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Social Communication and Language Deficits in Parents and Siblings of Children with ASD – A Short Review

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1. Introduction

Autism spectrum disorders (ASD) are a group of neurodevelopmental disorders associated with severe deficits in social communication, often accompanied by restricted patterns of behaviour, activity and interests [1]. ASD prevalence has been on the rise and is currently estimated to be 1:68 with higher rates for boys (1:42) than girls (1:189) [2].

Social, communication and cognitive deficits typical for ASD can affect individual sufferers with various severity and in many different combinations, which prompted the concept of autistic continuum, later replaced by autistic spectrum [3, 4]. Already in the earliest publications on autism, Kanner [5] and Asperger [6] identified certain similarities in the untypical severity of certain traits and behaviours in children with autism and their parents, such as tendency to social withdrawal, limited interest in people, late speech development and pedantry. Further research demonstrated that autistic traits are more prevalent in the closest relatives of individuals with ASD than in other groups [7-11].

Subtle, subclinical traits or characteristics that parallel the defining features of autism, present in non-affected individuals, in particular the first-degree relatives of people with autism, are referred to as “Broader Autism Phenotype” (BAP) [10-13]. The presence of specific characteristics in terms of social and communication skills, cognitive processes and personality in parents and siblings of individuals with ASD may suggest genetic involvement in the aetiology of autism, what is strongly supported by evidence obtained from twin and family studies. Research in this area may expand our knowledge of the nature of autism and the mechanisms underlying the emergence of its characteristic symptoms [14].

It has been estimated that BAP characteristics may be presented in at least 10-20% of parents and siblings of children with these disorders [12, 15]. For instance, Bolton et al. [12] found out

that 12,4% of siblings of the autistic probands compared to only 1,6% of the siblings of Down's syndrome were described as performing autistic traits, but of a less severe degree. In spite of many studies considering difficulties demonstrated by relatives of children with ASD, specific determination of characteristics included in BAP requires further research.

A number of publications have described the specific personality traits of parents and siblings of children with ASD: shyness, preference to be alone, insistence on sameness, reluctance to change and obsessive-compulsive behaviours [16, 17]. There are also data on the mental health problems in members of these families [18], although the results of studies in this area are not consistent (see: [19] for review).

In addition, the relatives of children with ASD demonstrate a specific cognitive characteristics. They achieved lower scores in attribution mental states based on facial expressions tasks [20], showed weaker central coherence (e.g. [16, 20, 21]), and a lower level of efficiency in planning, attention shifting and other executive function [22-26].

Several comprehensive reviews of the few dozen years of research on BAP have been published (e.g. [7, 14, 27, 28]). In this article we will be focusing on social communication problems such as understanding body language and emotional expression, as well as specific language characteristics in parents and siblings of people with ASD. Impaired language and social communication problems are defining elements of autism and include a delay or lack of spoken language that cannot be compensated by other means of communication, difficulties in initiation and maintenance of conversation as well as repetitive and stereotypic language patterns and expressions [29]. The deficits in these domains are recognized as the key features of broader autism phenotype [13, 30, 31].

This review was conducted in the first half of 2014 using the following electronic databases of international literature: Web of Science, MEDLINE/PubMed, SCOPUS, EBSCO. The articles reviewed were published from 1992 to May 2014. Keywords related to phenotype, endophenotype, autism, parents, siblings and family were used. The next step was to select studies meeting the following criteria: a) published in English; b) the social communication and language characteristics of autism in parents and siblings of individuals with autism were objects of study; c) original studies. We have excluded articles that did not explore the themes of social communication and language, those that discussed research on BAP in general population or more distant relatives of individuals with ASD rather than in their parents and siblings, as well as studies that did not include a control group.

2. Research on social communication and language in parents and siblings of individuals with ASD

Tab. 1 presents a summary of information about research on social communication and language in parents and siblings of individuals with autism. Descriptions of individual studies are limited to themes associated with social communication and language, although the majority of reviewed articles covered other aspects of BAP as well.

