Published in final edited form as:

Autism. 2010 November ; 14(6): 559–574. doi:10.1177/1362361310382107.

Narrowing the broader autism phenotype: A study using the Communication Checklist - Adult Version (CC-A)

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Abstract

This study investigated whether the Communication Checklist – Adult (CC-A) could identify subtypes of social and communication dysfunction in autism probands and their parents. The CC-A is divided into subscales measuring linguistic ability as well as two aspects of social communication: the Pragmatic Skills subscale assesses the level of pragmatic oddities (e.g., excessive talking), while the Social Engagement subscale picks up on those behaviours that reflect a more passive communication style (e.g., failure to engage in social interactions). CC-A data were collected for 69 autism probands, 238 parents of autism probands and 187 typical participants. The CC-A proved sensitive to the communication difficulties of autism probands and a proportion of their parents. The majority of parents who demonstrated the broader phenotype scored poorly on either the Pragmatic Skills or Social Engagement scale only. The Social Engagement scale was particularly sensitive to the difficulties of the parents, indicating that social-communicative passivity may be an important part of the broader autism phenotype. The findings provide evidence for the existence of more constrained pragmatic phenotypes in autism. Molecular genetic studies in this area may benefit from stratifying samples according to these phenotypes.

Keywords

autism; broad phenotype; genetic; communication; language; family

Genetic factors play a major role in the aetiology of autism spectrum disorders (ASD). Siblings of affected individuals are at an increased risk for developing the disorder, with identical twins at an even greater risk (Bailey et al. 1995). The genetic liability of ASD is also seen through those relatives of affected individuals who do not meet clinical standards for ASD diagnosis but nevertheless show mild to moderate features of the disorder, the so called ‘broader phenotype’ (Bolton et al., 1994; Dawson et al., 2002, Piven et al., 1997). Reports vary, but it is generally considered that around 10-20% of family members show at least some of the behavioural (e.g., social and communication impairments and repetitive behaviours), cognitive (e.g., executive dysfunction, face processing deficits), or biological characteristics (e.g., brain enlargement) reminiscent of ASD, but in much less severe form (for a review, see Dawson et al. 2002).

The strong support for a genetic involvement in autism has led to an intense international effort to identify candidate genes that may contribute to the onset of the disorder. However, while these studies have provided several interesting candidate chromosomal locations - most notably, regions on chromosomes 2q, 7q, 15q and 17q (for a review, see Abrahams and Geschwind, 2008) – replication of exact loci has been rare. The lack of reproducibility is

thought to reflect the complexity of the underlying genetic mechanism associated with autism; there are likely to be many interacting genes, each contributing only weak effect to the resulting phenotype. However some authors have postulated that at least part of the variance in findings may be attributed to differences in the definition of the autism phenotype (Skuse, 2007; Todd, 2005).

In traditional autism research, cases are identified according to standard criteria with instruments such as the Autism Diagnostic Interview – Revised (Lord et al., 1994) and/or the Autism Diagnostic Observation Schedule – Generic (Lord et al., 2000), and classified on a dichotomous scale as either being affected or unaffected. However, it has been suggested that a categorical approach to case identification lacks validity for two reasons (Todd, 2005). First, it does not take into account the wide variability of symptomatology experienced by those with an ASD diagnosis. ASD is diagnosed along a spectrum of severity and it is possible that there are different genetic mechanisms at play between those with different presentations. Second, recent evidence has emerged that the features characteristic of autism may be ‘fractionable’ (for a review, see Happé and Ronald, 2009), with largely independent genes operating on each part of the triad of autistic behaviours (social impairments, communication impairments, restricted repetitive behaviours and interests). However, this is a relatively new area of research and considerably more evidence needs to be collected in order to substantiate this proposal.

This notwithstanding, these views raise the possibility that using quantitative measures to identify specific behavioural profiles making up the inherited phenotype of ASD may serve to enhance the likelihood of identifying (sets of) genes underlying the condition. Indeed there is some evidence for this, with the incorporation of more tightly constrained phenotypes enhancing the strength of linkage in a number of molecular genetic studies (Buxbaum et al., 2001; Shao et al., 2002). Within this context, the current study provides an examination of the social and communicative ability of adults with ASD and their parents using a new assessment instrument, the Communication Checklist – Adult (Whitehouse and Bishop, 2009).

