

## The broad autism phenotype in parents of individuals with autism: a systematic review of the literature

O fenótipo ampliado do autismo em pais de indivíduos autistas: uma revisão sistemática da literatura

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### Abstract

The broad autism phenotype (BAP) is a milder manifestation of the defining symptoms of the syndrome in individuals without autism. This study conducted a systematic review of studies about behavioral characteristics of interpersonal relationships, communication and rigidity, as well as about three cognitive models, Theory of Mind, central coherence and executive function, in parents of individuals with autism. The indexed databases were LILACS, IBECs, Web of Science, and MEDLINE, and the studies retrieved were published between 1991 and March 2012. Parents of individuals with autism have more difficulties in interpersonal relationships and in pragmatic language use and have more rigidity traits. The inclusions of the cognitive theories in the group of BAP characteristics were inconclusive.

**Keywords:** Autism-spectrum disorder, phenotype, endophenotypes, parents.

### Resumo

O fenótipo ampliado do autismo (FAA) é a manifestação atenuada de características qualitativamente similares às que definem a síndrome, em indivíduos não portadores do transtorno. O objetivo deste artigo é realizar uma revisão sistemática de estudos que abordam as características comportamentais relacionadas a interação social, comunicação e rigidez, além dos modelos cognitivos Teoria da Mente (Theory of Mind, ToM), coerência central e funções executivas, em pais de indivíduos autistas. As bases de dados consultadas foram: LILACS, IBECs, Web of Science e MEDLINE; foram selecionados estudos publicados entre 1991 e março de 2012. Os resultados apontaram que os pais de indivíduos autistas possuem déficits na interação social, na linguagem pragmática e traços de rigidez. A inclusão dos modelos cognitivos do autismo no grupo de características que compõem o FAA permanece inconclusiva.

**Descritores:** Transtorno autístico, fenótipo, endofenótipos, pais.

### Introduction

Childhood autism is a behavioral syndrome characterized by a triad of symptoms: deficits in social interactions, deficits in verbal and non-verbal communication and inflexible, restricted and stereotyped behaviors and interests.<sup>1</sup> The intensity of these symptoms may vary from mild, in which individual have a high intellectual potential, to severe, in which there is mental retardation and lack of speech.<sup>2</sup>

For many years autism was believed to be a psychological disorder explained by relationship models. However, as genetic studies advanced, the understanding that children have autism due to their parents' affective distance is no longer accepted.<sup>3</sup> We currently know that it is a complex, highly hereditary neurobiological syndrome.

Studies have indicated that relatives of people with autism also have autism traits at a lesser degree than that found in high-functioning autism, but at an

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intensity that is very mild and does not characterize the disorder.<sup>4,5</sup> These mild expressions in individuals without the syndrome have been named the broad autism phenotype (BAP).<sup>6</sup> The prevalence of the BAP in families ranges from 12 to 30%,<sup>6-9</sup> and male parents have more BAP characteristics than mothers.<sup>7,10,11</sup>

The importance of studying phenotypes and endophenotypes of psychiatric syndromes has been highlighted in the literature. These characteristics may be useful in the identification of the genes that confer vulnerability to these disorders.<sup>12</sup> Additionally, studies about the BAP of parents may contribute to the definition of diagnostic criteria, may refine the understanding of some symptoms when subdomains are detected in each factor, and may contribute to the definition of treatments. Moreover, parents may also benefit from some interventions.

Although the term is BAP, this concept also includes the possible endophenotype of the syndrome. Phenotypes are the observable characteristics or behaviors that result from gene expression, environmental influences and the interaction between both,<sup>12</sup> and which correspond to the triad of core symptoms in the case of autism. Endophenotypes, in contrast, are manifestations of the genotype without the influence of the environment. They are subclinical inherited markers of the disease that may be neurophysiological, biochemical, endocrinological, neuroanatomic or cognitive. They involve fewer genes than those in the disorder itself, and are expressed in the person that has the gene even when the syndrome is not manifest. They are hereditary and more often found in the parents of patients with the disorder than in the general population.<sup>13</sup>

In autism, there is a cognitive abnormality (endophenotype) associated with behaviors for each behavioral symptom (phenotype). The models of cognitive deficits, possibly responsible for dysfunctional behaviors, are: Theory of Mind (ToM) deficit, weak central coherence and impaired executive functions.<sup>14</sup> These three theories are complementary, and each explains some core features of the disorder better than others.<sup>15</sup>

This study reviewed studies that investigated the behavioral characteristics of the triad of core symptoms, as well as the three main cognitive theories (ToM, central coherence and executive functions deficits) in the parents of children with autism to identify which features belong to the BAP in this group of individuals.

## Method

This review was conducted in the second half of 2012 using the following electronic databases: LILACS and IBECs – databases of Latin American and Caribbean health sciences literature; Web of Science and MEDLINE – databases of international literature. Studies published between 1991 and March 2012 were analyzed. The keywords used were all related to phenotype, endophenotype, autism, parents and family. The total number of studies retrieved in the search, as well as the keywords used in each database, is shown in table 1.

