

# TANGO2 Deficiency Disorder Fact Sheet

## What is TANGO2 Deficiency Disorder?

TANGO2 deficiency disorder (TDD) is a rare genetic condition caused by changes in the TANGO2 gene. Although there are some common symptoms and conditions across those affected by TANGO2 disease, there is also broad variability of presentation and severity.

### Acute Symptoms

- **Metabolic Crisis:** a serious condition caused by low blood sugar and the build-up of toxic substances in the blood.
- **Rhabdomyolysis:** a breakdown of muscle tissue that releases a damaging protein into the blood
- **Cardiac Arrhythmias:** life-threatening ventricular tachycardia (fast heart rate)



### Other Important Symptoms

- **Intellectual Disability:** present in almost all individuals with varying severity
- **Regression:** Loss of previously acquired motor, verbal and cognitive skills
- **Poor Coordination and Unsteady Gait**
- **Episodic Muscle Weakness:** recurrent episodes of muscle weakness in hands, limbs or trunk that can last minutes, hours or days
- **Benign Paroxysmal Torticollis (BPT):** periods of unusual, sustained posturing of the head and neck, during which the head tilts to one side
- **Seizures:** Seizures are observed in more than 75% of individuals

## What Treatments Are Available?

**B Vitamins-** While there is no known cure for TDD, there is evidence that shows that including a high daily dose of B vitamins has potential to greatly improve the health of individuals living with TDD. While B5 and B9 have specifically been referenced in TDD research, most TDD patients take a B Complex with all 8 B vitamins and an additional dose of B5 of at least 50mg. Consult your health care provider to help determine the best treatment options for your TDD warrior.

# Newly Diagnosed? 5 Things to Know

1

## YOU ARE NOT ALONE

While a TDD diagnosis can feel daunting, there is support, growing resources, and hope.

The information on this sheet is here to help you take the first steps. You are now part of a community that understands and is here to support you.

2

## LEARN & ASK QUESTIONS

There are several resources available to help you feel connected, informed, and supported as you navigate life with TDD (Use links or scan QR codes.)

- [The TANGO2 Research Foundation](#) website is a great place to start. It offers valuable insights about early diagnosis, potential treatments, managing symptoms, and preventing metabolic crises.
- [The TANGO2 Family Facebook Group](#) is a welcoming community where families from around the world connect, share experiences, and support one another.
- [TANGO2 Youtube Channel](#)
- [The TANGO2 Research Learning Network](#) provides a space for patients, families, physicians, and researchers to share knowledge and collaborate on advancing care and research for TDD.

3

## BUILD A CARE TEAM

Family members are an extremely important part of the TDD caregiving team, and they work with multiple healthcare specialists to help manage the care of individuals living with TDD.

- **Healthcare Specialists** - The range of symptoms and challenges for individuals living with TDD are variable but many TDD individuals see a range of medical specialists including; a Geneticist, Neurologist, Endocrinologist and Cardiologist.
- **Other healthcare professionals** - Others might include Dietician, Physical, Speech and Occupational Therapists and special educational service providers.

4

## BE PREPARED

Keep your resources handy in case of an emergency.

- **Emergency Protocol Letter** - Make sure you and all health care providers and caregivers have access to the TDD Emergency Protocol Letter. This is most effective when your child's doctor personalizes it, signs it and puts it on letterhead for their practice or hospital.
- **TDD Emergency Hotline** for medical professionals +1-860-598-0955. A TANGO2 hotline is available for any physician treating a TANGO2 patient. Doctors can call this emergency number at any time to request consultation from TDD medical experts. Please share this resource with your medical team.
- **Email:** For non-urgent questions, they can reach out via email at [tango2.research@bcm.edu](mailto:tango2.research@bcm.edu)

5

## GET INVOLVED

You can help find a cure for TANGO2 Deficiency Disorder.

- **TANGO2 Deficiency Disorder Natural History Study** - This study collects detailed information about TDD over time, tracking how it develops and affects patients. This data helps researchers better understand TDD, identify patterns, and find targets for future treatments or cures. By joining this study, patients and families contribute crucial information that accelerates research and advances care for TDD.
- **Volunteer** - Together, we can make a difference in the journey toward a cure.
- **Fundraise** - The TANGO2 Research Foundation relies on fundraising and donations to fund our research and work. Hosting your own fundraiser lets you tap into your talents and interests to inspire others to get in on the action.



T2RF Website



Facebook Group



Youtube Channel



Learning Network



Emergency Letter



Natural History Study



Volunteer

Visit [www.tango2research.org](http://www.tango2research.org) for more information