



Understanding motor acts and motor intentions in Williams syndrome

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ABSTRACT

Williams syndrome (WS) is a rare genetic disorder associated with unusually hyper-social demeanor and ease with strangers. These personality traits are accompanied by difficulties in social interactions, possibly related, at least in part, to a difficulty in understanding others' mental states. Studies on mentalizing capacities in individuals with WS have often led to contrasting results, some studies revealing specific impairments, others highlighting spared mentalizing capacities. So far, however, no study investigated the performance of individuals with WS in non-inferential understanding of others' motor intentions. In the present study we investigated this capacity by using a computer-based behavioral task using pictures of hand-object interactions. We asked individuals with WS first to describe what the other was doing (i.e. a task implying no kind of intention reading), and secondly, if successful in answering the first question, to describe the motor intention underlying the observed motor acts (i.e. why an act was being done, a task requiring non-inferential motor intention understanding). Results showed that individuals with WS made more errors in understanding what the other was doing (i.e. understanding a motor act) compared to both mental-age matched controls and chronological-age matched peers with typical development, while showing mental-age appropriate performance in understanding why an individual was acting (i.e. understanding a motor intention). These findings suggest novel perspectives for understanding impairments in social behavior in WS.

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1. Introduction

Williams syndrome (WS) is a genetic disorder caused by a hemizygous deletion in chromosome band 7q11.23, including the gene for elastin (ELN) and about 20 surrounding genes (Ewart et al., 1993; Korenberg et al., 2000). Incidence was evaluated as 1:20,000 live births (Morris, Demsey, Leonard, Dilts, & Blackburn, 1988), but more recent estimates speak of 1:7500 live births (Stromme, Bjornstad, & Ramstad, 2002). WS is associated with relatively low IQ (Mervis et al., 2000; Searcy et al., 2004; Vicari, Carlesimo, Brizzolara, & Pezzini, 1996) and a neuropsychological profile of strengths and weaknesses in both verbal and visuo-spatial domains (Bellugi, Lichtenberger, Jones, Lai, & St George, 2001; Brock, 2007; Levy & Bechar, 2003; Vicari, Caselli, Gagliardi, Tonucci, & Volterra, 2002; Volterra, Capirci, Pezzini, Sabbadini, & Vicari, 1996).

A consistent behavioral characteristic of individuals with WS is their hyper-social character (Bellugi, Adolphs, Cassady, & Chiles,

1999; Mervis & Klein-Tasman, 2000). Children with WS are often described as displaying a natural drive toward others (Doyle, Bellugi, Korenberg, & Graham, 2003), being gregarious, people-oriented and sensitive (Klein-Tasman & Mervis, 2003). Nonetheless, this outgoing personality does not result in appropriate behaviors in social contexts, as individuals with WS tend to approach others in an indiscriminate over-friendly way. Some studies ascribe this behavior to an overreliance on evident social signals, typically viewed as positive cues (e.g. a smiling face), and parallel inability to catch more subtle ones (e.g. furrowed eyebrows) (Bellugi et al., 1999; Jones et al., 2000). Other studies suggest that these hyper-social behaviors are due to the presence of a strong drive toward social stimuli and to a lack of inhibition (Frigerio et al., 2006; Mervis et al., 2003). Even if their cause is still unclear, these behaviors lead to lack of social adjustment in comparison to controls with typical development (TD) (Gosh & Pankau, 1997), anxiety (Leyfer, Woodruff-Borden, Klein-Tasman, Fricke, & Mervis, 2006) and social exploitation (Udwin, Howlin, Davies, & Mannion, 1998). This evidence has led researchers to investigate the mechanisms that may lead to impairment in social cognition in this syndrome (Frigerio et al., 2006; Riby & Hancock, 2008) and to consider that mental states attribution might be an area of weakness for individuals with

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WS (Porter, Coltheart, & Langdon, 2008; Santos & Deruelle, 2009; Sullivan & Tager-Flusberg, 1999; Tager-Flusberg & Sullivan, 2000; Tager-Flusberg, Sullivan, & Boshart, 1997).

Mental states attribution (also termed mentalizing) indicates the ability to make inferences about others' mental states and has been described as involving several brain regions, which allow inferring the goals and intentions of others' overt actions (Frith & Frith, 2006). Mentalizing abilities in our everyday social life, appear to be mediated by various mechanisms. On one hand recent studies have shown that observing others actions elicits a matching activation in the observer's parieto-frontal motor system, allowing for immediate, non-inferential motor intention understanding (see Rizzolatti & Sinigaglia, 2010 for a review). On the other hand this matching motor-activation does not appear to account for more complex mentalizing abilities (Frith & Frith, 2003; Saxe, Carey, & Kanwisher, 2004). For example, when an observed action is unfamiliar or bizarre, other higher-level, top-down structures are involved, among which the rostral cingulate cortex and the region around the superior temporal sulcus (Brass, Schmitt, Spengler, & Geyerly, 2007; Liepelt, Von Cramon, & Brass, 2008). Over the last years, brain-imaging studies have shown how non-inferential motor knowledge and inferential mentalizing mechanisms, may play distinct but complementary roles in understanding goals and intentions of others' actions (de Lange, Spronk, Willems, Toni, & Bekkering, 2008; Hari & Kujala, 2009).

In WS, behavioral studies have highlighted an uneven performance in mental state attribution (Porter et al., 2008; Santos & Deruelle, 2009; Tager-Flusberg & Sullivan, 2000), but we know of no study specifically dedicated to understanding of *motor* intention. The present study aims to better our knowledge of this type of intention understanding in youths with WS relative to TD children, by explicitly analyzing the understanding of observed actions.

