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Brief Report**Impaired communication ability in SOX11 syndrome****H. Smith,^{1,2} R. Al-Jawahiri,¹ L. Stokes,¹ M. Freeth,² S. Fricke,³ D. Matthews³ & A. McNeill¹ **¹ *Division of Neuroscience, The University of Sheffield, Sheffield, UK*² *Department of Psychology, The University of Sheffield, Sheffield, UK*³ *Human Communication Sciences, School of Allied Health Professions, Nursing and Midwifery, The University of Sheffield, Sheffield, UK***Abstract**

Background Speech and language skills are important for social interaction and learning. This study characterised the communication abilities of verbal individuals with SOX11 syndrome using a standardised parent/carer questionnaire, the Children's Communication Checklist (CCC-2).

Method Thirteen parent/carers of verbal individuals (aged 5–19 years) diagnosed with SOX11 syndrome completed the CCC-2. In order to contextualise findings, responses were compared to norms and to data from Noonan syndrome, a relatively well-known genetic diagnosis associated with communication impairment.

Results For all individuals, the CCC-2 composite score indicated significant communication difficulties. Language structure (speech, syntax, semantics and coherence), pragmatic language (inappropriate initiation, stereotyped language use of context and non-verbal communication) and autistic features (social relations and interests) scores were lower than typically developing norms. Subscale comparisons revealed relative difference in use of

context compared to other pragmatic domains (stereotyped language and inappropriate initiation). Individual scores showed substantial variation, particularly in regard to language structure profile. Differences were more pronounced than for Noonan syndrome, specifically in domains of speech, syntax, non-verbal communication and social relations.

Conclusions SOX11 syndrome is associated with communication impairment. It is important to assess communication abilities as part of the management of individuals with SOX11 syndrome and understand individual strengths and difficulties in order to provide targeted support.

Keywords communication, language, SOX11, speech, SRY-related high-mobility-group box 11

Introduction

The acquisition of language is an important skill for social interaction and learning. A language disorder can involve deficits in language structure (e.g. semantics, morphology and syntax) or pragmatic language (i.e. the appropriate use and interpretation of language in relation to situational context; Baird & Norbury 2016). Within an individual, any of the components of language can be differentially impaired. For example, pragmatic language

Correspondence: Dr Alisdair McNeill, Sheffield Institute for Translational Neuroscience (SITraN), The University of Sheffield, 385a Glossop Rd, Broomhall, Sheffield S10 2H, UK (e-mail: a.mcneill@sheffield.ac.uk).

impairment relative to structural language is recognised as a common feature of autism (Young *et al.* 2005; Schaeffer *et al.* 2023). Language disorders that are present in association with a known condition (such as a genetic condition diagnosis or autism) are differentiated from developmental language disorder (DLD), a term endorsed for use where language disorder is not associated with known biomedical aetiology (Bishop *et al.* 2017). This distinction is helpful as the presence of underlying biological factors is likely to have implications for the understanding of language disorder aetiology, prognosis and intervention. Speech sound disorders are also commonly diagnosed in genetic conditions and autism (Shriberg *et al.* 2011; Kent & Vorperian 2013). A speech sound disorder is distinct from a language disorder, referring to any difficulty or combination of difficulties with perception, motor production or phonological representation of speech sounds and speech segments (ASHA n.d.)

SOX11 syndrome is a rare genetic condition that is caused by deletions or de novo point mutations of the SOX11 gene. The prevalence of SOX11 syndrome is currently unknown. SOX11 is a transcription factor gene that plays an important role in brain development. The condition has been associated with medical issues including renal malformations and hypogonadism, in addition to intellectual disability and autism (Hempel *et al.* 2016; Al-Jawahiri *et al.* 2022). Previous case series have also reported that many individuals with SOX11 syndrome exhibit delayed speech and language, with some children exhibiting no speech at 3 years old (Hempel *et al.* 2016; Al-Jawahiri *et al.* 2022). This indicates that the SOX11 phenotype may include both verbal and minimally verbal individuals. However, to date, detailed information on communication ability has not been described. Characterising the strengths and difficulties associated with SOX11 syndrome would help to inform how best to support individuals with the condition.

Language differences have been associated with impairment in several areas of development including reading, general academic achievement and behaviour (Kastner *et al.* 2001; Yew & O'Kearney 2013). Furthermore, persisting language impairment has been found to be established early in life (Bornstein *et al.* 2018). However, it is unclear at present which individuals will go on to experience

persistent difficulties and which individuals will recover. Through investigating communication ability in a single-gene condition phenotype, such as SOX11 syndrome, it is possible to investigate communication impairment with greater precision and indication of risk of persisting difficulties specific to this condition.

