OB/GYN Associates of Miami

Thomas J. Dimino, M.D.
M. Victoria Lopez-Beecham,M.D
Luis P. Leyva Jr., M.D.
Ilya S. Johnson, M.D.
Manal S. Antoun, M.D.
Ana R. Hernandez, M.D.
Terry Reisman, M.D.
Paola Bordoni, M.D.
Dana Homer, M.D.

Informed Consent/Decline for SMA Carrier Screening

1. **What is Spinal muscular atrophy?** SMA is a hereditary disease that destroys the nerves responsible for controlling voluntary muscle movement, but does not affect intelligence. Muscles that control breathing, swallowing, head and neck control, walking and crawling are the most severely affected. SMA is a variable disease in terms of when the symptoms begin. Most often it shows up before a baby is two years old, but in some individuals the symptoms start before birth and in others not until after age 30. There is currently no cure or treatment for SMA.

2. **How is SMA inherited?** If both parents are carriers of an abnormal SMA gene, there is a chance that each parent will pass the abnormal gene on to their child. An individual with two abnormal SMA genes will be affected with the disease. With each pregnancy there's a :
    - 25% (1 out of 4) chance that the child will inherit two normal SMA genes, and will be neither a carrier nor affected with the disease
    - 50% (2 out of 4) chance that the child will inherit one normal and one abnormal SMA gene, and will be a carrier of SMA (but not affected)
    - 25% (1 out of 4) chance that the child will inherit two abnormal SMA genes – one from each parent – and will be affected with the disease.

3. **What is the purpose of SMA carrier screening?** The purpose of SMA carrier screening is to see if o couple is at increased risk for giving birth to a child who will be affected with SMA. The test is done on a sample of blood. If results show that a couple is at high risk, prenatal testing (chorionic villi sampling [CVS] or amniocentesis) during pregnancy or preimplantation testing before pregnancy can be done to see whether or not the baby has inherited two abnormal SMA genes. Neither carrier screening nor prenatal diagnostic testing can tell what type of SMA the child could have.
4. **If my test is negative, could I still be a carrier?** A negative result significantly lowers, but does not completely eliminate the risk of being a carrier of SMA. Carrie screening does not detect les common abnormalities (mutations) that cause SMA.

5. **Is there a cost for this test?** This is a routine clinical laboratory test and the results may aid in your diagnosis; thus, you or your health insurer will be billed for this procedure.

    - The decision to consent to or to refuse SMA testing is entirely mine.

- No test(s) will be performed and reported on my sample other than those authorized by my doctor; and any unused portion of my original sample will be destroyed within 2 months of receipt of the sample by the lab.

Patient Assentation of Informed Consent:

My signature below indicates that I have received information about this test, **Spinal muscular atrophy (SMA),** and that I have read and understood the material in this document. I have been given a full opportunity to ask questions that I may have about the testing procedure and related issues. I agree to undergo this testing.

_____

Patient Signature D.O.B. Date

_____

Signature of Parent/Guardian if Patient is a minor Print name of Parent/Guardian

Decline of SMA Testing

My signature below indicates that I have received information about this test, Spinal muscular atrophy (SMA), and that I have read and understood the material in this document but **I would like to decline** this test being done.

_____

Patient Signature D.O.B. Date

_____

Signature or Parent/Guardian if Patient is minor Print name of Parent/Guardian

For the Physician:

As the referring physician, I understand the benefits and limitations of this study and have requested that the above-named patient be tested. I attest to the fact that I have provided the patient with the information contained above and fully answered any questions. I believe that the patient understands the information and is voluntarily signing this informed consent.

_____

Signature of Physician/Health Care Professional Print Name of Physician/Health Care Professional