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The Broad Autism Phenotype Questionnaire: Prevalence and Diagnostic Classification

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Abstract

The Broad Autism Phenotype Questionnaire (BAPQ; Hurley et al, 2007) was administered to a large community-based sample of biological parents of children with autism (PCAs) and comparison parents (CPs) (n = 1692). Exploratory factor analysis and internal consistency parameters confirmed a robust three factor structure of the BAPQ, corresponding to the proposed aloof, pragmatic language and rigidity subscales. Based upon the distribution of BAP features in the general population, new normative cutoff values for BAPQ subscales were established that provide increased specificity relative to those previously reported (Hurley et al, 2007), and thus enhance the utility of the BAPQ for diagnostically classifying the BAP. These cutoffs were also used to estimate prevalence of the BAP and its three components, with rates ranging between 14 – 23% for PCAs and between 5 – 9% for CPs. Analysis of patterns of BAP characteristics within family members revealed that BAP features were more likely to co-occur in PCAs relative to CPs. Collectively, these findings extend the utility of the BAPQ and provide additional evidence that it is an efficient and reliable tool for disaggregating the heterogeneity of autism through the identification of meaningful subgroups of parents.

Keywords

Autism; Broad Autism Phenotype; Assessment; Prevalence; Genetics

Introduction

Autism is a heritable neurodevelopmental disorder defined by impairments in social interaction and communication, and repetitive and restricted behaviors. Considerable evidence has accumulated indicating that these characteristics extend below clinical threshold into the “Broad Autism Phenotype” (BAP), a term referring to the presence of

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milder but qualitatively similar forms of autism symptoms persisting outside the categorical boundary for diagnosis (Piven, 2001). Although BAP features are continuously distributed within the general population (Constantino and Todd, 2005; Happé et al., 2006; Robinson et al., 2011), they are most frequently and prominently found in first-degree relatives of individuals with autism (Bailey et al., 1995; Bolton et al., 1994; Piven et al., 1994, Piven et al., 1997a; Piven et al., 1997b; Sung et al., 2005). Prevalence estimates suggest that between 20% and 50% of family members exhibit at least one BAP feature (Bolton et al., 1994; Dawson et al., 2007), with higher rates occurring in multiple incidence families (Bernier et al., 2012; Losh et al., 2008). Indeed, a range of characteristics associated with autism have been reported at higher than chance levels for relatives of individuals with autism, including reduced quantity and quality of friendships (Piven et al., 1997b; Santangelo and Folstein, 1995), rigid, aloof and anxious personality (Austin, 2005; Murphy et al., 2000; Piven et al., 1997b), pragmatic language deficits (Landa et al., 1992; Piven et al., 1997b), weak central coherence (Happé et al., 2001), reduced emotional understanding (Szatmari et al., 2008), abnormal response to gaze (Adolphs et al., 2008; Scheeren and Stauder, 2008), and impairments in executive function (Hughes et al., 1997; Ozonoff et al., 1993) and social cognition (Baron-Cohen and Hammer, 1997; Losh and Piven, 2007).

A number of methods have been developed to quantify the BAP in relatives of individuals with autism. Initial efforts by Bolton et al (1994) used the Autism Family History Interview (AFHI) with informants to provide information about BAP characteristics within nuclear families and first- and second-degree relatives. Other interview methods such as the Modified Personality Assessment Schedule Revised (MPAS-R; Piven et al., 1997b), the Pragmatic Rating Scale (PRS; Landa et al., 1992) and the Friendship Interview (Piven et al., 1997b) further characterized the BAP by directly assessing parents in addition to informants. More recently, the Broad Autism Phenotype Symptom Scale (Sung et al., 2005) combined family history and direct assessment approaches to estimate heritability of BAP components. Together, these interview and assessment instruments constitute the gold standard for maximizing validity when identifying and quantifying the BAP (for a review, see Losh et al., 2011). However, because each is time-intensive and requires extensive training to administer and score, they do so at the expense of cost, making measurement of BAP traits in studies with limited resources and/or large samples prohibitive.

To provide an efficient, reliable and easily-administered assessment of the BAP requiring no clinical expertise, the Broad Autism Phenotype Questionnaire (BAPQ; Hurley et al., 2007) was developed based upon the direct assessment interview measures used by Piven and colleagues (i.e., the MPAS-R and the PRS). The BAPQ, which includes both self- and informant-report versions, consists of three subscales corresponding to the triad of characteristics associated with the primary diagnostic domains of autism: 1) social abnormalities, 2) pragmatic language difficulties and 3) rigid personality and a desire for sameness. Unlike other self-report measures of autism symptomology like the Autism Quotient (AQ) or the Social Responsiveness Scale (SRS), the BAPQ was specifically designed to assess BAP traits in unaffected individuals and is validated against direct clinical assessments of the BAP.

