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

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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:

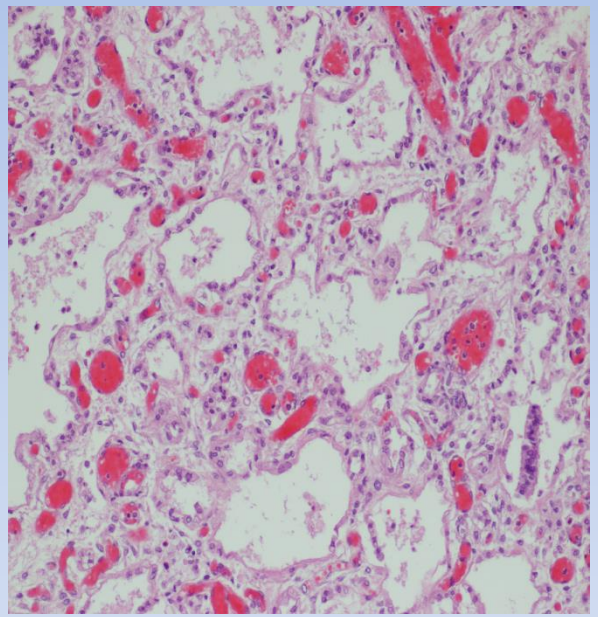
- chest X-ray displayed inflammatory changes in both lungs
- in echocardiographic assessment symptoms of PPHN with large ductus arteriosus and narrow pulmonary veins
- CoA was not confirmed

Applied therapies:

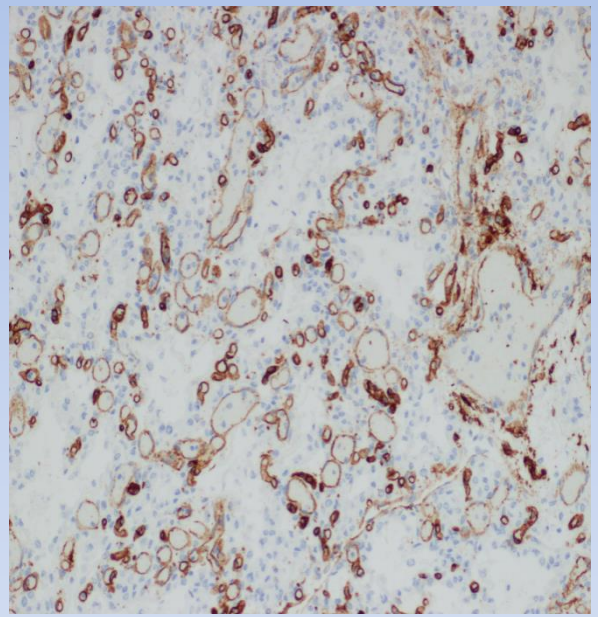
- initially noninvasive oxygen therapy
- continuous infusion of prostaglandins
- conventional ventilation
- broad-spectrum antibiotics
- inhaled nitric oxide
- catecholamines therapy
- high frequency ventilation

