Prevalence of blindness and cataract surgery in Gandaki Zone, Nepal

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WORLD VIEW

Prevalence of blindness and cataract surgery in Gandaki Zone, Nepal

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Background/aim: Himalaya Eye Hospital (HEH), established in 1993, is rendering eye care services in the Gandaki and Dhaulagiri zones. The hospital has extensive community outreach activities along with services for outpatients, emergencies, and subspecialties such as vitreoretinal, paediatric, and low vision. The operation theatre is well equipped to match the surgical needs including phaco surgery for cataract. The hospital has performed more than 14 000 cataract surgeries and 250 000 treatment services during this period. The aim of this study was to estimate the prevalence of blindness, visual impairment, and cataract surgical coverage among the older adult population of three districts of Gandaki Zone, where 80% of the hospital's service recipients reside.

Methods: People aged 45 years and older were enrolled in the study using a stratified cluster design. Subjects in 25 randomly selected clusters from the listed 806 were recruited through door to door visits. Each recruited subject had visual acuity (VA) and clinical examination conducted by an ophthalmologist. The survey was preceded by pre-pilot and pilot studies to refine the operational method. To assess quality assurance the interobserver variation in VA measurement was also carried out in five different clusters.

Results: Out of 5863 selected subjects 85.3% were examined. Blindness defined as presenting VA <6/60 in both eyes was found in 2.6% (95% confidence interval (CI): 2.2 to 3.9), whereas 16.8% individuals examined had vision <6/19 in one or both eyes. Cataract was the principal cause of blindness in 60.5%, and refractive error was the dominant cause of vision impairment (<6/19) 83.3%. Cataract surgical coverage was 59.5% among the cataract blind and associated with younger age, literacy, and male sex.

Conclusion: The finding suggests a positive impact of the HEH programme on the prevalence of blindness and cataract surgical services in the survey area. Strategies to further improve access and utilisation of facilities and increase cataract surgical coverage need to be developed.

Blindness and severe visual impairment remain leading causes of disability in the world. In 2002 the World Health Organization (WHO) made a global estimate that there are over 37 million blind and another 124 million people with low vision.[1]

In 1981, a national programme for the prevention and control of blindness was launched in Nepal.[2] The programme was preceded by a national epidemiological survey to determine the magnitude, causes, and regional distribution of blindness. The survey reported 0.84% of the population to be blind using the best corrected visual acuity (VA) cut-off of <3/60. The prevalence of blindness among those aged 45 years and older was estimated at 3.77%. Age related cataract was the major cause of blindness—83% among those >45 years (65.4% among all ages).[3]

These survey results were both established and dedicated to specific programme objectives for the prevention and treatment of eye diseases on a need based priority in different areas of the country. Resources from international and national non-governmental organisations were mobilised and coordinated to implement this national strategy. In the year 2002 alone, Nepal performed 111 740 cataract surgeries (Annual Report 2002, Nepal Netra Jyoti Sangh).

Subsequently, a 52 bed Himalaya Eye Hospital (HEH) was established in March 1993 at Pokhara, to serve the two million people of Gandaki and Dhaulagiri zones in western Nepal (two out of 14 zones of Nepal). The study area consists of the mainly rural, valley, hilly, and mountainous region with an altitude ranging from 1000–8000 metres. Agriculture is the main occupation of the population and the beautiful mountain terrain has provided the opportunity for the tourism industry. There are no other eye healthcare providers for the population surveyed. From 1993 to 2002 the hospital performed over 17 063 major surgeries (the majority being cataract) (Annual Report 2002, Himalaya Eye Hospital, Pokhara).

In 1995 a similar survey was carried out in adjacent Lumbini and Bheri zones.[4] The survey findings suggested that blindness prevalence may have decreased since 1981, both overall and cataract related. Severe blindness (all causes) was estimated as 3% versus approximately 3.8% in the 1981 Nepal-wide survey among more than 45 years of age.

The present evaluation was planned to assess the impact of 9 years of programme activities in three of the five districts of Gandaki Zone. This survey was carried out in the year 2002 to (a) estimate current prevalence and causes of blindness; (b) assess impact of hospital services with cataract surgical coverage as the indicator; and (c) assess visual outcomes of people who had undergone cataract surgery. This paper presents results pertaining to the first two objectives. Results of the outcomes of cataract surgery will be presented in a separate paper.

