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# Conference proceedings

# VISUAL-MOTOR INTEGRATION IN PRADER-WILLI SYNDROME: A LONGITUDINAL CASE STUDY

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## ABSTRACT

**Prader-Willi syndrome is a relatively rare, genetically determined complex neurodevelopmental disorder, which is caused by the absence of expression of one or more genes at the locus q11-q13 on chromosome 15. It is generally accepted that individuals with Prader-Willi syndrome (PWS) have a mild intellectual disability, and specific cognitive strengths and weaknesses. For example, they perform well on visuo-spatial tasks but they have poor motor skills due to hypotonia. The current study presents the longitudinal profile of visual-motor integration in a female with PWS. The Developmental Test of Visual-Motor Integration (VMI) has frequently been employed to evaluate visual-motor development in children and adolescents. The VMI test with its two supplementary Visual Perception and Motor Coordination tests was administered both to the female with PWS and to a group of 10 normally developing individuals matched for age and sex, by a certified neuropsychologist 3 times (at the ages of 8, 11 and 14 years). The results show that scores on the VMI test for the female with PWS increasingly diverge with age. Specifically, a marked downward shift was found on the Motor Coordination Test (total decrease 6.3% from 8 to 14 years old), while there was a marked upward shift on the Visual Perception Test (total increase 12.8%). It seems that cognitive strengths and weaknesses become more evident as individuals with Prader-Willi syndrome become older. This pronounced pattern across development can also be found in other genetic syndromes. The question of whether this is a systematic or a random effect requires further longitudinal studies.**

For years, researchers in mental retardation tended to group together individuals of various mental retardation etiologies who were similar in level of IQ. In light of previous research it has become evident that patterns of cognitive deficits and/or relative strengths differ by mental retardation etiology (1). The etiology-specific approach is useful not only for a basic understanding of mental retardation, but also for the development of educational interventions. Etiology-related phenotypes have increasingly been mainly documented in Down, Fragile X, Williams and Prader-Willi syndrome (PWS) (2).

PWS is a rare neurodevelopmental disorder affecting 1 in every 10,000 to 15,000 individuals (3). In most of the cases it is caused by the absence of the paternal genes at q11q13 on chromosome 15. This absence is produced by either a deletion at this region in the paternal chromosome or contribution of both chromosome 15s from the mother and none from the father (4). PWS is associated with developmental disabilities, hypotonia, hyperphagia, and characteristic physical and behavioral features (5). Behaviorally, many persons with Prader-Willi syndrome show persistent food seeking, due to hypothalamic abnormality, leading to life-threatening obesity (6). Other problems include temper-tantrums and obsessive-compulsive features such as skin-picking, hoarding, redoing things, and being overly concerned with symmetry and exactness (7).

Relative to maladaptive behavior, however, much less is known about the cognitive features associated with PWS (8). The average IQ typically is in the mild range of mental retardation (55-70). The distribution includes very few cases within the average range of intelligence or profound range of mental retardation (9). Cognitive strengths include long-term memory, visual-spatial perceptual abilities and simultaneous processing. Some individuals also show a great ability with jigsaw puzzles, which is included in the supportive findings for the diagnostic criteria (10). Academic strengths include reading decoding, acquired information and vocabulary (11). Cognitive weaknesses are seen in sequential processing deficits. Relative weaknesses may be apparent in short-term memory, including visual, motoric, and auditory short-term processing. Children with PWS score poorly on measures of motor impersistence and motor accuracy, mainly due to hypotonia (12). Math is often an academic weakness (13,14). Although findings suggest a profile, several warnings are in order. Not all persons with PWS show this profile, and studies are needed to identify the range of cognitive variability and possible sources of individual differences in cognitive patterns (14).

Strength in visual perception and weakness in fine motor skills in PWS can be assessed by standardized tests of Visual-Motor Integration (i.e., Developmental Test of Visual Motor Integration - VMI) (15). Tests of visual-motor integration are compound measures, calling upon visual-perceptual ability and fine motor coordination. Children with PWS show relative strengths on the VMI in that their scores were significantly higher than age- and IQ-matched peers with mixed mental retardation, but below those of age-matched normally developing children with average IQs (16). If a child with PWS performs poorly on the VMI, it could be because he or she has deficient visual perception and/or motor coordination abilities. This means that deficient visual-motor integration could be a function of suboptimal capacity in one or both of these component processes. According to the findings of specific motor weakness and specific visual-perceptual strength in PWS (9,12), it seems that deficient motor coordination may explain the low performance of children with PWS on tests of visual-motor integration, such as the VMI test.

