Carrier Testing for Spinal Muscular Atrophy (SMA)

FOR COUPLES OF PLAIN DESCENT

1 in 2,800 Mennonite and 1 in 250 Hutterite babies are born with SMA.

New treatments are available for SMA and are most effective when given early in life, ideally before children become symptomatic.

Although 1 out of every 25 Mennonites is a carrier of SMA, most are unaware of their carrier status. SMA is found in all Plain communities, yet many carriers do **not** have a family history of the condition.

All couples of Plain descent are encouraged to undergo carrier testing for SMA.

What is SMA?

Spinal Muscular Atrophy (SMA) is a progressive genetic condition affecting motor nerve cells (neurons) in the spinal cord and brainstem. These nerves control the muscles that enable us to move, breathe, and swallow. Over time in the disease course, these muscles become weak as a result of the loss of these nerve cells. SMA is a recessive disorder such that both parents must carry the genetic variant of SMA in order to have an affected child together. When both parents are carriers, each of their children has a 1 in 4 (25%) chance of being affected. SMA is the most common genetic cause of infant death worldwide and is found in all people around the world, including members of the Plain communities.

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COST OF TESTING

Carrier testing is available through the SMA Prevention Readiness Program free of charge.

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TREATMENT OPTIONS

SPINRAZA® (Nusinersen) is an approved medicine specific for SMA. Gene replacement therapy is currently in clinical trials for SMA.

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TIME TO WASTE

Children with SMA have the best outcomes when treatment is started soon after birth.

Clinic *for* Special Children

We Need Your Help!

THE SMA PREVENTION READINESS PROGRAM

Step 1 | Patient Education

- ✓ Share the educational SMA brochure with your patient.
- ✓ Explain that there are two targeted treatments, and that these are most effective when started early in life – ideally **before** a child shows any symptoms.
- ✓ Express that carrier testing is important because it allows babies at risk for SMA to be tested and receive treatment immediately, if affected. If both parents are not carriers, they don't have to worry about this condition for their children.
- ✓ Explain that SMA is **not** currently identified by the newborn screen in most states, but carriers can be identified by genetic testing.

Step 2 | Carrier Testing

- ✓ SMA carrier testing should be done prior to or during pregnancy.
- ✓ Through the SMA Prevention Readiness Program at the Clinic for Special Children, carrier testing is provided **free of charge.**
- ✓ A simple blood test, shipped to the Clinic, is all that is required using the standard molecular testing requisition and consent. Please contact the Clinic for specific instructions.

Step 3 | Personalized Counseling

- ✓ A staff member from the Clinic will follow-up with you and your patient to discuss the results and what they mean for their family.
- ✓ If a carrier couple is detected we will discuss treatment options with them, so they can think about their choices should they have an affected baby. We can test cord blood at birth to know if the baby is affected. If the baby is diagnosed with SMA, parents can begin treatment immediately, if they choose.

Our Goal

All couples of Plain descent know their SMA carrier status so all babies born with SMA have the best chance at a healthy life.

Who to Contact?

If you have any questions about SMA carrier testing, please contact a member of the SMA Prevention Readiness Team at the Clinic below.

Clinic *for* Special Children

SMA Prevention Readiness Team

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