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The Quality of Everyday Eye Contact in Williams Syndrome: Insights across Syndromes

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The Quality of Everyday Eye Contact in Williams Syndrome: Insights from Cross Syndrome Comparisons

Abstract

Past research shows that individuals with Williams syndrome (WS) have heightened and prolonged eye contact. Using parent report measures, we examined not only the presence of eye contact but also its qualitative features. Study 1 included individuals with WS (n=22, age 6.0–36.3). Study 2 included children with different neurodevelopmental (ND) conditions (WS, Autism Spectrum Disorder, Fragile X syndrome, Attention deficit hyperactivity disorder) and children with neurotypical development (NT) (n=262, age 4.0–17.11). Unusual eye contact features, including staring, were found in approximately half of the WS samples. However, other features such as brief glances were frequently found in WS and in all ND conditions, but not NT. Future research in ND conditions should focus on qualitative as well as quantitative features of eye contact.

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Introduction

Eye contact – the act of looking another person in the eyes – plays a powerful role in our everyday human social interactions. It signals mutual understanding and affiliation between people, and promotes social-emotional relationships and communication (Emery, 2000; Falck-Ytter et al., 2015; Kleinke, 1986). Experiences of eye contact also elicit a range of cognitive and affective reactions in the perceiver (for reviews see Conty et al., 2016; and Hietanen, 2018). In Western European societies, direct eye contact induces a range of positive evaluations (Kreysa et al., 2016; Willis et al., 2011). In contrast, a lack of eye contact may infer disinterest, whereas overly persistent eye contact may be deemed threatening and overly arousing (Akechi et al., 2013; Helminen et al., 2011). Therefore, when an individual's

eye contact is reduced or overly prolonged, or unusual in some way, this may adversely affect social impression-formation with consequences for the development of social relationships (Morrison et al., 2020; Sasson et al., 2017).

Several theoretical perspectives have been put forward to explain how eye contact modulates cognition and behaviour for those with neurodevelopmental (ND) conditions (for a review, see Senju & Johnson, 2009). The majority of these theoretical accounts apply particularly to the literature on Autism Spectrum Disorder (hereafter 'autism'[1]) and to the assumption by several different theories (e.g. social motivation theory Chevallier et al., 2012; hyperarousal model, Hadjikhani et al., 2017), that autistic individuals have diminished eye contact. One problem is that the evidence for this view rests mainly on studies that report reduced frequency or presence of eye contact. However, there has been remarkable neglect in considering the nature of the quality of eye contact, which could possibly lead to a different understanding of eye contact in individuals with ND conditions. One reason for the past focus on quantity rather than quality is that much of the research knowledge on eye contact stems from a broader laboratory-based research tradition on eye gaze more generally, which tends to equate looking at the eyes of computerised facial stimuli with 'eye contact'. While this paradigm affords a high level of experimental control, the passive viewing of socially-relevant stimuli is very different from how eye contact is experienced in everyday dyadic social interactions (see Kingstone, 2009). Research has shown that the realism of the stimuli used in social attention research (e.g. static versus dynamic images; isolated faces versus multiple faces in a social scene), impacts on eye contact (e.g. Hanley et al., 2013; Speer et al., 2007). Consequently, researchers have emphasised the importance of studying everyday situations to understand social attention in real-life interactive situations (e.g. Hanley et al., 2015; Kingstone, 2009; Risko et al., 2012). In the

[1] There is a growing literature emphasising the importance of adopting non-ablest language in academic articles and a need to move away from the term 'disorder' when describing Autism (Bottema-Beutel et al., 2020). In this article we use person-first language ("autistic person") in line with the preference of the majority of the autistic community (Kenny et al., 2016)

current investigation, we examine both the presence and quality of everyday eye contact of individuals with ND conditions, using the caregiver's perspective of eye contact.

Eye contact behaviour in Williams syndrome

Williams syndrome (WS) is a genetic ND condition commonly associated with a heightened desire for social contact (termed 'hyper sociability'; for a review of the WS social phenotype, see Thurman & Fisher, 2015). Indeed, WS is a really important ND condition to study various aspects of social behaviour because its genetic basis is well-defined (hemizygous deletion of ~25-28 genes on chromosome 7q11.23; Ewart et al., 1993), therefore research with this group has the potential to inform debate about genetic-brain-behaviour links and further our understanding of the "typical" social brain. Consequently, the WS social profile has garnered a significant amount of research attention at the level of both brain and behaviour. For example, evidence that WS is associated with structural and functional atypicalities in key areas of the 'social brain network' known to activate in response to eye contact, such as the amygdala (Haas et al., 2009; Martens et al., 2009) and fusiform face area (FFA; Golarai et al., 2010), has informed understanding of how different features of the WS social phenotype may be subserved by neural substrates (for a review see Haas & Reiss, 2012). At the behavioural level there has been a great deal of interest in capturing various aspects of social behaviour in WS, including eye gaze and eye contact behaviour. The predominant evidence of gaze behaviour in WS comes from face scanning and eye-tracking studies that have examined eye gaze behaviour towards images or movies on screen. These studies show that the face, particularly the eye region, attracts and holds the attention of individuals with WS for longer than is typical for young children, adolescents and adults (Porter et al., 2010; Riby & Hancock, 2008, 2009a, 2009b). This tendency for heightened, prolonged looking to faces and eyes has been linked to a lack of habituation to faces (Järvinen et al., 2012), to physiological reactivity and to attentional mechanisms related to arousal, suggesting the possibility of hypo-arousal in this group (Doherty-Sneddon et al., 2009; Riby et al., 2012; Skwerer et al., 2009, 2011).

