

A Case of an Epignathus with Intracranial Extension Appearing as a Persistently Open Mouth at 16 Weeks and Subsequently Diagnosed at 20 Weeks of Gestation

Pavel Calda, MD, PhD,¹ Michaela Novotná, MD,¹ David Čutka, MD,² Miroslav Břešťák, MD,¹ Lubomír Hašlík, MD,¹ Barbara Goldová, MD,³ Ivana Vítková, MD,³ Manuela Vaněčková, MD, PhD,⁴ Zdeněk Seidl, MD, PhD⁴

¹ Charles University in Prague, First Faculty of Medicine, Department of Obstetrics and Gynecology of the First Faculty of Medicine and General University Hospital, Apolinářská 18, 128 51, Prague 2, Czech Republic

² Centre of Medical Genetics, Novohradská 68, 370 08 Budweis, Czech Republic

³ Charles University in Prague, First Faculty of Medicine, Department of Pathology of the First Faculty of Medicine and General University Hospital, Studničkova 2, Prague 2, 128 00, Czech Republic

⁴ Charles University in Prague, First Faculty of Medicine, Department of Radiology of the First Faculty of Medicine and General University Hospital, U Nemocnice 2, Prague 2, 128 08, Czech Republic

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ABSTRACT: We report a rare case of oral mass (epignathus) with intracranial extension originally suspected antenatally at 16 weeks' gestation because of a persistent open mouth. Postmortem MRI and pathologic examination of the fetus confirmed an oral teratoma with bilateral ventricular dilatation, corpus callosum agenesis, and a neuroepithelial intracranial cyst. The relevant literature regarding this anomaly is reviewed. © 2010 Wiley Periodicals, Inc. *J Clin Ultrasound* 00:000–000, 2010; Published online in Wiley Online Library (wileyonlinelibrary.com) DOI: 10.1002/jcu.20762

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An epignathus (also called pharyngeal teratoma) is a rare form of benign congenital teratoma originating from the base of the skull, most commonly the palate or the mandible. Associated

malformations may be present, which commonly include cleft palate and bifid tongue or nose. Previous reports have noted the association of epignathus with the duplication of the pituitary gland.^{1–4} Sporadic chromosomal changes have also been reported,^{5,6} but usually epignathus is not associated with chromosomal aberrations. Most of these teratomas are unidirectional and protrude through the mouth. Cases with intracranial extension causing destruction of the brain tissue have poor prognosis.^{7,8} Prenatal sonographic diagnosis of large protruding oral tumors is easy.⁹ In most cases, the diagnosis was made in the late second or third trimester. However, one case of early diagnosis at 15 weeks' gestation has been described.¹⁰ We report a case of a small, primarily nonprotruding oral mass first suspected at 16w + 3d and subsequent 2D and 3D sonographic imaging, counseling, and postmortem MRI with pathologic correlation.

CASE REPORT

A healthy 29-year-old primigravida with no medical history underwent a first-trimester ultrasound examination at 12w 3d, which was normal. A

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Correspondence to: P. Calda

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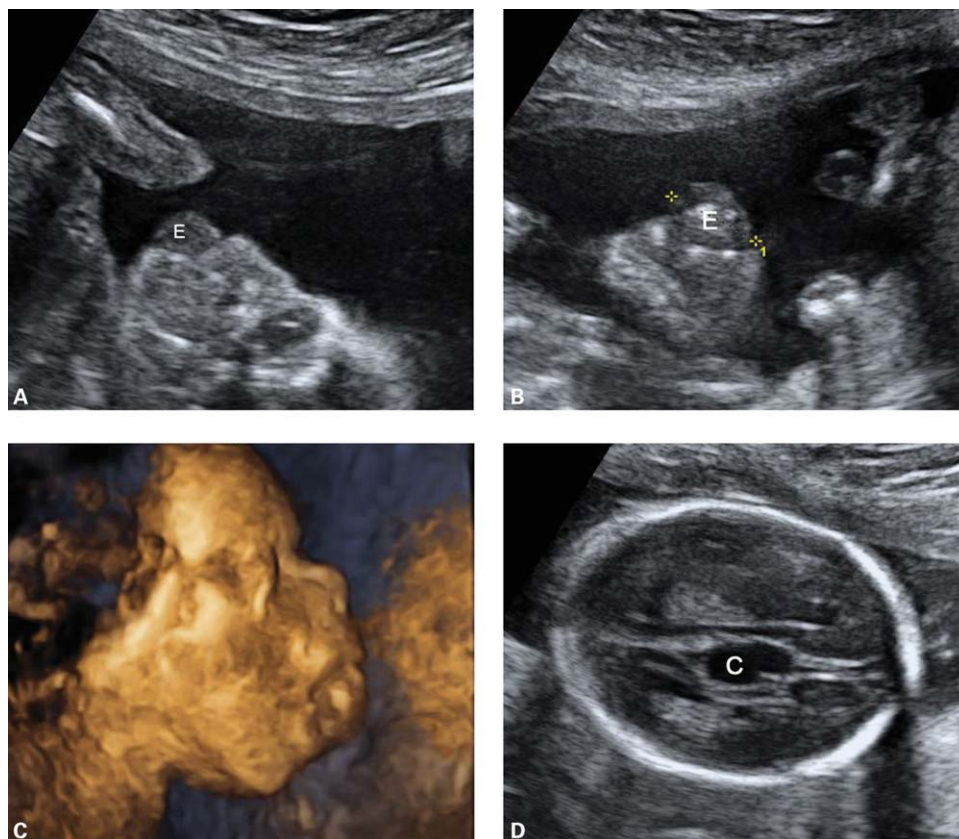


