Prevalence of Retinitis Pigmentosa in a Tertiary Eye Hospital of Nepal

Krishna Kant Gupta[1], Govind Gurung[1], Nitin Tulsyan[1]
[1]Ram Kumar Mahabir prasad Kedia Eye Hospital, Birgunj, Nepal

ABSTRACT

Introduction: Retinitis Pigmentosa (RP) is a group of diffuse retinal degenerative diseases predominantly affecting the rod and cone photoreceptors. The prevalence of retinitis pigmentosa seen in literature is approximately 1:4000. Retinitis Pigmentosa is one of the the most common causes of blindness in the age group of 20 to 40 years. The objective of this study was to determine the profile of retinitis pigmentosa in Terai and Nepal-India border region considering patients seeking care at a Tertiary level Eye Hospital in the terai region (southern part) of Nepal.

Materials and methods: A hospital-based, retrospective study was carried out at R. M. Kedia Eye Hospital. A total of 385 (83 males and 107 females from Nepal and 109 males and 86 females from India) diagnosed patients of Retinitis Pigmentosa were included in the study. Data was collected over a period of eleven years from 2008-2018.

Results: Out of 385 diagnosed Retinitis Pigmentosa patients, 192 (49.87%) were male and 193 (50.13%) were female with slightly female predominance. The prevalence of RP seen in our study was 0.03%. About 51% of the patients visited here were from India and nearby border areas/ villages which cover most of the rural areas of India. In this study it was found that 49.34% of the RP cases were from Nepal, of which 43.63% of cases were from Hindu community and 5.71% from Muslim community and about 50.66% cases of RP were from India, of which 37.67% from Hindu and 12.98% from Muslim community. The peak age of presentation of RP was at 30-39 years (29.09%), followed by 20-29 years (26.75%). The common marriage pattern of consanguinity was found in Muslim community in between the first cousins. In this study the hospital record did not show any evaluation for the syndromic disease in the hospital record, though RP is usually non syndromic and there are literatures where many syndromic forms have been identified.

Conclusion: The prevalence of RP seen in the study was 0.03% (A total of 1101299 sample population of which 385 patients had RP). Since RP is an inherited disease and is one of the non-treatable causes of blindness which runs in the families, a role of counseling to reduce consanguineous marriages should be brought forward to reduce the disease process.

Key words: India, Nepal, Prevalence, Retinitis pigmentosa.

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Corresponding Author
Dr. Krishna Kant Gupta
Department of Glaucoma,
Ram Kumar Mahabir Prasad Kedia Eye Hospital,
Birgunj, Nepal.
E-mail: kkg00720032@gmail.com

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INTRODUCTION

Retinitis pigmentosa (RP) is the most common form of inherited retinal dystrophies, with a prevalence of 1:4000 throughout the world. (Hamel, 2006), having the first symptom as nyctalopia, followed by the progressive loss in the peripheral visual field and eventually leading to blindness. (Boughman, 1980) The most common syndrome associated with RP is Usher syndrome (USH), with a prevalence of 1:100,000 transmitted as an autosomal recessive disorder associated with congenital sensorineural hearing loss and vestibular dysfunction. (Saihan, 2009)

Based on the prevalence of recessive retinitis pigmentosa, the carrier state is approximately 1:100. (Daiger, 2007) Retinitis Pigmentosa includes various hereditary disorders affecting the photoreceptors and retinal pigment epithelium (RPE) involving the entire fundus. (Donders, 1855)

RP can occur as a sporadic disorder, or be inherited as an AD, AR, or XLR pattern. (Bowling, 2016). Although few differences between each eye are likely, it typically affects both the eyes. (Massof, 1979) Unilateral retinitis pigmentosa has been seen in literature in which one eye insulates in degeneration, although both eyes invariably show involvement. (Henkes, 1966; Cogan, 1969)

Multiple gene mutations can cause RP, many of which encode proteins essential for photoreceptors structure and function. Pedigree analysis shows evidence of 25% autosomal dominant inheritance, 25% autosomal recessive inheritance and about 10% of X-linked recessive inheritance patients. (Yanoff, 2009). Mutation in Pro-23-his rhodopsin gene has been identified by molecular testing whereas atypical maculopathy along with peripheral retinal degeneration has been seen due to mutations in RDS/peripherin gene. (Weleber, 1993)

Retinitis pigmentosa affects the RPE and photoreceptors of the retina that cause progressive night vision loss which can be determined during the early stage of life. Early identification, awareness of the disease and its progression, genetic counseling, and visual rehabilitation is a must.

