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Prader Willi Syndrome: Case Report

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Introduction

Prader Willi Syndrome (PWS) is a common and complex disorder affecting multiple systems. Obesity typically begins after 1-2 years of age and is later exacerbated by hyperphagia with lack of satiety

Case Report

We present the case of a girl of 8.5 years old who was referred to the pediatric endocrinology clinic for exploration and management of morbid obesity associated with mental retardation and hyperphagia. In the perinatal period she had features of hypotonia with poor feeding and development in infancy associated with psychomotor retardation. On clinical examination her BMI was 37.3kg/m2 (morbid obesity) and her height: 131cm (-0.5SD). The patient has dysmorphic features with a narrow bifrontal diameter, almond-shaped palpebral fissures, narrow nasal bridge, and thin upper lip with downturned mouth. Genetic testing revealed a microdeletion in the region of 15q 11-13. Laboratory evaluation demonstrated low FT4, TSH, cortisol and ACTH levels, impaired fasting glucose and impaired glucose tolerance with an HbA1c of 6.6%. Growth hormone dynamic testing showed a partial growth hormone deficiency. Levothyroxine therapy was initiated alongside with nutritional counselling for good weight management that induced a modest improvement in weight status.

Conclusion

Much is yet to be learned about this complex disorder. If obesity could be more easily avoided, the major cause of morbidity and mortality would be eliminated and significantly improved health and quality of life could be expected.