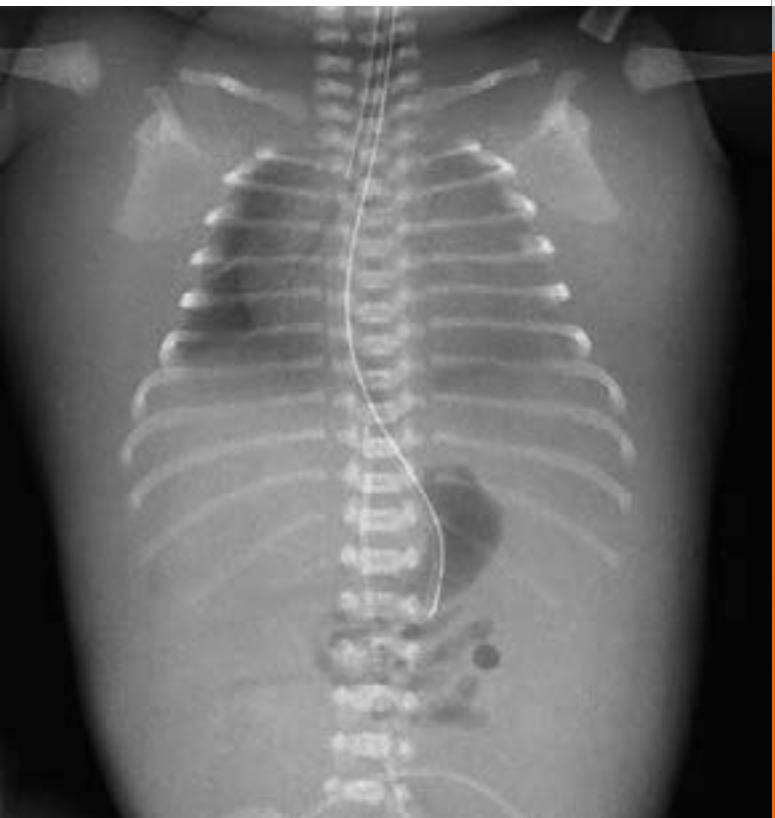
SWISS SOCIETY OF NEONATOLOGY

Endocardial fibroelastosis as a cause of hydrops fetalis



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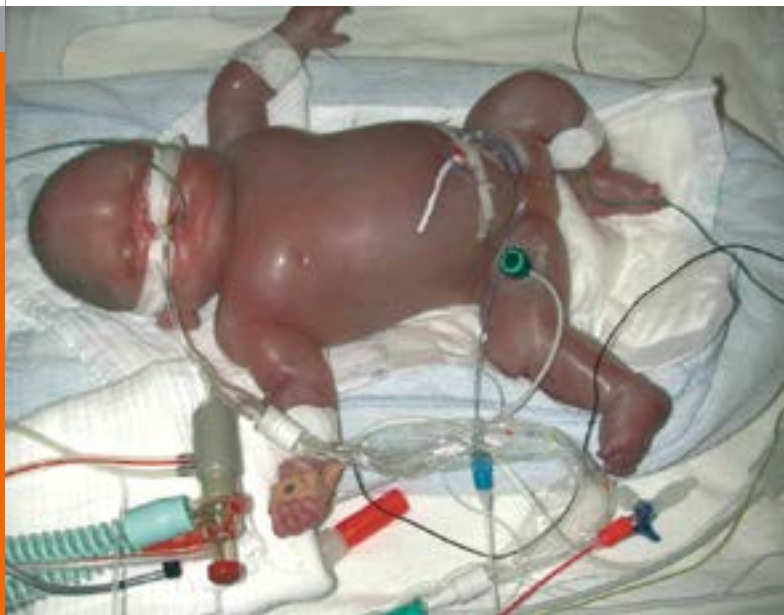
A premature female infant was born at 28 5/7 weeks gestational age by Caesarean section because of maternal preeclampsia. The mother, a 24-year-old G2/P2 had a history of weight gain of 10 kg in the previous two weeks of pregnancy. Prenatal ultrasound examination showed a fetus with severe hydrops with marked ascites, small bilateral pleural effusions, pericardial effusion with cardiomegaly and poor myocardial contractility, and oligohydramnios. Maternal serologies (HBV, CMV, EBV, HSV, parvovirus, rubella virus, toxoplasma, enteroviruses, lues, varicella, HIV) and fetal karyotype (46, XX) were unremarkable. The primary adaptation was poor, with Apgar scores of 1, 3, and 4 at 1, 5 and 10 minutes, respectively. Arterial cord pH was 7.33. The baby was intubated immediately after birth because of bradycardia and lack of spontaneous breathing. Epinephrine was administered intratracheally because of persisting bradycardia, following which the heart rate became normal. The baby remained cyanotic with saturations between 75% and 85% despite administration of 100% oxygen.

