

## Overview

### Method Name

Repeat Expansion Detection by PCR, Long Read Sequencing

### NY State Available

No

## Specimen

### Specimen Type

Whole Blood EDTA

### Specimen Required

**Specimen Type:** Whole Blood

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

**Specimen volume:** 8 mL

**Pediatric (0-3 years) Volume:** 2 mL (1 mL minimum)

#### Collection instructions:

1. Draw blood in a lavender-top (EDTA) tube.
2. Invert several times to mix.
3. Send ambient

**Note: Collection date is required.**

### Specimen Minimum Volume

6 mL

### Reject Due To

Hemolysis	OK
Lipemia	OK
Icterus	OK

### 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
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Z0101	DMPK DNA (DM1)	Not Provided
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