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ORIGINAL PAPER

The Broad Autism Phenotype Questionnaire

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Abstract The broad autism phenotype (BAP) is a set of personality and language characteristics that reflect the phenotypic expression of the genetic liability to autism, in non-autistic relatives of autistic individuals. These characteristics are milder but qualitatively similar to the defining features of autism. A new instrument designed to measure the BAP in adults, the Broad Autism Phenotype Questionnaire (BAPQ), was administered to 86 parents of autistic individuals and 64 community control parents. Sensitivity and specificity of the BAPQ for detecting the BAP were high (>70%). Parents of children with autism had significantly higher scores on all three subscales: aloof personality, rigid personality, and pragmatic language. This instrument provides a valid and efficient measure for characterizing the BAP.

Keywords Autism · Broad autism phenotype · Assessment

Introduction

Autism is a behavioral syndrome defined by the presence of social and communication deficits; ritualistic, repetitive behaviors; and a characteristic course. Family and twin studies showing a high recurrence risk and heritability, respectively, have strongly implicated hereditary factors in the etiology of this condition. Folstein and Rutter (1977) used twin data to suggest that the vulnerability for autism may be inherited as milder traits, qualitatively similar to the defining features of autism. This milder expression of the underlying genetic liability for autism, as manifest in non-autistic relatives of autistic individuals, has come to be known as the broad autism phenotype (BAP).

In his detailed prose descriptions of autistic individuals and their families, Kanner (1943) was the first to note that a number of parents of autistic children were "serious minded, perfectionistic individuals, with an intense interest in abstract ideas" who appeared to lack a genuine interest in developing relationships with others. In their twin study of autism, on the basis of finding a higher concordance rate for a BAP characterized by selected cognitive deficits, Folstein and Rutter (1977) proposed the idea that the genetic liability for autism might be expressed in non-autistic relatives in characteristics that were milder but qualitatively similar to those seen in autism. Studies from several independent research efforts support this idea. In both a twin and a large-scale family study, both a high MZ concordance and familial aggregation of a more broadly defined phenotype (which included social and communication deficits and/or ritualistic-repetitive behaviors) were noted using the family history method (Bailey et al., 1995; Bolton et al., 1994). Subsequent studies observed qualitatively similar deficits in the social, stereotyped-repetitive, and communication domains using direct assessment with standardized measures of personality and language

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(Landa et al., 1992; Piven et al., 1994, Piven, Palmer, Landa et al., 1997). Together, these studies have provided convincing evidence of the existence of this phenomenon, i.e., the expression in relatives of the underlying genetic liability for autism in characteristics that are milder than but qualitatively similar to the defining features of autism, referred to as the BAP.

A variety of methods have been used to characterize components of the BAP. Bolton et al. (1994) used the Autism Family History Interview (AFHI) to assess the BAP in relatives of autistic probands and controls. Specifically, an informant provided information about the nuclear family as well as first- and second-degree relatives. The results indicated a higher rate of social, communication, and ritualistic-repetitive behaviors, defined as a composite of several items in each of these domains. Subsequent studies provided additional support for the validity of the BAP and the AFHI (Piven, Palmer, Jacobi, Childress, & Arndt, 1997). For example, Piven, Palmer, Landa et al., (1997) used the Modified Personality Assessment Schedule (MPASR) [derived from the MPAS (Piven et al., 1994) and PAS (Tyrer & Alexander, 1979)], the Pragmatic Rating Scale (PRS), and the Friendship Interview (FI) to directly assess parents of autistic individuals for features of the BAP. Parents and informants (usually a spouse) were interviewed and consensus ratings were made for specific traits. Parents who had more than one child with autism (i.e., from multiple-incidence families) were found to have elevated rates of selected personality characteristics—aloof, rigid, untactful, anxious, and ''hypersensitive to criticism,'' in comparison to parents of children with Down Syndrome (DS). These parents also reported having fewer high-quality, intimate friendships as measured by the FI and were observed to have more deficits in social use of language as measured on the PRS. Most recently, Dawson and colleagues constructed the Broad Autism Phenotype Symptom Scale (BAPSS), an amalgam of the family history and the direct assessment approaches, and demonstrated the heritability of components of the BAP (.16) (Sung et al., 2005).

To date, at least two questionnaires originally designed to measure autistic traits have been used to measure the BAP: the Social Responsiveness Scale (SRS) (Constantino, 2002) and the Autism-Spectrum Quotient (AQ) (Baron-Cohen, Wheelwright, Skinner, Martin, & Clubley, 2001). The SRS was primarily designed to measure reciprocal social behavior, but it also includes items pertaining to communication/language and stereotyped behaviors and interests. The SRS measures symptoms along a single dimension, ranging from normal to autistic disorder. Using an informant-only design, in which each parent filled out the SRS about their spouse's behavior, Constantino and Todd (2005) found moderate correlations between SRS scores of non-autistic twins from the general population (via maternal report) and of their parents. Most recently, SRS scores were reported to be elevated in siblings of autistic children (Constantino et al., 2006). The AQ was developed based upon the core features of autism and contains five subscales: social skills, attention switching, attention to detail, communication and imagination (Baron-Cohen et al., 2001). Baron-Cohen and colleagues detected group differences in AQ scores between high-functioning autistic individuals and controls; between control males and females; and between college students majoring in mathematics and humanities. Bishop et al. (2004) administered the AQ to parents of autistic individuals and reported that the social skills and communication subscales distinguished parents of autistic individuals from parents of normal controls.

