International Journal of Science and Research (IJSR)
ISSN: 2319-7064
SJIF (2022): 7.942

Review Analysis of Fragile X Syndrome: From Molecular Insights to Clinical and Ethical Challenges

Doli Deori

Thae Assam Royal Global University, Royal School of Nursing, Betkuchi, ISBT, NH - 37, Guwahati - 35, Assam, India
Email: *dolly. deori[at]email. com*

Abstract: ***Fragile X Syndrome (FXS) is the most prevalent inherited cause of intellectual disability and a significant monogenic factor in autism spectrum disorders. This X - linked condition results from the silencing of the FMR1 gene due to abnormal CGG trinucleotide repeat expansions, leading to a deficiency in fragile X mental retardation protein (FMRP). The absence of FMRP disrupts synaptic development and plasticity, causing cognitive, developmental, behavioral, and physical challenges. Advances in molecular diagnostics, such as long - read sequencing, have improved the precision of identifying these mutations, although widespread clinical implementation remains limited. Clinically, FXS is characterized by a spectrum of symptoms, including intellectual disability, hyperactivity, anxiety, and distinct physical traits like a long face and prominent ears. Males are typically more severely affected than females due to their single X chromosome. Current therapeutic approaches focus on managing symptoms through pharmacological and behavioral interventions, while emerging therapies explore gene reactivation, protein replacement, and targeting disrupted pathways like mGluR5 and PI3K/AKT/mTOR. Despite scientific progress, challenges persist in diagnosis and treatment, particularly in low - resource settings. Ethical dilemmas arise regarding genetic screening, especially for newborns, raising concerns about consent, stigma, and societal discrimination. Recent research leveraging advanced animal models and patient - derived organoids provides deeper insights into FXS pathophysiology, offering hope for targeted and potentially curative interventions. This review underscores the need for a multidisciplinary approach to address the complexities of FXS, emphasizing early diagnosis, equitable access to treatments, and ethical considerations in genetic technologies. Collaborative research and healthcare initiatives are vital to improving outcomes and quality of life for individuals with FXS and their families.***

Keywords: Fragile X Syndrome, FMR1 gene, genetic screening, neurodevelopment

1. Introduction

Fragile X Syndrome (FXS) is the most common inherited cause of intellectual disability and a leading monogenic cause of autism spectrum disorder globally. This X - linked dominant condition arises from an abnormal expansion of CGG trinucleotide repeats in the FMR1 gene, resulting in the silencing of the gene and a deficiency in fragile X messenger ribonucleoprotein (FMRP). The absence of FMRP disrupts key neural processes, leading to a spectrum of developmental, cognitive, and behavioral challenges. [1]

Clinically, FXS is characterized by intellectual deficits, behavioral issues such as hyperactivity and social anxiety, and distinct physical features, including a long face, prominent ears, and post - pubertal macroorchidism. While males with the full mutation typically experience more severe symptoms, females with a heterozygous mutation often show milder impairments due to X - chromosome inactivation patterns. Advances in molecular diagnostics, such as long - read sequencing, have enhanced the precision of identifying CGG repeat expansions, though these techniques are not yet routine in clinical practice. [2]

Emerging research also highlights the societal and healthcare disparities affecting FXS diagnosis and management globally, underscoring the need for increased awareness, genetic counseling, and treatment access in under - resourced regions

2. Objective

The study aims to:

- Summarize the current knowledge on the genetic and molecular mechanisms of FXS.
- Evaluate the clinical features, diagnosis, and treatment strategies.
- Highlight recent advances in research, including emerging therapies.
- Address challenges in diagnosis, ethical considerations, and controversies surrounding genetic screening and treatment.

3. Methodology

This review - based analysis synthesized findings from peer - reviewed articles, clinical research papers, and expert guidelines. Key areas explored include:

- Molecular and genetic underpinnings of FXS.
- Clinical manifestations and behavioral impacts.
- Advances in therapeutic approaches and the use of animal models.
- Ethical issues in genetic testing and screening programs. Data were collected from databases such as PubMed, Frontiers, and institutional publications. The study employed a comparative analysis to identify gaps in research and practice.

