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# SOCIAL ATTENTION AND MEMORY IN YOUTH WITH ASD, ADNP SYNDROME, AND FOXP1 SYNDROME

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# SOCIAL ATTENTION AND MEMORY IN YOUTH WITH ASD, ADNP SYNDROME, AND FOXP1 SYNDROME

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**ABSTRACT**

SOCIAL ATTENTION AND MEMORY IN YOUTH WITH ASD, ADNP SYNDROME, AND FOXP1 SYNDROME

Megan L. Braconnier-Krupa

The current study used eye tracking and behavioral measures to examine social attention and recognition memory in two rare genetic disorders commonly associated with autism spectrum disorder (ASD): ADNP syndrome and FOXP1 syndrome. Impairment in social functioning is considered a core deficit in ASD. Although individuals with ADNP syndrome and FOXP1 syndrome typically present with symptoms of ASD, research suggests several dissimilarities in symptom presence and severity between the groups. Thus, there is a need to clarify disorder-specific patterns of social attention and their relationships to observed social skills. A visual paired comparison eye tracking task was used to assess social attention and memory in youth with ADNP syndrome ($n$ = 18) and FOXP1 syndrome ($n$ = 9) compared to youth with idiopathic ASD ($n$ = 90) and typically developing controls ($n$ = 43). Diagnostic groups demonstrated similar within-group attention to social and non-social stimuli but differences in engagement with the presented stimuli depending on stimulus type and image location. Rare genetic disorder groups also demonstrated differences in preferential looking patterns for novel versus familiar stimuli. Correlations with behavioral measures were examined for each group, with the social problems domain of the CBCL demonstrating the highest correlations with the ADNP group and the socialization domains of caregiver-report interviews (i.e., Vineland-3, ADI-R)

demonstrating the highest correlations with the FOXP1 group. These findings offer a characterization of social attention and memory patterns in youth with these rare conditions that yields insight into subtle differences in information processing across disorders.

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## TABLE OF CONTENTS

## LIST OF FIGURES

## Introduction

Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by deficits in social communication, as well as repetitive behaviors, interests, or activities (American Psychological Association [APA], 2013). Common social impairments in this population include reduced interest in peers, abnormal social approach, difficulty initiating and maintaining social interactions, unusual eye contact, and a lack of awareness and understanding of the thoughts and feelings of others (APA, 2013). While individuals with ASD exhibit substantial variability in symptom presentation and severity, some degree of impairment in social functioning is considered a core feature of the disorder (Tager-Flusberg, 2010).

Research has suggested that reduced attention to social stimuli is one contributing factor to distinctive impairments in social interaction skills in those with ASD compared to typically developing (TD) individuals (Chita-Tegmark, 2016). As many social skills require the ability to attend to others' facial cues (e.g., eye contact, joint attention, recognizing emotions), atypical attention to faces is thought to underlie many social impairments common in ASD (Dawson et al., 2005). Indeed, a recent review and meta-analysis of 38 eye-tracking studies concluded that individuals with ASD exhibit reduced attention to social stimuli compared to TD controls, particularly when the stimuli contain added social content (i.e., more than one person) (Chita-Tegmark, 2016). Differences in attention to social stimuli have been shown to include atypical gaze patterns (e.g., increased attention to the mouth compared to the eyes; Klin et al., 2002; Norbury et al., 2009), longer duration spent locating faces and shorter duration spent attending to faces compared to TD controls (Riby & Hancock, 2009), lack of preference for social content

compared to non-social content (Falck-Ytter et al., 2013), increased attention to background objects (Shic et al., 2011), and reduced recognition memory (Chawarska & Shic, 2009). These differences in social attention, which have been shown to predict eventual ASD diagnosis, have been detected as early as the second year of life (Chawarska et al., 2013; Jones et al., 2008). In addition to problems with social interactions, this atypical attention to faces can lead to problems with language acquisition, communication, and word recognition (Tenenbaum et al., 2014). Thus, it is important to understand patterns of social attention and memory in individuals with ASD in order to develop appropriate interventions to mitigate associated social and language deficits.

In the past decade, researchers have identified a number of autism candidate genes based on ASD symptomology associated with specific gene mutations or deletions. While there is a high prevalence of ASD diagnoses among those with the rare genetic disorders associated with these gene mutations, each exhibits a distinct clinical presentation or phenotype. For example, Phelan-McDermid syndrome (PMS), which is caused by a mutation or deletion in the chromosome region 22q13.3 and is currently one of the most frequent single-gene causes of ASD and ID (Leblond et al., 2014), is characterized by cognitive delays and intellectual disability, minimal or absent speech, hypotonia, and inattention (De Rubeis et al., 2018; Phelan & McDermid, 2011; Soorya et al., 2013). Despite high rates of ASD in this population, fewer individuals with PMS exhibit problems with social approach and engagement compared to idiopathic ASD (iASD; Richards et al., 2017). Conversely, individuals with PMS exhibit more problems with directing others' attention compared to individuals with iASD (Richards et al.,

2017). In this way, the overlap of social symptom presentation in ASD and related genetic disorders is not yet clear.

In addition to the *SHANK*3 gene disrupted in PMS (Harony-Nicolas et al., 2015), Activity Dependent Neuroprotector Protein (ADNP) and forkhead-box protein P1 (FOXP1) are among the most common autism risk genes (Arnett et al., 2018; Siper at al., 2017). ADNP syndrome (also known as Helsmoortel-Van der Aa Syndrome), caused by a mutation in the ADNP gene on chromosome 20q13.13, is characterized by broad medical problems (e.g., gastrointestinal, visual, cardiovascular), mild to severe delays in cognitive functioning, speech and motor delays, craniofacial dysmorphology, and unique biomarkers (i.e., early tooth eruption) (Gozes et al., 2017; Helsmoortel et al., 2014; Van Dijck et al., 2019). The ADNP gene has also been shown to play a role in object recognition and social memory in animal models (Malishkevich et al., 2015). While approximately 65% of individuals with ADNP syndrome meet criteria for ASD (Arnett et al., 2018; Van Dijck et al., 2019), there is considerable variability in the severity of ASD symptoms in this population (Arnett et al., 2018; Helsmoortel et al., 2014). Individuals with ADNP syndrome have been shown to exhibit less severe social affect symptoms and more use of nonverbal social communication (e.g., directing smiles, following another's gaze, expressing shared enjoyment, attempting to gain others' attention) compared to those with iASD, despite greater weaknesses in intellectual and verbal abilities (Arnett et al., 2018).

FOXP1 syndrome, caused by haploinsufficiency of the forkhead-box protein P1, is characterized by global developmental delay, intellectual disability, speech and motor delays, and mild dysmorphic features (LeFevre et al., 2013; Siper et al., 2017).

Additionally, individuals with FOXP1 syndrome have been shown to present with complex psychiatric symptoms, including anxiety, attention problems, externalizing behaviors, and obsessive-compulsive behaviors (Hamdan et al., 2010; Siper et al., 2017). Similar to ADNP syndrome, most individuals with FOXP1 syndrome exhibit ASD symptoms, yet symptom severity varies greatly, with only approximately 25% meeting full diagnostic criteria for ASD (Siper et al., 2017). ASD symptomatology in individuals with FOXP1 has been associated with lower language abilities (Siper et al., 2017). Additionally, individuals with FOXP1 who meet criteria for ASD show greater impairment in the domain of restricted and repetitive behaviors compared to the social affect domain (Siper et al., 2017). Thus, while the behavioral phenotype of these rare disorders often includes autism symptoms, there are noteworthy differences in the clinical presentations of each group, especially with regard to social deficits.