Study	Characteristics	Participants	Main results
Landa et al. (1992) [32]	Pragmatic language; verbal interactions	43 ASD parents (sex ratio not reported); 21 control adults (sex ratio not reported)	42% of ASD parents had some pragmatic language deficit, compared to 2% of controls
Szatmari et al. (1993) [33]	Cognitive impairments including language; developmental history	The unaffected siblings and parents of 52 PDD probands and 33 Down syndrome and low birth weight controls	No differences on the social and communication domains of the Vineland Adaptive Behavior Scales in ASD siblings compared to control siblings; No group differences in developmental history of language delays
Bolton et al. (1994) [12]	Social and communication impairments	ASD relatives (198 parents, 137 siblings); Control relatives (72 parents, 64 siblings)	20.4% of ASD siblings (and 3.1% of control siblings) showed communication atypicalities, social impairments, or restricted behaviors; The same pattern of results in parents, but to a lesser degree
Baron-Cohen and Hammer (1997) [34]	Reading emotions in the eyes	30 ASD parents (15 mothers and 15 fathers); 30 control adults (15 females and 15 males)	Parents of children with autism were slightly impaired in emotion recognition
Fombonne et al. (1997) [35]	Verbal intelligence, reading and spelling skills	99 first-degree ASD relatives; 36 relatives of individuals with Down syndrome	Slightly higher mean verbal IQ scores in relatives of ASD individuals; Siblings of ASD individuals, affected with the broad phenotype of autism, had significantly lower IQ scores, poorer reading and spelling abilities than unaffected siblings
Piven and Palmer (1997) [25]	Reading and spelling performance	25 mothers and 23 fathers from 25 multiple-incidence autism families; 30 mothers and 30 fathers from 30 Down syndrome families	ASD parents showed weaker reading performance (passage comprehension and rapid automatized naming) compared to parents of individuals with DS
Piven, Palmer, Landa, Santangelo, Jacobi, Childress (1997) [10]	Pragmatic language	39 multiple-incidence autism parents (having two children with autism); 58 parents of children with Down syndrome	Higher rates of speech and pragmatic language deficits in multiple-incidence autism parents
Piven, Palmer, Jacobi, Childress and Arndt (1997) [11]	Social and communication deficits	25 multiplex autism families; relatives of 30 Down syndrome probands	Higher rates of social and communication deficits in the families with multiple-incidence autism
Folstein et al. (1999) [36]	Pragmatic language; verbal IQ; reading and spelling performance	166 parents and 87 siblings of individuals with autism; 75 parents and 64 siblings of children with Down syndrome	No differences in verbal IQ scores, reading and spelling skills; More deficits in pragmatic language in parents of individuals with autism

Study	Characteristics	Participants	Main results
			as well as early language-related difficulties in that group
Hughes et al. (1999) [22]	Verbal fluency	31 siblings of children with autism; 32 siblings of children with developmental delay	Superior verbal span in siblings of children with autism; Bigger than expected part of that group of siblings achieved poor results in verbal fluency tasks
Pilowsky et al. (2003) [37]	Language abilities	27 siblings of children with autism, 23 siblings of children with mental retardation of unknown etiology, 22 siblings of children with developmental language disorders	Higher scores in siblings of children with autism on receptive, expressive, and total language scales of the Children's Evaluation of Language Fundamentals and on verbal IQ compared to siblings of children with developmental language disorders
Bishop (2004) [8]	Communication skills (self-report measure)	Children with ASD (59 with autism, 21 with PDD-NOS); ASD parents (65 mothers, 46 fathers); Control parents (48 mothers, 37 fathers)	Communication skills significantly lower in ASD parents (particularly fathers) compared to control parents
Bishop et al. (2004b) [38]	Phonological processing, communication	145 parents of children with ASD; 96 parents of typically developing children	No difference in phonological processing; In the group of parents classified as BAP there was a history of more language and literacy problems than in other ASD parents
Dorris et al. (2004) [39]	Mind-reading (Eyes Test)	27 siblings of children with Asperger syndrome (AS); 27 control children	Poorer performance of the AS siblings in the Eyes Test
Hill, Berthoz and Frith (2004) [40]	Emotion processing	27 high-functioning adults with autistic spectrum disorders, their biological relatives (n = 49), and normal adult controls (n = 35)	No significant differences between relatives of individuals with ASD and controls in identifying and describing feelings
Bishop (2006) [41]	Communication deficits (assessed by parents using Children's Communication Checklist-2, CCC-2)	43 ASD siblings; 46 control children	The only difference between groups in syntax; 23.8% of ASD siblings scored 2 SD below the control mean on CCC-2, compared to 2.2% of controls; Some differences in structural language skills
Palermo et al. (2006) [42]	Recognition of schematic displayed emotions	40 parents of children with autism, 40 control adults	Poorer performance in parents of children with autism; Fathers of children with autism had more difficulties than mothers
Chuthapisith et al. (2007) [43]	Language development	32 preschool siblings of children with autism (aged 2-6 years); 28 control children	Delayed language development in 8 of autism siblings; After excluded the siblings with ASD and DLD diagnosis, in the remaining 29 siblings

