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IMAGES IN CARDIOLOGY

Arterial Tortuosity Syndrome

Early Diagnosis and Association With Venous Tortuosity

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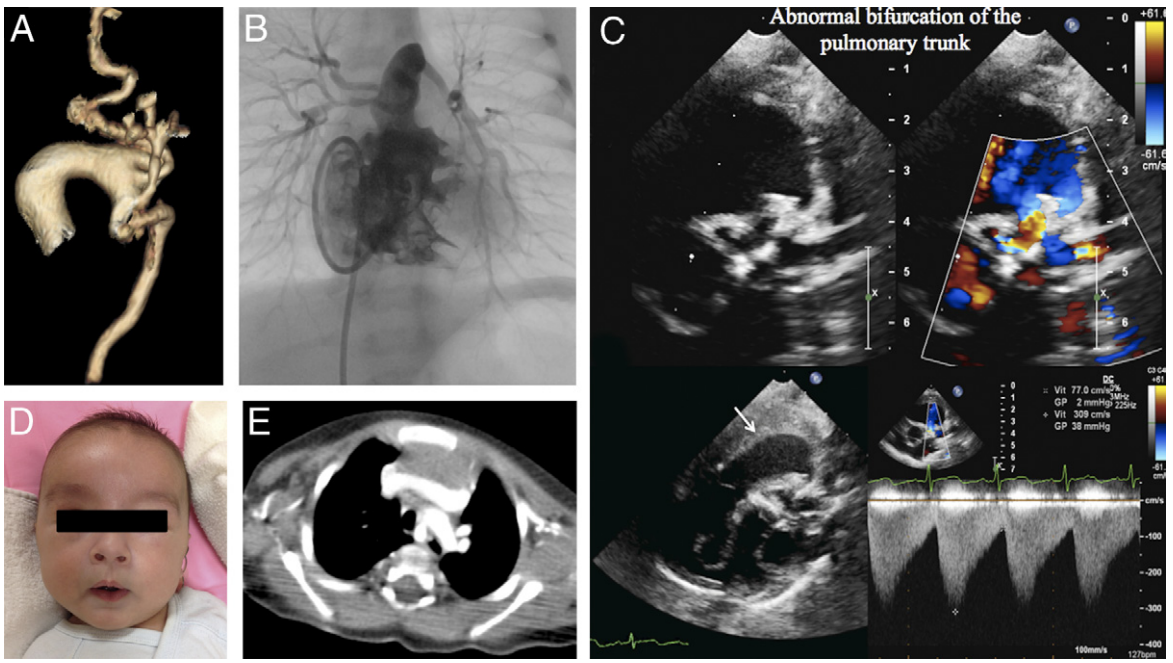

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A newborn, first child of consanguineous parents was admitted to intensive care because of persistent pulmonary hypertension. The diagnosis of arterial tortuosity syndrome (ATS) was suspected due to severe tortuosity of the aorta **(A)** and pulmonary arteries (**B and C**, Online Video 1). Despite initial hospitalization, the first months of life were uneventful. He progressively developed peripheral pulmonary stenosis and moderate coarctation of the aorta. His facial features were characteristic **(D)**, with elongated face, hypertelorism, and a long philtrum. He also presented with bilateral inguinal hernia, typical of connective tissue disease that is associated with ATS. The diagnosis was confirmed by genetic analysis: the patient was homozygous for a mutation in exon 2 (c.510G>A, p.Trp170X) of the *SLC2A10* gene. The particularity of this case is, for the first time to our knowledge, an early diagnosis of persistent pulmonary hypertension and an association of venous tortuosity (**E**; distorted innominate vein) with ATS.