RETINITIS PIGMENTOSA;
CLINICAL PRESENTATIONS AND ASSOCIATIONS

ORIGINAL
PROF-1807

DR. MOHAMMAD ALAM
Senior Registrar
Department of Ophthalmology
PGMI Lady Reading Hospital
Peshawar

DR. MOHAMMAD ISHAQ
Associate Professor Medicine
K.T.H, Peshawar

ABSTRACT: Objective: To review clinical presentations and associations in patients suffering from Retinitis Pigmentosa. **Design:** Descriptive study. **Period:** January 2005 to December 2009. **Setting:** KTH Peshawar, DHQ Hospital Karak and Group of Teaching Hospital Bannu. **Materials and Methods:** A proper performa was made for documentation of patients. After informed consent, history was taken properly. Ocular examination regarding visual status, anterior and posterior segment examination with direct, indirect ophthalmoscope and slit lamp bimicroscopy was done. IOP was checked with applanation tonometer. For systemic examination and associations opinion of physician was asked if needed. **Results:** Total 83 patients were examined out of which 49(59.03%) were male and 34(40.96%) were female. Regarding age factor 1(1.20%) patients were in age group up to 10 years, 3(3.61%) in age group 11 to 20 years, 12(14.45%) in age group 21 to 30 years, 21(25.30%) in age group 31 to 40 years, 27 (32.53%) in age group 41 to 50 years, 12(14.45%) in age group 51 to 60 years, and 7(8.43%) patients were in age group 61 to 70 years. In 79(95.18%) patients there was family history of R.P while in 4 (4.81%) patients no family history was present. All the patients have involvement of both eyes. 61(73.49%) patients presented with complaints of night blindness, 56(67.46%) patients with defect in visual acuity with range from 6/9 to perception of light, out of which 24 patients had refractive error and 32 patients had no refractive error. 21(25.30%) patients presented with field defect. Fundoscopy revealed, bone spicule pigments configuration in 83(100%) patients, attenuated blood vessels in 23(27.71%) patients and, waxy pallor disc in 12(14.45%) patients. Regarding associations, in systemic association group 82(98.79%) patients had no association while 1(1.20%) patients had bardet biedle syndrome. In ocular association group 24(28.91%) patients had myopia, 14(16.86%) patients had cataract. Primary open angle glaucoma was present in 3(3.61%) patients while 2(2.40%) had cystoid macular oedema. Keratoconus was present in 1(1.20%) patients. **Conclusions:** Retinitis Pigmentosa is a grave disease with irreversible loss of visual functions. Mostly it is bilateral. Severity increases with age. Familial onset is more.

Key words: Retinitis Pigmentosa (R.P), Visual Acuity (V.A), Intra ocular Pressure (IOP), Electroretinogram (ERG)

INTRODUCTION

Donder is generally credited with first describing R.P in 1855 to 1857[1,2]. .Although there were early observations of familial and complicated night blindness by other research workers as well as poor vision and pigmented lesions in the retina like by Schon[3] in 1828 and Von Ammon[4] in 1838. Von Grafe demonstrated the heriditary nature of disease to which he gave the name pigmentary degeneration in[5] 1858. In 1945, Karpe demonstrated that there was an abnormal to non-recordable ERG response in patients with RP and this electrophysiological response occurred in many patients before the appearance of clinical or ophthalmoscopic changes[6].

R.P is a hereditary disease caused by genetic abnormalities in a large number of genes expressed in retinal photo-receptor cells and in retinal pigment epithelial cells. A hallmark of this disease is the slowly progressive degeneration of photo-receptor cells leading to night blindness, visual field constriction and reduction in visual acuity. The prevalence of R.P ranges from 1/3000 to 1/5000[7,8,9] and about 1.5 million people are affected world wide[10].

R.P is the commonest retinal dystrophy affecting young individuals resulting in progressive loss of visual functions and making them visually handicapped. Degeneration in R.P is characterized by pigment deposits predominant in the peripheral retina and by a relative sparing of the central retina. In most cases there is primary degeneration of the photo-receptors rods with secondary involvement of the cones. This sequence of photo-receptors explains why patients initially present with night blindness and only in the later life would suffer from permanent visual loss in diurnal conditions.