The BAPQ was originally evaluated with a sample of 86 parents of children with autism (PCAs) and 64 comparison parents (CPs) (Hurley et al., 2007). PCAs were also assessed with the MPAS-R and the PRS to examine comparability of the BAPQ relative to direct interview measures. Internal consistency of the subscales was high (>0.85), and results suggested good sensitivity and specificity for identifying the BAP (both $>70\%$), with PCAs scoring significantly higher than CPs across all three subscales. More recent psychometric analysis of the BAPQ suggests it outperforms the AQ and SRS for assessing BAP traits within the general population due to the high internal consistency of its subscales that together reflect a factor structure corresponding to the full nature of the BAP (Ingersoll et al., 2011). Together, these initial analyses suggest that the BAPQ effectively identifies dissociable BAP traits in both family members of individuals with autism and the broader population.

The current study uses a large community-based sample to replicate previous BAPQ findings and provide a more stable estimate of its psychometric properties. Additionally, we extend beyond Hurley et al (2007) in a number of important ways. First, we identify new cutoff scores that expand the utility of the BAPQ. Although the BAPQ was initially devised to function as a screening tool for the BAP, the resulting high specificity and sensitivity raised the possibility of using it to diagnostically classify the BAP (e.g., to provide additional phenotypic information on unaffected individuals in molecular genetic studies searching for associated genetic markers). The original cutoffs reported by Hurley et al (2007), however, were not intended to differentiate between screening and diagnostic goals. We aim to do so here. Second, the larger sample used here enables the establishment of separate cutoff scores for self and informant versions of the BAPQ in addition to a composite rating combining the two. Third, we evaluate BAP prevalence rates in PCA mothers and fathers and establish normative rates of BAP prevalence in the general population. Finally, we examine whether multiple BAP features occur more frequently for PCAs relative to CPs.

Methods

Sample

Parents of children with a DSM-IV clinical diagnosis of Autistic Disorder, Asperger's Disorder, or Pervasive Developmental Disorder-Not Otherwise Specified, as determined by a licensed clinician with expertise in autism diagnosis and assessment, were recruited from the Autism Registry of the Carolina Institute for Developmental Disabilities (CIDD). The Autism Registry consists of approximately 5000 nuclear families of an individual with a diagnosis on the autism spectrum that have consented to be contacted regarding possible participation in research studies on autism. We mailed invitations and study materials to 1176 nuclear families who met the following inclusion criteria: parents were biological (i.e., adoptive and step-parents, and other guardians were excluded), aged 18–65 years, primarily English-speaking, and had a child 18 years old or younger who had received a clinical evaluation from TEACCH (Treatment and Education of Autistic and related Communication-handicapped Children) clinics or prior CIDD research projects. Parents were not pre-selected for BAP traits and should be generally representative of community

PCAs. Diagnoses of the children in the recruited families were 79.6% Autistic Disorder, 10.5% Asperger's Disorder and 9.9% PDD-NOS. Stamped, addressed envelopes were included in the mailing so that questionnaires were returned to the research team. Reminder postcards were mailed after three weeks to 744 non-responders and follow-up calls were also made to 150 randomly selected families. Parents were offered \$10 to participate. 60.5% of invited families had at least one parent participate, for a total of 711 parents and 344 children. Finally, to examine the sensitivity and specificity of the BAPQ relative to gold standard direct assessment measures, 35 PCAs were also assessed with the MPAS-R and the PRS. These parents were participants in concurrent CIDD studies that included these clinical assessments.

Nine hundred eighty one CPs participated in this study and were recruited through community mailings to families based on North Carolina birth records who had signed a form indicating an interest in participating in research studies (for more details, see Reznick et al., 2007). These families are largely demographically representative of the local population, though as is common in mailed surveys, they are disproportionately Caucasian and more highly educated. We did not screen these children or their parents for developmental disabilities (including autism or BAP). Thus, these parents should represent a community comparison group that reflects the rate of these characteristics within the general population. We specified that only biological parents should participate to ensure equivalence with the PCAs on characteristics related to mate acquisition. CPs were offered \$10 for participating.

Participating parents were asked to also have their spouses or close friends (when spouses were unavailable) complete the informant report version of the BAPQ. All participants provided informed consent, and the protocol for this study was approved by the UNC IRB Review Committee.

Measures

Assessment of the Broad Autism Phenotype in Parents—The BAPQ (Hurley et al., 2007) is a self and informant-report questionnaire consisting of 36 items spread across three 12 item subscales derived from direct assessment interviews (social aloofness and rigid personality from the MPAS-R and pragmatic language abnormalities from the Modified PRS). Items are rated along a six point Likert scale (ranging from “very rarely” to “very often”), which forces ratings to fall above or below a value of neutral on each question. Original internal consistency analysis of the subscales (Hurley et al., 2007) supported this three component model, which is consistent with traditional conceptualizations of domains characterizing autism: social, communication and restricted and repetitive behaviors. For further details of the development, design and initial validation of the BAPQ, please see Hurley et al. (2007).

