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# The Broad Autism Phenotype in Parents of Children with Autism Spectrum Disorder

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## Introduction

- Autism Spectrum Disorder (ASD) is a neurodevelopmental disability with complex underpinnings and phenotypic manifestations as reflected by its diagnostic classification.<sup>1</sup>
- ASD has a known genetic basis. Twin research reveals an estimated 70% concordance in monozygotic twins (range: 36%–95%) and 3% concordance in dizygotic twins (range 0%–31%).<sup>2</sup>
- The Broad Autism Phenotype (BAP) is a set of personality and linguistic characteristics that reflect mild traits of Autism Spectrum Disorder (ASD) observed in genetically-related individuals with ASD.<sup>3</sup>
- The BAP refers to the phenotypical expression of elevated, but diagnostically subthreshold, levels of ASD symptoms in relatives of individuals with ASD.<sup>4</sup>
- The BAP Questionnaire (BAPQ) is a valid measure used to detect the BAP in parents of children with ASD.<sup>2</sup>
- Research using the BAPQ is warranted. Information resulting from parental performance on the BAPQ is needed to contribute to the knowledge of behavioral markers underlying the genetic liability for ASD.



## Purpose

- The purpose of this investigation was to characterize the BAP in parents of children with ASD compared to normative data in order to better understand the genetic and heritable factors of ASD.

## Methods

- 30 parents (15 mothers and 15 fathers) of children with ASD were administered the BAPQ, which consists of three subscales (Table 1). Each subscale consists of 12 items for a total of 36 items. The average parental age at the time of the child's birth was 33 years (mother) and 37 years (father). All caregivers completed the BAPQ independently. Sensitivity and specificity of the BAPQ, and cutoff scores used to determine presence or absence of the BAP, are presented in Table 2.

➤ **Table 1. Components of the Broad Autism Phenotype**

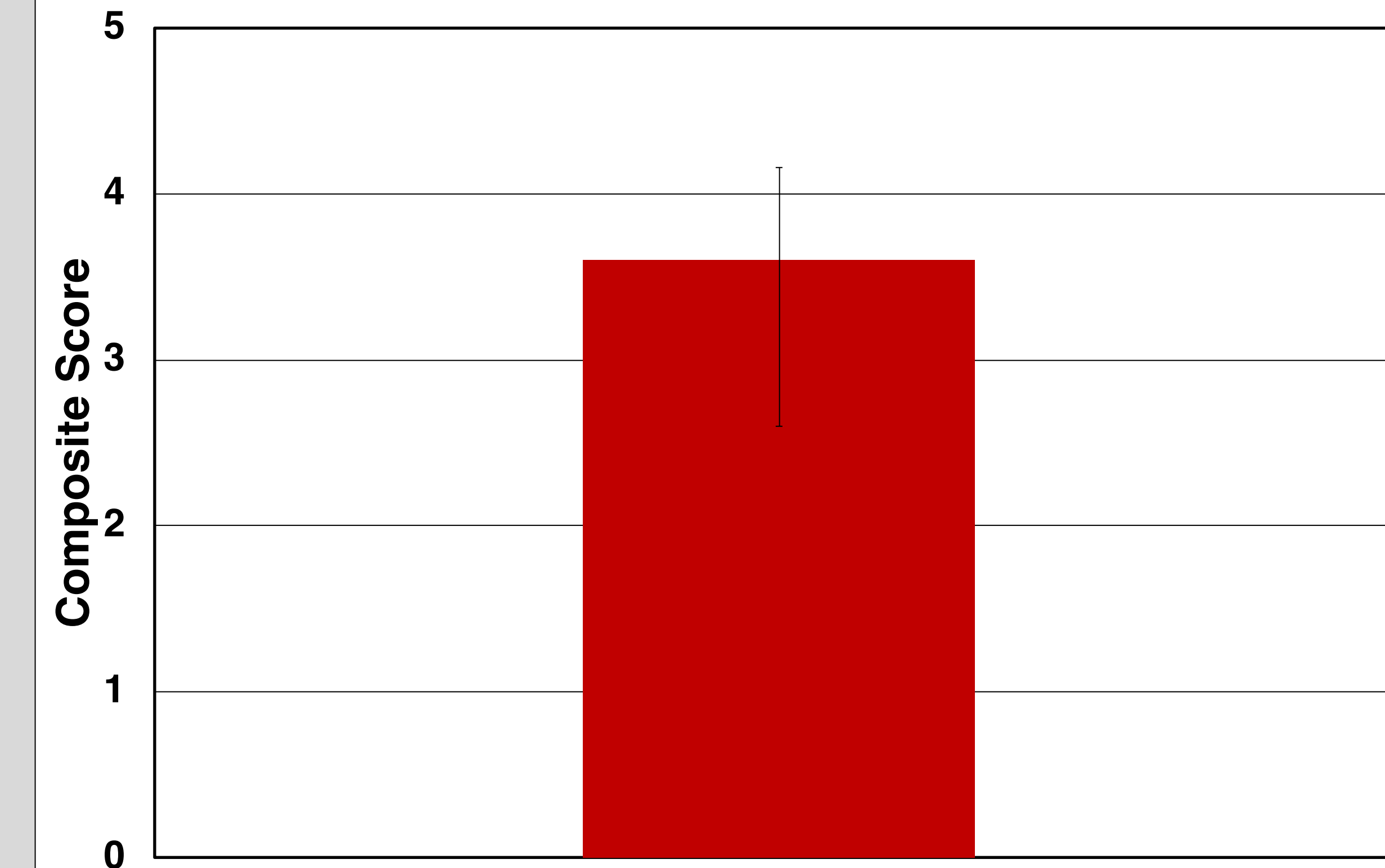
Subscale	Definition
Aloof Personality	Lack of interest in or enjoyment of social interaction
Rigid Personality	Little interest in change or difficulty adjusting to change
Pragmatic Language Deficits	Deficits in social aspects of language and difficulty communicating effectively or holding reciprocal conversation

