STUDENTS' CORNER
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# Frequency of nystagmus in retinitis pigmentosa patients

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## Abstract

**Objective:** To determine the frequency of nystagmus experienced by patients with retinitis pigmentosa.

**Method:** The descriptive study was conducted at the University of Lahore Teaching Hospital, Lahore, Pakistan, from May to August 2024, and comprised patients aged 5-50 years receiving care for retinitis pigmentosa. To assess the kind and degree of retinitis pigmentosa, the patients were subjected to anterior segment examination, cover-uncover test, and pen torch examination. Data was analysed using SPSS 20.

**Results:** Of the 25 patients with a mean age of 27.16±11.28 years, 16 (64%) were females. Overall, 15 (60%) patients were classified as blind, and 5 (20%) retained normal visual acuity. Nystagmus was present in 8 (32%) of the participants, predominantly among those with blindness ($p$= 0.377). Nystagmus was more frequent in females (6/16; 37.5%) than males. However, the difference was not statistically significant ($p$=0.734).

**Conclusion:** Bilateral blindness was the predominant outcome, with nystagmus affecting nearly one-third of patients regardless of visual acuity.

**Keywords:** Nystagmus, Patient, Retinitis pigmentosa. (JPMA 76: 991; 2026)

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## Introduction

Through the manipulation of light, the eye functions as an optical instrument to create a picture. The cornea of the eye has a clear layer through which light is refracted.

Together, the iris and pupil control how much light is transmitted during this process. A lens located beyond the pupil aids in image focus by focussing light on the retina, the back of the eye.[1] Rods and cones are specialised photoreceptor cells that line the inside of the retina. The eye's image is formed in part by these light-sensitive tissues. Rods that function in low light levels perceive peripheral vision. Conversely, cones are in charge of the image's colorectal and visual details.[2] The brain's visual processing centre receives signals from the rods and cones through the process of photo transduction, processes them, and interprets the information, followed by synthesis of the interpretation signals. This pathophysiology is what makes retinitis pigmentosa (RP) and other eye disorders and diseases more significant.[3] RP, or genetic retinal degeneration, gradually impairs vision, and, with more than 1.5 million cases worldwide, RP is the most common inherited retinal dystrophy (IRD). There are two forms of RP: syndromic and non-syndromic. Night blindness is the initial symptom of non-syndromic RP, which progresses to visual loss and finally reaches the peripheral vision range.[4] Following a few earlier reports of possible RP, including possibly the first fundus drawing of a patient with RP (or choroideremia) by Dutch physician Van Trigt, shortly after the invention of the ophthalmoscope, the term "retinitis pigmentosa" was first used by Dutch physician F.C. Donders in 1857.[5] Although digenic inheritance has also been reported, RP is mostly a monogenic illness, with the majority of genetic variations linked to the disease being expressed in photoreceptor or retinal pigment epithelium (RPE) cells. Since every gene serves a distinct purpose, genetic variations cause various biochemical alterations in the retina. RPE cells and photoreceptors eventually degenerate as a result of these alterations. Over 90 genes have been connected to RP so far, and, as diagnostic testing methods continue to advance, this number is probably going to rise.[6] RP is the name for the deterioration of the eye's photoreceptor rods and cones. It is an inherited eye condition affecting the retina.[7]

RP refers to a broad group of hereditary vision illnesses that cause the retina, the light-sensitive membrane lining the inside of the eyes, to gradually deteriorate[8] Peripheral vision, often known as side vision, progressively fades, and disappears in most people. Central vision is usually preserved in these situations until a later phase.[9] RP is associated with a number of systemic illnesses, including deafness, metabolic and neurological illnesses, kidney

