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Defining language phenotypes in autism

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Abstract

All children with autism spectrum disorders have deficits in pragmatic aspects of communication; however, formal language abilities are extremely heterogeneous, ranging from nonverbal to superior linguistic skills. Recent studies have focused on defining different language phenotypes among verbal children. One subtype has been compared to specific language impairment (SLI), a language disorder that is diagnosed on the basis of delays and deficits in language acquisition in the absence of hearing impairment, frank neurological damage or co-morbid psychopathology. Two behavioral studies address the question of whether children with autism and language impairment have specific language deficits that are similar to those found in SLI. These experiments focused on phonological processing in a nonsense word repetition task, and use of grammatical morphology in conversational speech. The findings from these studies are discussed in the context of recent neuroimaging and genetic studies of autism.

Keywords: Autism; Language; Phonological processing; Grammar

1. Introduction

One of the hallmark features of autism spectrum disorders (ASD) is wide variability in language skills [1]. Although deficits in pragmatic aspects of communication, specifically the ability to use both literal and non literal language appropriately in a range of social contexts, are universal symptoms even among adults with Asperger syndrome [2,3], formal language deficits are not core features of ASD [4]. [5] did not refer to language problems in his original discussion of the defining features of *infantile autism*; however, later he described some of the idiosyncratic clinical features of language use among the children he had diagnosed, including echolalia, pronoun reversals and stereotyped language [6].

There is growing interest among researchers and clinicians in delineating different subtypes in autism. For researchers, this would create more homogenous groupings that could facilitate finding the underlying causes of autism; for clinicians, subtyping would enhance the ability to tailor treatments to the specific needs of individuals based on their particular pattern of impairments. Rapin and her colleagues [7–9] were the first to describe the different types of language disorders found among children with autism, highlighting some of the similarities between autism and other developmental language disorders. More recently, [10] identified two language subtypes among verbal children with autism: children with normal linguistic abilities (phonological skills, vocabulary, syntax and morphology) and children with impaired language [10] argued that the language phenotype of the autistic children in the latter subtype was similar to the language phenotype that has been described for children with specific language impairment (SLI), a disorder characterized by delays and difficulties in language acquisition in absence of other sensory, cognitive or affective disorders [11]. Like autism, SLI represents a heterogeneous population [12]. There is no agreement among clinicians or researchers on uniform diagnostic criteria, including specific instruments, measures, or cut-off points that define this population. Nevertheless, in recent years, some consensus has been reached about language deficits that may be viewed as central to

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this disorder. Two clinical markers have been highlighted: deficits in phonological processing and grammatical morphology [12]. These clinical markers are now viewed as core features that define the phenotype of SLI, analogous to the social, communicative and restricted behavior patterns that are at the core of an autism diagnosis.

In an initial comprehensive study, [13] investigated language profiles in a large group of verbal children with autism using a battery of standardized language tests tapping articulation, phonological processing, vocabulary, and higher order grammatical and semantic skills. Two subgroups of children with autism were identified, based on their performance on these tests. About one quarter of the children had language test scores that fell within in the normal range (ALN subtype). The remaining children scored significantly below the mean on all the language tests except for the articulation measure. These children had a distinctive profile of performance across the language measures (moderate impairments in vocabulary and phonological processing and more severe impairments in higher order syntax and semantics), suggesting that they formed a subtype of children with autism and language impairment (ALI subtype). Although there was a moderate relationship between language scores and IQ, there were children with high and low IQ scores, and normal or delayed onset of language milestones, in both the ALN and ALI language subgroups. The profile of scores of the ALI children resembled closely the profiles reported for children with SLI [12]. The poor performance of these children on the test of phonological processing, a standardized nonsense word repetition test, was especially significant since, as noted earlier, this kind of measure is considered highly sensitive to the diagnosis of SLI [13,15–17]. However, it was not clear from this initial study whether the ALI children made the same types of errors when repeating nonsense words as do children with SLI, or whether deficits in nonword repetition are related to vocabulary knowledge, as has been found in SLI [17].

