

Giant epignathus with midline mandibular cleft: Insights in embryology and management

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ABSTRACT

A congenital teratoid tumor arising and protruding through the mouth is classified as epignathus or fetus in fetu. On review of literature, we found various reports of midline mandibular and lower lip cleft associated with flexion contracture of neck, midline cervical cord, but there is only one report of association with midline dermoid. We present an unusual case of midline cleft of mandible with an epignathus. A 2.3-kg male child, delivered transvaginally in the 38th gestational week, was referred to us for management of a large irregular growth hanging outside the mouth. On examination, he had a wide median cleft of the mandible with tongue adherent to the "V"-shaped defect in the area of lower lip. A midline irregular mass of size 12 × 8 × 5 cm with variegated consistency was arising in the midline from the floor of the mouth between the tongue and lower lip. X-ray and computed tomography scan showed a rounded soft tissue mass arising from the alveolus with multiple calcifications within it along with a large triangular calcification and absence of hyoid bone. The mass was excised by mobilizing the tip of tongue. Staged repair was planned for the defect in the mandible. Unfortunately, the baby succumbed postoperatively to complex congenital heart disease. Histopathology was suggestive of epignathus. We discuss hereby the embryology and current management strategies of the problem.

Keywords: Epignathus, Fetus in Fetu, Midline cleft mandible

INTRODUCTION

A congenital teratoid tumor arising and protruding through the mouth is classified as epignathus or fetus in fetu.^[1] The term "teratoma" is used for a mass comprising poorly organized tissues derived from each of three germ layers while "fetus in fetu" is reserved for well-differentiated teratoma with axial differentiation of limbs and organs.^[2,3] Midline mandibular clefts can be associated with midline clefts of the lips. The associations of upper lip cleft with nasal dermoid and lower lip cleft with flexion contracture of neck and midline cervical cord have been reported, but there is only one report so far of association with midline dermoid.^[4] We present an unusual case of midline cleft of mandible associated with an epignathus and complex congenital heart disease which has been never described before.

CASE REPORT

A baby boy weighing 2.3 kg was delivered transvaginally in the 38th gestational week by a young primipara. Antenatal screening did not yield any suspicious findings and pregnancy was uneventful. There was a large irregular growth hanging outside the mouth for which the baby was referred to us.

At presentation, the patient had tachypnea and respiratory distress. On examination, a wide median cleft of mandible was present with the tongue adherent to the "V"-shaped defect in the area of lower lip. The tip of the tongue was slightly bifid and fixed to the lowermost end of the defect. There was a midline irregular mass of size 12 × 8 × 5 cm of variegated consistency, arising from the floor of the mouth in the midline, between the tongue and lower lip. The mass was suspended with a broad pedicle,

causing outward and downward displacement of lower lip. There were few small globular swellings present on top of it resembling limb buds [Figure 1a].

The hemoglobin was 18.8 g% and total leukocyte count was 13,100. Serum electrolytes, C-reactive protein, and arterial blood gases were within normal limits. After stabilization, the child was further investigated. X-ray showed a rounded soft tissue mass arising from the alveolus which had multiple small calcifications and a large triangular calcification resembling a partly formed vertebra [Figure 1b]. Computed tomography showed similar findings with absence of hyoid bone and no intracranial, retropharyngeal, or parapharyngeal extension [Figure 1c]. Preoperative alpha-fetoprotein (α -FP) with β -HCG levels were 1,65,283 μ g/l and 15.35 μ g/l, respectively.

Staged correction of the deformity was planned. In the first stage, we excised the midline mass with the help of harmonic scalpel after mobilization of the tip of tongue. The repair of the defect in the mandible was planned for a later date [Figure 2a]. Postoperatively his α -FP and β -HCG, reduced significantly to 4964 μ g/l and 1.2 μ g/l, respectively.

