Syndrome/Diseases Name

- Introduction
- Clinical manifestation /Symptoms
- Causes
- Gene Panel
- Inheritance Pattern
- Diagnostics
- Mangement
- Future Options
- Reference

Fragile X syndrome (FXS)

Introduction

- Fragile X syndrome is a genetic condition involving changes in part of the X chromosome.
- This condition causes a range of developmental problems including learning disabilities and cognitive impairment.
- It is the most common form of inherited intellectual disability in males and a significant cause of intellectual disability in females.
- Fragile X syndrome (FXS) is the most frequent form of inherited intellectual disability and is also linked to other neurologic and psychiatric disorders

Fragile X syndrome (FXS)

Clinical manifestation /Symptoms

- long face, large prominent ears, flat feet
- hyperextensible joints, especially fingers
- low muscle tone
- males may have large testes after puberty

Fragile X syndrome (FXS)

Clinical manifestation /Symptoms

- intellectual disabilities,
- ranging from mild to severe attention deficit and hyperactivity,
- especially in young children anxiety and unstable mood autistic behaviors, such as hand-flapping and not making eye contact
- sensory integration problems,
- such as hypersensitivity to loud noises or bright lights speech delay,
- with expressive language more severely affected than receptive language
- seizures (epilepsy) affect about 25% of people with Fragile X syndrome

Fragile X syndrome (FXS)

Causes

❖Mutations in the FMR1 gene cause fragile X syndrome. The FMR1 gene provides instructions for making a protein called FMRP.

❖This protein helps regulate the production of other proteins and plays a role in the development of synapses, which are specialized connections between nerve cells. Synapses are critical for relaying nerve impulses.

❖In people with fragile X syndrome, however, the CGG segment is repeated more than 200 times. The abnormally expanded CGG segment turns off (silences) the FMR1 gene, which prevents the gene from producing FMRP.

Fragile X syndrome (FXS)

Inheritance Pattern

- Fragile X syndrome is inherited in an X-linked dominant pattern. A condition is considered X-linked if the mutated gene that causes the disorder is located on the X chromosome, one of the two sex chromosomes.

- In women who carry an FMR1 gene premutation (approximately 55 to 200 CGG repeats), the repeats can expand to more than 200 repeats in their cells that develop into eggs. This means that women with a premutation (or a full mutation) have an increased risk to have a child with FXS

- People with 59 to 200 CGG repetitions are described as having a fragile X pre-mutation and may not show any obvious signs or symptoms of fragile X syndrome. The number of CGG repetitions can increase when the gene is passed from generation to generation, increasing the chance of developing the full fragile X mutation.

Fragile X syndrome (FXS)

Gene Panel

FMR1

No of Genes : 1
Sample Type : EDTA-blood sample - 4 ml
TAT : 6 Weeks
Methodology : NGS

Diagnostic Tests

anderson
Clinical Genetics

Management

Fragile X syndrome (FXS)

Reference

https://www.nichd.nih.gov/health/topics/fragilex/conditioninfo/FAQs

https://ghr.nlm.nih.gov/condition/fragile-x-syndrome#diagnosis

https://www.health.harvard.edu/a_to_z/fragile-x-syndrome-a-to-z