Case Report

Bifid epiglottis with midline laryngeal hamartoma: a rare presentation of Pallister Hall syndrome

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ABSTRACT

Bifid epiglottis is a rare laryngeal anomaly that can be an isolated occurrence or as part of malformation syndromes, commonly the Pallister Hall syndrome (PHS). PHS presents with hypothalamic hamartomas, polysyndactyly, bifid epiglottis, imperforate anus and genitourinary abnormalities. We report a case of a 22 months old male child with features of PHS and Hirschsprung's disease. The presentations are with a hypothalamic mass, a bifid epiglottis and a midline mass in the larynx with symptoms of respiratory distress. The laryngeal lesion was endoscopically excised and its histology confirmed as hamartoma (laryngeal). The importance of an early diagnosis of PHS, genetic counselling and prompt management of a compromised airway and endocrine abnormalities is emphasized to have a better outcome.

Keywords: Bifid epiglottis, Pallister Hall syndrome, Laryngeal hamartoma

INTRODUCTION

Bifid epiglottis - a midline-cleft of the epiglottis, commonly presents as an isolated anomaly and can also be a part of complex malformation syndromes.¹ Common occurrence in Pallister–Hall syndrome (PHS) is because, in the embryo-the epiglottis, hypothalamus, and digital buds develop synchronously. Disturbances during this stage of development could account for the concurrence of bifid epiglottis, hypothalamic hamartoma, and polysyndactyly. However, bifid epiglottis with a laryngeal hamartoma presenting with airway compromise has not yet been described in PHS patients.

CASE REPORT

We report here a 22 months old male child with bifid epiglottis and a midline Epiglottic mass. The baby was born at 39 weeks to healthy, non-consanguineous parents. The length of the baby at birth was 45 cm; birth weight was 3000 g, and head circumference was 35.0 cm. From second day of birth baby presented with failure to pass stools, non-bilious vomiting and abdominal distention. The baby was clinically diagnosed to have Hirschsprung's disease. He had short digits with postaxial polysyndactyly of both hands and feet and type 3 syndactyly of the left hand. The distal phalanges of the hands were short with hypoplastic finger-nails (Figure 1b and 1c). The baby presented with dysmorphic facial features like frontal bossing, epicanthal eye folds, low set ears and upturned nose (Figure 1a). He also had duplicate anus and microphallus. Echocardiogram showed a small patent ductus arteriosus. The child underwent colostomy on day four of life. Anesthesia notes did not mention the upper airway abnormalities, and naso tracheal intubation was uneventful. Screening brainstem eoked
response audiometry showed no hearing in the left ear. Chromosomal analysis revealed 46 X, Y karyotype without evidence of cytogenetic abnormalities. Family history was noncontributory except for history of polydactyly in third degree relatives on the paternal side. In the next 12 months, the child was asymptomatic except for rare instances of aspiration while having liquids. There was associated progressive noisy breathing during sleep and upper respiratory infections. A pull through procedure (for the imperforate anus) and colostomy closure was planned when he was one and a half years old.

At intubation during this intervention, the anaesthesiologist noted a significant epiglottic mass blocking 80% of the supraglottic space. Transnasal flexible laryngoscopy by an ear, nose and throat (ENT) specialist showed a bifid epiglottis with a sessile midline mass between the two leaflets of the epiglottis. The posterior glottis, vocal cord mobility, subglottis and the lower airways were normal. The planned abdominal surgery was abandoned and the child was intubated with a small size endotracheal tube (ETT) to perform magnetic resonance imaging (MRI) of the neck and brain was done. The MRI showed an iso-dense non-contrast enhancing lesion in the supraglottis and a suprasellar hamartoma (Figure 2). The diagnosis of PHS was made in view of the classical findings of bifid epiglottis, polysyndactyly and hypothalamic hamartoma. Neurosurgical colleagues advised observation for the hypothalamic hamartoma as the child had no history of seizures or signs of space occupying lesion. Endocrinology opinion was sought to rule out hypo-pituitarism, but the rest of hormonal work-up was normal.

