A 29-year-old woman presented with dizziness. Physical and radiographic examinations showed skeletal hand malformations, i.e., digitalized triphalangeal thumbs and dys trophy of the carpal bones. When she was 15 years old, an atrial septal defect had been repaired. ECG recordings showed abnormalities of atrial excitation such as a wandering pacemaker, atrial ectopic activity, AV-nodal block, and sinus bradycardia of <60 bpm.

A clinical diagnosis of Holt-Oram syndrome was made. Holt-Oram syndrome is the prototype of heart-hand syndromes and has recently been mapped to the long arm of chromosome 12 (12q2). It must be differentiated from heart-hand syndrome type II (Tobatznik’s syndrome) and heart-hand syndrome type III (MIM No. 140450), which are phenotypically similar. The latter do not map to 12q2, and atrial septal defects do not occur in these conditions. Approximately 350 cases have been published. The autosomal dominant syndrome has a strong variance in phenotype expression. Approximately 50% of the patients have cardiac malformations. The limb malformations can vary from mild thumb abnormalities to complete absence of the arm. The presence of upper-limb abnormalities in a patient with cardiac disease can be the clue to the diagnosis of a complex congenital syndrome.

References

Figure 1. Photograph of patient’s hands.
Figure 2. Radiograph of both hands. Note triphalangeal thumbs, slightly hypoplastic thumb of right hand, and asymmetrical dysplastic carpal bones.

Figure 3. ECGs recorded on admission (A) and 6 (B) and 24 (C) hours after admission. Note differently shaped P wave marked with arrows at III in all recordings.
Holt-Oram Syndrome
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Circulation. 1998;98:2636-2637
doi: 10.1161/01.CIR.98.23.2636

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