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#### Alexithymia in children with and without Autism Spectrum Disorders

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#### Scientific Abstract

Alexithymia refers to pronounced difficulty in identifying and describing one's own emotions and is associated with an externally oriented focus of thinking. Alexithymia is known to be much more common in adults with autism spectrum disorders (ASD) compared to the typically-developing (TD) adult population. However, we know very little about alexithymia in young children with ASD and advancing our understanding of this topic may be of critical clinical and translation importance. Here we present the first study to examine alexithymia in children with ASD. We find that alexithymia is substantially elevated in ASD on both self- and parent-report measures. Despite both measures being sensitive to on-average group differentiation, we find no evidence of correlation between such measures, indicating that children and their parents may be using different sources of information. Parent-rated alexithymia is also associated with increasing levels of autistic traits. Discrepancy between self and other alexithymia ratings are also associated with autistic traits, but only in ASD. These results underscore the idea that assessing alexithymia in ASD at younger ages may help identify important subgroups that have particular difficulties in the domain of emotion processing.

Keywords: Autism, alexithymia, parent-report, self-report, autistic traits, children

#### Lay Abstract

Alexithymia refers to difficulty in labeling and describing one's own emotions and is associated with an outward focus of thinking. Alexithymia is more common in adults with autism spectrum disorders (ASD) compared to the wider adult population. However, we know very little about alexithymia in young children with ASD and advancing our understanding of this topic may be of critical importance for clinical practice. Here we present the first study to examine alexithymia in children with ASD. We find that alexithymia is higher in children with ASD on measures where parents respond about their children and also when children respond about themselves. Despite both measures being sensitive to differences between children with and without ASD, we find no evidence of correlation between parent and self-report measures, indicating that children and their parents may be using different sources of information. Parent-rated alexithymia is also associated with increasing levels of autistic traits. The discrepancy between self and other alexithymia ratings are also associated with autistic traits, but only in ASD. These results underscore the idea that assessing alexithymia in ASD at younger ages may help identify important subgroups of that have particular difficulties understanding and labeling emotions. Alexithymia is a word derived from ancient Greek and literally translates into "without words for emotion". This concept originally emerged in psychiatry through the work of Sifneos and colleagues on psychosomatic disorders, and refers to difficulties an individual has in identifying and describing one's own emotions or feeling, alongside difficulties in distinguishing feelings from bodily sensations, as well an externally oriented cognitive style of thinking (Nemiah, Freyberger, & Sifneos, 1976; Sifneos, 1973). In adult populations with ASD, elevated levels of alexithymia are a highly replicable and robust finding. Using a continuous metric of this elevation, our past work reported an estimated elevation of around 1.4 standard deviations (Lombardo, Barnes, Wheelwright, & Baron-Cohen, 2007). Case-control studies that use a categorical cut-off score, estimate that approximately 40-65% of adults with ASD could be considered alexithymic (Bird & Cook, 2013; Hill, Berthoz, & Frith, 2004; Lombardo et al., 2007; Nemiah et al., 1976). This estimate is much larger than the expected 10% of the general population that would fall above such cut-offs (Bird & Cook, 2013). In either case, it is clear that an important subset of adults with an ASD diagnosis are markedly different in how well they can identify, understand, and describe their emotions. Alexithymia may be an important clinical dimension to focus on, as well as become a domain that may be useful in parsing apart the wellknown and substantial heterogeneity in ASD (Bird & Cook, 2013; Lai, Lombardo, Chakrabarti, & Baron-Cohen, 2013).

As a separate construct to ASD, alexithymia may have some importance in explaining the marked heterogeneity in ASD, and the subsequent equivocal nature of some findings in the literature on emotion and empathy in ASD. A main hypothesis put forth by Bird and Cook (2013), is that alexithymia, and not ASD diagnosis, drives many of the supposed deficits in emotion and empathy in ASD (for review see Bird & Cook, 2013). A critical aspect of the design of Bird and colleagues' studies are that they first match ASD and control groups on alexithymia, and then subsequently test for deficits due to diagnosis, or whether variation on the dependent variable is accounted for by continuous variation in alexithymia. Across several studies, they have shown that no differences are apparent due to diagnosis, but that continuous variation in the dependent variable scales vary with continuous variation in alexithymia. One main implication of this work is that studies of ASD that do not account for more pronounced alexithymia in a subset of the ASD population, may interpret their results as a deficit due to ASD diagnostic status, when instead, the deficits may be driven by the onaverage higher levels of alexithymia within ASD. Aside from the experimental design implications of this work, a more general impact it could have for the study of ASD could simply be in how it may better account for a particular aspect of the marked heterogeneity in ASD. In other words, an important question to ask is why there are a substantial number of ASD cases that also have difficulties in the domain of alexithymia, and how are they different from ASD cases with no