After participants completed the self-report version of the BAPQ and provided demographic information, they were asked to complete an informant version about their spouse or partner. Participants were instructed to work independently, guess when unsure of an answer (to ensure completion of every item) and to provide responses that reference 1) their interactions with most people instead of a close relationship, 2) their behavior during most

of their adult life rather than select time periods. To reduce response bias, the BAPQ was presented to participants with an alternate title: The Personality Styles and Preferences Questionnaire.

Clinical Assessments

The MPAS-R (Piven et al., 1994) is a semi-structured interview for rating personality characteristics adapted from the Personality Assessment Schedule (PAS) (Tyrer, 1988) that was further revised to assess six personality characteristics: aloof, anxious, hypersensitive, overly-conscientious, rigid, and untactful; with aloof and rigid most reliably distinguishing PCAs from controls (Piven et al., 1997). Participants are assessed via separate self and informant interviews. Characteristics are rated as either present, absent or unknown (Losh et al., 2009) by two raters, with a third rater serving as a tiebreaker when needed. Inter-rater reliability trials for the characteristics of the MPAS-R range from 0.73 to 1.0.

The Modified Pragmatic Rating Scale (MPRS) is an abbreviated version of the PRS (Landa et al., 1992) that was developed to identify seven pragmatic language skills and four prosodic and grammatical speech errors in the PRS with more efficiency and less redundancy than the PRS. These eleven items were extracted from the PRS using logistical regression to determine those that most reliably predict pragmatic speech deficits. They best differentiated PCAs and CPs in the Iowa Family Study (Piven et al., 1997), are highly correlated to the PRS, and were validated during the Collaborative Linkage Study of Autism (Losh et al., 2008). Trained interviewers rate items 0, 1, or 2 based on a guided conversation incorporated in the MPAS-R that ensures that all rated behaviors have the opportunity to be observed. The PRS has good inter-rater reliability ($\kappa = .77$) and reliably differentiates PCAs from controls (Landa et al., 1992; Piven et al., 1997).

Data Analysis

Our first goal is to further validate the psychometric properties the BAPQ and replicate previous BAPQ findings in a large community-based sample that closely approximates the true population of PCAs and CPs. We examined the structure and content of the subscales beyond the reliability coefficients reported by Hurley et al. (2007) through the use of Exploratory Factor Analysis (EFA) on both self and informant ratings. In contrast to Principle Components Analysis, a technique used primarily for reducing variables, EFA assumes an underlying causal model and thus is an appropriate tool to empirically determine the number of latent constructs that underlie a set of items, and examine the theoretical structure of behavioral domains (DeVellis, 1991; Fabrigar et al., 1999). EFA was conducted using maximum likelihood discrepancy function with target rotation. Target rotation is a confirmatory procedure used to construct a target matrix that specifies the general pattern of expected loadings. This is similar to Confirmatory Factor Analysis (CFA) in that there is an a priori expectation for the number of factors and the particular items that will load to them, but differs from CFA in that EFA with target rotation makes loadings as close to zero as possible but does not force them, which allows determination of whether the data supports the presumed theory or target (Browne, 2001). Cronbach's alpha was also used to further confirm the internal consistency of the subscales. Next, to replicate previous BAPQ findings, an Analysis of Variance (ANOVA) was conducted to compare the total BAPQ

score and three subscale scores in mothers and fathers in the PCA group relative to the CP group. To mitigate bias involved with using only self or informant scores, these analyses used the scores averaged between the self- and informant-report versions for each parent.

Our second goal is to establish independent cutoff scores for PCA and CP mothers and fathers, and determine the sensitivity and specificity of these cutoff scores relative to gold standard direct clinical assessment measures. Whereas Hurley et al (2007) used BAP status diagnosed by direct clinical assessment (i.e., the MPASR and MPRS) to derive optimal BAPQ cutoffs, our community sample allowed us to establish normative cutoff scores for total BAPQ score and each component based upon their distribution within the general population. This approach enabled us to assess population prevalence of the BAP, which was not possible with the previous cutoff scores derived from a clinical sample known to present with BAP features (Hurley et al, 2007). We identified total and subscale BAPQ scores of 1.5 SD above the mean for men and women in our comparison sample (n= 981) separately on self and informant ratings, as well as the average between the two when both were available (73.4% of the sample). This threshold of 1.5 SD above the mean has been used in previous BAP studies to identify affected cases (Losh et al., 2008; Piven and Palmer, 1999). We assessed how these new BAPQ cutoff scores related to BAP status obtained via direct clinical assessments within our subsample of 35 PCAs who were administered the MPAS-R and MPRS. Based upon their BAPQ scores, we computed sensitivity (the proportion of parents based upon clinical assessments who were correctly classified as “present” by the BAPQ) and specificity (the proportion of parents based upon clinical assessments who were correctly classified as “absent” by the BAPQ).

