

# Current Symptoms of Autism in Fragile X Syndrome

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# Symptoms of Autism in FXS

- As many as 90% of males with FXS display symptoms of autism
- Major source of variability within the FXS phenotype
- What is the overlap in symptom profiles between idiopathic ASD and FXS?

# Symptoms of Autism in FXS

- In general, individuals with comorbid FXS and ASD have lower nonverbal IQ scores than individuals with FXS only
- Nonverbal IQ is negatively associated with autism symptoms in FXS but not in ASD
  - FXS  $r_{38} = -.532$
  - ASD  $r_{46} = -.147$
- If an individual with FXS meets diagnostic criteria for ASD, we do not know if the specific symptoms that lead to the diagnosis are the same across syndromes
- Is autism in FXS...
  - An independent comorbid condition?
  - Result of lower IQ scores?

# Research Question

- Do current symptoms of autism differ in boys with FXS or idiopathic ASD

# Participants (N = 84, 4 – 11 years)

|                          | <b>Fragile X<br/>Syndrome<br/>(n=38)</b> | <b>Autism Spectrum<br/>Disorder<br/>(n=46)</b> |
|--------------------------|--|--|
| <b>Chronological Age</b> | 7.23 years ( <i>SD</i> = 2.05)           | 7.53 years ( <i>SD</i> = 1.96)                 |
| <b>Nonverbal IQ</b>      | 59.71 ( <i>SD</i> = 13.79) *             | 74.61 ( <i>SD</i> = 19.93)                     |
| <b>Autism Severity</b>   | 6.21 (Range 2-10) *                      | 8.11 (Range 4-10)                              |

\* $p < .01$

# Participants with ASD

- Genetic testing to rule out fragile X syndrome prior to enrollment in current study
- Examined by physician during study visit to rule out other genetic conditions that could account for autistic symptomatology
- Met criteria for ASD diagnosis using revised algorithms
  - ADI-R: Risi et al. 2006
  - ADOS: Gotham et al., 2009

# Matched subgroups

- Participants were well-matched on chronological age, but not adequately matched on nonverbal IQ or severity of autism symptoms
- To facilitate a between-group comparison, we created subgroups matched on these characteristics
  - Participants matched pairwise for CA and Nonverbal IQ
    - n = 29 per group
    - Autism Severity (ASD>FXS)
  - Verbal participants matched pairwise for CA and Nonverbal IQ
    - N=21 per group
    - Autism Severity (ASD>FXS)
  - Participants matched pairwise for CA and Autism Severity
    - n = 25 per group
    - Nonverbal IQ (ASD>FXS)

# Assessments

- Autism Diagnostic Interview - Revised
  - Parent interview conducted by trained examiner
  - One of two “gold standard” instruments for diagnosing an autism spectrum disorder
  - Three domains – Social Reciprocity, Communication, Restricted and Repetitive Behaviors
  - Dependent measure: Questions querying current symptoms of autism



