



Fetal Oropharyngeal Teratoma: A Case Report

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Abstract Oropharyngeal teratomas are extremely rare fetal tumors and originate from pluripotent stem cells. The tumor typically arises from the palato-pharyngeal region around the basishenoid. These tumors are heterogenous in nature. The reported case was diagnosed at 20 weeks gestation. Ultrasound revealed a heterogenous, lobulated facial mass protruded from the oral cavity without any hypervascularity or intracranial extension. There was no evidence of polyhydromnios. After counseling, the parents opted for termination of pregnancy. Histopathological examination from the exophytic mass was consistent with immature teratoma containing components of all three germ cell layers. Genetic testing and fetal autopsy could not be arranged due to financial and infrastructural constraints.

Keywords Oropharyngeal teratoma · Facial tumor · Ultrasonography

Introduction

Teratomas generally constitute 25–35% of congenital tumors [1], with an incidence of 1/4000 births. Most commonly they affect the sacrococcygeal and gonadal regions. Head and neck teratomas are rare, of which less than 1% develop in the oropharynx [2], with an incidence of 1/35,000–1/200,000 births [3]. Early diagnosis with

ultrasound and magnetic resonance imaging (MRI) is essential to plan management.

Case Report

A healthy primigravida of 25 years presented to our fetal medicine unit in Kolkata for a second opinion at 20 weeks of gestation having had a routine anatomy scan elsewhere where a large tumor on the fetal face was found. She was “low risk” for trisomy 21 on first trimester combined screening with nuchal translucency and double markers at 12 weeks. There was no history of comorbidities and no known exposure to ionising radiation. There was no family history of congenital anomalies of this non-consanguineously married couple. Sonographic evaluation revealed a single live intrauterine fetus and fetal biometry measurements corresponded with gestational age. A normal appearing placenta was located on the upper posterior segment of the uterus with normal liquor amnii volume. A mass lesion of 49 × 50 mm covering the entire lower half of the fetal face was observed, containing both solid and cystic components (Fig. 1). The lobulated tumor seemed to have arisen from the buccal cavity (Fig. 2) and was avascular on colour doppler application. The soft tissue extended up to the lower margins of both orbits. However, fetal nasal bone (Fig. 1) and the maxillary border of the hard palate could easily be demonstrated on ultrasound (Fig. 3). Both orbits were well developed with clear margins (Fig. 4) but fetal lips could not be clearly delineated. Fetal brain structures were well demonstrated with no evidence of intracranial extension. Fetal stomach bubble seemed normal suggesting patent upper gastrointestinal tract. The rest of the fetal anatomy was also normal. The mass was provisionally diagnosed as an oropharyngeal

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Fig. 1 Profile view of fetal face showing an intact nasal bone and a complex mass arising out the fetal mouth

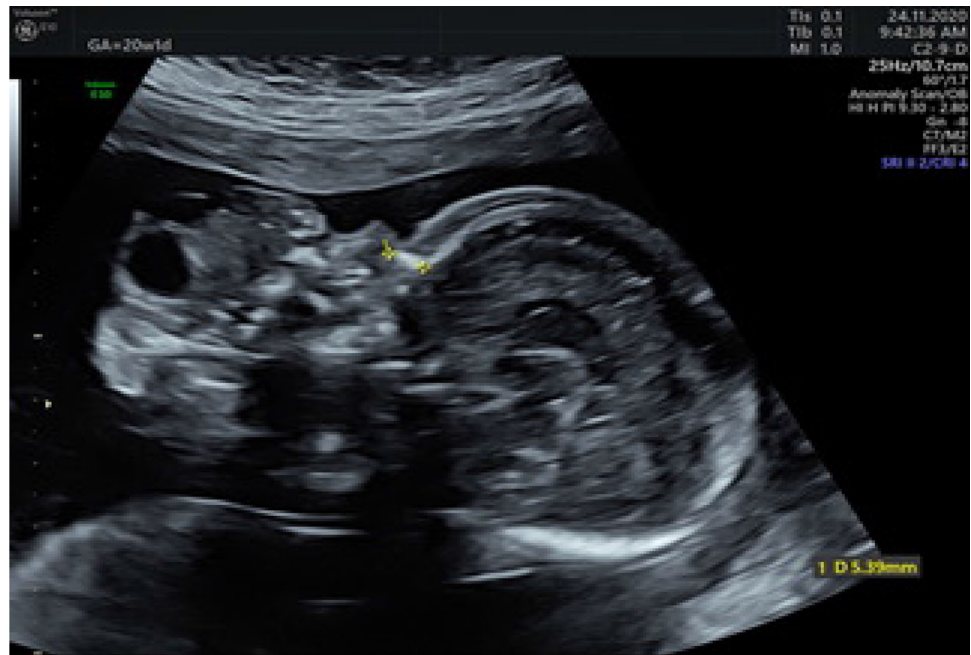


Fig. 2 3D sonography with HD Live rendering showing lobulated mass on fetal face



teratoma with differential diagnoses of hemangioma or a tumor of neural origin. After appropriate counseling about the findings and the high possibilities of poor prognosis, the parents opted for medical termination of pregnancy which was carried out at a peripheral unit with basic facilities only. A female fetus of 525 g was delivered with an exophytic tumor protruding through the oral cavity (Fig. 5). The tumor was dark red, lobulated, partly solid partly cystic and locally invasive into underlying skin and subcutaneous tissues. Specimen from the lesion was sent for

histopathological examination and a genetic counseling was arranged.

