

Myotonic Dystrophy (DM) Mutation Testing

DESCRIPTION:

- Myotonic Dystrophy (DM), an autosomal dominant disorder with 1 in 8000 incidence, manifests as muscle wasting, myotonia, as well as systemic effects.
- DM symptoms vary widely, from very mild weakness to severe and potentially fatal congenital hypotonia, and tend to increase in severity with age and in successive generations, especially with transmission through affected females.
- This test detects the genetic abnormality known to cause DM: unstable CTG trinucleotide expansion in the DNA of a noncoding region of the myotonin protein kinase gene at chromosome 19q13.3 (*Science*, 1992, 255:1256-1258).

REASONS FOR REFERRAL:

- Confirmation of diagnosis in a symptomatic patient; testing of children with questionable symptoms and signs in an affected family; testing of newborns with severe hypotonia and / or facial diplegia.

METHOD OF ANALYSIS:

- DNA from the patient is amplified by PCR using fluorescent primers. CTG allele sizes are determined by a high resolution laser-induced capillary electrophoresis system with internal standard.
- Southern hybridization of probe pMDY1 to a HindIII / BglI digest of genomic DNA confirms PCR results and detects larger expansions.
- Results are reported within 3 weeks or less of receipt of sample.

PLEASE CALL THE LAB WHEN TESTING A HYPOTONIC NEONATE. EXPEDITED CLINICAL RESULTS CAN USUALLY BE PROVIDED IN LESS THAN ONE WEEK.

REFERENCE RANGES:

- Normal: 30 or fewer CTG repeats. Indeterminate: 30 to 50 repeats.
- 50 or more CTG repeats are full mutations, however, individuals with 50 to 100 repeats do not always manifest symptoms.
- More than 800 repeats are usually associated with congenital DM.

SAMPLE REQUIREMENTS:

- For DNA testing, 5 to 10 milliliters of blood (minimum 1 ml) in EDTA (purple top) tubes should be sent by overnight carrier at room temperature.
- Prenatal testing: two (2) confluent T25 flasks of amniocytes. CVS cells not accepted.

TEST CPT CODES:

CPT 83891 DNA extraction highly purified
CPT 83892 DNA enzymatic digestion X 2
CPT 83894 DNA separation X 2
CPT 83896 DNA nucleic acid probe
CPT 83897 DNA nucleic acid transfer
CPT 83898 DNA amplification
CPT 83912 DNA interpretation and report

Discounts from list price are available for institutional billing under contractual arrangement with the laboratory. Contact Ellen Livers at 800-447-6614 ext 7523.