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Ophthalmologic Involvement in MNGIE may also affect the Central Visual Pathways

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LETTER TO THE EDITOR

We read with interest the article by Wang, *et al*. about a 33-year-old male with myo-neuro-gastrointestinal encephalopathy (MNGIE) due to the homozygous variant c.1159+1G>A in *TYMP1*, inherited from consanguineous parents [Wang, H. *et al*., 2023]. The patient presented clinically with gastrointestinal dysmotility, polyneuropathy, deafness, lactic acidosis, hepatic steatosis, diabetes, ptosis, ophthalmoparesis, leukoencephalopathy, severe visual impairment, dyschromatopsia, and electronegative electroretinograms [Wang, H. *et al*., 2023]. The study is impressive, but some points require further discussion.

Although optic atrophy has rarely been reported in MNGIE [Felhi, R. *et al*., 2019], we should know whether the index patient had optic atrophy. Orbital MRI is the most suitable tool for assessing optic nerve thickness. Optic atrophy could contribute to the index patient's visual impairment.

Another change that could have contributed to visual impairment is leukoencephalopathy. The central visual pathways may have been affected by leukoencephalopathy and therefore may have been partly responsible for the index patient's poor vision. Presentation of the cerebral MRI would be helpful to assess the degree of leukoencephalopathy and the degree of its contribution to visual impairment.

The third point is that there is no discussion about the genetic heterogeneity of MNGIE. MNGIE may be due not only to variants in *TYMP1*, but also to variants in *POLG1*, *RRM2B*, *LIG3*, *RRM1*, *MTTV1*, and *MT-RNR1* genes (MNGIE-like phenotypes) [Redha, N. *et al*., 2024]. It should be discussed whether the visual impairment depends on the mutated gene or even the specific variant.

The fourth point is that the treatment the index patient received for MNGIE was not reported [Wang, H. *et al*., 2023]. Knowing the type of treatments carried out is crucial, especially to know whether ophthalmologic manifestations of the disease benefited from the therapy applied. Known types of treatment include symptomatic treatment and causative therapies, Symptomatic therapy includes a change in diet, drugs to stimulate bowel motility, analgesics, antiemetics, antibiotics for bacterial overgrowth in the intestine as a complication of dysmotility, blockade of the splanchnic nerve or blockade of the coeliac plexus with bupivacaine in case of gastric pain, metoclopramide, domperidone, amitriptyline or bisacodyl for postprandial emesis and nausea, prokinetics for gastroparesis and parenteral nutrition [Bax, B. E, 2020]. Causative treatments include hemodialysis, allogenic liver transplantation, intestinal transplantation, allogeneic hematopoietic stem cell transplantation, and carrier erythrocyte entrapped thymidine phosphorylase [Bax, B. E, 2020].

In summary, the interesting study has limitations that put the results and their interpretation into perspective. Removing these limitations could strengthen and support the study's message. Reports of MNGIE patients should include the symptomatic or causative treatments used and whether these were beneficial, particularly for ophthalmologic involvement.

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