4. Historical Background of Fragile x Syndrome

Discovery of Fragile X Syndrome

Fragile X Syndrome (FXS) was first identified in 1943 by James Purdon Martin and Julia Bell in Britain. This pioneering study, documented in a family with multiple affected members, revealed an inherited condition characterized by intellectual disability and developmental challenges. Martin and Bell hypothesized that the disorder was sex - linked and heritable, marking the foundational understanding of FXS. Initially termed Martin–Bell Syndrome, their work laid the groundwork for subsequent research into the genetic mechanisms of the disorder. [3]

Evolution of Understanding FXS

Over the decades, research on FXS has evolved significantly. In 1991, scientists in the Netherlands identified the FMR1 gene as the cause of FXS. They discovered that a CGG trinucleotide repeat expansion within this gene leads to its silencing through methylation, disrupting the production of the fragile X messenger ribonucleoprotein (FMRP), which is crucial for neural development. This breakthrough enabled the classification of FMR1 mutations into intermediate, premutation, and full mutation types based on repeat counts .3

Advances in diagnostic technologies, including polymerase chain reaction (PCR) and long - read sequencing, have improved the accuracy of identifying the gene's status. Advocacy efforts, particularly by organizations like the FRAXA Research Foundation, have raised awareness and catalyzed research funding. In 2000, the U. S. Congress designated July 22 as National Fragile X Awareness Day to emphasize the need for ongoing study and improved support for affected individuals. [3]

This historical progression reflects a shift from descriptive observations to detailed molecular insights, offering a clearer understanding of FXS's etiology and paving the way for targeted therapeutic approaches. [3]

5. Genetics and Pathophysiology of Fragile x Syndrome

Genetics of Fragile X Syndrome

Fragile X Syndrome (FXS) arises from mutations in the FMR1 gene located on the X chromosome. The condition is most commonly caused by an abnormal expansion of the CGG trinucleotide repeat within the 5' untranslated region of the FMR1 gene. In individuals with FXS, the CGG repeat count exceeds 200 (full mutation), leading to hypermethylation of the FMR1 gene's promoter region. This epigenetic modification silences FMR1 expression, resulting in little or no production of fragile X messenger ribonucleoprotein (FMRP), a critical protein for synaptic plasticity and neuronal function. [4]

The number of CGG repeats classifies the FMR1 gene into different categories:

- Normal: Less than 45 repeats, with stable inheritance.
- Intermediate (Gray Zone): 45–54 repeats, often not associated with clinical symptoms but can expand in subsequent generations.
- Premutation: 55–200 repeats, leading to the risk of fragile X - associated conditions such as fragile X - associated tremor/ataxia syndrome (FXTAS) and fragile X - associated primary ovarian insufficiency (FXPOI).
- Full Mutation: Over 200 repeats, resulting in methylation, gene silencing, and FXS. [1]
- Inheritance is X - linked dominant. Men with the full mutation always pass the mutation to their daughters but not to their sons, while women with the mutation have a 50% chance of transmitting it to offspring of either sex.

Pathophysiology of Fragile X Syndrome

The pathophysiology of FXS is directly tied to the loss of FMRP, which is highly expressed in neurons and plays a key role in regulating synaptic function. FMRP acts as an RNA - binding protein involved in the transport and translation of specific mRNAs critical for synaptic development and plasticity. Its absence disrupts normal synaptic pruning and plasticity, leading to the over activation of pathways like the metabotropic glutamate receptor 5 (mGluR5) pathway. This over activation contributes to the cognitive, behavioral, and physical features of FXS. [4]

Additionally, FMRP regulates the expression of proteins involved in synaptic strength and maturation. Its deficiency leads to structural abnormalities in dendritic spines, including their elongation and immaturity, which correlate with intellectual disability and learning deficits. Molecular studies have also identified aberrant signaling in other pathways, such as PI3K/AKT/mTOR, that contribute to the phenotypic manifestations. [1, 2]

6. Prevalence and Incidence of Fragile x Syndrome (FXS)

Fragile X Syndrome (FXS) is the most common inherited cause of intellectual disability globally, with an estimated prevalence varying slightly across studies and populations.

- Global Prevalence: FXS affects approximately 1 in 7, 000 males and 1 in 11, 000 females. This difference reflects the genetic mechanism of the disorder, as males with one X chromosome are more affected by mutations in the FMR1 gene than females, who have two X chromosomes that can mitigate the impact of the mutation. [5, 6]
- Regional and Population Variations: Prevalence estimates can range due to differences in genetic screening, population genetics, and awareness. Studies report that in the U. S., up to 100, 000 individuals may have a full mutation, with an even greater number carrying the permutation. [5, 7]
- Carriers of Premutation: It is estimated that 1 in 150–300 women and 1 in 400–850 men carry a premutation in the FMR1 gene, which may not cause FXS but is associated with related disorders like fragile X - associated tremor/ataxia syndrome (FXTAS). [5, 7]

7. Clinical features and symptoms of fragile x syndrome (FXS)

Fragile X Syndrome (FXS) is a genetic condition that manifests a spectrum of intellectual, developmental, physical, and behavioral symptoms, which can vary based on the severity of the mutation and the sex of the individual.

