Clinical Study of Prevalence of Retinitis Pigmentosa in Tertiary Care Hospital

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ABSTRACT

Introduction: Retinitis pigmentosa is a slow and degenerative disease of the retina which leads to blindness. So the study was designed to know the prevalence of Retinitis Pigmentosa in patients attending tertiary care hospital, Guntur for blind certificate

Material and Methods: A total of 665 patients of all age groups attending to tertiary care hospital, Guntur for blind certificate during the period July 2016 to June 2017 were included and complete ophthalmological examination was done which includes best corrected visual acuity, slit lamp examination, fundus examination, visual field testing, intra ocular pressure measurement.

Results: Out of 665 patients, 86 patients had Retinitis Pigmentosa with involvement of both eyes. Of these 86 patients, 52 were males and 34 were females indicating that males were more commonly affected than females.47 patients had 100% disability, 27 patients had 75% of disability, 12 patients had 40% of disability.

Conclusion: Retinitis Pigmentosa,which is a degenerative retinal disease,results mostly from consanguineous marriages and is a cause of non treatable blindness.

Keywords: Retinitis Pigmentosa, Prevalence, Disability, Consanguineous Marriage, Ushers Syndrome

INTRODUCTION

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[1,2] 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 retina. In the majority of families it appears as a recessive trait and consanguinity of the parents is not infrequent. Occasionally it is associated with syndromes, such as syndrome of obesity, hypogonadism, mental defect and polydactyly (Laurence- Moon-Biedl-Bartum syndrome), deafness (Usher syndrome) cardiac conduction defects and abetalipoproteinaemia

Usher syndrome is the most frequent syndrome associated with RP, which accounts for about 14% of all RP cases.[7]

Bardet Biedl syndrome (BBS) is less frequent than Usher syndrome[8] and is characterized by obesity in childhood, mental retardation or mild psychomotor delay, polydactyly, hypogonadism and renal abnormalities that lead to renal failure,in association with RP (often of cone-rod dystrophy type). BBS is due to genetic mutations[9,10,] with cases of triallelic digenic inheritance.[11]

Retinitis pigmentosa is diagnosed clinically by the presence of night blindness and peripheral visual field defects, fundus changes, subnormal electroretinogram changes, and gradual worsening of these symptoms and signs.

Study aimed to record the prevalence of Retinitis Pigmentosa in patients attending tertiary care hospital, Guntur for blind certificate in relation to age, sex, percentage of disability and association with syndromes.

MATERIAL AND METHODS

The present study was conducted on 665 patients (randomly selected) who attended tertiary care hospital, Guntur for blind certificate during the period july 2016 to june 2017 and we included the patients of all age groups. All the patients underwent complete ophthalmological examination which included best corrected visual acuity by snellen chart[12], slitlamp examination, fundus examination with indirect ophthalmoscope[13], visual field testing by Humphrey field analyser, intra ocular pressure measurement by applanation tonometer.

STATISTICAL ANALYSIS

Descriptive statistics like mean and percentages were used for the analysis. Statistical analysis was done with the help of Microsoft office 2007.

RESULTS

The present study was conducted on 665 patients who attended regional eye hospital, Guntur for blind certificate during the period july 2016 to june 2017. Out of 665 patients, 86 patients had Retinitis Pigmentosa with involvement of both eyes. Of these 86 patients, 52 were males and 34 were females indicating that males were more commonly affected

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than females.47 patients had 100% disability, 27 patients had 75% of disability, 12 patients had 40% of disability.

Out of 86 patients who were diagnosed as cases of retinitis pigmentosa, 52 cases were males and 34 cases were females, showing male preponderance. All age groups were included in the study, and the peak age of presentation was 21 to 30 years age group [Table 1].

Depending on the above categories, the percentage of disability was calculated and among the 86 cases,12 patients had 40% disability, 27 patients had 75% of disability, 47 patients had 100% of disability [Table 2,3].

