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Prader-Willi Syndrome and the Use of Medical Nutrition Therapy

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Abstract

Title: Integrating Consistent Individualized Carbohydrate-Controlled Anti-Inflammatory Nutritional Plan (C-ICAN) in the Management of Prader-Willi Syndrome: A Case Report. Prader-Willi syndrome (PWS) is a rare genetic disorder caused by a loss of function of specific genes on chromosome 15. Patients with this disease present unique challenges in management, particularly regarding obesity and nutritional regulation as the disease symptoms change depending on the age of the patient and the phase of the disease. These challenges pose critical stressors to caregivers and their families. We present a case report of a 5-year-old Caucasian male diagnosed with PWS, exhibiting failure to thrive and uncontrolled weight gain. His caregiver was his elderly grandmother who, by her own admission, was ill-equipped to deal with the patient's physical symptoms and his behavior in response to dietary restrictions. Through a multidisciplinary approach involving medical nutrition therapy (MNT) involving the implementation of a Consistent Individualized Carbohydrate-Controlled Anti-Inflammatory Nutritional plan (C-ICAN), growth hormone supplementation, and behavioral interventions patient markedly improved physically and emotionally.

Keywords

Prader-Willi Syndrome, Growth Hormone, Medical Nutrition Therapy, Eating Control, Obesity

1. Introduction

Prader-Willi Syndrome (PWS) is a rare and complex genetic disorder considered one of the most common causes of life-threatening inherited obesity [1]. PWS was first described by Swiss physicians Andrea Prader and Heinrich Willi in 1956. The birth incidence of PWS is 1 in 10,000 to 1 in 30,000, with approximately 10,000 to 20,000 individuals living with the condition in the United States

[2]. The clinical presentation of PWS is due to the lack of expression of genes on the paternally inherited chromosome 15q11.2-q13 [3].

The diversity of the clinical presentation of PWS is due to the impaired development and function of the hypothalamic system. Growth hormone deficiency, hypogonadism, hypothyroidism, premature adrenarche, corticotropin deficiency, precocious puberty, and glucose metabolism are the most common endocrine dysfunctions observed in those affected by PWS [4]. The clinical manifestations of PWS present significant care challenges for patients' families and physicians, underscoring the necessity for individualized care plans to ensure successful management. Severe and early-onset genetic obesity is associated with frequent comorbidities that make the management of these diseases challenging. The current estimated prevalence of severe obesity in children is 5% - 10%, probably underestimated due to limited access to genetic diagnosis [5]. Management of genetic obesity focuses on lifestyle intervention, such as nutrition and physical activity. Medical nutrition therapy (MNT) continues to be a cornerstone of care for PWS treatment.

This case utilizes anthropometric measurements, like BMI, weight-for-age, and stature-for-age, and consistent adherence to a regimented MNT protocol in conjunction with growth hormone supplementation to shed light on integrating personalized dietary interventions with proven treatment regimens of growth hormone.

2. Methods

The hospital IRB approval was granted following written informed consent from the patient's caregiver, and de-identified medical records were obtained and utilized. MNT regimen, endocrinology progress notes, dietary progress notes, and growth curve evaluation were used dating back from the patient's birth to February 5th, 2017. A comprehensive retrospective review of this patient's medical history was examined to compile the discussion.

3. Case Summary

The patient is a five-year-old male who was diagnosed in early infancy with Prader-Willi Syndrome.

His past medical history includes being born at 37 weeks gestation with a birth weight of 2.56 kg (5 lbs. 10 oz), dysmorphic features, and poor oral feeding. He was treated for three weeks in the NICU. Diagnoses included lactose intolerance, GERD, and scoliosis. His surgical history included patent ductus arteriosus closure, gastrostomy tube placement, and left orchiectomy at age 18 months (about 1 and a half years). Both parents were healthy, with BMIs within normal limits and heights of 5'7". The feeding and growth clinic had been following this patient since his NICU discharge when he failed to thrive and worked with a multidisciplinary team to address his multiple developmental and nutritional needs as he grew into childhood. Treatment included growth hormone injections,

speech therapy, physical therapy, and occupational therapy. At the age five he was again re-evaluated at the feeding and growth clinic for increasing preoccupation with food, behavior issues and uncontrolled weight gain. The caregiver's stress regarding the patient's care was increasing. The established multidisciplinary treatment plan continued which included growth hormone injections, however, the MNT changed to an individualized dietary regimen of three meals and three small snacks with a low glycemic load pattern with strict portion control. Following the implementation of MNT, patient height, weight, BMI, and weight/age improved significantly. Percentiles were closer to age-appropriate norms with Consistent Individualized Carbohydrate-Controlled Anti-Inflammatory Nutritional Plan (C-ICAN) diet and growth hormone supplementation.