elevation in alexithymia? This distinction might prove useful in terms of practical clinical impact for patients (e.g., more personalized treatments) and might also prove useful in honing in on mechanisms that distinguish such potential ASD subgroups (Lai et al., 2013).

Given that alexithymia refers to a specific deficit in self-referential emotion processing, it is pertinent to discuss the larger relevant literature on non-emotion based deficits in self-referential cognition in ASD (Lombardo et al., 2007; Lombardo & Baron-Cohen, 2010, 2011). For example, a key facilitative effect of self-referential cognitive processing is the self-reference effect in memory. This effect manifests as increased memory for items that were previously encoded (i.e. processed) in a selfreferential manner and it has been shown in adults and children with ASD to be attenuated or absent (Grisdale, Lind, Eacott, & Williams, 2014; Henderson et al., 2009; Lombardo et al., 2007; Toichi et al., 2002). Children with ASD may also have difficulty in making self-other distinctions (Mitchell & O'Keefe, 2008) and this may be important for helping to explain early difficulties in theory of mind (Williams & Happe, 2009). Difficulty with self-other distinctions may also translate back to deficits within neural circuitry that typically makes such distinctions (Lombardo et al., 2010; Pfeifer et al., 2013). It is also noteworthy that many of the deficits in the domain of self-referential cognition in ASD mostly pertain to psychological aspects of the self, while physical aspects of the self are largely unaffected (Williams, 2010).

Because all measures of alexithymia are themselves self-report instruments, therein lies a question about how accurate are individuals with high levels of alexithymia and ASD at self-reporting about themselves? Because some individuals with ASD may have difficulties in self-referential cognition that affect self-insight and metacognitive processing (Lombardo & Baron-Cohen, 2010, 2011), it is possible that self-report ratings may not accurately measure alexithymia for such individuals. Therefore, it is necessary to evaluate alexithymia measures taken from close observers of the individual (i.e. parents), in order to examine whether individuals with ASD would still have higher alexithymia levels than comparison groups. In addition to enabling more accurate inferences to be made about alexithymia, the inclusion of another informant allows for measurement of discrepancies between self and other judgments, which itself may index difficulties in self-referential cognitive processing. Thus, a design innovation like using both self- and other-based ratings can allow further tests of how self-referential cognitive processing may relate to indices of socialcommunication impairment such as autistic traits.

A notable asymmetry in the literature on alexithymia in ASD, is that all the work exists within adult populations. It is important to be able to study this topic in early development, as there may be clinical utility in having information about alexithymia in assessments of young children. In this study, we set out to examine for the first time how alexithymia might manifest in children with ASD. We utilize both self- and parent-report measures of alexithymia that have been previously developed and utilized outside of ASD populations (Rieffe, Oosterveld, & Terwogt, 2006; Way et al., 2010). Much like the literature in adults, we predicted that elevations would be apparent. We also set out to characterize how alexithymia measures in children are related to intelligence and continuous measures of autistic traits. Finally, we examined how discrepancies in self versus other alexithymia ratings would relate to measures of autistic traits. Given existing ideas relating to how some children with ASD may have difficulties in self-referential domains that limit their self-insight and awareness, we specifically hypothesized that impaired self-insight resulting in higher parent-ratings compared to self-ratings of alexithymia would be related to increased levels of autistic traits in ASD, but would show no relationship within typically-developing (TD) children.

