Familial exudative vitreoretinopathy in a patient with choroidal coloboma

Vinod Kumar, Srikanta Kumar Padhy

Ophthalmology, Dr Rajendra Prasad Centre for Ophthalmic sciences, AIIMS, New Delhi, Delhi, India

Correspondence to

Dr Vinod Kumar,
drvinod_agg@yahoo.com

Accepted 30 January 2019

DESCRIPTION

A 10-year-old boy presented with diminished vision in his right eye noted for 1 month. There was no history of prematurity. Family and systemic history was unremarkable. Best-corrected visual acuity (BCVA) was 20/120 in the right eye with +6 dioptres sphere and 20/40 in the left eye with +2 dioptres cylinder at 180°. Anterior segments were normal apart from the presence of inferonasal iris coloboma in both the eyes. The dilated fundus examination showed type III choroidal coloboma (Ida-Mann classification) in the right eye and type II choroidal coloboma in the left eye (figure 1A,B). Both eyes also had peripheral avascular retina separated by demarcation lines (arrows, figure 1A,B). Ultra-wide field fluorescein angiography (figure 1C,D) of both eyes revealed straightening of vascular arcades, supernumerary vascular branching and temporal avascular retina in both the eyes. A closer look at the macula revealed subfoveal choroidal neovascularisation (CNV, figure 2A), which was confirmed on swept source optical coherence tomography (figure 2A,B) and fluorescein angiography (figure 1C, arrow). A diagnosis of both eye choroidal colobomata with familial exudative vitreoretinopathy (FEVR) with right eye CNV was made and intravitreal bevacizumab was advised in the right eye.

At 1 month after intravitreal bevacizumab, the CNV had regressed (figure 2C,D) and BCVA improved to 20/60. The parents declined further injections and was advised regular follow-up. The visual acuity was

figure 2 Colour fundus photograph (A) and swept source optical coherence tomography (SS-OCT) (B) show choroidal neovascularisation at the macula of the right eye. At 1 month after intravitreal bevacizumab injection, fundus photograph and SS-OCT (C,D) show resolution of intraretinal and subretinal fluid.

maintained at 6 months follow up with no signs of recurrence.

Familial exudative vitreoretinopathy is a bilateral asymmetric ocular hereditary disorder. It is characterised by peripheral avascular retina, neovascularisation and retinal detachment. About 50% of the cases can be linked to four causative genes *FZD4*, *NDP*, *LRP5*, and *TSPAN12* which affect 'wnt' signalling pathway.¹ Choroidal coloboma on the other hand occurs as a result of the failure of the embryonic fissure to close² which can occur due to various genetic defects and environmental factors. The coloboma gene network developed from the genes associated ocular coloboma has sonic hedgehog and paired box 6 genes in the pivotal role.³ However, disruption of 'wnt' signalling pathway could also cause coloboma.⁴ An interaction of these genes may explain the coexistence of the FEVR with coloboma. CNV is a known rare complication associated with choroidal coloboma.⁵ This case highlights the occurrence of these two anomalies in a single patient which, to the best of our knowledge, has not been reported before.

figure 1 UWF pseudo-colour photograph of the right (A) and the left eye (B) showing type-3 and type-2 choroidal coloboma respectively. The blue arrow marks line separating avascular peripheral retina from vascular retina. UWF fluorescein angiography (C,D) revealed straightening of vascular arcades, supernumerary vascular branching and temporal avascular retina consistent with familial exudative vitreoretinopathy. Arrow in (C) shows choroidal neovascularisation.

© BMJ Publishing Group Limited 2019. No commercial re-use. See rights and permissions. Published by BMJ.

To cite: Kumar V, Padhy SK. *BMJ Case Rep* 2019;**12**:e228711. doi:10.1136/bcr-2018-228711

Learning points

- ▶ Ultra-wide field angiography is useful for imaging of the peripheral retina, and it is particularly useful for providing panoramic images of the retina with a single click.
- ▶ Familial exudative vitreoretinopathy may also be associated with coloboma of the fundus.

Ultra-wide field angiography is useful for imaging of the peripheral retina, and it is particularly useful for providing panoramic images of the retina with a single click. This is especially relevant in paediatric patients⁶ or patients with poor vision, where creating a montage image may be time-consuming and a source of discomfort to the patient.

Contributors VK, SKP: acquisition of data, concept, writing manuscript, review.

Funding The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.

Competing interests None declared.

Patient consent for publication Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Poulter JA, Davidson AE, Ali M, *et al*. Recessive mutations in TSPAN12 cause retinal dysplasia and severe familial exudative vitreoretinopathy (FEVR). *Invest Ophthalmol Vis Sci* 2012;53:2873–9.
- 2 Pagon RA. Ocular coloboma. *Surv Ophthalmol* 1981;25:223–36.
- 3 Moosajee M, Gregory-Evans CY. Advances in the molecular genetics of ocular coloboma. *Expert Rev Ophthalmol* 2006;1:209–27.
- 4 Zhou CJ, Molotkov A, Song L, *et al*. Ocular coloboma and dorsoventral neuroretinal patterning defects in Lrp6 mutant eyes. *Dev Dyn* 2008;237:3681–9.
- 5 Bhende M, Suganeswari G, Gopal L, *et al*. Choroidal neovascularization associated with coloboma of the choroid: a series of three cases. *Indian J Ophthalmol* 2011;59:148–51.
- 6 Kang KB, Wessel MM, Tong J, *et al*. Ultra-widefield imaging for the management of pediatric retinal diseases. *J Pediatr Ophthalmol Strabismus* 2013;50:282–8.

Copyright 2019 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <https://www.bmj.com/company/products-services/rights-and-licensing/permissions/>
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow