
Lab Dept: Anatomic Pathology

Test Name: MELAS SYNDROME (MTTL1) SEQUENCING

General Information

Lab Order Codes: MELS

Synonyms: MTTL1 Mutation

CPT Codes: 81401 – Molecular pathology procedure, Level 2

Test Includes: Sequencing of the MTTL1 gene.

Logistics

Test Indications: Testing of patients with a confirmed or suspected diagnosis of MELAS.

Mutations in the MTTL1 mitochondrial gene (tRNA-Leu) have been identified in patients with MELAS. Mutations in MTTL1 can result in impaired mitochondrial respiratory chain complex I and cytochrome C oxidase activity. MELAS is characterized by mitochondrial encephalomyopathy, lactic acidosis, stroke-like episodes, often presenting in childhood. Other common symptoms include seizures, muscle weakness, recurrent headaches and vomiting, and exercise intolerance. Sensorineural hearing loss may also occur. Symptoms are highly variable, in part due to the amount of heteroplasmy. Symptoms may worsen during times of stress or illness.

Lab Testing Section: Anatomic Pathology - Sendouts

Referred to: EGL Genetics (EGL Test#: QA)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 3 weeks

Special Instructions: A complete [Emory general test request form](#). Select MELAS testing under Mitochondrial Disease.

Specimen

Specimen Type: Whole blood

Container:	Lavender top (EDTA) tube Alternate: Yellow-top (ACD) tube
Draw Volume:	Children <2 yrs of age: 2-3 mL blood Pediatrics: 3 – 5 mL blood Adults: 5 – 10 mL blood
Processed Volume:	1 – 10 mL whole blood
Collection:	Routine blood collection
Special Processing:	Lab Staff: Do Not Centrifuge. Specimen should remain in the original collection container. Store refrigerated until shipment. Ship overnight at room temperature to EGL Genetics. Forward promptly.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens.

Interpretive

Reference Range:	Interpretive report
Methodology:	The MTTL1 mitochondrial gene (tRNA-Lue) is PCR amplified and sequenced in both the forward and reverse direction. The 3234A>G mutation is analyzed by a sensitive allele-specific extension assay to reliably detect heteroplasmic mutations. Patient gene sequences are compared to a normal reference sequence. Sequence variations are then classified as previously described mutations, novel mutations, or variations of unknown significance.
References:	EGL Genetics Laboratory January 2018 (470) 378-2200 Fax: (470) 378-2250