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LETTER TO THE EDITOR(AUTHOR'S REPLY)

Authors' reply to "m.3243A>G carriers develop syndromic or non-syndromic multisystem phenotypes over time"

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To the Editor

We wish to thank Josef Finsterer for their letter commenting on our report. Our case emphasized the changes in renal pathology over 20 years that led to the development of chronic kidney disease. As renal diseases associated with mitochondrial cytopathy, we highlighted focal segmental glomerulosclerosis and tubulointerstitial nephropathy, which are the major findings in renal biopsy tissue of mitochondrial disorders. We agree that the clinical renal involvement in mitochondrial disorders is diverse, as reported elsewhere [1].

Although some of the examinations for multisystem involvement pointed out by Finsterer have already been mentioned in our report, we added other information on our patient. No abnormalities were found on his head MRI and ocular investigations. As a limitation of our report, his mother had already died and we could not contact his younger brother, making further genetic examination difficult. However, the diagnosis of maternally inherited diabetes and deafness (MIDD) in our patient was considerably definitive from the presence of diabetes and deafness, and a family history of these conditions in maternal relatives. Investigation of the muscle homogenate was impossible. We hope that our report will help physicians start appropriate management of MIDD as early as possible.

Declarations

Conflict of interest The authors declare that they have no conflicts of interest.

Informed consent Informed consent was obtained from the patient described in the present case.

Reference

1. Finsterer J, Scorza FA. Renal manifestations of primary mitochondrial disorders. Biomed Rep. 2017;6:487–94.

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