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Case Report

Young Boy's Battle with Childhood Obesity: Unmasking A Rare Case of Prader-Willi-Syndrome

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Abstract:

Prader-Willi syndrome (PWS) is one of the most prevalent causes of syndromic obesity. This genetic disorder begins with hypotonia and poor feeding in the neonatal period, later evolving into hyperphagia and rapid weight gain during early childhood, frequently leading to severe obesity in adolescence and adulthood. We report a case of a 14-year-old boy presenting with morbid obesity (BMI: 47.4) and related complications. Before admission, he was not properly evaluated for obesity. Along with obesity, the presence of hypogonadism, undescended testes since childhood, learning difficulties, poor academic performance, and behavioral abnormalities, including temper tantrums and emotional outbursts, raised a strong suspicion of Prader–Willi syndrome (PWS). We used the consensus criteria for the diagnosis of PWS clinically and focused on managing obesity with a dual GIP/GLP-1 receptor agonist; the patient achieved a remarkable weight loss of 25 kg over one year and marked reversal of obesity–related complications, significantly improving the patient's overall health and quality of life.

Keywords: *Obesity, Obesity related complications, Hyperphagia, Prader-Willi syndrome.*

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Introduction

Prader–Willi syndrome (PWS) is generally a sporadic disorder caused by lack of expression of paternally inherited imprinted genes in the chromosome 15q11–q13 region, typically due to a paternal deletion(65-70%), maternal uniparental disomy (25-30%) and there are also some rare causes like imprinting defects and translocations accounting for (~1%) ; this disorder is characterized by a recognizable pattern of dysmorphic features along with major neurologic, cognitive, endocrine, and behavioral/psychiatric disturbance [1].

PWS demonstrates a well-defined clinical course, though its manifestation may vary according to developmental phase, beginning with hypotonia and poor feeding in the neonatal period and later evolving into hyperphagia and rapid weight gain during early childhood, frequently leading to severe obesity in the absence of strict environmental control [2.] Despite these features, many affected individuals are not diagnosed until adulthood, as early signs may be subtle or missed, and later presentations—such as behavioral

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problems, metabolic derangements, or musculoskeletal deterioration—are often nonspecific and resemble other medical conditions, thereby delaying accurate diagnosis.[3]

Cognitive deficits are present in nearly all cases and usually involve mild to moderate intellectual disability, delayed speech and language development, and impairments in executive functioning and social behavior. Moreover, difficulties with behavior regulation—such as compulsive behaviors, emotional lability, repetitive skin picking, and inflexible thinking—are frequently observed and present significant challenges to long-term care and management [4,].

Renal involvement due to obesity is an increasing condition, parallel to the current obesity epidemic irrespective of the causes. Such individuals may develop a pathological syndrome known as obesity-related glomerulopathy (ORG), which is a consequence of hyperfiltration. Without therapeutic interventions, the usual clinical course of ORG is characterized by stable or slowly progressive proteinuria.[5,6]

Case Summary:

A 14-year-old boy, first child of non-consanguineous parents, was admitted to Dhaka Medical College Hospital (DMCH), Dhaka, Bangladesh, on 21st February, 2025 through emergency with the complaints of respiratory distress & daytime somnolence for one year along with progressively increasing body weight since early childhood. He also noticed pedal edema, gradual abdominal swelling, and passage of frothy urine for the last 3 months. According to his parents, he had feeding difficulties during infancy due to a poor sucking reflex. He was born at term with a birth weight of 3 kg & his growth was appropriate until the age of 3 years; thereafter, he developed rapid and excessive weight gain. After 4 years of age, the rate of weight gain increased markedly with progressive worsening of obesity. By the age of 8 years, his weight had reached approximately 75 kg (BMI: 34.7). His medical history was notable for hyperphagia, with a marked preference for high-calorie fatty foods. On query, his family members reported behavioral problems in the form of irrational rage and emotional outbursts with minor incidents, especially when the meal was restricted.

