Abstract

High prevalence of scoliosis in a large cohort of patients with Prader- Willi syndrome

Introduction

Data on the prevalence of scoliosis in Prader Willi syndrome are not unequivocal, ranging from 15% to 86%, but the role of growth hormone therapy (GHT) in the onset and progression of scoliosis remains controversial. in our study we investigated the prevalence and severity of scoliosis in a large group of patients with s. by Prader Willi, with and without hormone therapy (GH), evaluating the correlations with age, sex, body mass index (BMI) and genotype.

Methods

A cross-sectional study was conducted in 180 patients with genetically defined Prader Willi syndrome (99 deletion, 78 UPD, 3 not known), 96 females and 84 males, aged 17.6 + -12 years, (range 1, 3-49.7; median 15.5 years), BMI 29 + -011.2 (99 subjects (55%) with BMI> 2SDS, were considered obese, 148 subjects undergoing hormone therapy (prior to study and at time of recruitment) while 32 patients were never treated. Apart from anthropometric data, we performed an x-ray of the spine. scoliosis assessment, including COBB angle (CA) measurements, was performed by the spine surgeon himself vertebral. Scoliosis was defined as the curve with CA> 10 °. In addition, scoliosis was classified using the Scoliosis Research Society (2006) classification, mild (CA 10-20 °), moderate (CA 20-40 °) and severe scoliosis (CA> 40 °).

Results

148 subjects had scoliosis (82.2%), 80 females (54%) had scoliosis (mild n51, moderate n47, severe n50), predominantly with thoracolumbar curve, rarely with a double curve. The mean age at diagnosis of scoliosis was 7.1+ - 6.3 years (range 0.6-30.4 years). A brace was prescribed to 77 subjects aged 7.6 + - 3.9 years. And it was worn for 4.8 +/- 3.3 years. 26 subjects (14.4%) had severe scoliosis with a high risk of developing the curve and underwent vertebral surgery at the age of 12.8+ - 4.7 years (range 4-27 years). The mean age of initiation of hormone therapy was 4.7 +/- 5.6 years (0.1-32 years). Hormonal therapy was administered both to subjects with scoliosis (122/148 = 82.4%) and to patients without scoliosis (25/32 = 78.1. No correlation was found between scoliosis and genotype (deletion 83.8% 9 , UDP 82%) as well as with respect to gender (females 83.3%, males 80.9%). Scoliosis was present in 86% of patients over 18 years and in 79.2% of children and adolescents (P <0.05). 99 patients were obese and scoliosis was observed in 85% of cases, while the prevalence of non-obese was 79%.

Conclusion

In a large cohort of patients with Prader Willi, scoliosis affects many patients (148/180 = 82.2%) with a high prevalence of moderate-severe forms (68.2%). In this study 69.6% of the subjects underwent conservative or surgical treatment. Aside from age, our data indicate that scoliosis is intrinsic to the syndrome, regardless of sex, BMI, gender, and hormone therapy. Therefore we suggest that all subjects with Prader Willi do a regular radiographic examination of the spine, starting from the first years of life, especially when the clinical examination of the spine is difficult to perform due to the underlying obesity.

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The characteristics of scoliosis were investigated in a large cohort of children and adults with Prader-Willi syndrome (PWS), analysing the role of age, gender, puberty, body mass index (BMI), genotype and growth hormone therapy (GHT) on its onset and severity. A retrospective cross-sectional study was performed in 180 patients with genetically confirmed PWS (96 females), aged 17.6 ± 12 years. Eighty-five subjects (47%) were obese. One hundred and fifty subjects (83.3%) were on GHT, while 30 patients had never been treated. Overall, 150 subjects (83.3%) were affected by scoliosis, 80.2% of children and adolescents and 87.8% of adults. A mild degree of scoliosis was observed in 58 patients (38.7%), moderate in 43 (28.7%) and severe in 49 (32.6%). Median age at diagnosis of scoliosis was 6.3 years, while the severe forms were diagnosed earlier (median age: 3.8 years). The cumulative probability at 5 years of age was equal to 0.403 and almost doubled at 15 years. No significant associations were found between scoliosis and genotype, gender, pubertal stage, GHT and BMI. A corset was prescribed to 75 subjects (50%) at a median age of 7.5 years, while 26 subjects (17.3%) underwent surgery at a median age of 13.1 years. Our data indicate that scoliosis is one of the major concerns for PWS patients that increases with age, and therefore suggest the need for regular systematic monitoring of spinal deformity from paediatric age.