# The complexity of early diagnostic decision making: A follow-up study of young children with language difficulties

## Rianne Jansen and Jarymke Maljaars [ID]

Parenting and Special Education Research Unit, KU Leuven, Belgium; Leuven Autism Research (LAuRes), KU Leuven, Belgium

## Inge Zink

Department of Neurosciences, ExpORL, KU Leuven, Belgium; Department of Otorhinolaryngology, Head and Neck Surgery, UZ Leuven, Belgium

## Jean Steyaert

Department of Neurosciences, Developmental Psychiatry, KU Leuven, Belgium; Leuven Autism Research (LAuRes), KU Leuven, Belgium

## Ilse Noens

Parenting and Special Education Research Unit, KU Leuven, Belgium; Leuven Autism Research (LAuRes), KU Leuven, Belgium

## Abstract

**Background & aims:** Due to the complexity of early diagnostic decision making, we examined the predictive value of an early diagnostic classification and early abilities on later best estimate diagnosis for 22 clinically referred children with language difficulties.

**Methods and procedures:** Four years after initial evaluation (Time 1), the clinical files of these children were reviewed. A best-estimate (BE) diagnosis of language disorder (LD), intellectual disability (ID), or autism spectrum disorder (ASD) was established, with ASD being most common.

**Outcomes and results:** Early clinical classifications were relatively unstable or difficult to establish at a young age. The magnitude of children's cognitive and receptive language delay was a significant predictor of a later BE diagnosis of ID and LD respectively. A BE diagnosis of ASD, by contrast, could not be predicted from children's early social communication problems nor the presence of restricted and repetitive behaviors and interests.

**Conclusions:** Taken together, the results of this study suggest that language difficulties can be an early marker of a neurodevelopmental disorder which is often not identified at the age of first referral.

**Implications:** Eligibility for treatment should, therefore, be based on biopsychosocial case formulation rather than DSM or ICD diagnostic classification.

**What this paper adds?**

In this study a dimensional approach was used to characterize the abilities of young children referred with mild to profound receptive and/or expressive language difficulties. Later on, a categorical approach was adopted to establish best estimate diagnoses. Our clinical, broadly defined sample reflects the heterogeneous intake of young children referred for diagnostic assessment. Other studies on diagnostic stability often only focus on one diagnostic category (and are explicitly excluding children with specific other diagnoses), not taking into account the difficulties of early differential diagnostic decision making and stability across different categories over time. Investigations of differential diagnosis within a clinical group, instead of only differentiating children with a specific diagnosis from typically developing children, may be more informative for clinicians.

**Corresponding author:**
Jarymke Maljaars, Parenting and Special Education Research Unit, KU Leuven, Leopold Vanderkelenstraat 32 box 3765, 3000 Leuven, Belgium.
Email: jarymke.maljaars@kuleuven.be

## Introduction

Many clinicians working with toddlers and preschool-aged children have faced the dilemma of whether to defer diagnostic decision making in children with complex and rapidly changing ability profiles or providing a possibly inaccurate diagnosis that at least enables these children and their families to access intervention and education programs. The current study reports on the complexity inherent to early diagnostic decision making as it examined the predictive value of early abilities on later diagnosis in young, clinically referred children with language difficulties.

### The complex behavioral profiles of young children with developmental difficulties

Early childhood is characterized by periods of accelerated growth and relative stability which complicates the differentiation between normative variation and atypical development (Carter et al., 2004; Egger & Angold, 2006). Moreover, if children present with problems early in life, these can be indicative of several neurodevelopmental disorders. Nevertheless, these children are often referred to specialists or specialized services with expertise in one developmental domain or one specific neurodevelopmental disorder. They often only focus on the child's difficulties which are most obvious. Additional difficulties may, therefore, go unnoticed and children with similar ability profiles may be diagnosed differently depending on the educational background and the expertise of the clinician seen (Gillberg, 2010).

Language difficulties readily attract attention and are among the first concerns of parents of children later diagnosed with a neurodevelopmental disorder (Kozlowski et al., 2011). Hence, the heterogeneity among these children is used to illustrate the diagnostic complexity in young children with developmental difficulties.

### The example of young children with language difficulties

Young children with language difficulties do not only differ with respect to the type and severity of their language difficulties but also with respect to the presence of co-occurring problems and their development over time (Desmarais et al., 2008). Several studies, found that children with language difficulties can present

with co-occurring motor deficits (Finlay & McPhillips, 2013), limited nonverbal cognitive abilities (Buschmann et al., 2008), limited symbolic play capacities (Rescorla & Goossens, 1992), problem behavior (Henrichs et al., 2013), and/or problems in social relationships (Mok et al., 2014).

Results of longitudinal studies indicate that the majority of young children with language delay catch-up with their peers, though some continue to show language difficulties over time (e.g., Dale et al., 2003; 2014). Predicting the risk of persistent language difficulties from children's early abilities is difficult. Risk factors include a family history of language delay, limited use of alternative (nonverbal) communication strategies, and the presence of receptive as well as expressive language difficulties (Ellis & Thal, 2008). Language difficulties can also be an early marker of neurodevelopmental disorders other than a language disorder (LD), with autism spectrum disorder (ASD) and intellectual disability (ID) being particularly common (Ek et al., 2012; Miniscalco et al., 2006).