Although communication deficits are a diagnostic requirement for autism, there is considerable variability in the difficulties experienced by affected individuals. A substantial proportion of individuals experience moderate or severe problems with the linguistic or structural aspects of language (phonology, morphology, syntax), while others show little or no difficulty in acquiring these skills (Whitehouse et al., 2008). In contrast, difficulties with social use of language, also known as pragmatic language, are more pervasive in this population. The discrepancy in findings regarding structural and pragmatic language are mirrored in studies of the broader autism phenotype; while there is mixed evidence for an elevated rate of structural language deficits among family members (Piven et al., 1997), the vast majority of studies report pragmatic difficulties within this population (Williams et al., 2008). These findings have led to suggestions by Whitehouse et al. (2007) that social communication deficits, but not linguistic deficits, are associated with the heritable phenotype of ASD.

However, while it is clear many relatives of ASD probands exhibit pragmatic oddities, it remains uncertain as to the exact profile that may form part of the broader autism phenotype. Pragmatics encompasses a diverse range of behaviours, incorporating the quantity, quality and variety of discourse as well as the manner in which this is communicated. Existing pragmatic assessments, such as the Pragmatic Rating Scale (Landa et al., 1992), the Broader Phenotype Autism Symptom Scale (Dawson et al., 2007) and the Autism Quotient (Baron-Cohen et al., 2001), allow an examination of individual pragmatic behaviours, but do not differentiate between different social-communicative profiles. Wing (1997) made a

distinction between individuals with ASD whom she called ‘active but odd’ and others whose difficulties present as passivity. Individuals in the former case make active social approaches to others, but these are typically naïve, one-sided and inappropriate, with conversation often revolving around their own circumscribed interests. In contrast, individuals in the latter category do not spontaneously interact with others, preferring to keep their own company in social situations. Wing stresses that these do not necessarily represent mutually exclusive categories and are likely to show as much overlap as other ASD classification systems. Nevertheless, these categories provide a framework through which more specific ASD phenotypes can be identified.

The Communication Checklist – Adult (Whitehouse and Bishop, 2009) was developed as an adult extension of the Children’s Communication Checklist – 2 (CCC-2; Bishop, 2003). This questionnaire asks about communicative behaviour in everyday situations and is to be completed by an informant who knows the index case well. The CC-A has three subscales, derived from a factor analysis of data collected from the standardisation sample. Table 1 includes example items from each subscale. The first subscale, Language Structure, assesses the linguistic aspects of language (i.e., speech, syntax, semantics). The other two scales focus on social communication. The first of these scales, Pragmatic Skills, is sensitive to communicative behaviours that are demonstrably inappropriate, such as the interruption of others at inappropriate times, excessive talking, poor topic maintenance, and conversational ‘bluntness’. The second scale, Social Engagement, picks up on those behaviours that reflect a more passive communicative style. This scale is particularly sensitive to individuals with limited social skills and poor nonverbal communication, such as those who have difficulty expressing themselves in a group, seem distant in a social situation and/or have a blank facial expressions during interactions.

The Pragmatic Skills and Social Engagement subscales map well onto Wing’s (1997) distinction between ‘active but odd’ and ‘passive’ individuals and enables us to investigate whether qualitatively different forms of pragmatic behaviours are inherited in ASD. The current study used the CC-A to examine the presence of these two different social-communicative profiles in a large sample of adults with ASD as well as parents of ASD probands.

Method

Participants

ASD probands—Adults with ASD were recruited from their previous participation in studies conducted at the University of Oxford, UK (Whitehouse et al., in press) as well as from several sites participating in The Autism Simplex Collection (TASC): Trinity College, Dublin (Ireland), University of Utah (USA) and McMaster University (Canada). All of these participants ($n = 62$) had received a clinical diagnosis of autism. Our own testing found that all participants met criteria for autism on the Autism Diagnostic Observation Schedule – Generic (ADOS-G; Lord et al. 2000). A further seven participants were recruited from an autism support group located in Sydney, Australia. All of these participants had a diagnosis of autism, based on DSM-IV guidelines by at least two of a paediatrician, a psychologist and a speech pathologist. Because of the long-distance nature of this recruiting, ADOS-G was not administered to these participants.

Parents of ASD probands—Parents of individuals with ASD were recruited from the sites listed above (ASD-par: $n = 238$). ADOSG was administered to those parents where ASD may have been suspected by family members or the research team ($n = 4$). ASD was discounted in all of these cases. All participants in this group had a child (or, for two unrelated individuals, children) who met criteria for autism on the ADOS-G (Lord et al.

