REPORT

Nature and nurture: Williams syndrome across cultures

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Abstract

This study is concerned with ways in which children with Williams syndrome (WS), a rare neurodevelopmental disorder arising from a hemizygous deletion in chromosome band 7q11.23 including the gene for elastin (ELN) and approximately 20 surrounding genes, are affected by social mores of vastly differing cultures: the United States and Japan. WS presents a compelling model for the investigation because its genetic phenotype is well defined and results in an uneven cognitive profile as well as a social phenotype typical of the syndrome including overt over-friendliness toward strangers. While a number of research groups have been studying the cognitive strengths and weaknesses of individuals with WS in various countries, there have not been studies to date that explore the social phenotype in WS across different cultures. This study examines the ways in which social behavior in WS, stemming from specific genetic underpinnings, might be mediated by cultural expectations. We conducted a cross-cultural study using an instrument that measures aspects of sociability commonly found among people with WS. Quantitative analyses revealed a significant effect of diagnostic category in that in both countries, children with WS were rated as significantly higher in global sociability and more likely to approach strangers than were their normal counterparts. There was also an effect of culture, in that regardless of category, WS and normal children in Japan were rated lower than their counterparts in the US. We suggest that the excessively social phenotype of children with Williams syndrome, although markedly present across cultures, appears to vary in its intensity by culture. This is an intriguing illustration of interactions between nature and nurture.

Introduction

Williams syndrome (WS) is a rare neurodevelopmental disorder arising from a hemizygous deletion in chromosome band 7q11.23, including the gene for elastin (ELN) and approximately 20 surrounding genes (Ewart, Morris, Atkinson, Jin, Sternes & Spallone, 1993; Korenberg, Bellugi, Salandanan, Mills & Reiss, 2003; Korenberg, Chen, Hirota, Lai, Bellugi & Burian, 2000). It is estimated that more than 95% of individuals clinically diagnosed with WS have deletions that fall within the same breakpoints (Perez Jurado, Peoples, Kaplan, Hamel & Franke, 1996). Physical characteristics of WS include specific facial and physical anomalies; a variety of cardiovascular difficulties, commonly supravalvular aortic stenosis; mild to moderate mental retardation; failure to thrive in infancy; and small stature (Bellugi, Lichtenberger, Jones, Lai & St George, 2000; Morris & Mervis, 1999). In addition, adults with WS often display a characteristic pattern of cognitive strengths and weaknesses, that is, comparatively strong language abilities coupled with profound deficits in visuospatial functioning (Bellugi et al., 2000; Mervis, Robinson, Bertrand, Morris, Klein-Tasman & Armstrong, 2000).

In addition to the typical cognitive profile, a consistent behavioral characteristic of WS is heightened affiliative behavior as defined, in part, as being “overly friendly” [or “hypersocial”] compared to age-matched controls, and more “unreserved with strangers” as compared to children with non-specific mental retardation at the same chronological ages’ (see Doyle, Bellugi, Korenberg & Graham, 2004a; Jones, Bellugi, Lai, Chiles, Reilly & Lincoln, 2000; Klein-Tasman & Mervis, 2003; Mervis & Klein-Tasman, 2000, for reviews). Almost since its characterization as a syndrome, anecdotal observations have been made that persons with WS were outgoing.

