Congenital Tracheal Stenosis in a Patient with Cleft Lip

Qureshi AM[1], Nebayosi T[1], Devani P[2]

1 – Aga Khan University Hospital, Nairobi
2 – Gertrude's Children's' Hospital, Nairobi

Correspondence to: Dr Ayman M Qureshi; P.O Box 76553 – 00508 Kenya. Email: aymanqm@yahoo.com

Key words: Congenital Tracheal Stenosis, Cleft Lip

Abstract

Congenital tracheal stenosis (CTS) is a rare condition characterised by different patterns of tracheal narrowing. The pathological hallmark is the presence of complete tracheal rings, with or without associated anomalies. We present a case of asymptomatic CTS in a patient with unilateral cleft lip. Tracheal stenosis was discovered incidentally immediately prior to surgery for correction of the cleft lip, when several attempts at intubation proved unsuccessful. A CT scan of the neck and proximal chest demonstrated a localised region of airway narrowing of the distal trachea, in addition to a right sided cleft lip without cleft palate. Management of CTS depends on several factors, with surgery being the definitive form of treatment. We review the pathology of CTS, its relationship with craniofacial defects, the role of radiology and management of this disease entity.

Introduction

The rarity of congenital tracheal stenosis has led to several isolated case reports, or long term case series from various institutions (1), and the epidemiology, therefore, is not known. Cleft lip with or without cleft palate are the commonest craniofacial birth defects, and indeed, amongst the most common of all birth defects with incidences of between 1 and 7 per 1000 (2). High familial aggregation rates, and higher rates in monozygous versus dizygous twins implicate a strong genetic component (3). Cleft lip and palate may present as an isolated abnormality (non – syndromic) or with other phenotypic anomalies (syndromic). There are no reports of Cleft lip/palate with tracheal stenosis in the literature. We present a case of a five month old infant with a complete left cleft lip, whose intubation proved futile prior to surgery for cleft lip repair. A subsequent CT revealed localised narrowing of the distal trachea. A review of the literature does not reveal a strict cut-off, below which a trachea is said to be stenotic. Rather, the radiological diagnosis is descriptive, referring to an obviously narrowed trachea (in the patterns described later) or secondary to an extrinsic compressing structure or lesion. The natural history of the disease, is for catch up growth of the trachea to occur. Surgery is only considered in symptomatic patients where tracheal grafts or tracheoplasty are performed.

Case Report

A two and a half month old female infant presented to the surgical clinic with a defect of the left upper lip which extended through the philtrum and alveolus, to terminate in the ipsilateral nasal ala. The patient had no other medical problems and was breastfeeding well. A diagnosis of complete left sided cleft lip and alveolus was made. Plan of management included a thorough paediatric assessment, dietician review, and a review for lip moulding appliance.

The child was born at home by spontaneous vaginal delivery at term, which was uneventful. Pre, intra and post-partum history was unremarkable, and birth weight was 2.3kg. She was the 3rd born, both siblings alive and well. There was no family history of orofacial clefting, or other congenital abnormalities.

Examination revealed an infant in fair general condition, who had no pallor or jaundice, was afebrile, well hydrated and weighed 4.3 kg. Respiratory rate was 22/min and the chest was clear. No respiratory distress or cyanosis was

present. Heart sounds were normal. The child was alert, with normal CNS findings. Normal external female genitalia were noted and on musculoskeletal examination, the limbs appeared normal.

Follow up at the surgical clinic revealed that the child continued to do well, with progressive weight gain, normal feeding and sleeping habits, and normal developmental milestones. A booking for elective surgery to correct the cleft lip and alveolus was made for when the child was 5 months of age.

On the day of surgery, pre-operative evaluation was normal. Following induction of anaesthesia, several attempts were made at intubation using size 4, 3.5, 3 and 2.5 endotracheal tubes. The larynx appeared normal, however there was resistance on trying to pass the tube into the trachea. After failed intubation, anaesthesia was reversed and surgery therefore cancelled. The infant recovered well from anaesthesia, and no complications were observed following this. A plan was made to do a CT scan of the neck, to include the proximal chest.

The scan was performed with IV contrast and demonstrated the left sided cleft lip, with involvement of the alveolus on this side. The maxilla and palatine bone were of normal structure. No other orofacial abnormalities were present. The mid portion of the trachea, at the thoracic inlet, was stenotic for a length of 2.5cm. Tracheal diameter of the involved segment was 3.5mm (a 50% stenosis compared to the normal proximal trachea.) No obstructing, or externally compressing lesions were present, and both pharynx and larynx appeared normal. The visualised heart, its great vessels and their respective branches all appeared normal. A conclusion was made of a segmental tracheal stenosis in a patient with a unilateral complete cleft lip.

Currently, the child is doing well and is on follow up at the surgical clinic. A follow up CT scan performed at 17 months of age, showed tracheal stenosis, without change in diameter or length. The child remains asymptomatic for tracheal stenosis, which is expected to improve with time. Following discussion with the paediatric cardiothoracic surgeon, a decision was made to repeat a scan after 1 year. If no increase in calibre of the stenotic segment is seen to a level safe for surgery, anaesthesia under laryngeal mask airway (LMA) or IV anaesthesia with airway protection will be considered for cleft lip surgery to be carried out.

Figure 1: Coronal reformatted CT showing mid tracheal stenosis

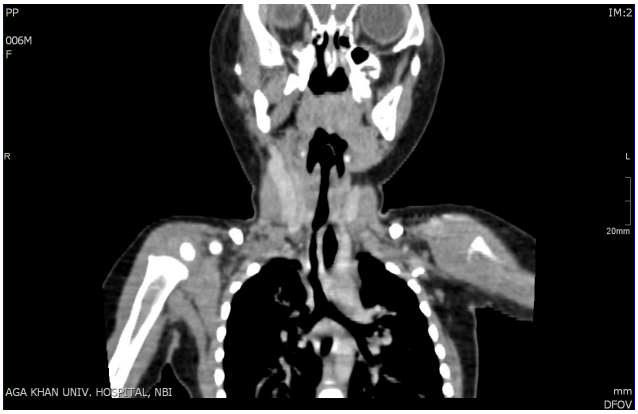
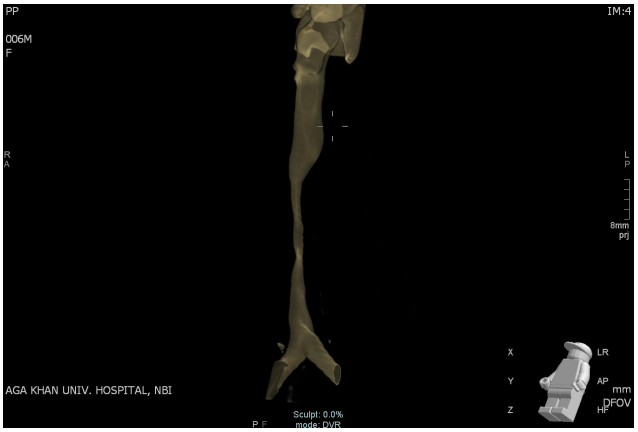


Figure 2: Volume rendered CT depicting tracheal stenosis



Discussion

Congenital tracheal stenosis (CTS) is a rare condition, characterised by narrowing of the trachea. Incidence of this condition is unknown due its rarity, with literature describing individual case reports, or long term experiences from medical institutions (1). Different patterns of stenoses exist: Type 1, generalised hypoplasia of the trachea, with sparing at the level of the cricoid and main bronchi constitute 30% of cases. Type 2, funnel type, results in gradual narrowing from the normal calibre cricoid cartilage to the carina, and make up 20% of cases. Type 3, accounting for half of all cases, and the variant seen in our case, demonstrates segmental stenosis of varying length and degrees (4, 5)

Anatomically, CTS is characterised by complete cartilaginous rings. The normal anatomical structure of the trachea comprises 16 – 20 incomplete cartilaginous rings, forming the anterior 2/3 of the trachea, which is completed posteriorly by elastic fibrous tissue and muscle fibres (trachealis muscle) (6). Associated malformations include vascular slings (commonly a pulmonary artery sling complex), trachea-oesophageal fistulae, pulmonary hypoplasia and Trisomy 21. The defect is thought to arise in utero during embryological development of the trachea. In the fourth week of development, an outgrowth in the ventral wall of the foregut, the laryngotracheal groove, progressively evaginates to form the laryngotracheal diverticulum. Longitudinal ridges/folds grow in from each side of the diverticulum and fuse in the midline forming the tracheo-oesophageal septum, which separates the laryngotracheal diverticulum from the dorsal aspect of the foregut, which will eventually form the oesophagus (7). Theories exist as to the embryological basis for tracheal stenosis. One such theory, proposed by Clements and Warner, postulates that a localised interruption in blood supply would cause stenosis of the trachea or bronchi, while normally perfused tissues would continue to grow(2).

Cleft lip, with or without cleft palate, represent the most common congenital craniofacial defects with incidences of 1 to 7 in 1000. Data from the WHO suggests lower rates of incidence in African populations (8). Complications associated with these conditions affect feeding, speech, hearing and psychological development, hence the need to repair the defects fairly early. Cleft lip and palate are classified into syndromic and non syndromic forms. Syndromic forms consist of 5 – 7% of all cases. Over 200 conditions are associated with the syndromic form of the disease, which is characterized by specific malformation patterns associated with other anomalies in addition to cleft lip/palate (9). No case of cleft lip with tracheal stenosis has been reported in literature.

The clinical presentation of tracheal stenosis depends on the degree of stenosis, age of the child and presence of associated anomalies. Most infants present within the first year of life with signs including biphasic stridor, tachypnoea, wheezing and even apnoea. These may be precipitated or aggravated by respiratory infections. In milder degrees of stenosis, infants may be asymptomatic, with symptoms occurring acutely following extubation after surgery for an associated anomaly. The onset of symptoms is attributable to mucosal oedema caused by the endotracheal tube, worsening the stenosis.