Beyond laboratory studies using eye tracking and measuring gaze to computerized images, a few other observational studies have also reported that young children with WS (< 4 years old) show intense and prolonged looking in real-world settings; during interactions in clinics (Mervis et al., 2003;) and with experimenters (Jones et al., 2000). Although studies using a clinical measure, the Autism Diagnostic Observation Schedule (ADOS; Lord et al., 2000), have reported up to 53% of children with WS had 'definite abnormality' with eye contact, we know little about the nature of the 'abnormality' as the ADOS assessment of eye contact does not capture quality features (Klein-Tasman et al., 2007, 2009). Given this limited evidence of prolonged, intense eye contact in naturalistic settings, it is still not established whether this quality of eye contact is common in individuals with WS, if it is a feature distinctive to WS or frequently found in other ND conditions. Research that examines eye contact behaviour in WS alongside other ND conditions will help to identify features of eye contact that may be particularly distinctive to WS (syndrome-specific) or shared across diagnostic groups (syndrome-general). See Asada and Itakura (2012) for further discussion.

Eye contact behaviour across neurodevelopmental conditions

While WS has been characterized by social interest associated with a heightened and prolonged presence of eye contact, other ND conditions, particularly Autism, in contrast have traditionally been associated with reduced presence of eye contact (Asada & Itakura, 2012; Senju & Johnson, 2009). Reduced eye contact, in turn, has been connected to a lack of social interest (Chevallier et al., 2012); an assumption that has been challenged by those with subjective, lived experience of autism (Jaswal & Akhtar, 2019) who argue that reduced quantity of eye contact does not necessarily equate with lack of interest. We propose that the clarification of this issue has been hampered by a single dimensional approach to the understanding of eye contact; that conflates presence and quality of eye contact. Characterizing eye contact by a single dimension leads to a view that reduced eye-contact is poor eye contact and increased eye contact is good eye contact; an assumption that tends to polarise the social phenotypes of ND groups into opposite profiles (see Asada & Itakura, 2012 for review of the Autism/WS distinction). By considering multiple qualitative features of

eye contact in everyday life contexts, across ND conditions, the current study attempts to move away from examining eye contact through a quantitative, single dimensional lens.

Like studies of WS, much previous research on eye contact in autism has also tended to focus on its presence or degree. Eye-tracking studies show that some autistic individuals spend less time than is typical attending to face areas (Sasson et al., 2007; Shic et al., 2011) and eye areas on a screen. For reviews of the Autism eye tracking literature see Guillon et al. (2014) and Papagiannopoulou et al. (2014). Both eye tracking studies (e.g. Hanley et al., 2014, 2015) and face to face observational studies (e.g. Leekam & Ramsden, 2006) also find differences in attentional orienting in autistic individuals compared to neurotypical and intellectually disabled peers and that reduced eye contact is very dependent on context (Jones et al., 2017; Kasari et al., 1993). Furthermore, reduced presence of eye contact has been associated with failure to automatically attend to the salience of social cues, rather than to active avoidance of others in several eye tracking studies (Hanley et al., 2013; Klin et al., 2002) and has been associated with over-arousal (Hadjikhani et al., 2017). First-hand insights from autistic adults also describe reduced eye contact as a strategy for arousal reduction (McGlensey, 2016; Trevisan et al., 2017) and report the use of qualitative strategies used such as non-eye fixation, blurring focus and strategic fixation (Trevisan et al., 2017). The perceived experience of unfocused eye gaze in these first-hand accounts however has not been measured from another person's perspective and the research reported here targets this by exploring parents' perspective of eye contact taken from their everyday experience.

While Autism and WS are two frequently studied ND conditions in the eye gaze and eye contact literature, these are not the only ND conditions that are associated with social difficulties related to eye contact. Like WS, Fragile X Syndrome (FXS) is a genetic condition associated with mild to moderate intellectual disability and impacts upon social functioning. The FXS social phenotype can be summarized as a mix of both social approach (Cornish et al., 2008) and social withdrawal behaviours (Roberts et al., 2007, 2019), alongside heightened social anxiety (Crawford et al., 2017). Studies to date consistently show that FXS

is associated with gaze avoidance (Hall et al., 2006, 2009, 2010; Hessl et al., 2006), which increases when the interlocutor is unfamiliar (Hall & Venema, 2017), but which may improve over the course of an interaction ('warm up effect'; Hall et al., 2009; Roberts et al., 2007). People with FXS show a tendency for shorter gaze episodes towards another person and for brief glances when the person is looking elsewhere rather than making direct eye contact (Cohen et al., 1991; Hall et al., 2006, 2015; Klusek et al., 2020).

Although social difficulties are not part of the diagnostic criteria for Attention Deficit Hyperactivity Disorder (ADHD), there is a growing literature reporting socio-cognitive difficulties, problematic peer relationships (for reviews see Gardner & Gerdes, 2015; Soucisse et al., 2015) and high rates of social vulnerability (Ridley et al., 2020). Studies reporting on aspects of gaze orienting and attention indicate impairments in attending to socially relevant information (Airdrie et al., 2018; Marotta et al., 2014, 2017; Muszkat et al., 2015), however everyday eye contact behaviors in this population have scarcely been documented. One relevant study using the ADOS found that unusual eye contact was reported statistically more frequently in a sample of autistic children compared to children with ADHD (Grzadzinski et al., 2016). Nevertheless, 31% of the ADHD sample were reported to have abnormal eye contact, yet the nature of the unusual eye contact was not described.

The current study

In this study, we explored the quality of everyday eye contact in individuals with WS in comparison with each of these ND groups using parent report. First, we studied the single dimension of 'presence' (or degree of presence). Second, we included a specific measure of different qualitative features that have been associated with different ND conditions. A two-stage approach was adopted. First, given the gap in the literature on the quality of eye contact in WS, particularly from a parent perspective, Study 1 used a set of standard interview questions to explore the qualitative features that parents might observe in their son/daughter's everyday eye contact. Although we expected a high presence of eye contact in WS, we also expected, given the findings of Mervis et al. (2003) and (Jones et al., 2000),

that parents might observe a quality of intense, prolonged eye-contact (equated with staring in this study). However, we did not know whether other qualitative features would be frequently seen or the extent to which staring would be found across all WS individuals and across all ages.

