Case Report

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Prenatal diagnosis and perinatal management of a giant fetal cervical mass with airway compromise: a case report

Abstract

We present the case of a 32-year-old pregnant woman, G3P2, with no relevant medical history, referred to a tertiary care center due to the prenatal detection of a fetal cervical mass. The initial diagnosis was made at 23 weeks + 5 days of gestation, revealing a large mass located at the lower mandibular region, with mixed components and marked vascularization on color Doppler, associated with significant fetal neck hyperextension. Serial ultrasound follow-up demonstrated progressive growth of the lesion, polyhydramnios, and suspected airway compression. These findings led to delivery planning by cesarean section with an EXIT (Ex Utero Intrapartum Treatment) procedure at 38 weeks of gestation, successfully securing the neonatal airway through orotracheal intubation at birth. In the postnatal period, the newborn was diagnosed with cystic hygroma and acyanotic congenital heart disease, requiring interventional management with serial drainages and bleomycin sclerotherapy, and remaining under mechanical ventilation. This case highlights the importance of prenatal diagnosis, multidisciplinary management, and perinatal planning in fetuses with congenital cervical masses and risk of airway obstruction.

Keywords: fetal cervical mass, cystic hygroma, cervical teratoma

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Introduction

A 32-year-old patient, G3P2, with two previous deliveries and no relevant medical history, received adequate institutional prenatal care at the Guatemalan Social Security Institute (IGSS), completing a total of 14 prenatal visits and six ultrasound examinations during pregnancy. Maternal infectious screening tests were negative. She was referred to a tertiary care center following the ultrasound detection of a fetal cervical mass. The initial diagnosis was established at 23 weeks + 5 days of gestation, identifying a mass located at the lower mandible measuring approximately 8 × 5 cm, with mixed echogenicity and marked color Doppler vascularization, causing significant fetal neck hyperextension (Figure 1). On follow-up ultrasound at 29.4 weeks, polyhydramnios was documented, with an amniotic fluid index of 33 cm. A heterogeneous multilobulated mass was observed at the lower mandibular region, predominantly cystic with solid components, measuring approximately 7.5 × 8.3 cm. Airway compression could not be ruled out (Figure 2). At 38 weeks of gestation, a new ultrasound revealed a multilobulated mass with mixed echogenicity located in the anterior neck, consisting of a larger predominantly anechoic lobe with thin septations and a smaller predominantly hyperechoic solid-appearing lobe, measuring approximately 12 × 10 × 8 cm in total. The mass caused probable displacement and compression of the airway and esophagus, with persistent fetal head hyperextension. Swallowing movements were observed during the examination.

Mild pulmonary hypoplasia was noted. The stomach contained fluid, cardiac anatomy appeared normal, and fetal genitalia were male. Polyhydramnios persisted, with an AFI of 34 cm, and fetal biometry was consistent with 38 weeks of gestation (Figure 3, 4). Due to the high risk of neonatal airway obstruction, delivery was planned by cesarean section at 38 weeks, performing an EXIT procedure to secure the airway at birth. Successful orotracheal intubation was achieved without immediate complications. The male newborn weighed 3,600 g at birth, measured 47 cm in length, and had Apgar scores of 9 and 9.

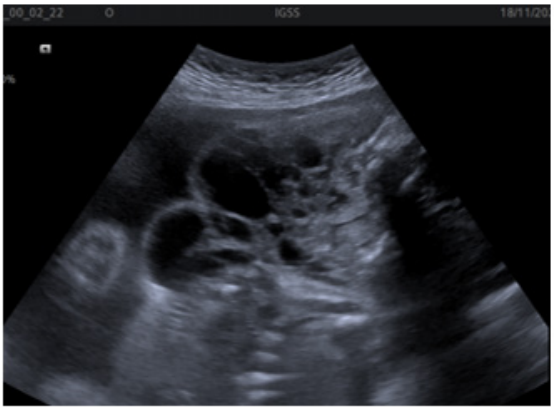

Figure 1 Multilobulated cervical mass at 29 weeks of gestation.