With all these complaints, he consulted several physicians since his early childhood & was advised for lifestyle modifications; symptomatic management was given, but all were unsuccessful. He also had delayed developmental milestones, learning difficulties, and late school enrollment at the age of 9 years. Currently, he is pursuing his education at a madrasa, but he has been skipping lessons due to illness, and his academic record has been poor. His past medical history was significant for undescended testes, for which he underwent orchidopexy at the age of 3 years. There was no other history suggestive of hypothyroidism, Cushing's syndrome, or medication-induced weight gain. There was no family history of obesity, diabetes mellitus, hypertension, or other significant chronic diseases

On examination, he was short-statured and morbidly obese with a body weight of 95.6 kg and a height of 1.42 m, BMI 47.4 kg/m2, which is class III obesity. Regarding vital signs: blood pressure was 130/85 mmHg, pulse was 80 beats/min, temperature was normal, but respiratory rate was 10-12 breaths/ min. He had small hands and feet and characteristic facial features, including almond-shaped eyes, a narrow nasal bridge, a thin upper lip, and downturned corners of the mouth. Features of hypogonadism were present, including micropenis and absent testes, with a lack of pubic and axillary hair. Skin examination revealed acanthosis nigricans and multiple striae. Neurological examination showed generalized hypotonia without focal neurological deficits. Pitting edema was present bilaterally, and a dipstick urine test was positive for proteinuria. Other systemic examinations were unremarkable.

We performed some laboratory tests for this patient. Hematological evaluation revealed a hemoglobin level of 11.3 g/dL and normal WBC and platelet count. Liver function tests showed: serum albumin was low at 2.3 mg/dl, with normal SGPT and PT. Urinalysis showed ++ proteinuria without hematuria and a normal serum creatinine level. We also performed polysomnography, and the impression was: no obstructive sleep apnea (OSA); apneas and hypoapneas index was 4.4, but frequent snoring was present, and hypoxemia was found during sleep time, making the diagnosis of obesity-induced hypoventilation. Glycemic assessment showed impaired glucose tolerance (IGT) on oral glucose tolerance testing, with a mildly elevated HbA1C level of 6.8%. The lipid profile demonstrated low high-density lipoprotein (HDL) cholesterol and raised triglyceride levels, with other parameters within acceptable limits. Hormonal analysis showed significantly reduced serum testosterone levels, along with suppressed luteinizing hormone (LH) and follicle-stimulating hormone (FSH), consistent with hypogonadotropic hypogonadism. Thyroid function tests (TSH, free T3, and free T4) and morning serum cortisol levels were within normal limits. Ultrasonography of the abdomen and pelvis demonstrated a normally sized kidney with absence of testicular tissue in the scrotum, with no evidence of ascites. Echocardiography showed preserved left ventricular systolic function. Chest radiography revealed no abnormalities. Cytogenetic analysis demonstrated a normal male karyotype. Fluorescence in situ hybridization (FISH)

for Prader–Willi syndrome was advised but could not be done due to economic constraints for the patient. Additional investigations, including insulin-like growth factor-1 (IGF-1), were normal; however, vitamin D level was mildly reduced (25 ng/ml).

Besides symptomatic treatment, management in this patient was mainly directed toward reducing obesity. A structured dietary plan, lifestyle modifications, and regular physical activity were advised. Furthermore, treatment with a GLP-1 receptor agonist and later dual glucose-dependent-insulinotropic polypeptide (GIP) and GLP-1 receptor agonist was initiated to facilitate weight reduction. An ACE inhibitor was initiated for its antiproteinuric effect, along with lipid-lowering drugs for dyslipidemia. The patient was followed up at 3-month intervals. Over the course of one year, he achieved a weight reduction of approximately 25 kg, accompanied by resolution of edema and proteinuria, normalization of plasma glucose levels, normal lipid profile, and marked improvement in oxygen saturation and daytime somnolence.