In Study 2 we used a parent questionnaire method to examine further the eye contact quality features used in Study 1 as well as other qualitative features, making cross-syndrome comparisons across children with WS, Autism, FXS and ADHD. In addition, we included a neurotypical comparison group to examine whether particular qualitative aspects of eye contact were specific to the presence of a ND condition. The research will contribute new evidence to an ongoing debate about the similarities and differences in eye contact in ND conditions, particularly between WS and Autism. The study will also add new findings to the literature on eye contact behaviour in FXS, and in ADHD; a topic that has received limited attention.

Study 1: Examining the nature of eye contact in Williams Syndrome

The first study explored the presence and quality of eye contact used by individuals with WS in their everyday life. A semi-structured set of interview items was used that enabled parents to describe both the presence of eye contact and qualitative features such as brief glances, staring behavior and unfocused gaze. The individual's developmental level of language and visuospatial ability was also recorded during the interview.

Participants

Twenty-two individuals with WS and their families were recruited throughout the North of England and Scotland following institutional ethical approval and study approval from the Professional Advisory Panel of the Williams Syndrome Foundation. Informed consent was given by all participants. The researcher conducting the interviews with parents X (blinded) was trained in its use by X (blinded). In all cases, it was the primary caregiver who completed the interview with the researcher, either at home or in the University.

Individuals were sampled across a wide age range. At the time of the parent interview, individuals with WS ranged between 6 years 0 months and 36 years 3 months of

age (male, 10, female 12), M_{age} 196 months (SD 98 months). All individuals were attending school, college or work placements; including five in mainstream school with support, 10 in special educational provision and five in supported work or college (two had information missing). All individuals had previously been diagnosed phenotypically by clinicians and their diagnosis had been confirmed with positive fluorescent in situ hybridization (FISH) testing.

Information on language delay, and current language and visuospatial ability was collected from parents during the interview. As Table 1 shows, the group was developmentally delayed. In terms of language delay, 78% of individuals (14/18, 4 missing) were late to use 2-3 phrases and 84% (16/19, 3 missing) were late to understand word meanings. In terms of current language ability, 21 participants (one missing) had sentence-level expressive language and all but one participant had sentence-level receptive language (simple or complex sentences). However, only two-thirds (14 individuals) used expressive language at the highest level (complex age-appropriate grammatical constructions) and only one third (seven) understood language at this level. Visuospatial data (two missing) showed that only three individuals (15%) had age-appropriate level of current skill.

Materials and procedure

A research form of the Diagnostic Interview for Social and Communication Disorders (DISCO; Leekam, 2020; Wing et al., 2002) was used. The DISCO is a semi-structured clinical interview used with parents and carers. It is most commonly used for parents of individuals on the autism spectrum of any age but is also suitable for use with individuals with other ND conditions and includes items applicable for ADHD, WS and FXS. The current interview followed the format of previous research that has used and published subsets of DISCO items (e.g. Prior et al., 1998). The eye-contact and language items used in Study 1 are included in the published DSM-5 algorithm (Kent et al., 2013) and DISCO ICD-10 Childhood Autism algorithm (Leekam et al., 2002), and the visuospatial skill item is a non-algorithm item in the DISCO (Wing et al., 2002). Each of the four eye contact items and each of the language and visuospatial items has a high level of inter-rater reliability ranging from k=.89 to k=1.00 (Wing et al., 2002).

Information on language delay and current language and visuospatial ability was collected using age-appropriate scales within the DISCO (see Table 1). Items from the current language scales have been published (Honey et al., 2007). Age- equivalent visuospatial skill was indicated by the ability to construct complex puzzles according to age group. Language delay (use of phrases, comprehension of word meanings without visible cue) was indicated by delay after 48 months old. Age-appropriate current sentence skills were recorded when complex grammatical constructions and past, present and future tense were present.

Information on the presence and quality of eye contact was collected using four eye contact items and scored using the DISCO syntax rules that have previously been applied in both interview (Kent et al., 2013) and questionnaire (Jones et al., 2020), research formats. The first item related to the presence of eye contact. The interviewer asked the caregiver whether it was easy to get eye contact with the individual. The item was scored as “eye contact present” if the answer was “yes”, even if the eye contact given was described as unusual in some way, and “no” if the parent reported little or no eye contact. The next three questions related to quality of eye contact seen as usual behaviour on an everyday basis. These were whether the individual (a) makes eye contact only in brief glances e.g. out of the corner of eyes, but not for the purpose of gaining another’s attention, (b) whether the individual has a blank, unfocussed gaze and (c) whether the individual stares too long and hard, perhaps holding another person’s face to make eye contact and/or looking closely into another’s eyes. Each item was sequentially assessed by the interviewer who established whether this was a typical behaviour for the individual (used routinely with adults and age peers) and whether it was marked (or frequent), occasional, or rarely/never seen. Following DISCO syntax rules, each item was scored as having a markedly unusual quality if judged to be “marked” (brief glances), “marked and frequent” (blank, unfocused gaze), and “marked staring or otherwise inappropriate” (staring) in that individual, but not if the feature was sometimes, rarely or never seen.

Results and Discussion

Case-by-case profiles of eye contact patterns are shown in Table 1 together with age and language/visuospatial level. The cells that include the plus symbol (+) indicate endorsement of a score for each individual (e.g. presence of eye contact or marked quality of eye contact) while the blank cells indicates non-endorsement. Results showed that 20 (91%) individuals (9 male, 11 female) gave eye-contact easily (even if inappropriately), while two (9%), gave little or no eye contact. Subsequent analyses focused on these 20 individuals, 13 of whom (65%; seven male, six female), had a 'marked' unusual quality of eye contact, as indicated by at least one out of three unusual features - brief glances, unfocused gaze, or stare. Brief glances at marked level were endorsed by eight (40%), unfocused gaze by eight (40%), and staring was endorsed by 10 (50%; see Table 1). Six individuals had marked scores for all 3 features.

INSERT TABLE 1 HERE

Further exploration was made of the characteristics of the 13 individuals with marked unusual quality of eye contact. More than half, nine of the 13 (69%), had early developmental delay in understanding of word meanings (two had no delay, two had missing data), and of these nine individuals, all but one (data missing) were also delayed in using two-to-three word phrases. The gender distribution was also approximately equal for endorsement of each of the three eye contact quality features.

