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Albert Riudor Guri Raquel Luna López Isabel Blanco

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Arterial Tortuosity Syndrome: A Rare Cause of Pulmonary Hypertension

Albert Riudor Guri[1], Raquel Luna López[2,3], Isabel Blanco[1,4,5,6]

1. Department of Pulmonary Medicine, Hospital Clínic, University of Barcelona, Barcelona, Spain

2. Department of Cardiology, Institut Cardiovascular, IDIBAPS, Hospital Clínic of Barcelona, Barcelona, Spain

3. Cardiology Department, Hospital Sant Joan de Déu, Barcelona, Spain

4. Fundació de Recerca Clínic Barcelona-Institut d'Investigacions Biomèdiques August Pi i Sunyer (IDIBAPS), Barcelona, Spain

5. Biomedical Research Networking Center on Respiratory Diseases (CIBERES); Madrid, Spain

6. ERN-LUNG, Hospital Clínic, Barcelona, Spain

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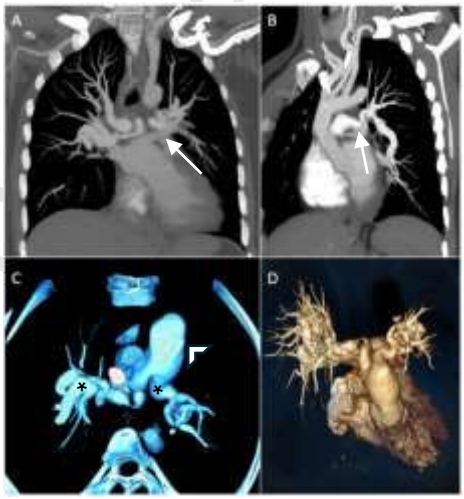

Figure 1. Coronal and sagittal section of the pulmonary CTA (A, B) show a decreased calibre of the main pulmonary arteries from their origin, which are elongated and have stenosis in their proximal third (arrows). Axial plane vascular window CT (C) shows a dilation of the pulmonary artery trunk (chevron arrow) and a significant tortuosity of the bilateral pulmonary lobar arteries (asterisks). Three-dimensional reconstruction image (D) of the pulmonary vascular tree.

We present a 19-year-old patient with a history of Arterial Tortuosity Syndrome (ATS). Neonatal right heart catheterization (RHC) revealed a dilated pulmonary artery with tortuous branches and stenotic, elongated aortic structures. By 2016, cardiac catheterization showed severe pulmonary hypertension (PH) due to focal stenosis (mPAP 51 mmHg, RAP 7 mmHg). A pediatric surgical attempt to resolve the stenosis was unsuccessful. Genetic testing confirmed an SLC2A10 mutation. In 2024, pulmonary CTA showed pulmonary artery dilation, proximal stenosis of main pulmonary arteries, marked tortuosity of pulmonary lobar arteries, and bronchial artery hypertrophy (Figure 1). ATS is a rare genetic disorder caused by SLC2A10 mutations, which encodes the GLUT10 protein, a cofactor in collagen and elastin biosynthesis; leading to weakened arterial walls and widespread vessel tortuosity. It shares features with Loeys-Dietz and Marfan syndromes. Pulmonary artery involvement can contribute to PH. Diagnosis relies on imaging and genetic testing, while management is supportive, with cardiovascular monitoring and surgery for severe cases. Beta-blockers and angiotensin-converting enzyme inhibitors may help reduce arterial stress; however, their efficacy in ATS remains unproven, warranting cautious use, especially in the presence of renal artery stenosis. Arterial tortuosity may serve as a prognostic marker for cardiovascular risk[1-5]

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ARG wrote the first draft. RLL helped with the literature review and writing. IB reviewed and edited the final version. All authors approved the final manuscript.

Ethics in publishing

1. Does your research involve experimentation on animals?:

No

2. Does your study include human subjects?:

No

3. Does your study include a clinical trial?:

No

4. Are all data shown in the figures and tables also shown in the text of the Results section and discussed in the Conclusions?:

Yes

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