

Spinobulbar Muscular Atrophy (Kennedy Disease), Molecular Analysis, Varies

Patient ID SA00001031	Patient Name SAMPLEREPDMGLM, VLD20150713A0292	Birth Date 1981-01-01	Gender M	Age 34
Order Number SA00001031	Client Order Number SA00001031	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 12 Jul 2015 19:23		

Spinobulbar Musc Atrophy, Kennedy's

Result Summary

MCR

NEGATIVE

Result

MCR

CAG repeat: 12 (Normal)

Interpretation

1 MCR

This result suggests that this individual is very unlikely to have spinal and bulbar muscular atrophy (SBMA). To date, alterations other than expansions within the AR gene that cause SBMA have not been identified. This result does not rule out the diagnosis of other inherited motor neuron diseases that have overlapping clinical features with SBMA.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

A PCR-based assay was utilized to detect CAG-repeat expansions within the androgen receptor gene. We estimate that the number of CAG repeats is correct to within $\pm 5\%$. Normal: 11–34; indeterminate: 35–37; Full penetrance: 38–62. An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

Test results should be interpreted in the context of clinical

findings, family history, and other laboratory data.

Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Bone Marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Specimen

MCR

WB Whole Blood

Released By

MCR

EMILY LAUER

Reason for Referral

MCR

Evaluation for a diagnosis of spinal and bulbar muscular atrophy (SBMA). Test for the presence of an expansion within the androgen receptor gene.

Received: 13 Jul 2015 20:29

Reported: 27 Jul 2015 10:24

Laboratory Notes

- 1 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 U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292