Imaging plays a key role in the diagnosis and evaluation of tracheal stenosis. Initial evaluation should include high kV frontal and lateral chest radiographs. These have the advantage of ready availability, negligible imaging time and invasiveness compared to modalities such as MRI and bronchoscopy, and lower radiation dose versus CT. The high kV technique allows adequate visualization of the trachea due to the high energy x-ray penetration though soft tissues, and particularly bone (spine, sternum, ribs) which are normally projected over this structure. CT and MRI of the chest are used to delineate anatomy, particularly detection of extrinsic causes of stenosis such as vascular slings. Rigid bronchoscopy is the gold standard of definitive diagnosis, with presence of complete cartilage rings the hallmark (10).This technique provides unparalleled spatial resolution in assessment of the airway lumen.

Management of congenital tracheal stenosis depends on the infants' clinical presentation. Asymptomatic patients may be managed conservatively, with chest physiotherapy, antibiotics to treat infections, and follow up. This group of patients continues to be managed expectantly until the age of 7 – 9 years where catch up growth of the trachea occurs (11). Symptomatic patients are usually managed surgically. Several surgical techniques exist. Short segment stenoses (less than 1/3 of the tracheal length) are amenable to resection and anastomosis. Longer segment stenoses are treated by several means. Tracheal grafts have been used to widen the airway following incision of the stenotic segment. Tissues used include costal cartilage, periosteum, pericardium and the oesophageal wall. This method has been associated with increased rates of restenosis, following scarring or granulation with re-epithelialisation (12). Slide tracheoplasty is a technique which widens tracheal calibre without the need for extensive mobilization, and results in less suture line tension. Most surgical repairs are currently carried out concurrently with cardiopulmonary bypass, which provides hemodynamic stability during surgery (13). Endoscopic procedures including dilatation and silicon stent placement are reserved for post-surgical complications such as stenosis and granulation tissue formation. Post surgical mortality is significantly increased in patients with co-existing intracardiac anomalies or secondary pulmonary diagnoses (14). Asymptomatic patients, like our case in point, do well on expectant management.

This case demonstrates an unusual presentation of a rare condition, and highlights the importance of a multidisciplinary approach to management of such patients.

References

1. Benjamin B, Pitkin J, Cohen D. Congenital tracheal stenosis. Ann Otol. 1981; 90:364–371.
2. Clements BS, Warner J. Pulmonary sequestration and related congenital bronchopulmonary-vasculture malformations: nomenclature and classification based on anatomical and embryological considerations. Thorax. 1987; 42:401 – 8.
3. P Stanier GE. Moore Genetics of cleft lip and palate: syndromic genes contribute to the incidence of non-syndromic clefts . Human Molecular Genetics. 2004;13(1):73–81.
4. Morris PJ, William C. Wood Oxford Textbook of Surgery 2nd edition. Oxford University Press. 2000;3:2604 – 2605.
5. Berrocal T, Madrid C, Novo S, et al. Congenital anomalies of the tracheobronchial tree, lung, and mediastinum. Embryology, radiology and pathology. Radiographics. 2004; 24(1):e17.
6. Standring, S. Gray's Anatomy 39th edition. Churchill Livingstone, Elsevier Chapter 62, Section 6 page 1060.
7. K. Moore, TVN Persaud. The developing human: clinically oriented embryology 8th edition. Saunders Elsevier. 2008. Chapter 10, page 198.
8. Mossey P, Castillia E. Global registry and database on craniofacial anomalies, World Health Organization, Geneva (2003) page 15 – 16. Retrieved on 25th July 2013 from http://www.who.int/genomics/anomalies/en/CFA-RegistryMeeting-2001.pdf.
9. Stuppia L, Capogreco M, Marzo G, et al. Genetics of Syndromic and Nonsyndromic Cleft Lip and Palate J Craniofac Surg. 2011; 22(5):1722-6.
10. Herrera P, Caldarone C, Forte V, et al. The current state of congenital tracheal stenosis. Pediatr Surg Int. 2007; 23(11):1033 – 44.
11. Cheng W, Manson DE, Forte V, et al. The role of conservative management in congenital tracheal stenosis: an evidence-based long term follow-up study. J Pediatr Surg. 2006; 41(7):1203–1207.
12. Cheng AT, Backer CL, Holinger LD, et al. Histopathologic changes after pericardial patch tracheoplasty. Arch Otolaryngol Head Neck Surg. 1997; 123(10):1069-72.
13. Chiu PP, Kim PC. Prognostic factors in the surgical treatment of congenital tracheal stenosis: a multicenter analysis of the literature. J Pediatr Surg. 2006 ; 41(1):221-5.
14. The Airway Reconstruction Team, The Hospital for Sick Children, Toronto, Ontario, Canada. Recent challenges in the management of congenital tracheal stenosis: an individualized approach J Pediatr Surg. 2005; 40(5):774-80.