CASE REPORT

Clubbing, cyanosis and a new diagnosis of atrial septal defect in a parturient: a straightforward diagnosis of Eisenmenger syndrome?

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SUMMARY

A parturient required emergency admission to intensive care following a major postpartum haemorrhage. She was hypovolaemic, hypoxic and cyanosed and was also noted to have digital clubbing. A transthoracic echo showed a large atrial septal defect (ASD) with right heart dilation and possible anomalous pulmonary venous drainage. Eisenmenger syndrome was diagnosed with cyanosis due to shunt reversal at atrial level (right atrium to left), as a consequence of an elevated pulmonary vascular resistance from chronically increased pulmonary blood flow. After a full initial recovery, she was referred to a specialist grown-up congenital heart disease unit for further investigation. Cardiac MRI confirmed an ASD but showed that the cause of the cyanosis was anatomical in origin. An elongated Eustachian valve was diverting desaturated inferior venacaval blood across the intra-atrial septum into the left atrium. Her congenital heart defect was therefore amenable to corrective surgery.

BACKGROUND

This case highlights how even a simple congenital heart disease (CHD) lesion such as an atrial septal defect (ASD) may be complex in terms of both the anatomy and physiology. It demonstrates the need for specialist referral of all patients diagnosed with CHD to an adult specialist unit, where they will be assessed and investigated by experts. In this case

MRI by congenital imaging specialists defined the intracardiac anatomy and physiology, providing an explanation for the cyanosis. Redefining the diagnosis meant that rather than having an inoperable defect (Eisenmenger ASD), she had a lesion which was amenable to surgical repair.

CASE PRESENTATION

A 29-year-old Afro-Caribbean Primip was admitted to intensive care following a major postpartum haemorrhage. Prior to this event she had been well and asymptomatic with an uncomplicated pregnancy. On examination she was hypotensive and hypoxaemic and she was therefore fluid resuscitated, intubated and ventilated. On examination it was noted that she had digital clubbing. Her predelivery haemoglobin was 14 g/day. Transthoracic echocardiography diagnosed a large (36 mm) secundum ASD with probable partial anomalous pulmonary venous drainage (PAPVD). The right heart was dilated and the tricuspid regurgitant (TR) jet estimated a peak pulmonary artery systolic pressure of 60 mm Hg. The initial diagnosis was therefore Eisenmenger syndrome (ES) secondary to ASD and PAPVD.

She was haemodynamically stable and remained hypoxaemic but was extubated after 24 h. She was then transferred to the cardiology unit and was discharged home well 10 days later.

Figure 1 (A) Sagittal MRI from a three-dimensional SSFP sequence showing superior and inferior vena cava draining into the right atrium; however, this region of the interatrial septum is absent and the abnormally elongated Eustachian valve is directing inferior vena caval flow towards the left atrial chamber. (B) Schematic view in the sagittal plane depicting the right to left shunt across the defect.

To cite: Butcher C, Plyman C, Hughes M, et al. *BMJ Case Reports* Published online: [Please include Day Month Year] doi:10.1136/bcr-2012-007992

In light of the new diagnosis of ES and underlying CHD, she was referred for follow-up to the specialist Grown-Up Congenital Heart Unit, The Heart Hospital. From the history it was noted that she was NYHA II prior to pregnancy. She denied any other symptoms and had no change in functional class during pregnancy. The pregnancy had been uncomplicated and she had remained well throughout. On examination it was noted that she clubbed with oxygen saturations of 88% in room air, jugular venous pressure was not elevated. Blood pressure was 126/70. First and second heart sounds were present with a fixed split of the second heart sound, in addition to a 2/6 ejection systolic murmur and 1/4 diastolic murmur, heard loudest at the upper-left sternal edge.

INVESTIGATIONS

A 12-lead ECG showed sinus rhythm with a rate of 100 beats/min and evidence of right ventricular hypertrophy.

Transthoracic echocardiography showed a large secundum ASD (36 mm) with probable anomalous drainage of the right pulmonary veins into the right atrium. The right ventricle was dilated with good systolic function. Left ventricular function and valvar function were normal.

Cardiac MRI confirmed the presence of a secundum ASD and anomalous drainage of the right lower pulmonary veins into the right atrium. The inferior vena cava straddled the ASD and Eustachian valve (figures 1A,B and 2). This resulted in diversion of desaturated blood from the inferior vena cava across the atrial septum via the defect into the left atrium.

OUTCOME AND FOLLOW-UP

Expert congenital cardiac MRI provided anatomical definition of the congenital defect, while at the same time showing how the unusual structural anatomy explained the presence of cyanosis. The diagnosis of ES was therefore incorrect and the defect was amenable to corrective surgery. She subsequently underwent surgery 10-month postpartum and made an excellent postoperative recovery (figures 3 and 4).

DISCUSSION

This case highlights, how what is often considered a 'simple' congenital heart lesion—an ASD, may be both anatomically and

Figure 3 Sagittal MRI SSFP image showing the postintervention appearance, with a patched interatrial septum.

physiologically complex. In this particular case the clinical findings of clubbing, cyanosis and ASD were not due to ES, but were explained by the unusual and complex intracardiac anatomy of the defect, which meant that it was entirely amenable to surgical repair.

This case demonstrates the need for referral of patients with even simple CHD to a specialist unit, where there they can be assessed by experts and there is access to specialist imaging with not only two-dimensional (2D) echo but also 3D MRI. It is recommended that all patients with a diagnosis of CHD, irrespective of lesion complexity, should be seen once in a specialist unit during their lifetime. This applies not only to newly diagnosed patients but also to those previously diagnosed. The

Figure 2 Preoperative two-dimensional echocardiogram (apical 4 chamber view) showing the atrial septal defect between the left and the right atrium (depicted by *).

Figure 4 Postoperative two-dimensional echocardiogram (apical 4 chamber view) showing the patched interatrial septum.

rationale for this is that diagnostic and particularly imaging techniques have improved hugely in the past decade and some historical diagnoses may be redefined in the current era.

This case also highlights the pitfalls of echo assessment of pulmonary artery pressure (PAP) estimation in the presence of intracardiac shunt lesions. Echo uses the TR jet to estimate PAP and it will often be elevated if there is a large shunt at atrial level due to increased pulmonary blood flow. If there is ever doubt about PAP/pulmonary vascular resistance, cardiac catheterisation should be performed to measure PVR.

It is also worth noting that Eisenmenger ASD is a rare diagnosis accounting for only 10% ES cases;¹ therefore, this diagnosis should be made with caution and specialist assessment is recommended.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCE

- 1 Gault JH, Morrow AG, Gay WA Jr, *et al.* Atrial septal defect in patients over the age of forty years clinical and hemodynamic studies and the effects of operation. *Circulation* 1968;37:261–72.

Learning points

- ▶ Even simple congenital heart disease (CHD) lesions can be anatomically and physiologically complex therefore all patients with a diagnosis of CHD should be seen in a specialist grown-up CHD unit for evaluation at least once during their lifetime.
- ▶ A diagnosis of Eisenmenger ASD is rare and should therefore be made with caution and investigated fully to confirm elevation of pulmonary vascular resistance, with exclusion of other aetiologies.
- ▶ MRI by congenital specialists provides anatomical and physiological information which complements other diagnostic modalities such as echo and cardiac catheterisation which are all often needed in the assessment of patients with CHD.

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