Studies were selected according to the following inclusion criteria: a) published in Portuguese, English or Spanish; b) object of study was the cognitive and behavioral characteristics of autism in parents of individuals with autism; c) original studies. Exclusion criteria were: a) studies about genetics that did not investigate autism phenotype or endophenotype; b) studies that included only siblings; c) studies that evaluated the BAP in the general population; d) studies that investigated endophenotype according to biological or neurofunctional dimensions; e) studies that evaluated only parents of individuals with pervasive developmental disorders other than autism; f) studies that evaluated the BAP using only personality traits of parents; g) studies that evaluated the BAP only according to presence of other psychiatric disorders in parents; h) opinion papers; i) studies that used non-standardized instruments; j) studies that did not include a control group.

**Table 1** - Keywords used in electronic databases to retrieve studies published between 1991 and March 2012

Database	Keywords used	Studies retrieved
LILACS	(Fenótip\$ OR Endofenotip\$ OR Fenotip\$ OR Endofenotip\$) AND ("Transtorno Autístico" OR Autismo\$ OR Autistic\$) OR ("Núcleo Familiar" OR Pais OR Padres OR Parents OR Famil\$).	04
IBECs		03
Web of Science	(Phenotype OR Endophenotype ) AND ("Autistic Disorder" OR Autism OR Autistic) AND (Parent OR father OR mother).	178
MEDLINE	("Phenotype" OR "Endophenotypes" OR Phenotype OR Endophenotype) wAND ("Autistic Disorder/etiology" OR "Autistic Disorder/psychology" OR "Autistic Disorder" OR Autism OR Autistic) AND ("Parents" OR Parent OR father OR mother).	232

According to the inclusion and exclusion criteria, the abstracts of all eligible studies were read by two independent examiners (LC and WC). Divergent opinions were discussed by the two until a consensus was reached. After the studies had been selected, their complete texts were read. Data about all studies were organized in tables for the analysis of objectives and hypothesis, methods, including the description of the sample and the instruments used, and, finally, results and conclusions. Therefore, the main ideas expressed in each study, as well as convergent and divergent findings, were analyzed qualitatively and descriptively, and data were interpreted and analyzed. Secondary references found in the studies retrieved from the databases were also used.

## Results

A total of 417 studies were retrieved: 368 were not included due to exclusion criteria or were not about the topic under study. Most of the studies excluded were about genetics (164 studies), syndromes that had some association with autism, such as the Rett syndrome (71 studies), or phenotype expression in individuals with autism (54 studies). The other 79 studies met other exclusion criteria, and 19 were repeated studies.

After reading the 27 studies originally selected, 8 other studies were found in their references. A total of 35 studies were included. Some also evaluated other family members, but their results were only about behavioral characteristics and cognitive models of parents of individuals with autism. Information about other relatives, as well as information about other constructs, was not included in the tables or text below. Two studies<sup>10,16</sup> are repeated in the two tables because they evaluated both behavioral and cognitive characteristics. Therefore, although only 35 studies were selected, the two tables show a total of 37 studies: 19 about behavioral characteristics and 18 about cognitive models.

## Behavioral characteristics in the triad of core symptoms

We reviewed what instruments were used to evaluate behavioral characteristics. Those that evaluated the BAP globally, that is, that included the three core domains of the syndrome, are shown in Table 2. In the 35 studies reviewed, only four instruments were used to evaluate the triad of core symptoms in parents: the Family History Interview (FHI),<sup>17</sup> the Autism Spectrum Quotient (AQ),<sup>18</sup> the Broader Phenotype Autism Symptom Scale (BPASS),<sup>19</sup> and the Broad Autism Phenotype Questionnaire (BAPQ).<sup>8</sup> FHI was the first instrument used for this purpose and was the most frequently used in studies up to 2000. Today, the questionnaire most often used in the AQ. Other instruments also evaluate isolated behavioral characteristics, and of the three domains, language is the one evaluated in most instruments.

The selected studies about behavioral characteristics of parents of individuals with autism are listed in Table 3.

## Changes in social interaction

Seven articles evaluated social relationships of parents of individuals with autism.<sup>7,8,11,20,24,26,28</sup> None of these studies evaluated only this domain, and social interaction was evaluated together with other behavioral characteristics. Moreover, social difficulties were approached according to different dimensions, and each study had a specific observation focus of the social functioning of parents of individuals with autism.

Some studies evaluated the interest of parents in social interactions,<sup>8</sup> and others evaluated social skills.<sup>7</sup> Results demonstrated that some individuals prefer not to interact and are aloof,<sup>8</sup> whereas others do want to interact, but are not able to do it satisfactorily.

