Syndrome/Diseases Name

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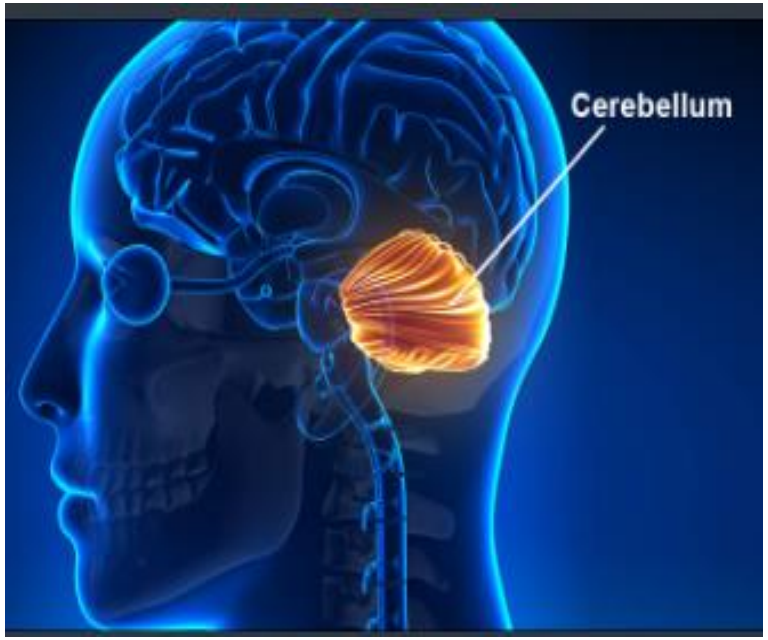
Comprehensive SCA Repeat panel

Introduction

- Spinocerebellar ataxia (SCA) is a term referring to a group of hereditary ataxias that are characterized by degenerative changes in the part of the brain related to the movement control (cerebellum), and sometimes in the spinal cord.

•There are many different types of SCA, and they are classified according to the mutated (altered) gene responsible for the specific type of SCA(SCA1 through SCA40).

•Most common type (Worldwide):SCA3



Comprehensive SCA Repeat panel

Clinical manifestation /Symptoms

- Problems with coordination and balance (ataxia)
- Uncoordinated walk
- Poor hand-eye coordination
- Abnormal speech (dysarthria)
- Involuntary back-and-forth eye movements (nystagmus)
- Vision problems
- Difficulty processing, learning, and remembering information

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Causes

- Damage, degeneration or loss of nerve cells in the part of your brain that controls muscle coordination (cerebellum), results in ataxia.
- Diseases that damage the spinal cord and peripheral nerves that connect your cerebellum to your muscles also can cause ataxia

Type of ADCA	Important SCA types	Clinical features
Type I	SCA1–SCA4, SCA8, SCA10, SCA12–SCA23, SCA25, SCA27, SCA28	Ataxia,Optic atrophy,Paralysis of extraocular muscles Dementia,Extrapyramidal symptoms
Type II	SCA7	Pigmentary maculopathy and subsequent blindness
Type III	SCA5, SCA6, SCA11, SCA26, SCA29, SCA30, and SCA31	Only ataxia; usually late onset

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Inheritance Pattern

- This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.
- An affected person usually inherits the altered gene from one affected parent. However, some people with SCA1 do not have a parent with the disorder.
- Individuals who have around 35 CAG repeats in the ATXN1 gene do not develop SCA1, but they are at risk of having children who will develop the disorder. As the gene is passed from parent to child, the size of the CAG trinucleotide repeat may lengthen into the range associated with SCA1 (40 repeats or more).

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Gene Panel

ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, BEAN1, CACNA1A, FMR1, FXN, NOP56, PPP2R2B, TBP

No of Genes : 14
Sample Type : EDTA-blood sample - 4 ml
TAT : 6 Weeks
Methodology : NGS

Genes associated with Comprehensive SCA Repeat panel

S.NO	Spinocerebellar ataxia types	Genes associated with Spinocerebellar ataxia
1	Dentatorubral-Pallidoluysian Atrophy	ATN1
2	Spinocerebellar Ataxia Type 1	ATXN1
3	Spinocerebellar Ataxia Type 10	ATXN10
4	Spinocerebellar Ataxia Type 2	ATXN2
5	Spinocerebellar Ataxia Type 3	ATXN3
6	Spinocerebellar Ataxia Type 7	ATXN7
7	ATXN8/ATXN8OS (Spinocerebellar Ataxia 8)	ATXN8, ATXN8OS

Diagnostic Tests

Management

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Reference

https://www.amboss.com/us/knowledge/Spinocerebellar_ataxia

https://ghr.nlm.nih.gov/condition/spinocerebellar-ataxia-type-1#

https://www.mayoclinic.org/diseases-conditions/ataxia/symptoms-causes/syc-20355652