FIGURE 1. Sagittal (A) and transverse (B) sonograms at 20w 3d gestation show a ball-shaped oral mass (E) with an opened mouth. (C) 3D image of the fetal face with the tumor. (D) Sonogram shows cystic brain lesion (C).

screening test performed by her gynecologist was positive, with an individual Down syndrome risk of 1:230 based on the following: nuchal translucency, 2 mm; hCG, 4.4 MoM; PAPP-A, 0.8 MoM; AFP, 0.9 MoM; estriol, 0.9 MoM. Amniocentesis was therefore performed at 16w 3d, which yielded a normal female karyotype. During ultrasound examination performed by an expert sonographer prior to amniocentesis using an Acuson Siemens S 2000, with 6C2 and 9L4 probes and a Voluson E8, with a RAB4-8-D probe, the peculiar finding of a persistently opened mouth was noted, for more than 30 minutes. With thorough re-evaluation, a 2×2 mm mass was seen inside the mouth. As this finding was very small, re-evaluation in 4 weeks was recommended. A repeat 2D/3D ultrasound examination at 20w 3d showed an appropriately grown female fetus with a biparietal diameter of 48 mm, head circumference of 170 mm, abdominal circumference of 152 mm, and femur length of 31 mm. However, we found a ball-shaped mass of mixed echogenicity measuring 22×17 mm, originating from the mouth and filling the oral cavity (Figure 1). Color Doppler examination revealed hypervascularity within the mass. Cranial examination showed asymmetry of the lateral ventricles. Agenesis or partial agenesis of

corpus callosum was suspected, but the sonographic appearance did not allow a definitive diagnosis (Figure 1D). The placenta was located on the posterior uterine wall and the volume of amniotic fluid was not increased. The fetus did not swallow or close the mouth throughout the whole ultrasound examination.

The tumor appeared to be growing rapidly and the intracranial changes were prominent. Our working diagnosis was epignathus with intracranial extension. Although there are case reports of delivery of viable fetuses by ex utero intrapartum treatment of cases with isolated epignathus,¹¹ in our case, the presence of intracranial extension made the prognosis very poor. After nondirective counseling, the parents decided to have the pregnancy terminated.

On gross examination, the 310 g female fetus showed a lobulated round vascular mass approximately 2 cm in diameter, visible through the open oral cavity (Figure 2A).

Postmortem MRI was performed the same day (5 hours had elapsed between the termination and postmortem examination of the brain) to determine the extent and characteristics of the mass. Imaging consisted of high-spatial resolution in the three orthogonal planes with T2-weighted images.



FIGURE 2. (A) Postmortem photograph of the fetal face shows epignathus. (B) Postmortem MRI. Sagittal T2W image shows mass with inhomogeneous signal intensity in the oral cavity, displacing the tongue posterior.

The postmortem MRI showed the mass in the anterior oral cavity, preventing the mouth from closing, causing a posterior displacement of the tongue. Intracranial extension was not described (Figure 2B). Another finding was mild bilateral ventricular dilatation caused by corpus callosum agenesis with a paramedian cyst.

Histopathological examination confirmed the presence of a congenital teratoma with neural, myofibrillar, and fibrous connective tissue (Figure 3) covered by nonstratified squamous epithelium. The neuroepithelial intracranial cyst measured $13 \times 10 \times 7$ mm and agenesis of corpus callosum was confirmed (Figure 4).

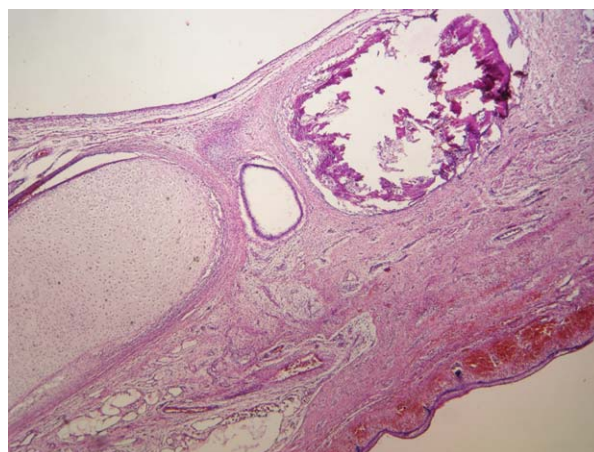


FIGURE 3. Histopathological examination of the teratoma shows cartilage, bone, and dermal tissue.

