

## Pattern of retinal degenerations and dystrophies in a hilly state of India

Anubhav Chauhan<sup>1</sup>, Kulbhushan Prakash Chaudhary<sup>1</sup>,  
Gian Chand Rajput<sup>1</sup>

<sup>1</sup>Department of Ophthalmology, Medical College, Nahan, District Sirmour, Himachal Pradesh, India.

**Corresponding author:** Dr. Anubhav Chauhan

Address: Pine Castle, Near Mist Chamber, Khalini, Shimla 171002, Himachal Pradesh, India;  
Telephone: +919816991482; E-mail: chauhan.anubhav2@gmail.com

### Abstract

**Aim:** To describe the pattern and distribution of Retinal Degenerations and Dystrophies in the hilly terrain of Himachal Pradesh (HP) (altitude ranging from 500-4500 meters above sea level).

**Methods:** This was an ambispective study of patients with retinal diseases attending the general ophthalmology clinic of a tertiary care facility at Shimla from August 2008 to April 2013. Out of 5600 subjects, 4323 were taken as a sample. The data was taken from the hospital records and thereafter analyzed to determine their age, sex distribution and diagnosis. All patients underwent visual acuity, refraction, slit lamp examination and fundus evaluation. The diagnosis was confirmed from fundus clinic records and evaluation of fundus photographic records retro-prospectively. The photographs were taken on the fundus camera (Kowa's Fundus Camera VX-10) and fundus fluorescein angiography (FFA) done where ever indicated.

**Results:** Amongst the 293 (6.77%) subjects of Retinal Degenerations and Dystrophies, retinitis pigmentosa (1.99%) was the most common condition, while choroideremia (0.02%), pattern dystrophy (0.02%), Alport syndrome (0.02%), Sjögren Larsson syndrome (0.02%), lattice degeneration (0.02%), and Wagner syndrome (0.02%) were the least common diseases.

**Conclusion:** Retinal disorders appear to be a major public health problem in India. The present study shall help us in planning the management of such disorders in the hilly state of Himachal Pradesh to reduce the visual morbidity arising out of such disorders.

**Keywords:** Himachal Pradesh, India, retina, retrospective.

## Introduction

Unlike the affections in anterior segment of the eye, the diseases affecting retina are mostly irreversible and often sight threatening. After the 1981 survey there have been several population based studies in Nepal that have reported the prevalence of blindness from retinal diseases within a range of 1% to 10.8% (1). Vitreo-retinal diseases as a group are one of the more common ocular morbidities leading to blindness in the adult population, while being the most common cause of blindness worldwide in children. In the developing world, which harbours almost 90% of the world's blind population, retinal diseases are among the leading cause of blindness after cataract (2).

Retinal diseases have had a low priority in prevention of blindness programmes in developing countries. Population-based surveys reported vitreo-retinal disorders to be responsible for 8.56% and 12.7% in Iran and India respectively (3). Recently, there has been a significant increase in the burden of vitreo-retinal disorders globally. In Nigeria, vitreo-retinal disorders constitute a significant cause of ocular morbidity and vision loss with reported hospital prevalence rates ranging from 4.5% to 13.0%. Elsewhere in Ethiopia, a 12.5% hospital prevalence of vitreo-retinal disorders was reported (4).

Retinal dystrophies are rare diseases defined by specific clinical and molecular features (5). Generalized retinal dystrophy is a frequent cause of visual impairment and blindness in younger individuals and a subject of new clinical intervention trials (6). Inherited retinal degenerations affect approximately 1 in 3000 people (0.2 million people worldwide) (7).

Retinitis pigmentosa (RP) is the most common form of inherited retinopathy, with a reported prevalence of approximately 1 in 3500 (8). Incidence of newly diagnosed cases per year is about six per 1,000,000 population according to a study (9). Available population-based studies on the prevalence of retinitis pigmentosa have been conducted mainly in populations in the Western countries but, for Asia,

only few studies have addressed and there is no specific information about India (10).

After undergoing an extensive literature search on the internet and to the best of our knowledge, no study on the pattern/distribution of all the retinal degenerations and dystrophies together has been carried out worldwide and only a few studies of isolated diseases classified under retinal degenerations and dystrophies have been carried out. In this ambispective study of retinal diseases at a tertiary care facility of Shimla hills, we determined the prevalence of retinal degenerations and dystrophies among the patients who reported for photographic evaluation.

## Methods

### *Subjects*

The present study was conducted in a tertiary care facility of Shimla hills. A total of 5600 subjects from all districts of H.P visiting the fundus clinic of a tertiary care institute were evaluated during a period from August 2008 to April 2013. From these 5600 patients, 4323 subjects were taken as a sample. It is a retrospective and prospective study. We confirm adherence to the guidelines of the Declaration of Helsinki as well as the hospitals ethics committee approval.

