

Prader Willi syndrome: A case report of a 10-year-old child

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Abstract- Prader-Willi syndrome is a complicated disorder (CVS, Respiratory illness), which has several symptoms related to hypothalamic insufficiency and affects many different systems such as respiratory, endocrine, neurologic and metabolic systems. Infantile hypotonia, developmental delays, mental impairment, behavioral problems, small stature, abnormality, distinctive facial features, obesity, hypogonadism, are significant results. The main causes of morbidity and mortality are behavioral issues and obesity. The absence of the typically active paternal genes in the proximal 15q imprinted region results in Prader-Willi syndrome, which is brought on by anomalies in this area. Such absence is brought about by maternal uniparental disomy, paternal interstitial deletion, mutations, or other abnormalities in the imprinting process. In recent years, diagnostic identification of all causes has become possible, enabling early detection and the implementation of effective management. Recent phenotypic variations among affected people of different races, as well as between those with deletions and uniparental disomy as a cause, have been identified through this testing. The treatment/management always depends upon the symptoms.

UBE3A gene in this location ^[5,6]. Significant hypotonia, feeding issues, and failure to thrive are all common in affected infants. Later in infancy or early childhood, excessive appetite with the gradual development of obesity, short stature and/or slowed growth, intellectual disabilities (average IQ of 65), and behavioral issues such as tantrums, outbursts, and skin picking also, appear ^[7]. Many indications of this disease, including hyperphagia, temperature instability, high pain threshold, sleep-disordered breathing, and other endocrine abnormalities, have been linked to hypothalamic dysfunction ^[8,9]. Symptomatic management is the only treatment for PWS. Obesity can be controlled by maintaining a diet. Osteoporosis and osteopenia are the most common and are prevented by providing adequate nutrition with calcium, vitamin D, and physical activities for improving bone health. Imbalance in Growth hormone and sex hormone are treated with Growth hormone therapies, antiepileptics are given for seizure conditions. Respiratory infections and coughs are more common and treated with third-generation cephalosporins and syrups. Gastrointestinal abnormalities like delayed gastric emptying, gastroparesis, constipation, abdominal distension, gastric inflammation to necrosis which is treated using antacids, proton pump inhibitors and H2 blockers.

I. INTRODUCTION

A multisystem illness called Prader-Willi syndrome (PWS) was originally identified in 1956 ^[1]. The fact that it was the first known microdeletion syndrome discovered by high-resolution chromosome research attracted the interest of geneticists 25 years later ^[2]. In both male and female gametogenesis, errors in genomic imprinting are paternal genes, which are typically expressed and are found on the chromosome 15q11–q13 region and are also responsible for PWS ^[3,4]. Conversely, Angelman syndrome (AS), a completely separate clinical disease, results from a lack of expression of the preferentially maternally expressed

II. CASE PRESENTATION

A 10-year-old male patient was admitted to the pediatric department with complaints of cough. During the vital examination, he showed normal vitals and lab parameters excluding TC (11982 cells/cumm). For symptomatic management, the pediatrician advised Syp. Ascoril LS (Ambroxol, Levosalbutamol, Guaifenesin) and the second-generation cephalosporine derivative such as Tab. Cefixime 200mg twice daily. The physical examination revealed that he had almond-shaped eyes with short size

and was obese with a weight of 83.6kg. On taking the medication history interview it was clear that the child was diagnosed with Prader-Willi syndrome when he was 4 months.

The foetal movement was not normal, but the scanning showed normal during the pregnancy time and the child was underweight (1.25kg) during delivery without abnormalities.

The first month showed an endogenous weight gain/obesity with reduced cardiac size. Cardiomegaly was diagnosed with the help of Chest X-ray. Echo revealed impaired RA/RV function with moderate PAH. Pulmonary angiography showed pulmonary embolism. D-dimer showed an elevation up to 6927mg/L and gradually decreased (737mg/L) as myocarditis got normal.

During the 6th month, the child was obese (5.65kg), mild hypotonia, partial ptosis, occipital plagiocephaly and edema on feet and hands were reported.

Normal hearing and vision were found in his 7th month.

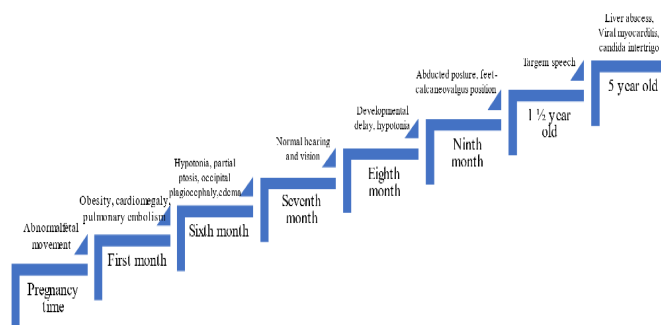
Growth delay (similar to 2nd month) was found along with hypotonia during his 8th month. His growth was similar to 2 months, head control therapy, prone positioning, and neck massaging was advised.