According to the DSM-5, LD is diagnosed when the individual has persistent difficulties in language comprehension and/or production and does not demonstrate age-appropriate language ability (American Psychiatric Association, 2013). In the current study, 'persistent' was defined as language difficulties despite language intervention. A large longitudinal study showed that it is difficult to diagnose LD before the age of five (Stothard et al., 1998).

Core symptoms of ASD include impairments in social-communication as well as the presence of restricted and repetitive behaviors and interests (RRBIs) (American Psychiatric Association, 2013). The overlap between ASD and LD has received considerable attention over the past few decades and tend to be most noticeable in the social-communication domain (Leyfer et al., 2008). Nevertheless, previous studies found hyper- or hyposensitivity reactions (Taal et al., 2013) as well as the presence of motor stereotypies (Goldman et al., 2009) in some children with LD. ASD is often diagnosed, on average, around the age of four years, although an earlier diagnosis is possible as well (Brett et al., 2016). Variable results have been found in studies on the stability of an early ASD diagnosis (Woolfenden et al., 2012; Zwaigenbaum et al., 2016). However, studies on diagnostic stability of for example LD or ASD often only focus on one diagnostic category. Therefore, these studies do not

take into account the difficulties of early differential diagnostic decision making and stability across different diagnostic categories over time.

Below-average cognitive abilities and impairments in adaptive behavior are the core symptoms of ID (American Psychiatric Association, 2013). Below-average cognitive abilities are often part of the ability profiles of many children with language difficulties (Buschmann et al., 2008). Although cognitive abilities tend to be relatively stable in typically developing children (Heller et al., 1996), they may be more variable in children with language difficulties (Benasich et al., 1993) and in children with ASD (Dietz et al., 2007).

### Aim of the study

Clinicians do experience challenges and uncertainties in early diagnostic decision making (Charest et al., 2019; McDonnell et al., 2019). Young children with developmental difficulties often develop rapidly and present with symptom patterns that can be indicative of several disorders. In the current paper we do not make any statements about which condition is causing another condition. Based on the DSM-5 criteria for LD, ASD, and ID, children may present with only LD, ASD or ID, or several condition may co-occur in one child, such as LD and ASD or ID and ASD. A diagnosis of LD is not possible when an ID is present. This paper focuses on how to distinguish between those different conditions in young children referred with language delays and how these classifications evolve over time. Early and accurate identification and diagnosis are important to facilitate access to intervention and to increase chances to positively influence developmental trajectories (e.g., Hampton & Kaiser, 2016; Walker et al., 2020).

In this exploratory study, a group of young children referred with language difficulties has been followed over a time period of four years. This study examines (1) whether an early diagnostic classification of ID, ASD and/or LD at Time 1 is stable when compared to a best-estimate (BE) diagnosis established four years later, and (2) whether early symptoms of ID, ASD, and LD at Time 1 correctly predicted later BE diagnosis.

## Methods

### Participants

At Time 1, the sample included 36 two-to four-year-old monolingual Dutch speaking children with language difficulties. The sample included 30 boys and 6 girls with a mean age of 33.36 months ($SD = 6.82$ months). The children were prospectively and consecutively recruited from two outpatient centers for children with developmental (language) difficulties in Leuven (Belgium) and scored either below the 16th percentile on the receptive and/or expressive language scale of a standardized language test or below the $3^{rd}$ percentile on one or more of its subscales. Children with nonverbal mental age equivalent scores below 15 months, chronic hearing deficits, uncorrectable visual impairments, severe motor difficulties, frank neurological signs, or a known genetic syndrome were excluded from the study. Based on these criteria only children with very severe impairments that prevented them to complete the assessment instruments used in this study were excluded. At Time 1 all children were assessed by a multidisciplinary team (MDT) consisting of at least a psychologist, speech language therapist, and a psychiatrist/child neurologist/pediatrician). Some of the participating children were diagnosed with ASD ($n = 4$), ID ($n = 7$), or ASD with co-occurring ID ($n = 2$) whereas this decision was deferred for others by the MDT.

One year later, all children were invited for follow-up assessment in order to track their development over time. Additional follow-up appointments were scheduled depending on the children's needs. We did, therefore, not have the same amount of data over a similar time frame for all children. Four years after Time 1, the first two authors - who were trained as clinicians and have several years of experience in assessing neurodevelopmental disorders in young children - reviewed the clinical files of all children of the original sample, including reports on developmental and medical history, test results, multidisciplinary diagnostic reports, and speech and language intervention reports. The second (but not the first author) was unfamiliar with all participants. Children were included in the current study if they made at least one return visit in the four years after Time 1 and if sufficient information was available to establish a best-estimate (BE) diagnosis for each of the three neurodevelopmental disorders of interest (see Table 1). Based on these criteria 14 children were excluded.

