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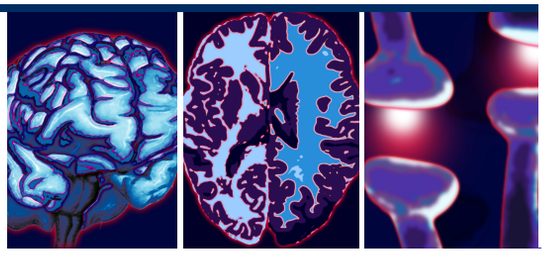

The developing language abilities and increased risks of 'unaffected' siblings of children with autism spectrum disorder

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Practice points

- Siblings of children with autism spectrum disorder (ASD) have a one in five chance of being diagnosed with ASD.
- This risk is over 16 times higher than children in the general population.
- First degree relatives are at an increased risk for subclinical ASD traits, referred to as Broader Autism Phenotype, and other neurodevelopmental and psychiatric disorders.
- Structural language refers to the lexical and syntactic abilities in receptive and expressive language.
- In ASD, individuals demonstrate a wide range of structural language abilities.
- Common features in ASD include atypical language and early language delays.
- Prospective longitudinal studies of infant siblings of children with ASD reveal that those who do not go on to have ASD are still at greater risk for early language delays than low-risk controls.
- Many studies have shown particular delays in receptive language in this group.
- Very few studies have examined structural language in siblings in middle childhood, and results are mixed; larger sample sizes are needed.
- Pragmatics refers to the appropriate use of language in a social context.
- Pragmatic abilities are universally impaired in children with ASD, making these abilities a strong candidate for investigation in high-risk siblings.
- However, measurement of pragmatics can be challenging.
- Despite strong research rationale, currently, there is a scarcity of research on pragmatic abilities in non-ASD siblings.

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Future Medicine part of fsg

- There are few longitudinal studies on language outcomes of non-ASD siblings in middle childhood and beyond.
- There is a need for research with larger sample sizes, careful case definitions, and studies that examine pragmatic language abilities in a wider range of contexts.
- The field has seen an increasing interest in non-ASD outcomes in the past few years, and this is expected to continue to grow.
- Ongoing prospective longitudinal studies will likely begin analyzing data on children into middle childhood in the next few years, hopefully producing replicable findings on the nature of language abilities and impairments, as well as the ability to link early predictors with language outcomes.
- Time will reveal whether the advent of DSM-5, with the newly devised ASD and social (pragmatic) communication disorder categories, will impact research into broader autism phenotype and language outcomes in siblings.

SUMMARY Siblings of children with autism spectrum disorder (ASD) are at a higher risk than the general population for ASD, ASD traits and other developmental differences. A review of prospective longitudinal studies that have followed siblings up to age 3 reveal that siblings with non-ASD outcomes are at increased risk for early language delays; however, there is a dearth of research on language development in these children through to middle childhood. Preliminary research into pragmatics (i.e., the contextually appropriate social use of language) suggests that non-ASD siblings may be at increased risk for impairment in this area, but this is a field ripe for further investigation. Understanding the nature of the increased risk for language and related learning challenges in non-ASD siblings of children with ASD is important for researchers, clinicians and parents, as early identification can lead to early intervention and better outcomes.

As the prevalence of autism spectrum disorder (ASD) has skyrocketed in the past couple of decades, so has the research surrounding this disorder [1], including evidence of high recurrence of ASD in siblings [2], and an increasing interest in the expression of subclinical, but ASD-related traits in some non-ASD family members (referred to as the 'broader autism phenotype'; BAP [3]). Despite increasing attention to the developmental differences in family members, we are still early in our discovery of the developmental risks and outcomes of non-ASD siblings of children with ASD. Language is one particular domain of concern and future inquiry, as emerging research suggests that ostensibly 'unaffected' siblings (i.e., those with non-ASD outcomes) may be at greater risk for delayed acquisition of language as well as pragmatic language impairment.

Understanding the nature of risks in language development for these siblings is so important, as language proficiency greatly impacts overall development and quality of life. Language is necessary for communicating needs and desires; it is the key to academic achievement and occupational performance; and it is integral to building social relationships as well as understanding the world around us. If research were able to uncover any risks that siblings of children with ASD may encounter during language development, it would have practical implications for these children to be screened and to be given access to timely language interventions.

