Case Report

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Prenatal Diagnosis of a Giant Epignathus in the Second Trimester and Immediate Successful Management at Birth: A Case Report

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Abstract

Epignathus is an extremely rare congenital oropharyngeal teratoma. Here, we report a case of epignathus without intracranial extension in a fetus. The mass was first found by ultrasonography at 22 gestational weeks. Serial ultrasound examinations and magnetic resonance imaging confirmed that the fetus had neither central nervous system involvement nor abnormal nose or tooth. The baby was delivered at 37 weeks and six days of gestation via cesarean section set up for ex-utero intrapartum treatment. The postnatal pathologic examination confirmed the presence of mature tissues predominantly containing ectopic central nervous tissue, osseous tissue, and bronchial mucosal tissue. Most cases of epignathus are associated with malformation and death. Ultrasound and magnetic resonance imaging prenatal assessments are very important to facilitate counseling and understand prognosis. In conclusion, the ex-utero intrapartum treatment procedure is a good approach to improve the survival of infants with epignathus.

Keywords: Prenatal diagnosis; Teratoma; Epignathus; Fetus.

Introduction

Epignathus is an extremely rare congenital oropharyngeal teratoma containing fetal organs and structures. It is estimated to occur in 1:35,000–1:200,000 live births with a high incidence in female fetuses compare with male fetuses (3:1-5.1:1).[1,2] The teratoma varies from a small pedunculated tumor that can be easily removed to a large mass involving the palate and sphenoid which is present at the mouth during birth, constituting an immediate threat to life.[3] Epignathus can cause hydramnios or fetal death in the ante partum period and respiratory distress due to tracheal obstruction in the post-delivery period.[4] Antenatal evaluation by ultrasonography and preparation of ex utero intrapartum treatment (EXIT) are necessary to reduce the risk of hypoxic brain injury and death at birth.

In this case report, we present the prenatal diagnosis of fetal epignathus and the immediate successful management of the large oropharyngeal teratoma. Written informed consent was obtained from the parents of the neonate for the publication of this case report and the related images.

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Case presentation

The mother was a 26-year-old woman, gravida 3 para 1, with a history of assisted reproductive technology treatment owing to excision of both fallopian tubes following two instances of past ectopic pregnancies. She was referred to our hospital at 24 gestational weeks. Her alpha-fetoprotein (AFP) level was elevated to 6.2 multiples of the median in the second trimester. She underwent ultrasonographic evaluation at 22 gestational weeks at a local hospital. The imaging results showed the presence of a large, mixed, solid and cystic protruding mass in the fetal oral cavity. The mother was referred to our hospital for further ultrasound evaluation which confirmed the presence of the oral mixed solid and cystic mass and the extension outside the fetus's oral cavity. The mass measured 4.03 cm × 3.23 cm × 4.38 cm. No blood flow was observed on Doppler ultrasound imaging. The fetus's upper lip skin was continuous without a cleft lip above degree II. Other structures of this fetus were normal (Fig. 1). Subsequently, a fetal teratoma was suspected.

The patient declined fetal karyotyping out of fear of losing the pregnancy because the fetus was very precious for her. She opted for more counseling about whether the mass could be removed after delivery. An oral and maxillofacial surgeon provided counseling and informed her that there were ways to remove the mass from the neonate. From then on, the patient underwent regular antenatal examinations at our hospital, and was followed-up with serial ultrasound examinations. The patient's pregnancy progressed normally until 37 weeks and six days of gestation. An ultrasound showed the amniotic fluid index increased to 24.28 cm, and the mass now measured 8.66 cm × 5.76 cm × 6.77 cm. Considering the fetus was mature and the mass would obstruct its airway, we performed a cesarean section to deliver the baby. Fetal magnetic resonance imaging (MRI) before the operation confirmed that there was neither central nervous system involvement nor abnormal nose or tooth in the fetus (Fig. 2).