METHODS

We used survey methods, sampling strategy, and examination protocol similar to that used for earlier studies in Nepal, China, and India.[4–7] The target population for the survey was people aged >45 years and resident of Kaski, Lamjung, and Syangja. These districts were chosen for survey as 80% of the clientele of the HEH (based on data from hospital records) were from these districts.

Abbreviations: HEH, Himalaya Eye Hospital; VA, visual acuity

Sampling clusters were created by grouping adjacent village wards with less than 850 people and subdividing those with more than 1700 people into segments, so that each sampling frame cluster had between 850 and 1700 people. The sampling frame contained 806 clusters with a total population of 1 007 070 and 193 634 of more than 45 years of age. The sample size was calculated based on estimating a cataract blindness prevalence (VA <6/60) of 8% within an error bound of 15% with 95% confidence. With an expected examination participation rate of 85% and cluster design effects of 2.0, the required sample size was 4619. We randomly selected 25 clusters from the 806 sampling units with approximately 170–340 people aged more than 45 years in each cluster.

The fieldwork was carried out from September to December 2002. House to house enumeration of all eligible people aged more than 45 years of age (who were living permanently in the cluster for at least the last 6 months) was carried out. Literate people were defined as those who attended at least 1 year of school. All enumerated people were invited for eye examinations in central location.

The eye examination team consisted of one ophthalmologist, two ophthalmic assistants, and one interviewer. Distance VA was measured using a retroilluminated logMAR tumbling E chart (Precision Vision, Villa Park, IL, USA) at 4 metres and at 1 metre if necessary, and was recorded as the smallest line read with one or no errors. A person unable to read the top line from 1 metre was tested for finger counting, hand movement, or light perception. Vision of each eye was measured separately, with their own glasses if used by the subject, and without glasses. Refraction (Streak retinoscopy and subjective) were performed in all people with presenting VA <6/19 in either eye, and in all subjects operated for cataract.

Basic eye examination of eyelid, globe, pupillary reflex, and lens was performed by an ophthalmologist using a torch, two times magnification binocular loupe, hand held slit lamp, and a direct ophthalmoscope. All eyes whose vision did not improve to 6/19 with refraction, except with a corneal cause or obvious cataract (defined as a lens opacity precluding view of the fundus), were dilated for detailed evaluation of the posterior segment. Intraocular pressure was measured using a Perkins hand held tonometer for cases suspected of having glaucoma on the basis of optic disc changes, primarily cup to disc ratios >0.5, and unhealthy optic nerve rim.

All eyes with VA <6/19 were assigned a principal cause of impairment/blindness by the examining ophthalmologist.

Subjects physically unable to come to the examination site were offered an examination at home. Treatment of minor ocular conditions was provided at the examination site free of charge. Those who required cataract surgery were referred to the HEH for free surgery.

A pilot study was carried out in two non-study clusters to standardise the enumeration/examination procedures.

Quality assurance on VA measurement was monitored during the survey period in five randomly preselected survey clusters. Independently, the two study ophthalmic assistants measured VA twice in those who presented with VA <6/19 and in 10% of people with normal vision. The data were analysed using the unweighted kappa statistic. Interobserver agreement for presenting VA between vision categories was 98.1% (unweighted kappa 0.92) and 91.2% (unweighted kappa 0.89) for best corrected VA.

Five vision categories, similar to those used in the previous survey[4] were defined for analysis and reporting: (1) normal or near normal vision >6/19 in both eyes; (2) visual impairment, unilateral or bilateral visual impairment <6/19 to 6/60 in the worse eye and >6/60 in the better eye; (3) unilateral blindness, VA<6/60 in the worse eye and >6/60 in the better eye; (4) moderate bilateral blindness/economic blind, <6/60 in worse eye, and <6/60 to >3/60 in better eye; (5) severe blindness/social blind, <3/60 in both eyes. Estimates (with 95% confidence intervals) of impairment and blindness prevalence were calculated along with that attributed specifically to cataract. Bivariate analysis and multivariate logistic regression was used to investigate potential associations with blindness.

The cause of blindness was analysed for each eye. The prevalence of cataract blindness and cataract surgery was estimated and potential associations with age, sex, and literacy explored in a multiple logistic regression model.