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overly social behavior in WS are well documented within the presence of excessive friendliness toward strangers or cognitively impaired; nor can they be attributed to developmentally unwarranted conventions governing others, as both WS and DS are resulting in a lack of understanding of the social world. These findings provide initial evidence that differences in approach toward strangers as assessed, with WS exceeding both comparison groups. In addition, children with WS were rated significantly more social overall than were those with DS or TD subjects. In the United States, the “good child” is assertive, socially competent with peers, and courteous (p. 158). Moreover, parental reports contrasting children in these countries of ‘self’ in the United States and in Japan, one might expect such differences to lead to variations in the ways in which children in the two countries are socialized. Hess, Azuma, Kashiwagi, Dickson, Nagano and Holloway (1986) note that ‘In Japan, a child is thought to be good if he or she is “obedient” (sunao), “mild and gentle” (otonasii), and “self-controlled” (jiseishin ga aru). In the United States, the “good child” is assertive, socially competent with peers, and courteous’ (p. 158). Moreover, parental reports contrasting children in these countries have shown that mothers in Japan rate their children as being shyer and less sociable than do mothers in the United States (Stevenson, Lee, Chen, Stigler, Hsu & Kitamura, 1990). (For further discussion on differences between US and Japanese socialization and child-rearing practices, see Conroy, Hess, Azuma & Kashiwagi, 1980; LaFreniere, Masataka, Butovskaya, Chen, Auxiliadora-Dessen & Atwanger, 2002; Lebra, 1994; Masataka, 2002; White, 1993; Zahn-Waxler, Friedman, Cole, Mizuta & Hiruma, 1996.)
Our previous studies suggest the involvement of a genetic predisposition in the expression of hypersociability in WS (Bellugi, Adolphs, Cassidy & Chiles, 1999; Doyle et al., 2004a; Doyle, Bellugi, Reiss, Galaburda, Mills & Korenberg, 2004b; Jones et al., 2000). Thus, exploring sociability across cultures can provide keen insight into the interplay of temperament (in a disorder with a known genetic basis) and culture. While a number of research groups have been studying the cognitive strengths and weaknesses of individuals with WS in various countries, there have been no studies that directly explore social behavior in WS across cultures. This study examines the ways in which social behavior in WS, which is thought to have a genetic predisposition, might be mediated by cultural expectations in both Japan and the United States.

Method

Participants

Participants included the parents of children living in Japan (24) and the United States (24). Twelve of the children in each sample had Williams syndrome (WS) and 12 were typically developing (TD). The children from each culture were matched individually with regard to age (between 3y, 3m and 13y, 7m) and gender (males and females are equally represented). Mean ages and standard deviations for each experimental group are displayed in Table 1.

The US participants were parents of children with WS attending a meeting of the Williams Syndrome Association, and the parents of TD children who attend school near the Salk Institute in California. The Japanese WS data were collected by Mr Sugimoto, a member of the Japanese Williams Syndrome Association, who ensured that each person completing the questionnaire understood the materials, including the informed consent form, and was willing to participate. Japanese TD data were collected through the laboratory of co-author Dr Nobuo Masataka of Kyoto University.

Procedures

The Salk Institute Sociability Questionnaire (SISQ) is an instrument specifically developed to assess specific aspects of social behavior commonly reported among people with WS first reported in Jones et al. (2000). The SISQ has been used in a variety of different contexts and across age groups as indicated previously. Moreover, the Salk Institute lab has collected data on over 80 adolescent and adult individuals with WS; of these, 44 had completed both the SISQ and another standardized parent-report instrument, the Multidimensional Personality Questionnaire (MPQ; Tellegen, 1985). The Salk studies find that the SISQ overall scores show high correlations with the MPQ in the WS cohort on social dimensions such as Social Potency and Social Closeness, whereas there is no correlation with other MPQ measures, nor with IQ. The MPQ has also been used by Klein-Tasman with a different cohort of individuals with WS (Klein-Tasman & Mervis, 2003). Similar to the findings from the Salk team, Klein-Tasman and Mervis report in using the MPQ that the distinctiveness of the WS personality appears to lie in the focus of WS on others, a pattern characterized by an eagerness to interact with others as well as high levels of tension and sensitivity. This distinctiveness of the WS social phenotype provides the groundwork for the present study of cross-cultural influences upon social behavior.

To ensure consistency between the English and Japanese versions of the SISQ for the cross-cultural comparison in this study, two individuals, fluent and literate in both the English and Japanese languages, independently translated the SISQ from English into Japanese and then back-translated from Japanese to English.

