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Letter to Editoe

Affection of the cochlear stria vascularis in MELAS suggests vascular involvement of the ear

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Abstract:

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COMMENTARY

With interest we read the article by Handzel *et al.,* 2020 about post-mortem investigations of the cochlea and the vestibulum by means of light microscopy in a patient with MELAS due to the variant m.3243A>G who died from respiratory failure at age 42y (Handzel, O. *et al.,* 2020). The authors concluded that hearing loss is a result of dysfunction of the stria vascularis and not due to loss of hair cells or neurons and that thus these patients may benefit from cochlear implants (Handzel, O. *et al.,* 2020). We have the following comments and concerns.

The wording is misleading. "Hearing loos" means deafness. According to the case description the patient had "hearing loss" at age 25y (Handzel, O. *et al.,* 2020). In the next sentence the authors state that "hearing loss" was "progressive" (Handzel, O. *et al.,* 2020). How can the inability to hear anything be progresssive? The term "hearing loss" should be replaced by "hypoacusis" or "impaired hearing". It is also unlikely that deafness at age 25y occurred suddenly. We should know if the index patient underwent cochlear implantation given the description of „hearing loss" since age 25y.

Death from respiratory failure at age 42y requires explanation. We should know if this was due to affection of the respiratory muscles and thus "muscular respiratory failure" or if there was cardiac involvement with heart failure secondarily leading to respiratory insufficiency. Furthermore, primary involvement of the lungs has been reported in mitochondrial disorders (Finsterer, J., & Zarrouk-Mahjoub, S. 2016). It is also conceivable that respiratory failure was unrelated to the underlying metabolic defect.

There is broad discussion about the genetics of MELAS but except for the m.3243A>G variant in the index case no further genetic data were provided. Missing particularly is the heteroplasmy rate. It would have been worthwhile to know if heteroplasmy rate of mtDNA copy number varied between the various structures such as saccules, spiral ganglions, stria vascularis, or ampulla.

Concerning the morphological affection of the vestibulum we should know if there was clinical involvement of the vestibulum. It should be mentioned if there was gait disturbance, vertigo, nausea, nystagmus, or vomiting. Primary involvement of the vestibulum has been previously reported in MELAS (Hougaard, D. D. *et al.,* 2019).

We do not agree with the statement that "this is an analysis of the oldest patient with MELAS syndrome to date". We recently investigated a 56yo male with MELAS. Niedermayr *et al.,*. recently

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reported a 68yo patient with MELAS (Niedermayr, K. *et al.,* 2018).

We do not agree with the statement that „hearing loss can be the first and initially the only manifestation of MELAS syndrome". Hearing impairment is usually not present at birth or in early childhood but develops later. However, what is present at birth or in early childhood is short stature. Thus, we should know the hight of the index patient and the time course of involvement of the other organs in the described patient would be interesting to know.

Since there is involvement of the arteries in mitochondrial disordes (Finsterer, J., & Mahjoub, S. Z. 2012), we should know if the arteries, arterioles, and capillaries of the stria vascularis showed any changes indicative of vascular involvement.

It would have been worthwhile to see ultrastructural findings of the cochlea and the vestibulum, particularly if morphology of mitochondria was compromised and if there abnormal storage of lipids or glycogen.

Overall, this interesting study has a number of shortcomings which should be adressed before drawing conclusions. Genetic work-up should be more profound since morphological abnormalities may be absent or non-specific. The stria vascularis should be screened for vascular abnormalities.

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