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Case Report
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Management of Newborn with Parder-Willi Syndrome: A Case Report

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Abstract

A case of Parder-Willi syndrome is described in a 2.5-year-old boy. He is positive with hypotonia and hyperphagia and failure to thrive with feeding problems in the neonatal period. Clinically he has dysmorphic features with widened nasal bridge, short stature, short hands and feet. The diagnosis was confirmed by gene mapping and DNA analysis. Early diagnosis was important for management of clinical signs, for parental counselling and for further investigations for stimulating the condition.

Introduction

Prader-Willi Syndrome (PWS) is a genetic disorder that occurs in approximately 1 out of every 15,000 births. PWS is recognized as the most common genetic cause of life-threatening childhood obesity and it affects males and females with equal frequency [1] (Figure 1).

However, the clinical characteristics defined are small hands and feet, abnormal growth and body composition (small stature, very low lean body mass, and early-onset childhood obesity), hypotonia (weak muscles) at birth, insatiable hunger, extreme obesity, and intellectual disability. There are two general stages of symptoms of Parder-Willi syndrome. Early stage and childhood and beyond stage [1,2].

Early life: Infants with PWS are hypotonic or "floppy", with very low muscle tone. A weak cry and a poor suck reflex are typical. Babies with PWS usually are unable to breastfeed and frequently require tube feeding. These infants may suffer from "failure to thrive" if feeding difficulties are not carefully monitored and treated. As these babies grow older, strength and muscle tone generally improve. Motor milestones are achieved, but are usually delayed. Toddlers typically enter a period where they may begin to gain weight easily, prior to having a heightened interest in food [1].

Childhood & beyond: An unregulated appetite and easy weight gain characterize the later stages of PWS. These features most commonly begin between ages 3 and 8 years old, but are variable in onset and intensity. Individuals with PWS lack normal hunger and satiety cues. They usually are not able to control their food intake and will overeat if not closely monitored. Food seeking behaviors are very common. In addition, the metabolic rate of persons with PWS is lower than normal. Left untreated, this combination of problems leads to morbid obesity in children with complications [1].

PWS is diagnosed by blood test that look for genetic abnormalities called- methylation analysis. A FISH (Fluorescent *in-situ* Hybridization) test identifies PWS by deletion.

Case Presentation

Antenatal history

A 2.5-year single term male born at 38 weeks of gestation to a second gravida mother with regular antenatal visits. There was a history of previous spontaneous abortion due to unknown cause.

Birth history

The present baby cried immediately after the birth with Apgar score of 7 at 1st minute and 8 at 5th minute and does not require any resuscitation.

Postnatal history

Previously the baby was having history of neonatal jaundice, infantile hypotonia, failure to thrive at 3 month of age, hyperphagia, recurrent typical febrile seizures.

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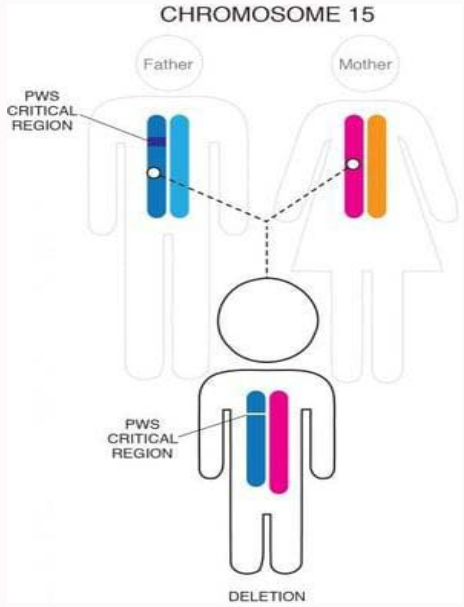

Figure 1: Prader-Willi syndrome.

Clinical manifestation

The appearance of the baby was depressed and wide nasal bridge, Obese according to the age, small eyes, dysmorphia, behavioral changes, Micropenis, short stature according to the age, hypopigmentation, small hand and feet are there, drooling of viscous saliva is present and presence of growth developmental delay in social and language domain of the baby. At present the baby was again admitted with a 3 episodes of abnormal body movement with frothing of saliva from the mouth and tachycardia. Currently the anthropometric measurement of the child was 26 kg of weight, Height was 62 cm, Head circumference was 52 cm, and Chest Circumference was 60 cm.

Confirmatory diagnosis

The baby looks obese according to the age so the baby was referred to genetic OPD for micro assay test and Gene Mapping and then there Parder-Willi syndrome was detected. Currently the baby was treated for the abnormal body movement tab phenobarbitone 7.5 mg and child was treated under child guidance clinical for behavioral issues.

Discussion

In this present study the baby 2.5-year-old was diagnosed with PWS was treated at Guru Tegh Bahadur Hospital New Delhi. The clinical characteristics were reported as small eyes, dysmorphia, behavioral changes, Micropenis, and short stature according to the age, hypopigmentation, small hand and feet. Literature also shows dental issues like maxillary lip and labial commissures facial down. Many studies resemble the findings of the present study i.e. Randomized clinical trials have demonstrated the favorable effect of Growth Hormone (GH) reposition to reduce fat mass and increase lean body mass, complementing nutritional guidance. Not with standing, GH seems not to have significant effect on controlling hyperphagia in these patients [3]. Corroborating Cortés et al., Olczak-Kowalczyk et al., and Vargas et al., the patient of this case report showed learning disorder related to writing; but with mild cognitive deficiency [4,5]. PWS patients also presented bone mineralization decrease, fact explaining the high osteoporosis incidence associated with the syndrome [2]. According to the patient's mother, episodes of irritability were rare, mainly related to dietary restrict ion. Prior studies also reported this situation [6]. Generally, this study's patient had a sociable and friendly behavior with all multidisciplinary team.

The peculiar facial characteristics of PWS patients were also observed in this case report: Almond eyes, thin maxillary lips, labial commissures facing down, and dysmorphic face. Among the oral manifestations reported by Carvalho et al., this case report found the presence of caries, enamel hypoplasia, and malocclusion. On the other hand, ogival palate, delay in tooth eruption, supernumerary teeth, microdontia, micrognathia, taurodontism and candidiasis were not seen [7,8].

Conclusion

Based on the above information, we concluded that the PWS has a stressful situation for parents and for patient so, constantly motivation was provided to them regarding maintain the nutritional status and specially the oral health which was not present in current patient but due to hyperphagia which is dormant to oral caries and obesity can be present in older children.

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