Cognitive and Developmental Features

- Intellectual Disability: A hallmark of FXS, ranging from mild to severe, is more pronounced in males due to their single X chromosome.
- Learning Difficulties: Impaired learning capabilities, including challenges in language acquisition and processing. [8, 10]

Behavioral and Emotional Features

- Autism Spectrum Disorder (ASD): Nearly 30% - 50% of males and 20% - 25% of females with FXS display ASD characteristics such as poor social interactions and repetitive behaviors.
- Anxiety and Social Avoidance: High levels of anxiety, social withdrawal, and avoidance of eye contact are common. [9, 10]
- Hyperactivity and ADHD: Attention deficits and hyperactivity are frequently reported, particularly in males. [9, 11]

Physical Features

- Distinctive Facial Characteristics: Individuals may exhibit a long face, prominent forehead and chin, and large ears, which become more apparent with age.
- Macroorchidism: Males often present with enlarged testes after puberty.
- Connective Tissue Issues: Features such as hyperflexible joints, flat feet, and mitral valve prolapse are associated with FXS. [8, 9]

Neurological Symptoms

- Seizures: Approximately 10% - 20% of individuals with FXS experience seizures.
- Sensory Sensitivities: Heightened sensitivity to environmental stimuli, leading to sensory overload. [8, 11]

Additional Features

- Sleep Disorders: Common disturbances include difficulty falling or staying asleep.
- Aggression and Self - Injury: Behavioral challenges such as aggression and repetitive self - injurious behaviors are more prevalent in individuals with co - occurring ASD. [8, 9]

The presentation of symptoms can vary between males and females, with females often exhibiting milder cognitive and behavioral impairments due to the presence of a second functional X chromosome. [10, 11]

8. Diagnosis and Screening of Fragile x Syndrome (FXS):

The diagnosis and screening of Fragile X Syndrome (FXS) involve genetic testing to detect abnormalities in the FMR1 gene, specifically the expansion of CGG trinucleotide repeats.

Diagnostic Methods

DNA Testing:
The gold standard for diagnosing FXS is molecular DNA testing, which identifies CGG repeat expansions in the FMR1 gene. A full mutation (>200 repeats) confirms the diagnosis of FXS. Testing can be performed using blood or saliva samples. [12, 13]

Techniques include polymerase chain reaction (PCR) and Southern blot analysis to assess repeat size and methylation status. [13]

Prenatal Testing:
For high - risk pregnancies, prenatal diagnostic procedures such as chorionic villus sampling (CVS) and amniocentesis can identify FMR1 mutations. These methods carry a small risk of miscarriage. [12]

Carrier Screening:
Genetic carrier testing is recommended for individuals with a family history of FXS, intellectual disability, or autism. This can be performed preconception or during early pregnancy to evaluate the risk of passing on the mutation. [12, 13]

Screening Programs:
Newborn Screening: Although not part of routine newborn panels, research initiatives are exploring its feasibility to facilitate early diagnosis and intervention. [12]

Early Check Programs: Voluntary newborn screening programs, such as those in the U. S., offer early detection of FXS and related conditions. [12]

Importance of Early Diagnosis:
Early identification allows families to access interventions like speech and behavioral therapy, improving developmental outcomes. Carrier testing is crucial for reproductive planning and understanding family risks. [12, 13]

9. Neurodevelopmental and Behavioral Impact of Fragile x Syndrome (FXS):

Fragile X Syndrome (FXS) significantly impacts neurodevelopment and behavior, with symptoms varying in severity. The absence of the Fragile X Mental Retardation Protein (FMRP), caused by FMR1 gene silencing, disrupts critical pathways in brain development and function.