A demographic study profile, gender and ethnicity relation with the disease progression study is necessary in this part of the region as most of the patients attending our hospital belong to different ethnic group, present with early symptoms of retinitis pigmentosa not only from terai region of Nepal but also from the Nepal-India border nearby cities. This type of study has not been carried out in this part of the region, it is necessary to better understand the prevalence of RP in patients seeking care at a tertiary eye hospital.

MATERIALS AND METHODS

This is a hospital-based, retrospective study done at R.M. Kedia Eye Hospital, data collected over a period of eleven years from 2008 to 2018. Ethical clearance from the Institutional Review

Committee (IRC-NNJS) was taken to carry out the study. Permission from the hospital record section was also taken to generate the data.

All the patients attending the hospital with complaints of night blindness included in the study were screened and diagnosed on the basis of their complaints, clinical findings, visual field defects, and progressive worsening of these symptoms and signs. A detailed family history, marriage history and a history of consanguinity were obtained from the patients with history of nyctalopia. All these data were collected from the hospital record section. All the patient's pupils were dilated with tropicamide 1% and a thorough fundus examination was done to look for the retinal signs of retinitis pigmentosa.

Diagnostic methods

Clinical diagnosis was made considering the patient's complaint of night blindness and a typical peripheral visual field defect, typical findings on fundus, hypovolted ERG, fundus photograph and Humphrey visual field perimetry (24-2) was done to look for the disease pattern and progression.

Diagnostic criteria

Functional signs

- Nyctalopia was the earliest symptom
- Photophobia appeared later
- The visual acuity was preserved in early and mid-stages

Visual field

- Presence of patchy losses of peripheral vision
- Presence of ring scotoma
- and finally tunnel vision

Fundus

- Presence of bony corpuscles initially seen in the peripheral retina
- Presence of attenuated retinal vessels
- Waxy pale optic disc

Statistics like mean and percentages were used for the data analysis that was collected from hospital resources and was divided by age, gender, ethnicity, and place. Statistical analysis was done with the help of Microsoft office 2010.

RESULTS

A Hospital data of eleven years from 2008-2018 was collected, whereby 11,01,299 patients (557379 from Nepal and 543920 from India) had attended R.M. Kedia eye hospital of which 385 patients had Retinitis Pigmentosa with involvement of both eyes. 190 (49.35%) of these patients were from Nepal while 195 (50.65%) patients were from India. Of the 190 patients from Nepal, 83 (43.68%) were male patients and 107 (56.32%) patients were female, and among 195 patients from India, 109 (55.90%) male and 86 (44.10%) were female. Thus the data from table: 1 indicates that there is female predominance of RP in Nepalese patients while that of India shows male predominance.

Table 1: Area of residence and gender wise distribution of Retinitis Pigmentosa patients.

	NEPAL		INDIA		
Year	Male	Female	Male	Female	Total
2008	5	5	9	3	22(5.71%)
2009	9	9	5	6	29(7.53%)
2010	13	9	17	8	47(12.2%)
2011	7	15	13	9	44(11.4%)
2012	9	25	8	7	49(12.72%)
2013	4	10	7	5	26(6.75%)
2014	13	14	11	11	49(12.72%)
2015	5	10	8	8	31(8.05%)
2016	8	6	11	15	40(10.3%)
2017	3	1	9	10	23(5.97%)
2018	7	3	11	4	25(6.49%)
Total	83(21.56%)	107(27.80%)	109(28.31%)	86(22.33%)	385(100%)

Three hundred and eighty patients diagnosed with Retinitis Pigmentosa were divided according to their year of presentation to hospital and age group as shown in table 2. The maximum number of the RP patients were seen in the 30-39 years of age group which was 112 patients (29.09%) followed by 20-29 years age group which was 103(26.75%) while the least number of patients belonged to age greater than 70 years i.e. only 7(1.81%).

Table 2: Age group and year wise distribution of Retinitis pigmentosa patients.

