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ORIGINAL ARTICLE

The Genetics and Clinical Presentation of Retinitis Pigmentosa A Tertiary Care Hospital Study

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ABSTRACT

Objective: This study aims to analyze the retinitis pigmentosa patients' Clinical Presentationand inheritance patterns. Research conducted at the tertiary care hospital

Material and methods: this tertiary care hospital study was carried out at a level I trauma center between 05 January 2018 to 05 January 2021. fifty-six patientsand members of their families underwent an ophthalmic evaluation to help identify those with the condition and define its phenotype. The family tree was traced. Several relatives also had optical gheremco tomography and fundus photography.

Results: at Presentation, 52 eyes (47%) were judged to be legally blind, whereas 38 (32%) had some degree of visual impairment. The visual field was narrowed in 22 instances (40%) due to the brightness of the interaction. In terms of inheritance patterns, we discovered that three cases were autosomal dominant (4%), 39 cases were autosomal recessive (70%), and three cases were x-linked (6%). Of the total cases, 11 (20%) were isolated incidents. There were 48 patients with a conventional retinitis pigmentosa image (86%) and eight patients with an unusual RP picture (12%), including three instances of pericentric RP (5%), 3 cases of Usher's disease (4%) and 1 case of retinitis punctata albescence (2%) bardet-biedl were identified in a single patient.

Conclusions: Retinitis Pigmentosa Patients Have A Very Significant Risk Of Becoming Blind. Autosomal Recessive sporadically follows the Most Prevalent Pattern Of Inheritance. Early Detection Of Retinitis Pigmentosa Is Important In Preventing Blindness From The Disease, As Is Encouraging Patients To Forego Marriages To First Cousins.

Keywords: Genetics and Clinical, Presentation, Retinitis Pigmentosa, kpc

INTRODUCTION

The term "retinitis pigmentosa" refers to a category of inherited eye diseases in which photoreceptors and the retinal pigment epithelium (RPE) gradually lose their ability to function. Roughly [01,05] million persons are Affected all over the world[1], with a frequency of about 01 in 9,000. In young people, retinitis pigmentosa is the most frequent retinal dystrophy, resulting in a gradual decline in visual acuity and visual field size and, therefore, a significant visual impairment[2,] randomly 50% Having no family Comparatively mild Hx and late-onset than xl/more ar's severe onset[3]. Various writers have classified retinitis pigmentosa in various ways, each based on the Mendelian inheritance pattern seen in the disease. Their mode of inheritance4,5 unquestionably influences these individuals' visual decline rate. This research aimed to examine how our population's clinical presentation and inheritance patterns stack up against those of other nations[6].

MATERIAL AND METHODS

This study was Conducted At Tertiary Care Hospital Ophthalmology outpatients at a tertiary hospital in Pakistan treated 56 [RP] patients between 05 January 2018 and 05 January 2021. Simple demographic and social information was collected using a standard form. All patients' family histories and infectious disease records were collected for genetic analysis. There was an attempt to look at everyone in the family. The patients were divided into the following main categories based on their manner of inheritance, or more people were affected by an autosomal recessive (AR) trait. Children's effects on their parents remain unaltered. This group also had patients who shared a parent. Because it is X-linked, only men are afflicted with XLRP. Pregnant women were the bearers. At least two generations of the characteristic were transmitted vertically. No progeny of infected males were affected.

Sporadic or Isolated (ISO): Persons with no discernible familial history were included in this group. There was just one person engaged. Snellen's chart visual acuity tests, retinoscopy for refractive error detection, color vision testing, confrontational visual field testing, applanation tonometry, and slit lamp biomicroscopy of the anterior and posterior segments were all components of the comprehensive ocular examination given to each patient. We used a direct and indirect ophthalmoscope, a Goldman triple mirror, and a +90 D. non-contact lens to inspect the fundus of the eye. In certain patients, fundus fluorescein angiography or colored fundus photography was done.SPSS (Statistical Package for the Social Sciences, USA) version 11.00 for Windows was used for data input and analysis.

RESULTS

A total of 56 people with confirmed retinitis pigmentosa were examined. The patients' ages varied from 5 to 75, with a mean of 29.80 (SD 18.45), and 41 (80%) were less than 42. (fig.1) There were more women than men, with 38 (65%) women and 19 (34%). (Fig.2). Twenty-six patients (45%) were residents of the immediate region, while thirty patients (52%) were referred from nearby communities or hospitals. The average level of visual acuity was [06/24], ranging from 06/06 to no light perception (Table 1). At the time of Presentation, 56 (45%) eyes were considered legally blind, whereas 38 (34%) were. (Table 1). In 22 instances, or 38%, the visual field narrowed due to the interaction. One instance (two percent) was determined to have elevated intraocular pressure. Myopia was detected in 24 (43%) individuals, hypermetropia in 08 (13%), and astigmatism in 09 (16%). (Table 2). Table 3 shows that only 30 patients (53%) had a positive family history, whereas 40.5% had a history of cousin marriage (Table 4). The percentages of autosomal dominant, autosomal recessive, and X-linked inheritance were 0.4, 38.5, and 3.5, respectively. There were 22 isolated incidents (20%). (Table 5). Patients with conventional retinitis pigmentosa number 46 (87%), while the remaining 088 (12%) exhibit an unusual appearance, including three instances of pericentric RP, 3 cases of Usher's disease, and 1 case of retinitis punctata albescence. It was determined that one individual had Bardet-Biedl syndrome and another had Cockayne syndrome (Table 6). atrophic maculopathy should be the top cause of blindness in 30 cases (53%), glaucoma in 1 (0%), atrophic

maculopathy in 36 (36%), cellophane maculopathy in 16 (27%), and a combination of the two in 5 (9%). (Table 7). Thirty-eight individuals, or 68%, had deteriorated vitreous. Just 1% of patients were discovered to have keratoconus, and 1% had drusen on the optic nerve head.