To explore how each of the unusual eye contact quality features was affected by other variables (current age, current language level, and visuospatial level), Mann-Whitney tests were carried out with the 20 participants who were reported by parents as giving eye contact easily. For each analysis, the group of individuals with 'marked' responses was compared with the group without marked features (scoring 'sometimes' or 'rarely/never'). Analyses were repeated to examine brief glances, unfocused gaze, and staring features separately and Bonferonni adjustment was applied to accommodate multiple comparisons (.05/3, p=.02). An age difference was found (see Table 1), as the group with marked staring features was older, having a mean age of 20 years 11 months (M_{age} = 251.20 months, *SD* = 108.37, *n* =10) while those without marked staring features had a mean age of only 12 years 6 months, (M_{age} =

150.60 months, *SD* = 56.44, *n* =10), *U* = 99.0, *p* < .010, however, there were no age differences for the other unusual quality features (unfocused gaze, *p* = .92; brief glances *p* = 1.00). No differences were found in visuospatial ability, current expressive and receptive language for those with marked unusual eye contact quality.

In summary, Study 1 used a set of parent interview questions for the first time, to explore the qualitative features of everyday eye contact in individuals with WS. The results showed positive presence of eye contact by 91%, together with an atypical quality of staring in 50%. This pattern supports previous evidence from laboratory and clinic studies. However, in addition, new evidence was found. Results showed that staring was more frequent among older ages. However, staring was not an exclusive or predominant quality feature and parents endorsed features of unusual quality of eye contact beyond staring, including brief glances and unfocused gaze. These were reported by parents in 40% of individuals with least one of these features often co-occurring alongside staring.

Study 2: Comparing eye contact in WS, other neurodevelopmental conditions and neurotypical development

To gather a larger sample of reports, Study 2 asked the same questions as in Study 1 but used a questionnaire measure with parents of children with WS. In addition, we adopted a cross-syndrome approach to examine potential syndrome-specific aspects of eye contact behaviour in WS, Autism, FXS and ADHD as well as neurotypical development (NT).

Given the research findings reviewed above and the results of Study 1, we predicted 1) a high presence of eye contact in WS compared with other ND groups 2) that unusual qualitative features would be found in WS and also in the other ND groups, with staring reported for children with WS (Klein-Tasman et al., 2007; Mervis et al., 2003), brief glances and avoidance reported for FXS children (Klusek et al., 2020) and a blurred or unfocused gaze (Trevisan et al., 2017) and/or avoidant gaze (Senju & Johnson, 2009) reported for autistic children. Given the results for the WS group in Study 1, we expected not only staring

304 but also other qualitative features to be reported. However it was not known whether other
305 ND groups might have particular distinctive and predominating qualitative features.

306 **Participants**

307 Parents/caregivers of children were recruited for this study as part of a larger
308 investigation of social interaction behaviours in children with and without ND conditions.
309 Survey responses were received for 276 caregivers/parents in total. Responses were
310 included for data analysis based on the child's primary diagnosis if the parent reported that
311 their child: (1) had a primary diagnosis of either WS, autism, FXS or ADHD, or had NT
312 development and did not have an intellectual disability or statement of Special Educational
313 Need (SEN), and (2) was aged 4–17 years. Of the 276 respondents, 262 met the
314 aforementioned inclusion criteria and fell in the following groups: WS (n=29), Autism (n=29),
315 FXS (n=18), ADHD (n=36) and TD (n=150). None of the participants in Study 1 were
316 included in the WS sample in Study 2.

317 Table 2 shows the child characteristics per group. Fifty-nine percent of the full
318 sample were males. The ND groups (apart from the WS group) included significantly more
319 males than the NT group. Of the ND groups, FXS included significantly more males than the
320 WS group. However there was no significant difference in the distribution of genders
321 between the other ND groups. The ND groups differed in parent-reported ID status as seen
322 in Table 2, $\chi^2(df = 3) = 50.98$, $p < .001$. As expected, the WS and FXS groups included a
323 significantly higher frequency of children with an ID compared to the Autism and ADHD
324 groups (but no difference in the frequency of ID-status between WS and FXS, or between
325 Autism and ADHD). For receptive language ability, the WS and FXS groups had a higher
326 frequency of children without full sentences compared to the Autism and ADHD groups.
327 Likewise, for expressive language ability, the WS and FXS groups had a higher frequency of
328 children without full sentences compared to the ADHD group, but no difference with the
329 Autism group.

330 *INSERT TABLE 2 HERE*

331 **Procedure**

Separate advertisements invited parents of (i) children with a diagnosis of WS, Autism, ADHD or FXS, and (ii) parents of children with NT, to complete an online survey about their child's social interactions and were distributed via a university research participation database for local families, social media, and UK charity networks. Informed consent was obtained from all participating caregivers/parents following positive ethical opinion from the University ethics committee. Parents did not receive financial remuneration.

Materials

Parents/caregivers reported on their child's eye contact behaviours as part of a larger bespoke survey on social interactions throughout development[2], via online survey software (www.onlinesurvey.ac.uk). In addition to the questions addressing the research aims, parents provided demographic information concerning the child's date of birth, gender, diagnostic status and intellectual disability status. To gather information about language abilities we included the following questions "does your child use language to communicate" (*none*; *single words*; *simple phrases*; *full sentences*), and "does your child understand language" (*none*; *single words*; *simple phrases*; *full sentences*).

