ISSN: 2581 – 3633
PubMed - National Library of Medicine - ID: 101745081

International Journal of Medical Science and Advanced Clinical Research (IJMACR)

Available Online at:www.ijmacr.com

Volume – 6, Issue – 1, February - 2023, Page No. : 500 - 505

Ocular morbidities in retinitis pigmentosa (rp) patients coming to tertiary health Centre in south Gujarat - A cross sectional study.

[1]Dr. Trupti M Solu, Additional Professor, Government Medical College, Surat.

[2]Dr. Mese Mero,Third Year Resident, Government Medical College, Surat.

[3]Dr. Aal Vijay, Second Year Resident, Government Medical College, Surat.

Corresponding Authors: Dr. Mese Mero, Third Year Resident, Government Medical College, Surat.

How to citation this article: Dr. Trupti M Solu, Dr. Mese Mero, Dr. Aal Vijay, "Ocular morbidities in retinitis pigmentosa (rp) patients coming to tertiary health Centre in south Gujarat - A cross sectional study", IJMACR- February - 2023, Volume – 6, Issue - 1, P. No. 500 – 505.

Type of Publication: Original Research Article

Conflicts of Interest: Nil

Abstract:

Purpose: To document macular pathologies, visual field changes and associated ocular pathologies in Retinitis Pigmentosa (RP) patients in a tertiary care Centre in South Gujarat.

Design: Cross sectional study.

Materials and Methods: It was a cross-sectional study which included patients coming to New Civil Hospital Surat from August 2021 to August 2022.

Results: A total of 40 patients were included in the study. Majority of (60%) patients were male. The majority of the participants belonged to the age group of 18-25 years (11, 28%) and 41-55 years (12, 30%).17 (43%) patients had family history of Retinitis Pigmentosa among them 7 had history of consanguineous marriage. Uveitis (2, 5%),Glaucoma (2, 5%) and Myopia was present among 11 (28%) of the patients. 61 eyes were not able to assess for the visual field, 13% of the right eye and 7% of the left eye had Grade 4 visual field defects. OCT examination of 80 eyes, 3% had within normal limit findings and in right eye and left eye around 90% of patients had decreased CFT findings. Macular pre-retinal fibrosis was present in 21 (53%) right eye and 18 (45%) of left eye.RPE atrophy was present in 38 (95%) of the right eye and 38 (95%) of the left eye.

Conclusion: Majority patients were male, and belonged to the age group 41-55 years. This study also established a significant association between consanguinity and positive family history of Retinitis Pigmentosa. Majority of patients had severe disease and were not able to assess their visual field and it has significant association with age of the Retinitis Pigmentosa patients. Several ocular comorbidities like posterior subcapsular cataract, myopia, open angle glaucoma and myopia among RP patients are present. On OCT majority of the patients

showed decreased CFT and almost all patients had RPE atrophy.

Keywords: RP, OCT, CFT, CME

Introduction

Retinitis Pigmentosa is the most common Inherited Retinal Dystrophy worldwide. It is characterized by the progressive degeneration and death of rod photoreceptors primarily, resulting in a nyctalopia and a progressive constriction of the visual field. [(1)] Global prevalence of retinitis pigmentosa (RP) is estimated to range from 1 in 9000 to 1 in 750 with an average of 1 in 4,000. [(2)] In India it is found to be much higher. Prevalence of Retinitis Pigmentosa in India is 1 in 372 in rural population and 1 in 930 in urban population. Around 1.4 million Indian carry genetic mutation for Retinitis Pigmentosa. [(3)]

Retinitis Pigmentosa is a Mendelian disease that is most commonly inherited in an autosomal recessive (arRP) (50–60% of cases), autosomal dominant (adRP) (30–40%), or X-linked (XLRP) (5–15%) manner. Studies have shown that among the three forms of Retinitis Pigmentosa, adRP typically presents with the mildest form of the disease while XLRP presents with the most severe. The average age of symptom onset is dependent on the genetic type. The autosomal recessive form will develop symptoms in the early teen years, but those affected with autosomal dominant RP will likely not have symptoms until well into their 20s. More than three-quarters of individuals with RP will be symptomatic and present for clinical evaluation and diagnosis of the disease by the time they are 30 years of age. [(4)] In India prevalence of Retinitis Pigmentosa is high with higher proportion of homozygous variants consistent with higher prevalence of consanguinity up to 24% in South India and 16% in North India. [(5)(6)]

