INTRODUCTION

Danon Disease (DD) is a rare, X-linked disorder of autophagy mediated by mutations in the Lysosomal-Associated Membrane Protein 2 (LAMP2) gene. LAMP-2 function is paramount to cellular autophagy, with its dysfunction resulting clinical manifestations of cardiomyopathy, skeletal myopathy, neurodegeneration, and retinopathy. [1,2] The true prevalence of DD is unknown, but DD is believed to underlie up to 5% of pediatric hypertrophic cardiomyopathy. [3]

METHODS

A retrospective review of pre-transplant echocardiographic data was performed in a global registry for DD from patients worldwide (United Kingdom, Australia and United States). Health information, laboratory results, and studies were supplied by the patients and/or their families. Prevalence comparisons of complications and outcomes were performed using Chi-square analysis. Comparisons of baseline echocardiographic parameters, demographics, and clinical outcomes were performed with unpaired t-test. For all statistical analyses, two-tailed p<0.05 was considered significant. Statistical analyses were performed with SPSS version 24 (Armonk, New York).

RESULTS

Records of 35 patients with DD were reviewed. 17 patients (49%) were male. Median age of cardiomyopathy diagnosis was younger in males compared to females (9 vs 24 yrs, p=0.005).

<table>
<thead>
<tr>
<th>Demographic</th>
<th>Male</th>
<th>Female</th>
<th>p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age at First Symptom, median years (IQR)</td>
<td>10 (7-15)</td>
<td>16 (13-20.5)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Age at Diagnosis, median years (IQR)</td>
<td>9 (6-12)</td>
<td>26 (31-52)</td>
<td>0.033</td>
</tr>
<tr>
<td>LAMP2-2-Cluster, No. (%)</td>
<td>5 (41.7%)</td>
<td>5 (41.7%)</td>
<td>1.000</td>
</tr>
<tr>
<td>Hypertrophic Cardiomyopathy (HCM), No. (%)</td>
<td>7 (53.8%)</td>
<td>10 (47.6%)</td>
<td>0.604</td>
</tr>
<tr>
<td>Dilated Cardiomyopathy (DCM), No. (%)</td>
<td>3 (23.1%)</td>
<td>11 (50.0%)</td>
<td>0.213</td>
</tr>
<tr>
<td>ECV, No. (%)</td>
<td>1 (7.7%)</td>
<td>1 (4.5%)</td>
<td>0.966</td>
</tr>
<tr>
<td>Stroke Volume, median years (IQR)</td>
<td>19 (16-21)</td>
<td>17 (13-20)</td>
<td>0.262</td>
</tr>
<tr>
<td>LV Ejection Fraction</td>
<td>59% (50-69%)</td>
<td>57% (35-88%)</td>
<td>0.596</td>
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</tbody>
</table>

Table 1. Demographics. Cardiac outcomes among males and females with DD.

CONCLUSIONS

Of 18 female patients, 8 (44.4%) patients had initial presentations with HCM and 2 (11%) patients with DCM. • Among females demonstrated profiles consistent with HCM: Mean EF, IVSd, and posterior wall thickness were 63%, 20.8mm, and 12.9mm respectively. • Among females with DCM on initial echocardiograms, Mean EF, IVSd, and posterior wall thickness were 20.5%, 16.1mm, and 8.4mm, respectively.

DISCLOSURES

This work was funded by Rocket Pharmaceuticals, EA is a shareholder in Rocket Pharmaceuticals. MB and MT are consultants for Rocket Pharmaceuticals.

REFERENCES


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