Complicating this cognitive picture still further is the idea of changing patterns of strengths and weaknesses with development. As a general rule, etiology-related strengths and weaknesses become more evident as children become older. For example, strength in visual memory tasks, in children with Down syndrome, becomes more advanced, compared to auditory memory tasks, as they grow older (17). A similar picture can be drawn for children with Williams syndrome. In this case, strengths involve levels of receptive vocabulary (on the Peabody Picture Vocabulary Scale) and weak-

nesses involve drawing measures such as the VMI test (18). As in Down syndrome, older compared to younger children with Williams syndrome showed more pronounced patterns of cognitive-linguistic strengths and weaknesses.

## AIMS AND OBJECTIVES

The current study aimed at further investigating the longitudinal profile of visual-motor integration in a female with PWS, using the VMI test. In line with previous research, it was expected to demonstrate the classic profile of the syndrome reflecting a motor weakness with well-developed visual-perceptual skills. The crucial matter is if this finding will further indicate more pronounced cognitive strengths (visual-perceptual skills) and weaknesses (motor skills) as she becomes older. Thus, the objective of the current study was to accomplish a longitudinal description of cognitive strengths and weaknesses of the Prader-Willi syndrome from age 8 to age 14, which has not been previously explored.

## METHOD

## PARTICIPANTS

**Case report**

Irene is now a 15-year old female with PWS, who was diagnosed at 4 years of age when cytogenetic testing demonstrated the classical deletion at 15q. Her mother had a full-term normal pregnancy. Irene had severe hypotonia. Her developmental milestones were severely delayed. She sat at 11 months, while she walked independently at 2 years 4 months. She was clearly obese until age 6. Her structured weight program, starting at age 7, faced a lot of difficulties due to medical problems (pneumonia instances made her stay at the hospital for a long time) and due to the presence of two normally developing brothers who were staying at home making Irene's diet difficult to be set. At the age of 14 her weight was 89 kilograms, and her height 149 cm. She suffers from scoliosis and never received hormonal replacement. Irene's behavior problems include skin picking and some mild temper tantrums. She also shows some kind of obsessive behavior, that is a need for order, exactness. She graduated from a special school for exceptional children at age 13. She now attends a vocational training center. She has always demonstrated strengths in working puzzles, and she used to fill out a puzzle book 24 hours a day. Cognitive testing shows a stable mild level of mental retardation (IQs =67, 69, 69 at the ages of 8, 11 & 14 respectively), according to the Test of Non-Verbal Intelligence (19).

Normally developing control comparison group

Ten normally developing individuals, matched for age and sex with Irene, were included in the control group. They were randomly selected from mainstream primary schools. Criteria for inclusion consisted of absence of known neurological abnormality, psychotropic medication, learning difficulties or a history of special needs as specified by parents. None of the control participants were receiving any special education.

## DESIGN

The design is a single subject study with an age-matched control comparison group. A single subject study methodology is adopted as in developmental cognitive neuropsychology it is dissociations within and between individual participants which are of interest, since averaging across subjects may mask the most relevant characteristics of performance (20).

## INSTRUMENTS

Visual-motor functioning was assessed by the VMI test with its two supplemental (Visual Perception and Motor Coordination) (15). The VMI is a developmental sequence of geometric forms to be copied with paper and pencil. In the Visual Perception Test, one geometric form that is exactly the same as each stimulus is to be chosen from among others that are not exactly the same as the stimulus. The task is to identify the exact much for as many of the stimuli as quickly as possible. This is a pure visual perceptual task; the motor requirements are reduced to a minimum by having the child simply point to her/his choices. In the Motor Coordination test, the task is to simply trace the stimulus forms with a pencil without going outside double-lined paths. Although visual perception cannot be entirely eliminated in such motor tasks, visual perceptual demands have been reduced greatly by providing examples, starting dots, and paths as strong visual guides for the required motor performance.

## PROCEDURE

The VMI test was administered to Irene by a certified psychologist 3 times (at the ages of 8, 11 and 14). She was tested individually in a quiet environment. Girls from the control group were tested individually 3 times (at the ages of 8, 11 and 14) in a room set apart from the class during school hours. Rooms were well illuminated and participants were seated across the table in front of the experimenter. During testing, children were not provided with any feedback on their responses, unless it was required by the procedure of the standardized test. The standardized VMI test was administered according to the stated procedure.

