

TANGO2-Related Metabolic Encephalopathy phenotyping

Name	
ID	
Gender	☐ Male ☐ Female
Date of birth	/ /
Ethnicity	
Referring Physician	
<u>Initial presentation</u>	
Onset of the first symptom	
Any dysmorphic features (e.g. hypodontia)	☐ Yes ☐ No
Diagnosis age	
Weeks of pregnancy	
Weight	
Height	
Head circumference	
Neonatal problems (birth - 28 days postpartum)	
First symptoms age (years)	
TANGO2 spell frequency	
First symptoms (description)	

If possible, add detailed description below:

<u>Neurological findings</u>	
Microcephaly	☐ Yes ☐ No
Episodic Weakness	☐ Yes ☐ No
Dystonic movements	☐ Yes ☐ No
Spasticity (modified Ashworth scale)	☐ Yes ☐ No
Hyperreflexia (+++ in neurological exam)	☐ Yes ☐ No

Seizures	Description:
classification if known	☐ Generalized tonic-clonic ☐ Tonic ☐ Clonic ☐ Absence ☐ Absence + eyelid myoclonia ☐ Myoclonic absences ☐ Atonic/astatic ☐ Myoclonic-atonic ☐ Focal with motor signs ☐ Focal with sensory symptoms ☐ Focal with vegetative symptoms ☐ Focal with impaired consciousness (dyscognitive) ☐ Febrile seizures ☐ Afebrile seizures
Are the seizures related to episodes of hyperglycemia or ventricular tachycardia?	☐ Yes ☐ No
Add detailed description below:	
MRI findings	☐ Normal ☐ Abnormal ☐ Unknown ☐ Not done
If possible, add details:	
EEG	☐ Normal ☐ Focal epileptiform ☐ Generalized epileptiform ☐ Other abnormality ☐ Unknown ☐ Not done
If possible, add details:	
Developmental delay	☐ Yes ☐ No

Severity of developmental delay		☐ Mild (difficulties at school) ☐ Moderate (special school) ☐ Severe (non-verbal)
Regression (lost skills)		☐ Yes ☐ No If yes, at which age?
Oromotor dysfunction (dysphagia and/or sialorrhea)?		
Details	Can walk	☐ Independently ☐ With some help ☐ Wheel-chair-bound
	Can talk	☐ Fluently ☐ Simple sentences ☐ Few words ☐ Non-verbal
	Muscle tone	☐ Increased (spasticity) ☐ Decreased, floppy (hypotonia)
For any further details, add below:		
Gross motor delay? IQ findings?		
<u>Musculoskeletal</u>		
Rhabdomyolysis	☐ Yes ☐ No ☐ Unknown	
Skeletal deformities	☐ Yes ☐ No	
<u>Ophthalmologic findings</u>	Description: ☐ Normal ☐ Abnormal	
If possible, add details:		
<u>Laboratory findings</u>		
Documented Hypoglycemia during acute metabolic crisis	☐ Yes ☐ No ☐ Unknown ☐ Not done	

Ammonia levels	☐ Elevated ☐ Normal ☐ Unknown ☐ Not done
Lactic acidosis during metabolic crisis	☐ Present ☐ Not present ☐ Unknown
Creatine kinase levels	☐ Elevated ☐ Normal ☐ Unknown ☐ Not done Amount (U/L):
Transaminases	☐ Elevated ☐ Normal ☐ Unknown ☐ Not done
Thyroid stimulating hormone abnormalities	☐ Yes ☐ No ☐ Unknown
Urine organic acids (Elevated urine dicarboxylic acids)	☐ Yes ☐ No
Acylcarnitine profile (elevated acylcarnitine)	☐ Yes ☐ No
If possible, add details:	
<u>Gastrointestinal findings</u>	
Feeding difficulties	☐ Yes ☐ No ☐ Unknown
Constipation	☐ Yes ☐ No ☐ Unknown
Daytime incontinence (> 5y and involuntary voiding)	☐ Yes ☐ No ☐ Unknown
If possible add details:	

Cardiac findings

Electrocardiogram in the setting of metabolic crisis

- ☐ Normal
- ☐ Abnormal
- ☐ Unknown
- ☐ Not done

If possible add details:

Other Organ Involvement

Hepatosplenomegaly

- ☐ Yes
- ☐ No
- ☐ Unknown

Lung problems

- ☐ Yes
- ☐ No
- ☐ Unknown

Immunodeficiency symptoms

- ☐ Yes
- ☐ No
- ☐ Unknown

Neurogenic hearing loss

- ☐ Yes
- ☐ No
- ☐ Unknown

If possible add details:

Treatment and Outcome

Medication

Please add medication details:

Outcome

Death

Is the patient deceased?

- ☐ Yes
- ☐ No

If yes add details on the cause of death: