

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