Reviewer's report

Title: Nephropathic cystinosis associated with cardiomyopathy: A 27-year clinical follow-up

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Reviewer: Tej Mattoo

Level of interest: A paper of limited interest

Advice on publication: Other (see below)

The case report describes a 33-year-old male who was diagnosed with nephropathic cystinosis at the age of six years. The patient had multiple surgeries and received two renal transplants. He died from a ruptured pseudoaneurysm and the cardiac autopsy revealed the presence of cystine crystals in interstitial cardiac histocytes and one myocardial cell, and the cysteine level in the heart was very high. Authors conclude that the patient suffered from cysteine cardiomyopathy.

This is an interesting case report that may highlight yet another long-term complication of untreated cystinosis. However, the authors need to convince the reader on the possible association of restrictive cardiomyopathy with cystinosis. They also need to give the details of cardiac evaluation, if available, and discuss the possible reasons for normal cardiac function in previously reported cases (ref.9 and 10) of nephropathic cystinosis with documented cysteine crystals in the myocardium.

Discretionary revisions:
Page 2, last sentence in the abstract: Need to mention that the 1000-fold increase in cardiac muscle was in comparison to........?

Page 7, first paragraph, last sentence: Clarify the sentence "The muscle cysteine levels............

Compulsory revisions:
Need to add a picture of the cardiac biopsy showing cysteine crystals, as this would greatly enhance the quality of the manuscript.

Competing interests:
None declared.