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Epignathus: Role of Fetal Magnetic Resonance Imaging and Histopathology—A Case Report

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Abstract

This case report describes a rare and life threatening fetal condition, oropharyngeal teratoma (epignathus), diagnosed prenatally at 26 weeks. The fetus presented with polyhydramnios and a large mass obstructing the airway. Despite multidisciplinary planning for an ex utero intrapartum treatment (EXIT) procedure, the baby died shortly after birth due to severe respiratory distress. Histopathological examination confirmed a high grade immature teratoma. The report highlights the importance of prenatal diagnosis, multidisciplinary management, and potential interventions like EXIT procedures for improved outcomes. It also emphasizes the role of fetal magnetic resonance imaging in characterizing the tumor and planning delivery strategies. In cases that have been diagnosed early enough, termination of pregnancy can be an option after counseling.

Keywords

- epignathus
- oropharyngeal teratoma
- fetal teratoma
- prenatal MRI
- prenatal interventions

Introduction

Teratomas are common fetal tumors formed by pluripotent cells of all three germ cell layers. These are predominantly benign tumors located in the body's midline, most commonly in the sacrococcygeal region. Oropharyngeal teratoma (epignathus) is an extremely rare congenital teratoma, occurring in 1 in 200,000 live births.[1] The bulk of the tumor tends to plug the oral cavity, often leading to severe obstruction of the upper airways and high neonatal mortality rates. Diagnosis is usually made in the second or third trimester of pregnancy by ultrasound, where they appear as masses of mixed echogenicity with solid cystic components and varying degrees of calcification and vascularity. Usually, they are not associated with genetic abnormality or recurrence.[2]

The prognosis and time of diagnosis depend upon factors such as the location, size, and tumor growth velocity. Associated complications during pregnancy may need interventions such as amnioreduction, intrauterine blood transfusions, radiofrequency ablation, pericardiocentesis, or ex utero intrapartum treatment (EXIT procedure).[3]

Case Report

A 24 year old $G_3P_1L_1A_1$ with a normal three generation pedigree presented at the 26th week of pregnancy with increasing abdominal girth and discomfort. Clinically, polyhydramnios was suspected. She had not undergone screening for aneuploidies. On ultrasound, a heterogeneously hypoechoic mass of size 7.66 × 7.21 cm with few intermixed hyperechoic areas was seen arising from the oral cavity of the fetus. It had areas of calcifications with minimal internal vascularity. In addition, there was associated polyhydramnios (Amniotic Fluid Index {AFI}: 33 cm) (►**Fig. 1**).

Fetal magnetic resonance imaging (MRI) was performed at 28 weeks' gestation for characterization of the tumor. On MRI, a large solid cystic mass of size 9.3 × 7.7 × 9 cm was seen arising from the lower face, mouth, and jaw region completely

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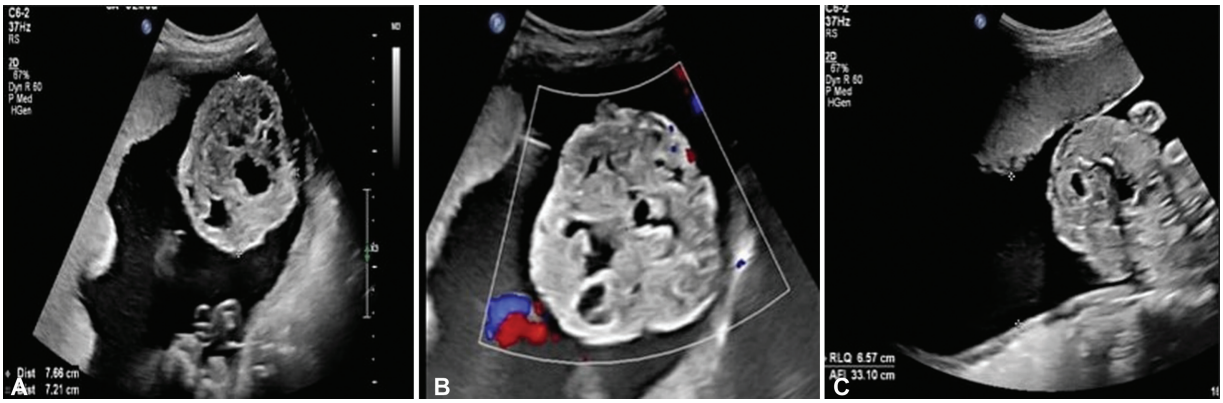

Fig. 1 Ultrasound done at 26 weeks showing (**A**) heterogenous mass of 7.66 × 7.21 cm with solid cystic areas and (**B**) minimal internal vascularity associated with (**C**) polyhydramnios (AFI: 33 cm).

