

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.

Reject Due To

Hemolysis NA

Lipemia NA

Icterus NA

Other NA

Specimen Minimum Volume

6 mL

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

Performing Laboratory Location

Athena Diagnostics

Fees & Codes**Test Classification****CPT Code Information**

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