

GENETIC TEST:**Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome (TANGO2 gene)**

FULL NAME:	Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome (TANGO2 gene)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Carrier diagnosis, Mutation confirmation, Post-natal Diagnosis, Pre-implantation genetic diagnosis, Prenatal diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, Skin biopsy, Muscle biopsy, Liver biopsy, Amniotic fluid, Chorionic villi, Cell culture, Skin fibroblasts

METHOD CATEGORY:	Sequence analysis: entire coding region Mutation screening and sequence analysis of selected exons
METHOD TECHNIQUE:	Next Generation Sequencing (NGS) Bi-directional Sanger Sequence analysis
RIZIV CODE:	565456-565460
EQA:	• DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger , • DNA Sequencing - Sanger
TURNAROUND TIME (MAXIMUM):	3 months (10 working days for prenatal diagnosis)
CREATED:	28 Aug 2019 - 14:28
CHANGED:	09 Mar 2023 - 16:21

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