Profile of Social Communication in Children with Fragile X Syndrome

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Background

Fragile X Syndrome (FXS) is a genetic condition that is the most common cause of inherited intellectual disability and is the most common known genetic cause of autism spectrum disorder

FXS has a higher prevalence in males and typically has a more severe presentation in males.

Features can include:
Behavioral: ADD/ADHD, ASD, sensory disorders
Speech: Phonological impairments, dysfluent, rapid rate
Language: Grammar and vocabulary delays
Pragmatics: Poor topic maintenance, difficulty initiating conversation, difficulty with abstract language [1]
Physical: large ears, long face, high-arched palate

According to the Fragile X Online Registry with Accessible Database (FORWARD), 40% of individuals in the database with FXS are diagnosed with ASD

"Most common known single gene cause of ASD" [2]

The purpose of this retrospective, descriptive review of children at the Rush University Fragile X Clinic is to evaluate the profile of social communication skills in children with FXS to look for skills that are more consistent across children in this population versus those that are more variable.

Methods

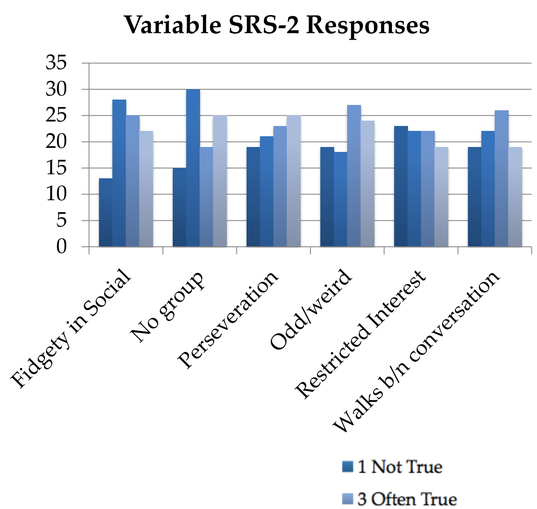
This review was completed via chart review of files present in the Rush University Fragile X Clinic, a clinic participating in FORWARD.

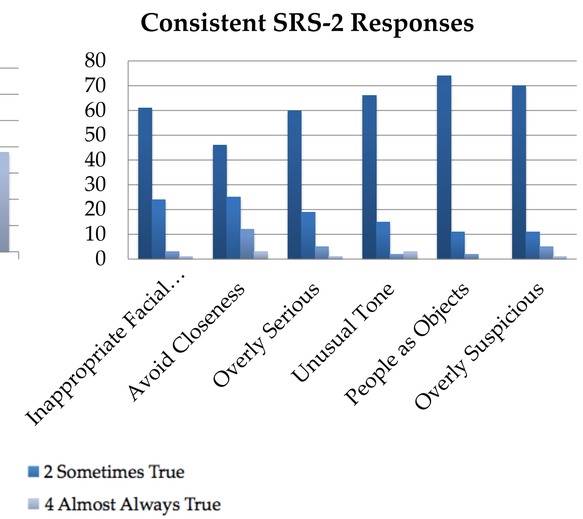
- Sample size: Total *N*= 84
  - Age range: 2;9 to 15;0
  - Majority of children were verbal
- Data obtained from the following parent questionnaires:
  - Social Communication Questionnaire: autism screener
  - Social Responsiveness Scale-2

Results

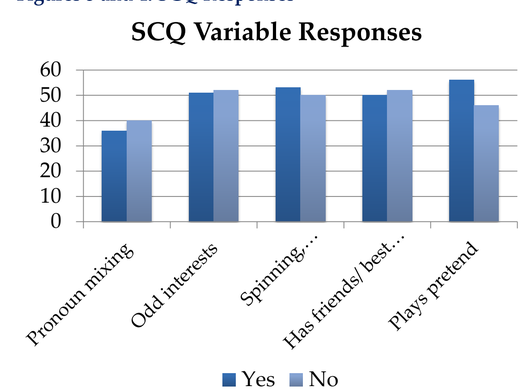
Responses on the SRS-2 and the SCQ were recorded and compiled to generate the total number of each type of response for each item. The figures below depict both the most variable and most consistent responses for each questionnaire.

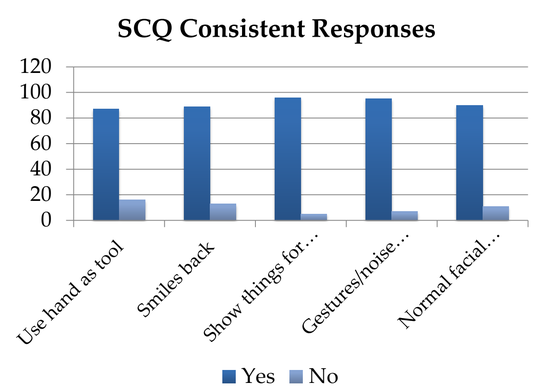
Figures 1 and 2: SRS-2 Responses





Figures 3 and 4: SCQ Responses





Conclusion

Consistent characteristics across both measures included:

- Appropriate facial expressions
- Social reciprocity via smile and showing objects to caregiver to gain attention
- Communicating via gesture or sounds to get help
- Using caregiver as object

However, there was a wide amount of variability on many items, including:

- Perseveration
- Social group or friends
- Odd or restricted interests
- Language impairments including pronoun mixing

This indicates that children with FXS may have varying presentations; therefore, thorough evaluation is necessary to determine the child's needs. Co-morbidity of ASD should also be considered when evaluating a child's pragmatic skills.

Limitations & Future Directions

This data represents only the portion of children with FXS whose files were accessible and who had the questionnaires present in their charts. The information represents preliminary trends only and has not been statistically analyzed; therefore, future statistical analysis is necessary.

References

Autism Spectrum Disorder and Fragile X Syndrome (n.d.). Retrieved February 19, 2017, from https://fragilex.org/

Roberts, J., Hennon, E. A., & Anderson, K. (2003). Fragile X syndrome and speech and language. *The ASHA Leader*, *8*(19), 6-7.

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