromosome. For this reason Down syndrome is also called "trisomy 21." After age 35, women are at increased risk for having a child with Down syndrome. Down syndrome causes mental retardation and various physical problems.

- **Gaucher Disease** is the result of defects in the genes that produce an enzyme called glucocerebrosidase. More than 80 genetic mutations can cause Gaucher Disease, but about 90% of the time the cause is one of four changes. Those descended from Norrbottniam Swedes or Ashkenazi Jews are most likely to carry genes for this rare disorder. Gaucher Disease can affect the spleen, liver, bones, lungs, kidneys, and nervous system.
- **Cystic Fibrosis** (CF) is the most common serious genetic disease in the U.S. One in 23 Americans carries one of the 700 gene mutations that can cause CF. Over 90% of all cases, however, are caused by a single mutation called DeltaF508. The gene mutations that cause CF interfere with the production of a protein called CFTR. Cystic fibrosis occurs in every ethnic population in the world. In the United States, those descended from Northern or Central Europeans or from Ashkenazi Jews are most likely carry genes that cause CF. CF is a life-shortening disease that affects breathing, digestion, and reproduction.
- **Sickle Cell Anemia** causes the body to manufacture red blood cells with a defective type of hemoglobin called hemoglobin S (for sickle). This disease is most prevalent among those whose ancestors lived in Africa, the Caribbean Islands, South and Central America, India, and Mediterranean countries such as Turkey, Greece, and Italy. Two million Americans carry the genetic trait for sickle cell anemia. One in 12 African Americans is a carrier. Sickle cell anemia causes pain, stroke, anemia, slow growth, and other problems.

Genetic testing can also detect Tay-Sachs disease, Caravan disease, Turner syndrome, congenital adrenal hyperplasia (CAH), Duchenne muscular dystrophy, and numerous other conditions.

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