ASD is a neurodevelopmental disorder characterized by social-communication deficits and the presence of repetitive behaviors, activities, or interests [4]. This lifelong disorder presents early in development and usually has diagnostic stability by 2–3 years of age [5,6]. For the purposes of this paper, the term ASD will be used to include the previous DSM-IV [7] categories of Autistic Disorder, Asperger Syndrome and Pervasive Developmental Disorder-Not Otherwise Specified. Additionally, when 'siblings' is used as a stand-alone term it will always refer to children who do not have ASD themselves, but do have

an older brother or sister (proband) with ASD.

Siblings of children with ASD: increased susceptibility

Part of the broader rationale for investigating the language abilities of siblings of children with ASD is the considerable evidence that these siblings are at an increased risk for developmental differences, particularly in domains that are affected in ASD itself. First degree relatives of individuals with ASD at greater risk for ASD, ASD traits, and other neurodevelopmental and psychiatric disorders [2,8–11]. Decades of research into the genetic architecture of ASD reveals that there is a substantial genetic contribution to ASD, with hundreds of ASD-risk genes implicated (see [12] for a review), though pre- and post-natal environmental factors are also seen as playing a noteworthy role [13]. Accordingly, ASD has a high recurrence rate within families and a high concordance rate in monozygotic twins, with the latter falling between 50–90% [13,14]. Regarding recurrence rates in younger siblings of children with ASD, the most recent estimate from a large-scale consortium of phenotyping studies (the 'Baby Sibs Research Consortium', funded by Autism Speaks) indicates a rate of 18.7%, which represents almost a one in five chance of being diagnosed with ASD [2]. This compares to a prevalence of one in 88 in the general population [101], indicating that siblings are over 16 times more likely to develop ASD.

The familial aggregation of ASD has led many researchers to investigate the presence of 'autistic-like' characteristics in family members. Evidence has been found for subclinical manifestations of the core features of ASD (social-communication impairment and repetitive behaviors/interests) in relatives and the general population [8]. These milder presentations of ASD symptomatology, referred to as the 'Broader Autism Phenotype', or BAP [3], are more common in biological relatives of children with ASD than in nonbiological relatives or the general population, and rates of BAP increase as the incidence of ASD in a family increases [3,15–17]. The majority of research into autistic traits has focused on the parents of children with ASD [18,19]; however, a growing body of literature is beginning to explore BAP in siblings.

Looking beyond ASD-specific symptomatology, parents and siblings of individuals with ASD also appear to be at greater risk for a range of neurodevelopmental and psychiatric difficulties and disorders, including but not limited to language impairments, dyslexia, attention deficits, obsessive-compulsive traits, social phobia and depression [9–11,20]. In early childhood, siblings of children with ASD are at greater risk for cognitive, temperament and behavior difficulties, including poor emotion regulation and sleep disturbance [21–25]. In a recent study, Georgiades *et al.* [26] studied 170 non-ASD siblings and 90 low-risk controls at 12 and 36 months of age. Cluster analysis was used on the combined sample, revealing a distinct cluster of children with reduced cognitive performance and elevated ASD symptomatology as measured by the Autism Observation Scale for Infants [27] as early as 12 months of age. The vast majority of this cluster (almost 90%) consisted of non-ASD siblings, representing 19% of the total non-ASD sibling sample. On average, at 3 years of age this subgroup of children displayed more social and communication challenges, lower levels of cognitive functioning and more internalizing problems, as measured by the Autism Diagnostic Interview-Revised (ADI-R [28]), Mullen Scales of Early Learning (MSEL [29]) and Infant-Toddler Social-Emotional Assessment [30], respectively.

Language abilities in children with ASD & their siblings

The research outlined above underscores that siblings of children with ASD are an at-risk group: not only for ASD and ASD-related symptoms, but also for other developmental difficulties. We now turn to look specifically at the research on language abilities. Both of the following sections will provide context by touching on the abilities of children with ASD before focusing on their non-ASD siblings.

■ Structural language development

Structural language (i.e., lexical and syntactic abilities in expressive and receptive language) is the basic building block of other linguistic skills. Across the spectrum of individuals diagnosed with ASD there exists an extremely wide range of structural language abilities. Some children have very limited expressive and receptive language and are essentially nonverbal; others have well-developed structural language and score in the average or above average range on standardized tests of language fundamentals [31,32]. Across the gamut of lexical and syntactic abilities, many children with ASD display atypical or idiosyncratic language, such as stereotyped

and repetitive speech, echolalia, and neologisms [33,34].