Given the established association between atypical social attention and social skills deficits, understanding the processing and attention patterns of individuals with rare genetic disorders may help to elucidate the nature and origin of social impairments in these unique populations. To date, research examining the social processing patterns of individuals with known genetic causes of ASD is scarce. Recently, Guillory and colleagues (2021) examined social attention and recognition memory in individuals with PMS compared to those with iASD and TD controls using a visual paired comparison eye-tracking task. While both iASD and PMS groups evidenced similar looking times and recognition memory, the authors found differences between groups on looking patterns and preferences for novel versus familiar stimuli; the iASD group demonstrated less active looking back-and-forth between images and the PMS group demonstrated the

lowest preference for novel stimuli compared to TD controls. Furthermore, whereas the iASD group demonstrated a greater novelty preference for non-social stimuli, the PMS group did not show the same preference. This study provides early evidence of differences in social attention between individuals with iASD and individuals with a rare genetic disorder associated with ASD, which is valuable for clarifying the mechanisms of observed social deficits and strengths in this population and informing interventions tailored to their specific needs.

**Present Study and Hypotheses**

Building on the findings regarding social attention and recognition memory in PMS (Guillory et al., 2021), this study examined social attention and recognition memory in youth with ADNP syndrome and FOXP1 syndrome compared to youth with iASD and TD controls to clarify disorder-specific patterns of social attention and their relationships to observed social skills. Due to advances in genetic testing, the number of individuals identified as having one of these known genetic causes of ASD will continue to increase. Although individuals with ADNP syndrome and FOXP1 syndrome typically present with symptoms of ASD and often meet criteria for an ASD diagnosis, research has demonstrated several dissimilarities in symptom presence and severity between the two groups, particularly with regard to social functioning (Arnett et al., 2018; Siper et al., 2017), as individuals with ADNP syndrome exhibit significant strengths with regard to nonverbal social communication compared to those with iASD (Arnett et al., 2018) and only 25% of individuals with FOXP1 syndrome demonstrate symptoms severe enough to meet full criteria for an ASD diagnosis (Siper et al., 2017). Thus, there is a need to better

understand the unique clinical phenotypes of these rare genetic disorder populations associated with ASD.

Research that characterizes the social strengths and deficits of these groups will be essential in order to inform appropriate and effective treatments for these individuals and their families. As younger child age at the start of treatment has been shown to predict greater intervention gains for children on the autism spectrum (Itzchak & Zachor, 2011), informing early intervention services for those with rare genetic disorders could promote better outcomes for these unique groups. Moreover, as individuals with ADNP syndrome and FOXP1 syndrome often present with a myriad of related health and psychiatric concerns requiring time-intensive interventions (Siper et al., 2017; Van Dijck et al., 2019), understanding the social strengths of these groups could reduce time spent on social skills treatment and improve quality of life for the family.

This study was the first to examine social attention and processing in individuals with ADNP syndrome and FOXP1 syndrome. I utilized data from a visual paired comparison eye tracking task (detailed below) to better understand attentional processes and memory in youth with ADNP syndrome and FOXP1 syndrome when presented with social and non-social stimuli. I then utilized various informant-report measures to better understand the presentation of social symptoms in these populations and how their clinical phenotype is related to or informed by their attentional processes. I hypothesized that the iASD and ADNP groups would demonstrate reduced social attention and memory compared to the TD control group due to characteristic social deficits, but that the iASD group would demonstrate more severe impairments in social attention compared to the ADNP group due to strengths in nonverbal social communication in

ADNP (including following another's gaze and attempting to gain another's attention through eye contact; Arnett et al., 2018). I further hypothesized that the FOXP1 group would demonstrate a global deficit in attention and memory (as opposed to an attention deficit for social material) due to documented broad attentional difficulties and a lower prevalence of ASD in this population compared to ADNP syndrome (Hamdan et al., 2010; Siper et al., 2017). Finally, due to the role of ADNP in object recognition and social memory in animal models (Malishkevich et al., 2015), I hypothesized that the ADNP group would demonstrate the greatest deficits in recognition memory.

## Method

### Participants

Data reported here represent a secondary analysis of data collected by larger studies aimed at phenotyping rare genetic disorders associated with ASD. Eye tracking data were examined from 160 youth ($M_{age}$ = 8.34 years, SD = 3.85 years, age range = 2-18 years) with iASD ($n$ = 90), ADNP syndrome ($n$ = 18), and FOXP1 syndrome ($n$ = 9), as well as typically developing youth (TD, $n$ = 43). Two participants were excluded due to lack of sufficient eye tracking data (i.e., an average proportion of looking time during the *familiarization* trial $\leq$ .25, indicating the participant did not attend to the images for at least 25% of the total task duration) and one participant was excluded due to a comorbid ocular condition causing involuntary eye movements. All excluded participants were in the ADNP group. Study procedures were approved by the Program for the Protection of Human Subjects at the Icahn School of Medicine at Mount Sinai and the Institutional Review Board (IRB) at St. John's University. Consent was obtained from the participants' legal guardians.

The Autism Diagnostic Observation Schedule, Second Edition (ADOS-2; Lord et al., 2012), as well as clinical consensus among licensed psychologists and psychiatrists, were used to confirm clinical diagnosis in the iASD, ADNP, and FOXP1 groups. All participants in the iASD group, five participants in the ADNP group, and three participants in the FOXP1 group met DSM-5 diagnostic criteria for ASD (APA, 2013). Genetic mutations in the ADNP and FOXP1 groups were validated by Clinical Laboratory Improvement Amendments (CLIA)-certified clinical genetics testing laboratories using whole exome sequencing (WES).

## Measures

### *Demographic Measure*

Legal guardians completed a patient information form containing items regarding the participant's age, gender, ethnicity, and diagnosis.

### *Diagnostic and Behavioral Measures*

**Vineland Adaptive Behavior Scale, Third Edition (VABS-3; Sparrow et al., 2016)**. The VABS-3 is a structured interview administered to caregivers that is used to assess adaptive behaviors. A clinician codes caregiver responses regarding current adaptive skills in the domains of communication, daily living skills, socialization, and motor skills, with the domain scores yielding an adaptive behavior composite score. All VABS-3 assessments were administered and scored by a clinical psychologist or a trainee (e.g., graduate students or postdoctoral fellows) under the direct supervision of a psychologist who met requirements for reliability. This instrument has demonstrated strong internal consistency for all domains (Sparrow et al., 2016). The current study will focus specifically on the socialization domain of the VABS-3.

**Child Behavior Checklist (CBCL; Achenbach & Rescorla, 2001).** The CBCL is a 113-item parent-report questionnaire used to assess emotional and behavioral problems in children and adolescents. The 2001 revision of the CBCL consists of eight syndrome scales, including anxious/depressed, withdrawn/depressed, somatic complaints, social problems, thought problems, attention problems, rule-breaking behavior, and aggressive behaviors, with the scales yielding total scores for internalizing behaviors, externalizing behaviors, and total problems. Caregivers rate their agreement with items on a 3-point Likert-type scale ranging from "not true" to "very true or often true." This

instrument has demonstrated strong internal consistency, sensitivity, and specificity (Achenbach & Rescorla, 2001). The current study will focus on the social and attention problems syndrome scales of the CBCL.

