Int. j. adv. multidisc. res. stud. 2023; 3(1):850-851

Received: 26-01-2023
Accepted: 03-02-2023

International Journal of Advanced Multidisciplinary Research and Studies

ISSN: 2583-049X

Letter to the Editor

Neuro-acanthocytosis requires an etiological assignment to optimize therapy an improve the outcome

[1] Sounira Mehri, [2] Josef Finsterer

[1] Biochemistry Laboratory, LR12ES05 "Nutrition-Functional Foods and Vascular Health", Faculty of Medicine, Monastir, Tunisia

[2] Neurology & Neurophysiology Center, Vienna, Austria

Corresponding Author: **Josef Finsterer**

We read with interest the article by Sogabe *et al* on a 32-years-old female with epilepsy who was admitted after a generalised seizure with tongue biting [1]. Her history was positive for hearing impairment in addition to epilepsy [1]. The patient had to be intubated due to an airway obstruction by blood [1]. Cerebral computed tomography was non-informative. Acanthocythosis was diagnosed and the patient benefited significantly from deep brain stimulation [1]. The study is excellent but has limitations that are objectionable and should be discussed.

The main limitation of the study is that the cause of acanthocytosis has not been elucidated. Acanthocytosis can generally be hereditary or acquired. The most common causes of acquired acanthocytosis are liver dysfunction, post-splenctomy syndrome, hypothyroidism, myelodysplastic syndrome, malnutrition (anorexia nervosa, cystic fibrosis), and medications. The most common genetic causes are autosomal recessive chorea-acanthocytosis, X-linked McLeod syndrome (MLS) due to mutations in the *XK* gene, paroxysmal, nocturnal hemoglobinurea, A-beta-lipoproteinemia, and mutations in the *VPS13A* gene [2, 3].

Another limitation of the study is that no results from electroencephalography (EEG) recordings were reported [1]. Because the patient was admitted after seizures, had perioral movements on hospital day 2, and had seizures on hospital day eight of hospitalisation, it is crucial that EEG results are presented. Since acanthocytosis plus epilepsy is most commonly found in McLeod syndrome, we should know whether any variants in the *XK* gene have been detected.

A third limitation of the study is that the presented cerebral imaging is of poor quality. We should know if there was bilateral atrophy of the caudate nucleus or the putamen on cerebral magnetic resonance imaging (MRI). Also lacking are FP-CIT-single photon emission computed tomography (SPECT) studies, which may reveal loss of striatal dopamine transporter availability and FDG-positron emission tomography (PET) studies, which can show reduced striatal glucose metabolism [4].

A fourth limitation of the study is that chorea acanthocytosis was not considered as a differential diagnosis. We should know if the involuntary, perioral movements described in the index patient were in fact choreatic movements. A video of the movement disorder should be provided.

Overall, the interesting study has limitations that call the results and their interpretation into question. Clarifying these weaknesses would strengthen the conclusions and could improve the study. Neuro-acanthocytosis requires an etiological assignment to optimize therapy and improve the outcome.

Acknowledgements

Funding: No funding was received.

Data access statement: All data are available from the corresponding author.

Ethics statement: Not applicable.

Author contribution: SM: design, literature search, discussion, first draft, critical comments, final approval. JF: design, literature search, discussion, critical comments, final approval.

Disclosures: The author declares that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

Data access statement: Not applicable.

Compliance with Ethics 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.

Keywords: Acanthocytes, Chorea, Seizures, Mcleod Syndrome, Deep Brain Stimulation

References

1. Sogabe T, Ishida K, Ogawa H, Ohnishi M. Tongue-biting ataxia that appeared to be a psychiatric disorder: a case of neuroacanthocytosis. Acute Med Surg. 2023; 10(1):e815. Doi: 10.1002/ams2.815.
2. Vaisfeld A, Bruno G, Petracca M, Bentivoglio AR, Servidei S, Vita MG, *et al.* Neuroacanthocytosis Syndromes in an Italian Cohort: Clinical Spectrum, High Genetic Variability and Muscle Involvement. Genes (Basel). 2021; 12(3):344. Doi: 10.3390/genes12030344.
3. Zheng Q, Zhu L, Zhang C, Jiao L. Nemaline Rods in a Patient of Chorea-Acanthocytosis with a Novel Pathogenic Mutation of VPS13A Gene. Neurol India. 2021; 69(6):1848-1849. Doi: 10.4103/0028-3886.333435.
4. Weber J, Frings L, Rijntjes M, Urbach H, Fischer J, Weiller C, *et al*. Chorea-Acanthocytosis Presenting as Autosomal Recessive Epilepsy in a Family with a Novel *VPS13A* Mutation. Front Neurol. 2019; 9:1168. Doi: 10.3389/fneur.2018.01168.