

CASE REPORT

Early Growth Hormone Therapy in A Child with Prader-Willi Syndrome: A Case Report

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ABSTRACT

This case report details the diagnosis and early management of a 3-year-old boy with Prader-Willi syndrome (PWS), a rare genetic disorder. He presented with characteristic features including neonatal hypotonia, cryptorchidism, and significant weight gain. Following a comprehensive evaluation, including a crucial pediatric sleep study due to the elevated risk of sleep-disordered breathing in this population, recombinant human growth hormone (rhGH) therapy was initiated. Notably, the child was also diagnosed with iron deficiency anaemia, an uncommon but important comorbidity requiring targeted nutritional intervention. The report underscores the critical need for pre-treatment screening, especially for sleep-disordered breathing, before commencing recombinant human growth hormone therapy in individuals with PWS. This case highlights the immense value of an integrated, multidisciplinary management approach encompassing endocrinology, nutrition, sleep medicine, and behavioural support. Early recognition and timely intervention with recombinant human growth hormone, coupled with comprehensive supportive care, are vital for significantly improving growth, body composition, and overall quality of life for individuals living with Prader-Willi syndrome.

Keywords: Growth Hormone Therapy, Hyperphagia, Cryptorchidism, Genetic Disorder

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1. INTRODUCTION

Prader-Willi syndrome (PWS) is a rare genetic disorder caused by the absence of paternally inherited genes on chromosome 15.(1) It is a rare genetic disorder characterized by hyperphagia, hypotonia, poor feeding, and delayed development, intellectual disability, behavioral issues, distinctive facial features and subsequent obesity in childhood.(2)It may also display characteristic facial features, short stature, and cognitive and behavioral challenges, including intellectual disability and temper tantrum stubbornness, and obsessive-compulsive behaviors. As children, they develop an insatiable appetite, leading to severe obesity if not carefully managed. Early diagnosis and comprehensive management, including dietary control, growth hormone therapy, and behavioral interventions, are crucial for improving quality of life for individuals with Prader-Willi Syndrome.(3).

As they grow older, a hallmark feature of Prader-Willi Syndrome emerges: hyperphagia, or excessive hunger. They also experience hypothalamic dysfunction, leading to various endocrine abnormalities, including hypogonadism, hypothyroidism, central adrenal insufficiency, and reduced bone mineral density. Lifelong endocrinological monitoring is crucial for the comprehensive management of these individuals.(4)

2. CASE REPORT

This A 3-year-old boy presented with progressive weight gain and was admitted for the initiation of growth hormone therapy. His clinical history was significant for early-onset hypotonia and feeding difficulties during infancy, which later evolved into marked hyperphagia and rapid weight gain after one year of age. (figure 1) In addition, a history of surgically corrected cryptorchidism further supported the diagnosis of Prader-Willi syndrome. On physical examination revealed no significant abnormalities apart from obesity, although his weight was notably elevated at 23.6 kg. (figure 2)

Laboratory investigations revealed iron deficiency anemia, as evidenced by low ferritin (7.30 ng/ml), reduced MCV (60.2 fL), and decreased hemoglobin (8.8 gm/dL), along with a mildly elevated cortisol level (25.7 mcg/dL) and a normal HbA1c (5.7%). A pediatric sleep study was also performed using a standardized questionnaire to assess for sleep-disordered breathing.

Based on these findings, growth hormone therapy was initiated, with the patient receiving daily subcutaneous injections of Eutropin (recombinant human growth hormone somatropin) at a dose of 0.5 ml at 9:00 PM for

five days during his inpatient stay, followed by a discharge regimen of Inj Headon, 2 IU (0.5 ml) administered subcutaneously at 9:00 PM daily for one month. This case exemplifies the classic presentation of Prader-Willi syndrome and highlights the necessity of a comprehensive, multidisciplinary approach that integrates endocrine management with supportive treatment for complications such as iron deficiency anemia and potential sleep-disordered breathing. Continued follow-up is essential to optimize long-term outcomes in this patient.