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.[7]

Out of 86 cases, 78 cases were not associated with any syndromes (non syndromic). 6 cases were diagnosed as Usher syndrome and 2 cases were diagnosed as Bardet Biedl syndrome [Table 4].

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[1,2] 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 retina. In the majority of families it appears as a recessive trait and consanguinity of the parents is not infrequent.

Bardet Biedl syndrome (BBS) is less frequent than Usher syndrome[8] and is characterized by obesity in childhood, mental retardation or mild psychomotor delay, polydactyly, hypogonadism and renal abnormalities that lead to renal failure,in association with RP (often of cone-rod dystrophy type

Diagnostic criteria

Symptoms

- Night blindness (nyctalopia) is the earliest symptom
- Tubular vision

Signs

- Loss of peripheral vision, Ring scotoma, and eventually Tubular vision
- Bony corpuscular pigment deposits, initially at the equator
- Arteriolar attenuation
- Consecutive optic atrophy with Waxy pallor of the optic disc

RP is one of the non treatable cause of blindness, with a high prevalence in Southern India.

In our study conducted on 665 patients, 86 patients were found to have Retinitis pigmentosa. Of these 86 patients 52(60.46%) were males and 34(39.53%) were females with male preponderance, indicating that RP is more common in males. Our results showed the peak age of presentation as 21 to 30 years (47.67%). A study conducted by Joshi et al in central India among patients attending the outpatient department for blindness certificate, reported that retinitis pigmentosa constituted 15.05% of blindness.[17] In our study, retinitis pigmentosa constituted 13% of blindness amongst patients attending outpatient department for visual handicap certification.

Regarding the percentage of disability, 12(13.95%) patients had 40% disability, 27(31.39%) patients had 75% disability and 47(54.65%) patients had 100% disability.

Out of 86 patients of RP, 78(90.69%) patients were non syndromic RP and 8(9.3%) patients were syndromic RP out of which 6 were diagnosed as Usher syndrome and 2 were diagnosed as Bardet Biedel syndrome.

Out of 665 patients,183 patients were from urban area and 482 were from rural area. The high prevalence of cases from

Age	Males	Females	Total
0-10	2	1	3(3.48%)
11-20	9	7	16(18.60%)
21-30	24	17	41(47.67%)
31-40	8	5	13(15.11%)
41-50	5	3	8(9.30%)
>50	4	1	5(5.81%)
	52(60.46%)	34(39.53%)	86(100%)

Table-1: Effect of age and sex

	40%	75%	100%	Total
Males	9	17	26	52(60.46%)
Females	3	10	21	34(39.53%)
	1 2(13.95%)	27(31.39%)	47(54.65%)	86(100%)

Table-2: percentage disability according to sex

Better Eye	Worse Eye	% Impairment
6/18-6/36	6/60 to Nil	40%
6/60-4/60 or field of vision 100-200	3/60 to Nil	75%
3/60 to 1/60 or field of vision < 100	F.C. at 1 ft. to Nil	100%
F.C. at 1 ft. to Nil or field of vision < 100	F.C. at ft. to Nil	100%

Table-3: Percentage of Disability by visual acuity and visual fields

Males	47	5
Females	31	3
	78(90.69%)	8(9.3%)
Table-4: Non syndromic RP Syndromic RP		

rural area is due to consanguineous marriages. Nirmalan et al.[18] 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.

In a study conducted by Sen et al[19] on prevalence of retinitis pigmentosa in south indian population aged above 40 years, retinitis pigmentosa was seen in 1 in 372 in rural population and 1 in 930 in urban population approximately.

CONCLUSION

Retinitis Pigmentosa is the most common pigmentary retinal dystrophy. Studies reported a much higher prevalence in South Indian population compared to western populations and population in the other parts of India. Role of consanguinous marriages in India must be brought forward in this context. Retinitis pigmentosa is an inherited disease which runs in the families. Since it is one of the non treatable cause of blindness, it is vital to reduce the prevalence, hence we need to concentrate on counselling to reduce consanguineous marriages.

