Right ventricular enlargement in a patient with hereditary haemorrhagic telangiectasia: a rare case of pulmonary arteriovenous malformations with concomitant atrial septal defect

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We describe a rare case (32-year-old woman) with hereditary haemorrhagic telangiectasia and concomitant atrial septal defect (ASD). Transthoracic saline contrast echocardiography (TTCE) revealed several microbubbles appearing early (third cardiac cycle) and significant microbubbles appearing later (seventh cardiac cycle) in the left atrium and ventricle (LA/LV) after opacification of the right atrium in the four-chamber view (panel A). Total left to right shunt was calculated at 3.9 L/min (Qp:Qs = 1.8:1). Despite right ventricular (RV) enlargement, search for intracardiac shunting was not undertaken because chest CT confirmed significant pulmonary arteriovenous malformations (PAVMs) in the right lower lobe (arrows in panel B). Following successful transcatheter coil embolization, TTCE demonstrated significant microbubbles within two cardiac cycles (panel C), consistent with intracardiac shunting. Owing to findings of RV dilatation and early positive microbubbles, a transoesophageal echocardiogram (TOE) was performed, identifying a secundum ASD in the bivacal view (panel D) and three-dimensional (3D) TOE en-face view of the interatrial septum from the LA aspect (arrow in panel E). Using a 3D cross-sectional area of 0.48 cm², the shunt across the ASD was estimated at 2.9 L/min (Qp:Qs = 1.6:1), implying intrapulmonary PAVMs flow of 1.0 L/min prior to closure. Coexistence of intracardiac and extracardiac malformations should be considered when early appearance of microbubbles is seen with TTCE even with known PAVMs. Furthermore, TOE should be performed in patients with RV dilatation of uncertain aetiology despite known extracardiac shunting. Intracardiac and extracardiac shunts are likely to co-exist when TTCE shows early appearance of microbubbles even if maximal microbubbles appear later.

Conflict of interest: none declared.

Supplementary data are available at European Heart Journal – Cardiovascular Imaging online.

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