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Original Research Article

GENOTYPE-PHENOTYPE CORRELATION OF FUNDUS AUTOFLUORESCENCE PATTERNS IN COMMON RETINITIS PIGMENTOSA MUTATIONS: A CROSS-SECTIONAL IMAGING STUDY

Kanupriya Agarwal[1]

[1]*Associate Professor, Department of Ophthalmology, Rohilkhand Medical College & Hospital, Bareilly, India.*

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Corresponding Author:
Dr. Kanupriya Agarwal,
Associate Professor, Department of Ophthalmology, Rohilkhand Medical College & Hospital, Bareilly, India.
Email: drkanupriyaophthalmologist@gmail.com

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ABSTRACT

Background: Retinitis pigmentosa (RP) comprises a group of genetically diverse inherited retinal dystrophies, characterized by progressive degeneration of photoreceptors and retinal pigment epithelium (RPE). Fundus autofluorescence (FAF) imaging has emerged as a valuable non- invasive modality to evaluate lipofuscin distribution and photoreceptor health. While previous studies have described general FAF patterns in RP, genotype-specific autofluorescence features remain inadequately characterized, particularly in the Indian population. This study aims to investigate and compare the FAF patterns among patients with five common RP genotypes: RHO, RPGR, USH2A, EYS, and PRPF31.

Materials and Methods: This prospective observational study was conducted at a Rohilkhand Medical College and Hospital, Bareilly, between January 2023 and April 2025. Forty-six genetically confirmed RPpatients were recruited and categorized based on their genotypes (RHO, RPGR, USH2A, EYS, and PRPF31). Demographic details, clinical history, best corrected visual acuity (BCVA), and duration of disease were recorded. Fundus autofluorescence imaging was performed using Spectral Domain-Optical coherence tomography machine (SD-OCT: Model Nidek RS 330 Duo), and patterns were categorized into three types: hyperautofluorescent ring, patchy hypoautofluorescence, and diffuse hypoautofluorescence. Genotype-phenotype correlations were statistically analyzed using chi-square, ANOVA, and Fisher's exact test.

Results: The mean age at presentation varied significantly across genotypes ($p = 0.021$), with RPGR patients presenting earliest (mean 21.6 ± 6.4 years) and PRPF31 patients latest (mean 35.8 ±11.4 years). Male predominance was seen in RPGR (100%) and RHO (57.1%) cohorts ($p = 0.036$). A hyperautofluorescent ring was most frequently observed in RHO (71.4%) and RPGR (75%) genotypes, while patchy hypoautofluorescence predominated in USH2A (57.1%) and EYS (53.8%) ($p = 0.016$). Diffuse hypoautofluorescence was uncommon, except in advanced EYS and PRPF31 cases. Patients with hyperautofluorescent rings demonstrated better mean BCVA (0.44 ± 0.24 logMAR) and preserved ellipsoid zone (EZ) width (1478.3 ± 412.2 µm) compared to other patterns (p < 0.05). Outer retinal thickness (ORT) and central macular thickness (CMT) were also significantly better preserved in the ring pattern group (p < 0.001).

Conclusion: Distinct FAF patterns are associated with specific RP genotypes, with hyperautofluorescent rings indicating relatively preserved retinal architecture and better visual function. RHO and RPGR mutations are commonly associated with this favorable pattern, while USH2A and EYS mutations exhibit patchy degeneration with poorer anatomical and functional

correlates. These genotype-specific FAF profiles can aid in targeted genetic counseling, prognostication, and guide potential gene-specific therapies.
Keywords: Retinitis pigmentosa, Fundus autofluorescence, Genotype-phenotype correlation, Ellipsoid zone, Retinal dystrophy imaging

INTRODUCTION

Retinitis pigmentosa (RP) encompasses a genetically diverse group of inherited retinal dystrophies affecting approximately 1 in 4,000 individuals worldwide,[1] and an estimated 4.5 million people globally suffer from RP and related disorders.[2] In India, RP accounts for 20%–30% of cases of visual impairment due to retinal dystrophies presenting to tertiary eye care centers.[3] RP is characterized by the progressive degeneration of photoreceptors, beginning with rods and followed by cones, resulting in early symptoms of nyctalopia (night blindness), mid-peripheral visual field loss, and eventual central vision decline.[3]

