Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins: An Autopsy Case Report

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CLINICAL HISTORY
This is the case of a 13-day-old female infant delivered at 34 2/7 weeks to a 38-year-old G1P0 mother with spontaneous rupture of membranes, via cesarean section on 2/26. Prenatally, the infant was diagnosed with an omphalocele and a 17q12 duplication encompassing the HNF1B gene; the karyotype was normal female.

At birth, the Apgar scores were 8 and 8 at 1 and 5 minutes respectively; her weight was 2620 grams. Continuous positive airway pressure was started at 4 hours of life due to cyanosis, increased work of breathing, and poor tracing on pulse oximetry. She experienced pulmonary failure and coded on 2/27 with desaturations in the low 60s with shunting, requiring extracorporeal membrane oxygenation (ECMO) cannulation; decannulation was performed on 3/7. On 3/8 she was diagnosed with pulmonary hypertension via echocardiogram and was consented to receive treprostinil (Remodulin) therapy on 3/9; she received prostaglandin and dopamine infusions as adjunctive therapies. Despite prostaglandin therapy a closed PDA was identified on echocardiogram on 3/10.

Over the course of the hospital stay, she received multiple units of fresh frozen plasma, platelets, and red blood cells. Overall, she had multiple desaturation events with several ventilator methods tried without success. On 3/11 the family decided to withdraw care and the infant was pronounced deceased at 16:14. The family requested a limited autopsy (in vitro exploration and removal of lungs only) that was performed on 3/12.

GROSS AUTOPSY FINDINGS
The external exam reveals multiple signs of therapeutic intervention and a firm, brown umbilical stump measuring 8.5 cm in length and 1.5 cm in diameter at its insertion to the abdominal wall.

The left lung weighs 61 g and shows an incomplete fissure while the right lung weighs 69 g and appears to be unilobular. There are 2 cc of bilateral serous pleural effusions.

The peritoneal cavity contains 6 cc of hemorrhagic fluid. A portion of mesentery is noted entering the umbilicus and extending up the midline to the liver.

Attached to the mesentery are loops of large and small bowel. The right colon and appendix are located in the left upper quadrant. The remainder of the colon is present in the left half of the abdomen. The large bowel appears to loop and enter/exit the retroperitoneum multiple times. The small bowel is present in the right half of the abdomen.

MICROSCOPIC FINDINGS
The lung parenchyma is premature for 34-36 weeks gestation and is more consistent with the beginning of the sacellar stage of pulmonary development. The alveoli are simplified with thickened septa containing centralized capillaries. The bronchovascular sheath contains thickened muscular arteries, bronchioles and veins/venules. Venous structures are absent from the intralobular septa.

At 1 year of age, this disease may present with persistent pulmonary hypertension leading to the necessity of ventilator support. The pathogenesis of this disease is unclear, and may be due to congenital heart defects, pulmonary hypertension, and bronchial atresia.

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