Mixed emotions:

The contribution of alexithymia to the emotional symptoms of autism

Geoffrey Bird Ph.D.1,2 & Richard Cook Ph.D.3

1MRC Social, Genetic & Developmental Psychiatry Centre, Institute of Psychiatry, Kings College London

2Institute of Cognitive Neuroscience, University College London

3Department of Psychology, City University London

*Corresponding author: geoff.bird@kcl.ac.uk
MRC Social, Genetic & Developmental Psychiatry Centre, Institute of Psychiatry PO80, London, SE5 8AF, UK.
Tel: +44 20 7848 5361
Fax: +44 20 7848 0866
Abstract (222 words)

It is widely accepted that autism is associated with disordered emotion processing, and in particular, with deficits of emotional reciprocity such as impaired emotion recognition and reduced empathy. However, a close examination of the literature reveals wide heterogeneity within the autistic population with respect to emotional competence. Here we argue that, where observed, emotional impairments are due to alexithymia - a condition that frequently co-occurs with autism - rather than a feature of autism per se. Alexithymia is a condition characterized by a reduced ability to identify and describe one’s own emotion, but which results in reduced empathy and an impaired ability to recognize the emotions of others. We briefly review studies of emotion processing in alexithymia, and in autism, before describing a recent series of studies directly testing this ‘alexithymia hypothesis’. If found to be correct, the alexithymia hypothesis has wide-reaching implications for the study of autism, and how we might best support sub-groups of autistic individuals with, and without, accompanying alexithymia. Finally, we note the presence of elevated rates of alexithymia, and inconsistent reports of emotional impairments, in eating disorders, schizophrenia, substance abuse, Parkinson’s Disease, Multiple Sclerosis, and anxiety disorders. We speculate that examining the contribution of alexithymia to the emotional symptoms of these disorders may bear fruit in the same way that it is starting to do in autism.

Key words: Autism, Alexithymia, Heterogeneity, Emotions, Expressions, Empathy
1. Introduction

Autism is a disorder characterized principally by social deficits: two of three main diagnostic features - impaired communication and reciprocal social interaction - lie within the social domain. While global emotional deficits are not currently a diagnostic feature of autism, problems of emotion processing, such as a lack of empathy and impaired recognition of emotion in others, are considered to be diagnostic markers of the condition.\(^1\),\(^2\) Despite the existing consensus that impairments of emotion processing are a feature of the autistic phenotype, empirical testing has yielded surprisingly equivocal results. In the present paper we argue that, where observed, emotional difficulties within the autistic population are actually attributable to alexithymia - a condition that frequently co-occurs with autism - rather than a feature of autism \textit{per se}.

Alexithymia is a sub-clinical condition characterized by difficulties identifying and describing one’s own emotional state.\(^3\) For example, an individual with alexithymia might know that they are experiencing an emotion, but be unaware whether that emotion is sadness, anger or fear. While the incidence of alexithymia in the typical population is estimated at 10\%, elevated levels of alexithymia are seen in a number of disorders such as anorexia nervosa, substance abuse, and post-traumatic stress disorder.\(^4\) The incidence of alexithymia is particularly high in the autistic\(^1\) population, with between 40\% and 65\% of adults with autism meeting criteria.\(^5\),\(^6\) Alexithymia has also been shown to be part of the “Broader Autism Phenotype”,\(^7\) the cluster of autism-like traits observed in parents of autistic children. Despite their frequent co-occurrence, alexithymia and autism are independent constructs. Alexithymia is neither necessary nor sufficient for an autism diagnosis, nor is it universal among autistic individuals. Conversely, many individuals show severe degrees of alexithymia without demonstrating autistic symptoms.

2. The emotional symptoms of autism

Difficulties in the emotional domain have been considered a feature of autism since the first cases reported by Kanner\(^8\) and Asperger.\(^9\) Reduced ‘emotional reciprocity’ is still considered to be a clinically significant indicator of autism,\(^2\) and some have argued that global emotional difficulties are a core feature of autism.\(^10\),\(^11\) However, a close reading of the literature reveals
that the empirical evidence in support of this claim is strikingly inconsistent.\textsuperscript{12-14} More consistent, is the substantial variability within the population of individuals with autism; while some individuals are clearly impaired, other autistic individuals perform at normal levels.

