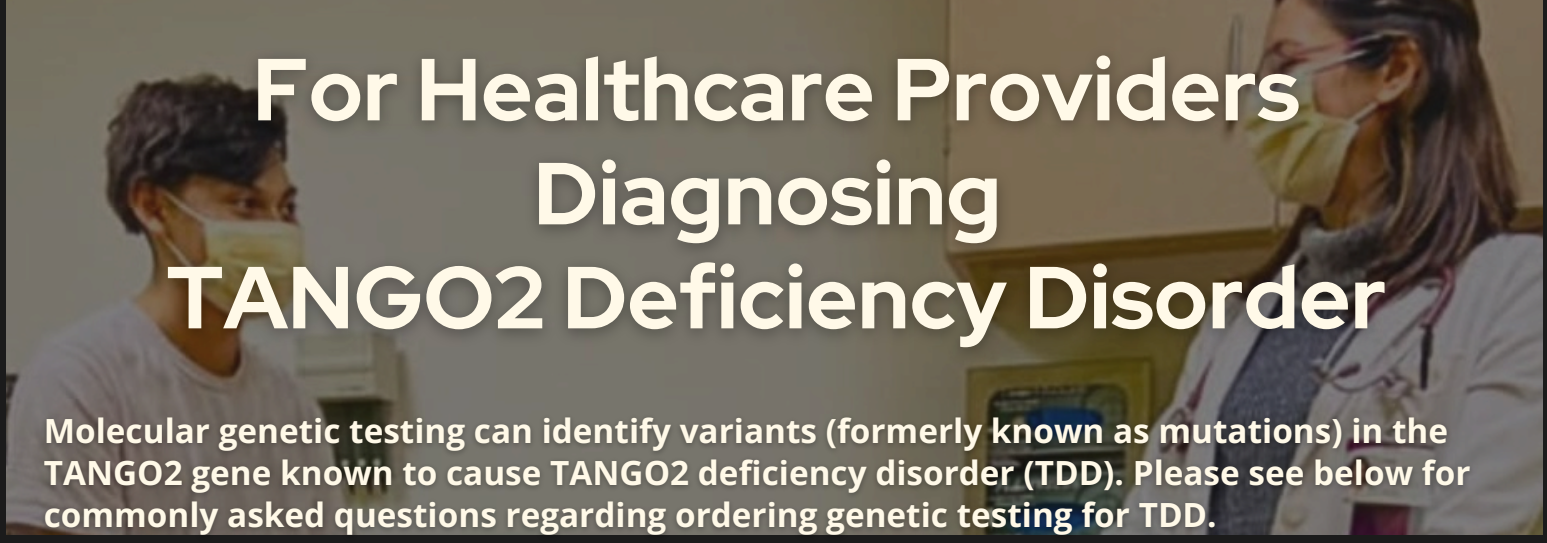

For Healthcare Providers Diagnosing TANGO2 Deficiency Disorder

Molecular genetic testing can identify variants (formerly known as mutations) in the TANGO2 gene known to cause TANGO2 deficiency disorder (TDD). Please see below for commonly asked questions regarding ordering genetic testing for TDD.

Which labs are offering TANGO2 gene testing? Genetic testing for TDD is typically offered by specialized genetic testing labs. For a list of labs currently offering TDD testing, we recommend searching for TDD on the Concert Genetics website.
The most comprehensive genetic testing methodology will include next-generation gene sequencing analysis as well as gene deletion and duplication studies.

How can I order TANGO2 genetic testing? TDD testing specifically can be ordered as a single-gene test or can be part of a multi-gene panel. A list of tests are available on the Concert Genetics website.
If you are ordering TDD testing as part of a multi-gene panel, it is important to check the assay methodology to determine whether both sequencing and deletion/duplication studies will be performed.

Through broad testing such as Whole Exome Sequencing or Genome Sequencing, a pathogenic variant in TANGO2 can be identified in most cases. However, you can contact the lab directly to report on any limitations in coverage of the TANGO2 gene on the particular test that was ordered.

If patient financial resources are limited, targeted analysis for TANGO2 pathogenic variants can be performed first in selected populations based on the ethnicities below:

- In individuals of Hispanic ethnicity from Latin America, targeted analysis for pathogenic variant c.460G>A (p.Gly154Arg) and the ~34-kb deletion encompassing exons 3-9 can be performed.
- In individuals of European ancestry, targeted deletion analysis for the ~34-kb deletion encompassing exons 3-9 can be performed.

Will TANGO2 testing be covered by my patient's health insurance? Coverage depends on the patient's insurance plan. Contact the insurer to confirm coverage, out-of-pocket costs, and any prior authorization. If not covered, most labs offer financial assistance or self-pay options. A Medical Geneticist or Genetic Counselor may also help identify research-based no-cost testing opportunities.

I have a patient with 22q11.2 deletion syndrome (DiGeorge Syndrome), should they be tested for TANGO2 deficiency? As the TANGO2 gene is located within the chromosomal 22q11.2 region, patients with this deletion are at a higher risk of having TANGO2 deficiency disorder. It is important to evaluate each patient with 22q11.2 deletion syndrome to determine whether they have clinical findings suggestive of TANGO2 deficiency disorder, especially if they have had an acute metabolic crisis or TANGO2 "spells" recognized by the sudden onset of hypotonia with loss of muscle control (e.g., clumsy gait, sudden falling while walking or sitting, inability to upright themselves after falling), head tilt (either to the side or back), body tilt, dystonia, abnormal posturing with hypertonicity, increased dysarthria, drooling, extreme fatigue, and disorientation.

Do patients with TDD show different traits according to the specific genetic abnormality in the TANGO2 gene? At this time, no genotype-phenotype correlations have been reported. Please check the TANGO2 GeneReviews page for the most updated information.

If you need assistance interpreting your patient's genetic testing result or would like to refer your patient to a genetics specialist, please contact a Genetics Clinic in your area.