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THEORY OF MIND IN THE BAP

Title: Cognitive Processes Predicting Advanced Theory of Mind in the Broader Autism

Phenotype

Short Running Title: Theory of Mind in the BAP

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Abstract

Little is known about executive functions (EF) associated with advanced theory of mind (ToM) abilities. We aimed to determine if advanced ToM abilities were reduced in individuals with sub-clinical traits of Autism Spectrum Disorder (ASD), known as the 'Broader Autism Phenotype' (BAP), and identify the EF that predicted unimpaired performance on an advanced ToM task, the Faux Pas test. We assessed 29 participants (13 males) with the BAP who were relatives of children with ASD. Thirteen participants showed reduced ability to understand a faux pas. A discriminant function analysis correctly classified 79% of cases as impaired or unimpaired, with high sensitivity (80%) and specificity (77%), which was best predicted by language-mediated EF, including verbal generativity, working memory, cognitive inhibition and flexibility.

Lay Summary:

Little is known about the complex cognitive processes that enable accurate interpretation of another person's thoughts and emotions, known as 'theory of mind'. In relatives of individuals with autism, who had mild traits of autism themselves, approximately half had difficulty interpreting situations involving a social faux pas. Cognitive inhibition and flexibility, working memory, and verbal generativity were related to, and appeared to be protective for, unimpaired understanding of a faux pas.

Key Words:

Theory of Mind
Social Skills
Faux Pas

Executive Function
Developmental Psychology
Broader Autism Phenotype
Autism Spectrum Disorder

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Introduction

‘Theory of mind’ (ToM) refers to an individual’s ability to attribute mental states, such as desires, beliefs, emotions, or intentions, to the self and to others, as well as the ability to explain and predict another’s behaviour based on their underlying mental states (Premack & Woodruff, 1978). A well-developed ToM is crucial for effective communication and empathy during everyday social interactions at school, work, and with friends and family. ToM lies at the core of social cognition (Ibañez & Manes, 2012), which is the ability to understand social situations and interpersonal interactions based on the verbal and nonverbal behaviours of others, such as gestures, facial expressions, eye contact, tone of voice, and body language (Carrington & Bailey, 2009).

ToM has been studied extensively in children in an attempt to understand its normal developmental trajectory. In typical development, ToM emerges through a natural progression of stages. By 4-5 years of age, typically developing children can easily pass traditional “first-order” ToM tasks (Korkmaz, 2011; Wellman, Cross, & Watson, 2001), and toddlers as young as 33 months can pass simplified versions of these tasks (Grosso, Schuwerk, Kaltefleiter, & Sodian, 2019; Setoh, Scott, & Baillargeon, 2016). Such tasks require individuals to predict the actions or thoughts of another person based on a false belief. The most common example of a first-order false belief task is the Sally-and-Anne test (Baron-Cohen, Leslie, & Frith, 1985; Wimmer & Perner, 1983). This test presents individuals with a story about two characters, Sally and Anne. Sally places a marble in a basket and then leaves the room. While Sally is away, Anne hides the marble in a box. Participants are asked where Sally will look for the marble upon returning to the room. To successfully pass the

task, participants must be able to take the perspective of Sally, who has a false belief about where the marble is. By 5-6 years of age, typically developing children are able to infer second-order false beliefs (Arslan, Verbrugge, Taatgen, & Hollebrandse, 2018; Korkmaz, 2011; Miller, 2009). These involve making inferences about a person's thoughts or feelings about someone else's mental state.

More advanced ToM processes including understanding irony, jokes, sarcasm, and faux pas occur later in development. It is not yet clear, however, exactly when these aspects of ToM develop (Filippova & Astington, 2010; Korkmaz, 2011; Miller, 2009). Studies of more advanced ToM abilities in school-aged children have produced mixed results, which may reflect that ToM is dependent upon differential cognitive processes. Furthermore, appropriately testing advanced ToM can be difficult as many tasks are adaptations of children's tests, which may not be sensitive to individual differences or subtle deficits in ToM (Dodell-Feder, Lincoln, Coulson, & Hooker, 2013). The Faux Pas test (Baron-Cohen, O'Riordan, Stone, Jones, & Plaisted, 1999; Stone, Baron-Cohen, & Knight, 1998) is an example of an advanced ToM task. It comprises social scenarios and conversations depicting awkward or inappropriate encounters, which reflect real-life situations and thus, are more socially and cognitively demanding than false-belief tasks.

ToM and Executive Functioning

ToM engages a range of psychological processes within a social context, including emotion recognition, empathy, mental state reasoning and executive functions (EF). Researchers across disciplines have created theoretical models to explain ToM abilities. One of the most supported of these highlights the relationship between ToM and EF (Bradford,

Jentsch, & Gomez, 2015; Hughes, 1998a; Ozonoff, Pennington, & Rogers, 1991). EF are higher-order cognitive processes that are required to complete goal driven tasks. They include planning, problem solving, attention switching, and concept formation, and regulate lower-order cognitive functions.

The link between EF and ToM has been consistently demonstrated across the lifespan, from early childhood (Cole & Mitchell, 2000; Doenyas, Yavuz, & Selcuk, 2018; Fahie & Symons, 2003; Fisher, Happé, & Dunn, 2005; Gao, Huang, Zhang, & Chen, 2019; Gordon & Olson, 1998; Hughes, 1998a; Hughes & Graham, 2002; Joseph & Tager-Flusberg, 2004; Marcovitch et al., 2015; Powell & Carey, 2017), middle childhood (Bock, Gallaway, & Hund, 2015; Kouklari, Tsermentseli, & Auyeung, 2018; Kouklari, Tsermentseli, & Monks, 2019; Lecce et al., 2019; Wilson, Andrews, Hogan, Wang, & Shum, 2018), through adulthood (Ahmed & Miller, 2011; Bradford et al., 2015; Bull, Phillips, & Conway, 2008; German & Hehman, 2006; McKinnon & Moscovitch, 2007; Saltzman, Strauss, Hunter, & Archibald, 2000), both in Western and non-Western cultures (Sabbagh, Xu, Carlson, Moses, & Lee, 2006). Longitudinal studies have consistently found that EF predict ToM over time (Austin, Groppe, & Elsner, 2014; Carlson, Mandell, & Williams, 2004; Derksen, Hunsche, Giroux, Connolly, & Bernstein, 2018; Doenyas et al., 2018; Hughes, 1998b; Kouklari et al., 2019; Lecce, Bianco, Devine, & Hughes, 2017; Marcovitch et al., 2015). A recent study took an experimental, rather than correlational, approach to understanding the role of EF in ToM in children 4-5 years of age (Powell & Carey, 2017). Using an EF depletion method during a false-belief task, the authors demonstrated that depleting EF resources negatively affected

children's ability to predict, as well as to explain, another's behaviour on the basis of their false belief.

