teminde hasar bulunduğunu düşündürmektedir. Ayrıca yavaş ventriküler taşikardi nedeniyle yapılmış olan radyofrekans ablasyon da ileti sistemindeki hasarı artırmış olabilir. Sağ ventrikül uyarılmasına ek olarak atriyoventriküler dissenkroninin de kalp yetersizliğini kötüleştirdiği düşünülmüştür. Hastanın cihazının CRT-D olarak değiştirilmesiyle sol ventrikül doluş basıncında düşüş ve kalp yetersizliği semptomlarında düzelme sağlanmıştır.

Sonuç

Miyokart enfarktüsü sonrası VT nedeniyle ICD implante edilmeden önce elektrofizyolojik çalışma ile ileti sistemindeki hasar bölgesinin belirlenmesi ve yüksek dereceli AV blok riskinin öngörülebilmesi mümkündür. Böyle bir hastada AV blok nedeniyle ICD'nin sağ ventrikül apikal uyarı yapması ve atriyoventriküler dissenkroni hastanın kalp yetersizliğini kötüleştirebilir. Sol ventrikül işlevlerinin kötüleşmesinin önüne geçilebilmesi amacıyla elektrofizyolojik çalışmada distal ileti sistemi kusuru gösterilen hastalarda CRT-D düşünülebilir.

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Renal coloboma syndrome associated with double- chambered right ventricle

Renal kolobom sendromu ile çift odacıklı sağ ventrikül birlikteliği

Introduction

Renal coloboma syndrome (RCS) (papillorenal syndrome) is an autosomal dominant entity characterized by hypodysplastic kidneys and optic nerve abnormalities ranging from optic pit to total optic disc coloboma (1, 2). The double-chambered right ventricle (DCRV) is a rare congenital heart abnormality caused by anomalous location of hypertrophic muscle bands creating an obstacle for the right ventricular ejection (3). In this paper we discuss clinical properties of a patient with papillorenal syndrome associated with congenital heart disease (CHD) including DCRV. To the best of our knowledge, this association has not been reported.

Case Report

A 21-year-old man without any ocular history presented to our clinic with loss of vision in his right eye for two months. His initial ophthalmologic examination revealed optic disc pit in both eyes and serous macular detachment in the right eye (Fig. 1). Slit lamp examination was unremarkable. His best-corrected visual acuity (BCVA) was 20/100 in the right and 20/20 in the left eye.

The patient had a history of surgery for atrial septal defect (ASD), pulmonary valve stenosis (PVS) and deformity of conus arteriosus when he was 3-year-old. Cardiac magnetic resonance imaging and echocardiography demonstrated DCRV, aneurysmal formation of the membranous septum, pulmonary and tricuspid regurgitation (Fig. 2). No treatment was given due to compensated cardiac disease.

He had also been suffering from vomiting, fatigue and spasms for two months. Blood work-up showed elevated serum creatinine (8.72

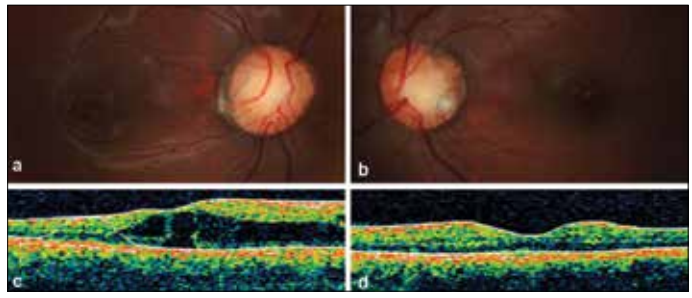

Figure 1. Dilated fundus exam revealing optic pit in both eyes (a, b). There is an optic pit-related serous macular detachment in the right eye (c)

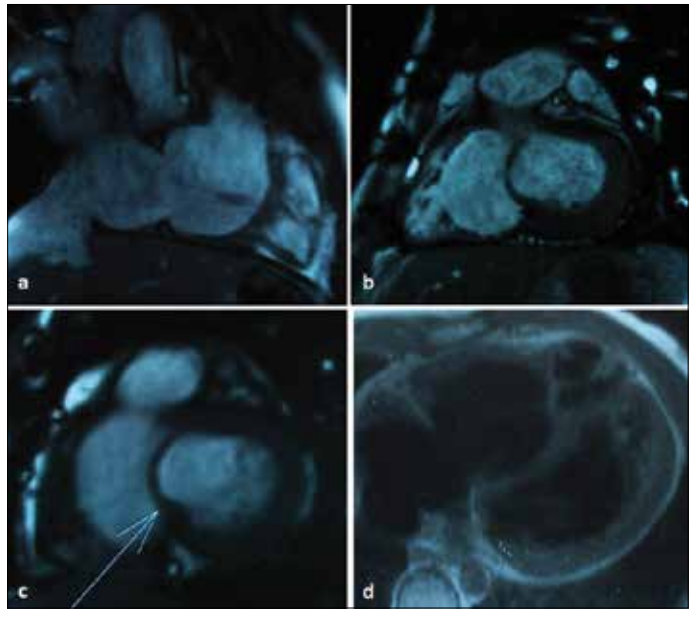

Figure 2. Cardiac MRI demonstrating anomalous muscle bundle dividing right ventricle into proximal and distal chambers (a, d), and membranous interventricular septal aneurysm (b, c)
MRI - magnetic resonance imaging

