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Retinitis Pigmentosa

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Abstract:
Background*: To study Retinitis Pigmentosa as a cause of blindness in patients attending MLB medical college and Hospital.*
Methods*: A total of 22 patients of all age groups attending to MLB medicalcollege and Hospital during the period April 2017 to January 2018 were included and examined which include visual acuity testing using Snellen chart, refraction, slitlampbiomicroscopy, intra ocular pressure measurement using Schiotz Tonometer, fundus examination with indirect ophthalmoscope, visual field testing using Humprey field analyser. Fundus picture was taken using Zeiss fundus camera. Patients having retinal pathology were exclusively included in the study and others were excluded.*
Results*: Out of 22 patients of retinitis pigmentosa 14 were males and 8 were females indicating that males were more commonly affected thanfemales.*
Conclusion*: Retinitis Pigmentosa is one among the non treatable cause of blindness which result due to consanguinity marriages as it is an inherited eye disease.*
Keywords*: Retinitis Pigmentosa, blindness, consanguinous marriage, inherited disease.*

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I. Introduction

Retinitis pigmentosa (RP) describes a heterogeneous group of inherited retinal dystrophies characterized by progressive photoreceptor cell degeneration that affects approximately 1 in 4000 in a general population [1]. The genetics of RP is varied; nonsyndromic cases may be inherited as an autosomal dominant (30%), autosomal recessive (20%), Xlinkedrecessive (15%), or sporadic/simplex traits (30%), and 5% may be early-onset and grouped as part of Lebercongenitalamaurosis [2]. Rarer forms also exist: X-linked dominant, mitochondrial, and digenic (due to mutations in two different genes). While RP is a disease usually limited to the eye, it may occur as part of a syndrome; as examples, Usher syndrome and Bardet-Biedl syndrome. Approximately 20%–30% of patients with RP have an associated nonocculardisease and would be classified as having syndromic RP. RP is characterized by progressive degeneration of the retina usually starting in the midperiphery of the fundusand advancing towards the macula and fovea. The most common form of RP is a rod-cone dystrophy in which night blindness is the first symptom, followed by progressive loss of peripheral visual field. Classic clinical findings include: bone spicule pigmentation or pigment clumping, retinal arteriolar narrowing, waxy pallor of the optic nerve, epiretinal membrane formation, atrophy of the RPE and choriocapillaris(starting at the midperipheryof the retina with preservation of the RPE in the macula until late in the disease), posterior subcapsular cataract, epiretinal membrane formation, and cystoid macular edema (CME) [1] (figure1,2,3,4).

II. Materials and Methods

The present study was conducted on 22 patients who attended MLB medical college and hospital during the period April 2017 to January 2018.Patients of all age groups were included in the study. All the patients who were included in the study underwent visual acuity testing using Snellen chart. Distance and nearvisual acuity, both presenting and best corrected after refraction, were measured for each eye separately usingSnellenchart(3). External eye examination, assessment of pupillary reaction, and anterior segment examination were done with slitlampbiomicroscope, intra ocular pressure measurement using SchiotzTonometer. All participantshad their pupils dilated and after dilatation the lens was examined with the slitlamp and cataract was graded Stereoscopic fundus examination, including assessmentof the vitreous, retina, and optic disc, was done with the indirectophthalmoscope using 20 diopter lens. Automated visual fields were done with the Humphrey visual field analyzer(11) using the central 30-2 threshold strategy in those participants assessed to have any suspicionof retinitis pigmentosa. Fundus picture was taken using Zeiss fundus camera in

patients with retinitispigmentosa.Stereoscopic fundus examination, including assessment of the vitreous, retina, and optic disc, was done at the slitlamp using 78 diopter lens and with the indirect ophthalmoscope using 20 diopterlens. Automated visual fields were done with the Humphrey visual field analyzer(4) using the central 24-2 threshold strategy in those participants assessed to have any suspicion of retinitis pigmentosa. Fundus picture was taken using Zeiss fundus camera in patients with retinitispigmentosa.

III. Results

Table 1 : Effect of age and sex

Age	Males	Females	Total
0-10	1	1	2(9.09%)
11-20	2	3	5(22.72%)
21-30	5	2	7(31.81%)
31-40	2	1	3(13.63%)
41-50	1	1	2(9.09%)
>50	3	0	3(13.63%)
	14(63.63%)	8(36.36%)	22 (100%)