The eye contact items corresponded exactly with interview items of Study 1 but the method was distinct as the items were presented in a fixed response format more suitable for a questionnaire. Items were presented as statements with options to select as follows: Item 1 "He/she makes eye contact (even if inappropriate, learned or occasional)" with a response option "yes/no". The next set of items relating to quality of eye contact, unlike Study 1, were not presented sequentially. Instead, they were presented as a forced choice format and caregivers could select only one item in response to the following question: "Please tell us more about the quality of eye-contact. Which of the following applies most usually?" Six response options were offered (shown in full in Table 3) In addition to the three items in Study 1 (staring, unfocused gaze, brief glance), two other items were offered to capture a wider range of qualitative features that might be seen in any of the children. These

[2] The data reported in the current paper were not included in XXX (blinded for review)

were (a) “always appropriate and natural”, and (b) “avoids eye contact”. One of the six (indicating the one that applies most usually) could be ticked. The next item, “If none of the above applies you can give more information here if you wish (this is optional)” allowed parents to elaborate on their child’s eye contact behaviour if it did not easily fit one of the pre-specified categories

Results and Discussion

The first hypothesis, that there would be a high presence of eye contact in WS compared with other ND groups, was not supported. Instead, results showed that the vast majority of all children with a ND condition engaged in eye contact. Although as many as 93% (n=27/29) of parents of children with WS endorsed this item, similar to Study 1, endorsement was also high for Autism: 86% (n=25/29), FXS: 72% (n =13/18) and ADHD: 86% (31/36). A Chi-Square test of Independence showed no significant difference between the four ND groups, χ^2 (3) =3.98, p=.264. Nevertheless, the strong presence of eye contact in all ND groups (96/112, 86%), was still lower than for the NT sample, virtually all of whom were endorsed as showing eye contact (146/149, 98%, one missing response), p <.001 (Fisher’s Exact Test).

The second hypothesis was that unusual qualitative features would be found in WS and in other ND groups. This hypothesis was examined in several ways. Table 3 presents the distribution of responses (i.e. children with endorsement of “yes” to Item 1 reporting presence of eye contact). First, taking the responses for “Eye contact always natural and appropriate” (Column 3 of Table 3), this was the most highly endorsed option for 87% of the parents of NT children and significantly higher than endorsement for the ND sample as a whole (31%; p < .001 Fisher’s Exact Test) or for the WS group alone (44%; p < .001 Fisher’s Exact Test). This evidence supports the prediction that even when children with a ND condition do give eye contact, the quality of their eye contact is not predominantly natural or appropriate. Nevertheless, the WS group did show a significantly higher frequency of

384 "appropriate" eye contact compared to the Autism group (12%; p =.01), but no difference
385 compared to FXS (15.4%; p =.09) or ADHD groups (42%; p = 1).[3]

386 *Insert Table 3 here*

387 Second, initial examination of the pattern of unusual qualitative features revealed that
388 the option "avoidant" was rarely selected for any of the ND groups. This was surprising,
389 given descriptions of avoidance in the Autism and FXS literature indicated by previous
390 literature (Hall et al., 2006; Senju & Johnson, 2009), but it demonstrates parents'
391 interpretation of their child's eye contact quality when selecting from different behavioural
392 options.

393 Subsequent analysis therefore focused on the three unusual quality descriptors from
394 Study 1 (staring, brief glances, and blank, focused gaze). Results showed that the majority
395 of parents in the ND sample selected one of these features as the most usual qualities of
396 their child's eye contact (ranging from 48% to 77% of each group and 54% of the total ND
397 sample) in comparison to only 8% of the NT group. A Fisher's Exact Test confirmed higher
398 endorsement any of these three (see Table 3) in the ND groups taken together (54%)
399 compared to the NT group ($p < .001$, Fisher's Exact).

400 Given the result of Study 1, we did not predict specificity or dominance in one
401 qualitative feature (e.g. staring) for the WS group. However, it was not known whether other
402 ND groups might have specific qualitative features that are distinctive or dominating. To
403 analyse this, a series of 2 x 2 Fishers Exact Chi square analyses were carried out, using
404 only the samples endorsed with brief glances, unfocused gaze or staring (totals from
405 columns 4-6 of Table 3 (i.e., WS n=13; Autism n=14; FXS n=10; ADHD n=15). The
406 categories "unfocused, blank gaze" and "stares" were collapsed together (due to small
407 expected frequencies) and compared with "brief glances"'. This confirmed a different
408 distribution of response: brief glances were more frequently selected for Autism (78.6%, p =

[3] Tested in a series of 2 x 2 Fishers Exact Chi Square analyses, with WS compared with each ND group for responses to the "appropriate" option versus the remaining response options.

409 .05), FXS (90%, p = .03) and ADHD (86.7%, p = .02) groups compared to the WS group
410 (5/13, 38.5%), while the presence of staring behaviour (with unfocused gaze) was more
411 frequently endorsed in the WS group (7/13, 61.5%) This finding supports previous
412 descriptions of “persistent” and prolonged eye contact in young children (Klein-Tasman et
413 al., 2007; Mervis et al., 2003), showing these behaviours are also found in older children and
414 adolescents. In summary, although dominance of one specific qualitative feature was neither
415 predicted nor found, the results indicate that when given a forced choice format, a small but
416 significant proportion of parents of children with WS tend to preferentially select
417 ‘staring/unfocused gaze’ in favour of ‘brief glances’ while the majority of parents of all other
418 ND groups select ‘brief glances’.
419 Only a very small minority of parents selected the option “none of the above apply”
420 (5.4% of the full sample: 5 NT, 8 ND sample), indicating that the options provided were
421 mostly consistent with the range of parent experiences. All of these parents also answered
422 “if none of the above apply please leave further information here (this is optional)”. The
423 majority of the free-text responses (4 NT and 5 ND) reported that the child might show more
424 than one type of eye contact behaviour according to situational or person context.
425 Follow-up analyses examined the relationship between eye contact behaviour, first
426 for presence and then for quality (“unfocused gaze” collapsed with “stares” as above) and
427 the demographic variables: Age, Gender, ID-status (yes/no) and language-status
428 (with/without full sentences) analysed using Chi-square tests. Small samples limited the
429 opportunities for finding significant associations with other demographic variables
430 throughout. No significant associations were found between type of unusual eye contact and
431 language ability (expressive or receptive), ID, gender or age and it was not meaningful to
432 test the comparison between staring and age found in Study 1 because of the sample sizes.