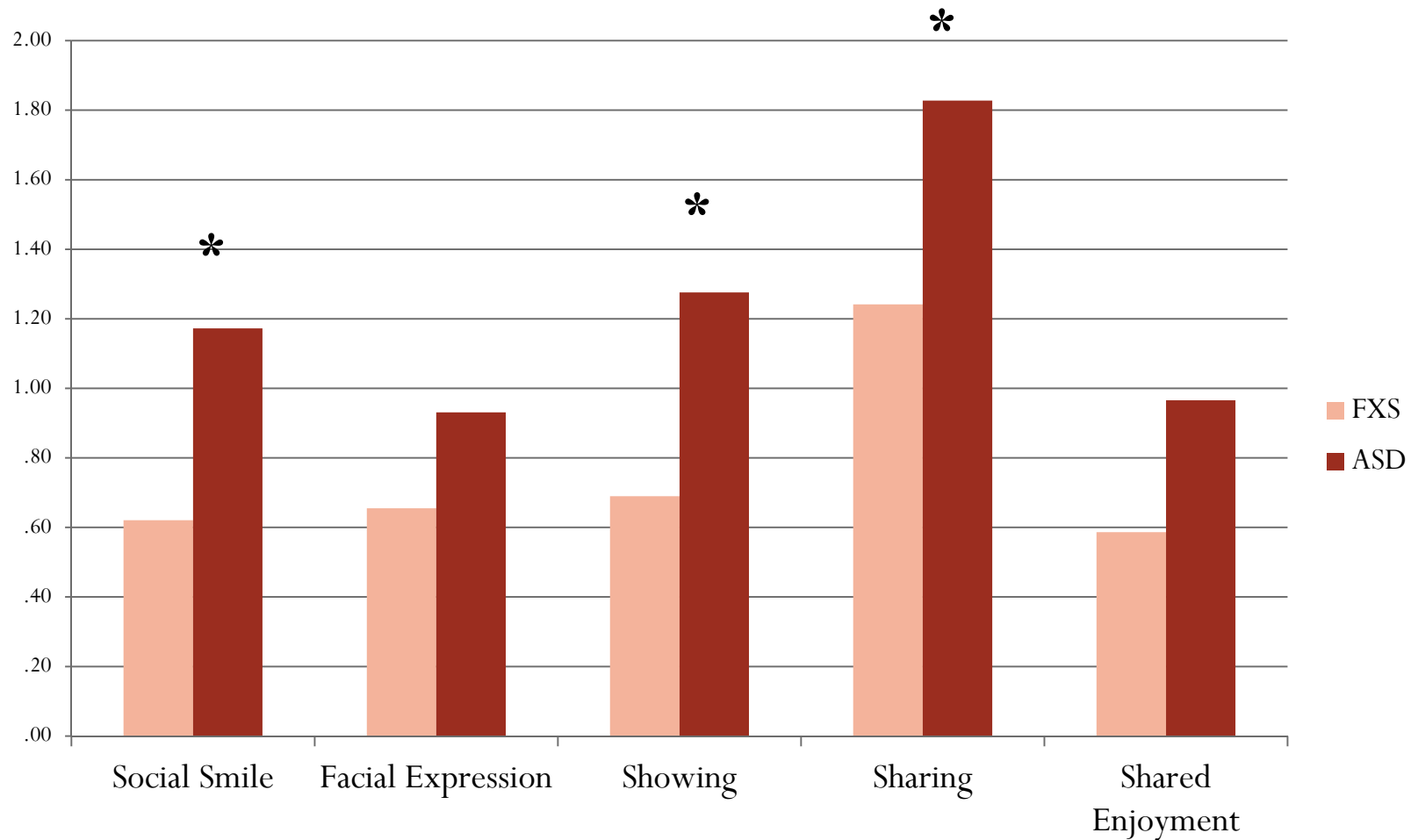
# Assessments

- Autism symptom severity
  - Continuous metric derived from ADOS
- Nonverbal IQ
  - Brief IQ (4 subtests from Leiter International Performance Scales)
- Language Ability
  - Receptive Vocabulary: PPVT-4
    - Standard score
  - Expressive Vocabulary: EVT-2
    - Standard score

