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Congenital absent epiglottis causing aspiration in infant

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Abstract

Congenital absent epiglottis is a rare phenomenon which has variable clinical presentation. Most of the cases are asymptomatic and go unnoticed to the adulthood. Some neonates present with stridor and respiratory distress and sometimes the absence give rise to obstructive sleep apnoea features. Further some infants with absent epiglottis may present with significant aspiration of feeds as well as own secretions which occasionally may give rise to life threatening chest infections and growth retardation.

Management of aspirating infant is by swallowing therapy, altering the enteral feeding route (Nasogastric tube or percutaneous endoscopic gastrostomy (PEG)) or in rare occasions may require tracheostomy and supraglottic closure. The case discussed here is of a infant with aspiration detected to have absent epiglottis managed by swallowing therapy and PEG insertion.

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Introduction

Swallowing and laryngeal competence is a complex process which involve multiple sensory inputs processing and effector mechanisms to prevent laryngeal penetration and aspiration. Aspiration in newborns and infants are caused by variety of factors. A common association being gastro oesophageal reflux. Cricopharyngeal dysfunction with increased tone in upper oesophageal sphincter as well as neuromuscular incoordination have all been considered as causes. Folding back of epiglottis with laryngeal elevation is considered a significant component in prevention of aspiration.

Congenital absence of epiglottis is an uncommon occurrence, and most cases go unnoticed to adulthood. Rarely this will result in gross aspiration of feeds in infants which has adverse outcomes with recurrent chest infections and associated morbidity.

Case Report

A 2 months old baby boy was referred to the Multi Disciplinary Cleft Clinic (MDCC) by the paediatrician with a suspected sub mucosal cleft palate. This baby was the second born in the family with parents having a history of consanguinity. Child was born by normal vaginal delivery with unremarkable perinatal period. Birth weight was 2.27kg. The mother noticed the child was showing some discomfort when being fed at the first month and sought medical advice, which eventually led her to the MDCC. The child was evaluated and the palatal abnormality was confirmed clinically as a high arched palate. Since the mother gave a history suggestive of episodic aspiration, child was referred for further evaluation by ENT team and Speech and language therapy. There was no history of stridor or stertor nor features to suggest Pierre Robin Sequence.

Fibreoptic nasal endoscopy was performed as a part of the evaluation, which showed normal nasal cavities, choanae, and nasopharynx as well as oropharynx. Larynx showed the abnormality of absent epiglottis with normal vocal cord motion. Aspiration of saliva was noticeable associated with pooling. The mother was explained about the condition and advised regarding the risk of aspiration with oral feeding and suggested nasogastric tube feeding. With the parental consent a nasogastric (NG) tube was inserted and trained the parents for tube feeding and care. Speech therapist also got involved and advised the mother further regarding the aspiration reduction and NG feeding. Clinical evaluation of swallowing by the speech therapist revealed, that the baby gets cough when saliva is collected in oral phase with high risk of aspiration.

The child was also diagnosed with hypothyroidism and endocrine team was referred accordingly. Thyroxine therapy was commenced, which is continued to date. The genetics counselling also was done but the parents were reluctant for genetic studies since they do not intend to have another child.

Child was scheduled for a microlaryngobronchoscopy to further visualization of the upper aero digestive tract. This revealed an absent epiglottis with no laryngeal cleft (Figure 1). Bilateral vocal cords showed normal motion with breathing. Subglottic larynx and trachea up to the carina was normal.

Child was started on oral proton pump inhibitors and domperidone as anti-reflux therapy to reduce aspiration of acidic stomach contents. Respiratory and paediatrics teams reviewed the baby and commenced on prophylactic antibiotics to minimize the risk of aspiration pneumonia.

Videofluroscopic swallowing study (VFSS) was performed which confirmed aspiration for thin fluids with maximum Penetration aspiration score (PAS) was 7 for liquids.

