Multimodal imaging in a case of Kranenburg syndrome

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DESCRIPTION

A man in his early 30s presented with a gradual and progressive decrease in vision in his left eye (LE) for the last year. The patient gave a history of retinal laser being performed 1 year previously, for which the records were not available but reported no improvement post-treatment. The best corrected visual acuity was 4/60 and the intraocular pressure was 12 mm Hg. The anterior segment examination was normal. Posterior segment examination revealed an oval, greyish crater-like depression in the temporal aspect of the optic nerve head and a large serous detachment at the posterior pole. Multimodal imaging was performed for a detailed evaluation of the case.

Multicolour imaging (MCI) of the LE (figure 1A) revealed a well-demarcated area in the temporal aspect of the optic nerve head, which was darker than the rest of the neuroretinal rim and was suggestive of an optic disc pit (ODP). Additionally, a large area of greenish hue at the macula corresponded to the area of retinal elevation, with a central orangish hue corresponding to serous retinal detachment.

Fundus autofluorescence (FAF) clearly highlighted two rows of circumferentially oriented

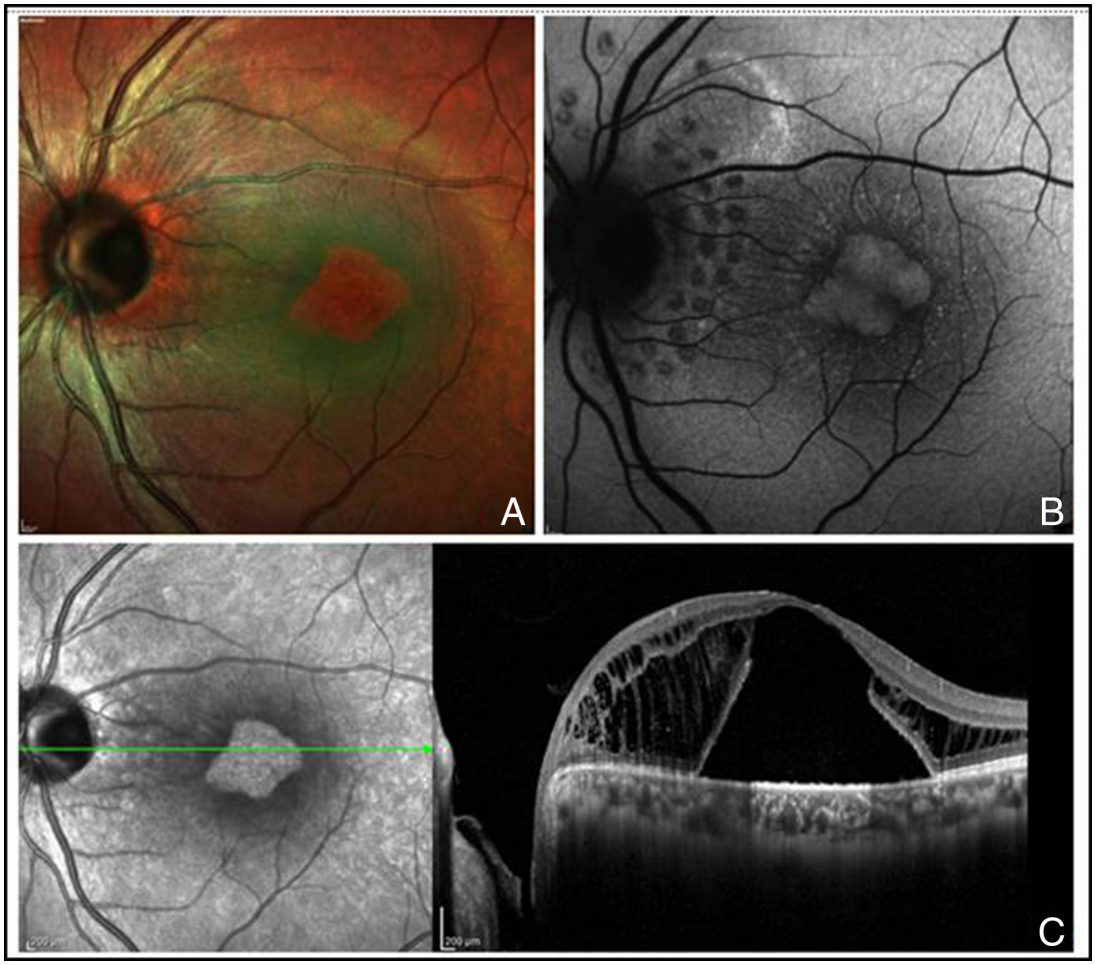

Figure 1 (A) Multicolour image showing a well-demarcated dark black area in the temporal aspect of the optic nerve head suggestive of an optic disc pit. The macular area appeared green due to retinal elevation with a central orangish hue due to increased light transmission from the overlying thinned-out retina and underlying subretinal fluid. (B) Fundus autofluorescence showing hypoautofluorescent lesions circumferentially around the optic disc suggestive of previous laser marks. Additionally, subretinal fluid and intraretinal schisis fluid gave hyperautofluorescent signals. (C) Spectral domain optical coherence tomography clearly demonstrates a large area of neurosensory detachment with multilayered intraretinal splitting (retinoschisis).

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peripapillary laser marks, which were seen as hypoautofluorescent marks (figure 1B). These laser marks were not well appreciated clinically but were clearly seen on FAF. They were suggestive of previous laser delimitation of ODP. Additionally, FAF revealed a central hyperautofluorescent zone, followed by a middle zone of hypoautofluorescence and outer radially oriented hyperautofluorescent areas. These corresponded to subretinal fluid at the centre and radially oriented intraretinal schisis cavities in the perifoveal region, respectively. Lack of hypoautofluorescent areas in the macula signifies a healthy Retinal pigment epithelium (RPE).