Blood gas analysis at 14 minutes of life revealed severe mixed acidosis (pH 7.00, pCO2 12.8 kPa, base deficit 12.8 mmol/l). Clinical examination showed massive hydrops (Fig. 1). Birth weight (2390 g) and head circumference (31 cm) were both above the 97th percentile, whereas the body length (39 cm) was between the 50th and 75th percentile. There

was radiological evidence of cardiomegaly (CT index 0.64) and ascites (Fig. 2). No relevant pleural effusions could be identified. Cardiomegaly was confirmed by echocardiography. The myocardium was thickened and edematous and characterized by very dense echoes. In addition, there was evidence of marked heart failure with enlargement of the right ventricle with tricuspid and pulmonary insufficiency, reduced left ventricular contractility with low cardiac output, and a moderately severe pericardial effusion (Fig. 3 and 4). Pathological laboratory values included a total protein of 15 g/l, an albumin of 8 g/l, a hemoglobin of 10.2 g/l, and platelets of 98'000/µl.

Preterm infant (gestational age 28 5/7 weeks, birth weight 2390 g) with severe hydrops.

Fig. 1



Because of poor oxygenation, the infant was switched from conventional to high frequency ventilation. Despite drainage of ascites, surfactant administration and circulatory support with volume and inotropes, the infant died at 10 hours of age. At autopsy, the cause of the hydrops fetalis was found to be extensive bilateral endocardial fibroelastosis, combined with dysplasia of the right ventricle (Fig. 5 and 6). Lung hypoplasia was also present.

Babygram showing generalized edema, cardiomegaly, as well as possible lung hypoplasia and ascites.