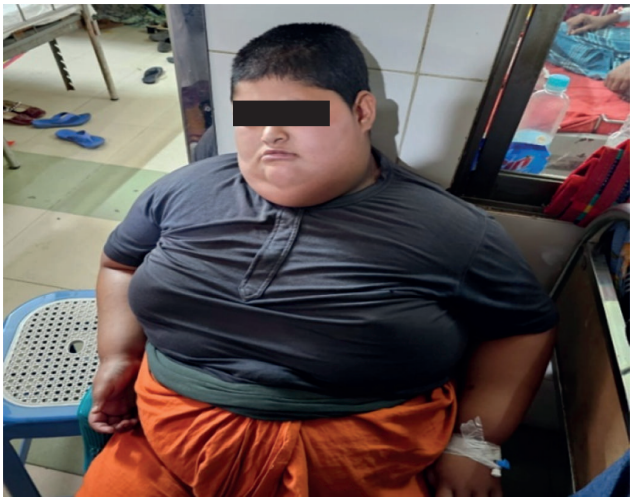

Figure: Patient with Prader-Willi Syndrome

Discussion

The clinical diagnosis of Prader–Willi syndrome (PWS) relies on a structured set of features stratified into major, minor, and supportive criteria according to their diagnostic weight. The major criteria encompass neonatal hypotonia, early feeding difficulties, characteristic craniofacial features, excessive weight gain during early childhood, hypogonadism, developmental delay, and hyperphagia culminating in obesity [7,8]. Minor criteria include reduced fetal movements, behavioral and psychiatric manifestations (such as stubbornness, obsessive–compulsive tendencies, hyperactivity, and aggression), self-injurious behaviors including skin picking, cognitive impairment, delayed speech and memory development, sleep disturbances, short stature, hypopigmentation of the skin and hair, small extremities (particularly hands), and decreased secretion of thick, viscous saliva. In addition, supportive findings—such as scoliosis, osteoporosis, and a relatively high pain threshold—provide further corroborative evidence and enhance the overall clinical suspicion for PWS [9].

Our patient demonstrated multiple features suggestive of Prader–Willi syndrome. When evaluated against the consensus clinical diagnostic criteria, the patient met seven of the eight major criteria—namely excessive weight gain, typical facial features, hypogonadism, learning difficulties, hyperphagia, feeding problems in infancy, and developmental delay. In addition, four minor criteria were satisfied, including characteristic behavioral disturbances, short stature, small hands, and sleep disturbance. 5 points are required for diagnosis, 4 of which should come from the major group. When these clinical features were collectively evaluated, the patient achieved a total diagnostic score of 9, which meets the threshold required to establish the diagnosis[10]. Although karyotyping was normal in this case, this does not exclude PWS, as most cases result from microdeletions or maternal uniparental disomy, which require molecular diagnostic techniques such as FISH or DNA methylation analysis for confirmation [11].

Management of Prader–Willi syndrome (PWS) is tailored to the patient's age and evolving clinical needs, encompassing both treatment of current manifestations and proactive anticipation of future complications. Optimal care is achieved through a coordinated multidisciplinary strategy. In addition, rehabilitative and developmental support from physical, occupational, and speech therapists, along with appropriate educational interventions, plays a crucial role in improving long-term outcomes [12].

Hormone replacement during adolescence and adulthood in Prader–Willi syndrome typically involves administration of testosterone, growth hormone, and levothyroxine, along with supplementation of calcium and vitamin D [2]. Nutritional management focuses on a balanced, calorie-restricted diet combined with regular physical activity and strict supervision to limit food-seeking behaviors and reduce the risk of obesity. Nevertheless, such lifestyle-based interventions often yield limited effectiveness, particularly in older individuals and in those diagnosed at a later stage [13].

Most currently available pharmacological therapies have shown limited efficacy in controlling hyperphagia and associated weight gain in Prader–Willi syndrome, with glucagon-like peptide-1 (GLP-1) receptor agonists

representing a notable exception[14]. A 2020 report demonstrated successful rapid weight reduction with liraglutide in an adolescent with morbid obesity and PWS[15], and a systematic review of ten studies involving exenatide and liraglutide reported improvements in body mass index ranging from 15 to 16.0 kg/m² [16].

In our patient, following treatment with dual GLP-1 receptor agonist and glucose-dependent insulinotropic polypeptide (GIP), the patient achieved a remarkable weight loss of 25 kg (BMI 34.7) and a marked reversal of obesity–related complications, significantly improving the patient's overall health and quality of life.

Conclusion

Adolescents and adults presenting with obesity or obesity related complications or abnormal weight gain that originated during childhood should be evaluated for potential genetic etiologies, with particular attention to Prader–Willi syndrome. In resource-limited settings where genetic testing is not readily available, clinical consensus criteria remain a valuable diagnostic tool. Timely implementation of lifestyle interventions, along with the use of effective anti-obesity medication when appropriate, may facilitate weight reduction and mitigate obesity-related complications**.**

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