Fairooz P. Manjandavida, Bangalore, India
Carol L. Shields, Philadelphia, USA

Peripapillary hypopigmented lesion in an infant

Case

A 1-year-old girl child suffering from infantile spasms was referred for fundus evaluation from department of pediatrics. The baby was a second offspring for the parents, born at full term by C-section, and assessed to have delayed developmental milestones. The child was following light and objects with both eyes. Ocular examination revealed normal anterior segments and reactive pupils in both eyes. Dilated fundus examination revealed anomalous disc with peripapillary atrophy and multiple hypopigmented lesions of <1 disc area size surrounding the disc in right [Fig. 1a] more than left eye [Fig. 1b].

What is your next step

A. Visual evoked potentials and electroretinogram
B. Laser delimitation of the chorioretinal lacunae
C. Magnetic resonance imaging (MRI) brain and pediatric neurology referral
D. Fundus fluorescein angiography.

Correct answer: C.

Findings

MRI brain revealed agenesis of corpus callosum with dilatation of the occipital and frontal horns of the lateral ventricles [Fig. 1c and d]. Electroencephalography (EEG) also revealed abnormal patterns. Aforementioned investigations, along with infantile spasms and chorioretinal lacunae on fundus examination completed the triad of Aicardi syndrome. The child was advised refraction and periodic fundus examination. Infantile spasms were managed by pediatric neurology team with oral steroids and antiepileptics. The child was also advised exercises and therapy for gait disorder.

Diagnosis

Aicardi syndrome

Discussion

Aicardi syndrome consists of triad of infantile spasms, agenesis of corpus callosum, and chorioretinal lacunae.[1] It predominantly affects girls with a prevalence of approximately 1 in 1,00,000. The mode of transmission is X-linked dominant pattern. Hallmark ophthalmologic feature is the presence of chorioretinal lacunae. Associated ocular features can be microphthalmos, persistent pupillary membrane, iris synechiae, cataract, dysplasia/coloboma of the optic nerve, anomalous retinal vessels, and retinal detachment.[2] Affected individuals can have developmental delay, intellectual disability, and skeletal abnormalities. They are also at risk of developing tumors, such as choroid plexus papillomas, arachnoid cysts, palatal hemangioma, angiosarcomas, and hepatoblastomas.

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.

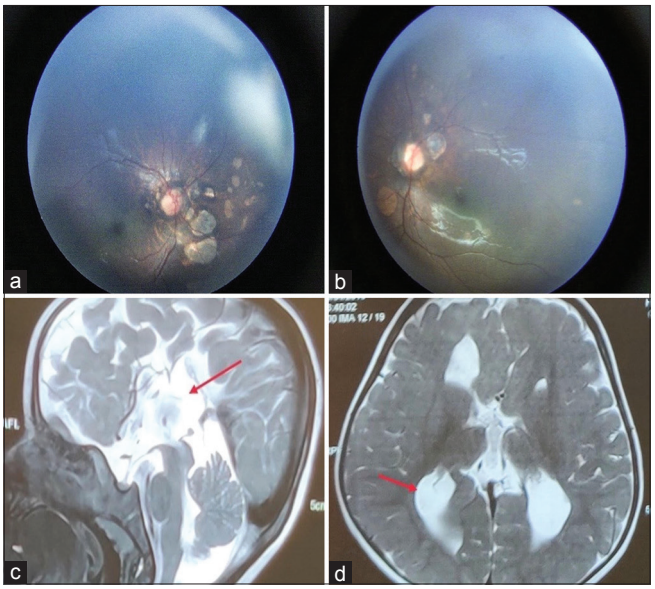

Figure 1: Fundus examination showing anomalous disc with peripapillary atrophy and chorioretinal lacunae around the disc in OD (a) and OS (b). T2-weighted MRI brain images: Sagittal view (c) showing agenesis of corpus callousum, axial view (d) showing widely spaced frontal horns with dilated trigones of lateral ventricles/ colpocephaly-racing car sign (arrow)

References

1. Aicardi J. Aicardi syndrome. Brain Dev 2005;27:164-71.
2. Fruhman G, Eble T, Gambir N, Sutton V, Van der Veyver I, Lewis R. Ophthalmologic findings in Aicardi syndrome. J AAPOS 2012;16:238-41.

Tejaswini Vukkadala, Shorya V Azad, Vinod Kumar

Vitreoretina Services, Dr. R. P. Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India

Correspondence to: Dr. Shorya V Azad, Vitreoretina Services, Dr. R. P. Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, Ansari Nagar, New Delhi - 110 029, India.
E-mail: shoryaazad@hotmail.com

Access this article online

Quick Response Code:

Website: www.ijo.in

DOI: 10.4103/ijo.IJO_1885_19

Cite this article as: Vukkadala T, Azad SV, Kumar V. Peripapillary hypopigmented lesion in an infant. Indian J Ophthalmol 2020;68:692.