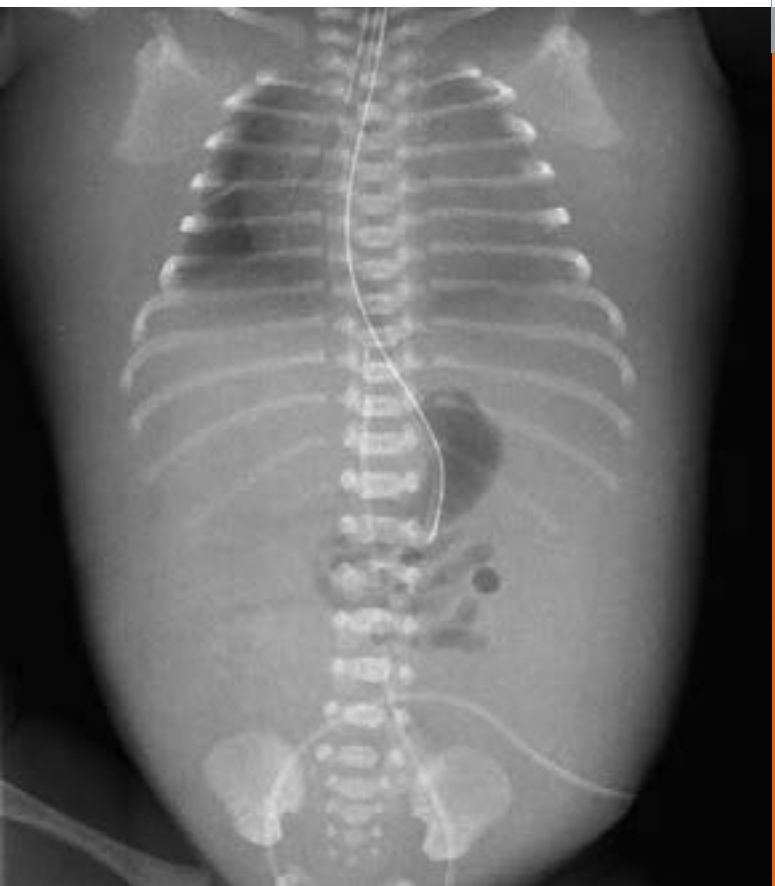


Fig. 2

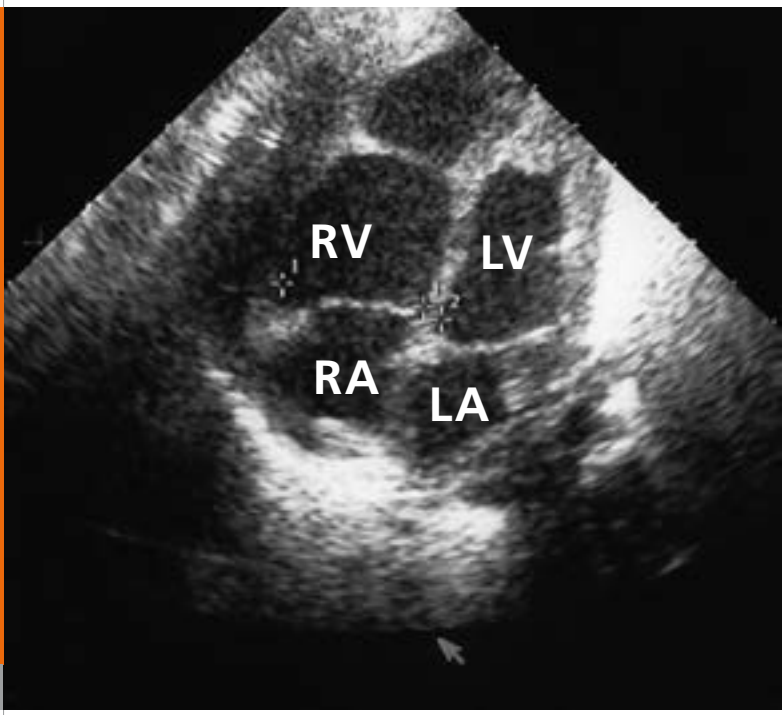


Fig. 3

Echocardiography: enlarged right ventricle (RV) and hyperechogenic myocardium (RA: right atrium, RV: right ventricle, LA: left atrium, LV left ventricle).