The cataract blindness burden was defined as the sum of those people already operated for cataract in both eyes and unoperated cataract blind. It was not possible to obtain the preoperative vision status of an already operated eye and we made an assumption that both eyes were blind preoperatively if both eyes were operated for cataract, or if one eye was operated and other eye was blind at the time of our examination. Surgical coverage was calculated as the number of bilaterally blind cataract cases operated divided by the number who could have been operated. The denominator includes the already operated bilateral blind (the numerator) plus the unoperated bilaterally blind with cataract being the principal cause of blindness in at least one eye.

Confidence intervals (CI) for prevalence estimates and odds ratios were calculated. We considered a p value <0.05 as significant. Missing values were assumed to be similar in distribution to the available data and were ignored during analysis.

Verbal informed consent was obtained before examination from all people. The examination protocol was the same as it was used in the earlier surveys in Nepal, India, and China and had been cleared by the World Health Organization Secretariat Committee on Research Involving Human Subjects and National Committee for Prevention of Blindness.[4–6]

RESULTS

We examined 5002 (85.3%) people aged 45 years or older out of 5863 enumerated people from 25 randomly selected clusters.

Table 1 shows the demographic characteristics of participants in the study. The mean age of participants was 58.7 (SD 10.9) years. Females were more likely to be examined (n = 2760, 55.2%, age adjusted OR:1.3, 95% CI:1.1 to 1.5). We did not find any significant difference in participation based on the literacy of the subject (age adjusted OR 1.1, 95% CI 0.9 to 1.3). Response rates were higher among those who farmed their own land (n = 3511, 87.0%), and agricultural labourers (n = 33, 94.3%) compared to manual labourers (n = 12, 63.2%) and professionals (n = 132, 62.0%). We could not examine 861 of the enumerated 5863 people, including 308 people who were temporarily unavailable despite repeated visits on the day of examination and 553 people who refused to participate for various reasons. We examined 207 people (4.1%) at their home.

The prevalence of moderate and severe bilateral blindness (vision better eye<6/60) was 2.6% (95% CI: 2.2 to 3.1) based on presenting VA (table 2) and 1.2% (95% CI: 0.9 to1.6) after best corrected VA (table 3). Older age and literacy were associated with vision impairment and blindness (table 4).

The adjusted odds ratios for older people to be blind (reference age group 45–49 years) was 1.65 (95% CI 0.6 to 4.9) for those aged 50–60 years, 4.68 (95% CI: 1.8 to 12.4) for those aged 61–70 years, and 24.01 (95% CI: 9.5 to 60.3) for those aged above 75 years. The adjusted odds ratio for illiterates to be blind (compared to literates) was 3.48 (95% CI: 1.7 to 7.1).

Table 1 Demographic comparison of enumerated and examined patients

	Enumerated (%)	Examined (%)	% Examined
Age group			
45–49	1498 (25.6)	1258 (25.1)	84.0
50–60	2184 (37.3)	1846 (36.9)	84.5
61–70	1291 (22.0)	1125 (22.5)	87.1
70+	890 (15.2)	773 (15.5)	86.9
Sex			
Male	2685 (45.8)	2242 (44.8)	83.5
Female	3178 (54.2)	2760 (55.2)	86.9
Literacy			
Literate	1496 (25.5)	1244 (24.9)	83.2
Illiterate	4366 (74.5)	3757 (75.1)	86.1
All	5863 (100)	5002 (100)	85.3

Table 2 Visual impairment and blindness by age, sex, and literacy based on presenting VA

	Normal/near (%)	Vision impaired (%)	Unilateral blindness (%)	Economic blind (%)	Social blind (%)	Total (%)
Age						
45–49	1202 (95.5)	31 (2.5)	20 (1.6)	3 (0.2)	2 (0.2)	1258 (100)
50–60	1405 (93.4)	49 (3.3)	41 (2.7)	5 (0.3)	5 (0.3)	1505 (100)
61–70	1020 (80.6)	139 (11.0)	82 (6.5)	13 (1.0)	11 (0.9)	1265 (100)
70+	537 (55.1)	227 (23.3)	121 (12.4)	38 (3.9)	51 (5.2)	974 (100)
Sex						
Male	1847 (82.4)	214 (9.5)	128 (5.7)	25 (1.1)	28 (1.2)	2242 (100)
Female	2317 (83.9)	232 (8.4)	136 (4.9)	34 (1.2)	41 (1.5)	2760 (100)
Literacy*						
Literate	1090 (87.6)	96 (7.7)	49 (3.9)	6 (0.5)	3 (0.2)	1244 (100)
Illiterate	3073 (81.8)	350 (9.3)	215 (5.7)	53 (1.4)	66 (1.8)	3757 (100)
All	4164 (83.2%)	446 (8.9%)	264 (5.3%)	59 (1.2%)	69 (1.4%)	5002 (100%)

*Literacy status missing in one case.

