ORIGINAL ARTICLE

Clinical Patterns of Retinitis Pigmentosa with special reference to maculopathy (A hospital based study)

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ABSTRACT

Aim: To describe clinical patterns of Retinitis Pigmentosa with special emphasis on optical coherent tomographic picture of maculopathy.
Study period and design: 2010 to 2014 descriptive, retrospective study.
Methods: We retrospectively reviewed ninety seven patients of Retinitis Pigmentosa (RP), presenting in two different centers of Lahore. The medical charts were analyzed. The data considered for the study was age, sex, ocular signs, symptoms, macular status and associated systemic problems of the patients. Optical Coherent Tomography was used where pattern of maculopathy was not discernable clinically. The patients were divided into three age groups (≤ 19 years, 20 to 40 years, >40 years). Clinical patterns were compared among three groups.
Results: The age range was from 1 to 75 years (mean 29.34). Male to female ratio was 1.2: 1. Family consanguinity was positive in 70.10 % of patients. The initial clinical presentation was night blindness in 89.7 % patients. Intra ocular pressures were normal in all cases. Commonest ocular finding other than typical triad of RP was posterior vitreous detachment (PVD). Usher syndrome was the commonest systemic association. Maximum number of patients had typical RP. Atrophic maculopathy had largest percentage in all groups.
Keywords: Retinitis pigmentosa, atrophic maculopathy, night blindness, bull's eye maculopathy,

INTRODUCTION

Retinitis Pigmentosa (RP) is a group of hereditary disorders characterized by progressive visual field loss and nyctalopia which, can later on involve the central vision. It is not a single disease. There are more than 40 genes for isolated RP and more than 50 different genes for syndromic RP that have been identified. RP has significant phenotypic variations. Even patients with same genetic mutation can present with very different clinical ocular findings. Recently, FDA has approved first retinal implant Argus II retinal prosthesis system for patients with advanced RP[1]. In this article, only clinical patterns of RP are discussed especially with reference to different age groups. Genetic evaluation was not carried out due to lack of facilities.

SUBJECTS AND METHODS

We retrospectively reviewed history charts of patients with RP. They were selected from patients presenting at Ghurki Trust Teaching Hospital and a private clinic from 2010 to 2014. Consent forms were signed by every participating individual. History and examination charts were completed. Visual acuity, intra ocular pressures, slit lamp examination and systemic evaluation was performed. Fundoscopy was done using indirect ophthalmoscopy. Data was compiled, analysed as a whole and then divided into three groups, depending upon age. Group 1 consisted of patients with less than 20 years. Group 2 included patients between 20 and 40 years. Group 3 had patients who were above 40 years of age. The Descriptive Statistics (cross tabulation) was calculated for different clinical entities.

RESULTS

In this series, the age range was from 1 to 75 years (mean 29.34). Male to female ratio was 1.2:1. Family consanguinity was positive in 70.10%. 53% of patients had no family history of RP. Night blindness was presenting symptom in 89.7%. Typical Retinitis Pigmentosa was the major presentation (Table 1). Other clinical findings (ocular and systemic) were compared between the three groups. Details of visual acuity are shown in table 2 & graph 1.

Intra ocular pressures were normal in all subjects. 22(22.68%) had lens opacities, 46(47.42%) presented with posterior vitreous detachment (PVD) and 36.08% cases out of total 97 patients had atrophic maculopathy. Usher's syndrome was the commonest systemic association of RP. For details refer to figures 2,3 and 4.

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Table 1: Different types of RP in different age groups.

Type	Group 1	Group 2	Group 3
Typical	57.7%	70.83%	78.3%
Retinitis punctata albescence	30.77%	12.5%	8.7%
Sine pigmento	11.5%	12.5%	8.7%
Sectoral	0%	2.08%	4.34%

Table 2: Visual acuity in different groups.

Visual acuity	Group 1	Group 2	Group 3
6/12 and better	19%	14.90%	17.40%
FC to 6/18	62%	60.42%	43.50%
Less than FC	19%	25.00%	39.10%

Fig. 1: Graphical representation of Comparison of visual acuity in different groups.