Although the majority of ToM studies use false-belief tasks (Bradford et al., 2015), there is increasing evidence from behavioural studies that there are distinct sub-components of ToM that engage unique cognitive processes (Ahmed & Miller, 2011; Bradford et al., 2015; Wilson et al., 2018). Ahmed and Miller (2011) assessed ToM in healthy adults using different ToM tasks, and EF was measured using the Delis-Kaplin Executive Function System (D-KEFS; Delis, Kaplan, & Kramer, 2001). Participants were administered the Faux Pas test (Baron-Cohen et al., 1999; Stone et al., 1998), which requires the ability to abstract and reason about another's mental state, as well as emotional empathy, and understanding of social etiquette. Performance on the Faux Pas test was best predicted by language-mediated cognitive flexibility, inhibition, verbal generativity, and working memory, as well as problem solving and sorting abilities, as measured by the Letter Fluency condition of the Verbal Fluency Test (referred to by the authors as 'verbal fluency') and confirmed correct sorts of the Free Sort condition of the Sorting Test (referred to as 'problem solving'). Gender also accounted for a significant amount of variance in Faux Pas test scores, with females outperforming males. Participants were also administered the Strange Stories test (Happé, 1994), which requires understanding of white lies, double bluff, and persuasion. Performance on the Strange Stories test was similarly predicted by language-mediated cognitive flexibility, inhibition, verbal generativity, and working memory as measured by the Verbal Fluency Test, as well as deductive reasoning, as measured by the total number consecutively correct of the Word Context Test.

Other correlates of ToM include IQ, gender and verbal ability (Carlson et al., 2004; Losh & Piven, 2007). Females generally perform better than males, and this gender effect may be partially mediated by verbal ability (Carlson et al., 2004). Other factors include interactions with parents and older siblings (Derksen et al., 2018), including the number of older children and siblings a child has daily contact with (Lewis, Freeman, Kyriakidou, Maridaki - Kassotaki, & Berridge, 1996; Ruffman, Perner, Naito, Parkin, & Clements, 1998), whether emotional status is discussed in the home environment and between friends (Dunn, Brown, Slomkowski, Tesla, & Youngblade, 1991; Hughes & Dunn, 1998), pretend play (Astington & Jenkins, 1995), and the extent to which a mother comments on her child's thoughts and feelings (Meins et al., 2002).

Impairments in ToM

Our understanding of the functional modularity of ToM can be advanced by studies of clinical populations who show fractionated profiles of ToM abilities. Deficits in ToM can result in misunderstanding the intentions of another person, leading to difficulties creating and maintaining relationships, social isolation, anxiety, and depression (Ahmed & Miller, 2011; Derksen et al., 2018). ToM deficits have been observed in disorders such as dementia, bipolar disorder, schizophrenia, and Autism Spectrum Disorder (ASD; Brüne & Brüne-Cohrs, 2006).

ASD is a life-long developmental disorder, beginning in early childhood, defined by pervasive impairments in social communication as well as restricted interests and repetitive behaviours (DSM-5; American Psychiatric Association, 2013). Children with ASD generally fail first-order false belief tasks until they reach the verbal ability of a typically developing

11-12 year old (Fisher et al., 2005; Happé, 1995). Studies of more advanced ToM abilities in school-aged children and young adults with high-functioning ASD (HFA; IQ >70) have provided mixed results, with some studies finding impairments compared to typically developing peers (Baron - Cohen, Jolliffe, Mortimore, & Robertson, 1997; Brent, Rios, Happé, & Charman, 2004; Kleinman, Marciano, & Ault, 2001), and others finding no differences (Ponnet, Buysse, Roeyers, & De Clercq, 2008; Roeyers, Buysse, Ponnet, & Pichal, 2001; Scheeren, de Rosnay, Koot, & Begeer, 2013; Senju, Southgate, White, & Frith, 2009; Spek, Scholte, & Van Berckelaer-Onnes, 2010). By adulthood, individuals with HFA or Asperger's Disorder often pass first- and second-order false-belief tasks (Dahlgren & Trillingsgaard, 1996). They usually struggle, however, with more advanced ToM tasks that require an understanding of sarcasm, irony, or bluff (Happé, 1994), as well as faux pas tasks (Baron-Cohen et al., 1999; Zalla, Sav, Stopin, Ahade, & Leboyer, 2009).

Individuals with ASD often have impairments in EF (Demetriou et al., 2018; Kouklari et al., 2018; Kouklari et al., 2019), particularly in inhibition and flexibility, with evidence to suggest that these may be associated with their deficits in ToM (Bradford et al., 2015; Cole & Mitchell, 2000; Gordon & Olson, 1998; Hughes, 1998a; Jones et al., 2018; Kouklari et al., 2018; Kouklari et al., 2019; Losh et al., 2009; McLean, Harrison, Zimak, Joseph, & Morrow, 2014; Ozonoff & McEvoy, 1994; Ozonoff et al., 1991; Sabbagh et al., 2006; Saltzman et al., 2000).

The Broader Autism Phenotype

It is well recognised that relatives of individuals with ASD may show some ASD features, commonly referred to as the Broader Autism Phenotype (BAP). Studies have

documented these features in 20-40% of first degree relatives of children with ASD (Gamliel, 2009; Georgiades et al., 2013; Messinger et al., 2013; Ozonoff et al., 2014; Rubenstein et al., 2019; Sasson et al., 2013). The BAP is characterised by traits that are qualitatively similar, but milder or reduced in amount, and do not cause sufficient impairment to warrant a diagnosis of ASD. Individuals with the BAP may have similar cognitive profiles to what has been found in some individuals with ASD, with deficits in performance IQ, EF, and reading comprehension (Piven, 2001). Although primarily found in first degree relatives of children with ASD, the BAP is also found in 2-9% of individuals from the general population (Bora, Aydın, Saraç, Kadak, & Köse, 2017; Constantino & Todd, 2005; Gökçen, Petrides, Hudry, Frederickson, & Smillie, 2014; Happé, Ronald, & Plomin, 2006; Robinson et al., 2011; Sasson et al., 2013).

However, BAP prevalence rates vary substantially depending on how the BAP is measured and defined (Rubenstein & Chawla, 2018). A recent systematic review (Rubenstein & Chawla, 2018) of studies of BAP prevalence in parents of children with ASD (n=41) found that self-report was the most common method of data collection (n=25), and that just over half of the studies (n=22) used multiple reporting methods (e.g., self-report and informant-report). The most commonly used instruments were the Family History Interview (Bolton et al., 1994), followed by the Autism Quotient (Baron-Cohen, Wheelwright, Skinner, Martin, & Clubley, 2001) and the Broader Autism Phenotype Questionnaire (Hurley, Losh, Parlier, Reznick, & Piven, 2007). Unsurprisingly, BAP prevalence was higher in studies using stricter criteria (i.e., defining BAP by meeting criteria on ≥ 1 domain versus ≥ 2 domains). The vast majority of studies screened for the review did not meet inclusion criteria (n = 380 excluded

out of 486 screened), as they only compared parents of children with ASD to controls without directly measuring the BAP.