**Apparatus**

Eye tracking data was collected using an EyeLink 1000 plus eye-tracker in head-free mode with a 17-inch LCD monitor and 1280 × 1024 pixel at 32 bits per color display, with a refresh rate of 60 Hz. Each participant completed a 5- or 13-point calibration prior to the start of the task. Data were acquired at 500 Hz. The EyeLink Experiment Builder software application was used to present the task.

**Testing and Experimental Procedure**

Eye tracking data were collected with participants seated in a chair or booster seat approximately 50 cm from the monitor and eye tracking apparatus in a dark, quiet room. Stimuli subtended a visual angle of 14.2°×10.2°. General verbal instructions were given prompting the participants to look at the images on the screen and to look at the dot in the center of the screen in between trials. In order to make the task accessible to participants with a range of cognitive and verbal abilities, comprehension of these instructions was not necessary, and the task proceeded as long as the participants were fixating on the images on the screen. The experimenter visually confirmed that the participant's gaze was fixated on the screen prior to starting each trial.

The visual paired comparison paradigm (Fagan, 1990; Rose et al., 2013) used in this study was comprised of social (i.e., achromatic faces) and non-social (i.e., multicolored shape patterns) visual stimuli against a black background developed by Rose and colleagues (2013) (see Figure 1). Following the procedure detailed by Rose and

colleagues (2013), the task consisted of a *familiarization* phase and a *test* phase. During an initial *familiarization* period, pairs of identical images were presented on the right and left halves of the monitor for either 5 (non-social) or 10 (social) sec, which excluded any time when the participant was not fixated on the monitor. In the *test* period, an image displayed during *familiarization* was then paired with a novel image for 5 sec, following which the familiar and novel images switched sides and remained on the screen for an additional 5 sec. The task included a total of nine trials (five social, four non-social) presented in a fixed order alternating between social and non-social image sets. A flashing target and a loud "spaceship" noise were used to reorient the participant's gaze to the monitor between trials.

**Plan for Data Analysis**

Areas of Interest (AOI) were defined as the rectangular area surrounding each image presented (AOI size: 14.2°×10.2° of visual angle). In the *familiarization* phase, total visit duration (TVD) in AOIs was calculated for both images and summed to determine total image exploration time. Social and non-social attention were then examined using a repeated measures analysis of variance (ANOVA) with image type (social, non-social) as the within-subjects variable and group (iASD, ADNP, FOXP1, TD) as the between-subject variable. Frequency of switching back and forth between the identical images during the familiarization period was examined using a repeated measures ANOVA. Global deficit in attention (defined by equal reductions relative to TD in *both* social and non-social attention as opposed to a significantly greater reduction in attention for social stimuli) was examined by comparing summed TVD in AOIs across image type (i.e., social and non-social) between the TD group and the rare genetic

disorder groups. Post-hoc analyses with Bonferroni corrections were conducted for significant results in order to examine which diagnostic group(s) drove effects and how the ADNP and FOXP1 groups differ from each other and from the iASD and TD groups.

In the *test* phase, TVD was calculated for each familiar and novel image both during the initial presentation and after the images switch sides. A preference score was calculated based on the average time spent in the AOI for novel vs. familiar to examine recognition memory. These scores were then examined using a repeated measures ANOVA. One sample *t*-tests were then conducted for each diagnostic group to examine preference for novel and familiar social and non-social stimuli relative to zero (i.e., no preference). Global deficit in recognition memory (defined by equal deficits in memory for *both* social and non-social images as opposed to a significantly greater deficit in memory for social material) were tested by comparing summed preference scores across image type (i.e., social and non-social) between the TD group and the rare genetic disorder groups. All analyses were Bonferroni corrected with a two-tailed $p < 0.05$ significance standard.

Finally, correlations between social attention and preference during the eye tracking task and various caregiver-report behavioral measures were examined separately for the ADNP and FOXP1 groups. The behavioral data was first tested for normality to determine whether parametric or non-parametric analyses were appropriate. Scores on behavioral measures were then correlated with TVD in AOIs during social image sets in the *familiarization* phase to examine the relationship between observed or reported social behavior and social attention. Scores on behavioral measures were also correlated with preference scores during the *test* phase to examine the relationship between observed or

reported social behavior and social recognition memory. Specifically, caregiver-interview data from the socialization domain of the VABS-3 and caregiver-report data on social and attention problems from the CBCL were examined.

## Results

### Preliminary Analyses

Data were first visually inspected for outliers and normality using Q-Q plots prior to analyses. Normalcy was then established through Kolmogorov-Smirnov tests.

### Demographic Measures

The final sample showed no significant difference among groups for age ($F_{3,156} = 0.18$, $p = 0.79$, $\eta_p^2 = 0.006$). A significant sex difference was found among the groups ($F_{3,156} = 5.84$, $p < 0.001$, $\eta_p^2 = 0.10$). Post-hoc analyses revealed that the iASD group had a greater proportion of males compared to the TD ($p < 0.001$), ADNP ($p < 0.001$), and FOXPI ($p < 0.001$) groups. This finding is consistent with observed population gender differences in rates of ASD (Baio et al., 2018).

### Social and Non-social Attention

Attention to social and non-social stimuli was compared among groups during the *familiarization* phase (see Figure 2). There was no significant main effect of stimulus type ($F_{1,156} = 0.47$, $p = 0.71$, $\eta_p^2 = 0.006$). Follow up within group tests of looking time for social versus non-social stimuli confirmed no differences in social attention in any group (ADNP: $t(17) = -0.65$, $p = 0.52$; FOXP1: ($t(8) = 0.35$, $p = 0.73$); ASD: ($t(89) = -1.24$, $p = 0.22$); TD: ($t(42) = 1.71$, $p = 0.10$). This finding indicates similar attention to social and non-social stimuli across groups.

Differences in gaze patterns were analyzed by examining the rate of saccade entries into images (see Figure 3). A significant main effect of group ($F_{3,156} = 5.38$, $p < 0.001$, $\eta_p^2 = 0.09$) and stimulus type ($F_{1,156} = 5.22$, $p < 0.001$, $\eta_p^2 = 0.08$), as well as a significant group by stimulus type interaction ($F_{3,156} = 5.84$, $p < 0.001$, $\eta_p^2 = 0.10$), was

detected for entry rate into AOIs during familiarization. Post-hoc analyses revealed that this effect was driven by reduced rate of AOI entry in the ADNP group in the non-social condition relative to both ASD ($p = 0.02$) and TD ($p < 0.001$), and in the social condition relative to ASD only ($p = 0.01$). Follow-up one sample t-tests examining the AOI entry rate difference between image types (i.e., social vs. non-social) revealed that the TD group demonstrated the largest difference in AOI entry rate between social and non-social stimuli (mean difference: 0.39; $p < 0.001$), favoring non-social images. The iASD group demonstrated the next largest difference in AOI entry rate (mean difference: 0.15; $p < 0.001$), followed by the ADNP group (mean difference: 0.08; $p = 0.04$), both favoring non-social images. The FOXP1 group did not demonstrate a significant difference in rate of entry in social compared to non-social AOIs (mean difference: 0.11; $p = 0.22$). There was no significant main effect of image location (i.e., right versus left; $F_{1,156} = 0.26$, $p = 0.72$, $\eta_p^2 = 0.20$; see Figure 4), or group by location interaction ($F_{3,156} = 0.45$, $p = 0.71$, $\eta_p^2 = 0.007$) Overall, engagement with the presented stimuli differed among diagnostic groups by stimulus type (i.e., social vs. non-social); however, dwelling times for social and non-social stimuli did not differ within groups.