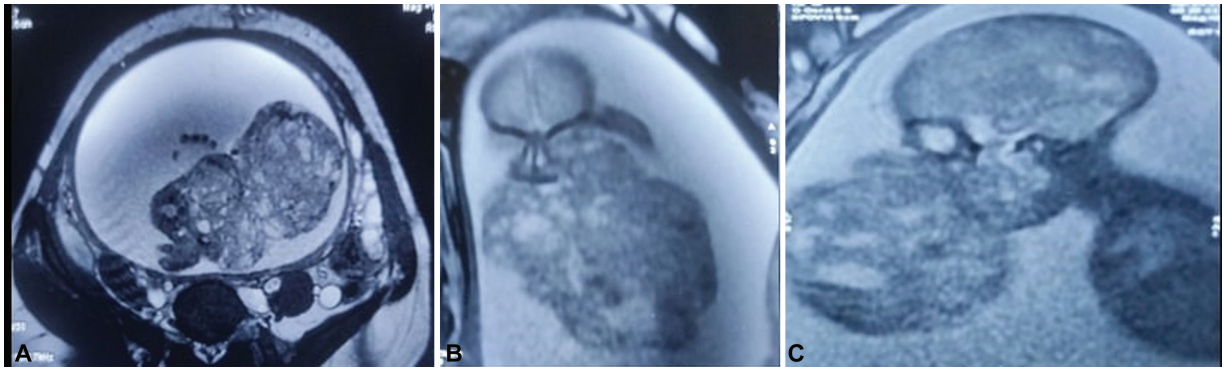

Fig. 2 Axial (**A**), coronal (**B**), and sagittal (**C**) sections on magnetic resonance imaging showing the extent of tumor.

filling the oral cavity and replacing the body of the mandible. Posteriorly, it was extending up to the posterior pharyngeal wall, causing complete obstruction of the oropharynx and nasopharynx airway. Superiorly, it involved the superior alveolar arch and maxilla with distortion of the face. There was no associated central nervous system anomaly or intracranial invasion by mass (►**Fig. 2**). With these findings, a probable diagnosis of fetal teratoma or microcytic type of slow flow congenital hemangioma was made. In follow up ultrasound at 30 weeks' gestation, the size of the mass increased to 10.3 × 8.7 cm along with fetal growth restriction, polyhydramnios (AFI: 40 cm), and fetal anemia (middle cerebral artery Peak systolic velocity (PSV): 1.7 Multiple of median (MoM)) (►**Fig. 3**).

Results

The case was monitored in antenatal outpatient department. Pediatric surgery registration was done, and multidisciplinary

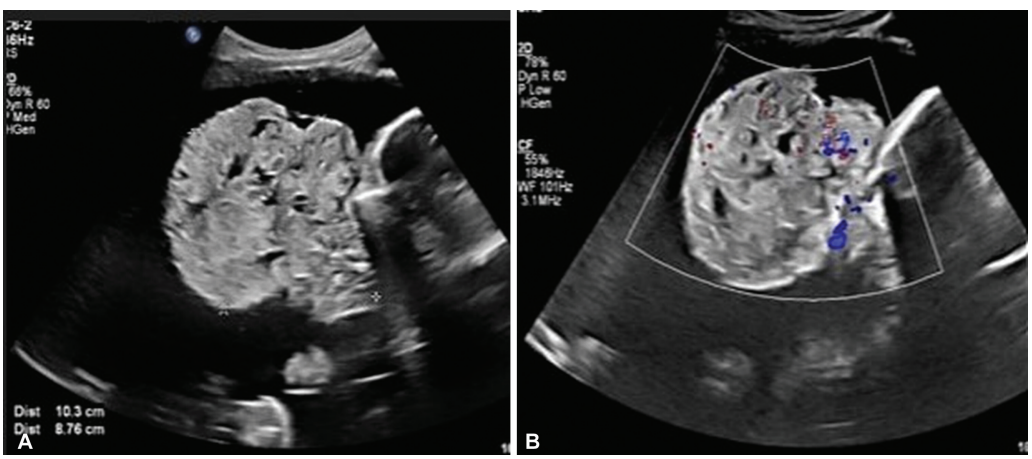

Fig. 3 Ultrasound done at 30 weeks showing (**A**) increase in the size of mass to 10.3 × 8.7 cm with (**B**) minimal internal vascularity associated with polyhydramnios (AFI: 40 cm).

