LUMEN ET VIRTUS

Cystic hygroma associated with multiple fetal malformations: Case report

https://doi.org/10.56238/levv15n38-098

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ABSTRACT

Cystic hygromas are fetal malformations related to the proliferation of lymphatic vessels associated with fibrosis, being a rare lymphangioma in neonates, and can be diagnosed during prenatal care. The relationship between cystic hygromas and other malformations is not direct, but it can be understood that there is a possibility of arising from chromosomal abnormalities. Thus, although very rare, there may be an association of this lymphangioma with other findings also arising from chromosomal diseases.

Keywords: Cystic hygromas, Single umbilical artery, Congenital heart disease.

INTRODUCTION

Cystic hygromas are fetal malformations related to the proliferation of lymphatic vessels associated with fibrosis, being a rare lymphangioma in neonates, and can be diagnosed during prenatal care. The embryonic origin of this malformation is not very well understood, but its course is benign, leading to believe that it occurs due to the dilation of these lymphatic vessels accompanied by cystic characteristics typically in the neck, clavicle and axillary regions in the form of nodules. Although it is benign, it has the potential for airway obstruction depending on the site of involvement1.

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The origin of the lymphatic vessel anomaly described is understood as the ability to proliferate from remains of embryonic tissue. Thus, although it is not a cause for concern in obstetricians and pediatricians, this condition is usually associated with chromosomal aneuploidies, hydrops fetus, and intrauterine death1.

The relationship between cystic hygromas and other malformations is not direct, but it can be understood that there is a possibility of arising from chromosomal abnormalities. Thus, although very rare, there may be an association of this lymphangioma with other findings also arising from chromosomal diseases.

The placenta is an organ developed during pregnancy with a vital role in the nutrition and oxygenation of the developing fetus, also acting in the excretion of waste products and performing as a protective barrier against pathogens. The maternal-fetal connection has the umbilical cord as a bridge between fetus and placenta, measuring about 50 centimeters in length, usually composed of two arteries (responsible for carrying venous blood) and one vein (responsible for carrying arterial blood), covered by the so-called Wharton's jelly2.

In some cases, the umbilical cord presents anomalies that alter its constitution, such as the absence of one of the arterial vessels, called the single umbilical artery (UAU). AUU is diagnosed with routine fetal ultrasound during the first trimester of gestation or up to the second trimester. Several theories seek to explain this anomaly, such as primary agenesis, secondary atrophy or even atresia of one of the umbilical arteries present3.

Some risk factors are associated with AUU, according to the literature by NUNES, A. F. et al., 20203, citing smoking, multiparity, advanced maternal age, and maternal metabolic diseases, relating the condition to chromosomal alterations, particularly trisomy 18 and trisomy 13, in addition to comparing them to adverse pregnancy outcomes. UAU is generally not associated with other fetal malformations, but it may be present in structural malformations, including congenital heart defects.

Cardiac malformation is one of the anomalies that can be associated with AUU and result in significant intrauterine functional impairment, being considered the main congenital malformation in terms of morbidity and mortality in early childhood, and may evolve symptomatically or asymptomatically, and can also be diagnosed in prenatal follow-up through fetal ECHO. The risk factors that trigger congenital heart diseases include genetic burden and maternal health, which may or may not be related to chromosomal alterations4.

This study aims to report the case of cystic hygroma diagnosed intrauterus, associated with fetal cardiac malformation and single umbilical artery, in order to help the community understand such pathology and contribute to scientific development.

METHODOLOGY

This is a case report study, whose information was collected through a review of medical records and a direct interview with the patient. In parallel, to support the ideas discussed in this article, a literature review was carried out in scientific databases such as PubMed, Scielo and Google Scholar. The production of this scientific article followed the regulations proposed by the National Research Council (CONEP).

CASE REPORT

A 23-year-old Caucasian, married female patient attends a consultation for prenatal care of a pregnancy diagnosed with pharmaceutical tests. He denies smoking, denies alcoholism, no use of continuous medication and/or history of family diseases.

G4P1A2, refers to a living daughter at three years of age born by cesarean section at term due to personal request, with an uneventful pregnancy resulting from a parent different from the current one. She reports that she suffered two miscarriages as a result of the same partner of the current pregnancy. Menarche at 13 years of age, regular previous menstrual cycle, with no relevant gynecological history. No history of obstetric past in first-degree relatives.

In a prenatal consultation, obstetric ultrasonography was requested, which showed 11 weeks and 4 days of gestation with a fetus with altered nuchal translucency. An investigation was initiated with morphological ultrasonography, which showed thickening of the fetal skin along the frontal and lumbar region, with its largest diameter in the nape of the neck, where it measures 16 mm, noting fine septations inside, hypodevelopment of the nasal bone and flow of the ductus venosus with negative wave A. Subsequent morphological ultrasonography performed at 17 weeks and 6 days of gestation showed a single umbilical artery and fetal heart, suggesting morphological alteration.

The last morphological obstetric ultrasonography performed at 26 weeks and 4 days of gestation showed negative ultrasonographic markers of aneuploidies, except for nasal bone (absent or hypoplastic) and positive structural anomalies, in addition to showing important ventricular septal defect, absent ductus venosus, and thickened frontal fold. Pregnancy currently in progress.

