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Differential language markers of pathology in Autism, Pervasive Developmental Disorder Not Otherwise Specified and Specific Language Impairment

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ABSTRACT

Language impairment is a common core feature in Pervasive Developmental Disorders (PDD) and Specific Language Impairment (SLI). Many studies have tried to define the specific language profiles of these disorders, some claiming the existence of overlaps, and others conceiving of them as separate categories. Fewer have sought to determine whether and how PDD-NOS language profile, including prosody, differs from those of Autistic Disorder (AD) and SLI. Here, 12 children with AD (mean age 9.75; sd 3.5), 10 with PDD-NOS (mean age 9.83; sd 2.17), and 13 children with SLI (mean age 9.17; sd 3.9) matched for age, sex and academic skills were explored for both receptive and expressive language skills. Prosody was also assessed with an intonation imitation task analyzed through automatic speech processing and compared to 70 typical developing controls matched for age and sex. A similar delay in phonology and vocabulary was observed in the three groups as were significant but variable differences between the groups in syntax, pragmatics and prosody. SLI showed correlations between chronological age and raw scores in all language tasks, while AD and PDD-NOS did not. Furthermore, SLI showed correlation within all raw scores in language tasks. Most of those correlations were also found in PDD-NOS but not in AD. In conclusion, these findings support the hypothesis that language skills in AD and SLI rely on different mechanisms, while PDD-NOS show an intermediate profile sharing some characteristics of both AD and SLI. They also suggest that expressive syntax, pragmatic skills and some intonation features could be considered as language differential markers of pathology, challenging the DSM-V proposal of broad criteria.

1. Introduction

Pervasive Developmental Disorder (PDD) is characterized by a triad of severe deficits and pervasive impairments in developmental areas such as reciprocal interactions, communication skills and stereotyped behavior. PDD is mostly seen as a "spectrum disorder" with several variants and gradients. DSM IV (APA, 2000) distinguishes three main disorders: Autistic

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Disorder (AD) with early onset, possible mental retardation, language impairment and symptoms in all areas characterizing PDD; Asperger Syndrome (AS) without language delay or mental retardation; and Pervasive Developmental Disorder Not Otherwise Specified (PDD-NOS) in which social, communicative and/or stereotypic impairments either do not coexist, or to a lesser degree resulting in that they do not fit other subtypes of PDD. Thus, diagnostic criteria for PDD-NOS are quite vague and mostly lead to a diagnosis by default (Volkmar, 2005) while being statistically the largest category (Fombonne, 2003).

1.1. Language in PDD

Language and communication impairment in PDD, especially in AD, has led to numerous studies over the last decades trying to specify profiles. Language in autism, when present, may show several varying subtypes within the spectrum (Kjelgaard & Tager-Flusberg, 2001; Rapin & Dunn, 1997; Tager-Flusberg, 2006). Some individuals may have *structural language disturbances:* (i) delayed phonology, especially in non-word repetition tasks (Bartolucci, Pierce, Streiner, & Tolkin Eppel, 1976; Kjelgaard & Tager-Flusberg, 2001; Tager-Flusberg, 1981; Whitehouse, Barry, & Bishop, 2007); (ii) poor comprehension skills, sometimes more impaired than the expressive ones (Bartak, Rutter, & Cox, 1977; Boucher, 2003; Tager-Flusberg, 1981), (iii) immature syntax and prevalence of syntactic errors (Kjelgaard & Tager-Flusberg, 2001). Functional deficits are characterized by: (i) a core pragmatics disorder (defined as the ability to use and understand the rules governing language as a communicative tool including tone of voice, facial expressions, communicative gesture and affect), accepted as universal in the whole spectrum and long-lasting, even in adult life (Lord & Paul, 1997; Rapin & Dunn, 1997, 2003); (ii) impairment regarding semantics, i.e. the linguistic meaning of utterances and bounds established between words/utterances and what they do/may represent (Boucher, 2003; Rapin & Dunn, 1997, 2003).

1.2. Overlapping with SLI?

Language was compared persons with autism and SLI participants. SLI, which excludes PDD, is defined as a "pure" impairment despite adequate hearing, intelligence or physical condition, encompassing a broad range of deficits regarding phonology, vocabulary, syntax, semantics and pragmatics. Rapin and Allen (1983) distinguished: (i) mixed receptive/expressive disorders, with comprehension and expression impairment, including verbal-auditory agnosia (word deafness) and phonologic-syntactic syndrome; (ii) expressive disorders, with spared comprehension, including verbal apraxia and phonologic disorder; (iii) higher order disorders, with spared articulation and phonology but disordered pragmatics and lexical-syntactic organization. This last category is controversial. Some authors consider it as a part of the ASD continuum while others refuse the systematic equation between pragmatic difficulties and ASD and identify intermediate characteristics (Bishop & Frazier-Norbury, 2002). Some studies conclude that the *structural disorders* observed in some AD children are similar to those of SLI (especially regarding phonology and syntax) and suggest overlaps between the disorders, hypothesizing a common phenotype (Bartak, Rutter, & Cox, 1975; Bartak et al., 1977). Conversely, studies focusing on non-word repetition, considered to be a psycholinguistic marker of SLI (Bishop, North, & Donlan, 1996; Conti-Ramsden, Botting, & Faragher, 2001), found similarities in AD and SLI regarding syntactic reception and expression, but differences in speech motor skills, verbal short-term memory, and error types in non-word repetition (Whitehouse, Barry, & Bishop, 2008). They thus rejected the hypothesis of an SLI subtype in Autism.

Some genetic studies suggest that the two conditions may be related (Folstein & Mankoski, 2000; Vernes et al., 2008) although others discuss an overlapping etiology and "phenomimicry" (Bishop, 2003, 2010). Moreover, although studies regarding ASD and SLI language and communication in first-relatives are consistent with the hypothesis that both disorders are highly inheritable, they do not support the hypothesis of one shared phenotype but rather suggest that language deficits in ASD and SLI have different origins (Lindgren, Folstein, Tomblin, & Tager-Flusberg, 2009; Whitehouse et al., 2007). A recent fMRI study found that SLI and Autistic children with Language Impairment (ALI) compared to normal controls showed abnormalities in the circuits that manage motor control and processing of language, cognition, working memory and attention. They also observed that cerebellar white matter was significantly larger in ALI than in SLI as well as regional volume differences between ALI and SLI groups in right hemisphere VIIA Crus I, suggesting similarities but also important language related developmental differences (Hodge et al., 2010).

1.3. Prosody in PDD

Prosody concerns the supra-segmental properties of the speech signal that modulate and enhance its meaning. It supports expressive language at several communication levels (McCann & Peppé, 2003). Abnormal prosody is identified as a core feature of individuals with autism (Kanner, 1943). The observed prosodic differences include monotonic or machine-like intonation, aberrant stress patterns, deficits in pitch and intensity control and a "concerned" voice quality. These inappropriate patterns related to communication/sociability ratings tend to persist over time even while other language skills improve (Paul, Augustyn, Klin, & Volkmar, 2005; Paul et al., 2005). Many studies have tried to define the prosodic features in patients with Autism Spectrum Disorder (ASD) (for a review see McCann & Peppé, 2003). With regards to intonation contours production and intonation contours imitation tasks, results are contradictory. In a reading-aloud task, Fosnot and Jun (1999) found that AD children did not differentiate questions and statements; all utterances sounded like