CASE REPORT

Laurence–Moon–Bardet–Biedl Syndrome and Obstructive Sleep Apnea

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ABSTRACT

Pediatric obstructive sleep apnea (OSA) often is underdiagnosed, particularly when associated with a syndrome when there are numerous and varied manifestations. However, a high index of suspicion and screening can help in early diagnosis and treatment for the same. We present a case of Laurence–Moon–Bardet–Biedl syndrome diagnosed since the age of 8 years presenting at the age of 16 years with severe OSA with good clinical recovery on therapeutic use of continuous positive airway pressure (CPAP).

Keywords: Laurence–Moon–Bardet–Biedl syndrome, Obstructive sleep apnea, Polysomnography, Positive airway pressure.

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INTRODUCTION

Laurence–Moon–Bardet–Biedl syndrome (LMBBS) is a rare autosomal recessive disorder characterized by short stature, ophthalmic manifestations including retinitis pigmentosa, polydactyly, obesity, mental retardation, and hypogonadism. Sleep-related breathing disorders in LMBBS are poorly understood and treated. We present a case of LMBBS diagnosed with severe OSA showing therapeutic response to continuous positive airway pressure (CPAP).

CASE DESCRIPTION

A 16-year-old boy was referred to the sleep clinic for complaints of excessive daytime sleepiness (EDS), tiredness, dull mentation, loud snoring, and irritable behavior. He was diagnosed with LMBBS. He had obesity, hypogonadism, nystagmus along with a history of developmental delay in childhood. He was the second child of a non-consanguineous marriage with no relevant family or sibling history. On examination, he had a neck circumference of 45 cm with Mallampati grade III on mouth opening (only soft palate seen and no uvula visible) as shown in [Figure 1](#). His body mass index was 31 kg/m². Epworth sleepiness score (ESS) was 18 out of 24 and STOP-BANG questionnaire satisfied six criteria (excluding hypertension and age which were otherwise also not relevant to the case). Thus, with EDS and a high-risk category for OSA in the aforementioned screening questionnaires, the patient underwent a diagnostic level I polysomnography ([Figs 2 and 3](#)), which revealed severe OSA with severe desaturations (minimum SO₂—79%). There was a rise in transcutaneous CO₂ by only 3 mm Hg thereby ruling out hypoventilation. A titration study with positive airway pressure was carried which, surprisingly revealed, abolishing all events at a CPAP of 6 cm H₂O ([Figs 4 and 5](#)). The patient was advised the same and had improvement in EDS, snoring with improved performance in school within 3 weeks.

DISCUSSION

LMBBS has originally been believed to be an amalgamation of Laurence–Moon syndrome (LMS) and Bardet–Biedl syndrome

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(BBS). Hypogonadism, pigmentary retinal degeneration, and some degree of mental retardation are common features, while spastic paraplegia is predominantly seen in LMS and polydactyly and obesity are often seen in BBS. However, in view of overlapping features, it is labeled as LMBBS.¹ Even though ophthalmic lesions (rod-cone dystrophy), polydactyly, obesity, learning disabilities, hypogonadism (males), and renal anomalies are primary features in the Beale's modified diagnostic criteria² for LMBBS, sleep-related breathing disorders (OSA) are not even included in the list of secondary features in this latest definition. Thus, OSA in LMBBS may remain unsuspected, undiagnosed, and untreated for several years affecting the physical and mental development of the child. The index case discussed earlier demonstrates the need to keep a high index of suspicion for OSA, diagnose it, and treat appropriately. Our case did not have any craniopharyngeal abnormalities requiring surgery. He had a therapeutic response in OSA by a simple CPAP and even demonstrated improvement in social and school performance.

CONCLUSION

LMBBS is a complex syndrome with varied manifestations. The OSA in LMBBS may be due to obesity alone (as in our case) or multifactorial including laryngeal web³ and craniofacial anatomical abnormalities. Thus, a multidisciplinary team for expert management of such patients is the key to favorable outcome.

Figs 1A and B: Lateral and front image of the patient showing cheek pad of fat and double chin with Mallampati grade III on mouth opening (only soft palate seen and no uvula visible)

Fig. 2: Hypnogram of diagnostic polysomnography revealing severe desaturations due to obstructive apneas and hypopneas

Fig. 3: A 10-minute window showing 20 epochs and numerous obstructive apnea and hypopnea with preserved respiratory efforts and falling saturation

Fig. 4: Hypnogram of titration polysomnography revealing easy titration with CPAP of 6 cm H₂O

Fig. 5: A 5-minute window showing 10 epochs with no residual sleep-related breathing disorders at a CPAP of 6 cm H₂O

CONTRIBUTIONS

All the six authors substantially contributed to the conception or design of the work; or the acquisition, analysis, or interpretation of data for the work; drafting the work or revising it critically for important intellectual content; final approval of the version to be published; and agreement to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

INFORMED CONSENT

Written informed consent was taken from the patient's parents.

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