ACD

H+E



CD34



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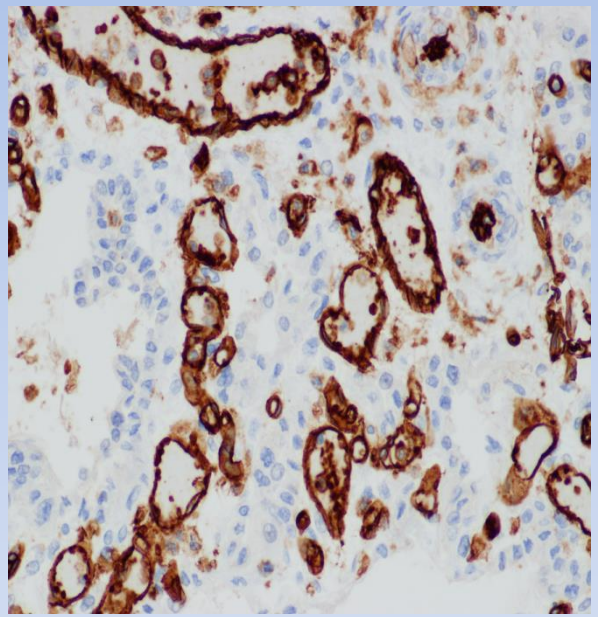
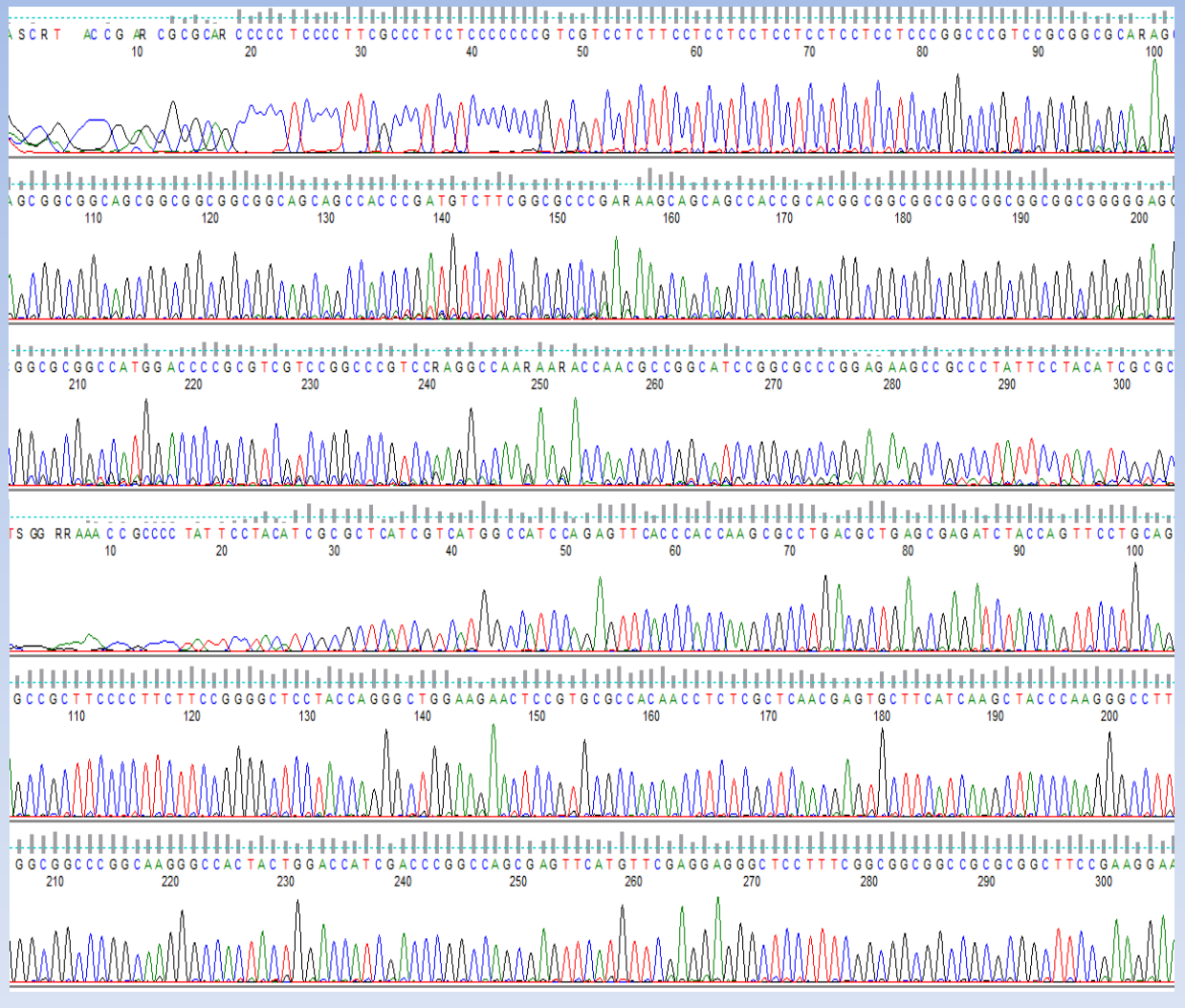