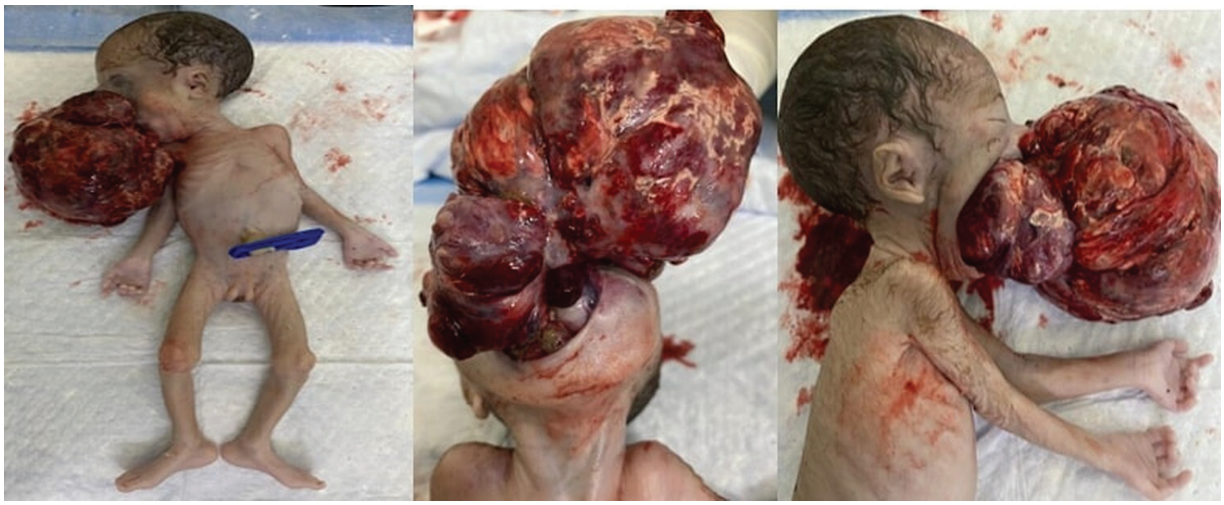

Fig. 4 Large multilobulated mass with calcifications and hemorrhage on the surface seen arising from the oral cavity.

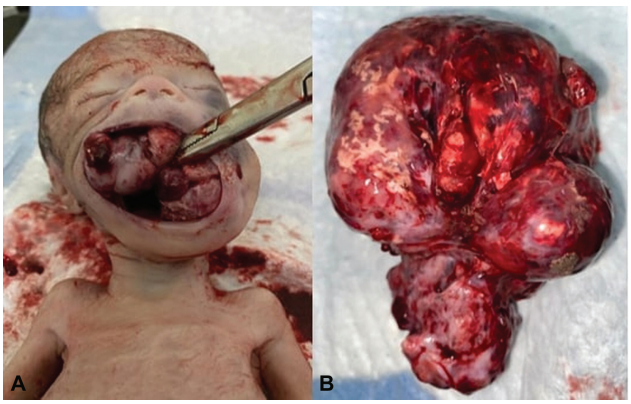

Fig. 5 (A) Oral cavity after removal of mass. (B) Mass after excision measuring 12 × 9 × 6 cm.

care was planned using an EXIT procedure. However, a week later, she had spontaneous preterm labor and delivered a baby girl of 900 g with an Apgar score of 2,1 who died after 10 minutes due to severe respiratory distress before any intervention could be instituted. Cord blood hemoglobin was 9 g%, suggestive of fetal anemia. An external autopsy was performed after taking written informed consent from the couple. A large, soft, multilobulated, and mobile mass of size 12 × 9 × 6 cm was seen protruding from the oral cavity, with evidence of hemorrhages and calcifications (►**Figs. 4**, **5**). No other gross fetal anomaly was identified. Subsequently, histopathological analysis of mass reported stratified squamous epithelium with a sebaceous gland, adipose tissue, salivary gland, intestinal epithelium, cartilage and bone, and immature neuroepithelium (►**Fig. 6**). These findings confirmed the diagnosis of high grade immature teratoma.

