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Editorial

Prader-Willi Syndrome: Diagnosis and Treatment

Jorge Sales Marques*

Pediatric Department, Centro Hospitalar Conde S. Januário, Macau, China

***Corresponding Author**: Jorge Sales Marques, Pediatric Department, Centro Hospitalar Conde S. Januário, Macau, China.

Received: November 01, 2016; **Published:** November 02, 2016

Prader-Willi syndrome (PWS) is the most common syndrome form of obesity. The syndrome is caused by lack of expression of the paternally active genes on the long arm of chromosome 15. The vast majority of cases occur sporadically. The ratio between male and female gender is the same. The prevalence rate is 1:10000 - 1:30000. About 70% of cases the origin is paternal deletion 15q11.2 - 13, 20 - 30% from maternal uniparental disomy and 1 - 3 % from defects in the imprinting center. In < 0.1 %, it 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 risk of recurrence is less than 0.1%.

Children with one and six years of age showed symptoms of hyperphagia with progressive development of obesity. Most patients with PWS have growth hormone deficiency. Other complications of obesity are hypogonadism and behavioral problems. For children three years of age or younger, the diagnosis of PWS is highly likely if five points are scored from among these criteria (four from major criteria). In children, older than three years of age and adults, eight points are required (five or more from among the major criteria). Major criteria (one point each): neonatal and infantile hypotonia, feeding problems during infancy, excessive weight gain after infancy, characteristic facial features, hypogonadism, global developmental delay or mild to moderate intellectual disability, and hyperphagia. Minor criteria (one half point each): decreased fetal movement, characteristic behavior problems (usually multiple), sleep disturbance or sleep apnea, short stature, hypopigmentation, small hands and/or feet, narrow hands with straight ulnar border, eye abnormalities (esotropia, myopia), thick viscous saliva with crusting at corners of mouth, speech articulation defects, and skin picking.

The first study is to 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. If the methylation is abnormal, other studies are done for the determination of the type of mutation: Deletion (75%) – FISH or microarray. Uniparental disomy (20 a 30%)– analyze of DNA polymorphism. Imprinting defect (2%) – if uniparental disomy not found, the suspect will be the mutation or deletion of the center of imprinting.

Linear growth, body composition, including fat-free mass and bone density abnormalities, appears to improve with GH treatment, only indicated in children older than the age of two years. Is contraindicated by the presence of severe obesity (eg, weight > 225 percent of ideal body weight), uncontrolled diabetes, respiratory compromise, severe sleep apnea or acute respiratory infection. Consensus guidelines suggest a starting dose of 0.5 mg/m2/day for infants and children with subsequent adjustments up to about 1 mg/m2/day as needed to achieve a target IGF-I level in the upper part of the normal range for age (+1 to +2 SD for age. Infants may require doses as high as 1.5 mg/m2/day. GH therapy may worsen obstructive apnea by stimulating adenotonsillar hypertrophy via IGF-1 signaling. GH treatment should not proceed until the sleep disordered breathing is effectively treated by weight loss in those with severe obesity and/or by adenotonsillectomy or other surgical intervention to treat the airway obstruction. Those with significant abnormalities on polysomnogram should have follow-up studies approximately one month after beginning GH treatment.

Volume 2 Issue 4 November 2016