# Results: Social Reciprocity

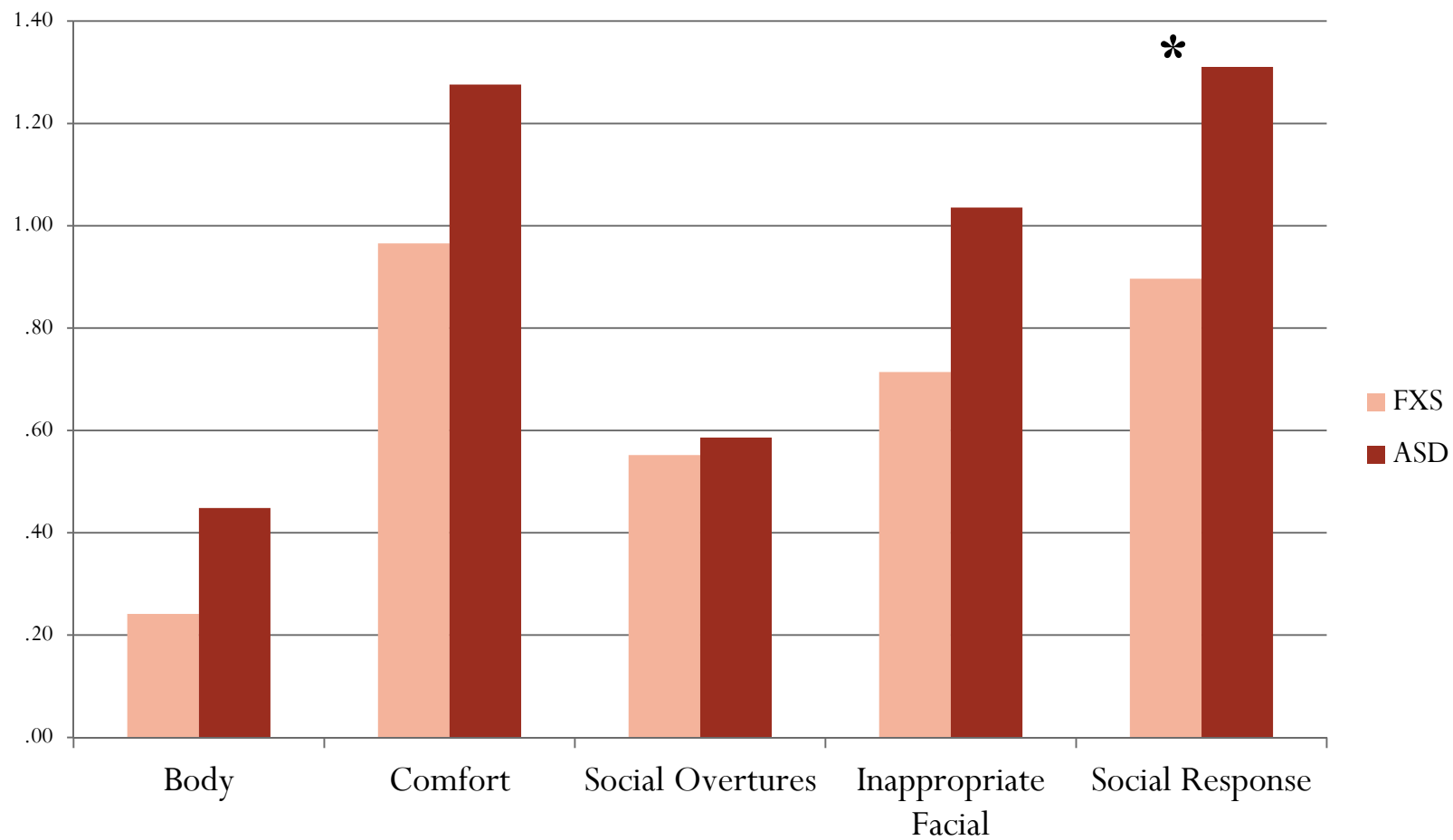
- **Boys with FXS less impaired**
- **Nonverbal IQ match**
  - Social Smiling
  - Showing and Directing Attention
  - Offering to Share
  - Appropriateness of Social Response
- **Autism Severity Match**
  - Offering to Share