Global deficits in attention were detected in the rare genetic disorder groups compared to the iASD and TD groups ($F_{3,156} = 15.04$, $p < 0.001$, $\eta_p^2 = 0.22$; see Figure 5). The ADNP group demonstrated significantly reduced attention in comparison to the iASD ($p < 0.01$) and TD groups ($p < 0.001$), while the FOXP1 group differed significantly from the TD group ($p = 0.04$), but not the ASD group ($p = 0.9$). The ADNP group evidenced the largest difference in overall attention compared to the TD group

(mean difference: 0.49 TVD in AOIs), followed by the FOXP1 group (mean difference: 0.27 TVD in AOIs).

**Social and Non-social Memory**

Preference for novel and familiar stimuli was evaluated during the *test* phase to assess for social and non-social recognition memory (see Figure 6). A significant main effect of test period (i.e., pre- vs. post-switch) was detected ($F_{1,156} = 145.21$, $p = < 0.001$, $\eta_p^2 = 0.56$), as well as a significant test period by stimulus type interaction ($F_{1,156} = 19.36$, $p = < 0.001$, $\eta_p^2 = 0.17$) and a significant group by stimulus type interaction ($F_{3,156} = 8.51$, $p = < 0.001$, $\eta_p^2 = 0.13$). Follow up analyses indicated that participants looked less at the novel image during the post-switch period in the TD (social mean difference: 0.33, $p < 0.001$; non-social mean difference: 0.28; $p < 0.001$) and iASD (social mean difference: 0.25, $p < 0.001$; non-social mean difference: 0.25; $p < 0.001$) groups. The FOXP1 group demonstrated this pattern for social images (mean difference: 0.36; $p < 0.01$), but not non-social images (mean difference: 0.27; $p = 0.06$), while the ADNP group demonstrated no preferences across test period (social mean difference: 0.13, $p < 0.22$; non-social mean difference: 0.19, $p < 0.08$). A significant effect of stimulus type was also detected ($F_{3,156} = 15.24$, $p < 0.001$, $\eta_p^2 = 0.23$). Across groups, participants spent more time looking at non-social novel images compared to social novel images (mean difference: 0.12; $p < 0.001$).

Bonferroni corrected one sample *t*-tests were conducted for each diagnostic group to examine preference for novel and familiar social and non-social stimuli relative to zero (i.e., no preference). Across groups, participants demonstrated novelty preference for both social and non-social stimuli in the pre-switch period; however, the rare genetic

disorder groups demonstrated reduced novelty preference for certain stimulus types in the post-switch period. It was found that the TD group had a significant novelty preference in the pre-switch and post-switch periods for both stimulus types (social, pre-switch: 0.53, $p < 0.001$; non-social, pre-switch: 0.65, $p < 0.001$; social, post-switch: 0.20, $p < 0.001$; non-social, post-switch: 0.36, $p < 0.001$). Similarly, the iASD group demonstrated a significant novelty preference in both test periods for both stimulus types (social, pre-switch: 0.31, $p < 0.001$; non-social, pre-switch: 0.44, $p < 0.001$; social, post-switch: 0.06, $p = 0.04$; non-social, post-switch: 0.19, $p < 0.001$). In the ADNP group, participants demonstrated a significant novelty preference in the pre-switch period for both stimulus types (social, pre-switch: 0.22, $p < 0.01$; non-social, pre-switch: 0.37, $p < 0.001$). During the post-switch period, the ADNP group showed a novelty preference for non-social stimuli (0.19, $p = 0.01$), but neither novelty nor familiarity preference for social stimuli (0.09, $p = 0.13$). The FOXP1 group also demonstrated a significant novelty preference in the pre-switch period for both stimulus types (social, pre-switch: 0.60, $p < 0.001$; non-social, pre-switch: 0.53, $p < 0.001$); however, dissimilar to the ADNP group, the FOXP1 group showed a novelty preference for social stimuli (0.24, $p = 0.03$) during the post-switch period. The FOXP1 group showed neither novelty nor familiarity preference for non-social stimuli (0.26, $p = 0.11$) in the post-switch period. No groups demonstrated familiarity preference for either type of stimulus during pre- or post-switch periods. Overall, all diagnostic groups demonstrated a preference for novel stimuli during the pre-switch periods, but the rare genetic disorder groups demonstrated reduced novelty preference for social (ADNP) or non-social (FOXP1) images during the post-switch

period. These findings suggest deterioration of memory in the rare genetic disorder groups by the post-switch test phase that may be stimulus class specific.

Finally, global deficits in recognition memory were detected in the ADNP and ASD groups, but not the FOXP1 group ($F_{3,156} = 13.39$, $p < 0.001$, $\eta_p^2 = 0.20$; see Figure 7). The ADNP group demonstrated significantly reduced attention in comparison to the TD ($p < 0.001$) and FOXP1 groups ($p = 0.05$), while the ASD group differed significantly from the TD group ($p < 0.001$). The FOXP1 group did not differ significantly in recognition memory compared to the TD group (p = 0.9). The ADNP group evidenced the largest difference in overall memory compared to the TD group (mean difference: 0.87), followed by the ASD group (mean difference: 0.74).

**Behavioral Measures**

Correlations between social attention and preference and caregiver-report behavioral measures were examined separately for the ADNP and FOXP1 groups. The data were examined for normality, and it was determined that non-parametric analyses were appropriate due to outliers. The findings are reported separately for each rare genetic disorder group below.

In the ADNP group, low to medium Spearman rho values were found between caregiver-report of social and attention problems on the CBCL and social attention and memory; however, no correlation was found between caregiver-report on the Vineland-3 of socialization skills in daily settings and eye tracking metrics of social attention and memory. More specifically, a medium, positive correlation was found between the social problems domain of the CBCL and both social attention ($\rho = 0.45$) and social memory ($\rho = 0.49$), and a low, positive correlation was found between the attention problems domain

of the CBCL and social attention ($\rho = 0.18$), but no correlation was found between the attention problems domain of the CBCL and social memory ($\rho = 0.04$). The socialization domain of the Vineland-3 was not found to be correlated with social attention ($\rho < 0.01$) or social memory ($\rho = -0.11$). Overall, the social problems domain of the CBCL demonstrated the highest correlation with eye tracking metrics of social memory and attention in the ADNP group.

In the FOXP1 group, caregiver-report of socialization skills on the Vineland-3 demonstrated higher correlations with eye-tracking metrics compared to a caregiver-report of social and attention problems on the CBCL. No correlation was found between the social problems domain of the CBCL and social attention ($\rho = -0.05$), but a medium, positive correlation was found between this domain and social memory ($\rho = 0.31$). A low, negative correlation was found between the attention problems domain of the CBCL and social attention ($\rho = -0.22$), but no correlation was found between the attention problems domain of the CBCL and social memory ($\rho = 0.09$). The socialization domain of the Vineland-3 was found to have a medium, positive correlation with social memory ($\rho = 0.45$), but no correlation with social attention ($\rho = 0.10$). Overall, the socialization domain of the Vineland-3 demonstrated the highest correlation with eye tracking patterns in the FOXP1 group.

