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Visual Vignette

A Boy With Progressive Cognitive Impairment and Darkening of Skin

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Case Presentation

A 15-year-old boy was brought to our hospital for evaluation of progressive cognitive impairment and darkening of skin for 3 years. He could not keep up with his peers in school, and there was hearing difficulty and dimness of vision as well. His parents noticed darkening of the skin along with the pigmentation of the tongue and oral mucosa (Fig. 1 and 2). There was no history of such illness in his family. His blood pressure was 100/60 mm Hg, with no postural drop. The magnetic resonance imaging scan of the brain revealed bilateral symmetrical large areas of change in signal intensity within the deep white matter of both the cerebral hemisphere and brainstem, suggestive of a white matter lesion (Fig. 3). His basal cortisol level was 1.70 μg/dL (reference range, 4.5-22.7 μg/dL), and the level of serum adrenocorticotropic hormone was 1207 pg/mL (reference range, 5-46 pg/mL). The levels of plasma very long-chain fatty acids and branched-chain fatty acids (very long-chain fatty acids, C26:0 and C26/C22) were elevated.

What is the diagnosis?

Answer

Adrenoleukodystrophy. The clinical course, imaging findings, and biochemical findings suggested a diagnosis of X-linked adrenoleukodystrophy. It is a rare, X-linked, peroxisomal disorder caused by mutations in the *ABCD1* gene that leads to high levels of very long-chain fatty acids in the plasma, which is found to be accumulated in the white matter of the brain, spinal cord, and adrenal cortex.[1,2] Supplementation of hydrocortisone was started at a physiologic dose. Except for the deteriorating cognitive function, the patient was stable. Hematopoietic stem cell transplantation and gene therapy are the only therapies in the early stages of cerebral disease.[3] The index patient in this case could not avail the therapy because of resource constraints.

Fig 1.

Disclosure

The authors have no multiplicity of interest to disclose.

Statement of Ethics

Written informed consent was obtained from the patient for his anonymous information to be published in this case study. This visual vignette has been approved by the ethics board of the National Institute of Neurosciences and Hospitals, Bangladesh.

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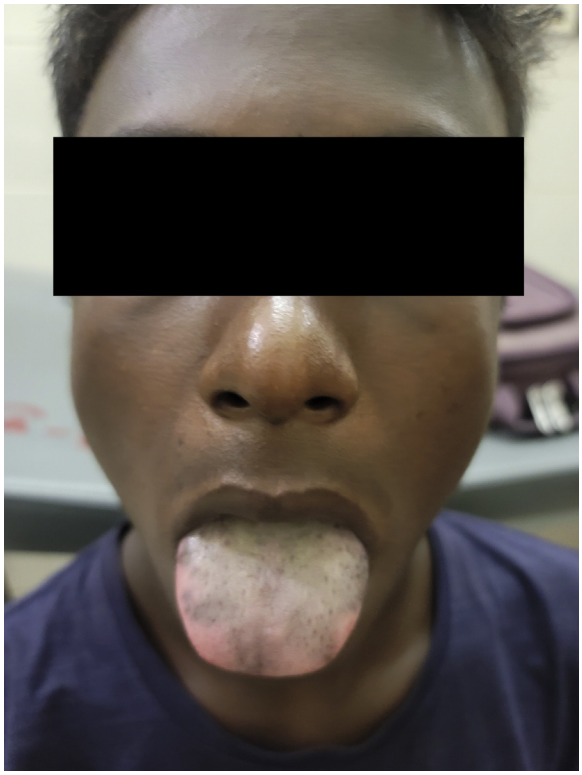

Fig 2.

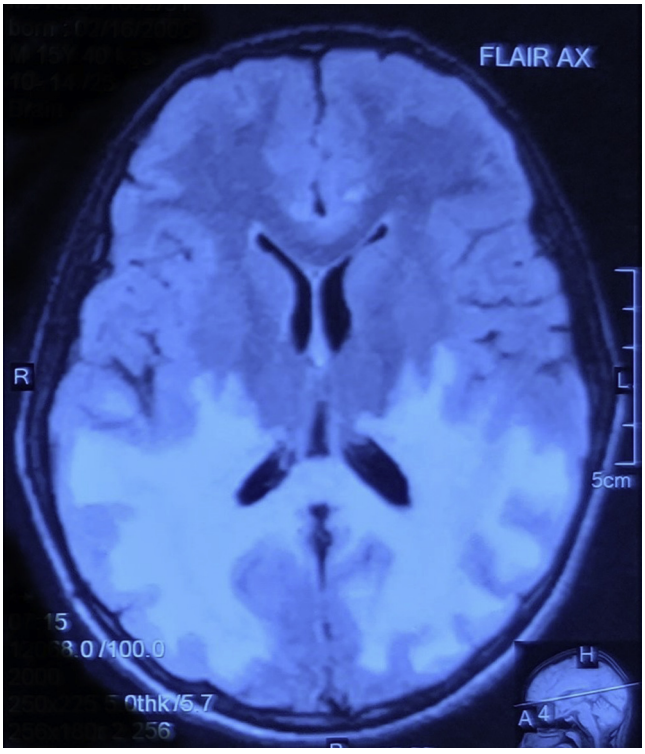

Fig 3.

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