



Testing that makes a difference

Athena Diagnostics: Shedding light on a devastating disease

Deborah and Chris Heine did everything they could to ensure that their second child would be born as healthy as their first. In addition to living a wholesome lifestyle, the California couple even got tested for genetic diseases like cystic fibrosis, Tay-Sachs disease and fragile-X condition before they conceived. In January 2004, the Heines joyfully welcomed Claire, a beautiful baby girl, into their lives.

Their joy was short-lived. When Claire was just nine weeks old, the couple received the heart-breaking news that their daughter had spinal muscular atrophy (SMA).

SMA is a genetic disease in which muscles involved in many vital functions – breathing, eating, movement – become progressively weaker and ultimately waste away and die. There are three forms of SMA: Types I, II and III. As the Heines so painfully learned, about seven in 10 babies with SMA have the most severe form, Type I, which usually leads to death before the age of two. Claire, who had Type I SMA, died when she was just nine months old.

Carrier testing

What makes the Heines' experience especially tragic is that it might have been avoided. For years, Athena Diagnostics, part of Thermo Fisher Scientific's Specialty Diagnostics Group, has been among a number of companies to offer a blood test that tells couples with a high degree of certainty whether they have a mutated (faulty or missing) gene called *SMN1*. This gene contains the code for a protein (*see sidebar*) that is critical to the survival and healthy functioning of specialized nerve cells, called motor neurons, that play a vital role in voluntary muscle movement.

If the test shows that both prospective parents have a mutated *SMN1* gene, they are SMA carriers and, while not affected themselves, have a one in four chance of having a child with SMA with each pregnancy. Because one in every 40 to 60 adults worldwide carries a mutated *SMN1* gene, about one in 10,000 infants is born each year with SMA, making it the second most common genetic disease and the leading cause of infant deaths.

"Had we known how common this disease is and that there was a test to screen for carriers, we would certainly have requested it," says Deborah Heine, who now heads a nonprofit foundation devoted to increasing awareness of SMA carrier testing (*see sidebar*).

New guidelines

In the future, it is likely that fewer couples will have to suffer the Heines' ordeal. In November 2008, the American College of Medical Genetics (ACMG) issued new guidelines recommending that all couples be offered carrier testing for SMA mutations before conception, rather than only couples with a family history of the disease, as was previously recommended. Fortunately, couples who are both SMA carriers have many reproductive options.

Athena Diagnostics, a state-of-the-art laboratory dedicated to the development and commercialization of testing for neurologic and other disorders, is ready to meet the anticipated increased demand for high-quality SMA testing. Based in Worcester, Massachusetts, the lab currently offers more than 200 tests for neurologic disorders, including SMA, to the medical community.

"Our innovative, advanced technologies, extremely high standards, and well-earned reputation for accuracy and reliability have helped us become a leading provider of tests for conditions that affect millions of people," says Dr. Dev Batish, Senior Laboratory Director at Athena Diagnostics. "SMA is a major focus of our company, and we currently offer the most comprehensive test available for this disease."

Diagnosis and prognosis

Product Manager Karla Gojne explains that, in addition to carrier testing, Athena Diagnostics offers prenatal diagnostic testing for SMA. This not only determines with a very high degree of accuracy whether a pregnancy is affected, but also provides information that suggests how severe the disease might be.

It does this by detecting the number of copies of a gene called *SMN2* – a sort of "back up" gene that has the remarkable capacity to perform some of the functions of *SMN1*. In general, the more copies of *SMN2* someone with SMA has, the milder his or her disease. For example, patients with Type II or III SMA, who despite many challenges frequently live long, productive lives, usually have more copies of *SMN2* than those with Type I. "We are the only company to offer information on *SMN2* copy number," says Gojne, adding that this test is also available to children and adults.

"The information provided by Athena Diagnostics' comprehensive SMA diagnostic test gives expectant couples and their doctors precious time to make important decisions early in the pregnancy," says Athena Diagnostics collaborator Dr. Thomas W. Prior, author of the new ACMG guidelines and Director of Molecular Pathology at Ohio State University. "In concert with genetic counseling, this is an additional and valuable tool for expectant parents and their physicians."

According to Athena Diagnostics' Batish, SMA will continue to be a priority in the future. In the coming year, for example, the company plans to introduce three new SMA tests that will be able to detect rare genetic causes of SMA that currently escape detection by existing methods.



LEARN MORE ABOUT SMA

To learn more about spinal muscular atrophy (SMA) and SMA testing, visit:

- athenadiagnostics.com
- clairealmanheinefoundation.org
- smafoundation.org
- fsma.org
- geneclinics.org (click on "Gene Reviews" and enter "SMA")

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