Reproductive Endocrinology and Infertility

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Genetic Disease Screening Information Sheet

If you or your partner are interested in Preconception Genetic Disease screening what are the steps to proceeding forward?

Carrier screening involves a blood test from one or both parents. All testing is optional and you can choose which tests are right for you based on your ethnic background and family history. The American College of Obstetrics and Gynecology (ACOG) and the American College of Medical Genetics (ACMG) make recommendations for testing based on the higher incidence of disease in the general population. Please watch the video produced by Dr. Schnorr reviewing the topic of Preconception Genetic Screening found at www.schnorrmd.com before proceeding on with this document.

Please note that treatment cannot proceed until you have made a decision regarding genetic screening, signed the informed consent form, and, if you decide to undergo genetic testing, discuss the test results with your physician.

If you would like to proceed forward with testing there are several steps you should take:

1. Consider seeing a Genetics Counselor to better understand the genetic risks you may have based on your ethnic backgrounds and your family histories. The cost of the genetics consult is approximately $100 and there are several options for genetic counselors:
    a. Medical University of South Carolina, 843-792-5300
    b. University of South Carolina, 803-779-4928
    c. LabCorp, 888-816-3862
2. If you want testing based on your ethnic background and family history without seeing a genetic counselor:
    a. Review recommended ethnic background testing information below, Table 1
    b. Review the family history questionnaire below, Table 2
    c. Review the test pricing information (Table 3) and call the financial coordinators if you have questions about your insurance coverage.
    d. Sign the consent form and select the testing you would like. The consent form can be found at www.schnorrmd.com.
    e. Fax, mail or hand deliver the signed consent form to your nurse coordinator. The fax phone number is 843-606-3937.

f. Feel free to call if you have questions

3. If you want to decline testing simply sign the consent form and fax, mail or hand deliver it to your nurse coordinator. The fax phone number is 843-606-3937. The consent form can be found at www.schnorrmd.com.

Ethnic Background Risk Factors

Table 1 is a list of the recommended carrier testing based upon ethnic background. These recommendations come from the American College of Obstetrics and Gynecology (ACOG) and the American College of Medical Genetics (ACMG). Costs for each of the test are found in Table 3.

Table 1: ACOG 12 and ACMG* 4 Screening Recommendations

Caucasian
- Cystic Fibrosis*
- Spinal Muscular Atrophy

Ashkenazi Jewish
- Cystic Fibrosis*
- Spinal Muscular Atrophy
- Tay-Sachs Disease
- Familia Dysautonomia
- Canavan Disease*
- Gaucher Disease
- Fanconi Anemia Type C
- Mucolipidosis IV
- Neimann Pick Type A
- Bloom Disease

Asian
- Cystic Fibrosis*
- Spinal Muscular Atrophy
- Beta Thalassemia

Hispanic
- Cystic Fibrosis*
- Spinal Muscular Atrophy
- Thalassemia

Mediterranean
- Cystic Fibrosis*
- Spinal Muscular Atrophy
- Thalassemia

African
- Cystic Fibrosis*
- Spinal Muscular Atrophy
- Sickle Cell Disease

Personal and Family History Risk Factors

Table 2 is a list of some of the common risk factors you or your partner may have for genetic disorders. It's important for you and your partner to evaluate this list to see if you answer yes to any of them. If you have answered yes you may benefit from speaking with a genetic counselor.

Table 2: Risk Factors for Genetic Disorders

- Will you be 35 years or older when your baby is due?
- Will the baby's father be 50 years or older when your baby is due?
- If you or your partner are of Mediterranean, Asian, or African descent, do either of you or anyone in your families have thalassemia (an inherited disorder that causes anemia)?
- Is there a family history of neural tube defects?
- Have you or the baby's father ever had a child with a neural tube defect?
- Is there a family history of congenital heart defects?
- Is there a family history of Down syndrome?
- Have you or the baby's father ever had a child with Down syndrome?
- If you or the baby's father are of eastern European Jewish, French Canadian, or Cajun descent, is there a family history of Tay—Sachs disease?
- If you or the baby's father are of eastern European Jewish descent, is there a family history of Canavan disease?
- If you or the baby's father are African American, is there a family history of sickle cell anemia or sickle cell trait?
- Is there a family history of hemophilia?
- Is there a family history of muscular dystrophy?
- Is there a family history of cystic fibrosis?
- Is there a family history of Huntington's disease?
- Does anyone in your family or the family of the baby's father have cystic fibrosis?
- Is anyone in your or the baby's father's family mentally retarded?
- If so, was that person tested for fragile X syndrome?
- Do you, the baby's father, anyone in your families, or any of your children have any other genetic diseases, chromosomal disorders, or birth defects?
- Do you have a metabolic disorder such as type 1 or type 2 diabetes or phenylketonuria?
- Do you have a history of pregnancy issues (miscarriages or a stillborn baby)?