Results

The SISQ consists of both quantitative and qualitative item types. Quantitative items ask parents to rate their child’s specific social behavior on a 7-point Likert scale. Qualitative items instead ask the parents to fill in a descriptive response. The items yield three subscales: tendency to approach strangers, tendency to approach familiars, and social emotional behavior (such as tendency to empathize with others, accuracy of emotional evaluations of others, eagerness to please others, and ability to remember names and faces of others). The three subscales together comprise Global Sociability. Items assessing social approach behavior consist of statements such as ‘How would you compare your child’s tendency to approach strangers with an average child of the same age?’ on a scale ranging from 1 (‘approaches much less’) to 7 (‘approaches much more’); or ‘How would you describe your child’s general behavior in social situations?’ on a scale ranging from 1 (‘very shy and inhibited’) to 7 (‘extremely outgoing’). Qualitative items include ‘Describe your child’s typical

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<th>WS</th>
<th>TD</th>
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<tr>
<td>Japan</td>
<td>Mean (SD)</td>
<td>7.22 (3.48)</td>
</tr>
<tr>
<td>United States</td>
<td>Mean (SD)</td>
<td>7.30 (3.04)</td>
</tr>
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reactions when meeting someone for the first time (please give examples); or ‘Give some examples of your child’s socializing with strangers.’ The social approach items were grouped for analysis into two types: those that assess the child’s tendency to approach family members or others encountered frequently (yielding an ‘Approach Familiars’ score) and those that assess the child’s tendency to approach people unknown to them (an ‘Approach Strangers’ score). The Social-Emotional score was the sum of four items; Approach Familiars score was the sum of three items; Approach Strangers was the sum of five items; and the Global Sociability score was the sum of all 12 items.

Quantitative data analysis

The quantitative data were analyzed by a 2 × 2 analysis of variance (ANOVA) with Diagnostic Category (Williams syndrome children versus Typically Developing children) and Culture (American versus Japanese) as independent variables and the summary Global Sociability score as the dependent variable. Wilks’ Lambda criterion was used to assess significance. This analysis revealed a significant main effect of Diagnostic Category (WS versus TD), $F(1, 44) = 18.07, p < .0001$, and a significant effect of Culture (Japanese versus American), $F(1, 44) = 11.10, p < .002$. The Diagnostic Category by Culture interaction failed to reach significance, $F = .42, p = > .05$, suggesting that the pattern of differences between the diagnostic groups was similar across the two cultures. Figure 1 shows the data distribution for the Global Sociability overall composite score combining all questionnaire items, and the data for each of the three subscales.

As can be seen from the figure, both the American and Japanese children with WS were rated significantly higher on Global Sociability than were the typically developing children; thus, there was a very strong effect for Diagnostic Category (WS or TD). At the same time, there was also a significant effect of Culture, in that parents of children in the US tended to rate their children as higher in Global Sociability than did parents of children in Japan, regardless of their diagnostic category (WS or TD). Comparison of scores across Cultures indicates a major difference for Approach Strangers, but...
Williams syndrome across cultures 759

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not for Approach Familiars (scores were nearly at ceiling) nor Social-Emotional items. Similarly, comparison of scores across Diagnostic Category indicates a major difference for Approach Strangers, but not for the other two subscales. Overall, these results illustrate that the significant between-group differences in Global Sociability were primarily contributed to by higher ratings of ‘Approach Strangers’ for both Diagnosis and Culture.

Qualitative analysis of sociability data

The SISQ also asks parents to provide qualitative descriptions of their child in various social situations with items such as, ‘Describe your child’s typical reactions when meeting a stranger for the first time (please give examples).’ Table 2 presents sample qualitative responses from the two cultural and diagnostic groups selected at different ages for this question.

Qualitative responses were further examined in order to address the problems of reference group effects (Heine, Lehman, Peng & Greenholtz, 2002) and cultural response biases that could be reflected in the use of a Likert-style questionnaire (Chen, Lee & Stevenson, 1995). For this analysis, all responses across categories and cultures were presented in randomized order not revealing their sources, and were translated into both languages. Consistent with the analysis reported in Doyle et al. (2004a), the raters were blind to the participants’ diagnostic classification and cultural membership. Two independent American and two independent Japanese raters, blind to the subjects’ identities, categorized all the responses into three behavioral groups: Shy, In-between or Social. While all parents (12) of the US-WS children provided an example to this questionnaire form, only five parents of US-TD children did so. There were 10 responses for the J-WS group, and 11 responses for the J-TD group. As a cross-cultural check, we also asked two Japanese raters to categorize the qualitative responses in the same way. Table 3 shows the frequencies of the ratings in percentages from both the American and Japanese raters for the four participant groups within and across the two cultures.