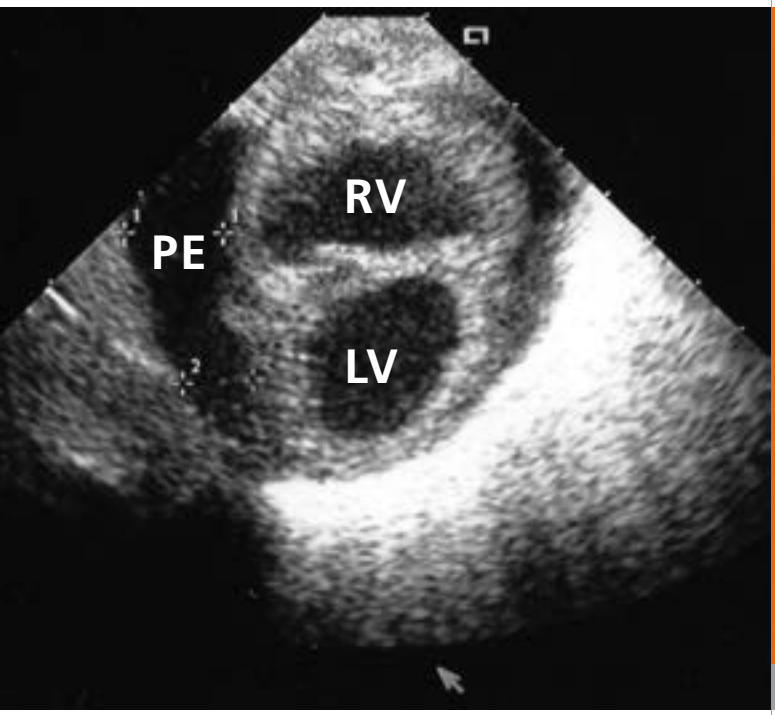


Fig. 4

Echocardiography: pericardial effusion (PE) (RV: right ventricle, LV: left ventricle).

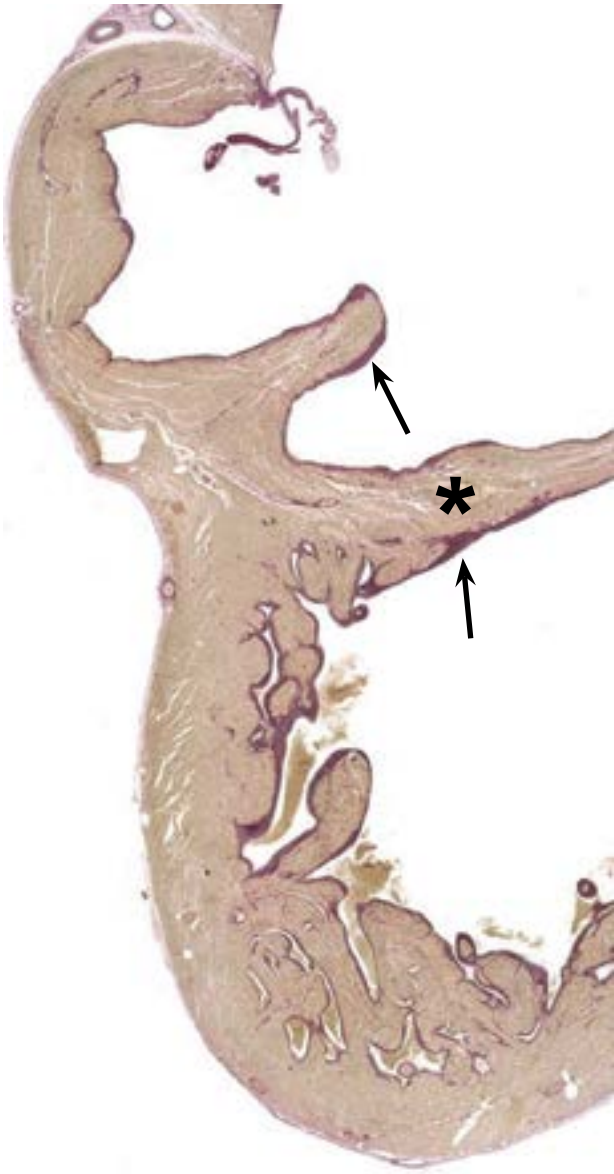
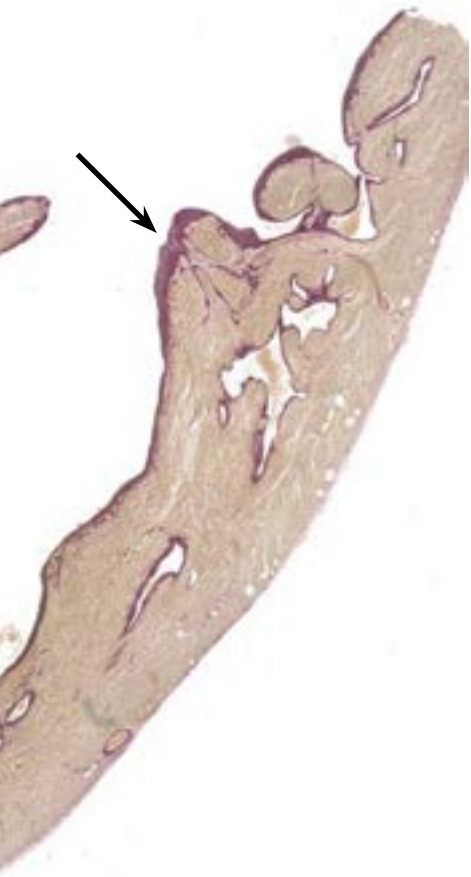