## RESULTS

The results show that scores on the VMI and its two supplemental increasingly diverge with age for the female with PWS (see Figure 1). Specifically, a downward shift, according to standard scores, was found on the VMI test (total decrease 4.94% from the ages of 8 to 14 years, which is 1.61% decrease from the ages of 8 to 11 years and 3.27% decrease from the ages of 11 to 14 years). There was a marked upward shift on the Visual Perception Test (total increase 12.83%, which is 7.35% increase from the ages of 8 to 11 years and 5.48% increase from the ages of 11 to 14 years). A marked downward shift was found on the Motor Coordination Test (total decrease 6.31% from the ages of 8 to 14 years, which is 8.06% decrease from the ages of 8 to 11 years and a small increase 1.75% increase from the ages of 11 to 14 years). On the contrary, there weren't any discrepancies, favoring visual perception or motor coordination, for the control comparison group (Figures 2,3,4 for the ages 8, 11 & 14 respectively).

In particular, the paired samples student t-test revealed no statistically significant differences between the ages of 8 and 11 years for the VMI, the Visual Perception Test and the Motor Coordination Test ($t = .95$ $p < .05$, $t = .59$ $p < .05$, and $t = -1.36$, $p<.05$ respectively). Additionally, no significant differences were found between the ages of 8 and 14 years for the VMI, the Visual Perception Test and the Motor Coordination Test ($t = .09$ $p < .05$, $t = .68$ $p < .05$, and $t = -.50$ $p < .05$ respectively). Finally no significant differences were reported between the ages of 11 and 14 years for the VMI, the Visual Perception Test and the Motor Coordination Test ($t = -.6$ $p < .05$, $t = .50$ $p < .05$, and $t = 1.09$ $p < .05$ respectively).

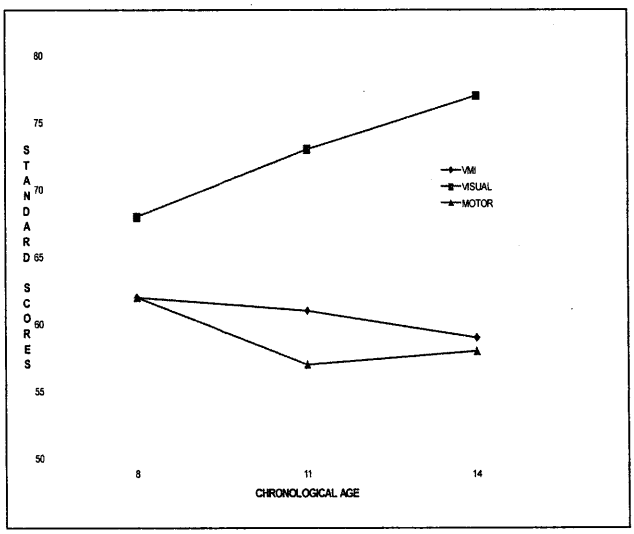


**FIGURE 1. VMI STANDARD SCORES OF PRADER-WILLI SYNDROME FEMALE AT THE AGES 8, 11 & 14 YEARS.**

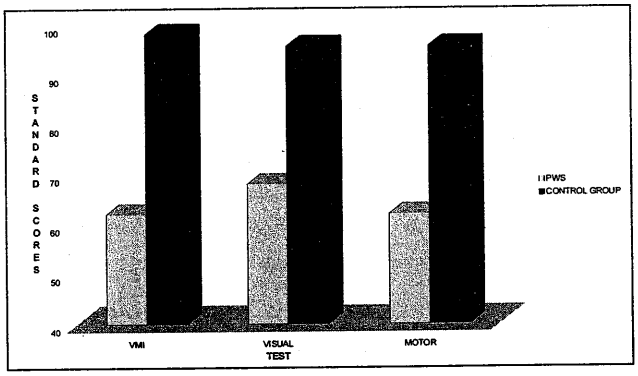


**FIGURE 2. STANDARDIZED SCORES OF THE FEMALE WITH PRADER-WILLI SYNDROME AND CONTROL GROUP ON THE VMI AT THE AGE 8 YEARS.**