Table 3 Visual impairment and blindness by age sex and literacy based best corrected VA by age, sex, and literacy

	Normal/near (%)	Vision impaired (%)	Unilateral blind (%)	Economic blind (%)	Social blind (%)	Total (%)
Age						
45–49	1229 (97.7)	9 (0.7)	17 (1.4)	3 (0.2)	0 (0.0)	1258 (100)
50–60	1445 (96.0)	16 (1.1)	38 (2.5)	2 (0.1)	4 (0.3)	1505 (100)
61–70	1149 (90.8)	34 (2.7)	70 (5.5)	3 (0.2)	9 (0.7)	1265 (100)
70+	741 (76.1)	76 (7.8)	119 (12.2)	11 (1.1)	27 (2.8)	974 (100)
Sex						
Male	2035 (90.8)	72 (3.2)	113 (5.0)	7 (0.3)	15 (0.7)	2242 (100)
Female	2529 (91.6)	63 (2.3)	131 (4.7)	12 (0.4)	25 (0.9)	2760 (100)
Literacy*						
Literate	1173 (94.3)	32 (2.6)	39 (3.1)	0	0	1244 (100)
Illiterate	3390 (90.2)	103 (2.7)	205 (5.5)	19 (0.5)	40 (1.1)	3757 (100)
All	4564 (91.2)	135 (2.7)	244 (4.9)	19 (0.4)	40 (0.8)	5002 (100)

*Literacy status missing in one case.

Cataract was the single most common cause of blindness (VA <6/60) (table 5).

Among the 128 blind, 95 (74.2%) were the result of cataract. Among the cohort that was blind because of cataract, 71 (74.7%) were blind in both eyes and 24 (25.3%) were blind in only one eye. The prevalence of blindness as a result of cataract was 1.9% (95% CI 1.6 to 2.3). If the already operated cataract blind cases are not included in the cataract blind cohort (the general protocol of the hospital is to operate only in blind eyes. As preoperative vision records of bilaterally operated people were not available, it was presumed that these eyes were blind before surgery) the prevalence of cataract blindness among the unoperated would drop to 1.7% (95% CI: 1.4 to 2.1). Uncorrected refractive error (most of them correctable aphakia) was found to be second highest cause of blindness (n = 23). Eleven (47.8%) people had bilateral uncorrected refractive errors and 12 (52.2%) had unilateral uncorrected refractive errors.

Together, unoperated cataract and refractive error account for 82 bilateral blind people and 26 people blind in one eye.

Table 6 shows the specific causes of blindness by eyes. Cataract is responsible for 60.5% of blind eyes. The total number of people operated for cataract was 195. The distribution of the 83 never operated cataract blind people by age, sex, and literacy distribution is shown in table 7.

Never operated cataract blindness was associated with increasing age and schooling (table 8).

We considered people with one eye operated for cataract and the fellow eye unoperated and not currently blind from

Table 4 Prevalence of presenting bilateral blindness (<6/60) by demographics

Age (years)	Number examined	Blindness prevalence No (%)	Adjusted odds ratio (95% CI)
45–49	1258	5 (0.4)	
50–60	1505	10 (0.7)	1.7 (0.6 to 4.9)
61–70	1265	24 (1.9)	4.7 (1.8 to 12.4)*
70+	974	89 (9.1)	24.0 (9.5 to 60.3)**
Sex			
Male	2242	53 (2.4)	
Female	2760	75 (2.7)	1.1 (0.8 to 1.7)
Literacy†			
Literate	1244	9 (0.7)	
Illiterate	3757	119 (3.2)	3.5 (1.7 to 7. 1)***
All	5002	128 (1.4)	

*p<0.01, **p<0.001, ***p<0.05.
†Literacy status missing for one case.

Table 5 Summary of people presenting bilaterally blind because of cataract, refractive error, or other causes

	Left eye			
Right eye	**Cataract**	**Refractive error**	**Other causes**	**All**
Cataract	71 (55.5) [0]	4 (3.1) [1]	7 (5.5) [5]	82 (64.1) [6]
Refractive error	6 (4.7) [3]	11 (8.6) [1]	1(0.8) [1]	18 (14.1) [5]
Other causes	7 (5.5) [3]	1 (0.8) [1]	20 (15.7) [5]	28 (21.9) [9]
All	84 (65.3) [6]	16 (12.5) [3]	28 (21.9) [11]	128 (100) [20]

Data are given as number (%) of people. [..] show number already operated for cataract in one or both eyes.

