Research in Developmental Disabilities 32 (2011) 1021–1029

Contents lists available at ScienceDirect

Research in Developmental Disabilities

Patterns and trajectories in Williams Syndrome: The case of visual orientation discrimination

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ARTICLE INFO

Article history:
Received 18 January 2011
Received in revised form 19 January 2011
Accepted 19 January 2011
Available online 21 February 2011

Keywords:
Williams Syndrome
Genetic disorder
Development
Visual orientation perception

ABSTRACT

Williams Syndrome (WS) is a developmental disorder typified by deficits in visuospatial cognition. To understand the nature of this deficit, we characterized how people with WS perceive visual orientation, a fundamental ability related to object identification. We compared WS participants to typically developing children (3–6 years of age) and typical adults in an orientation discrimination task with four stimulus types (small circular, large circular, collinear elongated and parallel elongated gratings). We measured orientation discrimination thresholds and the proportion of orthogonal errors (i.e., mirror-image reversal errors). We evaluated how these metrics (1) are modulated by stimulus condition, and (2) vary with chronological or mental age. We found that orientation perception in WS is comparable to that of typically developing children. Orientation discrimination thresholds were better for elongated gratings than circular gratings across all participant groups. For large circular gratings, the proportion of orthogonal errors was disproportionately greater in WS participants and typically developing 3–6 year old children than in typical adults. Moreover, we found that the ability to judge orientation in WS improves with increasing mental age, but not chronological age. These results suggest that orientation discrimination in WS is developmentally arrested, as opposed to abnormal or delayed.

1. Introduction

A foundational skill in visuospatial processing is orientation discrimination, which is an important component of scene perception, object recognition, and reading. Failing to detect and discriminate orientation information would result in misidentified objects. In the current study, visual orientation discrimination was evaluated in Williams Syndrome (WS), a genetic disorder that typically causes an unusual cognitive profile of impaired visuospatial abilities juxtaposed with relatively strong language abilities. Because orientation representation is fundamental in everyday "seeing", it is important to characterize its limits and understand its underlying mechanisms. Results from this study could lead to future work that can help ameliorate visuospatial difficulties in WS and other developmental disorders.

1.1. Williams Syndrome

WS is a genetic disorder that occurs in 1 out of 7500 live births (Stromme, Bjornstad, & Ramstad, 2002). It is associated with a microdeletion of about 20 genes on chromosomal region 7q11.23 (Lenhoff, Wang, Greenberg, & Bellugi, 1997) that

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doi:10.1016/j.ridd.2011.01.038

causes mild to moderate mental retardation (mean IQ of ~60). The cognitive profile of WS is remarkable because it can inform a genetic model of atypical visuospatial cognition.

People with WS have irregularities across multiple levels of the visual stream. They have ocular features that suggest that perceptual functions may be partially responsible for the deficits in visuospatial cognition. Winter, Pankau, Amm, Gosch, and Wessel (1996) reported that out of 152 WS individuals, 54% had a history of strabismus, and 74% had a stellate (star-shaped) pattern of the iris. They also reported that of the 46 people who had an ophthalmoscopic examination, 22% had retinal vascular tortuosity, which can cause vision loss due to inefficient oxygen transport.

Moreover, cortical structure and function of visual areas have been reported to be atypical. The primary visual cortex (V1) of WS participants has an abnormal shape and a smaller volume than typical controls (Reiss et al., 2000), which suggests that visuospatial deficits in WS individuals are likely linked to deficits in basic perceptual functions. The visuospatial deficits found in WS have been connected to abnormalities within the dorsal pathway of visual processing (Atkinson et al., 1997). Relative to typical adult controls, parietal areas in WS show hypoactivation (Meyer-Lindenberg et al., 2004). Parieto-occipital areas in WS have significant reductions in gray and white matter (Thompson et al., 2005), which correlate with reduced sulcal depth (Kippenhan et al., 2005).

1.2. Orientation discrimination

The cortical representation of orientation information begins in V1 (Ringach, Hawken, & Shapley, 1997), which contains neurons tuned to orientation. The orientation tuning of these neurons has been shown to match well with the orientation tuning measured from psychophysical behavior (Ringach, 1998). Nonetheless, extrastriate cortex has also been implicated in orientation processing. The occipito-parietal junction has been reported to encode object orientation, while the ventral temporo-occipital junction encodes object identity (Valyear, Culham, Sharif, Westwood, & Goodale, 2006). Moreover, inferotemporal cortical neurons (IT) might mediate left-right mirror-image reversal errors (Rollenhagen & Olson, 2000). Given that WS individuals have been shown to have abnormalities in primary and association visual areas, they could exhibit a deviant pattern of performance regarding orientation discrimination.

Initial studies of visual orientation processing in WS indicated no orientation selectivity at all, which would indicate profoundly deviant visual perception since sensitivity to orientation differences is found in young infants (Slater, Morison, & Somers, 1988). Many WS participants were reported to fail the pretest Benton judgment of line orientation task, JLOT (Wang, Doherty, Rourke, & Bellugi, 1995). In contradistinction, WS performance in the Benton face recognition task is very close to the performance of typical adults (Bellugi, Lichtenberger, Mills, Galaburda, & Korenberg, 1999; Wang et al., 1995).

However, the floor performance of WS individuals in JLOT may be due to the complexity of the task. The precision and accuracy of orientation discrimination in WS adults have been found to be similar to those of young children (Dilks, Hoffman, & Landau, 2008). Like typical children and adults, WS participants show the oblique effect, which is the relative imprecision of recognizing diagonal targets compared to horizontal or vertical targets. Like typical children of about 5 years of age, WS participants also confuse a diagonal target with its vertical axis mirror-image (Palomares, Landau, & Egeth, 2009). These results suggest that WS representation of orientation is functionally delayed or arrested, rather than functionally deviant.

While extensive research has been conducted on the mechanisms of orientation representation in typical adults, we sought to characterize this deficit in WS individuals by evaluating: (1) patterns of modulation by stimulus condition (Palomares, Landau, et al., 2009) and (2) developmental trajectories (Thomas et al., 2009). These independent analyses provide a comprehensive picture of WS abilities in judging visual orientation. In the current study, the size and shape of the stimulus gratings were varied in order to assess modulations in orientation discrimination precision and accuracy in typical children and adults and in people with WS. Participants' orientation thresholds and error patterns were measured for small circular, large circular, and elongated gratings. Whether increasing the size of a circularly symmetrical grating (Henrie & Shapley, 2001) or increasing its length would improve orientation discrimination across participant groups was evaluated. If orientation discrimination is better for collinear or parallel elongated gratings than for large circular gratings, then results would be consistent with the idea that processing of boundaries (i.e. the orientation of Gaussian envelope) is prioritized over processing of internal surfaces (i.e. the orientation of the carrier grating) (Alvarez & Cavanagh, 2008). More importantly, if WS individuals and typical participants show a different pattern of results as a function of size and length, then visual orientation processing in WS is developmentally deviant. Alternatively, WS individuals might show a similar pattern to typically developing children, which would mean that visual orientation processing in WS is developmentally immature. Lastly, WS individuals might show a similar pattern to typical adults, which would indicate a developmentally intact orientation representation.

The general ability to judge orientation has been shown to improve with increasing age in typical development. While it has been reported that orientation discrimination in WS is worse than that of typical adults (Palomares, Landau, et al., 2009), it is unclear whether orientation processing also improves with age in this disorder. Correlation analyses on thresholds and proportion of orthogonal errors (i.e. mirror-image reveral errors) against chronological age were conducted to assess developmental trajectories. If orientation discrimination reliably improves with chronological age in our WS sample of adolescents and adults, it would imply that this skill is develops more slowly in WS and in typical participants. If orientation processing in WS shows no improvement with age but is worse than in typical adults, it would suggest that this skill is developmentally arrested rather than developmentally delayed. Orientation discrimination data were also plotted against equivalent mental ages in WS to evaluate whether orientation processing is linked to general cognitive abilities.