Comparing the two, despite the small number of subjects, some trends emerge. American and Japanese raters both categorized the responses from WS individuals as more social regardless of culture, and both groups of raters categorized more of the TD responses as ‘Shy’. However,

<table>
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<th>Table 2</th>
<th>Sample responses to qualitative item 5: ‘Describe your child’s typical reactions on meeting a stranger for the first time’</th>
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<tbody>
<tr>
<td>(3–4 years)</td>
<td>She flinches and comes to parents.</td>
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<td></td>
<td>He says words that are used in greeting like ‘hello’ and ‘what are you doing?’ It happened frequently when he just learned the words. Although it is not happening as frequently as before, when he wants to brag about something, he still approaches strangers and starts talking to them.</td>
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<td>(5–8 years)</td>
<td>She often holds my hand tightly, stays behind me and observes the person. She will greet if told to do so. She observes how I, her mother, respond to the person and tries to correspond the situation.</td>
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<tr>
<td></td>
<td>She always greets them by saying, ‘Hi! I’m ___’ energetically. If someone talks to her, she happily starts to talk about different things.</td>
</tr>
<tr>
<td>(9–13 years)</td>
<td>She more likely watches the person from a distance rather than talking to him/her. She has a little difficulty greeting even after she is introduced to the person. She tries to minimize her words when she has to answer.</td>
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<tr>
<td></td>
<td>He starts talking about himself by looking straight into a person’s eyes. He says, ‘What are you doing.’ or ‘I’m K__.’ He still greets people by saying ‘hello’ to strangers passing by.</td>
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</tbody>
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<th>Table 3</th>
<th>Summary of data from Japanese and US raters for item 5. Responses were randomized across Diagnostic Category and Culture. Raters from US and Japan, blind to the purposes of the study, were asked to categorize each response as ‘Shy’, ‘Social’ or ‘In-between’</th>
</tr>
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<tbody>
<tr>
<td>Characterized as</td>
<td>Percentage of responses</td>
</tr>
<tr>
<td>US Raters</td>
<td></td>
</tr>
<tr>
<td>Shy</td>
<td>45</td>
</tr>
<tr>
<td>In-between</td>
<td>55</td>
</tr>
<tr>
<td>Social</td>
<td>0</td>
</tr>
<tr>
<td>Japanese Raters</td>
<td></td>
</tr>
<tr>
<td>Shy</td>
<td>41</td>
</tr>
<tr>
<td>In-between</td>
<td>45</td>
</tr>
<tr>
<td>Social</td>
<td>14</td>
</tr>
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</table>
even here, there is a trend toward cultural difference. The Japanese raters categorized some of the WS responses as ‘Shy’, and some of the J-TD responses as ‘Social’, whereas none of the American raters did so. Thus, the qualitative data complemented the quantitative data, in that there is a strong effect for Diagnostic Category, but some differences are observed among the cultural groups with respect to the frequencies of responses characterized as Shy, Social, and In-between.

Discussion

Williams syndrome provides a compelling model for investigating the effects of genotype, phenotype and environmental interactions. The genetic basis of WS is by now well known and documented in great detail (unlike, for example, the basis of autism). The genetic phenotype thus involves absence of one copy of a small set of genes on chromosome 7, occurring in nearly all clinically identified WS (Korenberg et al., 2003; Morris & Mervis, 1999). Currently several groups are working to begin to link genotype and phenotype in Williams, and the hunt is on to link specific genes within the WS region with the medical characteristics, brain development and behavioral functions (see Doyle et al., 2004a, 2004b; Korenberg et al., 2003; Meyer-Lindenberg, Hariri, Munoz, Mervis, Mattay & Morris, 2005; Meyer-Lindenberg et al., 2006, for examples).

