Holt-Oram Sendromu: Olgu Sunumu

HOLT-ORAM SYNDROME (HEART-HAND SYNDROME): CASE REPORT

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Özet

Holt-Oram sendromu üst ekstremite iskelet displazisi ve konjenital kardiyak defektlerle karakterize otozomal dominant geçişli herediter bir hastalıktır. Olgumuz her iki elde birinci parmak yokluğu, ikinci ve üçüncü parmaklarda basit inkomplet sindaktili ve torakal bölgede konjenital kifoskolyozu olan on yaşında erkek çocuktu. Hastanın her iki önkol ve el grafilerinde tip C radius aplazisi ve birinci ray yokluğu, torakal grafide ise kifoskolyoz saptandı. Ekokardiyografide geniş atriyal septal defekt vardı. Atriyal septal defekt kardiyopulmoner bypass kullanılarak perikard yama ile kapatıldı. Literatürde Holt-Oram sendromu ile birlikte şiddetli kifoskolyoza rastlamadığımız için bu olguyu yayınlamayı uygun bulduk.

Anahtar kelimeler: Holt-Oram sendromu, konjenital kalp defekti, kifoskolyoz, atriyal septal defekt

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Summary

Holt-Oram Syndrome is a hereditary disease transmitted by autosomal dominant inheritance characterized by skeletal dysplasia of upper extremities and congenital cardiac defects. Our case was a 10-year-old male patient with absence of first finger, simple incomplete syndactiliy in second and third fingers in both hands and congenital kyphoscoliosis of thoracal region. Type C radius aplasia was found in the X-ray films of both forearms and absence of first ray, kyphoscoliosis was identified in thoracal radiographs. There was a wide atrial septal defect in echocardiography which further was closed by cardiopulmonary bypass with pericardial patch. We find it interesting to present this case since there is no reported Holt-Oram Syndrome associated with severe kyphoscoliosis in the literature.

Keywords: Holt-Oram syndrome, Congenital heart defect, kyphoscoliosis

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Introduction

Holt-Oram Syndrome is a hereditary disease transmitted by autosomal dominant inheritance characterized by skeletal dysplasia of upper extremities and congenital cardiac defects. This syndrome has a wide clinical spectrum, from subclinic radiological findings to life threatening disease. Anomalies of upper extremities are usually present. Cardiac anomalies include atrial and ventricular septal defects and conduction disorders [1-5].

Cases

A 10-year-old boy presented with complaint of palpitation since 2 months. The patient had absence of first fingers, simple incomplete syndactyly in second and third fingers in both hands, and congenital kyphoscoliosis in thoracal region (Figure 1). Both forearm and hand radiographs of the patient showed type C radius aplasia and absence of first ray (Figure 2), and thoracal radiographs revealed kyphoscoliosis (Figure 3). There was systolic murmur on the left sternal margin and fixed splitting of the second heart sound on auscultation. Electrocardiogram (ECG) revealed no pathologies. Chest x-ray showed a mild dilatation of right atrium and right ventricle (Figure 4). Wide atrial septal defect was observed in echocardiography. Atrial septal defect (ASD) was closed with pericardial patch by cardiopulmonary bypass.

Discussion

Holt-Oram Syndrome was first described in 1960 as co-existence of ASD, conduction disorders on ECG and hand malformations [5]. The responsible gene for Holt-Oram Syndrome is reported to be localized in long arm of chromosome 12 (12p2). It has autosomal dominant inheritance. Its prevalence is calculated as 0.95 in 100.000 births. Eightyfive percent of cases are associated with new mutations. Basson and associates [1] reported two families with 19 and 18 affected members having Holt-Oram Syndrome. No pathology was found in family members of our case.

Upper extremity abnormalities are usually present which can be unilateral or bilateral. Typically these include structures originating from embriologic radial ray such as radial, carpal and thenar bones. Aplasia, hypoplasia, fusion and abnormal development of these structures include a wide spectrum of clinical disorders such as phocomelia, foreshortened arm, absence of first finger and triphalangia [1]. Our case had absence of first finger and simple incomplete syndactiliy of

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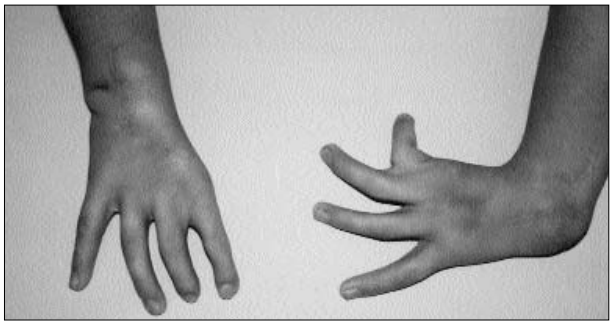

Figure 1. Patient's hands. Absence of first fingers, simple incomplete syndactiliy in second and third fingers.

