

CASE REPORT

Hyponatremic Seizure after Long Term Water Administration for Weight Control in a Patient with Prader-Willi Syndrome

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Abstract

Prader-Willi syndrome (PWS) is a genetic syndrome well known for morbid obesity due to hyperphagia that is difficult to treat. Strategies for weight management include strict dietary planning, growth hormone treatment, and bariatric surgery. Hyperphagia is sometimes accompanied by increased fluid intake in people with PWS. We present a patient who underwent dramatic weight loss after the family administered large amounts of water before each meal. She has maintained a normal BMI for over 15 years, but presented as a young adult with a single hyponatremic seizure. We suggest that using administration of water for appetite control in patients with PWS should not be done without medical supervision, including monitoring of electrolytes and dietary intake.

Keywords: Prader Willi Syndrome; Diet; Weight Control; Water Intoxication; Hyponatremic Seizure; Drinking Behavior

Introduction

Prader-Willi syndrome (PWS) is a genetic disorder caused by deletion or imprinting defect of 15q11.2-q13. It is characterized by infantile hypotonia and failure-to-thrive early in life, followed by hypogonadism, global developmental delay, and behavioral problems including temper tantrums and obsessive-compulsive disorders. PWS has been described as having two nutritional phases: poor feeding during infancy, and hyperphagia leading to obesity in later childhood. Strict dietary planning can help with weight control. In addition to unusual eating behaviors, some patients with PWS have been reported to have unusual drinking behaviors, where they may consume excessive amounts of liquid. The management of obesity in children with PWS is an important and difficult task. Some studies have shown that weight management can be achieved by optimizing food intake, and use of growth hormone may improve body composition. We present an adult who has achieved and maintained weight control for over 15 years by drinking large amounts of water. This was initiated and maintained by the family without medical or nutritional supervision.

Case Report

We present a case of a 26 year-old female with PWS who presented with an episode of water intoxication and a hyponatremic seizure at age 24 following excessive water intake, which had been used by the family as a mechanism for appetite control. She had no previous seizures. The patient had a history of hypotonia and poor sucking and feeding in infancy, leading to poor weight gain. Development was delayed due to hypotonia. She began to roll and sit at 1.5 years, and walked at 3 years. As an adult, her cognition is at the level of a 6 or 7-year-old, though she exhibits a high level of skill with puzzles. She cannot read but speaks fluently in two languages.

Her food-seeking behavior began at age 4. She had an uncontrollable appetite and she became aggressive when she was not given food. At times, she would pick her skin and suck her blood. At the age of 7, she was morbidly obese, and was reported to have weighed 100 kg, and at age 8, 127 kg. At that time, her parents initiated a dietary plan where they would give her a lot of water, at least 32 ounces prior to each meal, so that she would not eat as much during that meal. On average she took 32 ounces, four times per day. By the age of 9, she had lost around 45 kg, and the weight loss continued. For the remainder of her childhood and

early adulthood she weighed around 45 kg. She had further gradual weight loss in her early 20's, and on presentation at 24 years, she weighed 35 kg. Prior to presenting with hyponatremic seizure, she had begun desiring more water, up to sixteen to twenty 16-ounce glasses per day.

When she presented with seizure due to water intoxication, she was hospitalized to correct her sodium levels. After that episode, her parents tried replacing some of the supplemental water with a sports drink containing carbohydrates and electrolytes, but that was stopped due to weight gain of around 14 kg. Her parents tried giving a diet form of the same beverage, but it caused her to have sweet cravings. Currently, her diet consists of a nutritional shake replacing breakfast and sometimes a second meal, and food including rice with meat and vegetables for lunch, cereal or sandwich for dinner, fruit for a snack, and 8 ounces of water every hour from 10am to 7pm. She has obsessive-compulsive characteristics that contribute to her desiring the same food to which she has grown accustomed, so there is little variance to her meals from day to day. She takes a normal amount of salt in her food. She no longer exhibits food seeking behavior. The family keeps the food locked and if food is left out, she will still eat it. However, she does not eat nonfood items. At the most recent visit, she appeared healthy and proportionate, with a height of 4 feet 4 inches and weight of 80 pounds, equivalent to a BMI of 20.8.

Other medical issues include osteopenia and strabismus. ANA was checked due to malar rash and Raynaud's phenomenon and was normal. Cardiology evaluation showed functional tricuspid insufficiency. Ophthalmology evaluation revealed bilateral refractive amblyopia. She has hypogonadism but has never been treated for growth hormone deficiency or pubertal delay due to parental choice. Family history is negative for seizures.

