

Sonographic evaluation of epignathus

Tsitouridis I, Sidiropoulos D, Michaelides M

Department of Diagnostic and Interventional Radiology, Papageorgiou General Hospital, Thessaloniki, Greece

Abstract

Epignathus is extremely rare form of teratoid tumor arising from the palate or pharynx in the region of basisphenoid (Rathke's pouch), filling the buccal cavity and protruding from the mouth. This tumor usually causes death in neonatal life because of its location and because surgical removal is often impossible. Prenatal sonographic diagnosis is important for genetic counseling, obstetric management and in some cases neonatal surgery. We present a case of epignathus originating from the hard palate in a female child delivered at gestational week 33 by caesarian section due to increasing hydramnios. Hippokratia 2009; 13 (1): 55-57

Corresponding author: Michaelides M, Department of Diagnostic and Interventional Radiology, Papageorgiou General Hospital, Thessaloniki Ring Road, Nea Eukarpia, 56403 Greece, Tel: +302310693336, Fax: 00302310693320, e-mail: michaelidesm@yahoo.com

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Fetal teratoma originating from the mouth is extremely rare. Epignathus is a teratoma originating from the upper jaw, usually connected with the sphenoid bone or hard palate. This tumor can lead to death soon after birth, causing severe airway obstruction. Prenatal sonographic diagnosis is very important for surgical planning when operation is possible. The aim of the present study is to report a case of epignathus originating from the hard palate, in a newborn female child 33 weeks old.

Case report

A 35 year old healthy woman presented at Papageorgiou Hospital on May 2007 during her third pregnancy with no known malformations or other hereditary factors in her or her husbands family. She had two healthy children 8 and 4 years old. There were no problems in the first and second trimester and previous sonographic evaluation was normal. After week 29, signs of accelerating growth were seen and she complained of pain in the lower abdomen.

Sonographic evaluation revealed a mass 10x12 cm in the anterior neck area. Although its origin was difficult to establish, it appeared to originate from the mouth. The mass had complex echogenity, with solid and small cystic components (Figure 1A). Areas of calcifications with acoustic shadowing were noted (Figure 1B). There was also polyhydramnio (Figure 1C). Because of its appearance, a teratoma was considered. The posterior neck had no abnormalities and the fetal skull and vertebral column were intact. Due to accelerating hydramnios and uterine contractions, caesarian section was performed at week 33. The newborn was female, had severe difficulty in breathing, a low heart rate (<90) and died 15 minutes after birth. A large tumor was protruding from the mouth (Figure 2).

The child had no signs of malformations, with a birth weight of 1210 gr (without tumor) and a length of 32 cm. Skull perimeter was 27 cm. The polypoid mass (13x9x7 cm) was originating from the hard palate and different tissue structures were seen including bone, cartilage and skin, macroscopically signs of teratoma. Cleft lip and cleft palate were seen after the removal of the tumor (Figure 3).

In microscopic examination of the tumor, a variety of tissues originating from the three germ layers were seen, including neural tissue (ganglion cells and fibers), cartilage, bone, cavities lined by keratinized multilayered squamous epithelium, sebaceous glands and respiratory epithelium. All these elements were inside a fibrous stroma. These structures were typical teratoma, consistent with pharyngeal teratoma (Epignathus).

Discussion

Dermoid cysts or teratomas of head and neck occur almost exclusively in infants and young children¹. Teratomas are complex tumors, with varying amounts of tissues originating from the three germ layers. Malignant changes, which are common in teratomas in other parts of the body, have not been observed in these tumors².

The term epignathus (according to the name given by Geoffrey and Hillaire in 1837), implies a tumor arising from the jaw, though it is commonly used to describe tumors or teratomas of the mouth in the newborn³. Congenital teratomas have an incidence of 1 in 4.000 live births and those arising from oropharyngeal cavity comprise less than 2%. Sixty percent occur in the nasopharynx and predominately in girls⁴. To date, only 117 cases have been reported in the medical literature since Brown-Kelly (1918) made the first report³.



Figure 1: A. Polypoid mass with mixed echotexture (mostly solid) protruding from fetus mouth. Skull is intact. B. Acoustic shadowing due to calcifications (arrow). Note also cystic components of the tumor (arrowhead). C. Polyhydramnio.

Figure 2: Female child with a polypoid mass arising from the mouth. No other malformations are noted.

Figure 3: Cleft palate and cleft lip after removal of the tumor.

In 1940 Ewing⁵ classified these nasopharyngeal tumors into: a. Dermoids-consisting of epidermal and mesodermal germ layers, attached to soft/hard palate and/or pharynx near midline. Minimal or extensive intracranial extension may be present with skin cover. b. Teratomas-consisting of all three germ layers, with an indifferent degree of organization. It differs from type 1 by their greater structural complexities, earlier development and larger size. c. Epignathus, consisting of teratomas with high degree of organization and recognizable structures. There is presence of formed organs.

Nasopharyngeal teratomas are not true neoplasms but are developmental malformations in which the potential cells from one or more germ layers escape the physiological constraints which under normal circumstances govern the growth and development of an embryo. Instead, these embryonic cells proliferate, differentiate and form disorganized and disorderly conglomerates of various tissues that form a sessile or pedunculated mass that is attached to the nasopharynx².

Epignathia are spontaneously arising orofacial teratomas. They show a marked degree of somatic differentiation and organization and the term "fetus in fetu" is sometimes applied to these lesions because of their resemblance to neonatal fetal skeletal structures^{6,7}. No familial cases have been published and the genetic risk of having two or more children with epignathus in the same family is probably very small⁷. First or second trimester prenatal diagnosis of epignathus has not to our knowledge been reported. The complex distribution of these tumors in a teratoma, generates a sonographic image that may

be indistinguishable from other lesions that are simpler in composition and structure. This fact and the rareness of epignathus are the reasons for the difficulty of early diagnosis⁸.

Intrauterine diagnosis of epignathus using ultrasonic tomography has been first reported in 2 papers^{9,10}. Kang and his colleagues diagnosed epignathus on the grounds that a tumor, characterized by complicated echo patterns, was found to be sticking to the frontal surface of the fetal head. They estimated that this tumor had originated from malformed twins or from the placenta⁹. In the study of Chervenack et al., the coexistence of solid and cystic echo patterns and calcifications suggested a teratoma¹⁰. Shouno et al tried to rule out the possibility of encephalomeningocele or hygroma, but the large size of the tumor could not rule out the possibility that it was attached to the neck¹¹.

In our case the mixed echotexture of the mass, the presence of calcifications, hydramniosis and the relation to the mouth made epignathus the most possible diagnosis. Hydramniosis was probably due to obstruction of the fetal oropharynx, causing failure of fetal swallowing. It has also been suggested that polyhydramnio associated with epignathus may be secondary to excessive cardiac demands caused by an extensively vascular tumor mass¹². The tumor was unlikely to represent hygroma or a neural tube deficit, considering the features of the tumor and the absence of any deficit in the skull or the spine. Chervenack's group stated however that distinction of the tumor from hygroma or a neural tube deficit is difficult in the case of a large tumor covering the area of the head and neck¹⁰.

Early diagnosis is of great value for genetic counseling and obstetric management. In the present case, the newborn died 15 minutes after birth because it was impossible to operate on the tumor. However advances in neonatal and even antenatal surgery might provide good future opportunities for early handling of this malformation. It might be possible to diagnose this malformation even in the second trimester through an antenatal screening program using ultrasound, which can result in better results to children's survival.

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