

Autopsy findings of a child with left atrial isomerism associated with pulmonary agenesis

Achados de autópsia de uma criança com isomerismo atrial esquerdo associado à agenesia pulmonar

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ABSTRACT

Isomerism is a rare laterality defect. Our aim was to describe the autopsy findings of a child with left atrial isomerism (LAI) and pulmonary agenesis (PA), an association still not described in the literature. Fetal ultrasound revealed right renal agenesis, single umbilical artery and polyhydramnios. Echocardiography revealed a complex heart defect with LAI. The child died minutes after birth. Autopsy confirmed the prenatal findings and revealed the presence of unilateral PA, complex vascular abnormalities and polysplenia. Despite its rarity, health professionals should be aware for the possibility of such an association.

Key words: isomerism; heterotaxy syndrome; lung; congenital heart defects; autopsy.

CASE REPORT

Our aim was to describe for the first time the presence of pulmonary agenesis (PA) in a child with left atrial isomerism (LAI), whose identification was only possible after necropsy analysis. The patient's mother was a 24-year-old woman presenting fetal ultrasound with right renal agenesis and a single umbilical artery. Fetal ultrasound at 23 weeks of gestation also suspected of a congenital heart defect. Fetal echocardiography revealed LAI. The child was born by cesarean section at 38 weeks, weighing 2,840 g, and with Apgar scores of 2 and 0. She died shortly after birth. Autopsy revealed umbilical cord with two vessels; left PA with total absence of pulmonary tissue, bronchia and vascular supply, and tracheoesophageal fistula. In the topography of the right lung there was a nodular mass with bubbles, measuring $2.7 \times 2.5 \times 1.4$ cm, with a shiny grayish appearance at shear and without apparent lobulation, compatible with pulmonary parenchyma. There were two small vessels that are originated from it and that were connected to the heart. The main primary bronchus was connected to the pulmonary tissue located at right. There was LAI;

persistent left superior vena cava that drained into coronary sinus vein; absence of the supra-hepatic segment of the vena cava; direct drainage of hepatic veins and two right pulmonary veins draining to the left atrium that was located at right; ambiguous atrioventricular connection; hypoplastic and imperforate pulmonary valve; pulmonary trunk with no branches at left and severe hypoplasia of rights branches which communicated with the hypoplastic lung; absence of atrial septum and patent ductus arteriosus. There was also polysplenia, right renal agenesis and left ureteropelvic dilation. The liver was enlarged and located in the midline.

DISCUSSION

Isomerism is a rare defect in which a lateral symmetry becomes the mirror of the other. It is a kind of situs ambiguus (inversus) or heterotaxy⁽¹⁾. Pulmonary anomalies are common in LAI, in particular pulmonary segmentation abnormalities⁽²⁾. However, there is no description in the literature of patients presenting PA. PA is considered an extremely rare malformation.

Its morphological classification is based on the amount of missing bronchopulmonary tissue, and our patient presented an agenesis of unilateral type with complete absence of the bronchi. PA has been related to VACTERL association⁽³⁾, and Goldenhar⁽⁴⁾ and

Scimitar syndromes⁽⁵⁾, conditions not verified in our patient. In our case, the PA detection was performed after the birth, after necropsy. Perhaps, these cases, due to their severity, may be more observed among fetuses or stillborns.

RESUMO

O isomerismo é um defeito de lateralidade raro. Nosso objetivo foi descrever os achados da autópsia de uma criança com isomerismo atrial esquerdo (IAE) e agenésia pulmonar (AP), uma associação ainda não descrita na literatura. A ultrassonografia fetal revelou agenésia renal direita, artéria umbilical única e polidrômio; a ecocardiografia apresentou defeito cardíaco complexo com IAE. A criança morreu minutos após o nascimento. A autópsia confirmou os achados pré-natais e revelou a presença de agenésia pulmonar unilateral, anormalidades vasculares complexas e polisplenia. Apesar de sua raridade, profissionais da saúde devem estar cientes da possibilidade dessa associação.

Unitermos: isomerismo; síndrome de heterotaxia; pulmão; cardiopatias congênitas; autópsia.

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