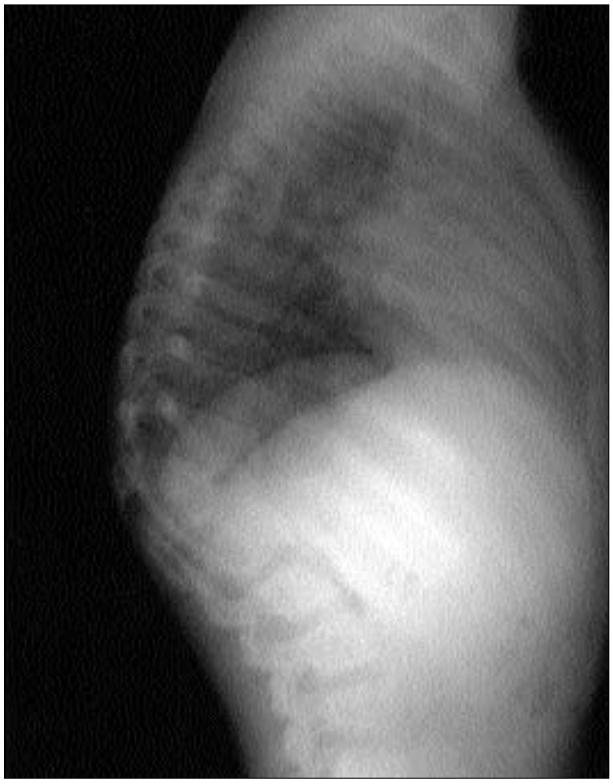

Figure 3. Kyphoscoliosis on the thoracal radiograph.

second and third fingers in both hands and x-ray films of both forearms and hands showed type C radius aplasia and absence of first ray. He also had congenital severe kyphoscoliosis in thoracal region.

There can be single or multiple atrial and ventricular septal defects with or without sinus bradycardia and atrioventriküler blocks at different degrees in Holt-Oram Syndrome [1,5]. Persistent left superior vena cava, patent ductus arteriosus, abnormal left coronary artery ostium and atrioventriküler canal defect are also reported [1,3,5]. Atrial septal defect was present in our case and was closed by cardiopulmonary by-pass using pericardial patch.

Holt-Oram Syndrome is a rare anomaly and it was suitable to report this interesting case having severe kyphoscoliosis besides cardiac and upper extremity anomalies.

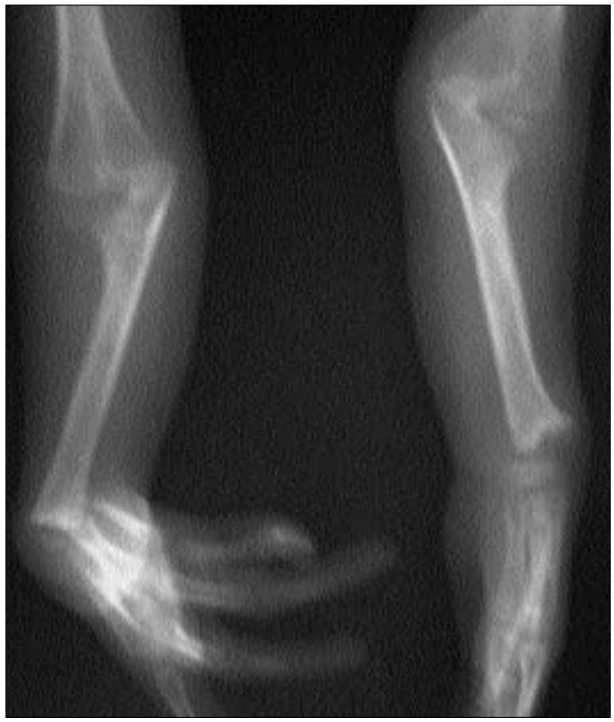

Figure 2. Radiograph of both hands. Note the type C radius aplasia and absence of first ray.

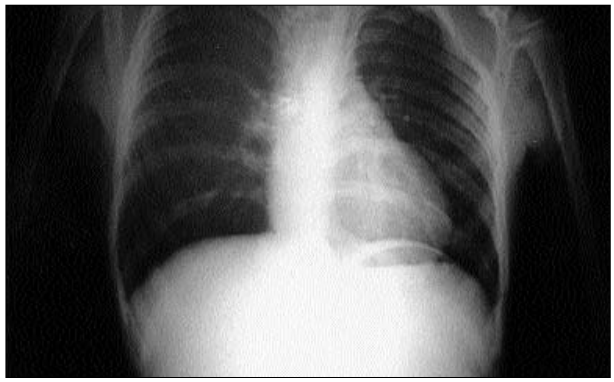

Figure 4. Chest X-ray. Right ventricule and right atrium are mild enlarged.

References

1. Basson CT, Cowley GS, Solomon SD, et al. The clinical and genetic spectrum of the Holt-Oram syndrome (Heart-Hand syndrome). N Engl J Med 1994;330:885-91.
2. Böhm M. Holt-Oram syndrome. Circulation 1998;98:2636-7.
3. Brockhoff CJ, Kober H, Tsilimingas F, Dapper F, Münzel T, Meinertz T. Holt-Oram syndrome. Circulation 1999;99:1395-6.
4. Ogur G, Gül D, Lenk MK, Imirzalioglu N, Alpay F, Ogur E. Variable clinical expression of Holt-Oram syndrome in three generations. Turk J Pediatr 1998;40:4:613-8.
5. Zhang KZ, Sun QB, Cheng TO. Holt-Oram syndrome in China: A collective rewiev of 18 cases. A Heart J 1986;111:572-7.