The majority of studies in first degree relatives of children with ASD have found deficits in social cognition (Dorris, Espie, Knott, & Salt, 2004; Gökçen et al., 2014; Gokcen, Bora, Erermis, Kesikci, & Aydin, 2009; Grove, Baillie, Allison, Baron-Cohen, & Hoekstra, 2014; Losh et al., 2009; Palermo, Pasqualetti, Barbati, Intelligente, & Rossini, 2006; Sucksmith et al., 2013; Wallace, Sebastian, Pellicano, Parr, & Bailey, 2010). For example, numerous studies have shown that parents and siblings of children with ASD exhibit impairments in emotion recognition, empathy, and ToM tasks compared to controls (Dorris et al., 2004; Losh et al., 2009; Palermo et al., 2006; Sucksmith et al., 2013; Wallace et al., 2010). A study of 3-year old siblings of children with ASD found that they performed worse on a false-belief task than children without siblings with ASD, regardless of whether they had ASD, BAP or were unaffected (Gliga, Senju, Pettinato, Charman, & Johnson, 2014). Parents of children with ASD have moderate impairments in empathy compared to typically developing adults, yet their impairments are less severe than in adults with ASD (Grove et al., 2014). Moreover, they have been found to perform worse in mental state reasoning and false-belief tasks compared to parents of typically developing children (Gokcen et al., 2009). ToM abilities have yet to be investigated in adult relatives of children with ASD who have the BAP, meaning that advanced ToM abilities have not yet been explored nor has their relationship to EF in this cohort.

The Present Study

The developmental trajectory of advanced ToM abilities is not fully understood in adolescents and adults, due in part to the differential role played by varying EF in advanced ToM. Individuals with the BAP represent an ideal cohort to examine the relationships between EF and ToM, as they have sub-clinical traits of ASD, and therefore likely have variable abilities in ToM. They also have fewer comorbidities and a higher IQ than ASD clinical samples often used (Yucel et al., 2014), making interpretation of underlying cognitive mechanisms more straightforward. In comparison, adults from the general population can easily pass even the most difficult ToM tasks and do not have impairments in EF, creating ceiling effects across tests of both ToM and EF. In healthy adults, language-mediated cognitive flexibility, inhibition, and verbal generativity, as well as problem solving and sorting abilities are correlated with performance on the Faux Pas test (Ahmed & Miller, 2011). Studying the predictive power of various EF on ToM in a population with variable ToM abilities will help identify EF that predict, rather than are merely associated with, successful performance of the Faux Pas test. Thus, we sought to determine the features of EF that best predict advanced ToM ability in a population known to have subtle deficits in social cognition.

Our aim was to determine if advanced ToM ability was impaired in the BAP, and to identify the components of EF that predict this ability. We hypothesised that 1) adolescents and adults with the BAP would show impairments in performance on the Faux Pas test, and 2) language-mediated EF, including verbal generativity, cognitive inhibition, flexibility and working memory would predict unimpaired performance in individuals with the BAP. The

current study is the first, that we are aware of, to test advanced ToM abilities in adult relatives of individuals with ASD, and the first to test individuals with the BAP using the Faux Pas test.

Methods

Participants

Participants were recruited to a study of the genetic causes of ASD through The University of Melbourne, Barwon Health, and the Royal Children's Hospital (Brown, 2014; Trevis et al., 2019). Participants belonged to two large multiplex families with at least eight 'affected' relatives (ASD or the BAP). Ethics approval was obtained through the Royal Children's Hospital and Barwon Health. The methods of this study have been previously described in detail (Trevis et al., 2019). In short, probands with ASD underwent screening tests including molecular karyotype, Fragile X DNA testing, and urine metabolic screen. Families of individuals with ASD who tested positive on this screen for a variant or condition likely to contribute to the development of ASD were excluded. A total of 62 individuals from two families were assessed ($n_{ASD} = 17$, $n_{BAP} = 32$, $n_{Unaffected} = 13$). We included 29 participants (13 males) with the BAP, aged 13-80 years ($M=46.69$, $SD=20.99$). Three participants were excluded due to incomplete D-KEFS data. On average, the participants' cognitive abilities fell within the average to high-average range, with males having a significantly higher Full-Scale IQ (FSIQ), Verbal IQ (VIQ), and Performance IQ (PIQ) than females (**Table 1**).

[Table 1]

*Measures and Procedure**Phenotypic Assessment*

In these families, individuals without ASD were assessed for the BAP using an extensive battery of tests administered across multiple sessions (**Figure 1**). Testing was conducted either in the participants' home, at Barwon Health, at The Royal Children's Hospital, or at the Melbourne Brain Centre, and took 6 to 8 hours to complete. Traits of the BAP were assessed using the Family History Interview (Bolton et al., 1994), a modified adult version of the Faux Pas test (Baron-Cohen et al., 1999; Stone et al., 1998), the Pragmatic Rating Scale (Landa et al., 1992), the Broader Autism Phenotype Rating Scale (BAPRS; adapted from Bailey et al., 1995), and a semi-structured interview (Brown, 2014). The Social Responsiveness Scale (SRS; Constantino & Gruber, 2005) was used to screen all individuals for potential ASD. Individuals with features suggestive of ASD were assessed using the Autism Diagnostic Observation Schedule (ADOS; Lord et al., 1999) and Autism Diagnostic Interview – Revised (ADI-R; Lord, Rutter, & Le Couteur, 1994) by a trained clinician (NB) with research level reliability. The final classification of all participants was reached at the conclusion of all assessments through expert clinical judgement and consensus agreement between a paediatric neurologist (IS), clinical neuropsychologist (SW), and ASD paediatrician (NB). Individuals were classified as having the BAP if the number of endorsed traits was above the BAP cut-off for at least one of the three domains on the BAPRS.

[Figure 1]

Theory of Mind

ToM was assessed using an adult version of The Faux Pas test adapted from Baron-Cohen et al. (1999) and Stone et al. (1998). The task was selected due to its psychometric properties (Faísca et al., 2016; Fernández-Modamio et al., 2018; Hayward & Homer, 2017; Smogorzewska, Szumski, & Grygiel, 2018; Söderstrand & Almkvist, 2012; Zalla et al., 2009). Although the psychometric properties of the Faux Pas test have not been formally assessed in adolescents, it has been shown to be able to discriminate between children and adults with high-functioning ASD from controls (Baron-Cohen et al., 1999; Zalla et al., 2009). Furthermore, it has not been used with individuals with the BAP. Participants were read aloud a series of scenarios depicting either a social faux pas or a control story, and were provided with the written text while they were asked questions about the actions, motivations, and beliefs of the characters in each scenario. Each participant was required to decide whether or not one of the characters said something awkward or inappropriate, and if so, to explain what was said, why the character made the awkward remark, and the character's understanding of the situation. The Faux Pas test is an advanced ToM task that requires participants to understand and make judgements about each characters' mental state and actions while considering the whole social situation (i.e., being aware of what each character knows and believes, and the social etiquette of the situation). Participants were presented with four faux pas stories and four control stories. Scores on the Faux Pas test ranged from 0-40, with a lower score indicating poorer ToM capabilities.

