

Retinal detachment with spontaneous dialysis of the ora serrata in a 13-year-old child with neurofibromatosis type 1: A case report

Access this article online	
Quick Response Code:	Website: www.ijo.in
	DOI: 10.4103/ijo.IJO_1895_19

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Received: 22-Oct-2019

Revision: 25-Dec-2019

Accepted: 26-Jan-2020

Published: 25-Jun-2020

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A 13-year-old child diagnosed with neurofibromatosis type 1 who on a routine control presented with rhegmatogenous retinal detachment associated to dialysis of the ora serrata in the left eye (OS). There were no clinical signs or history of contuse ocular trauma. Neurofibromatosis produces alterations in fibroblasts of the cortex of the vitreous base. This results in deficient production of the collagen fibers that anchor the vitreous base to the pars plana and the peripheral neurosensory retina. Thus,

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Cite this article as: Clemente-Tomás R, Ruíz-del Río N, Gargallo-Benedicto A, García-Ibor F, Hervas-Hernándis JM, Duch-Samper AM. Retinal detachment with spontaneous dialysis of the ora serrata in a 13-year-old child with neurofibromatosis type 1: A case report. *Indian J Ophthalmol* 2020;68:1473-5.

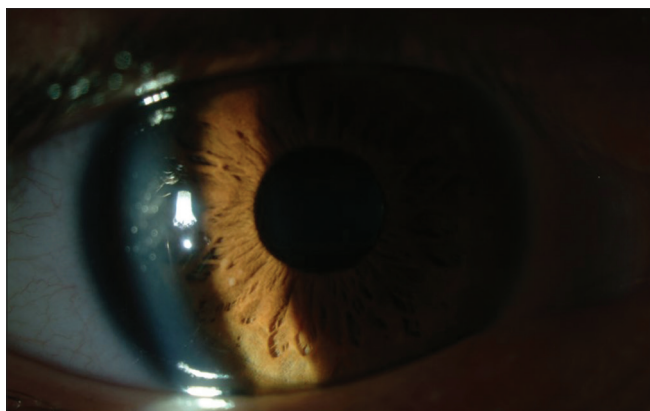


Figure 1: Anterior segment color photograph of the right eye showing iris Lisch nodules

suboptimal function of the fibroblasts explains spontaneous avulsion of the vitreous base. Such avulsion in turn is related to dialysis of the ora serrata.

Key words: Dialysis of the ora serrata, fibroblasts, neurofibromatosis type 1, rhegmatogenous retinal detachment, vitreous base

Neurofibromatosis type 1, also known as Von Recklinghausen's disease, is a multisystemic neurocutaneous disorder with an autosomal dominant hereditary pattern. It can present a great variety of ophthalmic manifestations such as iris Lisch nodules, neurofibromas of the ciliary body and choroid, optic nerve gliomas, sphenoid bone dysplasia, and plexiform neurofibroma of the eyelid. Retinal and choroid lesions are considered to be less frequent. Unusual ocular findings include multiple choroid nevi, combined hamartoma, choroid Schwannoma, myelinated nerve fibers, choroid melanoma, vasoproliferative retinal tumor, etc.

Case Report

We describe the case of a 13-year-old child diagnosed with neurofibromatosis type 1 and subjected to annual controls in the ophthalmology department. On occasion of a routine control, the patient presented with macula on rhegmatogenous retinal detachment associated to dialysis of the ora serrata in the left eye (OS). There were no clinical signs or history of contuse ocular trauma. The patient had no symptoms of any kind, and his best corrected visual acuity (BCVA) was 20/20 in both eyes (OU). The autorefractor (AR) reading under cycloplegia was +0.5 in the right eye (OD) and +1 in OS. Slit-lamp exploration revealed iris Lisch nodules OU [Fig. 1]. Ocular funduscopy following pupil mydriasis proved normal in OD, but OS showed nasal retinal detachment with dialysis of the ora serrata from the 7 to the 12 o'clock position [Fig. 2]. Upon repeated questioning, the patient again denied any history of trauma. The exploration yielded no clinical evidence suggestive of trauma. The patient underwent 23-G pars plana vitrectomy, scleral buckling, peripheral cryotherapy and endophotocoagulation.

Discussion

The vitreous humor is a clear material occupying approximately 80% of the volume of the eye. It is composed of collagen,

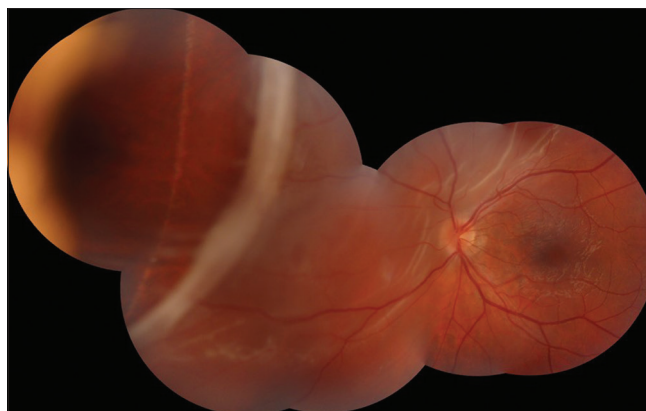


Figure 2: Posterior segment color photograph of the left eye showing nasal retinal detachment with dialysis of the ora serrata from the 7 to the 12 o'clock position. Retina is dis-inserted from the ora serrata and vitreous remain attached to the posterior margin of the break. There is no inversion of the posterior margin of the dialysis

hyaluronic acid, and water. The vitreous base is the region extending from 2 mm anterior to 3 mm posterior to the ora serrata. The cortex of the vitreous base contains numerous cells, almost all of which are fibroblasts.^[1] For this reason the collagen fibers are particularly dense in this region.

Neurofibromatosis has been shown to affect the fibroblasts, causing slow growth of the latter, with a lesser response to epidermal growth factor (EGF) and an abnormal morphology compared with healthy fibroblasts.^[2,3]

Our patient presented rhegmatogenous retinal detachment with spontaneous dialysis of the ora serrata in the absence of a history of trauma. The ocular examination revealed no evidence of anterior contuse trauma. Exhaustive exploration of the retinal periphery was carried out, and the existence of retinal degeneration or tumor disease capable of accounting for dialysis of the ora serrata was discarded. The medical history only reflected neurofibromatosis type 1.

Although spontaneous dialysis of the ora serrata has not been previously described in patients with neurofibromatosis, spontaneous avulsion of the vitreous base has been reported in a patient diagnosed with neurofibromatosis.^[4]

The existence of dialysis of the ora serrata is regarded as avulsion of the posterior margin of the vitreous base.^[5] Neurofibromatosis produces alterations in fibroblasts of the cortex of the vitreous base. This results in deficient production of the collagen fibers that anchor the vitreous base to the pars plana and the peripheral neurosensory retina. Thus, suboptimal function of the fibroblasts explains spontaneous avulsion of the vitreous base. Such avulsion in turn is related to dialysis of the ora serrata.^[6]

Conclusion

Our case emphasizes the importance of conducting annual ophthalmologic examinations in all patients diagnosed with neurofibromatosis type 1. The examination should include fundus under pupillary dilation and will be performed even if the patient is asymptomatic and has a visual acuity of 20/20.

In this way we will rule out retinal alterations related to neurofibromatosis that may go unnoticed.

To the best of our knowledge, this is the second reported case of spontaneous anomalies of the vitreous base in the context of neurofibromatosis.

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.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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