



Letter to the Editor

Comment on: Assessment of cardiac disease in MELAS requires comprehensive, prospective work-up



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Dear Dr. Finsterer,

We appreciate your interest in our work addressing cardiac involvement in MELAS syndrome [1]. However, while we find the aspects emphasised both relevant and worthy of further investigation, we disagree that these may significantly affect the reliability of our results. To date, a specific correlation between heteroplasmy rate (HR) and degree of cardiovascular involvement has not been demonstrated and a number of additional factors are likely involved in phenotypic expression [2]. Standard protocols for heteroplasmy detection have not been established, possibly limited by extreme intra-patient variability [3] or HR fluctuation over time [4]. Testing HR in multiple tissues remains undoubtedly useful for diagnostic purpose, especially in case of mild or late phenotype.

As reviewed by Arbustini et al. [5], the term LV hypertrabeculation / non-compaction describes a morphological trait rather than the functional or prognostic profile of a cardiomyopathy. Final outcome is driven by the type/severity of the underlying cardiomyopathy and on additional extra-cardiac involvement. In our cohort, only 2 of the 21 patients showed evidence of mild left ventricular hypertrabeculation, failing to meet the proposed diagnostic criteria for LVNC.

We acknowledge lack of systematic cardiac MRI data in our study. Of note, none of the two patients who underwent cardiac MRI showed areas of late gadolinium enhancement and it is unlikely that this test would have affected management in any of the study patients.

Finally, with regard to neurologic involvement, epilepsy was recorded in 3 of the 21 patients; none developed Takotsubo cardiomyopathy during the study.

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Authors contribution statement

All author have equally contributed in the design of the study, data collection, data analysis and interpretation, drafting the article and critical revision. All authors have approved the final version of the manuscript and its submission.

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¹ This author takes responsibility for all aspects of the reliability and freedom from bias of the data presented and their discussed interpretation.