IV. Discussion

Blindness defined as presenting visual acuity <6/60 in the better eye; Best corrected visual acuity <3/60and/or less than 10 degree visual field in better eye. Among the retinal causes of blindness, retinitis pigmentosa constitutes one of the non-treatablecauses of blindness. In our study conducted on 22 patients werefound to have retinitis pigmentosa. Of these 22 patients, 14 (63.63%) were males and 8 (36.36%) were femalesindicating that retinitis pigmentosa is more common among males compared to females. 7(31.81%) among 22persons were found between the age group 21-30 indicating that it was the most common age group to beinvolved.A study done on RP patients in various states of India has shown an autosomal-recessive, predominantinheritance pattern, and more than 92% of cases in autosomal-recessive category had positive history ofconsanguinity(5). The patients of AR(D) type of inheritance had characteristic symptoms of headache, giddiness flashes of light and worsening of symptoms after any stress or strain. Similar observations have been reported by others in RP patients.[6],[7],[8],[9] This group of patients could have congenital onset of the disease and also generally developed cataracts, as opposed to AR(R) group where cataracts were a rarity and congenital cases were not seen. The autosomal dominant patients had slowly progressive disease taking over 10-15 years.[10]

V. Conclusion

Retinitis pigmentosa as an inherited disease runs in the families need utmost care to reduce the burdenon prevalence of blindness as it is one among the non-treatable cause of blindness and as there are no specifictreatment modalities to cure. Hence we need to concentrate on genetic counselling to reduce consanguineous marriages.

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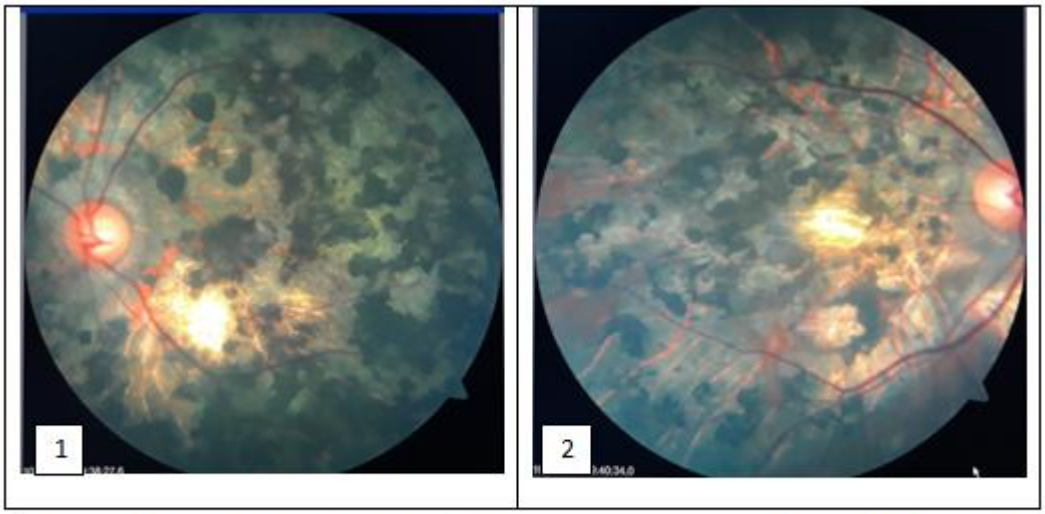

Figure1&2- Fundus photo of right and left eye showing picture of atypical RP(arteriolar narrowing and conglomerate bony spicules in the mid-peripheral region)along with atrophic patch seen over macula.

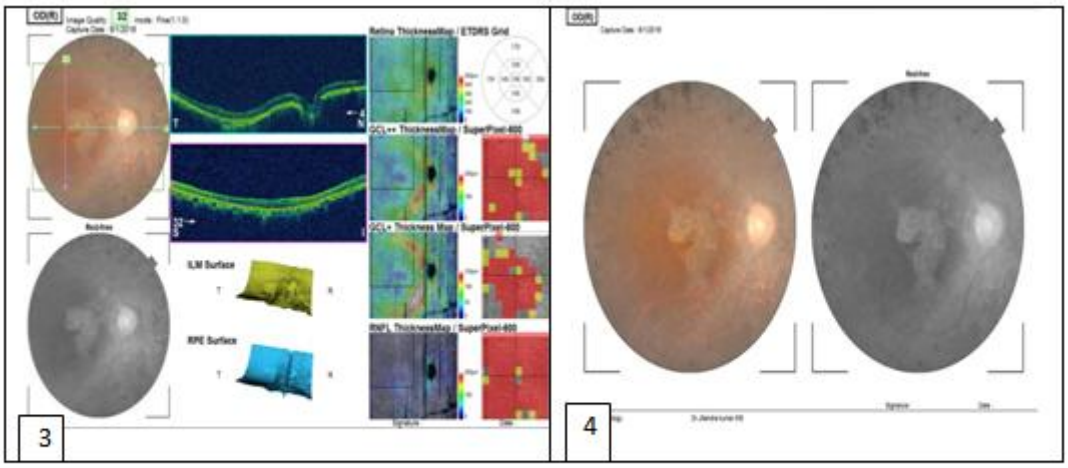

Figure 3&4-OCT image and fundus photo of typical RP showing pale disc, generalized arteriolar narrowing with bony spicules in periphery along with hypopigmented patch over macula.

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