Stroke-like episode relapse and concomitant exacerbation of autonomic and sensory neuropathy triggered by infection in pyruvate dehydrogenase complex deficiency

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Background

Pyruvate dehydrogenase complex (PDC) deficiency is a mitochondrial disorder of carbohydrate metabolism usually associated with neurological symptoms of various severity ranging from severe neonatal encephalopathy to adult-onset episodic ataxia. Stroke-like episodes and autonomic and sensory neuropathy have been rarely reported as a part of this disease phenotype.

Case Presentation

- 3-year-old boy with neonatal onset PDC deficiency;
- Two episodes of stroke-like episodes (SLEs) in the setting of upper respiratory infection; the second episode complicated by autonomic dysfunction attributable to acute-on-chronic neuropathy: acute urinary retention (no lesions of the spinal cord) + on ENMG: absent SNAPs, normal CMAPs, absent sympathetic skin response and reduced RR interval -> selective sensory and autonomic neuropathy;
- Imaging: on both occasions, magnetic resonance imaging (MRI) revealed T2-weighted fluid attenuated inversion recovery (FLAIR) hyperintensities with restricted diffusion involving primarily basal ganglia with a respective hyperemia on perfusion imaging + lipid/lactate peak on spectroscopy;
- Neuroradiological follow-up: progressive resolution of the lesions.

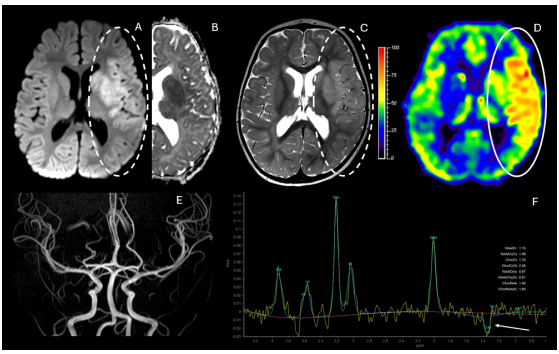

FIGURE 1
Axial (A) diffusion-weighted image (DWI) with corresponding (B) apparent diffusion coefficient (ADC) map; axial (C) T2-weighted image (T2WI), (D) pseudo-continuous Arterial Spin Labeling (pCASL), (E) 3D-TOF MR angiography, and (F) magnetic resonance spectroscopy (MRS). The MRI showed DWI and T2WI signal hyperintensity in the left striato-lenticular region and in the cortical-subcortical fronto-parieto-insular region on the same side (dashed circle). A corresponding area of hyperperfusion on pCASL (circle) and a normal visualization of the intracranial arterial vessels are also observed. MRS clearly showed the presence of a lipid/lactate peak (arrow).

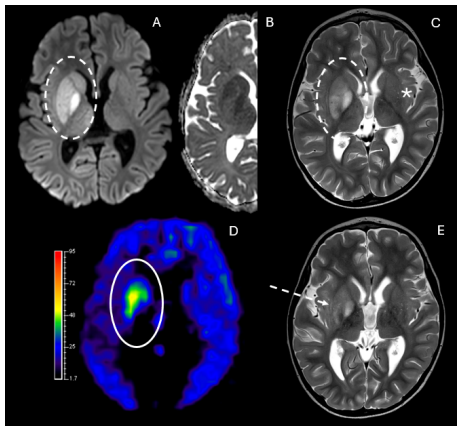

FIGURE 2
Axial (A) DWI with corresponding (B) ADC map, axial (C) T2WI and (D) pCASL. The MRI showed DWI and T2WI signal hyperintensity in the right striato-lenticular region (dashed circle) with corresponding area of hyperperfusion at pCASL (circle); complete resolution of the signal abnormalities previously depicted on the controlateral hemisphere were also noted (asterisk). Follow-up MRI (E) after two weeks showed initial resolution of the T2WI signal abnormality in the right basal ganglia (dashed arrow).

Conclusions

This report reliably extends the phenotypic variability of neurological symptoms in PDC deficiency including SLEs as its possible paroxysmal manifestation. Peripheral sensory and autonomic neuropathy may be underrecognized components of this mitochondrial disorder subjected to aggravation during periods of increased metabolic stress such as ongoing infection.

References

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