The final sample included 22 children (17 boys, 5 girls) with a mean age of 72 months [6 years] ($SD = 6.72$ months). The severity of the receptive ($z = -0.89$, $p = .384$) and expressive ($z = -0.02$, $p = .993$) language difficulties of the children who were included in the sample, did not differ significantly from their excluded counterparts. Nevertheless, given the level of attrition, the current sample cannot be considered fully representative of the original sample.

### Establishing a BE diagnosis

Based on DSM-5 classification criteria (American Psychiatric Association, 2013), the diagnostic categories of LD, ID, and ASD were divided into four

**Table 1.** Criteria per level of diagnostic confidence for each of the three diagnostic categories.

|  | LD | ASD | ID |
|---|---|---|---|
| Not applicable | Clinical diagnosis of ID *or* a receptive and/or expressive language score ≥ pc. 15 on a standardized language test at FU | No clinical signs of ASD at Time 1 *and* an ADOS score below the ASD cut-off at FU. | An IQ-score ≥ 80 or more on a standardized intelligence test at FU. |
| Possible | Children with a receptive and/or expressive language score < pc. 15 on a standardized language test at FU who: (1) received no language intervention prior to assessment, (2) showed rapid progression over time, or (3) were extremely shy, sick, or fatigued at the time of assessment. | No clinical signs of ASD at Time 1 but an ADOS score above the ASD cut-off at FU *or* clinical signs that did not meet diagnostic criteria for ASD at FU. | An IQ-score between 70 and 80 on a standardized intelligence test at FU. |
| Probable | A receptive and/or expressive language score between pc. 3 and pc. 7 on a standardized language test at FU despite intervention *or* children for whom the magnitude of their language delay increased over time despite intervention. | Clinical signs of ASD at Time 1 and an ADOS score above the ASD cut-off at FU. | An IQ-score ≤70 on a standardized intelligence test *and* a total adaptive behavior score ≤ pc. 5 on the Vineland Screener 0–6 yrs-NL at FU. |
| Applicable | Clinical diagnosis of LD (or a related diagnosis such as DLD or developmental dysphasia) *or* a receptive and/or expressive language score ≤ pc. 3 on a standardized language test at FU despite intervention. | Clinical diagnosis of ASD | Clinical diagnosis of ID |

FU = follow-up; LD = language disorder; ASD = autism spectrum disorder; ID = intellectual disability; pc. = percentile; DLD = developmental language disorder; ADOS = Autism Diagnostic Observation Schedule.

levels of diagnostic confidence: not applicable versus possibly, probably, or definitely applicable. Criteria for each level of diagnostic confidence were established in advance (see Table 1) and both raters independently completed a BE diagnosis form. BE diagnoses were based on follow-up data. With respect to the diagnostic category of ASD, however, Time 1 qualitative descriptions - but not the cut-off scores - from the Autism Diagnostic Observation Schedule (ADOS: Lord et al., 1999) and the Diagnostic Interview for Social and Communication Disorders - 11th edition (DISCO-11: Wing, 2006) were made available. This allowed the raters to take the presence of ASD-related characteristics in the past into account as - according to DSM-5 criteria - the onset of the symptoms characterizing this disorder is in the early developmental period (American Psychiatric Association, 2013). Children who presented with clinical signs at Time 1 and an ADOS score above the ASD-cut-off at follow-up qualified for diagnosis as did children who received a clinical diagnosis of ASD.

There was complete agreement between the raters, except for one child for whom consensus was reached after joint review of the information available. A BE diagnosis of LD, ID, or ASD was established in

probable or definite cases only. With respect to LD, care was taken to ensure that only those children with persistent language difficulties qualified for diagnosis which was operationalized as below-average receptive and/or expressive language abilities at two (or more) time-points despite intervention. Following DSM-5 criteria, a BE diagnosis of LD was not made if children met criteria for a BE diagnosis of ID.

## Instruments used at time 1

As a BE diagnosis of LD, ID, or ASD was predicted from children's abilities at Time 1, the instruments that were used to assess their abilities at that time are reported below.

**Language.** Language scores were derived from a developmental hierarchy of measures which captured the heterogeneity in receptive and expressive abilities among the participating children: (1) The Dutch version of the NonSpeech Test for Receptive and Expressive Language (NNST: Zink & Lembrechts, 2000) (for children with a language developmental age between 1 and 2 years) and (2) the Dutch version of the Reynell Developmental Language Scales (RTOS: Schaerlaekens et al., 2003) (for children with a

language developmental age between 2 and 5 years). Language ability scores for children who were not able to complete the NNST or RTOS were derived from the Dutch version of the MacArthur-Bates Communicative Development Inventories (N-CDIs: Zink & Lejaegere, 2003, 2007). Receptive language scores were derived from the NNST (27.3%) or RTOS (72.7%) whereas expressive language scores were derived from either direct assessment (NNST: 54.5%, RTOS: 22.7%) or parent report (N-CDIs: 22.7%). Parent-reported and assessed expressive abilities were highly correlated, at least for those children for whom both measures were available ($r_s = .88$). Different tests have to be used as none of the Dutch, norm-referenced language measures available covered the heterogeneity in ability among the participating children. All language tests used were appropriate for the language spoken at home, as only monolingual Dutch speaking children were included in the study.