There are several other ocular conditions associated with RP. Refractive error or nystagmus may occur in early-onset RP, whereas macular hole, epiretinal membrane formation, and cystoid macular edema (CME) may be present in more than 50% of RP patients. In addition, posterior subcapsular cataracts are present in up to 45% of RP patients, secondary retinal Vaso proliferative tumors, vitreous cysts in 6% of RP patients, and optic nerve head and fiber layer drusen in approximately 9% of RP patients. [(7)]

The natural course of RP involves an estimated loss of 4% to 12% of the visual field and 17% of ERG amplitude annually. RP is the leading cause of visual disability and blindness in subjects less than 60 years old. This accounts for 25% to 30% of all visual disability cases.

Individuals with RP experience a lower quality of life than healthy individuals, associated with reduced mobility and challenges with activities of daily living, increased financial burden due to difficulties staying in work, reduced autonomy, and often social isolation.

There are limited treatment options available for Retinitis Pigmentosa that too in resource limited country like India which has limited research available on the same. With higher prevalence of consanguinity lead to higher rate of Retinitis Pigmentosa among Indian population but there are lack of population screening and counselling for genetically transmitted ophthalmic disorders in India along with Mendelian disorder in general. There are many treatable ocular morbidities associated with Retinitis Pigmentosa. Early diagnosis and treatment of these treatable comorbidities is necessary to improve quality of life in Retinitis Pigmentosa patients. So, this study was conducted with

the aim to identify ocular morbidities associated with Retinitis Pigmentosa.

Materials and method

All patients above 18 years of age having typical RP attending out patient department at New Civil Hospital Surat, South Gujarat from August 2021 to August 2022 were included in the study. Patients with pre-existing retinal diseases and patients who had undergone PRP for diabetic retinopathy are excluded from the study. A detailed history was taken along with socio demographic data like age, gender, type of marriages of parents (consanguineous\ non consanguineous), family history recorded along with detailed clinical history like systematic illness, associated syndrome and pedigree chart. Detailed ophthalmic examination was done.

Visual acuity tests and refractive error was assessed on Snellen's chart. Color vision was assessed with the Ishihara chart. Slit lamp biomicroscopic examination was done followed by Intraocular tension measurement, fundus examination and visual field examination. Intraocular tension was documented with Goldman Applanation Tonometry. Patients was examined with the slit lamp biomicroscope (and 78D lens). The Fundus examination was carried out with an indirect ophthalmoscope and 20 D lens. Perimetry with Humphrey visual field analyzer (as and when required) was done. Spectral domain optical coherence Tomography subsequently was done as and when required.

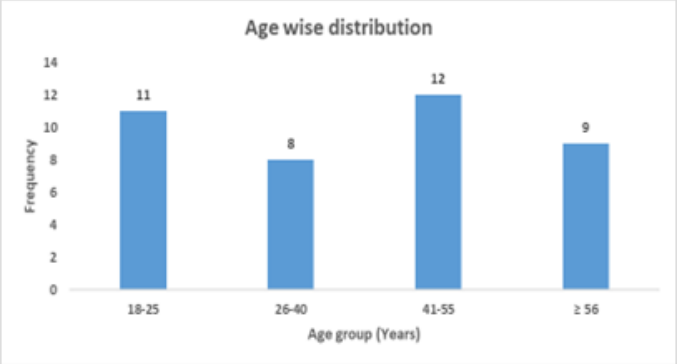
Results and discussion

Majority of (60%) patients were male. Parameswarappa DC et al. in their study from South India reported that 67% were males. [8]

The majority of the participants belonged to the age group of 18-25 years (11, 28%) and 41-55 years (12, 30%).

