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MELAS requires comprehensive work-up and treatment

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With interest we read the article by Quinn et al. about two paediatric MELAS patients with typical phenotypic and genotypic features. Quinn et al. (2016) We have the following comments and concerns.

MELAS may not only be inherited but also sporadic. Thus, we should be informed about the family history to outline the trait of inheritance. Were family members other than the two index cases clinically affected? Were the mothers of the two presented cases clinically affected and did they undergo genetic testing? Were the 2 reported cases related? Was the $tRNA^{(Leu)}$ mutation in case-1 inherited or sporadic?

Due to the multiorgan nature, MELAS patients may not only experience stroke-like episodes (SLEs) but also ischaemic stroke. In case-1 FLAIR sequences suggest previous ischaemic stroke. Quinn et al. (2016) Was the history positive for ischemic stroke in this patient and were there any cardiovascular risk factors for stroke/embolism present? SLEs manifest on MRI as stroke-like lesions (SLLs), which present as hyperintensities on DWI and ADC maps (vasogenic oedema). Were these typical lesions also found in the two patients during SLEs? Is it conceivable that the MRI lesion represents the endstage of a previously unrecognised SLEs? Generally, endstage of a SLL on MRI may be a normal brain, laminar cortical necrosis, white matter lesions, or cystic lesions associated with focal atrophy and deformation of the ventricles.

The authors make the interesting point that basal ganglia calcification (BGC) could result from seizure activity. Quinn et al. (2016) Are there any studies available which support this statement? We agree that BGC can be a manifestation of a MID Finsterer & Kopsa (Finsterer & Kopsa) but there are a number of MIDs with BGC but without seizures. Were electrolyte disturbances or hormonal dysbalances considered as the cause of BGC?

Not only valproic-acid but also carbamazepine, phenytoin, and barbiturates may be mitochondrion-toxic. Finsterer (2016) These compounds should be given with caution to MID patients and only if other antiepileptic drugs (AEDs) fail to be effective. Valproic-acid should be particularly avoided in patients carrying POLG1 mutations. Hynynen et al., (Hynynen et al.) How was the quality of seizure control in both patients?

L-arginine has been reported beneficial for SLEs in MIDs. Did the two patients receive L-arginine for SLEs? Were the two under a ketogenic diet, which has been shown beneficial, particularly for mitochondrial seizures? Did the two receive coenzyme-Q? What was the outcome of the SLEs in the two patients? Did they recover completely?

Overall, reporting of these two interesting cases could be supplemented by more extensive description of the phenotype, family history, instrumental investigations, and treatment these two patients received. Respiratory arrest upon administration of benzodiazepines suggests MID.

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