Prevalence rates were calculated by categorizing mothers and fathers in each group as “present” or “absent” on the total BAP and each component based upon whether their BAPQ values exceeded the new averaged cutoff scores. This process of dichotomizing parents into present versus absent is consistent with the direct clinical assessment techniques used to derive the BAPQ (e.g., the MPASR and the MPRS). Chi Square analyses were conducted to determine whether the prevalence of BAP components (i.e., the proportion of “present” cases) differed between PCA and CP mothers and fathers.

Our third goal is to determine if BAP features co-occur within PCAs to a greater degree than within CPs. Correlational analyses between BAP components were conducted for PCA and CP mothers and fathers. Significant correlations would suggest interdependence between the subscales. Next, to assess whether co-occurrence between BAP features is more likely for PCAs than CPs, dichotomized “present” scores on each of the three subscales were summed, resulting in possible scores of 0,1,2,3 (for example, a score of “2” would indicate that a person has two distinct BAP features) and the proportion of PCAs and CPs with multiple features were compared.

Results

Participant Characteristics

BAPQ data were obtained from 1692 parents (n=711 PCAs, n=981 CPs). Demographic information for each group appears in Table 1. PCAs and CPs differed on age and education

but not on gender or number of biological children. Age and education were therefore co-varied in all ANOVAs. Within the PCA group, 12.1% reported having more than one child with ASD. The mean age of the 344 children with autism was 9.52 (SD=3.72), and 78.8% were male. The 35 PCAs (18 mothers, 17 fathers; M age=41.41, SD age=5.97) who were also assessed with the MPAS-R and the PRS did not differ significantly from the larger PCA sample in gender, age, or education, but they did consist of a larger proportion of Caucasians.

BAPQ Psychometrics

To examine the subscale structure of the BAPQ, EFA with maximum likelihood discrepancy function and varimax rotation were performed for PCAs and CPs on all 36 items of the BAPQ for both self (n=1368) and informant (n=1383) ratings separately. Completed BAPQs with missing or invalid ratings (i.e., ratings given outside of the 1 to 6 scale) were excluded from analyses. The number of components to retain was guided by: (a) the scree plot method (Cattell, 1966), (b) eigenvalues above 1.0, and (c) interpretability. Solutions between 3 and 5 components were examined, and the three-component solution was selected as the best fit because it provided the simplest structure, in which high component loadings were highest and the other loadings were lowest (see Table 2). This three-component solution accounted for 45% of the variance for informant ratings (rotation sum of squared loadings: component 1=19%; component 2=14%; component 3=12%) and 39% of the variance for self-ratings (rotation sum of squared loadings: component 1=19%; component 2=12%; component 3=8%). Although minor differences emerged for the component structure of the self and informant ratings, all items loaded with their hypothesized subscale with the exception of items 7, 10, 11 and 21 for self-ratings only, resulting in a three component structure that is identical to the aloof, pragmatic language and rigidity subscale structure described by Hurley et al (2007). To further confirm the internal consistency of the subscales, Cronbach's alpha was used to measure the extent to which an item was correlated with the remaining items from the subscale from which it was drawn. The alpha values for the three subscales were as follows: aloof personality: self=0.92, informant=0.93; rigid behavior: self=0.86, informant=0.89; and pragmatic language difficulties: self=0.80, informant=0.76. All values are within or above the acceptable range for research purposes (Nunnally, 1967). Alpha values over 0.80 are generally considered moderately-high to high.

BAPQ Performance

Total BAPQ and subscale scores did not differ by age, race, marital status, or the number of biological children for either PCAs or CPs, but did by education, with higher education in both groups associated with lower BAPQ scores both overall and on each subscale. Self-reported BAPQ total and subscale scores did not differ between those with and without informant data.

Table 3 presents BAPQ composite and subscale scores for mothers and fathers in the PCA and CP groups. Separate ANOVAs conducted for mothers and fathers confirmed significantly higher scores on the BAPQ composite and all subscales for PCAs relative to CPs: PCA fathers had higher overall scores than CP fathers ($F(1,546)=17.47, p<.001$), and higher scores on all subscales: Aloof Personality, ($F(1,546)=9.17, p<.01$); Pragmatic

Language, ($F(1,546)=9.62, p<.01$), and Rigidity, ($F(1,546)=15.89, p<.001$). PCA Mothers had higher overall scores than CP mothers ($F(1,544)=23.71, p<.001$), and higher scores on all subscales: Aloof Personality, ($F(1,544)=13.34, p<.001$); Pragmatic Language, ($F(1,544)=23.57, p<.001$), and Rigidity, ($F(1,544)=11.09, p=.001$). Within PCAs, fathers and mothers did not significantly differ on overall BAPQ score ($p=.074$), pragmatic language ($p=.58$) or rigidity ($p=.51$), but fathers had higher scores on the aloof subscale than mothers ($F(1,373)=14.27, p<.001$). Results remained significant when covarying for demographic variables that differed between PCAs and CPs (i.e., age, education, race and marital status).