## Discussion

The present study examined social attention and memory in youth with two rare genetic disorders commonly associated with ASD, ADNP syndrome and FOXP1 syndrome, to better understand these aspects of social cognition in these diagnostic groups. While these aspects of cognition have been examined in individuals with ASD, this study is the first to examine social attention and memory in individuals with ADNP syndrome and FOXP1 syndrome. I found that diagnostic groups demonstrated similar attention to social and non-social stimuli (i.e., similar time spent looking and becoming familiar with social and non-social images) within each group; however, engagement with the presented stimuli during this looking time differed between diagnostic groups by stimulus type (i.e., social vs. non-social). Further, diagnostic groups demonstrated differences in preferential looking patterns for novel versus familiar stimuli. Specifically, while all diagnostic groups demonstrated a preference for novel stimuli during the time that the novel images were first presented, the rare genetic disorder groups demonstrated stimulus-type-specific reductions in novelty preference for social (ADNP) or non-social (FOXP1) images following switched image location, suggesting deterioration of memory. Global deficits in attention were found in both rare genetic disorder groups compared to typically developing controls, while global deficits in memory were found in the ADNP and iASD groups, but not the FOXP1 group, suggesting a strength in recognition memory for youth with FOXP1 syndrome compared to those with idiopathic autism. Correlations were conducted between patterns of social attention and memory on eye tracking tasks and parent-reported social impairment in the rare genetic disorder groups. Non-parametric analyses revealed that the social problems domain of the CBCL demonstrated

the highest correlation with eye tracking patterns in the ADNP group, while the socialization domain of the Vineland-3 demonstrated the highest correlation with eye tracking patterns in the FOXP1 group.

My hypothesis that the iASD and ADNP groups would demonstrate reduced social attention and memory compared to the TD control group, but that the iASD group would demonstrate more severe impairments in social attention compared to the ADNP group, was partially supported. The iASD and ADNP groups both favored non-social images compared to social images, with the iASD group demonstrating a greater preference for non-social images than the ADNP group. However, the TD group also favored non-social images, and the ADNP group demonstrated reduced social attention compared to the iASD and TD groups, suggesting more severe impairments in social attention than in the iASD group. This finding may indicate that established nonverbal social communication strengths in individuals with ADNP syndrome (i.e., following another's gaze, attempting to gain another's attention through eye contact; Arnett et al., 2018) may not be sustained in social interactions due to difficulties with attention. Further, the ADNP group demonstrated deterioration in recognition memory, while the iASD group did not. This finding was likely impacted by global deficits in attention and memory found in participants with ADNP syndrome (detailed below), which may supersede observed deficits to related to stimulus type (i.e., social vs. non-social).

My hypothesis that the FOXP1 group would demonstrate a global deficit in attention and memory (as opposed to an attention or memory deficit specific to social material) was partially supported. Global deficits in attention were detected in both rare genetic disorder groups compared to the TD groups. While the FOXP1 group evidenced a

global deficit in attention, the ADNP group demonstrated an even larger difference in overall attention compared to the TD group. This global deficit in attention is consistent with prior literature finding broad attentional difficulties in FOXP1 syndrome (Siper et al., 2017). As attentional difficulties have been found to be associated with social impairment across diagnoses (Mikami et al., 2019), attentional abilities represent an important target of intervention for individuals with rare genetic disorders and related attentional difficulties. Contrary to my hypothesis, the FOXP1 group did not differ significantly in social recognition memory compared to the TD group. Recognition memory for non-social images did differ between the FOXP1 and TD groups, suggesting a social memory strength in individuals with FOXP1 syndrome. This social strength may be related to the lower rates of ASD found in FOXP1 syndrome and may indicate a decreased need to provide interventions focused on memory skills in this population. Strengths in memory should be fostered to promote progress in other areas of cognitive functioning.

My hypothesis that the ADNP group would demonstrate the greatest deficits in recognition memory was supported. The ADNP group evidenced the largest difference in overall memory compared to the TD group, as well as reduced memory compared to the FOXP1 group. The ADNP group also demonstrated a reduced rate of AOI entry and both left and right looking time, indicating decreased overall exploration of the presented stimuli. This finding is consistent with previous research emphasizing the role of the ADNP gene in object recognition and social memory (Malishkevich et al., 2015) and suggests that recognition memory, especially over longer time periods, may be useful in differentiating ADNP syndrome from other rare genetic disorders associated with ASD

(e.g., FOXP1 syndrome). Understanding memory deficits in this population is important for guiding intervention practices, as youth with ADNP syndrome may require a greater focus on improving memory skills, more repetition, and accommodations for poor memory compared to youth with idiopathic ASD or other genetic conditions. Memory abilities should inform interventions provided to youth with ADNP syndrome to promote optimal cognitive, behavioral, and adaptive outcomes.

Behavioral measures demonstrated low to medium correlations with eye tracking metrics of social attention and memory. Several correlations were found to be consistent with expectations (i.e., attention problems were negatively correlated with social attention, social skills in daily activities were positively correlated with social memory). However, I found the direction of several observed correlations to be surprising, which may suggest that available measures for assessing social skills are not appropriate for rare disorder populations or that social challenges are not always related to social attention and memory for these groups. Specifically, social and attention problems were positively correlated with social attention and memory in the ADNP group, and social problems were positively correlated with social memory in the FOXP1 group. This suggests that increased social attention and memory are associated with increased social or attention problems. These inconsistent findings may reflect the small study sample (detailed under limitations) and a need to re-evaluate caregiver-report of social functioning with a larger sample size. However, this may also reflect that the CBCL does not adequately capture social strengths in these populations. These findings may also indicate that youth with ADNP and FOXP1 syndrome experience and social and attention problems related to factors other than their social attention and social memory abilities.

**Limitations**

This study was limited by factors inherent to examining rare genetic disorder populations. While the study sample represents a reasonable number of individuals identified as having these rare disorders and comparable samples to previous studies on individuals with FOXP1 (Arnett et al., 2018) and ADNP (Siper et al., 2017) gene mutations, analyses were limited by small sample sizes in the ADNP and FOXP1 groups. Further, small sample size precluded analyses examining differences in youth with rare conditions diagnosed with comorbid ASD versus those without an ASD diagnosis. As the quantity of individuals identified as having ANDP syndrome and FOXP1 syndrome continues to increase, future studies incorporating larger sample sizes should evaluate the extent to which cognitive functioning and diagnostic factors relate to patterns of social attention and memory.

The visual paired-comparison task was replicated from previous research in order to contextualize results against previously published studies. However, this study may have been affected by several task limitations. While Rose et al. (2013) calibrated the images in this task for equal attractiveness and habituation times within trials, it is possible that differences between stimuli for social and non-social images, as well as differences in presentation time, could affect results (Guillory et al., 2021). Previous work has also examined the limitations of novelty preference as an index of memory in visual paired-comparison tasks and suggested that other biological and environmental factors should be considered when characterizing recognition memory (Brown, 2007). Future research should utilize eye-tracking paradigms that minimize these task-specific concerns.