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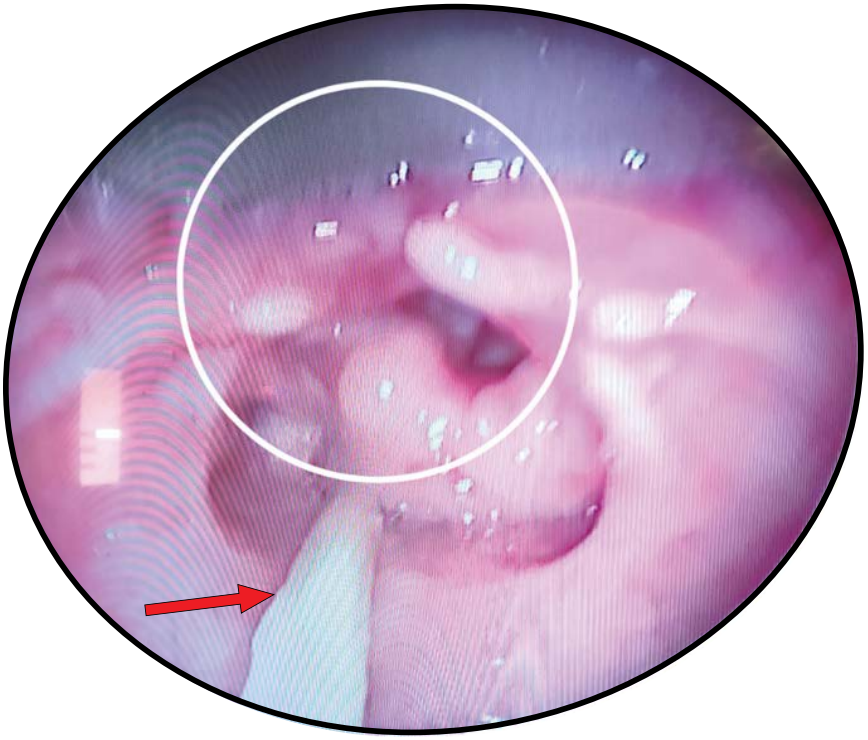

Figure 1: Endoscopic view of larynx with absent epiglottis. (marked by arrow is the nasogastric tube)

Later due to poor improvement of swallowing reflexes the child underwent percutaneous endoscopic gastrostomy (PEG) for enteral feeding. There were no complications from this procedure and the child continues to be fed via PEG. The child`s weight gain was initially poor currently gradual improvement is present. The baby boy is continued to be followed up at Lady Ridgeway Hospital for children for hypothyroidism and PEG tube care.

Discussion

Epiglottis is a component in supraglottic larynx developing from third and fourth branchial arches cranial to the embryonal laryngeal orifice during the 5th week of intrauterine life[1]. Absence of epiglottis is a rare phenomenon with most of the cases having an association with Pierre Robin Sequence [2]. The function of the epiglottis is debated.

Physiological studies have revealed the backward folding of epiglottis during the laryngeal elevation of swallowing plays a role in laryngeal competence. This occurs in association with vocal cord adduction and due to the structure of the epiglottis liquid bolus is directed towards pyriform sinuses aiding the swallowing process. Yet one study has shown dogs with surgically removed epiglottis swallow without difficulty contrary to the others[3].

The clinical presentation of congenitally absent epiglottis is quite variable. Some cases present with stridor with features suggestive of obstructive sleep apnoea[3]. Some reported cases of patients go unnoticed to the adulthood being completely asymptomatic and incidentally diagnosed with absent epiglottis. Further some cases present with gross aspiration as the index case.

The severity of the pathology depends on the dysfunction it produces, and management must be decided accordingly. Some neonates present with severe stridor requiring acute airway management. Aspirating infants must be evaluated and managed accordingly as is discussed in the index case.

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Aspiration can be diagnosed by clinical history and performance of bed side evaluations. Further confirmation can be done using instrumental assessments of swallowing including FEES (Fibreoptic endoscopic evaluation of swallowing) or VFSS (Videofluroscopic swallowing study). These are carried out in collaboration with the speech and language therapist. Both the studies have pros and cons and, in certain cases, can be complementary to one another. These also aid in management by identifying the suitability for consistency modifications and improvements by various manoeuvres.

The management options for aspirating infant must be commenced early since recurrent aspiration can result in infective pneumonias which can be a threat to life as well as resulting in poor growth. There can be long term sequalae in the lungs because of recurrent aspiration as well. Accordingly, the child should be commenced on altered route of enteral feeding initially with a NG tube which can later be converted to a PEG tube for longer term management. Prevention of reflux also plays an important role since aspiration of highly acidic gastric contents will result in further lung injury. Certain cases of severe aspiration may warrant surgical interventions [4].

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