MAYO CLINIC LABORATORIES

Test Definition: FSCA3

SCA3-MJD (ATXN3) DNA

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:Â Send 8 mL whole blood in original tube ambient

Specimen Minimum Volume

6 mL

Reject Due To

Hemolysis	NA
Lipemia	NA
Icteric	NA
Other	NA

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole Blood EDTA	Ambient (preferred)	10 days	
	Refrigerated	10 days	

Clinical and Interpretive

Clinical Information

Hereditary ataxias are a group of heterogeneous neurological disorders predominantly characterized by balance issues, progressive incoordination of gait and limbs, speech and eye movements (2, 3). Additional neurologic and systemic symptoms may be present based on the particular subtype (3). The overall prevalence of hereditary ataxias varies depending on the population but is estimated to be 1-9:100,000 (3).

Hereditary ataxias may be broken down into subtypes based on the mode in inheritance. Autosomal dominant ataxias are a clinically diverse group of disorders that consist of spinocerebellar ataxias (SCAs), episodic ataxias (EA), and some complex forms of ataxia (3). Autosomal recessive ataxias are also clinically heterogeneous but typically characterized by areflexia, peripheral sensorimotor neuropathy (often with loss of the sense of proprioception and vibration) and non-neurologic symptoms (2, 3).

Repeat expansions of CAG in the ATXN3 gene have been associated with spinocerebellar ataxia-3 (SCA3), also known as Machado Joseph disease (MJD). This disorder is characterized by eye lid retraction and infrequent blinking leading to a â€œstaring eyesâ€• phenotype, ophthalmoparesis, and impaired speech and swallowing (4).Â Other associated features including peripheral neuropathy, dystonia, and distal amyotrophy with areflexia may be present to varying degrees depending on the size of the repeat length (larger repeats being associated with more severe phenotypes) (4, 5). Age of onset follows an inverse relationship with the number of CAG repeats in the abnormal allele (4). Cases of SCA3 may exhibit anticipation from one generation to the next (4).

Reference Values

SCA3 CAG Repeat Ranges:

Normal: < = 44

Borderline 45 - 59

Positive > = 60

Clinical Reference

1. Durr, A. (2010) Lancet Neurol 9: 885-94. (PMID: 20723845)

2. Fogel, BL, et al. (2007) Lancet Neurol 6: 245-57. (PMID: 17303531)

3. Sandford, E, et al. (2014) Genes (Basel) 5: 586-603. (PMID: 25055202)

4. Paulson, HL. (2009) J Neuroophthalmol 29: 227-37. (PMID: 19726947)

5. Franca, MC, et al. (2008) Arch Neurol 65: 525-9. (PMID: 18413477)

Performance

PDF Report

Referral

Day(s) Performed

Monday through Friday

Report Available

14 to 32 days

Performing Laboratory Location

Athena Diagnostics

Fees and 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 Regional Manager. For assistance, contact Customer Service.

Test Classification

This test was developed and its analytical performance characteristics have been determined by Athena Diagnostics. It has not been cleared or approved by the U. S. Food and Drug Administration. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.

CPT Code Information

81180

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
FSCA3	SCA3-MJD (ATXN3) DNA	Not Provided

Result ID	Test Result Name	Result LOINC Value
Z0096	SCA3-MJD (ATXN3) DNA	Not Provided