Late diagnosis of a congenital apical ventricular septal defect with complete closure by right ventricular trabeculations

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Here, we report the case of a 48-year-old male who recently complained of unspecific symptoms such as sweating, palpitations, and atypical chest pain. Diagnostic work-up at the local hospital included exercise stress testing, which was found to be inconclusive. Cardiac catheterization showed normal coronary arteries. However, on levocardiography, the suspicion of a left ventricular (LV) aneurysm was raised (upper left Panel, black arrows) and the patient referred for surgical therapy to our tertiary cardiac care centre. At that time, the patient already lost his employment as an engine driver and was in serious fear of sudden rupture of the presumed aneurysm.

Diagnostic work-up at our institution including echocardiography and magnetic resonance imaging (MRI) revealed a large apical ventricular septal defect (VSD). Importantly, there was separation of the right ventricular (RV) apex from the remaining RV by excessive trabeculations and thereby elimination of any left to right shunt across the defect (upper mid and right Panel; lower panels demonstrating MRI short-axis views through the defect at an apical level (left and mid Panel) and 10 mm above (right Panel); in the mid panel, the morphological LV is coloured in red, the morphological RV in blue, and the morphological RV but functional LV in green; note the uncoloured bright blood pool between red and blue resembling the VSD).

This type of apical VSD constitutes a rare and distinct type of morphology and physiology and has to be considered in patients with suspected LV aneurysm, absent of coronary artery disease and lack of any heart murmur.

The patient was reassured that this condition has always been there, does not need any treatment and is not related to his unspecific symptoms.