In our studies, we highlight the consistency and variability of sociability in children with WS, which in turn allows for consideration of the expression of sociability not only in those Williams individuals with typical size deletions but also in Williams children with atypical deletions. Included in the large-scale study of development of sociability in young WS, DS, and TD described above were the data from a young child with WS who had a smaller deletion than is typical of the syndrome, retaining between one to three genes in the telomeric region that are almost invariably deleted in the ‘classic’ deletion. This child had the typical medical and cognitive diagnostic characteristics for WS. However, her sociability scores, especially those for approaching strangers, were significantly lower than the mean of the WS group, implicating specific genes in the emergence of this behavior in WS (Doyle et al., 2004a, 2004b). The relative lack of variability in hypersociability among children with WS gleaned both from parental report and experimental as well as observational measures, combined with the early age of onset, and the results obtained for the child with the atypical deletion, suggests that the behavioral feature of hypersociability in approaching strangers may be strongly influenced by the genetic deletion (see also Klein-Tasman & Mervis, 2003), as well as tempered by environmental factors.

To date, there have not been investigations of hypersociability across cultures in Williams syndrome. A cross-cultural study of expressiveness in language conducted by Reilly, Bernicot, Vicari, Lacroix and Bellugi (2003) with children in France, Italy, and the US found that the form and intensity of Williams’ social language behavior is influenced by the individual culture’s social tenets for expressing sociability. Reilly et al. reported that all groups of children with WS used more evaluative language than their TD controls; however, the effect was culturally specific. In other words, while individuals with WS across cultures and languages demonstrate atypically expressive use of language, this increase in evaluative language is relative to the baseline observed in the cultures of the respective individuals.

The results of the present study support a genetic ‘proportional stamp’ on the expression of social behavior in WS across cultures as children with WS in the US and in Japan showed more affinity for approaching strangers and were rated higher on Global Sociability than TD children in their respective countries. The present study was conducted to examine, using a common instrument, social behavior among children with WS in Japan and the United States, in order to investigate the ways in which social behavior is influenced by cultural expectations or mores. These results suggest that the WS social and genetic phenotype may influence the degree of expression of affiliative behaviors towards strangers, even among children for whom a cultural expectation of cautiousness towards strangers exists. We observed, by both quantitative and qualitative measures, nature’s stamp on culture’s milieu. In other words, the expression of sociability in WS may not be ‘either/or’ phenotype/culture, it is rather ‘both/and’.

Alternative explanations for the results reported here may still be made. It is interesting to note in Table 2 that the examples given for specific behaviors described by parents of children with WS to illustrate approaching and socializing with strangers are very similar, regardless of culture. Nevertheless, Japanese parents rated their children lower on the 7-point scale than did US parents; thus, an alternative explanation for the difference in quantitative scores obtained for Japanese children with WS as compared to their US peers may be that cultural influence is exerted most on parents’ ratings rather than on the expression of the behavior itself. Perhaps the stigma of having a ‘different’ child in Japan affected the ways in which parents ranked their child’s degree of sociability. Thus, further cross-cultural observational studies of children with WS interacting with strangers are needed to resolve possible discrepancies caused by
reliance on parental reports in order to gauge the interplay of phenotype and culture in WS.

This survey of WS in US and Japanese cultures examines the social behaviors that typify WS, and finds that despite the differences in upbringing between the two cultures, individuals with WS are found to be more sociable in approaching strangers than typically developing individuals, thus demonstrating the impact of the WS phenotype. The unique nature of WS allows us to investigate the dual influences of nature and nurture; thus, continuing to examine Williams syndrome across cultures will be an important avenue for further exploration.

Acknowledgements

We would like to thank first and foremost the families who contributed to this research. Without the continuing support of families living with Williams syndrome, our understanding of the interplay of nature, nurture and the human condition would not be nearly as advanced as it is today. One parent, Mr Sugimoto of the Japanese Williams Syndrome Association, was particularly helpful as he translated much of the materials into Japanese and acted as a liaison for the Japanese families who chose to participate. We particularly thank Dr Anna Jarvinen Pasley and Yvonne Searcy for their contributions to the analyses. All illustrations are copyrighted to Ursula Bellugi. This study was supported in part by the Oak Tree Foundation and by NIH grants HD33113 and NS 22343, which were awarded to Ursula Bellugi.

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