continuous with the distal portion of the left circumflex artery. We decided to continue her medical therapy and added a beta -blocker because we thought that this anomaly might directly induce myocardial ischemia. She had been asymptomatic at her last visit.

Discussion

Isolated SCA anomaly is one of the rarest coronary anomalies and constitutes 2-4% of all the coronary artery anomalies. SCA has been reported to be seen in 0.024% to 0.066% of the patients who undergo diagnostic coronary angiography (1-3). Our case is a very rare type of SCA anomaly and according to the Shirani et al. (4) classification, it can be categorized into the IA group which means that a solitary ostium in the left aortic sinus (I) is unassociated with an aberrant-coursing coronary artery (anatomic SCA) (A). This type has been reported in a few numbers in the literature (5, 6).

SCA anomalies are usually found incidentally during coronary angiography. Sudden death and myocardial infarction after exercise have been reported in patients whose left main or right coronary artery goes between main pulmonary artery and aorta (7). Shirani et al. (4) demonstrated that 15% of patients with SCA might have coronary ischemia due to the relation of coronary arteries with aorta or pulmonary artery. Thus, a coronary anomaly may itself cause myocardial ischemia without contribution of significant coronary stenosis.

Myocardial ischemia has been reported in 2 cases whose RCA originates from the left anterior descending or circumflex artery (8). In these cases, thinning of coronary arteries especially RCA was supposed to be responsible for cardiac ischemia. Herein, we presented the most benign type of SCA anomaly (2, 6) which was confirmed by MDCT. In our case, atherosclerosis, presence of which is an important prognostic factor in this type of SCA anomaly (2), was not present in the coronary arteries. We thought that ischemia caused by the SCA anomaly due to the thinning of RCA, was relieved by adding a beta- blocker.

Conclusion

This is the first case report on both conventional angiography and the MDCT images of a RCA arising from distal left circumflex artery.

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Char syndrome, a familial form of patent ductus arteriosus, with a new finding: hyperplasia of the 3rd finger

Ailesel patent duktus arteriyozus: Char sendromu ve yeni bir bulgusu; 3. parmak hipoplazisi

Introduction

Char syndrome is an autosomal dominant disorder characterized by patent ductus arteriosus (PDA), facial dysmorphism and abnormalities of the fifth finger of the hand (1). The prevalence of Char syndrome has not been determined but is believed to be quite low.

This report describes a Turkish family including five individuals affected by this disorder with an R236C mutation in the gene encoding the neural-crest-related transcription factor AP-2b. Affected family members had the typical facial, hand and foot anomalies and additionally presented case has rarely reported polythelia and non reported hypoplasia of the 3rd finger.

Case Report

A 15-day-old girl was referred because of a cardiac murmur. Consanguinity between the parents was denied. The respiratory and heart rates were 80/min and 160/min respectively, The patient had a flat midface, widely set eyes, mild ptosis, short philtrum and a triangular mouth; polythelia, foot and hand anomalies with clinodactyly were also noted (Fig. 1). Echocardiography revealed a large duct (6.5 mm) with unrestrictive ductal flow and predominantly left-to-right shunting, leading to left heart volume overload. The patient had an uneventful follow-up after surgical ligation and was discharged on the postnatal 45th day. The family history was suggestive for the presence of Char syndrome. His father, paternal uncle and a cousin were operated on for PDA. Similar phenotypic features and variable hand-foot anomalies were seen in them (Fig. 2). Additionally his paternal grandmother has typical facial dysmorphism, a small PDA, and polythelia. The pedigree is shown in Figure 1. Hypoplasia of the 3rd finger as a new finding in this syndrome was found in the proband and his father. Developmental, visual and hearing disorders were not detected in any members.

Genetic analysis of the *TFAP2B* coding exons and their flanking exons was performed as previously described (2). Analysis of the proband's genomic DNA revealed a coding region alteration in exon 4, a C-to-T transition at nucleotide 706 of the *TFAP2B* cDNA, which was present in heterozygosity. This sequence change predicted a substitu-