Mean age of the patients reported by Khanna et al. who conducted 15 years follow up study among Indian Retinitis Pigmentosa patients. 47.33 ± 10.89 years. [9]

Chart 1: Age wise distribution of the study population:



17 (43%) patients had family history of Retinitis Pigmentosa among them 7 had history of consanguineous marriage.

Consanguinity	**Our study(%)**	**Juyal et al (%)**	
		South India	North India
Consanguineous	23	24	16
Non-consanguineous	77	76	82

Table 1: comparison between consanguinity

Juyal et al. documented that Indian prevalence of Retinitis Pigmentosa is high with a higher proportion of homozygous variants consistent with higher prevalence of consanguinity up to 24% in South India and 16% in North India. [6]

In this study we found ocular comorbidities like Uveitis (2, 5%), Glaucoma (2, 5%) and Myopia was present among 11 (28%) of the Retinitis Pigmentosa patients. Dutta Majumder P et al stated that uveitis in RP patients is rare, but not uncommon. [10] Hung MC eta al. stated

that cumulative incidence of PACG in patients with Retinitis Pigmentosa was 1.61%and which was higher than control in their population-based cohort study. [11] Rishi et al reported 68% of patients with Myopia. [12]

We further examined 80 eyes for visual field defects, 61 eyes were not able to assess for the visual field, apart from that 13% of the right eye and 7% of the left eye had Grade 4 visual field defects.

Chart 2: Visual Field Defect among study population.

Xu M et al reported that the rate of VF loss varies among targets in patients with RP. Fifty percent of patients are not qualified to drive by the age of 37 and become legally blind by the age of 55. These results can be useful for counselling patients with RP as to their potential rate of VF decline. [13]

Table 2: Visual Field Defect among different age group

Variable	Group	Right Eye (%)	Left Eye (%)	
Visual Field Defect	Grade 1	0 (0)	0 (0)	0 (0)
	Grade 2	2 (5)	1 (2.5)	3 (3.75)
	Grade 3	1 (2.5)	2 (5)	3 (3.75)
	Grade 4	5 (12.5)	3 (7.5)	8 (10)
	Grade 5	2 (5)	1 (2.5)	3 (3.75)
	Grade 6	0 (0)	2(5)	2(2.5)
	Cannot be assessed	30 (75)	31 (77.5)	61 (76.25)
Total		40 (100)	40 (100)	80 (100)

On OCT examination of 80 eyes, only 3% had within normal limit findings and in right eye and left eye around 90% of patients had decreased CFT findings. Macular pre-retinal fibrosis was present in 21 (53%) right eye and 18 (45%) of left eye.

RPE atrophy was present in 38 (95%) of the right eye and 38 (95%) of the left eye.

Table 3: OCT findings among study population

Variable	Group	Right Eye (%)	Left Eye (%)	Total (%)
OCT Findings	Within Normal Limit	3(7.5)	3(7.5)	6(7.5)
	CME	0(0)	0(0)	0(0)
	ERM	1(2.5)	0(0)	1(1.25)
	Decrease CFT	36 (90)	37 (92.5)	73(91.25)
	VMT	0(0)	0(0)	0(0)
	Macular Hole	0(0)	0(0)	0(0)
Total		40(100)	40(100)	80(100)