The behavioral measures utilized in this study relied on caregiver-report of social functioning. While informant-report measures are generally more time- and cost-effective than direct observation, observation often provides more detailed and accurate information about the behaviors children exhibit. Further, questionnaires designed for typically developing youth and youth with ASD may not accurately capture social strengths and weaknesses in youth with rare disorders. Future studies should utilize observational measures of social behaviors to better characterize social functioning in rare disorders and its relationship to attention and memory patterns. Direct observational measures of social functioning, such as the Social Avoidance Scale (SAS; Roberts et al., 2007, 2009), or the Child Sociability Rating Scale (CSRS; Moss et al., 2013), have been used to characterize social skills in genetic disorder populations (e.g., fragile X, Angelman, Cornelia de Lange, and Cri du Chat syndromes) in previous research and could offer important information on specific social deficits in rare genetic disorder populations. In addition, behavioral measures should be normed for rare disorder populations to characterize functioning more accurately within the disorder, as well as relative to other conditions.

**Conclusions**

In conclusion, the results of this study represent beginning efforts to characterize patterns of social attention and memory in youth with rare genetic disorders associated with ASD. While both rare genetic disorder groups demonstrated memory deterioration for specific stimulus types (i.e., social memory in ADNP syndrome, non-social memory in FOXP1 syndrome), as well as global deficits in attention, findings suggest that social memory abilities were intact for the FOXP1 group. This finding highlights an important

strength in this group given established rates of social, cognitive, and attention problems in individuals with FOXP1 syndrome. As research continues to identify genes related to autism symptomology and varied emotional, behavioral, and medical phenotypes, such studies will prove important for understanding nuanced differences in social behavior and mechanisms and thereby guiding effective intervention practices. Further, unique patterns of attention and memory on biobehavioral tasks may differentiate genetic disorders from idiopathic autism in a way that indicates genetic testing is warranted for children diagnosed with ASD. In this way, specific gene alterations can be identified, allowing individuals and families to obtain accurate diagnostic and medical information. Continued work is needed to further elucidate the strengths and needs of this unique and important population.

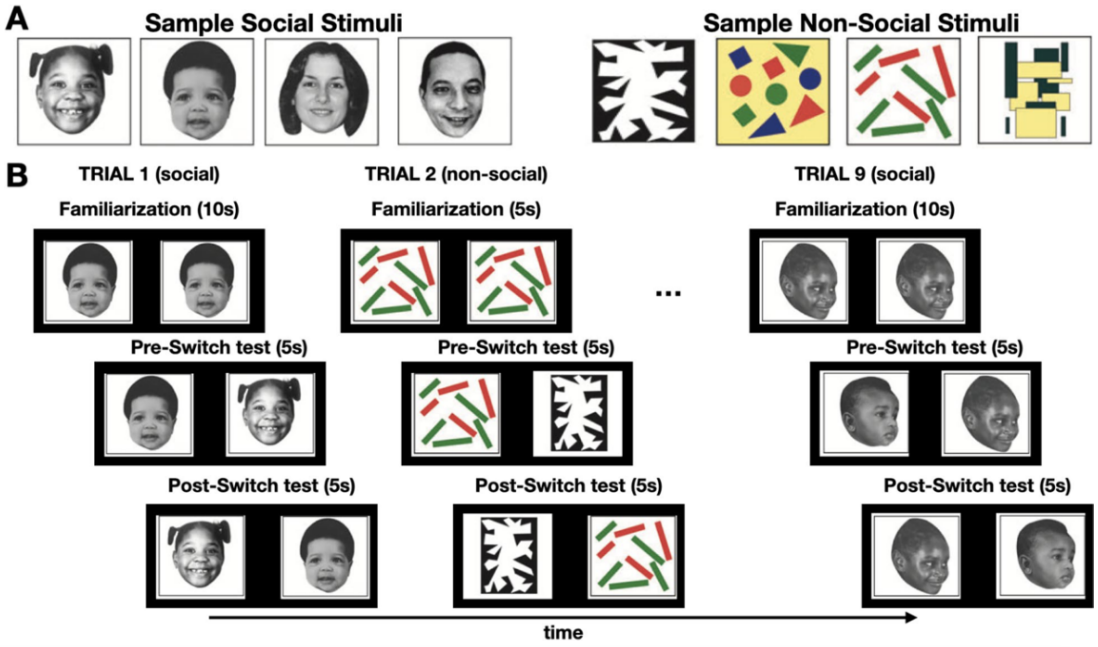


*Figure 1*. Visual paired-comparison paradigm sample from Guillory et al. (2021). (A) Sample of social and nonsocial images and (B) trial of visual-paired comparison task