Fig. 5

Pathology (Elastica-van-Gieson staining): dysplasia of the right ventricle with prominent muscular ridge (asterisk), dividing the right ventricle into two parts; evidence of endocardial fibroelastosis (arrows).



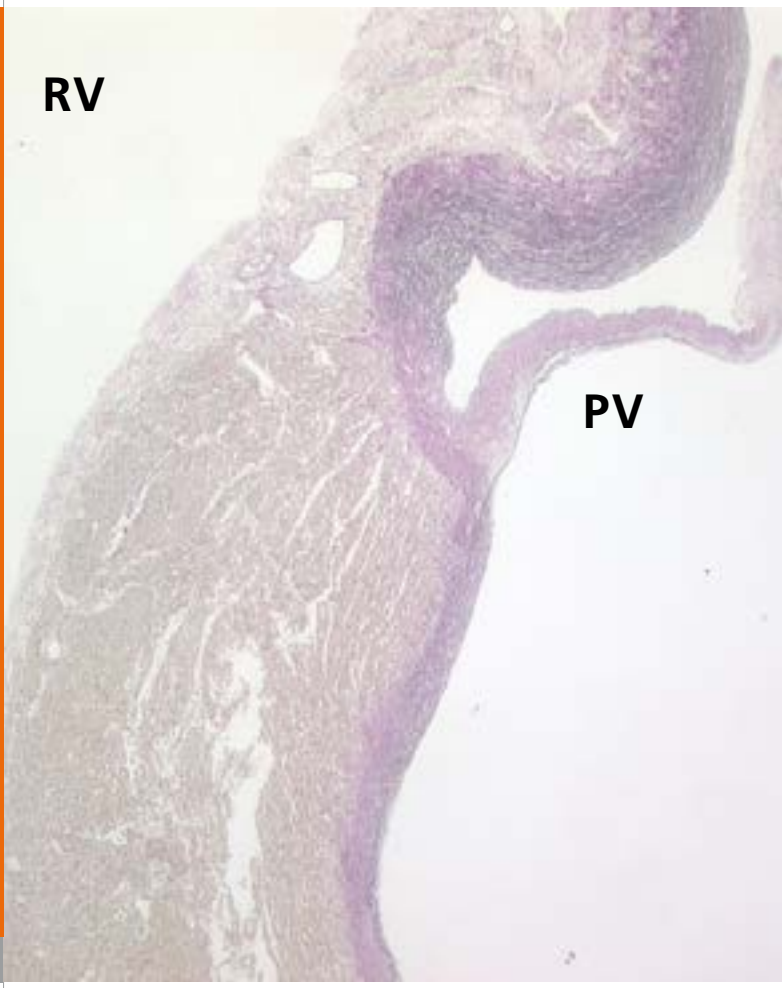
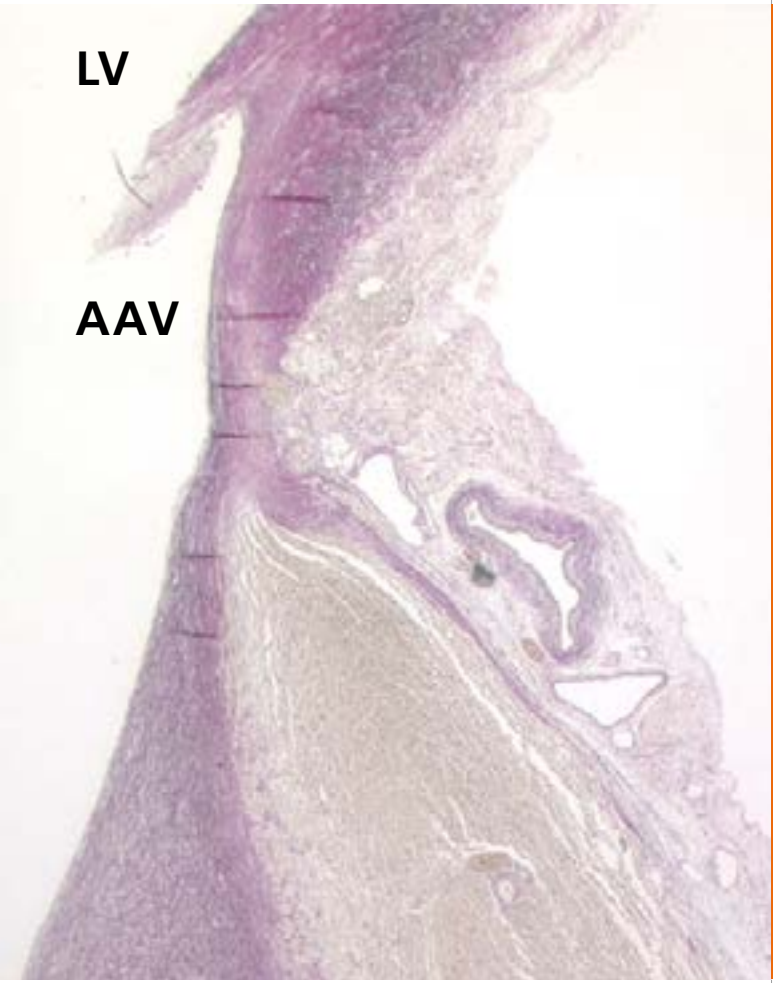


