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EPISODIC MUSCLE WEAKNESS IS RATHER ATTRIBUTABLE TO PYRUVATE DEHYDROGENASE DEFICIENCY THAN TO GUILLAIN-BARRE SYNDROME

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Letter to the Editor

With interest we read the article by Sen et al. about two patients with pyruvate dehydrogenase (PDC) deficiency (patient-1: 5yo male, patient-2: 39yo female, mother of patient-1) due to the variant c.821G > C; p.R274T in PDHA1 [1]. Patient-1 manifested with developmental delay, episodic muscle weakness, dystonia, epilepsy, and episodic lactic acidosis [1]. Patient-2 manifested with quadruparesis, ophthalmoparesis, and migraine [1]. It was concluded that PDC deficiency should be included in the differential diagnoses of alternating hemiplegia in childhood [1]. The study is appealing but raises the followin1g comments and concerns.

We do not agree that Guillain Barre syndrome (GBS) is a phenotypic manifestation of PDC deficiency [1]. GBS is an acquired, immunological disease, presenting with various different subtypes and diagnosed upon the clinical presentation, nerve conduction studies (NCSs) and elevated protein in the cerebro-spinal fluid (CSF) but normal cell count, known as “dissociation cyto-albuminque” [2]. In the vast majority of the cases, GBS is triggered by a bacterial or viral infection preceding the onset of the neurological compromise or by vaccination. However, patient-2 did not have dissociation cyto-albuminique at age 36y, when she developed an episode of tingling feet, sudden onset quadruparesis, dysphagia, and respiratory failure requiring artificial ventilation, tracheotomy, and placement of a gastrostomy tube. Results of NCSs were not reported. No antecedent infection was reported. Tough the clinical presentation was suggestive of GBS it is crucial to confirm the diagnosis according to established guidelines. One of the most widely applied diagnostic criteria for GBS are the Brighton criteria [3]. Since patient-2 was a manifesting carrier of PDC deficiency and since PDC deficiency can manifest with episodic weakness [4], it is conceivable that the episode at age 36y was rather attributable to PDC deficiency than to a GBS.

Since patient-2 had flaccid quadruparesis already prior to onset of “GBS” we should know the cause of pre-existing flaccid quadruparesis, particularly if it was attributed to the genetic defect or if the patient had chronic inflammatory demyelinating poly neuropathy (CIDP). Assuming that the patient had CIDP, it is conceivable that acute worsening at age 36y was rather a deterioration of CIDP than GBS. To differentiate between the two causes of flaccid quadruparesis we should know if the current medication of patient-2 was changed shortly before the onset of the episode or if she experienced a systemic viral or bacterial infection.

We do not agree with the classification of the bilateral lesions in the globus pallidus on DTI of patient-1 as metabolic stroke (stroke-like lesion (SLL)) [1]. Though SLLs may occur subcortically, they originate in the vast majority from the cortex and spread consecutively to the white matter [5]. SLLs are best diagnosed by multimodal MRI. There, they present as hyperintensity on T2-weighted images, on

diffusion weighted imaging (DWI), and on perfusion weighted imaging (PWI) [5,6], On oxygen-extraction fraction MRI SLLs appear as hypointensity and FDG-PET reveals hypometabolism within the lesion [7]. SLLs are dynamic in size and usually expand in the acute stage to regress in the chronic stage [5]. SLLs end up as white/gray matter lesion, cyst, laminar cortical necrosis, toenail-sign, or become invisible.

Overall, the study has shortcomings which should be addressed before drawing conclusions those presented. Established diagnostic criteria for diagnosing GBS should be applied, CIDP should be considered as a differential in patient-2, and the diagnosis metabolic stroke in patient-1 should be reassessed.

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References

1 Sen K, Grahame G, Bedoyan JK, Gropman AL. Novel presentations associated with a PDHA1 variant - Alternating hemiplegia in Hemizygote proband and Guillain Barre Syndrome in Heterozygote mother. Eur J Paediatr Neurol. 2021 Jan 22;31:27-30. doi: 10.1016/j.ejpn.2021.01.006.

2 Nguyen TP, Taylor RS. Guillain Barre Syndrome. 2020 Nov 19. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2020 Jan–.

3 Fokke C, van den Berg B, Drenthen J, Walgaard C, van Doorn PA, Jacobs BC. Diagnosis of Guillain-Barré syndrome and validation of Brighton criteria. Brain. 2014 Jan;137(Pt 1):33-43. doi: 10.1093/brain/awt285.

4 Debray FG, Lambert M, Gagne R, Maranda B, Laframboise R, MacKay N, Robinson BH, Mitchell GA. Pyruvate dehydrogenase deficiency presenting as intermittent isolated acute ataxia. Neuropediatrics. 2008 Feb;39(1):20-3. doi: 10.1055/s-2008-1077084.

5 Finsterer J, Aliyev R. Metabolic stroke or stroke-like lesion: Peculiarities of a phenomenon. J Neurol Sci. 2020 May 15;412:116726. doi: 10.1016/j.jns.2020.116726.

6 Kim JH, Lim MK, Jeon TY, Rha JH, Eo H, Yoo SY, Shu CH. Diffusion and perfusion characteristics of MELAS (mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episode) in thirteen patients. Korean J Radiol. 2011 Jan-Feb;12(1):15-24. doi: 10.3348/kjr.2011.12.1.15.

7 Finsterer J. The metabolic hypothesis is more likely than the epileptogenic hypothesis to explain stroke-like lesions. Wellcome Open Res. 2020 Jun 24;5:51. doi: 10.12688/wellcomeopenres.15758.2.