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Emergency tracheostomy in a patient with Melnick-Needles Syndrome and sleep apnoea

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Abstract
A six-year-old girl with a rare bone dysplasia (Melnick-Needles Syndrome) presented with a five month history of severe sleep apnoea, weight loss and failure of thrive. The syndrome is associated with craniofacial abnormalities, including micrognathia. Following a multi-disciplinary assessment an elective tracheostomy was considered the most appropriate treatment. The patient developed severe respiratory distress 10 days prior to the arranged date of surgery and required an emergency tracheostomy. This resulted in a dramatic return to health. The recognition of severe sleep apnoea in patients with craniofacial abnormalities and the role of initial tracheostomy are discussed.

Key words: Sleep apnoea syndromes; Tracheostomy; Osteochondrodysplasias, Melnick–Needles Syndrome

Introduction

In 1966 Melnick and Needles described a previously undiagnosed bone dysplasia. The syndrome is characterized by a diffuse bone dysplasia and abnormal facies. Diagnosis is based on clinical features with specific radiographic findings. The most striking changes occur in the long bones with bowing of the radius and tibia producing an S-shaped configuration. Sclerosis of the skull base and mastoid process are common along with hypoplastic changes in the mandibular coronoid process. The clinical signs include exophthalmos, full prominent cheeks, mild hypertelorism, micrognathia, large ears and misaligned teeth. Females are usually affected and the condition is inherited in an X-linked dominant pattern. The condition is lethal in males born to mothers with the syndrome (Donnenfeld *et al.*, 1987).

Previous reports of the condition have highlighted the obvious facial abnormalities including micrognathia without reference to sleep apnoea (Gorlin and Kneir, 1982; Van der Lely *et al.*, 1991). This case emphasizes the importance of the sleep apnoea syndrome when occurring in association with craniofacial abnormalities and the benefits of early tracheostomy.

Case report

A six-year-old female with Melnick–Needles Syndrome (MNS) presented with a five month history of progressive difficulty breathing at night, snoring and apnoeic attacks. She suffered from recurrent respiratory tract infections, day-time somnolence and was below the third centile for weight. The diagnosis of MNS was made at two years based on radiographic and clinical features.

Examination revealed severe micrognathia (Fig. 1), full prominent cheeks, exophthalmos and mild hypertelorism (Fig. 2). It was difficult to visualize the oropharynx. Examination under anaesthesia, following a difficult intubation, revealed normal sized unremarkable tonsils, a large floppy epiglottis and a normal larynx. Micrognathia was considered to be a major causative factor of her symptoms and she was referred for a plastic surgical opinion. In view of her age, any alteration to the mandible was considered unreasonable as this would interfere with mandible growth and dentition and ultimately worsen the situation.

A mini-sleep study demonstrated oxygen desaturation levels frequently below 80 per cent with greater than five apnoeic attacks during one hour of careful observation. The patient had a disturbed, restless sleep pattern associated with episodes of confusion. An elective tracheostomy was considered the most appropriate treatment because of the progressive nature and severity of her symptoms. Ten days prior to the arranged date of surgery she was admitted with severe dyspnoea and stertor and had emergency tracheostomy under general anaesthesia. She made a dramatic recovery and return to health. One year later she is tolerating a Size 3 Shiley tracheostomy® tube without difficulty and vocalizing well.