This study aimed to characterise the speech and language abilities of *verbal* individuals with SOX11 syndrome using a standardised parent/carer questionnaire, the Children's Communication Checklist (CCC-2; Bishop 2003). The study compared SOX11 syndrome CCC-2 data with TD children (normative CCC-2 data) and Noonan syndrome (NS) CCC-2 data (Selås & Helland 2016).

NS is a relatively common genetic condition with an estimated incidence of 1 in 1000–2500 births. It is primarily caused by pathogenic variants in *PTPN11* (Roberts *et al.* 2013). NS and SOX11 syndrome are comparable in general clinical features: ambulation is achieved, there is a degree of functional independence, developmental delay is mild–moderate rather than severe, and there is no malformation of the speech producing organs or epileptic encephalopathy (Selås & Helland 2016; Zenker *et al.* 2022). Selås & Helland (2016) reported significant differences in language ability in NS compared with typically developing children (as measured by the CCC-2). Specifically, differences were found in domains of language coherence, inappropriate initiation, stereotyped language, use of context, non-verbal communication, social relations and interests. Hence, NS provides a useful example of a genetic condition where communication is impaired in the absence of either malformations of the speech producing organs, severe developmental delay or epileptic encephalopathy. It is thus useful to compare SOX11 syndrome and NS, as differences in communication profile reflect neurodevelopment and not confounding variables such as malformations. Given that many clinicians will be familiar with NS, this comparison also provides a reference point for them to better appreciate the communication profile of SOX11 syndrome and to better counsel affected families.

Methods

The study received ethical approval from the Medical School Research Ethics Committee at the University of Sheffield (035843).

Participants and procedure

This study was part of a larger cross-sectional study investigating cognition and behaviour in SOX11 syndrome that recruited 20 parent/carers (hereafter 'parents'). Seven of these parents were not eligible to complete the CCC-2 as their child was either under 4 years old ($n = 2$) or unable to produce phrased speech ($n = 5$). Therefore, this article reports 13 parents of *verbal* individuals diagnosed with SOX11 syndrome (mean age: 10;7; $SD = 5;0$; age range: 5;1–19;11, male: 6, female: 7). Two individuals (aged 19;6 and 19;11) were out of age bounds for the CCC-2 (5;0–16;11). For these individuals, scaled scores were computed using the highest reference age category in the CCC-2 normative sample data (16–16;11). Nine families were from the United Kingdom, and four families were from the USA. The ethnicity of the vast majority of the sample was White (all but one family).

Parents were recruited via a closed SOX11 online family support group with worldwide membership. Parents completed the CCC-2 via online video meeting with a researcher. Remuneration was not provided.

Measures

The study used the CCC-2 (Bishop 2003), a 70-item parent-reported questionnaire measure of communication ability for children who are able to use phrased speech. Parent/carers indicate the frequency with which their child exhibits certain communicative behaviours on a scale of 0–3 (0 = less than once a week or never, 3 = several times a day or always). Raw scores are converted into scaled scores, adjusted for participant age. Four subscales (A. Speech, B. Syntax, C. Semantics, and D. Coherence) measure speech and structural language, four subscales (E. Inappropriate initiation, F. Stereotyped language, G. Use of context, H. Non-verbal communication) measure pragmatic language, and two subscales (I. Social relations and J. Interests) measure autistic features. In addition, the General Communication Composite (GCC) provides an overall measure of communication ability with scores <55 indicating significant communicative difficulties. The Social Interaction Deviance Composite (SIDC) identifies children whose pragmatic language abilities are disproportionately impaired relative to other aspects of language. Scores <0 are considered

consistent with an autistic communication profile, and scores ≥ 9 are considered consistent with a DLD communication profile. Two further composite scores for language structure and pragmatic language can be computed. Scores >24 represent 'typical functioning', 17–24 represent 'borderline functioning', and <17 represent 'impaired functioning'.

Results

Data for CCC-2 composite and subscale scores are shown in Table 1. GCC scores ranged from 9 to 43 (<1 st percentile–4th percentile). This suggests that all of the individuals with SOX11 assessed experienced significant communication difficulty.

Language structure in SOX11 syndrome

SOX11 mean scaled scores for speech, syntax, semantic and coherence were 2SD or more below the norm for typically developing (TD) children (Figure 1). A Friedman test indicated a statistically significant difference in language structure subscale scores, $\chi^2(3) = 9.09$, $P = 0.028$. *Post hoc* analysis with Wilcoxon signed-rank tests and Bonferroni correction found no significant differences between any of the subscales (significance level $P < 0.0125$). There was a notable variation in language structure composite scores (Table 1), with 1/13 individuals with SOX11 syndrome scoring as 'typical functioning', 2/13 as 'borderline functioning', and 10/13 as 'impaired functioning' (Figure S1).