Study	Characteristics	Participants	Main results
			verbal IQs were not significantly different from the control group
Di Michele et al. (2007) [44]	Pragmatic language (evaluation of the taped conversations)	23 parents of children with autism; 12 parents of children with Down syndrome and 23 parents of healthy children	More pragmatic language difficulties in parents of people with autism; Problems in identifying relevant and redundant, non-essential information
Losh and Piven (2007) [45]	Ability to read complex psychological states from viewing only the eye region of faces	48 parents of individuals with autism; 22 control parents, including parents of individuals with Down syndrome and typically developing children	No differences between parents of individuals with autism and Controls; There was an „aloof” subgroup identified among parents of individuals with autism (n = 13); The results of that group were lower than the results obtained by Controls in the Eyes Test
Ruser et al. (2007) [46]	Communicative competence; pragmatic language, over-talkativeness	47 parents of individuals with autism; 47 parents of children with specific language impairment (SLI); 21 parents of children with Down syndrome	Parents of children with autism and SLI presented lower communication abilities than parents of children with DS; Severe pragmatic language deficits in about 15% of autism and SLI parents
Pilowsky et al. (2007) [47]	Neurocognitive functioning	30 siblings of children with autism; 28 siblings of children with mental retardation, 30 siblings of children with developmental language delay	No differences between siblings of children with autism and the other groups
Whitehouse et al. (2007) [48]	Communication (self report measure: Autism Quotient by Baron-Cohen et al., 2001)	30 parents of children with autism; 30 parents of children with specific language impairment; 30 parents of children typically developing	Communication difficulties in parents of children with autism
Adolphs et al. (2008) [49]	Face processing	15 socially 'aloof' parents of individuals with autism; 27 'nonaloof' parents of children with autism; 20 control parents of neurotypical children	Socially 'aloof' parents showed poorer performance compared to 'nonaloof' parents and control parents
Scheeren and Stauder (2008) [50]	Communication (measured by AQ)	25 parents of children with HFA; 25 parents of typically developing children	No differences between groups
Schmidt et al. (2008) [51]	Phonological processing	22 parents of children with autism; 22 controls	ASD parents achieved lower scores on the nonword repetition task; No differences between groups in figurative language, receptive language, expressive language, verbal fluency and in history of reading difficulties
Losh et al. (2008) [13]	Pragmatic language	48 parents of individuals with autism (multiple-incidence	More pragmatic and speech errors in MIAF parents than in SIAF parents; SIAF parents

