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A R T I C L E   I N F O

Article history:
Received 26 February 2013
Received in revised form 11 May 2013
Accepted 13 May 2013
Available online 19 June 2013

Keywords:
Prader–Willi syndrome
Theory of Mind
Autism Spectrum Disorder
Maladaptive behavior
Social functioning

A B S T R A C T

In order to evaluate the social cognitive functioning in children with Prader–Willi syndrome (PWS), Theory of Mind (ToM) and symptoms of Autism Spectrum Disorder were evaluated. Sixty-six children with PWS aged 7–17 years were tested using the Theory of Mind test-R and the Diagnostic Interview for Social Communication disorders. We tested the correlation between Total ToM Standard Deviation Score (Total ToM SDS) and genetic subtype of paternal deletion or maternal uniparental disomy, and total IQ, verbal IQ and performal IQ. Prevalence and symptoms of Autism Spectrum Disorder were assessed. Median (interquartile range) of total ToM SDS of those aged 7–17 years was −3.84 (−5.73, −1.57). Their Total ToM SDS correlated with total IQ (β = 0.662, p < 0.001, adj.R² = 0.407), in particular with verbal IQ (β = 0.502, p = 0.001, adj.R² = 0.409), but not with performal IQ (β = 0.241, p > 0.05, adj.R² = 0.259). No difference in Total ToM SDS was found between children with deletion and maternal uniparental disomy (β = −0.143, p > 0.05, adj.R² = −0.016). Compared to the reference group of healthy children aged 7–12 years, children with PWS in the same age group had a median ToM developmental delay of 4 (3–5) years. One third of children with PWS scored positive for Autism Spectrum Disorder. Most prominent aberrations in Autism Spectrum Disorder were focused on maladaptive behavior. Our findings demonstrate a markedly reduced level of social cognitive functioning, which has consequences for the approach of children with PWS, i.e. adjustment to the child's level of social cognitive functioning.

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1. Introduction

Prader–Willi syndrome (PWS) is a neurogenetic developmental disorder caused by the absence of paternal expression of genes in chromosome 15 at the locus q11–q13, either by paternal deletion (DEL), maternal uniparental disomy (mUPD), imprinting defects or paternal chromosomal translocation (Cassidy, 1997). Its main characteristics are hypotonia, hypogonadism, short stature, obesity, intellectual disability and a specific behavioral phenotype.

Earlier studies described various behavioral characteristics, such as hyperphagia and food preoccupation, skin-picking, compulsive behavior, and psychotic disorders (Boer et al., 2002; Dimitropoulos, Feurer, Butler, & Thompson, 2001; Morgan et al., 2010; Vogels et al., 2004).

Differences in behavioral phenotype have been linked to the genetic subtype of individuals with PWS. Those with mUPD seem more prone to Autistic Spectrum Disorder (ASD) than individuals with DEL (Descheemaeker, Govers, Vermeulen, &
A recent study showed that impaired social functioning in subjects with mUPD was similar to that in subjects with Autism Spectrum Disorder (Dimitropoulos, Ho, & Feldman, 2012).

Children with ASD have an impaired Theory of Mind (ToM) development (Muris et al., 1999). The ToM describes the cognitive capacity to infer the mental states of oneself (Baron-Cohen, Leslie, & Frith, 1985) and others, and is an essential ability in social cognitive functioning and a core cognitive feature of ASD. The Theory of Mind has not been previously investigated in detail in children with PWS. One study tested three aspects of the ToM in a small group of young children with PWS, who were participating as one of the comparison groups in their major study of ToM in children with Williams syndrome (Tager-Flusberg & Sullivan, 2000). They found that children with PWS performed better in false belief, an element of the ToM, than children with Williams syndrome. Another study found that the level of social adjustment, using the Social Attribution Task, in adults with PWS were more poorly than in those with comparable intellectual disability but similar to those with a pervasive developmental disorder (Koenig, Klin, & Schultz, 2004).

In order to evaluate the social cognitive functioning in children with Prader–Willi syndrome, the Theory of Mind and Autism Spectrum Disorder and their correlation were evaluated.

We postulated that ToM development was delayed in most children with PWS, and expected that ToM scores would be less abnormal in children with DEL. We also hypothesized that children with DEL are less prone to ASD than children with mUPD. For that reason, we assessed the prevalence and symptoms of Autism Spectrum Disorder in these children.

2. Methods

For the present study, we invited participants aged 7–17 years who participate in the Dutch PWS Cohort Study, a study regarding the long-term effects of growth hormone treatment in children with PWS (de Lind van Wijngaarden et al., 2009). In the Dutch PWS Cohort Study, all participants were treated with Genotropin 1 mg/m²/day, cognitive functioning was measured biennially, and anthropometry was performed annually.

For the present study, a total of 76 children with PWS were eligible for participation, 66 of which agreed to participate, resulting in a response rate of 87%.

The genetic diagnosis of PWS was confirmed in all participants by methylation testing. The genetic subtype was known in all but 2 participants.

2.1. ToM

The Dutch ToM test-R (Steerneman, 2009) is a validated diagnostic instrument for healthy children between the age of 4–12 years old (Muris et al., 1999). It consists of 14 illustrated short stories, which are divided into 3 developmental stages. Stage 1 covers recognition of emotions and the difference of reality and surreality (e.g. “actually” cycling and “dreaming” about cycling). Stage 2 covers the first manifestations of “belief”: “first-order belief” and “false belief”. “First-order belief” is the ability to understand one’s own mental state as “I think”. “False belief” is the individual’s comprehension of another person’s mistaken belief; for example if a character has not been informed that an object has been moved to another place. An example for testing the “false belief” is the following situation: person 1 places an object in box 1 and leaves the scene. Person 2 transfers the object to box 2. When person 1 returns, the investigator asks the child where person 1 would look for the object. “False belief” is acquired if the child’s answer is box 1. If the child’s answer is box 2, it suggests that the child is not able to make a judgment about another person’s false expectation. Stage 3 consists of questions about the “second-order belief”, i.e. inference of someone’s belief about another’s belief (e.g. “John thinks that Mary thinks it is going to rain and therefore she is taking the umbrella when she is going outdoors…”).

The ToM-test-R includes 14 stories with a total of 33 questions and 3-pretense exercises. Scores are either 0 (failed) or 1 (passed), leading to a maximum of 36 points. A maximum of 12 points can be scored per stage. The ToM test-R has been validated for healthy children without intellectual disability in the age range of 4–12 years.

In order to search for a screening instrument for ASD in children with PWS, the ToM test-R was chosen to evaluate social cognitive functioning.

The ToM test-R was performed at the children’s home or residence and the scores were verified by another researcher, which resulted in similar scores.

2.2. DISCO

The Dutch translation of the Diagnostic Interview Social and Communication disorders (DISCO), 11th revision, was used to diagnose ASD. DISCO (van Berckelaer-Onnes, Noens, & Dijkxhoorn, 2008) is a standardized semi-structured, interviewer-based questionnaire for diagnosing ASD, and is based on the original validated DISCO of 2003 (Wing, Leekam, Libby, Gould, & Larcombe, 2002). It can accurately identify children with ASD, including children with intellectual disability (Maljaars, Noens, Scholte, & van Berckelaer-Onnes, 2011).

Only parents of children with PWS were interviewed for this study. DISCO was chosen because it diagnoses disorders within the broader “autism spectrum” (Wing et al., 2002). The diagnoses of the DISCO are based on DSM-IV-TR, the Diagnostic and Statistical Manual of Mental Disorders, fourth edition Text Revision (American Psychiatric Association, 2000).
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