Cutoff Scores and Prevalence Estimates

New cutoff scores appear in Table 4. To examine how these cutoff scores relate to gold standard clinical measures, sensitivity and specificity were calculated for the 35 PCAs who were administered the MPAS-R and MPRS (see Table 5). Specificity was high (80–100%), while sensitivity was good for the Aloof subscale and for the composite score for males, but low for the Pragmatic Language and Rigidity subscales. Compared to the lower cutoff scores reported by Hurley et al (2007), these new cutoff values maximize specificity at the cost of sensitivity. Recommendations for using each set of cutoff scores are included in the discussion.

Prevalence rates for the total BAP and each component were significantly higher for PCA mothers and fathers compared to their CP counterparts (see Table 6). Among PCAs, mothers had higher prevalence rates than fathers on Pragmatic Language ($p=.02$), but did not differ on Aloof ($p=.10$), Rigidity ($p=.52$) or total BAP ($p=.32$). Among CPs, fathers had higher prevalence rates than mothers on Pragmatic Language ($p=.05$), but did not differ on Aloof ($p=.61$), Rigidity ($p=.41$) or total BAP ($p=.70$).

Co-Occurrence of BAP Features

The three BAPQ subscale scores were all significantly inter-correlated for PCA mothers, PCA fathers, CP mothers and CP fathers (r values range from .31–.60, with all p 's<.001).

Co-occurrence was also examined by comparing the percentage of PCAs and CPs with multiple BAP features. The proportion of mothers and fathers in the PCA and CP groups with 0,1,2 or 3 BAP features can be viewed in Table 7. 18.3% of PCA mothers had more than one BAP feature compared to just 2.8% of CP mothers, a significant difference ($\chi^2(1,546)=39.78, p<.001$, odds ratio=.128, 95% CI [.062, .265]). 10.6% of PCA fathers had more than one BAP feature compared to 5.3% of CP fathers, also a significant difference ($\chi^2(1,548)=5.24, p=.022$, odds ratio=.472, 95% CI [.245, .901]). Among PCAs and CPs with only one BAP feature, no feature was significantly more common than another. Among PCAs with two BAP features, the Aloof/Pragmatic language pairing (44.4%) and the Aloof/Rigidity pairing (37.8%) occurred more commonly than the Pragmatic Language/Rigidity pairing (17.8%). Among CPs with two BAP features, the Aloof/Rigidity pairing was more common (66.7%) than the Pragmatic Language/Rigidity (20.8%) and Aloof/Pragmatic Language (12.5%) pairings.

Discussion

The current study replicates previous BAP findings using the BAPQ in a community sample of PCAs (n = 711) and CPs (n = 981) and extends them in several important ways. We first validated the psychometric properties of the BAPQ using exploratory factor analysis and replicated previous findings of higher BAPQ scores, both overall and on each subscale, for PCA mothers and fathers compared to their CP counterparts. These results supplement the original BAPQ analysis reported by Hurley et al (2007) and provide further support for findings by Ingersoll et al (2011) indicating that the three subscales of the BAPQ robustly and comprehensively capture the breadth of BAP characteristics. Collectively, these analyses indicate that the BAPQ is an efficient and reliable tool for identifying BAP features in PCAs and CPs, even in the absence of clinical evaluation.

New BAPQ cutoff scores derived from their distribution within the general population were also computed. This approach circumvented the ascertainment bias underlying cutoff scores in Hurley et al (2007), who selectively recruited parents previously known to exhibit BAP traits, which resulted in cutoff scores and sensitivity/specificity estimates tied to their sample and prevented the assessment of BAP prevalence rates.

The new cutoff values increase specificity compared to those reported by Hurley et al (2007). That is, the new cutoff values reduce false positives (i.e., cases identified as “positive” by the BAPQ who would not be identified as such by gold standard clinical assessment methods). Thus, because they maximize the likelihood that any “positive” cases identified by the BAPQ exhibit clinically observable BAP traits, the new cutoff scores are optimized for diagnostic classification of the BAP. Note that “diagnostic classification” in this context does not refer to “clinical diagnosis”, as most identified BAP cases will not exhibit impairment, but rather refers to the reliable classification of cases with BAP features. We recommend researchers use the new cutoff values when administering the BAPQ as their sole measure of the BAP (i.e., when no follow-up BAP assessments are used). For example, the new cutoff scores are optimized for 1) large-scale studies where clinical assessment is impractical, yet researchers want to ensure that those identified by the BAPQ are highly likely to have clinically identifiable BAP personality traits, and 2) studies in which researchers are intent upon excluding BAP-positive individuals from their sample (e.g., confirming that a control group excludes individuals with BAP traits).

