INTRODUCTION

• The prevalence and severity of cardiovascular disease in patients with mucopolysaccharidosis (MPS) is high and progressive, consisting in severe cardiac valve disease and ventricular hypertrophy; cardiac damage is a condition for survival in these patients.

• Coexistence of neurological disease leads to limitation of physical activity and clinical under-evaluation of their disease.

• Enzyme replacement therapy (ERT) in MPS may improve the organ impairment.

• ERT is available in Romania since 2005 for MPS type I and since 2011 for MPS type II and there are no reported results for Romanian patients for a consistent period of time.

AIM OF THE STUDY

The aim of the study was to characterize the cardiac disease and the results of enzyme replacement therapy in children with MPS type I and MPS type II.

PATIENTS AND METHODS

Patients

We evaluated 23 patients: 5 patients with MPS type I and 18 patients with MPS type II.

Methods

Enzymatic and molecular diagnosis was performed in all patients at diagnosis.

We assessed by Doppler echocardiography the function of valves, left ventricular chamber dimensions, septal and posterior ventricular wall thicknesses, systolic and diastolic function of the ventriles and pulmonary hypertension at every 6 months.

Interpretation was done according LV dimensions Z score and EAE/EAS recommendations for valve regurgitation and stenosis.

Treatment consisted in weekly administration of recombinant form of human alpha-L-iduronidase in dose of 0.58mg/kg/iv in MPS type I and recombinant form of human iduronate 2-sulfatase in dose of 0.5mg/kg/iv in MPS type II.

RESULTS

I. Echocardiographic alterations at diagnosis

<table>
<thead>
<tr>
<th>Type</th>
<th>No. of patients</th>
<th>Age at starting therapy (years)</th>
<th>Therapy duration (years)</th>
<th>Mitral valve thickening and moderate mitral regurgitation in a 6-years-old male patient with MPS type II</th>
</tr>
</thead>
<tbody>
<tr>
<td>MPS I</td>
<td>5</td>
<td>7.7 years</td>
<td>2 years</td>
<td>20% 80% 0% 0% → 100% 0% 0.05 p = 0.05</td>
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<tr>
<td>MPS II</td>
<td>18</td>
<td>5.8 years</td>
<td>4 years</td>
<td>100% 0% 0% 0% → 0% 100% 0% 0.05 p = 0.05</td>
</tr>
<tr>
<td>MPS I+ II</td>
<td>23</td>
<td>6.1 years</td>
<td>2 years</td>
<td>0% 100% 0% 0% → 0% 100% 0% 0.05 p = 0.05</td>
</tr>
</tbody>
</table>

II. Comparison of echocardiographic alterations at diagnosis with other studies

CONCLUSIONS

• Left valves lesions, ventricular hypertrophy, and pulmonary hypertension are the most common findings in children with mucopolysaccharidosis.

• ERT had little effect on cardiac valve disease.

• ERT progressively reduces myocardial GAG deposits.

• The age of initiation of therapy in patients with MPS type II do not represent a factor determining the effectiveness of this therapy.

LIMITS OF THE STUDY

• Small group of patients (MPS type I).

• Inhomogeneity of the groups, in terms of age at diagnosis and initiation of therapy.

• Difficulty in obtaining echocardiographic images due to associated skeletal and neurological involvement.