CASE REPORT

Obstructive sleep apnoea and arthrogryposis

Cindy Jon,¹ Ricardo Alberto Mosquera,¹ Sarah Mitchell,² Lynnette J Mazur³

¹Department of Pediatrics, Division of Pediatric Pulmonology, The University of Texas Health Science Center at Houston, Houston, Texas, USA

²Pediatric Sleep Center, Memorial Hermann Memorial City, Houston, Texas, USA
³Department of Pediatrics, The University of Texas Health Science Center at Houston, Houston, Texas, USA

Correspondence to

Dr Ricardo Alberto Mosquera, ricardo.a.mosquera@uth.tmc.edu

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SUMMARY

Arthrogryposis is a rare condition characterised by multiple congenital joint contractures. We present a case of a 10-year-old child with arthrogryposis and snoring. Polysomnography revealed significant obstructive sleep apnoea and hypoventilation that improved but did not completely resolve with adenotonsillectomy. With continuous positive airway pressure (CPAP) therapy, there was full resolution of all sleep disordered breathing. Initially, the patient admitted to difficulty tolerating nasal CPAP at home. However, she steadily improved adherence to therapy and admitted that with nasal CPAP use for the whole night, she felt more energised during the daytime.

airway pressure (CPAP) is considered an alternative therapy. As with adults, weight loss in obese patients may improve OSA.²

To date, there is limited published data regarding the prevalence and treatment of sleep apnoea in children with arthrogryposis. As such, when a child with arthrogryposis presents with typical sleep apnoea symptoms, there are no reference cases to assist the clinician towards the appropriate therapeutic interventions. We present a child with arthrogryposis with significant OSA that improved with T&A and completely resolved with nasal CPAP.

BACKGROUND

Arthrogryposis multiplex congenita is a non-progressive condition that is characterised by multiple joint contractures that are present at birth. Arthrogryposis is a clinical diagnosis which describes congenital contractures in two or more limbs. This rare condition stems from anomalous connective tissue, muscle, vascular and/or nerve development in utero leading to restricted joint mobility. In some cases, there may be associated pulmonary hypoplasia, poor neuromuscular tone and scoliosis. As such, patients have disabilities that affect activities of daily living.¹

The different types of arthrogryposis are distinguished by the degree of congenital contractures and neurological involvement. Patients with amyoplasia, or classic arthrogryposis, typically have internally rotated shoulders with extended elbows and flexed wrist as well as dislocated hips, extended knees and equinovarus foot contractures. Distal arthrogryposis spares proximal limb involvement and its subtypes may be associated with syndromes, such as Freeman-Sheldon, Gordon and Sheldon-Hall syndromes.¹

Obstructive sleep apnoea (OSA) prevalence is estimated to be present in 1–2% of healthy young children. History and examination findings alone cannot distinguish OSA from primary snoring. Polysomnography (PSG) identifies and quantitates sleep disturbances and is currently the standard method to diagnose OSA. Typically, apnoea hypopnoea index (AHI) >5/h is considered clinically significant.² In children, end-tidal CO₂ (ETCO₂) monitoring is important to detect sleep hypoventilation, which is defined as an ETCO₂ >50 Torr for >25% total sleep time (TST).³

In children, the first-line treatment for OSA is adenotonsillectomy (T&A). However, in patients who are not surgical candidates or have persistently elevated AHI after surgery, continuous positive

CASE PRESENTATION

A 10-year-old Hispanic girl with a history of arthrogryposis has been followed at Shriners Hospital for Children for joint contractures since 6 weeks of age. During consultation with the paediatrician for obesity, family disclosed a long history of snoring and apnoea with cyanosis during sleep that required stimulation. Her bedtime was 21:00 and she awakened at 5:00 feeling tired. However, she denied falling asleep at school or while watching television. She denied feeling tired or grumpy while awake.

