
Lab Dept: **Anatomic Pathology**

Test Name: **MYOTONIC DYSTROPHY**

General Information

Lab Order Codes: MYOD

Synonyms: Myotonic Dystrophy Molecular Analysis

CPT Codes: 81401 – Molecular Pathology procedure, Level 2

Test Includes: Only DM1 testing is performed. PCR and Southern blot analysis are used in combination to determine allele repeat size. PCR will detect up to approximately 100 CTG repeats while Southern blot is used to detect the larger repeats.

Logistics

Test Indications: Is useful to confirm the diagnosis and to identify the disease causing mutations within a family to allow for carrier testing and prenatal diagnosis.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: Greenwood Genetic Center

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: 24 hours

Turnaround Time: Results are reported within 3 weeks.

Special Instructions: N/A

Specimen

Specimen Type: Blood

Container: Lavender (EDTA) tube.

Draw Volume: 5 - 10 mL (Minimum: 3 mL) blood

Processed Volume: **Do Not** process or centrifuge whole blood. Submit specimen in original container.

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| Collection: | Routine venipuncture |
| Special Processing: | Lab Staff: Store unprocessed blood at room temperature. Ship at room temperature via overnight courier. If shipment is delayed by one or two day, the specimen should be refrigerated and shipped at room temperature. Do not freeze the specimen. Samples collected on Friday can be safely designated for Monday delivery |
| Patient Preparation: | None |
| Sample Rejection: | Frozen specimen; mislabeled or unlabeled specimens |

Interpretive

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| Reference Range: | Allele sizes are reported and assigned to be within the normal, intermediate or affected ranges. Normal (3-34 repeats), indeterminate (35-49 repeats), affected (\geq 50 repeats). |
| Critical Values: | N/A |
| Limitations: | N/A |
| Methodology: | Amplification of DNA to evaluate trinucleotide repeats, using Southern transfer technique when appropriate. |
| References: | Greenwood Genetic Center September 2012 |
| Updates: | 9/17/2012: Testing previously referred to Fairview University Medical Center 2/7/13: CPT update |