Study	Characteristics	Participants	Main results
		autism families - MIAF); 78 parents of individuals with autism (single-incidence autism families - SIAF); 60 parents of individuals with Down syndrome	committed significantly more pragmatic violations and speech errors than Down syndrome parents
Gamliel et al. (2009) [52]	Language development	37 siblings of children with ASD (SIBS-A); 47 siblings of typically developing children (SIBS-TD) (longitudinal study: from 4 months to 7 years)	At 7 years, 40% of the SIBS-A (and 16% of SIBS-TD) showed cognitive, language and/or academic difficulties (this subgroup was named SIBS-A-BP); Early language scores (14–54 months) were significantly lower in SIBS-A-BP compared to the language scores of SIBS-TD. Language was a major area of difficulty for SIBS-A during the preschool years
Lindgren et al. (2009) [53]	Syntax memory for language, Lexical comprehension, Semantics, Morphology, Reading abilities	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	Parents of people with autism and language impairment had a better performance in language tests than parents of children with specific language impairment
Losh et al. (2009) [54]	Reading complex psychological states from the eye region of faces	36 high-functioning individuals with autism, 41 controls (neurotypical individuals with no family history of autism), 83 parents of individuals with autism, 32 control parents (with no family history of autism or developmental delays)	There were three groups of parents of individuals with autism extracted: group of parents with social BAP characteristics (n = 22); group with the rigid/perfectionistic BAP traits (n = 34), and group without BAP features BAP (-) (n = 40). In Reading the Mind in the Eyes Test parents of individuals with autism with social BAP performed poorer than control parents. No difference between Controls and BAP (-) parents
Ben-Yizhak et al. (2011) [55]	Pragmatic language, school related linguistic abilities	School-age siblings of children with autism (SIBS-A), n=35; Controls (n = 42)	Lower pragmatic abilities in a subgroup of SIBS-A identified with BAP related difficulties; No differences between groups in school achievements and reading processes
Losh et al. (2010) [23]	Rapid automatized naming	Three samples: I: 48 parents of multiple children with ASD; 62 parents with a single child with autism; 53 parents of children with Down syndrome;	Longer naming times in parents of individuals with autism and in people with HFA compared to controls

Study	Characteristics	Participants	Main results
		II: 167 parents from multiplex families; III: 83 parents of individuals with autism, 32 parent controls, 36 high-functioning individuals with autism, 38 controls	
Wheelwright et al. (2010) [56]	Communication (self-report using AQ)	2,000 parents of children with ASD (571 fathers and 1429 mothers); 1,007 parents of typically developing children (349 fathers and 658 mothers)	Parents of children with ASD showed more communication difficulties
Whitehouse et al. (2010) [57]	Language (speech, syntax and semantics), pragmatic skills, communication style	238 parents of children with autism; 187 parents of typical individuals	No differences between groups in the language subscale
Levy and Bar-Yuda (2011) [58]	Language performance	Siblings of nonverbal children with autism SIBS-ANV (n=28); Controls (n = 27); aged 4–9 years	SIBS-ANV achieved lower scores on the Receptive Scale, Expressive Scale and the Total Language Scale of the Clinical Evaluation of Language Fundamentals; Differences in the language scores were associated with IQ
Neves et al. (2011) [59]	Facial emotion recognition	40 parents of children with autism; 41 healthy controls	Parents of children with autism performed worse in the facial emotion recognition test than controls
Bernier et al. (2012) [60]	Conversational skills	39 parents of multiple-incidence autism families (M-parents); 22 parents of single-incidence autism families (S-parents); 20 parents of children with developmental delay without ASD (DD); 20 parents of typically-developing children	Greater impairment in social communication skills in M-parents compared with S-parents, DD parents, and parents of typically developing children
Berthoz et al. (2013) [61]	Emotional impairments	High functioning adults with ASD (n = 38), parents of individuals with ASD (n = 87), typical adults (n = 47)	Parents differed from controls on social anhedonia; Higher proportion of parents were classed as alexithymic, compared with controls
Sucksmith et al. (2013) [62]	Emotion recognition	314 adults with ASD; 297 parents with children with ASD; 184 controls	No difference between parents of a child with ASD and controls at recognising the basic emotions (after controlling for age and non-verbal IQ)
Gizzonio et al. (2014) [63]	Verbal IQ	32 children with ASD, 21 siblings of these children,	No significant difference between Verbal Intelligence Quotient and Performance Intelligent Quotient scores between groups;

