

Overview

Method Name
Repeat Expansion Detection by PCR, Long Read Sequencing

NY State Available
No

Specimen

Specimen Type
Whole blood

Specimen Required
Specimen Type: Whole Blood
Container/Tube: Lavender top (EDTA)
Specimen volume: 8 mL
Collection instructions:
1. Draw blood in a lavender top (EDTA) tube(s).
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	Ambient (preferred)	10 days	
	Refrigerated	10 days	

Clinical & Interpretive

Clinical Information

Detects CCTG repeat expansions in the Zinc Finger Protein 9 (ZNF9) gene.

Typical Presentation: Individuals with a range of symptoms from cataracts to significant muscle wasting, cardiac complications, ptosis and myotonia.

Reference Values

A final report will be attached in MayoAccess.

Performance

PDF Report

Referral

Day(s) Performed

Monday through Friday

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

81187

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
FDMZ	CNBP DNA (DM2)	Not Provided

Result ID	Test Result Name	Result LOINC® Value
FDMZ	CNBP DNA (DM2)	Not Provided