Table 6 Principal cause of blindness in eyes

Principal cause	Eyes of bilaterally blind people No (%)	Eyes of unilaterally blind people No (%)	All blind eyes No (%)
Cataract	165 (64.5)	152 (57.6)	317 (60.5)
Refractive error	34 (13.2)	27 (10.2)	61 (11.7)
Neglected cataract/secondary glaucoma	1 (0.4)	2 (0.8)	3 (0.6)
PCO/after cataract	2 (0.8)	1 (0.4)	3 (0.6)
Corneal opacity	8 (3.1)	30 (11.4)	42 (8.0)
Surgical complication	1 (0.4)	1 (0.4)	2 (0.4)
Globe disorder	6 (2.3)	12 (4.6)	15 (2.9)
Glaucoma	1(0.4)	4 (1.5)	5 (1.0)
Optic atrophy	2 (0.8)	4 (1.5)	6 (1.2)
Vascular retinopathy	1 (0.4)	2 (0.8)	3 (0.6)
Macular degeneration	28 (10.9)	17 (6.4)	45 (8.7)
Amblyopia	5 (2.0)	5 (1.9)	10 (2.0)
Retinal detachment	2 (0.8)	4 (1.5)	6 (1.5)
Others	0	3 (1.1)	3 (0.6)
Total	256 (100)	264 (100)	520 (100)

PCO, posterior capsule opacification.

cataract as not being bilaterally blind at the time of cataract surgery. Thus, 122 of the 195 cataract operated people were possibly bilaterally blind at the time of surgery. Table 7 shows the surgical coverage 59.5% (95% CI: 52.4 to 66.2) among the cataract blind (VA<6/60). Surgical service coverage was less in elderly, higher in males, and among the literate. The difference found in sex and literacy is statistically significant (p <0.025), and (p<0.01), respectively.

DISCUSSION

The overall blindness with presenting VA of <6/60 in the better eye is 2.6% (95% CI: 2.3 to 2.9) among people aged 45 years or older in this region. We found age and illiteracy to be associated with blindness; this is similar to studies in other populations that used the same protocol.[4–7]

The prevalence of blindness in our survey (using a presenting VA criterion of <6/60) is much lower than the 5.3% reported from the Lumbini and Bheri zones of Nepal. After best correction, the prevalence reduces further to 1.2% (95% CI: 0.9 to 1.4) compared to 3.9% in the Lumbini and Bheri zones.[4] The prevalence of blindness (using a VA cut-off of <3/60 in the better eye) in our study (1.4%) is still lower than the prevalence reported from the Lumbini and Bheri zones (3%) in the western part of Nepal.[4] Similarly, the prevalence of moderate blindness (1.2%), unilateral blind (5.3%), and visual impairment (8.9%) is also lower than the

Table 7 Presenting cataract blindness (VA <6/60) and cataract surgery prevalence by age, sex, and literacy

	Number examined	Never operated cataract blind		Cataract operated				Cataract blindness burden		% Surgical coverage
				All operated		Presumed blind*				
		No	Prevalence†	No	Prevalence†	No	Prevalence†	No	Prevalence†	
Age										
45–49	1258	2	0.2	8	0.6	5	0.4	7	0.6	70
50–59	1505	4	0.3	23	1.5	14	0.9	18	1.2	62.9
60–69	1265	16	1.3	49	3.9	27	2.1	43	3.4	62.7
70+	974	61	6.3	115	11.8	76	7.8	137	14.1	54.9
Sex										
Male	2242	29	1.3	93	4.2	62	2.8	91	4.1	68.1
Female	2760	54	2.0	102	3.7	60	2.8	114	4.1	52.6
Literacy‡										
Yes	1244	6	0.5	49	3.9	25	2.0	31	2.5	80.6
No	3757	77	2.1	146	3.9	97	2.6	174	4.6	55.7
All		83	1.7	195	3.9	122	2.4	205	4.1	59.5

*Includes all bilaterally operated people and unilaterally operated people with a blind fellow eye.
†Crude prevalence per 100 examined subjects.
‡Literacy is missing for one case.

