Life-threatening congenital hydropericardium in a newborn with Down syndrome, transient abnormal myelopoiesis, Hirschsprung disease, and a ventricular septal defect

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Life-threatening congenital hydropericardium is an extremely rare condition. Common associated pathologies include infective intrauterine pericarditis, tumors, or hematological disorders, especially in children with coexisting Down syndrome (DS) [1, 2]. Genetic overexpression of critical loci on chromosomes q22.1 to q22.3 causes a significantly increased risk of acute megakaryoblastic leukemia. This may precede transient abnormal myelopoiesis (TAM) with mutations in hematopoietic transcription factor in the X-linked *GATA1* gene [3–5].

We present a 1-day-old male neonate with DS, congenital tamponade, and cardiopulmonary compromise. Fetal ultrasound diagnosed ventricular septal defect (VSD) with hydropericardium and enhanced nuchal translucency. Elevated pregnancy-associated plasma protein-A and human chorionic gonadotropin suggested DS, later confirmed on karyotype analysis.

The child was delivered *via* Caesarian section at 37 weeks' gestation weighing 2900 g and scoring 9–9–9–9 points on the Apgar scale at 1–3–5–10 minutes of life, respectively. However, the newborn's vital signs deteriorated prompting urgent examination: we found a dyspneic respiratory rate of 60 breaths/min, central cyanosis, tachycardia of 170 beats per minute with muffled heart sounds, low blood pressure of 46/32 mm Hg with mean arterial pressure of 36 mm Hg. A pulse oximetry test revealed low SaO_2 (80%) requiring oxygen delivery and respiratory support. Chest X-ray showed cardiomegaly with a cardiothoracic ratio of 1.0 (Figure 1A). Bedside transthoracic echocardiography (TTE) outlined 10 mm circumferential life-threatening tamponade (Supplementary material, *Videos S1, S2*), large VSD (Figure 1B–D, Supplementary material, *Video S3*), atrial septal defect (Figure 1E), and patent arterial duct with a bidirectional shunt indicating pulmonary hypertension. Urgent cardiac decompression using a 15G needle for pericardial drainage then pericardiocentesis was effective.

Hematology investigations showed a high blast count on blood smear (98%), hyperleukocytosis (73.7×10^3/μl), anemia, a reduced platelet count (15×10^3/μl), hyperuricemia (7.1 mg/dl), a normal protein level (5.9 g/dl), and elevated lactate dehydrogenase (1498 U/l) indicated TAM. After 24-hours, massive upper gastrointestinal bleeding appeared with hypotension and bradycardia requiring resuscitation, fresh frozen plasma, platelets, packed red blood cells, and phytomenadione. Intravenous vancomycin and meropenem, vasopressors, and inotropes (dopamine, milrinone) were used to treat clinical instability. Rasburicase was infused for tumor lysis syndrome prophylaxis and cytosine arabinoside (Ara-C) for cytoreduction. In follow-up, blood cell count showed remarkable recovery.

Within 2 weeks abdominal distention, flatulence, and constipation developed. Serial plain abdominal films showed dilated large intestine and a narrowed distal section. A suction biopsy of the rectal mucosal layers

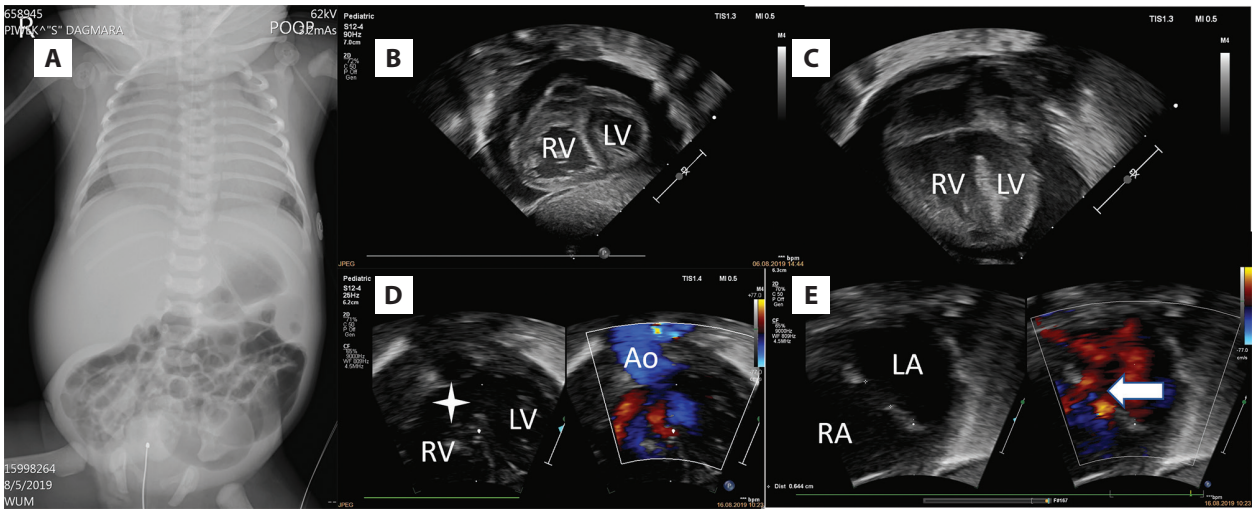

Figure 1. A. Chest and abdomen X-ray with an antero-posterior view showing extreme cardiomegaly, hepatomegaly, and distended intestines. **B.** TTE. A subcostal view showing cardiac tamponade. **C.** TTE. Four-chamber view showing cardiac tamponade with right atrial and ventricular enlargement and large ventricular septal defect. **D.** TTE. Five-chamber view showing large ventricular septal defect (the white star). **E.** TTE. Subcostal view showing atrial septal defect with two L-R shunts (the white arrow)

Abbreviations: Ao, aorta; LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle; TTE, transthoracic echocardiography

showed aganglionosis with a negative immunohistochemical calretinin test, confirming Hirschsprung's disease. Colon mapping with histopathological examination revealed aganglionosis in the distal segment of the sigmoid colon. Four weeks later, ileostomy was undertaken. Progressive signs of heart failure then required heart surgery at 7 weeks of age. Aortic cross-clamp and bypass circulation were used for VSD repair. The postoperative period was uneventful, and the patient was discharged home. Reverse ileostomy at 10 months successfully corrected Hirschsprung's disease.

At 1-year follow-up, the child was in a good condition but required multidisciplinary team support of a cardiologist, hematologist, endocrinologist (levothyroxine treated hypothyroidism), gastroenterologist, and physiotherapist.

Congenital hydropericardium with deranged hematology in DS is highly suggestive of TAM. Phenotype overexpression in DS provides a wide range of disorders with a need for early multidisciplinary therapy.

Article information

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