CASE REPORT

Ebstein cardiac anomaly, functional pulmonary atresia and isovaleric acidemia: A case report

Ammar M.H. Qadi *, Hussam K. Hamadah, Abdulraouf M.Z. Jijeh, Omar M. Hijazi, Mohamad S. Kabbani

King Abdul-Aziz Cardiac Center (KACC), King Abdul-Aziz Medical City (KAMC), King Saud University, Riyadh

[a] Saudi Arabia

In this report, we present a rare association between Ebstein anomaly (EA) and isovaleric acidemia (IVA) in a newborn who was admitted to our cardiac center. He underwent for PDA stenting to maintain adequate pulmonary blood flow, later he developed recurrent metabolic acidosis, prominent sweaty feet odor, neutropenia and thrombocytopenia. His organic acids profile in the urine confirmed the diagnosis of IVA.

To the best of our knowledge, there is no association between these two rare diseases. We are presenting this case report to highlight this rare association.

Keywords: Ebstein anomaly, Isovaleric acidemia, Congenital heart disease, Metabolic disorder

Introduction

Ebstein's anomaly (EA) is a congenital heart disease characterized primarily by abnormalities of the tricuspid valve (TV), right atrium (RA) and right ventricle (RV). The estimated risk of Ebstein's anomaly in the general population is one in 200,000 live births [1]. In newborns with this anomaly, tricuspid valve regurgitation may be significant as it can result in ineffective RV contractility that is unable to generate forward enough force to open the pulmonary valve in systole, thus producing 'functional' pulmonary atresia [2]. Isovaleric acidemia (IVA) is known as one of the 'classical' organic acidemias, and is caused by a genetic deficiency of isovaleryl-CoA dehydrogenase catalyzing the third step in leucine catabolism [3].

In this report, we describe one of the rare associations between EA and IVA diagnosed in an infant admitted to our center.

Case presentation

A two-day male newborn (3.9 kg) was admitted to our Pediatric Cardiac Intensive Care Unit.

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* Corresponding author. Address: Pediatric Cardiac ICU Division, Cardiac Science Department, King Abdul-Aziz Medical City, P.O. Box 22490, Mail Code 1423, Riyadh 11426, Saudi Arabia. Tel.: +966 18011111x13621; fax: +966 18011111x13679.
E-mail address: qadiam@ngha.med.sa (A.M.H. Qadi).

P.O. Box 2925 Riyadh – 11461KSA
Tel: +966 1 2520088 ext 40151
Fax: +966 1 2520718
Email: sha@sha.org.sa
URL: www.sha.org.sa

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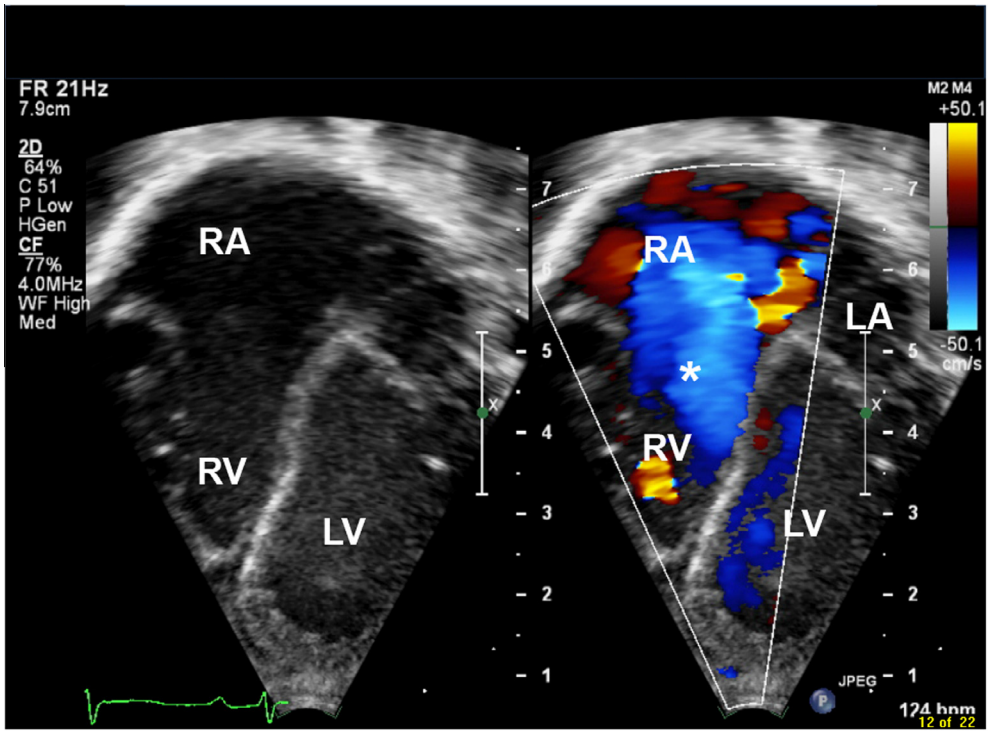

