Association between Apert Syndrome and Atrial Septal Defect

Pınar Dervişoğlu[1], Mustafa Kösecik[1], Serkan Kumbasar[2], Süleyman Salman[3], Bulat Aytek Şık[4], Baki Erdem[5], Bülent Tekin[6]

[1]Division of Pediatric Cardiology, Sakarya University School of Medicine, Sakarya, Turkey
[2]Clinic of Gynecology and Obstetrics, Sakarya Training and Research Hospital, Sakarya, Turkey
[3]Clinic of Gynecology and Obstetrics, Gaziosmanpaşa Taksim Training and Research Hospital, İstanbul, Turkey
[4]Department of Gynecology and Obstetrics, İstanbul Aydın University, İstanbul, Turkey
[5]Clinic of Gynecology and Obstetrics, Kanuni Sultan Süleyman Training and Research Hospital, İstanbul, Turkey
[6]Department of Gynecology and Obstetrics, İstanbul Yeni Yüzyıl University, İstanbul, Turkey

ABSTRACT

Apert syndrome, craniosynostosis, mid-face hypoplasia, symmetric syndactyly of the hands and feet, and varying degrees of mental retardation, and is characterized by congenital heart defects. Although the mode of inheritance is autosomal dominant, it creates new mutations in the majority of cases. In this study, an atrial septal defect cases of Apert syndrome, concomitant potential for congenital heart defects, in order to emphasize further investigation, we were willing to offer.

Keywords: Apert syndrome, newborn, atrial septal defect

INTRODUCTION

Apert syndrome is a rare congenital anomaly. It has an autosomal dominant inheritance. The fibroblast growth factor located on chromosome 10 develops as a result of the mutation of the receptor gene. There is no family history in most cases, and sporadic mutations have been shown. Progressive cutaneus and bone syndactyly, midfacial hypoplasia, and craniosynostosis are the most characteristic features (1). Various cardiovascular system anomalies may accompany Apert syndrome (2). The diagnosis is made through clinical, radiological, and genetic evaluations.

Here we present a rare case along with the review of the literature in order to emphasize on the detailed assessment in terms of the cardiovascular anomalies that may accompany.

CASE REPORT

A male baby who was born mature and weighing 4050 g to a 37-year-old mother at the 4th pregnancy through cesarean section delivery was admitted to our newborn service due to respiratory complaints. It was learned from the history that his father was 40 years old, there was no kinship between his mother and father, the other three siblings were healthy, and there were no syndromic individuals in the family. The patient's breathing rate was 60/min, cardiac apex beat was 140/min, and arterial blood pressure was 60/40 mmHg. Acrocephaly, high prominent forehead, depressed nasal bridge, beak nose, hypertelorism, proptosis in both eyes, downward deviation of the lateral edges of the eyes, bilateral low-set ears, bifid uvula, and complete syndactyly in both hands and feet were remarkable in the physical examination (Figure 1-4). His head circumference was 39 cm (97 p), height was 50 cm (50 p), and weight was 4050 g (97 p). The size of the front fontanel was 2×1 cm and that of the rear fontanel was 1×1 cm. Grade II/VI systolic murmur was detected in the pulmonary focus. Laboratory values were within the normal limits. In the atrial septum, a 5×6 mm secundum defect was detected through echocardiography. The urogenital system and abdominal examination findings were normal. There was no fusion between the cervical vertebrae. It was observed in the direct headgraphy that the cranial anterior–posterior diameter decreased, the nasal bridge was depressed, and there was flattening in the occipital bone. Dilatation in the third ventricle was observed in transfontanel ultrasonography. Pathology was not observed in the abdominal ultrasonography. Examination of the anterior segment and fundus showed normal results. Because he had a good general condition and the vital findings maintained stable, the patient, who was fed breast milk, was discharged on the fourth day of hospitalization after outpatient polyclinic follow-ups were planned. The patient is jointly followed by the departments of plastic and reconstructive surgery, pediatric neurology, and pediatric cardiology.

DISCUSSION

This syndrome, first described by Apert in 1906, is rarely found in craniosynostoses (1). Mutation in the FGF receptor gene was found in more than 90% of the patients. Mutations in this receptor have also been described in some other severe skeletal dysplasias. Most cases are sporadic, and the father's advanced age plays an important role in these patients. In a study conducted by Tolarova et al. (3) over a period of 10 years, the average age of fathers of 53 cases with Apert syndrome was found to be 34.1±6.2 years. In a study of 57 cases, it has been shown that all the mutations originate from the father in patients evaluated as having a new mutation (4). The father of our patient was also aged 40 years, and this was consistent with that reported in the literature, but we could not show the anticipated mutation because genetic examination could not be performed in our patient.