**Nonverbal cognition.** Because of the variety in nonverbal cognitive abilities among the participating children, nonverbal cognitive ability scores were also derived from a developmental hierarchy of measures: (1) The nonverbal scale of the Dutch version of the Bayley Scales of Infant Development (BSID-NL: Van der Meulen et al., 2002) (for children with a developmental age below 2;6 years) or (2) the revised version of the Snijders-Oomen Non-verbal Intelligence Test (SON-R 2$^1$/$_2$-7: Tellegen et al., 1998; Tellegen & Laros, 2013) (for children with a developmental age of 2;6 years and higher). The BSID-II-NL was administered to 45.5% of the children whereas the SON-R 2$^1$/$_2$-7 was administered to the remaining 54.5%.

**ASD-related characteristics.** The presence of ASD-related characteristics was assessed by means of the DISCO-11 and ADOS. The DISCO-11 is a semi-structured interview schedule that is administered to (one of) the children's parents and elicits information on an individual's developmental history and current present clinical picture (Wing, 2006). The DSM-5 diagnostic algorithm for ASD has satisfactory levels of both sensitivity and specificity and comprises a Social Communication and Repetitive Behavior domain (Kent et al., 2013). Calibrated algorithm raw domain scores based on current ratings were used in this study. The ADOS is a clinician-administered instrument that contains standardized activities and social presses to elicit communication as well as social behavior and promotes play as well as the imaginative use of objects. Different modules are available and the most appropriate module has to be selected based on the participant's expressive language level and chronological age (Lord et al., 2012). The diagnostic algorithm for ASD includes a Social Affect (SA) and Restricted and Repetitive Behaviors (RRB) domain. Calibrated

severity scores from raw domain totals were used in this study as they provide separate estimates of SA and RRB symptom severity which can be compared across ADOS modules 1-3 (Hus et al., 2014). Module 1 (no or some words) was used in 86.4% of the administrations whereas Module 2 (short sentences) was used in the remaining 13.6%.

## Ethical considerations

Parents received information on the content of the study and provided written informed consent. The design of the study was approved by the medical ethical board of the University Hospitals UZ/KU Leuven.

## Data analyses

Language and nonverbal cognitive mental age equivalent scores were converted into language and developmental quotients that provide an age-dependent estimate of the magnitude of the delay. Pearson's chi-square tests were used to examine the relationship between cluster membership at Time 1 and BE diagnosis at follow-up. Binary logistic regression analysis was conducted to examine whether early symptoms of LD, ID, and ASD at Time 1 correctly predicted later diagnosis. This technique enables the prediction of a categorical outcome from one or more continuous predictor variables which do not need to be normally distributed (Williams et al., 2013). Although the sample size in this study is smaller than generally recommended, the data met the assumptions for binary logistic regression analysis: (1) linearity of the logit, (2) the absence of multicollinearity (correlations between predictor variables < .71; variance inflation factors < 10), and (3) the independence of errors. Standardized residuals were checked to ensure that none of the children exerted an undue influence on the regression models. None of the children had absolute values above 2.5 and no more than 5% had absolute values above 2. In each of these case the value of Cook's distance was below one and the leverage values were in the expected range. Therefore, these children did not seem to have a large influence on the regression models (Stevens, 2002) and were included in the analysis.

## Results

After a multidisciplinary clinical assessment at Time 1, ten children received a clinical diagnostic classification of ID, ASD, or both (see Figure 1). For some children ($n = 6$) a possible or deferred classification of ASD was described in their clinical report. This means that several symptoms were clearly present, but that at the same time the clinical picture as a whole was still unclear, indicating that the specific diagnostic
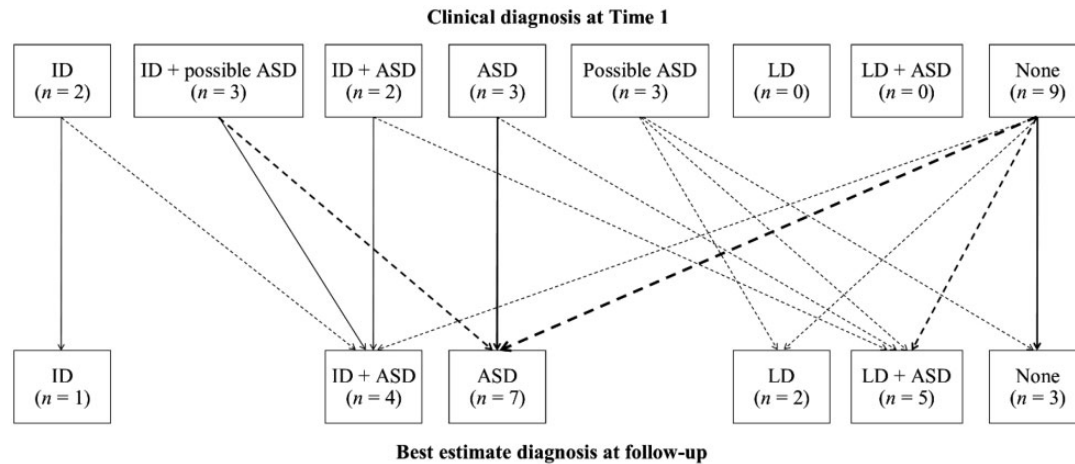
**Clinical diagnosis at Time 1**



