Not everything is always genetic – A prenatal presentation of tortuous and aneurysmal ductus arteriosus

Nem tudo é sempre genético – Apresentação pré-natal de canal arterial tortuoso e aneurismático

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Abstract

Tortuous and aneurysmal ductus arteriosus (TADA) is a relatively rare condition, especially in the first and second trimesters. The ductus arteriosus has functional importance in the fetal circulation, being its aneurysmatic format characterized by saccular dilatation of the vessel. Majority of cases have a benign evolution, although they can be associated with syndromic forms and with important complications. We report the clinical management and echocardiographic follow-up in a newborn with this very early finding in prenatal setting.

Keywords: Aneurysmal ductus arteriosus; Prenatal setting; Early diagnosis.

Resumo

O canal arterial tortuoso e aneurismático (CATA) é uma condição relativamente rara, especialmente no primeiro e segundo trimestres. O canal arterial tem importância funcional na circulação fetal, sendo seu formato aneurismático caracterizado pela dilatação sacular do vaso. A maioria dos casos tem evolução benigna, embora possam estar associados a formas sindromáticas e a complicações importantes. Relatamos a orientação clínica e o seguimento ecocardiográfico num recém-nascido com este achado muito precoce em contexto pré-natal.

Palavras-chave: Canal arterial aneurismático; Contexto pré-natal; Diagnóstico precoce.

CASE

Prenatal diagnosis of TADA is usually incidental in third trimester. The vast majority of cases has a good prognosis, though regular ultrasound follow-up is needed given the associated thrombotic risk[1]. Maternal diabetes and blood group A are considered high-risk factors[1,2]. TADA is related with trisomy 21 and 13, as well as, connective tissue diseases, as Marfan and Ehlers-Danlos syndromes[3], related with alterations in elastin expression or intima cushioning, or has multifactorial etiology[1,2].

We present a male with prenatal diagnosis of TADA. His mother, a 36-year-old woman, with blood group A, has a personal history of hypothyroidism and type 1 diabetes with no history of exposure to teratogens, namely consumption of polyphenol-rich substances, herbal infusions, or nonsteroidal anti-inflammatory drugs[4], and has a previous healthy daughter.

At 12 weeks of gestation, fetal ultrasound revealed a small omphalocele with intestinal content and invasive prenatal testing was performed through chorionic

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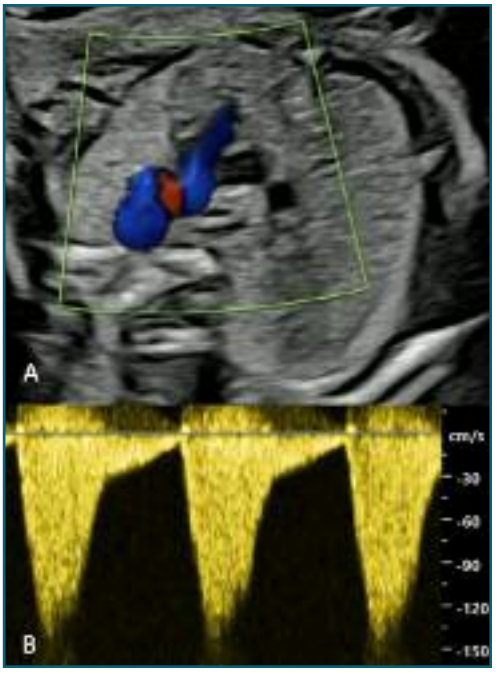

FIGURE 1. Prenatal ultrasound evaluation, twenty-four weeks gestation prenatal transabdominal echo: **A** - Three vessel view showing tortuous and aneurismatic *ductus arteriosus;* **B** - Flow Doppler with elevated velocity (120-130 cm/s).

villus sampling. The results of QF-PCR and arrayCGH *(CGX-HD 180K, PerkinElmer)* were both normal. MS-MLPA for Beckwith-Wiedemann syndrome was not possible to perform due to insufficient fetal DNA and early gestational age. Subsequent ultrasound evaluation showed regression of the omphalocele and adequate fetal growth.

Considering maternal diabetes background and the need to exclude other congenital malformations, fetal echocardiogram was performed at 24 weeks. It showed a tortuous ductus arteriosus with a turbulent high velocity flow, with a maximum velocity of 130 cm/s (Figure 1). Regarding the potential thrombotic or rupture risks, weekly echocardiogram was performed until birth, evaluating the ductus morphology and doppler flow, and searching for signs of right ventricular disfunction (tricuspid regurgitation and ventricular dilatation), showing no deterioration. The child was born at 38 weeks by vaginal delivery, with a birth weight of 3150 grams (25th centile) and Apgar score 9/10. Echocardiogram performed on the 3rd day of life, corroborated the prenatal findings, with complete resolution at 1 month of age (Figure 2).

This case highlights the unusual frequency of TADA in the earlier stages of pregnancy and recalls its most frequently observed follow-up and prognosis.

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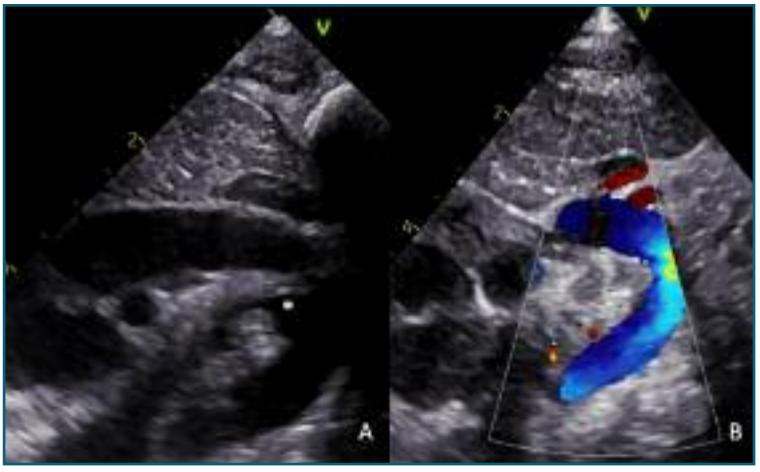

FIGURE 2. Postnatal ultrasound evaluation (Suprasternal views) **A** - Three days of life postnatal transthoracic echo: aortic arch with ductus arteriosus (large ampola*) and restriction at pulmonar side; **B** - One month of life postnatal transthoracic echo: normal aortic arch, no ductus arteriosus (closed).

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AUTHOR CONTRIBUTIONS

All authors made a substantial contribution to the information or material submitted for publication.

DECLARATIONS OF INTEREST

The authors have no conflicts of interest to declare.

INFORMED CONSENT

Patient consent for publication was obtained.

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