REFERENCES

1. Schémann JF, Leplège A, Keita T, Resnikoff S. From visual function deficiency to handicap: Measuring visual handicap in Mali. Ophthalmic Epidemiol 2002;9:133-48.
2. WHO study group. The prevention of blindness. WHO Technical Report Series No. 518, 1973. Available from: htt p://whqlibdoc.who.int/trs/WHO_TRS_518.pdf [last accessed on 2016. August 29].
3. "Bardet-Biedl (Laurence Moon)"
4. Koenekoop, Robert K; Loyer, Magali; Hand, Collette K; Al Mahdi, Huda; Dembinska, Olga; Beneish, Raquel; Racine, Julie; Rouleau, Guy A. Novel RPGR mutations with distinct retinitis pigmentosa phenotypes in French-Canadian families. American Journal of Ophthalmology. 2003;136: 678–87
5. Understanding Retinitis Pigmentosa
6. Farrar, G.J.; Kenna, P. F.; Humphries, P. NEW EMBO MEMBer's REVIEW: On the genetics of retinitis pigmentosa and on mutation-independent approaches to therapeutic intervention. The EMBO Journal. 2002;21: 857–64.
7. Boughman JA, Vernon M, Shaver KA: Usher syndrome: definition and estimate of prevalence from two high-risk populations. J Chronic Dis. 1983;36: 595-603.
8. Beales PL, Elcioglu N, Woolf AS, Parker D, Flinter FA: New criteria for improved diagnosis of Bardet-Biedl syndrome: results of a population survey. J Med Genet. 1999;36: 437-446.
9. Dollfus H, Verloes A, Bonneau D, Cossée M, Perrin-Schmitt F, Brandt C, Flament J, Mandel J-L: Le point sur le syndrome de Bardet-Biedl. J Fr Ophthalmol. 2005;28: 106-112.
10. Hichri H, Stoetzel C, Laurier V, Caron S, Sigaudy S, Sarda P, Hamel C, Martin-Coignard D, Gilles M, Leheup B, Holder M, Kaplan J, Bitoun P, Lacombe D, Verloes A, Bonneau D, Perrin-Schmitt F, Brandt C, Besancon AF, Mandel JL, Cossee M, Dollfus H: Testing for triallelism: analysis of six BBS genes in a Bardet-Biedl syndrome family cohort. Eur J Hum Genet. 2005; 13: 607-616.
11. Katsanis N, Ansley SJ, Badano JL, Eichers ER, Lewis RA, Hoskins BE, Scambler PJ, Davidson WS, Beales PL, Lupski JR: Triallelic inheritance in Bardet-Biedl syndrome, a Mendelian recessive disorder. Science. 2001;293: 2256-2259.
12. Ferris FL, Kassoff A, Bresnick GH, Bailey I. New visual acuity charts for clinical research. Am J Ophthalmol. 1982;94:91– 96.
13. Humphrey Field Analyzer II User's Guide. San Leandro: Humphrey Instruments Inc, 1994. 9
14. Kaplan J, Bonneau D, Fre'zal J. et al. Clinical and genetic heterogeneity in Retinitis pigmentosa. Hum. Genet 1990;85:635-42.
15. Foxman SG, Heckenlively JR, Bateman JB., et al. Classification of congenital and early onset RP. Arch Ophthalmo 1985;103:1502- 6.
16. Gawande AA, Donovan WJ, Ginsburg AP, et al.Photoaversion in RP. Br J Ophthalmol 1989;73:337-41.
17. Joshi RS. 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 2013;1:17-
18. Nirmalan PK, Krishnaiah S, Nutheti R, Shamanna BR, Rao GN, Thomas R. Consanguinity and eye diseases with a potential genetic etiology. Data from a prevalence study in Andhra Pradesh, India. Ophthalmic Epidemiol 2006;13:7-13.
19. Sen P, Bhargava A, George R, Ve Ramesh S, Hemamalini A, Prema R, Kumaramanickavel G, Vijaya L. Prevalence of retinitis pigmentosa in South Indian population aged above 40 years.

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