DISCUSSION

Teratomas are tumors derived from pluripotent cells and made of elements of different types of tissue, from one or more of the three germ cell layers. Brain tissue is the most frequent component along with cartilage, bronchial epithelium, and ependyma-lined cysts. An epignathus is an extremely rare form of teratoma in which the tumor is attached to the base of the skull, usually the hard palate, or the mandible. The incidence is approximately of 1 in 35,000 to 1 in 200,000 live births. They occur more frequently in females than males, with a ratio of 3:1.¹² Ultrasound can give a reliable diagnosis, when the finding has the typical appearance.¹³ In our case, the lesion was observed from the very early stage, when it was a very small oral soft-tissue mass, leading to a wide differential diagnosis. The differential diagnosis of a mass in the fetal or neonatal oral cavity should include teratoma, congenital epulis (rare benign oral cavity tumor

originating from anterior alveolar ridge), hemangioma, lymphatic malformation, sincipital cephalocele, dermoid cyst, and other benign or malignant soft-tissue masses (tongue teratoma).¹⁴ The parasitic twin syndrome has also been described in this location.^{11,15,16} Oral teratoma may show heterogeneous signal intensity on MRI due to variable contents such as cyst, calcifications, and fat. Hemangioma presents as a solid mass with disorganized vascularization of high-flow vessels and intense contrast enhancement on MRI. Lymphatic malformation and cephalocele present with cystic characteristics. Dermoid cyst has fat intensity signals, with preserved crista galli, contrary to the nasal cephalocele, where crista galli is not preserved.

The clinical presentation of an epignathus varies. The tumor is mainly diagnosed on a routine 2D sonographic examination. Polyhydramnios, hydrops, fetal death, and pre-eclampsia can accompany the epignathus. Impaired fetal

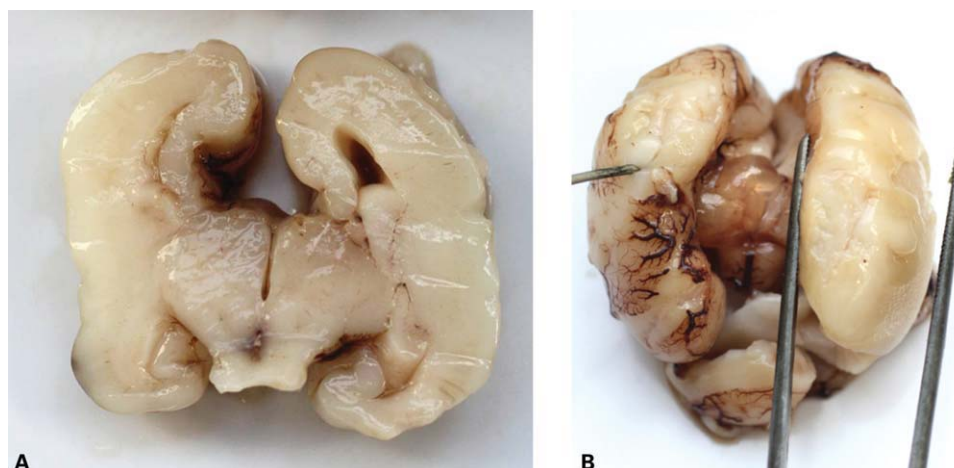


FIGURE 4. Pathologic examination confirmed agenesis of the corpus callosum (A) and the intracranial cyst (B).

swallowing can lead to polyhydramnios. Epignathus can be accompanied by elevated maternal serum alpha-fetoprotein, which requires a very detailed examination of the fetus.¹⁷ In our case, the alpha-fetoprotein level was normal. The size and exact location of the oral mass are important prognostic factors because larger tumors and nasopharyngeal involvement can cause both feeding difficulties and mechanical airway obstruction.^{18,19} These prognostic factors should be taken into consideration in determining the optimal management plan.²⁰ In potentially viable fetuses, when severe impairment is detected or suspected, the neonatal team should be prepared to rapidly secure the neonate's airway.

Precise determination of the spread of the tumor to the surrounding tissues is very important, because with intracranial spread, the prognosis is poor, ie, no surviving cases have been reported in the literature.²¹ Termination of pregnancy as an option for management should be discussed, as occurred in our case.

The prognosis of pharyngeal teratoma is very poor.^{22,23} Intrauterine fetal demise in up to 50% and neonatal demise of 50% of the survivors have been reported. Only six cases of infants surviving a pharyngeal teratoma have been reported over the last 10 years.

Some infants required plastic surgery for the cleft palate or lip, while others had just minor repair of luxation of the mandible. Currently there is no evidence to suggest epignathus is caused by environmental agents, Mendelian or polygenic inheritance, or chromosomal abnormalities. No cases of recurrent epignathus in one family have been reported in the literature.¹ This information is of essential importance when counseling the parents toward the next pregnancy. Our case illustrates that the suspicion

arising from an odd finding (an open mouth) seen early should trigger a sonographic follow-up.

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