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## Screening for Genetic Diseases in Ashkenazi Jews

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The incidence of inherited diseases varies greatly among different racial and ethnic groups. For example, the gene for sickle cell disease is more common among African-Americans and the gene for Tay-Sachs is more prevalent in the Ashkenazi Jewish population. Both of these diseases are inherited in a recessive manner, meaning they are caused by inheriting one abnormal gene from each parent. A person who inherits only one abnormal gene is called a “carrier” but is otherwise normal. However, when 2 carriers of an abnormal gene for the same disease have a child together, there is a 1 in 4 chance with each pregnancy that the child will develop the disease.

The American College of Obstetricians and Gynecologists recommends that Jewish patients (or the partners of Jewish patients) be offered carrier testing for a group of disorders that are carried with a greater frequency in this group than in other population groups. Examples of these disorders are:

Tay-Sachs disease—approximately 1 in 25 Ashkenazi Jews is a carrier. This is a lethal condition resulting in progressive nervous system deterioration, leading to death in childhood. Testing detects greater than 95% of Tay-Sachs carriers among Ashkenazi Jews.

Canavan disease—approximately 1 in 36 Ashkenazi Jews is a carrier. This is also a lethal progressive deterioration of the nervous system leading to childhood death. Testing detects greater than 95% of Canavan carriers among Ashkenazi Jews.

Cystic fibrosis (CF)—is not specifically a “Jewish related” disease, but 1 in 25 Caucasians of all ethnic origins is a carrier. CF results in chronic progressive lung disease and often digestive disease, leading to a shortened life span. Testing detects greater than 95% of CF carriers in Ashkenazi Jews.

Bloom’s disease, Fanconi anemia, Niemann-Pick disorder, mucopolysaccharidosis, and spinal muscular atrophy are some of the other diseases for which testing is available in the Ashkenazi Jewish heritage panel.

Please sign below to acknowledge that you have read and understood the above information and to indicate your choice.

Neither the baby’s father nor I are Jewish\_\_\_\_\_

We (or one of us) are Jewish and desire testing\_\_\_\_\_

We (or one of us) are Jewish and decline testing\_\_\_\_\_