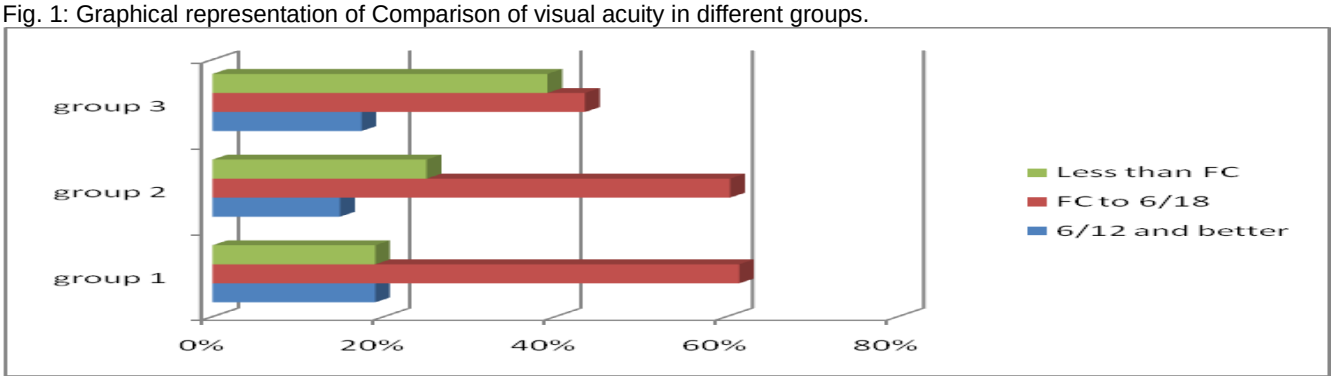


Fig. 2: Ocular and systemic associations of Retinitis Pigmentosa in different groups.

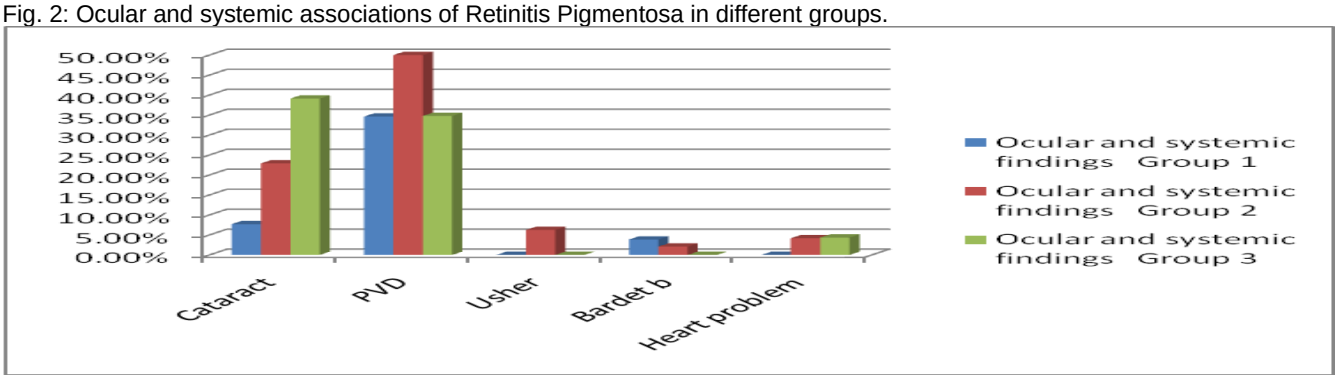


Fig. 3: Percentage of maculopathies

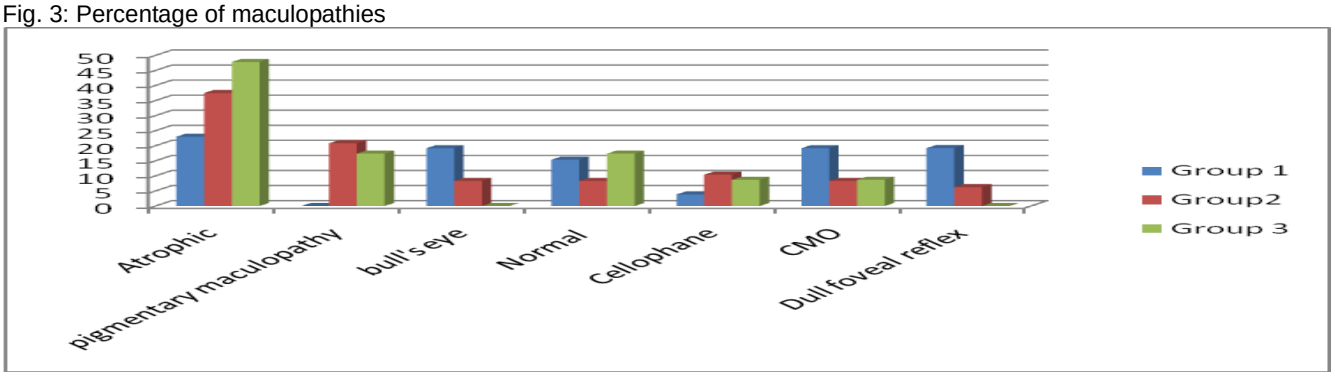


Fig. 4: Cystoid macular edema in a 9 years old child.