The BAPQ was designed to efficiently and reliably measure particular personality and language characteristics that we have previously postulated as defining features of the BAP, including social personality, rigid personality and pragmatic language deficits (Piven et al., 1997), in non-autistic parents of autistic individuals. The BAPQ was derived from our experiences with the MPASR and PRS, which are direct assessment measures of selected personality and pragmatic language characteristics. These three BAPQ subscales provide quantitative indices relevant to the three DSM-IV domains of autism (American Psychiatric Association, 1994): social deficits, stereotyped-repetitive behaviors and social language deficits. In this paper, we present data on the internal consistency and intercorrelations of the BAPQ subscales, comparison of responses among a sample of parents of autistic individuals and control parents of typically developing individuals from the community, and the sensitivity and specificity of the BAPQ for detecting BAP characteristics based on direct, clinically based assessment with the MPASR and PRS.

Methods

Sample

Fifty-four parents of individuals with autism were recruited from the Autism Registry of the UNC Neurodevelopmental Disorders Research Center as part of our ongoing Family Study of Autism (for details, see Losh & Piven, 2006). This sample was

expanded by the addition of 32 parents of autistic individuals, determined in previous studies by our research group [the Collaborative Linkage Study of Autism, 2001, and the Iowa Family Study (Piven, Palmer, Landa et al., 1997)] to have the BAP on direct clinical assessment (i.e., aloof and/or rigid personality on the M-PAS-R and/or pragmatic language deficits on the PRS; see detailed description of these instruments below). Thus the total sample of parents of autistic individuals was 86. Given this recruitment strategy of enriching for autism parents with the BAP, this sample cannot be considered representative of a random sample of autism parents selected from the community. Informants (including 75 spouses and five close friends where spouses were unavailable) were identified for each parent and asked to fill out the informant-report section of the BAPQ. Informants were not available for six subjects. Among the spouse informants, 68 were the biological parent of the autistic proband; seven were spouses from marriages subsequent to the birth of the autistic proband. These step-parents acted as informants only and did not fill out the self-report section of the questionnaire.

A group of 64 parents of typically developing children (i.e., biological, married parents of a child over 5 years of age, who did not have a history of autism or a related developmental disorder, e.g., specific language impairment) were recruited from the same community to serve as a comparison group. The control parents were recruited via a mass email advertisement to all employees at the University of North Carolina at Chapel Hill. Control parents had no history of autism or a related developmental disorder in first-degree relatives, based on self-report on a questionnaire administered at the time of completion of the BAPQ.

Participants were offered $10 as incentive for completing the questionnaire. All subjects and informants gave informed consent consistent with the guidelines of the UNC IRB Review Committee. Parents of children with autism (referred to as ''autism parents'') were defined as being the biological parent of at least one child with a DSM-IV autistic disorder (American Psychiatric Association, 1994) diagnosis, who additionally met criteria for autism on the Autism Diagnostic Interview (ADI) (Le Couteur et al., 1989; Lord, Rutter, & Le Couteur, 1994). Autistic individuals were also directly assessed with the Autism Diagnostic Observation Schedule-G (ADOS-G) (Lord, Rutter, DiLavore, & Risi, 2006; Lord et al., 1989), which functioned as a check on the child's current behavior as reported by the parents on the ADI. All raters were trained to a criterion of >90% on the ADI and >80% on the ADOS.

Design of the BAPQ

The BAPQ items were derived from the contents of direct assessment interviews that had been previously used by our group (MPASR, PRS) and our experiences conducting and rating these videotaped interviews in over 300 parents of autistic individuals over the last 20 years. The overarching goal was to produce scales corresponding to three primary components of the BAP (aloofness and rigid personality from the MPASR and pragmatic language problems from the PRS) that parallel the defining domains of behavior in autism (social deficits, stereotyped, repetitive behavior, and communication abnormalities). In this study, *aloof personality* is defined as a lack of interest in or enjoyment of social interaction; *rigid personality* is defined as little interest in change or difficulty adjusting to change; and *pragmatic language problems* refer to deficits in the social aspects of language, resulting in difficulties communicating effectively or in holding a fluid, reciprocal conversation. For several reasons, previous studies by our group have suggested that these three behaviors are key components of the BAP. First, they have strong theoretical relevance as they correspond to the three defining behavioral domains of autism (social, stereotyped-repetitive, and communication deficits) (Piven, Palmer, Landa et al., 1997), as contrasted with other behaviors that we have previously measured but that are not conceptually related to the defining features of autism (e.g., anxious/worrying, hypersensitive to criticism, and untactful), and providing some support for the face validity of these concepts as components of the BAP. Second, we have been consistently able to reliably measure these aspects of the BAP by our clinical direct assessment tools (MPASR, PRS). Third, we have shown that these three characteristics occur more commonly in autism parents than do other BAP traits (e.g., untactful) (Piven, Palmer, Landa et al., 1997). Finally, other instruments are available for adequately measuring other behavioral constructs that could be considered part of the BAP [e.g., the neuroticism scale of the NEO-PI, which appears to adequately measure anxious/worrying in autism parents (Piven, Palmer, Landa et al., 1997)].

