
Overview**Method Name**

Repeat Expansion Detection by PCR

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.**Reject Due To**

Hemolysis	NA
Lipemia	NA
Icteric	NA
Other	NA

Specimen Minimum Volume

6 mL

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

Performing Laboratory Location

Athena Diagnostics

Fees & Codes

Test Classification

CPT Code Information

81234

LOINC® Information

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

Result ID	Reporting Name	LOINC®
Z0101	DMPK DNA (DM1)	Not Provided