### *Procedures*

In brief the present study involved 4323 subjects residing in H.P (altitude ranging from 500-4500 meters above sea level). H.P is a hilly terrain and has a very distinct population that is composed of ethnolinguistic groups of tribals and socials. Most of the natives belong to Aryan origin while the people of Lahaul and Spiti district are essentially descendants of Mongols. Patients coming from all districts of H.P underwent visual acuity, refraction, slit lamp examination and pupil dilatation for detailed fundus evaluation. The diagnosis was confirmed from hospital records, fundus clinic records and evaluation of fundus photographic records retro-prospectively. Inclusion criteria included proper and

complete records of the patient with clear fundus photographs and FFA where as exclusion criteria included fundus photographs/FFA taken on fundus camera not clearly visible for making a diagnosis and patients presenting with opaque ocular media.

In all the subjects, ophthalmological examination was performed. Visual acuity was measured by using Snellen's chart, Slit lamp biomicroscopy was done to assess the ocular adnexa and the anterior segment of eye using a slit lamp biomicroscope (Haag Striet-900), Fundus examination was done by using the direct and indirect ophthalmoscope. Fundus photographs were taken on the fundus camera (KOWA'S FUNDUS CAMERA VX-10, KOWA Company Ltd, 4-14, Nihonbashi-honcho 3-chome, Chuo-ku, Tokyo 103-8433 Japan). Fluorescein Angiography was performed wherever indicated.

### Statistical analysis

Data collected was managed on an excel spreadsheet. Distributions were determined by using percentages.

### Results

During the period from August 2008 to April 2013, 5600 patients visiting the fundus clinic of the tertiary care institution were evaluated. From these 5600 patients, 4323 subjects were taken as a sample for the study. Since the present study was aimed to find out the pattern of retinal degenerations and dystrophies, other retinal disorders were not included.

Table 1 shows that of the total 4323 cases studied, there were more males 2563 (59.28%) than females 1760 (40.72%) with fundus diseases.

**Table 1. Gender distribution of cases**

Male / Female	Total	Percentage
Male	2563	59.28 %
Female	1760	40.72 %
Total	4323	100

**Table 2. Retinal Degenerations and Dystrophies**

Disease	Total	Percentage
RP	86	1.99 %
BD/VD	3	0.07 %
Choroideremia	1	0.02 %
GA	16	0.37 %
SD	19	0.44 %
FDD	37	0.86 %
PDS	1	0.02 %
COD	2	0.05 %
OHMD	19	0.44 %
FBFR	6	0.14 %
FA	4	0.09 %
Alport syndrome	1	0.02 %
PPCA	4	0.09 %
SLS	1	0.02 %
LAD	1	0.02 %
DCD	72	1.67 %
Wagner syndrome	1	0.02 %
GCD	19	0.44 %
Total	293	6.77 %

Table 2 depicts that out of the 293 (6.77%) Retinal Degenerations & Dystrophies, RP was present in 86 (1.99%), BD/VD (best disease/vitelliform dystrophy) in 3 (0.07%), choroideremia in 1 (0.02%), GA (gyrate atrophy) in 16 (0.37%), SD (Stargardt disease) in 19 (0.44%), FDD (familial dominant drusen) in 37 (0.86%), PDS in 1 (0.02%), COD (cone dystrophy) in 2 (0.05%), OHMD (other heredomacular degenerations) in 19 (0.44%), FBFR (familial benign fleck retina) in 6 (0.14%), FA (fundus albipunctatus) in 4 (0.09%), Alports syndrome in 1 (0.02%), PPCA (pigmented paravenous chorioretinal atrophy) in 4 (0.09%), SLS in 1 (0.02%), LAD in 1 (0.02%), DCD (diffuse chorioretinal degeneration) in 72 (1.67%), Wagner syndrome in 1 (0.02%) and GCD (generalised choroidal dystrophy) in 19 (0.44%) subjects.

## Discussion

Though Himachal Pradesh is a hilly terrain consisting of twelve districts, the different districts have different types of geographic and socioeconomic conditions. There is a diversity of culture, language, customs, food habits and way of life. Yet our study represents the patients residing in Himachal Pradesh. In the present study, a greater number of male patients with fundus diseases was observed as compared to females. These results are similar to other studies carried out (1,4,11,12). Prevalence of RP in population aged 30 years and above was estimated at 1:750 in a study from rural Central India (10). In another study, 7461 (95.9%) subjects had fundus details seen in both the eyes. Thirteen subjects (0.17%; 4 males, 9 females) were diagnosed as RP. RP in the urban population was seen in approximately 1 in 930 persons, while 1 in 372 of rural subjects had the disorder (13). A prevalence of 1:3996 for RP was found in another study. The mean age of the study population was

50.4 with percentage of female to male being 52% and 48% respectively (14). Prevalence of RP from a study came out to be 21 per 100,000 population or 1:4,756. Incidence of newly diagnosed cases per year was about six per 1,000,000 population (9). According to a study, prevalence of 1 in 6023 was estimated in the Slovene population. The highest prevalence of 1 in 1902 was found in the age group 65 years and older (15). Age distribution, sex predominance, rural/urban distribution and unilateral/bilateral ocular involvement of RP were not taken in our study.

