The handle http://hdl.handle.net/1887/19149 holds various files of this Leiden University dissertation.

Author: Maljaars, Janne Pieter Nella Wilhelmina
Title: Communication problems in children with autism and intellectual disability: Depicting the phenotype
Date: 2012-06-26
General introduction
General introduction

Children with autism spectrum disorder (ASD) and co-occurring intellectual disability (ID) are distinctively different from children with ASD or ID alone. The co-occurrence of ASD and ID is known to cause severe problems for the child, the parents, and other caregivers and is a strong predictor of poor prognosis (Boucher et al., 2008). There is a need for a better understanding of the challenges and impairments which characterize the co-occurrence of these disorders (Matson & Shoemaker, 2009). Communication problems are one of the core symptoms of ASD. Children with ASD and co-occurring ID form a particularly vulnerable group regarding communication problems. Severe communication problems evidently influence the child’s development and the child-rearing situation. Impairments in communication can lead to challenging behavior (Bott et al., 1997; Chamberlain et al., 1993; Sigafoos, 2000), and place a heavy burden on parents (Bebko et al., 1987). Valid and reliable diagnostic instruments and knowledge about strengths and weaknesses in communicative behavior are important to improve assessment and intervention for these children and their families.

Autistic disorder and intellectual disability

Autism spectrum disorders (ASDs) are lifelong, complex neurodevelopmental disorders, encompassing a wide spectrum of developmental disorders characterized by impairments in three behavioral domains: social interaction, communication, and range of interests and activities. In this thesis, the term ‘autism’ will be used to refer to autistic disorder (AD) specifically, whereas the abbreviation ASD will be used to refer to the broader spectrum of autistic disorder, Asperger syndrome, and Pervasive Developmental Disorder Not Otherwise Specified (PDD-NOS or atypical autism). The terms autism and autistic disorder are used interchangeably depending on journal policies. ASD corresponds to pervasive developmental disorder (PDD) as described in the Diagnostic and Statistical Manual of Mental Disorders, fourth edition – text revision (DSM-IV-TR; APA, 2000) and the International Classification of Diseases, tenth revision (ICD-10; WHO, 1992). Most of the literature used in this dissertation is based on the broader ASD, but given the classification difficulties in low-functioning children, the present study focused on AD, the core syndrome instead of the whole spectrum in order to create a more homogeneous group. According to the DSM-IV-TR (APA, 2000) AD is characterized by severe and pervasive impairments in social interaction and communication skills, and by the presence of restricted and stereotyped patterns of behavior, interests, and activities with onset prior to the age of three years. Criteria for these impairments are based on both quantitative (absence of normative, developmental milestones) and qualitative differences (presence of aberrant behaviors) from typical development (Klin et al., 2005). Each of these core features have to be present in order to diagnose AD. However, the clinical manifestations and severity of
symptoms of AD are often very heterogeneous (Beglinger & Smith, 2001; Hill & Frith, 2003).

Estimates of the prevalence of ASDs have gradually increased over the past decades. The estimates for ASDs from recent studies have been considered to be just over 1% (Harrington, 2010; Lord, 2011), and the average prevalence for AD is approximately 0.2% (Fombonne, 2009). Furthermore, ASDs are four times more common in males than in females, although some recent studies are questioning this ratio (Gould & Ashton-Smith, 2011; Kopp & Gillberg, 2011). Although ASDs are considered to be among the most heritable developmental disorders, the underlying genetic factors remain largely unknown (Glaser et al., 2010; Noor et al., 2010). Recent studies have led to the identification of several autism susceptibility genes and assessment of copy number variation has discovered numerous potentially important novel candidate loci (Abrahams & Geschwind, 2008). Many of these genes and genomic loci have been associated with ID, which suggests that both developmental disorders share a common genetic etiology (Betancur, 2011; Noor et al., 2010).

Epidemiological studies report varying percentages for the rate at which ID is present in individuals with ASD, ranging from about 15% to 70%, depending on diagnostic criteria and level of functioning (Bhaumik et al., 2010). The highest prevalence rates of ID are found in the more strictly defined group with AD (Charman et al., 2011). Furthermore, increased severity of ID is positively correlated with incidence of AD (Vig & Jedrysek, 1999). Conversely, about 3 to 50% of individuals with ID have ASD (De Bildt et al., 2004b). ASDs are one of the most frequently co-occurring disorders in individuals with ID (Matson & Shoemaker, 2009). ID is defined by significant limitations in intellectual functioning and adaptive functioning (two or more standard deviations below the population mean) (APA, 2000; Schalock et al., 2010). Particularly for research purposes, classification of the level of severity of ID is often based on IQ-scores only. Mild ID is defined by an IQ range of 50-55 to approximately 70, moderate ID by 35-40 to 50-55, and severe to profound ID by an IQ below 35-40 (APA, 2000).