2000). The majority of participants in this group had a child with ASD who was under the age of 17 and therefore too young to be assessed with the CC-A. Because of this, there were roughly only a quarter of individuals who had a child in the ASD group described above ($n = 63$, 26.47%).

Control group—The entire normative sample used in standardisation of the CC-A was included as a control group. Individuals were recruited from around the UK based upon criteria for age (between 17 and 79 years), ensuring a range of socioeconomic backgrounds, while excluding those with frank neurological impairment, a history of special educational needs or from a non English-speaking background. Preliminary analyses indicated no age effects for CC-A scores and data from all participants were combined into one group. Participant data were excluded if there were missing data on two or more items ($n = 29$), leaving a total of 187 participants with a complete or near-complete data set. Table 2 provides the chronological age and sex ratio of the three participant groups, as well as information on the respondents who provided ratings on the index cases.

Communication Checklist – Adult (CC-A (Whitehouse and Bishop, in press))

CC-A maintains the same format as the CCC-2, but with modifications to make the item content more appropriate for adults. There are 70 behavioral statements and informants must judge whether the individual in question demonstrates that behavior: less than once a week or never (a score of 0), at least once a week but not every day (1), once or twice a day (2), or several times a day or always (3). Fifty items relating to communicative weakness are grouped at the beginning of the checklist (items 1-50) and positive items are presented in a block at the end (items 51-70). For scoring, the positive skills items were reversed so that, as with the communicative weakness questions, higher scores indicated worse communicative skill.

Factor analysis was carried out to identify natural groupings in item responses, and this revealed three main factors. The first factor, Language Structure, has loadings predominantly from items concerned with language structure (phonology, grammar and vocabulary) plus some loading from items assessing comprehension. The second factor, Pragmatic Skills, has especially high loadings on items assessing coherence, inappropriate initiation, stereotyped language and interests. The third factor, Social Engagement, maps almost completely onto the distinction between the negatively and positively worded items, but also has loadings from items assessing use of context in comprehension, nonverbal communication, and social interaction. Note that the polarity of the positively worded items had been reversed before the analysis, so the high loading here indicates that this factor is picking up on a tendency to engage infrequently in positive communicative behaviours. All but two of the 70 items loaded onto these three factors with absolute values $> .3$ (the final two items, B36 and G15, were allocated to their relevant scale based upon face validity). Closely similar findings were obtained by Geurts et al. (in press) in a factor analysis of the original CCC in a sample of Dutch children, where positive items grouped together in a single factor.

In the process of norming the CC-A, standard scores equivalents were derived for each subscale, based around a mean of 10 and a SD of 3, after transforming the data to reduce non-normality. On average, males had higher (poorer) raw scores than females on the both the Pragmatic Skills and Social Engagement subscales, a finding consistent with the well-established sex-differences in social-communication functioning (Baron-Cohen et al., 2001). Because of this, separate standard score arrays for males and females were calculated for these two measures. A total score for the CC-A was derived by summing raw score ratings from all 70 items. Once again, to account for sex differences, z-scores for this measure were

calculated separately for males and females. Note, however, that the strong skew in the distributions of the subscales and total scores means it is not possible to interpret z-scores greater than zero in terms of statistical frequency.

Results

CC-A was normed in the UK, but there were some participants in the ASD and ASD-par groups who were recruited from the US and Australia. Analyses of the ASD sample found no differences between the UK ($n = 12$) and non-UK participants ($n = 57$) on the Language Structure, $t(67) = 1.1$, $p = .28$, Cohen's $d = 0.38$, Pragmatic Skills, $t(67) = .94$, $p = .35$, $d = 0.33$, and Social Engagement subscales, $t(67) = 1.62$, $p = .11$, $d = 0.61$. Comparisons in the ASD-par group also revealed no statistically significant difference between UK ($n = 54$) and non-UK participants ($n = 184$) (Language Structure: $t(236) = 1.78$, $p = .08$, $d = 0.27$; Pragmatic Skills: $t(236) = .81$, $p = .78$, $d = 0.13$; Social Engagement: $t(236) = 1.73$, $p = .09$, $d = 0.25$). We therefore combined participants from UK and non-UK in both the proband and parent groups. It is important to note that, where there was a trend for nationality differences, it was always in the direction of the UK sample showing greater level of impairment. This suggests that, if anything, the current study would reveal a conservative estimate of impairment in combined samples.