Pragmatic language in SOX11 syndrome

SOX11 mean scaled scores for inappropriate initiation, use of context and nonverbal communication were 2SD or more below the norm for TD children and mean scaled scores for stereotyped speech were 1SD or more below the norm for TD children (Figure 1). A Friedman test indicated a statistically significant difference in pragmatic language subscale scores, $\chi^2(3) = 16.05$, $P = 0.001$. *Post hoc* analysis with Wilcoxon signed-rank tests and Bonferroni correction found a significant difference between stereotyped language and use of context ($Z = -2.96$, $P = 0.003$) and inappropriate initiation and use of context ($Z = -2.82$, $P = 0.005$; significance level $P < 0.0125$). There were no significant

Table 1 CCC-2 scores for SOX11 and Noonan syndrome (NS)

	SOX11			NS		
	N	Mean (SD)	Range	N	Mean (SD)	t test
General Communication Composite (GCC)	13	24.77 (11.98)	9–43	17	44.53 (19.93)	* $t(28) = -3.16, P = 0.004$, Cohen's $d = 1.20$
Language structure composite	13	11.00 (8.72)	0–28	17	Data not available	
A Speech	13	2.92 (3.80)	0–12	17	7.18 (3.59)	* $t(28) = -3.14, P = 0.004$, Cohen's $d = 1.15$
A Syntax	13	1.54 (2.07)	0–6	17	6.71 (3.57)	* $t(28) = -4.65, P < 0.001$, Cohen's $d = 1.77$
A Semantics	13	3.00 (2.58)	0–7	17	4.75 (3.07)	$t(28) = -1.65, P = 0.109$, Cohen's $d = 0.62$
A Coherence	13	3.54 (2.26)	0–7	17	5.29 (3.12)	$t(28) = -1.71, P = 0.099$, Cohen's $d = 0.64$
Pragmatic language composite	13	13.77 (5.29)	6–24	17	26.12 (13.05)	* $t(28) = -3.21, P = 0.003$, Cohen's $d = 1.24$
A Inappropriate initiation	13	3.92 (1.55)	2–7	17	4.88 (3.00)	$t(28) = -1.05, P = 0.303$, Cohen's $d = 0.40$
A Stereotyped language	13	4.31 (1.38)	2–7	17	5.82 (3.13)	$t(28) = -1.62, P = 0.117$, Cohen's $d = 0.62$
A Use of context	13	2.00 (1.58)	0–5	17	3.53 (2.63)	$t(28) = -1.85, P = 0.074$, Cohen's $d = 0.71$
A Non-verbal communication	13	3.54 (3.26)	0–13	17	6.59 (2.62)	* $t(28) = -2.84, P = 0.008$, Cohen's $d = 1.03$
Autistic features						
A Social relations	13	3.46 (2.73)	0–9	17	6.76 (3.78)	* $t(28) = -2.66, P = 0.013$, Cohen's $d = 1.00$
A Interests	13	5.54 (2.79)	1–10	17	5.06 (2.59)	$t(28) = 0.49, P = 0.630$, Cohen's $d = 0.17$

NS data from Selås & Helland (2016).

Subscale scores (A, B, C, D, E, and F) are mean scaled scores. Composite scores (GCC, language structure, and pragmatic language) are means of summed subscale scores.

*Statistical significance using $P \leq 0.05$.