mg/dL), urea (200 mg/dL), phosphorus (5.84 mg/dL), parathyroid hormone (1196 pg/mL) and decreased levels of Fe^{+} (26 µg/dL), red blood cell ($3.83x10^{6}$/pL) and hemoglobin (11.6 g/dL). Urinalysis revealed proteinuria. Abdominal ultrasonography and computed tomography showed bilateral renal hypoplasia (Fig. 3) and a diagnosis of end-stage renal failure was made. Renal transplantation was performed, followed by six-month hemodialysis treatment. Two months after the successful renal transplantation, serous submacular fluid reduced and macular anatomy recovered but BCVA remained at the same level. The patient received no ophthalmologic treatment for the optic pit.

Discussion

Renal coloboma syndrome is inherited autosomal dominantly and PAX2 is the only gene known to be associated with RCS (4). PAX2 has critical roles in eye and renal embryogenesis.

Primitive eye is separated into two parts (later forms a fissure) by sonic hedgehog (SHH). During 3th week of gestation SHH upregulates PAX2 which is leading to fissure closure (5). Optic pit or coloboma can occur if this fissure fails to close between 3th and 7th weeks. Coloboma is frequently associated with other congenital abnormalities one of which is kidney. As with most organs, differentiation of the kidney involves epithelial mesenchymal interactions. During the 5th week PAX2 and WNT proteins promotes these interactions leading to formation of renal tubules (5).

In contrast to renal development, WNT proteins act as an inhibitor role in cardiac development. During the 4th to 7th weeks, the heart undergoes into a typical four-chambered structure with endocardial cushions, which have critical role in many cardiac deformities (5). DCRV is relatively rare as an isolated anomaly (approximately 0.5-2% of CHD) with no inheritance pattern (6). Patients with DCRV frequently have other congenital cardiac anomalies. The most common associated cardiac lesions include ventricular septal defect, PVS and subaortic stenosis (7). The embryological basis of DCRV has not been clearly described and DCRV coexisting ASD and PVS may be the result of conus arteriosus deformity. Development of aneurysmal formation of the membranous septum in our patient was probably caused by septal infarct secondary to abnormal septal hypertrophy rather than an embryological defect.

In our case there might be a defect concerning those molecular mediators that mentioned above in any stage of development (at most probably between 3-7 weeks) and PAX2 does not appear to be the only factor in this association. Therefore identifying PAX2 mutation is not necessary for definite diagnosis which is particularly based on clinical evaluation. Mutations in PAX2 have been identified in 50% of persons with RCS (1, 8) and it is estimated that about half of individuals with RCS do not have a known genetic basis (9). In our patient PAX2 gene were amplified from extracted DNA by using polymerase chain reaction primers. Mutation screening was performed by single strand conformation polymorphism. No pathologic allelic variant was observed. We consider that other gene mutations different from PAX2 may also cause this clinical entity.

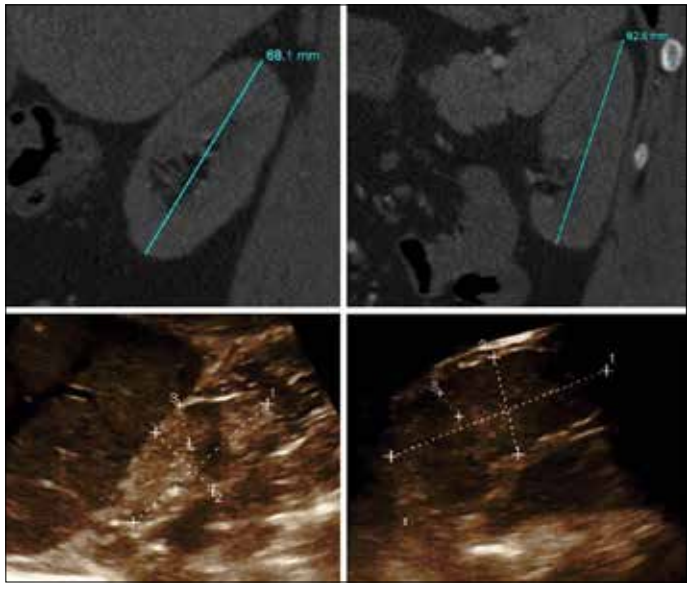

Figure 3. Abdominal ultrasonography and computed tomography views of bilateral renal hypoplasia

Conclusion

Renal coloboma syndrome is a multisystem disorder and a collaborative approach including specialists in ophthalmology, cardiology nephrology and medical genetics is necessary for a definite diagnosis. To the best of our knowledge, this is the first case report of RCS coexisting with DCRV.

Patient consent: We confirm that the patient has given written informed consent to the publication of this original article.

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