\textit{Emotion Recognition}

Many empirical findings have been published consistent with the hypothesis that individuals with autism find it harder to recognize, interpret and describe emotional stimuli. Several studies have reported that autistic individuals show poor recognition of emotional facial expressions, in particular those with negative valence, relative to matched controls.\textsuperscript{15-18} Some autistic individuals also seem to be impaired at recognizing the emotional valence of verbal and non-verbal vocal cues,\textsuperscript{19,20} body movements,\textsuperscript{20-22} and describing the emotional content of music.\textsuperscript{23} Moreover, some individuals with autism have difficulties matching the emotional cues conveyed by faces, voices and body postures across domains.\textsuperscript{11,24} Complimentary neuroimaging results have been reported showing atypical responses in regions of the face-processing network when autistic individuals view emotional expressions.\textsuperscript{25-27}

However, the picture is strikingly equivocal. Numerous studies have found no evidence of facial emotion recognition deficits in autism.\textsuperscript{28-30} A quantitative meta-analysis of the impact of autism on facial emotion recognition indicated a small effect size from a very mixed literature.\textsuperscript{14} Independent qualitative reviews have concluded “behavioral studies are only slightly more likely to find facial emotion recognition deficits in autism than not”\textsuperscript{12, p.317} and “most studies fail to show any deficits at all in the perception of simple emotions by children with [high-functioning autistic spectrum disorder]”.\textsuperscript{13, p.348} Several studies have also reported intact recognition of vocal affect,\textsuperscript{31-33} including one conducted with a large sample ($N = 99$) of autistic adolescents and matched controls.\textsuperscript{32} Various factors have been cited as possible causes of this heterogeneity, including sample demographics – e.g. potential differences due to the use of children/adults and the use of high/low-functioning autistic participants - and methodologies – e.g. the nature of the control groups used, the types of emotional stimuli, and the recognition procedures vary widely throughout the literature.\textsuperscript{12,14}
Empathy

It is frequently asserted that individuals with autism not only have difficulties recognizing emotions, but also lack emotional empathy.34-39 Defining empathy is no trivial matter, but our working definition here will be that empathy occurs when the perception of another’s emotional state causes the empathizer to experience that state. This definition distinguishes empathy from Theory of Mind40 (also known as ‘cognitive empathy’) which refers to the ability to represent, but not necessarily share, another’s mental state.ii

Consistent with this characterization, several studies have found that individuals with autism score lower on self-report empathy questionnaires including the Empathy Quotient36,41 and the Interpersonal Reactivity Index of empathy (IRI).37,42 Additionally, when presented with vignettes describing other children’s emotional experiences, some children with autism report less empathy with the characters described.43 It has also been reported that some individuals with autism show less corticospinal excitability when observing a painful stimulus being applied to another person,39 and a weaker tendency to imitate the emotional facial expressions of others.38 Studies using functional neuroimaging have found atypical neural responses when autistic individuals view emotional stimuli in the brain regions thought to mediate empathy44,45 and in the mirror neuron system46 which may mediate imitation of emotional facial expressioniii.

However, the evidence for an empathy deficit in autism is again surprisingly mixed.47 Several studies have found no group differences in empathy using self-report measures.48,49 Moreover, reports of group differences on self-report measures of empathy often obscure the fact that a significant proportion of those with autism, and their families, report equal or super-normal degrees of empathy.50 Some authors have found that autistic children demonstrate entirely typical emotional responses during social interactions and to recorded emotional expressions of others.51 Several studies also report perfectly intact automatic tendencies to imitate emotional facial expressions in individuals with autism52 and increased electromyographic (EMG) signals – an index of covert imitative motor activity - to vocal and facial emotion.53 Finally, several neuroimaging studies have failed to find differential mirror neuron system activation when autistic participants view emotional facial expressions.27,54
3. The alexithymia hypothesis

The alexithymia hypothesis suggests that, where observed, the ‘emotional symptoms of autism’ are in fact due to the greater proportion of individuals with severe alexithymia in the autistic population. Alexithymia is characterized by difficulties identifying and describing one’s own emotional state. Nevertheless, a considerable body of evidence suggests that individuals with high degrees of alexithymia also have difficulties recognizing emotional facial expressions, particularly those with negative valence. Moreover, it is also well-established that alexithymia is associated with reduced empathy. Such findings accord well with the growing consensus that the mechanisms responsible for our subjective experience of emotions are recruited when recognizing the same emotions in others.