Address for Correspondence: Dr. Süleyman Salman, E-mail: ssalman8640@yahoo.com

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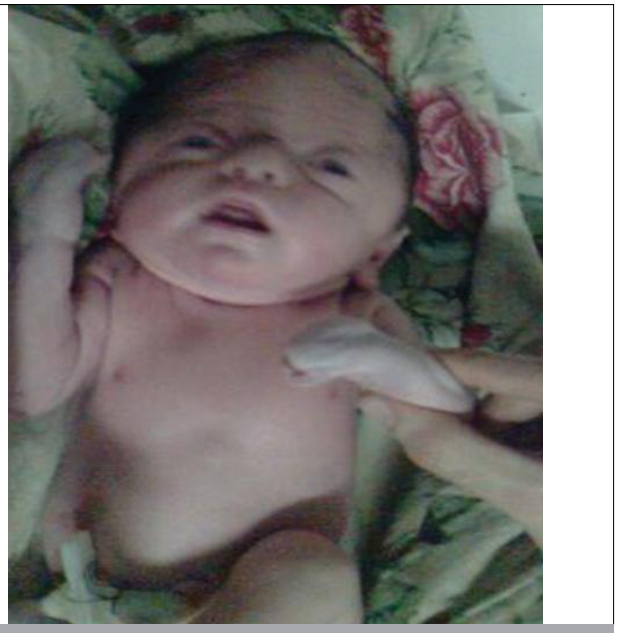

Figure 1. Acrosephaly

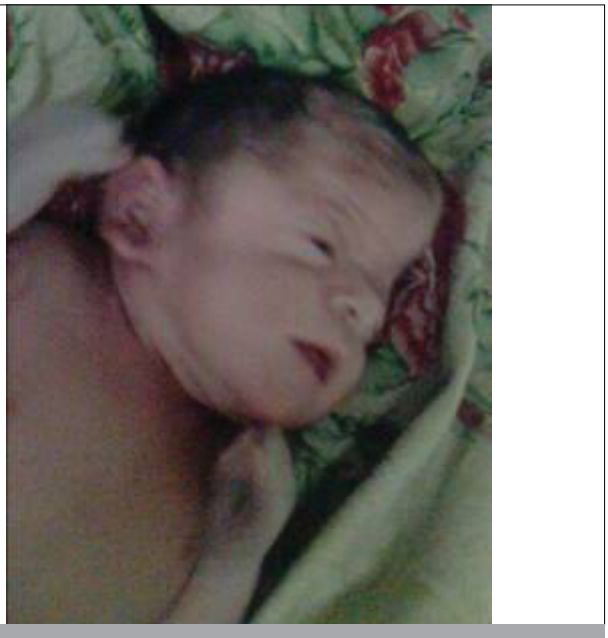

Figure 2. High prominent forehead, depressed nasal bridge

Figure 3. Complete syndactyly in both hands and feet

Figure 4. Complete syndactyly in both hands and feet

The most prominent clinical findings of this syndrome are cranial anomalies and symmetrical syndactyly in the hands and feet (5). Because of the early closure of the cranial sutures, the growth of the skull stays perpendicular to the axis of the closing suture. As a result, because of the growth of brain and the increasing pressure in the skull, complications such as visual anomalies, hydrocephalus, and mental retardation may develop (6). Hypertelorism and exophthalmos are the eye findings that are detected in all reported cases. In some cases, hippocampal anomalies and progressive hydrocephalus have been reported. Cerebellar, gyral, or cortical defects, lissencephaly, corpus callosum hypoplasia, or agenesis have been defined. A large face, smooth occiput, craniosynocytosis, depressed nasal bridge, low ears, high palate, and hypertelorism were found in our case, which was consistent with the cases reported in the literature (7, 8). Cervical fusion can be seen at a rate of 68%. The cervical vertebral graph of our patient was evaluated as normal (9). Symmetrical syndactyly occurs in Apert syndrome and is most commonly found in the second, third, and fourth interdigitals, and the first and fifth fingers are generally free. The formation of fusion in fingers other than the thumb is the second most common case (10). There was a total syndactyly in all extremities of our patient (Figure 1, 2).

Prenatal diagnosis is mainly made by ultrasonographic imaging of the craniosynostosis and cytokinesis. The gestational age in which the findings are seen varies, and beginning at the 16th week, it may last up to the 32nd week (11). There was no finding in supporting Apert syndrome in the prenatal follow-ups of our patient.

Cohen and Kreiborg (2) examined internal organ anomalies in 136 patients with Apert syndrome. They found the rate of cardiovascular anomalies consisting of the tetralogy of Fallot and ventricular septal defect to be 10%. Quinteno-Rivera et al. (11) found that ASD, VSD, and PDA were common defects in Apert syndrome and that early mortality was seen in patients in whom congenital heart diseases accompanied. The case with Apert syndrome accompanied by VSD reported by Demirpençe et al. (12) in 2009 also supported our case.

CONCLUSION

Congenital heart defects can be detected in Apert syndrome, though not as frequently as trisomies. Therefore, we would like to emphasize once again that the cases considered to have Apert syndrome should be evaluated in detail in terms of cardiac anomalies.

Informed Consent: Due to the retrospective design of the study, informed consent was not taken.

Peer-review: Externally peer-reviewed.

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