INTRODUCTION

- Danon Disease (DD) is a rare X-linked disorder due to mutations in the Lysosomal Associated Membrane Protein 2 (LAMP-2), which is important for cell autophagy [1,2].
- Penetration of disease-causing mutations in DD is nearly 100%. Given its inheritance pattern, DD has been assumed to be more severe in males, but there is still considerable uncertainty about the natural history of the disease. [3]
- DD is associated with severe cardiac, predominantly hypertrophic cardiomyopathy, and skeletal myopathies as well as cognitive impairment and hepatic abnormalities. [5]
- Prevalence of DD is estimated to be as high as 5% of all cases of pediatric cardiomyopathy. [4,5]

METHODS

- A retrospective registry for DD was created to understand the natural history of the disease.
- The diagnosis of DD is typically made by identification of LAMP-2 mutation or by clinical findings. As a result, delays in diagnosis or misdiagnoses are common.[3, 5]
- Comparisons of demographics, comorbidities and outcomes were performed using Chi-square analysis.
- For all statistical analyses, two-tailed p<0.05 was considered significant. Statistical analyses were performed with SPSS version 24 (Armonk, New York).

RESULTS

- A total of 35 patients (51.4% female) were included with the majority Caucasian (94.3%) and from the United States (80%).
- Irrespective of the organs involved, initial symptoms occurred in males and females at ages 8.5 (2-13) vs 15 (13.3-25.5) years, p = <0.001 and the diagnosis of DD was made earlier in males (age 9 (4-12) vs 24 (12-32) years, p = 0.005).
- Hypertrophic cardiomyopathy was more frequent in males compared to females (88.2% vs 44.4%, p = 0.006).
- Dilated cardiomyopathy (DCM) was the first manifestation of cardiac disease in 2 (11.1%) females.
- Heart transplantation (HTx) occurred in 4 (13.3%) males compared to females (88.2% vs 44.4%, p = 0.006).
- Skeletal myopathy was present in 82.4% of males and 56.6% of females with earlier onset in males compared to females (10 (3-12) vs 21.5 (10-46) years, p = 0.034).

DISCLOSURES

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- MB and MT are consultants for Rocket Pharmaceuticals.

REFERENCES