Alexithymia is thought to be the product of developmental dysfunction of, or reduced connectivity between, limbic structures, including the anterior insula (AI) and anterior cingulate cortex (ACC); regions implicated in the subjective experience of emotion, affect recognition and empathy. Early studies suggested that both of these regions failed to show the typical modulation of cerebral blood flow when alexithymic participants viewed angry faces. More recently, Moriguchi et al. found that alexithymic individuals showed less activation in the left caudal ACC, when viewing hands and feet depicted in painful situations. FeldmanHall et al. demonstrated that individuals with high levels of alexithymia have reduced neural activation within the AI when seeing others in pain, and are less motivated to act altruistically to relieve another’s distress. Complimentary findings of structural differences within the AI and ACC have recently been revealed using voxel-based morphometry.

The past two decades have seen growing acceptance that, i) alexithymia is associated with deficits of emotional experience, interpretation and recognition, and ii) that the incidence of severe alexithymia is substantially elevated in the autistic population. Nevertheless, studies of autism – even those investigating putative emotional symptoms – rarely use control groups matched for alexithymia, and typically fail to report the levels of alexithymia present in samples. This raises the very real possibility that, where observed, deficits of emotion recognition and empathy reflect disproportionate levels of alexithymia in
autistic samples, rather than being characteristic of autism (Figure 1a). Support for the alexithymia hypothesis has been provided by a series of recent studies.

Figure-1

Emotion recognition
Several studies have recently investigated emotion recognition deficits, in individuals with and without autism, with varying degrees of alexithymia.\textsuperscript{75-77} In two psychophysical experiments, Cook et al.\textsuperscript{75} separately assessed the contribution of autism and alexithymia to difficulties of facial emotion recognition. In one experiment participants were asked to assign an emotion label to morphed face stimuli expressing combinations of different emotions (two stimulus sets blended incrementally disgust and anger; happiness and fear). In a second experiment participants simply had to identify whether sequentially presented expressions were identical. Alexithymia was found to be closely associated with imprecise recognition of emotion: Although able to distinguish different facial expressions, individuals with severe alexithymia were unable to consistently label the emotions depicted (Figure 1b). Autism severity, however, did not correlate with any of the perceptual measures, nor was it associated with any deficit once differences in alexithymia were accounted for.\textsuperscript{iv}

Using a similar paradigm, Heaton et al.\textsuperscript{76} investigated the relative contribution of autism and alexithymia to difficulties recognizing vocal affect. Participants were asked to identify the emotion conveyed in verbal (three-digit-numbers being read aloud) and non-verbal (e.g., laughing, crying) vocalizations. While recognition accuracy was reduced in the autistic group, further analyses revealed substantially elevated rates of alexithymia in the autistic sample and highly significant correlations ($r > .60$) between alexithymia scores and recognition accuracy. The authors concluded that, when differences in low-level vocal processing were taken into account, residual differences in emotion recognition accuracy were due to co-occurring alexithymia.

The contribution of alexithymia and autism to the recognition of musical affect has also been studied.\textsuperscript{77} Participants were presented with pieces of music and asked to indicate which of 28
words described the emotional content. A group difference was identified; autistic participants chose fewer words, than matched controls, to describe the pieces. However, subsequent mediation analysis revealed that the effect was again due to co-occurring alexithymia: the presence of autism was no longer predictive of impoverished descriptions once alexithymia had been accounted for. In contrast, alexithymia continued to be a significant predictor once autism was accounted for.

**Empathy**

The contribution of alexithymia to the ‘emotional symptoms of autism’ is not only observed in behavior, but also in fMRI studies of empathic processes. Bird et al. measured the degree of neural activity evoked by a loved-one’s pain, in those areas of the brain active when personally receiving pain, as an objective measure of empathy. Participants were again individuals with and without autism, with varying degrees of alexithymia. The authors observed that the degree of empathic brain activity in the AI was predicted by alexithymia in both autistic and non-autistic participants, and that once alexithymia was accounted for, that there were no group differences in empathy related to autism.