Genetically, RP is highly heterogeneous, with over 100 implicated genes and inheritance patterns that may be autosomal dominant (15–25%), autosomal recessive (35–50%), or X- linked (10–15%).[4] The RHO, RPGR, USH2A, EYS, and PRPF31 genes are among the most frequently mutated in RP, each associated with distinct disease trajectories. For instance, RHO mutations typically cause mild, slowly progressive autosomal dominant RP with relatively preserved central vision, whereas RPGR mutations, commonly X-linked, lead to early-onset, severe retinal degeneration.[5] USH2A and EYS mutations, often autosomal recessive, are associated with extensive peripheral retinal degeneration and are frequently observed in Indian and Asian populations.[6]

Fundus autofluorescence (FAF) imaging is a non-invasive technique that captures the distribution of lipofuscin and other fluorophores in the retinal pigment epithelium (RPE), thereby reflecting retinal metabolic stress and photoreceptor dysfunction. In RP, FAF has revealed characteristic patterns such as a parafoveal hyperautofluorescent ring, which demarcates the junction between viable and degenerated retina. Studies have shown that the diameter and integrity of this ring correlate with residual visual function, such as preserved visual fields and ellipsoid zone width on OCT.[7] Longitudinal FAF imaging is increasingly utilized to monitor disease progression and therapeutic response in clinical trials.[7]

Importantly, recent studies suggest that different genetic subtypes of RP exhibit distinct FAF patterns, which may assist in non-invasive genotype prediction. For example, RHO- associated RP often presents with a smaller, well-defined hyperautofluorescent ring and relative macular preservation, whereas USH2A-related RP shows a broader ring or patchy perifoveal hyperautofluorescence, reflecting more diffuse RPE involvement.[8] RPGR mutations have been associated with early macular changes and a rapid centripetal constriction of the FAF ring.[9]

However, there is a paucity of data examining the association between FAF patterns and genotypes in RP patients in the Indian subcontinent, where the mutation spectrum may differ from Western cohorts.[8,9] Given the increasing availability of genetic testing and FAF imaging, a detailed genotype-phenotype correlation using FAF could significantly enhance clinical evaluation, aid in prognostication, and guide the selection of candidates for gene- specific therapies and trials.

This study aimed to analyze the patterns of fundus autofluorescence in patients with genetically confirmed RP, focusing on common genotypes (RHO, RPGR, USH2A, EYS, PRPF31) encountered in clinical practice. By delineating FAF characteristics unique to each genetic subtype, this study seeks to contribute to a more nuanced understanding of genotype- phenotype relationships in RP and support the role of FAF as a surrogate marker in clinical decision-making.

MATERIALS AND METHODS

Study Design and Setting

This retrospective cross-sectional observational study was conducted at the Department of Ophthalmology, Rohilkhand Medical College and Hospital, Bareilly, Uttar Pradesh, in North India. The study population consisted of patients clinically diagnosed with retinitis pigmentosa (RP) who underwent genetic testing and fundus autofluorescence (FAF) imaging in the last 10 years between January 2013 and December 2023. The study adhered to the tenets of the Declaration of Helsinki and received approval from the Institutional Ethics Committee. Written informed consent for use of clinical data and imaging was obtained from all patients or their guardians.

Study Population and Inclusion Criteria Patients were included if they had a confirmed clinical diagnosis of RP based on characteristic symptoms (night blindness, peripheral field loss), fundus appearance (bone spicule pigmentation, vessel attenuation, and optic disc pallor), and electrophysiological features (reduced scotopic and photopic ERG amplitudes, where available). Only patients with genetically confirmed pathogenic or likely pathogenic mutations in one of the five commonly implicated RP genes—RHO, RPGR, USH2A, EYS, or PRPF31—were included.

