DEPARTMENT OF GENOMICS AND MOLECULAR DIAGNOSTICS

Patient Name	**Mr. PIYUSH KHADA KRUPAKAR**	**Visit ID**	**A0085756**	**Collection Date**	**05-03-2024 04:56 PM**
Age / Gender	**33 Y / MALE**	**UHID**	**-**	**Registration Date**	**06-03-2024 04:56 PM**

SCA Panel Testing

Referral Reason

I/V/O family history of SCA ,he was referred to genetic screening test for SCA panel.

Test Result

Heterozygous pathogenic (approximately 57) CAG repeats in *PPP2R2B* gene responsible for SCA12.

Interpretation

This individual shows the presence of heterozygous pathogenic CAG repeats at the *PPP2R2B* gene locus responsible for SCA12.Clinical correlation is recommended.

Methodology

End point PCR was performed from the isolated DNA to identify the triplet repeats of the SCA subtypes (1,2,3,6,7,8, and 12).

Recommendations

- Clinical correlation and Genetic counseling are recommended.

Clinical Information

Cause

SCAs are caused by mutations in one of numerous genes. See the table below for the subtypes of SCA and the associated genes tested in this panel.

Inheritance

SCAs are inherited in an autosomal dominant pattern. Children of an individual with an SCA have a 50% chance of inheriting the mutation. Anticipation is also observed in some of the SCAs. This means that as the disease passes through generations, the severity can increase and the age of onset can decrease.

Diagnosis

SCA12 is a late-onset, autosomal dominant, slowly progressive disorder. Action tremor is the usual presenting sign.Subsequent development of ataxia and hyperreflexia suggests spinocerebellar ataxia.Spinocerebellar ataxia type 12 (SCA12) is an uncommon forms of autosomal dominant spinocerebellar ataxia which is most commonly reported from India. At present it is understood that

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this form of degenerative ataxia is confined to the *Agarwal* community in India [8-9]. There is only limited data on the prevalence of SCA12 patients from non-*Agarwal* community. The cut-off for pathogenic trinucleotide repeats length in SCA12 is also unclear. SCA12 is caused by CAG repeat expansion mutation in PPP2R2B gene, that encodes Bβ, a regulatory subunit of protein phosphatase . CAG repeats number 7-28 in normal individuals and 55-78 in SCA12 patients. The mechanism by which this mutation leads to SCA12 has not been determined. The CAG expansion in *PPP2R2B* has promoter function in vitro. CAG length correlates with increased Bβ expression. There is no evidence that this CAG expansion results in polyglutamine production. Action tremor, anxiety, and depression in SCA12 have responded to usual treatments for these disorders. SCA12 may be considered in patients who present with action tremor and later develop signs of cerebellar and cortical dysfunction.

Disclaimer

- 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.
- The repeat regions other than indicated genes in the SCA panel are not are not evaluated by this assay.
- The accuracy of triplet repeat length may vary by +/- triplet for smaller normal alleles and +/-3 for expanded alleles and for SCA6 the accuracy of triplet repeat length may vary by +/- triplet for smaller normal alleles and for expanded alleles as per the guidelines (Sequeiros etal PMID: 20179742).
- Although all precautions are taken while conducting these tests, there is a standard error rate of approximate 1% in all genetic tests and this should be taken into consideration before any clinical decision. 1
- It is presumed that the specimen used to perform the test belongs to the patient specified above, such verification having been carried out at the collection level of sample.
- The exact number of repeats cannot be determined by this test.

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SCA Panel Testing

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***** End Of Report *****

Suggested clinical correlation & follow up

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