A new entity of hypomyelination with atrophy of basal ganglia and cerebellum-like syndrome with bilateral developmental cataract

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Key words: Bilateral developmental cataract, hypomyelination with atrophy basal ganglia and cerebellum, leukoencephalopathy

A 6-year-old girl presented with defective vision for distance. She was born at term to healthy, nonconsanguineous parents. The antenatal and immediate neonatal period were uneventful. She had developmental delay with a history of tonic-clonic seizures.

On examination, slurred speech, truncal ataxia with generalized spasticity, no facial dysmorphic features, or dental abnormalities were noted. She walked with support. Her right eye had posterior subcapsular cataract and the left eye had total cataract [Fig. 1].

On MRI brain, hyposignal in T1, hypersignal in T2, and flair of symmetrical distribution, compromising diffusely the brainstem, cerebral, and cerebellar white matter were noted with lateral ventricle enlargement, basal ganglia and corpus callosum dimension reduction, and cerebral sulci and cerebellar vermis accentuation [Fig. 2]. Thus, it is indicative of diffuse hypomyelination with atrophy of the basal ganglia and cerebellum, a classical finding noted in H-ABC syndrome.[1] The child subsequently underwent bilateral cataract extraction with IOL implantation.

Postwritten informed consent targeted exome sequencing was performed for genetic analysis. However, TUBB4A mutation was not found.

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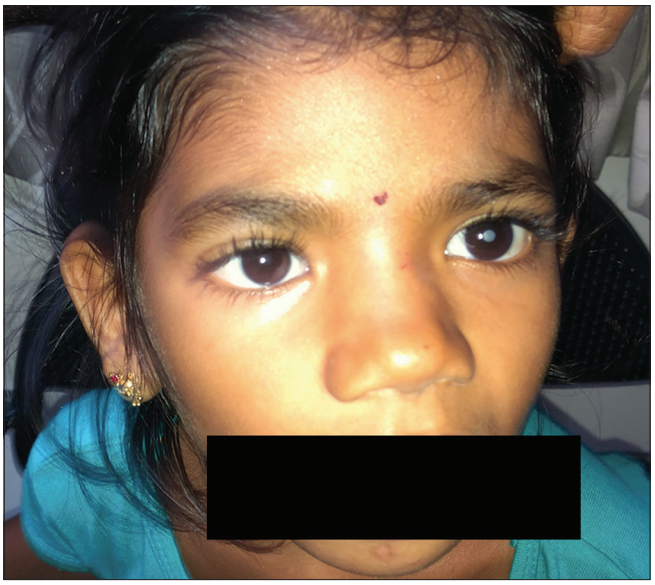

Figure 1: Right eye showing posterior subcapsular cataract and left eye total cataract

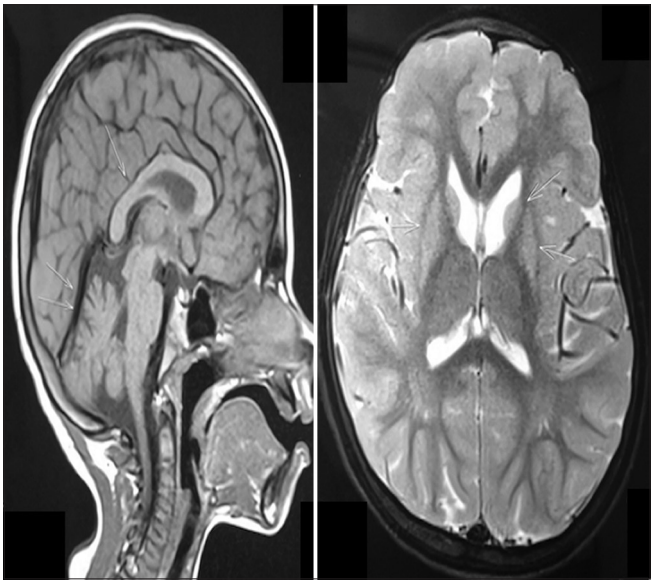

Figure 2: MRI brain showing hyposignal in T1, hypersignal in T2 and flair of symmetrical distribution, compromising diffusely the cerebral and cerebellar white matter, corpus callosum and internal capsule, as well as the brain stem, besides a mild enlargement of lateral ventricles, important reduction of the dimensions of the basal ganglia, preserved thalamus, thin corpus callosum, discrete accentuation of cerebral sulci and of cerebellar vermis

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Discussion

H-ABC syndrome is an autosomal dominant progressive leukoencephalopathy caused by mutations in the Tubulin Beta 4A Class IVa (TUBB4A) gene. However, sporadic de novo mutations have also been detected.[2] Age at presentation can vary characterized initially by normal to delayed motor development, subsequently pyramidal signs and extrapyramidal signs with speech disorders.

The ocular findings include nystagmus secondary to an oculogyric crisis and optic nerve involvement.[3] Bilateral developmental cataracts have not been reported in H-ABC syndrome though reported mainly in leukoencephalopathy with vanishing white matter[4] and in hypomyelination and congenital cataract syndrome. These have an autosomal recessive inheritance with MRI suggestive of hypomyelination with cerebellar and not basal ganglia involvement.[5] The classical triad of generalized hypomyelination with basal ganglia and cerebellar atrophy is highly indicative of H-ABC as described by Van der knapp.[1]

We thus present a unique case of H-ABC-like syndrome with developmental cataract having a clinical and MRI presentation suggestive of H-ABC syndrome in the absence of TUBB4A mutation.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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