Executive Function

The Delis-Kaplin Executive Function System (D-KEFS; Delis, Kaplan, & Kramer, 2001) is a standardized assessment of executive functioning for use in individuals from ages 8-89 years. It consists of a comprehensive battery of nine subtests of executive functioning (**Table 2**), with each subtest comprising multiple tasks. Two subtests, Twenty Questions and Word Context, were not administered. This was done to minimise participant fatigue, and because the full D-KEFS was not essential to achieve the primary aim of the broader overall study, which was to identify genetic variants associated with BAP traits and/or endophenotypes. The primary scaled scores from each of the seven remaining subtests were used in the current analyses.

[Table 2]

Data Analyses

Given that there is no cut-off score to indicate impaired performance on the Faux Pas test, a cluster analysis was used to classify participants as having either impaired or unimpaired performance; a k-means cluster analysis and a two-step cluster analysis were performed. To test our second hypothesis, a discriminant function analysis was used to identify the EF that best predicted group membership for the unimpaired group, and thus, predict unimpaired performance on the Faux Pas test. A logistic regression, as well as the descriptive statistics, yielded similar results to the discriminant function analysis; therefore, the results from the discriminant function analysis are presented as the most robust analysis. All analyses were conducted using SPSS (version 22), with a criterion level of $p \leq .05$.

Outliers (n=4) were winsorised to within 2.5 SD of the mean for each variable to minimise their impact on the mean score.

Results

Faux Pas Test Performance in the BAP

The participant Faux Pas test scores ranged between 24-40 ($M = 35.45$, $SD = 4.77$), with a mean score consistent with previous studies that have found that healthy control adults score at or near ceiling (Lee, 2010; Stone et al., 1998; Zalla et al., 2009). Faux Pas test scores were significantly correlated with FSIQ ($r_s = .517$, $p = .004$), VIQ ($r_s = .457$, $p = .013$), and PIQ ($r_s = .513$, $p = .004$). Faux Pas test scores were not correlated with age ($r_s = -.231$, $p = .228$).

A k-means cluster analysis split the participants into two clusters representing impaired and unimpaired performance on the Faux Pas test. Participants in the first cluster had Faux Pas test scores between 24-33 (n=8), and participants in the second cluster had scores between 34-40 (n=21). A two-step cluster analysis was also performed, which split the Faux Pas test scores into two clusters. Participants in the first cluster had scores between 24-35 (n=13) while participants in the second cluster had scores between 37-40 (n=16). The classification of the second cluster (i.e., better performance) from the two-step cluster analysis most closely resembled findings from previous studies of healthy control participants (Lee, 2010; Stone et al., 1998; Zalla et al., 2009), and was therefore more likely to accurately represent unimpaired performance. Thus, participants were classified as having either impaired (n=13) or unimpaired (n=16) performance based on two-step cluster analysis

(**Figure 2**). Participants in the impaired group answered at least 5 questions incorrectly across eight faux pas scenarios, suggesting moderate to significant ToM difficulties. Errors reflected difficulties in identifying the occurrence of a faux pas and understanding its effect, as well as understanding the beliefs and intentionality of the person committing the faux pas.

[Figure 2]

The difference in median scores between the impaired ($Md = 32$) and unimpaired ($Md = 39$) groups was significant using a Mann-Whitney U test ($U = 0.00$, $z = -4.612$, $p = .000$), with a very large effect size ($r = -0.86$). There were no significant differences between the groups in regards to age or gender (**Table 3**).

[Table 3]

Predicting Impaired Faux Pas Test Performance

Because each D-KEFS subtest has multiple conditions, the conditions chosen as predictor variables for the discriminant function analysis were selected based on three factors: 1) its significant correlation with the Faux Pas test (**Table 4**); 2) its ability to tap into the core cognitive processes required to successfully complete the D-KEFS subtest; and 3) meeting the assumptions required for analyses (normality, linearity, homoscedasticity, multicollinearity, and singularity). The four D-KEFS variables that met these three criteria and were therefore selected as predictor variables for the discriminant function analysis were

Verbal Fluency (Category Switching Total Accuracy), Design Fluency (Total Correct Composite), Color-Word Interference (Inhibition/Switching), and Sorting (Free Sorting Confirmed Correct Sorts). The distribution of scaled scores for the selected D-KEFS variables (**Table 5**) indicated that the majority (66%) of the sample's scores fell within one standard deviation of the mean and were similar to a normal distribution ($M = 10$, $SD = 3$), but were negatively skewed. There were no significant differences in performance between males and females, except for Sorting, where males performed better than females. Individuals with impaired performance on the Faux Pas test had lower FSIQ, VIQ, PIQ, Verbal Fluency, Design Fluency, and Color-Word Interference scores than those with unimpaired performance (**Table 6**).

[**Table 4**]

[**Table 5**]

[**Table 6**]

A discriminant function analysis was used to test our second hypothesis that verbal generativity, language-mediated cognitive inhibition and flexibility, and working memory predict successful completion of the Faux Pas test, and to identify the variables that best predicted group membership (impaired vs unimpaired Faux Pas test performance). The variables entered into the discriminant function analysis using a step-wise procedure included: FSIQ, Verbal Fluency, Design Fluency, Color-Word Interference, and Sorting. For the discriminant function, the largest standardized coefficient was Verbal Fluency (.944)

followed by Color-Word Interference (.803), while the largest correlation was Verbal Fluency (.660) followed by Sorting (.478) and Color-Word Interference (.469) (**Table 7**).

Using Verbal Fluency and Color-Word Interference, the model correctly classified 79% of cases and predicted group membership ($\Lambda = .583$, $\chi^2(2) = 12.96$, $p = .002$) with a high degree of sensitivity (80%) and specificity (77%) (**Figure 3**).

[**Table 7**]

[**Figure 3**]

Discussion

In this study, we have shown for the first time that advanced ToM abilities, as measured by the Faux Pas test, are reduced in adolescents and adult relatives of children with ASD who have the BAP. We also identified specific EF that predict successful performance on an advanced ToM task, namely language-mediated cognitive flexibility and inhibition, verbal generativity, and working memory. Approximately half (45%) of our BAP sample showed impaired performance on the Faux Pas test, with successful classification of impaired performance in 79% of individuals. The EF predictors of Verbal Fluency (Category Switching Total Accuracy) and Color-Word Interference (Inhibition/Switching) showed a high sensitivity (80%) and specificity (77%) of classification.

During the Verbal Fluency test, participants are asked to generate a list of words while following a set of rules (e.g., no names of places, no repeating words, etc.). For the

Category Switching condition of the Verbal Fluency test, participants must verbally generate a list of words while switching back and forth between categories (i.e., furniture and fruit), thus exhibiting a high degree of cognitive flexibility and inhibition, working memory, and verbal generativity. The Category Switching Total Accuracy score includes the switching error rate, and therefore reflects a combination of cognitive flexibility and inhibition. The Inhibition/Switching condition of Color-Word Interference similarly targets cognitive flexibility and inhibition, as participants are asked to read a page of written colour names that at times appear in a different colour ink (e.g., the word 'red' printed in blue ink) while following specific rules depending on how the words are presented (i.e., either read the word or label the ink colour). Interestingly, the Inhibition condition of Color-Word Interference was not significantly correlated with the Faux Pas test, nor was Category Switching Total Correct (indicative of verbal generativity, working memory, and inhibition, but not of cognitive flexibility as the score equals the sum of the items that correctly fall within either category irrespective of set-shifting).