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problems, and many more abnormalities.[10] RP affects 1 in 4,000 healthy individuals worldwide.[11] There are three types of hereditary diseases for RP: X-linked (5-15%), autosomal dominant (30-40%) and autosomal recessive (50-60%).[12] Cones are in charge of the colour perception of vision on the retina.[13] Two varieties of cones exist. Some cones are responsive to short wavelength S and the blue colour of the visual spectrum, while, according to the visual spectrum, some cones are sensitive to medium wavelengths, which are green.[14] There is a connection between distinct cones with different genetic bases and different opsins. The S cone opsins are autosomal connected to chromosome 7. The M and L cones, on the other hand, are connected to X chromosomes. The two cones are nearly the same. Their homologous recombination process accounts for the variation in green-red colour blindness severity. It highlights deviations from typical spectral sensitivity. Because colour blindness is gender-linked, it affects both male and female karyotypes. Cone opsins M and L are more prevalent, and it has been observed that by the time they are two months old, newborns can see colour.[15] Two months later, the nervous system begins to produce normal colour perception, which carries out visual transduction and sends data from the brain's visual cortex. It is crucial for distinguishing orange, red and yellow colours. Having a normal mechanism is essential for performing domestic tasks.[16] The most common cause of blindness and visual impairment in those aged <60 years is RP,[17] which includes a class of IRDs that are characterised by progressive vision loss.[18] Since there are currently no therapies to halt the course of the illness or restore vision, RP is regarded as incurable. The goals of therapy are to alleviate problems, prevent vision loss, and assist patients in managing the psychological effects of their illness.[17] The rods and cones along retinal cells may degenerate in RP patients. This deterioration leads to a reduction in visual acuity (VA), which results in night blindness and vision loss. The central nervous system's impairment of vision is the cause of this. The brain uses visual data to cause an involuntary eye movement as a compensation strategy. Nystagmus is the term for this involuntary eye movement.[19] Nystagmus is described as having involuntary eye movements as well as rhythmic, repetitive eye movements. These motions can be in a circular, horizontal or vertical direction.[20] In RP, nystagmus develops to improve or maintain the eye's visual fixation. The purpose of the eye movement is to help focus on areas where vision is still clear. This adaptation approach is established to perceive the remaining visual input and the impact of a disturbance in vision.[21] Depending on the severity and course of the RP, nystagmus has been observed in some patients. RP patients do not immediately experience nystagmus; rather, it develops as a secondary cause when the visual impairment worsens.[22] Seesaw nystagmus is a rare kind of nystagmus characterised by cyclical eye movements, frequently including the extorsion and depression of one eye and the intorsion and elevation of the other. Seesaw nystagmus is thought to be brought on by injury to the interstitial nucleus of cajal, which is situated in the meso-diencephalic region, and/or its fibres connect to the spinal cord, oculomotor, trochlear and vestibular nuclei.[23] The intensity, frequency, and direction of a person's vision as well as their perception of it are all affected by nystagmus. RP causes fixation instability, which reduces the amount of visual input and raises the risk of developing nystagmus. Furthermore, nystagmus and decreased central vision make reliance on peripheral vision worse. This makes it difficult for an RP person to concentrate on visual targets.[24]

The current study was planned to ascertain the prevalence of nystagmus in RP patients, and to assess the role that fixation instability plays in its development.

## Patients and Methods

The descriptive study was conducted at the University of Lahore Teaching Hospital, Lahore, Pakistan, from May to August 2024. After approval from the institutional ethics review committee, the sample was raised using non-probability convenience sampling technique. Informed consent was obtained before data collection from all the subjects. The sample size was determined using G*Power with a 95% confidence interval (CI).[25] Those included were patients of either gender aged 5-50 years receiving RP care, supported by a detailed family history indicating the presence of RP in relatives such as siblings, cousins or grandparents. Those with a history of ocular surgery, systemic or vascular conditions, or neurodegenerative diseases, like multiple sclerosis, that could affect orthotic assessment were excluded. Additionally, patients with pituitary adenomas were excluded.

In addition to basic demographics, ocular history and family history of RP were recorded. Along with the basic history, previous nystagmus history was also noted. Refraction was performed to evaluate the best-corrected VA (BCVA) and VA, or eyesight. Both the near and far vision were recorded. The pen torch fixation test was used to assess nystagmus. The pen torch was to be the patient's only focus. The patient's fixation on the pen torch was evaluated to determine whether or not the patient had involuntary eye movements. A cover and uncover test was used to look for hidden nystagmus symptoms. An occluder was used, which involved holding it in front of the eyes for a short while. After taking off the occluder, the eye's fixation

was examined. An ophthalmoscope was used to view the fundus during the anterior eye examination portion of the fundus examination process.

Data was analysed using SPSS 26. Data was expressed as frequencies and percentages or as mean±standard deviation. The chi-square test was used to examine the association between nystagmus and VA in both eyes. P<0.05 was considered significant.

## Results

The detailed results of the study demonstrate that out of the 25 patients included in the study, the mean age was 27.16±11.28 years. The majority of participants were female 16 (64%), whereas males comprised 9 (36%). In terms of visual acuity, 15 (60%) patients were classified as blind, 5 (20%) had normal visual acuity, 2 (8%) had moderate visual impairment, and 3 (12%) had severe visual impairment, as detailed in Table 1.

