

TANGO2 Deficiency Disorder Fact Sheet

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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 TDD, there is also broad variability of presentation and severity.

Symptoms at Baseline

- Developmental delay
- Intellectual disability: in most individuals with varying severity
- Poor coordination and gait abnormalities
- Speech difficulties
- Strabismus or lazy eye
- Seizures (40-50%)
- Hypothyroidism (40-50%)
- TANGO2 Spells: can include head tilt, body tilt, abnormal posturing, loss of muscle control, drooling, extreme fatigue
- Typically no laboratory abnormalities are seen during these episodes



Acute Symptoms

- **Metabolic Crisis:** can be triggered by fasting, infection, dehydration, exposure to excessive heat, etc. Symptoms can be disorientation, worsening gait abnormality, swallowing difficulties, muscle weakness, and red urine. Laboratory abnormalities and abnormal EKG are seen during these episodes. Muscle breakdown can result in high levels of CK in blood. High levels of lactate and liver enzymes can be seen. Low glucose level can occur at times. Heart tracing can show prolonged QTC which can develop into serious heart arrhythmias.
- **Cardiac Crisis:** About $\frac{2}{3}$ of patients in acute metabolic crisis can develop cardiac crises which are characterized by serious ventricular tachycardia (fast heart rate), or cardiomyopathy (weakness of heart muscle), or stopping of the heart (cardiac arrest).



What Treatments Are Available?

B Vitamins – While there is no known cure for TDD, evidence suggests 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 vitamin complex with all 8 B vitamins plus an additional dose of vitamin 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.

- [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, clinicians, 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

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 healthcare and medical specialists including; a Geneticist, Neurologist, Endocrinologist, Cardiologist, Dietician, Physical, Speech and Occupational Therapists as well as special educational service providers.

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BE PREPARED

Keep your resources handy in case of an emergency.

- **Emergency Protocol Letter** Make sure you, caregivers and all health care providers have access to the TDD Emergency Protocol Letter. This is most effective when your child's primary care physician personalizes it, signs it and puts it on letterhead for their medical practice or hospital.
- **TDD Clinical Consult Emergency Hotline +1-860-598-0955.** This hotline provides life-saving consultation to clinicians treating patients with TDD including those that are hospitalized, currently in crisis, or at risk of crisis. Go to tango2hotline.org for more details.
- **Email** For non-urgent clinical questions, contact TDD medical experts via email at tango2.research@bcm.edu

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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 clinicians and 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 individuals with TDD.
- **Volunteer** - Together, we can make a difference in the journey toward a cure.
- **Fundraise** - The TANGO2 Research Foundation depends on fundraising and donations to support our mission critical initiatives. Hosting your own fundraiser allows you to leverage your talents and passions to engage and inspire others to contribute to this important cause.



T2RF Website



Facebook Group



Youtube Channel



Learning Network



Emergency Letter



Natural History Study



Volunteer

Visit www.tango2research.org for more information