

## Overview

### Method Name

Repeat Expansion Detection by PCR, Southern Blot

### NY State Available

Yes

## Specimen

### Specimen Type

Whole Blood EDTA

### Specimen Required

**Specimen Type:** Whole Blood

**Container/Tube:** Lavender top (EDTA)

**Specimen volume:** 8 mL

**Collection instructions:** Send 8 mL whole blood (lavender-top) EDTA tubes

**Note:** Collection date is required.

### Specimen Minimum Volume

6 mL

### Reject Due To

|           |    |
|-----------|----|
| Hemolysis | NA |
| Lipemia   | NA |
| Icteric   | NA |
| Other     | NA |

### Specimen Stability Information

| Specimen Type    | Temperature         | Time    | Special Container |
|------------------|---------------------|---------|-------------------|
| Whole Blood EDTA | Ambient (preferred) | 10 days |                   |
|                  | Refrigerated        | 10 days |                   |

## Clinical & Interpretive

**Clinical Information**

Detects CTG repeat expansions in the muscle protein kinase (DMPK) gene.

Typical presentation: Adults may present with a range of symptoms from cataracts to significant muscle wasting, cardiac complications, ptosis and myotonia, infants may present with severe hypotonia, skeletal deformities, developmental delay and mental retardation.

**Reference Values**

A final report will be attached in MayoAccess.

**Performance****PDF Report**

Referral

**Day(s) Performed**

Monday, Thursday

**Report Available**

14 to 32 days

**Performing Laboratory Location**

Athena Diagnostics

**Fees & Codes****Fees**

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact [Customer Service](#).

**CPT Code Information**

81234

**LOINC® Information**

| Test ID | Test Order Name | Order LOINC® Value |
|---------|-----------------|--------------------|
| FDM1    | DMPK DNA (DM1)  | Not Provided       |

| Result ID | Test Result Name | Result LOINC® Value |
|-----------|------------------|---------------------|
| Z0101     | DMPK DNA (DM1)   | Not Provided        |