CASE REPORT

# Congenital Epignathus with Transposition of Great Arteries!

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## Abstract

Epignathus of a fetus is a rare type of oropharyngeal teratoma rarely reported in the literature, especially from the Indian subcontinent. Antenatal diagnosis of this condition is even rarer. The overall survival rates with these lesions have been between 17 and 87.5%. However, survival in the setting of antenatally diagnosed teratomas has only been described anecdotally. Transposition of the great arteries (TGA) is an isolated abnormality in about 90% of the cases and rarely is associated with extracardiac malformation. Gallbladder duplication resulting in two separate gallbladders is a rare congenital anomaly. We present an unusual case of antenatally diagnosed massive oropharyngeal teratoma, which was associated with transposition of great arteries and duplicated gallbladder. To conclude, the antenatal diagnosis of TGA decides the postnatal outcomes and hence requires planned delivery and further management. A combination of these conditions may cause airway obstruction and respiratory compromise, like in our case, which necessitated termination of pregnancy.

**Keywords:** Airway obstruction, Neonate, Teratoma, Transposition of great vessels.

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## Introduction

Congenital germ cell tumors of the oropharyngeal region are a rare antenatal finding. The most common site of teratoma is in the sacrococcygeal region. It constitutes <10% of reported cases in the head and neck region.[1–5] Few reports of cardiac anomalies (septal defects) and facial deformities with oral teratomas have also been observed.[6–8]

The basic classification is histological and is widely used.

- Dermoid tumors: It is the most common form of teratoma composed of ectoderm and mesoderm derivatives.
- Teratoids: Poorly differentiated tumors containing all three germ layers.
- Teratoma: Well-differentiated tumors containing tissues from all three germ layers.
- Epignathus: Highly developed rare oral tumors possessing a high mortality rate. They have developmental fetal organs and limbs.[9–12]

Masses with solid and cystic components are more in favor of teratoma.

We present a case of oropharyngeal teratoma associated with congenital heart disease and gallbladder duplication. TGA is the second most common cyanotic congenital heart disease associated with significant mortality and morbidity. It is characterized by atrioventricular concordance and ventriculoarterial discordance. It is rarely associated with chromosomal anomalies or extracardiac malformations. However, our case was a rare combination. To improve the antenatal diagnosis of TGA, it is vital to identify the key ultrasound (US) markers of TGA. The presence of a parallel course of TGA, two vessels instead of three in the three-vessel trachea (3VT) view, and identification of the origin of arteries are the key markers on US. Other antenatal US markers such as an abnormal right convexity of the aorta, an I-shaped aorta, and the "boomerang sign" may also be used to diagnose TGA.[13–15]

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**Conflict of interest**: None

## Case Description

A 24-year-old female with two pregnancies, one delivery, and one living child with no history of consanguinity came for her first antenatal scan to US at 24 weeks of gestation. During the targeted imaging for fetal anomalies scan, we detected a cystic mass with few calcifications measuring 19, 11, and 20 mm in anteroposterior, transverse, and craniocaudal dimensions, respectively. Mass is seen arising from the oropharynx. In addition, multiseptated cystic mass was noted arising from mandibular and upper neck measuring 29, 37, and 47 mm in anteroposterior, transverse and craniocaudal

dimensions, respectively (Fig. 1). Lesion is seen causing mass effect on submandibular neck and trachea. The protruded mass is seen to cause a significant mass effect on the nasopharyngeal airway and impedes fetal swallowing, which leads to maternal polyhydramnios. Fetal echocardiography revealed transposition of great arteries and a small inlet type of ventricular septal defect (VSD) (Fig. 2). In addition, duplication of the gallbladder was noted (Fig. 3).

Fetal magnetic resonance imaging (MRI) was performed to reveal the extensions of mass and rule out other head and neck anomalies (Fig. 4). Lips and hard palate were visualized as intact by both US and MRI. In view of a combination of anomalies not compatible with life, termination was performed (Fig. 5). Abortus was subjected to an autopsy, which confirmed findings. Histopathology revealed immature teratoma (Fig. 6). Peripheral blood karyotype was normal. The couple was informed about the possible risks in the subsequent pregnancies.