Along with atypical language, another common characteristic of ASD is delayed language acquisition. On average, toddlers with ASD produce their first words at around 38 months of age, instead of by the typical 8–14 month mark [35]. Howlin [35] collected retrospective reports of early language from parents of older children. Almost all research into early language development in ASD is limited by this method of data collection; however, a study by Ellis Weismer, Lord and Esler [36] recently confirmed the severity of early language delays with direct assessment measures and with a very large sample of toddlers (aged 24–36 months, mean age = 30.6 months) with ASD (n = 257) and a control group with non-ASD developmental delay (DD; n = 69). In this study, toddlers with ASD displayed significantly greater language delays than the DD group. Toddlers with ASD also showed an atypical pattern of lower receptive to expressive language scores on the MSEL – a language profile that is opposite to the profile in typically developing (TD) children as well as in the DD control group in this study. Likewise, in a sample of 294 preschool children with ASD, Volden *et al.* [37] found a pattern of more impaired receptive language relative to expressive language on the Preschool Language Scale-4 [38], another direct assessment measure of structural language.

Turning to the group of four out of five younger siblings who do not receive a diagnosis on the autism spectrum, research has shown that delays in early language acquisition reliably differentiates this group from toddlers with no family history of ASD. The strongest research design we have to study early language development of siblings is prospective, longitudinal investigation of younger siblings of probands with confirmed ASD, often compared with children with no family history of ASD (herein referred to as low-risk controls; e.g., [39,40]). This study design allows for babies to be recruited before or shortly after birth, and infants' development can be followed from very early in life. In the last decade, large-scale longitudinal studies of this nature have been running in Canada, the USA, the UK and Israel [2,41]. Research from these studies provides a long-awaited opportunity to examine the risks and outcomes for siblings – in language and many other domains – paired with very solid methodology and growing sample sizes. To date, most prospective studies of this kind have focused on development from 0–36 months of age.

Zwaigenbaum, Bryson and colleagues have been following one of the largest (and growing) longitudinal cohorts of siblings for over a decade. One of the earliest studies from this cohort [39] compared 23 low-risk controls with 65 high-risk siblings (19 with ASD and 46 non-ASD, as determined by Autism Diagnostic Observation Schedule (ADOS) diagnostic cutoff scores at age 2). At 12 months, non-ASD siblings displayed lower receptive language scores on the MSEL, as well as fewer phrases and gestures as reported by parents on the MacArthur Communicative Development Inventory [42], in comparison to low risk controls. Non-ASD siblings from this cohort were also found to display fewer play-related gestures compared with controls at 18 months, as reported by parents on the MacArthur Communicative Development Inventory [40].

As this cohort of infant siblings has grown older, the diagnostic status of all participants is determined by an independent best estimate diagnostic assessment at 3 years of age, informed by the two gold-standard diagnostic measures – the ADOS [43], and the ADI-R – as well as by the judgment of an expert clinician, blind to family status and previous assessment information. This diagnostic model results in a very rigorous methodology that follows best clinical and research practice. In the investigation of 'unaffected' siblings, this level of systematic categorization of participants into ASD and non-ASD groups is vitally important. Otherwise, children who actually have ASD could artificially inflate the scores of the so-called non-ASD group.

In an American cohort of siblings, Toth and her colleagues [44] studied 42 non-ASD siblings and 20 low-risk controls, aged 18–27 months old. The non-ASD siblings displayed lower receptive language scores on the MSEL, analogous to the findings of Zwaigenbaum *et al.* [39], and matching the pattern of lower receptive than expressive language seen with children with ASD, as described by Ellis Weismer *et al.* [36] and Volden *et al.* [37]. Non-ASD siblings used fewer words and distal gestures, lower overall rates of social communication on the Communication and Symbolic Behavior Scales Developmental Profile [45], and a significant number of siblings displayed below average expressive language abilities on the MSEL. However, it is

noteworthy that the non-ASD status of participants was determined by clinical assessment at this same time point, particularly because their sample was as young as 18 months. Diagnostic status is not considered stable at this age, and thus the sibling group means could have been lowered by participants who later met criteria for ASD.