Discussion

The prognosis of epignathus is usually extremely grave. It primarily depends upon the bulk of the lesion, growth velocity, and the involvement of surrounding structures. In a combined review of 33 neonates with prenatally diagnosed oropharyngeal teratoma, 12 died in utero or shortly after birth, mostly due to airway obstruction.[4] In the current case also, early neonatal demise occurred due to severe respiratory distress following severe mechanical airway obstruction. Simonini et al followed 79 cases of prenatally diagnosed teratoma over a period of 17 years, among which six cases (7.6%) had epignathus. The mean age of diagnosis in these cases was 27 weeks and 4 days (ranging from 21 weeks 1 day to 36 weeks 5 days). Five of the epignathus were complicated

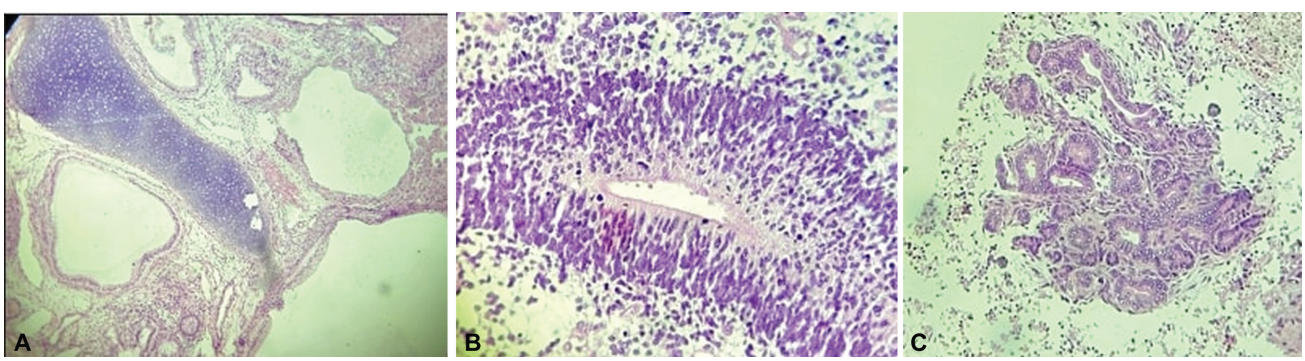

Fig. 6 Histopathological examination of mass. (A) Cartilage with ciliated columnar epithelium. (B) Immature neuroepithelium. (C) Eccrine glands

by polyhydramnios, three needed amnioreduction and an EXIT procedure was performed in one case in the prenatal period. Pregnancy was terminated in two cases; one resulted in stillbirth, another one in neonatal death, and two are alive and healthy. Three cases were delivered by cesarean section, one of them was stillborn. The mean age of delivery in these cases was 35 weeks and 2 days. Among those two who are alive, one underwent tumor resection (R_1) followed by plastic reconstruction, whereas another one underwent complete tumor resection with cleft and dental surgery. In one case of a liveborn fetus with epignathus, the parents opted for palliative care due to an unfavorable prognosis. Both were followed up till 30 months and 7 years of age, respectively, till the publication of literature. Mature teratoma was confirmed on histopathological analysis in both cases. On long term follow up, one case was complicated by cerebral venous sinus thrombosis, whereas another one with duplication of the pituitary gland syndrome.[5] In another reported case of fetal immature orofacial teratoma detected at 13 weeks 1 day of gestation in the first trimester on ultrasound, pregnancy was terminated after extensive couple counseling.[6]

Conclusion

Oropharyngeal teratomas are a rare fetal tumor that can be diagnosed in utero by ultrasound examination. To achieve favorable outcomes for children with teratomas, it is important to recognize specific associated complications during the course of pregnancy and to manage them with a multidisciplinary team approach. Various interventions may have to be performed during the antepartum and intrapartum periods, ranging from fetal pericardiocentesis and amnioreduction to the EXIT procedure, to facilitate delivery. Fetal MRI should be taken into consideration in tumors of the neck and oropharynx, which offers very high sensitivity in the diagnosis and assessing spatial extension of the tumor. MRI is also useful in confirming airway patency and particularly helpful in determining the need to perform an EXIT procedure. Intracranial tumor extension can severely worsen prognosis; thus, detailed fetal neurosonography, as well as additional fetal MRI, is also advocated. Termination of pregnancy remains an option after thorough couple counseling when diagnosed in early pregnancy.

Conflict of Interest
None declared.

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