The increased specificity of the new cutoff scores came at the cost of reduced sensitivity relative to those derived by Hurley et al (2007). That is, the new cutoffs may mistakenly exclude some true positives (i.e., cases identified as “positive” by clinical assessments that are not identified by the BAPQ). Sensitivity of the new cutoff scores was particularly low for females on total score. Thus, the Hurley et al (2007) cutoffs should be used when researchers want to maximize sensitivity, as they likely will capture more true positives compared to the new cutoff scores. They will, however, also capture more false positives. Samples identified with the cutoff scores reported by Hurley et al (2007) will be larger than those identified with the new cutoff scores, but because they also include more false positives, they result in a less “pure” BAP sample. We therefore recommend using the Hurley et al (2007) cutoffs for screening purposes, when researchers want to catch as many

true positive BAP cases as possible but can follow-up with additional BAP assessments to exclude any false positives.

The new cutoff values were used to report prevalence rates of the BAP and its components in PCAs and CPs. Higher prevalence rates were found in PCA mothers and fathers across all BAP traits, with prevalence rates on each subscale ranging between 14–23% for PCAs and between 5–9% for CPs. This prevalence gap between PCAs and CPs may represent a conservative estimate, as we deliberately did not screen CPs for a family history of developmental disability in order to more accurately represent heterogeneity found in the general population.

To address whether the BAP is better characterized as consisting of independent or interdependent features, we examined co-occurrence of BAP traits within parents. Similar to results reported for the SRS (Constantino and Todd, 2005), the three BAPQ subscales were moderately correlated for both PCAs and CPs, suggesting some degree of interdependence between individual BAP components. We also found that PCA mothers and fathers were more likely than their CP counterparts to have multiple BAP features. Although this suggests that co-occurrence of BAP features may constitute one factor underlying increased risk for autism in offspring, most of the PCAs in the present study who exhibited a BAP feature exhibited only one, a finding that is consistent with previous investigations of the BAP (Piven et al., 1997a; Losh et al., 2008). Indeed, just 18.2% of mothers and 10.6% of fathers in the PCA group presented with multiple BAP features, and only 4.8% of mothers and 1.1% of fathers exhibited all three. This proportion stands in marked contrast to their children with autism, who by definition of diagnosis exhibit co-occurrence of all features. Consistent with theories advocating the disaggregation of the autism phenotype (Happé et al., 2006), the relatively low percentage of PCAs presenting with multiple BAP features may indicate a degree of independence in BAP subcomponents that could potentially correspond to separable underlying mechanisms.

Several limitations should be considered while interpreting the results reported here. First, although our PCA group was comparable to the CP group in both gender breakdown and in their number of biological children, CPs were younger, more likely to be married and more highly educated, and had a higher proportion of Caucasian participants. These demographic differences likely emerged because of differences in the ascertainment strategy for the two groups. Importantly, however, our goal was to obtain a comparison sample that approximated the distribution of BAP traits within the local community population rather than a case control group matched to our PCA group, and thus group differences reported here should be interpreted in the context of this distinction. Further, because PCAs and CPs differed on education and age, we co-varied them in our ANOVAs to ensure group effects remained independent of these influences. Second, a small proportion of the children of PCAs had Asperger Disorder (10.5%) or PDD-NOS (9.9%), and thus the sample cannot be considered exclusive to parents of autism alone. Third, the prevalence and sensitivity/specificity data reported here are based upon averaging subject and informant versions of the BAPQ, which is not equivalent to a “best estimate” commonly derived from clinical assessment. Clinical assessments can determine a “best estimate” of BAP characteristics by evaluating the relative validity of information offered by subjects and informants, especially

in cases in which discrepant information is provided. Unfortunately, when using the BAPQ in the absence of additional clinical information, evaluating the validity of subject versus informant reports is impossible. For the current study, we chose to err on the side of conservatism by averaging subject and informant ratings, while acknowledging that this is not equivalent to a true best estimate. We did, however, conduct several post-hoc analyses to explore whether prevalence and sensitivity/specificity findings would substantively differ if calculated based upon subject or informant versions only, and differences were minimal. Nevertheless, we provide separate gender-specific subject and informant cutoff scores in addition to the average, which affords individual studies the flexibility to modify the criteria to better reflect their conceptualization of a clinical “best estimate”. For example, one might code parents as “positive” if they exceed *either* the subject or the informant cutoff for an individual BAP component, rather than relying upon the average.