A multidisciplinary team was assembled to secure the airway and carry out EXIT. The team included specialists from

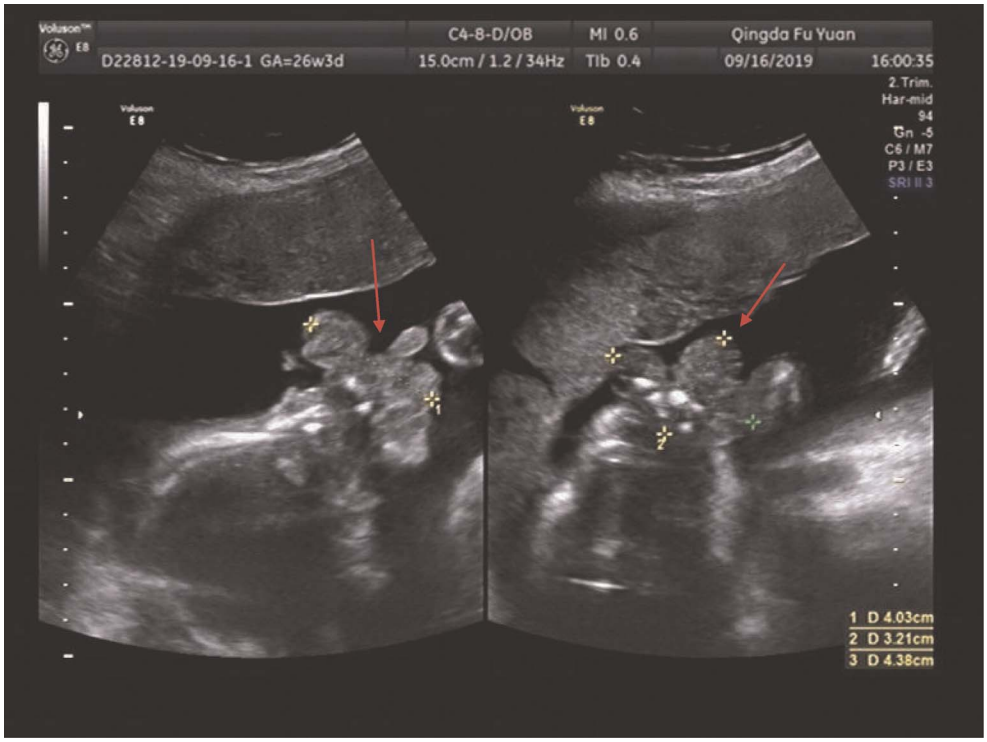

Figure 1. Well-defined mass measuring 4.03 cm × 3.23 cm × 4.38 cm by ultrasonography and arising from the fetal oral cavity at 24 gestational weeks. The arrows indicate the mass.

the maternal-fetal medicine department, neonatology department, anesthesia department, oral and maxillofacial surgery department, and ear nose and throat department. The plan was that as soon as the fetal head and shoulders were delivered, the neonate would be provided with a tracheal cannula or tracheostomy performed with keeping the placental circulation intact. After that, the neonate would undergo a further thorough evaluation or operation (as needed). Fortunately, the infant cried immediately after its head was delivered. At that moment, it was confirmed that there was no severe airway obstruction and a live female infant was delivered via cesarean section.