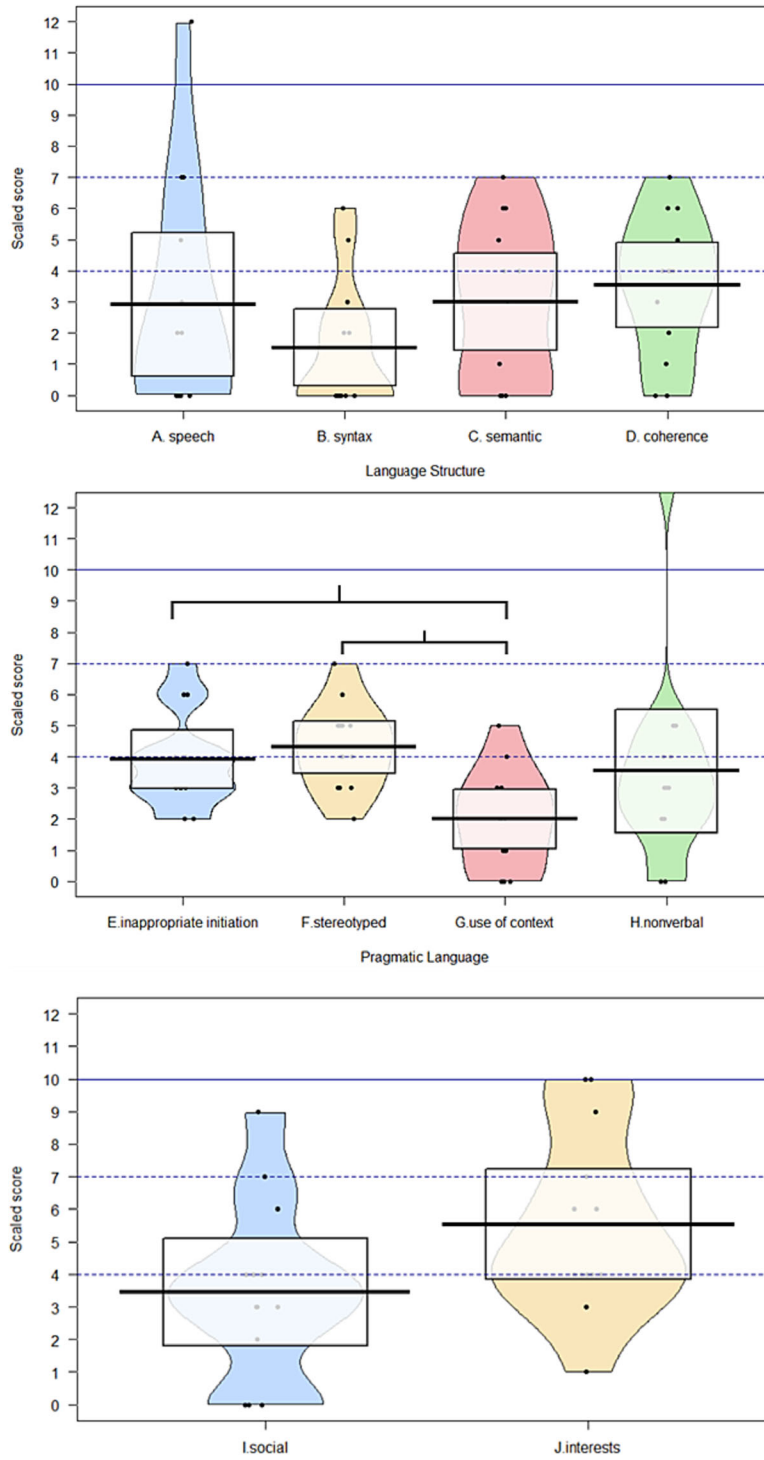


Figure 1. SOX11 CCC-2 scaled scores. Group mean is indicated by the bold black solid line. CI is indicated by the transparent box. Mean for each subscale for typically developing standardised norms = 10 (indicated by the solid line) and SD = 3 (1SD and 2SD from the mean indicated by the dashed lines). *Significant difference between mean scaled scores ($P < 0.0125$).

differences found between the other subscale pairs. This suggests that individuals with SOX11 syndrome had a varied pragmatic language profile with relative impairment in use of context. For the pragmatic language composite, 4/13 individuals with SOX11 syndrome scored as 'borderline functioning' and 9/13 scored as 'impaired functioning' (Figure S2).

Autistic features in SOX11 syndrome

The SOX11 mean scaled score for social relations was greater than 2SD, and for interests greater than 1SD, below the norm for TD children. No significant difference was found between social relations and interests ($Z = -1.79$, $P = 0.073$; Figure 1). Individual data are presented in Figure S3. Two out of the 13 individuals with SOX11 syndrome had a SIDC score consistent with an autistic communication profile, and 4/13 had an SIDC score consistent with a DLD communication profile. The remaining 7/13 scored between these categories.

Comparison with Noonan syndrome

The communication ability of individuals with SOX11 syndrome was compared with a sample of $N = 17$ children with NS (mean age: 10;2, SD: 3;1; age range: 6–15 years; male: 12, female: 5) obtained from Selås & Helland (2016) (Table 1). Compared with the NS group, the SOX11 group had significantly more impaired GCC and pragmatic language composite scores. With regard to subscale scores, the SOX11 group had greater impairment in speech and syntax, non-verbal communication and social relations.

