Alveolar capillary dysplasia as a rare cause of severe respiratory failure in two newborns

Zuzanna Owsiańska, MSc¹; Zuzanna Kozłowska¹; Tomasz Szczapa, MD PhD¹; Joanna Wróblewska, MD PhD²,³; Apolonia Kałużna, MSc²,³; Dorota Janczewska, MSc²; Andrzej Marszałek, MD PhD²,³; Janusz Gadzinowski, MD PhD¹

1 Department of Neonatology, Poznań University of Medical Sciences
2 Department of Cancer Pathology, Greater Poland Cancer Centre
3 Department of Pathology and Prevention of Cancer, Poznań University of Medical Sciences
ACD – Alveolar capillary dysplasia

Alveolar capillary dysplasia (ACD) is a rare neonatal lung disease characterized anatomically by a defective and hypoplastic development of pulmonary alveoli leading to persistent pulmonary hypertension (PPHN) and finally lethal respiratory failure.
CASE 1

• Male sex
• Birthweight: 2920g
• GA: 39 weeks
• C-section
• In prenatal examination: polyhydramnios, omphalocele, hydronephrosis of the right kidney and ventricular septal defect (VSD)
• Apgar scores: 1’ – 9, 5’ - 10
CASE 1

Results of the tests:
• chest X-ray was suggestive of pneumonia
• echocardiography revealed persistent pulmonary hypertension (PPHN), atrial and ventricular septal defect (ASD/VSD)

Applied therapies:
• initially noninvasive oxygen therapy
• surgical repair of omphalocele
• conventional ventilation
• broad-spectrum antibiotics
• PPHN was treated with inhaled nitric oxide
• surfactant administration
• catecholamines infusion
• high frequency ventilation
CASE 2

- Male sex
- Birthweight: 2400g
- GA: 39 weeks
- Vaginal delivery
- Prenatally: intrauterine growth restriction (IUGR) and coarctation of the aorta (CoA)
- Apgar scores: 1’ – 8, 5’ - 10
CASE 2

Results of the tests:
• chest X-ray displayed inflammatory changes in both lungs
• in echocardiographic assessment symptoms of PPHN with large ductus arteriosus and narrow pulmonary veins
• CoA was not confirmed

Applied therapies:
• initially noninvasive oxygen therapy
• continuous infusion of prostaglandins
• conventional ventilation
• broad-spectrum antibiotics
• inhaled nitric oxide
• catecholamines therapy
• high frequency ventilation
Sequence FOXF1

CASE 1 and 2

Exon 1
FISH analysis

control reaction

the correct variant
Translocation

CASE 1 and 2

[Images of FISH analysis results]
• Described patients are first confirmed by geneting testing in Poland.
• ACD should be considered in neonates with idiopathic severe hypoxic respiratory failure who fail to respond to pulmonary vasodilator therapy.
• Histopathological examination of lung tissue remains the gold standard for ACD diagnosis, however genetic testing may allow potentially faster confirmation of ACD, and can be performed in vivo.