Special Issue

Endocrinology and Metabolic Diseases: Prader-Willi Syndrome

Message from the Guest Editor

Prader-Willi syndrome (PWS) is the most common syndromic form of obesity, occurring in approximately one in 10,000–30,000 live births, without sex differences in prevalence. PWS results from the loss of expression of paternal alleles in the PWS region of chromosome 15. It is most commonly caused by a paternal deletion (65-75%) or a maternal uniparental disomy 15 (mUPD, 20-30%). The syndrome is characterized by hypotonia, behavioural challenges, typical dysmorphic features and hypothalamic dysfunction, resulting in hyperphagia and morbid obesity with its comorbidities, pituitary hormone deficiencies (growth hormone, with sexual hormones as the most common), abnormal temperature regulation, autonomous nervous system alterations and high pain threshold. Other characteristics that may require surgery are criptorchydia, strabismus and scoliosis..... Thus, all original articles or systematic reviews on topics related to PWS health and wellbeing are welcome in this Special Issue.

Guest Editor

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