➤ **Table 2. Psychometric Properties of the Broad Autism Phenotype Questionnaire**

Subscale	Cutoff	Sensitivity	Specificity
Aloof Personality	3.25	77.8	81.4
Rigid Personality	3.50	70.0	81.8
Pragmatic Language Deficits	2.75	76.2	73.8
Composite Score	3.15	81.8	78.1

## Results (Cont.)

Figure 4. Broad Autism Phenotype



## Conclusions

- The present investigation utilized the BAPQ to characterize the BAP in parents of children with ASD.
- Parents of children with ASD tested positive for the BAP across all subscale scores and composite BAP scores.
- The scores gathered from the current sample are markedly increased compared to controls, as based on existing normative data.<sup>3-4</sup>
- The BAPQ is simple to administer and may be a viable screening tool to identify presence/absence of the BAP.
- The BAP can provide genetic and phenotypic insights into ASD.

## Results

Figure 1. Aloofness

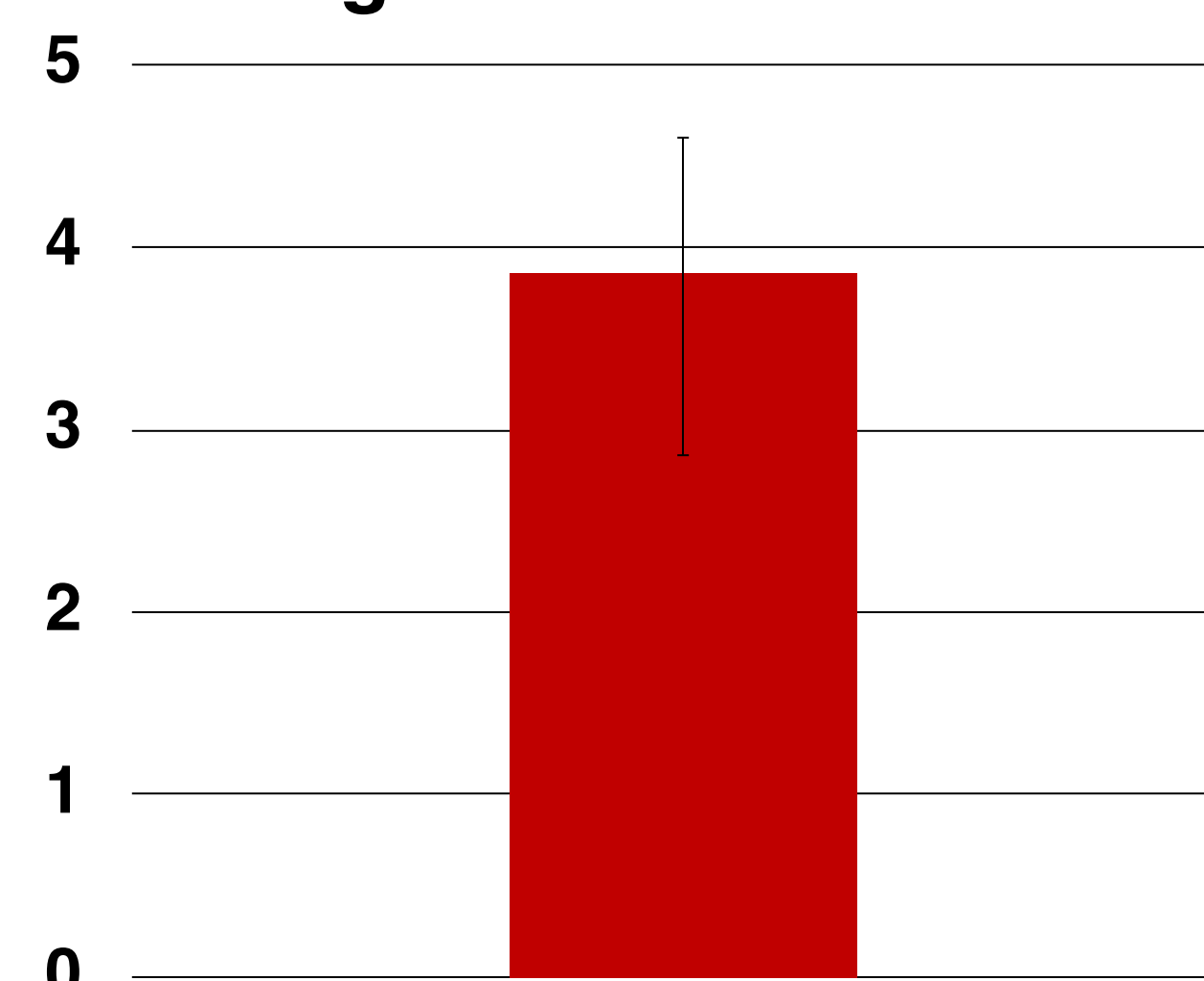


