



Juvenile Neuronal Ceroid Lipofuscinoses: A Rare Case

Juvenil Nöronal Seroid Lipofusinozlar: Nadir Bir Olgu

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Keywords: Vision loss, seizures, mental retardation, optic atrophy
Anahtar Kelimeler: Görme kaybı, nöbetler, zeka geriliği, optik atrofi

Dear Editor,

A 19-year-old male with a known 13-year history of mental retardation and seizure disorder presented with uncontrolled seizures and altered behavior, which he had had for 3 days. From the age of 7 years, the patient had a history of uncontrolled generalized tonic clonic seizures for which he was prescribed antiepileptic drugs. With time, there was progressive loss of vision, intelligence, and previously acquired physical and mental abilities. There was no past history of any other chronic illness. The patient had normal birth and developmental history. Family history revealed that his elder brother had a similar illness and died at the age of 18 years.

In the general examination, the patient was drowsy. Vitals and temperature were normal. In the central nervous system examination, the patient was disoriented to time, place, and person. Higher mental functions could not be assessed. No signs of meningitis were present. Power was 5/5 in all four limbs. All superficial and deep tendon reflexes were normal bilaterally. Plantar reflexes were flexor on both sides. Respiratory, cardiovascular, and abdominal examinations were essentially normal.

In the laboratory examination, the complete hemogram and biochemical parameters were normal. A peripheral blood smear revealed vacuolated lymphocytes. In a fundus examination, bilateral optic atrophy was seen with severe attenuated vessels, bony spicules, and cystoid macular edema. There were circular bands of different shades of pink and orange at the optic nerve

and retina resembling a bull's eye in the back of the eye (Figure 1). An electroencephalogram showed spike and wave discharges suggestive of seizure activity.

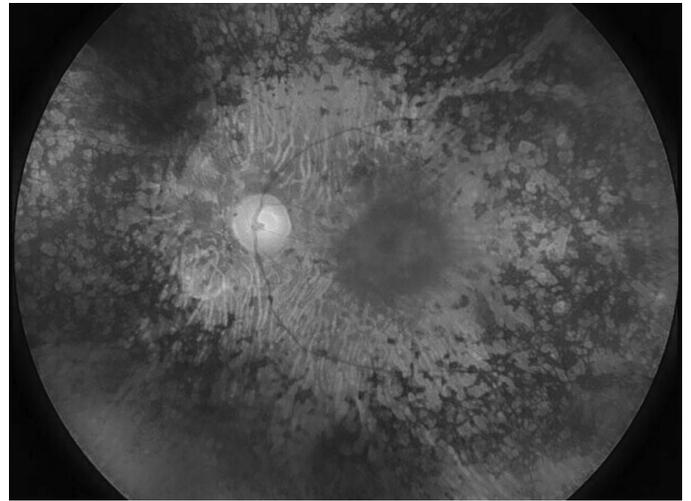


Figure 1. Fundus examination showing bilateral optic atrophy with severe attenuated vessels, bony spicules and cystoid macular edema. Circular bands of different shades of pink and orange at the optic nerve and retina also present.

