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Danon disease with typical early-onset cardiomyopathy in a male: Focus on a novel LAMP-2 mutation

Authors: Bui, Yen K.[1]; Renella, Pierangelo[2]; Martinez-Agosto, Julian A.[3]; Verity, Anthony[4]; Madikians, Andranik[5]; Alejos, Juan C.[2]

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Abstract:

Bui YK, Renella P, Martinez-Agosto JA, Verity A, Madikians A, Alejos JC. Danon disease with typical early-onset cardiomyopathy in a male: Focus on a novel LAMP-2 mutation.
Pediatr Transplantation 2008. © 2008 Blackwell Munksgaard Abstract:
We report a case of a 16-yr-old male with Danon disease caused by a novel mutation in the LAMP-2 gene. Mutations in the LAMP-2 gene result in the absence of LAMP-2 on immunohistochemical staining of muscle tissue, thus defining Danon disease, a rare X-linked myopathy. It is characterized clinically by HCM or left ventricular hypertrophy, a WPW pattern on ECG, variable degrees of muscular weakness (skeletal myopathy), mental retardation, and retinal changes. The patient presented with severe skeletal muscular weakness and respiratory failure. He also had a history of two OHTs, the first one for severe HCM and the second for allograft rejection. The patient's myopathy was initially presumed to be exclusively related to steroid-induced “critical care myopathy.” However, further evaluation with a thigh muscle biopsy revealed autophagic vacuoles with sarcolemnal features suggestive of a lysosomal storage disorder. DNA analysis ultimately identified a previously unreported hemizygous IVS6+3_+6delGAGT splice site deletion mutation in the LAMP-2 gene located within the 5′ splice site of intron 6, consistent with Danon disease.

Keywords: cardiomyopathy; myopathy; lysosomal storage disease; orthotopic heart transplantation

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Affiliations: 1: Department of Pediatrics, Moffitt Children's Hospital, UCSF, San Francisco, CA **2:** Division of Pediatric Cardiology Mattel Children's Hospital, UCLA Medical Center, Los Angeles, CA **3:** Department of Human Genetics and Division of Medical Genetics, David Geffen School of Medicine, UCLA, Los Angeles, CA **4:** Department of Pathology, David Geffen School of Medicine, UCLA, Los Angeles, CA **5:** Department of Critical Care Medicine, Mattel Children's Hospital, UCLA Medical Center, Los Angeles, CA, USA

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