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Review

The phenotype and neural correlates of language in autism: An integrative review

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ABSTRACT

Although impaired communication is one of the defining criteria in autism, linguistic functioning is highly variable among people with this disorder. Accumulating evidence shows that language impairments in autism are more extensive than commonly assumed and described by formal diagnostic criteria and are apparent at various levels. Phenotypically, most people with autism have semantic, syntactic and pragmatic deficits, a smaller number are known to have phonological deficits. Neurophysiologically, abnormal processing of low-level linguistic information points to perceptual difficulties. Also, abnormal high-level linguistic processing of the frontal and temporal language association cortices indicates more self-reliant and less connected neural subsystems. Early sensory impairments and subsequent atypical neural connectivity are likely to play a part in abnormal language acquisition in autism. This paper aims to review the available data on the phenotype of language in autism as well as a number of structural, electrophysiological and functional brain-imaging studies to provide a more integrated view of the linguistic phenotype and its underlying neural deficits, and to provide new directions for research and therapeutic and experimental applications.

Contents

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Abbreviations: AS, Asperger syndrome; ASD, autism spectrum disorder; DTI, diffusion weighted imaging; ERP, evoked response potentials; fMRI, functional magnetic resonance imaging; HFA, high-functioning autism; MEG, magnetoencephalography; MMN, mismatch negativity; PDD-NOS, pervasive developmental disorder not otherwise specified; SLI, specific language impairment; ToM, theory of mind; WCC, weak central coherence.

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1. Introduction

Autism is a neurodevelopmental disorder associated with lifelong handicaps in social adaptation. The disorder is defined by a description of symptoms in three domains: social interaction, verbal and non-verbal communication and stereotyped behaviour (American Psychiatric Association, 1994). Autism is currently considered to be one of the autism spectrum disorders (ASD), which also include the Asperger syndrome (AS) and pervasive developmental disorder not otherwise specified (PDD-NOS). The term *spectrum* refers both to the heterogeneity of the ASD disorders and the wide variety in functional ability. Although not universally agreed upon, the definition of AS provided in DSM-IV entails impaired social interaction and stereotyped behaviour in the absence of language and cognitive delay (Klin et al., 2005). Individuals with PDD-NOS fail to meet the full criteria for autism. This is the case when the number of criteria met is sub-threshold, or the onset occurs over the age of 3, or atypical symptoms are present, or a combination of the above (Buitelaar and van der Gaag, 1998; Buitelaar et al., 1999).

Linguists commonly describe language and language disorders in terms of phonology, semantics, syntax and pragmatics, and since the early seventies, researchers have been analysing linguistic deficits in autism using these categories (Cromer, 1981). Whereas phonology deals with the perception and production of sound units whose concatenation produces words, semantics deals with the meaning of lexical items, syntax with the structure of words in sentences, and pragmatics with the conventions and rules governing the use of language for communication (Boucher, 2003). From a neuroscientific perspective, however, there is no clear-cut relation between linguistic categories and cortical function. Recent functional imaging studies have furthered the idea that the language system is organised in a large number of small but tightly clustered modules in both the left and right hemisphere with unique contributions to language processing. There is also increasing evidence that cortical language regions are not specific to language, but involve more reductionist processes that give rise to language as well as non-linguistic functions (Bookheimer, 2002).

Given the absence of a clear-cut relation between the linguistic categories and cortical function, it is unfeasible to deduce neurobiological deficits in autism from the high-order language deficits characterising the disorder. Instead, several psychological theories, such as the weak central coherence (WCC) theory (Frith, 1996) and the impaired theory of mind (ToM) (Baron-Cohen et al., 1985), have attempted to explain the high-order language deficits in autism. The weak central coherence theory predicts that, since people with autism are biased towards local versus global processing, their ability to integrate contextual information into a composite whole is diminished. The high-order core deficit in central processing supposedly results in altered low-level processing. Several studies have indeed demonstrated a reduced ability to infer word-meaning from sentence context (Happe, 1997) or to infer global meaning from sentences (Jolliffe and Baron-Cohen, 2000), yielding empirical evidence for the WCC account for at least the semantic and pragmatic language deficits in autism. However, WCC would also predict a superior performance on single word tasks, as is the case in hyperlexia. Yet, hyperlexia is only rarely seen in autism. The majority of people with autism have difficulties with the meaning of isolated words as well as whole sentences.

ToM refers to the specific cognitive ability to infer other people's mental states and to understand that others have beliefs, desires and intentions that are different from our own. It has been argued that early stages of ToM are necessary for the ability to use symbols such as words (Tager-Flusberg, 2000), and that impairment in ToM in autism therefore causes an inability to comprehend the meaning of words. Furthermore, acquisition of language may be mediated by shared or joined attention, which, in case of an impaired ToM, would be impaired as well (Kuhl et al., 2003). Semantic ability and false belief have indeed been found to correlate in children with autism (Tager-Flusberg, 2000).

The psychological framework provided by these top-down theories assumes that an impaired high-level cognitive function is causing the impairments in autism. This assumption has been criticised for several reasons. Firstly, converging evidence suggests that abnormalities of the processing of low-level sensory information may lead to impairments in higher-order cognitive functions, rather than the other way around (Happe and Frith, 2006; Bertone et al., 2005). That is to say, altered low-level perceptual processing in autism should not be considered a by-product of weak central coherence. Quite on the contrary, perceptual abnormalities give rise to weak central coherence. Secondly, these neuropsychological top-down theories are descriptive rather than explanatory, and finding the neural correlates of these theories has been proven difficult since their predictions of cortical functioning are too general to be falsifiable.

Nevertheless, there have been converging efforts from different disciplines to document the neural correlates underlying the symptoms of ASD (Volkmar et al., 2004). Although most of the research has focused on impairments in social cognition (mostly using visual stimuli such as faces), language impairments in autism are increasingly recognised. More and more findings on the clinical phenotype and the neural substrates of language and communication in autism are being added to the literature, but the broad field of autism research and the many different analytical approaches make it difficult to oversee the current literature. This paper therefore aims to review recent evidence from structural, electrophysiological and functional studies on the neural correlates of linguistic abnormalities in autism. Findings on the phenotype of language impairments in autism will also be addressed here. We will argue that the linguistic features in autism cover a wider range of impairments than described in the DSM-IV criteria for autistic disorder and are more linked to the neural architecture in autism than earlier behavioural studies have suggested. More specifically, language ability varies greatly among people with autism. Although most individuals with autism have semantic, syntactic and pragmatic language deficits, there is also a number that have phonological difficulties. Functional brain-imaging data show aberrant neural activation in semantic, syntactic and pragmatic tasks of higher-order language functions, as well as in low-level sensory processes. Furthermore, we will argue that the abnormalities of low-level sensory processing of linguistic stimuli can be interpreted in the light of connectivity models in autism. Finally, we will discuss the relationship between language impairments and the other functional impairments in autism (social interaction and stereotyped and rigid behaviour patterns), as well as the relationship between autism and specific language impairment (SLI), to provide a more integrated view of the linguistic phenotype and its underlying neural deficits. An integrated review may be useful for both clinicians and researchers, as it will allow further congruity between the observed language deficits and their putative causes and could lead to important therapeutic and experimental applications in the future.

2. Methods

We conducted an extensive internet search of the English literature published on the MEDLINE and PsycInfo databases in the past two decades, using the keywords (autism, autistic disorder, and Asperger syndrome) AND (language, language disorders,