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CASE REPORT

A Case of Prader-Willi Syndrome with Obstructive Sleep Apnea

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ABSTRACT

Background: Prader-Willi syndrome (PWS) is a genetic disease caused due to defect in DNA methylation within the Prader-Willi region (PWCR) at 15q11q13. A young female presenting with complaints of sleep disturbance, burning micturition, and pain in abdomen is a case in point. Patient was subjected to polysomnography and was diagnosed as Obstructive sleep apnea (OSA). Result: The relationship between Prader-willi syndrome and obstructive sleep apnea is established and hence monitoring of the patients of PWS is of significance in patients of OSA.

Keywords: *Genetic disorder, Obstructive sleep apnea, Prader-Willi syndrome, Polysomnography.*

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INTRODUCTION

Obstructive Sleep apnea is defined by repetitive episodes of apnea and hypopnea in addition with symptoms such as excessive daytime sleepiness, behavioural worsening and cardiovascular abnormalities.[1] PWS have a characteristic deletion of the proximal part of the long arm of chromosome 15 {del15(q11q13)}.[2] Diagnosis of PWS is often clinical. There are'nt any biological marker available in the market for diagnosing of PWS despite advance research in the field of molecular genetics.

CASE REPORT

A female patient, aged 10-years-old, came to Out patient department with complaints of sleep disturbances for 15 days, she wakes up in the middle of night and has difficulty going back to sleep. No history of disruption of daily activities due to sleep. Burning micturition for 7 days which was associated with increased frequency and urgency. Patient also complained of pain in abdomen for 2 days, localised to periumbilical region, mild to moderate in intensity, spasmodic in nature, associated with food intake and relieved on taking medication. Patient also had history of snoring since the age of 7-8 months of age.

Past history- No History of any previous hospitalisation. Patient was earlier diagnosed for Attention deficit hyperactive disorder (ADHD) and was taking treatment for the same but now patient is not taking any treatment for the past 3-years.

Family History- History of Anti-tubercular treatment (ATT) intake presents in mother for which she completed her treatment of 9 month. Patients mother also has a history of type-2 Diabetes mellitus (T2DM) for which she is currently on medication.

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Prenatal history– Patient was born at 7 months of gestational age due to premature rupture of membranes (PROM) by a normal vaginal delivery. Baby cried immediately after birth. history of delayed milestones present. (neck holding was present at 12 months of a

ENT evaluation (Figure 1 & 2) was done for complaints of snoring i/v/o adenoid hypertrophy. But ENT evaluation showed no signs of any adenoid hypertrophy. Patient was later subjected to overnight Polysomnography test. Which showed an apnea-hypoapnea index (AHI) of 6.6, which was diagnostic of mild OSA.

CONCLUSION

The relationship between PWS and sleep related breathing disorder (SRBD) is well established with OSA being the most common in such patients. A retrospective study showed that 76% patients with PWS has history of abnormal sleep.[3] A estimate of 70–80% of children with PWS have OSA,[5] contrast to 2–3% of all children globally.

Children prader willi syndrome are at increased risk of developing OSA due to coexisting factors like severe obesity and upper airway narrowing. Some other factors contributing to OSA are hypotonia, craniofacial abnormalities

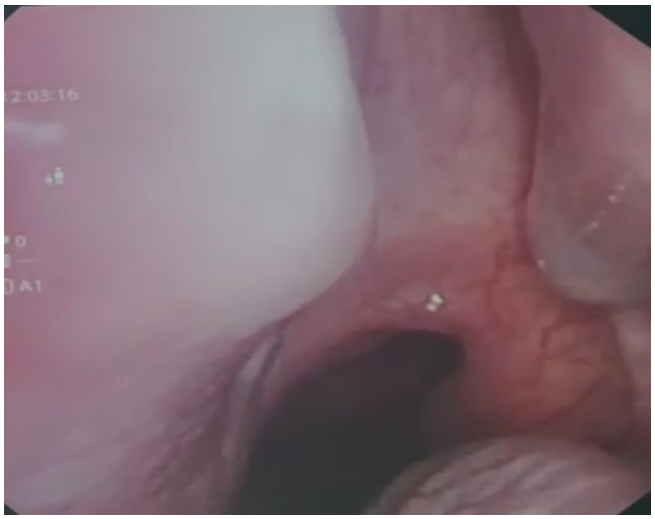

Figure 1: Endoscopy

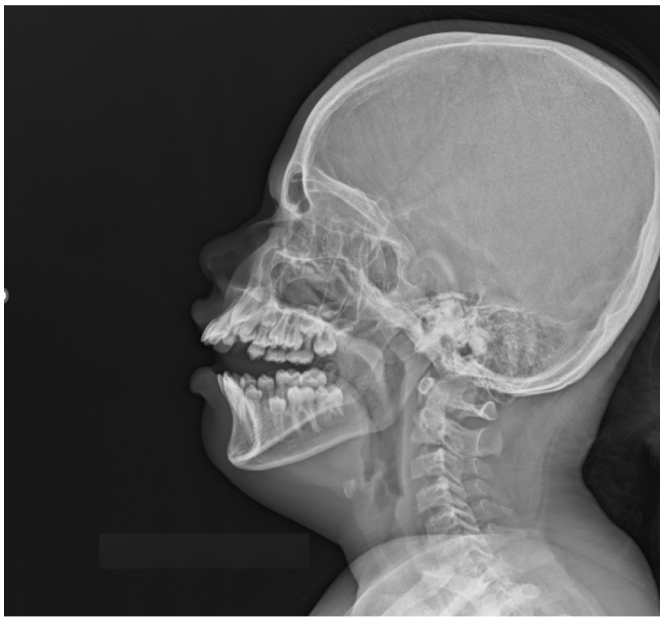

Figure 2: X ray - Lateral Nasopahynx

(high arched palate, micrognathia, etc), endocrine dysfunction, increased adeno-tonsillar tissue and abnormal ventilatory control (leading to hypoxia and hypercapnia). Polysomnography is a standard tool for diagnosis of OSA and it also helps in distinguishing OSA from central causes of sleep apnea. Symptoms can be subtle a can cause behavioural abnormalities Untreated OSA can worsen symptoms like cognitive impairement, behavioural, and metabolic health in PWS. Hence screening of patient is necessary before starting Growth hormone Therapy in all PWS patient.

Treatment of OSA helps in improving quality of life, cognition and behavioural symptoms. Positive airway pressure by CPAP devices (continuous positive airway pressure) is the mainstay of the treatment. Treatment may involve supportive management like weight reduction and sleep positioning. Some refractory cases may also need surgical interventions like adenotonsillectomy and growth hormone therapy. This case demonstrates the importance of screening patients of PWS with Polysomnography.

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