ASD probands

Because the ASD group contained five females only (7.25% of participants in this group), sex differences were not examined in this sample. Separate independent samples t-tests compared scores on each CC-A subscale in the ASD and control group. The ASD participants obtained lower standard scores than the normative group on each subscale (see Table 3). Scores on the Social Engagement subscale were particularly low for the ASD group, with the mean score around 2 SDs below the average of the control group.

To provide a comparison between groups for the total CC-A score, we calculated the proportion of individuals in each group scoring 1 SD or more below the mean on this measure. A large group difference was found, with around 90% of ASD probands meeting this criterion ($n = 62$), in comparison with 16% of the control group ($n = 30$), $\chi^2 = 119.28$ $df = 1$, $p < .001$, $\Phi = 0.68$. When we restricted the criterion to scoring 2 SD or more below the mean, the proportional difference between the ASD ($n = 47$, 68.1%) and control group ($n = 4$, 2.1%) remained highly significant, $\chi^2 = 137.53$ $df = 1$, $p < .001$, $\Phi = 0.73$. These findings confirm that the CC-A is sensitive to identifying social and communicative dysfunction in adults with ASD.

Parents of ASD probands

ANOVA then compared the scores of the ASD-par and normative samples on the three CC-A subscales. Participants' sex was included in the analysis as a second independent variable. There was a main effect of group, $F(3,419) = 13.04$, $p < .001$, Wilks' $\lambda = .92$, partial $\eta^2 = .09$, but no effect of sex ($p = .74$, partial $\eta^2 < .01$) nor an interaction between these two variables ($p = .89$, partial $\eta^2 = .01$). No group difference was found on the Language Structure subscale, $F(1,421) = .16$, $p = .69$ (see Table 4). There was a trend for the ASD-par group to score lower than the Typical group on the Pragmatic Skills subscale, but this did not reach statistical significance, $F(1,421) = 3.28$, $p = .07$. However, there was a significant difference in the same direction on the Social Engagement subscale, $F(1,421) = 19.32$, $p = .05$. Analysis of the total score of the two groups, found that proportionately more ASD-par ($n = 61$, 25.63%) than control participants ($n = 30$, 16%) scored 1 SD or more below the mean on this measure, $\chi^2 = 5.72$, $df = 1$, $p < .02$, $\Phi = 0.12$.

Previous studies have adopted a criterion of scoring 2 SDs or more below the mean of a given assessment as an indication of the broader autism phenotype (Bishop et al., 2006). Table 5 shows the rates of individuals in the ASD-par and control groups meeting this criterion on each CC-A measure, together with odds ratios. Effect sizes for each odds ratios were calculated using the formula $\ln(\text{odds ratio})/1.81$ (Chinn 2000). There was no significant difference in the rate of impairment on the Language Structure or Pragmatic Skills subscale or the CC-A total score. There was, however, a clear difference in the rate of impairment on the Social Engagement scale (ASD-par > controls), with a calculated odds ratio of around 2.5.

Phenotypic division?

We compared the distribution of impairments on the three subscales in both the ASD and ASD-par groups. This analysis was restricted to participants who had marked impairment (2SD below mean) on at least one subscale, leaving 57 participants in the ASD group (82.61%) and 31 participants in the ASD-par group (13.03%). The control group, which did not feature in this analysis, had 15 participants (8.02%) who scored 2SDs below the mean on one or more subscales (the comparison between the ASD-par control group approached significance, $\chi^2 = 2.72$, $df = 1$, $p = .1$, $\Phi = 0.13$). Figure 1 shows that there was a tendency for participants in the ASD group to score in multiple subscales ($n = 43$, 75.44%) rather than in a single subscale only ($n = 14$, 24.56%). This was significantly different to participants in the ASD-par group, who were more likely to score poorly on a single ($n = 22$, 71% of ASD-par group) rather than multiple subscales ($n = 9$, 29%), $\chi^2 = 17.89$, $df = 1$, $p < .001$, $\Phi = 0.45$.

