#30 Clinical profile in adolescents and adults with Prader-Willi Syndrome

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Introduction

Prader-Willi Syndrome (PWS) is a rare neurodevelopmental disorder caused by the lack of expression of a 15q11q13 gene. Previous descriptions include infantile hypotonia, poor suction, alterations in development, short stature, hypogonadism and hypogentaism, hyperphagia and excessive weight gain, cognitive and behavioral problems including tantrums, skin-picking among others (Butler, Manzardo, & Forster, 2016; Goldstone, Holland, Hauffa, Hokken-Koelega, Tauber, & Speakers, 2008).

Although clinical manifestations of this syndrome have been studied, there are no such descriptions in the Argentine population, and neither in patients undergoing transdisciplinary treatment.

Aim

This study aim to describe the clinical profile of a sample of adolescents and adults with PWS, comparing a group of patients that attends to transdisciplinary treatment at Fundación SPINE versus a control group.

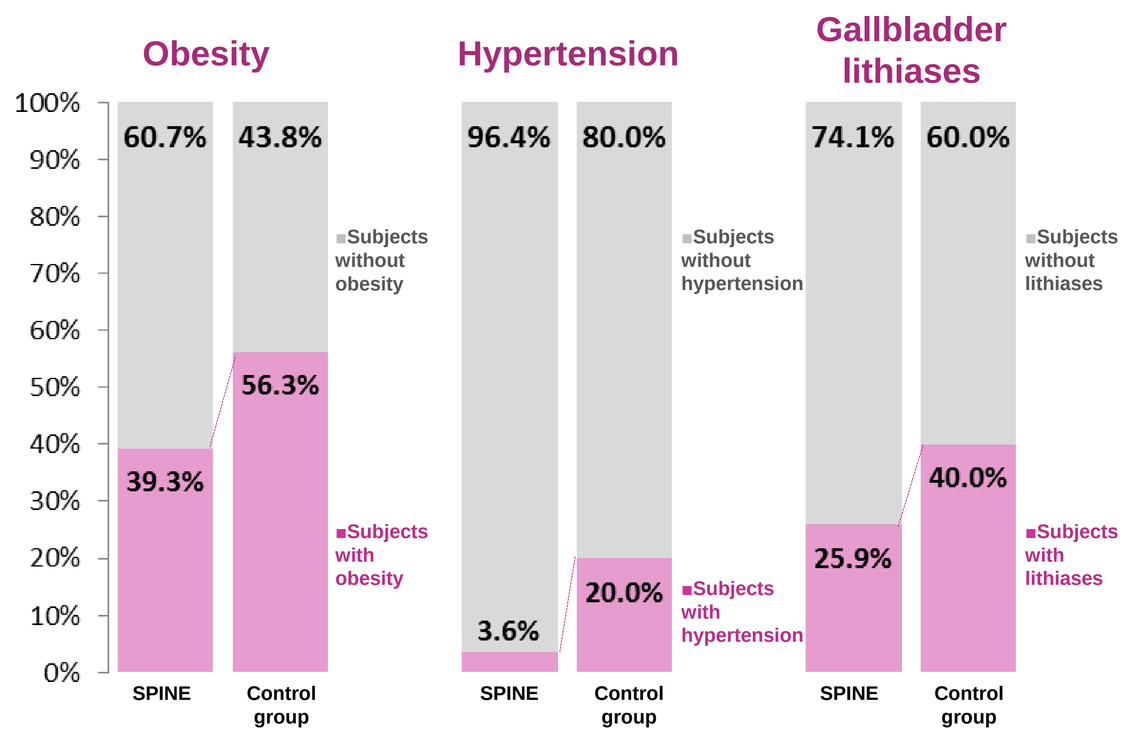
Methods

This study has a descriptive, cross-sectional, non-experimental design. The sample consist on 44 adolescents and adults with PWS, without growth hormone therapy, 63,6% are patients and receive transdiciplinary treatment at Fundación SPINE, and 36,4% where a control group. The following studies where analyzed: Ambulatory Monitoring Blood Pressure, Holter Cardiac, Polysomnogram (PSG), Hepato Biliary Pancreatic Ultrasound (BPH) Total Body Bone Densitometry (BMD).

Results

It was found that 56,3% from control group has obesity, versus 39,3% from SPINE patients group. Regarding blood pressure, 20% from control group has hypertension, versus only 3.6% from SPINE group. Regarding heart rate, 25% from control group has bradycardia, versus 21.4% from SPINE group. Abdominal ultrasound studies found that 40% from control group has gallbladder lithiasis, versus only 25.9% from SPINE group.

Regarding bone densitometry, 2 cases of the control group have made the study, 50% of them (n = 1) presents some type of complication (osteopenia). Of the 19 cases of the SPINE group, 63.2% presented some type of complication (47.4% osteopenia and 15.8% osteoporosis) while the remaining 36.8% obtained a score within a normal range. With respect to the rate of apnea events per hour, results are similar between groups, 33.3% from control group and 35.3% from SPINE group has altered index.



Conclusions

This study analyzed the prevalence of obesity, hypertension, gallbladder lithiasis, sinus bradycardia, osteopenia and apneas and hypoapneas in adolescents and adults with PWS.

Due to the few cases studied in the control group, it is not possible to make a conclusion about bone densitometry, it will be necessary to continue studying this aspect. Differences in the presence of obesity, hypertension and gallbladder lithiases were found between the control group and the SPINE group. This would indicate the possibility of making improvements in the clinical profile of patients with PWS under trans-disciplinary treatment. The absence of differences between both groups in relation to sinus bradycardia suggests that it could be a characteristic of the syndrome.

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Tratamiento e Investigación de Enfermedades Poco Frecuentes

References:

- Goldstone, A. P., Holland, A. J., Hauffa, B. P., Hokken-Koelega, A. C., Tauber, M., & Speakers Contributors at the Second Expert Meeting of the Comprehensive Care of Patients With PWS. (2008). Recommendations for the diagnosis and management of Prader-Willi syndrome. The *Journal of Clinical Endocrinology & Metabolism, 93*(11), 4183-4197.
- G Butler, M., M Manzardo, A., & L Forster, J. (2016). Prader-Willi syndrome: clinical genetics and diagnostic aspects with treatment approaches. *Current pediatric reviews, 12*(2), 136-166. .