Table 2 shows the prevalence of nystagmus among the various visual acuity groups of oculus dexter (OD). Nystagmus was also observed more in the blind patients (6/15), with none having this condition in the patients with normal visual acuity. OD statistical analysis indicated that there was no significant correlation between nystagmus and visual acuity ($p$=0.377), as indicated in Table 3. Table 4

**Table-1:** Descriptive data of best-corrected visual acuity (BCVA) oculus dexter (OD).

| BCVA-OD | | n (%) |
|---|---|---|
| **Valid** | blindness | 15 (60.0) |
| | moderate visual impairment | 2 (8.0) |
| | Normal | 5 (20.0) |
| | Severe visual impairment | 3 (12.0) |
| | Total | 25 (100) |

**Table-2:** Descriptive data of nystagmus oculus dexter (OD).

| Crosstab Nystagmus | | | | |
|---|---|---|---|---|
| **Count** | | **Nystagmus** | | **Total** |
| | | **No** | **Yes** | |
| **BCVA-OD** | blindness | 9 | 6 | 15 |
| | moderate visual impairment | 1 | 1 | 2 |
| | Normal | 5 | 0 | 5 |
| | Severe visual impairment | 2 | 1 | 3 |
| **Total** | | 17 | 8 | 25 |

BCVA: Best-corrected visual acuity.

**Table-3:** Chi-square data of oculus dexter (OD).

| Chi-Square Tests | Value | df | *p*-value |
|---|---|---|---|
| Pearson Chi-Square | 3.094a | 3 | 0.377 |
| N of Valid Cases | 25 | - | - |

**Table-4:** Descriptive data of best-corrected visual acuity (BCVA) oculus sinister (OS).

| | BCVA- OS | n (%) |
|---|---|---|
| **Valid** | blindness | 15 (60.0) |
| | moderate visual impairment | 2 (8.0) |
| | Normal | 5 (20.0) |
| | Severe visual impairment | 3 (12.0) |
| | **Total** | 25 (100) |

**Table-5:** Descriptive data of nystagmus oculus sinister (OS).

| Crosstab Nystagmus | | | | |
|---|---|---|---|---|
| **Count** | | **Nystagmus** | | **Total** |
| | | **No** | **Yes** | |
| **BCVA-OS** | blindness | 8 | 7 | 15 |
| | moderate visual impairment | 2 | 0 | 2 |
| | Normal | 5 | 0 | 5 |
| | Severe visual impairment | 2 | 1 | 3 |
| **Total** | | 17 | 8 | 25 |

**Table-6:** Chi-square data of oculus sinister (OS).

| Chi-Square Tests | Value | df | *p*-value |
|---|---|---|---|
| Pearson Chi-Square | 4.779a | 3 | 0.189 |
| N of Valid Cases | 25 | - | - |

**Table-7:** Descriptive data of nystagmus.

| Nystagmus | n (%) |
|---|---|
| No | 17 (68.0) |
| Yes | 8 (32.0) |
| **Total** | 25 (100) |

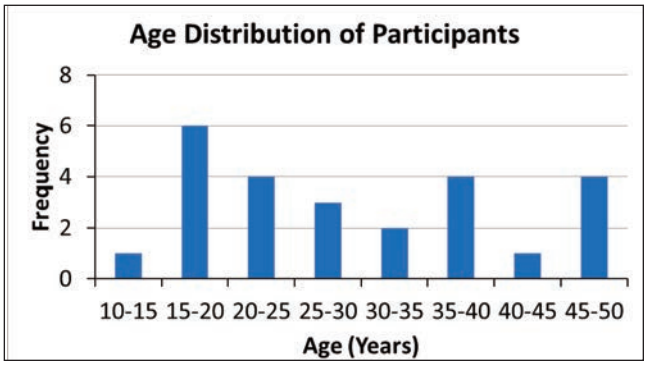


**Figure-1:** Descriptive data of age.

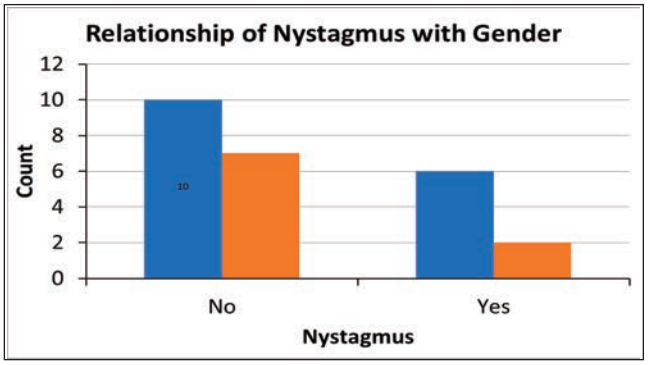


**Figure-2:** Relationship of nystagmus with gender.

presents the distribution of best-corrected visual acuity of oculus sinister (OS), with most of the patients categorised as blind (60%). Table 5 gives the relationship between nystagmus and visual acuity in OS, and again, the majority of cases are found in blind patients (7/15), and no case had normal visual acuity. The statistical analysis of OS, however, also did not reveal any significant association ($p$=0.189) as presented in Table 6. In all, 8 (32) patients were found to have nystagmus, as summarised in Table 7.