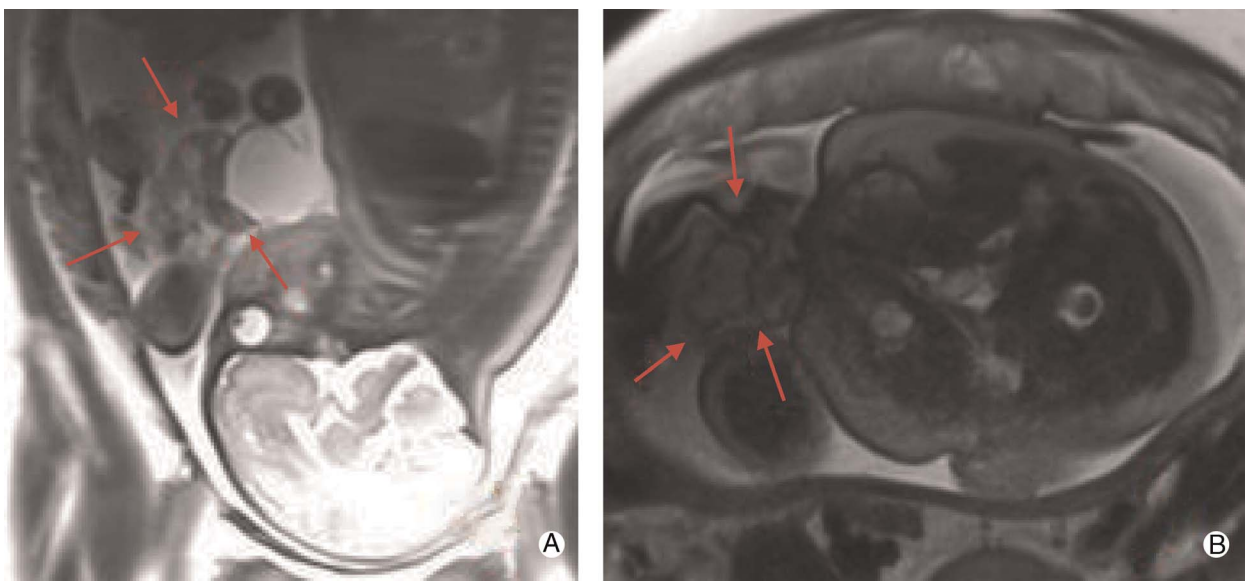

Figure 2. Magnetic resonance imaging at 36 gestational weeks showed no central nervous system involvement or serious associated malformations. The arrows indicate the mass. A Sagittal section. B Coronal section.

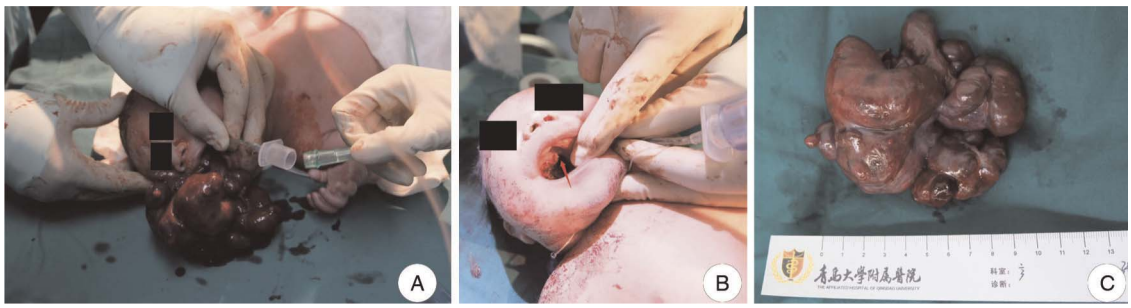

Figure 3. The neonate immediately underwent oropharyngeal mass resection at birth. Written informed consent was obtained from the parents of the neonate for the publication of these figures. A Appearance of the neonate with trachea cannula before resection. B Appearance of the neonate with trachea cannula after resection. The arrow indicates a cleft palate. C Appearance of the tumor.

The neonate immediately underwent oropharyngeal mass resection with trachea cannula and local anesthesia. Most of the mass was removed. A small cleft palate was diagnosed after the resection (Fig. 3). The excised mass underwent histopathological microscopic examination after the resection, which confirmed the presence of mature tissues that predominantly contained ectopic central nervous tissue, osseous tissue, and bronchial mucosal tissue in most areas (Fig. 4). However, only a few cells were similar to primitive neurons in the focal areas. Immunohistochemistry showed GFAP(+), Olig-2 (+), IDH1(-), p53(-), CD34 vessel(+), H3K27M(-), Calretinin(+), NeuN(+), NSE(+), and Ki-67(+)1%.

The neonate was admitted to the neonatal department and discharged after one week. MRI showed that there was a tumor in the right posterior palate. This baby requires reoperation in the future.

Discussion

Oropharyngeal teratoma was classified by Ehrich (1945) based on the location of origin as episphenoid, epipalatine or epiranus, and epignathus tumors.[4] Teratomas can be classified into four groups according to histopathological microscopic examination: dermoid cysts, teratoid cysts, teratomas, and epignathi.[5] However, the term epignathus tumor is generally understood to denote neonatal oropharyngeal cavity teratomas without specifying the original location.[6] The histopathological microscopic examination of this case showed the presence of three highly differentiated germ-layer derivatives.

The etiology of epignathus is still poorly understood. Many studies have reported that fetuses with epignathus had no chromosomal abnormalities.[7,8] However, in a few studies, chromosomal abnormalities such as trisomy 13, ring X-chromosome, mosaicism with inactive ring X-chromosome, gonosomal

