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Case Report

Echoic intracardiac focus and down syndrome: about a case report.

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Abstract:

Background: The study focuses on the intracardiac echoic focus (IEF), a result of excessive mineralization within fetal heart muscles. Diagnosis through second-trimester ultrasound aids in identifying this marker, often linked weakly to Trisomy 21.

Observation: Patient JA, with a history of pregnancies and complications, presented with a current pregnancy complicated by gestational diabetes. Initial screenings suggested a low Trisomy 21 risk, but an ultrasound in the second trimester revealed an IEF and minimal pericardial effusion. Further tests were suggested but not carried out due to financial constraints.

Results: Despite a calculated intermediate risk of Trisomy 21 based on the likelihood ratio and serum markers, the patient eventually gave birth to a confirmed Trisomy 21 newborn via cesarean section.

Conclusion: The case demonstrates the challenges in diagnosing Trisomy 21 prenatally. A comprehensive approach using ultrasound markers and serum screenings is essential, as Trisomy 21's varied presentation limits reliance on ultrasound alone. Integrating both methods remains pivotal for accurate risk assessment and informed decision-making in prenatal care.

Keywords: intracardiac echoic focus; trisomy 21; prenatal diagnosis; serum markers; perinatal outcomes; fetal abnormalities

Introduction:

The intracardiac echoic focus or intraventricular hyperechoic nodule results from excessive mineralization and calcification of papillary muscles and chordae tendineae.[1]

Diagnosis is easily made through mandatory morphological ultrasound in the second trimester, using the four-chamber view of the heart, which is an integral part of the ultrasound screening.[2]

The intracardiac echoic focus appears as an area of echogenicity similar to that of bone within the region of the papillary muscle in either one or both ventricles of the fetal heart.[3]

80% of IEFs are found in the left ventricle.
5% in the right ventricle.
7% in both ventricles.
Moreover, it serves as a weak marker for trisomy 21.

If the IEF is located in the right ventricle, is bi-ventricular, multiple, or not isolated, the risk of trisomy 21 becomes more significant.[4]

If the IEF is isolated, its likelihood ratio is 1.1, but rises to 6.4 if other signs of trisomy 21 are present.

Observation:

This concerns patient JA, aged 36, with no significant medical history, previously operated on in our department in 2014 for a myomectomy.

She is G5P3A2.

1st pregnancy (2016) + 2nd pregnancy (2017) = 2 spontaneous miscarriages.

3rd pregnancy (2019) = Pregnancy induced by ovarian stimulation, complicated by a twin pregnancy, carried to term under Enoxaparin and Kardegic, with a term cesarean delivery (due to a precious pregnancy) of a newborn who passed away in the 2nd month of life (cause of death unknown).

4th pregnancy (2021) = Spontaneous pregnancy complicated by gestational diabetes, managed under Enoxaparin + Kardegic, fetus deceased in the 8th month due to a post-traumatic retroplacental hematoma (traffic accident) with cesarean delivery.

5th pregnancy = CURRENT

This is a spontaneous pregnancy managed under Enoxaparin + Kardegic, complicated by gestational diabetes. During the first-trimester serum marker screening, a low risk for Trisomy 21 was discovered, equal to 1 in 1534.

During the morphological ultrasound in the second trimester at 23 weeks of gestation, a minimal anterior pericardial effusion with a 2-millimeter intraventricular left spot was identified.

A test for Trisomy 21 screening using circulating fetal DNA was proposed to the patient but not performed due to financial constraints, as well as an amniocentesis, which was declined.

Ultimately, our patient delivered via cesarean (due to contraindication for vaginal delivery: bi-cicatricial uterus) at a gestational age of 38 weeks and 6 days, giving birth to a confirmed Trisomy 21 newborn.

Discussion:

Since the late 1980s, efforts have been made to identify morphological signs and fetal biometric data detectable through ultrasound that could justify an amniocentesis. Chromosomal anomalies can often have an early phenotypic expression accessible via second or third-trimester fetal ultrasound.[5] This can manifest as a highly suggestive poly-malformative syndrome indicative of aneuploidy. However, Trisomy 21 rarely presents itself as a clear-cut ultrasound tableau.[6]

Various studies have sought to identify a range of minor signs associated with Trisomy 21. The most significant advancement in this field occurred in the early 1990s with Szabo and Gellén describing increased nuchal translucency in trisomic fetuses, nonspecific but visible from the 1st trimester.[7]

More recently, new signs have been evaluated in the first trimester. Antenatal ultrasound screening for Trisomy 21 is crucial. Among the indicative signs in the first trimester are increased nuchal translucency, absence of nasal bones, underdevelopment of the upper maxilla causing a flattening of the face (measured as the fronto-maxillary facial angle), a reversed 'a' wave in the Doppler of the ductus venosus, and tricuspid regurgitation in the tricuspid valve Doppler.[8]

Furthermore, notable signs in the second and third trimesters include major malformations like atrioventricular canal defects and duodenal atresia, along with minor signs such as thickened nuchal fold, short humerus and/or femur, hyperechoic bowel loops, pyelectasis, absence or hypoplasia of nasal bones, brachymesophalangia, clinodactyly of the 5th finger, ventriculomegaly, macroglossia, and intracardiac echoic focus - as observed in our patient.[9]

The likelihood ratio (LR) is a risk-modulating coefficient attached to a test. The post-test odds are equal to the pre-test odds multiplied by the likelihood ratio of the test. Several likelihood ratios can be used simultaneously or successively, provided the independence of the tests has been demonstrated.[10]

Therefore, antenatal ultrasound screening for Trisomy 21 coupled with the results of first-trimester serum markers or potentially second-trimester markers constitutes a fairly reliable means of early detection of this common chromosomal aberration and one of the leading causes of intellectual disability.[11] It is important to multiply the likelihood ratio of the weak Trisomy 21 markers identified through ultrasound by the risk of Trisomy 21 obtained from the first-trimester or, if unavailable, second-trimester serum marker screening to guide further screening or diagnostic steps.[12]

Recall that a risk lower than 1/1000 is considered low, between 1/1000 and 1/50 is intermediate, and above 1/50 is high, indicating an immediate need for amniocentesis. In our patient, the isolated 2mm intracardiac echoic focus in the left ventricle associated with minimal anterior pericardial effusion yields a likelihood ratio of 6.4, multiplied by the calculated risk from the first-trimester serum markers of 1/1534, ultimately resulting in an intermediate risk value. This is why a test for Trisomy 21 screening using circulating fetal DNA was proposed to the patient but ultimately not performed.[13]

Conclusion:

The case illustrates the complexity and challenges in prenatal diagnosis of Trisomy 21. Utilizing a combination of ultrasound markers, serum screenings, and understanding the likelihood ratios associated with various indicators becomes pivotal in stratifying risks and guiding further diagnostic steps. Despite advancements in identifying potential markers, the nuanced and variable presentation of Trisomy 21 underscores the importance of comprehensive assessment and the limitations of relying solely on ultrasound findings. The interdisciplinary approach, integrating both ultrasound and biochemical markers, remains crucial for more accurate risk assessment and informed decision-making in prenatal

care.

Consent for publication : Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

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