The work of Yirmiya and colleagues [47,48] in Israel reveals a similar language profile among a group of 30 siblings and 30 low-risk controls, aged 4–36 months old. These two studies continue to highlight the prevalence of early language delays in the sibling group: they were characterized by lower scores on the language domains of the Bayley Scales of Infant Development [49] and the Reynell Developmental Language Scale [50]. However, both of these studies lacked a systematic method for assessing ASD symptomatology beyond a short parental questionnaire (the Checklist for Autism in Toddlers [51], or the Social Communication Questionnaire [52]). Moreover, only one participant received a diagnosis of ASD, but this child's data were not removed from those of the remaining, non-ASD siblings. Nonetheless, it is unlikely that the observed group differences could be entirely explained by one participant's scores, and thus the suboptimal performance of the sibling group likely provides evidence of developmental concerns in at least some members of this group.

Hudry *et al.* [53] and the British Autism Study of Infant Siblings team provide further support for early language delays in siblings of children with ASD and lack of 'receptive advantage' as seen in children with ASD, but not in TD children. This study included 53 high-risk siblings (17 with ASD and 36 non-ASD, as determined by a clinical researcher at age 3), who were assessed at 7, 14, 24 and 38 months. A third of the non-ASD siblings in this sample were determined to have atypical outcomes at age 3, operationalized as displaying ASD traits or poor cognitive/language outcomes (i.e., high scores of the ADOS or the ADI-R; low scores on the MSEL). As a group, these atypical non-ASD siblings showed persistent language delay and alack of the 'receptive advantage'. Another noteworthy finding of this study is that the typical outcome non-ASD siblings displayed language delays at 14 months, but by 24 months these had resolved and they were on par in receptive and expressive language with the low-risk control group.

Landa and colleagues were the first to use latent class analysis to examine developmental trajectories of siblings of children with ASD [54]. Their sample of 204 siblings, whose cognitive, language and motor development was followed every 6 months from 6–36 months using the MSEL, yielded four class trajectories: accelerated development, normal development, early motor/receptive language delay and developmental slowing. Siblings were also divided into 'unaffected', BAP and ASD groups based on performance at their 3-year assessment. Here again we see a different definition of BAP; in this study, participants were classified as BAP if they had social or language delay (i.e., siblings could be defined as BAP if they displayed early language delay, even if they showed no atypical social communication features representing ASD traits). Membership in each class was generally as expected, with the majority of unaffected siblings falling into classes one and two, the majority of BAP siblings falling into two or three, and the majority of ASD siblings falling into two, three or four. Overall, 18.4% of non-ASD siblings fell into class three, revealing early receptive language and motor delay in a substantial proportion of this group. In the same longitudinal cohort, Landa *et al.* [55] reported a one-time raw score decline on the MSEL in 2% of non-ASD siblings, but no further detail or analysis was provided to elucidate the specific nature of this finding.

In the largest sibling sample to date (based on combined data collected from the Baby Siblings Research Consortium), Messinger *et al.* analyzed the 3-year outcomes of 507 non-ASD siblings and 324 low-risk controls [56]. As a group, non-ASD siblings displayed lower verbal functioning, as measured by a combined receptive and expressive language score on the MSEL, as well as lower cognitive scores on the MSEL and higher ASD traits on an ADOS severity metric. As hypothesized, not all siblings demonstrated developmental difficulties; group differences were driven by the performance of a subset of siblings. A latent class analysis revealed that the majority (65%) of non-ASD siblings occupied classes typified by low ASD severity and average developmental functioning. A further 14% of non-ASD siblings occupied a class typified by elevated ASD severity and high developmental functioning – although a similar proportion of controls fell into this class. The remaining 21% of non-ASD siblings – disproportionate to controls – fell into classes of high ASD severity with

low-average developmental functioning and low ASD severity with low developmental functioning, revealing that a substantial minority (i.e., a third) of non-ASD siblings display BAP characteristics, at 3 years of age, with or without developmental delays.

Very few longitudinal studies have been published examining the abilities of siblings beyond 3 years of age. In a study of 37 non-ASD siblings and 22 low-risk controls between ages 4 and 7, Warren *et al.* [57] surprisingly found no group differences on a direct measure of structural language, the Clinical Evaluation of Language Fundamentals, Preschool Edition [58]. They also found no group differences on measures of cognition and behavioral regulation. One significant finding was lower executive functioning abilities in the non-ASD siblings on the Developmental Neuropsychological Assessment [59].