Table 8 Relation of age and sex with cataract blindness and cataract surgery for people with without schooling

	Never operated cataract blindness	All cataract operated	Cataract blindness burden
Age			
45–49	1.0	1.0	1.0
50–59	1.7 (0.3–9.3)	2.5 (1.1–5.6)*	2.2 (0.9–5.3)
60–69	8.1 (1.9–35.5)**	6.7 (3.1–14.2)**	6.3 (2.8–14.2)**
70 +	43.1 (10.5–177.3)**	22.7 (10.9–47.2)**	29.5 (13.6–64.0)**
Sex			
Male	1.0	1.0	1.0
Female	1.6 (1.0–3.6)	1.3 (0.9 –1.8)	1.2 (0.9–2.1)
Schooling			
⩾1 year	1.0	1.0	1.0
None	2.7 (1.1–6.4)*	0.6 (0.5–1.0)	1.4 (0.9–2.1)

Data are given as adjusted odds ratios (95% confidence intervals), obtained by multiple logistics regression. Separate models were fitted for those with versus those without schooling. *p<0.01, **p<0.05.

finding of 1995 survey in Lumbini and Bheri zones (2.3%, 7.4%, and 13.4%, respectively). Age related cataract still remains the principal cause of blindness in this population despite the focus on providing cataract surgery to the needy.

The cataract surgical coverage in this population is 59.5%, higher than the 42.0% reported from the Lumbini and Bheri zones.[4] We find that the cataract surgical coverage decreases with increase in age even as the risk of blindness from cataract increases. Younger cataract blind people who could be more physically active are more likely to be operated for cataract than the elderly cataract blind population, although recruitment was not different between young (50–60) and old (60+). This finding is consistent to the findings of studies conducted earlier in China[5] and India,[6 7] where cataract surgery was found to be associated with younger age group. Although there is no difference in prevalence of blindness between sexes, cataract surgical coverage was higher in males (68.1%) than females (52.6%, OR 1.92, 95% CI 1.0 to 3.6).[5] This may possibly be because of differential access to services with males travelling more and thus having better access to cataract surgery than females. These findings are similar to previous studies from Nepal[4] and India.[5–9] A higher cataract surgical coverage among literates is also consistent with previous findings from Nepal[4] and India.[7–9] However, the principal study outcomes are from the area best served by HEH. The surgical coverage, although very encouraging and substantial, may well be better than other areas that are not so well covered by the hospital. The best served areas of HEH which were included in the study are within 30 km radius of the hospital location and also have a reasonably good transport service.

Results from our study suggest a positive impact of the HEH programme on the prevalence of blindness and cataract surgical services in the survey area. Strategies to further improve access and utilisation of facilities and increase cataract surgical coverage need to be developed.

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ECHO

Juvenile ARRP and LCA have common mutations

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A sequencing study has shown mutations in common genes in juvenile autosomal recessive retinitis pigmentosa (ARRP) and Leber's congenital amaurosis (LCA), suggesting that the diseases are closely related and explaining their clinical similarity.

Mutations in the commonest genes causing LCA were found in a panel of unrelated patients with juvenile ARRP, juvenile isolated retinitis pigmentosa (IRP), and LCA but not in controls. Mutations in CRB1 and GUCY2D were found in RP and LCA; in RPGRIP1 in RP; and in RPE65 in LCA only. The RPGRIP1 mutation was formerly known only in LCA

Two new mutations were discovered: deletions causing frameshift mutations in GUCY2D, usually occurring in LCA, in a patient with IRP and in RPGRIP1 in a patient with IRP. A new combination of two mutations in CRB1, was detected in a patient with ARRP. In all, nine mutations were found in 12 of 35 patients and seven new, 15 known, and three possible pathogenic polymorphic sequence changes. Clinical characteristics overlapped among patients, 17 of whom had juvenile ARRP, nine juvenile IRP, and nine LCA.

The study hinged on whether clinical and genetic similarities between ARRP and LCA might indicate mutations in common genes. RP affects 1:4000 worldwide; juvenile RP overlaps with LCA, which occurs at a rate of 1: 35000 worldwide. No fewer than 17 genes have so far been identified in ARRP and eight in LCA, accounting for only half of cases. However, all but three LCA genes (AIPL1, CRX, and RPGRIP1) have already been implicated in ARRP.

▲ Booij JC, *et al*. *Journal of Medical Genetics* 2005;**42**:e67.doi10.1136/jmg.2005.035121