Study	Characteristics	Participants	Main results
		43 children with typical development	Not significant, a predominance of performance over verbal abilities observed in siblings group
Kadak et al. (2014) [64]	Recognition of emotion (face expression)	36 mothers and 36 fathers of children with ASD; 19 mothers and 19 fathers of typically developing children	Poorer recognition of emotional expressions in ASD parents
Oerlemans et al. (2014) [65]	Recognition of facial emotion and affective prosody	90 children with ASD (43 with and 47 without ADHD), 79 ASD unaffected siblings, 139 controls aged 6-13 years	The worse performance of unaffected siblings than the controls and better than the ASD probands in recognition of facial emotion and affective prosody tasks

Table 1. Social communication and language characteristics in parents and siblings of individuals with ASD

As it is shown in Table 1, many authors have found social communication and language deficits in first-degree relatives of individuals with ASD. Both receptive and expressive language is affected [37]. Difficulties include, among others: pragmatic language deficits (e.g. [10, 12, 13, 32, 36, 44, 46, 55]), verbal fluency [22], reading abilities [25, 35], delay of language development and problems in language developmental history [11, 38, 43, 52], conversational skills [60] and syntax [41].

Some researchers, however, found no differences between first degree relatives of people with ASD and the comparison groups in the social communication and language (e.g. [33, 47, 51]). No such differences were found, among others, in the language development history [33], verbal fluency [51] and reading and spelling abilities [36, 55]. Similarly, there were no differences between parents or siblings of individuals with autism and control groups in terms of phonological processing [38, 51] and structural language [53, 57].

Findings on the development of verbal and non-verbal intellectual skills in first-degree relatives of people with autism are less consistent. Some comparisons have shown that first-degree relatives of individuals with ASD had lower verbal IQ compared to control groups (e.g. [37]), while other studies found no such differences [36, 63]. Fombonne with colleagues [35] even reported slightly higher verbal IQ in relatives of individuals with autism than in relatives of people with Down syndrome.

Studies using self-reported measures to assess difficulties in communication experienced by parents of individuals with ASD have also failed to provide a clear picture. In the majority of those studies parents reported more severe difficulties in that area compared with adult controls (e.g. [8, 48, 56]). Scheeren and Stauder [50], however, found no differences when comparing parents of high-functioning individuals with autism and parents of typically developing children.

In a similar way several studies have indicated also that parents or siblings of children with ASD showed lower scores in recognize emotions tasks [34, 39, 42, 59, 61, 64, 65] than Down syndrome or typically developing children relatives. However, Sucksmith with colleagues

[62], after controlling for age and non-verbal IQ, did not detect differences in recognizing the basic emotions between parents of children with ASD and controls.

It should be noted that in some studies in which differences between first-degree relatives of individuals with autism and controls were not significant it was possible to identify subgroups of participants demonstrating traits similar to those seen in individuals with ASD. Subgroups with BAP characteristics are significantly more numerous in the groups of parents or siblings of individuals with ASD than subgroups with similar problems in control groups. For instance, Landa with colleagues [32] stated that 42% of parents of children with ASD had some pragmatic language deficits, compared to 2% of control parents. Findings from research where it has been shown that among parents or siblings of people with autism there were the subgroups that manifested some difficulties in social communication and language, but it does not apply to these groups as a whole (e.g. [43, 45, 49, 54, 60, 66]) may be particularly relevant to further research on genetic involvement in BAP. Schmidt with colleagues [51] showed impairments in phonological processing in parents of children with low functioning autism. In their study on emotion recognition, Adolphs with colleagues [49] found difficulties in parents identified as “socially aloof”, while “nonaloof” parents were similar to controls. Folstein et al. [36] found that only those parents of individuals with autism who showed cognitive deficits associated with language in childhood performed worse than parents of individuals with Down syndrome in reading and writing tasks. By controlling for a variety of variables, including autism severity and developmental characteristics individuals with autism, as well as the number ASD cases in the extended family (e.g., taking into account the siblings of both parents of an individual with autism, as well as their children) we are likely to find out more about BAP.

