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Unilateral choanal atresia in siblings – a rare occurrence

ABIR K. BHATTACHARYYA, M.S., F.R.C.S., VALERIE J. LUND, M.S., F.R.C.S., F.R.C.S.ED.

Abstract
The genetic aspects of choanal atresia have not been clearly defined though it is probably a multifactorial trait as in cleft lip and cleft palate. The appearance of the condition in both single and successive generations supports this contention.

Choanal atresia can occur as an isolated anomaly, but is more commonly associated with one or more concomitant congenital anomalies. In this report two sisters both in their teens presented with unilateral choanal atresia as an isolated anomaly. Endoscopic trans-nasal repair of choanal atresia was performed in both of them as this offers excellent visualisation and access.

Key words: Choanal atresia; Endoscopy; surgery, Sibling relations

Introduction

Atresia of the choanae was first described by Roederer in 1775 (Schwartz, 1942). Since then many comprehensive reviews of the embryology, clinical features, investigation and management have been published (McGovern, 1961; Evans and Maclachan, 1971; Pirsig, 1986; Brown, 1987; Crockett *et al.*, 1987; Morgan and Barley, 1990).

There have been fewer reports on its hereditary aspects. Choanal atresia has been reported in single and successive generations (Lang, 1912; Wright, 1922; Phelps, 1926; Ransome, 1964) but the genetic relationship is still poorly understood.

Most reports of multiple affected relatives have concerned siblings, most commonly in sisters (Grahne and Kaltiokallio, 1966). Although a familial incidence of choanal atresia must still be considered rare, with the increasing use of endoscopy in routine examination of the nose and postnasal space the suggestion that if it were looked for more often it might be found more frequently (Ransome, 1964) may be vindicated.

Case report

Two sisters aged 11 and 18, presented with right-sided nasal obstruction and nasal discharge. The nasal obstruction had been present since birth. As both sisters could breathe only through the left side they thought that this was 'normal' and did not seek any medical advice but the presence of unilateral mucopurulent nasal discharge eventually prompted referral. Nasal endoscopy with a 0° rigid Storz Hopkins endoscope revealed right-sided choanal atresia in both of them. They did not have any other congenital anomalies reported in association with choanal atresia. CHARGE - (C: Colobamatous blindness, H: Heart disease, A: Atresia of the choanae, R: Retarded growth and development including the central nervous system, G: Genital hypoplasia in males, E: Ear deformities including deafness) was specifically sought for and excluded.

On examination they did not have asymmetry of the facial skeleton, high arched palate or orthodontic anomalies. There was no family history of choanal atresia in their parents, grandparents or close relations.

Axial computed tomography (CT) scan revealed a membranous atresia of the right choanae in the younger child (Figure 1) and a bony atretic plate on the right side in the older one (Figure 2). Mucosal thickening of the right maxillary sinus and obstructed ostiomeatal complex was also present in this individual.

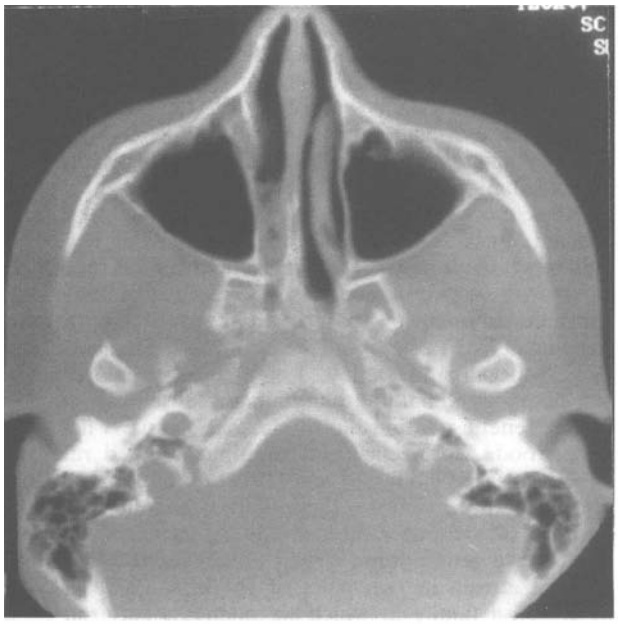

FIG. 1

Axial CT scan suggesting a largely membranous atresia of the right choana in the younger child. Secretions that have pooled in the back of the nasal cavity can often mask the thickness of the membranous atresia.

From the Professorial Department of Rhinology, The Royal National Throat, Nose and Ear Hospital, 330-332 Gray's Inn Road, London, UK.
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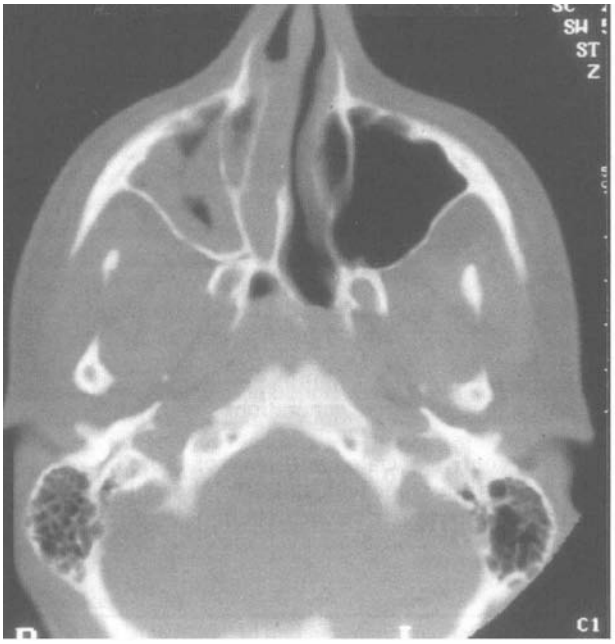

FIG. 2

Axial CT scan demonstrating a bony atresia of the right choana in the older girl, with associated thickening of the mucosa of the right maxillary sinus.

