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Cases

Centre for Eye Health

CFEH Facebook Case #3

A 7 year old Chinese boy was referred to the Centre for investigation of an unusual macula appearance. His maternal uncle has had very poor vision for most of his life. Both eyes showed a similar appearance with VA's of 6/60. Retinal photos, Optomap images and OCT images of the left eye are shown below.

What is your diagnosis? How would you manage this case?

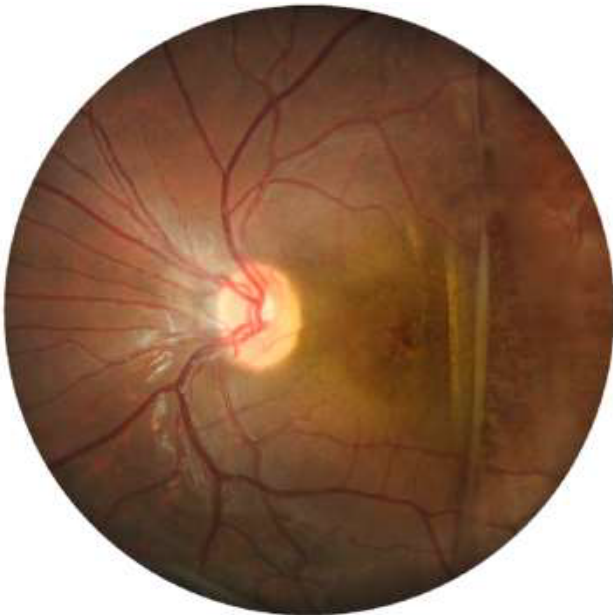


Image 1 Left posterior pole photograph

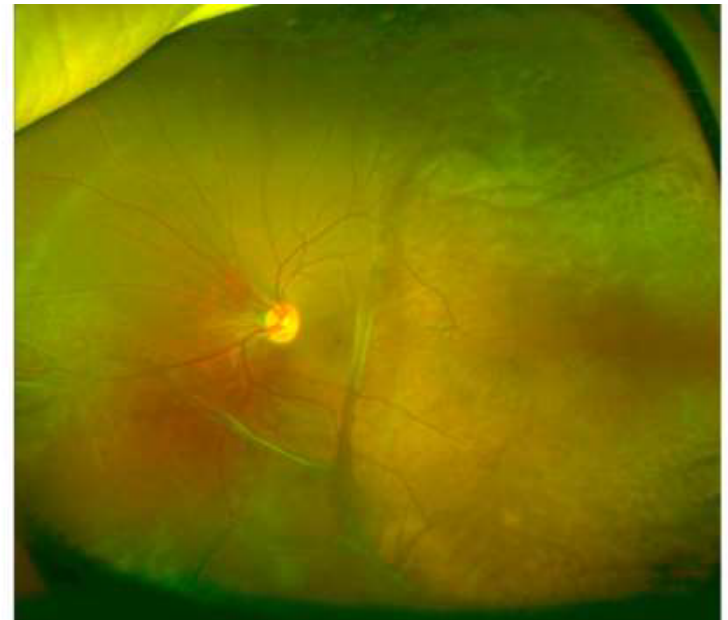


Image 2 Left Central Optomap Image

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An initiative of Guide Dogs NSW/ACT and The University of New South Wales

ABN 85 132 812 143

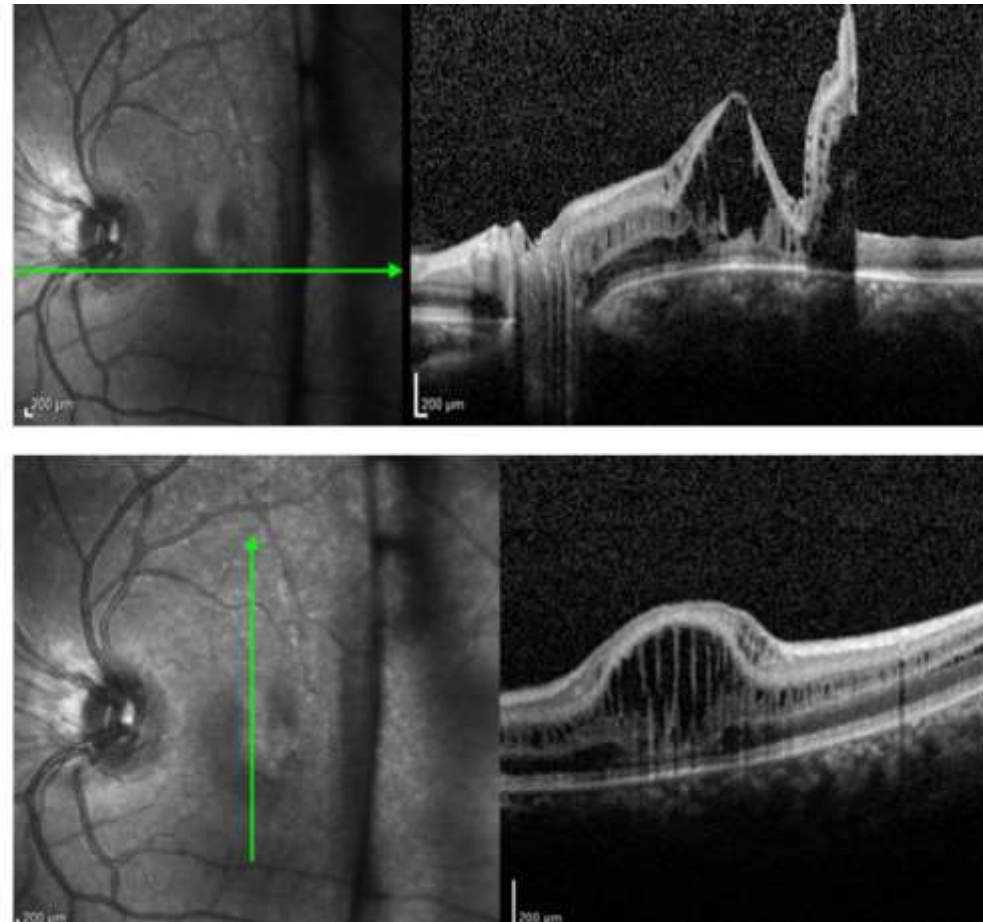


Image 3 Macula OCT Line Scans

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ANSWER

X-linked Juvenile Retinoschisis

Visual acuity can range from 6/6 to light perception. VA typically reduces during first 2 decades with a then slow decline until the 5th and 6th decades. Foveal changes are seen in all cases and peripheral schisis in approximately half. Funduscopically, the macula often shows folds radiating out from the foveolar.

The macula changes associated with x-linked juvenile retinoschisis are best viewed with OCT. This patient's images show characteristic cystic spaces within the retina in the inner nuclear layer and the outer nuclear/outer plexiform layer which is typical of this condition. There is a marked elevation of the retina adjacent to the fovea extending to the inferior retina known as a vitreous veil. These can lead to retinal detachment and vitreous haemorrhage.

A defect in the XLR51 gene is responsible for this condition. This gene encodes an amino acid protein called retinoschisin which mediates the interactions and adhesions between the photoreceptor, bipolar and Muller cells.

Review by a retinal specialist is advised with electrophysiology testing sometimes useful in confirmation of the diagnosis. Prophylactic treatment of the retinoschisis is not generally advised as there is a high risk of retinal detachment, however genetic counselling and low vision aids / management is recommended.