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ASOCIACIÓN MADRILEÑA
PARA EL SÍNDROME DE
PRADER-WILLI



IMPORTANCE OF THE MINOR CRITERIA FOR A POSITIVE CLINICAL DIAGNOSIS OF PRADER WILLI SYNDROME PATIENTS

C. Skrypnik¹, M. Bembea¹, V. Belengeanu², E. Tomescu³, P. Grigorescu Sido⁴, I. Pascanu⁵, M. Covic⁶.

¹ University of Oradea, Faculty of Medicine, Genetics Department, Oradea, Romania.

² University of Medicine and Pharmacy, Genetics Unit, Timisoara, Romania. ³ Institute for Mother and Child Care, Genetics Unit, Bucharest, Romania. ⁴ University of Medicine and Pharmacy, Pediatric Unit I, Cluj Napoca, Romania. ⁵ University of Medicine and Pharmacy, genetics Unit, Targu Mures, Romania. ⁶ University of Medicine and Pharmacy, Genetics Unit, Iasi, Romania.

INTRODUCTION: Prader-Willi syndrome (PWS), a genetic disorder that involves chromosome 15, is the most common form of obesity caused by a genetic syndrome. Diagnosis often is delayed until early childhood because the clinical findings are relatively nonspecific, particularly in infancy, and the dysmorphism often is subtle.

MATERIALS AND METHODS: We evaluated 16 patients with age between 8 month and 27 years old, clinically diagnosed with PWS following the consensus major and minor criteria. All patients had a positive clinical score for PWS, having dysmorphic features, obesity associated with childhood hyperphagia, hypogonadism and mental retardation.

RESULTS: All patients had unrelated healthy parents and resulted from a physiological pregnancy, with normal birth weight and normal Apgar score. The medium age of diagnosis was 8 years old, based on the suggestive phenotype. The main reasons for a genetic evaluation were neonatal hypotonia, obesity and mental retardation. The minor criteria had a variable frequency. Decreased fetal movement and infantile lethargy were found in only 4/16 (25%) and 3/16 (18.75%) of patients, respectively. Small hands and feet characterized all patients. Short stature (compared with family members) and hair hypopigmentation had a low frequency being found in 3/16 (18.75%) cases. 9/16 (56.25%) of patients had high pain threshold and 40% of patients had skin picking and sleep disturbance. Sleep apnea characterized 7/16 (43.75%) of patients and was correlated with the weight gain. A distinctive behavioral phenotype were found in 11/16 (68.75 %) of cases.

DISCUSSION: Sensitivities of the minor criteria ranges from 18.75% (infantile lethargy) to 100% (small hands and feet). The minor criteria can sometimes reveal the diagnosis before a major sign, and their association with the major criteria, allow a positive diagnosis of PWS. The minor criteria can be important to avoid over diagnosis and to make a correct selection for the molecular diagnostic tests.