Indian Journal of Pediatrics (October 2024) 91(10):1092
https://doi.org/10.1007/s12098-024-05120-2

CORRESPONDENCE

Recurrent Mononeuropathy, Stroke and Deafness in a Teenage Boy

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Received: 14 January 2024 / Accepted: 27 March 2024 / Published online: 10 April 2024

To the Editor: Monogenic autoinflammatory disorders pose a diagnostic challenge due to their variable age of onset and protean manifestations. We report an intriguing case of a 17-y-old boy who began experiencing recurrent neurological symptoms at age 10, starting with an acute-onset left ulnar axonopathy, recovering with antitubercular therapy and steroids. At 15 y, he suffered a brainstem ischemic stroke. Basic stroke workup identified positive anti-nuclear antibodies (1:100 titre, speckled pattern) with rest procoagulant/vasculitis workups being negative. The patient recovered with aspirin and rehabilitative measures within three weeks. A year later, he developed an acute-onset right foot drop (peroneal axonopathy), resolving completely with prednisolone. Ten months later, recurrent acute-onset hearing loss occurred, partially recovering with steroids. Extensive investigations over seven years (Supplementary Fig. S1) including vasculitis workups and digital subtraction angiography, yielded inconclusive results. Suspecting an inherited vasculitis syndrome, whole exome sequencing was sent, which revealed a likely pathogenic, homozygous missense variant (c.139G>C, p.Gly47Arg) in exon 2 of *ADA2* gene, confirming the diagnosis of deficiency of adenosine deaminase 2 (DADA2). The patient was started on weekly etanercept and aspirin was discontinued given the risk of hemorrhagic strokes. The patient has been on follow-up for 1 y without experiencing additional flares.

DADA2 is a recently described monogenic autoinflammatory disorder encompassing hematologic, immunologic, neurologic and dermatologic features with elevated inflammatory markers [1, 2]. Neurological manifestations like early-onset stroke, mononeuritis multiplex, cranial neuropathies and cerebral vessel aneurysms are seen in 50–80% of patients. The index case exhibited a purely neurological phenotype, harboured a pathogenic missense variant (p.G47R) and also belonged to the Aggarwal community, echoing a noteworthy observation from the published Indian cohort [3]. Consensus statements advocate lifelong tumor necrosis factor inhibitor (TNFi) therapy for DADA2 with vasculitis, stroke or ongoing systemic inflammation [4]. Recognition of this treatable entity is crucial for pediatricians and specialists, ensuring timely diagnosis and initiating TNFi to avert potential morbidity and mortality.

Supplementary Information The online version contains supplementary material available at https://doi.org/10.1007/s12098-024-05120-2.

Declarations

Research Ethics and Informed Consent Written informed consent for publication of their clinical details and/or clinical images was obtained from the patient.

Conflict of Interest None.

References

1. Maccora I, Maniscalco V, Campani S, et al. A wide spectrum of phenotype of deficiency of deaminase 2 (DADA2): a systematic literature review. Orphanet J Rare Dis. 2023;18:117.
2. Barron KS, Aksentijevich I, Deuitch NT, et al. The spectrum of the deficiency of adenosine deaminase 2: an observational analysis of a 60 patient cohort. Front Immunol. 2022;12. Available at: https://www.frontiersin.org/articles/10.3389/fimmu.2021.811473. Accessed on 2 Dec 2023.
3. Sharma A, Naidu G, Sharma V, et al. Deficiency of adenosine deaminase 2 (DADA2) in adults and children: experience from India. Arthritis Rheumatol. 2021;73:276–85.
4. Lee PY, Davidson BA, Abraham RS, et al. Evaluation and management of deficiency of adenosine deaminase 2: an international consensus statement. JAMA Netw Open. 2023;6:e2315894.

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