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Romanian Prader-Willi Association

ASOCIACIÓN MADRILEÑA  
PARA EL SÍNDROME DE  
PRADER-WILLI



## SOMATIC AND BEHAVIORAL DIFFERENCES IN CHILDREN WITH PRADER-WILLI SYNDROME

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**INTRODUCTION:** Phenotypic differences among patients with Prader-Willi syndrome are based on their genetic background. These differences may be somatic or behavioral and may have important implications for the disease's outcome.

**OBJECTIVE:** To compare clinical features and behavioural characteristics of two patients with Prader-Willi syndrome

**PATIENTS AND METHODS:** Two patients, an 8 year and 7 months old boy and a 16 year old girl at the age of the diagnosis, previously considered as having idiopathic encephalopathy, were clinically evaluated. Methods of evaluation consist of history, physical examination, anthropometric and metabolic assessment with plasma glucose, total cholesterol and triglyceride measurement by enzymatic methods, and blood gases measurement, ECG recording and psychological evaluation.

**RESULTS:** Diagnosis was based on the consensus clinical criteria with a weighted score of 14 for the male and 11 for the female patient. The common diagnostic features were: neonatal and infantile hypotonia, feeding problems and failure to thrive followed by excessive weight gain after three years of age, mental retardation, compulsive hyperphagia and characteristic facial features. Both patients had an increased body mass index (BMI) of 40.26 kg/m<sup>2</sup> and 42.12 kg/m<sup>2</sup>, the measure of severe progressive obesity. Short stature, small hands and feet, learning disability, behavioral problems as minor criteria were also present in both cases. Hypogonadism, longer period of infantile hypotonia and tube feeding, more severe speech articulation defects, more complex obsessive - compulsive disorders with skin picking and severe obesity were present in the male patient. The family had no control on his food intake. Obstructive sleep apnea and hypoventilation syndrome, achanthosis nigricans and other components of the metabolic syndrome were associated at the age of ten in this patient with a lethal outcome. Oligomenorrhea and dyslipidemia were present and short stature and acromicria were more evident in the girl patient, with comparable BMI but at a different age and with better learning performance.

**CONCLUSIONS:** In both our patients' diagnosis was based on consensus clinical criteria. Severe obesity in the younger child was associated with increased number of life - threatening comorbidities. Some characteristic somatic features of Prader-Willi syndrome became evident with age. The difference between the symptoms' severity in the two cases may be genetic and even environmentally determined.