Losh et al.<sup>28</sup> found that 23% of the parents of multiple-incidence autism families do not have friends, in contrast with 11% of parents of single-incidence autism families

**Table 2** - Description of the instruments used to evaluate the BAP globally among parents of individuals with autism

Scale	Psychometric features	Studies that used instrument
FHI <sup>17</sup>	Categorical assessment instrument used with children and adults. Semistructured interview that assesses deficits in the three areas of autism.	Pickles et al. (2000) <sup>9</sup> ; Piven et al. (1997) <sup>20</sup> ; Fombonne et al. (1997) <sup>21</sup> ; Folstein et al. (1999) <sup>22</sup>
AQ <sup>18</sup>	Self-report dimensional scale; 50 items divided into five domains: social skills, communication skills, attention to detail, attention switching and imagination It is used with children and adults.	Bishop et al. (2004) <sup>7</sup> ; Scheeren & Stauder (2008) <sup>10</sup> ; Whitehouse et al. (2007) <sup>23</sup> ; Wheelwright et al. (2010) <sup>11</sup>
BPASS <sup>19</sup>	Dimensional scale. It is divided into four domains: social motivation, flexibility and variety of interests, social expressiveness and conversational skills. It may be used to evaluate adults and children.	Bernier et al. (2012) <sup>24</sup>
BAPQ <sup>8</sup>	Likert-like dimensional scale with 36 items. It has three domains: social interests, pragmatic language, and rigid personality. It is used with adults.	Hurley et al. (2007) <sup>8</sup>

AQ = Autism Spectrum Quotient; BAPQ = Broad Autism Phenotype Questionnaire; BPASS = Broader Autism Phenotype Symptom Scale; BAP = broad autism phenotype; FHI = Family History Interview.

**Table 3** - Studies about the behavioral characteristics of parents of individuals with autism

Reference	Sample of parents	Behavioral characteristics	Instruments used	Results
Landa et al. (1992) <sup>25</sup>	43 parents of children with autism spectrum disorder 21 parents of children with Down syndrome or adults without children with autism	Disinhibited social communication Inadequate expressions Odd verbal interactions	PRS	42% of the parents of individuals with autism had some pragmatic language deficit, compared with 2% of parents in the control group
Fombonne et al. (1997) <sup>21</sup>	160 parents of individuals with autism 42 parents of individuals with Down syndrome	Reading and writing skills	FHS Form A of GORT Short version of ERT NART The Schonell Graded SST	No significant differences were found in reading and spelling
Piven et al. (1997) <sup>6</sup>	48 parents of individuals with autism (multiple-incidence autism families) 60 parents of individuals with Down syndrome	Pragmatic language Social interaction Personality traits	MPAS-R PRS FS	Parents of children with autism had more characteristics of rigidity, aloofness, hypersensitivity to criticism and anxiousness They had more speech and pragmatic language deficits Moreover, they had more limited friendships
Piven et al. (1997) <sup>20</sup>	48 parents of children with autism 60 parents of children with Down syndrome	Social interactions Communication Rigid and stereotyped interests and behaviors	FHI	Parents of children with autism had greater social and communication deficits and more rigid and stereotyped interests and behaviors
Baron-Cohen et al. (1998) <sup>26</sup>	275 math students 266 engineering students 100 physics students 652 literature students	Work interests	Questionnaire about incidence of 6 conditions: autism, anorexia, schizophrenia, language delays, manic-depression and Down's syndrome	Autism was significantly more frequent in families of physics, engineering and math students than in control group of literature students
Folstein et al. (1999) <sup>22</sup>	166 parents of individuals with autism 75 parents of children with Down syndrome	Pragmatic language Social interaction	FHI PRS FI PAS	Parents of individuals with autism had early language-related difficulties and more difficulties in reading, writing and pragmatic language use However, they did not have greater difficulties in evaluation of social-related component of BAP
Bishop et al. (2004) <sup>7</sup>	69 parents of people with autism-spectrum disorder 59 parents of children without autism	Social skills Communication Imagination Attention to detail Attention switching	AQ	Parents in clinical group had more autism characteristics in communication and social skills than parents in control group There were no differences between groups in other factors
Bishop et al. (2004) <sup>32</sup>	145 parents of children with autism spectrum disorder 96 parents of children with typical development	Phonological processing Social skills Communication Imagination Attention to detail Attention switching	AQ Nonword repetition Nonsense passage reading	Parents with more BAP characteristics according to AQ reported history of more language and literacy problems However, they did not have poorer scores in phonological processing
Di Michele et al. (2007) <sup>16</sup>	23 parents of children with autism 35 parents in control group (12 of children with Down syndrome and 23 of healthy children)	Pragmatic language	Experimental tasks of Gricean conversational maxims	Parents of people with autism had more pragmatic language errors than controls when evaluating taped conversations: difficulty in identifying relevant, redundant and non-informative information
Ruser et al. (2007) <sup>27</sup>	47 parents of children with autism 47 parents of children with specific language impairment 21 parents of children with Down syndrome	Emotional expressiveness and awareness of other Communicative performance Over-talkativeness Pragmatic language	Modified version of the FHI of Developmental Disorders of Cognition and Social Functioning PRS-M	Parents of people with autism and specific language impairment did not differ from each other according to PRS-M Both groups had a significantly poorer performance in communicative skills than parents of children with Down syndrome