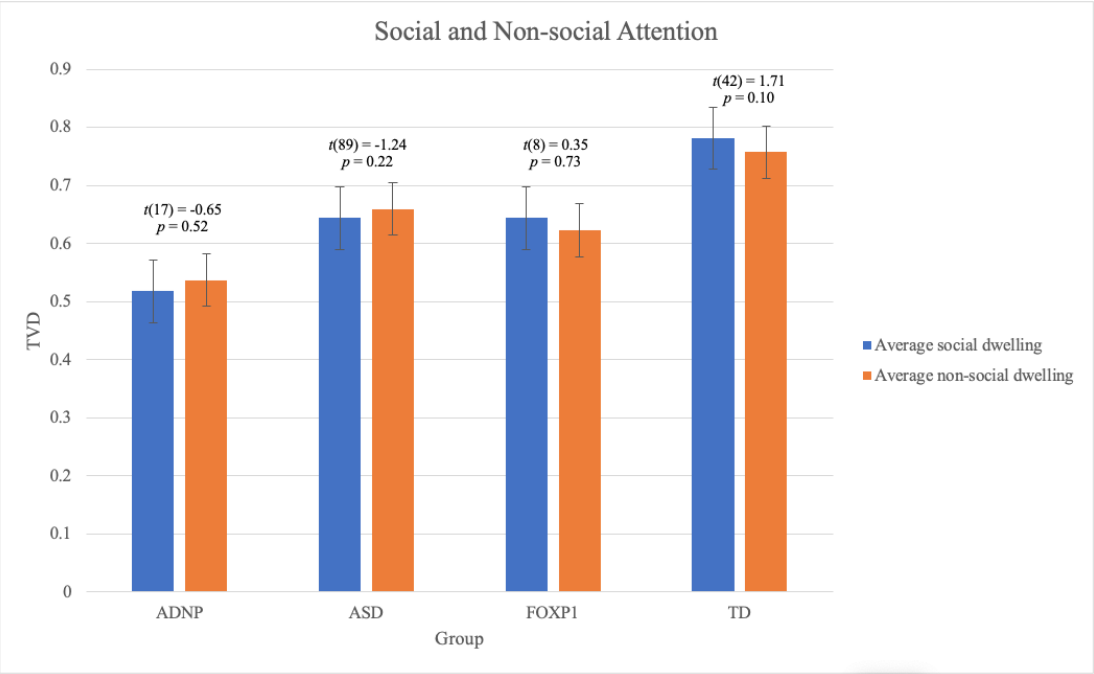


*Figure 2*. Average attention to social and non-social stimuli by group

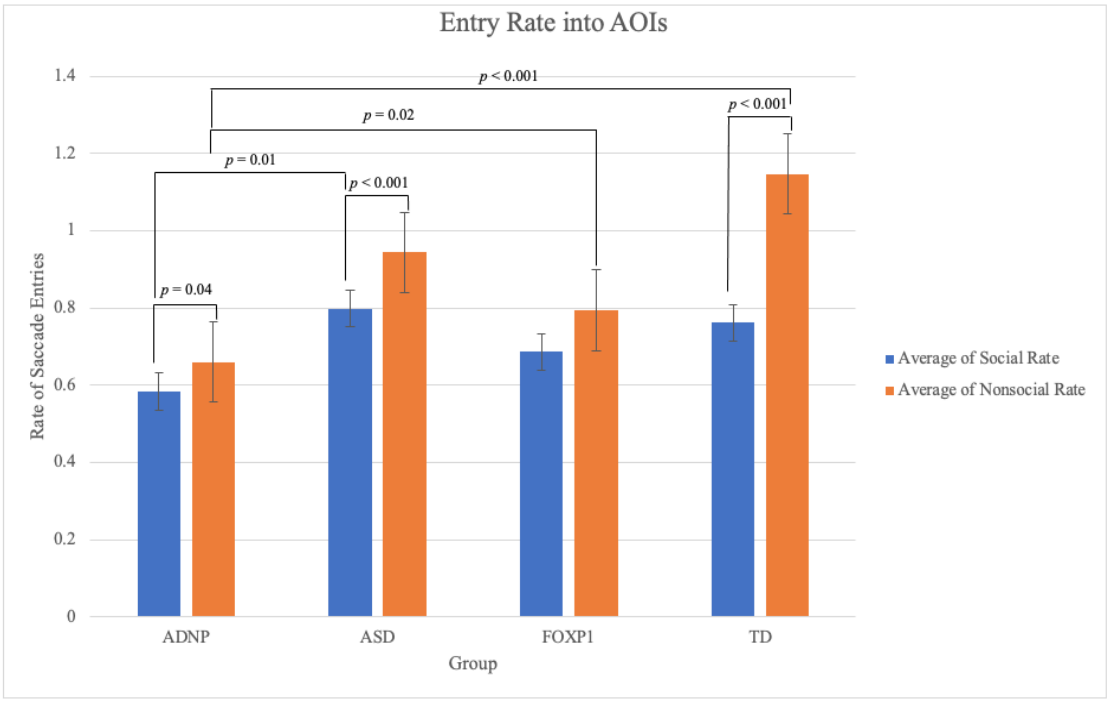


*Figure 3*. Average entry rate into AOIs by group

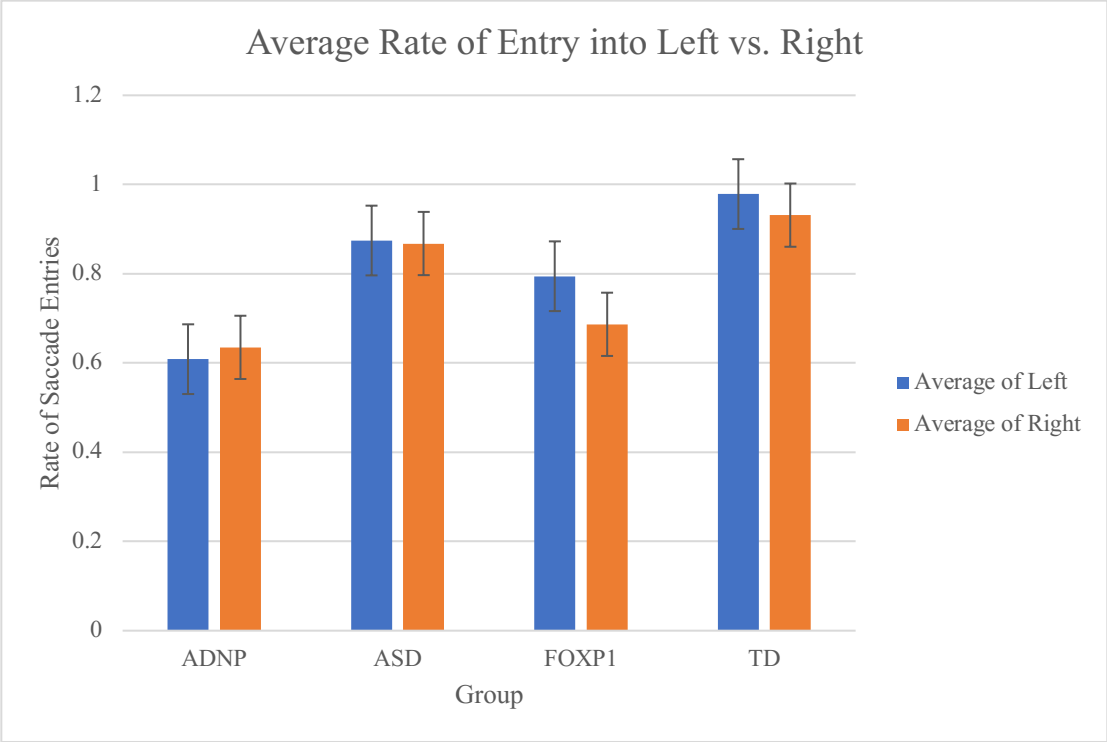


*Figure 4*. Average entry rate into AOIs on left and right sides by group

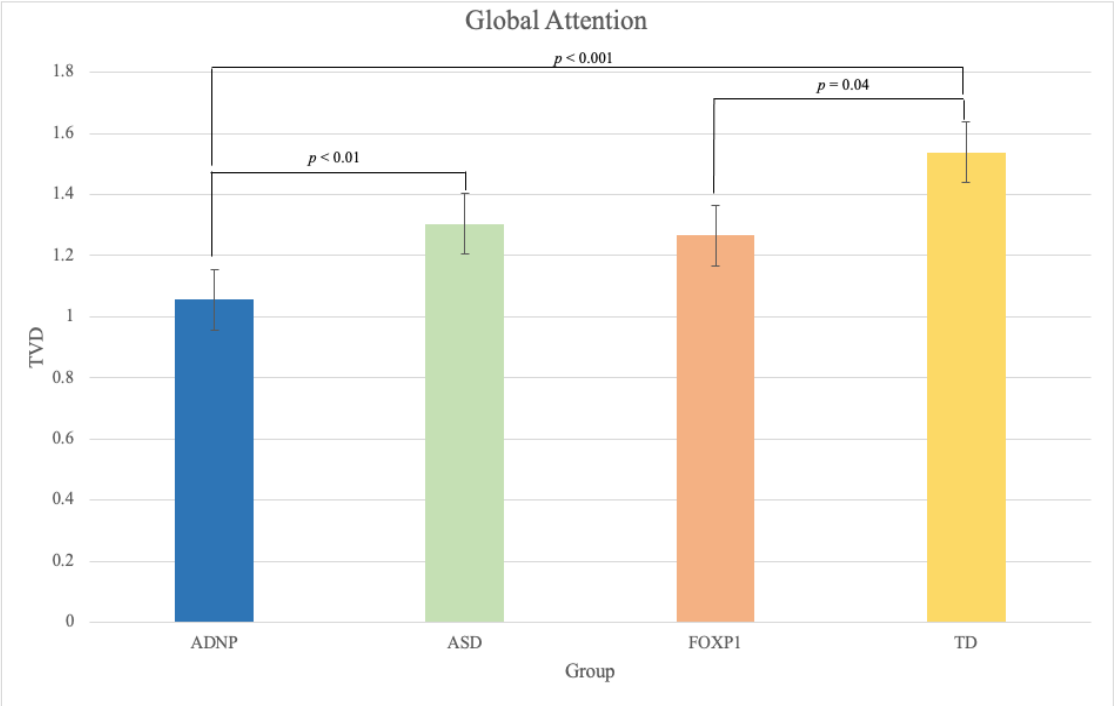


*Figure 5*. Global attention (i.e., attention to both social and non-social stimuli) by group

