A rare cause of dilated cardiomyopathy: Alström syndrome

**Dilate kardiyomiopatini nadir bir nedeni: Alström sendromu**

Alström syndrome (AS) is a recessively inherited genetic disorder characterized by: pigmentary retinal dystrophy, sensorineural hearing loss, obesity, non-insulin dependent diabetes mellitus. Other features reported in some but not all subjects include acanthosis nigricans, hypogonadism, short stature, hepatic, renal and cardiac failure (1) (Table 1). The gene mutated in AS patients has recently been identified as ALMS1 (2).

We present a 21-year-old man who was referred to the hospital for routine examination before applying to military services. He had five brothers and three sisters. According to the history, one of his brothers also had almost the same features but could not be examined because of social causes. There was no history of consanguinity and drug use in pregnancy. From birth, our patient suffered from progressive vision loss and nystagmus. On admission his blood pressure was 130/80mmHg, pulse rate 110/min. Weight was 55kg, height 144cm and body mass index (BMI) 26kg/m². He had thin hair and frontal hair loss. Eye examination showed nystagmus, bilateral posterior subcapsular cataract. Vision was at the level of perception but no projection in both eyes. His fundus examination showed pale discs and retinal pigmentary changes. He had acanthosis nigricans. Physical examination disclosed orthopnea, dyspnea, bilateral jugular venous distention and bilateral pretibial pitting edema. On auscultation, tachycardia, coarse lung sounds and bilateral rales below the scapulae were detected. He had gynecomastia, bilateral testicular atrophy and pretibial edema.

Laboratory analyses were as following: fasting blood glucose - 255mg/dl, insulin -36IU/mL, HbA1c - 11.7% (range, 4.8-8.0), creatinine - 1.5mg/dl (range, 0.6-1.2), serum glutamic-oxalacetic transaminase - 52 (range, 8-40), serum glutamic-pyruvic transaminase - 83U/L (range, 10-40), gamma-glutamyl transpherase -486 (range, 0-49U/L), alkaline phosphatase -127U/L (range, 38-94), thyroïd stimulating hormone - 1.56 mcIU/mL (range, 0.27-4.2), luteinizing hormone - 30.28miU/mL (range, 1.7-8.6), follicle stimulating hormone - 33.72miU/mL (range, 1.5-12.4), prolactin 10.44ng/mL (range, 4.04-15.2), testosterone - 1ng/mL (range, 2.4-9.5) 24-hour urinary albumin excretion- 250mg/24h. Electrocardiogram

Table 1. Clinical features and complications of Alström syndrome

<table>
<thead>
<tr>
<th>Presentation</th>
<th>(*) Literature, %</th>
<th>Patient in the present study</th>
</tr>
</thead>
<tbody>
<tr>
<td>Retinal degeneration</td>
<td>98</td>
<td>+</td>
</tr>
<tr>
<td>Sensorineural deafness</td>
<td>89</td>
<td>+</td>
</tr>
<tr>
<td>Diabetes</td>
<td>82</td>
<td>+</td>
</tr>
<tr>
<td>Obesity</td>
<td>98</td>
<td>-</td>
</tr>
<tr>
<td>Acanthosis nigricans</td>
<td>68</td>
<td>+</td>
</tr>
<tr>
<td>Hypergonadotropic hypogonadism</td>
<td>78</td>
<td>+</td>
</tr>
<tr>
<td>Hypothyroidism</td>
<td>17</td>
<td>-</td>
</tr>
<tr>
<td>Short stature</td>
<td>98</td>
<td>+</td>
</tr>
<tr>
<td>Hepatic dysfunction</td>
<td>92</td>
<td>+</td>
</tr>
<tr>
<td>Dilated cardiomyopathy</td>
<td>60</td>
<td>+</td>
</tr>
<tr>
<td>Renal dysfunction</td>
<td>49</td>
<td>+</td>
</tr>
</tbody>
</table>

* 1. kaynakta nayılanmıştır.
The use of renal stents in percutaneous treatment of very large coronary arteries

Çok geniş çaplı koroner arter darlıklarında renal stent kullanımı

In current, absence of the large size coronary stents poses significant challenges to the operator. Both early and late outcomes after drug-eluting stents (DES) implantation are limited only by the available caliber of the DES (3.5 mm in the United States) and the mechanical limitations placed on current DES overexpansion (4.75 mm for Cypher, 4.25 mm for Taxus). This is also validated for bare metal stents (BMS) (3.5 to 5.0mm) and so, there is a significant need for a large size stent that addresses this clinical challenge. We considered that the renal “vascular” stents, which have larger size, might be used in percutaneous treatment of coronary lesions in large-size vessels.

Case 1 was a 65-year-old male with a history of severe chronic obstructive pulmonary disease who was transferred to our institution for coronary angiography, which revealed the significant ostial left main coronary artery lesion and proximal left anterior descending artery stenosis (Fig. 1A). Because he represented a poor operative risk due to severe pulmonary disease, it was decided to perform percutaneous cardiac intervention. Therefore, the left main artery was stented with 5.5x12mm renal stent with postdilatation (Fig. 1B-C) and proximal left anterior descending artery was stented with 4.0x11mm bare-metal stent (Fig. 1D-E). Final angiographic appearance was normal with TIMI 3 flow (Fig. 1E). The patient was angina free at 3 months follow-up.

Case 2 was a 61 years old male patient with stable angina pectoris was referred to coronary angiography, which demonstrated significant middle left circumflex coronary artery stenosis (Fig. 2A-D). The other coronary arteries had mild atherosclerosis only. Percutaneous transluminal coronary angioplasty, with implantation of one 8.0x12mm renal stent, was successfully performed (Fig. 2E-F). The patient did well, without symptoms over the following three months.

Whereas larger stents induce more trauma to vessels and therefore more intimal hyperplasia, more edge dissections and more coronary ruptures; underexpanded stents increase both the risk of restenosis and the likelihood of stent thrombosis. Therefore, stent size must be carefully matched with reference vessel diameter, generally aiming for a 1:1 balloon to artery ratio. Since standard coronary angioplasty balloons or stents have generally not been available in diameters exceeding 5 mm, placing coronary stents may still remain challenging when vessels are extremely large. Consequently, angioplasty of larger arteries and grafts is commonly performed with undersized balloons or stents. The observational data support the use of adjunctive balloon postdilatation following stent deployment in the great majority of patients.