What are genetic diseases?
Genetic diseases are caused by genes that do not work properly. Genes are the basic units of inheritance that determine many characteristics of a developing baby, such as eye color, height, and blood type. Genes are also very important in determining a baby's health. Genes that do not work correctly are responsible for genetic diseases such as thalassemia, sickle cell diseases, cystic fibrosis, Tay-Sachs disease, Canavan disease, and familial dysautonomia (see table on other side of pamphlet).

All of our genes come in pairs; we get one from each parent. In order to have one of the above disorders, a baby must receive two non-working genes for the same disease, one from each parent. A person who has only one non-working gene for one of these diseases is called a carrier.

Who carries genetic diseases?
We all have some genes that do not work properly, so everyone is a carrier for some genetic diseases. However, some genetic diseases, such as the ones mentioned here, occur more often in certain ethnic and racial groups than in the general population. You don't have to have a family member with one of these diseases to be a carrier. Usually, there is no family history, so most people do not know whether or not they are carriers.

What does it mean to be a carrier?
Being a carrier does not cause any health problems. However, if both parents are carriers for the same genetic disease, there is a chance their child will have the disease. A child will have one of these diseases only if he or she receives the same non-working gene from each carrier parent.

What does a negative result mean?
A negative test result means you are most likely not a carrier for that particular disease. Therefore, no further testing is needed. These tests are very accurate but they will not identify all carriers.

What does a positive result mean?
A positive test result means that you are a carrier for that particular disease. If your test result is positive, a genetic counselor will contact you and the father of the baby will be offered testing. If his test is negative, there is only a very small chance that your baby will have the disease for which you are a carrier, and no more testing will be done.

If the father of the baby is also found to be a carrier, a genetic counselor will meet with you to discuss the option of testing the baby before it is born (prenatal diagnosis). Prenatal diagnosis can tell you whether or not the baby will have the disease. Some people who learn that their baby will have one of these diseases may choose not to continue their pregnancy. Others may use this information to help them prepare for taking care of a child with special medical needs. If you choose not to have prenatal testing, the baby can be tested shortly after it is born. As of July 2007, newborn screening for cystic fibrosis, sickle cell, and thalassemia disease is performed on all babies born in California. A genetic counselor can help you make these decisions and give you ongoing support.
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| Thalassemia             | - Asian                                                    | Varies from 1 in 4 to < 1 in 100 depending on disease type and ethnicity | • Alpha thalassemia disease can range from mild anemia to a severe anemia that causes babies to die during the pregnancy. Mothers of an affected baby may develop serious health problems during the pregnancy.  
  • Beta thalassemia disease causes severe anemia and poor growth beginning in infancy/early childhood. Lifelong transfusions are often required and there may be a shortened life span.  
  • Hemoglobin E/beta thalassemia disease is a variable condition that causes moderate to severe anemia.  
  • Generally cannot be cured. | Blood test that detects most but not all carriers. |
|                         | - Southeast Asian                                          |                           |                                                                                                              |                   |
|                         | - Mediterranean                                            |                           |                                                                                                              |                   |
|                         | - African American/Black                                   |                           |                                                                                                              |                   |
| Sickle Cell Diseases    | - African American/Black                                   | About 1 in 12             | • Blood disorders beginning in infancy/early childhood that cause anemia, bone pain, and frequent serious infections. There may be a shortened life span.  
  • Treatment may include frequent hospital stays, medications, blood transfusions.  
  • Severity varies. Some people live without serious illness.  
  • Generally cannot be cured. | Blood test that detects most but not all carriers. |
|                         |                                                            |                           |                                                                                                              |                   |
| Cystic Fibrosis         | - Caucasian/White, (Non-Hispanic)                          | About 1 in 25             | • Disease of the lungs and digestive system, developing in early childhood.  
  • Thick mucus clogs the lungs, causing difficulty breathing and frequent lung infections. Lung disease worsens over time. Problems with digesting food, resulting in poor weight gain.  
  • Treatment may include daily chest physical therapy, medications, frequent hospital stays.  
  • Severity varies. Some people live without serious illness. Average life span is about 35 years.  
  • Currently no cure. | Blood test that detects about 90% of carriers in the Caucasian (non-Hispanic) population. |
|                         |                                                            |                           |                                                                                                              |                   |
| Tay-Sachs Disease       | - Jewish (Ashkenazi/Eastern European)                      | About 1 in 30             | • Disease of the brain and nerves, developing in infancy.  
  • Causes muscle weakness, mental retardation, and blindness. Greatly worsens over time.  
  • Death occurs by about 3 to 5 years of age.  
  • No treatment or cure. | Blood test that detects about 95% of carriers in the Ashkenazi Jewish population. |
|                         |                                                            |                           |                                                                                                              |                   |
| Canavan Disease         | - Jewish (Ashkenazi/Eastern European)                      | About 1 in 40             | • Disease of the brain and nerves, developing in infancy.  
  • Causes muscle weakness, mental retardation, and seizures. Worsens over time.  
  • Death usually occurs by 10 years of age.  
  • No treatment or cure. | Blood test that detects about 97% of carriers in the Ashkenazi Jewish population. |
|                         |                                                            |                           |                                                                                                              |                   |
| Familial Dysautonomia   | - Jewish (Ashkenazi/Eastern European)                      | About 1 in 30             | • Disease of the nervous system beginning in infancy.  
  • Can lead to pain insensitivity, unstable blood pressure and/or temperature, problems with speech and movement, no tears when crying, and difficulty swallowing.  
  • Average life span is 30 years.  
  • Currently no cure. | Blood test that detects over 99% of carriers in the Ashkenazi Jewish population. |