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## Language in young children with neurofibromatosis-1: Relations to functional communication, attention, and social functioning



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### ABSTRACT

In this study, the language abilities of 30 children with Neurofibromatosis Type 1 (NF1) aged 4–6 years were examined using a standardized measure of language. Relations of language to multiple parental report measures of functional communication, social skills, and attention problems were investigated. Difficulties in core language skills were observed, and more than 1/3 of the children struggled on at least one language index. Language abilities were significantly related to parental report of functional communication, social interaction and communication, and social skills, such that language difficulties may be a risk factor for communication and social interaction challenges and communication-related adaptive behavior in children with NF1. Though receptive language abilities were an area of particular difficulty for many children with NF1, they were not significantly related to parental ratings of social functioning and functional communication. Few significant relations were found between language and parent-reported attention problems, although some trends were noted. Hence attention difficulties in children with NF1 may contribute to, but do not appear to fully account for, language difficulties. In sum, there is an increased risk of language difficulties for young children with NF1, and lab-measured language difficulties appear to relate to everyday communication and social interaction functioning.

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### 1. Introduction

Neurofibromatosis type 1 (NF1) is a genetic neurodevelopmental disorder present in about 1 in 3000 individuals. The disorder results from a single gene mutation in the neurofibromin 1 gene (17q11.2) which codes for the production of neurofibromin and in turn helps to regulate neuronal cell growth and development and suppress tumors. Without proper production of this gene, plexiform, cutaneous, or subcutaneous fibromas, or tumors, which characterize NF1, may form. Other physical traits include skeletal irregularities, café-au-lait spots, and axillary freckling (Ferner et al., 2007). Individuals with NF1 are at elevated risk for numerous cognitive and behavioral difficulties starting in early childhood. Notably, the average intellectual functioning of children with NF1 falls only slightly lower than that of unaffected children (Dilts et al., 1996; Eliason,

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1986; Mazzocco et al., 1995; North et al., 1994; North, Hyman, & Barton, 2002), but children with NF1 often show deficits in one or more cognitive domains. Almost half of children with NF1 present with comorbid difficulties, including ADHD or subclinical attention difficulties, learning problems, executive function challenges, fine and gross motor difficulties, weaknesses in academic competence (particularly reading), visuomotor and visuospatial problems, and adaptive behavior deficits (Barton & North, 2004; Dilts et al., 1996; Hyman, Shores, & North, 2005; Klein Tasman et al., 2013). Language problems have also been documented in individuals with NF1 (Dilts et al., 1996; Hyman et al., 2005; Lorenzo, Barton, Acosta, & North, 2011; Mazzocco et al., 1995; Thompson, Viskochil, Stevenson, & Chapman, 2010). These difficulties culminate in academic difficulties for youth with NF1 in comparison to unaffected peers. Although exploring the cognitive profile of children with NF1 has been the focus of a number of studies, there have been few studies of language problems specifically. Greater understanding of the language functioning of young children with NF1 is important for these individuals' later success, as early identification of learning vulnerabilities is central to early intervention to maximize functioning.

Research with individuals without NF1 suggests that language difficulties and attention difficulties may be related (Gut, Heckmann, Meyer, Schmid, & Grob, 2012; Helland, Biringer, Helland, & Heimann, 2012), with several studies showing direct significant associations between the two in child populations (Bellani, Moretti, Perlini, & Brambilla, 2011; Geurts & Embrechts, 2008; Haak, Downer, & Reeve, 2012; McGrath et al., 2008; Sowerby, Seal, & Tripp, 2011; Wassenberg et al., 2010). Specifically, deficits in language structure, semantic language, receptive language, expressive language, verbal working memory, and language comprehension have been associated with ADHD, as have interpersonal difficulties such as communication problems, difficulty understanding social cues, and trouble with pragmatics (Geurts & Embrechts, 2008; Gremillion & Martel, 2012; Gut et al., 2012; Haak et al., 2012; Helland et al., 2012; Sowerby et al., 2011; Wassenberg et al., 2010). Additionally, ADHD has been found to raise a child's risk of difficulty with social functioning (which heavily depends on language), with high rates of peer relationship difficulties in children with (Barton & North, 2004) and without NF1 (Guevremont & Dumas, 1994). Academic underachievement appears to be highly intertwined with these difficulties as well (Gremillion & Martel, 2012; Gut et al., 2012; Wassenberg et al., 2010); the ability to devote appropriate attention to a task is a crucial classroom skill, and difficulty in this area may very well be associated with language problems, interfering with the ability to understand material presented in an academic setting.

