Unilateral focal choroidal excavation in cone dystrophy

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DESCRIPTION

An 18-year-old man presented with gradual and painless loss of vision in both eyes (BE) since the last 2 years. The symptoms were worse in day light and less in dim light. Best corrected visual acuity was 20/160, N 24 with a spherical equivalent of −6 D in BE. Colour vision showed complete red-green deficiency on Ishihara’s test. Anterior segments and intraocular pressures were normal. Fundus examination of BE revealed scleral crescent at temporal disc margin, prominent choroidal vessels at posterior pole and symmetric well circumscribed atrophic lesions in both macula (figure 1A,B). Right eye (RE) additionally showed thinning in central macula bordered by hyperpigmentation. Fundus autofluorescence (AF) showed absence of flecks, hypo-AF centrally in BE corresponding to the fundus lesions, but much more prominent in the RE. There was a ring of hyper-AF surrounding these lesions (figure 1C,D). Optical coherence tomography (OCT) of RE showed central conforming subfoveal focal choroidal excavation (FCE). Outer retinal atrophy was noted involving the ellipsoid zone of BE with hyper-reflective areas below the external limiting membrane in BE (figure 2). Full field electroretinogram (ERG) of BE showed intact scotopic 3.0 responses, with absent single flash photopic 3.0 response (figure 2). Visual field tests for the central 10 degrees were not reliable due to excessive fixation losses in BE, though right eye showed a centre involving scotoma while BE had higher mean deviation thresholds (p < 1%). The patient was diagnosed to have BE generalised cone dystrophy and high myopia with FCE in RE.

On leading questions, one of the four siblings of the patient (elder sister) was found to have similar visual difficulties. Two preceding generations did not have known ocular complaints. The sister on examination was found to have phenotypic features of cone dystrophy similar to the index case, but without FCE. Genetic evaluation was declined by them. Counselling about fair prognosis of the condition, need for monitoring due to risk of choroidal neovascular membrane (CNVM) and close follow-up was informed to the patient.

FCE is a relatively newer described association of retinal dystrophies and represents posteriorly excavated choroid without any staphyloma or scleral changes. It can be due to failure of embryonic choroidal development or due to sequelae of any chronic choroidal pathology that ensues atrophy.
FCE was found to be associated with macular dystrophies like Best disease in two large retrospective series.\textsuperscript{1,2} It may be present in up to 6% of cases of Best disease, and the vitelliform deposits and fibrotic pillars have been considered to be the aetiology in such cases.\textsuperscript{1} FCE may also rarely accompany Stargardt disease and Pattern dystrophy.\textsuperscript{2} Diagnosing FCE by careful clinical evaluation of macula and OCT becomes relevant as these cases are prone to development of CNVM.\textsuperscript{2}

Presence of FCE in cone dystrophy is an unexpected finding as choroidal changes are generally absent in this disease. Cone dystrophy typically involves only the outer retina while sparing the choroid.\textsuperscript{3} Myopia is a known association of FCE.\textsuperscript{2} To our knowledge, there is a single report of FCE in cone dystrophy prior to the current case. Roy et al have reported bilateral FCE in a case of cone dystrophy, but refractive error was not mentioned.\textsuperscript{3} In our case only one eye was affected though high myopia was in both eyes. Further, FCE was absent in his sibling with cone dystrophy. For these reasons, FCE appears to be a coincidental finding rather than a resultant of cone dystrophy in our case.

Learning points

► Focal choroidal excavation can be a coincidental occurrence in cases of cone dystrophy.
► Optical coherence tomography imaging is of essence in diagnosis of such cases. These patients should be monitored closely for complications like choroidal neovascular membrane.

Contributors

AB: Collected images of fundus photo, autofluorescence images, optical coherence tomography and electroretinogram. BT: Managed the case and writing manuscript. RN: Writing manuscript. SJ: Writing manuscript.

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.

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