The term motor intention has been used by developmental psychologists to indicate understanding a *single motor act* as goal-directed as well as understanding a *series of motor acts* as determined by overarching goals (Gergerly & Csibra, 2003; Gerson & Woodward, 2010; Rochat, 2007; Sommerville, Hildebrand, & Crane, 2008; Sommerville & Woodward, 2005; Sommerville, Woodward, & Needham, 2005; Woodward & Sommerville, 2000; Woodward, Sommerville, Gerson, Henderson, & Buresh, 2009). However a conceptual distinction may be drawn between these two conditions. For example, imagine observing a person grasping a mug. We may perceive the goal-relatedness of each one of his motor acts (e.g. reaching for the mug, grasping it, picking it up, etc.). Furthermore, depending on how the mug is being grasped (e.g. shaping the hand as a hook around its handle or wrapping thumb and four fingers as a claw around its upper edge) and in which context it is being grasped (e.g. close to a teapot or close to a dish drainer), these motor acts may be understood as part of a motor act sequence with an overarching goal (e.g. drinking tea or putting the mug away). In the first case we have an immediate understanding of *what* the other is doing (i.e. reaching, grasping, picking up, etc.), while in the second case we grasp the motor intention underlying the observed motor act, i.e. the *why* of the other's actions (i.e. drinking, putting away, etc.).

Neurophysiological research on the functional properties of the parieto-frontal hand grasping/manipulations circuit sustained the importance of the *what/why* distinction in immediate, non-inferential, understanding of others' actions. In particular, research on the properties of mirror neurons in the parieto-frontal circuit of non-human primates, showed that these neurons encode the observed motor acts in relation to their goals, i.e. they encode *what* the others are doing (e.g. grasping, holding, or breaking) (Gallese, Fadiga, Fogassi, & Rizzolatti, 1996; Rizzolatti, Fadiga, Gallese, & Fogassi, 1996; Umiltà et al., 2008). Subsequent studies uncovered a chained organization in motor acts sequence both during action

execution and observation. Namely, the same goal-related motor acts (e.g. grasping) are encoded by different neurons depending on the over-arching goal of the action sequence in which they are embedded (e.g. grasping-to-eat or grasping-to-place). Thus, according to the context in which they are performed and other variables, the observation of an initial motor act of a sequence activates the whole motor sequence in the parieto-frontal areas of the observers, allowing them to understand, from the very beginning, the motor intention underlying the overarching action, i.e. *why* an action is performed (Fogassi et al., 2005; Iacoboni et al., 2005). Ensuing neuro-imaging and EEG studies have traced similar circuits in humans. These circuits become active at the mere sight of static stimuli showing specific hand-object interactions. Different parts of these circuits encode the *what* and the *why* of an observed action (Iacoboni et al., 2005; Johnson-Frey et al., 2003). As stated above these are not the only circuits involved in mentalizing abilities. For example a recent fMRI study by Spunt and colleagues has shown how other brain areas (i.e. the dorsomedial and the ventromedial pre-frontal cortex, the temporal poles bilaterally and the posterior cingulate cortex) may play a role in understanding the actions of others. These authors highlight how the mirror neuron system encodes immediate, motor aspects of others' intentional actions, while other mentalizing systems interpret these aspects in terms of unobservable mental states and traits (Spunt, Satpute, & Lieberman, 2010).

Following these developmental and neurophysiological studies a recent behavioral study (Boria et al., 2009) has evaluated *what* and *why* understanding in a group of high functioning children with autistic spectrum disorders (HFA) and matched TD controls. The task used static images showing different hand-object interactions to discriminate understanding the goal-relatedness of a single action (i.e. touching vs. grasping) and understanding the motor intention with which it was being performed (i.e. grasping to use vs. grasping to put away). Interestingly, results highlighted a specific impairment in *why* understanding as opposed to *what* understanding in children with HFA, which could be overcome through contextual cueing.

Summing up, increasing evidence from developmental psychology, neurophysiology and research on neuro-developmental disorders, has highlighted the relevance of distinguishing between understanding the *what* and the *why* of an observed act. However, so far no behavioral study has attempted to assess these basic abilities in WS. Therefore we attempted to gain better knowledge of motor acts and motor intention understanding in youths with WS relative to typically developing controls, by focusing on the following questions: (1) Are individuals with WS able to recognize different observed motor acts (i.e. touching or grasping)? (2) Are they able to interpret these acts in terms of overarching goals (i.e. grasping-to-use or grasping-to-place)?

To address these issues we studied the performance of a group of youths with WS and a TD control group, individually matched on mental age and lexical comprehension, on a task inspired by Boria et al. (2009). Further, to fully examine motor intention understanding in WS and to discriminate between impairment and mental-age appropriate performance we also included a group of TD youths individually matched for chronological age and gender.

2. Materials and methods

2.1. Participants

A total of 55 participants were recruited to the study: 19 youths with WS, 18 children with TD individually matched on mental age, lexical comprehension and gender (TDMA) and 18 youths with TD individually matched on chronological age and gender (TDCA). As 1 child with WS was excluded from the study for failing to comply with task requirements during training phase, the final sample comprised: 18 youths with WS (9 females and 9 males with mean chronological age 13;7, range 5;1 to 30;0 years, and mean mental age 6;5, range 4;1 to 11;2), 18 children with

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