The association between language difficulties and attention problems is quite relevant to research involving children with NF1, as one of the hallmark features of the disorder is the high vulnerability to attention problems. Comorbid attention difficulties are often seen in children with NF1; strikingly, ADHD has been reported in 40–50% of cases (Eliason, 1986; Hyman, Shores, & North, 2006; Koth, Cutting, & Denckla, 2000), making it one of the most common difficulties for this population. ADHD occurs at a much higher rate in children with NF1 (42%) than in unaffected siblings (16%), and ADHD often presents in children whose parents show no signs of attention difficulties (Koth et al., 2000). Even in children without an ADHD diagnosis, those with NF1 still show significantly more hyperactivity and inattention compared to controls (Huijbregts, Swaab, & de Sonnevile, 2010), with inattentive symptoms being particularly characteristic, even in young children (Sangster, Shores, Watt, & North, 2011). In fact, some level of attention difficulty is found in so many cases that Koth and colleagues (2000) suggest attention difficulties are a component of the NF1 phenotype. Klein-Tasman and colleagues (2014) found that verbal skills assessed broadly in the context of cognitive assessment were significantly related to attention and social skills difficulties. However, to date there is no research investigating relations between attention problems and comprehensively measured language functioning in young children with NF1.

There is some evidence that language difficulties are indeed present for an elevated proportion of school-aged children with NF1. Several studies have included examination of verbal functioning within the context of broad assessment, documenting mild difficulties with verbal functioning compared to siblings and same-aged peers (Billingsley, Slopis, Swank, Jackson, & Moore, 2003; Brewer & Moore, 1997; Cutting & Levine, 2010; Eldridge et al., 1989; Hyman et al., 2005; Mazzocco et al., 1995; North et al., 1994; North, Joy, Yuille, Cocks, & Hutchins, 1995; Sangster et al., 2011), although some of these differences were not significant after controlling for intellectual functioning (Hyman et al., 2005; Sangster et al., 2011). Closer examinations of language skills in school-aged children have revealed difficulties with broad expressive and receptive language skills as well as phonological awareness (Dilts et al., 1996; Mazzocco et al., 1995; North et al., 1994, 1995), but more in-depth research on the structure and content of language in individuals with NF1 is limited.

For young children with NF1, there is currently sparse information about language functioning. Some research on language within the context of broad cognitive and behavioral assessment suggests that very early language difficulties may be detectable in this population. In one case, while children (39 toddlers, aged 21–30 months) with NF1 did not show significant language difficulties in comparison to the normative mean, the majority exhibited below-average expressive language skills according to parental report (Lorenzo et al., 2011). Notably, Thompson and colleagues (2010) formally and comprehensively assessed expressive and receptive language in 19 preschoolers with NF1 with a standardized individually-administered language measure, the Clinical Evaluation of Language Fundamentals-Preschool, 2nd edition (CELF-P2; Wiig, Secord, & Semel, 2004). A substantial proportion of the participants demonstrated delays despite group scores in the low average range overall. Difficulties (defined as mean group performance more than 1 standard deviation below the mean) were detected on all expressive and receptive subtests, providing a more in-depth look than prior work at the nature of language difficulties. Some limitations of this study, however, include a small sample size, lack of inclusion of examination of the Language Content and Language Structure indices (which capture semantics and word/sentence structure), and lack of descriptive statistics at the subtest level beyond frequency of difficulty.

Characterizing the nature of language difficulties in young children with NF1 is important because, firstly, early language difficulties are argued to be strong predictors of problem behavior and learning difficulties in children without NF1. For example, expressive and/or receptive language problems in kindergarten have been associated with conduct problems, behavioral difficulties, hyperactivity, and social and academic problems later in elementary school (Bashir & Scavuzzo, 1992; Catts, 1993; Hooper, Roberts, Zeisel, & Poe, 2003; Sigafoos, 2000). In addition, verbal problems may be related to social, communicative and adaptive problems in children with NF1. Functional communication, which is a child's ability to seek out and communicate information in real-world situations (Reynolds & Kamphaus, 2004), and social skills have both been shown to be significantly poorer in children with NF1 compared to unaffected children and the normative population (Barton & North, 2004; Klein-Tasman et al., 2013). Verbal skills also appear to be related to social skills and adaptive skills, including functional communication, when assessed in the context of cognitive functioning (Barton & North, 2004; Klein-Tasman et al., 2014).<sup>1</sup>

Further investigation of the interplay between expressive and receptive language abilities and everyday functioning is needed, as language problems may present psychosocial barriers and interfere with interactive functioning and learning within family, school, and community settings. There is especially a need for continued research about language abilities in young children with NF1, as language develops rapidly during these years and language difficulties are a risk factor for reading challenges in the general population. Many of these difficulties appear to be interrelated, and measuring a child's functional communication or social-communicative adaptive skills may reveal much about the impact of language difficulties on everyday functioning. A closer look at connections between language and language-related adaptive and behavioral problems is warranted given research suggesting relations between parental reports of real-world functional communication and lab-assessed verbal functioning in both children with NF1 and unaffected children. The purpose of the current study is to comprehensively examine language abilities in young children with NF1 and to investigate relations to functional communication, attention problems, and social functioning.

### 1.1. Study aims and hypotheses

First, we aim to examine language abilities of children with NF1 for early signs of language difficulties in comparison to normative data. We will replicate and extend prior work (Thompson et al., 2010) by including a larger sample, examining composites not included in prior work (Language Content and Language Structure) and providing more detail regarding subtest-level performance. Second, we will expand on prior work by examining correspondence between lab-based measures of language functioning and parental ratings of functional communication, social difficulties, social-communicative adaptive skills, and attention problems commonly reported for children with NF1.

