Prader- Willi Syndrome: A general picture of 51 cases

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INTRODUCTION

Prader Willi Syndrome (PWS) is the most common genetic cause of obesity. It is characterized genetically by the absence of paternally expressed genes from the chromosome 15, either by paternal deletion, maternal uniparental disomy or microdeletion or epimutations of the imprinting center in the 15q11-q13 and clinically presents with hypotonia, short stature, satiety disorder and decreased basal metabolism.

OBJECTIVE

The aim of this study is to describe the morphological characteristics of patients with PWS who have been followed in a Pediatric Endocrinology Outpatient Clinic.

METHOD

We performed a retrospective study on 51 patients evaluating the age of diagnosis, genetic mutation, use of growth hormone (rhGH), age of beginning of follow-up, and Z-score of weight, height and body mass index (BMI). Data on their first and latest visit to our clinic were compared.

RESULTS

The mean age of diagnosis was 3.43 (±3.28) years old. The mean age of their first appointment was 4.95 (±4.26) years old and the average time of follow up was 6.45 (±5.24) years.

Table 1. Patients Characteristics

24 Males	**27** Females		
23 Deletion	**20** Uniparental Disomy	**6** Methylation*	**2** Imprinting Defects
18 with rhGH	**18** without rhGH	**15** With irregular use of rhGH	

*Conducted initial testing of methylation without mutation type specification

Table2. Comparison between initial and final evaluations

	Initial	Final
Median Age (Range)	3.3 years (0.1-16.8years)	10.8years (1.9-28.3years)
Stature	Z -1.34 (±1.4SD)	Z -1.41 (±1.52SD)
Z BMI	Z +2.26 (±2.61SD)	Z +2.97 (±1.58SD)
Mean follow up time: 6.45(± 5.24)years		

CONCLUSION

Despite the early diagnosis of PWS, it is noteworthy the delay between the diagnosis and the start of follow-up, postponing the measures to minimize the weight gain. An adequate coping since the time of diagnosis could introduce the basic concept of the disease in order to avoid obesity and raise adherence to accomplish diet restriction and effective rhGH treatment.

PWS is a rare disease that needs specialized attention and a multidisciplinary team struggling to minimize the deleterious effects of obesity, which is the cause of bad quality of life and early death in these patients.

ESPE 2015 836--P3

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DOI: 10.3252/pso.eu.54espe.2015

Poster presented at: 54th Annual Meeting 1-3 October ESPE 2015 BARCELONA European Society for Paediatric Endocrinology