Comments on: Outer retinal tubulation and inner retinal pseudocysts in a patient with maternally inherited diabetes and deafness evaluated with optical coherence tomography angiogram

Dear Editor:

With interest we read the article by Tripathy *et al*. about a 47-year-old female with hypoacusis, diabetes, and ischemic heart disease attributed to the mtDNA variant m.3243A>G in *MT-TL1*.[1] Ophthalmologic investigations revealed areas of foveal atrophy and hypoautofluorescence, depression of inner retinal surface, pseudocysts, thinning of the outer retina, outer retinal tabulation, loss of external limiting membrane and thinning of retinal pigment epithelium, and choriocapillaris.[1] Retinal and choreal abnormalities were interpreted as subclinical involvement in the mitochondrial disorder (MID).[1] We witnessed a number of shortcomings in the study which have been stated as follows:

Firstly, no heteroplasmy rates in any affected or non-affected tissue were provided. We should know heteroplasmy rates since they can be helpful to predict the disease trajectory and are required for genetic counselling.[2]

Secondly, the mother of the index patient was not genetically investigated. Since 75% of the mtDNA mutations are maternally inherited,[3] it is conceivable that the culprit variant was transmitted via the maternal line. Phenotypic expression in the mother at variance from that in her daughter is not unusual since the phenotypic expression of an mtDNA variant can be highly heterogeneous between family members.[4] We should be aware if the mother of the index patient also manifested with hypoacusis and diabetes in addition to the cataract.

Thirdly, neither the index patient nor her mother were prospectively investigated for multisystem disease, frequently found in m.3243A>G carriers.

In addition, we should be informed about the cause of ischemic heart disease,[1] that is, whether it was due to coronary heart disease from diabetes or a primary manifestation of the underlying genetic defect, as has been recently demonstrated.[5]

Overall, this interesting case report has a number of shortcomings which need to be addressed before the conclusions drawn can be adopted.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Josef Finsterer

Krankenanstalt Rudolfstiftung, Messerli Institute, Vienna, Wien, Austria

Correspondence to: Prof. Josef Finsterer, Postfach 20, 1180 Vienna, Austria, Europe.
E-mail: fifigs1@yahoo.de

References

1. Tripathy K, Sarma B, Mazumdar S. Outer retinal tubulation and inner retinal pseudocysts in a patient with maternally inherited diabetes and deafness evaluated with optical coherence tomography angiogram. Indian J Ophthalmol 2020;68:250-3.
2. McCann BJ, Tuppen HA, Küsters B, Lammens M, Smeitink JA, Taylor RW, *et al*. A novel mitochondrial DNA m. 7507A>G mutation is only pathogenic at high levels of heteroplasmy. Neuromuscul Disord 2015;25:262-7.
3. Poulton J, Finsterer J, Yu-Wai-Man P. Genetic counselling for maternally inherited mitochondrial disorders. Mol Diagn Ther 2017;21:419-29.
4. Liu G, Shen X, Sun Y, Lv Q, Li Y, Du A. Heteroplasmy and phenotype spectrum of the mitochondrial tRNA (Leu (UUR)) gene m. 3243A>G mutation in seven Han Chinese families. J Neurol Sci 2019;408:116562. doi: 10.1016/j.jns. 2019.116562.
5. Zhang Z, Liu M, He J, Zhang X, Chen Y, Li H. Maternally inherited coronary heart disease is associated with a novel mitochondrial tRNA mutation. BMC Cardiovasc Disord 2019;19:293.

Access this article online	
Quick Response Code:	Website: www.ijo.in
	DOI: 10.4103/ijo.IJO_2358_19

Cite this article as: Finsterer J. Comments on: Outer retinal tubulation and inner retinal pseudocysts in a patient with maternally inherited diabetes and deafness evaluated with optical coherence tomography angiogram. Indian J Ophthalmol 2020;68:1509-10.