In view of progressive respiratory symptoms, occasional aspiration and anticipating further surgery for Hirschsprung’s disease, an excision biopsy of the laryngeal lesion was planned under suspension laryngoscopy. The child was intubated nasotracheally with a 3.5 portex soft blue line tube under endoscopic vision. A Parson laryngoscope with side slot (Storz, ref. 8576 B) was inserted to expose the epiglottic leaflets and the supraglottic mass. The ETT was protected using moist gauze. A carbon dioxide laser fiber of 2 watts (G3 lasers private limited) was used to incise the mucosa and raise submucosal flaps in the vertical plane to egg-shell and fully expose the mass. A hyoid saving complete submucosal excision of the mass done. The mucosal flaps and thus the bifid epiglottis were endoscopically sutured using 4.0 vicryl sutures and tissue glue (TISSELM®). The child was extubated on table and had an uneventful post-operative period, with no complaints of aspiration or respiratory distress.

Histopathological examination of the mass revealed hyaline cartilage, enchondral ossification, with bone marrow tissue, highly suggestive of a hamartoma (Figure 3c). At the fourth month follow up, the child has had no symptoms of aspiration or noisy breathing and post-operative laryngoscopy showed optimal healing (Figure 3 b). Few months later, the child underwent the pull through procedure for imperforate anus and the colostomy closure was done a month later. At the last follow up, the child had an age-appropriate growth with normal hormonal levels. The findings in this report fall in the realm of routine clinical care and do not relate to basic human research. Informed consent was obtained from the patient’s parents to participate in this report and allow publication of his photographs and the clinical information.
DISCUSSION

Bifid epiglottis (BE) is a congenital malformation defined as a midline-cleft of the epiglottis. It presents as an isolated anomaly, a component of multiple anomalies or as a syndromic constituent of malformation syndromes. Digital anomalies are the most common association with BE.1,2 The second most common is hypothalamic hamartoma and/or hypopituitarism. The concurrence of BE with PHS, a rare malformation syndrome characterized by hypothalamic hamartoma, polydactyly, and imperforate anus are noteworthy. The combination of these malformations may be due to the synchronous embryonic development of the epiglottis, hypothalamus, and the digital buds. Other features in PHS may include abnormal lung lobation, renal abnormalities including cystic malformations, renal hypoplasia, ectopic ureteral implantation, genitourinary anomalies and Hirschsprung’s disease,4 Rarely cochlear anomalies have also been reported, and is usually caused by truncating frame shift/ nonsense and splicing mutations in the middle third of GLI3.7 The clinical course of the PHS ranges from mild to lethal in the neonatal period. The malformations in PHS are due to mutations in GLI3 located on chromosome 7p14.1.4 GLI3 codes for a zinc finger transcription factor that regulates genes downstream in the sonic hedgehog (SHH) signaling pathway which can affect multiple organs like central nervous system, limbs, and craniofacial structures.

Bifid epiglottis is a rarely reported laryngeal anomaly, often presents in the neonatal period with symptoms of stridor with or without aspiration. The diagnosis is confirmed by direct laryngoscopy. In the case reported here, the child had many features of PHS-like polydactyly, hypothalamic hamartoma, Hirschsprung’s disease, duplicate anus, microphthalmus, uni-aural hearing deficit, bifid epiglottis, and facial deformities. He also had a midline laryngeal hamartoma which was removed surgically to manage progressive compromised airflow and aspiration. Transoral microscopic approach allowed us to remain axial to the supraglottis and keep the airway secured under vision during surgical manipulation of the mass. Use of the fiber-driven CO₂ laser allowed a precise and blood-less dissection and hyoid preservation. Finally, endoscopic suturing of the dissected mucosa allowed adequate mucosal healing of the airway, re-suturing of the bifid epiglottis and thus effectively treat the supraglottic obstruction. As against the endoscopic approach – an open transcervical approach would have meant neck incision, extensive infra- and suprahypoid dissection and complete excision of the hyoid body that can cause short- or long-term dysphagia. Re-establishing a favorable neo epiglottis via a neck approach would have been difficult and in addition, there would have been an increased risk of a pharyngo-laryngo-cutaneous fistula.

CONCLUSION

This report adds to the existing literature by describing a supraglottic hamartoma with progressive airway compromise in a setting of PHS. Clinical management of these patients should be reserved to a multi-specialty care unit.

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