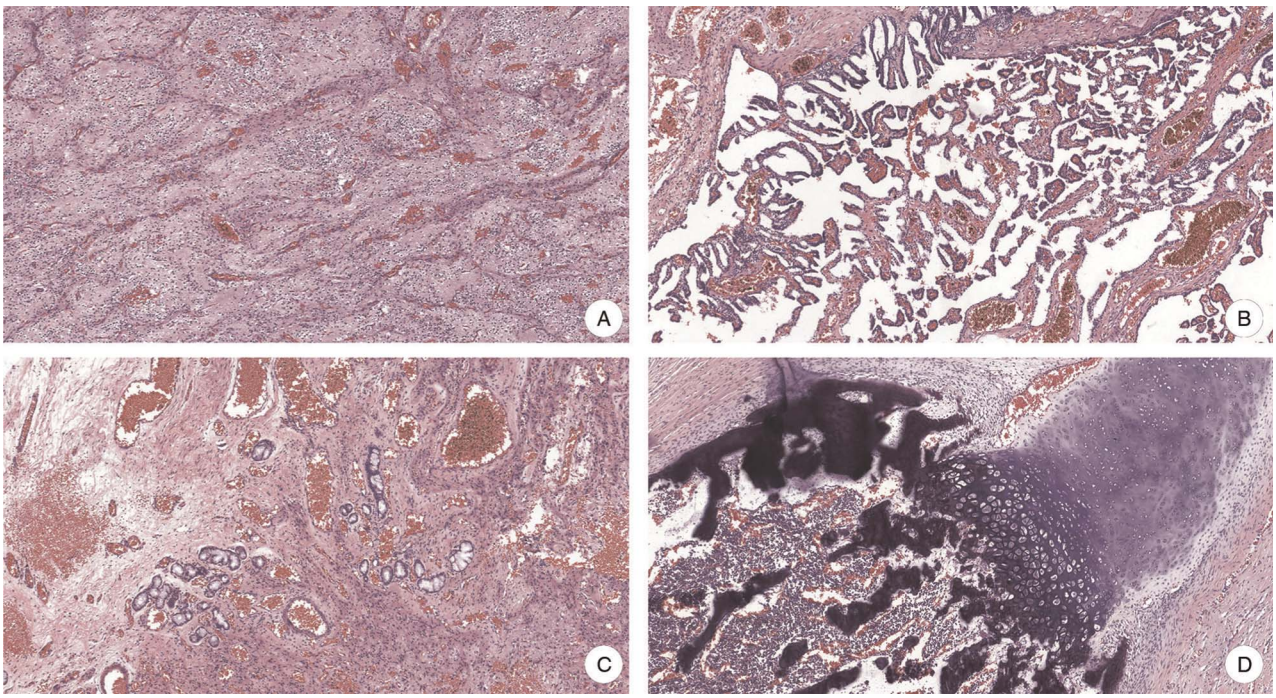

Figure 4. Microscopic examination of histopathological staining (hematoxylin-eosin staining, ×100). A Central nervous tissue-neuroglia. B Central nervous tissue-choroid plexus. C Salivary gland. D Osseous and cartilage tissue.

pentasomy 49, and other gene mutations have been found in neonates with epignathus.[9-12] Ten percent of epignathus may exhibit associated abnormalities.[13] The most common associated malformation is cleft palate, and the likely reason is that the palate is prevented from closing because of the tumor. In our patient, only a small cleft palate was diagnosed, whereas other reported associated malformations such as bifid tongue and/ or bifid nose, diaphragmatic hernias, glossoptosis, and inguinal hernias were absent.[14] Fusion of the primitive tongue buds can be impaired by the early development of the epignathus with subsequent anterior positioning of the tongue; this process is known to cause bifidity and glossoptosis. [5]

The prognosis depends on the location, volume, pathological classification of the mass, and associated malformations of the fetus. Given that the umbilical cord and placenta perfuse the fetus, a giant epignathus may not cause many significant problems during pregnancy, but it may influence fetal swallowing and result in polyhydramnios However, as the umbilical cord is clamped immediately after delivery, a giant nasopharyngeal teratoma may cause airway obstruction and neonatal mortality. Some authors have reported that epignathus is infrequently associated with an intracranial extension, and prenatal intracranial teratomas have been reported to result in poor prognosis. [14–16]

It is necessary to identify chromosomal abnormalities and associated malformations to avoid lethal malformation in neonates. Hence, prenatal diagnosis is extremely important. In this case, we recommended the mother to undergo amniocentesis in the early second-trimester for fetal karyotyping, but she refused. Therefore, we offered a complete ultrasound and MRI examination to evaluate the fetus's condition, which confirmed that there was no central nervous system involvement or serious associated malformations.

