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Diagnosing Genetic Eye Disease Requires Genetic Testing

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LETTER TO THE EDITOR

We read with interest the article by Pawar, *et al*., about a study on five patients with rare eye diseases [Pawar, N. *et al*., 2022]. The study is excellent but has limitations.

The major limitation of the study is that the diagnosis Kearns-Sayre syndrome (KSS) in patient-1 and patient-2 was not genetically, confirmed [Pawar, N. *et al*., 2022]. Although the phenotype in both patients was suggestive of KSS, this diagnosis needs to be confirmed by genetic testing. In the vast majority of cases, KSS is due to single mtDNA deletions, but mtDNA point mutations have also been reported to cause KSS. mtDNA point mutations that are phenotypically manifesting as KSS include the variants m.3249G>A in the tRNA(Leu) gene, m.3255G>A in the tRNA(Leu) gene, or m.3243A>G in the tRNA(Leu) gene [Finsterer, J .*et al*., 2016]. Knowing the genetic background in KSS is critical not only for assessing the disease progression and outcome, including retinopathy, but also for genetic counselling. mtDNA point mutations are maternally inherited in 75% of cases, but mtDNA deletions are maternally inherited in only 4% of cases [Poulton, J. *et al*., 2017].

A second limitation is that no explanation was provided as to why repetitive nerve stimulation in patient-2 was indicative of post-synaptic myasthenia. Did the patient have KSS plus myasthenia or was the result false positive? Knowing the correct diagnosis is crucial as the treatment for the two differs significantly. We disagree that myasthenia was ruled out by a negative test for acetyl-choline receptor antibodies. The negativity of these antibodies does not rule out myasthenia, since it can also be due to antibodies against MUSK, LRP4, or titin. There are also myasthenia cases without antibodies.

A third limitation is that Hallervorden Spatz disease (HSD) in patient-4 was not genetically confirmed. Since mutations in the *PANK2* gene are responsible for HSD in most cases [Sriram, N. *et al*., 2023], it is critical that patient-4 undergoes genetic testing.

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Ethical compliance statement: The authors confirm that the approval of an institutional review board or patient consent was not required for this work. We confirm that we have read the Journal's position on issues involved in ethical publication and affirm that this work is consistent with those guidelines. This article is based on previously conducted studies and does not contain any new studies with human participants or animals performed by any of the authors.

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