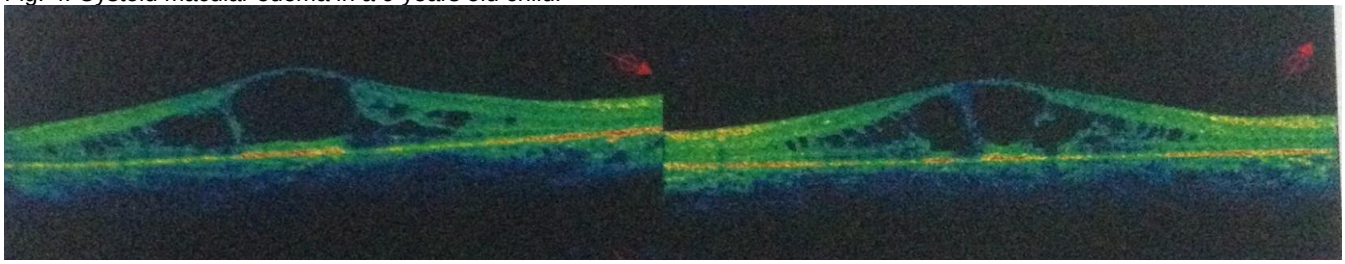


Fig. 5: Surface wrinkling maculopathy in a patient with RP

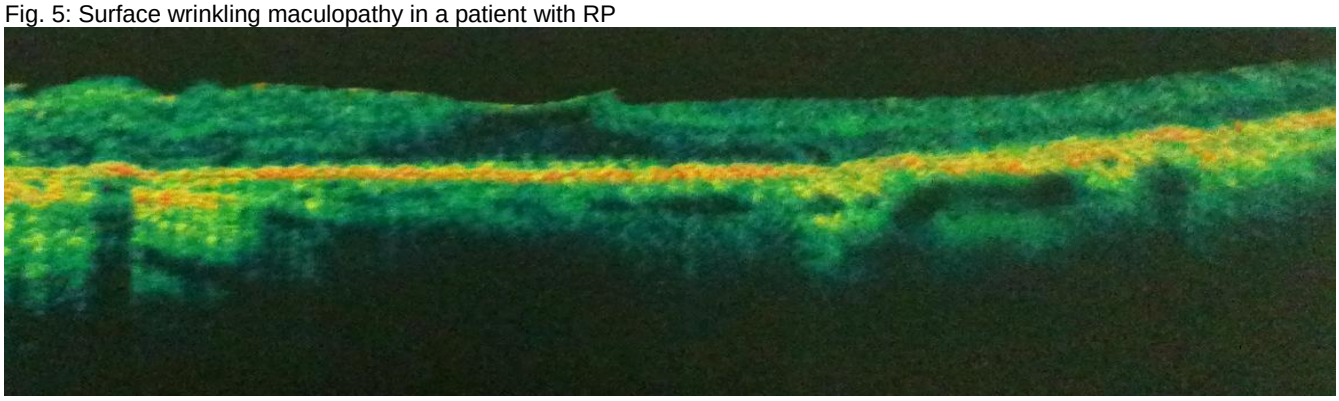
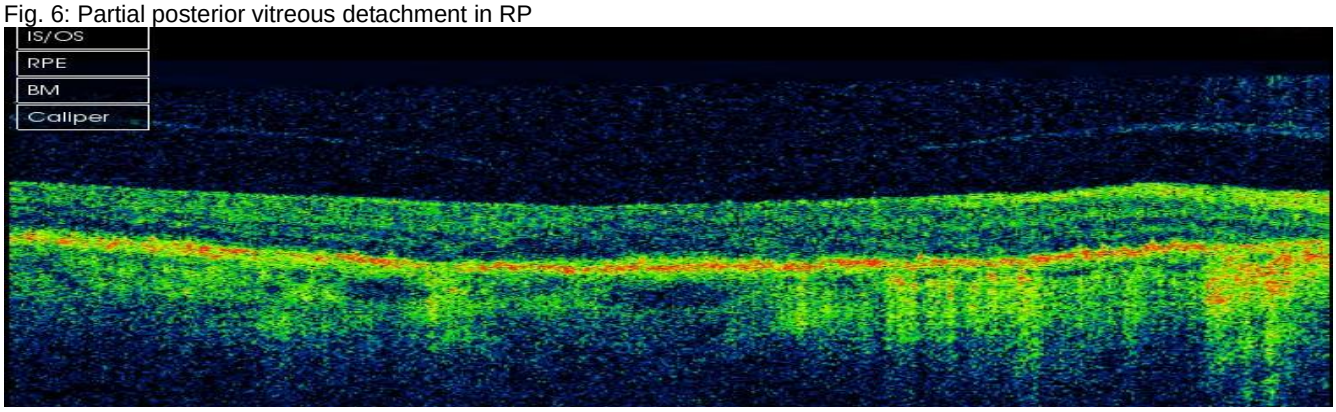