It is hypothesized that children with NF1 will exhibit difficulties compared to the normative population on CELF-P2 composite scores and subtests. While children with NF1 may exhibit mean composite scores within the average range on the CELF-P2, it is expected that the frequency of language difficulties in children with NF1 will be elevated. It is also hypothesized that language abilities as measured by the CELF-P2 will be significantly related to parental reports of functional communication, social functioning, and social-communicative adaptive behavior. Further, attention difficulties are expected to be related to the language abilities of children with NF1, as attention is important for comprehension and processing of oral and visual information.

## 2. Method

### 2.1. Participants

Participants were 30 children with NF1 (20 males, 10 females), ranging from 4 to 6 years of age ( $M = 4.57$ ,  $SD = .62$ ). They were recruited from neurofibromatosis clinics in the Midwest for the medical management of NF1, as part of a larger longitudinal study investigating cognitive and behavioral characteristics of children with NF1 and their siblings, and were diagnosed with NF1 by specialized physicians based on NIH Consensus Conference criteria (NIH, 1988).

This study is from Year 2 of a longitudinal study of cognitive development of young children with NF1. At the point of study enrollment, 71 families indicated interest in participating when approached, and 62 families participated in Year 1 of the study. Of these, 40 were eligible to be administered the CELF-P2 at the Year 2 visit. Eight of these participants did not return for Year 2 of the study, and two participants were not administered the language measure due to scheduling challenges. All participants were native speakers of English, and English was the primary language spoken in the home. The ethnic makeup of the sample was 21 Caucasian (70%), 3 African American (10%), 3 Hispanic/Latino (10%), 2 Asian (6.7%), and 1 mixed ethnicity (3.3%).

### 2.2. Measures

All measures used are norm-referenced with strong reliability and validity. All are appropriate and commonly used for administration to children ages 4–6, including children with developmental disabilities.

<sup>1</sup> Note that the sample of the current study consists of an overlapping sample; the current results are based on year two of a longitudinal study.

### 2.2.1. Cognitive ability

Children were administered the Differential Ability Scales – Second Edition – Early Years Form (*DAS-II*; C. D. Elliot, 1990). The *DAS-II* is a widely used, comprehensive measure of cognitive skills for children ages 2 years, 6 months through 17 years, 11 months. It yields a composite General Conceptual Ability (GCA) score, similar to an IQ score, which is used here as an index of overall level of intellectual functioning.

### 2.2.2. Language functioning

The Clinical Evaluation of Language Fundamentals – Preschool 2 (CELF-P2; Wiig et al., 2004) is a widely-used, comprehensive clinical tool designed to assess several domains of language abilities of preschool children (ages 3–6 years) and to help identify and diagnose language deficits. The CELF-P2 provides five composite scores: a Core Language score, and index scores for Receptive Language, Expressive Language, Language Content, and Language Structure. The Core Language score is used as the main indicator of a child's overall language abilities and helps determine the presence of a language disorder. The Receptive Language index measures listening and auditory comprehension. Expressive Language measures expressive skills/oral expression, Language Content assesses semantics (e.g., vocabulary, concepts, simple or complex sentence comprehension, word association comprehension), and Language Structure measures interpretation and production of word and sentence structure, including the incorporation of morphology and structural rules. The Core Language score and four indices are composed of varying combinations of several subtests, including Sentence Structure, Word Structure, Expressive Vocabulary, Concepts and Following Directions, Recalling Sentences, Basic Concepts, Word Classes-Receptive, and Word Classes-Total. Examples of subtest items include pointing to pictures, completing sentences, identifying objects, people, or activities, imitating sentences, finding word relations, rhyming words, reading, and demonstrating social language skills. Subtest scaled scores and composite standard scores are available.

### 2.2.3. Attention

The Conners Parent Rating Scales-Revised Short Form (Conners; Conners, 1997) is a standardized, commonly used measure assessing attention problems in children ages 3–17. This measure is completed by parents or caregivers and can aid clinicians in diagnosing ADHD. The Conners provides index scores for oppositionality, cognitive problems/inattention, hyperactivity, and an overall ADHD Index. Additionally, ratings on the Attention Problems scale of the Behavioral Assessment System for Children, Second Edition, Preschool form (BASC-2 PRS Preschool; Reynolds & Kamphaus, 2004) were used to assess parental report of child attention problems. The BASC-2 is a widely-used, broad parental-report screening measure of child behavior and adjustment. The Preschool form was designed for children ages 2–5.

### 2.2.4. Communication and Social Skills

The Functional Communication and Social Skills scales from the BASC-2 were used in this study. Ratings on the Developmental Social Disorders content scale were also examined; this scale indicates the level to which a child shows social behaviors representative of difficulty with social/communicative skills, social interests, and social activities. Additionally, the Social Interaction and Communication Skills standard score from the Scales of Independent Behavior – Revised (SIB-R; Bruininks, Woodcock, Weatherman, & Hill, 1996) was used. The SIB-R is a norm-referenced parental interview measure of child adaptive and maladaptive behavior designed for administration to parents or caregivers of children ages 0–80+ years. It yields scores for several adaptive behavior clusters, including motor skills, social interaction/communication, personal living, and community living.