BAPQ items ask participants to rate how frequently each statement applies to them along a dimension in which a rating of one means that the statement very rarely applies, a rating of two indicates it applies rarely, three applies occasionally, four applies somewhat often, five applies often, and a rating of six indicates that the statement applies very often. This 6-point scale provides a range of possible responses and forces

ratings to fall above or below a value of neutral on each question. Selected items were worded to be reverse-scored, to limit the potential for a response set bias. Two versions of the BAPQ, a self- and informant-report versions were administered and were identical except for appropriately modified first- and third-person pronouns (e.g., ''He enjoys trying new things'' rather than ''I enjoy trying new things'').

A pool of potential items was generated, and pilot versions of the BAPQ were administered to a convenience sample of University of North Carolina employees and other acquaintances of the researchers in order to explore the distribution of responses provided by typical adults. During the first pilot study, 116 items from seven different subscale domains were administered to 27 individuals. Seven pilot subscales were determined a priori based on MPASR and PRS constructs (e.g., aloof personality), and items were derived from typical questions used during interviews. Items that were rarely endorsed (i.e., less than 10% of those sampled) were of particular interest. Items with distributions not conforming to this pattern were eliminated or revised. During a second pilot study, the remaining 105 items were administered to a convenience sample of 91 individuals. Four subscales aimed at aspects of the BAP that were not considered key features and that did not parallel defining characteristics of autism (e.g., anxious/worrying; hypersensitive to criticism, untactful and overly conscientious) were eliminated at this stage to focus the questionnaire on the three core and defining behavioral domains of primary interest in autism: social behavior (i.e., aloof personality), stereotyped-repetitive behavior (i.e., rigid personality) and communication (i.e., pragmatic language deficits). This resulted in a final set of 36 items (12 for each of three subscales) that were rarely endorsed among the pilot sample participants. These items (see self-report version of the BAPQ in the Appendix A) were administered to all participants in the sample reported in this study.

BAPQ Administration and Scoring

Autism parents were contacted via telephone and given the option of taking electronic or paper versions of the BAPQ. Control parents (UNC employees and parents of typically developing children) were contacted via a mass campus email and encouraged to enter their contact information, age, gender and preference for taking a paper or electronic version of the BAPQ on a secure registration web site. Parents who preferred paper copies were sent packets including questionnaires and instructions through the mail. Parents who preferred to take the BAPQ electronically were sent an email including a link to the web page where the questionnaire was hosted, a username and a randomly generated password necessary to log in.

To avoid potential response bias among parents, participants were asked to complete a Personality Styles and Preferences Questionnaire rather than the Broad Autism Phenotype Questionnaire, which explicitly refers to autism in the title. All participants were given three instructions to keep in mind while filling out the questionnaire: (1) to reference interactions they had with *most* people rather than special relationships such as those with immediate family members; (2) to think about the way they behaved the majority of their adult life rather than during selected time periods or transitory phases; and (3) to guess if they were unsure how to answer (thus insuring completion of all 36 items). After filling out the self-report section, participants were asked to complete an informant version about their spouse's or partner's behavior. Spouses/partners were instructed to work independently to avoid influencing each other's responses.

Summary scores for each of the three subscales were computed by reverse scoring the appropriate items (see Appendix), averaging across the 12 items for each subscale and averaging across all 36 items to create a total score. All summary scores therefore had a range 1–6. This procedure was carried out separately for the self- and informant-report scores for each participant, resulting in two sets of values for most parents. In order to combine the two versions into a single set of scores, best-estimate scores were computed by averaging the corresponding self- and informant-report values for each subscale. When participants only had scores on one version (self-report or informant), those values were used as the best estimate.

Direct Clinical Assessment of the BAP

Autism parents were examined in a face-to-face clinical interview to determine the presence or absence of components of the BAP. The Modified Personality Assessment Schedule Revised (M-PAS-R) and PRS are included in a two-hour semi-structured interview assessment for the BAP (Collaborative Linkage Study of Autism Group, 2001; Losh & Piven, 2006; Piven, Palmer, Landa et al., 1997). The interview was designed to elicit the subject's life story, including major life events, current and past social functioning and behavior at home, with friends and at work. For a more detailed description of this interview, see Piven, Palmer, Landa et al., (1997). Parents were interviewed about themselves and also served as informants for

their partners (usually a spouse and parent of the autistic child). All interviews were videotaped and rated by two independent raters trained to a reliability criterion of greater than 80%. Results from these interviews led to the identification of a sample of autism parents with the BAP (see definitions above) and participation in the current study.