Some empirical data suggest that families may differ in terms of genetic liability to autism. Losh et al. [13] compared three groups of individuals: 25 parents from multiple-incidence autism families, 40 parents from single-incidence autism families, and 30 parents from Down syndrome families. They found that autistic characteristics were most pronounced in parents from multiple-incidence autism families, less pronounced in single-incidence autism families, and weakest in parents of children with Down syndrome. In the majority of families with two children with autism, both parents demonstrated autistic characteristics; by contrast, in families with one child with autism the likelihood of both parents showing those characteristics was the same as for one parent or neither parent to have autistic traits. Gerdtts and Bernier [66] showed that mothers, fathers, and siblings from multiplex ASD families were less expressive in their use of nonverbal communication compared to mothers, fathers and siblings from simplex families. Thus, it appears that research on multiple-incidence autism families can provide valuable information with respect to the hereditary mechanisms underlying autism. Schwichtenberg et al. [67] found that children from multiplex autism families had greater BAP traits than simplex siblings, and ASD multiplex infant siblings were more likely to develop ASD than ASD simplex and control. Findings from research on BAP in monozygotic and dizygotic twins are also interesting. It was shown [68] that concordance for BAP was much greater in MZ pairs than DZ pairs.

Recently there has been a surge in research on infants at high familial risk for ASD (see [69] for review). An estimated 10-20% of at high risk infant siblings may be affected by sub-clinical

ASD symptoms or other developmental impairments [70]. These studies are not included in Table 1 because participants included children with ASD. Nevertheless, their findings with respect to social communication and language are relevant to the understanding of BAP. A number of those research projects have shown that some siblings of individuals with ASD demonstrate observable communication deficits already in the first three years of life and that these impairments can change over time. They include, among others, lower receptive language scores, delayed receptive and expressive language [15, 71-74], requesting behavior [75, 76], understanding words and phrases, gesture use, and social-communicative interactions with parents [77]. The important question is how early are those problems manifested. It was shown that at 6 months of age there were no statistically significant group differences in language development between high risk (HR, children having sibling with autism) and low risk (LR) infants (no autism history in family) [15, 78]. No differences in gaze following were found in children of 7 and 13 months between HR and LR groups [79]. Georgiades et al. [80] concluded that pragmatic language deficits were not relevant traits of BAP at 12 months. Obviously, this does not mean that HR children experience no deficits in language development at that age. Ozonoff with colleagues [78] found atypical language development in high-risk infants of 12 months of age. Differences in language between HR and LR infants of 24 months of age are found much more often [15, 73, 74]. Longitudinal studies are the most desirable as they offer insight into the dynamics and changes in the development of these children. While problems are overcome in some, in others they persist at later stages. As demonstrated by Gamliel et al. [71], expressive and receptive language deficits were still present in HR children at 54 months of age despite the resolution of some other developmental problems. Another important issue is to find out how many of 24-months old HR siblings demonstrating language difficulties are eventually diagnosed with ASD. Hudry et al. [81] found that reduced receptive vocabulary advantage in high risk infants at 14 months, maintained to 24 months only in the subgroups of ASD or other atypicality outcome. This suggests a close to typical development of other children in the HR group. The results of these sample studies on HR infants expose gaps in the knowledge on the issue.

3. Conclusions

Currently, it is difficult to identify universal, clear regularities relating to social communication and language deficits in parents or siblings of children with autism, but they have been found in some subgroups. A more complete knowledge in that area can contribute to a better understanding of autism. It can also provide hints for future research, by focusing attention on selected subgroups of parents and siblings.

There are many reasons for the variation in empirical results discussed in this section. Specific ones include methodological considerations such as sample size, research methods, enrolment criteria, as well as specifics of control groups (Cf. [19]).

It would be difficult to identify the components of BAP in terms of social communication and language based on currently available data. The best documented aspect of BAP appear to be

pragmatic language deficits. Other characteristics analysed in the above studies as BAP components, such as delay of language development, difficulties in reading, spelling and writing, difficulties in structural language use or verbal fluency, remain controversial.

A number of studies on BAP focus on parents, and there are also many that analyse HR infants. In other studies on siblings, groups tend to be very heterogeneous, e.g. in terms of age. Longitudinal research on the development of social communication and language deficits in preschool and school age siblings are particularly necessary, especially that, as shown by Gamliel et al. [52], language may be a major area of difficulty for siblings of individuals with autism during the preschool years.

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