Some studies reported that elevated maternal AFP levels can be detected in the first-trimester[3,8,17] as in our case. High serum levels of maternal AFP indicate the presence of a tumor that can be confirmed by ultrasonography.[18] The ultrasound can identify the lesion even at a gestational age of 15–17 weeks.[19] The reconstructed images from 3D ultrasonography can allow spatial evaluation of the lesion, provide more detailed information about the location and extent of the epignathus, and assess its positional relationship with the anatomical structures of the face and head. Fetal MRI is helpful to assess the relationship between the mass and airway structures and any likely intracranial invasion that can lead to poor prognosis of the fetus.[20] Comprehensive assessment during pregnancy provides more details about the developing fetus and allows parents to better understand the condition affecting their unborn child and further counseling to understand the prognosis.[15] In our case, the MRI examination did not show any evidence of fetal intracranial extension or central nervous system-related anomalies.

Differential diagnoses of the epignathus include nasal glioma, oro- or nasopharyngeal cystic lymphangioma, giant epulis, sphenoid meningoencephalocele, nasoeth-moid meningoencephalocele, and rhabdomyosarcoma.[21] Although it is difficult to distinguish these conditions by prenatal examination, the mode of treatment is rarely affected.

Because the tumor mass can rupture, bleed, or block the birth canal, it is best to perform cesarean section to deliver the baby. If airway obstruction is suspected prenatally, an EXIT procedure should be considered. The EXIT procedure can preserve the intact placental circulation to the fetus and provide enough time to manage the fetal airway. At the time of cesarean section, tracheal intubation or tracheostomy should be performed before the umbilical cord is clamped,[13] and the mass can be resected on placental support. This process is called operation on placental support.[19] However, during this procedure, placental abruption, placental laceration, or uterine relaxation because of general anesthesia can lead to maternal and fetal hemorrhage.[22,23] The duration of placental support to the partially delivered fetus may be less than 60 min.[24] The prognosis of the fetus depends on tumor resectability, even though the EXIT procedure can successfully manage the fetal airway. Only cases wherein the tumors are resected after birth can survive until discharge.[25]

Prenatal assessment of airway status is essential to plan an EXIT procedure and is still not effectively addressed. As in this case, the EXIT procedure was prepared for but was eventually not required, as the neonate cried immediately after its head was delivered. This confirmed that there was no severe airway obstruction. Prenatal ultrasound and MRI are primary methods to assess fetal airway obstruction. The trachea-esophageal index is defined as the degree of displacement between the esophagus and the trachea and can predict the degree of difficulty in securing the airway. A trachea-esophageal index score of <5 predicts a difficult airway.[26] Ultrasound static imaging and visualization of the fetus swallowing and further confirmation by fetal MRI can also be used to determine airway patency.[27] Mohammad *et al.* reviewed several specific cases in an attempt to determine whether certain malformations or airway obstruction characteristics could help predict certain airway interventions. They did not observe a clear trend for most of the malformations. The size or anatomical distortion degree of the cervical teratoma did not predict a single type of airway intervention.[28] However, prenatal imaging that shows anatomic compression, solid mass, or polyhydramnios is linked with potential airway intervention.[29] In this case, the MRI examination did not show any evidence of anatomic compression, which increased our and the parents' confidence in the successful rescue of the fetus.

In this case report, we discuss the characteristics and outcome of epignathus with a small cleft palate in a fetus. The fetus was diagnosed to have a giant epignathus by ultrasonography in the second trimester. Regrettably, fetal karyotype analysis was not conducted owing to the mother's refusal. The MRI performed before the operation excluded central nervous system involvement and abnormal nose or tooth. Prenatal ultrasound and MRI evaluation did not show any evidence of anatomic compression. For the safety of the neonate, EXIT was prepared for but was eventually unnecessary. The neonate successfully survived because of immediate and successful management postpartum. Immediate oropharyngeal mass resection with tracheal cannula was performed under local anesthesia, and most of the mass was removed. Nonetheless, the baby still needs a second surgery in the future. More methods for prenatal assessment of airway obstruction would be helpful for prenatal counseling and prognosis assessment of the fetus.

Conclusions

Epignathus is a rare congenital oropharyngeal teratoma that in most cases is associated with malformation and considered fatal death. Prenatal assessments by ultrasound and MRI are

very important to facilitate counseling and improve the understanding of prognosis. The EXIT procedure is a good approach to improve survival in neonates with epignathus.

Funding

None.

Conflicts of Interest

None.

Data Availability

Data sharing not applicable to this article as no datasets were generated or analyzed during the current study.

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