Myotonic Dystrophy: CTG Repeat Analysis

**Test Code:** MD  
**Turnaround time:** 3 weeks  
**CPT Codes:** 81404 x1

### Condition Description

Myotonic dystrophy (DM) is the most common adult muscular dystrophy. It is inherited as an autosomal dominant disorder. DM presentation ranges from congenital to adult onset. The congenital form typically presents with hypotonia and respiratory distress often followed by early death or severe mental retardation. In adults, the characteristic findings include:

- Progressive weakness
- Muscle wasting
- Myotonia
- Lenticular opacities
- Frontal balding
- Testicular atrophy.

DM has been associated with an amplification of an unstable CTG repeat in the *DMPK* gene.

### Genes

**DMPK**

### Indications

### Methodology

Testing for DM includes PCR amplification and fragment size analysis, as well as triplet-primed PCR analysis. Large size expansions may require Southern analysis for accurate size estimation.

### Detection

Nearly all CTG expansions in the *DMPK* gene will be detected by this assay.

### Reference Range

Normal individuals carry 5-34 CTG repeats. Abnormal individuals carry >50 repeats.

### Specimen Requirements

**Submit only 1 of the following specimen types**

#### Type: Whole Blood (EDTA)

**Specimen Requirements:**
- EDTA (Purple Top)  
  - Infants and Young Children (< 2 years of age to 10 years old): 3-5 ml  
  - Older Children & Adults: 5-10 ml  
- Autopsy: 2-3 ml unclotted cord or cardiac blood

**Specimen Collection and Shipping:**
Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

#### Type: DNA, Isolated

**Specimen Requirements:**
- Microtainer  
  - 20µg  
- Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

**Specimen Collection and Shipping:**
Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

### Special Instructions

Please call the laboratory genetic counselor to arrange prenatal testing prior to collecting a prenatal sample.