# Leber's hereditary optic neuropathy: mind the heart!

Thomas Morris Hey, MD, Ph.D., Jens Mogensen, MD, D.M.sc, Ph.D.

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# Leber's hereditary optic neuropathy: mind the heart!

Short title: LHON – mind the heart!

MD, Ph.D. Thomas Morris Hey[1,2*], Jens Mogensen, MD, D.M.sc, Ph.D.[3,4]

*: Corresponding author: Email: thomas.morris.hey@rsyd.dk; Address: Lillebaelt Hospital – Vejle, Kabbeltoft 25, 7100 Vejle, Denmark, tel. +45 21128022

1: The department of cardiology, Lillebaelt Hospital- Vejle , Denmark

2: The department of cardiology, Odense University Hospital, Denmark

3: The department of cardiology, Aalborg University Hospital, Denmark

4: The health faculty, The University of Southern Denmark, Denmark

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**Letter to the Editor**

We appreciate the interest by Finsterer and Mehri in our recent report on Leber's hereditary optic neuropathy (LHON).[1]

In mitochondrial disease the heteroplasmy rate (HR) express the ratio of abnormal/total number of mitochondria in a specific tissue. The ratio is often tissue specific which imply that HR of myocardial tissue may differ significantly from HR's of other tissues. Since the severity of the disease expression in mitochondrial disease is associated with the tissue specific HR it is of little clinical significance to establish HR. Furthermore, it may be harmful to the patient to obtain myocardial tissue due to the risk of myocardial perforation during the sample procedure. In our study the genetic investigations included only part of mitochondrial-DNA harboring recognised pathogenic variants. However, as pointed out by Finsterer and Mehri information about copy number variants and haplotype may be of scientific interest to better understand the pathophysiology in LHON.[1]

As described in our report the index-patient had involvement of multiple organs including demyelination of the posterior portion of the optical nerves consistent with LHON. He had an otherwise normal neurological investigation and no signs or symptoms of Hardings disease.[2, 3]

Recently, Finsterer and Mehri suggested that left ventricular hypertrabeculation (LVHT) is a specific disease expression associated with LHON based on the clinical findings in two patients carrying a m.3460G>A mutation.[1] However, it is well recognised that LVHT may be part of the normal left ventricular anatomy as well as part of the disease expression in both dilated- and

hypertrophic- cardiomyopathy (HCM).[4] Therefore, it is less likely that LVHT represent a specific disease entity with a well-defined prognosis.

Finsterer and Mehri suggested implantation of loop recorders in LHON patients with LVHT due to sudden cardiac death (SCD) in one patient in their report.[1] We appreciate these considerations although it is our opinion that management of patients should be individualized and based on their clinical disease expression, which was benign in the patients in our report. There was no family history of SCD, they did not experience syncopes and had repeated Holter recordings without arrhythmias. Loop recorder implantation was therefore not indicated.

Previous studies of LHON patients have also reported cardiac manifestations similar to HCM caused by pathogenic sarcomeric variants, which suggests that routine cardiac investigations should be offered to all LHON patients. Furthermore, it seems well indicated that at least *ND1* should be part of the genetic screening of HCM patients.[1, 2]

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