CASE REPORT

Neonatal lingual choristoma with thyroid hemiagenesis

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SUMMARY

A 45-day-old infant presented with choking spells and cyanosis. Examination revealed a lingual cyst. Contrast-enhanced CT confirmed the diagnosis of lingual cyst with incidental thyroid hemiagenesis. The child underwent excision of the lesion, which was reported as lingual choristoma.

BACKGROUND

The presence of respiratory and feeding problems in a neonate is an alarming issue. The presence of a mechanical obstruction as an aetiology is a surgical emergency warranting immediate relief of obstruction and prevention of impending complications. The various differential diagnoses causing mechanical obstruction are 10-fold, and careful diagnosis is a must, especially in a neonate. We present a 45-day-old infant with choking spells; and our management of a mechanical obstruction.

CASE PRESENTATION

A 45-day-old baby girl presented to our outpatient department with choking spells and blood stained frothing from the oral cavity. She was born as a second child to her parents at term as a low birth weight baby (2½ kgs). Choking spells were present on crying. Her mother had pregnancy-induced hypertension and had needed a caesarean section to deliver the baby at term due to her uncontrolled hypertension. The baby cried immediately after birth and her sucking reflex was good. Following birth, the baby had several episodes of breathlessness and bluish discolouration of the face on crying, and was referred from a rural health centre to our tertiary care centre. There was no history of fever, difficulty swallowing or excessive crying.

On examination, the baby's heart rate was 170/min and respiratory rate was 66/min. Preductal SpO₂ was 76% and postductal SpO₂ was 85%. Examination of the oral cavity revealed a 1×1 cm cystic lesion over the base of the tongue with prominent blood vessels over its surface (figure 1). The rest of the local examination was unremarkable. Systemic examination revealed reduced breath sounds on the right side of the chest and bilateral fine basal crepitations with subcostal retraction. The rest of the systemic examination was unremarkable.

INVESTIGATIONS

Contrast-enhanced CT revealed a 9×8×10 mm thin walled, well-defined, smooth, vertically oval cystic lesion at the midline in the base of the

Figure 1 1×1 cm cystic lesion over the base of the tongue with prominent blood vessels over its surface.

tongue, without any obvious contrast enhancement. Anteriorly, the cystic lesion was insinuating in the midline base of the tongue and posteriorly it was abutting the lingual surface of the epiglottis (figures 2 and 3). Incidentally, there was absence of the left lobe of the thyroid and isthmus (figures 4 and 5). Neonatal thyroid stimulating hormone (TSH) was normal.

DIFFERENTIAL DIAGNOSIS

- Mucocoele
- Lingual thyroid
- Lymphoepithelial cyst
- Haemangioma
- Venolymph malformation
- Salivary tumour
- Melanotic neuroectodermal tumour of infancy
- Dermoid
- Epidermoid
- Teratoma
- Epulis
- Cystic hygroma

TREATMENT

The neonate underwent excision of the cyst under general anaesthesia. Following excision, her airway was maintained for 24 h and feeds were initiated. The cyst was sent for histopathological examination.

OUTCOME AND FOLLOW-UP

Histopathological examination revealed a thin-walled cyst lined by pseudostratified ciliated

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Figure 2 Axial CT scan showing the cystic mass in the midline.

columnar epithelium and areas of squamous epithelium (figure 6). There was also evidence of mucous glands and skeletal muscle bundles in the cyst wall (figure 7), thereby confirming the diagnosis of a lingual choristoma with thyroid

Figure 3 Sagittal CT scan showing the cystic mass embedded in the posterior one-third of the tongue.

Figure 4 Contrast-enhanced axial CT scan showing absent left thyroid lobe.

hemigenesis. The child followed up 1-week later and was completely free of the symptoms. Postoperatively, thyroid function tests were normal.

DISCUSSION

Choristoma is defined as the proliferation of normal tissue at an abnormal site. It denotes cystic benign masses usually lined by upper aerodigestive epithelial cells.^{1 2 3 4} Hamartoma is another entity that denotes the abnormal growth of normal tissue.^{1 3} Hamartomas are usually haemangiomas and lymphangiomas, whereas choristomas can be glial, cartilaginous, osseous, sebaceous, thyroid or glandular in origin or they may simply be proliferation of respiratory or gastrointestinal mucosa.^{1 2} Various terminologies have been used in the literature to describe lingual choristomas, such as anterior median lingual cyst, lingual bronchogenic cyst, median lingual cyst, foregut duplication cyst,

Figure 5 Contrast-enhanced coronal CT scan showing absent left thyroid lobe.

