Korean J Ophthalmol 2023;37(1):93-94
https://doi.org/10.3341/kjo.2022.0130

Letters to the Editor

Letter to the Editor: Comments on "Acute Partial Oculomotor Nerve Palsy and Optic Neuritis Preceding Juvenile Idiopathic Arthritis: A Case Report"

Dear Editor,

Chung et al. [1] reported a case of recurrent cranial nerve palsy and subsequent optic neuritis preceded juvenile idiopathic arthritis. Further systemic evaluations and following ocular examinations revealed the history of skin rash, lymph node enlargement, fever, small intestine perforation, facial nerve palsy, and nonsterile arthritis (Fig. 1). Given the unusual phenotype, we decided to perform trio whole exome sequencing. The whole exome sequencing revealed that compound heterozygote mutation (p.Gly358Arg) on the adenosine deaminase 2 (ADA2) results in autosomal recessive monogenic vasculitis syndrome.

Deficiency of ADA2 (DADA2) is a rare, recessively inherited monogenic autoinflammatory disease characterized by systemic inflammation, vasculitis, early-onset stroke, cytopenia, bone marrow failure, and immunodeficiency [2]. Inflammation of blood vessels most characteristically involves the central nervous system, causing recurrent strokes that are either ischemic or hemorrhagic and often begin in early childhood [2]. DADA2 involving multiple organs engages a differential diagnosis that includes other forms of the various autoinflammatory disease, such as juvenile idiopathic arthritis, polyarteritis nodosa, antiphospholipid syndrome, or systemic lupus vasculitis [3–5]. DADA2 also has various ocular manifestations, such as a reduction in choroidal thickness, paralytic strabismus, retinal vasculitis, and optic neuritis [2]. The diagnosis of DADA2 should be suspected in young children who present with recurrent stroke related to central nervous system vasculitis, and unusual ocular inflammations, especially in the presence of livedoid skin lesions, recurrent fever, and cytopenia without evidence of infection or autoimmunity. Currently, tumor necrosis factor inhibitors are the treatment of choice for vasculitis and inflammatory manifestations and for preventing strokes [2].

We suggest that inherited autoinflammatory disease, including variants in ADA2, should be differentiated when pediatric patients had recurrent cranial nerve palsy associated with stroke, ocular inflammations, and systemic inflammatory signs.

Conflicts of Interest: None.
Acknowledgements: None.
Funding: None.

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Received: September 21, 2022
Accepted: December 18, 2022

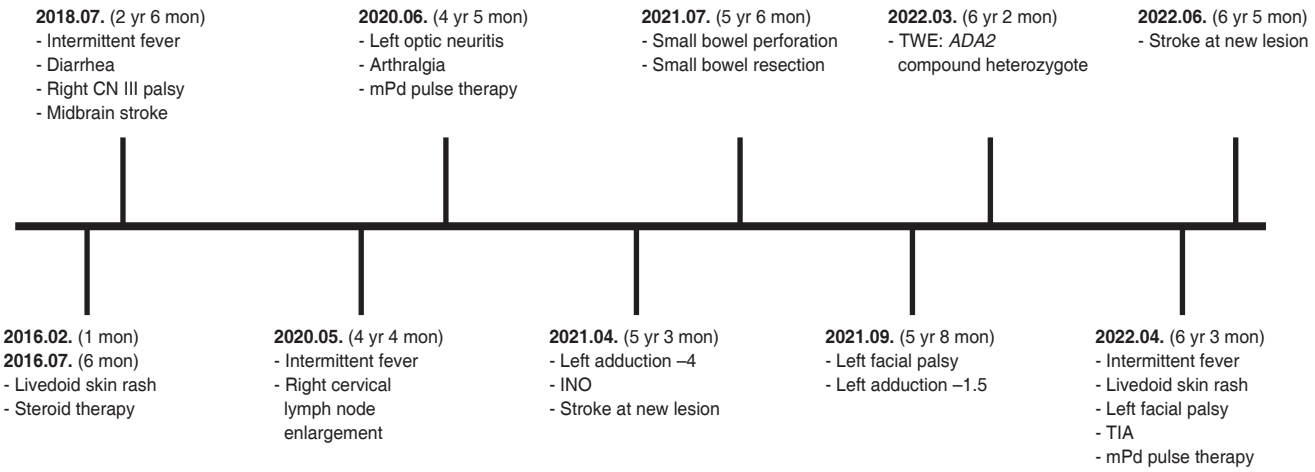

Fig. 1. Clinical course of the patient with deficiency of adenosine deaminase 2 (ADA2). CN = cranial nerve; mPd = methylprednisolone; TWE = trio whole exome; INO = internuclear ophthalmoplegia; TIA = transient ischemic attack.

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