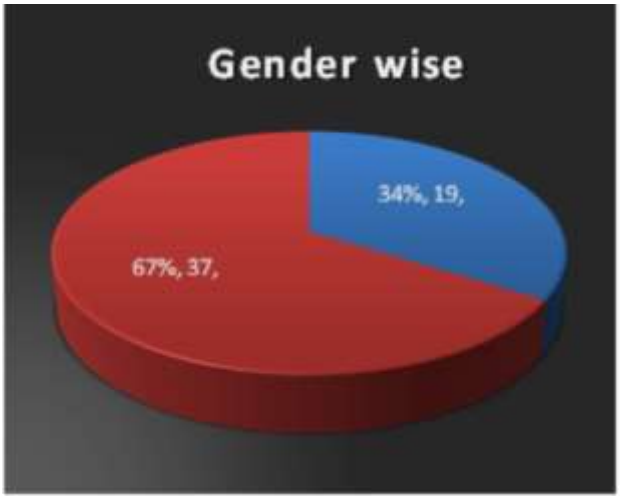

Fig. 1: Patients Gender female to high

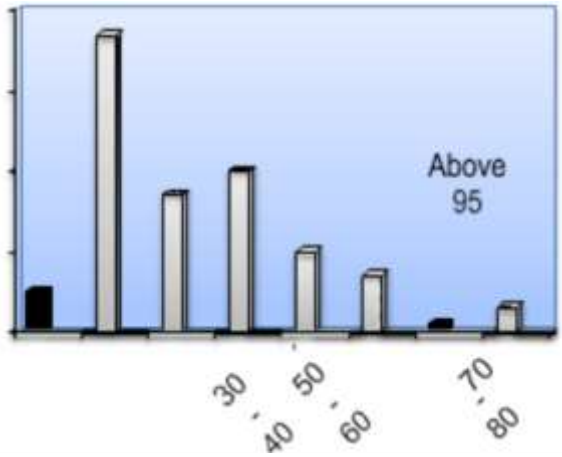

Fig. 2: Definitions of Age Groups

Table 1: Retinal regions in each eye

Hypermetropia	08 (13)
Myopia	24 (43)
Total	56 (100)

Table 2: Family history

	Frequency n (%)
Positive	30 (54)
Negative	26 (45)
Total	56 (100)

Table 3: Marriages between close relatives

[Visual acuity]	[Frequency n (%)]
[03/60 or Below]	52 (45)
[03/60 to 6/60]	16 (12)
[06/60 to 6/18]	22 (19)
[06/18 to 6/6]	22 (20)
Total	56 (100)

Table 4: Distribution of refractive errors

Refraction	Frequency n (%)
Emmetropia	16 (28.6)
Astigmatism	08 (15.2)

Table 5: Distribution of frequency

	Frequency n (%)
Absent	16 (29)
Present	40 (71)
Total	56 (100)

Table 6: Distribution of modes of inheritance

Modes of inheritance	Frequency n (%)
Autosomal dominant	02 (02)
Autosomal recessive	18 (35)
X-linked	02 (03)
Sporadic	06 (10)

Table 7: Retinitis pigmentosa: a description of the subtypes

[Types of RP	Frequency n (%)
[Typical RetinitisPigmentosa]	47 (85)
[Atypical RetinitisPigmentosa] 1	07 (12)
Pericenteric RP	03 (05)
2 Usher's Syndrome 3Retinitis	02 (04)
Punctataalbescence	01 (02)
4Laurance moon BiedlSyndrome	01 (01)

Table 8: State of maculae

Maculopathy	Frequency n (%)
Atrophic	24 (44)
Cellophane and atrophic	08 (13)
Cellophane maculopathy	07 (13)
Normal maculae	14 (24)
Pigmentary	03(05)
Total	56 (100)

DISCUSSION

The most prevalent retinal dystrophy, retinitis pigmentosa, causes visual impairment in working-age adults due to its detrimental effects on retinal function[7]. Our analysis found a significantly higher mean age of Presentation of 22 to 95 years (P=0.002) compared to the [24] years reported in western literature. That's a sign that our demographic tends to appear late. The gender imbalance in this research may result from the specific social and cultural context in which it was conducted[8]. Most patients in this research (83%) are under 45, suggesting that those in their prime earning years are the primary target[9]. Most patients (70%) were found to have an autosomal recessive pattern of inheritance[10]. Various studies report varying percentages (Table 8). Possible causes include variations in how this disease is classified and recorded and the increasing prevalence of cousin marriages in our culture. Compared to the 24% reported in western literature, our research found that 45% of eyes were legally blind at Presentation. Similar to what was described in the literature, we discovered 53% of patients required glaucoma or lens extraction. [11].

CONCLUSION

Retinitis pigmentosa is a genetic eye disorder that may lead to blindness. At a minimum, tertiary care institutions should offer electrophysiologic facilities for early detection of this condition. Avoiding marriages within families might lessen the risk of contracting this disease.

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