**Best estimate diagnosis at follow-up**

**Figure 1.** BE diagnosis at follow-up compared to clinical diagnosis at Time 1.
ID = intellectual disability; ASD = autism spectrum disorder; LD = language disorder;
*Note.* Weight of line indicates number of cases; solid line = stable diagnosis; dotted line = change in diagnosis.

classification had to be reconsidered at a later stage. No children were diagnosed with LD at Time 1, as persistency of language problems could not yet be determined.

Four years later, 19 (86%) of the 22 children who participated in this study, received a BE diagnosis of ID, ASD, and/or LD, with ASD being most common ($n = 16$). Three children did not meet criteria for any of the diagnoses of interest at follow-up. BE diagnosis and clinical diagnosis at follow-up corresponded in 61% of the cases (LD 43%; ID 80%; ASD 63%). In the remaining cases, a BE diagnosis was established in the absence of a clinical diagnosis.

Information on the stability and change in diagnosis from Time 1 to follow-up is depicted in Figure 1. In only five cases (23%) clinical diagnosis or the combination of two diagnoses at Time 1 remained exactly the same at follow-up. Ten children (45%) received a new or additional diagnosis at Time 2. Three children (14%) switched diagnostic categories and in two cases (9%) a diagnosis was withdrawn. A diagnosis of ASD at Time 1 turned out to be more stable at follow-up (9 out of 11 children with a (possible) ASD diagnosis at Time 1) compared to a diagnosis of ID (4 out of 7 children with an ID diagnosis at Time 1). However, more children received a later ASD diagnosis at Time 2 without being diagnosed at Time 1 (7 new diagnoses) compared to ID (1 new diagnosis).

### Predicting a BE diagnosis of LD

A BE diagnosis of LD was established in 7 of the 22 children. Five of the children in the LD subgroup also received a BE diagnosis of ASD. The receptive and expressive language quotients at Time 1 (Table 2) did not differ significantly between children with and

without a BE diagnosis of LD (receptive language: $z = -1.16$, $p = .130$; expressive language: $z = -1.55$, $p = .127$). A binary logistic regression analysis was performed to examine whether a child's receptive and/or expressive language quotient at Time 1 predicted a BE diagnosis of LD at follow-up. Children's developmental quotient was included as a predictor variable as well, as ID and LD cannot be diagnosed concurrently according to DSM-5 criteria (American Psychiatric Association, 2013). The regression model was marginally significant (model $\chi^2_{(3)} = 7.76$, $p = .051$) and pointed to the magnitude of children's cognitive ($b = 0.13$, $SE = 0.07$, Wald's $\chi^2 = 3.54$, $p = .060$) and receptive language ($b = -0.07$, $SE = 0.04$, Wald's $\chi^2 = 2.82$, $p = .093$) delay as possible predictors. The analysis was, therefore, rerun with only these two predictor variables included.

The adapted regression model significantly predicted children's diagnosis (model $\chi^2_{(2)} = 6.82$, $p = .033$) and accounted for between 26.7% and 37.4% of the variance in diagnostic status. Overall, the model correctly classified 77.3% of the children. Nevertheless, the identification of children with a BE diagnosis of LD was only slightly better than chance level (57.1%). Table 3 displays the coefficients, Wald statistics, and odd ratios for each of the two predictor variables. The magnitude of children's receptive language delay was a significant predictor of diagnostic status meaning that the greater the delay at Time 1, the greater the likelihood of obtaining a BE diagnosis of LD at follow-up.

### Predicting a BE diagnosis of ID

Five of the 22 children received a BE diagnosis of ID at follow-up. Four of the children in the ID subgroup also

**Table 2.** Means (and standard deviations) for predictor variables per BE diagnosis at follow-up.

| | Best estimate diagnosis | | | | | |
| --- | --- | --- | --- | --- | --- | --- |
| | LD | | ID | | ASD | |
| | No (n = 15) | Yes (n = 7) | No (n = 17) | Yes (n = 5) | No (n = 6) | Yes (n = 16) |
| Developmental quotient | 89.56 (18.46) | 94.31 (10.33) | 96.96 (12.70) | 71.05 (9.32) | | |
| Receptive language quotient | 83.76 (24.13) | 71.57 (15.82) | | | | |
| Expressive language quotient | 64.19 (13.48) | 56.39 (8.06) | | | | |
| ADOS SA domain (severity score) | | | | | 2.67 (0.82) | 2.94 (1.53) |
| ADOS RRB domain (severity score) | | | | | 6.67 (1.21) | 7.13 (1.26) |
| DISCO domain A (social communication, raw score) | | | | | 3.17 (2.48) | 6.25 (3.84) |
| DISCO domain B (repetitive behavior, raw score) | | | | | 1.00 (1.27) | 1.94 (2.57) |

