

DISEASE:

Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome

NAME:	Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome
DESCRIPTION:	Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome is a rare, genetic, neurodegenerative disease characterized by episodic metabolic encephalomyopathic crises (of variable frequency and severity which are frequently precipitated by an acute illness) which manifest with profound muscle weakness, ataxia, seizures, cardiac arrhythmias, rhabdomyolysis with myoglobinuria, elevated plasma creatine kinase, hypoglycemia, lactic acidosis, increased acylcarnitines and a disorientated or comatose state. Global developmental delay, intellectual disability and cortical, pyramidal and cerebellar signs develop with subsequent progressive neurodegeneration causing loss of expressive language and varying degrees of cerebral atrophy.
ORPHACODE:	480864
SYNONYMS:	TANGO2-related metabolic encephalopathy-arrhythmia syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	TANGO2
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