In another longitudinal study, Gamliel and colleagues [60] published a follow-up evaluation of non-ASD siblings at 7 years of age. They compared three groups, determined at age 7, using multilevel growth curve analysis: siblings displaying BAP, siblings without BAP and low-risk controls. BAP was defined as one or more scores 1.5 standard deviations below average on the Clinical Evaluation of Language Fundamentals (CELF-3 [61]), Wechsler Intelligence Scales for Children [62] and/or Wide Range Achievement Tests [63] and/or parental reports of difficulties. Note, however, that ASD symptomatology was not considered in this definition of BAP. The results of this study reaffirm language as a core area of concern for non-ASD siblings: the sibling-BAP group had significantly lower structural language scores than controls at 7 years, and showed a pattern of lower language (but not cognitive) scores in early childhood. Interestingly, the majority of these BAP siblings demonstrated functional difficulties for the first time at 7 years, having not exceeded the threshold for concern at 4 years. This delayed onset of impairment may be due to the increasing psychosocial and academic demands that children face as they grow older. However, no assessment of ASD symptomatology was employed at the 7-year mark and the authors define BAP without assessing autistic traits, revealing a lack of consensus in the literature over case definitions. It also raises the possibility that some of the BAP (non-ASD) siblings in this study unknowingly met criteria for ASD. As the study relies on delineating a relatively small group of high-risk children into those with ASD, those with BAP, and those without either of the above, if any children were misclassified, their performance could artificially affect the group means and affect the interpretation of findings.

In contrast with Gamliel *et al.*'s [60] findings at 7 years, Ben-Yizhak *et al.* [64] did not find evidence of impaired structural language in non-ASD siblings at 9–12 years. Moreover, there was not even a trend in this direction; on average, siblings classified as BAP received the highest receptive and expressive language scores, followed by non-BAP siblings, and lastly the low-risk control group. Of note, these two conflicting studies used the same language measure (CELF-3) and the same cohort of siblings. Two notable differences between these two studies are: the age of the siblings and the definition of BAP used to dichotomize the sibling group (Ben-Yizhak *et al.* operationalized BAP as an ADOS algorithm score ≥4, whereas Gamliel *et al.* defined BAP as poor performance on measures of structural language, cognitive ability and/or school achievement). It is important to bear in mind the impact different case definitions may have on results. As well, the specific nature of language difficulties may evolve over the course of development, so there is a need to be mindful of the effects of both increasing demands and opportunities for improvement.

■ Pragmatic language

Although structural aspects of language may be intact in children diagnosed with ASD, pragmatic abilities are universally impaired [64–66]. Pragmatics refers to the appropriate social use of language as a communicative tool, and a diagnosis of ASD requires a deficit in social-communicative behavior [4]. Hymes [67] described pragmatics as 'a speaker having knowledge of when to say what to whom, and how much to say' – for example, deciding when to use a pronoun (e.g., 'he jumped') versus a pronominal expression (e.g., 'the dog jumped') based on a listener's need for reference. The speaker must give enough information, but not too much information, in order to transmit their message appropriately in relation to the communicative context. In turn, the listener must rely on this communicative context in order to successfully interpret the message. In the pragmatics domain, 'information' includes two types of cues: linguistic (e.g., word choice and amount of background information) and paralinguistic (e.g., tone of

voice, volume modulation, emphasis, prosody and gestures).

Within pragmatics, individuals with ASD have well-documented difficulties with conversational back-and-forth, conversational repair, adjusting the register of speech to a given social situation, as well as nonverbal components, such as appropriately integrating eye contact, facial expressions and gestures [32,68,69]. However, measurement limitations are one of the major barriers to research on pragmatic language. Pragmatics is defined as context-appropriate behavior; as such it makes most sense to study it in context, but this can be challenging. It is a difficult balance between measurement reliability (i.e., collecting standardized, quantifiable data) and external validity (i.e., capturing the wide variety of impairments that occur spontaneously in the complex social world). One approach has been to examine speech produced during discourse or narrative tasks. Participants are presented with a standard task, such as telling a story from a wordless picture book, and their responses are analyzed for a variety of pragmatic markers. Narrative and discourse tasks have been able to demonstrate pragmatic impairments in individuals with ASD [70–72], although there has been mixed agreement for the type and extent of impairment [73]. Some of the discrepancy may be explained by how strictly the interpretation focuses on pragmatics or confounds general story-telling, syntax and semantics. Compared with matched controls, individuals with ASD show less effective and less efficient use of cohesive ties of reference, such as the over-use of pronominal expressions when maintaining reference – a type of nonelliptical speech (e.g., 'The boy went to the lake. The boy took his dog.'), and the use of unresolved anaphors leading to ambiguous pronouns (e.g., 'Mike and Henry went to the mall. He really liked it.') [70,74]. Of note, children who have an earlier history of ASD but who no longer meet the criteria for a diagnosis show persistent pragmatic impairments in narrative tasks [75].