*Note.* LD = language disorder; ID = intellectual disability; ASD = autism spectrum disorder; SA = Social Affect; RRB = Restricted *and Repetitive Behaviors.*

**Table 3.** Binary logistic regression analysis: Predicting the odds of obtaining a BE diagnosis of LD, ID, or ASD at follow-up.

| BE dx | Model | Predictor | b (SE) | Wald's $\chi^2$ | p | OR | 95% CI |
| --- | --- | --- | --- | --- | --- | --- | --- |
| LD | 1 | Developmental quotient | 0.11 (0.06) | 3.74 | .053 | 1.12 | 1.00–1.26 |
| | | Receptive language quotient | −0.09 (0.04) | 4.61 | .032 | 0.92 | 0.84–0.99 |
| ID | 1 | Developmental quotient | −0.17 (0.07) | 5.71 | .017 | 0.84 | 0.73–0.97 |
| ASD | 1 | ADOS Social Affect domain | 0.35 (0.47) | 0.57 | .450 | 1.42 | 0.57–3.53 |
| | | ADOS RRB domain | 0.46 (0.45) | 1.06 | .303 | 1.59 | 0.66–3.81 |
| | 2 | DISCO Social Communication domain | 0.27 (0.18) | 2.24 | .134 | 1.31 | 0.92–1.86 |
| | | DISCO Repetitive Behavior domain | 0.16 (0.30) | 0.29 | .591 | 1.17 | 0.66–2.10 |

*Note.* BE dx = best estimate diagnosis; LD = language disorder; ID = intellectual disability; ASD = autism spectrum disorder; RRB = Restricted and Repetitive Behaviors.

received a BE diagnosis of ASD. The children with a BE diagnosis of ID had a lower developmental quotient than their non-ID counterparts at Time 1 (Table 2) which indicated a significantly greater impairment in nonverbal cognitive abilities ($z = -2.98$, $p < .001$). A binary logistic regression analysis was performed to examine whether a child's developmental quotient at Time 1 predicted a BE diagnosis of ID at follow-up. The regression model significantly predicted children's diagnosis (model $\chi^2_{(1)} = 12.67$, $p < .001$) and accounted for between 43.8% and 66.6% of the variance in diagnostic status. The model correctly classified 80% of the children who did and 94.1% of the children who did not receive a BE diagnosis of ID. Overall, 90.9% of the predictions were accurate. The magnitude of children's nonverbal cognitive delay was a significant predictor of diagnostic status (Table 3) meaning that the greater the delay at Time 1, the greater the likelihood of obtaining a BE diagnosis of ID at follow-up.

## Predicting a BE diagnosis of ASD

A BE diagnosis of ASD was established in 16 of the 22 children. Some of the children in the ASD subgroup also received a BE diagnosis of ID ($n = 6$) or LD ($n = 5$). Of the 16 children who received a BE diagnosis of ASD, seven scored above the ASD cut-off on the ADOS at Time 1, as did three children who did not receive a BE diagnosis of ASD. None of the children in our sample scored above the ASD cut-off on the DISCO. Nevertheless, children with a BE diagnosis of ASD obtained higher scores on the Social Communication domain of the DISCO at Time 1

than their non-ASD counterparts (Table 2), which indicated that the parents of these children reported significantly more difficulties in social interaction and communication ($z = -1.74$, $p = .042$). These differences were particularly salient for those items assessing social-emotional reciprocity (ASD group: $M = 3.13$, $SD = 2.03$; non-ASD group: $M = 1.00$, $SD = 1.27$, $z = -2.20$, $p = .012$).

A binary logistic regression analysis was performed to examine whether children's ADOS or DISCO domain scores at Time 1 predicted a BE diagnosis of ASD at follow-up. In order to limit the number of predictor variables included in the regression model, analysis were performed separately for each diagnostic instrument. Neither model resulted in an accurate prediction of children's diagnostic status (ADOS model $\chi^2_{(2)} = 1.31$, $p = .521$; DISCO model $\chi^2_{(2)} = 3.87$, $p = .145$). Hence, children's social-communicative abilities or the presence of RRBIs at Time 1 - either observed or reported - did not predict a BE diagnosis of ASD at follow-up (Table 3).

## Discussion

Young children with language difficulties form a heterogeneous group, not only with respect to the type and severity of their language difficulties but also with respect to the presence of co-occurring problems and their development over time. This exploratory study examined in a group of clinically referred children with language difficulties whether BE diagnosis at follow-up was related to early clinical classification at Time 1 and if early symptoms of ID, ASD, and LD correctly predicted later diagnosis.

### Stability of early diagnostic classification in young children with language difficulties

For most of the participating children, a clinical diagnosis was established only after repeated, multidisciplinary biopsychosocial assessment and not at the age of first referral. Moreover, many of the children who did receive a clinical classification at Time 1, switched diagnostic categories or received an additional diagnosis over time. Based on the comparison between diagnosis at Time 1 and at follow-up, early diagnostic classifications in young children referred with language difficulties seem to be rather unstable before the age of four.