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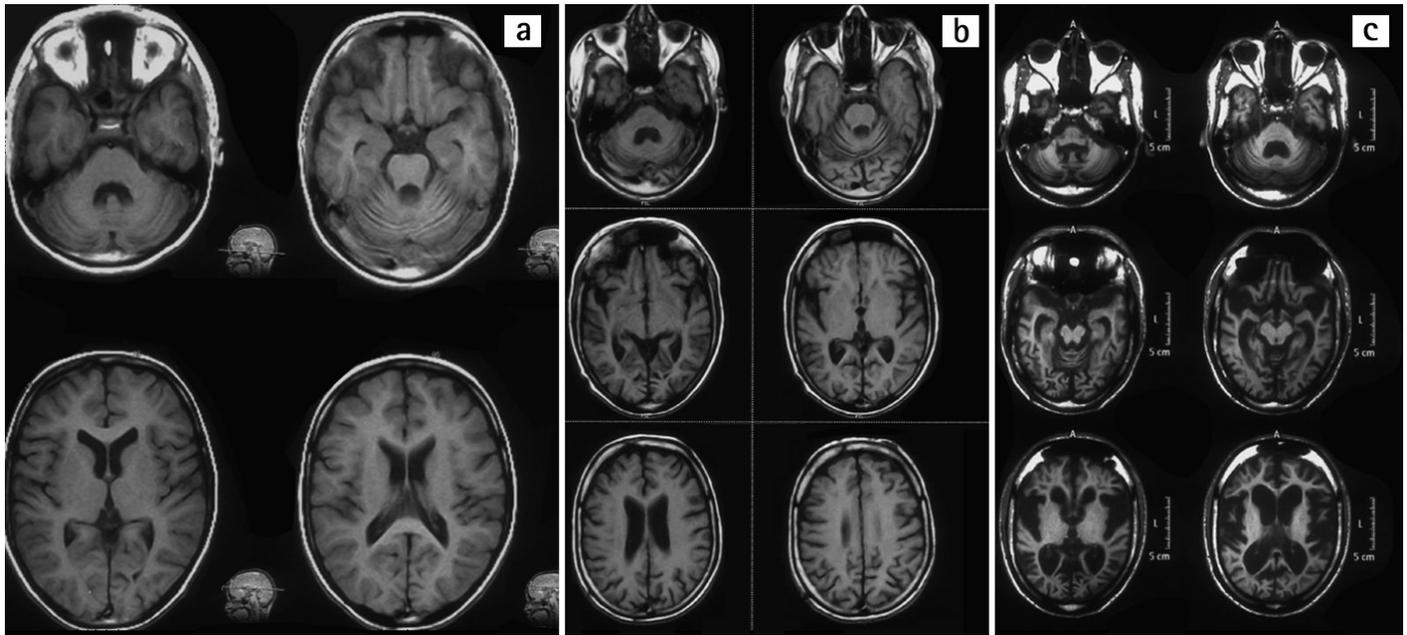


Figure 2. a) Normal magnetic resonance imaging brain done at the age of 7 years, b) magnetic resonance imaging at age of 11 years showed cerebral and cerebellar atrophy, c) magnetic resonance imaging brain at age of 19 years revealed diffuse cerebral and cerebellar atrophy with hypodensity in thalamus and few white matter changes in bilateral frontal area.

Previous magnetic resonance imaging (MRI) of the brain performed when the patient was aged 7 years was normal (Figure 2a). Repeat MRI at age 11 years showed cerebral and cerebellar atrophy (Figure 2b). Repeat brain MRI in the present admission revealed diffuse cerebral and cerebellar atrophy with hypointensity in the thalamus and a few white matter changes in the bilateral frontal area suggestive of a neurodegenerative disorder (Figure 2c).

Based on the clinical history of seizure and rapid deterioration of vision resulting in blindness with classic fundus changes and collaborating MRI changes, and the background of similar symptoms in the elder brother with a fatal outcome, a diagnosis of juvenile neuronal ceroid lipofuscinoses was considered. The patient was started on sodium valproate and clobazam and the seizures were controlled. He was discharged after a week and is being followed up routinely. Genetic studies could not be performed because the tests are not available in our institution and patient's family could not afford them.

Juvenile neuronal ceroid lipofuscinoses is inherited as an autosomal recessive trait and occurs because of mutations of the *CLN3* gene located on chromosome 16 (1). It is characterized by abnormal accumulation of certain fatty, granular substances such as ceroid and lipofuscin within neurons of the brain, as well as other tissues of the body (2). Neuronal degeneration commences in the dendritic tree and proceeds to final neuronal loss, which predominates in cortical regions of the cerebrum and cerebellum, resulting in progressive atrophy of the brain and neurologic impairment. The presence of vacuolated lymphocytes on peripheral smear, bull's eye maculopathy on

fundus examination, and progressive brain atrophy on MRI are characteristic (3,4).

Ethics

Informed Consent: An informed consent form was taken from the patient.

Peer-review: Peer-review: Internally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices: Deepak Jain, H.K. Aggarwal, Pulkit Chhabra, Avinash Rao, **Concept:** Deepak Jain, H.K. Aggarwal, **Design:** H.K. Aggarwal, **Data Collection or Processing:** Deepak Jain, Avinash Rao, **Analysis or Interpretation:** Deepak Jain, H.K. Aggarwal, **Literature Search:** Pulkit Chhabra, Avinash Rao, **Writing:** Pulkit Chhabra, Avinash Rao, Deepak Jain.

Conflict of Interest: No conflict of interest was declared by the authors.

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