The Holt-Oram syndrome is an autosomal dominant heritable disorder characterized by skeletal upper-limb dysplasias and congenital cardiac defects. We describe a 43-year-old woman who presented with paroxysmal tachycardia and progressive heart failure. Both ring fingers were abnormally short as a result of dysplasia of metacarpal IV (Figure 1). Auscultation revealed a loud systolic murmur at the left sternal margin and a widely split $S_2$. The ECG was consistent with right ventricular hypertrophy (Figure 2). The chest x-ray showed severe right atrial and right ventricular dilatation and marked bilateral central pulmonary arterial distension with small peripheral pulmonary vessels (Figure 3, left). Echocardiography showed a large secundum-type atrial septal defect with signs of severe right ventricular volume overload. At right heart catheterization, oximetry showed a large left-to-right shunt at the atrial level with a Qp/Qs ratio of 2.3. The mean pulmonary arterial pressure was 53 mm Hg, and the wedge pressure was 13 mm Hg. A pulmonary arteriolar resistance of 851 dyn·s·cm$^{-5}$ was calculated. Operative closure of the atrial septal defect was undertaken despite markedly elevated pulmonary arteriolar resistance. Three months after the procedure, the patient reported excellent clinical improvement: there was no murmur, the size of the heart and the radiological signs of pulmonary hyperperfusion had decreased significantly (Figure 3, right), pulmonary artery mean pressure was 14 mm Hg, and pulmonary arteriolar resistance had decreased to 412 dyn·s·cm$^{-5}$.

Figure 1. Patient’s hands. Both ring fingers are abnormally short as a result of dysplasia of metacarpal IV.

Figure 2. ECG showing P pulmonale, right axis deviation, and qR pattern in V1 and V2, thus being compatible with right atrial and right ventricular hypertrophy.
Figure 3. Left, Chest x-ray before surgery. Right ventricle, right atrium, and central parts of pulmonary artery are massively enlarged. Right, Chest x-ray after operative closure of atrial septal defect, showing significant reduction of right atrial and right ventricular dimensions and of central pulmonary artery distension.