

GET INVOLVED

The TANGO2 Research Foundation is a team of parents, family members, medical professionals and other volunteers - all dedicated to the core mission of the foundation.

We are always looking for more caring, dedicated people to volunteer their talents and time to help create a more hopeful future for those living with TANGO2 disease. Please visit our web site to learn more.



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tango2research.org

**BETTER
UNDERSTANDING
THROUGH
RESEARCH.**



TANGO2
RESEARCH FOUNDATION



OUR MISSION IS TO LEAD THE WAY IN FINDING A CURE FOR TANGO2 RELATED DISEASE BY HELPING TO FUND, COORDINATE AND GUIDE SCIENTIFIC RESEARCH THAT LEADS TO A BETTER UNDERSTANDING OF TANGO2 DISEASE.

WHAT IS TANGO2 DISEASE?

TANGO2 itself is a protein coding gene on chromosome 22 (22q11.21). This gene belongs to the transport and Golgi organization family, whose members are predicted to play roles in secretory protein loading in the endoplasmic reticulum.

Individuals with TANGO2-related metabolic encephalopathy and arrhythmias can present in acute metabolic crisis (hypoglycemia, hyperlactacidemia, mild hyperammonemia) or with developmental delay, regression, and/or seizures. The acute presentation varies from profound muscle weakness, ataxia, and/or disorientation to a comatose state. The majority of individuals present with intermittent acute episodes of rhabdomyolysis.

PRIMARY SYMPTOMS

Metabolic Crisis

- May include hypoglycemia, hyperlactacidemia, and mild hyperammonemia
- Urine organic acids can show marked ketoacidosis and lactic acidosis

Rhabdomyolysis

- Myoglobinuria and profound lower-extremity weakness can develop
- CPK significantly elevated in some individuals (>200,000 U/l).
- Elevated aldolase and transaminases

Cardiac Arrhythmias

- During acute illness, transient electrocardiogram (ECG) changes can be seen, most commonly QT prolongation and rarely Brugada type I pattern
- Life-threatening recurrent ventricular tachycardia (VT) or torsade de pointes can occur during times of acute illness

Other Acute Symptoms

- Acute presentation can vary from profound muscle weakness, ataxia, and/or disorientation to a comatose state

OTHER SYMPTOMS

CONTRIBUTING TO DIAGNOSIS

- Intellectual disability
- Regression of motor and cognitive skills
- Hypothyroidism
- Poor coordination and unsteady gait
- Increased tone
- Episodic muscle weakness
- Benign paroxysmal torticollis (BPT)
- Hyperreflexia
- Clonus
- Unexplained seizures (> 75%)
- Myopathic facies
- Brain imaging abnormalities
- Intermittent exotropia

DIAGNOSIS & TESTING

The diagnosis of TANGO2-related metabolic encephalopathy and arrhythmias is established in a proband by identification of biallelic pathogenic variants in TANGO2 on molecular genetic testing.