#### Method

#### Participants

The ASD participant group consisted of 25 children (23 male, 2 female) with a diagnosis of ASD and/or Asperger's Syndrome using DSM-IV criteria (APA, 2000). Participants ranged in age from 8-13 years (M=10.21, SD=1.53). A typically-developing (TD) participant group consisted of 32 children (15 males, 17 females) ranging in age from 8-12 (M=10.00, SD=1.34). ASD and TD groups did not differ in age (t(54)=0.54, p=0.59). Children in the TD group had never been suspected of, or received a diagnosis for, any significant developmental disorder. Participants were recruited from an array of sources including the University of Edinburgh Developmental Psychology participant database, national autism awareness and support organizations, through online parenting forums, and via primary schools.

Exclusion criteria included the presence of a diagnosed psychiatric illness and/or history of neurological illness or brain injury (n=1) as these could be interpreted as evidence of alexithymia. These criteria led to the exclusion of one case in the ASD Group, resulting in a final ASD sample size of 24. Parents gave informed consent before partaking in the study, in accordance with the University of Edinburgh Psychology ethics committee.

#### Measures

#### The Wechsler Abbreviated Scale of Intelligence (WASI)

This scale was used to measure full-scale IQ (FSIQ) and to ensure no disproportionate differences existed between groups. We utilized an abbreviated 2- subscale version of the WASI comprised of

the verbal reasoning tests and matrix reasoning, which provides full-scale IQ only. This was done in order to reduce testing time for participants. The WASI is a widely used measure of intelligence, which has been used extensively in studies involving children with and without ASD with high reliability (Minshew, Turner, & Goldstein, 2005). The TD group (M=114.81, SD=9.97) was significantly higher in FIQ compared to the ASD group (M=102.20, SD = 14.43) (t(54)=3.86, p=0.0003).

#### Children's Alexithymia Questionnaire - Self Report

The Children's Alexithymia Questionnaire for children (CAQ-SR) was developed as an accompaniment to the original adult questionnaire for alexithymia (TAS-20) (Bagby, Taylor, & Parker, 1994). This is a self-report questionnaire that consists of 20 items. Children are asked to score each item on a three-point response scale (not true; sometimes true; often true). The internal reliability of CAQ-SR has shown to have an alpha coefficient of .75 (Rieffe et al., 2006). A higher score indicates more symptoms of Alexithymia.

#### Children's Alexithymia Measure - Parent Report

The Children's Alexithymia Measure (CAM-PR) is a parent-report measure of alexithymia. It is comprised of 14-items measured on a 4-point Likert scale (almost never, sometimes, often, and almost always). The CAM-PR has shown strong internal reliability, with an alpha coefficient of .92 (Way et al., 2010). Higher scores indicate more symptoms of Alexithymia.

#### Autism Spectrum Quotient – Child Version (AQ-Child)

The AQ-Child is a 50-item parent-report questionnaire developed to detect autistic traits in children 4–11 years of age. Higher scores indicate a greater number of autistic traits. AQ-Child items are answered on a 4-point Likert scale (definitely agree, slightly agree, slightly disagree, and definitely disagree). The AQ-Child has shown good test–retest reliability (r=.85, p<.001), high sensitivity (95%) and high specificity (95%) (Auyeung, Baron-Cohen, Wheelwright, & Allison, 2008).

#### Social Responsiveness Scale–Second Edition (SRS-2)

The SRS-2 is a 65-item questionnaire that measures deficits in social behavior associated with ASD. items are scored on a 4-point Likert scale (not true, sometimes true, often true, and almost always true ). The SRS-2 has shown good test–retest reliability ranging from .88 to .95, high sensitivity (92%) and high specificity (92%) (Bruni, 2014).

#### Procedure

Children were tested in the University of Edinburgh Developmental Psychology Laboratory or in a school setting. Care was taken to ensure experimental conditions were closely aligned in each setting. Written, informed consent was obtained from parents with the child's consent being given verbally and/or through observation of outward affect. Occupational status was recorded in order to test SES, and any co-occurring medical or psychological issues were noted.

The WASI was administered to all subjects. The child was then asked to complete the CAQ-SR about his/herself, with a researcher present to provide clarification or help understanding any items. During this time the parent or guardian completed the CAM-PR, the AQ-Child and the SRS-2 School-Age parent-report versions of the questionnaires. The full testing session lasted from between one to one and a half hours for each child including a short break if required. Participants were informed they could discontinue at any stage. Each participant completed the experiment in full.