Taken together, there are two possible conclusions that can be drawn from these findings: 1) That cognitive flexibility best predicts Faux Pas test performance, not inhibition; or 2) That cognitive flexibility in conjunction with higher-level/more advanced inhibition best predict Faux Pas test performance. While inhibition is required for all four conditions, the difficulty or level of inhibition required varies. For example, in Category Switching Total Correct, participants only need to inhibit repeating a word (which is likely to be more of a function of working memory). In comparison, during the Inhibition condition of Color-Word Interference, participants need to inhibit reading a word (an automatic process) in order to

label the colour, which is a much more cognitively demanding task. Both the Category Switching Total Accuracy score and Inhibition/Switching require cognitive flexibility, which inherently also requires a degree of inhibition to successfully set-shift. The most cognitively demanding task for both cognitive flexibility and inhibition is the Inhibition/Switching condition. These findings suggest that while ‘low-level’ inhibition is not associated with Faux Pas test performance, ‘high-level’ inhibition that is intertwined with cognitive flexibility, is.

During the Faux Pas test, participants must incorporate information presented to them to make inferences about each character’s knowledge, emotional state, and intentionality. To successfully do so, they must simultaneously inhibit some information in order to flexibly consider each character’s mental state, and articulate their thoughts to the examiner. Our findings support the notion that while basic ToM tasks rely heavily on working memory and cognitive inhibition, advanced ToM tasks recruit additional cognitive resources, such as cognitive flexibility and verbal generativity. These findings suggest that for individuals with the BAP, cognitive flexibility and inhibition are strongly related to, if not essential for, identifying and understanding awkward or inappropriate statements and actions during everyday social interactions.

A unique strength of our study is that we have interrogated the impact of different cognitive processes on ToM in individuals with intact intellectual and EF abilities, but mild impairments in social cognition. Contrary to previous studies which found that relatives of children with ASD showed impairment in EF (Sucksmith, Roth, & Hoekstra, 2011), we found no evidence of this in the group analysis. Generally, the participant mean D-KEFS scaled scores (**Table 4**) were within 1 SD of the average when compared to normative data

($M=10$, $SD=3$). The sample can therefore be characterised as ‘high-functioning’ adolescents and adults with sub-clinical traits of ASD, half of whom have noticeable difficulties with ToM. Our participants with unimpaired performance on the Faux Pas test (**Table 6**) had mean D-KEFS scaled scores within or above the average range and their language-mediated cognitive flexibility and inhibition abilities were well above average. Considered together, we speculate that, while EF itself may not be impaired in individuals with the BAP, having EF abilities above average may be a protective factor for ToM abilities.

Our findings reflect those observed in healthy non-BAP adults, in whom similar EF are predictive of ToM ability (Ahmed & Miller, 2011). Therefore, our findings and those of typically developing adults suggest that impaired ToM in the BAP may be related to EF regulation and integration within a social setting, rather than recruitment of impaired EF per se. These findings may not be restricted to ASD or the BAP, but may apply to other clinical populations who have been found to have impairments in ToM, such as Schizophrenia and Bipolar Disorder. If similar results were found in other populations, this would provide insight into the fundamental cognitive processes underlying social impairment.

Our study had an even ratio of males to females, which is often not the case in clinical studies comprising participants with ASD. We explored possible gender differences and their impact on ToM in individuals with the BAP. Contrary to previous research our female participants did not outperform males on our ToM task (Ahmed & Miller, 2011; Losh & Piven, 2007). This may reflect that gender differences in social cognition in the BAP may be different to what is found in typically developing individuals, if they are present at all. Alternatively, it has been suggested that the gender differences found in previous studies may

have been verbally mediated (Carlson et al., 2004), with females typically having stronger verbal skills than males. To explore this possibility, we compared cognitive and EF abilities between males and females. Interestingly, males had a significantly higher FSIQ, VIQ, and PIQ than females (**Table 1**), and scored significantly higher on the Sorting subtest of the D-KEFS (**Table 5**). This suggests that although males in our study had a significant cognitive advantage over females, there were no gender differences in ToM.

Unlike most studies, which rely on a self-report questionnaire to determine the presence of BAP features, participants in our study underwent rigorous clinical phenotyping over 6-8 hours. Finding simple tests sensitive enough to detect the subtle features seen in individuals with the BAP is challenging, as they often perform at ceiling on measures designed for ASD. The Faux Pas test offers a method of identifying subtle differences in advanced ToM. Our results show that there is a subgroup of individuals with the BAP who have impaired ToM abilities. This may serve as a useful marker in understanding the clinical genetics of ASD in future studies.

In contrast to many studies of the BAP, our study focused on individuals who were members of families with at least eight affected (ASD or BAP) individuals. Evidence suggests that there appears to be a graded expression of ASD traits according to genetic liability to ASD. Losh et al. (2008) found more ASD traits in parents who had multiple children with ASD than parents with only one child with ASD, who in turn had more ASD traits than parents of children with Down Syndrome who served as a comparison group. In the general population Gökçen et al. (2014) found that adults with more ASD traits performed worse on tasks of emotion recognition, cognitive shifting, and emotional cognitive

shifting, compared to adults with fewer ASD traits. Therefore, our sample with high genetic liability provides an ideal population to further understand the cognitive and genetic mechanisms of social cognition in ASD and the BAP.

Despite the strengths of the study, there are limitations to bear in mind when considering the current findings and their application to future research. One such limitation is the lack of a control group consisting of adolescents and adults without the BAP or a family history of ASD. This would allow for direct comparison of Faux Pas test performance between individuals with and without the BAP from the same population. Other studies of Faux Pas test performance in control participants have used different versions of the Faux Pas test and have not provided median scores, making direct comparison to the current sample difficult. Additionally, while the Faux Pas test has been regularly used in children and adults, it has been used to a lesser extent in adolescent samples; thus, the psychometric properties of the Faux Pas test in adolescents have not yet been established. Interpretation of our findings, which includes results from adolescents ($n = 5$) should be considered within this context; however, there is evidence to suggest that the ability to understand a faux pas is present before adolescence (Banerjee & Watling, 2005; Baron-Cohen et al., 1999), between 7-9 years of age in boys and 9-11 years in girls (Baron-Cohen et al., 1999).

Furthermore, the sample size was prohibitive of analyses that may provide a more nuanced picture of the relationship between EF and advanced ToM. While EF has consistently been found to predict ToM, other factors have also been associated with ToM (e.g., early childhood environment and experiences). A larger sample size would allow for the identification of potential mediators and/or moderators within this relationship. For

example, it is possible that a third factor (e.g., having older siblings or experience with older children when younger) may be moderating the link between EF and ToM, or alternatively, that EF may be both a predictor and a moderator. If EF moderated the link between advanced ToM and another predictor, this would provide further evidence that EF may play a protective role for unimpaired ToM abilities.