The partner does not have a history of pathological genetic alterations in the family, but reports that the mother used alcohol and drugs during her pregnancy.

Pregnant woman reached 38 weeks of gestation and was admitted to the emergency room with a ruptured sac, being opted for cesarean delivery due to the risk to the fetus, referred to the neonatal ICU after birth.

DISCUSSION

Lymphatic malformations are congenital anomalies of the lymphatic system with an estimated incidence of 1 in 200,000 live births. They are characterized as macrocystic (greater than 1 centimeter) and microcystic (less than 1 centimeter), and are commonly found in the head and neck region, but may appear in other locations5. The fetus in question was diagnosed intrauterine, presenting a macrocystic lesion of 16 mm in the nuchal region, thus, the presentation of this hygromas corroborates the literature that describes similar cases, although the intrauterine diagnosis is less frequent.

The treatment of lymphatic malformations involves a multidisciplinary approach, since they can evolve with breathing difficulties in infants. Individualized treatment depends precisely on these complications and other associated symptoms, including surgery, laser therapy, or drug forms5. This outcome should be evaluated after the birth of the child in question, but the mother did not present complications during her pregnancy.

The fact that draws attention to the case is that, despite being a benign form, lymphatic malformation is associated with other potentially dangerous fetal malformations. The fact that the fetus has a single umbilical artery and cardiac malformation brings to light a possible genetic syndrome.

In the case of the single umbilical artery, by itself, it is a major triggering factor for premature birth, which is one of the main causes of infant morbidity and mortality, characterizing a robust risk of negative outcome[6]. It can be diagnosed through obstetric ultrasound with doppler, which allows the visualization of a cord that has only two vessels, instead of three, from the 12th week of gestation7.

Physiologically, the umbilical cord is made up of two arteries and one vein, seen in cross-sections in three separate circular echonegative images. In cases of AUU, the visualization of a cord that has only two vessels formulates the definitive diagnosis of the condition7. Fortunately, the delivery of this case described was carried out at term and there were no major complications, in addition to the referral to the neonatal ICU already expected for a newborn with cardiac alterations. However, as described in all literature, the diagnosis of AUU came during prenatal care with ultrasound in the second trimester of pregnancy.

In a study conducted in Cuba, it was observed that maternal smoking is a major risk factor for the development of a single umbilical artery, but it differs from other studies that state that there is no great relationship between smoking mothers and fetuses with UAU7. In this case, smoking was not present in the mother's history and therefore cannot be pointed out as a risk factor for this condition. Another factor pointed out was Diabetes Mellitus, which is also not present in this case. This same study points to the presence of a single umbilical artery related to other fetal malformations.

Congenital heart disease (CHD) is present in 40% of fetal malformations and is responsible for numerous cases of infant mortality worldwide, requiring intensified natal care. Some of these pathologies are milder, not causing major hemodynamic repercussions, others, however, are intense to the point of requiring intervention within the first days of life, since they have repercussions on child development8.

The work of Rosa R. C. M. et al (2013)[9] brought to light a literature review of the last 20 years on the association of congenital heart diseases with extracardiac malformations, evidencing their technological limitations, however, citing authors such as Miller et al (2011)[10] who studied 7,984 cases of cardiac malformations of babies born between 1968 and 2005 in the United States, including stillbirths and elective terminations after 20 weeks of gestation. Of these, 71.3% had isolated heart diseases, 13.1% were associated with some syndrome, and 13.5% were associated with extracardiac malformations. Large studies show us that the prevalence of congenital heart diseases is high, as previously mentioned, but its impacts are mainly restricted to the repercussions of the disease itself and are not usually genetically syndromic. For this fetus in question, reported in this study, there is clearly a genetic syndrome, because in addition to extracardiac malformations, there are also markers of aneuploidies, such as hypoplasia of the nasal bone. This literature review concludes that mortality is higher when associated with abdominal malformations, which does not occur in this case.

Nasal bone hypoplasia is a condition characterized by inadequate or undersized development of the nasal bone, which can lead to changes in facial appearance and, in some cases, respiratory problems. This condition can be associated with genetic syndromes, commonly trisomy 21, 18, 13 and other rarer ones or it can occur in isolation; Maternal ethnic origin is also a factor to be taken into account, as it impacts the dimensions of the nasal bone. A higher rate of diagnoses of genetic alterations occurs when hypoplasia of the nasal bone is associated with other congenital malformations11. Also present in this report, hypoplasia of the nasal bone is associated with both cardiac malformation and alterations in the formation of the umbilical cord. The definitive syndromic diagnosis, however, must come together with genetic tests on the newborn, but this occurs invasively, requiring special maternal authorization.

It is essential that when detecting anomalies and markers of chromosomal diseases, the mother is referred for proper follow-up and can perform the karyotype of the fetus, since up to 30% have a syndromic diagnosis. Obstetric ultrasonography is still the least expensive exam and is capable of detecting simple alterations, opening up an important investigative parameter12. Other exams, such as morphological ultrasonography, are more sensitive to measure such changes, but have a higher cost. In any case, prenatal care is extremely important to ensure gestational success and less morbid impact on newborns and their families.

CONFLICTS OF INTEREST

The authors state that there is no potential conflict of interest that could compromise the impartiality of the information presented in this scientific article.

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