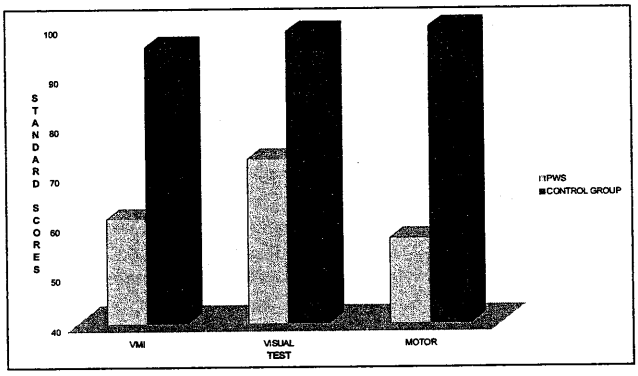


**FIGURE 3. STANDARDIZED SCORES OF THE FEMALE WITH PRADER-WILLI SYNDROME AND CONTROL GROUP ON THE VMI AT THE AGE 11 YEARS.**

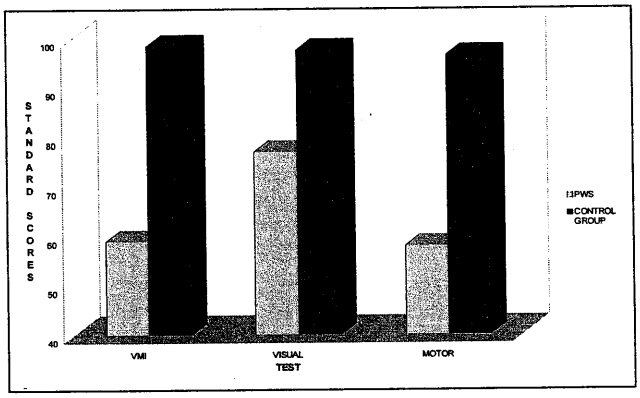


**FIGURE 4. STANDARDIZED SCORES OF THE FEMALE WITH PRADER-WILLI SYNDROME AND CONTROL GROUP ON THE VMI AT THE AGE 14 YEARS.**

Further, age equivalent scores on the VMI test increasingly diverge with age for the female with PWS (see Table 1). With increasing age her strength in the visual perception test becomes more advanced compared to the motor coordination test. At the age of 8 years, the difference was about 8 months, at the age of 11 years discrepancies reached 18 months. By age of 14 years, "visual over motor" age-equivalent scores were 32 months apart, favoring visual perception.

**TABLE 1. VMI RAW SCORE AGE EQUIVALENTS FOR THE FEMALE WITH PRADER-WILLI SYNDROME AT THE AGES OF 8, 11 & 14 YEARS.**

| | | |
|---|---|---|
| VMI (8 yrs) 4:6 | VMI (11yrs) 5:2 | VMI (14 yrs) 5:10 |
| VISUAL (8 yrs) 4:9 | VISUAL (11 yrs) 6:6 | VISUAL (14 yrs) 8:3 |
| MOTOR (8 yrs) 4:1 | MOTOR (11 yrs) 4:10 | MOTOR (14 yrs) 5:7 |

## DISCUSSION

These results clearly demonstrate the classic profile of the syndrome reflecting a motor weakness (Motor Coordination Test) with a visual-perceptual strength (Visual Perception Test) on the VMI. Perhaps the most intriguing finding is that the female with PWS shows more pronounced patterns of cognitive strengths and weaknesses, as she grows older, as in Down and Williams syndromes (17,18). The results of the current longitudinal case study confirm that visual-perceptual and motor coordination abilities develop at different rates. The present results have significant theoretical implications for basic research on visuo-motor behaviour of PWS, as well as important applied implications for neuropsychological assessment and educational interventions. The question of whether this is a systematic or a random effect requires further longitudinal investigation. One should, therefore, be wary of suggesting the existence of a clear dissociation between visual and motor abilities, although the current results do suggest that this dissociation exists and will become more apparent as development progresses. Information generated from this study could be utilized in an educational or remedial context to enable more theory-driven techniques. Additional research is also necessary to identify patterns of cognitive functioning across development in persons with other genetic syndromes.

Why do such gaps widen as the PWS individual gets older? One theory (namely, emergentism) holds that gaps widen because the child's learning is deeply and continuously embedded within the child's learning environment (21). Thus, while many children with PWS show a propensity or disposition to display specific strengths and weaknesses due to genetically mediated neurochemical and brain developmental abnormalities (22,23,24), the children's "learning environment" (i.e., parents, teachers. peers) may then reinforce such propensities (21, 25). Specifically, families may play to the child's strengths -encouraging what the child is able to do more successfully- while avoiding behaviors or interactions that focus on the child's weaker areas. In the same way, children themselves may gravitate toward performing behaviors reflecting relative strengths as opposed to relative weaknesses.

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