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Case Study

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RETINITIS PIGMENTOSA IN TWO SIBLINGS WITH DIFFERENT PRESENTATION: A CASE STUDY

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ABSTRACT

We report a case of retinitis pigmentosa in two siblings, with inverse retinitis pigmentosa in one and typical retinitis pigmentosa in another. This is a case report of a 10 year boy who presented with diminution of vision in both eyes. On examination he had inverse retinitis pigmentosa. similar complaints were found in his elder sister, 27 yrs of age, who had Advanced Typical Retinitis pigmentosa involving macula. Retinitis pigmentosa in 50–60% of cases has autosomal recessive inheritance; however the type of retinitis pigmentosa in the siblings is generally same.

KEYWORDS: Retinitis Pigmentosa, Inverse Retinitis Pigmentosa, Pericentric Retinitis Pigmentosa.

INTRODUCTION

Retinitis pigmentosa is the most common hereditary dystrophy of fundus.[1,2,3] Around 1.5 million people are estimated to be affected all over the world.[4] It can occur either sporadically or hereditary. It has varied inheritance pattern with autosomal recessive being the most prevalent type (50-60%), followed by autosomal dominant (30-40%) and lastly X linked recessive (5-15%).[5,6,7] X linked recessive has the worst prognosis while Autosomal dominant has the best prognosis of disease.[8,9,10,11] It presents usually within first 30 years of life with complaints of nyctalopia and tubular vision with symmetric visual impairment in both the eyes. It is characterized by pallor of the disc, arterial attenuation, and pigmented bony spicules, which typically start from mid periphery and progress towards posterior pole. Electrodiagnostic tests like electroretinogram can detect the disease even before the features appearing clinically.[2,3,12] Majority of RP cases are non syndromic, 20–30% are associated with systemic association termed as syndromic RP.[13] Here, we report a familial case of Retinitis pigmentosa with atypical inverse RP in boy and typical RP in his elder sister.

CASE REPORT

A boy of 10 yrs presented with complaint of gradual painless progressive diminution of vision which was more during night time since childhood. There was a history of similar complaints in his elder sister, who was 27 yrs of age. No any other family member was affected. Systemic examination was within normal limits. On ocular examination his Snellen's visual acuity was 4/60 in right eye (RE) and 5/60 in left eye (LE) and that of his elder sister was RE 6/36p and LE 3/60 with no improvement with pin hole in any of them. On retinoscopy of male child the reflex got neutralized at -5.00 dioptre sphere in both meridians in both the eyes. His sister also showed myopia of around -1.50 dioptre sphere in both the eyes. Horizontal nystagmus was present in boy with alternate divergent squint while left eye was in divergence in elder sister. Anterior segment examination was normal with angles wide open. Fundus examination revealed arteriolar attenuation, waxy pallor of disc and pigmentary bony spicules in the macula of the boy along with few bony spicules in the mid peripheral retina. A small intraretinal haemorrhage was observed at the region of posterior pole in his left eye (Figure 1a and 1b). His elder sister showed presence of bony spicules in the mid peripheral retina, which progressed to posterior pole along with macular scarring. An ill defined yellowish area was seen in posterior pole of the right eye suggestive of choroidal neovascular membrane in right eye.(Figure 2a and 2b) Visual field examination could not be performed due to visual acuity less than 6/60 in both the eyes. On OCT done via Spectralis HD OCT, posterior segment of the boy revealed posterior bowing of the retina in both the eyes suggestive of high myopia. Rest of the foveal profile was normal apart from loss of normal foveal dip. A zone of

hyperreflectivity was seen in left eye suggestive of haemorrhage in posterior pole.(Figure 3a and 3b) OCT of girl child revealed foveal thinning in both the eyes along with hyperreflective area at the level of Bruch's membrane in right eye suspective of choroidal neovascular membrane (CNVM). (Figure 4a and 4b) Fundus fluorescein angiographic images of girl child confirmed the diagnosis of CNVM in her right eye.

DISCUSSION

Retinitis Pigmentosa is a hereditary retinal degeneration involving photoreceptors. Findings in favour of our diagnosis were diminution in vision more in the night, classical triad of the disease in fundus and supportive ancillary investigations like OCT and FFA. Electrodiagnostic tests could not be performed as they were not available. It can be classified on the basis of clinical picture as Typical and Atypical. Typical Retinitis Pigmentosa is characterized by triad of waxy pallor optic disc, generalized arteriolar attenuation and pigmented bony spicules in mid-peripheral retina. Later on these perivascular bony spicules extend both anteriorly and posteriorly.[15] It can also be classified as Simple retinitis pigmentosa or Syndromic Retinitis pigmentosa. Retinitis pigmentosa inversa or pericentric retinitis pigmentosa is an atypical form of Retinitis pigmentosa which is a rare finding.[15] Unlike the typical RP, there is marked diminution of vision in early course of disease as the disease process involves posterior pole and macula from the very beginning.[14] In this case one of the sibling had inverse RP hence explaining the marked diminution of vision at an early age. On the contrary the elder sibling had typical RP that has progressed at a very early stage to involve posterior pole hence deteriorating the central visual acuity. The disease forms a part of various syndromes. Majority of the cases of syndromic RP are autosomal recessively inherited.[15,16] We excluded systemic involvement in our cases by detailed history and systemic examination.