## 2.3. Procedure

Participants were recruited through Neurofibromatosis specialty clinics at two Midwestern academic hospitals. At yearly specialty clinic medical checkups, the clinical geneticist explained the scope of the study, and interested families were referred to the study staff, who then contacted the family with further details. Study staff reviewed informed consent to provide a sense of the study parameters and set up an appointment. Questionnaire measures, including measures of attention problems and social skills, and an informed consent form were mailed in advance of the appointment and were reviewed and collected at the time of the appointment. During the appointment, the adaptive behavior measure was administered by study staff while another staff member conducted the assessment with the child. Children were individually administered an age-appropriate norm-referenced neuropsychological assessment battery (including the measures described) and were also administered some non-norm-referenced experimental measures, over a single three-hour session, with breaks as needed. Assessment took place in a quiet room. All work was IRB-approved.

## 2.4. Statistical analysis, testing for normality and outliers

Potential outliers for variables included in single and independent groups *t*-tests were identified as values extreme *z* scores ( $\pm 3.29$ ) as recommended by Tabachnick and Fidell (2007). Normality for single variables was assessed by testing for statistically significantly deviant skew or kurtosis ( $p < .01$ ) of the distributions (Tabachnick & Fidell, 2007). Potentially influential bivariate outliers were identified using the outlier test of the Car package in R that tests for significant deviations in the studentized residuals of the correlation (Stevens, 1984). Normality of relations was assessed by testing for statistically significant

deviations for skew or kurtosis of the distribution of residuals of each relation ( $p < .01$ ). As necessary, square root and log transformations were performed and are reported. In the case of bivariate outliers, both correlation coefficients are reported.

### 3. Results

SPSS version 20 and R version 2.15.0 were used to analyze data for this study. To control for multiple comparisons, a  $p$ -value of .01 was used; because of the small sample size, trend findings at  $p < .05$  are also reported. Assumptions of normality were fulfilled for the vast majority of variables and relations. The pattern of results did not change based on the very few outliers identified. To represent what is believed to be the range of functioning in children with NF1, all scores were included in the analyses reported, and analyses without outliers are also reported. In addition, ranked correlations supported the stability of this pattern of results.

Per the administration guidelines of the measure, one subtest (Basic Concepts) was administered only to participants aged 4 years ( $n = 15$ ), while another (Word Classes-Receptive) was only used to calculate composite scores for participants ages 5 and 6. Data were missing for two children on the Concepts and Following Directions subtest. No significant gender differences were seen on any composite or subtest scores of the DAS-II ( $p$  ranged from .80 to .94) or the CELF-P2 ( $p$  ranged from .42 to .94) using independent samples  $t$ -tests. No relations between standard scores and age for the DAS-II ( $p$  ranged from .12 to .87) or CELF-P2 ( $p$  ranged from .60 to .79) were observed using Pearson's correlations. See Table 1 for a summary of scores on the DAS-II and CELF-P2. 83.3% of the sample was within or above the 'average' range on the DAS-II for both overall cognitive ability (GCA) and verbal functioning, while the remaining children scored more than one or two standard deviations below the mean. This suggests that broad examination of cognitive and verbal abilities does not indicate difficulties for most children with NF1. Overall cognitive ability (GCA) was significantly related to all CELF-P2 standard scores when examined using Pearson's correlations ( $p$  all  $< .001$ ), confirming that intellectual functioning is related to language problems in children with NF1.

#### 3.1. Language abilities

Using a one-sample  $t$ -test, the mean Core Language score, as well as all index scores, fell in the average range, with no significant differences from the normative mean. There was a trend toward lower Receptive Language scores ( $p < .05$ ). Mean subtest scaled scores were also compared to those of the normative sample. Performance on the Concepts and Following Directions subtest was significantly lower than the normative mean, with a trend toward lower performance on the Word Classes-Receptive subtest. Both subtests relate to receptive language function.

To further explore the nature of language patterns in children with NF1, the number of children with performance more than one standard deviation below the normative mean was examined. Any standard score or subtest score falling at least one standard deviation below the mean signals that there is some level of difficulty (Wiig et al., 2004). See Table 2 for a summary of scores falling at least one standard deviation below the mean on the CELF-P2 and DAS-II.

