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Commentary

Pediatric Mitochondrial Disorders can be Mistaken for Neuromyelitis Optica if not Examined Properly

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We read the article by Kim *et al.* [1] with interest, who reported on two patients, a 15 months-old female (patient-1) and a 5 year-old female with Leigh syndrome due to the mtDNA variants m.8344A>G in *MT-TK* (patient-1) and the variant m.13094T>C in *MT-ND5* (patient-2) [1]. Both patients were initially misdiagnosed as neuromyelitis optica (NMO) spectrum disorder (NMO-SD) [1]. It was concluded that early genetic diagnosis is important for reorientating care and avoiding potentially harmful immunosuppressant therapies in patients with a mitochondrial disorder (MID) [1]. The study is excellent but has limitations that are cause of concerns and should be discussed.

We disagree with the diagnosis of Leigh syndrome in patient-2 [1]. Since the magnetic resonance imaging (MRI) lesions have been described as asymmetric [1], the diagnosis is not justified. Leigh syndrome is typically characterised by symmetrical T2-hyperintense lesions of the basal ganglia, thalamus, brainstem, or cerebellum.

A limitation of the study is that magnetic resonance spectroscopy (MRS) was not performed on either patient-1 or patient-2. MRS can reveal increases in lactate or other metabolites, as previously described in patients with Leigh syndrome [2].

Although the degree of heteroplasmy was reported to be high in patient-1 [1], the exact degree of heteroplasmy was not specified. In patient-2, the heteroplasmy rate was not mentioned at all [1]. In addition, mtDNA copy number and haplotype should be given. Knowledge of heteroplasmy rates, mtDNA copy number, and haplotype is crucial as they determine the clinical presentation, disease course, and outcome of these patients.

Ptosis in patient-2 suggests metabolic myopathy or myasthenia rather than NMO-SD. There have been no reports of patients with NMO-SD presenting with ptosis.

It is not clear why patient-2 received methyl-prednisolone and plasma exchange. Myelin-oligodendrocyte glycoprotein (MOG) antibodies, aquaporin-4 antibodies, and oligoclonal bands were negative in this patient [1]. Although steroids can have a beneficial effect on some MIDs, they can also be harmful, so they should be administered with caution [3].

Since the m.8344A>G variant commonly manifests itself phenotypically as myoclonic epilepsy with ragged-red fiber (MERRF) syndrome and MERRF syndrome is clinically characterised by myocloni, generalised epilepsy, ataxia and myopathy [4], we should know if patient-1 presented with myocloni or generalised seizures. It would be also interesting to know if this patient had epileptiform discharges recorded on the electroencephalography (EEG).

Patient-2 presented with acute visual loss [1]. Was impaired visual acuity due to retinitis pigmentosa, optic atrophy, anterior, ischemic optic neuropathy (AION), posterior, ischemic optic neuropathy (PION), demyelination of the optic nerve, or a stroke-like lesion?

Surprisingly, neither in patent-1 nor in patient-2 were visually evoked potentials (VEPs) recorded [1]. P100 latencies are usually prolonged in patients with NMO-SD [5].

In summary, the interesting study has limitations that put the results and their interpretation into perspective. Addressing these issues would strengthen the conclusions and could improve the status of the study. Because MIDs often mimic other neurological diseases, such myasthenia, myotonic dystrophy, multiple sclerosis, ADEM, or neuromyelitis optica, it is crucial to rule out these differential diagnoses as early as possible to avoid MID patients receiving inadequate or potentially harmful treatment.

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Statement of Ethics: a) The study was approved by the institutional review board (responsible: Finsterer J.) at the 4th November 2022. b) Written informed consent was obtained from the patient for publication of the details of their medical case

and any accompanying images.

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