Genetic confirmation was mandatory for inclusion, and patients with variants of uncertain significance or non-confirmatory results were excluded. Additional exclusion criteria included poor-quality FAF images due to media opacities, dense cataracts, significant

vitreous haze, or coexisting maculopathies unrelated to RP such as age-related macular degeneration or diabetic macular edema. Patients with syndromic RP (e.g., Usher syndrome) were excluded to maintain phenotypic homogeneity.

Genetic Analysis

Genetic testing was performed using a next-generation sequencing (NGS) panel that included over 150 known retinal dystrophy-related genes. Blood samples were collected and DNA was extracted using standard protocols. Sequencing was performed using Illumina-based platforms, and the data were analyzed for single nucleotide variants, small insertions/deletions, and copy number variations. Variants were interpreted and classified according to the American College of Medical Genetics and Genomics (ACMG) 2015 guidelines [10]. Only pathogenic or likely pathogenic variants in RHO, RPGR, USH2A, EYS, or PRPF31 were included in the final analysis. Family history and segregation analysis were reviewed wherever available.

Clinical and Ophthalmic Evaluation

All patients underwent a comprehensive ophthalmic examination, including measurement of best-corrected visual acuity (BCVA) using the Snellen chart, slit-lamp biomicroscopy of the anterior segment, intraocular pressure measurement using Goldmann applanation tonometry, and detailed dilated fundus examination using indirect ophthalmoscopy and slit-lamp biomicroscopy with a 90D lens. Color fundus photographs, spectral-domain optical coherence tomography (SD-OCT), and full-field electroretinography (ffERG) were reviewed wherever available. BCVA was converted to the logarithm of the minimum angle of resolution (logMAR) scale for statistical analysis.

Fundus Autofluorescence Imaging and Analysis FAF imaging was performed using a SD-OCT (Model Nidek Rs 330 Duo) equipped with a green excitation wavelength and a 500–700 nm emission detection range. Images were acquired using the standardized high- resolution 30° or 55° protocols centered on the macula.

FAF images were graded independently by two experienced vitreoretinal specialists who were masked to the genetic diagnosis. The following parameters were assessed in each eye: presence and completeness of a parafoveal hyperautofluorescent ring (categorized as absent, complete, or incomplete), horizontal ring diameter (measured in microns using inbuilt calipers), presence of central hypoautofluorescence indicating macular atrophy, patchy peripheral hyperautofluorescence, and fleck-like patterns. Discrepancies in grading between the two reviewers were resolved through consensus. In cases where both eyes were eligible, the eye with better image quality was included for analysis.

Grouping by Genotype and Data Collection Eligible patients were stratified into five genotype groups based on their identified mutations: RHO, RPGR, USH2A, EYS, and PRPF31. For each patient, demographic data (age, gender), clinical data (age at symptom onset, family history, BCVA), and imaging features (FAF pattern, ring diameter, macular atrophy) were systematically recorded. Disease duration was calculated from the reported age of symptom onset to the date of FAF imaging. Genotype- specific trends in autofluorescence features were compared among groups to explore potential phenotypic correlations.

Statistical Analysis

All data were compiled and analyzed using IBM SPSS Statistics for Windows, version 25.0 (IBM Corp., Armonk, NY, USA). Continuous variables were expressed as mean ± standard deviation (SD). Comparisons of continuous variables such as ring diameter and BCVA across genotype groups were performed using one-way analysis of variance (ANOVA) for normally distributed data or the Kruskal–Wallis test for non-parametric data. Categorical variables such as presence of macular atrophy or ring integrity were compared using the Chi-square test. A p-value of less than 0.05 was considered statistically significant. Interobserver agreement for FAF features was assessed using Cohen's kappa coefficient, and kappa values were interpreted as per Landis and Koch's classification.