Transmission of R.P is genetic and may be by various modes i.e, Autosomal dominant, autosomal recessive or X.linked[11]. In has been reported that autosomal dominant inheritance type, has the least severe impairment while X-linked recessive has the most severe impairment in visual acuity[12,13]. R.P is a leading cause of

blindness and incurable visual impairment owing to its irreversible evolution towards loss of central vision.

R.P has ocular associations like myopia, cataract, open glaucoma, keratoconus, age-related macular degeneration, macular hole etc and systemic associations. Important systemic associations with R.P are usher syndrome[14], bardet biedle syndrome[15]. R.P is mostly bilateral but unilateral cases have also been reported. First unilateral R.P was described by Pedralgia in 1865[16]. However this clinical entity still represents an enigma and its existence and relationship to the bilateral form are debated.

This study was based on the clinical presentations and associations in patients suffering from R.P.

MATERIALS AND METHODS

This study was conducted on patients suffering from R.P from January 2005 to December 2009 in KTH Peshawar, DHQ Hospital Karak and Group of Teaching Hospital Bannu. A proper performa was made for evaluation of the patients. After informed consent proper history was taken. Visual acuity with and without pin hole was checked. Retinoscopy was done in patients who had refractive error. Anterior segment and posterior segment examination were done with direct and indirect ophthalmoscope, slit-lamp bimicroscopy. Intra ocular pressure was checked with applanation tonometer. Perimetry was done with confrontation method. Diagnosis of RP was made on the basis of examination of the fundi using direct and indirect ophthalmoscope and indirect slit lamp bimicroscopy, the criteria being bone spicule pigments configuration, vascular attenuation, and waxy pallor disc. Automatic visual field, retinal angiography, electro-physiological test like ERG and genetic tests were not performed in our study due to inadequate level of technical expertise and facilities. In some patients for systemic involvement opinion of physician was asked.

The variables analyzed were gender, age, familial / non familial, main complaints, intra ocular pressure, ocular and systemic associations.

Regarding age, patients were divided into 7 groups from A to G. Group A patients had age up to 10 years, Group B 11 to 20 years, Group C 21 to 30 years, group D 31 to 40 years, group E 41 to 50 years, group F 51 to 60 years and group G had patients from 61 to 70 years.

It is important to mention that most of the patients had mixed types of clinical presentations and with different findings.

RESULTS

Total 83 patients suffering from RP were examined out of which 49(59.03%) were male and 34(40.96%) were female Table I. Regarding age 1(1.20%) patients were in group A, 3(3.61%) patients in group B, 12(14.45%) patients in group C, 21(25.30%) patients in group D, 27(32.53%) patients in group E, 12(14.45%) patients in group F while 7(8.43%) patients were in group G. table II.

Family history of RP was present in 79(95.18%) patients while in 4(4.81%) patients no family history was present table III.

Table-I. Showing gender distribution

Gender	Number	%age
Male	49	59.03%
Female	34	40.96%

All the patients had bilateral involvement. Regarding presentation, 61(73.49%) patients presented with night blindness, 56(67.46%) presented with defective vision, 21(25.30%) patients had visual field defect table IV. In patient with defective vision i.e 56, all had no refractive error. Out of 56 patients 24 patients had refractive error while 32 patients had no significant error.

Fundoscopy revealed bone spicule pigments configuration in 83(100%) patients, attenuated blood vessel in 23(27.71%) patients while waxy pallor disc in 12(14.45%) patients table V. Regarding ocular associations, 24(28.91%) patients had refractive error (myopia), 14(16.86%) patients had cataract mostly posterior sub capsular, 3(3.61%) patients had glaucoma, 2(2.40%) patients had cystoid macular oedema while 1(1.20%) patients had keratoconus. table VI.

Table-II. Age distribution

Category	Number	No. of patients	%age
A	Up-to 10 years	1	1.20%
B	11-20 yrs	3	3.61%
C	21-30 yrs	12	14.45%
D	31-40 yrs	21	25.30%
E	41-50 yrs	27	32.53%
F	51-60 yrs	12	14.45%
G	61-70 yrs	7	8.43%

Table-III. Familial distribution

Category	No. of patients	%age
Family History	79	95.18%
No Family History	4	4.81%

Table-IV. Clinical Presentation

Category	No. of patients	%age
Night blindness	61	73.49%
Vision defect	56	67.46%
Field defect	21	25.30%

Systemically only 1(1.20%) patients had bardet biedle syndrome while there was no systemic association in 82(98.79%) table VII.