The MPASR involves assessment of several personality constructs that define aspects of BAP, including having an aloof and rigid personality. Subjects (and informants interviewed separately) are asked to give specific behavioral examples illustrating the presence or absence of these characteristics. Subjects are asked to avoid focusing on periods when they may have suffered from an episode of a psychiatric disorder (e.g., major depression) but instead to give examples that were most indicative of their general style of functioning. Best-estimate ratings were based on behavioral examples provided by the subject and an informant (usually the subject's spouse) in response to probes and were not determined on the basis of observations or interpretations made by the interviewer. Ratings were made independently for each trait: a score of two indicated clear and compelling examples of behaviors specific to that trait, a score of one was assigned when the trait was endorsed but no specific examples were elicited and a score of zero was assigned when the behavior was not endorsed. Acceptable inter-rater agreement was achieved for both aloof (95% agreement, $\kappa = .81$) and rigid (86% agreement, $\kappa = .79$). Two independent raters made all ratings from videotapes of the interviews. Disagreements between these two raters were resolved by a third rater (JP, ML). To determine the sensitivity and specificity of the BAPQ for detecting aloof and rigid personality, scores on the MPASR and PRS were dichotomized into ''present'' or ''absent,'' with 0–1 on the MPASR resulting in the characteristic being classified as absent, while scores of two indicated the characteristic was present. According to this criterion, 27 autism parents were classified as having aloof personality and 20 with rigid personality. Fifteen parents were rated as both aloof and rigid.

The PRS is a measure of pragmatic language based on observations made during review of the videotaped subject interview on seven behavioral items, including whether the individuals provided excessive detail during conversation, frequently lost track of the conversation and engaged in conversational tangents (Landa et al., 1992; Piven, Palmer, Landa et al., 1997). Behaviors on the PRS were rated on a 3-point scale, with zero indicating normal behavior, one indicating moderate abnormality not considerably disruptive to the conversation and two indicating that the behavior was strikingly abnormal, causing the conversational partner to use compensatory strategies to maintain the flow of conversation. Ratings of one or two required the examiner to provide at least one example. Items were summed to produce a total PRS score, with a range of 0–33. High inter-rater reliability was established with these scores (90% agreement, $\kappa = .77$).

In order to determine clinically significant cutoffs, we reviewed the PRS ratings from our library of tapes (blind to participants' scores on the BAP-Q). Whereas scores of 3–4 were judged to reflect abnormal pragmatic language use, individuals scoring within this range typically displayed very mild impairments (rated as one) across a number of items, requiring refined clinical judgment to score reliably. More clear-cut pragmatic language difficulties, detectable by lay raters, were represented by scores of five or greater, and this cutoff was therefore set as the criterion for pragmatic language impairment. Twenty-one autism parents in this study had pragmatic language problems according to this criterion. The presence of pragmatic language problems often overlapped with the presence of aloof and rigid personality; seven of the 21 parents with pragmatic language impairments were also rated as aloof or rigid, with five parents rated as having all three characteristics present.

A composite diagnosis of the BAP was defined as the presence of at least two of three directly assessed BAP characteristics (aloof, rigid or pragmatic language deficit). A previous report by our group suggested that the presence of two or more BAP traits was highly specific for autism parents (from multiple-incidence autism families) versus parents of DS controls (Piven, Palmer, Landa et al., 1997). Composite scores were computed by summing the dichotomized (zero or one) scores from each of the three directly assessed characteristics (range 0–3). The 22 autism parents with scores of two or three were considered to meet criteria for a composite diagnosis of the BAP.

Results

Demographics

Eighty-six autism and 64 community control parents were included in the current sample. Autism and control parents were comparable on age (mean 47.9 years, SD 7.2; and 47.6 years, SD 7.3, respectively) and gender (43% versus 50% males, respectively). Thirty-eight (45%) autism parents and 36 (56%) of controls had an annual income over $80,000 annually; 38 (45%) autism parents and 28 (44%) of controls had

income between $40,000 and $80,000; eight (10%) autism and no control parents had an annual income less than $40,000. Twenty (23%) autism and three (5%) of control parents had a high school education; 39 (45%) autism and 30 (47%) of control parents attended college; and 27 (32%) autism versus 31 (48%) control parents had a graduate degree. Seventy-eight (91%) autism and 56 (88%) control parents had both self- and informant-report versions of the BAPQ. Thirty (35%) autism and 57 (89%) control parents completed an electronic version of the BAPQ, while the remainder completed paper versions. One reason for this differential was that many autism parents participated in a concurrent family study of autism and were given paper copies as part of that study, while control parents were recruited only via a mass email to employees of the University of North Carolina. There was no difference in BAPQ total scores in autism or control parents based on method of administration (autism parents [electronic version mean (SD) = 2.85(73); paper version mean (SD) = 2.93 (72); $t(84) = .5$, $p = .619$]; control parents: [electronic version mean (SD) = 2.76(55); paper version mean (SD) = 2.55(.6); $t(62) = .93$, $p = .356$]).

Internal Consistency

Inter-item reliability for each subscale was examined. Cronbach's α coefficient was .94 for the aloof subscale, .91 for the rigid subscale, .85 for the pragmatic language subscale and .95 across all 36 items. Item-total correlations for each individual item relative to the rest of the items in that subscale (with that item removed) were ≥.39. Inter-item reliability did not differ between self- and informant-report versions, between male and female participants or between autism and control parents. Therefore, best-estimate ratings employed an average of self- and informant-report ratings. Inter-item reliability was also examined separately for the positively–negatively valenced items to assess for a potential response bias based on polarity. Cronbach's α was 0.9 for both the 21 positively scored (mean [SD] = 2.79 [.61]), and 15 reversed-scored (mean [SD] = 2.89 [.8]) items.

