Episodic ataxia panel

Introduction

- Episodic ataxia is one type of ataxia among a group of inherited diseases of the central nervous system.

•Episodic ataxia may be the result of genetic defects that lead to impairment of specific nerve fibers that carry messages to and from the brain to control movement of the body.

•There are at least eight recognized types of EA. All are hereditary, though different types are associated with different genetic causes, ages of onset, and symptoms. Types 1 and 2 are the most common.

Episodic ataxia panel

Clinical manifestation /Symptoms

•Episodic ataxia type 1 (EA1) often associated with muscle twitching or stiffness

• Episodic ataxia type 2 (EA2) often associated with involuntary jerky eye movement

• Episodic ataxia type 3 (EA3) in one Mennonite family for which the genetic defect maps to 1q42

• Episodic ataxia type 4 (EA4) with onset between 3rd to 6th decade not yet mapped

• Episodic ataxia type 5 (EA5) with seizures

• Episodic ataxia type 6 (EA6) associated with seizures,hemiplegia, migraine

• Episodic ataxia type 7 (EA7) of adult onset in one family for which the genetic defect maps to 19q13

• Episodic ataxia type 8 (EA8) of infantile onset in one family for which the genetic defect maps to1p36.13-p.34.3

• Episodic ataxia with paroxysmal choreoathetosis and spasticity

• Episodic ataxia of late onset after the 6th decade typically with no family history, slow progression, and poor responsiveness to acetazolamide

Episodic ataxia panel

Causes

- Episodic ataxia can be caused by mutations in several genes that play important roles in the nervous system
- Diseases that damage the spinal cord and peripheral nerves that connect your cerebellum to your muscles also can cause ataxia
- Although changes in chemical signaling in the brain underlie the recurrent attacks seen in people with episodic ataxia, it is unclear how mutations in these genes cause the specific features of the disorder.
- The genetic causes of episodic ataxia types 3, 4, and 7 have not been identified.

Episodic ataxia

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.

Episodic ataxia panel

Gene Panel

KCNA1,AEMK,EA1,CACNA1A,CANCNL1A4,SCA6,EA3,EA4,PATX,CACNB4,EJM6,EA5,EIG9,SLC1A3,EAAT1,EA6,EA7,UBR4

No of Genes : 18
Sample Type : EDTA-blood sample - 4 ml
TAT : 6 Weeks
Methodology : NGS

Genes associated with Episodic ataxia panel

S.NO	Episodic ataxia types	Genes associated with Episodic ataxia	Phenotype /GeneMIM Number
1	Episodic ataxia type 1/Myokymia syndrome	KCNA1,AEMK,EA1	160120/176260
2	Episodic ataxia type 2	CACNA1A,CANCNL1A4,SCA6	108500/601011
3	Episodic ataxia type 3	EA3	606554/606554
4	Episodic ataxia type 4	EA4,PATX	606552/606552
5	Episodic ataxia type 5	CACNB4,EJM6,EA5,EIG9	613855/601949
6	Episodic ataxia type 6	SLC1A3,EAAT1,EA6	612656/600111
7	Episodic ataxia type 7	EA7	611907/611907
8	Episodic ataxia type8	UBR4	616055

Diagnostic Tests

anderson
Clinical Genetics

Management

Episodic ataxia panel

Reference

https://ghr.nlm.nih.gov/condition/episodic-ataxia#inheritance

https://www.healthline.com/health/episodic-ataxia#type-2

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