In addition to impairments in phonological processing SLI is characterized by grammatical deficits, especially in marking verbs for tense [18,19]. Long after their age-matched peers have stopped making errors, children with SLI continue to omit grammatical morphemes in their speech; for example, marking the past tense (e.g., *She play-ed in the garden*). Studies by Rice and her colleagues showed that for English-speaking children with SLI children's grammatical marking of tense distinguished preschool children with SLI from unimpaired children in both conversational speech as well as on experimental probes [19–21]. Drawing on part of the same sample that had participated in [14] study, [22] investigated grammatical tense marking in children with autism. About half the children had the ALN phenotype and half the ALI phenotype. The children were given experimental probes to elicit past tense and third person present tense (e.g., *A firefighter put-s out fires*), using the same tasks designed by [20]. The ALI children performed poorly on these probes (30% correct on past tense and 37% correct on third person present tense) compared to the ALN children (76% and 64%, respectively). The most common error pattern among the ALI children was to omit any morphological marking on the verb stem, the error that is most frequently reported for children with SLI.

The studies reviewed here provide support for the view that there are parallels in language profiles between a subgroup of children with autism, ALI, and children with SLI; however, it is not clear whether these parallels are misleading in that the language deficits stem from different underlying mechanisms or whether they reflect more substantive overlap in the phenotypes of these syndromes. The following studies were conducted to follow up on the findings from our earlier research on phonological processing and grammatical morphology among children with autism.

2. Study 1: Phonological processing

The goal of this study was to follow up on the findings of [14], who found that children with ALI had low scores on a standardized test of nonsense word repetition (NWR), in order to examine the kinds of errors made by children with ALI and to investigate the whether NWR performance is correlated with vocabulary and other phonological processing tasks in children with autism, as has been found for children with SLI [18].

2.1. Methods

2.1.1. Participants

A group of 35 children (6 girls and 29 boys) aged between 7 and 14 years ($\underline{M} = 10;4$; $SD = 1;9$) participated in this study. All the children met DSM-IV criteria for a diagnosis on autism, which was confirmed on the ADI-R and ADOS – Module 3. The participants were relatively high functioning (average nonverbal IQ score = 83), and were recruited through a variety of community sources (including schools, parent support groups, etc.). Children with Rett syndrome, Childhood Disintegrative Disorder, or with autism-related medical conditions (e.g., neurofibromatosis, tuberous sclerosis, Fragile-X syndrome) were not included in this study.

2.1.2. Procedures

The children were administered a battery of standardized tests to assess IQ, vocabulary knowledge, nonsense word repetition and other phonological processing skills. IQ was assessed using the Differential Ability Scales (DAS), which yields a full scale as well as separate verbal and nonverbal IQ scores. Expressive and receptive vocabulary knowledge was assessed with the Expressive Vocabulary Test and the Peabody Picture Vocabulary Test. Three subtests of the Comprehensive Test of Phonological Processing (CTOPP) were administered to assess memory for digits, rapid automatic naming, and NWR. For the NWR subtest, all 18 items were administered to the children, even if they reached ceiling before the final items on the test, in order to provide a full set of stimuli on which to carry out error analyses. The children's performance on the NWR subtest was taped and later transcribed by two experts in phonetic transcription. These transcripts were used to explore the types of errors made by the children.

2.2. Results and discussion

In this sample of 35 children, 15 children scored in normal or borderline range on the NWR (Standard Score of 7 or higher) and 20 children were significantly impaired (SS of 6 or lower). Based on their performance on NWR and other language test scores obtained from these children, these children were designated as having the ALN and ALI phenotypes.

Across both groups of children, performance on NWR was significantly correlated with rapid automatic naming and expressive vocabulary, after partialing out non-verbal IQ scores ($r(32) = .44$, $p < .01$ and $r(32) = .50$, $p < .01$, respectively). Thus, NWR is closely related to expressive fluency (rapid automatic naming), as well as to expressive vocabulary knowledge.

The error analyses of the phonetic transcripts of the children's responses revealed that all the children, including the ALI subgroup, repeated the correct number of syllables (ranging from 1 to 5) for the stimulus items (98% correct for ALN and ALI subgroups). Errors were classified into two groups: phoneme deletion and phoneme substitution. For all children, substitution errors were more prevalent; however, for the ALI children, 20% of their errors involved phoneme deletion, compared to 11% of the errors made by ALN children.

