DIAGNOSTIC JIGSAW: UNREVEALING AN UNUSUAL CASE OF PEDIATRIC LEFT VENTRICULAR DYSFUNCTION

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INTRODUCTION

Ventricular systolic dysfunction in children can represent a diagnostic challenge. While certain etiologies—such as myocarditis—are more common, the clinical presentation can be atypical. In some cases, the standard diagnostic algorithms may fall short, and clinicians must be willing to question initial assumptions. This case highlights the importance of remaining open to less conventional diagnostic possibilities.

CASE HISTORY

A 10-year-old girl, with unremarkable medical history, normal neurological development, mild short stature, regularly engaged in physical activity, was referred to the emergency department due to the recent onset of fever, cough and incidental finding of a heart murmur by her General Pediatrician. On initial evaluation, she had normal BP, no signs of hypoperfusion, T 38.2°C, eupneic without the need for oxygen therapy. Physical examination revealed a grade 2/6 systolic murmur. A chest X-ray showed parenchymal consolidation of the left upper lobe. Laboratory tests indicated an inflammatory response, with elevated CRP and leukocytosis.

The initial ECG revealed abnormal findings: LAH, LVH and significant T-wave inversions in the anterior and high lateral leads. So, a transthoracic echocardiogram was performed, which demonstrated left ventricular systolic dysfunction with a suspected coronary distribution: akinesia of the apical region and mid-segments of the anterior wall and interventricular septum. The patient was admitted with a working diagnosis of pneumonia and left ventricular dysfunction of uncertain etiology.
To rule out myocarditis—considered the most common cause of ventricular dysfunction in this age group—biomarkers for myocardial necrosis were tested repeatedly and remained persistently negative.

Due to the challenging nature of the case, a second echocardiography was conducted. Notable findings that helped guide the differential diagnosis included: left ventricular dilatation and systolic dysfunction with a coronary distribution, increased echogenicity of the endocardium and posterior papillary muscle (suggestive of endocardial fibroelastosis), moderate mitral regurgitation and evidence of developed collateral circulation at the level of the interventricular septum. These features strongly suggested an ischemic etiology.

Subsequent detailed evaluation of the coronary arteries was undertaken. A dilated right coronary artery (RCA) was seen originating from the right sinus of Valsalva, with blood flow away from the aorta. The left coronary artery (LCA), however, was not visualized arising from the left sinus of Valsalva. Using a modified high parasternal short-axis view, the LCA was identified as originating from the posterior aspect of the pulmonary artery (PA), with flow directed toward the PA.

The clinical and instrumental findings were consistent with the diagnosis of Anomalous Left Coronary Artery from the Pulmonary Artery (ALCAPA).
A more detailed history revealed no prior symptoms suggestive of myocardial ischemia, such as chest or epigastric pain, dyspnea, or syncope—making the clinical presentation particularly atypical for the underlying condition.

The case was discussed in a multidisciplinary team meeting with the Pediatric Cardiology Department of the regional reference Center, with an indication to advanced diagnostic imaging (coronary CT angiography) to confirm the diagnosis and define the optimal therapeutic strategy.

CONCLUSIONS

This case underscores the importance of considering coronary anomalies, such as ALCAPA, in the differential diagnosis of pediatric ventricular dysfunction—even in the absence of overt ischemic symptoms. The atypical presentation, likely modulated by the development of robust collateral circulation, allowed the patient to remain clinically compensated despite significant myocardial ischemia.

The absence of typical clinical signs highlights the role of comprehensive echocardiographic evaluation, where subtle yet specific findings—such as regional wall motion abnormalities, endocardial echogenicity, mitral regurgitation, and septal collateral vessels—can guide clinicians toward the correct diagnosis. In this case, careful integration of these elements prompted a focused assessment of coronary artery origins, ultimately revealing a life-threatening but treatable congenital anomaly.