Mitochondrial Genome Deletions/Duplications: Southern Blot

**Test Code:** ML  
**Turnaround time:** 4 weeks  
**CPT Codes:** 81479 x1

### Condition Description

**Indications**

Mitochondrial disorders such as Kearns Sayre, Chronic Progressive External Ophthalmoplegia (CPEO) and Leigh Disease.

**Methodology**

Southern analysis of mitochondrial genome following digestion with EcoRV, BamHI and SnaBI.

**Detection**

Over 95% of large mitochondrial deletions.

**Reference Range**

Qualitative assay.

**Specimen Requirements**

Submit only 1 of the following specimen types

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) or ACD (yellow top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Type: Muscle Biopsy**

Specimen Requirements:

1-2 mm in length or > 100 mg is acceptable.

Flash freeze sample upon collection using liquid nitrogen. If storage is required, store sample at -80°C or colder.

Specimen Collection and Shipping: Ship frozen sample on dry ice with overnight delivery.