Fig. 5

Histology: endocardial fibroelastosis of right and left ventricle (fibrosis: pink, elastosis: grey) (LV: left ventricle, RV: right ventricle, PV: pulmonary valve, AAV: attachment of aortic valve).


LV
AAV

DISCUSSION

Hydrops is characterized by the presence of a generalized subcutaneous edema of the fetus or newborn, usually accompanied by ascites, pleural and pericardial effusions (1). Before the introduction of anti-D prophylaxis and intrauterine transfusions, the most common cause was rhesus incompatibility. Today, non-immunological causes are responsible for the majority of cases. These include cardiovascular disorders, pulmonary malformations, chromosomal defects, intrauterine infections, renal and gastrointestinal pathologies, placental anomalies, metabolic disorders, anemia of non-immunological origin and maternal diseases. The etiology remains unclear in approximately 20% of cases (2-5).

Endocardial fibroelastosis is one of the possible causes of hydrops fetalis. In this entity, there is a thickening of the endocardial layer with the presence of collagen and elastic fibers. The exact pathogenesis remains unclear. Endocardial fibroelastosis can occur without associated cardiac defects (primary form) or it develops as a secondary form due to structural heart anomalies (6, 7). In our case, there was a combination of endocardial fibroelastosis and right ventricular dysplasia; the concomitant presence of fibroelastosis in the left ventricle, however, suggests primary endocardial fibroelastosis.

The two major factors determining the overall prognosis in hydrops fetalis is the underlying etiology and the time of diagnosis (the earlier the diagnosis in pregnancy, the poorer the outcome). Furthermore, the mortality rate is significantly increased in the presence of pleural effusions when associated with consecutive lung hypoplasia. If a chromosomal defect or structural anomalies are excluded, symptomatic therapy of the fetus (e.g. intrauterine transfusion, fetal thoracocentesis) can markedly improve chances of survival (8, 9, 10). For long-term prognosis, particularly with respect to neurological development, gestational age at birth plays an important role (11).

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