Neurodevelopmental Impact

1) Cognitive Impairments: FXS is a leading cause of inherited intellectual disability. Cognitive challenges

Paper ID: SR241208112548 DOI: https://dx.doi.org/10.21275/SR241208112548

range from mild to severe, with deficits in attention, memory, and executive functioning. [14, 15]

2) Affected individuals often exhibit learning delays and struggle with adaptive skills needed for daily living. [15]
3) Sensory Processing Issues: Heightened sensitivity to stimuli, often leading to sensory overload, is common. This contributes to anxiety and difficulties in social and environmental interactions. [16]
4) Motor Development: Delayed motor milestones and coordination difficulties are often observed, complicating physical activities and fine motor skills. [16]

Behavioral Impact

1) Autism Spectrum Disorder (ASD): About 30% - 50% of males and 20% - 25% of females with FXS display behaviors consistent with ASD, including social deficits, repetitive behaviors, and communication challenges. [14, 16]
2) Anxiety and Emotional Regulation: Anxiety disorders are highly prevalent, manifesting as social withdrawal, shyness, and heightened fear responses. Emotional dysregulation often exacerbates behavioral challenges. [14, 16]
3) Behavioral Inflexibility and Aggression: Individuals with FXS frequently exhibit repetitive questioning, difficulty adapting to changes, and episodes of irritability or aggression. These behaviors often stem from underlying frustration or sensory discomfort. [14, 15]
4) Hyperactivity and Attention Deficits: Hyperactivity and attention deficit hyperactivity disorder (ADHD) symptoms are particularly prominent in males with FXS. [15]

Long - term Implications

Behavioral challenges and cognitive delays in FXS can affect educational attainment, employment opportunities, and independence in adulthood. Early interventions targeting these domains significantly improve outcomes. [14, 16]

10. Management and Therapeutic Interventions for Fragile x Syndrome (FXS):

1) **Pharmacological Treatment: Pharmacological management focuses on alleviating specific symptoms rather than curing the condition:**

- ADHD and Hyperactivity: Medications like stimulants (methylphenidate) or non - stimulants (guanfacine) are used to manage hyperactivity and impulsivity.
- Anxiety and Emotional Regulation: Selective serotonin reuptake inhibitors (SSRIs) like fluoxetine are often prescribed for anxiety and mood dysregulation.
- Behavioral Symptoms: Atypical antipsychotics, such as aripiprazole and risperidone, are used for aggression and self - injurious behaviors. [17, 18]

2) **Behavioral and Educational Interventions**

- Behavioral Therapies: Applied Behavior Analysis (ABA) and other behavioral techniques help manage challenging behaviors, promote social skills, and improve adaptive functioning. Tailored interventions address triggers like sensory sensitivities and communication difficulties. [17, 18]
- Educational Support: Individualized education programs (IEPs) are essential for addressing learning needs. Speech and language therapy targets communication challenges, while occupational therapy aids in improving sensory integration and motor skills. [19]

3) **Emerging Therapies**

- Gene Therapy: Researchers are exploring interventions targeting the reactivation of the silenced FMR1 gene. Experimental approaches like CRISPR are being studied for potential application.
- Pharmacological Innovations: Drugs like metformin and lovastatin are under investigation for their roles in mitigating synaptic dysfunction and improving cognitive outcomes. [17, 18]
- Neuroplasticity - Enhancing Drugs: Medications targeting mGluR5 pathways show promise in preclinical trials to address neurodevelopmental deficits. [18]

4) **Role of Early Intervention and Supportive Therapies:**

- Early diagnosis is crucial for initiating interventions during critical developmental windows. Supportive therapies, such as:
- Physical Therapy: To enhance motor skills and coordination.
- Speech and Language Therapy: To support communication development.
- Parent Training and Support: Families benefit from training to manage behaviors and navigate educational resources effectively. [17, 19]

11. Recent Advances in Research on Fragile x Syndrome (FXS):

1) **Advances in Molecular and Genetic Research: Recent studies have expanded our understanding of the molecular underpinnings of FXS:**

- Epigenetic Mechanisms: Research has highlighted the role of epigenetic silencing in the FMR1 gene, not only affecting FMRP production but also disrupting a broader network of genes. Techniques such as CRISPR have demonstrated potential for reducing the CGG repeat expansion in the FMR1 gene, reactivating gene expression, and reversing some symptoms. [20, 21]
- Protein Replacement Strategies: Innovative approaches like using mRNA technology or truncated FMRP have shown promise in restoring protein levels, which are essential for synaptic function. This technology might bypass the genetic mutation itself. [22, 24]
- GSK3 Inhibitors: A selective inhibitor targeting the GSK3 alpha isoform has shown success in preclinical trials by reducing synaptic abnormalities and improving cognitive outcomes without the side effects observed in earlier mGluR5 inhibitors. [24]

2) **New Insights from Animal and Cellular Models**

- Advanced Animal Models: While traditional mouse models have limitations, new "knock - in" models better mimic human FMR1 gene silencing. These models are crucial for testing therapies like CRISPR and gene reactivation. [22, 23]

