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**FOR IMMEDIATE RELEASE**

## **Spinal Muscular Atrophy (SMA) Plain community carrier screening program yields impactful results**

*The program identified carriers in the Plain community to deliver innovative treatments presymptomatically to affected newborns*

STRASBURG, PA- The Clinic for Special Children (CSC) has completed its Spinal Muscular Atrophy (SMA) Prevention Readiness program. The program, launched in 2018, offered free carrier testing with the goal of identifying Plain (Amish or Mennonite) individuals and couples who are SMA carriers. SMA is the most common genetic cause of infant death worldwide and causes progressive degeneration of motor nerve cells in the spinal cord and brainstem. Historically the only treatment for SMA was care of the symptoms. However, recently approved targeted therapies have been shown to be effective in treating the disease. They are best administered before a patient shows significant symptoms of the disease so genetic diagnosis is key. One of the effective ways to identify families with a risk for the disease is to identify adult carriers.

Originally planning to collect 2,000 samples over 3 years, CSC conducted 2,177 tests in just 15 months across 15 states. There were 318 individuals identified as carriers of SMA, exceeding the 1 in 25 carrier frequency of the general Mennonite population but reflecting the effectiveness of the program's cascade testing (i.e., testing of blood relatives of identified carriers).

A total of nine (9) carrier couples were identified (both parents were carriers for SMA, having a 25% chance of having an affected baby). Furthermore, three (3) affected infants were identified at birth and all were treated presymptomatically with gene therapy. In addition, six (6) at-risk infants were tested as newborns and were not affected.



# Clinic for Special Children

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This program was funded by AveXis, a biotechnology company that launched Zolgensma® - a gene therapy for pediatric patients with SMA.

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## **About the Clinic for Special Children**

The Clinic for Special Children (CSC) is a non-profit organization located in Lancaster County, PA, which provides primary clinical care and advanced laboratory services to children and adults who have genetic or other complex disorders. Founded in 1989, CSC provides services to about 1,100 active patients and is recognized as a world leader in translational and precision medicine. The organization is primarily supported through community fundraising events and donations. For more information, please visit [www.ClinicforSpecialChildren.org](http://www.ClinicforSpecialChildren.org)