



## FXS and ASD

Fragile X syndrome (FXS) has been referred to as the leading known genetic form of autism, primarily due to reports of high co-morbidity with autism spectrum disorders (ASD).

Reports of co-morbidity of ASD diagnoses range considerably, with a common estimate of 50% (Abbeduto, Mcduffie, & Thurman, 2014), with males being diagnosed at a higher frequency than females.

## DSM-IV and DSM-V

### DSM-IV Autistic Disorder

At least 6 symptoms  
At least 2 **Social Interaction** symptoms (e.g. nonverbal behaviors, peer relationships, shared enjoyment, reciprocity)  
At least 1 **Communication** symptom (e.g. delay or lack of speech, ability to maintain conversation, repetitive speech, lack of imitation/make-believe play)  
At least 1 **Restricted, Repetitive Behavior** symptom (e.g. preoccupations, rigid routines, repetitive movements)  
Before age 3

### DSM-5 Autism Spectrum Disorder

Must have all three from **Social Communication/Interaction** domain  
Deficits in social emotional  
Deficits in communicative behaviors  
Deficits in relatedness  
AND  
At least two from **Restricted, Repetitive Behaviors or Interests**  
Stereotyped speech, movement, use of objects  
Rigidity with routines  
Fixated interests  
Hyper or hypo-responsiveness to sensory input

An initial look at how symptoms of ASD in FXS align with *DSM-IV* vs. *DSM-V* criteria using parent-report only suggested a decrease in eligibility for co-morbid ASD when *DSM-V* criteria are used (Wheeler et al., 2014).

A recent analysis of this data (DeRamus et al., 2016) demonstrated that more individuals with FXS met criteria for ASD on the *ADOS-2* when mapped to the diagnostic framework of *DSM-IV* than that of *DSM-V*.

## Methods

The symptoms of ASD in 143 individuals with FXS (72 male and 71 female) from the *Autism Diagnostic Observation Schedule, Second Edition (ADOS-2)* were examined with regards to the criteria of SCI and RRBI.

In addition, frequencies of those meeting criteria for ASD based on a relaxed diagnostic criteria of the *DSM-V* standards were examined.

Relaxed version of *DSM-V* criteria: reducing the number of qualifying symptoms by one in social communication and interaction (SCI) and/or repetitive and restricted behaviors and interests (RRBI) criteria.

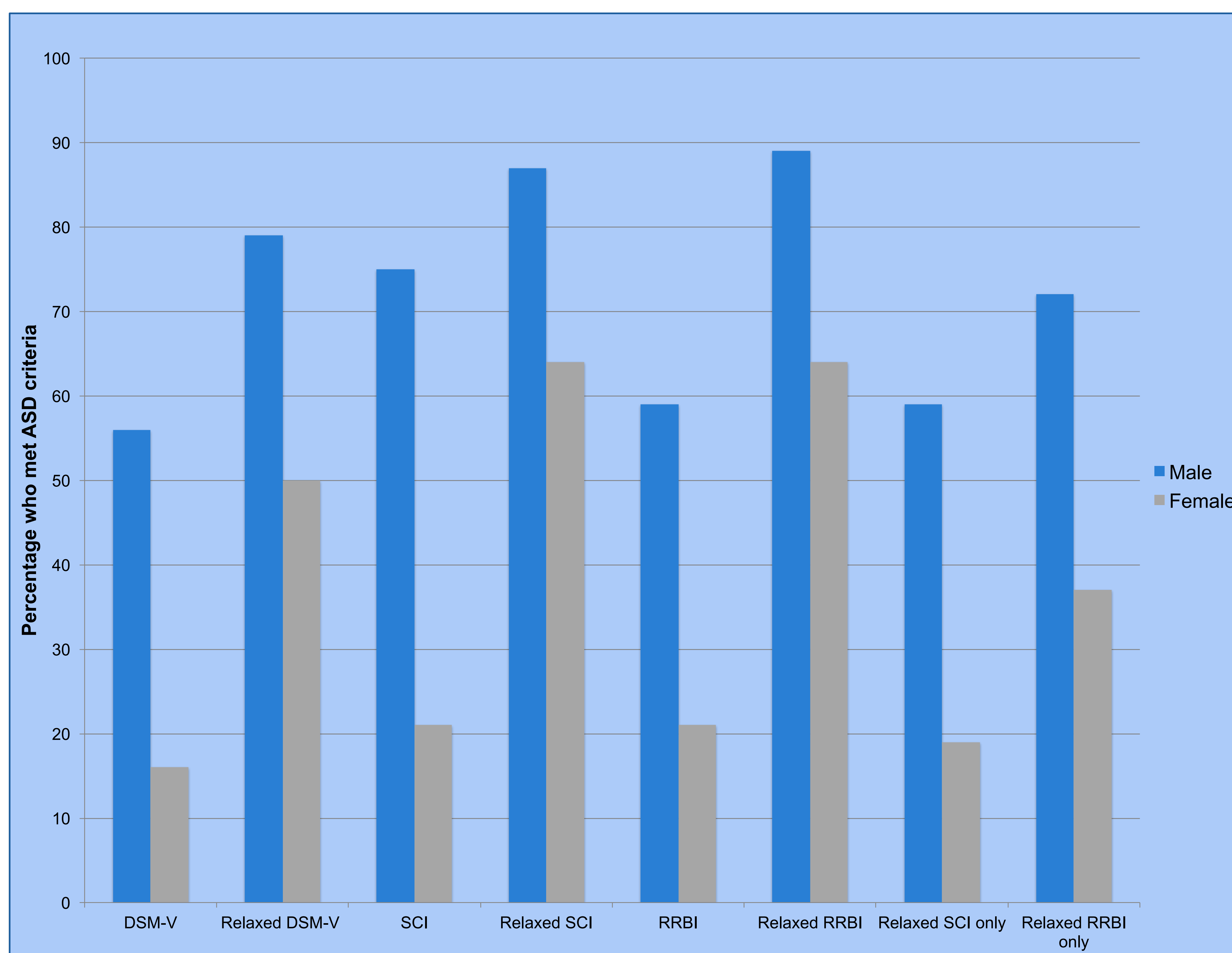
## Results

### DSM-IV

- **66% of males and 21% of females met criteria for ASD**

### DSM-V Overall

- 56% of males and 16% of females met criteria for ASD
- 79% of males and 50% of females met criteria for ASD under relaxed *DSM-V* criteria.
- **59% of males and 19% of females met criteria for ASD when only SCI was relaxed**
- 72% of males and 37% of females met criteria for ASD when only RRBI was relaxed



## Discussion

There is a larger increase for both males (16% higher) and females (21% higher) meeting criteria when the RRBI criterion is relaxed by one qualifying symptom as compared to when the SCI criterion is relaxed by one qualifying symptom (3% higher for male and females).

Further analysis will be done to determine the significance of these differences and to look into the sub-criteria within the SCI and RRBI domains.

Examination of the ASD symptomatic profile of individuals with FXS is important to the understanding of diagnostic trends and the focus of further treatment due to funding constraints on services covered by insurance.