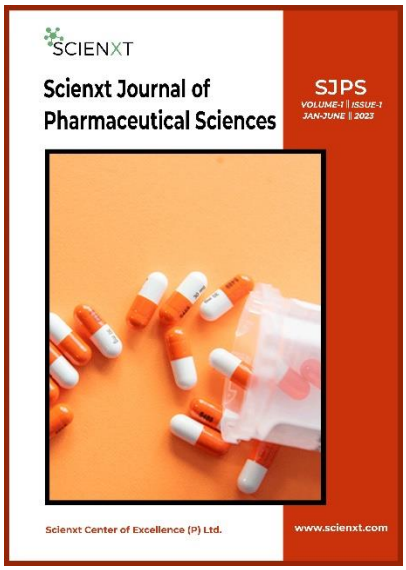

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A case report on Prader Willi Syndrome

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Abstract:

Prader Willi syndrome is a genetic disorder caused by the loss of a portion of chromosome 15 which is handed down from father to son. 14-year-old boy patient was hospitalized after complaining of having a fever and shortness of breath for the previous month. Thyroxin 75mg 1-0-0 was prescribed in the past for K/C/O hypothyroidism. Patient started experiencing breathlessness, began subtly and worsened over time. Patient was unable to hold at his trunk and couldn't walk by 2 years. The patient started to gain weight by 8-9 years. Diagnosis was done based on criteria for PWS. They obtained an overall score of 9+ based on both major and minor criteria and patient was treated with medications based on signs and symptoms of the patient.

Keywords:

Prader Willi syndrome, chromosome 15, hypothyroidism.

1. Introduction:

Prader Willi syndrome is a genetic disorder usually caused by the loss of a portion of chromosome 15. In chromosome no: 15, one of the pair is found to be different in case of PWS [1]. The chromosome 15 abnormality is found to impact the growth and function of the hypothalamus, an area of the brain which performs many body functions such as hormone production and appetite management [2].

The usual symptoms of Prader Willi syndrome include behavioral problems, short height, and delay in puberty, speech, development with small stature, excessive hunger or decreased hormone production throughout the body. Flaccid paralysis or abnormalities, infertility, overweight, poor baby feeding, excessive sleepiness, sleep apnea or small feet are also frequent [1]

2. Case Presentation:

14-year-old boy patient was hospitalised after complaining of having a fever and shortness of breath for the previous month. Thyroxin 75mg 1-0-0 was prescribed in the past for K/C/O hypothyroidism. Up until a month prior, the patient appeared to be in good health. At that point, however, the patient started experiencing breathlessness, which began subtly and worsened over time. Patient was unable to hold at his trunk and couldn't walk by 2 years. The patient started to gain weight by 8-9 years. According to the mother's history, the child was born via lower uterine segment section (LDCS) and did not cry right away. He had a history of meconium aspiration, NICU admission, neonatal jaundice, difficulty milk sucking, hypotonia, poor academic performance, hyperphagia, and tantrums. Personal history revealed a diverse diet, regular sleep patterns, increased appetite, regular bowel movements, and a regular bladder. Family history was not significant. On physical examination, the patient's conically curved arm made it challenging to fit a rectangle (cylindrical when worn) cuff to the arm, and PR was 98 bpm, RR18 com, SPO2- 96 percent with 3L O2. BP could not be measured. The peripheral smear report shows normal WBC counts and normocytic hypochromic RBCs. It appeared to be normocytic hypochromic with relative neutrophilia. A study of the urine revealed the presence of urine protein together with 4-6 pus cells. ESR 60 mm at the end of 1 st hour. Reticulocyte count showed 0.1. Diagnosis was done based on criteria for PWS. They obtained an overall score of 9+ based on both major and minor criteria and patient was treated with medications based on signs and symptoms of the patient.

Figure. 1: Patient with Prader Willi Syndrome

3. Diagnosis:

It was done based on criteria for PWS. They obtained an overall score of 9+ based on both major and minor criteria.

3.1. Major Criteria:

Neonatal hypotonic (+)

Loss of muscle tone represents an indication of a deeper medical issue [3].