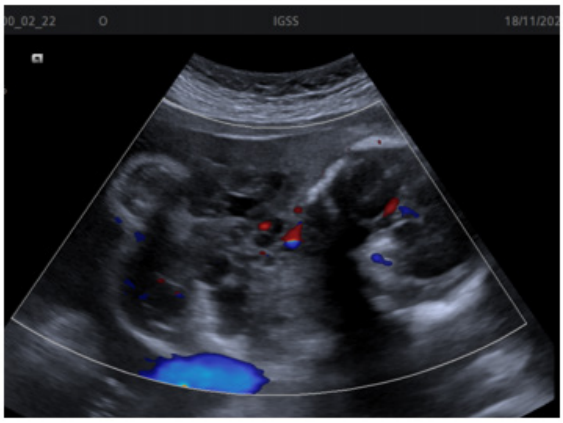

Figure 2 Multilobulated cervical mass at 29 weeks with minimal Doppler flow.

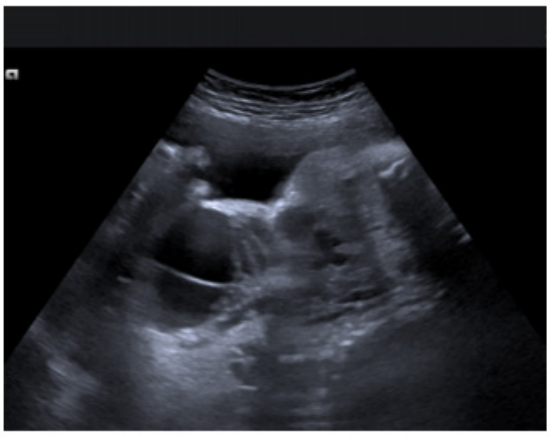

Figure 3 Multilobulated cervical mass at 38 weeks of gestation.

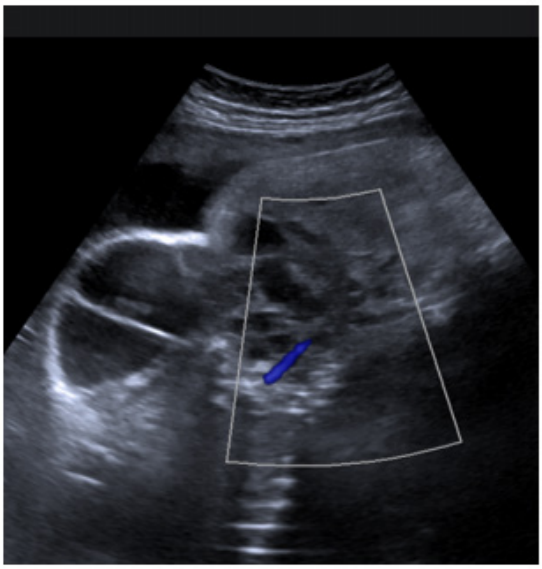

Figure 4 Multilobulated cervical mass at 38 weeks with minimal Doppler flow.

Neonatal findings

Physical examination revealed a large cervical mass involving the right hemiface, neck, and right anterior thoracic wall. The mass measured approximately 10 × 12 cm at the mandibular and cervical level and 9 × 6 cm at the anterior thorax. On palpation, the lesion was soft, covered by intact skin, with superficial vascularization and tenderness, without ulceration or skin changes. Based on clinical and imaging findings, the differential diagnosis included a congenital cervical mass, primarily considering cervical teratoma and cystic hygroma, associated with airway obstruction risk, polyhydramnios, and probable pulmonary hypoplasia.

Postnatal evolution showed that at 15 days of life, the newborn was diagnosed with cystic hygroma and a cyanotic congenital heart disease, consisting of a 3.5 mm ventricular septal defect and a 2 mm patent ductus arteriosus. Drainage of the cystic hygroma was performed, yielding approximately 70 cc of Sero hematic fluid. A second drainage procedure was subsequently performed, combined with bleomycin sclerotherapy. At the time of manuscript preparation, the patient remained under mechanical ventilation with a guarded prognosis.

Differential diagnosis of fetal cervical masses

The differential diagnosis of fetal cervical masses is broad; however, the combination of ultrasound characteristics, Doppler pattern, and anatomical location allows prenatal diagnostic orientation and anticipation of perinatal implications.

Cystic Hygroma / Lymphangioma

Cystic hygroma is a type of lymphangioma, also referred to as nuchal edema or increased nuchal translucency when detected in the first trimester. It represents a vascular anomaly of the lymphatic system characterized by fluid accumulation, primarily located in cervicofacial and axillary regions. Its incidence is approximately 1 in 6,000 live births and increases to 1 in 750 spontaneous abortions.[1] Ultrasound findings include multiloculated cystic collections with thin walls, usually located in the cervical region. Color Doppler typically shows no internal flow, aiding differentiation from hemangiomas. Differential diagnoses include encephalocele and cervical teratoma. Polyhydramnios, although suggestive of neural tube defects, has also been described in cystic hygroma.[1]

The prognosis of cystic hygroma is unfavorable in approximately 77.8% of cases and is influenced by associated structural malformations and chromosomal abnormalities, most commonly trisomy 21 and Turner syndrome, as well as trisomy's 18 and 13 and triploids.[1] Anatomical location influences prognosis: nuchal lymphangiomas show a higher association with chromosomal abnormalities compared to axillary lesions. Anterior cervical lymphangiomas are clinically significant due to the risk of airway obstruction and associated perinatal complications.[1]

Cervical teratoma

Fetal teratomas are congenital tumors derived from more than one embryonic germ layer. Although they are the most common fetal

tumors, cervical teratomas represent less than 5% of cases, with an estimated incidence of 1 in 35,000 to 200,000 live births. Prenatal diagnosis is often associated with polyhydramnios, non-immune hydrops, cardiac failure, and preterm birth.[2] Cervical fetal teratomas are generally benign; however, they are associated with significant perinatal morbidity and mortality, mainly due to fetal airway obstruction and distortion of adjacent soft tissues.[2] Associations with structural anomalies have been reported, including imperforate anus, cystic fibrosis, fetal chondrodysplasias, and maxillary deformities. Genetic associations include trisomy 13, X chromosome mosaicism, and monosomic pentasomy (49); nevertheless, cervical teratomas may also occur in fetuses with normal karyotype.[2]

Ultrasound characteristics include a solid or mixed well-defined mass that may extend beyond the midline, with calcifications present in more than half of cases. Color Doppler may demonstrate varying degrees of intralesional vascularization, which is a key feature in differential diagnosis.[2] From a topographic standpoint, cervical teratomas are most frequently located in the anterior and midline neck, whereas cystic hygroma, lymphangioma, hemangioma, and bronchogenic cysts are usually found in the lateral or posterior neck regions.[2] Anomalies of the thyroglossal duct should also be considered, as they are the most common congenital neck malformations and may occur anywhere along the thyroid gland migration pathway.[3] Prognosis after surgical management is excellent, although recurrence rates of up to 10% have been described.[4]

Conclusion

Fetal cervical masses represent a diagnostic and therapeutic challenge due to their wide heterogeneity and the potential risk of neonatal airway obstruction. This case demonstrates that early prenatal diagnosis, supported by serial ultrasound evaluation and color Doppler, allows appropriate differential diagnosis, anticipation of perinatal complications, and timely obstetric and neonatal management. Furthermore, it highlights the importance of a multidisciplinary approach and individualized perinatal planning, including consideration of the exit procedure in selected cases to secure the airway at birth and improve neonatal outcomes. Finally, this case underscores the need for close postnatal follow-up, as definitive diagnosis and prognosis may evolve after clinical and imaging correlation.

Acknowledgments

None.

Conflicts of interest

The authors declare no conflicts of interest.

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