Outros

(21479) - CASE REPORT: PRENATAL LOEYS-DIETZ SYNDROME - DILATATION OF THE MAIN PULMONARY ARTERY AND ASCENDING AORTA

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Introdução
Loeys-Dietz syndrome (LDS) is an autosomal dominant connective tissue disease with important cardiovascular manifestations, which can lead to death at a young age due to aortic aneurysm dissection. This is a rare case of prenatal diagnosis of Loeys-Dietz syndrome, which is usually diagnosed after birth.
Objectivos

The main purpose of this case report is to make clinicians aware of a rare finding on fetal echocardiography: the dilatation of the great vessels.

Metodologia

Case Report: A 34-year-old primipara was referred to the fetal cardiology clinic at 24 weeks' gestation. The baby's father suffers from Loeys Dietz syndrome with an identified TGFBR2 mutation (LDS type 2). He has had an aortic root replacement and has a strong family history of sudden cardiac death from acute aortic root dissection.Given the paternal diagnosis, the couple opted to do a NIPT which confirmed that the fetus had the same TGFBR2 mutation.

First trimester screening was low risk and the 20-week anomaly scan was reported as normal. There were no signs of skeletal and craniofacial affection, typical manifestations of the syndrome. At 24 weeks, the fetal echocardiography showed a dilatation of the ascending aorta measuring 6.5 mm (z-score +3.39) and a dilatation of the main pulmonary artery measuring 7 mm (z-score + 4.10).

Resultados e Conclusões

The latest published cohort study on fetal ascending aortic dilatation showed no association between this and connective tissue disorders. On the other hand, there are some case reports describing this condition in fetuses with connective tissue diseases, the most common of which is Neonatal Marfan Syndrome. Five cases have been reported associated with Loeys-Dietz syndrome

In conclusion the presence of a dilated main pulmonary artery and/or ascending aorta, referral to a fetal cardiologist and genetic counseling is recommended - these genetic conditions are serious.

Palavras-chave : Loeys-Dietz Syndrome, Prenatal diagnosis, Dilated ascending aorta, Dilated Main Pulmonary artery