Comment on: Childhood optic atrophy in biotinidase deficiency

Sir,

We read with interest the article titled "Etiology and clinical profile of childhood optic nerve atrophy at a tertiary eye care center in South India" by Chinta *et al*.[1] In this article, authors have highlighted the importance of investigations to rule out serious systemic conditions causing optic atrophy. Authors have also mentioned about socioeconomic constraints precluding extensive investigations. We appreciate authors' effort and research work.

We would like to highlight a few points regarding biotinidase deficiency (BD), which is known to cause optic atrophy. This condition is treatable with oral biotin supplementation. BD may be profound or partial.[2] Profound deficiency manifest between 3 and 6 months of age with neurological, cutaneous, and pulmonary features. Older children present with limb weakness, deafness, optic atrophy, and scotomas. The onset may be acute or insidious, with either a steady progression or a series of acute incidents interspersed with periods of apparent normality. The latter is seen with partial BD. Partial BD may cause mild symptoms during the period of stress (e.g., infection). There are few case reports of BD initially presenting with optic neuropathy and atrophy.

Hayati *et al*.[3] studied seven BD patients, who presented with ocular features. Six of their patients had optic atrophy. Their age ranged from 5 to 15 years. Apart from optic atrophy, optic neuritis and retrobulbar neuritis are recognized clinical manifestation of BD in older children.

A very high prevalence of inborn errors of metabolism (IEM)[4] to the extent of 1 in every thousand newborns was observed in Expanded Newborn Screening Programme, 2000, conducted at Hyderabad. Lodh *et al*.[5] documented 91 newborns with IEM. Thirty nine (42.8%) patients had BD. The regional incidence and prevalence rates of IEM may provide a useful guideline for the selection of appropriate investigation, thereby reducing the economic burden of the patient's family. To conclude, BD should be ruled out in older children presenting with optic atrophy, acute visual loss, scotomas, deafness, neurological symptoms, seizures, and cutaneous scars.

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Conflicts of interest

There are no conflicts of interest.

N Venugopal, Sherin Kummararaj[1]

Department of Neuro-Ophthalmology, Clinic and Glaucoma Service, [1]Pediatric Ophthalmology and Perimetry Centre, AG Eye Hospital, Puthur, Tiruchirappalli, Tamil Nadu, India

Correspondence to: Dr. N Venugopal, No. 19, Mathuram Apartments (Behind YMCA), Officer's Colony, Puthur, Tiruchirappalli - 620 017, Tamil Nadu, India.
E-mail: natavenu@yahoo.com

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