A similar result was observed in a related study involving introspection upon emotion. Participants with and without autism and with varying degrees of alexithymia, were asked to judge the emotion evoked in them by pictorial stimuli drawn from the International Affective Picture System (IAPS) including images of illness, death and injury, and to report their level of empathy. The process of judging one’s emotional response to the distressing images resulted in significant AI activity, and the degree of this activity correlated with participants’ self-reported degree of alexithymia and empathy.

**4. Implications**

*Research methods & procedures*

If correct, the alexithymia hypothesis has important implications for the methodology adopted in future studies of autism. When emotion-related processes are investigated, studies should examine whether impairments are a primary feature of autism or a consequence of co-occurring alexithymia. Alexithymia is typically assessed using short self-report measures.
that may be completed in a few minutes. Matching autistic and control samples for alexithymia should therefore become as routine as matching for age, gender and IQ. Moreover, by ensuring that both autistic and control samples include individuals with varying degrees of alexithymia\textsuperscript{75,78}, the effect of the latter can be accurately quantified and, if necessary, controlled for statistically (i.e., entered as a co-variate or partialled out in regression). Such designs also allow the investigation of interactions between alexithymia severity and autism.

In addition, extreme caution is required when interpreting the results of all studies of autism using ‘emotional’ stimuli (e.g., emotional facial expressions), even those that purport to study ‘non-emotional’ processes (e.g., ‘Theory of Mind’, language, imitation, mirroring). For example, the “Reading the Mind in the Eyes Test”\textsuperscript{83,84} is frequently characterized as a measure of ‘Theory of Mind’ or ‘mentalizing’. In this task, participants are presented with stimuli consisting of the eye regions of a face, and asked to infer the person’s mental (e.g., interested or disinterested?) and affective states (e.g., relaxed or worried?). It is typically reported that individuals with autism tend to assign different states than typically developing individuals. However, because substantial proportions of the statements pertain to emotions, it is likely that co-occurring alexithymia will contribute to the poor performance of autistic samples. Accurate estimates of non-emotional deficits associated with autism thus require instruments that do not confound these abilities with emotion processing.

*Clinical Practice*

If future research shows the alexithymia hypothesis to be correct, the diagnostic criteria for autism may require revision. If it is found that individuals with autism, but without alexithymia, exhibit no emotional impairments, problems of emotion processing (e.g., a lack of empathy and impaired recognition of emotion in others) should no longer be considered diagnostic markers of autism.\textsuperscript{1,2} The trend towards amalgamating the social and emotional symptoms of autism may need to be reversed – under our hypothesis social and emotional impairments should be viewed as distinct, with autism being associated with the former, but not the latter.
A greater appreciation of the association with alexithymia may also improve interventions for autism. Not only is alexithymia associated with a wide-range of emotional impairments but it is known to reduce the effectiveness of some therapies. The alexithymia hypothesis suggests the existence of sub-groups within the autistic population, defined by the presence or absence of alexithymia. Given the ease with which alexithymia severity may be estimated using self-report instruments, intervention strategies could be tailored to meet the specific needs of each subgroup. Alternative approaches could thereby address the emotional problems of individuals with alexithymia, or build on the emotional competence of those autistic individuals without alexithymia to ‘scaffold’ social interaction.

5. Future challenges

Discussion of the alexithymia hypothesis brings into focus a number of outstanding questions and unresolved issues that demand further research attention. First, little is known about the origins and causes of alexithymia. Structural and functional neuroimaging have repeatedly implicated a number of regions, including the AI and ACC. Together, these areas constitute a neural circuit by which individuals are likely to become aware of, and reflect upon, their emotions.69,70 Weaker neural connectivity within this network, and between this network and higher-order sensory areas may therefore result in difficulties identifying one’s own emotions, and those of others. Nevertheless, further empirical work is needed to identify the aetiology of alexithymia, particularly with respect to the contributions of genes and environment. The initial work being conducted in this area suggests both genetic and environmental contributions.

