

## Overview

### Method Name

Repeat Expansion Detection by PCR, Southern Blot

### NY State Available

Yes

## Specimen

### Specimen Type

Whole blood

### 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
Icterus	NA
Other	NA

### 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