Association of Primum-Type Atrial Septal Defect and Patent Foramen Ovale in a Patient with Holt-Oram Syndrome

Holt-Oram Sendromlu Hastada Primum Tip Atriyal Septal Defekt ve Patent Foramen Ovale Birlikteliği

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A 49-year-old woman with dwarfism reported signs of shortness of breath. A physical examination of the patient revealed congenital deformities in the hands and feet (Figure 1). Radiographs showed that the extremities of both her hands had metatarsal polydactyly; moreover, with her feet seem to have epiphyseal dysplasia and polysyndactyly were determined (Figure 2). Telecardiography showed cardiomegaly, and electrocardiography showed right axial deviation and right ventricular hypertrophy. Transthoracic echocardiography revealed primum-type atrial septal defect in the interatrial septum, dilatation of the right heart cavities, and myxomatous mitral valves (Figure 3A). Transesophageal echocardiography showed a 2.5 cm-wide, primum-type atrial septal defect and a wide patent foramen ovale (Figure 3B). A pulmonary arterial pressure of 47/14 (average: 28) mmHg, a systemic blood flow (QS)/ pulmonary blood flow (QP) of 2.88, and a PVR of 2.15 Wood units were measured in cardiac catheterization. The patient with skeletal dysplasia and congenital cardiac defect was diagnosed with Holt-Oram syndrome and was referred for surgery. After median sternotomy, right atriotomy was performed by cardiopulmonary bypass under moderate hypothermia using the aortic-bicaval cannulation technique; the primum-type atrial septal defect was repaired using the pericardial patch, and the patent foramen was primarily repaired. The patient did not have any postoperative problems and was discharged on the 5th day.

Holt-Oram syndrome is a hereditary disease with an autosomal-dominant, inherited skeletal system dysplasia and congenital cardiac anomalies[1,2]. This syndrome, associated with upper extremity anomalies and congenital heart lesions, was first identified by Holt and Oram in 1960 by detecting it in nine affected individuals belonging to four generations of a family[3]. In this rare case report, we present a case of Holt-Oram syndrome that was diagnosed with primum-type atrial septal defect and patent foramen ovale and was treated using corrective heart surgery.

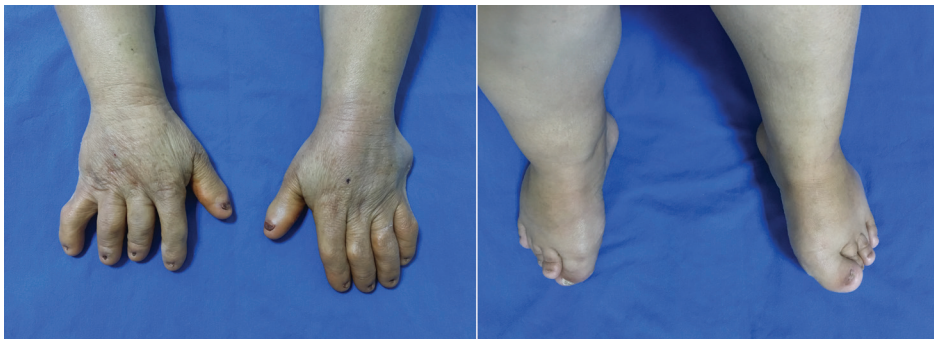

Figure 1. Congenital deformities in the hands and feet of the patient.

Cite this arcticle as: Kaya H, Coşkun MS, Yavuz C, Ertaş F. Association of primum-type atrial septal defect and patent foramen ovale in a patient with holt-oram syndrome. Koşuyolu Heart J 2019;22(2):141-2.

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Submitted: 01.04.2019
Accepted: 19.05.2019