This study confirmed the presence of phonological processing deficits among children with ALI. As in SLI, the children with ALI made more deletion errors than the ALN group, whose performance was equivalent to typically developing children. There was also a close relationship between phonological processing and vocabulary. This replicates SLI findings by [17] who hypothesized that previous vocabulary knowledge enhances phonological short-term memory processes, and therefore repetition accuracy. Taken together, these findings mirror what has been reported in the literature for children with SLI [16], and underscore the similarities in phonological processing deficits in a subgroup of autism, children with ALI.

3. Study 2: Grammatical morphology

The goal of this study was to follow up on the experiment study by [22] who found that children with ALI tended to omit tense morphology on experimental tasks that were designed to elicit sentences that included verbs marked for either third person present singular or past tense. Most studies investigating grammatical morphology deficits in SLI have relied on conversational speech samples to explore grammatical deficits in everyday settings. These studies have found that younger children with SLI omit both third person present tense (e.g., *she sing-s*) and past tense (e.g., *he walk-ed*) morphemes in sentence contexts in which they are required to be marked [19]. For children with SLI, grammatical morphology deficits are specific to aspects of tense-related morphemes and no impairments are found on other kinds of morphemes (e.g., noun-related). In this study, children with autism (both ALI and ALN) were compared to children with SLI in their use of grammatical morphology in conversational speech.

3.1. Methods

3.1.1. Participants

This study included 29 children with autism, divided into ALN ($N = 9$) and ALI ($N = 20$) subtypes, based on their performance on standardized language tests, and a comparison group of 13 children with SLI. The children with autism were diagnosed in the same way as those in Study 1 and the same exclusionary criteria were applied. The children with SLI all had a documented history of language delay and impairment, and scored more than 1 standard deviation below the mean on standardized language tests. The three groups were matched on age (average age = 7 years 5 months). The ALI and SLI children were also well matched on nonverbal IQ, using the DAS (average nonverbal IQ = 89 for ALI and 90 for SLI) and mean length of utterance (MLU), which is an index of productive language based on conversational speech (average MLU for ALI = 3.0 and for SLI = 3.06). The ALN group had significantly higher nonverbal IQ (average IQ = 116) and MLU (average MLU = 3.88) scores than the other children.

3.1.2. Procedures

Conversational speech samples were taped during a parent-child play interaction and an examiner-child interaction that was part of the administration of the ADOS. The speech samples were later transcribed using standard procedures.

Following the methods of earlier studies [19], the transcripts were coded for two sets of grammatical morphemes: verb morphemes (third-person singular present -s, past tense -ed) and control noun-related morphemes (plural -s, prepositions in/on). Each child utterance was first coded for whether it included obligatory contexts for any of these target morphemes. Following this, all obligatory contexts were then coded for whether the correct target morpheme was supplied by the child.

3.2. Results and discussion

The average percent correct use of each of the coded morphemes, defined as the percent supplied in obligatory contexts, by each group is presented in Table 1. As can be seen, performance across all the groups was high for the noun morphemes: all were well over 90% correct, which

Table 1
Mean percentage of use of verb and noun morphology in obligatory contexts

	ALN	ALI	SLI
Verb morphemes			
Third person present	92	85	74
Past tense	95	91	100
Noun morphemes			
Plural	99	100	97
Prepositions	100	94	97

is generally taken as an indicator of mastery of a grammatical structure. For the verb morphemes, all three groups were above 90% on the past tense; however, the ALI and SLI groups were lower on the third person present tense.

The performance of the ALI and SLI children in this study was generally high – most had mastered the use of grammatical morphemes in conversational speech. This is not surprising given the relatively older ages of the subjects compared to the ages of children with SLI who had been investigated, for example, by [19] or by other research teams. In more recent studies, [23] reported that by age 8, most children with SLI have finally caught up with their unimpaired peers who had mastered tense morphology by age 4. Interestingly, in this study, both the ALI and SLI children are still showing some difficulty with the third person present tense. These children had no difficulties using the control noun-related morphemes, confirming the specificity of the lingering grammatical deficits to verb morphology, particularly third-person singular -s. Across all the morpheme analyses, the children with SLI were indistinguishable from the ALI children.