All three subscales were significantly ($p < .001$) correlated with each other, among both autism (aloof and rigid $-r = .72$, aloof and pragmatic language $-r = .61$, and rigid and pragmatic language $-r = .61$) and control (aloof and rigid $-r = .54$, aloof and pragmatic $-r = .53$, and for rigid and pragmatic language $-r = .51$) parents. Subscale intercorrelations were comparable for male and female subjects and for self- and informant-report versions.

Sensitivity and Specificity

The number of autism parents classified by direct clinical assessment as present or absent for the three BAP components and for the composite rating, and the sensitivity and specificity of the BAPQ subscales and total score, are presented in Table 1. Receiver operator curves were calculated to estimate sensitivity and specificity for predicting the presence of the BAP (as characterized by direct clinical assessment measures) across every possible value of the predictor variable, the BAPQ subscale score. Scores on each BAPQ subscale were used to predict the presence or absence of their corresponding direct clinical assessment traits (e.g., BAPQ aloof to predict MPASR aloof), and BAPQ Total Score was used to predict composite diagnosis scores (dichotomized into present vs. absent as described above). Sensitivity refers to the proportion of autism parents with the BAP on direct clinical assessment who were correctly classified as present by the BAPQ. Specificity refers to the proportion of autism parents without the BAP on direct clinical assessment who were correctly classified as negative by the BAPQ. As cutoff values become more conservative, specificity is increased and sensitivity is decreased. Optimal cutoffs for each subscale were defined as the point where both sensitivity and specificity were maximized, with the goal of maximizing sensitivity and specificity. The cutoff values for the BAPQ threshold determined by this method are presented in Table 1 along with the sensitivity and specificity values. Sensitivity and specificity were at or above 70% for all subscales and over 80% for two of the three subscales. Sensitivity and specificity were approximately 80% for the total BAPQ score.

BAPQ Scores in Autism Parents with and Without the BAP and Control Parents

Mean BAPQ subscale scores for control parents and for autism parents with and without BAP components (aloof, rigid, pragmatic language) on direct clinical assessment appear in Fig. 1 and Table 2. Mean differences between the three groups were examined using ANCOVA, including education and household income as covariates. Significant differences were found among the three groups on all three BAPQ subscales and the total BAPQ score: $F(2,143) = 24.78$, $p < .001$, $F(2,143) = 5.09$, $p < .01$, and $F(2,143) = 10.21$, $p < .001$, $F(2,143) = 22.25$, $p < .001$, for aloof, rigid, pragmatic language, and total scores, respectively. Bonferroni-adjusted contrasts were employed to assess the pair-wise post-hoc comparisons for controls vs.

Table 1 Sensitivity and specificity of the best estimate BAPQ[a]

BAPQ subscale	BAPQ cutoff	BAP absent[b]	BAP present[b]	Sensitivity (%)	Specificity (%)
Aloof	3.25	59	27	77.8	81.4
Rigid	3.50	66	20	70.0	81.8
Pragmatic language	2.75	65	21	76.2	73.8
Total score	3.15	64	22	81.8	78.1

[a] Best estimate from self-report and informant ratings

[b] Frequency on direct assessment measures (MPASR and PRS)

BAPQ Broad Autism Phenotype Questionnaire; *BAP* Broad autism phenotype; *MPASR* Modified Personality Assessment Schedule; *PRS* Pragmatic Rating Scale

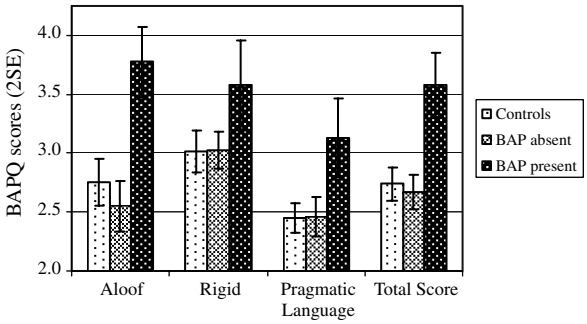

Fig. 1 BAPQ Subscale Scores in Autism Parents with and without the BAP, and Controls

BAP absent, BAP absent vs. BAP present, and BAP present vs. controls on each subscale and for the total BAPQ score. There were no significant mean BAPQ differences ($p > .05$) between control and BAP-negative parents on any of the subscale or total BAPQ scores. Autism parents with the BAP present had significantly higher BAPQ scores than BAP-negative and control parents on all three subscales and on total BAPQ score.