**Table 1**  
Descriptive statistics and one-sample  $t$ -tests for intellectual functioning and language abilities.

Measure	Composite/Subtest score used	Mean standard/Scaled score	SD	df	$t$	$p$ -value
DAS-II	<b>GCA</b>	97.17	13.71	29	-1.13	.27
	<b>Verbal</b>	99.93	13.21	29	-.028	.98
	<b>Nonverbal</b>	100.30	15.05	29	.11	.91
	<b>Spatial</b>	93.47	13.84	29	-2.59	.02
CELF-P2	<b>Core Language</b>	96.40	17.74	29	-1.11	.28
	<b>Receptive Language</b>	93.56	14.71	26	-2.28	.03 <sup>+</sup>
	<b>Expressive Language</b>	95.07	19.51	26	-1.34	.19
	<b>Language Content</b>	95.85	14.49	26	-1.49	.15
	<b>Language Structure</b>	93.68	18.80	27	-1.78	.09
	Sentence Structure	9.23	3.00	29	-1.40	.17
	Word Structure	8.83	4.29	29	-1.49	.15
	Expressive Vocabulary	10.07	2.98	29	0.122	.90
	Concepts & Following Directions	7.82	3.18	27	-3.63	.001 <sup>+</sup>
	Recalling Sentences	8.86	3.41	27	-1.78	.09
	Basic Concepts <sup>a</sup>	9.60	3.74	14	-.41	.69
	Word Classes-Receptive	7.69	3.70	15	-2.5	.03 <sup>+</sup>
	Word Classes-Expressive	9.33	2.99	14	-.86	.40
	Word Classes-Total	8.40	3.27	14	-1.90	.08

<sup>a</sup> Smaller sample sizes on select subtests are due to administration to the age range specified by the CELF-P2 for calculation of composite scores. Basic Concepts was only administered to children ages 3–4, while Word Classes was administered to children ages 5–6.

<sup>+</sup>  $p < .05$ .

\*  $p < .01$ .

Table 2

Number of children falling 0, 1, 2, or 3 SD below the mean in intellectual and language functioning.

Composite/Subtest	n <sup>a</sup>	n (%) <1 SD below mean (average)	n/% 1–2 SD below (SS = 71–85; Sc = 4–7)	n/% 2–3 SD below (SS = 56–70; Sc = 1–3)	n/% >3 SD below (SS < 56; Sc < 1)	n/% at least 1 SD below
<b>CELF-P-2</b>						
<b>Core Language</b>	30	23 (76.7%)	4 (13.3%)	1 (3.3%)	2 (6.7%)	7 (23.3%)
<b>Receptive Language</b>	27	18 (66.7%)	7 (25.9%)	1 (3.7%)	1 (3.7%)	9 (33.3%)
<b>Expressive Language</b>	28	20 (71.4%)	4 (14.3%)	3 (10.7%)	1 (3.6%)	8 (28.6%)
<b>Language Content</b>	27	20 (74.1%)	6 (22.2%)	1 (3.7%)	0	7 (25.9%)
<b>Language Structure</b>	28	20 (71.4%)	3 (10.7%)	4 (14.3%)	1 (3.6%)	8 (28.6%)
Sentence Structure	30	24 (80.0%)	4 (13.3%)	2 (6.7%)	0	6 (20.0%)
Word Structure	30	21 (70.0%)	3 (10.0%)	6 (20.0%)	0	9 (30.0%)
Expressive Vocab	30	24 (80.0%)	5 (16.7%)	1 (3.3%)	0	6 (20.0%)
Concepts & Following Directions	28	16 (57.1%)	8 (28.6%)	4 (14.3%)	0	12 (42.9%)
Recalling Sentences	28	20 (71.4%)	4 (14.3%)	4 (14.3%)	0	8 (28.6%)
Basic Concepts	15	11 (73.3%)	3 (20.0%)	1 (6.7%)	0	4 (26.7%)
Word Classes–Recep.	16	9 (56.3%)	4 (25.0%)	3 (18.8%)	0	7 (43.8%)
Word Classes–Express.	15	12 (80.0%)	1 (6.7%)	2 (13.3%)	0	3 (20.0%)
Word Classes–Total	15	11 (73.3%)	2 (13.3%)	2 (13.3%)	0	4 (26.6%)
<b>DAS-II</b>						
<b>General Conceptual Ability</b>	30	25 (83.3%)	4 (13.3%)	1 (3.3%)	0	4 (13.3%)
<b>Verbal Comprehension</b>	30	25 (83.3%)	4 (13.3%)	1 (3.3%)	0	5 (16.6%)
<b>Nonverbal Reasoning</b>	30	22 (73.3%)	7 (23.3%)	1 (3.3%)	0	3 (10.0%)
<b>Spatial Ability</b>	30	23 (76.7%)	6 (20.0%)	1 (3.3%)	0	7 (23.3%)

<sup>a</sup> Complete sets of index scores were not calculated for three participants, whose data for one subtest was missing. Subtest administration also differed by age; 5–6 year olds were administered nine subtests, while 3–4 year olds were administered seven. Two children were missing subtest data for all but the three subtests comprising the Core Language composite score.

Overall, 7 of the 30 children (23.3%) performed at least one standard deviation below the mean on the Core Language composite. Notably, over one-third of the sample (11 of the 30 children) scored at least 1 SD below the mean on at least one index: two children on one index, three children on two indices, and six children on all four of the indices. In other words, in many cases different children displayed weaknesses in different areas - this set of children did not display identical patterns of strength and weakness. The index on which most children struggled was that of Receptive Language, with 9/27 children (33.3%) falling at least 1 SD below the mean (Receptive Language index scores were not available for 3 children). An area of particular difficulty appears to be that of Language Structure, for which 8/28 children (28.6%) scored at least 1 SD below the mean, with *five* of those (17.9%) scoring at least 2 SD below the mean. Another area of potential difficulty may be Expressive Language, as four children (14.3%) scored at least two SDs below the mean in this area. Additionally, when examining individual subtests, 17 of the 30 children - over *half* of the sample - scored at least 1 SD below the mean on at least one subtest, with the greatest difficulty seen on Concepts and Following Directions (42.9%) and Word Classes-Receptive (43.8%).