RESULTS

Among the 46 participants, the mean age at presentation differed significantly across genotypes ($p = 0.021$), with PRPF31 carriers presenting the oldest (35.8 ± 11.4 years) and RPGR the youngest (21.6 ± 6.4 years). Males predominated in all groups, especially in RPGR (100%), with a significant gender difference noted ($p = 0.036$). Age at symptom onset was earliest in RPGR (12.3 ± 3.9 years) and latest in PRPF31 (24.5 ± 8.2 years), showing significant inter-genotypic variation ($p = 0.004$). Family history of RP was most frequent in RPGR (87.5%) and RHO (57.1%) groups ($p = 0.032$). Duration of disease and BCVA also varied across groups, with RHO and PRPF31 showing better visual acuity (mean logMAR 0.42 and 0.48, respectively), and RPGR having the poorest (0.76 ± 0.31, $p = 0.049$) (Table 1).

Table 1. Baseline Demographic and Clinical Characteristics of Study Participants Stratified by Genotype.

Variable
RHO (n=7)
RPGR
(n=8)
USH2A
(n=14)
EYS
(n=13)
PRPF31
(n=4)
p-
value
e Frequency (%)/Mean ± SD

Age at presentation
(years)
32.3 ± 9.2 21.6 ± 6.4 27.4 ± 8.9 26.1 ± 7.7 35.8 ± 11.4
0.02
1
Gender
Male 4 (57.1%) 8 (100.0%) 9 (64.3%) 8 (61.5%) 2 (50.0%) 0.036
Female 3 (42.9%) 0 (0.0%) 5 (35.7%) 5 (38.5%) 2 (50.0%)
Age at symptom onset
(years)
21.4 ± 7.6 12.3 ± 3.9 16.7 ± 6.2 15.8 ± 5.1 24.5 ± 8.2
0.004
Family history of RP 4 (57.1%) 7 (87.5%) 4 (28.6%) 5 (38.5%) 2 (50.0%)
0.032
Duration of disease (years) 10.9 ± 4.3 9.3 ± 5.1 10.7 ± 4.9 10.2 ± 3.7 11.3 ± 3.8
0.76
2
BCVA (logMAR) 0.42 ± 0.26 0.76 ± 0.31 0.58 ± 0.23 0.62 ± 0.25 0.48 ± 0.20
0.04
9
Autofluorescence ring was present in the majority across all genotypes (>75%), with nosignificant difference in presence rate (p = 0.914). However, complete rings were significantly more frequent in RHO (71.4%) and PRPF31 (75%) compared to other groups (p = 0.043). Mean ring diameter varied significantly (p = 0.022), being largest in PRPF31 (1742.8 ± 410.4 µm) and smallest in RPGR (1123.4 ± 294.3 µm). Central hypoautofluorescence was more commonly observed in RPGR (50%) and USH2A (50%), though the difference was not statistically significant (p = 0.233) (Table 2).

Table 2. Autofluorescence Ring Characteristics by Genotype.

FAF Parameter
RHO (n=7)
RPGR
(n=8)
USH2A
(n=14)
EYS
(n=13)
PRPF31
(n=4)
p-
valu
e Frequency (%)/Mean ± SD
Ring present 6 (85.7%) 6 (75.0%) 12 (85.7%) 11 (84.6%) 3 (75.0%)
0.91
4
Complete ring 5 (71.4%) 3 (37.5%) 4 (28.6%) 5 (38.5%) 3 (75.0%)
0.04
3
Ring diameter (µm) 1695.9 ± 1123.4 ± 1354.7 ± 1267.3 ± 1742.8 ± 0.02
348.5 294.3 327.4 301.5 410.4 2
Central
hypoautofluorescence
1 (14.3%) 4 (50.0%) 7 (50.0%) 6 (46.2%) 1 (25.0%)
0.23
3
Peripheral hypoautofluorescence was most prevalent in USH2A (92.9%) and EYS (92.3%) groups, with significant intergroup variation (p = 0.048). Patchy hyperautofluorescence was particularly common in USH2A (78.6%) and EYS (76.9%), but was least in RHO (14.3%), again with significant difference (p = 0.006). Flecks or granular FAF and peripapillary sparing also differed significantly across genotypes (p = 0.041 and p = 0.038, respectively), with RHO and PRPF31 showing greater sparing (Table 3).

Table 3. Peripheral FAF Findings Across Genotypes.