At follow-up, a best-estimate (BE) diagnosis of LD, ID, or ASD was established in approximately 86% of the children. BE diagnosis and clinical diagnosis at follow-up corresponded in the majority of cases. As we do not have data on the representativeness of our sample for all children who visited the collaborating outpatient centers, it may be possible that parents who chose to participate had more or different concerns about their child's development than non-participants. This, in turn, may have contributed to the high rates of neurodevelopmental disorders found within the sample. Previous studies, however, also found high rates of neurodevelopmental disorders in children previously diagnosed with language difficulties (Ek et al., 2012; Miniscalco et al., 2006; 2018).

### Predicting children's BE diagnosis from their early abilities

There was a significant relationship between the magnitude of children's receptive language delay at Time 1 and a BE diagnosis of LD at group level, though some children with severe receptive language difficulties at Time 1 showed (near) average linguistic abilities at follow-up. Clinicians who identify children with language difficulties have the difficult task of deciding whether or not intervention is necessary to ameliorate the established problems. Language pathways are fluid and the majority of children will spontaneously catch-up with their peers before entering primary school (Dale et al., 2003). Some, however, continue to experience language difficulties and these children should access intervention programs as early as possible. The intervention model for children with language difficulties described by Ebbels et al. (2019) offers clinicians a stepwise approach of language intervention with increasing levels of intensity and individualization based on the child's needs to facilitate clinical decision-making. With regard to diagnostic classification, only children with persistent language difficulties should receive a diagnosis of LD (American Psychiatric Association, 2013). DSM-5, however, offers no criteria to determine 'persistency' which may be thought of as below-average receptive and/or expressive language abilities at at least two time-points despite intervention.

The magnitude of children's nonverbal cognitive delay at Time 1 was a significant predictor for a BE diagnosis of ID at follow-up. Although nonverbal IQ scores appeared to be relatively stable at group level, some children showed a considerable drop in IQ scores over time whereas others moved from the below-average to the average ability range. Although nonverbal IQ tests are frequently used, it remains unclear what these tests are exactly assessing for children with language difficulties (Gallinat & Spaulding, 2014). Although limited abilities in both the language and cognitive domain may have a shared etiology, alternative explanations should be considered as well. As the difficulty of test-items increases, the performance of children with language difficulties may be hampered by: (1) a limited understanding of the verbal instructions that often accompany an examiners nonverbal

behavior and (2) less developed self-directed speech which enhances the performance of typically developing children in novel problem solving tasks (Gallinat & Spaulding, 2014; Lidstone et al., 2012). Increased language proficiency and on-task behavior, on the other hand, may account for the rise in some children's IQ scores over time.

Children with ASD were overrepresented in the current sample as a BE diagnosis of this disorder could be established in almost 75% of the children. Language development is delayed as well as deviant in many preschool-aged children with ASD (Boucher, 2012) and language difficulties have been part of the diagnostic criteria up to DSM-IV-TR. DSM-5, however, has listed language impairment as a specifier which should inform treatment planning. In this study, early symptoms of ASD within the social-communication and behavioral domain were assessed by means of an interview schedule as well as direct observation. Although children in the ASD subgroup scored somewhat higher on indices of both instruments at Time 1, the presence of the disorder could not be predicted from children's social-communication problems nor the presence of RRBIs. This may be due to unequal subsample sizes though the possibility of behavioral overlap between the ASD and non-ASD group should be considered as well. This became particularly salient as three children who did not receive a BE diagnosis of ASD scored above the ASD cut-off on the ADOS at Time 1.

Many children with language difficulties experience social problems which can persist over time (Mok et al., 2014) and may resemble the problems of children with ASD on standardized clinical measures for ASD symptoms, such as the ADOS. Some individual behaviors, however, tend to discriminate between children with and without ASD as the former seem to be less inclined to initiate and engage in reciprocal social interactions (Mildenberger et al., 2001; Ventola et al., 2007), and show more socially withdrawn behavior compared to children with LD (Richard et al., 2019). This was supported by the results of the current study as parents of children with a BE diagnosis of ASD reported higher levels of impairment on DISCO items assessing social-emotional reciprocity.

The children with and without a BE diagnosis of ASD did not differ significantly on the RRBI domain of either the ADOS or DISCO in this study. RRBIs encompass both lower- (e.g., motor stereotypies) and higher-level (e.g., circumscribed interests, adherence to routines and rituals, and repetitive language) behaviors (Turner, 1999). Young and less verbal children with ASD will mainly display lower-level RRBIs, as these RRBIs require less advanced language and cognitive abilities. These behaviors are also common in typically developing children (Larkin et al., 2017; Leekam et al.,

2007) and in children with various other neurodevelopmental disorders (Leekam et al., 2011). The importance of RRBIs as a stable predictor of ASD may increase with age (Camarata, 2014) and/or may be limited to higher-level RRBIs (Honey et al., 2008; Mildenberger et al., 2001). Moreover, it is not only the topography (i.e., types of RRBIs), but mainly the frequency, duration, and/or associated level of impairment that differentiates RRBIs in children with ASD from their non-ASD counterparts (Honey et al., 2008; Leekam et al., 2011; Watt et al., 2008).