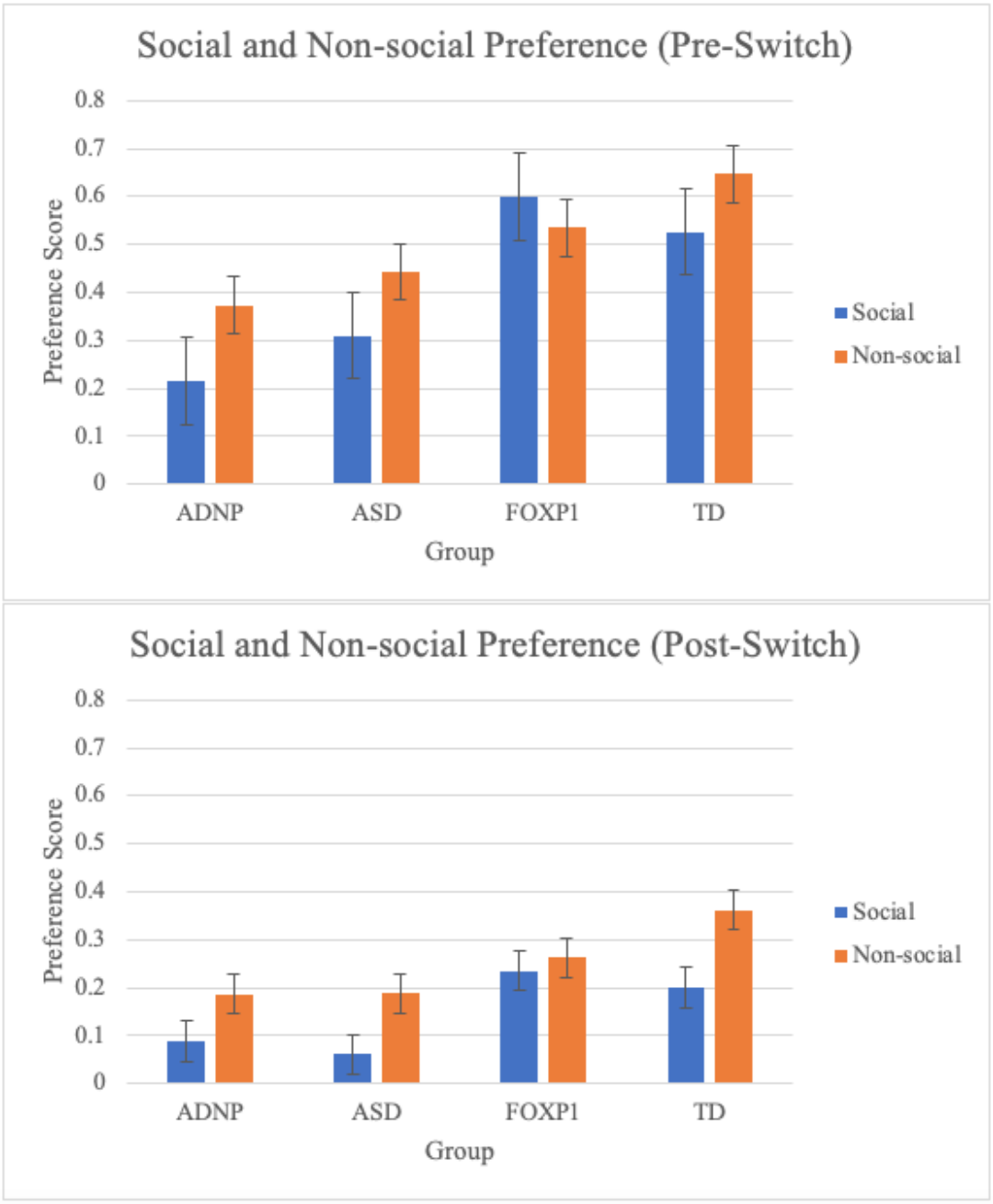


*Figure 6*. Social and non-social preference at pre- versus post-switch by group

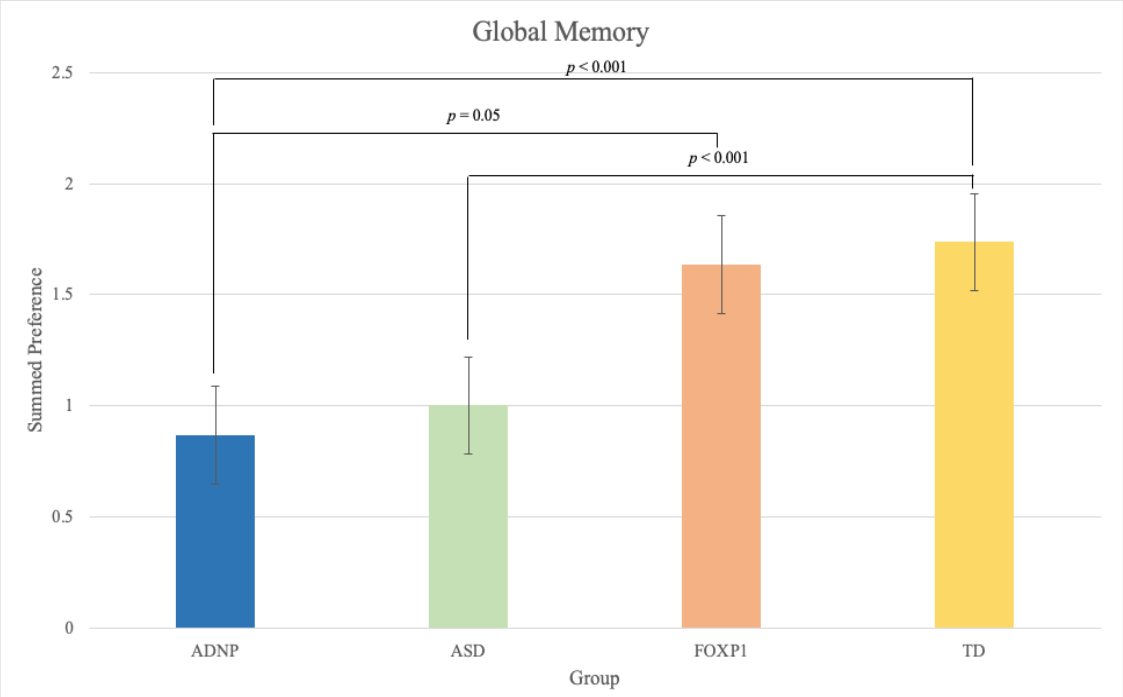


*Figure 7*. Global memory (i.e., recognition memory for both social and non-social stimuli) by group

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