Visual Loss in Biotinidase Deficiency

We read with interest the article titled "Subacute Myelopathy: Think Beyond Neuromyelitis Optica Spectrum disorder" by Kasinathan *et al.*^[1] Authors have highlighted the significance of high index of suspicion in diagnosing late onset Biotinidase deficiency (BD). We appreciate the author's effort and research. We would like to highlight few points on late onset BD from neuro-ophthalmic perspective. Older children and adolescents with BD may present with limb weakness, hearing loss, optic atrophy (OA), and scotomas.^[2] Acute incidents precipitated by stress (infection) interspersed by periods of apparent normality is a feature of partial BD.

Chinta *et al.*^[3] evaluated the etiology of optic atrophy (OA) in 324 children (583 eyes) <16 years of age. In 98 children etiology of optic atrophy was not known. Twenty-three children (OA of unknown etiology) either had history of delayed milestones or seizures. Neuroimaging was normal in this subset of children. Our assumption is, probably late onset BD should be ruled out in older children and adolescents of this subset. Long-term treatment with antiepileptic drugs may further cause biotin deficiency.

Khadse *et al.*^[4] evaluated clinical features of 40 children (62 eyes) diagnosed as optic neuritis (ON). Nutritional optic neuropathy was not an exclusion criteria in this study. In two cases of recurrent ON, aetiology of ON was unknown. Seven ON eyes had final visual outcome of <20/200. Probably late onset BD should be considered in older children developing recurrent pediatric ON of unknown aetiology associated with poor visual outcome. Children with borderline malnutrition/vitamin deficiency, develop frank vitamin deficiency following a bout of illness.

To conclude, BD should be considered in older children presenting with cutaneous stigmata, optic atrophy, acute loss of vision, scotomas, deafness, neurological symptoms, and seizures. High index of suspicion^[5] is necessary to diagnose BD.

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Conflicts of interest

There are no conflicts of interest.

Nataraja P. Venugopal

Neuro-Ophthalmology Clinic and Glaucoma Service, AG Eye Hospital, Puthur, Trichy, Tamil Nadu, India

Address for correspondence: Dr. Nataraja P. Venugopal, Neuro-Ophthalmology Clinic and Glaucoma Service, AG Eye Hospital, Puthur, Trichy, Tamil Nadu - 620 017, India. E-mail: natavenu@yahoo.com

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