

# Atrial septal defect type II and upper limb malformation in 40-year-old male as a manifestation of Holt-Oram syndrome

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The present study is the case of a 40-year-old Caucasian male with bilateral, symmetric malformation of the upper limbs; absence of I metacarpals and thumbs (Fig. 1D, E). He was admitted to the Department of Cardiology because of impaired exercise tolerance (New York Heart Association [NYHA] II/III) and recurrent palpitations. His mother died when he was an infant at the age of 36 years, and two of his brothers died at the age of 3 and 15 years because of untreated congenital heart disease — none of them presented with skeletal abnormalities.

On electrocardiogram sinus rhythm was 70 bpm, first-degree atrioventricular block and incomplete right bundle branch block were found. Transthoracic and transesophageal echocardiography revealed normal left ventricular function and right ventricular overload due to large, an unusually elongated (elliptic, 27 × 13 mm in size) type II

atrial septal defect with hemodynamically significant left-to-right shunt (TAPSE 20 mm, S' RV 13 cm/s, RVIT 52 mm, RAA 24 cm<sup>2</sup>, Fig. 1A, B). Based on four-dimensional echocardiographic area sizing, an atrial septal defect nitinol occluder (Memopart 26 mm, Lepu Medical) was implanted percutaneously via femoral vein (Fig. 1C) with complete atrial septal defect closure. Control echocardiography after 3 and 6 months showed a good result of occluder implantation with significant clinical improvement (NYHA I).

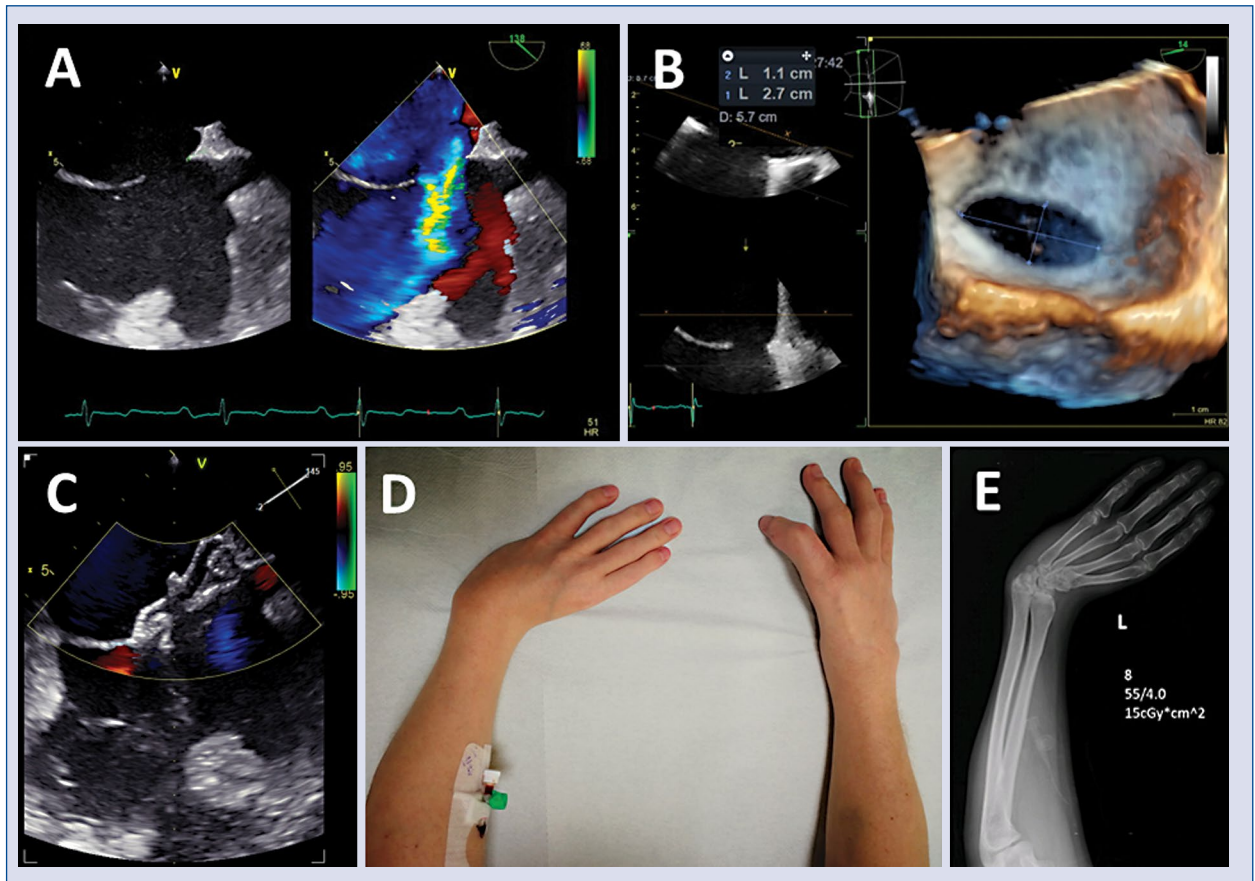
This case report is an example of Holt-Oram syndrome (heart-hand syndrome) which is an autosomal dominant disorder characterized by upper limb malformations in association with congenital heart lesions and increased risk for cardiac conduction abnormalities. Life expectancy for Holt-Oram syndrome varies among affected individuals and predominantly depends on the severity of the congenital heart defect.

**Conflict of interest:** None declared

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**Figure 1.** A. Two-dimensional transesophageal echocardiography with color Doppler — type II atrial septal defect with hemodynamically significant left-to-right shunt; B. Three-dimensional transesophageal echocardiography — elongated (elliptic, 27 × 13 mm in size) type II atrial septal defect; C. Two-dimensional transesophageal echocardiography with color Doppler — good result of Memopart 26 mm occluder implantation; D, E. Malformation of upper limbs (real photo and X-ray) — absence of I metacarpals and thumbs.