

CASE REPORT

Upper airways abnormalities and tracheal problems in Morquio's disease

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Morquio's disease is a metabolic disorder that can cause various respiratory abnormalities. Patients who live into adulthood are likely to develop upper airway problems and respiratory failure. With advances in home ventilation, these patients are increasingly likely to be referred to specialist respiratory units. We describe our experiences with two such patients.

Morquio's disease is caused by a deficiency of galactosamine-6-sulfate sulfatase which leads to a build up of keratin sulfate.^{1, 2} It is inherited in an autosomal recessive manner.² The disorder causes short stature, spinal deformity, odontoid hypoplasia, corneal opacities, cardiac abnormalities, hepatomegaly, acoustic deafness, and dental abnormalities.³ Mental retardation is not a feature of the condition. Life expectancy is normally less than 30 years but isolated cases of long survival have been documented.⁴⁻⁶

The respiratory problems in Morquio's disease can consist of a restrictive defect due to thoracic cage deformity,^{6, 7} upper airway obstruction during head flexion,⁵ or both. There have been isolated reports of sleep disordered breathing.⁸

We present two further patients with Morquio's disease who were both over 40 years of age and who developed problems with progressive type 2 respiratory failure. Tracheal problems encountered in their assessment for ongoing ventilation are discussed.

CASE REPORTS

Case 1

A 42 year old man with classical clinical features of Morquio's disease was admitted with progressive respiratory problems. He had an older sister who was reported to have had the same condition who had died with respiratory failure aged 42. He was of short stature and had marked thoracic, spinal, and limb abnormalities. Lateral radiographs of his cervical spine showed a rudimentary odontoid peg.

The patient was admitted for assessment. Arterial blood gas analysis confirmed type 2 respiratory failure. Lung function tests on admission indicated a mixed obstructive and restrictive defect. Formal sleep studies were not performed but he became deeply cyanosed and hypoxic during sleep and developed frequent position-independent apnoeas suggestive of significant upper airway obstruction. Radiography indicated a narrow trachea in all head positions and showed the trachea deviating posteriorly and to the right as it entered the thoracic inlet.

Initially the patient remained stable but in the third week after admission he began to deteriorate, in part due to a lower respiratory tract infection. Specialist respiratory review was sought and it was suggested that non-invasive ventilation was commenced and a tracheostomy considered. Attempts at CPAP ventilation were made but the patient was unable to tolerate this and his respiratory failure continued to worsen. ENT

advice was sought and a tracheostomy was considered but not attempted because of perceived technical difficulties with deviation and laxity of the trachea and major bronchi and the risk of spinal cord compression during a general anaesthetic. The patient deteriorated further and died 4 weeks after admission.

Necroscopic examination showed a narrowed laryngeal airway due to tissue thickening within the larynx. The subglottic trachea was stenosed and the more distal trachea and bronchi were crescent shaped due to flattening of the cartilage and stretching of the non-cartilaginous portion. Necroscopic tissue showed a marked deficiency of galactosamine-6-sulfate sulfatase consistent with Morquio's disease type A.

Case 2

A 44 year old man was referred with acute on chronic respiratory failure. Before admission he developed features consistent with a right sided pneumonia. He was intubated with a size 7 cuffed endotracheal tube and ventilated shortly after admission. The upper airway was crowded and narrowed but the intubation proceeded uneventfully. Due to a lack of intensive care beds he was transferred to another district general hospital on day 1.

He had the classical features of Morquio's disease and was of normal intellect. His younger sister who had similar features had died suddenly aged 10 years of presumed cardiac disease. No other family members were affected.

He responded well to initial antibiotic therapy but was slow to wean from the ventilator so a tracheostomy and CT scan were performed on day 7. A very narrow floppy trachea was noted and a size 5 tracheostomy tube was inserted. CT scanning showed the trachea to be globally reduced in size. On day 20 the tracheostomy tube was changed to a size 7. Bronchoscopic examination at this time showed gross swelling and erythema of the trachea, carina, and main bronchi. The right bronchial tree was twisted and distorted and all of the airways collapsed easily. ENT input was received but regional specialist respiratory opinion was not sought during this period. The airway problems were considered to be partly related to acute inflammation and no specific treatment was organised.

Weaning from the ventilator proved extremely difficult and progress was punctuated by ventilator associated infections. He was transferred back to the original unit on day 67. By day 86 he was stable on CPAP with a F_{iO_2} of 0.35. He was transferred to the regional centre for assessment for laryngotracheobronchial measures and long term ventilation. At the centre he received respiratory, ENT and neurological review. MRI scanning showed virtual closure of the oropharynx and a tiny tracheal diameter (fig 1). Tracheal stenting was considered technically possible when all airways inflammation had settled, but concern was raised about the laxity of the airways beyond the trachea. He returned to the original unit after 2 days at the regional centre but he developed another severe pneumonia and died during this episode. His parents refused permission for a necroscopic examination, having already previously refused removal of any tissue.

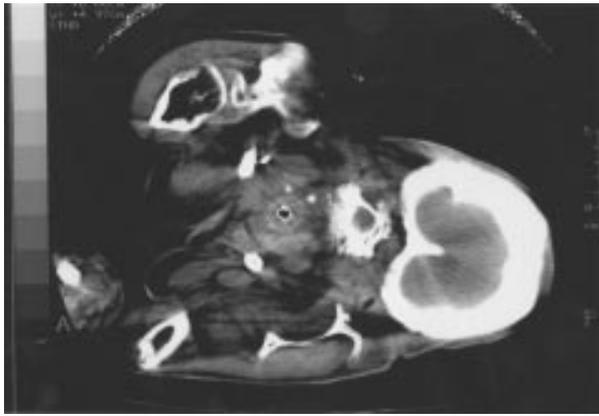


Figure 1 MRI scan image showing tracheal and upper airway narrowing by abnormal deposition of soft tissue.

DISCUSSION

There are now a number of case reports of patients with Morquio's disease living beyond 30 years.⁴⁻⁶ The cases presented here illustrate that, as life expectancy increases, these patients are likely to develop type 2 respiratory failure caused by the abnormalities of their thoracic cage.

The frequency of upper and large airway obstruction in Morquio's disease is variable.⁵⁻⁷ Both our patients had upper airway obstruction and marked tracheal abnormalities, independent of head position. The tracheal distortion seen in both patients is an unsurprising consequence of the shortened spinal height. Combined with the laxity of the tracheal tissue, this can lead to frequent total airway collapse. Tracheal stenting is one of a number of therapeutic options, although our experience shows that careful consideration should also be given to any abnormalities in the main bronchi before this is undertaken.

Both of our patients had abnormalities in the upper airway secondary to deposition of abnormal tissue. Combined with the relative inflexibility of the neck and the need to keep the cervical spine stable, this can make intubation very difficult.⁹

With increased survival time and current advances in home ventilation, respiratory specialists are increasingly seeing adult patients with Morquio's disease and similar metabolic disorders. Both patients described here illustrate a number of complex clinical problems that may be encountered. These are usually difficult to treat and, in this group of patients, it is imperative that such problems are anticipated and referral to a specialist centre made at an early stage.

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