

## Case Reports

### Can Cystinosis Cause Coronary Artery Dilatation?

S. Olgar, K. Nisli, A. Dindar, R.E. Omeroglu, T. Ertugrul

Department of Pediatric Cardiology, Faculty of Medicine, Istanbul University, Istanbul 34200, Turkey

**Abstract.** In children, dilated coronary arteries are usually caused by Kawasaki's disease. We report four children with dilated coronary arteries and nephropathic cystinosis.

**Keywords:** Nephropathic cystinosis — Coronary artery dilatation — Cardiomyopathy

Cystinosis is an autosomal recessive disorder characterized by the excessive accumulation of cystine in several organs, including the kidneys, spleen, liver, lymph nodes, cornea, and thyroid gland. Dilated cardiomyopathy and aortic root aneurysm have been reported in patients with cystinosis. We report four patients with nephropathic cystinosis and dilated coronary arteries.

## Case Reports

### Case 1

A 13-year-old girl presented with chronic renal failure caused by cystinosis. Cystinosis was diagnosed from renal and bone marrow biopsies, which were taken in the nephrology department, and confirmed by ophthalmological examination. She was put on a routine hemodialysis program. Her parents were consanguineous, and she had two healthy sisters.

A cardiac murmur was found on routine cardiologic examination, and a chest x-ray revealed cardiomegaly. Echocardiographic examination showed bilateral coronary artery dilatation. Left coronary artery and right coronary artery diameters were found to be 4.9 and 4.8 mm (Fig. 1A and 1B), respectively. Biventricular hypertrophy with a low left ventricular ejection fraction (58%) was also present.

### Case 2

A 9-year-old girl, in whom cystinosis had been diagnosed at 1 year of age, was admitted. She had experienced severe growth retardation since the age of 5 months and was found to have hypertension,

Correspondence to: S. Olgar, email: [erfolgar@yahoo.com](mailto:erfolgar@yahoo.com)

hypothyroidism, and proteinuria. The diagnosis was confirmed from renal and bone marrow biopsies. The patient needed control hemodialysis twice weekly due to chronic renal insufficiency. She was the first child of a 37-year-old mother and 50-year-old father. She had two healthy sisters and a brother. The parents were first cousins; genetic analysis of the patient found no karyotypic abnormality.

Echocardiography revealed mild mitral valve insufficiency, slight tricuspid valve insufficiency, left ventricular dilatation, decreased left ventricular ejection fraction (44%), and increased right ventricular pressure (35 mmHg), estimated from tricuspid valve regurgitation. Both coronary arteries were dilated. The left coronary artery and right coronary artery diameters were 3.2 and 4.1 mm (Figs. 2A and 2B), respectively. A thallium myocardial perfusion scan was normal.

### Case 3

An 11-year-old girl with chronic renal failure was admitted to the nephrology unit suffering from growth retardation, irritability, and constipation. Nephropathic cystinosis was diagnosed by renal biopsy, and hemodialysis with medical treatment was started. Her parents were nonconsanguineous, and no karyotypic abnormality was found on genetic analysis.

Echocardiography revealed mild mitral and tricuspid valve insufficiency, left ventricular dilatation, with a left ventricular end diastolic diameter (LVEDd) of 3.82 mm, a decreased left ventricular ejection fraction (52%), and slightly increased right ventricular pressure (37 mmHg), estimated from tricuspid valve regurgitation. Her coronary arteries were found to be dilated. The left coronary artery and right coronary artery diameters were 4.7 and 3.8 mm (Figs. 3A and 3B), respectively. A thallium myocardial perfusion scan was normal.

### Case 4

A 9-year-old girl at the time of echocardiography was diagnosed with cystinosis on the basis of an ocular examination, and the diagnosis was confirmed by bone marrow and renal biopsies. She was followed up for approximately 6 years without dialysis, but after this period of time dialysis was initiated.

Echocardiography revealed slight mitral valve insufficiency, left ventricular dilatation (LVEDd of 3.69 mm), and a normal left ventricular ejection fraction (76%). Her left coronary artery and right coronary artery diameters were 3.6 and 3.2 mm (Figs. 4A and 4B), respectively.