Arterial Tortuosity Syndrome

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Abstract

A rare autosomal recessive condition, Arterial tortuosity syndrome (ATS) presents with ectactic blood vessels, cutaneous laxity, and bowel rupture. We report a case of an asymptomatic infant with arterial tortuosity syndrome who presented with left ventricular hypertrophy without any obvious obstruction to the outflow tract.

TITLE AND AUTHORS

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Arterial Tortuosity Syndrome - Meandering Aorta and branches in an infant

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Manuscript

An asymptomatic 10 month old male child born out of a consanguineous marriage was referred for evaluation of a cardiac murmur. History was not contributory and physical examination was unremarkable. Transthoracic echocardiogram (TTE) revealed concentric left (LVH) and right ventricular hypertrophy (RVH) with preserved LV function, with dilated root and ascending aorta (supplemental video 1). The aortic valve was tricuspid with mild aortic regurgitation and no aortic stenosis. The tortuous arch, neck vessels and loops of serpentine descending thoracic and abdominal aorta were noted (Supplemental video 2). The LVH prompted a search for coarctation of aorta, which was not evident on the TTE. The lower limb had bounding pulses and the child was well perfused with normal blood pressure. A diagnosis of ATS was considered, and a cardiac computed tomography (CT) was performed. CT (panel A – D) confirmed the diagnosis and it showed ectactic and tortuous aorta and its branches. Proximal ascending aorta was dilated (29 x 31 mm) with tapering at the distal arch with no evident obstruction. The pulmonary arteries were dilated. Mesenteric and bilateral renal arteries were tortuous, with no evident obstruction. The LVH in this child was probably due to a high afterload and the ventriculo-vascular impedance imposed by both the tapering at the distal arch and the subsequent tortuous and meandering aorta. (1) Aortic tortuosity syndrome (ATS) is a rare, autosomal recessive connective tissue disorder caused by a loss of function mutation in the*SLC2A10* gene causing fragmentation of tunica media of great arteries. (2) It causes tortuous, aneurysmal aorta and pulmonary arteries, bowel rupture, cutaneous and joint laxity, and intracranial aneurysms. (3) The child was followed up medically but passed away a month from evaluation due to unknown causes - which could be postulated to be a consequence of intra cranial bleed from the ectactic arteries (4), artery to artery thromboembolism from the sites of aneurysmal dilatation, or acute vessel or bowel rupture.

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