CASE REPORT

Acute upper airway obstruction and emergency front of neck access in an achondroplastic patient

Craig James McCaffer,¹ Catriona Douglas,² Matthew H Wickham,² Gerard L Picozzi²¹Department of Otolaryngology and Head and Neck Surgery, NHS Lanarkshire, Airdrie, Lanarkshire, UK²Department of Ear Nose and Throat, Monklands District General Hospital, Lanarkshire, UK

Correspondence to

Craig James McCaffer, cmccaffer@nhs.net

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SUMMARY

Dwarfism is defined as a failure to attain a height of 148 cm in adulthood. Achondroplasia is the most common form of short-limbed dwarfism. Although this condition is relatively rare, with an incidence of 0.5–1.5 per 10 000 live births, most medical professionals will come across the achondroplastic dwarf (AD) during their career. Faulty endochondral ossification produces the characteristic short stature phenotype, as well as severe craniofacial, central nervous system, spinal, respiratory and cardiac anomalies. These unusual characteristics may present airway management difficulties in elective as well as emergency situations. Within the literature there is very little information regarding the emergency insertion of a surgical airway in an adult AD. We present our experience of this situation in the form of a case report and a review of the relevant literature.

BACKGROUND

Awareness of the anatomical abnormalities in achondroplasia that can cause severe upper airway compromise and how these can complicate its management are points that are looked at in this report.^{1–5}

CASE PRESENTATION

A 67-year-old woman measuring 133 cm in height and weighing 44.3 kg was admitted to a general medical receiving ward with a 3-day history of feeling generally unwell with poor mobility, diarrhoea and vomiting. Medical history included achondroplasia with kyphoscoliosis. She had no previous airway problems and had never had a general anaesthesia. Seven days into the admission, the patient developed stridor. Treatment with oxygen, nebulised epinephrine and intravenous steroids was initiated, and an ENT (ear, nose and throat) opinion was sought. Flexible laryngoscopy showed mild supraglottitis and no other significant source of stridor or airway compromise. The vocal cords were mobile and the subglottis appeared clear. The patient was transferred to an ENT theatre for further endoscopy and assessment.

TREATMENT

Despite ongoing medical management the patient quickly developed worsening stridor and airway compromise. This rapidly progressed to a hypoxic respiratory arrest, resulting in an asystolic cardiac arrest. The patient was successfully resuscitated by the cardiac arrest team. During resuscitation, it was noted that airway manoeuvres and simple adjuncts,

such as an oropharyngeal airway, were ineffective. A decision was made to achieve a definitive airway.

General anaesthesia was induced using a rapid sequence induction and cricoid pressure was applied. Laryngoscopy by a consultant anaesthetist revealed a Cormack and Lehane grade 4 view and tracheal intubation was unsuccessful. Rigid laryngoscopy was also unsuccessful with no view of the larynx again due to the patient's anatomy. Emergency surgical tracheostomy was then performed by the ENT consultant. During the tracheostomy, the patient's tracheal diameter was noted to be very small. Therefore, a 6 mm cuffed endotracheal tube (ET) was sutured in place.

OUTCOME AND FOLLOW-UP

One month post arrest, the patient was progressing well with an uncuffed, fenestrated tracheostomy tube. She was considered stable enough for decannulation. Full decannulation in recovery, with appropriate consultant ENT and anaesthetic support, was, however, unsuccessful. Re-insertion of the tracheostomy tube was required. Flexible endoscopic examination of the fenestrated tube identified that due to anatomical abnormalities, such as kyphoscoliosis, the fenestrations were flush with the tracheal wall and, therefore, normal breathing and speech were inhibited. This anatomical abnormality, along with the reduced vital capacity, meant that the safest option for the patient's airway was long-term tracheostomy.

DISCUSSION

Achondroplasia is the most common skeletal dysplasia,⁶ with an incidence of 0.5–1.5 per 10 000 live births.² There are an estimated 651 000 people with dwarfism in the world and achondroplasia accounts for approximately 70% of these cases. It is caused by mutations in fibroblast growth factor 3 (FGFR3).⁷ This gene normally codes for constitutively active growth factor receptor. As a result of this mutation, faulty endochondral ossification results,⁴ which produces the characteristic phenotype of short stature and short proximal limb segments.

The first step in the management of any critically ill patient is to establish an airway and administer supplementary oxygen.⁸ This is especially urgent in the context of acute airway compromise or respiratory arrest. If airway manoeuvres, adjuncts or endotracheal intubation attempts fail, a surgical airway is required, ideally a tracheostomy.

In achondroplastic dwarfs (ADs), abnormal bone growth leads to a disproportionate body and head structure, in addition to the presence of an

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unusually collapsible larynx, trachea and/or bronchi.⁹ ADs often have a relatively small airway, kyphoscoliosis, mid face hypoplasia and/or oromotor hypotonia.¹⁰ It would be reasonable to anticipate that these features may complicate an already difficult emergency airway and this case also seems to suggest this. Interestingly, however, a review of the anaesthetic literature suggests the contrary. Several studies have reported no difference in either obtaining a seal for mask ventilation or in orotracheal intubation in ADs.^{11–13} Despite these findings from the literature, we suggest that in critically unwell ADs, anticipation of airway difficulties due to these features should be considered early. This early anticipation may avoid progression towards potential respiratory compromise or respiratory arrest.

There are no studies or guidelines regarding emergency surgical methods of airway management in ADs. One case report,⁸ examining trauma in ADs, suggests following the Advanced Trauma and Life Support (ATLS) criteria when considering such procedures. This study also suggests sizing the ET or tracheostomy tube (TrT) in an AD child by basing the estimate on the child's weight. For an adult AD, a 7.0–7.5 mm TrT is suggested. In our case, a size 5.5–6.0 TrT was required. Therefore, it is clear that predicting the appropriate size of an airway tube in ADs is difficult.

ENT manifestations in achondroplasia are commonly otological or respiratory. It has been reported that patients with achondroplasia have a greater rate of hearing loss compared to patients with other skeletal deformities.¹⁴ With regard to respiratory problems in ADs, these are well described, ranging from upper to lower airway problems. In addition to their smaller stature, individuals with achondroplasia have been identified to have reduced vital capacity, which is thought to be due to decreased chest wall compliance. They have high rates of adenotonsillar hypertrophy with resultant obstructive sleep apnoea

(OSA) and may also have midface hypoplasia. The normal management of OSA is adenotonsillectomy, but a certain number of patients with OSA treated with adenotonsillectomy do not improve. Tracheostomy, the gold standard for severe upper airway obstruction and OSA, has been described in patients with achondroplasia.¹⁵ Midface distraction, based on the principle of distraction osteogenesis, has been shown to improve upper airway obstruction in two patients with achondroplasia who were tracheotomy dependent. Distraction of the midface by 25 mm in each case allowed for resolution of OSA and decannulation.¹⁶

In summary, it is clear that in emergency situations ADs pose significant airway challenges. Early anticipation of specific anatomical features in ADs and how these may complicate airway management is important. This awareness may avoid progression towards respiratory compromise or arrest.

Competing interests None.

Patient consent Obtained.

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Learning points

- Achondroplasia is the most common genetic form of dwarfism.
- The most common ENT (ear, nose and throat) manifestation in achondroplasia patients is conductive hearing loss.
- Respiratory problems are well described in patients with achondroplasia, spanning from upper to lower airway problems.
- There are a number of anatomical abnormalities in the achondroplasia patient that could result in upper airway problems—reduced vital capacity, adenotonsillar hypertrophy, midface hypoplasia and kyphoscoliosis.

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