Fig. 6: Partial posterior vitreous detachment in RP



DISCUSSION

RP is a genetical group of diffuse retinal dystrophies initially predominantly affecting the rod photoreceptors with subsequent degeneration of cones. Its worldwide prevalence is approximately 1:5000[2]. The clinical triad of RP includes arteriolar attenuation, bone spicule pigmentation, and waxy disc pallor.

In this particular study, family consanguinity was positive in 70.10 %. It was reported in a previous research that almost 60% marriages in Pakistan were consanguineous. Among those, 80% were between first cousins[3]. This explains high prevalence of this disease in our part of the world. Average age in our study was 29.34 years which was quite similar to another study by Naz et al (average age 25.56 years)[1]. Research by Jamshed Ahmed had shown a mean age of 28.86 years (SD±16.52)[4].

If we analyze international data, it reveals that in Japan average age of Retinitis pigmentosa is 35.1 years[5]. In Nigeria it is 36.7 years[6].

Male to female ratio in our series was 1.2: 1 (54.6% males). Eballe et al had shown equal male to female ratio[7]. Male preponderance was reported in the researches of Kaya-Ganziami et al[8] and Ukponmwan et al[6] in Nigeria, where male predominance of 63.6% and 66.6% respectively had been reported.

Majority of our RP patients (89.7%) had night blindness as a first symptom of disease. Similar findings were depicted in the previous studies[8,9,10].

Retinitis Pigmentosa is not only an isolated disease but there are other systemic associations. In our study, Usher syndrome was the commonest type of syndromic RP. It was seen in 3.09% cases. Earlier studies had estimated that 10–30% of patients with retinitis pigmentosa (RP) also had some form of hearing impairment[11,12]. Hearing loss in Usher syndrome can be variable and is also associated with some vestibular dysfunction[13]. Other syndromes included Bardet biedl syndrome and Kearns Sayre syndrome.

There are different presentations of Retinitis Pigmentosa. Commonest type of RP in our series was Typical RP. Others were Retinitis punctata albescence, sectoral and sine pigmento. Sectoral RP was the least common. PVD was seen in 47.42% patients, even in the younger age group (group 2). Atrophic maculopathy was the commonest cause of severe visual loss. All these findings were consistent with the findings of Jamshed et al[4], with the exception that they had patients with keratoconus and optic disc drusen associated with RP which we did not have in our series.

Macular findings were compared between

different groups. Atrophic maculopathy was the commonest maculopathy in all three groups. Second commonest type was Pigmentary maculopathy in groups 2 and 3. In group 1, Bull's eye maculopathy and cystoid macular edema were second commonest. Such comparison in different age groups is so far not found in the literature.

Role of Optical coherence tomography (OCT) in macular diseases cannot be overlooked. There were patients in which clear categorization of type of maculopathy could not be done. We performed OCT in such cases. For examples refer to figure 4, 5 and 6.

In this patient, clinical picture of macula did not correlate with the fall in visual acuity. OCT showed large cystoids spaces especially the thickness of the neurosensory retina at the center of the fovea. Studies have shown that these cystoid spaces do not directly correspond with the angiographic leakage but there is a very close relation of thickness of neurosensory retina with vision loss[14].

Some researchers have described a measurable degree of RNFL thinning on OCT, in patients with RP[15]. Another feature not seen in our series and described by others is total or nearly total vascular obliteration, and abnormal choroidal vessels[16].

Although a very old hereditary disease, Retinitis pigmentosa is still under study with new technologies emerging day by day. Work on genetic engineering is under way. With the advent of latest diagnostic tools, new doors in studying pathogenesis are opening which will in turn help in treating vision loss in such diseases. OCT is a very good gadget in cases where clinical picture of macula does not correlate with the fall in visual acuity.

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