

# Prader-willi syndrome

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## Abstract

**Background:** Prader willi syndrome is a multi-systemic genomic imprinting disorder caused by lack of expression of genes on paternally inherited chromosome 15<sup>1</sup> Here, we present a case of PWS in a 5-year-old male child who had presented with excessive sleepiness and rapid breathing.

**Keywords:** Prader willi syndrome, genomic imprinting, chromosome 15

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## Developmental History: Mild intellectual defect

Table 1: Anthropometry

	Observed	Expected	Percentile
<b>Weight</b>	45kg	18kg	> 97
<b>Height</b>	104cm	110cm	< 3rd
<b>Head Circumference</b>	47cm	50cm	< 3rd
<b>BMI</b>	41.6		Morbid Obesity

## On Examination

Patient was afebrile with heart rate of 170/min, RR of 50/min, and BP of 90/60 mmhg. Mild pallor and edema present. Spo2 was 78% at room air and 97% with oxygen by mask. Short stature, microcephaly, short limbs and neck RS: bilateral vesicular breath sounds heard. Breath sounds decreased bilaterally at inframammary and mammary areas.



Figure 1: morbid obesity

## INTRODUCTION

PWS is a multi-systemic complex genetic disorder. It occurs because of absence of expression of paternally inherited genes in 15q11-q13 region. It may be due to paternal deletion, maternal uniparental disomy, imprinting defect.<sup>1,2</sup>

## CASE PRESENTATION

A 5-year-old male child born as 1st issue of nonconsanguineous marriage. presented with excessive sleepiness and loss of appetite for 7 days and rapid breathing in the last 3 days. Child developed excessive sleepiness associated with loss of appetite in the last 7 days and rapid breathing for 3 days.

### Past History

Child was diagnosed with PWS at the age of 6 months by DNA methylation analysis technique. Child was hospitalized multiple times for lower respiratory tract infection.

### Birth history

LSCS done due to non-progression of labour and his birth weight is 2.9kg.

No history of NICU admission.

**Lab Investigations:** CBC done with Hb-9.8, wbc- 16,300, platelet- 2,82,000. LFT done with SGOT (60), SGPT (552) raised, Sr bilirubin, Sr protein, RFT, Electrolytes are normal. TFT was normal.

**Lipid profile:** Triglyceride -88(raised), Cholesterol -95, HDL - 36, VLDL -17.6, LDL -41.4, LDL/HDL= 1.15, Chol/HDL = 2.64.

Chest X-Ray done showing highly placed diaphragm.

USG Abdomen and Chest showed no pathology

**2D-ECHO:** moderate to severe tricuspid regurgitation with RVSP-68mmhg, RA/RV dilated, mild pulmonary regurgitation LVEF-60%.

**Pulmonary artery:** moderate PAH.

**Diagnosis:** known case of Prader willi syndrome with secondary pulmonary hypertension, central sleep apnea, morbid obesity.

**Treatment:** Oxygen by Prong. Tab ALDACTONE 25mg Tab AMBRISANTAN 5mg. Syp TONOFERON 5ml. CPAP machine advised during sleep.

**Advice:** Regular Physiotherapy, Dietary control to decrease obesity.

## DISCUSSION

PWS is a highly variable genetic disorder affecting multiple body systems. PWS was first discovered by Prader *et al.* in 1956<sup>6</sup> and now recognised as genomic imprinting disorder. Estimated prevalence of PWS is 1/10,000 to 1/30,000<sup>1</sup>. The errors in genomic imprinting, which occur during both male and female gametogenesis, depending on the parent of origin, the loss of expression of paternal genes on chr15q11-q13 region leads to PWS and maternal genes in this region leads to Angelman syndrome, which is an entirely different clinical disorder.<sup>7,8,9</sup> DNA methylation analysis is the technique to diagnose and differentiate PWS from Angelman syndrome. PWS is most common syndromal cause of life threatening obesity and first recognized disorder related to genomic imprinting in humans<sup>7</sup>. Mothers often have decreased fetal activity and mostly baby found in breech positional the time of delivery<sup>2</sup>. Early diagnosis is important for effective long term management. Clinical manifestations change with age. Most common manifestations include Hypotonia and poor suck resulting in failure to thrive during infancy. As the individual ages, other features such as food seeking with excessive weight gain, unique behavioural characteristics and cognitive impairment become evident<sup>1,2,3</sup>. The phenotype is likely due to hypothalamic

disfunction which is responsible for hyperphagia, hypersomnia, temperature instability, high pain threshold and multiple endocrine abnormalities include Growth hormone deficiency causing short stature for family, TSH deficiencies and hypogonadism. Obesity and its complications are the major cause of morbidity and mortality in PWS<sup>3</sup>

Management is symptomatic and supportive. Parental education about appropriate parenting techniques (structure and consistency) emphasising control of food intake and behaviour management which correlates with prognosis.<sup>4,5</sup> Hormone replacement therapies like GH treatment in early age improves growth, body composition and muscle strength<sup>4</sup>.

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