433 **General Discussion**

434 Eye contact strengthens the communication process during human social interaction
435 and shapes our judgements about others (Conty et al., 2016; MacDonald, 2009). For this
436 reason, it is important to understand how eye contact manifests in everyday life for those

with WS and with other ND conditions. The results of Study 1 and 2 show that parents of individuals with WS, nearly all of whom described their child as making eye contact, also described their child's eye contact as unusual rather than natural and appropriate. Our findings support previous evidence showing prolonged and intense looking in individuals with WS and Study 1 also found first evidence of an association between staring and increased age. However, importantly, staring was not the only type of unusual feature as many parents also reported the use of brief glances and blank unfocused gaze.

The cross-syndrome comparison with other ND groups in Study 2 revealed surprising insights. First, the research literature for Autism and FXS, often describes individuals as having reduced or avoidant eye contact. But parents of these children, who must be looking at their children's eyes on an everyday basis, tend not to describe a lack of eye contact. Like the parents of children with WS, most parents of children with Autism, FXS and ADHD reported that their child does make eye contact; however, when given different options to indicate the quality of that eye contact, they indicate an unusual quality to it. The most frequently endorsed feature for parents of all three groups was brief glances, whereas this was not the case for the parents of the WS group who more frequently than the other groups, selected stares or unfocused gaze in this forced choice question format. However, staring/unfocused gaze was not unique to WS and many parents also endorsed brief glances in their children with WS.

This study contributed to the literature by moving beyond the conventional measurement of eye contact as being either present or absent, in varying degree. By separating the measurement of 'presence' from an additional measurement of 'quality', we found different results from studies that have used a single measure of presence of eye contact as an indicator that eye contact is good versus poor. In contrast, our results suggest that nearly all individuals with WS (study 1), and nearly all children whether WS, Autism, FXS or ADHD (study 2), do make eye contact even if in an unusual manner. The type of this unusual quality also seems to be consistently identified by parents as taking the form of brief glances, unfocused gaze or staring, as evidenced by the fact the 'none apply' was rarely

endorsed in Study 2. In Study 2 we also found that the option of "avoids eye contact" was rarely endorsed by parents in preference to these other three items. However, it is not clear why they made this preference. Possibly, the choice of one of six forced choice options constrained them and resulted in few cases of "avoids eye contact". Further research is needed to test out why parents did not choose 'avoids' in preference to other items and to evaluate whether this is because it is not a feature of eye contact according to caregiver perspective, or whether it is because other types of contact behaviour are merely more common.

We learn from the cross-syndrome comparison design of Study 2 that unusual eye contact is found across multiple ND conditions, rather than specific designs being associated with specific patterns of eye contact. It is unclear the extent to which this is due to direct yet variable effects of the ND condition on eye contact, or whether these behaviours are differently acquired through factors which may vary but show commonalities across ND conditions, along with external and internal environment. To disentangle this further, the next stage of research enquiry may benefit from moving towards a more transdiagnostic design. In a recent review on the transdiagnostic model for understanding neurodevelopment, Astle et al. (2021) outline a spectrum of study designs that can offer transdiagnostic insights, which vary in the emphasis placed on diagnostic status. Based upon this classification, studies like ours that test for syndrome-specific associations offer value in elucidating where aspects of cognition and behaviour crossover different ND conditions, or are distinctive. However, this traditional, categorical approach is problematic as it rests on the assumption that ND conditions are homogenous and have clear-cut boundaries; an assumption that does not match up with the clinical reality. Consequently, researchers have argued for the need to reconceptualise neurodevelopment and embrace more transdiagnostic features of design throughout the research process (Astle et al., 2021; Casey et al., 2014; Sonuga-Barke & Thapar, 2021). In the case of research on eye contact, there would be value in following a model similar to that used in research areas of cognition and learning (e.g. Bryant et al., 2020; Mareva et al., 2019) by recruiting a large heterogeneous sample of individuals

with ND conditions known to impact on social attention and social interaction, and stratifying on the basis of particular eye contact styles (the 'diagnostic-blind' approach in Astle et al., 2021).

An important consideration for studies such as ours that do compare groups according to diagnostic label, is that children and adults who receive a diagnosis of any neurodevelopmental condition may also receive other associated diagnoses (Cleaton & Kirby, 2018). Autism frequently co-occurs with other conditions and as atypical eye contact is a diagnostic feature of Autism, this might explain unusual eye contact differences in other conditions as well. As information on co-occurring Autism diagnoses had been collected at the time of recruitment, we were able to carry out further analysis of those with associated diagnoses (WS, n=2, FXS 9 children, ADHD=9). The pattern of results for presence of eye contact and for unusual quality of eye contact remained unchanged, therefore significant effects of an associated autism diagnosis were not evident in this study, but given the small sample sizes, future research designs should test more fully for the effect of co-occurring diagnoses on eye contact presence and quality (see model of study designs outlined in Astle et al., 2021).

Limitations

There are several important limitations to this study. While the results from parent reports in these studies appear striking, it should also be remembered that there are problems using subjective methods of this kind. Parents were aware that this was an interview or questionnaire studying social interactions in those with ND conditions and responses could be attributed to a response bias. Therefore, a recommendation for future research would be for the inclusion of different measures that combine insights from direct observations and experiments, along with multi-informant reports of everyday eye contact. Teacher insights would make a valuable addition given teachers are interacting with children on a regular basis but within a different setting compared to parents.

Another limitation was that the measure adapted from Study 1 for use in Study 2, did not use exactly the same format. Parents were given a forced choice which did not include

options for reporting overlapping types of eye contact quality, as measured in Study 1. This means we cannot make exact comparisons between the measures. Nevertheless, despite differences in the presentation format, the measurement of common behaviour indicators of quality of eye contact (staring, unfocused gaze, brief glances) in each of the two studies contributes new evidence to this sparse literature on the quality of eye contact within WS and across other ND groups. Further testing and replication is still a priority however. Although we might be encouraged by the endorsement rates for Study 2 across the options linked to Study 1, with few choosing the option 'none of these apply', still further validation of the Study 2 method is needed. For example, we recommend further testing of internal, convergent and discriminant validity as has been carried out for other questionnaires using DISCO items (e.g. Jones et al., 2020).