A goal of this study was to determine whether the CC-A was able to identify qualitatively different social communication profiles in parents demonstrating the broader autism phenotype. As shown in Figure 1, six of 31 parents had impairment unique to the Pragmatic Skills Subscale, while just under half of the parents scored poorly on the Social Engagement subscale only. Under one-third of participants scored poorly on both of these subscales (29.03%). In contrast, selective impairments were much rarer in ASD probands: over two-thirds of participants scored poorly on both the Pragmatic Skills and Social Engagement subscales.

Discussion

This study confirmed that the CC-A is a valid measure of the social and communicative difficulties experienced by ASD probands. While close to a third of the ASD group scored poorly on all three scales, it was the subscales assessing social communication in which difficulties were most evident. The CC-A was also found to have some sensitivity in measuring the broader autism phenotype in parents of ASD probands. The proportion of parents demonstrating marked impairment on CC-A subscales (13%) was numerically higher than that in the control sample (8%), and similar to that found by other studies of ASD parents using instruments such as the Family History Inventory (11.3%, Bolton et al., 1994) and the Autism Spectrum Quotient (17.5%, Bishop et al., 2004). Once again, the predominant areas of difficulty were in the subscales of social communication, in particular the Social Engagement subscale, where the prevalence of significant difficulties was over twice the level of that reported in the control group.

Although the CC-A was sensitive to the broad social communication difficulties characteristic of ASD, it was not particularly able at identifying pragmatic subgroups within the ASD group. There was considerable overlap between the Pragmatic Skills and Social Engagement subscales for the ASD probands; all but one participant with marked impairment on the former subscale had similarly poor performance on the latter subscale. This finding could be considered unsurprising, given that individuals with ASD are

diagnosed on the basis of having a broad range of social and communication deficits. However, note that there was a significant minority of the ASD sample (19.3%) who scored poorly on the Social Engagement only. We suggest that these individuals correspond to the passive subtype of ASD proposed by Wing (1997), while ASD probands who fit the active but odd profile appear more difficult to identify using the CC-A.

The CC-A proved much better at distinguishing between subtypes of social-communicative impairment among parents showing the broader autism phenotype. Around 70% of the current sample scored poorly on either the Pragmatic Skills or Social Engagement subscales only, suggesting that different pragmatic profiles exist within the broader autism phenotype. These findings are supported by previous studies of ASD relatives. Murphy et al. (2000) administered a modified version of the Personality Assessment Schedule (Tyrer and Alexander, 1979), a standardised personality interview, to a large sample of adult relatives of children with ASD. A factor analysis of participants' scores yielded three prominent factors, two of which – 'difficult' (tactless and impulsive) and 'withdrawn' (undemonstrative, aloof and unresponsive) – appear also to map onto the two subtypes of ASD outlined by Wing (1997). It has been suggested that the social and non-social behaviours that are characteristic of ASD may result from different causal pathways (Happé and Ronald 2009). The current findings (and those of Murphy et al.) provide support for this view, in suggesting that distinct and aetiologically important phenotypes may exist *within* the social-communicative domain. Note, however, the finding that some parents of ASD probands show active but odd behaviours while others exhibit a passive communication style, speaks only to the familiarity of these characteristics. Twin studies, which are able to separate genetic from environmental influences, will be important in extending the data reported here.

Whitehouse et al. (2007) proposed that pragmatic but not structural language problems make up part of the broader autism phenotype. The finding that a lower proportion of ASD parents had impairment on the Structural Language subscale relative to the normative sample provides some support for the latter part of the hypothesis. Although conclusive evidence for this theory can only be obtained by examining the concordance of structural language abilities between generations, it is worth noting that all probands included in this study met full criteria for autism and therefore structural language difficulties are likely to have been present in at least some of these individuals (Whitehouse et al., 2008). If linguistic difficulties were part of the heritable phenotype of ASD, then we would have expected a greater proportion of individuals in the ASD-par group relative to the normative sample to score poorly on the Structural Language subscale. This was not the case. The conclusions we draw from the current data that structural language deficits are not part of the broader autism phenotype are in agreement with previous studies that have found little evidence for these difficulties among relatives of ASD probands (for a review, see Williams et al., 2008).