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Whitehouse et al. (2007) <sup>23</sup>	30 parents of children with autism 30 parents of children with specific language impairment 30 parents of typically-developing children	Reading and spelling Social skills Communication Imagination Attention to detail Attention switching	TROG-2 Test of Word Reading Efficiency Dictation task AQ	Parents of children with autism had similar scores to those of typically-developing children The two groups had scores higher than those of parents of children with specific language impairments However, parents of children with autism had communication deficits according to AQ
Losh et al. (2008) <sup>28</sup>	48 parents of individuals with autism (multiple-incidence autism families) 78 parents of individuals with autism (single-incidence autism families) 60 parents of individuals with Down syndrome	Aloofness and social behavior Rigid personality Pragmatic language Personality traits	MPAS-R FI The NEO- Personality Inventory PRS	BAP expression was gradual in the groups of families: parents of multiple-incidence autism families had more BAP characteristics than parents of single-incidence autism families, who, in turn, had more BAP characteristics than parents in the control group
Scheeren & Stauder (2008) <sup>10</sup>	25 parents of children with high-functioning autism 25 parents of typically-developing children	Social skills Attention switching Attention to detail Communication Imagination	AQ	Results did not reveal differences between groups
Schmidt et al. (2008) <sup>29</sup>	22 parents of children with autism 22 individuals in control group	Phonological processing	PPVT-III EVT DK-EFS TOLC-E Nonword repetition subtest of CTOPP WAIS subtests (vocabulary, similarities, block design)	Parents of people with autism had a worse performance in repetition of nonword syllables (nonword repetition test) In other phonological tests, there were no differences between groups
Lindgren et al. (2009) <sup>30</sup>	62 parents of children with autism and language impairment 39 parents of children without autism and language impairment 70 parents of children with specific language impairment	Semantics Morphology Syntax Memory for language Lexical comprehension Reading abilities	Comprehensive Test of Phonological Processing Clinical Evaluation of Language Fundamentals - Third Edition The Peabody Picture Vocabulary Test - Third edition The Woodcock-Johnson Psycho Educational Battery - Revised	Parents of people with autism and language impairment had a better performance in language tests than parents of children with specific language impairment
Wheelwright et al. (2010) <sup>11</sup>	2,000 parents of children with autism 1,007 parents of typically-developing children	Social skills Communication Imagination Attention to detail Attention switching	AQ	Parents of individuals with autism had higher AQ scores in 4 of 5 factors than parents of typically-developing children, which indicated that the first had more BAP characteristics Fathers had more characteristics than mothers
Whitehouse et al. (2010) <sup>31</sup>	238 parents of children with autism 187 typical individuals with no history of autism in the family	Language (speech, syntax and semantics) Pragmatic skills (active communication) Social engagement (passive communication style)	CC-A	There were no differences between groups in results of language subscale However, parents of individuals with autism had higher scores in the other two subscales, particularly in social engagement subscale
Bernier et al. (2012) <sup>24</sup>	39 parents of multiple-incidence autism families 22 parents of single-incidence autism families 20 parents of children with developmental delay without ASD 20 parents of typically-developing children	Social motivation Social expressivity Conversational skills Flexibility/range of interests (restricted interests)	BPASS	Parents of multiple-incidence autism families had significantly more autism phenotype characteristics than parents in other groups Moreover, parents of single-incidence autism families did not differ from those of typically-developing children

AQ = Autism Spectrum Quotient; BAP = broad autism phenotype; BPASS = Broader Autism Phenotype Symptom Scale; CC-A= Communication Checklist – Adult; CTOPP = Test of Phonological Processing; DK-EFS = The Verbal Fluency Subtest from the Delis Kaplan Executive Function System; ERT = Edinburgh Reading Tests; EVT = The Expressive Vocabulary Test; FHS = Family History Schedule; FI = Friendship Interview; FS = Friendship Scale; GORT = Gray Oral Reading Test; MPAS-R = Modified Personality Assessment Schedule Revised; NART = National Adult Reading Test; PAS = Personality Assessment Schedule; PPVT-III = Peabody Picture Vocabulary Test; PRS = Pragmatic Rating Scale; SST = Spelling Test-B; TOLC-E = The Figurative Language Subtest from the Test of Language Competence – Expanded Edition; TROG-2 = Test for Reception of Grammar-2; WAIS = Wechsler Adult Intelligence Scale.

and 3% of parents of children with Down syndrome. High rates of social difficulty in multiple-incidence autism families were also found in the studies conducted by Bernier et al.<sup>24</sup> and Piven et al.<sup>6,20</sup> Moreover, parents of children with autism had limited friendships<sup>6</sup> and often worked in fields that require little social understanding, such as math, engineering and physics.<sup>26</sup>

### Language and communication impairments

Sixteen studies evaluated the communicative characteristics of parents of individuals with autism.<sup>6-8,11,16,20-25,27,29-32</sup> Seven studies evaluated this domain together with others (social domain and rigidity),<sup>6-8,11,20,22,24</sup> and nine were specially designed to evaluate communicative skills.<sup>16,21,23,25,27,29-32</sup> Seven evaluated structural language abnormalities<sup>21,22,27,29-32</sup> and nine, the social use of language (pragmatic language)<sup>6-8,11,16,22,24,25,31</sup>; some evaluated these two communicative characteristics. Although structural language abnormalities are not classified as behavioral characteristics, they were included in this section because they lead to communicative deficits.