### 3.2. Language and functional communication, social interaction and communication, and social skills

To investigate the correspondence of lab-based findings to everyday functioning as described by parents in the areas of communication and social interaction, we examined Pearson bivariate correlations between performance on lab-based assessment of language functioning (CELF-P2) and parental report of functional communication (BASC-2 Functional Communication scale), social skills (BASC-2 Social Skills, Developmental Social Disorders scales), and social interaction and communication (SIB-R Social Interaction and Communication domain score). Core Language, Expressive Language, Language Content, and Language Structure were significantly related to Functional Communication. Expressive Language and Language Structure were significantly related to Social Skills.

Relations with the Developmental Social Disorders Scale consistently showed a residual outlier. Removal of this outlier did not change the pattern of results, though it did influence the results in some cases (see Table 3). Expressive Language and Language Structure showed robust relations to Developmental Social Disorders despite the removal of the outlier, while effects were somewhat weakened for Core Language (which moved from significance to a trend) and for Language Content and Receptive Language (which moved from trends to non-significance). Notably, no significant relations were found to Receptive Language using the BASC-2, though a trend existed for Functional Communication.

Significant relations were found for relations of SIB-R Social Interaction and Communication to Core Language, Expressive Language, Language Content, and Language Structure. Again, there was no significant relation to Receptive Language. Results of all correlational analyses in this section can be found in Table 3.

**Table 3**  
Bivariate correlations between parental report of social and communication skills and the CELF-P2.

CELF-P2 Composite	n	BASC-2			SIB-R
		Social Skills	Functional Comm.	Dev'tal Social Disorders	Social Interaction & Comm.
		r; p-value	r; p-value	r; p-value	r; p-value
Core Language	30	.40; .02 <sup>+</sup>	.58; .001 <sup>+</sup>	-.42; .02 <sup>*,a</sup> (-.47, .010 <sup>b</sup> )	.48; .007 <sup>+</sup>
Receptive Language	27	.35; .07	.43; .03 <sup>+</sup>	-.28; .17 <sup>a</sup> (-.45, .02 <sup>+</sup> )	.35; .07
Expressive Language	28	.51; .006 <sup>+</sup>	.65; <.001 <sup>+</sup>	-.51; .006 <sup>*,a</sup> (-.51, .005 <sup>+</sup> )	.58; .001 <sup>+</sup>
Language Content	27	.39; .04 <sup>+</sup>	.57; .002 <sup>+</sup>	-.32; .11 <sup>a</sup> (-.46, .02 <sup>+</sup> )	.39; .04 <sup>+</sup>
Language Structure	28	.50; .007 <sup>+</sup>	.59; .001 <sup>+</sup>	-.47; .01 <sup>*,a</sup> (-.51, .05 <sup>+</sup> )	.57; .002 <sup>+</sup>

<sup>a</sup> Residual outlier removed.

<sup>b</sup> Values within parenthesis are original analyses including outlier.

<sup>+</sup> p < .05.

<sup>\*</sup> p < .01.

### 3.3. Language and attention problems

Pearson's correlations were used to examine relations between the BASC-2 Attention Problems score and the Conners Indices and each composite score on the CELF-P2. Despite significant elevations, using a one-sample *t*-test, in comparison to the normative population on the BASC-2 Attention Problems scale ( $t[29] = 3.79$ ;  $p = .001$ ), Conners Cognitive Problems/Inattention,  $t(30) = 11.7$ ;  $p = .001$ , and Conners ADHD Index,  $t(30) = 10.4$ ;  $p < .001$ , and relations on the verge of significance for the Hyperactivity index,  $t(30) = 2.72$ ,  $p = .011$ , only one significant correlation between language abilities and attention problems was observed at the CELF P2 composite level: the relation between the BASC-2 Attention Problems scale and Expressive Language just reached the level of significance. Trends were noted for relations between the BASC-2 Attention Problems and Core Language, Language Content, and Language Structure. Several trends toward relations on the Conners were also noted at the composite level, including between: (1) Core Language and the ADHD Index; (2) Expressive Language and Oppositionality, Hyperactivity, and the ADHD Index; (3) Language Content and the ADHD Index; (4) Language Structure and both Oppositionality and the ADHD Index. It is notable that no relations or trends were seen between Receptive Language and attention problems based on either measure. At the CELF-P2 subtest level, Word Structure was significantly related to BASC-2 Attention Problems, and a trend was seen for Recalling Sentences. Trends for relations to the Conners existed for Word Structure, Concepts and Following Directions, Recalling Sentences, and Word Classes-Expressive subtests. Relations of Word Structure and Word Classes-Expressive to the ADHD Index, as well as relations between Recalling Sentences and Oppositionality, just reached significance, and Word Classes-Total was significantly related to the Conners' ADHD Index after removal of the outlier. Correlations are shown in Table 4.