Due to lack of resources, we could not investigate the case further to gather information which could help us in prognosticating the case and predicting the chances of recurrence in future pregnancies. MRI may have added more value for better visualisation of tumor extension and guide further management [1]. Fetal autopsy would have been helpful to find out the origin of the tumor and involvement of adjacent fetal parts. The risk of recurrence could not be ascertained, as no genetic testing was done.

Fig. 3 Clear maxillary border of hard palate



Fig. 4 Well developed orbits and lenses with clear margins



Discussion

Teratomas are true neoplasms originating from the cells of all three germ lines. They may contain fat, cartilage and bone. Oropharyngeal teratomas appear as heterogenous masses with solid and cystic components and are believed to be the result of migration and entrapment of mesoderm and endoderm with ectoderm during embryogenesis. The tumor typically arises from the palato-pharyngeal region around the basisphenoid (Rathke's pouch). With progressive growth it fills the buccal cavity and finally protrudes out of the mouth [4]. Depending on histopathology, teratomas can be classified as immature or malignant and mature or benign. Congenital teratomas are usually benign and have a predominance to female genders.

Oropharyngeal teratoma may be isolated or present in combination with other craniofacial malformations, most commonly cleft palate. They may also be seen in association with frontonasal dysplasia and/or various degrees of craniofacial duplications.

Clinical presentation of such cases may vary and such neoplasms may appear in different stages of fetal development. High levels of maternal serum alpha-fetoprotein (AFP) on second trimester screening tests can provide a clue for such fetal anomalies [5]. In our case, parents denied the AFP test due to financial constraints. Instead, they preferred histopathological examination following termination. These tumors frequently cause increased amniotic fluid volume due to inability of the fetus to swallow. However, in our case the amniotic fluid volume



Fig. 5 Photograph of aborted fetus with lobulated midline facial mass

was normal because of preserved swallowing mechanism as the palate and maxilla were unaffected by the tumor.

Teratomas generally appear as solid-cystic mass on sonography. The solid components are often nonhomogeneous. On microscopic analysis, the tumor comprises of ectodermal, mesodermal and endodermal components containing squamous and glandular epithelium, mature bones and cartilages and immature neural elements. Similar sonographic, macroscopic and microscopic findings in our case lead us to reach the diagnosis in our case. 2D sonography can diagnose the presence of the tumor as well as its extension with a few exceptions [6]. In the present case, 3D ultrasound was also helpful in assessing external facial anatomy. It provided images that were understandable by the parents and facilitated counseling.

No specific chromosomal abnormalities or inherited disorders are found to be associated with oropharyngeal teratoma, often called epignathus. They can either be isolated, like in our case, or can be associated with other anomalies or part of a syndrome. A very few case studies have indicated the association with chromosomal aberrations like 49 XXXY (XXXY syndrome), 1q, 19q duplications [7]. Furthermore, HLXB9 homeobox gene (*MX1* gene) mutation on chromosome 7q36, associated with Currarino Syndrome or Currarino Triad (OMIM #176450)

has been identified with fetal teratoma [8]. Due to the inconsistent evidence, it is difficult to narrow down the testing options. Though no specific copy number variation (CNV) has been established as yet, a high-resolution chromosomal microarray on the tumor tissue DNA was planned to be the first step of investigation to rule out any obvious CNV. Based on the outcome of the first investigation, whole-exome sequencing or germline cancer panel on the stored somatic DNA could have been offered to confirm involvement of any single gene defect, if any.

The prognosis of oropharyngeal teratoma is generally poor and mortality is attributed to airway obstruction [9]. Prognosis depends on the size of the lesion, the involvement of vital structures and other associated anomalies. A multidisciplinary approach is needed for successful management and surgery remains the mainstay of treatment. Intracranial extension must be excluded before surgery. In case of hydrocephaly or intracranial involvement neonatal surgery is avoided because of a poor prognosis [10]. No recurrence was reported after complete resection [11]. Ex-utero intrapartum treatment (EXIT) procedure, during the cesarean section has also been reported to secure the airway with intraoperative intubation or tracheostomy before cutting the umbilical cord [12].

Conclusion

Antenatal diagnosis of oropharyngeal teratoma is useful for parental counseling and helps them make an informed choice. The final diagnosis should be based on histology. Termination, where permissible, may be recommended but that would also depend on parental choice. Finally, it is important to reassure parents about extremely low risk of recurrence in subsequent pregnancies.

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