- Organoid Research: Patient - derived brain organoids (mini - brains) and stem cell models now allow researchers to study FXS - specific neuronal defects, such as altered synaptic connectivity and neurodevelopmental trajectories. [23]
- BREACH Mechanism Discovery: The identification of "BREACHes" (Beacons of Repeat Expansion Anchoring Contacting Heterochromatin) reveals how large heterochromatin domains disrupt critical neuronal genes, offering new therapeutic targets beyond the FMR1 gene. [21, 24]

12. Challenges and Controversies in Fragile x Syndrome (FXS):

a) Limitations in Current Diagnosis and Treatment

- Diagnostic Delays: Many individuals with FXS are diagnosed late due to the variability in symptoms and overlap with other conditions like autism spectrum disorder (ASD). Current diagnostic tools rely heavily on molecular testing, which is not universally accessible, particularly in low - resource settings. [25, 26]
- Treatment Gaps: FXS treatments focus on managing symptoms, such as anxiety or hyperactivity, rather than addressing the root genetic cause. Many pharmacological interventions, like mGluR5 inhibitors, have failed in clinical trials due to side effects and limited efficacy. [25, 27]

b) Ethical Considerations in Genetic Screening

- Newborn Screening: Including FXS in newborn screening raises ethical concerns. Early identification of an "untreatable" condition can cause parental anxiety, influence bonding, and create societal stigma. [25, 26]
- Voluntary Consent: Screening programs, especially in prenatal and newborn phases, must ensure informed and voluntary consent. Families may face dilemmas about continuing pregnancies when FXS is diagnosed prenatally. [26]
- Potential Discrimination: Knowledge of carrier status or genetic predisposition could lead to discrimination in employment or insurance, raising broader ethical concerns. [25]

c) Controversial Therapies

- Unproven Interventions: Some alternative therapies, such as dietary supplements or experimental behavioral methods, lack robust scientific validation. These can divert families from evidence - based approaches and lead to financial and emotional burdens. [25, 26]
- Gene Editing Technologies: Emerging techniques like CRISPR present ethical challenges, particularly regarding long - term impacts, access equity, and the potential for unintended genetic modifications. [26]

13. Summary

Fragile X Syndrome is caused by the silencing of the FMR1 gene due to CGG trinucleotide repeat expansions. The absence of FMRP affects neuronal development, leading to intellectual disability, behavioral issues, and physical anomalies. Advances in molecular genetics, such as CRISPR technology, and innovative therapies like GSK3 inhibitors, offer hope for improved outcomes. Despite these advances, limitations in diagnosis, accessibility of treatments, and ethical dilemmas in genetic screening remain significant hurdles. Emerging research on epigenetics and patient - derived organoids further expands the understanding of FXS.

14. Conclusion

This analysis underscores the complexity of managing FXS, requiring a multidisciplinary approach. While recent research offers promising therapeutic avenues, addressing diagnostic challenges and ensuring ethical implementation of genetic technologies are critical for advancing care. Collaborative efforts in research, policy, and clinical practice are vital to improve the quality of life for individuals with FXS and their families.

Conflict of Interest: Nil

Acknowledgment:
I thank the higher authorities of The Assam Royal Global University for Support and encouragement.

References

[1] Ciobanu, C., Nucă, I., Popescu, R., Antoci, L., Caba, L., Ivanov, A. V., Cojocaru, K., Rusu, C., Mihai, C., & Pânzaru, M. (2022). Narrative Review: Update on the Molecular Diagnosis of Fragile X Syndrome. *International Journal of Molecular Sciences*, *24* (11), 9206. https: //doi. org/10.3390/ijms24119206
[2] Mbachu, C. N., Mbachu, I. I., & Hagerman, R. (2024). A Comprehensive Review of Fragile X Syndrome and Fragile X Premutation Associated Conditions in Africa. *Genes*, *15* (6), 683. https: //doi. org/10.3390/genes15060683
[3] Centers for Disease Control and Prevention. Fragile X syndrome. Available from: https: //www.cdc. gov/ncbddd/fxs/data. html
[4] Xie, L., Li, H., Xiao, M., Chen, N., Zang, X., Liu, Y., Ye, H., & Tang, C. (2024). Epigenetic insights into Fragile X Syndrome. *Frontiers in Cell and Developmental Biology*, *12*, 1432444. https: //doi. org/10.3389/fcell.2024.1432444
[5] Fragile X Syndrome: Understanding Prevalence and Diagnosis. Fragile X Association. Available from: https: //www.fragilex. org
[6] Centers for Disease Control and Prevention (CDC). Data and Statistics on Fragile X Syndrome.2024. Available from: https: //www.cdc. gov
[7] Hagerman RJ, et al. Fragile X Syndrome: Causes, Diagnosis, Mechanisms, and Therapeutics. J Clin Invest.2012; 122 (12): 4314 - 4322. Available from: https: //doi. org/10.1172/JCI63141
[8] Cleveland Clinic. Fragile X Syndrome (FXS): What it is, symptoms, and treatment. Cleveland Clinic.2024. Available from: https: //my. clevelandclinic. org.
[9] Frontiers in Genetics. Epigenetic insights into Fragile X Syndrome. Frontiers.2024. Available from: https: //www.frontiersin. org.
[10] Centers for Disease Control and Prevention (CDC). Fragile X Syndrome (FXS): Symptoms and related

