Obesity in a Child with Short Stature and Development Delay: What Diagnosis?

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According to the WHO, the international epidemic of obesity is considered the most significant contributor to ill health. More than 30% of adults are obese, i.e. body mass index (BMI) > 30 kg/m², and one in three children born in the early 21st century will develop diabetes with a consequent reduction in lifetime expectancy.

Around 95% of the patients have nutritional obesity and in this case, the stature is increased. When we have an obese child with short stature, associated with development delay, we need to exclude - Prader-Willi syndrome.

Prader-Willi syndrome (PWS), was first described in 1956 by Prader et al. This syndrome is caused by deletion of chromosome 15 (15q11-q13) from paternal origin in approximately 70% of patients. Another 30% are from maternal uniparental disomy and 1-3% from defects in the imprinting center. In less than 0.1 %, is caused by balanced translocation.

There is a correlation between phenotype and genotype. Children with uniparental disomy showed less physical symptoms and behavioral problems. They also are more intelligent than children with deletion. The neonatal period is characterized by hypotonia, eating difficulties and poor weight development that may persist during the first year of life. Later on, affected children initiate hyperphagia behaviors that, together with delayed growth, lead to the progressive development of obesity. The early diagnosis of this clinical entity is important not only to prevent parents from the challenges inherent in this syndrome but also to guide an early intervention plan.

When we suspect of PWS, the first study is analyze the methylation which detected the abnormal imprinting of the parents in the cryptic region of 15q11.2 -13 chromosome, using southern blot or PCR.

Short stature and progressive obesity, associated with delayed psychomotor development, are characteristic of children with PWS. These features are more evident from the age of 4, which sometimes makes early diagnosis more difficult. The growth chart showed at the beginning, length and weight in the 3rd percentile. Lately, the weight will start to increase around the age of one year and at 2 years old, will be more than 97 percentile. On the other hand, the length will keep on the 3rd percentile. The prevention of obesity and its complications, such as diabetes, hypertension and respiratory problems, is a priority in the follow-up of these patients. Several studies have shown deficiency of growth hormone as well as the benefits of its replacement not only in the final stature but also in the body mass of the patient, decreasing complications and improving physical performance. It is contraindicated in children with a history of sleep apnea and polysomnography should be performed prior to any treatment, since elevation of IGF-1 levels cause hypertrophy of the adenoids, increasing the risk of sudden death.

Consensus guidelines suggest a starting dose of 0.5 mg/m²/day of growth hormone for infants and children with subsequent adjustments up to about 1 mg/m²/day as needed to achieve a target IGF-1 level in the upper part of the normal range for age (+1 to + 2 sd for age).

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