DISCUSSION

R.P is a grave disease with irreversible damage and loss of visual status. It has dreadful complication. If detected early and treated properly the quality of life can be made better to some extent.

Our study was focused on clinical presentations and associations of patients suffering from R. P. Gender, age, clinical presentations and R. P related associations were the variables to be analyzed. It is eminent from our study that male preponderance was more as compared to female showing 59.03% male and 40.93% female and

Table-V. Fundoscopy Findings

Category	No. of patients	%age
Bone spicule pigments	83	100%
Attenuate blood vessel	23	27.71%
Waxy pallor disc	12	14.45%

Table-VI. Ocular Associations

Category	No. of patients	%age
Myopia	24	28.91%
Cataract	14	16.86%
Glaucoma	03	3.61%
Cystoid macular oedema	02	2.40%
Keratoconus	01	1.20%

Table-VII. Systemic association

Category	No. of patients	%age
No association	82	98.79%
Bardet Biedle Syndrome	01	1.20%

most of the patients affected were in the age range of 41 to 50 yrs. Study of Andre Omgbwa Eballe, Godfroy Koki et al shows similar results with high prevalence of R.P in male and more patients affected in forties[17].This also shows bilateral involvement in all patients which is identical to our study. This study also reveals that myopia was the most common refractive error like in our study. But the prevalence of glaucoma was 7.50% as compared to 3.61% in our study. The above mentioned study shows drop in visual acuity in 87.49% patients and night blindness in 70% patients, but our study shows it in 67.46% and 73.49% respectively. Gorover et al found in their US series study an average age on presentation 41 years and worsening of the gravity of the disease with increasing age which is similar to our study[18]. Study of UK Ponmwan[19] et al in Nigeria and Tsujikawa[20] et al in Japan reported average age at diagnosis of 36.7 and 35.1 years respectively.

This difference may be explained by delayed presentation of our patients. Our study shows in the fundi findings bone spicule pigments configuration in 100% patients, attenuated blood vessels in 27.71% patients and waxy pallor disc in 14.45% patients and cystoid macular oedema inn 2.40% patients. Study of Sun Ho Lee[21] et al reflects bone spicule pigments in 88.8% patients, attenuated retinal blood vessels in 76.2% patient's waxy pallor disc in 12.6% patients and cystoid macular oedema in 0.5% patients. This study also shows high prevalence of cataract and myopic refractive error as evident from our study. R.P has many systemic associations. Our study reveals bardet biedle syndrome in 1(1.20%) patients. Study of Jamshed Ahmed Shows this association in 0.9% patient[22]. This study also reveals that male are more affected and mostly under age forty years. This study shows keratoconus in 0.9% patient but we noted it in 1.20% patients. Study of Peng in China shows that myopia has close association with R.P. This study also demonstrates 2.3% prevalence of glaucoma in R.P[23].

R. P is mostly bilateral. Our study shows bilateral involvement in 100% patients. There may be unilateral involvement. Study of Mohammad Naqaish Sadiq et al has revealed R.P unilaterally[24].

We endorse some limitations in our study. They include inadequate technical expertise and facilities which prevented some relevant and important tests being performed particularly ERG which is of paramount importance in patients with R.P. It can detect the RP in early stages. There is no relationship between ERG and VA because ERG may be off in the presence of good central visual acuity. Fluorescein angiography has also diagnostic as well as prognostic importance. Automatic visual field analyzer has significant role but was not easily accessible.

CONCLUSIONS

RP is a grave disease. Visual impairment in RP is very common. The clinical features and complications get worse with increasing age. It has many ocular and systemic associations. Myopia, cataract and glaucoma have high prevalence in RP patients. Detailed study and proper education of the patients regarding the presentation, complications and familial occurrence should be addressed. Proper debat in primary, secondry and tertiary level health institutions involving school teachers and civil society should be carried out.

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Correspondence Address:
Dr. Muhammad Alam
Senior Registrar Ophthalmology
C/O Khattak Pharmacy Opposite Gynae OPD Gate
Lady Reading Hospital, Peshawar
malamktk@gmail.com

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"Be nice to people on your way up because you meet them on your way down."

(Jimmy Durante)