In a pioneering study, Landa et al.<sup>25</sup> evaluated communication of parents and found three groups of pragmatic abnormalities measured by means of the Pragmatic Rating Scale (PRS): disinhibited social communication, awkward/inadequate expressions and odd verbal interactions. Since that study, pragmatic language skills have been broadly investigated, which corroborates the idea that parents of children with autism have social communication difficulties.<sup>6-8,11,16,24,27</sup>

Abnormalities associated with structural language have also been evaluated in groups of parents. Whitehouse et al.<sup>23</sup> evaluated language deficits and found that parents of children with autism and parents of typically-developing children had similar performances, and that both groups had a better performance than that of parents of children with specific language impairments (SLI). Therefore, the difficulties associated with structural language are not found in the BAP of parents. This finding has been confirmed by the studies conducted by Bishop et al.,<sup>32</sup> Whitehouse et al.,<sup>31</sup> and Lindgren et al.<sup>30</sup> However, other studies have found phonological processing deficits among parents of individuals with autism, which supports the hypothesis that these deficits are part of the BAP.<sup>29</sup>

The analysis of reading and writing acquisition revealed that the difficulties of some parents of children with autism seem to be associated with an abnormal development of language rather than with autism.<sup>21</sup> In the study conducted by Folstein et al. in 1999,<sup>22</sup> only the parents with a history of cognitive deficits associated with language in childhood had a poorer performance

in reading and writing tasks than parents of individuals with Down syndrome. A history of language delays and deficits in childhood in the group of individuals with autism was also reported by Piven et al.<sup>20</sup>

Pragmatic language deficits may be seen as a reliable characteristic of the BAP. However, other language deficits, such as delays and difficulties in reading and writing acquisition and difficulties in structural language use, although present in some of the parents of children with autism, have not been confirmed to be part of the BAP,<sup>23,30,31</sup> and further studies should be conducted to investigate this topic.

### Restricted interests and repetitive and stereotyped behavior

Of the three core symptoms of autism, this is the one with the smallest number of studies that evaluated parents. Only five studies were retrieved.<sup>6,8,20,24,28</sup> Moreover, this domain has only one category, divided into different characteristics: intense restricted interests; adherence to routines and rituals; stereotyped and repetitive motor mannerisms; and persistent preoccupation with parts of objects.<sup>1</sup> Although it is a single category, the behaviors described here are substantially heterogeneous, but the main studies using questionnaires to detect BAP characteristics in parents have not defined this subdomain classification.

In the studies conducted by Piven et al.,<sup>6</sup> Hurley et al.,<sup>8</sup> and Losh et al.,<sup>28</sup> rigidity describes resistance to changes and a strong attachment to routines. Bernier et al.<sup>24</sup> evaluated behavioral flexibility and range of interests. Piven et al.,<sup>20</sup> in turn, evaluated restricted interests and rigidity, or perfectionism. No study included stereotyped and repetitive motor mannerisms in the evaluation of parents. Questionnaires evaluated the same domain in different ways.

The hypothesis that parents also have a rigid personality, with little interest in what is new, as well as with difficulties in dealing with routine changes, has been confirmed in some studies.<sup>6,8</sup> In the study conducted by Bernier et al.<sup>24</sup> only parents of multiple-incidence autism families had characteristics of rigidity. Losh et al.,<sup>28</sup> however, found that parents of multiple-incidence autism families had more characteristics of rigidity than parents that had only one child with autism, who, in turn, had more autism traits than parents in the control group.

### Cognitive models

Eighteen studies evaluated central coherence, executive functions and ToM in groups of parents of individuals with autism, and most included more than one cognitive model. Table 4 describes these studies.

**Table 4** - Studies about cognitive models: executive functions, central coherence and ToM among parents of children with autism

