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.

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	
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 (=/> 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 refered to Fairview University Medical Center 2/7/13: CPT update