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Effect of congenital heart defects on language development in toddlers with Down syndrome

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Abstract

Background—Down syndrome (DS, OMIM #190685) is the most commonly identified genetic form of intellectual disability with congenital heart defect (CHD) occurring in 50% of cases. With advances in surgical techniques and an increasing lifespan, this has necessitated a greater understanding of the neurodevelopmental consequences of CHDs. Herein, we explore the impact of CHD on language development in children with DS.

Methods—Twenty-nine children with DS were observed systematically in parent–child interactions using the Communication Play Protocol to evaluate their language use; they also completed the Mullen Scales of Early Learning and MacArthur Communication Development Inventory. Mean ages were 31.2 months for children with DS and CHD (DS + CHD, $n = 12$) and 32.1 months for children with DS and a structurally normal heart (DS – CHD, $n = 17$).

Results—Compared with the DS – CHD controls, the DS + CHD group revealed lower scores in multiple areas, including fine motor skills and expressive and receptive vocabulary. Whereas most differences were not statistically significant, the Communication Development Inventory word count and symbol-infused joint engagement differed significantly ($P < 0.01$) and marginally ($P = 0.09$) between groups.

Conclusions—Finding that CHDs may account for part of the variation in language delay allows us to consider the specific mechanisms underlying the impact of CHDs on language acquisition in children with DS. Conclusions from this first study on early language outcomes of children with DS + CHD may be useful for clinicians in providing developmental surveillance and early intervention programmes with specific emphasis on language therapy as part of long-term follow-up for children with DS + CHD.

Keywords

congenital heart defects; Down syndrome; intellectual disability; language

Introduction

Down syndrome (DS, OMIM #190685) is the most common genetic cause of intellectual and developmental disabilities and results from an extra chromosome 21 (Trisomy 21). The incidence is approximately one in 691 live births (Parker *et al.* 2010), which suggests that approximately 6000 of the ~4 million infants born each year in the USA have DS. Although characteristic facial appearance, intellectual disability, and hypotonia are present in virtually

all individuals with DS, the expression of these features vary considerably. Likewise, congenital and acquired medical complications are variable among individuals with DS, but present at a higher frequency than the general population. For example, 41–56% of all individuals with DS have congenital heart defects (CHDs) (Freeman *et al.* 1998, 2008; Stoll *et al.* 1998; Torfs & Christianson 1998), 5% manifest various gastrointestinal defects, and 0.6% develop leukaemia (Roizen 2001). Additionally, language impairments are common in DS, particularly in expressive language, phonology, articulation and grammar (Chapman *et al.* 2000). Although it is well recognized that hearing and oral structure abnormalities contribute to such language difficulties, the impact of CHD on language development in children with DS has not been described.

Studies of CHD in children without DS indicated that a range of skills including language may be impaired even when the CHD is repaired (Miller *et al.* 2007; Hovels-Gurich *et al.* 2008; Majnemer *et al.* 2008). Despite a significantly higher frequency of CHDs in DS, research on the neurodevelopmental outcome of individuals with DS and CHD is to date limited to one study (Visootsak *et al.* 2011). That study compared the neurodevelopmental outcome of 17 children with DS and no CHD to the outcome of 12 children with DS and atrioventricular septal defects (AVSD), the most common CHD in DS. Children with DS + AVSD (mean age = 14.5 ± 7.3 months) had greater developmental delay than children with DS and no CHD (mean age = 14.1 ± 8.4 months). The DS + AVSD cases exhibited lower composite scores in all domains relative to DS – CHD controls. Although the motor domain was the only domain that showed a statistically significant difference between groups ($P < 0.05$), both cognitive scores ($P = 0.63$) and language composite scores ($P = 0.10$) were marginally lower in the DS + AVSD cases compared with the DS – CHD controls.

The study reported here extends Visootsak *et al.*'s (2011) work by elucidating the effect of CHD on the language of children with DS. In addition to comparing the performance of children with DS + CHD to children with DS – CHD on standardized tests, we compared observations of language use during parent–child interactions. Prior studies suggest that although young children with DS are as able to sustain periods of joint engagement during which they attend to a shared object or event with their partners as mental age matched typically developing children, they are significantly less able to infuse these periods with symbols, including language (Adamson *et al.* 2009). Our primary question is whether, among children with DS, having a CHD is related to lower scores of language skills and less use of language during social interactions. As this is the first study to examine early language outcomes of children with DS + CHD, our findings would have important implications because early intervention may foster prelinguistic and early linguistic skills, and possibly increase potential for community inclusion and independent living.