Reference	Sample of parents	Cognitive models	Instruments used	Results
Szatmari et al. (1993) <sup>33</sup>	96 parents of individuals with autism 54 parents of children with Down syndrome or low birth weight	Executive functions Central coherence	Subtests of the Wechsler Adult Intelligence Scale (digits, blocks, vocabulary and comprehension)	There were no differences between groups
Piven & Palmer (1997) <sup>34</sup>	48 parents of multiple-incidence autism families 60 parents of individuals with Down syndrome	Executive functions (planning)	Tower of Hanoi	Parents of individuals with autism had more difficulties in planning tasks than parents in control group
Hughes et al. (1997) <sup>35</sup>	40 parents of children with autism 40 parents of learning disabled children 36 adults of unaffected families	Executive functions (attentional-shifting skills, planning, working memory)	CANTAB Wisconsin Card Sorting Task Tower of London Spatial working memory task Short-term spatial working memory	Parents of children with autism had good memory skills, but impaired planning and attentional-shifting skills
Happe et al. (2001) <sup>36</sup>	43 parents of individuals with autism 30 parents of individuals with dyslexia 20 parents of typically-developing children	Central coherence	WRAT reading subtest EFT Sentence completion task Unsegmented block design task Visual illusions task	Parents of children with autism, particularly fathers, had weak central coherence This was seen in semantic tasks (sentence completion), perceptual judgment (illusions), visuospatial construction and problem solving
Bölte & Poustka (2003) <sup>37</sup>	54 parents of individuals with autism (single incidence) 28 parents of multiple-incidence autism families 31 parents of individuals with schizophrenia (single incidence) 4 parents of multiple-incidence schizophrenia families 22 unaffected individuals	Facial expression (ToM)	Test to recognize facial affect	Results did not reveal significant differences between groups
Bölte & Poustka (2006) <sup>38</sup>	62 parents of children with autism 36 parents of children with schizophrenia 30 parents of children with mental retardation	Central coherence Executive functions	Embedded figures test Unsegmented block design task Wisconsin Card Sorting Task Tower of Hanoi Trail-making test	Parents of children with autism had a significantly faster performance in EFT than parents in other two groups There were no other differences between groups
Jonge et al. (2006) <sup>39</sup>	51 parents of children with autism 54 parents of typically-developing children	Central coherence	EFT	Parents of children with autism were not different from matched controls in locating shapes However, they made fewer incorrect attempts before finding the right shape
Palermo et al. (2006) <sup>40</sup>	40 parents of children with classical autism 40 parents of typically-developing children	Facial expression (ToM)	HVOT Facial expression decoding test Experimental emotional labeling tasks in response to schematic human facial patterns	Parents of children with autism performed worse than control parents in reasoning facial patterns, particularly sadness and disgust In the other two tasks, there were no differences between groups
Wong et al. (2006) <sup>41</sup>	145 parents of children with autism spectrum disorder 96 parents of typically-developing children	Executive functions (planning, set shifting, inhibition and generativity)	WAIS subtests (vocabulary, similarities, picture completion and object assembly) Tower of London IDED set-shifting task RIL task Pattern meanings	Parents of children with autism performed worse than controls in verbal and non-verbal generativity tests, but had no difficulties with planning or inhibition

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Delorme et al. (2007) <sup>42</sup>	39 parents of individuals with autism 53 parents of individuals with COD 47 healthy individuals	Executive functions (planning and working memory)	Family interview for genetic studies Tower of London Trail-making test Verbal fluency test Non-verbal fluency test	In the Tower of London test, parents of individuals with autism and with COD had lower scores and were slower than individuals in control group No difference was found between groups in the other four tasks
Di Michele et al. (2007) <sup>16</sup>	46 parents of individuals with autism 23 parents of typically-developing children 12 parents of individuals with Down syndrome	ToM	False belief task (smarties task; Sally-Anne task)	Parents of individuals with autism performed worse than controls in false belief tasks
Losh & Piven (2007) <sup>43</sup>	48 parents of children with autism 16 parents of typically-developing children 6 parents of children with Down syndrome	ToM	Eyes test MPAS-R	There were no differences between groups However, parents defined as aloof had social-cognitive deficits
Scheeren & Stauder (2008) <sup>10</sup>	25 parents of children with high-functioning autism 25 parents of typically-developing children	Central coherence	WAIS block design subtest	Groups had similar scores in the block design test
Gokcen et al. (2009) <sup>44</sup>	76 parents of children with autism 41 parents of typically-developing children	Executive functions ToM	WAIS subscales (comprehension, block design, similarities and figure completion) Benton Face Recognition Test – Short Form Stroop test ACT UOT Hinting task	Parents of children with autism had verbal working memory deficits and difficulty in reasoning about other's emotions They also performed poorly in mental state reasoning task
Jonge et al. (2009) <sup>45</sup>	25 parents of children with autism 30 parents of children with Down syndrome	Central coherence	WAIS block design subtest	Groups had similar scores in the block design test
Losh et al. (2009) <sup>46</sup>	41 typical individuals with no history of autism in the family 83 parents of individuals with autism 32 typical parents with no history of autism in the family 24 parents that participated without their children 15 single fathers and mothers of a child with autism	ToM Executive functions Central coherence	Eyes test Morphed faces task Trustworthiness of faces Movie stills task Point light task Tower of Hanoi EFT Sentence completion task Trail-making test Block design test	Parents with BAP differed from controls in social cognition measures There were no differences between groups in measures of central coherence or executive functions
Wallace et al. (2010) <sup>47</sup>	22 parents of children with autism spectrum disorder 26 typically-developing adults	Facial expression (ToM)	Computer-assisted face processing tasks (unfamiliar face and object discrimination, facial expression recognition, judgment of eye-gaze direction)	Parents of individuals with autism were less capable of discriminating subtle differences between faces and had more difficulties in identifying expressions of fear and disgust than typically-developing individuals
Neves et al. (2011) <sup>48</sup>	40 parents of individuals with autism 41 healthy controls	Facial expression (ToM)	ER-40 PFMT	Parents of children with autism performed worse than controls in facial emotion recognition tasks, particularly anger, fear and happiness

ACT = auditory consonant trigrams; BAP = Broad Autism Phenotype; CANTAB = Cambridge Neuropsychological Test Automated Battery; COD = compulsive obsessive disorder; EFT = Embedded Figures Test; ER-40 = Penn Emotion Recognition Test; HVOT = Hooper Visual Organization Test; MPAS-R = Modified Personality Assessment Schedule Revised; PFMT = Penn Facial Memory Test; RIL = response inhibition and load; ToM = Theory of Mind; UOT = Unexpected Outcomes Test; WAIS = Weschler Adult Intelligence Scale; WRAT = Wide Range Achievement Test.