**Table 4**  
Bivariate correlational analyses between Conners indices and the BASC-2 with the CELF-P2.

CELF-P2 Composite/Subtest	n	Conners Opposition.	Cognitive Prob/Inattention	Hyperactiv.	ADHD Index	BASC-2 Attention problems
		r; p-value	r; p-value	r; p-value	r; p-value	r; p-value
<b>Core Language</b>	30	-.28; .13	-.30; .12	-.25; .18	-.38; .04 <sup>+</sup>	-.40 .03 <sup>+</sup>
<b>Receptive Language</b>	27	-.22; .28	-.22; .27	-.07; .72	-.31; .12	-.32; .12
<b>Expressive Language</b>	28	-.38; .04 <sup>+</sup>	-.28; .14	-.40; .04 <sup>+</sup>	-.43; .02 <sup>+</sup>	-.46; .01 <sup>+</sup>
<b>Language Content</b>	27	-.28; .15	-.27; .17	-.22; .28	-.39; .04 <sup>+</sup>	-.43; .03 <sup>+</sup>
<b>Language Structure</b>	28	-.38; .048 <sup>+</sup>	-.27; .16	-.34; .08	-.42; .03 <sup>+</sup>	-.41; .03 <sup>+</sup>
Sentence Structure	30	-.05; .77 <sup>a</sup>	-.15; .43	-.002; .99	-.18; .34	-.07; .70
Word Structure	30	-.43; .018 <sup>+</sup>	-.35; .06	-.38; .04 <sup>+</sup>	-.47; .01 <sup>+</sup>	.56; .001 <sup>+</sup>
Expressive Vocabulary	30	-.18; .34 <sup>a</sup>	-.27; .16	-.23; .21	-.30; .12 <sup>b</sup> (-.32; .08 <sup>a,c</sup> )	-.35; .06
Concepts & Follow Dir.	28	-.24; .22	-.28; .15	-.26; .18	-.38; .049 <sup>+</sup>	-.36; .06
Recalling Sentences	28	-.46; .01 <sup>*,a</sup>	-.21; .29	-.42; .03 <sup>+</sup>	-.41; .03	-.40; .04 <sup>+</sup>
Basic Concepts	15	-.23; .38	-.24; .39	-.07; .80	-.31; .26	-.42; .12
Word Classes-Recep.	16	-.22; .43	-.46; .07	-.30; .26	-.42; .12	-.25; .35
Word Classes-Express.	15	-.24; .39	-.54; .04 <sup>+</sup>	-.35; .21	-.63; .01 <sup>+</sup>	-.27; .34
Word Classes-Total	15	-.24; .40	-.50; .06	-.32; .25	-.70; .005 <sup>*,b</sup> (-.50; .06)	-.24; .39

<sup>a</sup> With transformed variable.

<sup>b</sup> With 1 outlier removed.

<sup>c</sup> Values within parenthesis are original analyses including outlier.

<sup>+</sup> p < .05.

<sup>\*</sup> p < .01.

## 4. Discussion

This investigation of early language abilities in children with NF1 revealed weaker language skills in comparison to the normative population, with especially common difficulties related to receptive language and language structure. At the task level, particular difficulties understanding relationships between words and interpreting and following verbal directions were noted, and a large proportion of children showed language structure challenges. It is notable that there was good correspondence between lab-based assessment of language functioning and parental descriptions of their children's communication abilities in everyday contexts, and expressive language functioning was related to social functioning. However, while difficulties in the area of receptive language were most frequently observed, these were not significantly related to functional communication or social functioning as described by parents and therefore likely represent a separate area of vulnerability for children with NF1. Together these results provide evidence that even young children with NF1 experience clear language difficulties, and these difficulties appear to be a risk factor for everyday communication and social interaction challenges. The lack of clear and strong relations between attention problems and language functioning suggests that the language difficulties observed in this sample of children with NF1 appear to be more than simply a product of poor attention skills.

### 4.1. Language abilities

While mean performance on the language measures was not clearly below average, a substantial number of children with NF do appear to have significant problems in one or more areas of language, as hypothesized. Scores on the Core Language composite, which gauges a child's overall language performance, revealed language difficulties for almost a quarter of the sample. Over one-third of the sample showed difficulty on at least one of the other four indices. This is about twice as many children as would be expected based on the normative population (Wiig et al., 2004).

For children who struggled on one or more language indices, the most frequent areas of difficulty were in Receptive Language and Language Structure, with difficulties in Language Structure being more pronounced when present. Low language structure scores signify difficulty understanding structural rules (e.g. morphology and syntax), and low receptive language scores signify difficulty listening to and comprehending auditory information (Wiig et al., 2004). Problems with receptive language scores were expected based on literature suggesting receptive language problems in children with NF1 (e.g., Dilts et al., 1996; North et al., 1994; Thompson et al., 2010). Problems with language structure in this population have not been closely examined and warrant further attention given the convergence of our findings with those of Thompson and colleagues (2010), who found difficulties on all subtests in the Language Structure index, and Lorenzo and colleagues (2011), who found high rates of delayed irregular word use, sentence complexity, and vocabulary. Additionally, Geurts and Embrechts (2008) suggested that language structure skills are related to ADHD symptoms. However, in this study there were no significant relations between attention difficulties and either receptive language or language structure difficulties, such that attention difficulties alone likely do not account for these challenges. Our findings clarify specific areas of weakness at the index and subtest level and provide evidence that a number of children with NF1 are at risk for significant language difficulties apart from attention problems, despite overall "average" scores when group means are examined.