A second, related, question concerns the substantially increased incidence of severe alexithymia in the autistic population. To-date, little progress has been made explaining this association. To offer some explanation, we speculate that i) a genetic vulnerability to suboptimal neural connectivity may be a feature of several disorders; ii) phenotypes characterized by particular cognitive and behavioral deficits reflect the locus of such poor connectivity. Where the affected networks relate principally to social cognition and cognitive control, a ‘pure’ autism phenotype may emerge. In contrast, a ‘pure’ alexithymic phenotype may result from suboptimal connectivity confined to limbic structures (e.g., AI, ACC). More
typically, however, poor connectivity may be diffuse across many of these networks - as a consequence of common genetic / environmental risk factors - resulting in the co-occurrence of autism and alexithymia. While we can cite little evidence in support of this speculation, it is striking that weaker neural connectivity has been implicated in both autism\textsuperscript{92,93} and schizophrenia;\textsuperscript{94,95} another disorder associated with elevated rates of alexithymia.

A further challenge for future research is to identify objective measures of alexithymia. At present self-report questionnaires are typically used to estimate symptom severity. These measures have the advantage of being quick to complete and have been shown to have predictive validity. Nevertheless, self-report instruments are likely to underestimate levels of alexithymia in severely-affected individuals due to the fact that accurate reporting is likely to require a basic degree of emotional awareness. The use of self-report questionnaires may therefore underestimate the strength of the relationship between alexithymia and disordered emotion processing in certain clinical groups. It is important then that tools are developed to measure alexithymia that do not rely on accurate self-report or interoceptive awareness. Neuroimaging paradigms offer a potential solution, but remain costly and inefficient. Consequently, the use of other indices of physiological arousal, in addition to self-report, may offer a solution to this problem. Indeed, initial data suggests that the degree of alexithymia in autism, as measured by self-report questionnaires, correlates well with the degree of correspondence between objective measures of emotional arousal, as measured by galvanic skin response and heart-rate variability, and self-report.\textsuperscript{96} This technique both validates the use of self-report questionnaires in the alexithymic population, does not require accurate self-report, and provides a method for the detection of alexithymia even in the absence of awareness of emotional difficulties.

Finally, it also remains to be seen whether there are different subtypes of alexithymia. The inability to identify and describe emotions - the characteristic deficit of alexithymia - may arise from several neuro-cognitive impairments. Consequently, some authors have hypothesized the existence of two types of alexithymia; Type I, associated with reduced physiological arousal, and Type II, in which typical physiological arousal is present but disconnected from higher cognition.\textsuperscript{97,98} However, this position remains controversial and
some have argued that a two-factor solution does not describe well the variance observed in the alexithymic population. Whether subtypes of alexithymia exist remains an open question, but one that must be addressed as a matter of priority.

6. Extending the argument?
In the foregoing sections we have made a big claim; that the deficits of emotional processing sometimes seen in autism, are not a primary feature of this disorder, but are instead due to comorbid alexithymia. However, we have chosen to round off our review, by alluding to an even bigger possibility: Can co-occurring alexithymia also explain the emotional symptoms seen in other psychiatric disorders?

Elevated rates of alexithymia are seen in a number of disorders including schizophrenia, eating disorders, Parkinson’s disease, and social anxiety. Interestingly, deficits of emotional processing, including problems recognizing the emotions of others, are also seen in many of these conditions (e.g. Parkinson’s Disease, Schizophrenia, alcohol abuse, eating disorders, and social anxiety.) Moreover, several of these conditions are characterized by atypical experience of emotion. For example, ‘negative symptoms’ - one of the five domains outlined in the diagnostic criteria for schizophrenia - consists of disordered emotional expression and diminished motivation and pleasure. These symptoms are supported by somewhat mixed empirical reports of atypical facial expression, emotion-related neural activity, and emotional experience.

To date, only a handful of studies have assessed the contribution of alexithymia to the emotional difficulties in disorders other than autism, but these generally support an extended alexithymia hypothesis. For example, Pedrosa Gil et al. found significant impairments of emotion recognition in patients with somatoform disorders. Crucially, however, this group deficit was no longer evident once alexithymia was accounted for statistically, suggesting that emotion recognition difficulties were due to elevated levels of alexithymia, and not somatoform disorder per se. Similarly, in a study of non-clinical individuals with a trend towards disordered eating, alexithymia severity was associated with emotion recognition difficulties over and above disordered eating symptomatology.
7. Conclusions

In the present paper, we have advanced the view that the inconsistent emotional symptoms of autism, including a lack of empathy and problems of emotion recognition, are due to the substantially elevated incidence of severe alexithymia present in the autistic population, and not autism per se. This ‘alexithymia hypothesis’ is supported by a series of recent studies conducted on individuals with and without autism, and with varying degrees of alexithymia. In every case, whether behavioral studies of facial emotion recognition, recognition of vocal or musical affect, fMRI studies of empathy or emotional introspection, alexithymia, but not autism, has been associated with emotional deficits.

