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Original research article

Clinical study of retinitis pigmentosa among patients coming to tertiary center for visually handicapped certificates

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Abstract

50 patients of Retinitis pigmentosa cases attended to ophthalmology outpatient department of tertiary care centre i.e., Government General Hospital, Guntur for visually handicapped certificates from January 2023 to May 2023 were inquired about age, sex, presenting features, history of presenting complaint, family history, examined for visual acuity, refraction, colour vision, slit lamp examination, fundus examination.

RESULTS: In this study, 58% males & 42% females are diagnosed as Retinitis pigmentosa. 58% have nyctalopia, 26% have defective vision, 40% have poor vision, 20% have constricted fields.

CONCLUSION: Prevalence of Retinitis Pigmentosa was seen high in 4th – 5th decade, male preponderance.

Keywords: Retinitis pigmentosa, nyctalopia.

Introduction

- Retinitis Pigmentosa (RP), classified as rod cone dystrophy, is a genetically heterogeneous disorder considered to be a final common pathway arising from rod photoreceptor degeneration and RPE abnormalities.
- With an incidence of 1: 3,500-1: 4,000 in the USA and Europe, RP is the most common form of IRD.
- Inheritance of RP can be either AD, AR, XL, or mitochondrial, and over 60 genes are known to cause RP. AR inheritance accounts for 5-20% of RP and of these, 10-15% are due to variants in the USH2A gene. AD inheritance accounts for 15-20% of RP and of these, 20-30% are due to variants in the Rhodopsin (RHO) gene. XL inheritance is considered to be the most severe form of RP with onset in childhood and accounting for 5-15% of RP.
- Of these, 70-90% are due to variants in the RP GTPase regulator (RPGR) gene. There is no known family history in 40-50% of patients, with the majority of these, have AR RP.
- A rare form of RP known as 'digenic' occurs when simultaneous variants arise in both PRPH2 (previously known as RDS) and ROM1 genes.
- Additional terminology includes 'simplex' where an isolated case of RP occurs with an absence of family history and 'multiplex' where isolated RP occurs in 2 or more family members (such as siblings) with no preexisting family history
- Modes of transmission-dominant, recessive, X-linked, digenic, mitochondrial, uniparental isodisomy.
- Symptoms-abnormal adaptation, night blindness, loss of mid-peripheral and then far-peripheral visual field, tunnel vision, eventual loss of visual acuity.
- Signs on the ocular examination-posterior subcapsular cataracts, vitreous cells, attenuated retinal

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vessels, bone spicule pigment around the mid-periphery, waxy pallor of the optic discs, cystoid macular edema in some cases.

Aims

- The purpose of the study is to evaluate the clinical picture of Retinitis Pigmentosa among patients coming to tertiary centers for blindness certificates.

Objectives

- To assess the frequency of occurence of Retinitis Pigmentosa in hospital settings.
- To correlate Retinitis Pigmentosa with their clinical disease profile in a tertiary care center for blindness certificate.
- Identify causes/risk factors of Retinitis Pigmentosa.
- To health educate the cases about low vision aids.

Materials and Methods

- A prospective observational study was done on 50 patients coming to tertiary centre i.e., GOVERNMENT GENERAL HOSIPTAL, GUNTUR for vision loss from JANUARY 2023 TO MAY 2023.
- With age between 20-55 years and patients diagnosed with Retinitis Pigmentosa with informed consent were included in the study & patients with other retinal pathologies were excluded from the study.

History

- At visit, a detailed history was obtained from each patient. This included the patient signing informed consent details of ocular complaints, duration, laterality, family history of blindness. Information about each patient was collected and entered in a proforma specially designed for the case series.

Following ophthalmological examinations are done

1. Best-corrected visual acuity with Snellen's chart.
2. Near vision test plates for near vision.
3. Colour vision with Ishihara's color vision plates.
4. Anterior segment examination using a slit lamp.
5. Fundus-slit-lamp biomicroscopy with 78/90D or indirect ophthalmoscopy.

Inclusion criteria

- Patients with age between 20-55 years.
- Patients diagnosed with Retinitis Pigmentosa.
- Results of perimetry for each eye showing narrowing for 15 degrees or more.
- Patient signed informed consent.

Exclusion criteria

- Patients age less than 10 years.
- Patients with other retinal pathologies.
- Patients refused participation.

Results

- The present study was conducted on 100 patients who attended tertiary care hospital, Guntur for visually handicapped certificate during period JANUARY 2023 TO MAY 2023. Of these 50 patients diagnosed as RETINITIS PIGMENTOSA, 29 were Males & 21 were Females indicating that male were more commonly affected than females. 21 patients had 100% disability, 17 patients had 75% disability, 12 patients had 40% disability.