# Nonverbal Behaviors/ Shared Enjoyment



Participants Matched on NVIQ

# Social Emotional Reciprocity

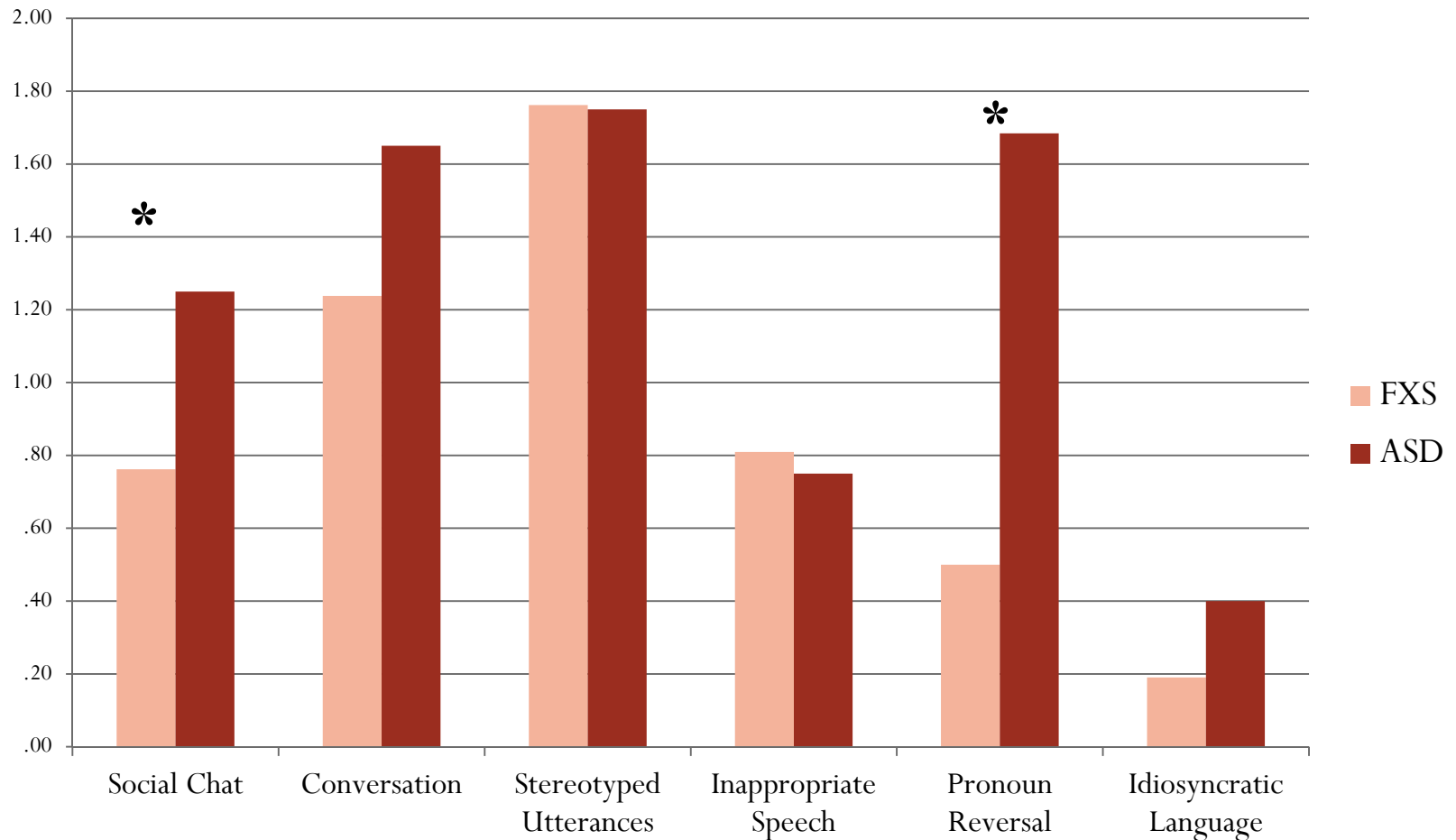


Participants Matched on NVIQ

# Results: Communication

- Differences for verbal participants in subgroup matched on NVIQ
- Boys with FXS less impaired
  - Social Verbalization / Chat
  - Pronominal Reversal

