

A 32-year-old woman with Down's syndrome and central cyanosis

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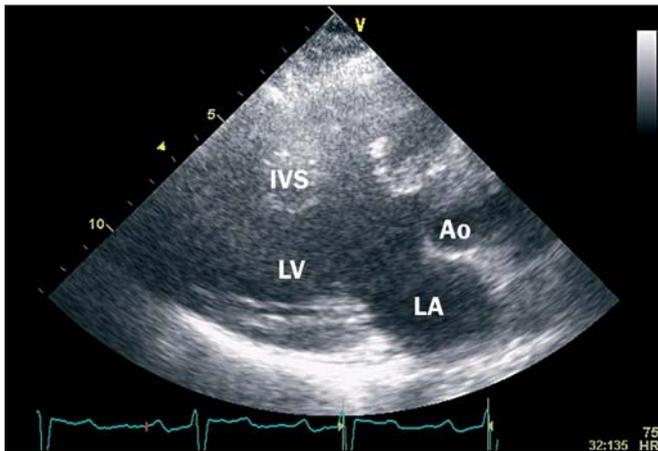


Figure 1. Parasternal long axis view. LA=left atrium, LV=left ventricle, IVS=interventricular septum, Ao=aorta.

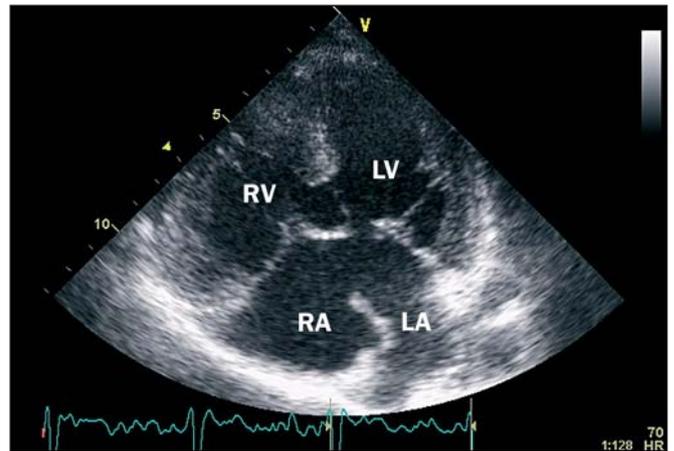


Figure 2. Four-chamber view. LA=left atrium, LV=left ventricle, RA=right atrium, RV=right ventricle.

A 32-year-old woman was referred to our hospital because of a progressive decline in her physical condition. Her medical history revealed a congenital heart defect and Down's syndrome. In the past, surgery for her heart defect had been considered, but ultimately was not performed. On physical examination central cyanosis was evident. Subsequently echo-

cardiography was performed (figures 1 and 2; see also movies 1 and 2 on the journal's website at www.cardiologie.nl).

What is your diagnosis and how would you treat this patient? ■

Answer

You will find the answer on page 181.

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In this section a remarkable 'image' is presented and a short comment is given.

We invite you to send in images (in triplicate) with a short comment (one page at the most) to Bohn Stafleu van Loghum, PO Box 246, 3990 GA Houten, e-mail: l.meester@bsl.nl.

'Moving images' are also welcomed and (after acceptance) will be published as a Web Site Feature and shown on our website: www.cardiologie.nl

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Answer to the imaging on page 171

The echocardiographic findings are consistent with complete atrioventricular septal defect, see also movies 1 and 2 on the journal's website at www.cardiologie.nl. Down's syndrome is strongly associated with this congenital heart defect.¹ An essential morphological hallmark is the presence of a common atrioventricular junction: there are five leaflets, two of which are bridging leaflets.¹ Nowadays the recommended treatment is surgical repair, preferably before the age of six months.¹

Pulmonary artery pressure could not reliably be assessed with echocardiography, but because of the central cyanosis an Eisenmenger reaction with pulmonary hypertension is likely. In the BREATHE-5

trial treatment of patients with Eisenmenger syndrome with bosentan, an endothelin receptor antagonist, has been shown to improve exercise capacity and haemodynamics.² Treatment with bosentan 62.5 mg twice a day was started; after four weeks the dose was increased to 125 mg twice a day. Unfortunately, after four months of treatment the six-minute walking distance showed no significant improvement. ■

References

- 1 Graig B. Atrioventricular septal defect: from fetus to adult. *Heart* 2006;**92**:1879-85.
- 2 Galiè N, Beghetti M, Gatzoulis MA, et al. Bosentan therapy in patients with Eisenmenger syndrome: a multicenter, double-blind, randomized, placebo-controlled study. *Circulation* 2006;**114**: 48-54.