Comparison of Self- and Informant-report Versions of the BAPQ

Self- and informant-report versions of the BAPQ were compared in the subset of autism parents ($N = 98$) and control parents ($N = 56$) where both self- and informant-report data were available. Mean self-report, informant-report and best-estimate scores for each BAPQ subscale appear in Fig. 2. Informant scores were slightly higher than self-report scores on all subscales and the total BAPQ score, although differences were not significant. Sensitivity and specificity were computed for self- and informant-report versions and are presented in Table 3. ROC analyses were used to determine optimal cutoffs for each version. Sensitivity and specificity of the informant version of the BAPQ, for direct assessment diagnosis of the BAP, were uniformly higher than the self-report version for all subscales and total score (with the exception of the characteristic ''rigid,'' where self- and informant-report versions were equally sensitive). Differences were most striking for specificity on the rigid subscale where self- and informant-report specificity were 49% and 74%, respectively, and for the pragmatic language subscale, where self- and informant-report sensitivity were 56% and 72%, respectively. Sensitivity and specificity for informant and best-estimate versions were comparable, with the exception of a slightly better performance of the best-estimate versions for the rigid and pragmatic language subscales. Self- and informant-report correlations, while significant for both autism parents (aloof: $r = .55$, $p < .001$; rigid: $r = .32$, $p < .01$; pragmatic language $r = .31$, $p < .01$; total BAPQ:

Table 2 Comparison of BAPQ subscale scores in autism parents with and without the BAP[a] and control parents

BAP-Q subscale	Controls M (SD)	BAP absent M (SD)	BAP present M (SD)	F-test: DF Bet (within)	F	*p*	Contrasts: significance (*p*)		
							control vs. BAP absent	control vs. BAP present	BAP present vs. BAP absent
Aloof	2.75 (.78)	2.55 (.83)	3.77 (.77)	2 (147)	22.81	<.001	.262	<.001	<.001
Prag. language	2.45 (.51)	2.46 (.68)	3.13 (.76)	2 (123)	10.58	<.001	1.00	.000	.000
Rigid	3.02 (.71)	3.03 (.63)	3.58 (.86)	2 (136)	5.45	.005	1.00	.018	.007
Total score	2.74 (.55)	2.67 (.58)	3.58 (.65)	2 (123)	21.42	<.001	.799	<.001	<.001

[a] As determined by direct assessment (MPASR and PRS)

BAPQ Broad Autism Phenotype Questionnaire; *BAP* Broad autism phenotype; *MPASR* Modified Personality Assessment Schedule; *PRS* Pragmatic Rating Scale

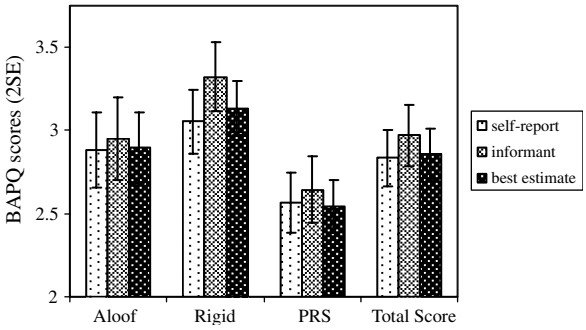

Fig. 2 Comparison of Self-report, Informant-report, and Best Estimate BAPQ Scores in Parents of Autistic Individuals

$r = .39$, $p < .001$) and control parents (aloof: $r = .70$, $p < .001$; rigid: $r = .57$, $p < .001$; pragmatic language $r = .48$, $p < .001$; total BAPQ: $r = .66$, $p < .001$) were greater for controls overall and for the characteristic "aloof."

Gender Differences

Sensitivity and specificity of the best estimate version of the BAPQ were examined separately for males and females and are presented in Table 4. Optimal ROC cutoffs were calculated. Sensitivity and specificity are roughly similar for males and females with the exception of an increased sensitivity for males with aloof personality and for females with pragmatic language deficits and an increased specificity for females with aloof personality. Sample size was too small for a meaningful separate analysis of self- and informant-report gender effects.

Discussion

The BAPQ was designed to measure aloof personality, rigid personality and pragmatic language deficits: key personality and language components of the BAP. These three components parallel the social deficits, stereotyped-repetitive behaviors and social language deficits that define the syndrome of autism (American Psychiatric Association, 1994). The BAPQ was developed on the basis of 20 years of experience characterizing the BAP through family studies using direct, clinically based assessment tools. The clinical assessment tools and definitions are therefore thought to have substantial face validity as measures of the BAP. The present data demonstrate that the subscales of the BAPQ have internal consistency; have high sensitivity and specificity for the direct, clinical assessment ratings of the BAP; and differentiate autism parents with a clinically defined BAP from both autism parents without direct clinical evidence of the BAP and from community control parents (not expected to have high rates of the BAP).

Our original goal was to develop a screening tool for efficient, valid and reliable detection of autism parents

Table 3 Sensitivity and specificity for self-report vs. informant-report versions of the BAPQ

BAPQ subscale	Version	BAPQ cutoff	BAP absent	BAP present	Sensitivity (%)	Specificity (%)
Aloof	Self-report	3.00	55	23	74	71
	Informant	3.50	55	23	78	80
Rigid	Self-report	2.90	61	17	71	49
	Informant	3.60	61	17	71	74
Pragmatic language	Self-report	2.60	60	18	56	62
	Informant	2.70	60	18	72	67
Total score	Self-report	3.00	60	18	67	63
	Informant	3.30	60	18	78	82

BAPQ Broad Autism Phenotype Questionnaire; *BAP* Broad autism phenotype

Table 4 Sensitivity and specificity of the BAPQ by gender

BAPQ subscale	BAPQ cutoff	BAP absent	BAP present	Sensitivity (%)	Specificity (%)
Males					
Aloof	3.25	19	16	88	68
Rigid	3.65	22	13	69	77
Pragmatic language	2.95	22	13	69	77
Total score	3.35	21	14	71	81
Females					
Aloof	3.00	36	7	71	78
Rigid	3.25	39	4	75	82
Pragmatic language	2.70	38	5	80	79
Total score	3.25	39	4	75	87

