Congenital Giant Epignathus with Intracranial Extension in a Fetal

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Epignathus is a rare congenital orofacial teratoma. The incidence of epignathus is between 1:35,000 and 1:200,000 live births with a female predominance. We reported an uncommon male fetal case of epignathus originating from the right maxillofacial region with upward to intracranial.

A 355 g male fetus was aborted at gestation age of 17 weeks and 6 days by a 31 years old healthy woman with her first gravidity at Haidian Maternal and Children Health Hospital. An ultrasound scan showed no anomaly at 12th gestational weeks (GWs). Down syndrome screening test showed a high risk of open neural tube defects at 16th GW. Three-dimensional ultrasound scan indicated an irregular anechoic mass of the size 7.0 cm × 4.6 cm arising from the fetus's right maxillofacial region at 17th GW [Figure 1a]. Chromosomal analysis showed a normal male karyotype.

The couple was not consanguineous marriage, and the pregnancy was uncomplicated with any infection or teratogenic exposure. After a detailed consultation, they opted for termination of pregnancy and signed an informed consent regarding to the autopsy.

Fetal autopsy examination showed a large complex mass of the size 6.7 cm × 6.5 cm × 5.0 cm protruding from the right maxillofacial region. It was dark red and the surface was uneven and multi-lobulated [Figure 1b]. Facial features were deformed due to the compression of the mass. Part of the mass that connected to the hard palate was visible through the open oral cavity.

On cut section, the mass was mainly cystic and contained soft gray-white tissue like brain. The craniocerebral tissue was dissected after formaldehyde fixation, and the mass was found growing upward to intracranial. The diameter of intracranial extension section was 3.5 cm, which mainly located in middle and posterior of cranial fossa [Figure 1c]. No cerebellum and brainstem were detected.

Microscopic examination showed that the majority of the tumor was immature neural epithelial tissue. The main components were tubules lined by dark hyperchromatic cells with stratification [Figure 1d]. Surrounding the immature neuroepithelial tubules, there were mature tissues, which included cartilage, hepatic tissue, salivary components, and squamous epithelium. The final pathological diagnosis supported congenital epignathus (immature teratoma) with intracranial extension.

Epignathus is a rare congenital teratoma located in oropharyngeal region. Exact etiology of epignathus is not known. It may arise from pleuripotential cells in the region of Rathke's pouch.[1] Epignathus teratoma may vary in structure and degree of differentiation and be classified as mature, immature, and malignancy. The immature and malignant types have more significantly aggressive behavior. In addition to the size and exact location, fast-growing speed is also a risk factor associated with poor prognosis.[2]

Most epignathus teratomas were detected during the second and third trimester by two-dimensional or three-dimensional ultrasound.[3] Early radical excision

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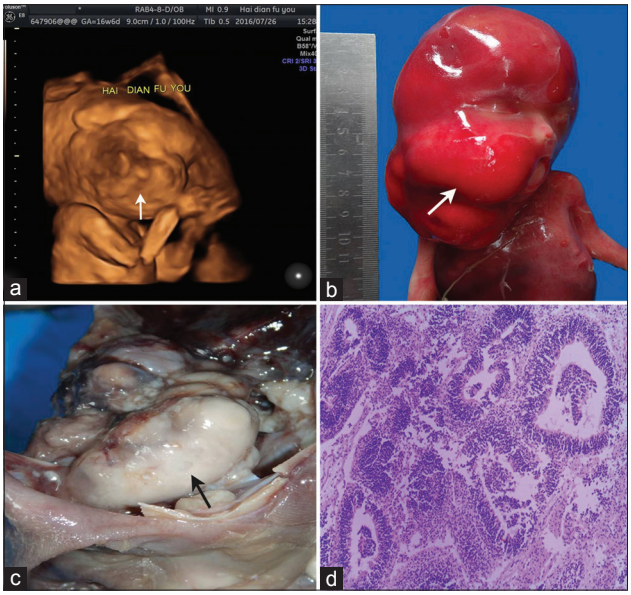

Figure 1: (a) Three-dimensional ultrasound scan showed an irregular anechoic mass at the fetus's right maxillofacial region. (b) A large complex mass protruding from the right maxillofacial region with uneven and multilobulated surface. (c) The intracranial extension section of the mass mainly located in middle and posterior of cranial fossa. (d) The main components of the tumor were immature neuroepithelial tissue, which presented many tubules lined by dark hyperchromatic cells with stratification.

is the treatment of large head and neck teratoma without intracranial extension. There were reports of successful delivery of live fetuses by *ex utero* intrapartum treatment of fetus with isolated epignathus.[4]

There is no evidence suggesting that epignathus caused by environmental agents, mendelian or polygenic inheritance. Sporadic chromosomal changes had been reported, but usually, it has not reported to be associated with epignathus.[5] Chromosomal analysis of this case revealed a normal male karyotype.

No evidence showed epignathus has genetic predisposition. The probability of having more than one child with epignathus in the same family is very low. This information is critically important when providing counseling to the parents regarding to the next pregnancy.

In conclusion, this case was very rare. In case of an early detection of fast-growing fetal epignathus, especially with intracranial extension, pregnancy termination should be considered.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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