INFORMATION ABOUT ETHNICITY-BASED PRENATAL GENETIC SCREENING

The following lab slip offers pregnant women the opportunity to choose testing for several genetic diseases that are more common in certain ethnic groups. The lab slip is also used to order routine tests required for your prenatal care. Please read the following information about genetic diseases your baby could be at higher risk to have because of ethnicity. You will learn what tests you are eligible to have, along with basic information about the diseases, so you can decide whether you want testing. A table with more detailed information about each condition is on the back of this page. Genetic testing for these conditions is done by an optional blood test that you choose by filling out the attached lab slip.

A baby can only have the genetic conditions listed below if both parents are “carriers” of the same condition. Carriers of these genetic conditions do not have any symptoms and usually do not have relatives with the disease. If both parents are found to be carriers, their baby will have a 25% (1 in 4) chance to have the condition. If you are found to be a carrier, the father of your baby will be offered testing. If he is also a carrier, you will be offered a prenatal diagnostic procedure (CVS or amniocentesis) to determine if the baby inherited the condition. If prenatal testing determines that your baby has the condition, you will have the option to continue or terminate the pregnancy. Couples can also decline prenatal testing and have the baby tested at birth. Genetic testing cannot predict the exact symptoms an individual baby will have, and there is no cure for the conditions described below.

If you have Southeast Asian ancestry (Cambodian, Thai, Laotian, Vietnamese, Hmong):
You are eligible to have screening for hemoglobin E/beta thalassemia disease. Southeast Asians have a 1/60 (~2%) to 1/4 (25%) chance to be a hemoglobin E carrier.

If you or the father of the baby have any African American ancestry:
You are eligible to have screening for sickle cell disease. African Americans have about a 1/12 (8%) chance to be a sickle cell disease carrier.

If you or the father of the baby have any Caucasian (White, not Hispanic) ancestry:
You are eligible to have screening for cystic fibrosis. Caucasians have about a 1/25 (4%) chance to be a cystic fibrosis carrier.

If both you and the father of the baby have any Ashkenazi Jewish ancestry:
You are eligible to have screening for Tay-Sachs disease, Canavan disease, and familial dysautonomia. Ashkenazi Jews have about a 1/30 (3%) chance to be a carrier for Tay-Sachs disease or familial dysautonomia, and a 1/40 (2.5%) chance to be a Canavan disease carrier. Ashkenazi Jews are considered Caucasian, so cystic fibrosis screening can also be selected.

If the ethnicity of you and your partner is not listed above, you are at lower risk to be a carrier for the above conditions and testing is not as effective, so you will not be screened. If you have a family history of any of the above conditions, contact your Genetics Department as soon as possible so you can have appropriate testing (phone numbers on back).

All pregnant women are screened for certain types of thalassemia. This test is not optional. Thalassemia screening will occasionally identify sickle cell and hemoglobin E carriers.
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| Thalassemia | • 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. | Detects most but not all carriers |
| Sickle Cell Diseases | • Blood disorders beginning in infancy/early childhood that cause anemia, bone pain, and frequent serious infections. Life span may be shortened.  
• Treatment may include frequent hospital stays, medications, and blood transfusions.  
• Severity varies. Some live without serious illness.  
• Generally cannot be cured. | Detects most but not all carriers |
| Cystic Fibrosis | • Disease of the lungs and digestive system beginning in infancy.  
• Thick mucus clogs the lungs, causing difficulty breathing and frequent lung infections. Lung disease worsens over time. Problems digesting food results in poor weight gain.  
• Average life span is 35 years.  
• Treatment may include daily chest physical therapy, medications, and frequent hospital stays.  
• Severity varies. Some live without serious illness.  
• Currently no cure. | Detects about 90% of Caucasian (non–Hispanic) carriers, and about 98% of Ashkenazi Jewish carriers |
| Tay-Sachs Disease | • Disease of the brain and nerves beginning 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. | Detects about 95% of Ashkenazi Jewish carriers |
| Canavan Disease | • Disease of the brain and nerves beginning in infancy.  
• Causes muscle weakness, mental retardation, and seizures. Greatly worsens over time.  
• Death usually occurs by 10 years of age.  
• No treatment or cure. | Detects about 97% of Ashkenazi Jewish carriers |
| Familial Dysautonomia | • 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. | Detects over 99% of Ashkenazi Jewish carriers |

For additional information, contact your local Genetics Department:

Fresno: 559-324-5330  
San Francisco: 415-833-2998  
Oakland: 510-752-6298  
San Jose: 408-972-3300  
Sacramento: 916-614-4075
ETHNICITY-BASED GENETIC SCREENING: Please answer the following questions and check all the boxes that apply. More than one ethnic background may apply to you and/or the father of the baby, so be sure to mark all the boxes that apply (for example: someone who is Ashkenazi Jewish is also Caucasian, so both boxes should be marked). Please note: If you have had genetic testing for any of the conditions listed below, you do not need to be retested.

1. Do you have any Southeast Asian background?
   - [ ] NO
   - [ ] YES Myself [ ]
     - If yes, do you want testing for hemoglobinE-beta thalassemia disease? [ ] Yes [ ]
     - No

2. Do you OR the father of the baby have any Black/African American background?
   - [ ] NO
   - [ ] YES Myself [ ] Father of baby [ ]
     - If yes, do you want testing for sickle cell diseases? [ ] Yes [ ]
     - No

3. Do you OR the father of the baby have any Caucasian (White, not Hispanic) background?
   - [ ] NO
   - [ ] YES Myself [ ] Father of baby [ ]
     - If yes, do you want testing for cystic fibrosis? [ ] Yes [ ]
     - No

4. Do you AND the father of the baby have any Eastern European Jewish (Ashkenazi) background?
   - [ ] NO
   - [ ] YES Myself and the father of baby [ ]
     - If yes, do you want testing for Tay-Sachs disease? [ ] Yes [ ]
     - No
     - If yes, do you want testing for Canavan disease? [ ] Yes [ ]
     - No
     - If yes, do you want testing for familial dysautonomia? [ ] Yes [ ]
     - No

Please write the countries your ancestors are from (before coming to the United States) and your ethnic background:

You: ___________________________

Father of the baby: ___________________________