

## Myotonic Dystrophy type 1 DNA Screen

### Background

Over 99% of myotonic dystrophy type 1 (DM1) cases are caused by expansion of a CTG trinucleotide repeat in one allele of the *DMPK* gene at chromosome 19q13.3. *DMPK* alleles with >55 repeats are abnormal; alleles with 38-54 repeats are normal but potentially mutable upon transmission; alleles with <37 repeats are normal. The number of CTG repeats does not reliably predict the age of onset, the rate of disease progression, or the severity of symptoms for an individual patient. Affected children born to women who are affected or who carry an abnormally expanded *DMPK* allele are at increased risk for having the severe congenital form of the disorder.

### Indications for Testing

- Confirm a clinical diagnosis of myotonic dystrophy
- Evaluate suspected myotonic dystrophy
- Presymptomatic testing of adults at-risk for myotonic dystrophy (in conjunction with genetic counseling)
- Prenatal diagnosis of an at-risk pregnancy
- Differential diagnosis of neonates with unexplained hypotonia and/or respiratory distress.

### Restrictions/Exclusions from Testing

- Presymptomatic testing of minors (< age 18) (including evaluations for adoption)

### Genetic Counseling

Genetic counseling can be useful to patients and families considering genetic testing. The laboratory can provide referrals to genetics clinics in the patient's locale or a listing can be found at [www.genetests.org](http://www.genetests.org)

### Ordering

1. Obtain blood sample.
  - For prenatal testing, call the Genetics Lab with information on fetal sample and expected date of receipt (blood sample of affected parent may also be required).
2. Fill out a Clinical Lab Request - Genetics for each patient  
Available at <http://depts.washington.edu/labweb/Divisions/MolDiag/MolDiagGen/index.htm>  
-Request: "Myotonic dystrophy"
3. Provide information needed for test interpretation:
  - Reason for ordering test (e.g., confirm; rule out; presymptomatic)
  - Clinical history (muscle weakness; myotonia; abnormal EMG)
  - Family history / pedigree (relatives with DM1 or suspected myotonic dystrophy)
4. Call Laboratory Medicine Community Services (206) 598-6066 to arrange the best method of shipment.

### Sample Requirements and Specimen Handling

Whole blood - EDTA (purple top) - 5 mL. (Note: Heparin (green top) tubes are not acceptable.)  
Samples should be received within 72 hours of collection.  
Samples may be refrigerated until shipped.  
For prenatal diagnosis specimens, consult laboratory.

### Test Frequency and Reporting

Test results usually within 2-3 weeks of specimen receipt. A written interpretative report is issued.

## **References**

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