# Initiate and Sustain Conversation/ Stereotyped Speech

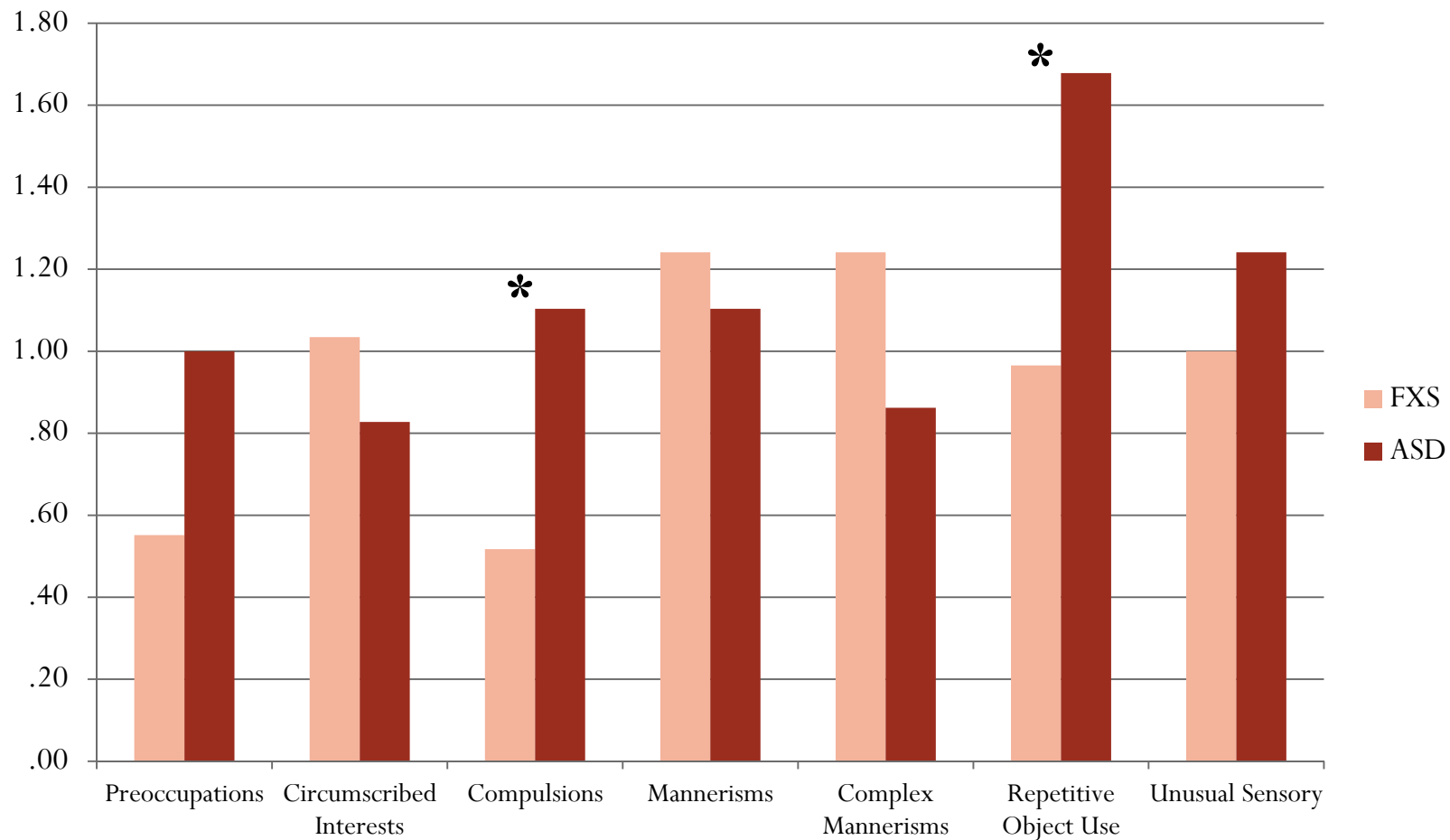


Verbal Participants Matched on NVIQ

# Results: Restricted Interests/Repetitive Behavior

- Boys with FXS less impaired than both matched subgroups...with one exception
- **Nonverbal IQ match**
  - Repetitive Use of Objects
  - Compulsions and Rituals
- **Autism Severity Match**
  - Unusual Preoccupations
  - Compulsions and Rituals
  - **Complex Mannerisms\***

