Overview

Useful For
Molecular confirmation of clinically suspected cases of sporadic or familial spinobulbar muscular atrophy (SBMA)
Presymptomatic testing for individuals with a family history of SBMA and a documented expansion in the androgen receptor (AR) gene

Testing Algorithm
See Inherited Motor Neuron Disease Testing Algorithm in Special Instructions.

Special Instructions
• Informed Consent for Genetic Testing
• Molecular Genetics: Neurology Patient Information
• Inherited Motor Neuron Disease Testing Algorithm
• Informed Consent for Genetic Testing (Spanish)

Method Name
A Polymerase Chain Reaction (PCR)-based assay is utilized to detect expansion-type mutations (CAG repeats) within the androgen receptor gene.

NY State Available
Yes

Specimen

Specimen Type
Varies

Shipping Instructions
Specimen preferred to arrive within 96 hours of draw.

Specimen Required
Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.
Specimen Type: Whole blood
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube.

Forms
1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on
Clinical Information
X-linked spinal and bulbar muscular atrophy (spinobulbar muscular atrophy; SBMA; or Kennedy disease) is characterized by onset of progressive muscle weakness, atrophy, and fasciculations typically in the fourth or fifth decade of life. Affected patients also have signs of androgen insensitivity such as gynecomastia, reduced fertility, and testicular atrophy. The clinical severity and age at onset can be quite variable, even within families. Because this is an X-linked disease, males manifest this disorder and females are generally asymptomatic carriers. However, there have been reports of female carriers who exhibit symptoms such as muscle weakness and cramping.

SBMA is caused by an expansion of the CAG trinucleotide repeat in exon 1 of the human androgen receptor (AR) gene. This trinucleotide repeat is polymorphic in the general population, with the number of repeats ranging from 11 to 34. The number of repeats found in affected individuals can range from 38 to 62. There is no consensus as to the clinical significance of alleles of 35 CAG repeats and literature suggests that alleles of 36 to 37 CAG repeats may be associated with reduced penetrance. As with other trinucleotide repeat disorders, anticipation is frequently observed and larger CAG expansions are associated with earlier onset and a more rapid clinical progression.

Reference Values
Normal alleles: 11-34 CAG repeats
Abnormal alleles: 36-62 CAG repeats
An interpretive report will be provided.

Interpretation
An interpretive report will be provided.

Cautions
For predictive testing, it is important to first document the presence of a CAG-repeat amplification in the androgen
receptor (AR) gene in an affected family member to confirm that molecular expansion is the underlying mechanism of disease in the family. We strongly recommend that patients undergoing predictive testing receive genetic counseling both prior to testing and after results are available. Predictive testing of an asymptomatic child is not recommended. Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete. Current evidence suggests that the majority of individuals with spinobulbar muscular atrophy (SBMA) have a CAG-repeat expansion. However, we cannot eliminate the possibility that another type of mutation not detected by our assay is present within the AR gene.

Clinical Reference

Performance

Method Description

PDF Report
No

Specimen Retention Time
Whole Blood: 2 weeks (if available) Extracted DNA: 3 months

Performing Laboratory Location
Rochester

Fees & Codes

Test Classification
This test was developed, and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information
81204-AR (androgen receptor)(eg, spinal and bulba muscular atrophy, Kennedy disease, X chromosome inactivation) gene analysis; characterization of alleles (eg, expanded size or methylation status)