Spectral-domain optical coherence tomography (SD-OCT) through the fovea revealed a large area of retina elevation corresponding to neurosensory detachment (figure 1C). Additionally, areas of multilayer retinoschisis could be appreciated both on the nasal and temporal aspects of the macula.

Multimodal imaging provided a detailed analysis of ODP maculopathy and a diagnosis of Kranenburg syndrome was made. Kranenburg syndrome is a rare congenital anomaly characterised by the presence of ODP with serous retinal detachment.[1] Disc pit is considered to be a colobomatous abnormality of the optic disc, caused by an embryonic disturbance of the primitive epithelial papilla. The origin of subretinal fluid in ODP is unclear. Potential sources of this fluid include vitreous fluid, cerebrospinal fluid entering via the subarachnoid space, leakage from blood vessels of the optic nerve head or extravasation of fluid from the choroid via Bruch's membrane or peripapillary atrophy.[2] Serous detachment is seen in 25–75% of ODP, and if untreated can have a poor visual prognosis.[3] Treatment options include laser delimitation, pars plana vitrectomy with or without internal limiting membrane peel, macular buckle surgery and performing inner retinal fenestrations.[3]

Though the ODP was appreciated clinically, a detailed evaluation by multimodal imaging (MCI, FAF and OCT) helped us identify previous laser marks and delineate the areas of intraretinal schisis and neurosensory detachment clearly. FAF additionally provided information about the health of the underlying retinal pigment epithelium and the patient was advised to have surgical intervention in the LE.

Learning points

- Kranenburg syndrome refers to the presence of optic disc pit (ODP) associated with neurosensory detachment.
- Multimodal imaging can provide useful clues in detecting ODP, delineating the extent of schisis and neurosensory detachment and determining the health of underlying RPE.
- On multicolour image, ODP appears dark black, the retinal elevation appears green and the subretinal fluid with overlying retinal thinning appeared orange.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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REFERENCES

1 KRANENBURG EW. Crater-like holes in the optic disc and central serous retinopathy. *Arch Ophthalmol* 1960;64:912–24.
2 Moisseiev E, Moisseiev J, Loewenstein A. Optic disc pit maculopathy: when and how to treat? A review of the pathogenesis and treatment options. *Int J Retina Vitreous* 2015;1:13.
3 Rizzo S, Belting C, Genovesi-Ebert F, *et al*. Optic disc pit maculopathy: the value of small-gauge vitrectomy, peeling, laser treatment, and gas tamponade. *Eur J Ophthalmol* 2012;22:620–5.

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