Physical examination revealed contractures of the elbows and knees with equinovarus deformities of the feet (figure 1). She had an arm span of 150 cm (95th centile), weight 57.3 kg (>97th centile) and body mass index (BMI) 23 kg/m² (95th centile). Tonsils were mildly enlarged. No facial deformities or scoliosis were detected. She required a wheelchair and assistance with daily activities.

INVESTIGATIONS

The patient was referred to the Memorial Hermann Pediatric Sleep Center for diagnostic PSG, which revealed significant OSA with an AHI of 13.3/h and elevated ETCO₂ diagnostic of sleep hypoventilation (table 1).

She underwent T&A followed by another PSG which demonstrated persistent OSA with an AHI of 5.8/h; however, the hypoventilation resolved (table 1). Owing to persistent OSA, she had a CPAP titration PSG which resulted in a normal AHI of 1.7/h on CPAP of 5 cm H₂O via a nasal mask with heated humidification.

Spirometry (table 2) was performed in the clinic and showed a mild obstructive airflow pattern that did not improve after bronchodilator treatment. Since she was wheelchair user and had significant equinovarus abnormalities, height was measured using arm span.

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Figure 1 Arthrogryposis examination findings: contractures of the hands, elbows and knees with equinovarus deformities of the feet.

OUTCOME AND FOLLOW-UP

Although she initially had significant difficulty tolerating night nasal CPAP, she had improved adherence to therapy over time. Five months after initiation of therapy, the patient used nasal CPAP with heated humidification for five nights a week. When she is adherent to nasal CPAP therapy, she felt more energised and rested the next day. When asked, the patient stated that the nasal mask irritated her face. At the clinic visit, we provided

several alternative mask options until the patient was satisfied with the mask interface. Five months after the CPAP titration PSG, her weight increased by 4.5 to 61.8 kg. She was counselled regarding the morbidity associated with excessive weight gain, including worsening of sleep-disordered breathing.

DISCUSSION

We present a patient with baseline OSA that improved but did not resolve after T&A. Arthrogryposis may contribute to the residual sleep-disordered breathing due to possible association with pulmonary hypoplasia, scoliosis or poor neuromuscular tone.¹ However, it is difficult to isolate a specific cause since there are many phenotypic variations and disorders associated with arthrogryposis.

Kohyama and Shiiki⁴ reported two teenagers with Freeman-Sheldon syndrome who had OSA during rapid eye movement sleep only. However, Freeman-Sheldon is also associated with oropharyngeal abnormalities which may compromise the upper airway. Our patient had arthrogryposis without an associated syndrome, and her upper airway examination only revealed mildly enlarged tonsils.

Obesity may be another contributor to the patient's persistent OSA after T&A. In children, obesity is defined as BMI ≥ 95 th centile for the patient's corresponding age and gender. Obesity increases the risk of OSA by fourfold in children. Costa and Mitchell⁵ reported only half of their obese patients who underwent T&A had resolution of OSA, as defined by an AHI $< 5/h$. Fat deposition may lead to narrowing of the upper airway and thus residual OSA, even after T&A.⁶

Hypoventilation during sleep, as defined by $ETCO_2 > 50$ Torr for $> 25\%$ TST in children, may be secondary to upper airway obstruction, lung or chest wall disease, and neuromuscular disorder. Elevated CO_2 may lead to significant morbidities, such as heart failure, cor pulmonale and hypoxaemia.⁷ Marcus *et al* demonstrated that children are more likely to have partial airflow obstruction as increasing negative pressures were applied during sleep, when compared with adults who had a distinct critical closing pressure at approximately -20 cm H_2O . As such, children are more susceptible to hypoventilation and hypopnoeas (partial airflow obstruction) as opposed to adults who tend to have apnoeas (complete cessation of airflow).⁸

Our patient had multiple risk factors for hypoventilation, including enlarged tonsils, neuromuscular impairment from arthrogryposis and chest wall impairment due to obesity. We surmise that CO_2 monitoring during a PSG is important to detect sleep apnoea and hypoventilation in patients with arthrogryposis.