Despite these limitations, the current study increases our understanding of the BAP and extends the utility of the BAPQ. We not only provide further psychometric validation that the BAPQ is a reliable instrument for measuring behavioral BAP characteristics, but we also use a large-scale community comparison sample to derive new normative cutoff values that expand the research functionality of the BAPQ. The increased specificity of the new cutoff values introduce the opportunity to use the BAPQ diagnostically, though we continue to recommend using the original cutoff values reported by Hurley et al (2007) for screening purposes due to their higher levels of sensitivity. We also specify higher prevalence rates of BAP characteristics for PCAs relative to CPs, and present evidence that BAP features co-occur more often within PCAs than CPs. In sum, this study provides further evidence BAP traits exist at elevated rates in PCAs, and confirms that the BAPQ is an efficient and reliable measure for disentangling autism-related heterogeneity in parents, a process that can facilitate efforts to uncover pathophysiological mechanisms of autism.

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Table 1

Demographic Information

	Parents of Children with Autism (N = 711)	Comparison Parents (N =981)	<i>p</i> -value
<i>Male</i>	50.5%	49.9%	ns
<i>Age (Mean, sd)</i>	40.39 (7.25)	35.95 (5.30)	<.001
<i># of Biological Children (Mean, sd)</i>	2.26 (.98)	2.17 (1.16)	ns
<i>Caucasian</i>	81.7%	86.7%	.003
<i>Marital Status</i>			<.001
Married	68.9%	87.1%	
Divorced	8.7%	1.4%	
Single/never married	4.1%	3.0%	
Widow(er)	0.7%	--	
Did Not Specify	17.6%	8.6%	
<i>Education</i>			<.001
Grade 1 - High School Graduate	37.5%	13.2%	
Some college–College Graduate	46.5%	48.4%	
Masters or Doctorate Degree	15.9%	37.9%	

Table 2

Exploratory Factor Analysis on Self and Informant Ratings

	BAIQ ITEMS	SELF				INFORMANT				
		C1 Aloof	C2 Rigid	C3 PL	C1 Aloof	C2 Rigid	C3 PL	C1 Aloof	C2 Rigid	C3 PL
1.	I like being around other people	.751	.221	-.021	.824	.233	-.032			
2.	I find it hard to get my words out smoothly	.302	.088	.419	.229	.138	.474			
3.	I am comfortable with unexpected changes in plans	.172	.534	.077	.189	.559	.103			
4.	It's hard for me to avoid getting sidetracked in conversations	.021	.036	.540	-.050	.102	.565			
5.	I would rather talk to people to get information than to socialize	.518	.124	.148	.535	.157	.098			
6.	People have to talk me into trying something new	.225	.512	.185	.223	.647	.138			
7.	I am "in-tune" with the other person during conversation	.469	.148	.421	-.384	-.077	-.589			
8.	I have to warm myself up to the idea of visiting an unfamiliar place	.335	.493	.181	.267	.580	.157			
9.	I enjoy being in social situations	.810	.260	.041	.837	.267	-.007			
10.	My voice has a flat or monotone sound to it	.354	.078	.296	.280	.156	.396			
11.	I feel disconnected or "out of sync" in conversations with others	.518	.114	.482	.446	.179	.509			
12.	People find it easy to approach me	.574	.108	.199	.545	.130	.262			
13.	I feel a strong need for sameness from day to day	.099	.680	.132	.077	.745	.085			
14.	People ask me to repeat things I've said because they don't understand	.220	.136	.534	.151	.129	.604			
15.	I am flexible about how things should be done	.232	.522	.122	.212	.540	.280			
16.	I look forward to situations where I can meet new people	.751	.292	-.056	.802	.306	-.022			
17.	I have been told that I talk too much about certain topics	-.062	.169	.486	-.149	.117	.590			
18.	When I make conversation it is just to be polite	.458	.168	.198	.399	.167	.135			
19.	I look forward to trying new things	.443	.557	.022	.380	.594	.133			
20.	I speak too loudly or softly	.054	.165	.495	-.004	.063	.509			
21.	I can tell when someone is not interested in what I am saying	.170	.122	.189	.164	.117	.505			
22.	I have a hard time dealing with changes in my routine	.105	.735	.229	.088	.774	.214			
23.	I am good at making small talk	.663	.140	.140	.687	.139	.187			
24.	I act very set in my ways	.126	.590	.160	.186	.612	.234			
25.	I feel like I am really connecting with other people	.731	.162	.232	.694	.143	.273			
26.	People get frustrated by my unwillingness to bend	.183	.372	.310	.207	.417	.346			
27.	Conversation bores me	.583	.151	.198	.574	.125	.229			