Future research into the cognitive processes underlying advanced ToM abilities may provide insight into key target areas for intervention for young children with ASD (especially those who are higher-functioning) and focused early learning strategies for their siblings who are at a substantially higher risk of having the BAP with mild impairments in ToM. Based on our findings, we can speculate that in addition to natural experiences that promote development of ToM (Astington & Jenkins, 1995; Dunn et al., 1991; Hughes & Dunn, 1998; Lewis et al., 1996; Meins et al., 2002; Ruffman et al., 1998), educational exercises that place a greater cognitive demand on EF such as cognitive flexibility and inhibition may lead to improved social outcomes later in life. Future research into potential mediators and moderators of EF and advanced ToM, as well as the relationship between EF and ToM within other clinical populations, may provide further insight into the deeper mechanisms underlying social impairments.

Conclusion

This study found that approximately one in two adolescents and adults with the BAP exhibited a reduced ability to detect and interpret a social faux pas. Language-mediated cognitive flexibility and inhibition, verbal generativity, and working memory predicted unimpaired advanced ToM abilities. While general intellect and EF abilities were unimpaired

in the sample, overall increased EF abilities appeared to play a protective role for unimpaired ToM abilities. Although males had a significant cognitive and verbal advantage over females, there were no gender differences in advanced ToM abilities, suggesting that males may be more severely affected. The overall results suggest that impaired advanced ToM abilities may be a key endophenotype in a subgroup of individuals with the BAP.

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Tables

Table 1

Mean Wechsler Abbreviated Scale of Intelligence (WASI) Scores of Participants

	<i>Overall Sample (N=29)</i>		<i>Males (n=13)</i>		<i>Females (n=16)</i>		<i>t</i>	<i>p</i>
	<i>M (SD)</i>	Range	<i>M (SD)</i>	Range	<i>M (SD)</i>	Range		
FSIQ	109.45 (16.90)	67-133	118.46 (8.81)	104-133	102.13 (18.53)	67-132	3.12	.005
VIQ	110.10 (16.35)	78-135	118.31 (10.98)	100-135	103.44 (17.24)	78-134	2.69	.012
PIQ	106.38 (17.11)	58-127	114.08 (8.69)	102-127	100.13 (19.82)	58-126	2.53	.019

Note. FSIQ = Full-Scale IQ; VIQ = Verbal IQ; PIQ = Performance IQ. Significant results ($p \leq .05$) are in bold.

Table 2*Executive Functions Measured by the Delis-Kaplin Executive Function System (D-KEFS)*

Subtest	Executive Functions
Trail Making	Flexibility, working memory
Verbal Fluency	Flexibility, inhibition, working memory, verbal generativity
Design Fluency	Flexibility, inhibition, working memory, non-verbal generativity
Color-Word Interference	Flexibility, inhibition, working memory
Sorting	Flexibility, working memory, problem solving
Twenty Questions	Working memory, problem solving
Word Context	Flexibility, working memory, deductive reasoning
Tower	Inhibition, working memory, spatial planning, rule learning
Proverbs	Verbal abstraction

Note. The Twenty Questions and Word Context subtests were not administered.

Table 3*Characteristics of Participants based on Performance on the Faux Pas Test*

	Unimpaired			Impaired			<i>U</i>	<i>z</i>	<i>p</i>
	<i>n</i>	<i>Md (SD)</i>	<i>Range</i>	<i>n</i>	<i>Md (SD)</i>	<i>Range</i>			
Age	16	39.00 (19.45)	13-78	13	60.00 (21.99)	15-80	73.50	-1.338	.181
								<i>X</i> ^{2†}	<i>p</i>
Gender	16	-	-	13	-	-		.060	.806
Male	8	-	-	5	-	-		-	-
Female	8	-	-	8	-	-		-	-

Note. The participants' scores on the Faux Pas test ranged from 24-40 ($M=35.45$, $SD=4.77$). Significant results ($p \leq .05$) are in bold.

[†] A Chi-square test for independence with Yates Continuity Correction was used to test for gender differences between groups.

Table 4*Correlations between the Faux Pas Test and D-KEFS Scores*

Subtest	<i>M (SD)</i>	Pearson		Spearman	
		<i>r</i>	<i>p</i>	<i>r_s</i>	<i>p</i>
Trail Making					
Visual Scanning	8.36 (3.92)	.172	.383	.259	.184
Number Sequencing	8.93 (3.72)	.368	.050	.402	.031
Letter Sequencing	9.52 (3.45)	.519	.004	.448	.015
Number-Letter Switching	9.28 (3.47)	.223	.245	.193	.316
Motor Speed	10.64 (1.73)	.138	.484	.181	.358
Verbal Fluency					
Letter Fluency	11.17 (3.71)	.225	.240	.256	.179
Category Fluency	11.79 (3.68)	.182	.344	.172	.372
Category Switching Total Correct	12.48 (3.55)	.136	.480	.290	.127
Category Switching Total Accuracy *	13.54 (2.66)	.392	.039	.539	.003
Design Fluency					
Filled Dots	10.76 (2.97)	.169	.380	.255	.181
Empty Dots	10.62 (3.23)	.222	.247	.204	.288
Fluency Switch	11.48 (3.11)	.281	.140	.383	.040
Total Correct Composite *	11.29 (2.51)	.366	.056	.444	.011
Color-Word Interference					

Color Naming	9.83 (2.32)	.428	.021	.465	.011
Word Naming	9.72 (3.05)	.289	.128	.207	.281
Inhibition	10.79 (2.40)	.087	.655	.163	.397
Inhibition/Switching *	9.41 (2.95)	.401	.031	.340	.071

Table 4 Continued

Sorting

Free Sorting Confirmed Correct Sorts *	10.66 (2.91)	.308	.104	.425	.022
Free Sorting Description	10.90 (4.40)	.366	.051	.445	.016

Tower

Total Achievement	10.18 (1.93)	.304	.115	.361	.059
Mean First-Move Time	12.77 (3.56)	-.479	.098	-.398	.178
Time per Move Ratio	9.31 (3.33)	-.082	.789	.020	.948
Move Accuracy Ratio	8.08 (2.36)	.425	.148	.491	.089
Total Rule Violations	54.92 (44.78)	.421	.152	.385	.195

Proverb

Total Achievement - Free Inquiry	12.46 (2.45)	.234	.234	.194	.363
Total Achievement - Multiple Choice	70.24 (38.59)	.172	.412	.140	.506
Accuracy	12.8 (2.51)	.374	.086	.240	.282
Abstraction	11.59 (1.74)	-.023	.921	.100	.657

Note. Significant results ($p \leq .05$) are in bold.