Sequence FOXF1

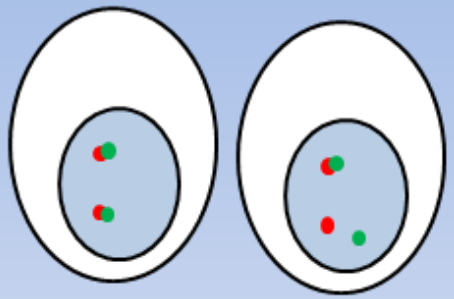
CASE 1 and 2

Exo
n 1

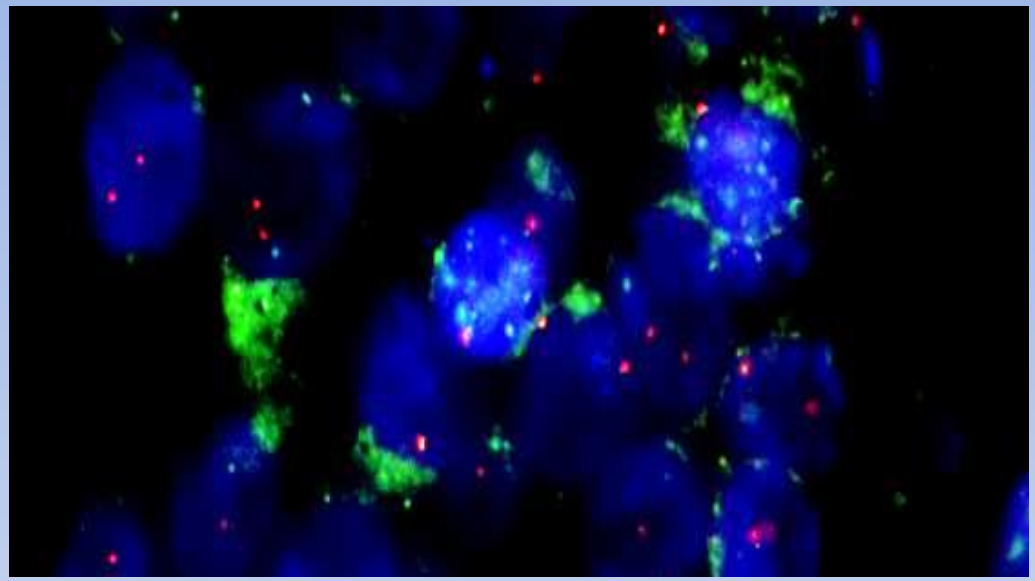


FISH analysis

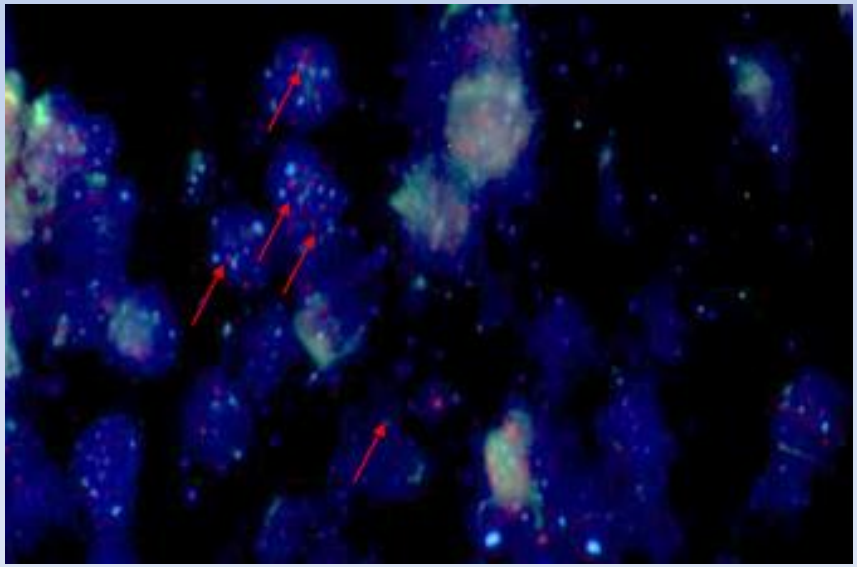
control reaction



the correct variant
Translocation



CASE 1 and 2



- Described patiens 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.