Method

Participants and procedure

The participants were 29 children (12 DS + CHD and 17 DS – CHD) who also participated in a study of the joint engagement and emergence of language in children with autism compared with those with DS (Adamson *et al.* 2009). There were 19 men and 10 women; 79% were European American, 21% were African American, and 83% had completed college. Participants were referred to Georgia State University by their healthcare providers or were self-referred after learning about the research study at local parent support events. English was the primary language in all homes. The mean age at the time of the parent–child observation was 31.2 months for children with DS + CHD ($SD = 3.2$, range = 27.5–39.4) and 32.1 for DS – CHD controls ($SD = 3.7$, range = 25.9–40.5), not a significant difference.

The CHD status was determined through parent report. Controls were patients with DS who had a structurally normal heart. The severity of the CHD ranged from minor defects ($n = 7$), such as atrial septal defects, ventricular septal defects, or mitral valve dysfunction that did not require surgery, to major defects ($n = 5$) such as ventricular septal defects, AVSD, and/or congestive heart failure requiring surgical repair.

Measures

Three language measures were employed. A count of words in the child's expressive vocabulary was made using the *MacArthur Communication Development Inventory* (CDI) (Fenson *et al.* 1993) that the mothers completed within a week of the observation session. Although originally intended for typically developing children, it is highly reliable and well validated for children with DS (Miller *et al.* 1995; Caselli *et al.* 1998; Zampini & D'Odorico 2009). A standardized norm-referenced assessment was made using the *Mullen Scales of Early Learning* (Mullen 1995) that produced scale scores for expressive and receptive language as well as scale scores for visual reception and fine motor skills. The assessment of language use was derived from observations of parent-child interactions that were made using the Communication Play Protocol (CPP) (Adamson *et al.* 2004), a procedure that produces video records during which the parent encourages child communication in six 5-min scenes that focus on three common communication functions: interacting, requesting and commenting. Trained coders reliably used an 11-code scheme to characterize the child's attention to people, objects and symbols (Adamson *et al.* 2004). For the current study we derived four variables: *total joint engagement* (the per cent of time the child actively focused on an object or event with the partner), *coordinated joint engagement* (the per cent of time the child actively attended to both partner and a shared object or event), *supported joint engagement* (the per cent of time the child actively focused on an object or event without acknowledging the partner), and *symbol-infused joint engagement* (the per cent of time the child actively attended to symbols, most often language, as well as focusing on an object or event with the partner).

Results

As seen in Table 1 and Fig. 1, parents reported significantly smaller vocabularies on the CDI for children with DS + CHD [median = 9 vs. 44 words, $P = 0.01$; Cohen's $d = 0.74$, an effect size in a range – 0.5–0.8 – that Cohen (1988) characterized as moderate]. On the Mullen Scales (see Table 1 and Fig. 2), both expressive ($P = 0.12$) and receptive language ($P = 0.19$) scores were relatively lower for the DS + CHD group compared with DS – CHD, although not statistically significant in this small sample, Cohen's $d = 0.54$ for both – while the visual ($P = 0.88$) and fine motor ($P = 0.84$) scores were not affected. Because most scores were skewed, medians are given in Table 1 and Mann-Whitney tests were used to test for group differences; t -tests gave essentially the same results.

During the parent-child interactions observed with the CPP, children with CHD spent less time in symbol-infused joint engagement ($P = 0.09$, Cohen's $d = 0.85$; see Table 1 and Fig. 3), a state during which they use language as well as focus on shared objects. Total joint engagement and non-language dependent forms of joint engagement did not differ.

Discussion

As advances in surgical techniques have improved the survival rates and lifespan in children with DS and CHD, studies to understand the course of their neurodevelopmental profile will have important implications. This study replicates the previous finding that children with DS + CHD score lower on standardized assessments of language than children with DS – CHD (Visootsak *et al.* 2011). Moreover, it documents that this pattern is evident as well when

language is evaluated using parent reports of early expressive vocabularies and during parent–child interactions.