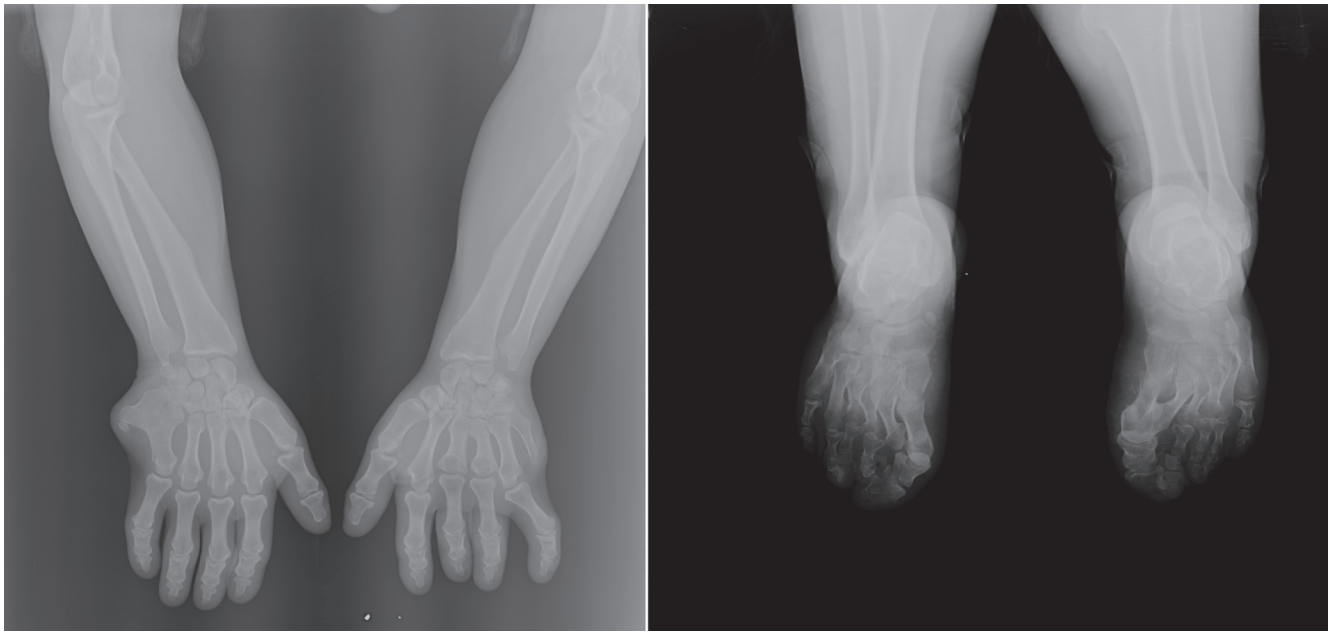

Figure 2. Radiographs of the hands and feet of the patient.

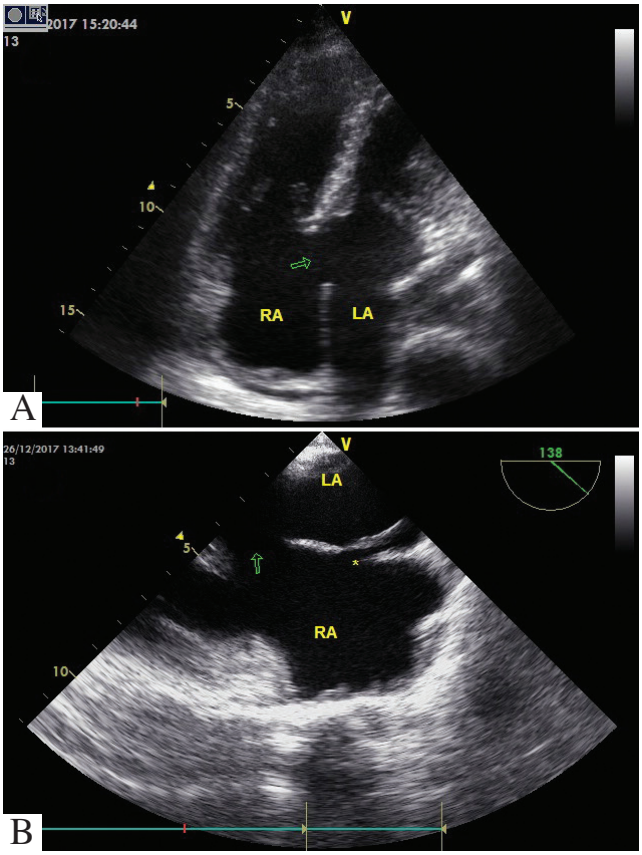

Figure 3. (A) Transthoracic echocardiography revealed primum-type atrial septal defect (arrow) **(B)** Transesophageal echocardiography showed primum-type atrial septal defect (arrow) and patent foramen ovale (asterisk) (LA: Left atrium, RA: Right atrium).

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