Year	0-9	10-19	20-29	30-39	40-49	50-59	60-69	>70
2008	2	8	3	5	1	3	0	0
2009	0	6	5	10	3	2	2	1
2010	0	20	9	8	2	1	5	2
2011	1	11	8	10	3	3	5	3
2012	0	4	19	19	1	2	2	1
2013	1	5	10	9	0	0	1	0
2014	0	13	18	18	1	0	0	0
2015	0	7	11	12	1	0	1	0
2016	2	10	8	12	4	0	3	0
2017	2	4	10	6	0	0	1	0
2018	5	9	2	3	0	2	4	0
Total	13(3.37%)	97(25.19%)	103(26.75%)	112(29.09%)	16(4.15%)	13(3.37%)	24(6.23%)	7(1.81%)

Table 3: Diversity wise distribution of Retinitis pigmentosa patients.

	Hindu community	Muslim community	Total
Nepal	168(43.63%)	22(5.71%)	190(49.34%)
India	145(37.67%)	50(12.98%)	195(50.66%)
Total	313(81.29%)	72(18.70%)	385(100%)

Based on the above stratification, as this hospital is located near the Nepal-India border, it was found that about half of the patients visiting the hospital were from India. As terai has mixed community, Table:3 shows the diversity in the distribution of RP patients from Hindu community (81.29%) of which 43.63% were from Nepal and 37.66% were from India, while 5.71% from Nepal and 12.98% from India were from Muslim community.

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 or 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 retrospective study conducted from 11 years of hospital records on 11,01,299 patients, 385 patients were found to have Retinitis Pigmentosa. Of the 385 patients 192 (49.87%) were male and 193 (50.13%) were female with slightly female predominance.

Since this hospital is located near the Nepal-India border, about 51% of the patients visiting the hospital have Indian origin and come from nearby border areas/ villages. Most of the terai area population belongs to rural area and have mixed community and in this study it was found that 49.34% of the RP cases were from Nepal, of which 43.63% of cases were from Hindu community and 5.71% from Muslim community and about 50.66% cases of RP were from India, of which 37.67% from Hindu and 12.98% from Muslim community. Nirmalan et al. have previously studied the effect of consanguinity on eye diseases with potential genetic etiology in Andhra Pradesh where parental consanguinity was reported by 1822 rural subjects and 782 urban subjects, (Nirmalan, 2006) which coincides with our study.

The peak age of presentation of RP in this study showed at 30-39 years (29.09%), followed by 20-29 years (26.75%), A study conducted by Joshi et al, causes of visual handicap amongst patients attending outpatient department of a medical college for visual handicap certification in central India showed the average age of presentation of RP was from 20-44 years (Joshi, 2013) which is similar to this study.

In a study conducted by Suman S Thapa et al. about Prevalence and pattern of vitreo-retinal diseases in Nepal: the Bhaktapur glaucoma study,(Thapa et al, 2013) the population prevalence of retinitis pigmentosa was 0.12%,while in this study it was found that the prevalence of RP was 0.03% among other retinal diseases, which must be due to the fact that the Bhaktapur Glaucoma Study was a population based cross-sectional study involving 4800 subjects aged 40 years and over from one district only, while this study is a retrospective study and has shown that peak age of RP presentation is from 20-39 years age group.

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 multiple loci.

Zafar S. et al, 2017 in their study, "Retinitis pigmentosa genes implicated in South Asian populations: a systematic review" did a comprehensive literature search using MEDLINE and CINAHL databases and all relevant articles on causative mutations for non-syndromic Retinitis pigmentosa from 1999 till 2015. Overall, 41 articles were identified involving 66 families; 28(68%) from Pakistan, 12(29%) from India and 1(2.4%) from Bangladesh. No data was available from the rest of the countries in the region. Our study found significant gaps in knowledge of the disease and the absence of cost-effective screening programs in this region.

RP is usually non syndromic but there are also many syndromic forms, the most frequent being Usher syndrome. Usher syndrome is the most frequent syndrome associated with RP, which accounts for about 14% of all RP cases. (Boughman, 1983), Zhonghua Yan Ke and Za Zhi, 2012 in their study on analysis of clinical phenotype and mode of inheritance in retinitis pigmentosa patients with consanguineous marriage, found that the incidence of RP with consanguineous marriages was high in Ningxia Region and the mode of inheritance of RP patients with consanguinity is autosomal recessive. The common marriage pattern of consanguinity was between the first cousins, the age of onset of RP was early and the ocular fundus changes of some patients were atypical. There was no record found in the hospital data showing any evidence for the syndromic evaluation for the

disease, this study does have some limitations on the systemic evaluation for the disease in our record and is totally dependent on the record section.