Support from this study for the former part of Whitehouse et al.'s (2007) theory – that pragmatic impairments are heritable in ASD – is not as clear-cut. While the Social Engagement subscale was sensitive to the communicative style of the parents of ASD, there was no difference between the ASD-par and normative samples on the Pragmatic Skills subscale both in terms of the mean group score or the proportion of individuals showing deficit. Hence, while the current study provides evidence for the heritability of a passive pragmatic phenotype in ASD, this was not observed for more explicitly odd behaviours. This finding has implications for our understanding of the social-communication traits that make up the broader autism phenotype, and requires further investigation in a range of different populations. For example, this finding may be an artifact of our study focusing on *parents* of affected children. It is possible that passive behaviours are less disruptive to relationships than the overtly odd communicative style measured by the Pragmatic Skills

subscale. Accordingly, individuals with the latter characteristics would be less likely to form stable romantic relationships and have children than individuals with the former characteristics. This hypothesis can be tested by using the CC-A to compare the social-communicative characteristics of relatives of ASD probands with- and without- a long-term romantic partner. This notwithstanding, the current study provides clear evidence that difficulties with social engagement, possibly reflecting a passive communication style, can occur independently of pragmatic difficulties, which reflect an 'active but odd' communication style, as well as structural language impairment. These features are less obvious than other pragmatic difficulties and it is possible that they may be overlooked in a research or clinical evaluation. However, the data presented here suggests that these are a quantifiable feature of this population and may form an important part of the broader autism phenotype.

The current study involved multiple research groups from different countries. However, while this approach enabled the recruitment of a large sample, it prevented the same investigator from administering the ADOS-G to every participant. It is possible that the inclusion of multiple testing sites introduced a degree of measurement error to the current findings. However, please note that all investigators had received training on ADOS-G administration and scoring, and that there were no statistically significant differences in CC-A scores between UK and non-UK samples.

In summary, the current study provides evidence for differences in the social and communication difficulties experienced by parents of ASD probands. While a minority of parents were reported to display overt pragmatic oddities, around half of parents meeting criteria for the broader autism phenotype showed a more passive style of communication. We suggest that molecular genetic studies of ASD may be facilitated by use of this scale, as it provides a quantitative measure of subthreshold autistic communicative behaviours and allows for detailed phenotypic differentiation between those individuals showing the social and communicative difficulties characteristic of the broader phenotype. Furthermore, it is quick and inexpensive to apply to the large numbers of participants that are needed for these types of investigations.

Acknowledgments

The authors would like to thank Christina Chrysler, Louise Gallagher, Miriam Law Smith and Ann Thompson, who generously shared their data for this study and to Ken Towbin for advice on US compatibility. Thank you also to David McDonald for collecting the normative data, Emma Whitehouse for providing feedback on this manuscript and to all of the participants for taking part in this research.

References

- Abrahams BS, Geschwind DH. Advances in autism genetics: on the threshold of a new neurobiology. *Nature Reviews Genetics*. 2008; 9:341–355.
- Bailey A, Le Couteur A, Gottesman I, Bolton P, Simonoff E, Yuzda E. Autism as a strongly genetic disorder: evidence from a British twin study. *Psychological Medicine*. 1995; 25:63–77. [PubMed: 7792363]
- Baron-Cohen S, Wheelwright S, Skinner R, Martin J, Clubley E. The autism-spectrum quotient (AQ): Evidence from Asperger syndrome/high-functioning autism, males and females, scientists and mathematicians. *Journal of Autism and Developmental Disorders*. 2001; 31:5–17. [PubMed: 11439754]
- Bishop, DVM. The Children's Communication Checklist, version 2 (CCC-2). Psychological Corporation; London: 2003.
- Bishop DVM, Maybery M, Maley A, Wong D, Hill W, Hallmayer J. Using self-report to identify the broad phenotype in parents of children with autistic spectrum disorders: a study using the Autism-