4. General discussion

The main findings from both these studies confirmed earlier research indicating that in autism, there are different language phenotypes – here referred to as ALN and ALI – reflecting subgroups in which structural, rather than pragmatic, aspects of language are quite different. Thus, we found group differences in the first study in the distribution of errors on a nonword repetition task, and in the second study in the marking of third person present tense morphology. Moreover, these studies provide further support for the hypothesis that the language phenotype of the ALI subtype is the same as the phenotype for SLI. These studies complement other research which has found that not only are there similarities in the linguistic deficits found in ALI and SLI, but these groups also show the same pattern of impairments in aspects of language use that extend into pragmatics, including the ability to use context to process ambiguous words [24], narrative production [25] and comprehension [26] and general communicative impairments as measured on a parental checklist [27].

The behavioral findings on similarities in the phenotypes of autism and SLI have been extended to investigations of brain morphology. For example, one aspect of brain morphology that has been the focus of several studies is hemispheric asymmetry in language regions. In the majority of non-language impaired right-handed individuals, the inferior frontal region in the left hemisphere (Broca's area) is larger than in the right hemisphere [28]. Similar left hemisphere asymmetry is also found in the more posterior *planum temporale* [29]. Studies of children with SLI show reduced or reversed asymmetries in the frontal regions corresponding to Broca's area [30,31].

A few studies have investigated hemispheric asymmetry in children with autism and children with SLI. Herbert and her colleagues compared boys with autism to matched normal controls [32]. They found that the autistic boys had significant reversed asymmetry in the inferior frontal cortex, which was 27% larger in the right hemisphere compared to 17% larger in the left hemisphere for the normal controls. There were also significant differences between the autism and control groups in the asymmetry patterns in the *planum temporale*. While both groups showed a left hemisphere asymmetry, this was more extreme in the autistic boys (25% leftward asymmetry for autism compared to 5% in the controls).

These findings were replicated by [33]. In this study, the boys with autism were divided into ALN and ALI subgroups and were compared to matched groups of boys with SLI as well as normal controls. De Fosse et al. found that atypical asymmetry patterns (right asymmetry) in inferior frontal cortex; more extreme left asymmetry in *planum temporale* were found for the ALI and SLI groups; however, the ALN group showed the same asymmetry patterns as the control children.

The behavioral evidence and the neuropathological findings presented here support the claim that there is a subtype among children with autism who have a language phenotype that is the same as in SLI [10]. The similarities in the phenotypes of ALI and SLI may also suggest some shared aspects of etiology, particularly genetics. Both SLI and autism are considered to be disorders with a relatively strong genetic basis [34]. They are both disorders with heterogeneous phenotypes that are likely to have a complex genetic basis. Evidence from family and twin studies shows that SLI clusters in families and has a high heritability rate [35–37]. Similar findings have been obtained for autism. Twin studies suggest a heritability rate of over 90% for autism [38]. Numerous studies have demonstrated that parents and siblings of children with autism are more likely to exhibit traits that are conceptually similar but milder in form to the symptoms that define autism, known as the broader autism phenotype [39].

Behavior genetic studies have found interesting and significant overlap between families with an autistic child and families with an SLI child. Studies of the broader autism phenotype have found that among first degree relatives of children with autism, there are elevated rates of histories of language delay and language-based learning deficits [40,41], indicating that in families identified on the basis of a proband with autism, there are unusually high rates of language impairment. Similarly, one study of families identified on the basis of a proband with SLI found elevated rates of autism among the siblings compared to the base rate in the population [42]. These studies suggest familial co-morbidity for autism and SLI, providing evidence for the hypothesis that there is some shared genetic etiology for these disorders.

Although at this time no specific risk genes have yet been found for either autism or SLI, several genetic studies of autism have incorporated information about the language phenotype of probands in their genetic analyses. For example, when the Collaborative Linkage Study of Autism (CLSA) restricted their genetic analyses to a subgroup of probands with autism who had no language or clearly impaired language, linkage signals on both 7q and 13q were significantly increased, suggesting that these signals might be attributable to the language impaired subtype within autism [43]. Similar findings for a locus on 7q were obtained by Alarcon and colleagues using the Autism Genetic Resource Exchange (AGRE) families [44], and more recently for a locus on 17q [45]. These genetic findings hold out some promise that defining language phenotypic subtypes within the autism population may provide important benefits to genetic studies.

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