Figure 6 H&E staining showing a thin-walled cyst lined by pseudostratified ciliated columnar epithelium and areas of squamous epithelium.

enterogenous cyst, oral alimentary cyst and gastric mucosal choristoma.^{1 5} However, Manor *et al*⁶ suggested that the term lingual cyst of epithelial origin is best used to denote a lingual choristoma to avoid implications in the aetiology.

Lingual choristoma is seen in newborn and ranges up to the fourth decade. It is common in early childhood and the literature places the mean age at 5½ years and median age at 6 months. The male to female sex ratio is 1.6:1.^{1 2 7 8} Average cyst size range is 1 to 6.5 cm with a mean of 2.3 cm.^{1 2 7} The commonest site for a lingual choristoma is the anterior two-thirds of the tongue and floor of the mouth. Rarer sites include the posterior one-third of the tongue, pharynx and hypopharynx.^{1 2 3} The existence of other coexisting congenital anomalies with lingual choristomas has not been reported in the literature,¹ however, in our case we found thyroid hemiagenesis as an incidental finding coexisting with a lingual choristoma. Thyroid hemiagenesis is usually diagnosed during assessment for another clinical condition, and the aetiopathology and development of hemiagenetic thyroid, and their impact on hormone function, and presence of other thyroid disorders, is highly debatable, as noted in the literature. The prevalence of thyroid hemiagenesis is 0.2–0.25%; it occurs commonly in the female population with predominance in the left lobe of the thyroid. Thyroid hemiagenesis is predominantly euthyroid at birth but may present in a hyperthyroid state. Constant overstimulation of the remaining lobe by TSH may produce compensatory

Figure 7 H&E staining showing mucous glands and skeletal muscle bundles in the cyst wall.

hypertrophy with progression of age, and may be associated with other thyroid disorders such as cyst, goitre, well-differentiated carcinoma and autoimmune pathology, warranting close follow-up.^{9 10}

The aetiopathogenesis of a lingual choristoma is based on two theories. The developmental theory is based on the fact that pluripotent stem cells of the first and third pharyngeal arches are responsible for the development of the tongue, and foramen caecum is the site of development of thyroid. It has been postulated that remnants of undescended thyroid or heterotopic epithelial cells of the respiratory and digestive tract can get embedded in the developing tongue and predispose to the development of a choristoma. The reactive theory suggests that constant irritation and inflammation of the posterior lingual surface can predispose to the development of a choristoma.^{1 2 5 7 8}

In our case, we had the development of a choristoma in the posterior one-third of the tongue and thyroid hemiagenesis. The coexistence of both can be based on the developmental theory, but the absence of thyroid tissue in the cyst still questions the theory.

On microscopy, there is presence of hyperkeratotic, basal hyperpigmentation and hypergranulosis in the cyst wall that replaces the normal oral stratified squamous epithelium. There is also presence of submucosal sweat glands, hair follicles, sebaceous glands and adnexal structures depending on the tissue of origin of the choristoma. Absence of a true cyst wall with presence of columnar epithelium with mucus and fundic glands is not uncommon.^{2 4}

Clinical presentation varies depending on the size of the choristoma and age. In the newborn, the most common presentation is impaired feeding and choking spells. In children and adults, presentation is usually in the form of an asymptomatic lump in the throat, dysphagia, gagging, nausea and throat irritation.^{1 3 7 11} Clinical examination usually reveals a mass at the site of lesion. It is, however, difficult to identify the tissue of origin on examination of the mass.

MRI is more specific than CT scan in the imaging of a lingual choristoma. On T1-weighted imaging there is an impaired intensity (due to protein, infection, blood or mucus) and T2-weighted imaging shows a hyperintensity. Dermoid, lymphatic malformation and choristoma usually form a radiological differential diagnosis with such findings.^{3 4 11} Histopathological examination clinches the diagnosis and immunohistochemistry can be carried out to pinpoint the tissue of origin.^{11 12}

Surgical excision is the mainstay of treatment. Needle aspiration has been known to produce recurrence. Care has to be taken to maintain the airway postoperatively due to presence of oedema and possible airway obstruction. Normal diet can be resumed 1–3 days following the operative procedure.^{1 3 7 8 11 12}

Complications of choristomas are extremely rare. Malignant transformation in a long-standing cyst in an adult has been reported in the literature. Gastric or intestinal mucosa as tissue of origin can produce ulceration and bleeding, and long-

Learning points

- ▶ Choristomas are defined as the proliferation of normal tissue at an abnormal site.
- ▶ MRI is more specific than CT scan in the imaging of a lingual choristoma.
- ▶ Surgical excision is the mainstay of treatment. Needle aspiration has been known to produce recurrence.

standing presence of mucus glands in choristomas can induce fistula formation. It has been observed that there is no evidence of recurrence when a choristoma is completely removed.^{1 3 8 11}

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