One norm-referenced measure of pragmatics is the Test of Pragmatic Language (TOPL [76]). The TOPL is able to capture a wide range of expressive pragmatic abilities through direct assessment, although the standardized nature of the assessment means that children answer hypothetical questions in a structured setting with examiner support. A few studies have demonstrated that children with ASD perform more poorly than their TD peers on the original version of the TOPL [77,78], and their low scores are not accounted for by difficulties with structural language or nonverbal intellectual reasoning [66]. In one study [79], over half of the participants with ASD received a standard score of less than 80 (i.e., >1 standard deviation below the mean) on the TOPL; however, a few participants received scores in the average range, highlighting the variability in abilities of this group, and also the limitations of this structured question-and-answer measure. In 2007, a new version of the TOPL was released (TOPL-2 [80]). Whether the revised TOPL-2 will be a more sensitive measure of pragmatic language in ASD remains an open question.

Given the widespread nature of pragmatic impairments in ASD, they are a natural candidate for investigation in siblings of children with ASD. Despite a strong rationale for investigation, there is a notable lack of systematic research on this topic. We know that many children with ASD display structurally intact language, and yet, the investigation of language in siblings usually only extends to the assessment of language fundamentals. Research in pragmatic abilities in siblings is minimal, due, at least in part to the aforementioned measurement challenges associated with pragmatics. Even research using structured assessments, such as the TOPL or narrative tasks, is lacking. While these measures are appropriate for school-aged children, much of the research examining structural language in siblings has focused on children up to age 36 months. This is a sensitive developmental period where pragmatic abilities are beginning to develop, but there are very few measures of pragmatic language appropriate for this age group. One such measure is the Language Use Inventory [81], a parent-report questionnaire for children as young as 18 months, but the development of additional standardized measures targeting the early development of pragmatic language is warranted.

There has been some limited investigation of the pragmatic abilities of siblings in middle childhood, though only two recent noteworthy studies were uncovered in our review. In 2006, a study by Bishop, Maybery, Wong, Maley and Hallmayer [82] examined the pragmatic abilities of 42 school-aged non-ASD siblings and 46 low-risk controls using the Children's Communication Checklist (CCC-2 [83]), a parent-report questionnaire. Four of the subscales of the

CCC-2 examine pragmatics and four examine structural aspects of language. An advantage of the CCC-2 is that it was designed to capture the variety of pragmatic difficulties that are common along the autism spectrum (along with identifying children with pragmatic language impairment and specific language impairment). There is strong evidence that the CCC-2 is a sensitive measure of pragmatic deficits in ASD [80,84–87], but Bishop *et al.*'s [82] study is the first investigation of the CCC-2 in non-ASD siblings. Surprisingly, no significant group differences were found for the pragmatics scales; however, when looking at the total score of the CCC-2, 24% of the non-ASD siblings of children with ASD scored more than two standard deviations below the controls' mean. These siblings were more likely to have fathers who scored high on the social and communication scales of the Autism Quotient [88], a self-report questionnaire. The authors suggest that the CCC-2 could have utility as a brief screening device for BAP, but further investigation of pragmatic abilities in non-ASD siblings is needed. Given that differences in pragmatic abilities may not be obvious to parents and may be exacerbated by differences in frames of reference (e.g., sibling families have a child with ASD to compare to, while control families do not), direct-observation measures might yield more informative data.

In 2011, Ben-Yizhak and colleagues [64] published the first study of non-ASD siblings of children with ASD that directly assessed both structural language and pragmatics. Siblings from a longitudinal cohort were followed-up in middle-childhood (32 siblings, 38 low-risk controls; 9–12 years old). Structural language and cognitive abilities were evaluated by the CELF-3 and Wechsler Intelligence Scales for Children [89], respectively. By comparing the scores from a subset of items from the ADOS, the authors found evidence that siblings classified as BAP had significantly more pragmatic impairments than siblings classified as typically developing. Although this is a starting point, the use of the ADOS as the measure of pragmatic language has substantial limitations. The authors used items from the ADOS both to create their BAP/non-BAP subgroups, and to create their variable of pragmatic impairment. Extrapolating items from the same measure for both variables reduces the independence of the data, especially given evidence of high intradomain correlations [90], and thus it is hard to interpret the findings of this study. There is very sound rationale for proposing that siblings may be at increased risk for pragmatic language impairment; however, more systematic research is needed to explore this area.