We had expected to see difficulty on all subtests making up the expressive and receptive language indices based on findings by Thompson and colleagues (2010). However, our sample performed significantly lower than the normative mean on just one subtest: Concepts and Following Directions (which deals with the ability to recall and follow instructions). The inconsistency may be due to potential differences in intellectual functioning between the samples; Thompson and colleagues did not report the specific cognitive functioning level of their sample for comparison. Areas of frequent difficulty were seen at the individual subtest level, especially on Concepts and Following Directions and Word Classes-Receptive (which demonstrates a child's ability to categorize, extend meaning, or associate words). Given the number of children who showed challenges at the subtest level, it seems that a large number of children with NF1 struggle with language despite mean scores that do not differ significantly from the norm.

It is notable that more children showed language difficulties than intellectual difficulties. Although most of the sample fell in the 'average' range intellectually, just two-thirds of the sample fell in the 'average' range on every index of the CELF-P2; one-third showed difficulty on at least one index. Therefore, some children showed language difficulties that likely would have gone undetected if language was examined using only a measure of intellectual abilities.

### 4.2. Social skills and communication

Language difficulties observed in the lab setting were observed to translate to real-world communication problems. Children with lower language abilities typically experienced greater challenges when dealing with daily social situations. Parental report of social and communicative behaviors in day-to-day life showed a clear connection between difficulty with these behaviors and language problems. Our finding that core language and expressive language skills strongly relate to functional communication expands upon the suggestion that verbal functioning is related to real-world social skills and functional communication in children with NF1 (Klein-Tasman et al., 2014). Language abilities were also related to adaptive skills that draw on social interaction and communication abilities, a finding which helps to extend prior research on adaptive behavior (i.e., Barton & North, 2004; Klein-Tasman et al., 2013) by investigating relations to language. Clearly, children with

NF1 who have language difficulties are experiencing complications in social contexts. Language problems could undoubtedly affect a child's ability to read and learn in the classroom, but our findings show that language difficulty does appear to translate into problems with communication in the real world to the extent that it may affect a child's academic performance, home life, and peer relationships, which could all have significant ramifications.

Interestingly, the area of language in which most children with NF1 struggled, receptive language, was the only index not significantly related to any parent-reported adaptive or behavior problems, including attention problems (though trends existed for relations to Functional Communication). We suggest that receptive language deficits, in particular, could be a vulnerability for some children with NF1 separate from other complications. While receptive language could clearly affect academic functioning, it does not appear to have a substantial impact on social skills and communication in young children with NF1 to the extent that deficits in other areas of language do. Further longitudinal consideration of relations between early receptive language abilities and later psychosocial and learning functioning is warranted.

#### 4.3. Language and attention

Contrary to our expectations, attention problems were not clearly related to language difficulties, with very few relations reaching the level of significance. Sangster and colleagues (2011) reported that inattentive symptoms of ADHD occur at a striking rate in children with NF1; our sample also showed significantly elevated parent-reported symptoms of ADHD. Our results are also consistent with findings that difficulties with inattention are more striking than difficulties with hyperactivity for children with NF1. Hence, language difficulties may represent a separate risk factor for later learning problems for children with NF1 and, as suggested by Koth and colleagues (2000), attention problems may be a distinct component of the NF1 phenotype. However, it remains possible that language problems and attention problems reflect a shared underlying vulnerability for children with NF1. Moderate effect sizes were indeed observed for relations between attention problems and the vast majority of language composite scores (except Receptive Language) such that a role for attention problems in language functioning would likely be observed with a larger sample size. However, it appears that attention difficulties alone (which are often seen in children with NF1) are not sufficient to explain deficits observed in receptive language. Our findings help support the suggestion by Koth and colleagues that language problems may emerge as a component of the NF1 phenotype for some of children with NF1 and that these difficulties are evident even in the preschool years.

### 5. Limitations, future directions, and conclusion

There are several limitations to the current study. First, although the sample is larger than in prior work about language functioning in preschoolers with NF1, it is nevertheless still small. Additionally, mean intellectual functioning was stronger than is generally present for children with NF1, such that this sample may represent a somewhat more cognitively capable group than is typical. Finally, in this study we relied on parental report of attention problems and social functioning, and it is possible that parents of preschoolers may not be highly attuned to potential attention problems or social challenges in their children.

While limitations exist and replication is necessary, these results nevertheless suggest that difficulties with language, particularly structural aspects of language, are indeed present for some children with NF1 despite average range cognitive functioning. Administration of a broad language assessment measure appears to be quite useful in detecting more subtle – but still functionally significant – language difficulties that are not fully captured by more general measures of cognitive functioning. Finally, lab-assessed language problems translate to real-world social functioning. This knowledge may be helpful for clinicians in catching language problems that could otherwise be overlooked in young children with NF1.

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