

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

Z0101	DMPK DNA (DM1)	Not Provided
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