The most serious limitation of the study was that the lack of associations with ID, age and gender, were likely due to a lack of power due to small samples distributed across the ND groups. Although the sample size for the WS group in both studies was the same as the sample size for other studies (Klein-Tasman et al., 2007, 2009) there were limitations in making group-wise comparison for each ND condition and in drawing conclusions on the effects of ID, age and language level. As this was compounded by the constraint on caregivers to select only one of six options to describe their child's eye contact, further replication is needed by comparing larger participant groups and testing different research designs.

Future directions and implications

The relationship between older age and staring behaviour in Study 1 is an intriguing finding. One explanation is that staring behaviour emerges throughout development in WS. Another interpretation is that the reporting of marked staring in adults relates more to a change in the perception of this behaviour. From the perspective of the interlocutor, an adult showing staring behaviour may be more striking and deemed less socially acceptable compared to a child staring. However, it is important to note this association with age was

548 not found in the child-only sample of Study 2, therefore future research should help to
549 corroborate differences and similarities across age and ND groups.
550 Future cross-syndrome comparisons will also benefit from a fine-grained analysis of
551 the differential qualitative aspects of unusual eye contact in relation to social interaction and
552 communication. Klein-Tasman et al. (2007, 2009) noted findings of 'abnormal eye contact' in
553 young children with WS as measured within the ADOS domain of reciprocal social
554 interaction. Common difficulties were also found in the ADOS domains of declarative
555 pointing, showing and giving objects reciprocal social interactions and social communication
556 and cognition. However, as the qualitative nature of abnormal eye contact (e.g. specific type
557 of qualitative features) is not recorded by the ADOS, follow up research using ADOS, DISCO
558 or other assessment measures could help to clarify the relation between particular qualitative
559 types of eye contact and other social interaction, communication and social cognition
560 difficulties. The prediction would be that unusual qualitative features have particular
561 implications for other aspects of social interaction and for social cognition as the flow of
562 interaction is affected.
563 Our findings may also prove useful in future trans diagnostic research, with respect to
564 (1) separating out the cognitive processes involved in attention and arousal, (2) elucidating
565 the neural circuitry associated with eye contact, and (3) the psychosocial factors associated
566 with qualities of eye contact. In terms of the cognitive processes, it may be possible to test
567 whether unfocused gaze is related to slow allocation of automatic attention (Kuhn et al.,
568 2010), whether staring is related to attentional shifting and hypo arousal (Riby et al., 2011),
569 and whether brief glances are linked to gaze aversion strategies during information
570 processing (Doherty-Sneddon et al., 2012). In the case of neural processes, a more trans
571 diagnostic analysis would be particularly informative for revealing the neural processes
572 associated with qualities of eye contact in people with genetic and non-genetic ND
573 conditions. Not only is there a dearth of research documenting how the brain circuitry
574 responds to eye contact in people with ND conditions, to our knowledge, no research has
575 examined how qualitative features of eye contact are sub served by neural substrates.

Indeed the characteristic use of qualitative features of eye contact early in life may itself have a role in neural development indicating bi-directional biology-behaviour relations, rather than a simple underpinning of neural processes driving eye-contact quality. The results also address psychosocial influences on eye contact and how different qualitative features may serve as adaptive functions to increase or avoid social contact when eye contact is experienced as overly stimulating, distracting in some way, or not as socially rewarding. With respect to brief glances for example, for some people who find it aversive to look in the eyes of others (hyperarousal), brief glances may serve to reduce the uncomfortable sensation, as indicated by evidence of increased activation of the subcortical system when focusing on the eye region (Hadjikhani et al., 2017) and first-hand insights from autistic people (McGlensey, 2016; Trevisan et al., 2017). However, brief glances may also indicate an opportunity for information processing during gaze aversion (Doherty-Sneddon et al., 2012). Collecting further parental data on the quality of eye contact used by their child in varying contexts (e.g. interaction partners, social situations) would add valuable insights into the psychosocial factors that may influence eye contact behaviour.

The findings also point to the direction for future research priorities in the areas of FXS and ADHD. Our findings regarding brief glances support previous research with children with FXS. However, the previous research has largely referred to brief glances made while the individual looks elsewhere rather than as part of making eye contact, therefore further fine-grained observational research is needed to examine the extent to which the well documented finding of brief glances in FXS (e.g. Hall et al., 2015) provides a communication strategy for eye contact, at least as far as parents are concerned. At the same time, the results open a new direction of research in ADHD; a ND condition in which eye contact difficulties have previously been neglected. The fact that only 42% of this group showed eye contact that is always appropriate and natural, and similarities in the pattern of unusual eye contact quality to that seen in other ND conditions, should be investigated in relation to their known challenges establishing and maintaining friendships (Normand et al., 2011, 2013) and broader socio cognitive skills (Bora & Pantelis, 2016; Sibley et al., 2010;

Uekermann et al., 2010). Further research is also needed with this group to understand eye contact patterns in those with co-occurring ADHD and autism.

From a clinical and societal perspective, the findings emphasise that eye contact given by people with ND conditions may look different from the neurotypical preference of direct, steady gaze, but that the observable qualities may vary across individuals with the same diagnosis. Difference from a neurotypical pattern of eye contact should not be interpreted as a call for intervention, given these behaviours likely serve an adaptive role. One important consideration however, is the potential impact that different eye contact behaviours may have on the wider social interaction, in terms of impression formation and potential stigma (Morrison et al., 2020); Sasson et al., 2017. Unusual qualities of eye contact may miscommunicate information about the intentions and attitudes of people with ND conditions. For example, brief glances may infer that the person is disinterested in the interaction. Equally, being on the receiving side of prolonged eye contact may be an uncomfortable experience. Prolonged staring at a time of greater social independence during adolescence and young adulthood is particularly important given the vulnerability issues that have been emphasised in people with ND conditions (Fisher et al., 2013; Jawaid et al., 2012; Ridley et al., 2020).

