OPEN ACCESS Freely available online

Journal of Genetic Syndromes & Gene Therapy

Short Communication

Fragile X Syndrome: A Novel Frontier for Healthcare and Research Field

Carol Hartney*

Division of Developmental Medicine, Boston Children's Hospital, Boston, Massachusetts, USA

DESCRIPTION

Fragile X Syndrome (FXS) is a rare genetic disorder that affects approximately 1 in 4,000 males and 1 in 8,000 females worldwide. It is the most common inherited cause of intellectual disability and autism spectrum disorders. First described in the 1940s, FXS is caused by a mutation in the *FMR1* (Fragile X Mental Retardation 1) gene on the X chromosome. This article aims to explore the complexities of FXS, is symptoms, genetic mechanisms, diagnosis, and potential treatments.

FXS results from a mutation in the *FMR1* gene, which leads to a lack or reduced production of the FMRP (Fragile X Mental Retardation Protein). FMRP plays a crucial role in the development and functioning of the brain, particularly in regulating the production of other proteins needed for proper brain development. Without sufficient FMRP, various developmental processes are disrupted, leading to the characteristic features of FXS. These features include intellectual disability, learning difficulties, social and behavioral challenges, language delays, sensory sensitivities, and physical characteristics such as a long face and large ears [1].

Genetic mechanisms

The mutation responsible for FXS is an expansion of a repetitive DNA sequence known as CGG repeats within the *FMR1* gene. Normally, this sequence is repeated between 5 and 55 times. However, in individuals with FXS, the CGG repeats exceed 200. This expanded repeat causes the gene to become "methylated," resulting in the gene being switched off and the absence or reduction of FMRP. Additionally, the expanded repeats can lead to instability and fragility of the gene, hence the name "fragile X." The severity of the disorder often correlates with the number of CGG repeats [2].

Symptoms and associated challenges

The symptoms of FXS can vary widely in severity. In addition to intellectual disability, affected individuals often display social and emotional challenges, including anxiety, hyperactivity, attention deficit, and autism spectrum disorders. Language and communication difficulties, such as speech delay and echolalia, are common. Sensory sensitivities, such as sensitivity to noise or touch, are also observed. Moreover, individuals with FXS may exhibit certain physical features, including a long face, prominent ears, and hypermobile joints. It is important to note that females with FXS may have milder symptoms due to the presence of a second X chromosome that can compensate for the affected X chromosome [3].

Diagnosis and treatment

Diagnosing FXS involves genetic testing to identify the mutation in the *FMR1* gene. Prenatal testing can also be performed for families at risk. Early diagnosis is crucial for implementing appropriate interventions and therapies. While there is no cure for FXS, various interventions can help manage symptoms and improve quality of life. Educational support, speech and language therapy, occupational therapy, behavioral interventions, and medications targeting specific symptoms, such as anxiety or hyperactivity, can be beneficial. Recent research has also focused on developing targeted treatments that aim to reactivate the silenced *FMR1* gene and restore FMRP production [4].

CONCLUSION

Fragile X Syndrome is a complex genetic disorder that affects individuals in diverse ways. Understanding the underlying genetic mechanisms and associated challenges can aid in early diagnosis and appropriate intervention strategies. Ongoing research continues to shed light on potential treatment options, offering hope for improved outcomes and better quality of life for those affected by this condition. Increased awareness, support, and resources are essential for individuals with FXS and their families.

REFERENCES

1. Bach S, Shovlin S, Moriarty M, Bardoni B, Tropea D. Rett syndrome and fragile X syndrome: different etiology with common molecular dysfunctions. Front Cell Neurosci. 2021:474.

Correspondence to: Carol Hartney, Division of Developmental Medicine, Boston Children's Hospital, Boston, Massachusetts, USA, Email: carol.hartney@childrens.harvard.edu

Received: 26-May-2023, Manuscript No. JGSGT-23-24689; **Editor assigned**: 30-May-2023, PreQC No. JGSGT-23-24689 (PQ); **Reviewed:** 13-Jun-2023, QC No. JGSGT-23-24689; **Revised:** 20-Jun-2023, Manuscript No. JGSGT-23-24689 (R); **Published:** 27-Jun-2023, DOI: 10.35248/2157-7412.23.14.400.

Citation: Hartney C (2023) Fragile X Syndrome: A Novel Frontier for healthcare and Research Field. J Genet Syndr Gene Ther.14:400.

2. Cregenzán-Royo O, Brun-Gasca C, Fornieles-Deu A. Behavior problems and social competence in Fragile X Syndrome: A systematic review. Genes(Basel). 2022;13(2):280.
3. Faundez V, De Toma I, Bardoni B, Bartesaghi R, Nizetic D, de La Torre R, et al. Translating molecular advances in Down syndrome and Fragile X syndrome into therapies. Eur Neuropsychopharmacol. 2018;28(6):675-690.
4. Bourgeois JA. Neuropsychiatry of fragile X-premutation carriers with and without fragile X-associated tremor–ataxia syndrome: implications for neuropsychology. Clin Neuropsychol. 2016;30(6):913-928.