- Spectrum Quotient. *Journal of Child Psychology and Psychiatry*. 2004; 45:1431–1436. [PubMed: 15482503]
- Bishop DVM, Maybery M, Wong D, Maley A, Hallmayer J. Characteristics of the broader phenotype in autism: a study of siblings using the Children's Communication Checklist-2. *American Journal of Medical Genetics B: Neuropsychiatric Genetics*. 2006; 141:117–122.
- Bolton P, Macdonald H, Pickles A, Rios P, Goode S, Crowson M, et al. A Case - Control Family History Study of Autism. *Journal of Child Psychology and Psychiatry and Allied Disciplines*. 1994; 35:877–900.
- Buxbaum JD, Silverman JM, Smith CJ, Kilifarski M, Reichert J, Hollander E, et al. Evidence for a susceptibility gene for autism on chromosome 2 and for genetic heterogeneity. *American Journal of Medical Genetics B: Neuropsychiatric Genetics*. 2001; 68:1514–1520.
- Chin S. A simple method for converting an odds ratio to effect size for use in meta analysis. *Statistics in Medicine*. 2000; 19:3127–3131. [PubMed: 11113947]
- Constantino JN, Todd RD. Intergenerational transmission of subthreshold autistic traits in the general population. *Biological Psychiatry*. 2005; 57:655–660. [PubMed: 15780853]
- Dawson G, Estes A, Munson J, Schellenberg GD, Bernier R, Abbott R, et al. Quantitative assessment of autism symptoms in autism probands and parents: Broader Phenotype Autism Symptom Scale. *Journal of Autism and Developmental Disorders*. 2007; 37:523–536. [PubMed: 16868845]
- Dawson G, Webb S, Schellenberg GD, Dager S, Friedman S, Aylward E, et al. Defining the broader phenotype of autism: Genetic, brain, and behavioral perspectives. *Development and Psychopathology*. 2002; 14:581–611. [PubMed: 12349875]
- Happé F, Ronald A. The 'fractionable autism triad': A review of evidence from behavioural, genetic, cognitive and neural research. *Neuropsychology Review*. 2009; 18:287–304.
- Geurts HM, Hartman CA, Verté S, Oosterlaan J, Roeyers H, Sergeant JA. Pragmatics fragmented: The factor structure of the Children's Communication Checklist. *International Journal of Language and Communication Disorders*. in press.
- Landa R, Piven J, Wzorek MM, Gayle JO, Chase GA, Folstein SE. Social language use in parents of autistic individuals. *Psychological Medicine*. 1992; 22:245–254. [PubMed: 1574562]
- Lord C, Risi S, Lambrecht L, Cook EH, Leventhal BL, Dilavore PC, et al. The Autism Diagnostic Observation Schedule-Generic: A standard measure of social and communication deficits associated with the spectrum of autism. *Journal of Autism and Developmental Disorders*. 2000; 30:205–223. [PubMed: 11055457]
- Lord C, Rutter M, Le Couteur A. Autism Diagnostic Interview - Revised: A revised version of a diagnostic interview for caregivers of individuals with possible pervasive developmental disorders. *Journal of Autism and Developmental Disorders*. 1994; 24:659–685. [PubMed: 7814313]
- Murphy M, Bolton PF, Pickles A, Fombonne E, Piven J, Rutter M. Personality traits of the relatives of autistic probands. *Psychological Medicine*. 2000; 30:1411–1424. [PubMed: 11097081]
- Norbury CF, Nash M, Bishop DVM, Baird G. Using a parental checklist to identify diagnostic groups in children with communication impairment: A validation of the Children's Communication Checklist – 2. *International Journal of Language and Communication Disorders*. 2004; 39:345–364. [PubMed: 15204445]
- Pilowsky T, Yirmiya N, Shalev RS, Gross-Tsur V. Language abilities of siblings of children with autism. *Journal of Child Psychology and Psychiatry*. 2003; 44:914–925. [PubMed: 12959499]
- Piven J, Palmer P, Jacobi D, Childress D, Arndt S. Broader autism phenotype: evidence from a family history study of multiple-incidence autism families. *American Journal of Psychiatry*. 1997; 154:185–190. [PubMed: 9016266]
- Shao Y, Raiford KL, Wolpert CM, Cope HA, Ravan SA, Ashley-Koch AA, et al. Phenotypic homogeneity provides increased support for linkage on chromosome 2 in autistic disorder. *The American Journal of Human Genetics*. 2002; 70:1058–1061.
- Skuse DH. Rethinking the nature of genetic vulnerability to autistic spectrum disorders. *Trends in Genetics*. 2007; 23:387–395. [PubMed: 17630015]
- Todd RD. Genetic advances in autism hinge on the method of measuring symptoms. *Current Psychiatry Reports*. 2005; 7:133–7. [PubMed: 15802090]

