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Is reduced global longitudinal strain in m.3243A>G carriers truly attributable to the mutation?

Josef Finsterer, MD, PhD[1]*; Sinda Zarrouk-Mahjoub, PhD[2]

[1]Hospital Rudolfstiftung, PO Box 20, 1180 Vienna, Austria

[2]University of Tunis El Manar and Genomics Platform, Pasteur Institute of Tunis, Tunisia

***Corresponding Author(s): Josef Finsterer**

Hospital Rudolfstiftung, PO Box 20, 1180 Vienna, Austria

Tel: +43-1-71165-72085; Fax: +43-1-4781711

Email: fifigs1@yahoo.de

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Letter to the Editor

In a recent article, Koene et al. reported about a study of 30 carriers of the m.3243A>G mutation who were retrospectively evaluated for their findings on 2D speckle tracking echocardiography [1]. Global Longitudinal Strain (GLS) was reduced in 56-70% of the patients. We have the following comments and concerns.

Table 1 of the article shows that 10 out of 30 patients had cardiac involvement other than a reduced GLS [1]. Which type of cardiac disease did the authors detect? A frequent finding in patients with mitochondrial disorders is Left Ventricular Hypertrabeculation/noncompaction (LVHT) [2]. How often did the authors detect it in their 30 patients? Was extension of LVHT correlated with the leucocyte or urinary epithelial cell heteroplasmy rate? How many patients had hypertrophic and how many dilative cardiomyopathy? Were ventricular arrhythmias recorded on long-term ECG? Patients carrying the m.3243A>G variant may develop myocardial fibrosis. How many had late gadolinium enhancement on cardiac MRI or fibrosis on endomyocardial biopsy?

Heteroplasmy in the urinary epithelial cells ranged from 7 to 96% [1]. A patient with a heteroplasmy rate of 7% is unlikely to manifest clinically. Which were the clinical manifestations in this patient? Did he present with reduced GLS or another cardiac manifestation? How do the authors explain the discrepancy between table 1 and table 3. In table 3 no patient with a heteroplasmy rate of 7% is listed. Why does table 3 only contain

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27 and not 30 patients? Were the 3 excluded patients those in whom GLS could not be determined? Was the heteroplasmy rate among the 10 patients with cardiac involvement higher than that in carriers without cardiac disease? There are altogether 8 patients with a heteroplasmy rate <50% [1]. Did these patients manifest clinically or were these 8 patients those with reduced GLS. Did heteroplasmy rates only correlate with GLS at baseline or also at follow-up?

Concerning the follow-up data we should be informed how many patients in addition to the ten patients developed cardiac involvement, particularly heart failure. Did patients with reduced GLS receive cardiac treatment? Which type of medication was applied? How did the authors exclude coronary atherosclerosis as the cause of reduced GLS?

The number of patients with reduced GLS differed substantially depending on the reference limits applied (15 vs. 19 patients)? How to explain the variable reference limits? Why did the authors not use their own reference limits based on their own control group?

Overall, this interesting study could profit from providing more details about the cardiac findings, from explanation why low heteroplasmy rates were associated with cardiac disease, from providing data how coronary artery disease was excluded as a cause of reduced GLS, and from application of their own reference limits to assess GLS.

Author contributions:

JF: design, literature search, discussion, first draft, SZ-M: literature search, discussion, critical comments

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