Figure 1



Figure 2

3. DISCUSSION

The Prader-Willi syndrome (PWS), a rare genetic condition caused by the absence of paternally expressed genes on chromosome 15q11-q13, is typically brought on by a paternal deletion or maternal uniparental disomy.(5) The normal clinical progression of PWS is presented in this case report. Early hypotonia and feeding issues in infancy, followed by hyperphagia and rapid weight gain—characteristics of PWS that usually appear between the ages of one and six—were displayed by the patient.(6) Furthermore, a history of surgically treated cryptorchidism is consistent with typical endocrine symptoms seen in male PWS patients.

Based on established evidence that recombinant human growth hormone (rhGH) therapy improves body composition, increases muscle mass and linear growth, and may improve cognitive outcomes in children with

PWS, the patient began recombinant human growth hormone therapy.(2)(7)recombinant human therapy is recommended in early childhood following appropriate clinical and respiratory screening, is currently advised as a routine intervention for children with genetically confirmed PWS. (3) Because of the increased risk of central and obstructive sleep apnea in this population, which recombinant human growth hormone medication may exacerbate, screening for sleep-disordered breathing is an essential part of pre-treatment evaluation. By including a pediatric sleep evaluation prior to starting treatment, our case complied with these recommendations.(8)

This patient also presented with iron deficiency anemia, an uncommon but clinically relevant findings in PWS. In order to promote healthy growth, anemia should be continuously checked and handled, even when nutritional shortages are widespread as a result of

restrictive diets or feeding difficulties.(9)A lifelong, multidisciplinary approach encompassing endocrinology, nutrition, sleep medicine, behavioral therapy, and genetics is necessary for the diagnosis and treatment of Prader-Willi Syndrome. Parental education and early behavioral treatments are frequently necessary for behavioral issues such as obsessive-compulsive traits, food-seeking behavior, and temper outbursts. (4)Since endocrine abnormalities such hypogonadism, hypothyroidism, and central adrenal insufficiency are frequent in PWS and add to the complexity of management, the patient in this report will need ongoing monitoring.(10)

In conclusion, this case illustrates the traditional clinical range of Prader-Willi syndrome and highlights the significance of early detection, recombinant human growth hormone medication beginning, nutritional support, and continuous multidisciplinary care in order to maximize patient outcomes.

4 CONCLUSION

The This case highlights the classical clinical presentation of Prader-Willi Syndrome and underscores the importance of early diagnosis and multidisciplinary management. Initiation of recombinant human growth hormone therapy, alongside targeted management of associated complications such as iron deficiency anemia and potential sleep-disordered breathing, reflects current best practices in the multidisciplinary care of Prader-Willi Syndrome. The patient's clinical course reinforces the necessity for ongoing endocrinological, nutritional, behavioral, and developmental surveillance. A coordinated, lifelong management strategy remains imperative to optimize functional outcomes and enhance overall quality of life in individuals affected by this complex genetic disorder.

5.ACKNOWLEDGEMENT

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6. DECLARATION OF PATIENT CONSENT

The authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given her consent for her images and other clinical information to be reported in the journal. The legal guardian understands that their name and initials will not be published and due effort will be made to conceal identity, but anonymity cannot be guaranteed

7. DATA AVAILABILITY STATEMENT

All the authors had full access to study data and take responsibility for the integrity of the reported information

8. AUTHOR CONTRIBUTION

All authors contributed significantly to the development of this case report. Sai Keerthana Puthiyedath conceived the idea for the case report and contributed to methodology and final proofreading. Ajas Asharaf and Aloshya joy were involved in data collection, and manuscript drafting. Sona Sagi and Muhammed Shahzin and Lasima jabeen assisted with literature review and critical revision of the manuscript. Diya Elizabeth Manoj,Ebenz Chalson K and Shahla P assisted with critical revision, organizing the data and refining the manuscript. All authors have read and approved the final manuscript

KEY CLINICAL MESSAGE

Early initiation of recombinant human growth hormone therapy following appropriate screening for sleep-disordered breathing can significantly improve growth and metabolic outcomes in children with Prader-Willi Syndrome. Multidisciplinary management is essential for addressing endocrine, nutritional and developmental complications

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CONFLICTS OF INTEREST

There are no conflicts of interest among authors.

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