## Discussion

Teratomas are well-differentiated tumors with tissues derived from all three germ layers, which are foreign to the anatomical site of origin. Most of the teratomas are benign, but reports of malignant teratomas do exist.

The exact pathogenesis of its development is unknown. According to one of the accepted theories, it is speculated that the totipotent embryonic tissues adjacent to the primitive streak and notochord are displaced by some unknown mechanism during ontogenesis.[9]

The tissues often found in these consist of cartilage, bronchial epithelium, and brain tissues. Another pathological variant of epignathus is "*fetus-in-fetu*" occurs due to the incomplete twinning of monozygotic twins of development.[11,12]

Epignathus is an uncommon tumor attached to the skull base, usually to the hard palate or the mandible.[4,6] Incidence approximately 1:35000–1:200000 live births.[1–3] The clinical presentation is variable depending on the size of the mass. The newborn may be either asymptomatic or may present with feeding difficulty, stridor, or recurrent apnea.[7,8,10] Giant epignathus fill the oral cavity and protrude from the mouth resulting in respiratory compromise.[4,7] In our case, the teratoma was huge enough to cause respiratory compromise along with feeding problems.

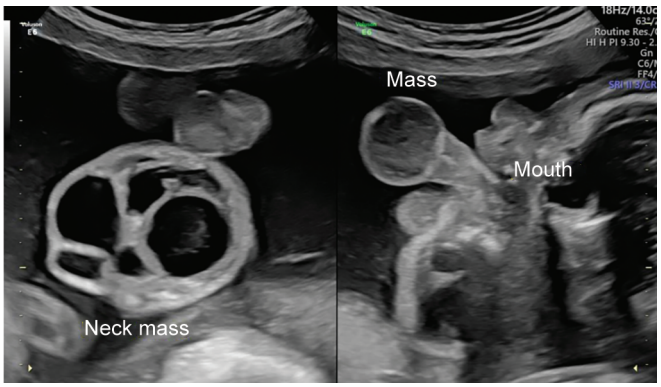


**Fig. 1:** Ultrasound of the fetus revealed a cystic mass arising from the oropharynx, submandibular region, and anterior aspect of the neck

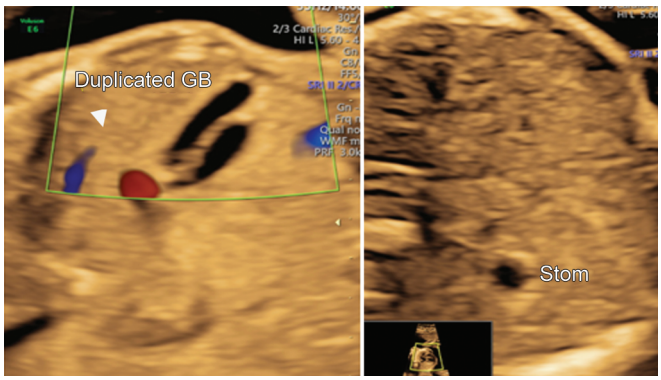


**Fig. 3:** Ultrasonogram revealed a duplication of the gallbladder and suboptimally distended stomach due to impeded fetal swallowing