As the first-line of therapy for OSA in children, T&A improves the quality of life, behaviour and PSG findings. However, Marcus *et al*⁹ reported no improvement in attention or executive function using neuropsychological testing after T&A. In our patient, T&A improved PSG findings but did not lead to full resolution of OSA. CPAP is a second-line of therapy in children. As with adults, CPAP is an effective therapy but adherence to treatment is suboptimal. Optimisation of mask fitting and appropriate therapeutic pressures as well as addition of special features, such as heated humidification and slow ramping of pressures, may improve adherence.¹⁰

Given the multiple extremity contractures, we could not attain an accurate height which affected the predicted spirometry values. Regardless, the spirometric pattern revealed mild airflow obstruction that did not resolve with bronchodilator therapy. Interestingly, there was no restrictive pattern even with the multiple musculoskeletal abnormalities on examination and excessive weight.

Table 1 Summary of PSG data

Clinical information	First PSG Baseline diagnostic	Second PSG After adenotonsillectomy	Third PSG CPAP titration study
TST (min)	431.5	364	253.5
REM sleep (% TST)	16	19.5	13.8
N1 sleep (% TST)	0.9	3.9	1.7
N2 sleep (% TST)	40.6	38	25
N3 sleep (% TST)	41.2	33.3	30.8
Apnoeas			
Obstructive	23	13	0
Mixed	4	5	0
Central	6	3	6
Hypopnoeas	63	13	1
RERA	0	0	0
AHI (events per hour of TST)	13.3	5.8	1.7
Obstructive AHI (obstructive events per hour of TST)	12.5	4.8	0.2
End tidal CO_2	> 50 Torr for 30.7% TST	> 50 Torr for 0.1% TST	–
Oxygen saturation nadir (%)	79	86	95
Periodic limb movement			
Events	29	0	30
Index (events/h)	4	0	7.1

AHI, apnoea hypopnoea index; CPAP, continuous positive airway pressure; PSG, polysomnography; REM, rapid eye movement; RERA, respiratory effort-related arousal; TST, total sleep time.

Table 2 Spirometric measurements

Measurements	Predicted	Prebronchodilator		Postbronchodilator		% Change
		Actual	% Predicted	Actual	% Predicted	
FVC (L)	1.60	2.42	85	2.42	85	0
FEV1 (L)	1.43	1.64	63	1.68	65	3
FEV1/FVC (%)	89	68	74	69	76	2
FEF 25–75 (L/s)	1.83	1.32	43	1.32	43	0
PEF (L/s)	3.48	2.08	35	2.41	40	16

FEF 25–75, forced expiratory flow between 25% and 75% of vital capacity; FEV1, volume exhaled during the first second of an FVC manoeuvre; FVC, forced vital capacity; PEF, peak expiratory flow.

Regardless of the source of the airway obstruction resulting in OSA, T&A improved the AHI and CPAP led to resolution of residual OSA in our patient. In children with arthrogryposis and sleep disorders, PSG evaluation should be considered. If sleep-disordered breathing is detected, T&A and CPAP are appropriate therapies to treat the abnormalities. Close clinical evaluation of CPAP tolerance and appropriate mask fitting may improve adherence to therapy.

Learning points

- ▶ Arthrogryposis is a rare condition that presents with multiple musculoskeletal contractures due to anomalous connective tissue, muscle, vascular and/or nerve development in utero.
- ▶ Patients with arthrogryposis and snoring should be screened for sleep-disordered breathing with polysomnography, which includes CO₂ monitoring.
- ▶ Arthrogryposis may lead to obstructive sleep apnoea and sleep hypoventilation that may require multiple therapeutic interventions; therefore, serial clinical evaluations are warranted.
- ▶ Adenotonsillectomy and continuous positive airway pressure are effective treatments for obstructive sleep apnoea and sleep hypoventilation in children with arthrogryposis.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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