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Vacant mitochondria in the myocardium of a patient with mitochondrial cardiomyopathy and diabetes mellitus

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DESCRIPTION

A 67-year-old woman was referred for heart failure, New York Heart Association class 2. Her medical history included: 25 years of diabetes, 7 years of hearing disorder and 3 years of mild cognitive dysfunction, with a family history of diabetes of her mother. She had been diagnosed with a point mutation of mitochondrial DNA 3243 (A→G) 3 years ago. Echocardiography revealed systolic and diastolic dysfunction with concentric left ventricular hypertrophy (figure 1A). Endomyocardial biopsy was performed, and light microscopy showed disarranged myocardial fibres with vacuolation (figure 1B). Electron microscopy revealed a marked increase of mitochondria that were vacant inside without a cristae structure, disarranged along with the myocardial fibres, and deformed into various sizes (figure 2A,B). The final diagnosis was mitochondrial cardiomyopathy along with maternally inherited diabetes and deafness. The identification of vacant mitochondria in this patient made us choose the energy-sparing therapy using a β -blocker with 5 mg a day of carvedilol and an ACE inhibitor with 5 mg a day of enalapril for the treatment of heart failure at first.

Considering the increasing number of patients with diabetes mellitus and about 1% of the diabetic population being thought to have point mutation of a mitochondrial gene in different races,¹ the importance in mitochondrial cardiomyopathy due to mitochondrial gene mutation has been more strongly recognised. This type of mitochondrial diabetes, which is called maternally inherited diabetes and deafness, is characterised by deafness in more than 60%,² cognitive impairment, and heart failure, though generally long after the onset of diabetes.

Learning points

- ▶ About 1% of the diabetic populations have point mutation of mitochondrial gene.
- ▶ Mitochondrial cardiomyopathy is one of differential diagnoses if patients had deafness, cognitive impairment and diabetes.
- ▶ Vacant mitochondria and other structural abnormalities were observed in mitochondrial cardiomyopathy.

Figure 1 (A) Representative images of echocardiography. Systolic dysfunction with 42% ejection fraction, hypertrophy of a 12-mm wall thickness, and a restrictive filling pattern of transmitral flow with an E/A of 2.54 and 142-ms deceleration time was observed. (B) H&E staining.

Figure 2 Vacant mitochondria observed by electron microscopy. M, mitochondria; arrowhead, a condensed Z band with the concentration of abnormal mitochondria; black arrow, the area replaced by glycogen. Magnification, (A) 3000 $\times$ and (B) 12 000 $\times$.

Competing interests None.

Patient consent Obtained.

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Please cite this article as follows (you will need to access the article online to obtain the date of publication).

Kato T, Hirose S, Nohara R. Vacant mitochondria in the myocardium of a patient with mitochondrial cardiomyopathy and diabetes mellitus. *BMJ Case Reports* 2012;10.1136/bcr-2012-006645, Published XXX

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