Conclusion & future perspective

In sum, prospective studies of non-ASD siblings clearly highlight language as a core area of concern for these children. In early childhood, non-ASD siblings are at an increased risk for delays in language acquisition, and often do not display the receptive-over-expressive advantage seen in typical development. They are also at an increased risk for ASD traits, but with the current state of the literature, there is variability with respect to whether this includes impairments in pragmatic language. In longitudinal studies where all siblings are assessed by trained staff and/or expert clinicians, the risk levels have been revealed to be higher than previously expected: nearly 20% receive a diagnosis of ASD [2], and in the remaining group, over 20% display language delays, ASD traits, or developmental concerns [26,54,56].

Currently there is relatively sparse research on structural language in non-ASD siblings past age 3, and results in preschoolers and young school-aged siblings have been mixed. Fortunately, the field has seen a substantial increase of interest in non-ASD sibling outcomes in the past few years, and this is likely to continue and expand. We expect the research to grow beyond examination of the infants and toddler developmental stages, into the exploration of language and social-communication outcomes of siblings in middle childhood and beyond. Future studies would benefit from larger sample sizes and rigorous diagnostic procedures for group classification (e.g., [26]). Movement toward a universal definition of the broader autism phenotype (which is often used to characterize subgroups of non-ASD siblings) will facilitate these efforts, as the current state of research involves considerable heterogeneity, from requiring subthreshold ASD traits, to including children with language impairment only.

As children grow older, they meet with increased academic and psychosocial demands, and a host of developing language abilities, beyond lexical and syntactic skill, become increasingly important. This means that a wider range of language abilities can be assessed, from reading and writing to pragmatic language. It is

our hope that future research will further examine whether non-ASD siblings are at increased risk in the development of this array of language abilities, as difficulties in these areas (e.g., reading disability, pragmatic language impairment) have the potential to create significant functional impairment. Given the current dearth of research on pragmatic language in non-ASD siblings of children with ASD, this is a field ripe for further investigation. Future studies would benefit from employing more sensitive and independent direct assessment tools, and gathering information from a wider range of contexts in order to conclude whether non-ASD siblings are at risk for difficulties with the social use of language. It will be essential that future research carefully consider the selection of assessment measures most suited to the characterization of impairments in the social use of language. This endeavor will greatly benefit from the creation of more developmentally attuned measures that capture the breadth of pragmatic language abilities that emerge throughout the course of childhood.

Although many siblings demonstrate a typical developmental trajectory, it is important for researchers, clinicians, teachers and parents to better understand the nature of increased risk for siblings, as early detection of emerging developmental concerns can result in earlier intervention, increased supports at home and at school, and better outcomes. Prospective longitudinal studies of younger siblings of children with ASD have been at the vanguard of research on non-ASD outcomes. As the participants in these ongoing longitudinal studies grow older, the studies will be well-placed to extend our knowledge of the developmental trajectories of these siblings into middle childhood and, importantly, to link early predictors with later language outcomes. Earlier identification of risk for language-related learning challenges can improve access to earlier therapy. Given that language and communication difficulties are amenable to intervention, access to appropriate intervention has the potential to improve outcomes in language development and school achievement.

Finally, with the advent of DSM-5 [4], it will be interesting to see whether the changes in diagnostic criteria will affect research unfolding in this area. Due to the very recent release of the new DSM, all research reviewed in this article followed the DSM-IV nomenclature. In the DSM-5, the diagnoses previously named under the category of Pervasive Developmental Disorder have been collapsed into one, ASD, and the diagnostic criteria have been revised. There has been much speculation as to whether the new ASD criteria will be more or less inclusive, which may have an impact on whom researchers classify as 'non-ASD' siblings or as BAP. Another relevant change in DSM-5 is the addition of Social (Pragmatic) Communication Disorder. This is the first time a disorder relating to impairments in pragmatic language has been included in the DSM. It is possible that this may encourage more research into pragmatic language or aid in creating consistent case definitions for non-ASD siblings who experience functional impairment in the pragmatic domain.

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