

CASE REPORT

# Sleep Apnoea in Postaxial Acrofacial Dysostosis (Miller) Syndrome

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**Abstract**

We report a 13-year-old girl with postaxial acrofacial dysostosis (POADS) also called Miller Syndrome with sleep disordered breathing due to bilateral temporomandibular joint (TMJ) ankylosis and micrognathia/retrognathia, which was effectively treated with surgical correction.

**Keywords:** Acrofacial dysostosis, Sleep disordered breathing, Temporomandibular joint ankylosis.ecessary intervention should be done at the earliest to remove the causative factor.

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## Introduction

The acrofacial dysostoses (AFD) are a group of disorders characterised by defects in craniofacial and limb development. The craniofacial abnormalities include malar hypoplasia and micrognathia/retrognathia combined with variable limb malformations, which are “pre-axial” if located on the radial/tibial side and “post-axial” if located on the ulnar/fibular side. The preaxial form of AFD is called Nager AFD and the postaxial form is called Miller syndrome.<sup>1</sup> In this report, we discuss a case of postaxial acrofacial dysostosis (POAD) with obstructive sleep apnea, effectively treated by surgical correction of the craniofacial defect.

## Case Report

A 13 year old girl was referred for pre-operative evaluation prior to surgery for bilateral temporo-mandibular (TM) joint ankylosis. She had history of loud snoring,

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associated with choking episodes during sleep for the past 10 years. However she denied excessive daytime sleepiness. On examination we noticed micrognathia, retrognathia, malar hypoplasia, normally formed ears but with posterior angulation (Figure 1) and short left arm (Figure2) and small left fifth digit (Figure 3). Radiograph of both upper limbs showed shortening of the left humerus and ulna and bowing of radius (Figure 4). Hematological and biochemical investigations, chest roentgenogram, arterial blood gas analysis, 2–dimensional echocardiography and ultrasonography of abdomen revealed no abnormality. Computed tomography (CT) showed bony ankylosis of both TM joints with dysplasia of the mandibular condyles and coronoid processes. The patient was subjected to an overnight limited sleep study. The parameters recorded were airflow with oro-nasal flow sensors, snoring by microphone, oxygen saturation by pulse oximetry, thoracoabdominal movements and body position by sensors using Densa DMS 200 respiratory polysomnography (M/S Ferraris Medical Limited, U.K.). The study showed presence of both apnoea and hypopnoeas, which were obstructive in nature with an apnoea-hypopnoea index (AHI) of 10 per hour, loud snoring, frequent limb movements and associated oxygen desaturation to 69 percent (Figure 5). She underwent surgical correction of TM joint ankylosis for each side separately followed by mandibular advancement procedure. Sleep study repeated 26 months after the surgical procedure showed reduction in snoring and AHI

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(from 10 per hour to 1.2 per hour) while saturation remained above 90 percent throughout the night.



Fig 1: Photograph showing retrognathia and micrognathia



Fig 2: Photograph showing short left forearm.



Fig 3: Photograph of both hands showing short left fifth digit



Fig 4: Radiograph of forearm showing shortening of ulna with bowing of radius.

## Discussion

Miller Syndrome also called Genee-Wiedemann syndrome is a rare congenital autosomal recessive disorder.<sup>2</sup> Miller syndrome is characterized by facial features that are very similar to the more common Treacher-Collins syndrome, but with the addition of limb abnormalities not seen in that syndrome. It is also similar to Nager's syndrome, except that the latter has preaxial extremity defects instead of postaxial. The craniofacial abnormalities include malar hypoplasia, cleft palate, cup-shaped ears and lower lid ectropion. Micrognathia is the rule and may improve with age. Most have shortened forearms and radiologic evidence of ulnar hypoplasia, as seen in our case. In addition bilateral absence of the fifth digit, including the fifth metacarpal, or unilateral aplasia or hypoplasia of the fifth digit is observed frequently.

Temporomandibular joint ankylosis is known to cause sleep disordered breathing.<sup>3</sup> Airways obstruction and obstructive sleep apnea syndrome have been reported in Nager syndrome<sup>4</sup> and Treacher Collins Syndrome,<sup>5</sup> which are phenotypically similar to Miller syndrome. Although sleep apnea have not been previously reported in Miller syndrome, previous studies have shown that these patients have significant upper airway obstruction due to micrognathia.<sup>6</sup> Our patient had

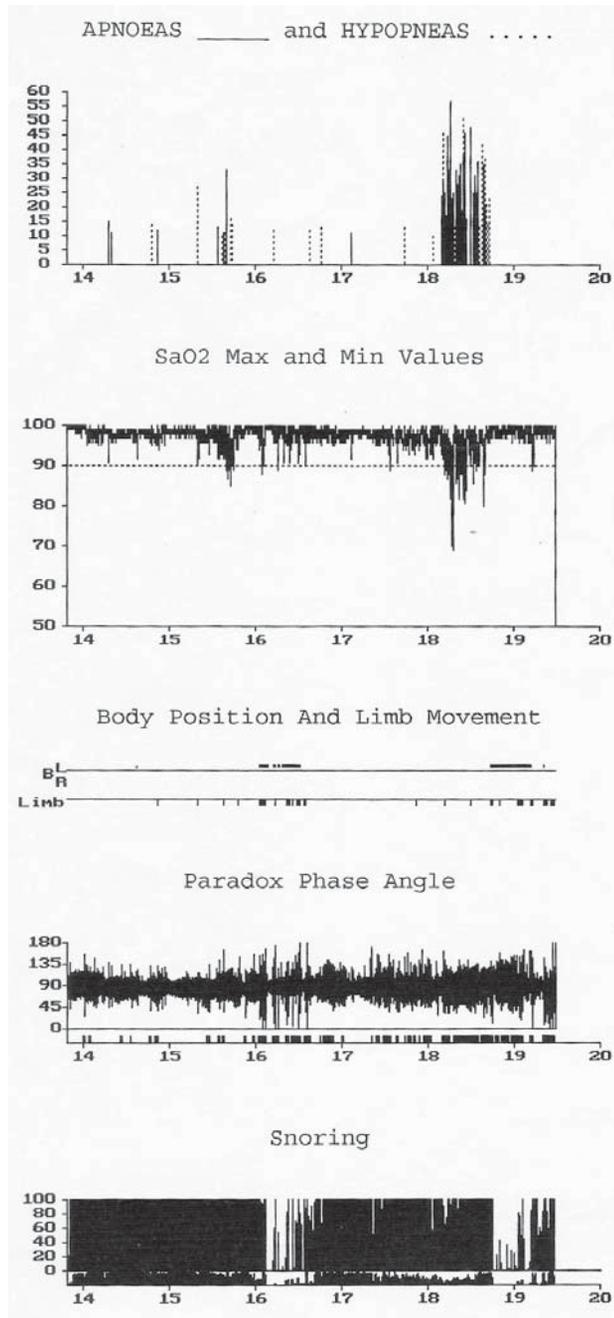


Fig 5: Limited polysomnography showing apnoea hypnoea index of 10 per hour with oxygen desaturation

temporomandibular joint ankylosis in addition to micrognathia and retrognathia which contributed to upper airway obstruction and obstructive sleep apnea. The obstructive sleep apnea was corrected following surgery.

In conclusion, sleep disordered breathing should always be considered in patients with acrofacial dysostosis as it is treatable with surgery.<sup>7</sup>

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