Discussion

This study investigated the communication ability of verbal individuals with SOX11 syndrome using the parent-reported CCC-2. A key finding is that individuals had lower reported abilities in language structure and pragmatic language and exhibited differences in autistic features, relative to TD norms. For some individuals, reports suggested an autistic communication profile, but for others, the profile was more similar to that of DLD, with most children fitting neither criteria. In addition, individuals with SOX11 syndrome had greater impairment in communication ability compared to NS, specifically

in domains of speech, syntax, non-verbal communication and social relations.

While the present study offers new evidence, the generalisability of findings is limited by the relatively small cohort included. Furthermore, the assessment relied on a single time point, parent-reported measure, and thus subjective perceptions of the affected individual's communicative ability. It is also critical to highlight that this study only applies to *verbal* individuals with SOX11 syndrome. It will be important for future studies to investigate speech and language ability in larger samples of individuals with SOX11 syndrome, including both verbal and minimally-verbal individuals, and using direct, standardised assessments of speech and language ability that encompass a broader range of speech and language domains.

In their case series of 53 individuals with SOX11 SNVs, Al-Jawahiri *et al.* (2022) reported that just 40% had begun to speak by 30 months and that several individuals were non-verbal, providing initial evidence to suggest that speech and/or language ability is impaired in this condition. The low speech and language scores reported in this prospective study provide additional evidence of communication impairment in SOX11 syndrome and may indicate the domains of communication that are most affected. In addition, the reports of impaired communication in older individuals indicate that differences persist in SOX11 syndrome individuals beyond early childhood.

In terms of language structure skills, syntax, semantics and coherence were clearly different relative to TD norms. There was a notable variation here, however. This variation indicates that assessing individual strengths and difficulties in language is likely to be important for individuals with SOX11 syndrome. Future research should explore sources of this variation.

While all areas of pragmatic language ability were impaired, the findings suggest that individuals with SOX11 syndrome may have relative weakness in use of context compared with other aspects of pragmatic language (inappropriate initiation and stereotyped language). This difficulty could relate to physical context (what is around them and their communication partner), the discourse context (what has already been said) or the cultural context (what is in common knowledge between people;

Matthews 2021). Similarly, the use of context was the most impaired pragmatic language domain in NS (>2SD below the norm of TD children), and similar weakness in use of context has been observed in other genetic conditions: SETBP1 (Morgan *et al.* 2021), Down syndrome (Smith *et al.* 2017) and Sotos syndrome (Lane *et al.* 2019). Pragmatic development can be supported by access to interaction with others (Matthews 2021). Yet, in many genetic conditions, interaction with others is limited by lifestyle factors associated with the condition, such as missed time in education or missed time with peers. It may therefore be important to consider the potential contributing role of interaction opportunity when supporting pragmatic language development in individuals with SOX11 syndrome.

The association between communication impairment and impaired adaptive function has been well documented in autism and in genetic conditions such as Down syndrome and Fragile X (Shaffer *et al.* 2020; del Hoyo Soriano *et al.* 2022; Miranda *et al.* 2023). Despite evidence of communication impairment in NS, this does not appear to be highly pervasive in terms of impaired adaptive function; the majority of children with NS grow to live independently as adults (Noonan Syndrome Guideline Development Group 2010). Contrastingly, the ubiquitous and more severe communicative impairment reported in SOX11 syndrome in the present study indicate that individuals with SOX11 syndrome will likely require substantial support in their day-to-day lives.

Findings from this study indicate that verbal individuals with SOX11 syndrome do not demonstrate a consistent autism or DLD pattern with most individuals falling between these criteria. While this could suggest limited association between SOX11 and autism, it should be noted that the SIDC score is not recommended for use as a diagnostic tool but rather as a tool to yield contrasting subgroups in a research sample (Bishop 2003). The use of more comprehensive and multidisciplinary assessment of autistic traits, language and cognition is necessary to provide stronger evidence in regard to the association between SOX11 syndrome and autism.

The findings indicate that communication impairment is a prominent feature of SOX11 syndrome with verbal individuals experiencing differences in language structure, pragmatic language

and autistic features. Findings highlight the importance of assessing communication abilities as part of the management of individuals with SOX11 syndrome and understanding individual profiles of strengths and difficulties in order to provide individualised support.

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Conflict of interest

No conflicts of interest have been declared.

Ethics approval statement

Ethical approval was obtained from the Medical School Research Ethics Committee at the University of Sheffield. Written informed consent was obtained from participants.

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Data availability statement

Data are available from the lead author upon reasonable request.

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Supporting Information

Additional Supporting Information may be found online in the supporting information tab for this article.

Figure S1. Language structure in SOX 11 syndrome
Figure S2. Pragmatic language in SOX 11 syndrome
Figure S3. Autistic features in SOX 11 syndrome