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ASOCIACIÓN MADRILEÑA
PARA EL SÍNDROME DE
PRADER-WILLI



EFFECTS ON SCOLIOSIS IN FOUR PATIENTS WITH PRADER-WILLI SYNDROME TREATED WITH GROWTH HORMONE FOR FIVE YEARS

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INTRODUCTION: Prader-Willi Syndrome (PWS) is a genetic disease with an average incidence rate of 1/25.000, which is due to a common genetic defect that abolishes the expression of imprinted paternal genes in the 15q11-q13 chromosomal region. It is characterized by severe hypotonia, obesity, short stature, small hands and feet, hypogonadism, and feeding disorders.

The objective of our study is to examine orthopedic aspects of PWS, as well as to evaluate how growth hormone (GH) intake might affect the trend of scoliotic bend.

METHODS: We examined 14 PWS patients (9 females and 5 males), aged 2 to 23 years, diagnosed with a methylation test and FISH. All patients received the Adam's Forward Bend Test. The amplitude of the scoliosis curvature was evaluated using the Cobb's method and MEHTA index, and the degree of skeletal maturity using Risser's index.

RESULTS: Only 71.43% (namely, 10) out of the above-mentioned sample presented with primary idiopathic scoliosis and no other orthopaedic complication. Over a period of 5 years, the characteristics of scoliosis in GH off-treatment PWS patients remained stable, as evaluated with Cobbs method and MEHTA index.

DISCUSSION: The scoliosis in PWS patients overlaps with the clinical features encountered in idiopathic scoliosis. It is present from an early age and remains stable during childhood, but progresses in 15 to 20% of cases during adolescence. GH treatment in PWS normalizes height and increases lean body mass, which is beneficial to weight management. In our sample there was no significant difference in scoliosis progression and spine curve measurements with Cobbs angle method and MEHTA index during the GH treatment.