Age	Male	Female	Total
20-30 years	5	1	6
31-40 years	5	7	12
41-50 years	8	10	18
51-60 years	11	3	14
Total	29	21	50

Out of 50 patients who were diagnosed as cases of Retinitis Pigmentosa, 29 cases were males i.e.,58% & 21 cases were females i.e., 42%, showing male preponderance. The peak age of presentation is 41 – 50 years of age group i.e., 36% for visually handicapped certificate.

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BCVA	Total
6/12-6/36	30
6/60-1/60	35
CF-PL+	33
NO PL	2

Out of 100 eyes examined, 30 eyes have visual acuity of 6/12-6/36 i.e., 30% 35 eyes have visual acuity of 6/60-1/60i.e., 35%, 33 eyes have visual acuity of CF-PL+ i.e., 33%, 2 eyes have visual acuity of NO PL i.e., 2%.

Categories	Better eye	Worse eye	Percentage
Category I	6/18-6/36	6/60 to Nil	40%
Category II	6/60-4/60 or field of vision 10^0 to 20^0	3/60 to Nil	75%
Category III	3/60 to 1/60 or field of vision $<10^0$	F.C. at 1ft. to Nil	100%
Category IV	F.C. at 1ft. to Nil or field of vision $<10^0$	F.C. at 1ft. to Nil	100%

	20%	40%	75%	100%	Total
Males	3	4	9	13	29
Females	2	4	8	7	21
	5	8	17	20	50

Out of 50 patients examined, 29 patients were males of which 3 patients are 6% i.e.,10%, 4 patients are 40% i.e.,8%, 9 patients are 75% i.e.,18%, 13 patients are 100% i.e.,26% & where as 21 patients were females of which 2 patients are 20% i.e.,4%, 4 patients are 40% i.e.,8%, 8 patients are 75% i.e.,16%, 7 patients are 100% i.e.,14%.

Symptoms	Number of Patients
Nyctalopia	29
Defective vision	13
Poor vision	20
Constricted fields	10

Out of 50 patients with retinitis pigmentosa, 29 patients have symptom of nyctalopia i.e., 58%, 13 patients have symptom of defective vision i.e., 26%, 20 patients have symptom of poor vision i.e., 40% & 10 patients have symptom of constricted fields i.e., 20%.

DISCUSSION:

Retinitis pigmentosa is a slow, degenerative disease of the retina, almost invariably occurring in both eyes, beginning in childhood and often resulting in blindness in middle or advanced age, which is a major public health problem in developing countries like India. (Schémann, 2002) The degeneration affects primarily the rods and cones, particularly the former, and commences in a zone near the equator of the eye gradually spreading both anteriorly and posteriorly. The symptoms are characteristic, the most prominent being defective vision in the dusk (night blindness, nyctalopia) The visual fields show concentric contraction, especially marked if the illumination is reduced. In early cases a partial or complete annular ring scotoma is found corresponding to the degenerated zone of the retina. In the majority of families it appears as a recessive trait and consanguinity of the parents is not infrequent.

In this prospective observational study conducted in tertiary care centre, from January 2023 to May 2023 for visually handicapped certificates, 50 cases of Retinitis Pigmentosa cases were diagnosed presented with nyctalopia in 58% cases, defective vision in 26% cases, poor vision in 40% cases & constricted visual fields in 20% cases. With male preponderance i.e., 58% males & 42% females are affected. Bhattarai D. et al 2015 reported a case of Unilateral Retinitis Pigmentosa in a 70 year old female. Though the frequency of Unilateral RP is reported to be around 5% of bilateral retinitis pigmentosa. (Farell, 2009). Till now the etiology of Unilateral RP is unknown and its inheritance is unclear. However, as shown by some studies, the genetic inheritance behind the unilaterality of the disease may be due to different unidentified mutations at the single loci or non-linked mutations in m

Conclusion

This study was undertaken in the Department of Ophthalmology, Government General Hospital, Guntur, during the period of JANUARY 2023 TO MAY 2023. 50 cases of Retinitis Pigmentosa were studied for disc changes.

Age

Highest occurrence was seen in 41-50 years of age group followed by 51-60 years of age group, Retinitis Pigmentosa peaks between 4^{th}-5^{th} decade.

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Gender: In the present day, males are more affected than females, with a male to female ratio 1.38:1.

- That ocular abnormalities associated with RP are many and have high prevalence, especially cataracts and macular abnormalities.
- RP is often complicated by cataracts which is a leading cause of visual disability.
- Complicated cataracts affect the visual acuity and life quality in RP patients.
- For macular abnormalities, CME may negatively affect BCVA in RP patients with eyes with clear lens. It is essential to evaluate the macular structure with OCT when accessing RP patients' visual function.

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