#### Statistical Analysis

All statistical analyses were performed in Matlab (R2014b) using functions from the Statistics toolbox. We first tested distributional assumptions of CAQ-SR and CAM-PR measures in each group using a Kolmogorov-Smirnov test (kstest.m). This analysis determined that all alexithymia measures in both groups did not come from a standard normal distribution (all p<3.08e-17). Therefore, our main hypothesis tests examining between-group differences were implemented using a nonparametric permutation t-test (10,000 iterations) and using FIQ, sex, and age as a covariates. Next, we used robust regression (using Tor Wager's Robust Regression Matlab Toolbox; http://wagerlab.colorado.edu/tools; (Wager, Keller, Lacey, & Jonides, 2005)) to be insensitive to bivariate outlying data points and computed correlation matrices for each group across IQ measures, autistic trait measures, and alexithymia measures. Only correlations passing Bonferroni-correction for all 20 comparisons across both groups (i.e. 0.05/20 correlations; p < 0.0025) were considered significant. Finally, to compare CAQ-SR and CAM-PR scores, we first transformed the data into zscores and then computed a difference score as CAQ-SR minus CAM-PR, to index degree of selfother discrepancy in alexithymia ratings. Higher values on this difference score indicate where selfratings were larger than parent-ratings. Lower values indicate where parent-ratings were higher than self-ratings. We then computed correlations (via robust regression) between these difference scores and SRS and AQ. To test the difference between TD and ASD correlations we used the paired.r function within the psych R library, and computed one-tailed p-values, given the directional hypothesis that self-other discrepancy scores would be negatively correlated in ASD and not correlated in TD.

#### Results

Table 1 presents means, standard deviations and ranges for each group separately as well as combined. Our main hypothesis tests examined whether children with ASD would show elevated alexithymia scores on both self- and parent-report measures. ASD children self-reported significantly higher scores on the CAQ-SR (ASD median=23.5, IQR=8, mean=21.667, SD=5.87; TD median=16, IQR=8, mean=16.5, SD=5.38; t=2.40, p=0.009, Cohen's d=0.94) (Figure 1A-B). Similarly, though with a much larger effect size, parent-report on the CAM-PR also indicated substantially elevated alexithymia in ASD (ASD median=18, IQR=12.5, mean=19.70, SD=10.20; TD median=4, IQR=4.5, mean=5.71, SD=6.24; t=4.74, p<0.0001, Cohen's d=1.74) (Figure 1C-D).

#### Insert Table 1 and Figure 1 here

We next examined correlations between alexithymia measures and other autistic trait measures and IQ for each group separately. Figure 2A-B shows correlation matrices for each group. Correlations between SRS and AQ survive in Bonferroni-correction for 20 comparisons (i.e. p<0.0025), thus confirming that these two measures of autistic traits are similar. Within ASD, CAM-PR but not CAQ-SR, correlated with both AQ and SRS. Within the TD group, the only correlation between alexithymia and autistic traits was between SRS and CAM-PR. All CAM-PR associations with autistic trait measures can be interpreted as increases in parent-rated alexithymia co-vary with increases in autistic traits.

#### Insert Figure 2 here

Finally, we examined self-other discrepancy in alexithymia ratings. To achieve this aim, we computed an index of self-other discrepancy by converting CAQ-SR and CAM-PR into z-scores and then computing the difference between the two. High values on this index indicate self-ratings that are higher than parent-ratings, while negative values indicate individuals where the parent-ratings are higher than self-ratings. Given prior notions of lack of self-insight and deficits in self-referential cognition for some ASD individuals, the cases that score low on this index are of particular interest, since these individuals likely under-rate their levels of alexithymia compared to observer-ratings, and this may likely be consistent with the fact that these individuals are indeed more affected in terms of symptom severity. Thus, the prediction is that this self-other discrepancy index would be negatively correlated with SRS and AQ scores in ASD, but show no relationship in TD, and that the difference in relationships in this specific direction would be significant. Confirming this finding, we find that across both SRS and AQ, self-other discrepancy index is negatively correlated in ASD (SRS r=-0.54, p=0.01; AQ r=-0.42, p=0.06), but is not correlated in the TD group (SRS r=-0.04, p=0.86; AQ r=0.01, p=0.94). The difference between-groups in correlations was significant for both SRS (z=1.98, p=0.02) and AQ (z=1.62, p=0.05) (Fig 3A-B).

