Low speech rate but high gesture rate during conversational interaction in people with Cornelia de Lange syndrome.

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Acknowledgements: The authors would like to thank all the participants and families who took part in this research. This research was funded by Cerebra and Cornelia de Lange Foundation, UK and Ireland.

Disclosure: The authors have no conflicts of interest. We confirm that we have read the Journal's position on issues involved in ethical publication and affirm that this report is consistent with those guidelines.

Data availability: The data that support the findings of this study are available on reasonable request from the corresponding author. The data are not publicly available due to privacy or ethical restrictions.

Abstract

Background: Cornelia de Lange syndrome (CdLS) is a rare genetic syndrome with notable impaired expressive communication characterised by reduced spoken language. We examined gesture use to refine the description of expressive communication impairments in CdLS. Method: During conversations, we compared gesture use in people with CdLS to peers with Down syndrome (DS) matched for receptive language and adaptive ability, and typically developing (TD) individuals of similar chronological age. Results: As anticipated the DS and CdLS groups used fewer words during conversation than TD peers ($p<.001$). However, the CdLS group used twice the number of gestures per 100 words compared to the DS and TD groups ($p = .003$). Conclusions: Individuals with CdLS have a significantly higher gesture rate than expected given their level of intellectual disability and chronological age. This result indicates the cause of reduced use of spoken language does not extend to all forms of expressive communication.

Key words: Communication, gesture, speech, social engagement, Cornelia de Lange syndrome, Down syndrome, social anxiety

Cornelia de Lange syndrome (CdLS) is a rare genetic syndrome (estimated prevalence 1:10,000 – 1:62,000; Kline et al., 2007; Barisic et al., 2008) caused by mutations of the NIPBL, SMC1A, SMC3, HDAC8 and RAD21 genes (Deardorff et al., 2007, 2012a,b; Dorsett & Krantz, 2009; Gillis et al., 2004; Krantz et al., 2004; Yan et al., 2006). Genetic aetiology for a proportion of people with confirmed clinical diagnosis remains unknown. Intellectual disability (ID) ranges from severe to mild (Kline et al., 2018).

Physical phenotype includes distinctive facial features, upper limb abnormalities, vision and hearing problems (Kline et al., 2018; Liu & Krantz, 2009) with compromised health (Berg et al., 2007; Hall et al., 2008). Phenotypic behaviours include self-injury (Basile et al., 2007; Berney et al., 1999, Oliver et al., 2009), autistic characteristics (Moss et al., 2008; Oliver et al., 2011; Richards et al., 2015), and social impairments including selective mutism, social anxiety, social avoidance and extreme shyness (Crawford et al., 2017; Moss et al., 2016; Nelson et al., 2017; Richards et al., 2009). Impairments and delay of communication are evident (Ajmone et al., 2014; Hoddell et al., 2011; Sarimski, 1997, 2002) with expressive language more impaired than receptive, and written language (Goodban, 1993; Oliver et al., 2008; Ajmone et al., 2014).

The specificity of communication impairments in CdLS has not been explicated (Kline et al., 2007; Liu & Krantz, 2009). The wide variability from people speaking no words to use of full sentences (Sarimski, 1997, 2002; Goodban, 1993) is associated with level of ID (Liu & Krantz, 2009), birth weight, hearing impairments, upper limb abnormalities, developmental milestones and social relatedness (Goodban, 1993). Some communication impairments independent of chronological age or ID include specific impairments in motor imitation (Hoddell et al., 2011) and fewer intentional and joint attention communicative acts (Sarimski, 2002).

Selective mutism is evident (Moss et al., 2016; Nelson et al., 2017; Richards et al., 2009), with demonstrably less speech when individuals are expected to initiate conversation (Crawford et al., 2020; Nelson et al., 2017; Richards et al., 2009). It is unclear whether this is due to reduced motivation for social engagement (Nelson et al., 2017; Crawford et al., 2020) or impairment of spoken language exacerbated by the social context. Examination of gesture use in a social context, in which communication is expected to be initiated, could indicate the specificity of reduced expressive communication in that context. If reduced motivation for social engagement drives selective mutism it would be expected that use of gestures would be reduced.

Given the incomplete profiling of communication impairments in CdLS, we describe the rate of gestures in CdLS in a social context in contrast to people of typical development (TD) comparable for chronological age and people with Down syndrome (DS) of comparable chronological age and degree of ID. Using these contrasts we address the question of whether the rate at which people with CdLS use gestures is comparable to or different from the rate for TD peers and/or by people with another genetic syndrome with comparable degree of ID. A contrast group of individuals with DS was used because of similarity to CdLS for developmental quotients (Reid et al., 2017; Richman et al., 2009) and expressive language deficits (Goodban, 1993; Martin et al., 2009).