concerns. CDC.2024. Available from: https: //www.cdc. gov.
[11] UC Davis MIND Institute. Fragile X Research and Treatment Program. UC Davis Health.2024. Available from: https: //health. ucdavis. edu.
[12] Fragile X Association of Australia. Testing and Screening for Fragile X Syndrome.2024. Available from: https: //www.fragilex. org. au
[13] Centers for Disease Control and Prevention (CDC). Testing and Diagnosis of Fragile X Syndrome. CDC.2024. Available from: https: //www.cdc. gov.
[14] Frontiers. Epigenetic insights into Fragile X Syndrome. Frontiers.2023. Available from: https: //www.frontiersin. org
[15] Brain Communications. Neurodevelopmental effects of Fragile X Syndrome. Brain Communications.2024. Available from: https: //academic. oup. com/braincomms/article/6/1/fcad353/7517738
[16] National Fragile X Foundation. Behavior and Fragile X Syndrome. FragileX. org.2023. Available from: https: //fragilex. org
[17] National Fragile X Foundation. Treatment and intervention recommendations for Fragile X. FragileX. org.2024. Available from: https: //fragilex. org
[18] NICHD. What are the treatments for Fragile X Syndrome? Eunice Kennedy Shriver National Institute of Child Health and Human Development.2024. Available from: https: //www.nichd. nih. gov
[19] National Fragile X Foundation. Behavioral challenges in Fragile X Syndrome. FragileX. org.2023. Available from: https: //fragilex. org
[20] National Institute of Mental Health. Researchers expand understanding of genetic mechanisms underlying Fragile X Syndrome. NIMH.2024. Available from: https: //www.nimh. nih. gov.
[21] Penn Medicine. Researchers uncover unexpected molecular patterns in Fragile X Syndrome. Penn Medicine.2024. Available from: https: //www.pennmedicine. org.
[22] FRAXA Research Foundation. Breakthrough discoveries in Fragile X research: insights from Banbury Meeting on curative therapies. FRAXA.2024. Available from: https: //www.fraxa. org.
[23] Gordon Research Conferences. Insights from human cellular models of Fragile X and ASD. GRC.2024. Available from: https: //www.grc. org
[24] MIT News. Scientists find a new way to reverse symptoms of Fragile X. Massachusetts Institute of Technology.2024. Available from: https: //news. mit. edu
[25] American Academy of Pediatrics. Ethical, legal, and social concerns about expanded newborn screening: Fragile X syndrome as a prototype. Pediatrics.2024. Available from: https: //publications. aap. org.
[26] RTI International. Ethical, legal, and social concerns about expanded newborn screening: Fragile X syndrome and emerging issues. RTI.2024. Available from: https: //www.rti. org.
[27] MIT News. Scientists find a new way to reverse symptoms of Fragile X. Massachusetts Institute of Technology.2024. Available from: https: //news. mit. edu

Author Profile

Doli Deori, a seasoned nursing professional, holds a B. Sc. in Nursing and an M. Sc. in Nursing. Currently, she is an Associate Professor at The Assam Royal Global University, where she inspires future nurses. Her passion for knowledge is evident in her numerous research and review articles published in esteemed journals, along with her authorship of six books on various nursing subjects. Her dedication to excellence has been recognized with the "Best Associate Professor" award from the UIJIR Academic and Research Foundation. Notably, she has also contributed to innovation by securing a patent. As she pursues her Ph. D., Doli remains committed to pushing the boundaries of nursing knowledge.