#### Insert Figure 3 here

#### Discussion

To our knowledge, the present study is the first to empirically examine alexithymia in children with ASD. Our study identifies that similar to the literature in adults (e.g., Bird & Cook, 2013; Hill et al., 2004; Lombardo et al., 2007), children with ASD also show elevations in alexithymia. These elevations are observed both in child self-report as well as parent-report ratings, and thus illustrate the robustness of the effect across raters. Parent-ratings did indicate a much larger effect size than self-report ratings. Given that there are some questions regarding how accurate self-report measures could be in children with ASD and alexithymic difficulties, it is important to see confirmation in the parent-reports. One possible explanation for the enhanced effect size in parentreport ratings could be because a subset of ASD children may not be as accurate in their self-ratings, due to certain difficulties with self-insight and/or self-referential cognitive deficits. Nevertheless, these results should help point to clinical considerations for a subgroup of children with ASD with the highest levels of alexithymia. Clinically, it would be important for future research to characterize whether it is this subgroup with high levels of alexithymia that demonstrate some of the exemplar behavioral characteristics in the domain of emotion processing that are typically regarded as characteristic symptoms that some but not all cases of ASD exhibit. It may also be important in the future to potentially use assessments of alexithymia to tailor interventions or treatments in a more personalized manner and to target specific domains of difficulty that may be specific to such a subgroup of ASD with additional alexithymia difficulties.

In this study we have also characterized how measures of alexithymia in children with ASD relate to continuous measures of autistic traits. Here we find that only parent-rated alexithymia correlates with autistic traits. In ASD, this correlation is apparent across both AQ and SRS, while in the TD group it exists only with SRS. These findings are generally consistent with the results in adults that also show a correlation between TAS-20 scores and the AQ (Lombardo et al., 2007). Given the correlation between the measures of autistic traits and alexithymia, the items comprising each measure were compared to examine whether there was overlap in the face validity of the characteristics measured by the AQ-Child and SRS-2 with the alexithymia scales. The autistic trait and alexithymia measures

were observed to be quite distinct. However, future studies specifically designed to examine the psychometric properties and relatedness of the scales would be useful.

Interestingly, alexithymia measures for self- and parent-report are not correlated. Thus, while on average there seems to be similar directionality of group differences across self- and parent-ratings (e.g., ASD>TD), this evidence of a lack of any relationship between these measures may be important. One explanation behind this result could be that parents and children use different sources of information to make such ratings, and/or some children with ASD may not be as accurate in rating their level of alexithymia.

Finally, we found some evidence for differentiation between ASD and TD in the correlation between autistic traits and self-other alexithymia rating discrepancies. This analysis was motivated by the ability to use self-other alexithymia rating discrepancies as an index of individual differences in selfreferential cognitive abilities such as insight and self-awareness. Because lower values on this type of self-other discrepancy index indicate individuals who possess self-ratings that are below how their parents would rate them, we suspected that it is within this lower range of values on this index that we would find the most impaired ASD individuals. This logic follows along the idea that selfreferential cognitive abilities are facilitative of many other social-communicative abilities (Lombardo & Baron-Cohen, 2010, 2011). These expectations set up the specific prediction that there would be a negative correlation between this self-other alexithymia rating discrepancy index and autistic traits that is specific to ASD. Confirming this hypothesis, we indeed find that those ASD individuals with the highest levels of autistic traits are also those whereby parents tend to rate them higher in alexithymia than self-ratings. No such relationship was apparent in the TD group, and the correlations were significantly different across the groups. These results suggest that even on a measure of self-referential difficulty in the domain of emotion, such as alexithymia, there will be a tendency for some ASD individuals to potentially under-rate themselves compared to close other ratings (e.g., parent-rating) and this indicator of potential lack of self-insight or self-awareness may index their level of social-communicative impairment measured on quantitative measures of autistic traits.