During the 9th month, he was 9.3kg with an abducted posture, placing his feet in the calcaneovalgus position. Targem's speech was found when he was 1 ½ years old.

At the age of 5, he was admitted under the pediatric department with severe abdominal pain and fever, USG Abdomen and Pelvis showed a hypoechoic right hepatic lesion. CECT of the Abdomen and Pelvis was taken and reported as a peripherally enhancing cystic lesion in the right lobe of the liver which was further diagnosed as Liver abscess.

A couple of months later, he was admitted with tiredness, poor activity, pallor, respiratory distress, cough, and hence diagnosed as Viral Myocarditis RVH+RAH+PAH RV dysfunction and candidal Intertrigo.

DISEASE PROGRESSION



III. DISCUSSION

The most prevalent type of obesity brought on by a genetic abnormality is Prader-Willi syndrome (PWS), a genetic illness that typically affects chromosome 15. Because the clinical signs are generally nonspecific, particularly in infancy, and the dysmorphism is frequently modest, diagnosis is frequently postponed until early childhood^[10].

Infantile hypotonia, which causes decreased mobility and lethargy with diminished spontaneous arousal, weak cries, and impaired reflexes, including a poor suck, is a practice common finding. The cause of the hypotonia is central, and when performed for diagnostic purposes, neuromuscular investigations, including muscle biopsy, are typically normal or exhibit vague symptoms of disuse. Early infancy fails due to poor sucking and laziness, and gavage feeding or the use of special nipples is frequently necessary for a variable amount of time, usually weeks to months^[11].

Even in slender newborns with PWS, the ratio of fat mass to lean body mass is high. This is likely due to hypotonia and the resulting loss of muscle mass. However, considerable obesity typically develops between the ages of 1 and 6 years after the initiation of hyperphagia. Food-seeking behaviours are widespread and include hoarding or hunting for food, consuming unattractive foods including rubbish, pet food, and frozen food, and stealing food or cash to pay for food. Binges on damaged food from the trash, as well as products like boxes of sugar or frozen raw meat, may be more difficult for people with a high threshold for vomiting, and toxicity from inadequate Ipecac used to induce vomiting has happened^[12].

In PWS, obesity is the main factor in morbidity and mortality, and if it is prevented, longevity may be almost normal^[13]. Excessive weight gain can lead to cardiopulmonary impairment, type II diabetes, hypertension, thrombophlebitis, and chronic leg edema. Sleep apnea happens more frequently^[14]. A dysfunction in the hypothalamus that prevents satiety is what causes the hyperphagia. Additionally, there is a reduced caloric need, which is most likely brought on by hypotonia and a decline in activity^[15].

One of the key characteristics of people with PWS is short stature. Without therapy, the mean final height for girls with PWS is 148 cm, and for boys with PWS is 155 cm^[16]. Children with PWS do not experience the growth acceleration associated with puberty. For newborns and kids with PWS who are not receiving GH treatment, growth charts have been created^[17]. Obese children who are not PWS have lower GH secretion while still retaining normal height and serum insulin-like growth factor-1 (IGF-1) levels. True GHD is taken into consideration in children with PWS because of their small stature and low serum GH and IGF-1 levels. Children with PWS who were given GH treatment throughout their childhood were able to grow to a typical adult height. investigations investigating the effects of GH unrelated to exercise ability were driven by the positive effects on body weight, body composition, and exercise capacity revealed in these investigations^[18, 19, 20].

Although most patients' verbal abilities are their greatest strength, their speech is frequently poorly articulated and has a nasal or slurred tone. The majority of patients are slightly retarded and there are clear cognitive impairments. 20% of people have significant retardation, whereas 40% have borderline retardation or poor normal intellect. Cognitive ability does poorly in academics^[21].

IV. CONCLUSION

Infants with PWS typically already experience hypoventilation. Hypoventilation is most likely the result of weak respiratory muscles, some pharyngeal narrowing, and a decline in CO₂ sensitivity. Obesity and kyphoscoliosis both play a significant impact in catastrophic outcomes and may further exacerbate the breathing issue. Hypoventilation also increases infection susceptibility, which can exacerbate respiratory insufficiency on its own. A combination of excessive obesity and a pre-existing breathing issue may not have received the attention it deserved in the majority of patients who died during GHT. Before GHT was started, all of these kids had hypoventilation and poor respiratory control. A molecular organization disturbance in a crucial region of 15q causes this rare condition, which has significant effects on the central nervous system as a whole. The right diagnosis is crucial, and it can be made using a combination of clinical diagnostic standards and molecular genetic techniques. The cornerstone of managing obesity is still dietary control. The brains of PWS patients should be studied using modern diagnostic techniques like positron emission tomography and magnetic resonance imaging.

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Nil

INFORMED CONSENT

Before taking this case the patient and their families were informed and informed consent was acquired.

CONFLICTS OF INTEREST

Nil

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