If proven correct, the alexithymia hypothesis has a number of substantive implications for autism research and clinical practice. Studies addressing emotion processing in autism should examine whether impairments are a primary feature of autism or a consequence of co-occurring alexithymia. Crucially, matching autistic and control samples for alexithymia should therefore become as routine as matching for age, gender and IQ. Caution is also required when interpreting the results of all studies of autism using ‘emotional’ stimuli, even those that purport to study ‘non-emotional’ processes; for example Theory of Mind or imitation. If it is found that individuals with autism, but without alexithymia, exhibit no emotional impairments, ‘emotional symptoms’ should no longer be considered diagnostic markers of autism.

Although an ever-increasing body of work is consistent with the alexithymia hypothesis, a number of key questions remain. Further empirical work is urgently needed to identify the aetiology of alexithymia, particularly with respect to the contributions of genes and environment. A related question concerns the cause of the dramatically increased incidence of severe alexithymia in the autistic population and the potential role of genetic vulnerability to suboptimal neural connectivity. Finally, it remains to be seen whether an extended alexithymia hypothesis can explain the heterogeneity of emotional symptoms seen in other psychiatric disorders, such as schizophrenia and Parkinson’s disease. Nevertheless, one thing is clear; this intriguing condition demands considerably more research attention than it currently receives.
Acknowledgements and Conflict of Interest Statement

The authors gratefully acknowledge the comments and advice of Rebecca Brewer, Jennifer Cook, Francesca Happé, and Punit Shah on earlier drafts of the manuscript. GB was supported by a grant from the Baily Thomas Charitable Fund (3089/1).

The authors declare there to be no conflict of interest with respect to this manuscript.
References:


94. Cook J, Barbalat G, Blakemore SJ. Top-down modulation of the perception of other people in schizophrenia and autism. *Front Hum Neurosci* 2012; **6**.


Figure 1

Figure 1: (a) Illustration of the alexithymia hypothesis. Samples drawn from the autistic population may contain a high proportion of individuals with co-occurring alexithymia (e.g., Sample A), in which case the majority of tested individuals will exhibit emotional symptoms such as difficulties recognizing others’ emotions and reduced empathy. Samples may also contain relatively few individuals with co-occurring alexithymia (e.g., Sample C) yielding little evidence of emotional impairment in the majority of individuals tested, leading to the conclusion that autism is not characterized by emotional symptoms. More typically, however, samples drawn from the autistic population are likely to be heterogeneous; comprising a mix of individuals with varying degrees of co-occurring alexithymia (e.g., Sample B), leading to weak inconsistent trends towards emotional deficits across studies and wide heterogeneity within experimental samples. (b) Representative data from Cook et al\textsuperscript{75} demonstrating that increasing degrees of alexithymia is associated with increased attribution thresholds (indicative of poor emotion recognition) for emotional facial expressions. After accounting for alexithymia there was no effect of autism.

\textsuperscript{i} We use the terms “autistics” and “individuals with autism” to refer to the population of individuals with an autism spectrum condition as these terms are most favored by that population\textsuperscript{115}

\textsuperscript{ii} Several authors would argue that this definition of empathy is not sufficient, that in order for full empathy to be demonstrated a degree of self-other distinction is necessary (in contrast to emotional contagion). Here we are referring only to the process by which recognition of another’s emotional state causes that state to be experienced by the self, rather than to self-other distinction which may or may not be affected by alexithymia.

\textsuperscript{iii} While imitation and activation of the mirror neuron system are conceptually distinct from empathy, in the special case of emotional expressions imitation may lead to emotional contagion and ultimately empathy.

\textsuperscript{iv} Neither autism nor alexithymia was predictive of identity recognition thresholds or performance on a sequential matching task, confirming that alexithymia is not associated with low-level visual impairments. Moreover, these findings accord with the view that atypical processing styles and deficits of low-level vision, associated with autism (reviewed by\textsuperscript{116}), often have negligible effects on tasks assessing the perception of faces\textsuperscript{117}. It remains an interesting possibility that autism is associated with disproportionately impaired memory for faces.