RP was detected in 32 (0.3%) subjects in one study (16). While in another study, out of 4027 subjects, RP was diagnosed in four subjects (all males) (17). Out of 10,100 subjects, 44 (0.44%) had RP (18). In our study, it was detected in 86 (1.99%) patients. The difference between the previous studies and our study might be due to difference in the sample size or geographic variations. RP was the most frequent (47%) retinal dystrophy found in a study while 30% subjects had Stargardt disease and 11% had Best disease (5). In the present study also, RP was the most common disease encountered.

The findings of my study cannot be compared to, or benchmarked against similar other studies owing to the fact that no study material could be traced on the internet and similar public domains despite my dedicated efforts to this effect. Nevertheless, we consider that more studies for pattern/distribution of all retinal degenerations and dystrophies should be carried out so that an in-depth knowledge regarding the diseases pattern is known.

In conclusion, retinal disorders appear to be a major public health problem in India. The results of this study gave an insight into the pattern of retinal degenerations and dystrophies in the hilly state of Himachal Pradesh.

**Conflicts of interest:** None declared.

## References

1. Karki DB, Malla OK, Byanju RN, Shrestha S. Analysis of 400 cases of posterior segment diseases visiting retina clinic of Nepal eye hospital. *Kathmandu Univ Med J* 2003;1:161-5.
2. Thapa SS, Thapa R, Paudyal I, Khanal S, Aujla J, Paudyal G, et al. Prevalence and pattern of vitreo-retinal diseases in Nepal: the Bhaktapur glaucoma study. *BMC Ophthalmol* 2013;13:9.
3. Khan A, Riaz Q, Soomro F, Qidwai U, Qazi U. Frequency and Patterns of Eye Diseases in Retina Clinic of a Tertiary Care Hospital in Karachi. *Pak J Ophthalmol* 2011;27:155-9.
4. Eze BI, Uche JN, Shiweobi JO. The burden and spectrum of vitreo-retinal diseases among ophthalmic outpatients in a resource-deficient tertiary eye care setting in South-Eastern Nigeria. *Middle East Afr J Ophthalmol* 2010;17:246-9.
5. Bocquet B, Lacroux A, Surget MO, Baudoin C, Marquette V, Manes G, et al. Relative Frequencies of Inherited Retinal Dystrophies and Optic Neuropathies in Southern France: Assessment of 21-year Data Management. *Ophthalmic Epidemiol* 2013;20:13-25.
6. Bertelsen M, Jensena H, Bregnhøj JF, Rosenberga T. Prevalence of Generalized Retinal Dystrophy in Denmark. *Ophthalmic Epidemiol* 2014;21:217-23.
7. Bessant DA, Ali RR, Bhattacharya SS. Molecular genetics and prospects for therapy of the inherited retinal dystrophies. *Curr Opin Genet Dev* 2001;11:307-16.
8. Sullivan LS, Bowne SJ, Birch DG, Hughbanks-Wheaton D, Heckenlively JR, Lewis RA, et al. Prevalence of Disease-Causing Mutations in Families with Autosomal Dominant Retinitis Pigmentosa: A Screen of Known Genes in 200 Families. *Invest Ophthalmol Vis Sci* 2006;47:3052-64.
9. Bunker CH, Berson EL, Bromley WC, Hayes RP, Roderick TH. Prevalence of Retinitis Pigmentosa in Maine. *Am J Ophthalmol* 1984;97:357-65.
10. Nangia V, Jonas JB, Khare A, Sinha A. Prevalence of retinitis pigmentosa in India: The Central India Eye and Medical Study. *Acta Ophthalmol* 2012;90:e649-50.
11. Teshome T, Melaku S, Bayu S. Pattern of retinal diseases at a teaching eye department, Addis Ababa, Ethiopia. *Ethiop Med J* 2004;42:185-93.
12. Oluleye TS, Ajaiyeoba AI. Retinal diseases in Ibadan. *Eye (Lond)* 2006;20:1461-3.
13. Sen P, Bhargava A, George R, Ve Ramesh S, Hemamalini A, Prema R, et al. Prevalence of retinitis pigmentosa in South Indian population aged above 40 years. *Ophthalmic Epidemiol* 2008;15:279-81.
14. O'Neill JJ, McKay GJ, Simpson DA, Silvestri G. The Epidemiology of Retinitis Pigmentosa in Northern Ireland. *Invest Ophthalmol Vis Sci* 2007;48:3724.
15. Peterlin B, Cankl-Klain N, Morela V, Stirn B, Rainer S, Cerar V. Prevalence of retinitis pigmentosa in Slovenia. *Clin Genet* 1992;42:122-3.
16. You QS, Xu L, Wang YX, Liang QF, Cui TT, Yang XH, et al. Prevalence of retinitis pigmentosa in North China: The Beijing Eye Public Health Care Project. *Acta Ophthalmol* 2013;91:e499-500.
17. Xu L, Hu L, Ma K, Li J, Jonas JB. Prevalence of retinitis pigmentosa in urban and rural adult Chinese: The Beijing Eye Study. *Eur J Ophthalmol* 2006;16:865.
18. Tous HM, Izquierdo NJ. Retinitis pigmentosa in Puerto Rico. *P R Health Sci J* 2006;25:315-8.