#### Limitations, Caveats, and Future Directions

One of the most salient limitations of the current work is the relatively small sample size in each group. However, despite the relatively smaller sample size, it is possible that the effect size we have estimated in the current study would suggest that the true effect size for detecting a group difference in alexithymia in ASD children is quite large and thus can be detectable in smaller

samples. Future work obtaining much larger sample sizes from community settings will help the field gauge the full generalizability of our initial results. This work will be important for several reasons. Larger-scale work will ultimately help in determining how important it is in clinical settings to measure alexithymia in children with ASD. As noted above, there are potentially important clinical reasons to assess this aspect of children's functioning, as there may be a subgroup of ASD individuals that show very high levels of alexithymia, and knowing such information could help tailor more appropriate and personalized intervention for those children.

Another limitation of this study is that the administration of gold standard diagnostic instruments for ASD such as the Autism Diagnostic Interview – Revised (ADI-R) (Lord, Rutter, & Le Couteur, 1994) or the Autism Diagnostic Observation Schedule (ADOS) (Lord et al., 2000) was not possible due to constraints on resources. Rather, clinical diagnoses were confirmed using diagnostic letters for each participant. To measure symptom severity, the AQ-Child and SRS-2 were administered, which are not diagnostic tools but were designed to quantitatively measure characteristics associated with ASD. It would be important for future studies to examine the relationship between alexithymia measures, ADI-R and ADOS scores and what specific behaviors and measures might discriminate individuals with ASD who have alexithymia from those who do not.

A design issue that the current study highlights for future work is that it will be of critical importance to obtain both self- and other informant-reports (i.e. parent, teacher) in ASD children. It is known that some children with ASD may have remarkable difficulties with self-insight and other aspects of self-referential cognition that will ultimately make assessment of alexithymia via self-report challenging. Our results show that while on-average one can make similar statements about ASD as a group, any individual level inferences must take into account both self- and informant-reports. Correlations between self- and parent-report of alexithymia in this study were non-existent, indicating that the source of information that children and parents use to rate alexithymia is likely different. However, one limitation of the current study's use of alexithymia measures is that the self-report versus parent-report measures incorporate different items. To make more direct comparisons of self- versus informant-reports it would be beneficial to develop one measure with the same items that can be applied to different informants.

Another potential implication of this work for future studies is how it may help in parsing heterogeneity in emotional aspects of functioning in ASD. As Bird and Cook highlight, the literature on this topic is very mixed (Bird & Cook, 2013), and one potential explanation for this is that each individual study includes different mixtures of individuals that might include individuals with both ASD and alexithymia versus ASD with no alexithymia. If alexithymia drives many of the deficits

observed in the domain of emotion processing in ASD, then it will benefit the field to utilize such assessment measures of alexithymia in ASD children in order to better parse apart whether any emotion-related deficits are indeed driven by a subgroup of ASD children with pronounced alexithymia.

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### Table 1

	All (N= 56)			NT Group (N= 32)			ASD Group (N= 24)		
Variable	<u>M</u>	<u>SD</u>	<u>Range</u>	M	<u>SD</u>	Range	<u>M</u>	<u>SD</u>	Range
CAQ-SR	18.71	6.12	6-33	16.50	5.38	6-26	21.67	5.87	7-33
CAM-PR	11.71	10.69	0-38	5.72	6.24	0-27	19.71	10.21	4-38
Child Age	10.09	1.42	7-13	10.00	1.34	8-12	10.21	1.53	7-13
IQ	109.41	13.52	76-136	114.81	9.98	86-136	102.21	14.43	76-136

Means, standard deviations and ranges for each group

### **Figure Captions**

Figure 1: Boxplots and permutation null distributions for alexithymia self-report (CAQ-SR; panels A-B) and parent-report (CAM-PR; panels C-D). Dots within the boxplots indicate individual data points. The grey histogram in panels B and D represent the null distribution estimated from the permutation test, and the solid black line indicates the true t-stat in the unpermuted data.

Figure 2: Correlation matrices for alexithymia, autistic trait, and IQ measures in ASD (A) and TD (B) groups. The star indicates correlations that pass Bonferroni-correction at p<0.0025.

Figure 3: Scatterplots depicting the relationship between self-other alexithymia rating discrepancy scores and autistic traits measured by the SRS (A) or AQ (B). Blue dots in panels C and D represent ASD individuals, while red dots represent TD individuals. Correlation estimates are computed using robust regression.