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FOR IMMEDIATE RELEASE

Amish Nemaline Myopathy Natural History Study Finds Promise for Gene Therapy Treatment

STRASBURG, PA- A new comprehensive natural history study about Amish nemaline myopathy (ANM) in the Old Order Amish population focuses on the promise of gene therapy for this lethal disorder. Amish nemaline myopathy (ANM) is an infantile-onset muscle disease linked to a mutation of the TNNT1 gene. The study summarizes genealogical records, clinical data, and molecular reports of one hundred and six ANM patients born between 1923 and 2017 and was led by researchers from the Clinic for Special Children in Strasburg, PA. It appeared this month in the journal *Human Molecular Genetics*.

All ANM patients in the study were born at a normal birth weight, failed to thrive by 9 months of age, and died at a median age of 18 months from respiratory failure. Progressive deformity of the chest wall has led to the disorder to commonly be referred to as “chicken breast disease”. Symptoms of ANM shortly after birth include low muscle tone, hip and shoulder stiffness, and tremors, followed by progressive muscle weakness, degeneration and joint contractures. ANM has a carrier frequency of 6.5% in the Old Order Amish population and because the community is reluctant to use costly or invasive life-sustaining technologies for an otherwise lethal disorder, the natural history of the disease was observed without interventions.

Muscle biopsies from two ANM patients showed an abnormal pattern of muscle fibers with preserved nerve function. Researchers compared the human ANM muscle biopsies with those from a transgenic mouse model; they were very similar, suggesting the mice can serve as a good model of the human disease. At the Clinic for Special Children, genetic testing allows practitioners to confirm the diagnosis of ANM within days of birth. The findings in this study provide a strong platform for exploring gene replacement therapy in newborns diagnosed with ANM.

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The research was conducted by a team including the study's joint lead authors, Michael D. Fox, MD from Diagnostic Referral Division, Nemours/Alfred I. duPont Hospital for Children in Wilmington, DE, Department of Pediatrics, Sidney Kimmel Medical College at Thomas Jefferson University in Philadelphia, PA and the Clinic for Special Children in Strasburg, PA and Vincent J. Carson MD, from the Clinic for Special Children; senior author Kevin A. Strauss, MD from the Clinic for Special Children; Han-Zhong Feng, MD, PhD and J.-P. Jin, MD, PhD from Department of Physiology, Wayne State University School of Medicine in Detroit, MI, Michael W. Lawlor, MD, PhD from Department of Pathology and Laboratory Medicine and Neuroscience Research, The Medical College of Wisconsin in Milwaukee, WI, John T. Gray, PhD from Audentes Therapeutics in San Francisco, CA, and Karlla W. Brigatti, MS, LCGC from Clinic for Special Children.

About the Clinic for Special Children

The Clinic for Special Children (CSC) is a non-profit organization located in Strasburg, PA, which provides primary pediatric care and advanced laboratory services to those who suffer from genetic or other complex medical disorders. Founded in 1989, the organization provides services to over 1,050 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