* Selected as a predictor variable for discriminant function analysis

Table 5*Mean D-KEFS Scaled Scores Selected for Analyses*

	Overall Sample		Males (n=13)		Females (n=16)		<i>t</i>	<i>p</i>
	<i>M (SD)</i>	Range	<i>M (SD)</i>	Range	<i>M (SD)</i>	Range		
VF	13.54 (2.66)	8-18	13.50 (2.75)	8-18	13.56 (2.68)	9-18	-.060	.952
DF	11.29 (2.51)	6-16	11.75 (1.36)	10-14	10.94 (3.11)	6-16	.934	.361
CW	9.41 (2.95)	2-14	9.54 (3.07)	2-14	9.31 (2.94)	2-14	.202	.842
S	10.66 (2.91)	6-18	12.23 (2.49)	8-18	9.38 (2.63)	6-15	2.978	.006

Note. Significant results ($p \leq .05$) are in bold.

VF = Verbal Fluency; DF = Design Fluency; CW = Color-Word Interference; S = Sorting.

Table 6*Differences in Cognition and Executive Functioning between groups with Unimpaired and Impaired Performance on the Faux Pas Test*

	Unimpaired (n=16)		Impaired (n=13)		<i>t</i>	<i>p</i>
	<i>M (SD)</i>	Range	<i>M (SD)</i>	Range		
FSIQ	116.7 (12.47)	88-133	100.46 (17.69)	67-132	2.905	.007
VIQ	116.94 (13.19)	91-135	101.69 (16.36)	78-134	2.780	.010
PIQ	112.38 (14.59)	70-127	99.00 (17.60)	58-121	2.239	.034
VF	14.80 (2.43)	9-18	12.08 (2.18)	8-15	3.104	.005
DF	12.25 (1.98)	8-16	10.00 (2.63)	6-116	2.586	.016
CW	10.50 (1.63)	14-18	8.08 (3.66)	2-14	2.214	.042
S	11.56 (2.83)	6-12	9.54 (2.70)	9-15	1.957	.061

Note. The participants' scores on the Faux Pas test ranged from 24-40 ($M=35.45$, $SD=4.77$), with a total possible score of 40. Significant results ($p \leq .05$) are in bold. FSIQ = Full-Scale IQ; VIQ = Verbal IQ; PIQ = Performance IQ. VF = Verbal Fluency; DF = Design Fluency; CW = Color-Word Interference; S = Sorting.

Table 7*Standardized Coefficients and Loading Matrix*

	Pooled Within-Groups Correlations [†]	Standardized Canonical Discriminant Function Coefficients
VF	0.660	.944
DF	0.478	-
CW	0.469	.803
S	0.164	-
FSIQ	0.138	-

Note. Absolute values > 0.4 are in bold.

VF = Verbal Fluency; DF = Design Fluency; CW = Color-Word Interference; S = Sorting; FSIQ = Full-Scale IQ

[†] Pooled within-groups correlations between discriminating variables and standardized canonical discriminant functions

Figure Legends

Figure 1. Phenotyping process for individuals participating in the broader study. Participants without a clinical diagnosis of ASD were assessed for the BAP. Individuals with clinical diagnoses of ASD, or who were suspected of having ASD, were assessed for ASD.

Figure 2. Distribution of the Faux Pas test scores. The dashed line indicates the cut-off value for impaired versus unimpaired performance as determined by a two-step cluster analysis. Individuals with scores of 37 or above were considered to have unimpaired performance. Possible Faux Pas test scores range from 0-40, with higher scores indicating better performance.

Figure 3. Classification of performance on the Faux Pas test based on the Verbal Fluency and Color-Word Interference subtests of the D-KEFS. Individuals with impaired performance on the Faux Pas test are indicated by dark circles, while those with unimpaired performance are indicated by empty circles. Those with impaired performance tend to have VF and CW scores below the line.

Figure 1 Top

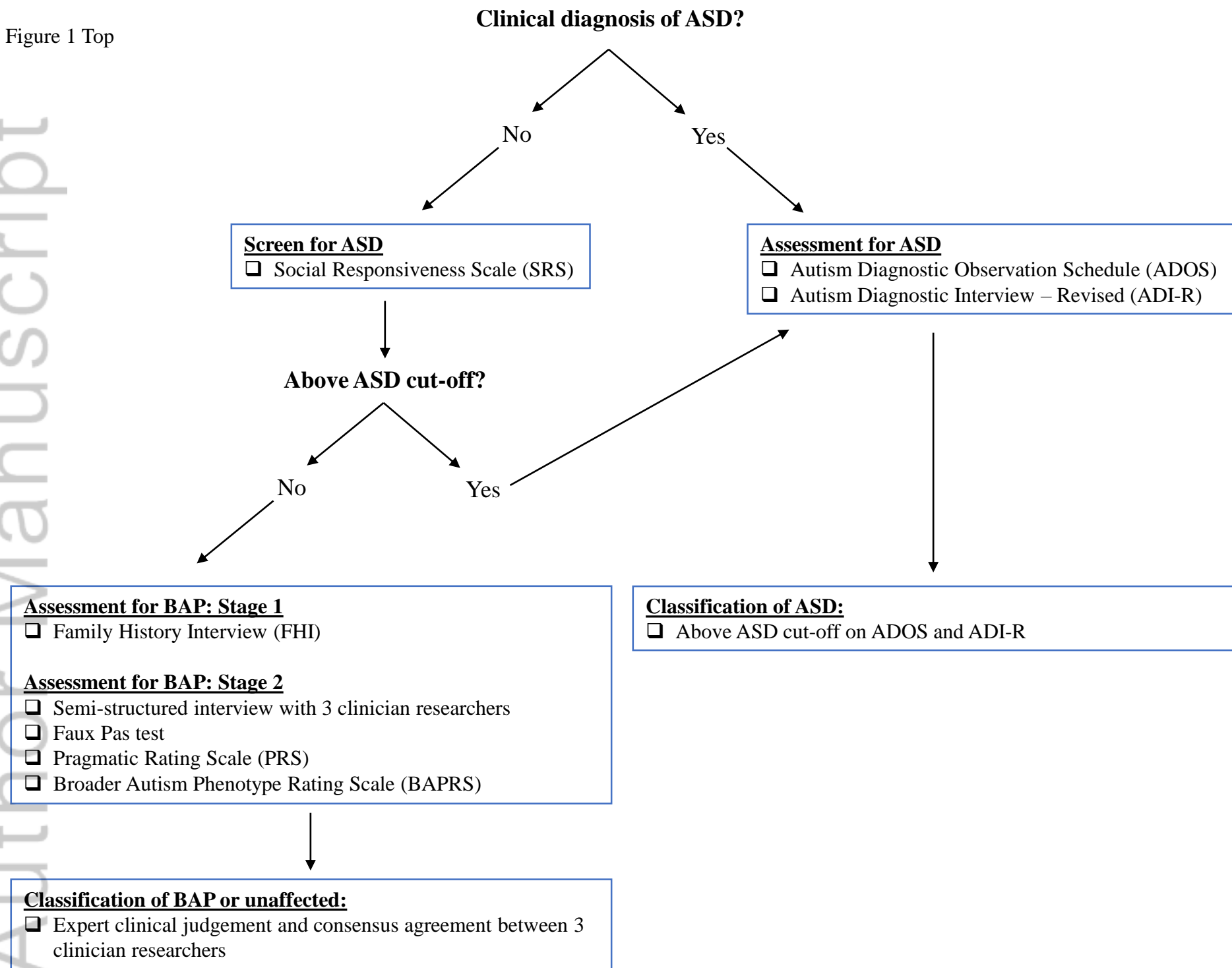


Figure 1. Phenotyping process for individuals participating in the broader study. Participants without a clinical diagnosis of ASD were assessed for the BAP. Individuals with clinical diagnoses of ASD, or who were suspected of having ASD, were assessed for ASD. This article is protected by copyright. All rights reserved.

