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【 LETTERS TO THE EDITOR 】

Authors' Reply to 'Heteroplasmy of the m.3243A>G Mutation May Influence Phenotypic Heterogeneity'

Key words: mitochondrial cardiomyopathy, family history, heteroplasmy, non-compaction

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The Authors Reply A physical examination, laboratory test and computed tomography scan revealed sensorineural hearing loss, cerebellum atrophy, exophoria and intellectual disability. There was no involvement of the eye, gastrointestinal tract, kidney, bone marrow or skin. Unfortunately, we did not perform a biopsy of any tissues other than the myocardium and so cannot compare the heteroplasmy rates.

The family history of the patient is listed in Figure. His brothers and son are, at present, healthy. His mother suffered from diabetes mellitus, and her cardiac function, serum lactate and pyruvate levels are normal. However, his family declined any further examinations, including a genetic analysis and cardiac scintigraphy. Therefore, the inheritance of the m.3243A.G mutation remains unclear.

The cardiac images of echocardiography and magnetic resonance imaging (MRI) findings were not consistent with the definition of non-compaction in Jenii et al. (1, 2), but were similar to those of hypertrophy cardiomyopathy. The phenotype of mitochondrial cardiomyopathy, in general, demonstrates a wide range of morphologic patterns.

The preserved uptake of ^{99m}Tc-MIBI is an important finding in the present case. As Fisnterer et al. pointed out, it is reasonable that diffuse fibrosis results in a decreased uptake of ^{99m}Tc-MIBI. However, the total uptake of ^{99m}Tc-MIBI and ^{123}I-BMIPP is determined by the balance between the number of viable myocytes, blood flow and metabolic changes. In this case, the number of viable myocytes decreased, but the blood flow might have been maintained or even increased. The inhomogeneous heteroplasmy rate in each myocyte may also affect the image patterns. Diverse uptake patterns of ^{99m}Tc-MIBI and ^{123}I-BMIPP can be observed in individual cases of mitochondrial cardiomyopathy.

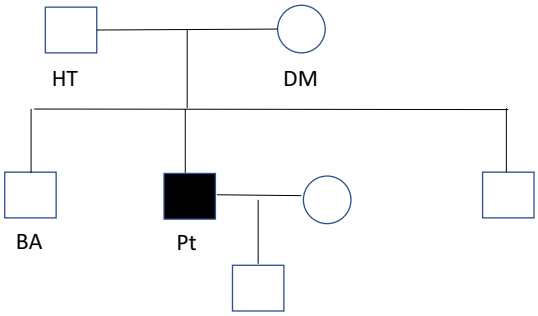

Figure. **Family tree. HT: hypertension, DM: diabetes mellitus, BA: bronchial asthma**

We are now following up with the patient at an outpatient clinic. Further studies of cardiac scintigraphy are important to reveal the progression process of mitochondrial cardiomyopathy.

The authors state that they have no Conflict of Interest (COI).

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