Making an accurate diagnosis of ASD in ID is difficult, especially in those with severe or profound ID (Bhaumik et al., 2010; Vig & Jedrysek, 1999). The main reason for this is the overlap in behavioral characteristics between children with ASD and children with ID. Especially, the criteria formulated as quantitative impairments, i.e. absence of normative, developmental milestones, are difficult to apply in low-functioning children. Delays in social and communicative development and absence of specific behaviors may also be explained by low levels of cognitive functioning (Bhaumik et al., 2010). This makes some diagnostic criteria from the DSM-IV-TR difficult to apply. Individuals with severe or profound ID, with ASD as well as without ASD, may have no or limited expressive language abilities. In these cases, diagnostic criteria concerning atypical use of language and impairments in conversations are not applicable. Since symbolic abilities typically emerge just around 18 to 24 months (Thiemann-Bourque et al., 2011), symbolic play can also be absent due to cognitive level of functioning. Furthermore,
stereotyped and repetitive behaviors can also be present in individuals with ID without ASD (Bodfish et al., 2000; Bradley et al., 2004). In addition, common medical comorbidities in the ID population, such as deaf blindness, epilepsy, or cerebral palsy, may obscure the clinical presentation of ASD symptoms, and can result in a limited behavioral repertoire and restricted possibilities to express themselves (Bhaumik et al., 2010). Nevertheless, there are also clear distinct symptoms present in individuals with ID and co-occurring ASD compared to individuals with ID without ASD, mainly in severity of stereotyped and repetitive behaviors and relative delay in social and communicative development regarding their general level of functioning (Matson et al., 2008). In the case of ID without ASD, social and communicative skills are usually corresponding with the child’s overall developmental level (Klin et al., 2005). The combination of ASD and ID results in a mutual aggravation of the impairments involved in both disorders and has consequences for the well-being of the individuals themselves and their families (Boucher et al., 2008). A correct diagnosis is important to choose the most indicated intervention strategies. Individuals with ID and ASD need to be treated in a different way than individuals with ID without ASD (Matson & Shoemaker, 2009). Although ASD and ID do co-occur, research in ASD is generally focused on individuals with intelligence in the normal range. An IQ below 70 or even 85 is frequently used as an exclusion criterion for autism research. Therefore, relatively little is known about children with ASD and co-occurring ID.

**Communication in low-functioning children with autistic disorder**

Impairments in communication comprise an important dimension of the ASD phenotype and involve both verbal and nonverbal communication problems (Landa, 2007; Weismer et al., 2010). Communication and language skills are extremely variable in individuals with ASD (Kjelgaard & Tager-Flusberg, 2001; Thurm et al., 2007). A significant part of mainly low-functioning individuals with ASD do not develop language or only a limited number of communicative words or gestures (Boucher et al., 2007). Particularly in case of severe or profound ID, often no verbal or symbolic communication is present. These individuals communicate through idiosyncratic gestures, vocalizations, or other behaviors (Snell, 2002). When children with AD do not gain language by the age of 6, they frequently remain nonverbal (Prior & Ozonoff, 1998). The levels of language development and communicative competence acquired by children with ASD have been found to be important predictors of later outcome in terms of adaptive functioning, intelligence, academic achievement, and level of independence in adulthood (Lord et al., 2004). Several studies in individuals with co-occurring ID reported that receptive communication is overestimated by caregivers of low-functioning individuals, resulting in a too high level of approach (Barlett & Bunning, 1997; Bradshaw, 2003; Cascella, 2004; McConkey et al., 1999). Overestimation of communication skills has consequences for quality of life, learning potential, and level of independence (Barlett & Bunning, 1997; Bradshaw, 2002). Communication problems
in children with ASD and ID are severe, resulting in increased caretaker demands, and may lead to stress in parents and professionals (Lecavalier et al., 2006). In addition, a relation between communication problems and the presence of challenging behavior is evident. Previous studies showed that problem behaviors increase in frequency, intensity, and duration, when communication problems are more severe (Bott et al., 1997; Chung et al., 1995; Van Berckelaer-Onnes et al., 2002). Despite these disconcerting results, relatively little is known about communicative abilities and impairments in children with ASD and ID, in particular in children who are either nonverbal or minimally verbal (Tager-Flusberg et al., 2005).

Exploring patterns in communicative strengths and weaknesses can contribute to a better understanding of the nature of these problems in low-functioning children with ASD. Moreover, it may lead to recommendations for assessment, development of more effective interventions, and suggestions about which children may benefit from specific intervention approaches. However, most standardized assessment measures focus primarily on formal aspects of language, which can only provide information on a limited number of aspects of communication for children with ASD and therefore are not sufficient, especially for low-functioning children with ASD (Lord & McGee, 2001; Twachtman, 1995). Communication problems in ASDs are often examined in a narrow way by focusing on the level of expressive language and abnormalities in language use. There is a need for both qualitative and quantitative information on a broad range of communicative skills. When assessing communication in children with ASD, it is useful to combine standardized and norm-referenced instruments with more descriptive and qualitative observational procedures to be able to identify developmental discontinuities and provide complementary information about more subtle differences between children (Klin et al., 2005). Assessment of communication can be divided in three important aspects, namely communicative functions, forms, and content. By integrating information gathered from the assessment of competences and impairments in the use and / or understanding of function, form, and content of communication, it should be possible to outline a picture of overall communicative competence (Twachtman, 1995).