- Tyrer P, Alexander J. Classification of personality disorder. *British Journal of Psychiatry*. 1979; 135:163–167. [PubMed: 497619]
- Whitehouse AJO, Barry JG, Bishop DVM. The broader language phenotype of autism: A comparison with Specific Language Impairment. *Journal of Child Psychology and Psychiatry*. 2007; 48:822–830. [PubMed: 17683454]
- Whitehouse AJO, Barry JG, Bishop DVM. Further defining the language impairment of autism: Is there a specific language impairment subtype? *Journal of Communication Disorders*. 2008; 41:319–336. [PubMed: 18295779]
- Whitehouse, AJO.; Bishop, DVM. 'The Children's Communication Checklist - Adult version (CC-A). Pearson; London: in press
- Whitehouse AJO, Line EA, Watt HJ, Bishop DVM. Qualitative aspects of developmental language impairment relates to language and literacy outcome in adulthood. *International Journal of Language and Communication Disorders*. in press.
- Williams D, Botting N, Boucher J. Language in autism and specific language impairment: Where are the links? *Psychological Bulletin*. 2008; 134:944–963. [PubMed: 18954162]
- Wing L. The autistic spectrum. *Lancet*. 1997; 350:1761–1766. [PubMed: 9413479]

Figure 1.

The number (and proportion) of ASD probands and parents of ASD probands (ASD-par) with marked impairment (≥ 2 SDs below mean) on the various combinations of the Language Structure, Pragmatic Skills and Social Engagement subscales of the CC-A

Table 1

Examples of items from each CC-A subscale

CC-A subscale	Item	
Language Structure	38	Makes mistakes in pronouncing long words; e.g. says “vegebable” rather than “vegetable” or “trellistope” rather than “telescope”
Pragmatic Skills	26	Moves the conversation to a favourite topic, even if others don’t seem interested in it
	35	It’s difficult to stop him/her from talking
	42	Includes over-precise information (e.g. exact date or time) in his/her talk, e.g. when asked “when did you go on holiday” may say “13th July 1995” rather than “in the summer”
Social Engagement	7	In social situations, s/he seems inattentive, distant or preoccupied
	56 [*]	Makes good use of gestures to get his/her meaning across
	67 [*]	Talks about his/her friends; shows interest in what they do and say

^{*} These are positively phrased items, which require the normal scoring system to be reversed (i.e., a rating of 3 would indicate deficit, while a rating of 0 would suggest appropriate communicative skill).

Table 2

Characteristics of the normative, ASD and ASD parents (ASD-par) groups. Information on the relation of respondents to the index case is also included

	Normative (N = 187)	ASD (N = 69)	ASD-par (N = 238)
Age	48.05 (19.56)	26.67 (7.98)	43.08 (9.97)
Range	17.4-79.9	17-65.9	26.4 – 77.4
Sex (M: F)	90:97	64:5	123:115
Respondent			
Parent	41 (21.93%)	65 (92.2%)	7 (2.94%)
Partner	128 (68.45%)	2 (2.9%)	222 (93.28%)
Child	13 (6.95%)	0 (–)	9 (3.78%)
Sibling	5 (2.67%)	0 (–)	0 (–)
Other	0 (–)	2 (2.9%)	0 (–)

Table 3

Mean standard scores for the three CC-A subscales for the ASD and normative samples. Standard deviations are in parentheses. Lower scores indicate greater levels of difficulty

	ASD (N = 69)	Normative (N = 187)	t	p	Effect size (d)
Language Structure	5.74 (3.19)	10.18 (2.85)	10.7	<.001	1.47
Pragmatic Skills	5.01 (3.08)	10.06 (2.84)	12.31	<.001	1.7
Social Engagement	3 (3.96)	10.05 (2.81)	13.58	<.001	2.05

Table 4

Mean standard scores (and SD) for the three subscales for the ASD-par and norm group. Effect sizes (Cohen's d) are presented for each comparison between the two groups

	ASD-par (n = 238)	Normative (n = 187)	Effect size (d)
Language Structure	10.31 (3.02)	10.18 (2.85)	0.04
Pragmatic Skills	9.55 (2.98)	10.06 (2.84)	0.18
Social Engagement	8.72 (3.33)	10.05 (2.81)	0.43

Table 5

Proportion (%) of ASD relatives and the normative sample with marked impairment (> 2 SDs below mean) on the three subscales and total score of the CC-A. Odds ratio (OR), p value and effect size of the comparisons are also included

	ASD-par	Normative	OR	p	Effect size
Language Structure	2.1	3.7	0.55	.31	0.33
Pragmatic Skills	6.3	4.3	1.5	.36	0.22
Social Engagement	10.1	4.3	2.51	.02	0.51
Total score	3.4	2.1	1.59	.45	0.26