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

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple 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.

## Special Instructions

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