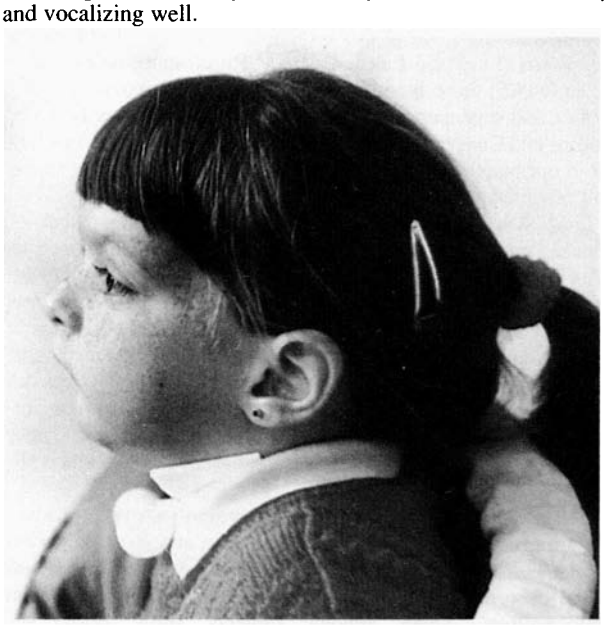

Fig 1

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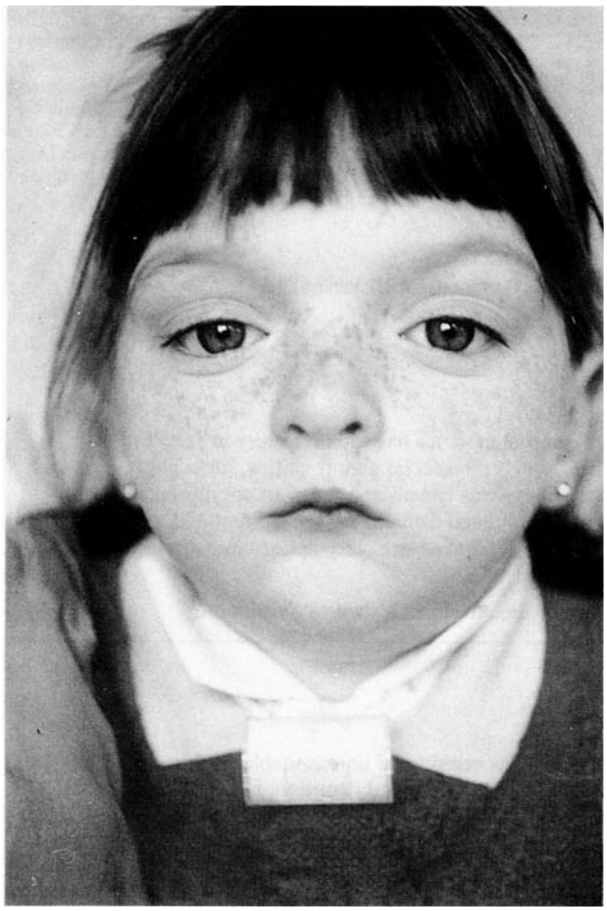

Fig 2

Discussion

Since 1966 twenty cases of Melnick–Needles Syndrome have appeared in the literature. Our case represents the first description of Obstructive Sleep Apnoea Syndrome (OSAS) in a patient suffering from the condition. In the vast majority of children with OSAS adeno-tonsillar hypertrophy plays a major aetiological role and removal of this lymphoid tissue usually improves symptoms (Lind and Lundell, 1982). Rarer abnormalities also cause OSAS; these include micrognathia, glossoptosis, macroglossia and severe craniofacial malformations, such as Treacher Collins and Crouzon Syndromes. Micrognathia was a major factor in our patient's symptoms which were aggravated by recurrent respiratory tract infections.

In OSAS, oxygen desaturation occurs with each episode of apnoea, producing hypoxaemia, eventually leading to respiratory failure, pulmonary hypertension, cor pulmonale and death (Kreiger *et al.*, 1987). Interestingly, Klint *et al.*, (1970) described a patient with MNS and with pulmonary hypertension without intrinsic cardiac defects. Wendler and Kellerer (1975) reported a case of MNS with right ventricular hypertrophy and overload. Melnick and Needles (1966) described a patient with cor pulmonale and respiratory failure in their original report. These patients may have suffered from unrecognized OSAS since all had severe micrognathia.

The importance of craniofacial abnormalities in the production of sleep apnoea is receiving increasing attention (Rojewski *et al.*, 1984; Lowe *et al.*, 1986). In a review by Powell *et al.* (1983) six per cent of OSAS cases had clear craniofacial abnormalities. Sleep apnoea subjects show posteriorly positioned maxilla and mandible, over-erupted maxillary and mandibular teeth, an anterior open bite and a posteriorly placed pharyngeal wall. The relative significance of each of these alterations in craniofacial form remains unclear. Surgical correction of abnormalities of the maxillofacial area are usually performed on adults. These procedures include mandibular advancement, surgical lysis of the tempero-mandibular joint and Le Fort 1 maxillary advancement osteotomy (Colmenero *et al.*, 1991).

Children with a combination of craniofacial abnormality and sleep apnoea are rare, and few people have extensive experience in their management. Le Fort 111 midface advancement has been used in these cases but no consensus has been reached as to the effectiveness of this procedure (Lauritzen *et al.*, 1986). Midface advancement to improve the size of the pharynx is ineffective unless the advanced position can be maintained post-operatively and this can be impossible in a child. Lauritzen experienced two fatalities of the seven patients he operated on. He believed these deaths would not have happened had they had a tracheostomy.

In our patient, the small tracheostomy tube is presently well tolerated and is not interfering with speech development. The potential risks of future difficult intubations has been circumvented.

In view of its minor morbidity, we recommend that tracheostomy be performed initially in children with craniofacial abnormalities and sleep apnoea. Further surgery can be delayed and performed at a later stage if necessary.

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