CONCLUSION

The prevalence of Retinitis Pigmentosa in this study was 0.03%. Retinitis Pigmentosa is the most common pigmentary retinal dystrophy. Studies have reported a much higher prevalence in the south Indian population compared to western populations and populations in other parts of India.

REFERENCES

Bhattarai D, Paudel N, Adhikari P, Gnyawali S, Joshi S (2015). Unilateral retinitis pigmentosa. Nepalese Journal of Ophthalmology, 7(1), pp.56-59. doi: 10.3126/nepjoph.v7i1.13171

Boughman J, Conneally P, Nance WE (1980). Population genetic studies of retinitis pigmentosa. Am J Hum Genet 1980; 32:223-235. Pmid:7386458

Boughman J, Vernon M, Shaver K (1983). Usher syndrome: Definition and estimate of prevalence from two high-risk populations. Journal of Chronic Diseases, 36(8), pp.595-603. doi: 10.1016/0021-9681(83)90147-9

Cogan D (1969). Pseudoretinitis Pigmentosa. Archives of Ophthalmology, 81(1), p.45. doi: 10.1001/archopht.1969.00990010047007

Daiger S (2007). Perspective on Genes and Mutations Causing Retinitis Pigmentosa. Archives of Ophthalmology, 125(2), p.151. doi: 10.1001/archopht.125.2.151

Donders F (1855). Beiträge zur pathologischen Anatomie des Auges. Albrecht von Graefes Archiv für Ophthalmologie, 2(1), pp.106-118. doi: 10.1007/bf02720791

Farrell (2009). Unilateral retinitis pigmentosa and cone-rod dystrophy. Clinical Ophthalmology, p.263. doi: 10.2147/opth.s5130

Hamel C (2006). Retinitis pigmentosa. Orphanet Journal of Rare Diseases, 1(1). doi: 10.1186/1750-1172-1-40

Henkes H (1965). Does Unilateral Retinitis Pigmentosa Really Exist?. Ophthalmologica, 149(3), pp.202-203. doi: 10.1159/000304765

Joshi R (2013). Causes of visual handicap amongst patients attending outpatient department of a medical college for visual handicap certification in central India. Journal of Clinical Ophthalmology and Research, 1(1), p.17. doi: 10.4103/2320-3897.106275

Kolb H, Galloway N (1964). Three cases of unilateral pigmentary degeneration. British Journal of Ophthalmology, 48(9), pp.471-479. doi: 10.1136/bjo.48.9.471

Massof R, Finkelstein D, Starr S, Kenyon K, Fleischman J, Maumenee I (1979). Bilateral symmetry of vision disorders in typical retinitis pigmentosa. British Journal of Ophthalmology, 63(2), pp.90-96. doi: 10.1136/bjo.63.2.90

Nirmalan P, Krishnaiah S, Nutheti R, Shamanna B, Rao G, Thomas, R (2006). Consanguinity and Eye Diseases with a Potential Genetic Etiology. Data from a Prevalence Study in Andhra Pradesh, India. Ophthalmic Epidemiology, 13(1), pp.7-13. doi: 10.1080/09286580500473795

Saihan, Z, Webster A, Luxon L, Bitner-Glindzicz M (2009). Update on Usher syndrome. Current Opinion in Neurology, 22(1), pp.19-27. doi: 10.1097/wco.0b013e3283218807

Schemann, J, Leplege, A, Keita, T, Resnikoff, S (2002). From visual function deficiency to handicap: Measuring visual handicap in Mali. Ophthalmic Epidemiology, 9(2), pp.133-148. doi: 10.1076/opep.9.2.133.1519

Thapa, S, Thapa, R, Paudyal, I, Khanal, S, Aujla, J, Paudyal, G et al (2013). Prevalence and pattern of vitreo-retinal diseases in Nepal: The Bhaktapur glaucoma study. BMC Ophthalmology, 13(1). doi: 10.1186/1471-2415-13-9

Weleber R (1993). Phenotypic Variation Including Retinitis Pigmentosa, Pattern Dystrophy, and Fundus Flavimaculatus in a Single Family with a Deletion of Codon 153 or 154 of the Peripherin/RDS Gene. Archives of Ophthalmology, 111(11), p.1531. doi: 10.1001/archopht.1993.01090110097033

Zafar S, Ahmad K, Ali A, Baig R (2017). Retinitis pigmentosa genes implicated in South Asian populations: a systematic review. Journal of the Pakistan Medical Association, 67(11). Pmid: 29171570