Figure 2 top

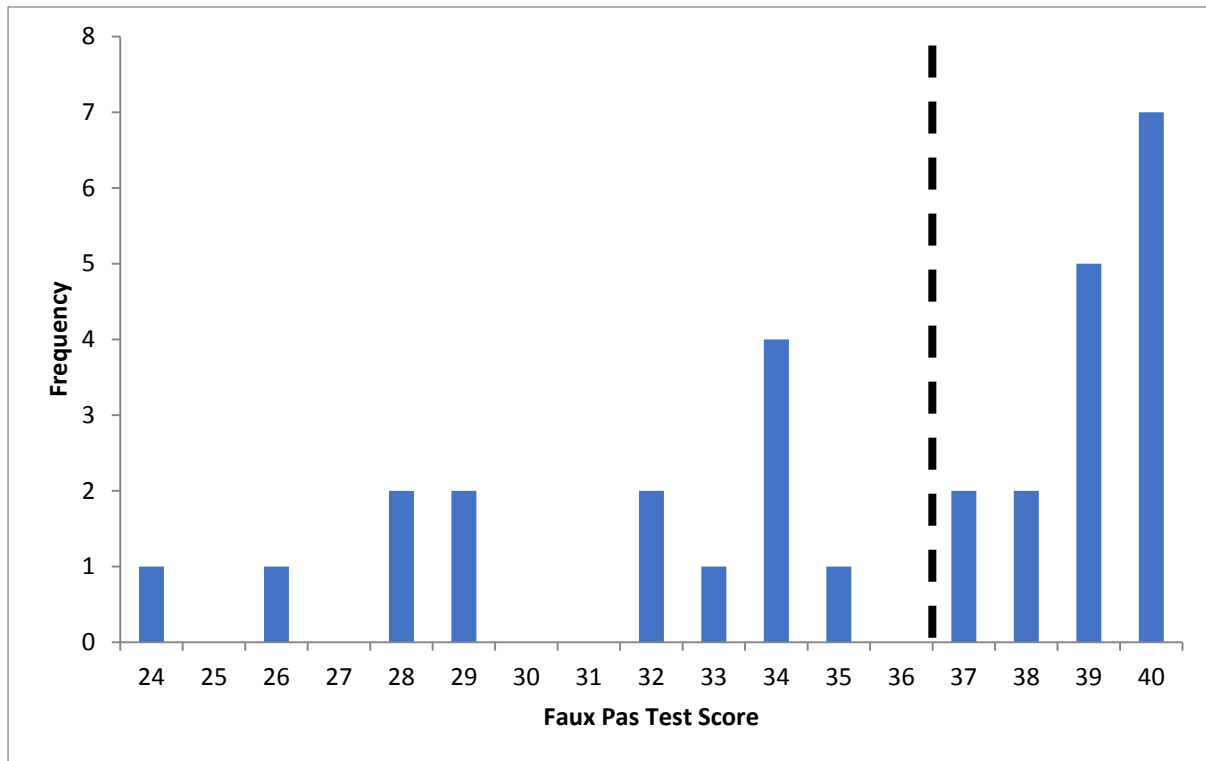


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Figure 3 top

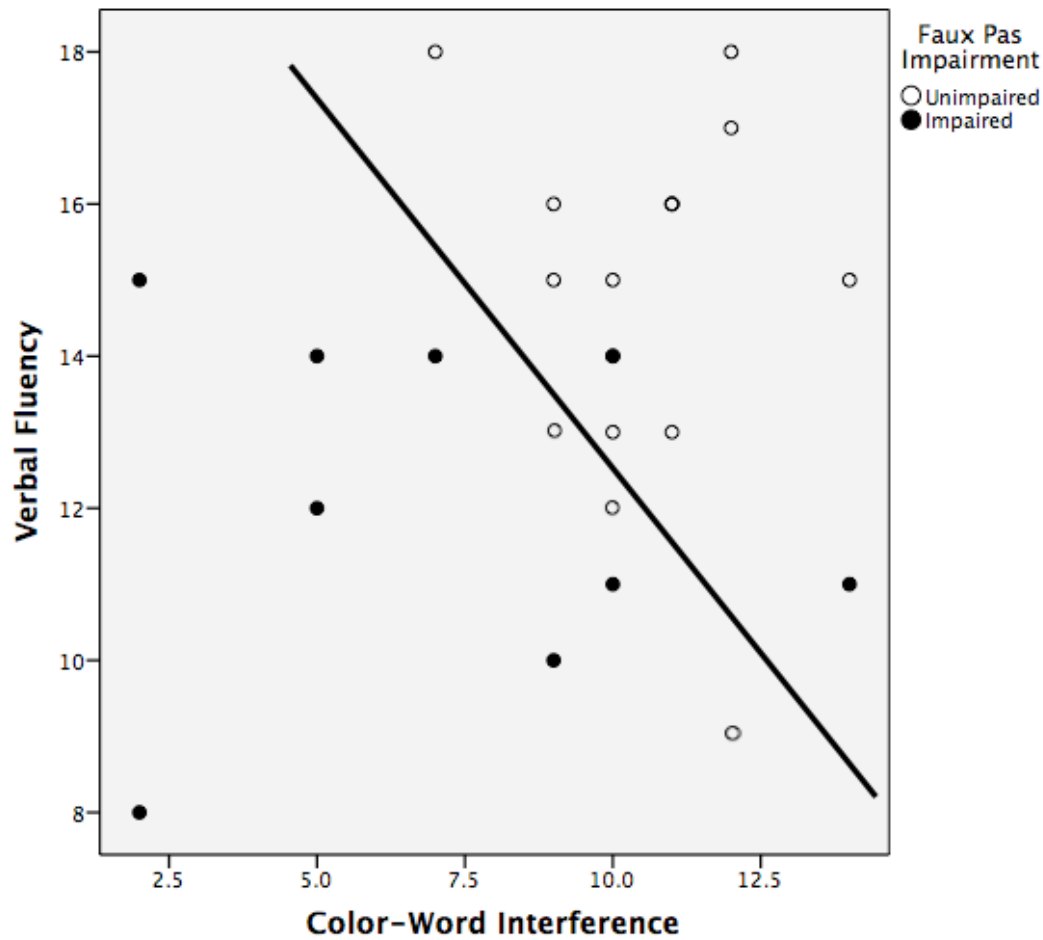


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