The child developed stridor immediately after extubation. The tongue was kept on traction suture which improved the breathing pattern and the patient was shifted to neonatal surgical intensive care unit. After 24 hours, the child developed respiratory acidosis and was kept on conventional ventilator support for 2 days. The patient was extubated on postoperative day 3 but had persistent respiratory distress. The position was changed to prone suspecting obstruction due to epiglottis or backward fall of the base of tongue. The baby had repeated episodes of fall in O_2 saturation even with ventilatory support and developed pansystolic murmur with cardiomegaly and signs of pulmonary edema. Hence, tracheotomy was postponed and focus was shifted for management of congestive cardiac failure (CCF). He was started on digitalis with frusemide with dopamine and was shifted on high-frequency oscillatory ventilation. The complex congenital heart disease was diagnosed as atrioventricular canal defect. Despite the best possible efforts and measures, the baby succumbed to complex congenital heart disease (Atrio-Ventricular canal defect).

The excised specimen measured 12 × 8 × 5 cm and cut surface revealed the majority of the lesion to consist of soft with fleshy areas, one being cystic with few areas of calcifications [Figure 2b]. Microscopic examination showed presence of neural tissue, cartilage, bone, epididymis, and vas deferens with intestinal epithelium [Figures 2c and 2d]. There was layered arrangement of derivatives all three germ layers from outward to inward (suggestive of fetus in fetu) differentiating it from mature teratoma.

DISCUSSION

Previously all teratomas arising in the mouth of newborns were classified as Epignathus. However, nowadays the term “true epignathus” is used for monstrosities containing well-formed organs and limbs, which should therefore be regarded as a parasitic fetus.^[5]



Figure 1 (a): Midline irregular mass arising from the floor of the mouth, between the tongue and lower lip with overlying multiple small globular swellings resembling limb buds

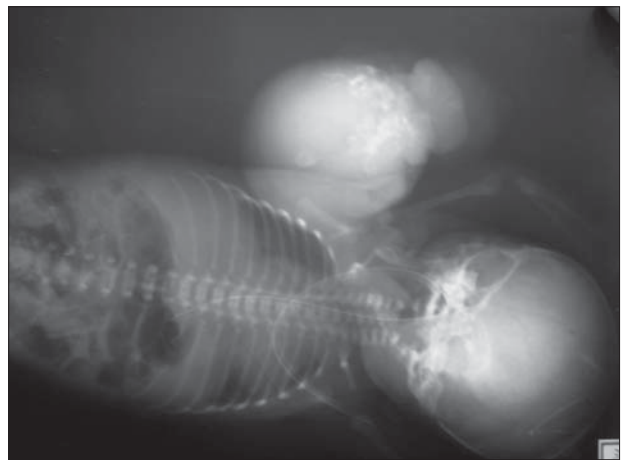


Figure 1 (b): X-ray showing rounded soft tissue mass arising from the alveolus with multiple calcifications within it in addition to a large triangular calcification suggestive of partly formed vertebra

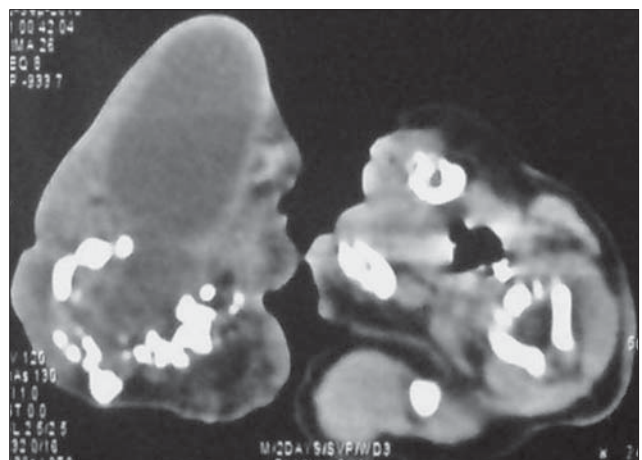


Figure 1 (c): Computed tomography scan showing heterogenous mass and absence of hyoid bone without any intracranial, retropharyngeal or parapharyngeal extension of the lesion