Chart 3: OCT findings among study population

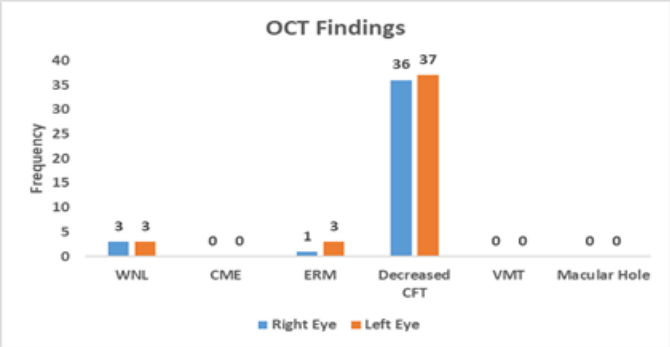
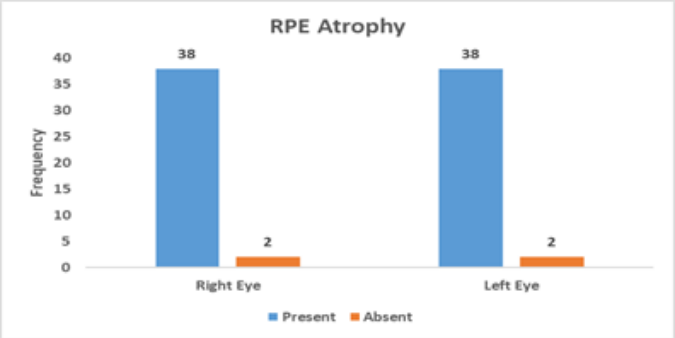


Table 4: Macular Pre-Retinal Fibrosis among study population

Variable	Group	Right Eye (%)	Left Eye (%)	
Macular Pre-Retinal Fibrosis	Present	21 (52.5)	18 (45)	39 (48.75)
	Absent	19 (47.5)	22 (55)	41 (51.25)
Total		40 (100)	40 (100)	80 (100)

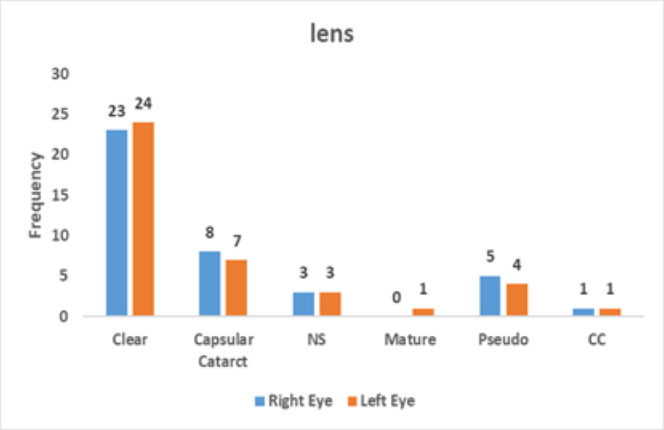
Chart 4: RPE Atrophy among study population.



Miura et al reported that Retinal Pigment Epithelial atrophy is very common in patients with Retinitis Pigmentosa. Which align with the findings reported by our study. [14]

On examining the lens 23 (58%) of the right eye had clear lens and 24 (60%) of the left eye had clear lens.

Chart 5: Findings of lens among study population



A study conducted by Faujiwara et al documents that there is an association of inflammation and the pathogenesis of subcapsular cataract formation in patients with Retinitis Pigmentosa. [15]

Results of the study conducted by Alexander S et al. showed that 72% of patients with Retinitis Pigmentosa reported benefit from cataract surgery, as cataract surgery improve vision among Retinitis Pigmentosa patients and in our study majority of the elderly patients presented with cataract and it was statistically significant. [16]

Conclusion

In this study, we identified ocular prevalence of comorbidities among Retinitis Pigmentosa patients. All 40 Retinitis Pigmentosa presented with its Triad. The majority of patients had severe diseases as were not able to assess their visual field and it has significant association with age of the Retinitis Pigmentosa patients. Patient education and psychological support is necessary. Proper rehabilitative management and measures can greatly improve their quality of life. On OCT majority of the patients showed decreased CFT. Almost all patients had RPE atrophy present. This study also reports a significant association between positive family history and consanguinity among Retinitis Pigmentosa patients. Premarital genetic counselling needs to be done to prevent Retinitis Pigmentosa.