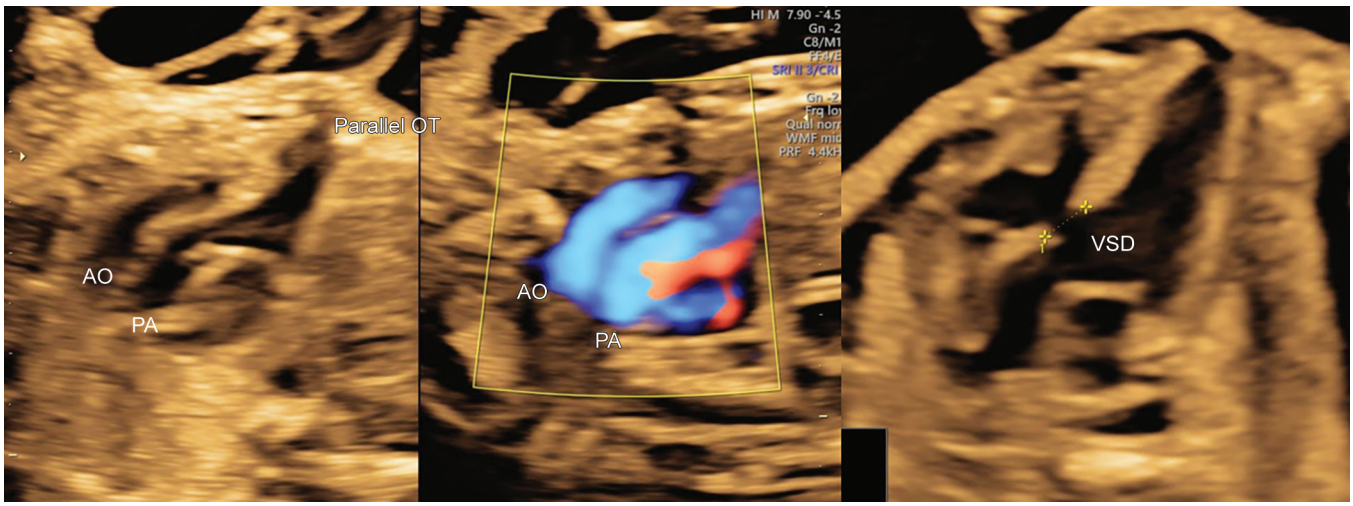


**Fig. 2:** Fetal echo revealed VA discordance and the parallel alignment of the great arteries, 3VT view showing altered anatomical relationships features of transposition of great arteries and small inlet type of VSD

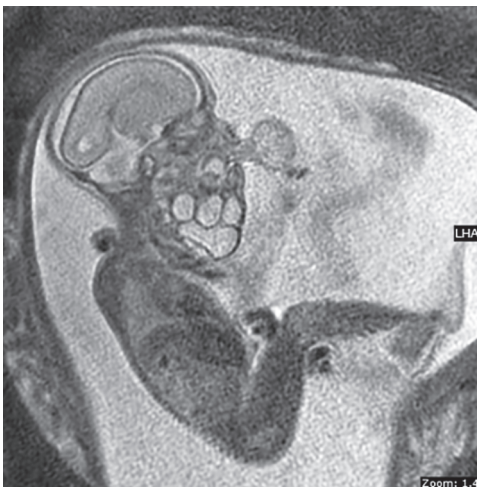

**Fig. 4:** Fetal MRI: sagittal T2 weighted images showing multiseptated cystic lesions arising from the oropharynx, submandibular region, and upper neck

**Fig. 5:** Image of abortus showing multiple cystic lesions arising from the oropharynx

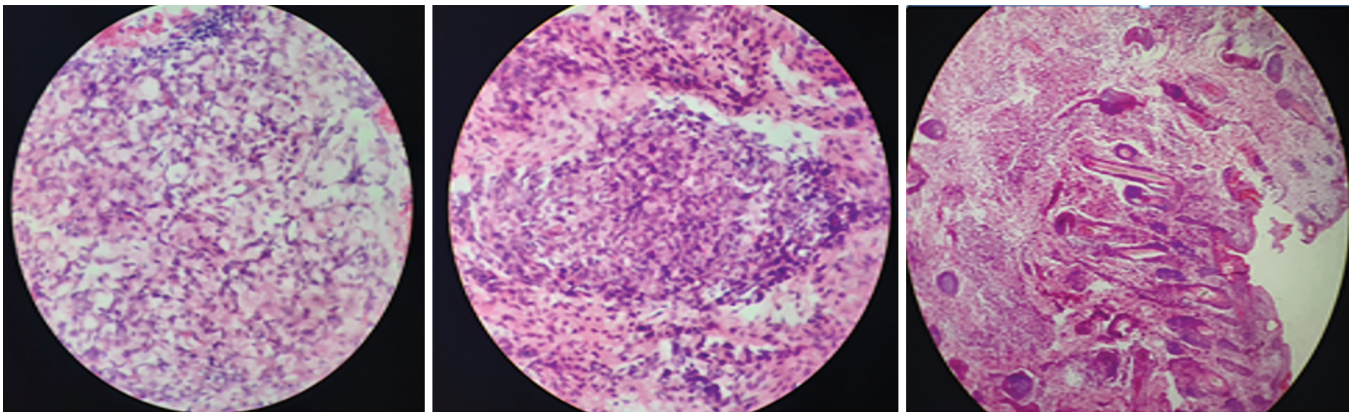

**Fig. 6:** Photomicrograph showing cells of ectoderm, mesoderm, and endodermal origin. Multiple immature cells noted. No signs of malignancy

