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**Alveolar capillary dysplasia with misalignment of the pulmonary veins: two cases associated with congenital heart disease**

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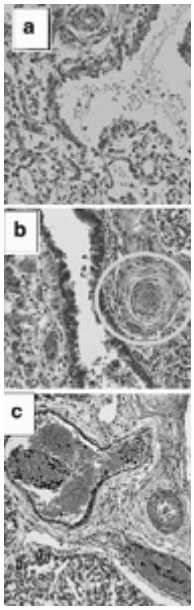
Introduction: alveolar capillary dysplasia with misalignment of the pulmonary veins (ACD/MPV) is a rare and lethal cause of pulmonary hypertension (PH) in newborns, frequently associated with cardiovascular and other systems anomalies. We present two cases of neonates with congenital heart diseases and refractory PH with post-mortem diagnosis of ACD/MPV.




Methods: retrospective review of patient records regarding clinical findings, image and genetic tests, and autopsy data.

Results: the first patient was a full-term male who developed respiratory distress requiring mechanical ventilation within his first hours of life. Extubated on day three, he began suffering hypoxic episodes. Echocardiogram and cardiac CT showed coarctation of the aorta with arch hypoplasia, signs of pulmonary hypertension, large patent ductus arteriosus and a levoatriocardinal vein. Non-invasive respiratory support and prostaglandin infusion were started. Aortic arch repair and levoatriocardinal vein ligation were performed on day 15. After surgery, he suffered severe pulmonary hypertensive crises refractory to maximal ventilatory parameters and pulmonary vasodilator treatment. Cardiac catheterization demonstrated suprasystemic pulmonary artery pressure with normal PWP. ACD/MPV was suspected, but the patient developed multiorgan failure and died on day 22 before performing pulmonary biopsy.

The second case was a full-term male with prenatal diagnosis of complete AVSD initially discharged without treatment. He was admitted again on day 29 due to hypoxic episodes, starting oxygen therapy and anticongestive treatment. Echocardiograms showed unbalanced AVSD and signs of severe PH. His clinical status progressively worsened, requiring mechanical ventilation on day 35. A cardiac CT ruled out pulmonary vein stenosis, and cardiac catheterization demonstrated suprasystemic pulmonary artery pressure with normal PWP. Pulmonary vasodilators were initiated, with initial but unsustained response. As ACD/MPV was suspected, genetic test was performed and pulmonary biopsy was programmed, but he suffered a left ACM stroke on day 59 and limitation of the therapeutic effort was performed.

Both necropsies and genetic tests were compatible with ACD/MPV ***(Table 1).***

Conclusions: ACD/MVP must be suspected in neonatal cases of refractory pulmonary hypertension without typical risk factors. The association with congenital heart disease must not mislead the clinical suspicion. Lung biopsy should be performed to consider limiting invasive procedures and provide palliative care.

| | Case 1 | Case 2 |
|---|---|---|
| Pulmonary findings | Underdeveloped alveoli and diffuse expansion of the interstitium (a); capillaries away from alveolar basement membranes. Thick and tortuous arterioles, with a marked narrowing of the lumen by intimal hyperplasia and fibrinoid necrosis (b, circle). Large and dilated veins, abnormally located beside the central bronchiolar-vascular bundles (c). ***Figure 1 (Patient 1)*** | |
| Associated anomalies | Aortic coarctation with arch hipoplasia and bicuspid aortic valve. Bilobed right lung, symmetric liver and intestinal malrotation. | Complete unbalanced AVSD. Intestinal malrotation. |
| Genetic findings | De novo hemizygous deletion of chromosome 16q24.1-q24, containing FOXF1, FOXC2, FOXL1 and JPH3 genes. | De novo heterozygous variant in FOXF1: c.257G>C (p.Arg86Pro). |