RESPONSE TO LETTER
TO THE EDITORS

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From rarity to reality: Poland's first case of neurological Erdheim-Chester Disease with cerebellar manifestations

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To the Editors

We thank Finsterer and Mehri [1] for their interest in our paper [2] and their constructive comments. We welcome this chance to clarify and expand upon the points they have raised.

In reply to the first statement on our report of the first case of ECD with neurological involvement in Poland, we hereby acknowledge the case described by Chrostowska et al. in 2022 [3]. Their patient showed a massive infiltration of the ocular adnexa of the eye with compression of the optic nerve and presented with severe limitation of eye motility and episodes of diplopia. While we were aware that the optic nerve is anatomically part of the CNS, we regret not citing this case, and thank Finsterer and Mehri for bringing it to our attention. We also acknowledge that we selected a somewhat provocative title to underline the importance of recognising ECD's neurological manifestations. Our aim was to raise awareness among neurologists that, in Poland (population 38 million) and other European countries, other cases are likely to exist. Our intention was to highlight a case involving cerebellar involvement, representing a different spectrum of CNS manifestations in ECD that required neurological assessment and management, thereby contributing to the neurological understanding of ECD in Poland.

Finsterer and Mehri also highlighted the endocrinological manifestations of ECD [1]. We fully agree that such symptoms are of interest and should be included. Due to journal limitations, and since our patient did not exhibit endocrine symptoms or abnormalities indicating endocrine disorders — and was indeed thoroughly examined in this regard — we did not emphasise this aspect of the disease in our paper.

However, we agree that in the discussion we should have included information that endocrine symptoms may occur within the spectrum of ECD manifestations. As delineated in the consensus recommendations by Goyal et al. [4], endocrine dysfunction is a common feature in ECD, and deserves a place in the comprehensive management of the disease. We shall keep this aspect in focus in future discussions, and work towards interdisciplinary cooperation to bring about the best outcomes for patients.

The authors of the letter also referenced neurological symptoms cited from the research by Jezierska et al. [5], which is linked to Langerhans cell histiocytosis (LCH) in the paediatric population. LCH and ECD are both histiocytoses, but they are very different in their clinical manifestations and demographic distributions. ECD is found predominantly in older males, and is comparatively rarely seen in the paediatric age group. It is important to distinguish between these entities so as to avoid inappropriate extrapolation of symptoms from LCH patients to ECD-diagnosed patients. LCH patients might present with more pronounced neurological symptoms than ECD individuals. However, this remains an unanswered question as it is also known that there might be a possible overlap between ECD and LCH, as underlined in the study by Pegoraro et al. [6], which could have consequences on the evolution and symptoms of ECD.

To conclude, we appreciate the chance to become part of this debate and believe that expert dialogues such as this will contribute substantially to better understanding regarding ECD. We hope that this response has clarified our position, and that we have emphasised the utmost importance of interdisciplinary approaches in the diagnosis and treatment of rare diseases like ECD.

Article information

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