Method

Participants

Footage and background data for participants with CdLS and DS were taken from a previous study of social behaviour and executive functioning (see Nelson et al., 2017; Reid et al., 2017). Participants were included if they: 1) had a diagnosis of CdLS or DS, 2) were aged 12 or over so hearing problems were likely resolved, 3) could speak at least thirty words in

two-to-three-word phrases, 4) scored at least six on the self-help subscale of the Wessex Scale (Kushlick et al., 1973) and 5) had a receptive vocabulary age equivalence of 40 months or above on the Vineland Adaptive Behavior Scale (VABS; Sparrow, Balla & Cicchetti, 1984).

Of the 45 individuals recruited, 15 were excluded because in video footage they spoke too infrequently or gestures were obscured. Consequently, 15 participants with each syndrome were included and matched for chronological age, receptive language as measured by the British Picture Vocabulary Scale 2 (Dunn et al., 1997), and adaptive behaviour measured by the VABS. The groups did not differ on gender or hearing status. See supplementary information for statistics.

A chronological age-matched, TD paid participant group (n = 15) provided a benchmark of typical performance. Exclusion criteria were diagnosis of a developmental disorder or special educational needs school attendance. The TD group was comparable to the CdLS and DS groups for gender (see supplementary information for statistics). Participant characteristics are reported in table 1.

++++++++++++++++++++INSERT TABLE 1 ABOUT HERE++++++++++++++++++

Procedure

Video footage across groups ranged from four to eight minutes in length. To standardise clips, three-minute segments that began thirty seconds after the first conversational question (enabling warm-up) were coded.

Measures

Social tasks

Clips from the *Required Social Interaction* condition with an unfamiliar adult (experimenter) from Nelson et al., (2017) were used. During this condition, an adult initiated

conversation by asking questions. The range of topics included holidays, free time, family and sports. This condition was chosen as it emulated everyday social interaction.

Examiner variables

No significant differences across groups were found for the number of words spoken by the experimenter ($F(2)= 1.90$, $p = .163$, partial $\eta^2 = .08$). However, the examiner asked fewer questions of TD participants ($F(2) = 4.07$, $p = .024$, partial $\eta^2 = .16$). The CdLS and DS groups did not differ on this variable ($t(28) = .383$, $p = .706$, $d = 0.14$).

Gesture coding

Gestures were defined as communicative hand and head movements expressing a message in coordination with speech (McNeill, 1992). Gesture coding using ELAN 3.9.0 (Max Planck Institute for Psycholinguistics, 2010), focussed on the stroke phase of gestures only (i.e. the most forcefully executed movement segment; Kita et al., 1999) without preparation and retraction phases. This ensured the meaning encoding part of the gesture (McNeill, 1992) was recorded. Types of gestures were combined into an overall 'Gesture' code. Inter-rater reliability assessed for 20% of the data indicated 90% agreement for occurrence of gestures.

Spoken language coding

Spoken language was transcribed with each conversational turn, and utterance within a turn, separated for analysis. Hesitations (e.g. "um", "err") were counted as words for the calculation of MLU (Mean Length of Utterance) and gesture rate because they co-occur with gestures (Butterworth & Beattie, 1978; Navretta, 2015) and therefore inclusion affords the opportunity for gesture. Percentage agreement for 20% of the data for the identification of individual words, utterances and turns was 97.7%, 90.1% and 96.7% respectively.

Data Analysis

Dependent variables were spoken language and gesture rate. A gesture rate (the number of gestures / 100 words) indicates gesture use and standardises frequency for participants with CdLS or DS who would speak less than TD participants. For spoken language, transcription data were used to calculate the mean number of words produced, MLU, utterances and turns during the segment. Unintelligible speech was not counted.

Shapiro-Wilk tests showed almost all data were normally distributed. One-way ANOVAs and post-hoc t-tests were conducted to identify group differences and explore significant results, respectively. Effect sizes were calculated for post-hoc tests using Cohen's d (small=.2, medium=.5, large=.8) (Cohen, 1988).

Results

Spoken language

Table 2 summarises characteristics of the groups' expressive language. Groups did not differ significantly in the number of conversational turns or utterances. As anticipated, the TD group's MLU was significantly longer than the other two groups (CdLS ($t(20) = -4.98$, $p<.001$, $d = 1.82$), DS ($t(16) = -5.44$, $p<.001$, $d = 1.99$)). MLU did not differ significantly between the CdLS and DS groups ($t(28) = -.37$, $p = .714$, $d = 0.06$).