** ACOG Education Pamphlet AP094*

Description of Diseases

Cystic Fibrosis primarily involves the respiratory, digestive and reproductive systems. Symptoms include pneumonia, diarrhea, poor growth and infertility. Some people are only mildly affected but others with severe disease may die in childhood. CF does not affect intelligence. If BOTH parents are carriers, there is a 1 in 4 (25%) chance to have a child with CF. Carrier frequency varies by ethnicity but averages 1 in 30 in the U.S.

Spinal Muscular Atrophy destroys the nerve cells that affect voluntary movement. Infants with SMA have problems breathing, swallowing, controlling their head and neck and have difficulty crawling or walking. The most common cause of SMA affects infants and can cause death between 2-4 years of age. Less commonly the disease starts later in adulthood. SMA does not affect intelligence. If BOTH parents are carriers, there is a 1 in 4 (25%) chance to have a child with SMA. Carrier frequency varies by ethnicity but ranges from 1 in 35 to 1 in 117 in the U.S.

Fragile X Syndrome is a genetic condition involving changes in the X chromosome. It can affect boys and girls. It is the most common form of mental retardation in boys. It can cause developmental delay, mental retardation, autism and hyperactivity. If a mother is a carrier, there is a 50% chance of having a child with fragile X syndrome.

Sickle Cell Anemia is characterized primarily by chronic anemia and periodic episodes of pain. It is caused by a defect in the HBB gene. The hemoglobin molecule is responsible for carrying oxygen from the lungs to the body organs and tissues and carbon dioxide back to the lungs. This abnormal or "sickle" shaped hemoglobin cannot squeeze through small vessels and therefore often stack up and cause blockages that then deprive the body of its needed oxygen carrying blood. This is a lifelong disease and regular health maintenance is critical.

There are many other inheritable diseases. Some are more prevalent in Hispanic, Jewish, Asian or Mediterranean backgrounds. If you or your partner has any of these backgrounds you may need to consider additional testing that has not been specifically listed above. Please, feel free to ask your doctor if you think additional testing should be considered based on your own ethnicity.

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Table 3: ACOG 12 and ACMG* 4 Screening Recommendations with LabCorp Codes

Below is a list of the recommended preconception genetic screening tests based solely upon ethnic background assuming a normal family history. Please note these are estimated prices for the respective tests. You may have a financial benefit based upon your insurance carrier to having your testing done through LabCorp rather than Coastal Fertility Specialists. We are happy to provide a lab order if that is in your best interest. If you have questions please feel free to contact one of our financial coordinators at 843-883-5800.

Recommended Test	Estimated CFS Price	LabCorp Test Code
Caucasian		
Cystic Fibrosis*	$280	450020
Spinal Muscular Atrophy	$240	450010
Ashkenazi Jewish		
Cystic Fibrosis*	$280	450020
Spinal Muscular Atrophy	$240	450010
Tay-Sachs Disease	$100	511246
Familia Dysautonomia	$100	511352
Canavan Disease*	$130	511147
Gaucher Disease	$150	511048
Fanconi Anemia Type C	$100	511212
Mucolipidosis IV	$100	511386
Neimann Pick Type A	$100	511329
Bloom Disease	$100	512145
Asian		
Cystic Fibrosis*	$280	450020
Spinal Muscular Atrophy	$240	450010
Beta Thalassemia	$17	121679
Hispanic		
Cystic Fibrosis*	$280	450020
Spinal Muscular Atrophy	$240	450010
Thalassemia	$17	121679
Mediterranean		
Cystic Fibrosis*	$280	450020
Spinal Muscular Atrophy	$240	450010
Thalassemia	$17	121679
Sickle Cell		
Cystic Fibrosis*	$280	450020
Spinal Muscular Atrophy	$240	450010
Sickle Cell	$17	121679