A communicative function refers to the intention, goal, or purpose communication is used for. With respect to functions of communication, this study is confined to the most basic functions, because of our focus on low-functioning children. These functions concern communicative acts for behavior regulation, social interaction, and joint attention. Usually, intentionality in communication develops during the first year of life and is an important precursor for language development (Travis & Sigman, 2001; Watt et al., 2006). Children with ASD have problems in the development of intentionality or pragmatics, but severity can differ from no intentional communication of any kind to problems in complex pragmatic functions, such as using sarcasm or the intention to deceive another person (Twachtman, 1995).
Several forms or means can be used for communication, of which language is the most common and conventional form. Language involves a complex communication system that is based on a set of rules relating symbols to referents (Twachtman-Cullen & Twachtman-Reilly, 2007). The most basic intentions, such as behavior regulation and joint attention, can be communicated by either non-symbolic or symbolic communication forms. Language or other symbolic communication systems are required to express more complex communicative functions (Coupe O’Kane & Goldbart, 1998). Children with ASD have been found to use fewer vocalizations (Tager-Flusberg et al., 2005) and develop language at a slower pace compared with typical development (Eigsti et al., 2011; Weismer et al., 2010). Furthermore, the use of gestures is also limited. They often use only primitive presymbolic motoric gestures, such as pulling or manipulating another’s hand, and lack the use of more conventional gestures, such as waving, pointing and nodding one’s head (Lord & McGee, 2001; Shumway & Wetherby, 2009). The absence of conventional communicative means often leads to the development of idiosyncratic or undesirable behaviors, such as automutilation and aggression, to communicate (Wetherby et al., 2000).

Content of communication refers to the meaning of words or other communicative forms that is conveyed. Noens and Van Berckelaer-Onnes (2005) conceptualized communication problems in ASDs as a problem in sense-making or the perception of meaning. Sense-making can take place at different levels: sensation, presentation, representation, and metarepresentation (see Chapter 5 for a more detailed explanation). The levels of sensation and presentation refer to non-symbolic levels of understanding of communication. At these non-symbolic levels, children do not comprehend that a word or a picture can refer to something else. At the levels of representation and metarepresentation, the symbolic or referential meaning of communicative forms is understood (Noens & Van Berckelaer-Onnes, 2005). Children with ASD have difficulty acquiring conventional and symbolic aspects of communication. The use and comprehension of symbols require an understanding of the relationship between a symbol and its referent. A symbol stands for and is separate from its referent (Bates, 1979). Symbols enable people to exchange information efficiently by representing something else even beyond the here and now (Namy, 2005). During the first few years of life most children achieve important milestones in symbolic development (DeLoache, 1995).

In summary, assessment of communication in children with ASD should determine the communicative functions a child is currently expressing as well as the means, i.e. the repertoire of behaviors and level of language, by which the child attempts to do so. It is also important to assess the level of language comprehension in order to provide information which parents and professionals can use to adjust their communication level towards the child with ASD. Furthermore, assessment of the level of symbolic development or sense-making is of critical importance in planning intervention when assessing low-functioning children with minimal or no language.
**Objectives and overview of the dissertation**

The main objective of the current thesis is to provide insight into the communication problems of children with AD and co-occurring ID in order to improve assessment and intervention strategies in this vulnerable group. The five chapters that follow aim to shed light on the function, form, and content of communication in children with AD and ID. Each chapter concerns a published manuscript, which can be read separately. Due to this, some overlap between each of the chapters is inevitable. The study in Chapter 6 is based on a different research group than the other chapters. Figure 1.1 presents an overview of the different components that were examined in the various studies.

![Figure 1.1 Overview of different components of communication examined in the current study](image)

The figure can be elucidated as follows. Since a diagnosis of AD in ID is complicated, in Chapter 2 the validity of a semi-structured interview for the identification of ASD in children with ID was evaluated. The main focus of Chapter 3 is to examine functions and complexity of forms used to communicate in both verbal and nonverbal low-functioning children with AD. Chapter 4 extends the current knowledge on the most common form of communication, namely language in children with AD and ID. Particularly, attention is paid to the relation between expressive and receptive language and the role of joint attention and symbol formation in language development. In Chapters 5 and 6 sense-making of visual (and tactile) communication forms in low-functioning children or adults with AD is studied. The relation with adaptive behavior and autism symptomatology is examined in Chapter 5. Chapter 6
presents the findings of sense-making in specific subgroups of low-functioning individuals with AD. In this chapter, the influence of co-occurring deafness in individuals with AD and ID is explored. The discussion of Chapter 5 is mainly focused on clinical implications, while in Chapter 6 the theoretical implications are the focus of attention.