++++++++++++++++++++INSERT TABLE 2 ABOUT HERE++++++++++++++++++++

The CdLS, DS and TD groups spoke a mean of 167.5 (range = 39 – 423), 178.4 (range = 68 – 307) and 434.4 (range = 239 – 552) words, respectively (Figure 1, upper panel). Groups differed significantly for number of words produced ($F(2,42) = 39.63$, $p<.001$, partial $\eta^2 = .654$). Post hoc analyses revealed a difference between the TD group and both

the CdLS, and DS groups, with participants with CdLS ($t(28) = -7.26$, $p<.001$, $d = 2.65$) and DS ($t(28) = 8.39$, $p<.001$, $d = 3.06$) producing significantly less words than the TD group, while not differing statistically from each other ($t(28) = -.32$, $p = .753$, $d = 0.12$).

++++++++++++++++++++INSERT FIGURE 1 ABOUT HERE++++++++++++++++++++

Gesture rate

The mean number of gestures produced per 100 words for the CdLS, DS and TD groups was 28.5 (range = 5.80 – 64.65), 15.34 (range = 0.40 – 27.06) and 13.08 (range = 6.94 – 19.76) respectively (see Figure 1, lower panel)

There was a significant group difference for gesture rate ($F(2,42) = 6.70$, $p = .003$, partial $\eta^2 = .242$). The CdLS group evidenced a significantly higher gesture rate (approximately double that of other groups) than both the DS group ($t(17) = 2.39$, $p = .028$, $d = 0.87$) and TD group ($t(15) = 2.93$, $p = .010$, $d = 1.07$). TD and DS groups did not differ statistically ($t(20) = 1.12$, $p = .277$, $d = 0.04$).

Discussion

We investigated the use of gestures by people with CdLS in comparison to participants with DS and chronological age-matched TD peers during social interactions. This is the first study directly investigating gestures in CdLS. As anticipated, both the CdLS and DS groups spoke significantly less than the TD group. This is consistent with the well-documented impairment in expressive language related to the degree of ID present in both syndromes and delayed with respect to chronological age (Abbeduto et al., 2001; Ajmone et al., 2014; Chapman & Hesketh, 2000; Goodban, 1993; Sarimski, 2002). Participants with CdLS produced gestures at a rate almost double that for either comparison group, with individuals with DS producing gestures at a rate comparable to TD peers. This is surprising given it is

reported that individuals with Down syndrome have relative strengths in gesture (Caselli et al., 1998; Singer-Harris et al., 1997). Importantly, on measures of spoken language no differences were found between the CdLS and DS groups, suggesting comparability for spoken language ability. Consequently, the significantly higher rate of gestures in the CdLS group cannot be explained by the degree of associated ID or spoken language ability. However, this study only included verbal individuals with CdLS, so this finding may not extend to non-verbal individuals.

This higher gesture rate in CdLS warrants explanation. Interestingly, Bell et al. (2018) described attenuated behaviour (see Breen & Hare, 2017) in people with CdLS and Richards et al. (2009) described physical movements preceding instances of speech in CdLS. In combination these findings might indicate that speech production is attenuated and movements preceding speech could be a strategy to help initiation. A higher rate of gestures might, therefore, compensate for attenuated speech under conditions of social demand. This interpretation requires further investigation with wider sampling of gesture use but is consistent with the observation that, whilst people with CdLS speak less during social demands, motivation for social engagement is evident and hence reduced spoken language is unlikely to be driven by social avoidance (Nelson et al., 2017; Crawford et al., 2020).

The findings have wider implications for understanding the relationship between other characteristics of CdLS. For example, reduced spoken language, previously argued to result from social anxiety, might instead be the driver of anxiety, with compromised spoken language production provoking anxiety in social settings; a stance adopted by Nelson et al. (2017). Given the previous argument that the higher rate of gestures in CdLS indicates motivation for social engagement, this provides additional evidence of this hypothesised direction between spoken language and anxiety. Further research is required to confirm the direction of this relationship.

This study has produced novel and insightful findings regarding the profile of communication impairments in CdLS. Whilst results need to be replicated in larger samples with larger sampling periods, the findings evidence the rate of gestures is higher than expected given level of ID and expressive language abilities in CdLS. This likely indicates a syndrome-related spoken language impairment. In combination with other evidence, it appears reduced spoken language during social interaction is not driven solely by reduced social motivation and anxiety.