The conceptualization of RRBIs as a (multi)dimensional construct ranging into the typically developing child population, questions the validity of social (pragmatic) communication disorder (SCD) as a new diagnostic category in the Communication Disorder section of the DSM-5. SCD is defined by persistent pragmatic difficulties in the absence of other ASD symptomatology in particular RRBIs (American Psychiatric Association, 2013). Nevertheless, children with social communication difficulties are likely to display RRBIs at some point during development (Norbury, 2014). As these RRBIs may not be severe enough to meet the ASD threshold, these children are likely to fall between diagnostic categories.

## Limitations and directions for future research

The study was limited by the use of different instruments to measure receptive and expressive language skills and non-verbal cognition, as none of the instruments available covered the variety in the participant's abilities. Because of the small sample size and the possible overrepresentation of children with persistent and more encompassing developmental problems, the results of this study should be interpreted with caution. Moreover, as the number of and time between follow-up assessments depended largely on the children's needs, we did not have the same amount of data over a similar time frame for all children. It is important to acknowledge that clinical diagnosis and BE diagnosis are not equivalent as BE procedures force raters to make a decision which in clinical practice may be deferred. Nevertheless, care was taken to ensure that only clinically relevant problems qualified for a BE diagnosis of LD, ASD, or ID. Further research should include a more in-depth examination of the abilities of young children with language difficulties. Attention to motor, behavioral, and affective functioning may reveal important dimensions of functioning and further psychopathology in children with early language difficulties. Moreover, additional research into the validity of the DISCO algorithm for young children is needed as none of the participating children with a clinical diagnosis or BE diagnosis of ASD reached the

cut-off. This can be due to limited suitability of the current DSM-5 criteria for young children in general or the selection of algorithm items from the DISCO in particular. Many of the items included in the current algorithm require children to have a certain age or level of language proficiency in order to be met.

## Clinical implications

Although parents may present to practitioners with specific developmental concerns, co-occurring problems are likely to be present or tend to develop over time (Gillberg, 2010). Children with developmental difficulties in any domain of functioning should, therefore, be referred for biopsychosocial assessment by an interdisciplinary team. In a biopsychosocial assessment, biological (e.g. gender, physical illness), psychological (e.g., behaviors, emotions) and social (e.g., family background, cultural background) factors that could be contributing to the observed difficulties are taken into account. Parental information as well as assessment and observation in structured and unstructured, more naturalistic situations will result in a holistic picture of children's strengths and weaknesses in interaction with their environment (O'Keeffe & Macaulay, 2012). An assessment conducted by clinical experts from different disciplines (e.g., speech language therapists, psychologists, pediatricians/child psychiatrists/child neurologists/other medical doctors) offers different perspectives and will avoid 'tunnel vision'. It is essential that an interdisciplinary team meets to share, integrate, and synthesize information and to provide recommendations for intervention together (Guralnick, 2000). By stating this we do not necessarily argue in favor of a dimensional as opposed to a categorical approach to diagnostic decision making as both have their value. It is the order in which they are used that is of uttermost importance. Dimensional assessment and thoughtful case formulation are a necessary first step. A categorical clinical diagnosis can be established second, if at all (O'Keeffe & Macaulay, 2012).

Many early intervention programs are restricted to children with a specific clinical diagnosis. Such an allocation model assumes that neurodevelopmental disorders can be reliably identified in all children likely to benefit from treatment and that (the families of) children with a clinical diagnosis are in greater need than those without (McDowell & O'Keeffe, 2012). The results of this study indicate that diagnostic classification is often only possible after repeated multidisciplinary, biopsychosocial assessment and not at the age of first referral. Nevertheless, early intervention provides children with enriched experiences, diminishes symptom severity, prevents secondary problems from occurring and enhances sensitive parenting as well as

parental well-being (Koegel et al., 2014). These benefits may lead clinicians to err on the side of a false-positive diagnosis in cases of diagnostic uncertainty in order to enable intervention for these children and their families (Skellern et al., 2005). The interrelatedness of diagnosis and intervention seems no longer sustainable (McGorry & Van Os, 2013; O'Keeffe & Macaulay, 2012). Disentangling diagnosis from symptoms and impairments will - although admittedly complex - be a fruitful way forward to ensure early intervention services for children whose clinical phenotype is (still) unclear (Rapee et al., 2012).

## Conclusions

This exploratory study revealed some of the complexity inherent to diagnostic classification in young children. Although a clinical diagnosis can be established for some of them, many present with mixed symptom patterns and/or subthreshold symptomatology in a developmental period where intervention is considered most likely to be effective but often requires a clinical diagnosis.

## Highlights

- Early diagnosis in children with language difficulties seem to be unstable before the age of 4.
- Language disorder and intellectual disability classifications can be predicted based on early abilities.
- However, large inter-individual differences were seen in developmental trajectories.
- A clinical differential diagnosis was established only after repeated assessments.

### ORCID iD

Jarymke Maljaars (iD) https://orcid.org/0000-0002-8555-4584

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