BAPQ Broad Autism Phenotype Questionnaire; *BAP* Broad autism phenotype

with the BAP. On the basis of this screening, identified parents of autistic individuals might then undergo subsequent direct clinical assessment to confirm the presence of the BAP. We hoped that the BAPQ would have high sensitivity and at least adequate specificity, both characteristics of a good screening tool. However, ROC analyses of the BAPQ, based on information from both self- and informant-report scores, revealed both high sensitivity and specificity for the three BAP components we measured and for a composite diagnosis of the BAP. This finding suggests that the BAPQ may be useful as an efficient and valid diagnostic tool for identifying individuals with the BAP. This may be particularly valuable in circumstances where direct assessments are not practical. The cutoffs provided from the ROC analyses provide a rational approach to categorically defining individuals with and without components of the BAP, and the ordinal nature of the BAPQ total and subscale scores affords a quantitative index of the severity of the BAP. Both approaches (as a screening and a diagnostic tool) are likely to be useful applications of this instrument.

Information from self- and informant-report was examined separately and combined into a best-estimate rating. Given the effort required to get information from both subjects and informants, an obvious question is whether this two-pronged approach is necessary. Although the sensitivity and specificity of the best-estimate BAPQ was consistently better than informant-only scores, for the most part these two approaches yielded comparable results, suggesting that informant information may be sufficient. However, the size of the present sample was insufficient to fully examine the characteristics of self-report and informant ratings (e.g., gender of both subject and informant). Without further data on the BAPQ, it would seem prudent to collect both self- and informant-report ratings when possible.

The BAPQ also appears to be valid for identification of the BAP in both males and females, although, as noted above, further studies are needed to examine the effect of informant gender and informant BAP status on sensitivity and specificity. The sensitivity and specificity of self-report scores were consistently lower than both best-estimate and informant-only ratings (especially in the case of specificity for the characteristic ''rigid,'' sensitivity for pragmatic language deficits and specificity for total BAP score), suggesting that self-report-only ratings are less valid than either best-estimate or informant-only information. The literature on reporter effects in personality disorders is consistent with this: self-report scores on personality-disorder inventories tend to be weakly related to clinician diagnoses (Hyler et al., 1989), and in direct assessment personality-disorder interviews, ratings between separate informants are more strongly correlated than ratings between subjects and informants (Perry, 1992).

Comparison with Other Instruments Employed to Measure the BAP

The SRS (Constantino, 2002) and the AQ (Baron-Cohen et al., 2001) are questionnaires that have been employed with first-degree relatives of individuals with autism to measure BAP characteristics. Both were designed to measure behaviors ranging from those seen in autism to that characteristic of normal functioning. While the SRS primarily measures social behavior, the AQ measures five constructs, including the three behavioral domains that define autism and the domains of imagination and attention switching. Both instruments are also designed for assessment of characteristics in children and adults (there are both 4–18-year-old and adult versions of the SRS, and the AQ has been given to individuals ranging from 16 to 58). In contrast, the BAPQ was designed specifically to measure the BAP in adults. The questions were based upon the often subtle way BAP traits are manifested among parents of individuals with autism who display aspects of the BAP, and the BAPQ has been shown in the present study to have high sensitivity and specificity for a clinically derived direct assessment of traits previously shown to aggregate in parents of autistic individuals.

Like the BAPQ, the BPASS (Sung et al., 2005) was specifically designed to measure BAP symptoms, and there are strong similarities among many of the constructs being measured; both the BPASS and the MPASR and PRS derive from measures (the AFHI and MPAS) developed for the Baltimore and British Family Studies of Autism (Bolton et al., 1994; Piven et al., 1994). Unlike the BAPQ, the BPASS is a lengthy tool that includes both direct interview and observation portions; it requires special training for valid administration. In contrast, the BAPQ is inexpensive to administer, can be delivered remotely, and can be administered and scored with minimal instruction. Like the AQ and SRS, the BPASS is aimed at a wider developmental scope than the BAPQ, with items appropriate for both children and adults. Also like the SRS, heritability has been demonstrated for the BPASS.

Strengths, Limitations, and Future Directions

The BAPQ is unique in being specifically designed to measure the BAP, using content based upon clinical observation of BAP characteristics in parents of

autistic individuals and using direct assessment tools that have been validated in several case–control family studies. The BAPQ includes subscales aimed at components of the BAP that are conceptually distinct and that map onto the corresponding behavioral domains that define autism. The high sensitivity and specificity of the BAPQ suggest that it may be useful as both a screening tool for affected individuals in the population and as a diagnostic tool, particularly when more labor-intensive direct assessment methods are not practical. Finally, the BAPQ is simple to administer and requires no clinical expertise.