When stratified by gender, nystagmus was more frequently observed in females (6/16; 37.5%) compared to males (2/9; 22.2%). However, this difference did not reach statistical significance ($p$=0.734), as illustrated in Figures 1 and 2.

## Discussion

The current study demonstrated a 32% (8/25) prevalence of nystagmus in RP patients, mostly unrelated to visual acuity. An earlier study, aimed at determining the alteration in eye motility among 23 RP patients, showed that both genders had VA 6/10, and that RP patients showed involuntary eye movement. However, severe visual impairment or high refractive error alone may not fully explain the occurrence of nystagmus.[26] It also reported absent vestibular function with pendular high-frequency oscillations (~6Hz), suggesting a vestibular origin. This difference indicates that nystagmus in RP may arise from multiple mechanisms rather than solely from visual impairment.[27] A study in 2019 was done among RP patients who had visual impairment over the preceding three months. Patients undergoing Quatrix P12 intraocular lens (IOL) of the right eye and Rayner 620H IOL of the left eye were included. On examination, subluxation of IOL and dense opacities were seen within the IOL. Results showed positive nystagmus signs among these patients with a VA of 20/200. It was concluded that there was an increase in cases of IOL opacification and nystagmus rate among RP patients.[28] The current study showed a 32% prevalence of nystagmus in RP patients, primarily among those with severe vision loss, with no significant link to VA. In contrast, a study reported preserved CA (20/32 or better) and reduced optokinetic response (OKR) gain in subjects with Usher syndrome type 2A (USH2A) gene (USH2A) variants, indicating impaired ocular motor function despite relatively good vision, suggesting that nystagmus in RP may not always correlate with the degree of visual impairment, but could also reflect underlying genetic or neuro-ophthalmic factors.[29] The current study reported a 32% prevalence of nystagmus in retinitis pigmentosa (SRP, predominantly in those with profound vision loss (60% blind), while a study on Phosphodiesterase 6A (PDE6A) gene-associated RP demonstrated progressive, but less severe visual decline over an average of 4.8 years.[30] Unlike the current cohort with early, profound impairment, patients in the earlier study had a wider age range (12-76 years) and slower progression, with structural changes, such as reduction in ellipsoid zone width and persistent cystoid macular oedema.[30] This contrast highlights the heterogeneity of RP, where certain genetic subtypes exhibit gradual degeneration, while others lead to early severe vision loss and higher nystagmus prevalence.[30]

The current study has several limitations that may influence the interpretation of the findings. The relatively short study duration limited participant recruitment, preventing the inclusion of a broader and more diverse population. Resource constraints further restricted the scale of data collection, thereby reducing the statistical power and depth of analysis. Additionally, the rarity of RP posed significant challenges in accessing a larger patient pool, which may have impacted the representativeness of the sample and limited the ability to draw more generalised conclusions.

Despite the limitations, however, the current study has several implications. In terms of clinical management, the high prevalence of bilateral blindness (60%) and notable occurrence of nystagmus (32%) in RP patients underscore the need for early screening, visual rehabilitation, and low-vision interventions to enhance functional outcomes and quality of life. Besides, the variability in nystagmus prevalence and severity of visual impairment suggests a possible genetic or phenotypic influence, which may guide personalised management strategies. Early onset of profound vision loss indicates significant socioeconomic and educational impacts, warranting tailored support systems and policies for affected individuals.

Future studies should investigate how specific genetic mutations influence the severity of visual loss and nystagmus development in RP patients. Longitudinal studies with long-term follow-up are needed to track the progression of nystagmus and visual impairment. There is also a need to explore the role of vestibular and ocular motor dysfunction in RP-related nystagmus through advanced imaging and electrophysiological studies. Studies should also assess how nystagmus and vision loss collectively affect mobility, balance and quality of life to inform rehabilitation strategies.

## Conclusion

Nystagmus occurrence was notable in RP patients. Bilateral blindness was the predominant outcome, with nystagmus affecting nearly one-third of patients regardless of visual acuity.

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**MH:** Design and writing.
**MM, MS & MI:** Data collection.
**UE:** Final review and editing.
**MZ:** Statistical analysis and final approval.