CONCLUSION

Inverse retinitis pigmentosa is a rare form of RP and leads to marked diminution of vision in early stage of disease. In this case two siblings have different forms of retinitis pigmentosa hence making it difficult to recognise the genetic basis of inheritance of the disease in two of them.

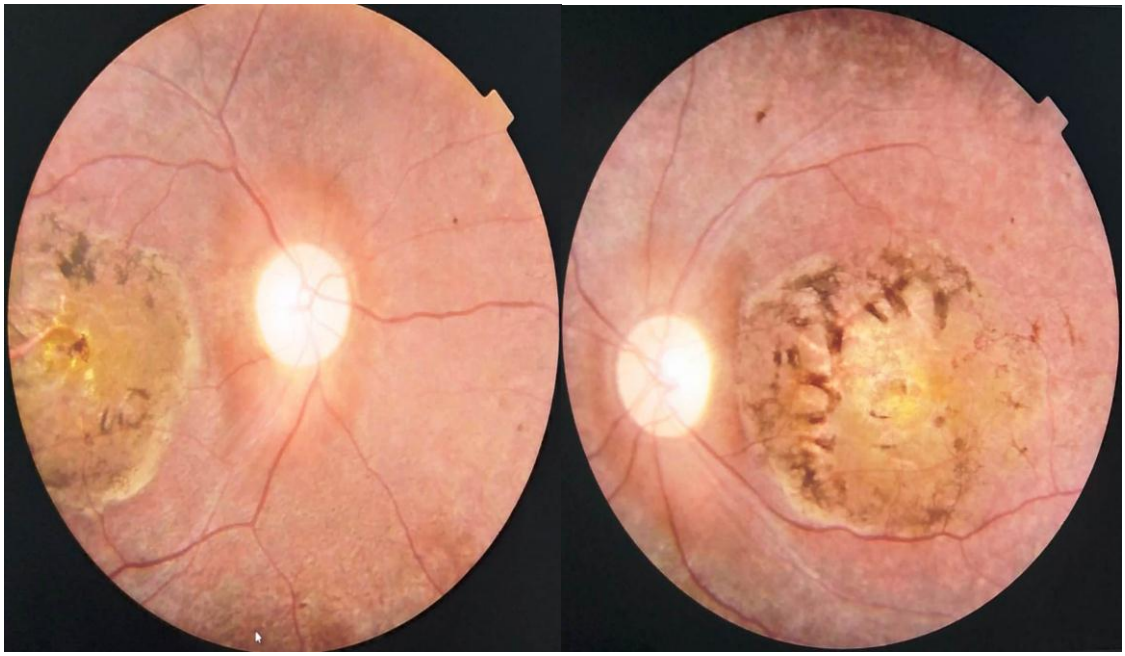

Figure 1: a & b showing right and left fundus images of boy.

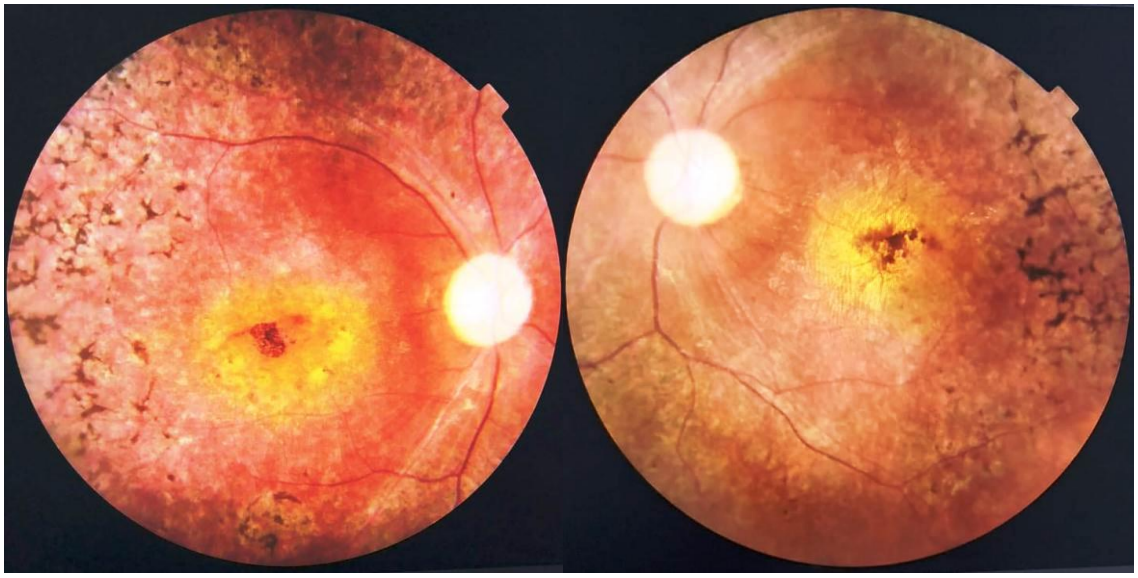

Figure 2: a & b showing right and left fundus images of girl.