## Central coherence

The theory that individuals with autism have a weak central coherence, developed by Frith in 1989,<sup>49</sup> suggests that most people that do not have autism-spectrum disorder have a natural and spontaneous tendency to integrate details into a global entity and to process information received according to its context, observing and recalling more of the general picture than

of the details. In contrast, people with autism-spectrum disorder seem to have a deficit in local processing and often miss the general meaning. However, different from other cognitive theories, the theory of a weak central coherence does not define deficits; rather, it claims that strong and weak central coherence have advantages and disadvantages and are merely different cognitive styles.<sup>50</sup> Initially, this theory attempted to explain all the core symptoms of autism. However, after several studies

that supported, or not, this hypothesis, it is currently seen as one of the cognitive styles found in the disorder, which contributes, therefore, to the view that autism includes multiple deficits.<sup>15</sup>

In addition to visual and spatial skills, the central coherence cognitive models also include verbal skills. Individuals with a weak central coherence have more difficulties in understanding words by means of their context, which, therefore, contributes to a deficit in communication.<sup>51</sup> Some studies found abnormalities in central coherence tests only when evaluating verbal skills and not, therefore, in tests of visual skills.<sup>46</sup>

The theory of a weak central coherence, although logical and convincing, remains inconsistent in the case of parents of individuals with autism. Six studies evaluated this cognitive model,<sup>10,36,38,39,45,46</sup> but only two evaluated verbal characteristics.<sup>36,46</sup> Results have been inconclusive, as authors constantly apply the same instruments to different samples and find different results.<sup>36,45</sup> Bölte & Poustka<sup>38</sup> evaluated central coherence skills of parents of children with autism and found a better performance in the Embedded Figures test, but not in the block design test. They found that the improved skill of people with autism to identify simple forms embedded in complex figures seems to be the main characteristic of local processing. Therefore, they suggest that this skill may be a characteristic of the BAP. Jonge et al.<sup>39</sup> found that fathers performed better than control parents only in number of attempts to find the correct answer in the Embedded Figures test. However, the same authors conducted a study to evaluate central coherence of parents using the block design test and did not find differences between groups.<sup>45</sup>

Happé et al.<sup>36</sup> evaluated parents of children with autism using a battery of tasks and found that fathers had a weak central coherence according to visual and verbal tasks. However, in the same study, mothers had abnormal results only in verbal tasks. Their findings of unequal results for different sexes are inconclusive, according to the authors, because of the small sample size.

Other studies did not find any differences between parents of individuals with autism and other groups of parents, and their authors suggest that parents of individuals with autism have deficits in local processing, or a weak central coherence.<sup>10,45</sup> However, their studies included only tests that evaluated visual skills. Losh et al.<sup>46</sup> also did not find any differences in visual skills between groups of parents, but difference were detected in verbal skills.

### **Executive functions**

Executive functions are a set of integrated skills

that allow individuals to direct their behavior to goals, to monitor efficiency and adequacy of these behaviors, to drop inefficient strategies and, therefore, to solve immediate, medium-term and long-term problems.<sup>52</sup> Individuals with autism have deficits in these cognitive skills, which may be responsible for a high level of rigidity and perseveration,<sup>53</sup> as well as for the performance of rituals and difficulties in dealing with changes in their routines. According to the seven studies retrieved,<sup>33-35,41,42,44,46</sup> parents of individuals with autism also have deficits in executive functions, particularly those associated with planning<sup>34,35,42</sup> (however, see Wong et al.<sup>41</sup> and Losh et al.<sup>46</sup>); difficulties in operational memory<sup>44</sup> (however, see Wong et al.<sup>41</sup>); cognitive flexibility<sup>35</sup> (however, see Bishop et al.,<sup>33</sup> Delorme et al.,<sup>42</sup> Losh et al.<sup>46</sup>); and verbal and non-verbal fluency<sup>41</sup> (however, see Delorme et al.<sup>42</sup>).

Although several studies confirm the presence of deficits in executive functions of parents of individuals with autism and their children, these deficits are not associated only with this disorder,<sup>53</sup> which raises questions about what role they may have in explaining the behavioral abnormalities found in autism.

### **Theory of Mind**

Theory of Mind (ToM) is the main cognitive model of the social impairments of individuals with autism-spectrum disorder. Defined as the capacity to impute states of mind to oneself and others,<sup>54</sup> individuals with a ToM deficit may have serious deficits in their interpersonal relationships, as the understanding of the mental state of others may guide one's own behavioral and emotional responses. In addition to social interactions, ToM deficits may lead to pragmatic language impairments, absence of pretend play and lack of empathy.<sup>14</sup>

ToM may be divided into two domains: decoding mental state, which involves the capacity to perceive the mental state of others in observable information, such as facial expression and gestures; and reasoning other's mental states, which involves the capacity to integrate information in the context with information about the history of people (attitudes, knowledge, experiences) to understand their behavior.<sup>55</sup>

Eight studies evaluated ToM in parents of individuals with autism.<sup>16,37,40,43,44,46-48</sup> In the analysis of decoding tasks, parents of individuals with autism had more difficulties in reasoning the emotions of others' than parents in the control group in the studies conducted by Palermo et al.<sup>40</sup> (sadness and disgust), Wallace et al.<sup>47</sup> (fear and disgust) and Neves et al.<sup>48</sup> (fear, anger and happiness). However, results were similar to those found for the control groups in the studies conducted by Gokcen et al.<sup>44</sup> and Bölte & Poustka.<sup>37</sup> In the analysis of mental state reasoning tasks,

parents of individuals with autism performed worse than the control group in the study conducted by Di Michele et al.<sup>16</sup> and Gokcen et al.<sup>44</sup> The instruments used in the evaluations were not the same.