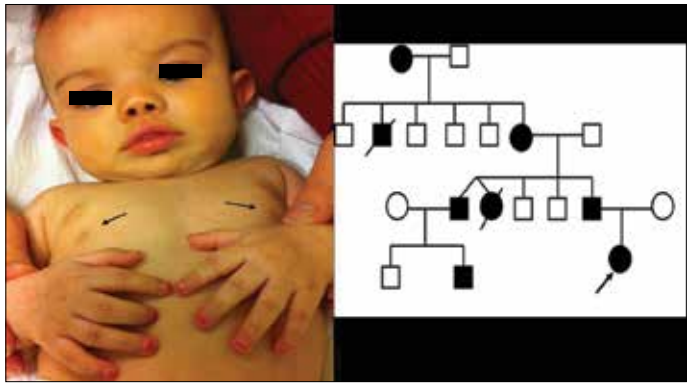

Figure 1. The propositus. Typical facial dysmorphism of Char syndrome are noted, including wide-set eyes, flat midface, flat nasal bridge and broad flat nasal tip, short philtrum resulting in a triangular mouth and thickened averted lips, as well as polythelia and clinodactyly of 5th finger and Pedigree of the family inheriting Char syndrome

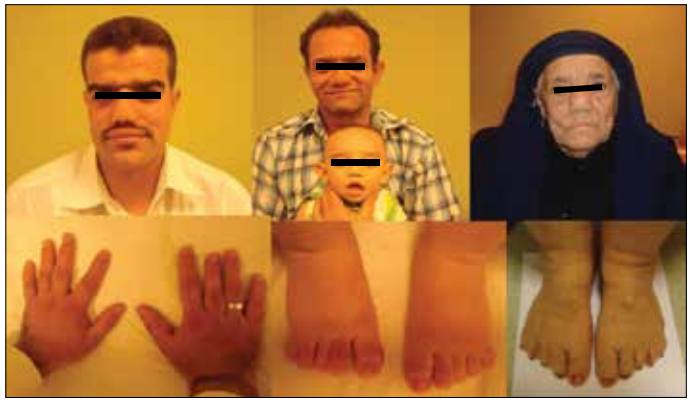

Figure 2. Affected family members. Upper panels: Frontal view of the family members showing typical facial dysmorphism of Char syndrome including a broad, high forehead, wide profile, down slanting palpebral fissures, hypertelorism, a short nose with a broad, flattened tip, short philtrum and prominent lips. Lower panel: hand and foot anomalies, hypoplasia of 3rd finger of hand and broad 2nd finger of foot and incomplete syndactyly of toes 4 and 5

tion of an arginine by a cysteine at position 236. This change had not been previously identified in >340 control chromosomes. The R236C was also identified in the proband's affected father. Genetic analysis was not performed on other family members.

Discussion

The incidence of isolated PDA in full-term infants is about 1 in 2,000 live births (3). However familial occurrence of PDA is quite rarely observed. Char syndrome has subsequently been reported by several investigators (4-7). No information is available concerning the likelihood of spontaneous closure of a PDA associated with Char syndrome, but it is likely to be rather low. Less common features associated with Char syndrome are polythelia, foot anomalies (interphalangeal joint fusion or clinodactyly, syndactyly), hearing abnormalities, visual impairment, development delay, parasomnia and other cardiac defects (4-6). Our proband had dysmorphic features, polythelia, foot and hand anomalies with clinodactyly and hypoplasia of the 3rd finger. Hypoplasia of the 3rd finger was found also in his father. This finding was not described previously in the literature to our best knowledge.

Char syndrome was mapped to a narrow region of chromosome 6p12-p21 (8). Further studies showed that *TFAP2B* mutations cause Char syndrome (6). Zhao et al. (2) reported six *TFAPB2* mutations that cause Char syndrome identified among 10 patients with the disorder and their families. An R236C mutation was detected in our propositus and his father. To the best of our knowledge, this is the first Turkish family in which Char syndrome has been detected.

The triad of Char syndrome has been variable in all families reported to date. The penetrance of Char syndrome has not been determined formally. One asymptomatic individual with a disease-causing *TFAP2B* mutation has been described (5). In our study, although the family members had all the characteristic features of this disorder, anomalies of hand/foot were variable. Moreover, the propositus and grandmother had polythelia. Zannoli et al. (7) first described polythelia as a feature of Char syndrome in 2000. We presented the second report of a family with Char syndrome exhibiting this feature. In the light of all these knowledge, we think that it can be described different features of this syndrome in the future.

Conclusion

Although rare, Char syndrome should be part of the differential diagnosis for patients with a family history of PDA, dysmorphic features, hand/foot anomalies and also polythelia. It is important to establish the diagnosis because the recurrence risk for the offspring was 50% in affected parents.

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