Figure 2 (a): Defect in the mandible after excision of mass



Figure 2 (b): Excised specimen

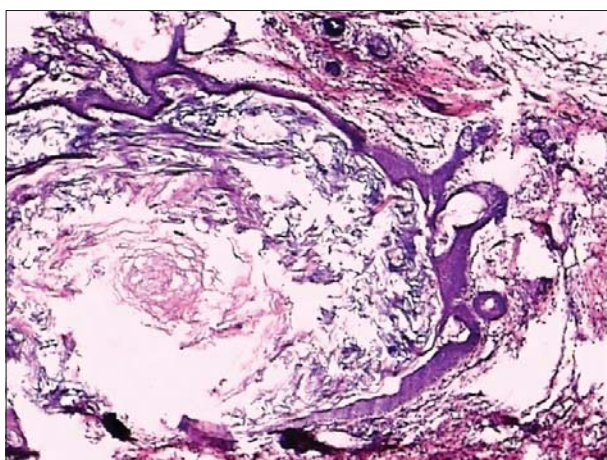


Figure 2 (c): Histopathological examination of specimen showing neural tissue, cartilages, bone, and intestinal epithelium (H&E, 10x)

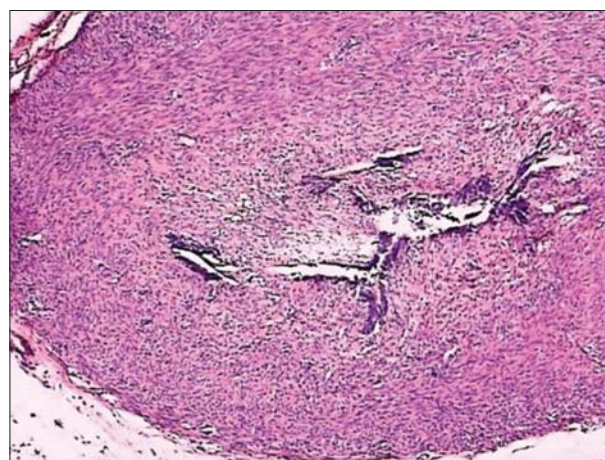


Figure 2 (d): Histopathological examination of specimen showing vas deferens and epididymis (H&E, 4x)

Teratomata of the head and neck are uncommon and those arising within oral cavity attached to mandible, palate, or base of the skull are still rarer.^[6] Ewing classified oropharyngeal teratoma into dermoids, teratomata with teratoids, and epignathi.^[7] Dermoids are the most common and tend to affect adults.^[8] Teratomata consist of poorly organized derivatives of three germ layers and generally affect infants.^[8,9] Epignathus have a parasitic fetus which is attached to the palate of autosite. They fill the oral cavity and protrude from the mouth causing respiratory obstruction. The pedunculated epignathus is attached either to the nasopharynx in the region of the basisphenoid (Rathke's pouch) or to the dorsal surface of the palate.^[10] The term fetus in fetu is reserved for masses in which the differentiation of the teratoma is carried to a high degree with presence of axial differentiation of limbs and organs. It is difficult to make a difference between epignathi with structures resulting from abortive attempts of twinning because of existence of highly organoid appearance.^[2,3]

Couronne first described the median cleft of the lower jaw.^[11] The cleft of the lower lip may present as a notch. Frequently, the cleft extends to bony mandibular symphysis. The anterior portion of tongue is often bifid and bound to the divided mandible.^[4] The case we describe here had a midline cleft mandible with

epignathus (or fetus in fetu) and bifid tongue. Such an abnormality has not been reported before.

