



Contact: Kelly Cullen  
P | 717-687-9407  
E | kcullen@clinicforspecialchildren.org

**FOR IMMEDIATE RELEASE**

## **INTERNATIONAL STUDY OF TNNT1 MYOPATHY ESTABLISHES OPERATIONAL FRAMEWORK FOR FUTURE CLINICAL TRIALS**

STRASBURG, PA. September 2023 - A new study summarizes WiTNness – a hybrid prospective/cross-sectional observational study of TNNT1 myopathy, a rare and lethal neuromuscular disease that causes progressive muscle weakness and stiffness, while cognitive and social development remain normal. The study aimed to identify clinically meaningful endpoints for future therapeutic trials following recent guidelines on natural history studies to support clinical trial readiness. The study was a broad collaborative effort led by clinicians and researchers at the Clinic for Special Children that included participants from around the globe and appears in the current issue of *Annals of Clinical and Translational Neurology*.

The study followed the clinical course of 16 participants in the cross-sectional cohort and six children in the prospective cohort. The study examined the endpoints of ventilator-free survival and “thriving” or the ability to swallow and grow normally without non-oral feeding support. The median ventilator-free survival age was 20.2 months and probability of death or permanent mechanical ventilation was 100% by 60 months, which demonstrates the severity of the disease. All six children (100%) in the prospective cohort failed to thrive by 12 months of age.

In addition to using motor milestone checklists and validated motor scales adapted to this patient population, the study team used a novel motor assessment tool – the Ability Captured Through Interactive Video (ACTIVE-mini) system that records and quantifies movement of uniquely colored tags on the infant’s limbs. This data, along with laboratory and other imaging tools, suggests that primary symptoms of TNNT1 myopathy are restricted to the skeletal muscle.



# Clinic for Special Children

535 Bunker Hill Road, PO Box 128, Strasburg, PA 17579 T 717.687.9407 F 717.687.9237

The WiTNness study established a sound operational framework for future clinical trials with the goal of accelerating therapeutic development for this devastating and ultrarare disease. A similar hybrid study could be applied to other similar rare disorders to economize and democratize the development of critical therapies around the world.

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The research was conducted by a team including the study's first author Kevin A. Strauss from the Clinic for Special Children, Strasburg, PA, Department of Pediatrics, Penn Medicine-Lancaster General Hospital, Lancaster, PA, Department of Pediatrics, UMass Chan Medical School, Worcester, MA, and Department of Molecular, Cell & Cancer Biology, UMass Chan Medical School, Worcester, MA; Vincent J. Carson from the Clinic for Special Children, Strasburg, PA and Department of Pediatrics, Penn Medicine-Lancaster General Hospital, Lancaster, PA; Emilienne Bolettieri, Mariah Everett, Ashton Bollinger, Lauren E. Bowser, Keturah Beiler, Millie Young, and Karlla W. Brigatti from the Clinic for Special Children, Strasburg, PA; Simon Edvardson and Nitay Fraenkel from ALYN Hospital Pediatric and Adolescent Rehabilitation Center, Jersulam, Israel; Adele D'Amico and Enrico Bertini from the Unit of Muscular and Neurodegenerative Disorders, Department of Neurosciences, IRCCS Bambino Gesù Children's Hospital in Rome, Italy; Lokesh Lingappa from the Department of Pediatric Neurology, Rainbow Children's Hospital, Hyderabad, India; Devyani Chowdhury from Cardiology Care for Children, Lancaster, PA and Department of Cardiology, Nemours Children's Health, Wilmington, DE; Linda P. Lowes, Megan Iammarino, and Lindsay N. Alfano from Center for Gene Therapy, Nationwide Children's Hospital, Columbus, OH.

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

The Clinic for Special Children (CSC) is a non-profit organization located in Strasburg, PA, which provides comprehensive medical care and advanced laboratory services to those who live with genetic or other complex medical disorders. Founded in 1989, the organization provides services to over 1,400 individuals 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)