He was also being followed by endocrinology, pediatrics, and a registered dietitian. He received physical therapy, occupational therapy, and speech therapy to aid in steady developmental progress. MNT for the patient included implementation of the C-ICAN. Specifically, his dietary regime consisted of a low glycemic diet with strict meal patterning: three meals and three small snacks with an even spread of lean protein. He avoided fried foods, sweetened beverages, and milk products. Physical activity was encouraged along with increased daily water intake (five cups per day). The patient's legal guardian supervised his feeding times, and strict portion control regulations were followed. Although PWS is typically associated with early childhood obesity, the patient displayed improved growth patterns with the dietary and therapeutic interventions. This case emphasizes the need for individualized care plans to manage PWS effectively.

4. Results

The implementation of the C-ICAN diet in the management of Prader-Willi Syndrome (PWS) resulted in notable improvements in the patient's growth patterns. Challenges, such as periods of weight gain due to family difficulties, were addressed through ongoing support and education for caregivers. However, following adherence to the C-ICAN diet, along with growth hormone supplementation and behavioral interventions, considerable progress was observed. The patient's growth parameters improved, with percentiles reaching levels more typical for their age group.

Anthropometric measurements, including BMI, weight-for-age, and stature-for-age, showed notable advancements over treatment. Importantly, the patient's food-seeking behaviors became manageable for the caregivers. Overall, the results suggest the overall, the results suggest that integrating personalized dietary interventions, such as the C-ICAN plan, alongside conventional treatments, holds promise in individuals with PWS. The caregiver was relieved after behavior stabilization and showed how treatment implementation can help with family stress from patients' disease state [5]-[7]. Further studies are warranted to assess the long-term efficacy and feasibility of the C-ICAN plan in managing PWS and related disorders.

5. Discussion

Obesity in people with PWS differs from the normal obese population. People with PWS in comparison to the normal population with obesity with a similar BMI have less lean body mass and more body fat. The energy expenditure is lower with PWS due to their low fat-free mass or low lean body mass, low level of physical activity and low baseline metabolic rate. Our hypothesis is because of the C-ICAN diet protein-carbohydrate meal pattern and strict timing of meals, the patient's food-seeking behavior decreased significantly which led to the caregiver not needing to lock the food pantry [5]-[7].

The patient has been followed since birth and was showing normal pace weight gain up until the age of 2. At that point, the patient showed an increased interest in food, and he started gaining weight. By the age of 5, behavior problems such as sneaking food became problematic for the family, so the team initiated a new nutritional strategy, which included implementing the C-ICAN diet. The C-ICAN diet, involving structured, low glycemic load meals, offered a viable dietary strategy. By moderating carbohydrate intake and focusing on anti-inflammatory foods, this plan aims to stabilize hunger levels and mitigate enhancing weight management and quality of life for PWS patients. Following a strict MNT protocol with consistent growth hormone supplementation and MNT, the patient's BMI, stature, and weight compared to age were held within an appropriate range compared to the PWS population.

6. Conclusions

The presented case highlights the successful integration of the Consistent Individualized Carbohydrate-controlled Anti-Inflammatory Nutritional plan (C-ICAN) in the management of Prader-Willi Syndrome (PWS) [8]. Through a comprehensive approach involving medical nutrition therapy, growth hormone supplementation, and behavioral interventions, significant improvements in the patient's growth patterns were achieved [9]. The C-ICAN diet, with its focus on regulating glycemic levels and managing hyperphagia, played a pivotal role in optimizing the patient's nutritional status and overall well-being [10].

Challenges encountered underscored the importance of ongoing support and education for caregivers to maintain adherence to dietary and lifestyle interventions [10]. This case underscores the efficacy of individualized and multidisciplinary care approaches in addressing the complex needs of individuals with PWS, emphasizing the importance of tailored interventions for optimal outcomes [11]. Further research and clinical experiences are warranted to validate the long-term efficacy and feasibility of the C-ICAN plan in the management of PWS and potentially other related disorders [11].

As our patient progresses in age, we expected the projected trend to be held constant. We believe that these biophysical parameters are of utmost necessity in order to reduce the future likelihood of developing other chronic and debilitating disease processes like Type 2 diabetes and cardiovascular disease. The bene-

fits of weight management and the maintenance of a healthy BMI have long been recognized. Overall, this case provides compelling evidence for the efficacy of individualized and comprehensive care approaches in managing PWS. Further research and clinical experiences are warranted to validate the long-term effectiveness and feasibility of the C-ICAN plan, not only in PWS management but also in addressing similar genetic obesity-related disorders.

A caveat to our conclusion is that this is a single case and the patient lost follow-up at age 7 due to the relocation of the family; so the C-ICAN diet used with a multidisciplinary team should be replicated in a study with a PWS population to better understand its metabolic effectiveness.

Informed Consent

The patient in this study provided written informed consent prior to participation.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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