Test Name & Code:

- **Test Name:** Melas Syndrome (A3243G, T3271C, A3252G)
- **Test Code:** 4624

Related Diseases & Genes:

• "MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES" (OMIM #: 540000) || Gene: *MTTL1* (OMIM #: 590050)

Synonyms:

- Mitochondrial Myopathy
- A3243G Mutation
- T3271C Mutation
- A3252G Mutation

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 3 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 5 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

• Neurogenetics