Figure 1. Trans thoracic echocardiography with Doppler showing tricuspid valve regurgitation. ∗ Tricuspid valve regurgitation, LV: Left ventricle, RV: Right ventricle, RA: Right atrium.

Diagnosed with cyanotic congenital heart disease, he was placed on prostaglandin E_1 (PGE_1) for further diagnosis and management, and intubated shortly after that. His chest X-ray examination showed cardiomegaly. Trans-thoracic echocardiography (TTE) confirmed diagnosis of Ebstein's anomaly, functional pulmonary atresia, and patent ductus arteriosus (PDA) with systemic to pulmonary shunt through PDA (Fig. 1). The patient was found to be dependent on PDA, and a trial of weaning from PGE_1 lead to significant desaturation. He was hemodynamically stable with PGE_1 infusion, and was maintained on 70–85% oxygen saturation.

The patient underwent PDA stenting by interventional cardiac catheterization on day 10 of life in order to maintain adequate pulmonary flow through PDA and to allow liberation from PGE_1 infusion. Following successful PDA stenting and liberation from PGE_1, the patient was weaned from the ventilator, and PGE_1 infusion was stopped completely with no hemodynamic compromise or desaturation. Saturation was maintained with normal pulmonary blood flow and normal lung vascularity (Fig. 2).

During the first 2 weeks of life, attempts to initiate feeding were unsuccessful due to both hemodynamic instability and to intolerance of enteral feeding. After extubation, feeding was established with regular baby formula. However, within 48 h of achieving full feeding, the baby started to have

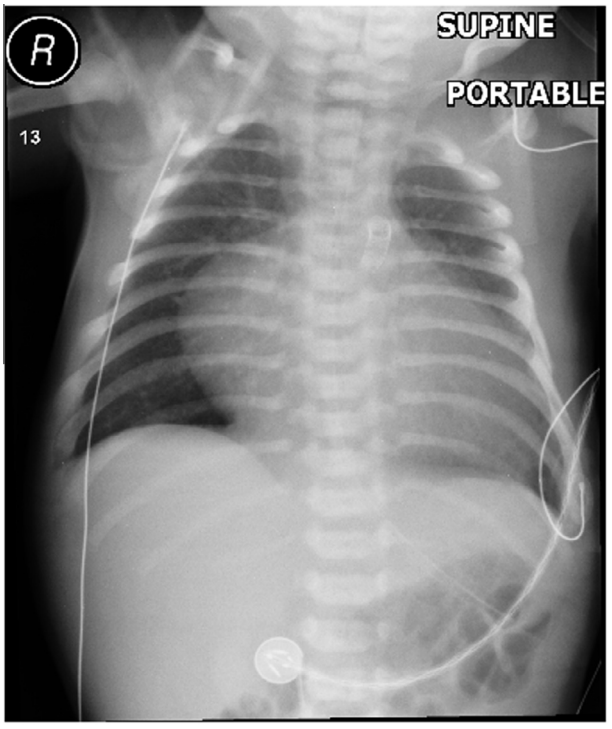

Figure 2. CXR after PDA stenting.