ISO 9001 : 2008 CERTIFIED LAB

KOS Diagnostic Lab
(A Unit of KOS Healthcare)

KOS™
EXCELLENCE IN HEALTHCARE & DIAGNOSTICS

Dr. Vinay Chopra
MD (Pathology & Microbiology)
Chairman & Consultant Pathologist

Dr. Yugam Chopra
MD (Pathology)
CEO & Consultant Pathologist

240111107810493

Mr. SAHIL GUPTA
..

PID NO: P33724535397653
Age: 35 Year(s) Sex: Male

Reference: DR.VINAY CHOPRA

Sample Collected At:
Dr Vinay Kumar Chopra
Dr Vinay Kumar Chopra Kos Diagnostic Lab 6349/1 Nicholson Road Ambala Cantt Hry 133001. 06-Hr 13
Processing Location:- Metropolis Healthcare Ltd,Unit No409-416,4th Floor,Commercial Building-1,Kohinoor Mall,Mumbai-70

VID: 240111107810493
Registered On:
11/11/2024 08:54 AM
Collected On:
10/11/2024 8:52AM
Reported On:
17/11/2024 10:01 AM

Test Name : **Spinocerebellar Ataxia Type 12 (SCA12) DNA Test**

Specimen : Whole Blood

Results

CAG Repeats	Interpretation
10 on both the alleles	**Not Detected - Diagnosis of SCA12 excluded**

Medical Remarks :

Method : DNA PCR

The CAG repeat expansion in *PPP2R2B* gene for diagnostic confirmation of SCA12 is determined by PCR and Capillary Electrophoresis on DNA sequencer followed by Fragment Analysis using GeneMapper software.

The accuracy of CAG repeat length may vary by +/- 1 triplet for smaller normal alleles and by +/- 3 for expanded alleles as per the reported guidelines. (Sequeiros et al; 2007)

Interpretation

CAG Repeat Length Interpretation in SCA12:

Disease	Number of CAG Repeats		
	Normal	Uncertain	Full penetrance
SCA12	4-32	40-45	51-78

Clinical implications of SCA12 DNA Test:

- Autosomal Dominant Cerebellar Ataxia (ADCA) is heterogeneous group of neurodegenerative disorders with variable expression and phenotypic overlap.
- Spinocerebellar ataxia type 12 (SCA12) is an autosomal dominant (AD) neurologic disorder in which tremor and ataxia are prominent signs.
- The world-wide frequency of SCA12 is quite low, but its prevalent is high in India. It is now believed that it is the third common cause of ADCA in India.
- The age of onset of SCA12 tremors is highly variable and ranges between 8-55 years of age, but seems to cluster primarily between third and fifth decade of life.
- SCA12 has been linked to a trinucleotide CAG repeat expansion mutation in the gene PPP2R2B.
- Individuals with PPP2R2B CAG repeats in the range of 4-32 are negative for SCA12 and those with repeats ≥51 have high probability of having SCA12.

Dr. Talat Khan
MD PATHOLOGY

Dr. Monisha Banerjee
Ph.D
Senior Consultant Molecular Pathology

NOTE:
This Sample was outsourced

240111107810493

Mr. SAHIL GUPTA
..

PID NO: P33724535397653
Age: 35 Year(s) Sex: Male

Reference: DR.VINAY CHOPRA

Sample Collected At:
Dr Vinay Kumar Chopra
Dr Vinay Kumar Chopra Kos Diagnostic Lab 6349/I Nicholson Road Ambala Cantt Hry 133001. 06-Hr 13
Processing Location:- Metropolis Healthcare Ltd,Unit No409-416,4th Floor,Commercial Building-1,Kohinoor Mall,Mumbai-70

VID: 240111107810493
Registered On:
11/11/2024 08:54 AM
Collected On:
10/11/2024 8:52AM
Reported On:
17/11/2024 10:01 AM

- Diagnosis is based on the clinical picture, familial history and ultimately on genetic testing. Differential diagnosis is broad and includes other types of SCA which may have similar features.

- Genetic counseling is recommended in symptomatic patients or those with a family history of the disorder due to known SCA mutation, and pre-symptomatic testing should be discussed in adults.

Indications for SCA12 DNA Test:

- Individuals with a family history of SCA12 who want to determine their risk.
- To differentiate individuals with SCA12 from other ataxias

Note:

This test has been developed and its performance is validated at Metropolis Healthcare Ltd.

Limitation of the Assay:

- Presence of PCR inhibitors in the sample may prevent DNA amplification. Paradoxical results may arise due selection of inappropriate specimens and contamination during specimen collection.
- Genetic Counseling is recommended for the patient and family members.

References:

- Sequeiros et al., 2010, European Journal of Human Genetics, 18, 1188–1195
- Sumathipala et al., 2013, BMC Neurology 2013, 13:39
- Spears eta al., 2010, Labmedicene, Vol. 41, No.1

-- End of Report --

Tests marked with NABL symbol are accredited by NABL vide Certificate no MC-2139

Dr. Talat Khan
MD PATHOLOGY

Dr. Monisha Banerjee
Ph.D
Senior Consultant Molecular Pathology

This Sample was outsourced

KOS Central Lab: 6349/1, Nicholson Road, Ambala Cantt -133 001, Haryana
0171-2643898, +91 99910 43898 | care@koshealthcare.com | www.koshealthcare.com