Feeding problems in infants (+)

At first, they are slow eaters and seem malnourished. After infancy, the feeding issues get better [4].

Excessive rapid weight gain (+)

Starting above the age of 2 year old, a persistent appetite causes rapid weight gain which is an indication of PWS [5].

Characteristic facial features (-)

It includes short height, tiny hands and feet and unusual facial features such as a small forehead, almond-shaped eyes, triangular mouth and unusual pale complexion and light hair [7].

Hypogonadism(+)

Genital underdevelopment and infertility and at birth, genital hypoplasia is visible [6].

Global developmental delay(+)

GDD, hyperphagia and the onset of morbid obesity gradually around the age of 3. Strabismus, facial characteristics and other musculoskeletal issues can all help identify it [8].

Obsession history(+)

Children and adult start engaging in repetitive or obsessive behavior along with mental illnesses such as anxiety and skin plucking [9].

Deletion of 15q 11 13(+)

It happens as a result of absence of genes expressed from father and are only regularly expressed when the paternally given copy of chromosome 15 is present [10].

4. Minor Criteria:

Behavioral problems with tantrums (+)

Sleep apnea (+)

4.1. Major Indications in the patient:

4.1.2. Hypogonadism (PWS):

The hypothalamus-pituitary-gonadal axis is involved in both central and peripheral pathways that contribute to hypogonadism in PWS [11].

4.2. Obesity:

Primary regulatory mechanisms in the hypothalamus, persistent rise in plasma ghrelin causes an increase in hunger and food consumption. Plasma pancreatic polypeptide-a little protein produced by the pancreas that aids in regulating the release of other chemicals from the organ. Reduced PP and PYY are inability to modulate satiety. Reductions in muscle tissue and an increase in body fat are caused by GH deficiency and hypogonadism [5].

4.3. Obstructive sleep apnea:

OSA is hypothesized to be caused by a combination of altered facial characteristics, low muscle tone, which can cause respiratory muscles to become insufficient and obesity which can cause pharyngeal narrowness in PWS individuals [2].

4.4. Hypothyroidism:

Hypothyroidism is diagnosed due to which thyroid gland itself is the source of the malfunction.

5. Discussion:

The prevalence of PWS in the United States is between 10,000 and 20,000 with a birth incidence of 1 in 10,000 to 1 in 30,000 [13]. Most cases arise due to loss of genetic information from the copy of chromosome no. 15, inherited from the father.

Based on the clinical characteristics, PWS is classified into five stages as shown in Table 1. [14].

Table. 1: Clinical characteristics of nutritional phases

Phase 0	**Fetal movement is reduced, and growth is restricted.**	**In utero**
Phase 1a	The infant develops hypotonic and may fail to thrive.	0- 9 months
Phase 1b	Infant starts feeding and develops consistently following a growth curve.	9- 25 months
Phase 2a	Weight gain occurred with no discernible change in appetite or calorie consumption.	2- 4 years of age
Phase 2b	Continuous weight gain accompanied with increasing food interest	4- 8 years of age
Phase 3	Hyperphagia, increased food craving, and a lack of satiety	8 years of age
Phase 4	Loss of insatiable appetite and can feel full	Adulthood

Clinical signs are typically used by a doctor to make a suspected diagnosis of Prader-Willi syndrome (PWS). Methylation analysis is the test of choice since it can identify all of the main genetic subtypes of PWS and discovers >99 percent of cases [12].

6. Clinical Approach:

A balanced diet, hormonal therapy, and treatment based on the specific patient's symptoms are typically part of the treatment plan. Here, the patient received symptomatic treatments like oxygen support, bronchodilators and antibiotics for breathlessness and infections respectively. They also received proper calorie intake control, metformin use for weight loss and for hypothyroidism they used Tab. Tyronorm.

7. Conclusion:

PWS is a chromosome 15 genetic abnormality that results in an aberrant HGP axis pathway. Obesity, OSA, behavioral issues, and intellectual incapacity are some of its symptoms. The basic course of treatment entails hormonal therapy together with a well-planned nutritional diet to reduce obesity and treatment based on the patient's indications and symptoms, in order to improve the patient's quality of life.

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