To conclude, it is known that measurement differences lead to particular interpretations of eye contact (Jongerius et al., 2020). We argue that the previous single dimension interpretation, based on measurement of the degree or strength of eye contact, has led to the oversimplified assumption that reduced eye contact equates to poor eye contact while eye contact that is not reduced equates to good eye contact. This in turn, has led to an interpretation that polarises different ND groups such as WS and Autism and makes the incorrect assumption about underlying social motivational and cognitive factors. Given our findings on similarities across ND conditions, we think it is time to focus on describing eye-contact profiles more in terms of different qualitative styles, and less in terms of a single dimension (i.e. degree of presence/absence). This new perspective would have implications for research on psychological and neural mechanisms related to eye-contact as

it indicates that quality of eye contact subtypes may be studied independently of traditional diagnostic groupings and divisions.

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Table 1

Data for each individual with WS for DISCO items assessing eye contact and language and visuospatial skill level

Age in months	M/F	Language Delay		Age-appropriate level of current skill			Eye contact present	Unusual quality of eye contact (marked or frequent)		
		Late to use 2-3 phrases	Late to understand word meanings	Expressive language Level 1-9 [a]	Receptive language Level 1-7 [b]	Visuospatial skill Level 1-12 [c]		Brief glances	Blank unfocused gaze	Stares
72	F	Yes	Yes	8	5	9	+	+	+	+
89	M	Yes	Yes	8	5	5				
100	F	No	No	9	6	9	+			
101	M	–	–	9	3	12	+			
106	M	Yes	Yes	9	7	10	+	+		
115	F	Yes	Yes	7	6	10	+			
124	F	Yes	Yes	8	7	10	+			
153	M	Yes	Yes	9	7	9	+		+	
159	M	Yes	Yes	9	7	9	+			
161	F	Yes	Yes	9	5	12	+			
172	F	Yes	Yes	9	4	10	+	+	+	+
193	M	No	–	9	4	10	+	+	+	+
193	M	–	Yes	8	5	8	+	+	+	+
205	F	Yes	Yes	8	4	–				
206	M	Yes	Yes	9	4	12	+		+	+
210	F	No	Yes	9	7	6	+			
258	F	Yes	Yes	8	5	8	+			
277	M	Yes	Yes	9	5	6	+	+		
286	F	–	No	9	7	8	+	+	+	+
301	M	Yes	Yes	9	7	3	+			+
396	F	No	No	9	6	8	+			+
435	F	–	–	–	6	–	+	+	+	+

Note. Dash sign (–) = parent data was not available. Cells with plus sign (+) indicate endorsement of either (a) presence of eye contact and (b) unusual quality of eye contact at a marked level.

[a] Language expression: 0-2 = No speech or babbles, 3-4 = Says names for things only, 5 = says phrases of 2 words only, 6 = Says longer phrases, 7 = Uses spontaneous sentences, present tense only, 8 = Uses sentences/phrases including 'but' and 'because', 9 = Uses past, present and future tenses and complex grammatical constructions.

[b] Language comprehension: 0-1 = No response or responds to name only, 2 = Understands simple words from phrases in context (learned from gestural cues, e.g. time for bed), 3 = Knows the meaning of some words and can responds e.g. 'give me your cup', 4 = Follows instructions involving 2 new objects "Put the doll on the chair", 5 = Can reliably follow instruction to fetch 2 or more objects from outside of the room, 6 = understands a sequence of commands, 7 = Understands instructions involving decisions (conditionals) "see if my phone is in my bedroom and if not look for it in the bathroom".

[c] Visuospatial skill: 0 = does not hold objects in hands, 1 = holds objects in hands, 2 = examines objects, 3 = handles objects, 4 = rolls toys on floor, 5= builds tower of 2-5 bricks, 6 = builds tower of 6 bricks, 7 = arranges objects in size order, 8 = completes puzzle 6 pieces, 9 = completes puzzle 10 pieces, 10 = completes puzzle 20-30 pieces, 11 = completes puzzle 50 pieces, 12 = completes puzzle 150 pieces.

Table 2

Demographic characteristics of the sample (% reported) split by diagnostic group

Demographic variables	Autism (*n* = 29)	WS (*n* = 29)	ADHD (*n* = 36)	FXS (*n* = 18)	NT (*n* = 150)
Males/females/prefer not to say	72/28/0	59/41/0	78/19/3	94/6/0	48/51/1
Age (months)					
M (*SD*)	127 (28.4)	100 (36.3) [a]	127 (38.8) [b]	118 (36.9)	107 (45.8) [c]
Range	59-187	48-204	54-179	54-197	48-215
Presence of an intellectual disability	21	90	28	89	0
Expressive language					
None	3	7	0	11	1
Single words	3	7	0	17	0
Simple phrases	7	24	6	33	0
Full sentences	86	62	94	39	99
Receptive language					
None	0	0	0	6	0
Single words	0	7	0	0	0
Simple phrases	17	28	6	28	0
Full sentences	83	66	94	67	100

[a] Missing data (*n* = 1). [b] Missing data (*n* = 1). [c] Missing data (*n* = 1).

Table 3

Quality of eye contact behaviour endorsed by parents in each group

Group	*n* [a]	Quality of eye contact applied most usually					
		Eye contact always appropriate and natural	Brief glances	Blank, unfocused gaze	Stares	Avoids eye contact	None of these apply
WS	27	12 (44.4)	5 (18.5)	1 (3.7)	7 (25.9)	1 (3.7)	1 (3.7)
Autism	25	3 (12)	11 (44)	2 (8)	1 (4)	3 (12)	5 (17.2)
FXS	13	2 (15.4)	9 (69.2)	0 (0)	1 (7.7)	0 (0)	1 (7.7)
ADHD	31	13 (41.9)	13 (41.9)	1 (3.2)	1 (3.2)	2 (6.5)	1 (3.2)
Neurotypical	143[a]	124 (86.7)	11 (7.7)	1 (0.7)	0 (0)	2 (1.4)	5 (3.5)
Total ND	96	30 (31.3)	38 (39.6)	4 (4.2)	10 (10.4)	6 (6.3)	8 (8.3)

Note. Percentages are presented in parentheses.

[a] Parents who reported “yes” to Q1 about the presence of eye contact. [b] Of the 146 TD parents who reported yes to Q1, 3 data points were missing.