Figure 2. Rigidity

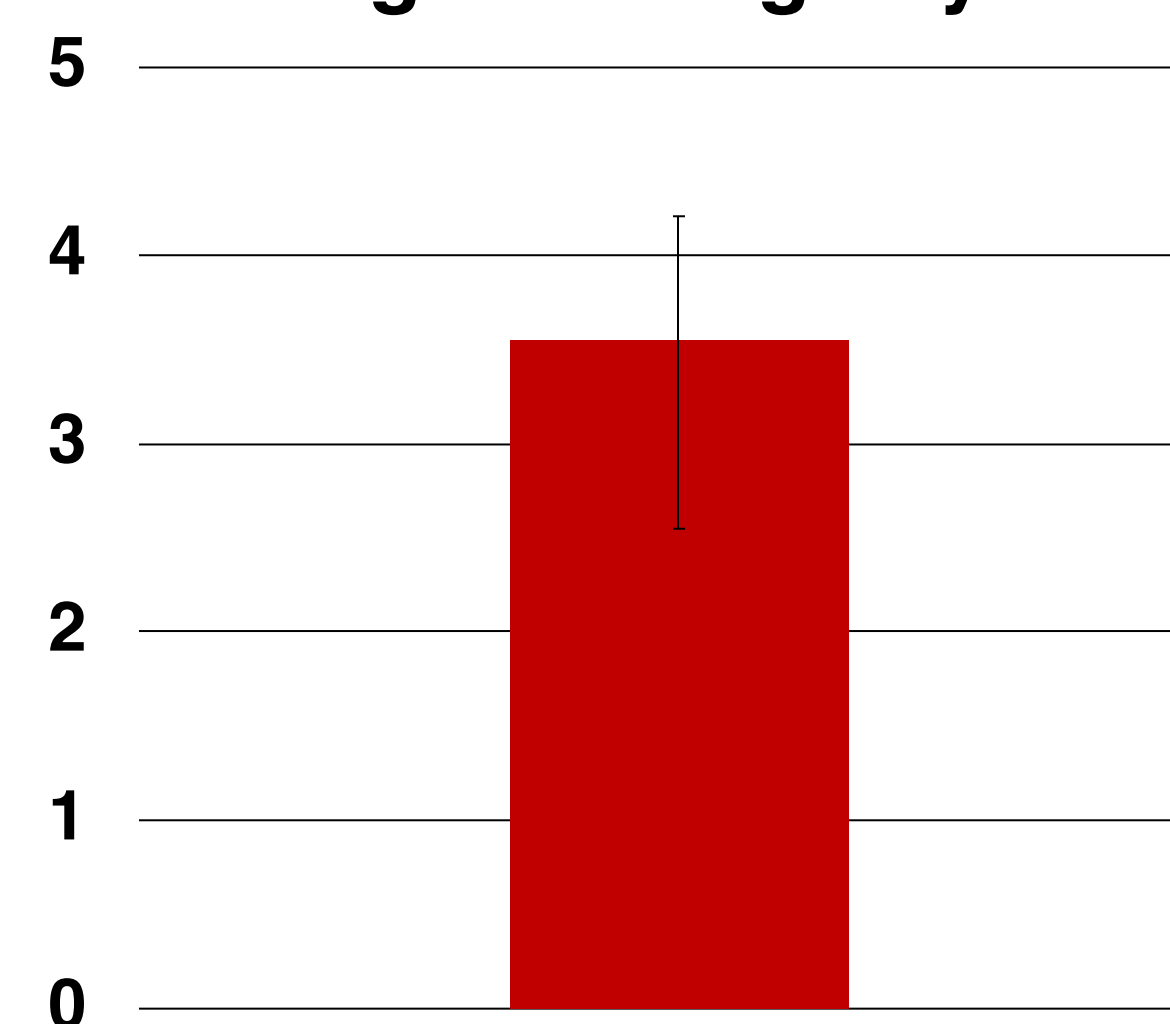
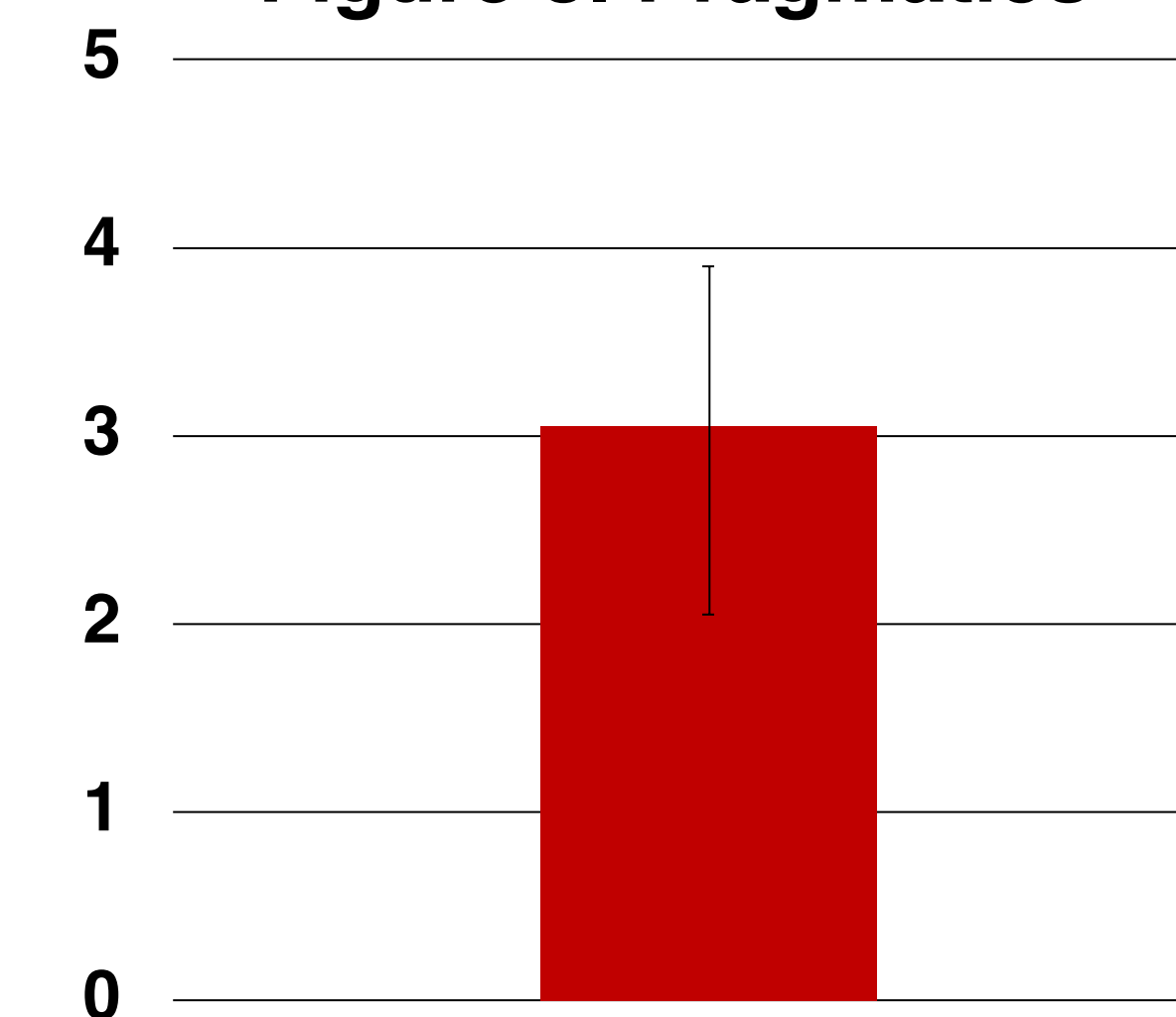


Figure 3. Pragmatics



Subscale Scores on the Broad Autism Phenotype

## References

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3. Hurley, R. S., Losh, M., Parlier, M., Reznick, J. S., & Piven, J. (2007). The broad autism phenotype questionnaire. *Journal of autism and developmental disorders*, 37(9), 1679-1690.
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