It is well known that language is among the most impaired domains of functioning in DS, particularly in language production, syntax and intelligibility, which may be further complicated by their dysmorphic facial features, including a small oral cavity, a narrow, high-arch palate, and a large tongue that protrudes forward (Roberts *et al.* 2007). Children with DS produce their first words at a much later age than typically developing children. Whereas the latter begin to produce their first words by 12 months of age, the average age for this milestone in children with DS is approximately 21 months, although many do not produce their first words until much later (Stoel-Gammon 2001). Once they begin to produce words, children with DS continue to make slow progress compared with their typically developing peers (Yoder & Warren 2004). What is far less studied are variations in their well-documented language delay. Finding that CHDs account for part of the variation in language delay is interesting because it may prompt consideration of the specific mechanisms underlying the negative impact of CHDs, both for children with DS and for those without DS, on language acquisition. In previous studies of other CHD subtypes in typically developing children, perioperative management strategies and specific patient characteristics have been identified as potential factors influencing neurodevelopmental outcomes, including language development (Bellinger *et al.* 1999, 2003; Mahle & Wernovsky 2001; Gaynor *et al.* 2003, 2007; Mahle *et al.* 2006). We recognize that our small sample size makes it impossible to evaluate any of these factors. Nevertheless, our findings set the foundation for further longitudinal studies with a larger sample size to evaluate the possible contributions of a variety of perioperative variables (e.g. cardiopulmonary bypass, deep hypothermic circulatory arrest, haemodilution, hypoxaemia, low cardiac input, length of hospital stay) and patient-related determinants (e.g. parental education, gender, race, presence of a genetic syndrome) that may play a role in the neurodevelopmental outcomes, particularly in the language domain, of children with CHDs.

We recognize that our DS + CHD cohort was not homogenous in terms of type of defect. The small sample makes it impossible to correlate the severity of the CHD to language outcome. Nonetheless, our findings are relevant with regard to the delineation and interventional implications of distinct language outcomes in children with DS + CHD. Of particular note is the impact of CHD on the infusion of symbols during episodes of joint engagement during parent–child interactions. The findings that the total amount of joint engagement and the amount of the coordinated or supported forms of joint engagement were not associated with CHD suggests that the preverbal attentional foundation for communication is not as vulnerable in DS as the expansion of this attentional structure that occurs as language is acquired. This conclusion supports current efforts to provide early language interventions, including those that specifically supplement speech with augmented and alternative modes of symbolic communication that can facilitate symbol-infused joint engagement as often and as early as possible (Adamson *et al.* 2010).

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Figure 1.
 Open circles represent children with DS – CHD, gray circles children with DS + CHD.
 Solid horizontal lines represent group medians for the CDI. CDI, Communication
 Development Inventory; CHD, congenital heart defect; DS, Down syndrome.

Figure 2.

Open circles represent children with DS – CHD, gray circles children with DS + CHD. Solid horizontal lines represent group medians for the Mullen variables. CHD, congenital heart defect; DS, Down syndrome.

Figure 3. Open circles represent children with DS – CHD, gray circles children with DS + CHD. Solid horizontal lines represent group medians for the joint engagement variables. CHD, congenital heart defect; DS, Down syndrome.

Table 1

Descriptive statistics for joint engagement and language scores

Variable	DS + CHD (<i>n</i> = 12)				DS - CHD (<i>n</i> = 17)				<i>d</i>	<i>P</i>
	50th	(25th–75th)	min–max		50th	(25th–75th)	min–max			
CDI	9	(5–37)	0–129	44		(19–88)	8–273	0.74	0.01	
Mullen composite	51	(49–58)	49–68	56		(50–63)	49–89	0.45	0.32	
Mullen visual	20	(17–25)	16–27	20		(18–23)	12–32	0.12	0.88	
Mullen motor	20	(18–22)	14–24	20		(18–22)	12–27	0.08	0.84	
Mullen receptive	14	(12–18)	11–24	18		(14–25)	8–27	0.54	0.19	
Mullen expressive	15	(13–15)	10–18	16		(14–17)	6–29	0.54	0.12	
Total joint engagement	74	(64–79)	54–82	72		(68–79)	18–93	0.04	0.96	
Supported joint engagement	53	(48–59)	22–65	49		(41–57)	18–78	0.32	0.25	
Coordinated engagement	17	(12–22)	1–59	18		(15–28)	0–67	0.22	0.45	
Symbol-infused joint engagement	1	(0–4)	0–5	5		(1–15)	0–39	0.85	0.09	

Note. Scores are percentiles because of generally skewed distributions; *d* = Cohen's *d*. *P* values are per a Mann–Whitney *U*-test. CDI, Communication Development Inventory; CHD, congenital heart defect; DS, Down syndrome.