Both were treated by an endoscopic trans-nasal approach without stenting as described by Cumberworth *et al.* (1995), with specific attention to removal of the vomerine septum to achieve adequate permanent fenestration at one year follow-up.

Discussion

Choanal atresia was first described by Roederer in 1755 (Schwartz, 1942) and in 1829 Otto defined the anatomic anomaly (Otto, 1830). Unilateral atresia is more common than bilateral atresia in the proportion 3:2 (Kaplan, 1985), the incidence of bilateral choanal atresia being 5/7000 live births. Females appear to be affected twice as often as males (Maniglia and Goodwin, 1981). The right side is affected more often than the left. Although bilateral choanal atresia is a true medical emergency in a newborn, unilateral atresia may present much later. In this report the sisters were aged 11 and 18. The reports of associated congenital anomalies vary from 43 per cent to 72 per cent (Kaplan, 1985; Leclerc and Fearon, 1987; Duncan *et al.*, 1988; Morgan and Bailey, 1990), and are more often associated with bilateral than unilateral disease.

The genetic relationship of choanal atresia is not fully understood. Most reports of multiple affected relatives have concerned siblings. Lang (1912) noted right choanal atresia in three siblings, their mother and her aunt, an instance of extremely rare hereditary incidence in three consecutive generations. Wright (1922) reported bilateral choanal atresia in two pairs of children. Phelps (1926) described a family in which six members suffered from choanal atresia in two consecutive generations. Ransome (1964) found 12 families with two or more members affected in one of whom four of five siblings were affected. More recently Grahne and Kaltiokallio (1966) observed choanal atresia in sisters.

Choanal atresia is a feature of the CHARGE association. Posterior choanal abnormalities occurred in 56 per cent (28/50) in a comprehensive evaluation of otolaryngological abnormalities in 50 patients with CHARGE syndrome (Morgan *et al.*, 1993). Facial palsy, cleft palate and dysphagia are also commonly associated. Koletzko and Majewski (1984) besides validating the CHARGE association suggested the inclusion of orofacial clefts and oesophageal atresia in their report of six cases with choanal atresia. They also noted a certain degree of dysmorphism (low-set and dysplastic ears, retrogenia, antimongoloid slant of palpebral fissures and anteverted nares) in each of their six patients. Davenport *et al.* (1989) described 15 cases and concluded that CHARGE is a recognisable pattern of malformations and a true syndrome rather than an association. Interestingly external ear malformations and a 'wedge shaped audiogram' were unique features valuable in diagnosis. Congenital cardiac anomalies like tetralogy of Fallot, atrial septal defect, ventricular septal defect, parachute mitral valve, supravalvular and peripheral pulmonary stenosis have been reported in patients with CHARGE syndrome (Cyran *et al.*, 1987; Metlay, 1987). The presence of central nervous system malformations predominantly forebrain anomalies, particularly arrhinencephaly and holoprosencephaly have been strongly associated with choanal atresia (Lin *et al.*, 1990).

The diagnosis of choanal atresia may readily be made by a combination of nasal endoscopy and CT scanning. The latter will determine the extent of bony atresia and should be performed after careful preparation of the nose using vasoconstrictor drops and nasal suction to obtain optimum imaging. A child with secretions and neo-natal rhinitis may be misdiagnosed as having choanal atresia as secretions may pool in the back of the nasopharynx in the absence of adequate vasoconstriction (Morgan and Bailey, 1990).

Surgical management has been undertaken by four main approaches: (1) trans-nasal, (2) trans-palatal, (3) trans-septal and (4) trans-antral, of which the first two are in common use today. Pirsig (1986) reviewed more than 100 papers dealing with choanal atresia which revealed much controversy regarding the optimum approach (Table I). Although the trans-palatal route allows good access it is associated with a longer operating time, greater blood loss and longer convalescence than some of the other approaches and may lead to disruption of palatal growth. By contrast the trans-nasal approach preserves the developing hard palate and can be achieved in a shorter time with minimal blood loss. Morgan and Bailey in 1990 reported that the serious potential neurological complications of this approach could be minimised by visualizing the postnasal space with a 120° Hopkins Rod telescope from beneath and using a curved dilator.

A combination of intra-oral and trans-nasal approaches was described by Benjamin (1985) in which he alluded to the use of endoscopes, both in the diagnosis and treatment of the condition. In 1990, Stankiewicz advocated an entirely trans-nasal endoscopic fenestration of choanal atresia which he described in four cases, two of these required revision endoscopic surgery due to re-stenosis which he attributed to inadequate removal of the bony margins, both of the vomerine septum medially and the lateral wall. This has also been emphasised by Cumberworth *et al.* (1995) as essential to long-term success, an opinion which we would endorse.

TABLE I

SUCCESS RATES OF REPAIR OF CHOANAL ATRESIA USING DIFFERENT APPROACHES

1.	Trans-antral approach	80%
2.	Trans-palatal approach	84%
3.	Trans-septal approach	83%

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Conclusion

Choanal atresia is an uncommon condition in which familial incidence is itself rare. However, with the availability of routine outpatient endoscopy, partial and even unilateral atresia may be more frequently encountered and should certainly be excluded in any individual complaining of unilateral nasal obstruction at whatever age.

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Address for correspondence:
Professor Valerie J. Lund, M.S., F.R.C.S., F.R.C.S.Ed.,
Professorial Unit,
The Royal National Throat, Nose and Ear Hospital,
330–332 Gray's Inn Road,
London WC1X 8DA.