	SELF			INFORMANT		
	C1 Aloof	C2 Rigid	C3 PL	C1 Aloof	C2 Rigid	C3 PL
BAPO ITEMS						
28. I am warm and friendly in my interactions with others	.615	.132	.221	.632	.123	.299
29. I leave long pauses in conversations	.319	.100	.389	.292	.140	.422
30. I alter my daily routine by trying something different	.248	.520	.009	.253	.544	.060
31. I prefer to be alone rather than with others	.616	.159	.105	.679	.256	.011
32. I lose track of my original point when talking to people	.090	.107	.544	.002	.125	.663
33. I like to closely follow a routine while working	-.003	.527	.051	.073	.594	.003
34. I can tell when it is time to change topics in conversation	.349	.114	.370	.218	.123	.594
35. I keep doing things the way I know, even if another way might be better	.083	.385	.271	.107	.460	.348
36. I enjoy chatting with people	.845	.174	.026	.868	.139	-.004

Table 3

Averaged Subject-Informant BAPQ Scores for PCA and CP Mothers and Fathers

	Aloof Personality Mean (SD)	Pragmatic Language Mean (SD)	Rigidity Mean (SD)	Total Score Mean (SD)
<i>Mothers</i>				
PCA (n = 186)	2.68 (.85)	2.48 (.57)	3.14 (.73)	2.77 (.58)
CP (n = 360)	2.44 (.63)	2.27 (.42)	2.95 (.60)	2.55 (.43)
<i>Fathers</i>				
PCA (n = 189)	3.02 (.91)	2.51 (.55)	3.09 (.72)	2.87 (.59)
CP (n = 359)	2.79 (.83)	2.37 (.48)	2.84 (.71)	2.66 (.54)

Note: Total BAPQ score and scores on each subscale were significantly higher for PCA mothers relative to CP mothers, and PCA fathers relative to CP fathers (all $ps < .01$).

Abbreviations: PCA = Parents of Children with Autism; CP=Comparison parents.

Table 4

Cutoff Scores for BAPQ Self and Informant Ratings, and the Average of the Two, for Males and Females

	Male	Female
ALOOF	Self: 4.13	Self: 3.45
	Informant: 4.19	Informant: 3.64
	Average: 4.03	Average: 3.39
PRAGMATIC LANGUAGE	Self: 3.23	Self: 2.94
	Informant: 3.29	Informant: 3.19
	Average: 3.09	Average: 2.90
RIGIDITY	Self: 3.91	Self: 3.70
	Informant: 4.20	Informant: 4.30
	Average: 3.90	Average: 3.85
TOTAL SCORE	Self: 3.55	Self: 3.17
	Informant: 3.63	Informant: 3.46
	Average: 3.47	Average: 3.19

Note: Original "best estimate" cutoff scores reported by Hurley et al. (2007) were as follows: **Male:** Aloof 3.25, Pragmatic Language: 2.95, Rigid: 3.65; Total Score: 3.35 **Female:** Aloof 3.00, Pragmatic Language: 2.70, Rigid: 3.25; Total Score: 3.25.

Table 5

Sensitivity and Specificity of Amended Cutoff Scores by Gender

	Male (n=17)	Female (n=18)
ALOOF		
Sensitivity	60%	67%
Specificity	100%	93%
PRAGMATIC LANGUAGE		
Sensitivity	33%	33%
Specificity	82%	93%
RIGIDITY		
Sensitivity	38%	20%
Specificity	100%	92%
TOTAL SCORE		
Sensitivity	83%	33%
Specificity	100%	100%

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Table 6

Prevalence Rates of BAP and BAP Components in Parents of Children with Autism (PCA) and Comparison Parents (CP)

	PCA	CP	Odds Ratio (95% CI)	p-value
<i>Aloof</i>				
Mothers	22.6%	9.4%	2.8 (1.7–4.6)	<.001
Fathers	15.9%	8.4%	2.1 (1.2–3.6)	.007
<i>Pragmatic Language</i>				
Mothers	23.1%	4.7%	6.1 (3.3–11.0)	<.001
Fathers	13.8%	8.4%	1.8 (1.0–3.1)	.047
<i>Rigidity</i>				
Mothers	16.7%	7.2%	2.6 (1.5–4.5)	<.001
Fathers	14.3%	8.9%	1.7 (.99–2.9)	.05
<i>Total Score</i>				
Mothers	23.2%	8.1%	3.4 (2.1–5.7)	<.001
Fathers	19.0%	8.9%	2.4 (1.4–4.0)	.001

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Table 7

Proportion of PCA and CP Mothers and Fathers with 0,1,2,3 BAP Features

# of BAP Features	<u>PCA</u>		<u>CP</u>	
	Mothers	Fathers	Mothers	Fathers
0	60.8%	67.8%	81.9%	80.5%
1	21.0%	21.7%	15.3%	14.2%
2	13.4%	9.5%	2.2%	4.5%
3	4.8%	1.1%	0.6%	0.8%

Abbreviations: PCA = Parents of Children with Autism; CP=Comparison parents.

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