Physical Exam

On physical exam she is thin with short stature, and has mild facial dysmorphism with long facies, bitemporal narrowing, overcrowded teeth, mild retrognathia, and almond-shaped eyes. She has a malar rash. She is Tanner 1-2 stage for breasts and Tanner 2 stage for pubic hair. She has small hands and small feet.

Testing

Microarray revealed a loss of approximately 5.05Mb on chromosome 15q11.2q13.1. This involves over 100 genes and 18 OMIM genes, including those responsible for Prader-Willi Syndrome and Angelman Syndrome. She is also a carrier of a deletion of the OCA2 gene, but does not have apparent hypopigmentation.

Discussion

Water intoxication has been described in isolated cases in patients with PWS. The Prader-Willi Association has a "Water Intoxication Alert" since experientially this has been a concern. The water intoxication in other cases has been related to medications and SIADH, but in this case was utilized by the family for appetite control. The use of large volumes of water given to the patient in this case successfully controlled her weight. It was only after 17 years of this method and a brief period of increased water intake that she developed a hyponatremic seizure.

Water intoxication in the general population has been studied in great detail, with the majority of cases attributing intoxication to vigorous hydration, psychiatric disorders, infantile formula mismanagement, and freshwater drowning. The kidneys in a normal adult can excrete approximately 800 to 1000 ml/hour. When the amount of water intake exceeds the amount that can be excreted by the kidneys, water is retained and can cause hyponatremia. Concentration gradients between intracellular and extracellular compartments can cause severe swelling and edema, particularly dangerous in brain tissue. Severe manifestations of hyponatremia include seizures, coma, respiratory arrest, and death [13].

Water intoxication in the PWS population has been demonstrated in a few studies, but has not been deeply explored in the medical literature. In one study of water intake in people with PWS, the majority of infants with PWS drank an unusually small amount, but as they aged, there were episodes of consumption of excessive amounts of water. It was reported in one cohort that 23% of the PWS individuals were drinking excessive water by age 25. Two of these individuals experienced water intoxication resulting in severe hyponatremia. In both cases, the excessive water intake was attributed to psychiatric medication and in one case SIADH was demonstrated [1]. While abnormal drinking behavior is not a diagnostic criterion for PWS, it has been noted and reported by families of patients with PWS [1,7,4]. In one study with a large adult PWS cohort, water intoxication was one of the most common causes for hospitalization for individuals over age 40, and a documented cause for younger adults as well [14].

There are a number of strategies to assist with weight loss in individuals with PWS including behavioral management, strict dietary planning, exercise, bariatric surgery, and growth hormone administration. One study was successful at managing weight and lowering body fat with an energy-restricted diet with a well-balanced macronutrient composition and fiber intake [11]. A fat-reduced and modified carbohydrate diet has been described to have efficacy when started in childhood or introduced in adolescence [3]. Liquid diets and water administration are not commonly used dieting strategies in PWS, but are represented in the dieting strategies by the general population. Some published studies have reported that liquids are less satiating than solids, whereas other studies have reported that solids are less satiating than liquids. The context may be important in that water in between meals has less of an impact on satiety than does water intake just before or during meals [2]. One study suggests that water load may increase

the energy expenditure by stimulation of sympathetic dependent thermogenesis, which will aid in weight reduction [8].

This report highlights anecdotal success in weight control when a family implemented free water in larger than recommended amounts. Her diet had not been managed by a dietician or physician to assure adequate intake of calories, nutrients, and electrolytes, though this has been recommended for the future. We acknowledge that it is not standard practice to give excessive water and can lead to complications such as hyponatremia, which may be associated with cerebral edema and even death in the most severe cases. We do not have longitudinal sodium measurements but only once was this patient known to be symptomatic due to hyponatremia, when she presented with a seizure in young adulthood. She did not experience dysphagia, choking, or other GI symptoms when she drank large volumes of water during the 17 years that she was on this diet.

Conclusions

PWS can cause unusual eating and drinking behaviors, with too low intake in infancy and excessive intake starting in childhood. This case report suggests that water administration was an effective method of weight loss and maintenance in this particular patient. However, electrolyte supplementation and monitoring would be crucial to avoid the complication of water intoxication, and a dietician should help assure adequate nutrition. This case may be informative in future research on weight control in patients with PWS.

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