References

1. Henderson RH. Inherited retinal dystrophies. Paediatrics and child health. 2020 Jan 1;30(1):19-27.

2. Bruninx R, Lepièce G. Retinitis pigmentosa. Revue Medicale de Liege. 2020 Feb 1;75(2):73-4.

3. (Bansal M, Tandon R, Saxena R, et al. Ophthalmic genetics practice and research in India: Vision in 2020. Am J Med Genet Part C. 2020;1–10. https://doi.org/10.1002/ajmg.c.31827)

4. Jauregui R, Takahashi VK, Park KS, Cui X, Takiuti JT, Lima de Carvalho JR, Tsang SH. Multimodal structural disease progression of retinitis pigmentosa according to mode of inheritance. Scientific reports. 2019 Jul 24;9(1):1-7.

5. Yohe S, Siva Sankar M, Ghosh A, Ghosh A, Holle J, Murugan S, Gupta R, Schimmenti LA, Vedam R, Thyagarajan B. Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India. Molecular Genetics & Genomic Medicine. 2020 Feb;8(2): e1081

6. 6Juyal G, Mondal M, Luisi P, Laayouni H, Sood A, Midha V, Heutink P, Bertranpetit J, Thelma BK, Casals F. Population and genomic lessons from genetic analysis of two Indian populations. Human genetics. 2014 Oct;133(10):1273-87.

7. 7Loukovitis E, Anastasia S, Tranos P, Balidis M, Asteriadis S, Thanos V, Thanos S, Anogeianakis G. A

review of recent developments in retinitis pigmentosa genetics, its clinical features, and natural course. Medical hypothesis discovery and innovation in ophthalmology. 2020;9(4):231-54.

8. Parameswarappa DC, Das AV, Thakur PS, Takkar B, Multani PK, Padhy SK, Doctor MB, Agarwal K, Jalali S. Retinitis pigmentosa in Laurence-Moon-Bardet-Biedl syndrome in India: Electronic medical records driven big data analytics: Report II. Indian Journal of Ophthalmology. 2022 Jul 1;70(7):2533-8.

9. Kanna RC, ParameswarAppa D, Mar amula S, Mettla AL, Girdhar P, Banerjee S, Shekhar hK, Chakrabarti S, Jalali S, Rao GN. Incidence, Visual Impairment and Blindness due to Retinitis Pigmentosa in Rural Population in India: 15 Year Follow-up of the Andhra Pradesh Eye Disease Study cohort. Investigative Ophthalmology & Visual Science. 2022 Jun 1;63(7):2156-A0184.

10. Dutta Majumder P, Menia N, Roy R, Sen P, E. George A, K. Ganesh S, Biswas J. Uveitis in patients with retinitis pigmentosa: 30 years' consecutive data. Ocular immunology and inflammation. 2018 Nov 17;26(8):1283-8.

11. Hung MC, Chen YY. Association between retinitis pigmentosa and an increased risk of primary angle closure glaucoma: A population-based cohort study. Plos one. 2022 Sep 9;17(9): e0274066.

12. Rishi E, Rishi P, Bhende M, Koundanya VV, Sidramayya R, Mai tray A, Rao C, Susvar P, Bhende P, Sharma T. Retinal detachment in 31 eyes with retinitis pigmentosa. Ophthalmology Retina. 2018 Jan 1;2(1):10-6.

13. Xu M, Zhai Y, MacDonald IM. Visual field progression in retinitis pigmentosa. Investigative Ophthalmology & Visual Science. 2020 Jun 3;61(6):56-.

14. Miura G, Baba T, Yamamoto S. Two cases with retinitis pigmentosa that developed severe retinal atrophy long after vitreo-retinal surgery. American Journal of Ophthalmology Case Reports. 2020 Jun 1; 18:100716.

15. Fujiwara K, Ikeda Y, Murakami Y, Fun Atsu J, Naka take S, Tachibana T, Yoshida N, Nakao S, HI Satomi T, Yoshida S, Yoshitomi T. Risk factors for posterior subcapsular cataract in retinitis pigmentosa. Investigative ophthalmology & visual science. 2017 May 1;58(5):2534-7.

16. Alexander S, Moore E, Cushley L, Silvestri G. Is cataract extraction and intraocular lens implant surgery a benefit for patients with Retinitis Pigmentosa. Investigative Ophthalmology & Visual Science. 2019 Jul 22;60(9):4527-.