The antenatal diagnosis of TGA requires planned delivery and thus influences postnatal outcomes. Hence it is important to identify the US markers of TGA to improve the antenatal diagnosis and consequently provide perinatal support. The presence of classic US markers and ancillary findings helps in diagnosis.

## Conclusion

The detailed analysis of the outflow tract views was the key to the antenatal diagnosis of TGA as in our case. Although benign, congenital teratomas are potentially lethal due to the risk of airway obstruction. However, our case was a rare combination of congenital epignathus, TGA, and duplication of the gallbladder, which necessitated termination of pregnancy.

## References


1. Lalwani AK, Engel TL. Teratoma of the tongue: a case report and review of the literature. Int J Pediatr Otorhinolaryngol 1992;24(3):261–268. DOI: 10.1016/0165-5876(92)90024-j
2. Abemayor E, Newman A, Bergstrom L, et al. Teratomas of the head and neck in childhood. Laryngoscope 1984;94(11 Pt 1):1489–1492. DOI: 10.1288/00005537-198411000-00020
3. Kountakis SE, Minotti AM, Maillard A, et al. Teratomas of the head and neck. Am J Otolaryngol 1994;15(4):292–296. DOI: 10.1016/0196-0709(94)90099-x
4. McMahon MJ, Chescheir NC, Kuller JA, et al. Perinatal management of a lingual teratoma. Obstet Gynecol 1996;87(5 Pt 2):848–851.
5. Kaplan C, Perlmutter S, Molinoff S. Epignathus with placental hydrops. Arch Pathol Lab Med 1980;104(7):374–375.
6. Lionel J, Valvoda M, Hadi KAAA. "Giant epignathus"- a case report. Kuwait Med J 2004;36(3):217–220.
7. Too S, Sarji SA, Yik Y, et al. Malignant epignathus teratoma. Biomed Imaging Interv J 2008;4(2):e18. DOI: 10.2349/biij.4.2.e18
8. Hu R, Jiang RS. The recurrence of a soft palate teratoma in a neonate: a case report. Head Neck Onco 2013;5(2):16.
9. Shah A, Latoo S, Ahmed I, et al. Head and neck teratomas. J Maxillofac oral Surg 2009;8(1):60–63. DOI: 10.1007/s12663-009-0015-x
10. Kontopoulos EV, Gualtieri M, Quintero RA. Successful in utero treatment of an oral teratoma via operative fetoscopy:

case report and review of the literature. Am J Obstet Gynecol 2012;207(1):e12–e15. DOI: 10.1016/j.ajog.2012.04.008
11. Mirshemirani A, Khaleghnejad A, Mohajerzadeh L, et al. Congenital nasopharyngeal teratoma in neonate. Iran J Pediatr 2011;21(2):249–252.
12. Chauhan DS, Guruprasad Y, Inderchand S. Congenital nasopharyngeal teratoma with a cleft palate: case report and a 7 year follow up. J Maxillofac Oral Surg 2011;10(3):253–256. DOI: 10.1007/s12663-010-0140-6
13. Abuhamad A, Chaoui R. Genetic aspects of congenital heartdefetcs: a practical guide to fetal echocardiography: normaland abnormal hearts. Philadelphia: Lippincott Williams &Wilkins 2010. P. 11–29.
14. Udelsman R, Sugarbaker PH. Congenital duplication of the gallbladder associated with an anomalous right hepatic artery. Am J Surg 1985;149(6):812–815. DOI: 10.1016/S0002-9610(85)80193-8
15. Nichols DM, Li DK. Superior mesenteric vein rotation: a CT sign of midgut malrotation. AJR Am J Roentgenol 1983;141(4):707–708. DOI: 10.2214/ajr.141.4.707