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Letter to the Editor

Assessment of cardiac disease in MELAS requires comprehensive, prospective work-up☆

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With interest we read the article by Brambilla et at. about a follow-up investigation over 8.5 y of cardiac involvement in 21 patients <21 years with MELAS syndrome due to the variant m.3243A > G [1]. We have the following comments and concerns.

The main shortcoming of the study is that heteroplasmy rates of the m.32343A > G variant in various tissues, such as hair follicles, skin fibroblasts, muscle, buccal mucosa, lymphocytes, urinary epithelial cells, were not provided. Heteroplasmy rates may be a main denominator of the phenotype of the mutation and thus the degree of cardiac involvement [2]. Only in the limitations it is mentioned that heteroplasmy rates were determined in different tissues but were thus not comparable.

A further shortcoming is that left ventricular hypertrabeculation (LVHT, noncompaction) was not addressed. Since mitochondrial disorders are the disease in which LVHT can be most frequently found [3], it is crucial to look for this myocardial abnormality, which has prognostic implications.

Another shortcoming is that cardiac MRI was not applied [1]. Cardiac MRI may not only detect LVHT in case echocardiography fails to detect it, but may also show late gadolinium enhancement, an indicator of myocardial fibrosis [4].

Since MELAS frequently manifests with epilepsy [5], and since seizures may be complicated by Takotsubo cardiomyopathy (TTS), we should be informed how many of the 21 included patients had epilepsy and how many developed TTS during any of the seizures.

Overall, this interesting study has a number of shortcomings which may impede a reliable interpretation of the results.

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