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Table 1. *Participant group characteristics*

		CdLS (n = 15)	**DS (n = 15)**	**TD (n = 15)**
Age (in years)	Mean (range)	26.0 (13 - 42)	24.2 (15 - 33)	24.9 (16 - 38)
Gender	M:F	8:7	5:10	7:8
Hearing status[a]	% Normal	78.6[b]	73.3	N/A
BPVS-II Age Equivalence	Mean (range)	6.8 (4 – 12)	6.9 (4 – 13)	N/A
VABS Adaptive Behavior Composite Standard Score	Mean (range)	56.8 (20 – 95)	53.5 (25 – 93)	N/A

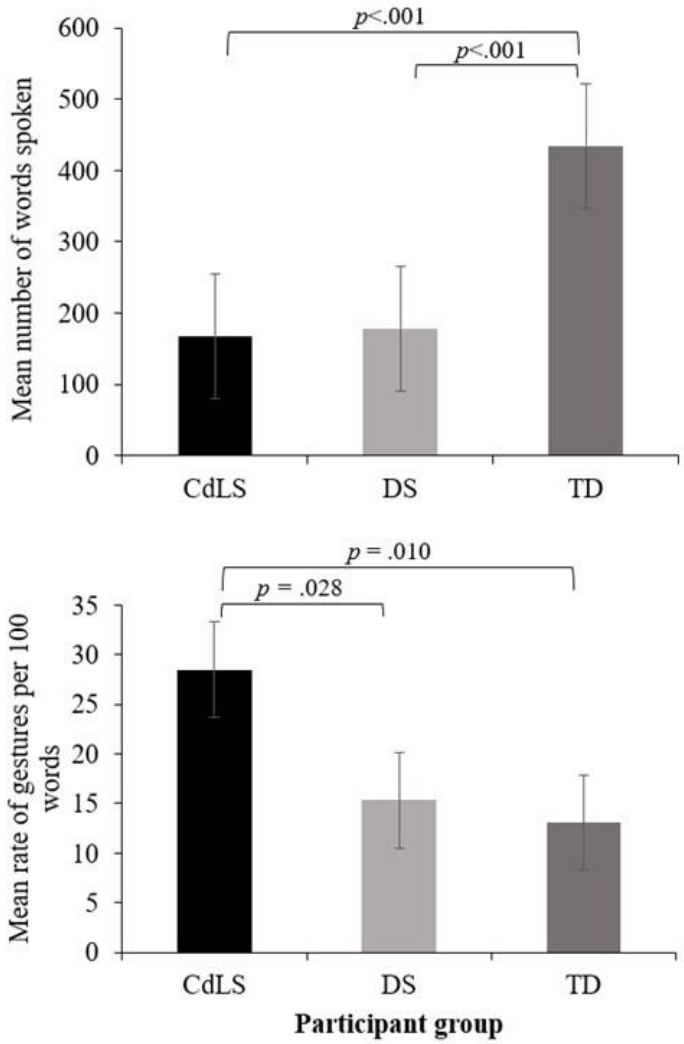
[a]measured by parental report on the Wessex Scale (Kushlick, Blunden, & Cox, 1973). [b]data not available for 1 participant

Table 2. The mean number (SD) and range of conversational turns, number of utterances, and MLU (Mean length of utterance, i.e. the number of words per utterance), in the analysed three-minute recordings.

				Statistical analyses		
	CdLS (n = 15)	DS (n = 15)	TD (n = 15)	F	*p*	Post-hoc analyses
Number of turns	32.4 (9.2) 11 - 48	33.5 (7.4) 21 - 46	29.4 (8.6) 16 – 46	.95	.394	ns
Number of utterances	43.4 (7.7) 33 - 59	47.1 (10.2) 32 - 70	43.8 (17.5) 21 – 81	.40	.672	ns
Mean length utterance	3.8 (2.6) 1.1 - 9.6	3.7 (1.3) 1.5 – 5.9	11.5 (5.4) 4.5 – 21.2	23.98	<.001	TD > CdLS, DS

Figure Legends

Figure 1. Lower panel: Mean number of words spoken by each participant group during the three minutes of coded conversation. Upper panel: Rate of gestures produced per one-hundred words spoken by each participant group.


p<.001
p<.001
Mean number of words spoken
600
500
400
300
200
100
0
CdLS
DS
TD
p = .010
p = .028
Mean rate of gestures per 100 words
35
30
25
20
15
10
5
0
CdLS
DS
TD
Participant group

Supplementary Material

Statistics for matching criteria

15 participants with Cornelia de Lange and Down syndromes were matched for chronological age (t(28) = -.619, p = .541, d = 0.23, variance ratio (VR) = 2.22), receptive language (t(28) = .072, p = .943, d = 0.03, VR = 0.60) as measured by the British Picture Vocabulary Scale 2 (Dunn et al., 1997), and adaptive behaviour (t(25) = -.074, p = .942, d = 0.03, VR = 1.30) measured by the VABS. The syndrome groups did not differ on gender (X^2(1) = 1.222, p = .269, Cramer's V = .20) or hearing status (X^2(2) = .166, p = .920, Cramer's V = .08). The TD group was comparable to the CdLS and DS groups for chronological age (F(2) = .209, p = .812, partial η^2 =.01) and gender (X^2(2) = 1.260, p = .533, Cramer's V = .17).