There were several limitations to the current study. The number of parents with BAP characteristics present was not large enough to fully examine the role of self- and informant-report characteristics such as gender or BAP status of the rater. The ascertainment strategy employed in this study (i.e., enriching our sample by adding autism parents known to have the BAP) limited our ability to assess population rates of the BAP in parents of autistic individuals. Comparison of the BAPQ with direct-assessment measures employed sensitivity and specificity estimates that are directly tied to the number of affected individuals in the sample. These estimates are therefore biased by our strategy to selectively recruit parents known to have BAP traits. Clearly, a large-scale study of rates of BAPQ characteristics in the population of parents with autistic children, as well as in the general population, would be of great interest and would circumvent these ascertainment biases. Normative data would allow the examination of the quantitative nature of BAPQ traits in autism relatives as well as the general population. A large-scale epidemiologic study would be helpful to determine population prevalence rates of BAP characteristics as measured by the BAPQ. Examination of the relationship of the BAPQ subscales to other existing instruments (e.g., the NEO-PI and SRS) would add to the validity of the BAPQ. Future studies should also examine the test-retest reliability of BAPQ.

In summary, the BAPQ is a simple, efficient and valid instrument for diagnosis and characterization of the BAP in adult relatives of autistic individuals. Although additional studies are warranted to elucidate the psychometric properties of this instrument, to document reliability and to provide further evidence of its validity, the present data suggest that the BAPQ is likely to find numerous uses in the increasing number of studies aimed at characterizing the milder expression of the genetic liability for autism in non-autistic relatives of autistic individuals.

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Appendix A

Instructions

You are about to fill out a series of statements related to personality and lifestyle. For each question, circle that answer that best describes how often that statement applies to you. Many of these questions ask about your interactions with other people. Please think about the way you are with most people, rather than special relationships you may have with spouses or significant others, children, siblings, and parents. Everyone changes over time, which can make it hard to fill out questions about personality. Think about the way you have been the majority of your adult life, rather than the way you were as a teenager, or times you may have felt different than normal. You must answer each question, and give only one answer per question. If you are confused, please give it your best guess.

1—Very rarely	2—Rarely	3—Occasionally
4—Somewhat often	5—Often	6—Very often

Questions:

1.	I like being around other people	1 2 3 4 5 6
2.	I find it hard to get my words out smoothly	1 2 3 4 5 6
3.	I am comfortable with unexpected changes in plans	1 2 3 4 5 6
4.	It's hard for me to avoid getting sidetracked in conversation	1 2 3 4 5 6
5.	I would rather talk to people to get information than to socialize	1 2 3 4 5 6
6.	People have to talk me into trying something new	1 2 3 4 5 6
7.	I am ''in-tune'' with the other person during conversation***	1 2 3 4 5 6
8.	I have to warm myself up to the idea of visiting an unfamiliar place	1 2 3 4 5 6
9.	I enjoy being in social situations	1 2 3 4 5 6
10.	My voice has a flat or monotone sound to it	1 2 3 4 5 6
11.	I feel disconnected or ''out of sync'' in conversations with others***	1 2 3 4 5 6
12.	People find it easy to approach me***	1 2 3 4 5 6
13.	I feel a strong need for sameness from day to day	1 2 3 4 5 6
14.	People ask me to repeat things I've said because they don't understand	1 2 3 4 5 6
15.	I am flexible about how things should be done	1 2 3 4 5 6
16.	I look forward to situations where I can meet new people	1 2 3 4 5 6

17. I have been told that I talk too much about certain topics	1 2 3 4 5 6
18. When I make conversation it is just to be polite***	1 2 3 4 5 6
19. I look forward to trying new things	1 2 3 4 5 6
20. I speak too loudly or softly	1 2 3 4 5 6
21. I can tell when someone is not interested in what I am saying***	1 2 3 4 5 6
22. I have a hard time dealing with changes in my routine	1 2 3 4 5 6
23. I am good at making small talk***	1 2 3 4 5 6
24. I act very set in my ways	1 2 3 4 5 6
25. I feel like I am really connecting with other people	1 2 3 4 5 6
26. People get frustrated by my unwillingness to bend	1 2 3 4 5 6
27. Conversation bores me***	1 2 3 4 5 6
28. I am warm and friendly in my interactions with others***	1 2 3 4 5 6
29. I leave long pauses in conversation	1 2 3 4 5 6
30. I alter my daily routine by trying something different	1 2 3 4 5 6
31. I prefer to be alone rather than with others	1 2 3 4 5 6
32. I lose track of my original point when talking to people	1 2 3 4 5 6
33. I like to closely follow a routine while working	1 2 3 4 5 6
34. I can tell when it is time to change topics in conversation ***	1 2 3 4 5 6
35. I keep doing things the way I know, even if another way might be better	1 2 3 4 5 6
36. I enjoy chatting with people ***	1 2 3 4 5 6

***Casual interaction with acquaintances, rather than special relationships such as with close friends and family members.

Scoring Instructions

Reverse scored items (1 becomes 6, 5 becomes 2, etc.): 1, 3, 7, 9, 12, 15, 16, 19, 21, 23, 25, 28, 30, 34, 36.

Items by Subscale:

Aloof (1, 5, 9, 12, 16, 18, 23, 25, 27, 28, 31, 36)

Pragmatic Language (2, 4, 7, 10, 11, 14, 17, 20, 21, 29, 32, 34)

Rigid (3, 6, 8, 13, 15, 19, 22, 24, 26, 30, 33, 35)

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