Some studies evaluated parents of individuals with autism using behavioral and cognitive measures. Parents that had abnormal results in a behavioral domain (social interest) had a poorer performance in the ToM tests than parents of individuals with autism who had abnormal results in other behavioral domains, such as rigidity and language pragmatics, and parents of typically-developing children.<sup>43,46</sup>

## Discussion

This review investigated what traits may be part of the BAP of parents, particularly the triad of core symptoms and the major cognitive models. Although BAP is a topic of great interest for investigators in this field and numerous studies have been published (over 35), the quality of studies according to topic is low when domains are analyzed individually. Except for the language and communication domain, the one most often studied (16 studies), the mean number of studies per topic was seven. The constructs analyzed are heterogeneous, and the number of publications is even lower when an isolated sub-item is investigated, such as planning as a sub-item of executive functions. Therefore, further studies should be conducted before there is a definition of which traits are undoubtedly part of the group of BAP characteristics.

The results of the studies that evaluated behavioral characteristics in a global context are convergent, as parents of individuals with autism have more abnormal results than other groups of parents in the three domains. However, the analysis of subdomains reveals differences of opinion between authors, particularly in the inclusion of abnormal language structure use as part of the BAP.

Although they all belong to the same domain, subdomains may have different cognitive origins, and the combination of traits may lead to biased conclusions in the understanding of these disorders. For example, the third domain of the triad includes both repetitive and stereotyped mannerisms and specific and intense interests. Therefore, as the number of traits included in one single domain increases, more contradictory findings will be reported. The instruments used in the evaluation of behavioral characteristics often include several different characteristics within each domain. To ensure less discrepancy between results, future studies should use instruments that evaluate the three behavioral domains individually and include the subdomains of each factor.

The analysis of the cognitive models of autism in the literature revealed that there is a cognitive abnormality associated with each behavioral symptom and responsible for it.<sup>15</sup> According to some hypotheses, ToM deficits may be associated with social deficits and deficits in communication<sup>36</sup>; the deficits in executive functions may be associated with the high level of rigidity and perseveration<sup>53</sup>; and weak central coherence may be associated with communication difficulties<sup>51</sup> and visual and spatial differences.<sup>50</sup> However, as these models are not universally adopted in autism, they are also not expected to be in the BAP. All three theories were investigated in studies that supported or refuted their inclusion in the group of BAP characteristics, and the results are, therefore, inconclusive.

Cognitive abnormalities, as well as behavioral symptoms, are also substantially heterogeneous and divided into subdomains. Therefore, an individual may have deficits in one area, but not in another. ToM involves the ability to decode and understand mental states, whereas executive functions make up a spectrum of competences, such as the capacity of organization, planning and working memory, among others. Central coherence may also be divided, as it includes both visual and verbal skills. Therefore, when a deficit in a construct is said to be part, or not, of a group of BAP characteristics, the subdomain to which it refers should be specified.

The evaluative instrument should be examined to define whether they are capable of detecting subtle changes, because the BAP is, by definition, made up of milder characteristics than those that define diagnostic criteria. For example, the block design test, which can detect superior performance in individuals with autism,<sup>56</sup> did not detect any differences between groups of parents, which left a question open: is the test unreliable for this evaluation, or is weak central coherence actually not found in parents of individuals with autism? As the studies that used the Embedded Figures test found results with significant differences,<sup>38,45</sup> we believe that weak central coherence is, in fact, one of the endophenotypes of autism, and that the block design test is not reliable for this type of evaluation.

Most of the studies retrieved were conducted in developed countries. Of the four instruments used to evaluate behavioral characteristics, two have been translated to Portuguese: BAPQ (manuscript being prepared by authors) and AQ.<sup>57</sup> However, neither has been validated for use with Brazilian populations.

One of the limitations of this review was the exclusion of studies that evaluated the BAP only in siblings. As the literature about it is extensive, it would not be possible to analyze the major traits in both groups and report

all findings in a single review. Therefore, we chose to evaluate only those studies that included parents or both, parents and siblings. However, we believe that studies about the BAP in groups of siblings would bring a great contribution to the understanding of the possible BAP characteristics and would increase the number of studies for each domain.

## Conclusion

Parents of individuals with autism have attenuated behavioral abnormalities in the triad of core symptoms that characterizes autism.

The evidence of participation of the cognitive theories of ToM deficit, executive functions deficit and weak central coherence in the group of characteristics of the endophenotype of autism is not conclusive. Further studies should be conducted using instruments that detect subtle cognitive abnormalities and evaluate the different subdomains of cognitive models.

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