MAYO CLINIC LABORATORIES

Test Definition: FDM1

DMPK DNA Test (DM1)

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