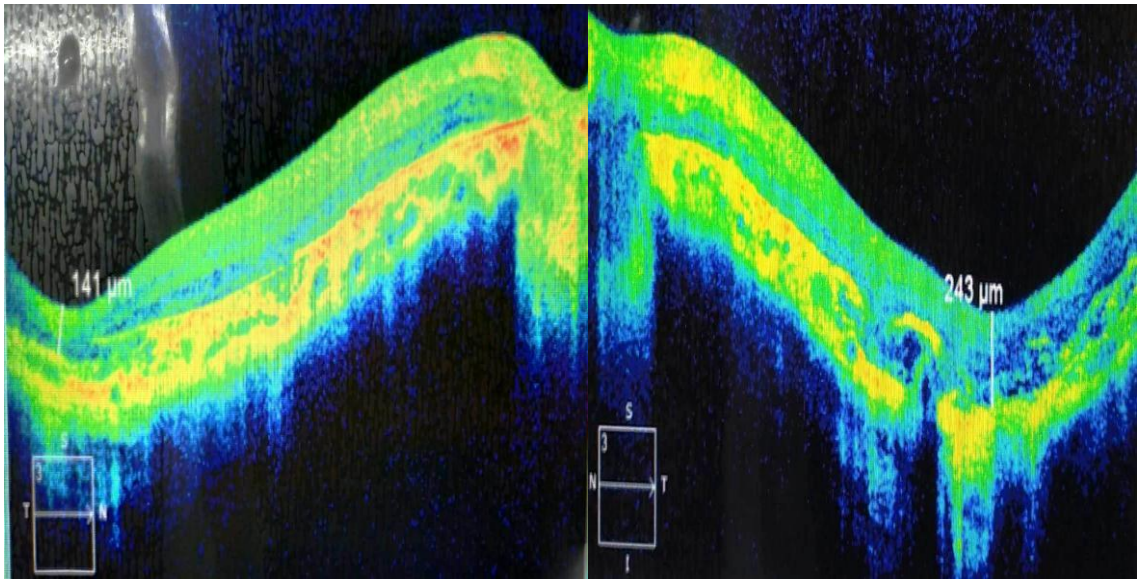

Figure 3: a & b showing OCT images of right and left eye of boy.

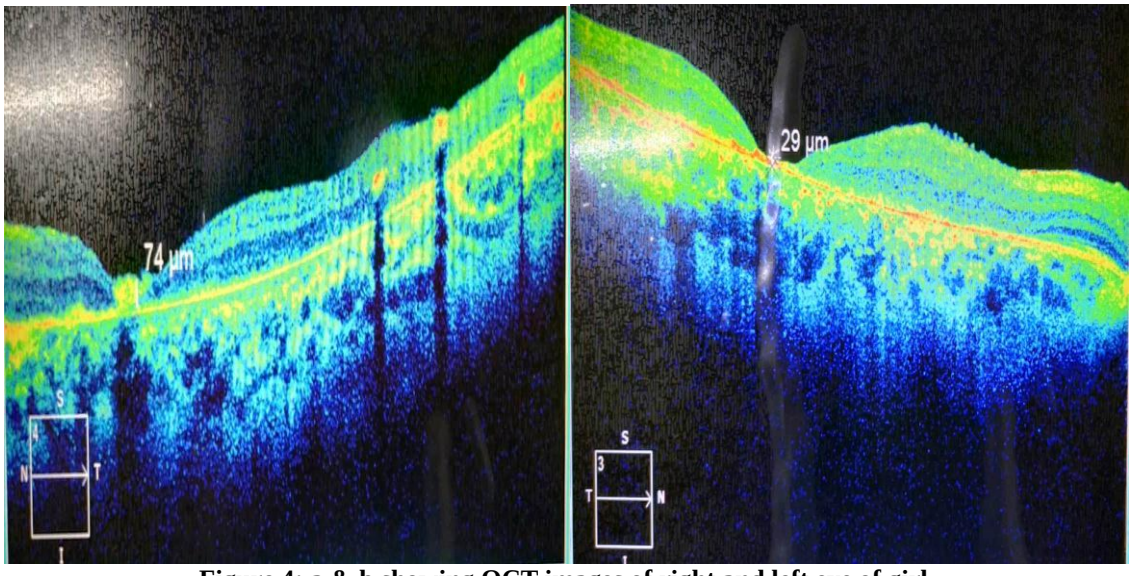

Figure 4: a & b showing OCT images of right and left eye of girl.

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