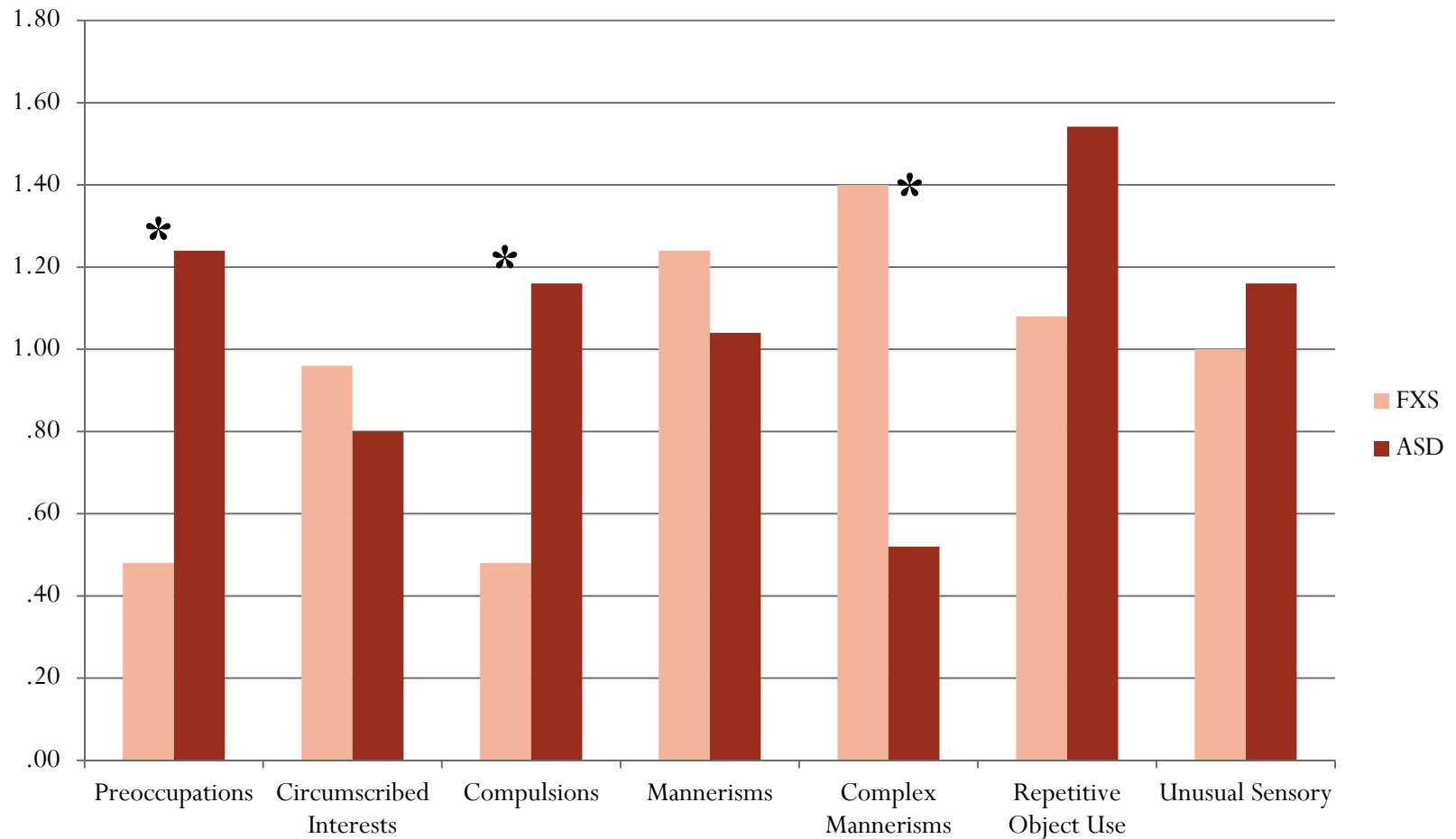
# Restricted Interests/ Repetitive Behaviors



Participants Matched on NVIQ



# Restricted Interests/ Repetitive Behaviors



Participants Matched on Autism Severity

# Summary of Findings

- There are differences in symptoms of autism between boys with FXS and boys with ASD
  - Even when matched on Nonverbal IQ or Autism Severity
  - Boys with FXS were less impaired than boys with ASD (with one exception)
- **Conclusion:** Characteristics of autism in FXS may represent a different cognitive mechanism than in idiopathic autism
- **Future Directions:**
  - Examine between group differences in concurrent correlations between other child characteristics and symptoms of autism
  - Between groups: Compare current behaviors measured with direct observation using the ADOS
  - Within groups: Compare symptoms of autism measured directly compared symptoms reported by parent informant

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