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【 PICTURES IN CLINICAL MEDICINE 】

Heart-hand Syndrome

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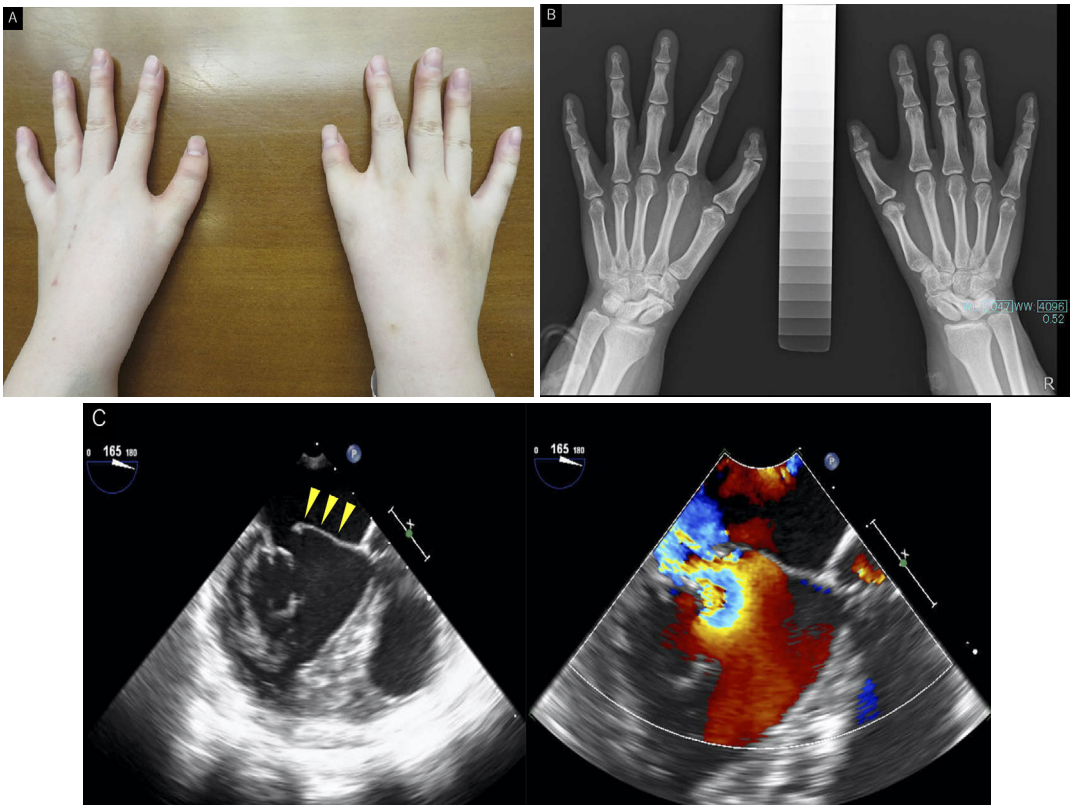

Picture.

A 30-year-old woman with a history of atrial septal defect disclosure, polydactyly (extra digits resected) and sick sinus syndrome was referred to undergo echocardiography. Her cardiac family history was unremarkable. Her thumb was attached to the upper rim of the palm, aligning the first interdigital web parallel with other web lines (Picture A). Hand radiography showed that the distal phalanges of the thumbs deflected ulnarly (Picture B). Transthoracic and transesophageal echocardiography revealed mitral regurgitation due to prolapse (Picture C). Her polydactyly along with cardiovascular comorbidities prompted suspicion of Holt-Oram Syndrome (HOS). The patient underwent genetic testing for HOS, and a heterozygous pathogenic tryptone bile X-glucuronide (TBX):c.148-1G>C variant was identified in intron 2 of the *TBX5* gene. HOS, also known as heart-hand syndrome, is a rare autosomal heritable disorder characterized by skeletal upper-limb deformities and cardiac defects due to *TBX5* variants (1). Atrial and ventricular septal malformations are prevalent, although left-sided abnormalities, including mitral valve prolapse and conduction system de-

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fects, have also been reported (2).

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