Embryological explanation of epignathus and cleft mandible

Exact etiology of epignathus is not known. It may arise from pleuripotential cells in the region of Rathke's pouch.^[5] Several hypotheses about the pathogenesis of median clefts of the lip and mandible have been proposed. According to Surendran *et al.*, failure of the paired first branchial arches to unite normally results in midline clefts of lower lip with mandible along with anomalies of anterior two thirds of the tongue.^[4] In severe deformities, even second and third branchial arches can be involved resulting in cleft of lower lip with mandible, contracture of neck in midline with absence of hyoid bone.^[4]

However, according to Catharina *et al.*, instead of paired branchial arches, only one first branchial arch develops during the early embryonic period (≤ 17 mm crown-rump length).^[12] Two mandibular processes grow within this first branchial arch, separated by a groove in the midline. These mandibular processes do not fuse but merge during the late embryonic period (≥ 17 to ≤ 60 mm crown-rump length). In the same developmental period, there is formation of the lip and the alveolar process

with the anlage and outgrowth of one membrane bone center in each mandibular process. This results in the formation of the mandible with its symphysis. They also proposed that hypoplasia of the mandibular processes during the early embryonic period will lead to the most severe cleft of the mandible extending into the neck. During the late embryonic period, median clefts with lesser severity develop. Disturbances of the outgrowth of bone centers of the mandible, resulting in nonformation of its symphysis, cause cleft of the mandible with involvement of all related soft tissues. Defects in the merging process produce just a notch of the vermilion or a higher cleft of the lower lip with or without involvement of the alveolar process of the mandible.

Embryological explanation of our case

On the basis of above theories we propose that in present case, origin of the mass can be explained by pluripotential growth of cells derived from all three germ layers. The mass on the other hand prevented the mesodermal migration and fusion of mandibular process resulting into cleft of mandible with forward and downward displacement of lower lip.

Management

The treatment of these tumors depends on the extent and site of origin. Some epignathi which have a small site of origin from buccal mucosa are easily excised by cautery, snare, or harmonic scalpel as in our case. Although complete excision of a sacrococcygeal teratoma is required to prevent malignant degeneration, this may not appertain to the treatment of epignathus as there is no case on record of malignant dissemination.^[9] Surgical excision may consist of an oral approach to excise the tumor at its base once intracranial extension has been ruled out.^[13] Radical disfiguring surgery, which would result in impairment of speech and deglutition, is contraindicated in neonate. The airway should be secured by endotracheal intubation or if necessary by tracheotomy. Airway obstruction and craniofacial deformities resulting from mandibular deficiency are challenging and complex clinical problems.

Previously, the operative procedure in early infancy was limited to the excision of mass with repair of soft tissue to avoid interference with normal growth pattern due to trauma to the bony structures.^[4] Recently, mandibular distraction osteogenesis is rapidly gaining popularity. Early bilateral mandibular distraction has obviated tracheostomy in most newborns, with low operative morbidity in addition to early decannulation and resolution of obstruction. Moreover, refinements in distraction technique have advanced treatment of non-airway-related mandibular deformities.^[14]

The treatment should be started as early as possible.^[14] In the median clefts of the lower lip, a V excision of the cleft with closure in layers is the procedure of choice. When tongue is bifid which is tied down to the cleft, Z-plasty is done to free the tongue. Prolonged follow-up is necessary to correct speech impediments,

facial deformities, and dental irregularities with malocclusion of the jaws.^[6] Serial estimation of serum α -FP should be done to rule out the reactivation of the tumor. If levels increase, a CT scan is necessary to define the extent. In the event of recurrence, further local surgery prevents feeding difficulties.^[15] Chemotherapy is indicated if local surgery fails but radiotherapy should be avoided to avoid serious long term complications.

In our case, the mass was composed of intestine-like structures, skin, bone (part of vertebra), vas deferens and epididymis with glans-like structure along with limb bud like structures. This level of differentiation indicates that mass could be fetus in fetu, but absence of organized skeleton and broad attachment to autosome as in our case goes against it.^[2,13,16] We have therefore classified this case as giant epignathus with midline cleft of mandible and this association has not been reported previously in English literature.

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