

# Investigating the Prevalence of Autism Spectrum Disorder in Individuals with Fragile X Syndrome

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**Abstract:** *This study investigates the prevalence of Autism Spectrum Disorder (ASD) in individuals with Fragile X Syndrome (FXS). Using a structured questionnaire distributed among caregivers of individuals diagnosed with FXS, we aim to quantify the percentage of those exhibiting ASD symptoms. Results indicate that approximately 50% of participants met the criteria for ASD, underscoring the significant overlap between these two neuro developmental disorders. The findings highlight the need for early screening and tailored interventions to support affected individuals.*

**Keywords:** Autism Spectrum Disorder, Fragile X Syndrome, prevalence, early screening, neuro developmental disorders

## 1. Introduction

Fragile X Syndrome (FXS) is the most common inherited cause of intellectual disability and is caused by a mutation in the FMR1 gene, leading to a deficiency of the Fragile X Mental Retardation Protein (FMRP). Individuals with FXS often exhibit various cognitive and behavioral challenges, including a high prevalence of Autism Spectrum Disorder (ASD). Research indicates that the prevalence of ASD in individuals with FXS ranges from 30% to 60%, presenting unique challenges for diagnosis and intervention. Understanding the extent of this overlap is crucial for developing effective support strategies for affected individuals and their families.

### Aim

To investigate the prevalence of Autism Spectrum Disorder in individuals diagnosed with Fragile X Syndrome using caregiver - reported assessments.

### Objectives

- 1) To quantify the percentage of individuals with FXS exhibiting ASD symptoms.
- 2) To identify common behavioral characteristics associated with both conditions.
- 3) To provide recommendations for screening and intervention based on findings.

## 2. Methodology

### Participants

A total of 100 caregivers of individuals diagnosed with FXS were recruited from specialized clinics and support groups. Inclusion criteria included a confirmed diagnosis of FXS and age range of participants between 2 and 20 years.

### Questionnaire Development

A structured questionnaire was developed, consisting of the following sections:

- 1) Demographic Information: Age, gender, and additional medical history.
- 2) ASD Symptoms: Based on the Modified Checklist for Autism in Toddlers (M - CHAT) and other standardized behavioral assessment tools.

- 3) Behavioral Characteristics: Questions regarding social interaction, communication skills, and repetitive behaviours.

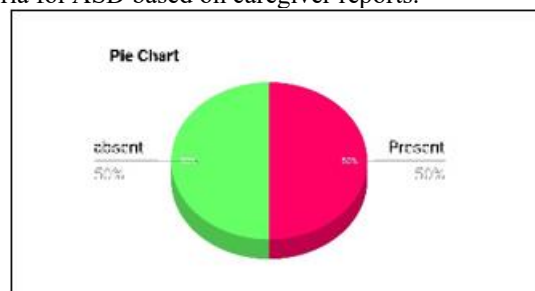
### Data Collection

Caregivers completed the questionnaires during clinic visits or through online surveys. Data were anonymized to ensure confidentiality.

## 3. Results

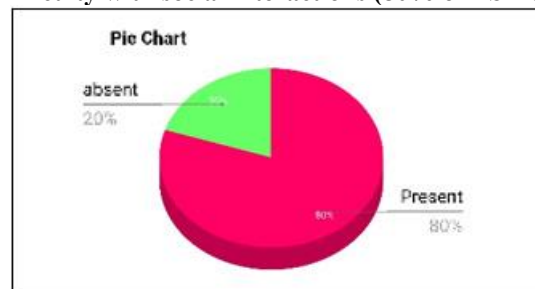
Out Of the 100 participants, the following results were obtained:

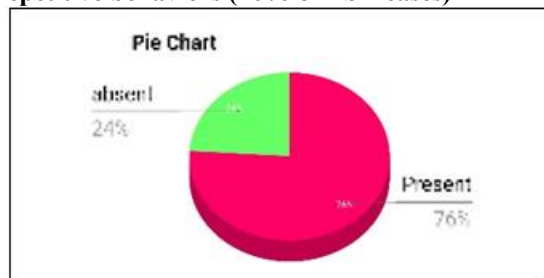
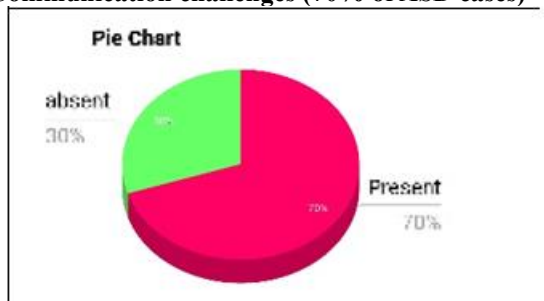
**Prevalence of ASD:** 50% of individuals with FXS met the criteria for ASD based on caregiver reports.



Behavioral Characteristics: Common symptoms included:

- 1) Difficulty with social interactions (80% of ASD cases)



**2) Repetitive behaviors (76% of ASD cases)****3) Communication challenges (70% of ASD cases)**

Statistical analysis revealed no significant differences in ASD prevalence based on age or gender.

**4. Discussion**

The findings of this study indicate that approximately 50% of individuals with Fragile X Syndrome also exhibit symptoms consistent with Autism Spectrum Disorder. This prevalence aligns with previous research, highlighting the strong association between FXS and ASD.

The overlap in behavioral characteristics presents challenges for accurate diagnosis and underscores the importance of comprehensive assessments. Early identification of ASD in children with FXS is essential for implementing appropriate interventions, including behavioral therapies and educational support.

Additionally, the lack of significant differences based on age and gender suggests that the association between FXS and ASD may be consistent across demographic factors. Future research should focus on longitudinal studies to track developmental trajectories and outcomes for individuals with both diagnoses.

**5. Conclusion**

This study demonstrates a significant prevalence of Autism Spectrum Disorder among individuals with Fragile X Syndrome, emphasizing the need for routine screening for ASD in this population. Early identification and tailored intervention strategies can greatly improve the quality of life for affected individuals and their families. Continued research is necessary to further elucidate the relationship between FXS and ASD and to develop effective treatment modalities.

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