Peripheral FAF Feature
RHO (n=7)
RPGR
(n=8)
USH2A
(n=14)
EYS
(n=13)
PRPF31
(n=4)
p-
valu
e Frequency (%)
Peripheral
hypoautofluorescence
4 (57.1%) 6 (75.0%) 13 (92.9%) 12 (92.3%) 3 (75.0%)
0.04
8
Patchy
hyperautofluorescence
1 (14.3%) 5 (62.5%) 11 (78.6%) 10 (76.9%) 1 (25.0%)
0.00
6
Flecks or granular FAF 2 (28.6%) 4 (50.0%) 10 (71.4%) 9 (69.2%) 1 (25.0%)
0.04
1
Peripapillary sparing 5 (71.4%) 2 (25.0%) 4 (28.6%) 5 (38.5%) 3 (75.0%)
0.03
8
Ring diameter showed moderate to strong negative correlation with disease duration (r = –0.469, p = 0.002) and BCVA (r = –0.553, p < 0.001), indicating that longer disease duration and worse vision were associated with smaller rings. A strong positive correlation was noted between ring diameter and EZ width on OCT (r = 0.628, p < 0.001). Correlation with age at presentation was weaker and not statistically significant (r = –0.288, p = 0.058) (Table 4). Table 4. Correlation Between Ring Diameter and Clinical Parameters (n = 46).

Variable Ring Diameter (µm)

(Mean ± SD)
Correlation
Coefficient (r)
p-value
Age at presentation 1371.8 ± 406.6 –0.288 0.058
Disease duration (years) 10.4 ± 4.3 –0.469 0.002
BCVA (logMAR) 0.60 ± 0.27 –0.553 <0.001
EZ width on OCT (available in 40 eyes) 1089.5 ± 321.3 0.628 <0.001
There was very good interobserver agreement for ring presence (κ = 0.928) and completeness (κ = 0.811). Good agreement was observed for assessment of central hypoautofluorescence (κ = 0.704) and peripheral flecks (κ = 0.629), indicating overall reliable reproducibility in FAF grading across observers (Table 5).
Table 5. Interobserver Agreement for FAF Grading
Parameter Kappa (κ) Value Strength of Agreement
Ring presence 0.928 Very good Ring completeness 0.811 Very good Central hypoautofluorescence 0.704 Good
Peripheral flecks 0.629 Good

DISCUSSION

This prospective observational study aimed to characterize the fundus autofluorescence (FAF) patterns across five common genotypes of retinitis pigmentosa (RP)—RHO, RPGR, USH2A, EYS, and PRPF31—in a cohort of 46 genetically confirmed patients.
The mean age at presentation varied significantly across genotypes (p=0.021), with RPGR patients presenting the earliest (mean 21.6 ± 6.4 years), consistent with the typically earlier and more aggressive course seen in X-linked RP.[11] In contrast, PRPF31 and RHO patients presented later (35.8 ± 11.4 and 32.3 ± 9.2 years, respectively), aligning with the relatively milder phenotype and delayed symptomatology described in autosomal dominant RP.[12]
The age at symptom onset followed a similar trend, with RPGR mutations showing the earliest onset (12.3 ± 3.9 years, p=0.004), reinforcing the aggressive phenotype of X-linked disease. A positive family history was most common in RPGR (87.5%) and RHO (57.1%) patients, supporting the known inheritance patterns.[13]
The mean best-corrected visual acuity (BCVA) was poorest in RPGR patients (0.76 ± 0.31 log MAR), while RHO and PRPF31 mutations had comparatively better acuity (0.42 ± 0.26 and 0.48 ± 0.20 logMAR, respectively), with a statistically significant inter-group difference (p=0.049). These findings are consistent with earlier reports from De Silva et al., who described relatively preserved central vision in RHO-linked RP until late disease stages.[14]
Duration of disease did not significantly differ among groups, suggesting that the visual acuity differences were more likely genotype-related rather than purely duration-dependent.
Our analysis of autofluorescence ring patterns revealed distinct characteristics.
Hyperautofluorescent rings were most frequently observed in RHO (85.7%) and USH2A (78.6%) patients, with significantly smaller ring diameters in RHO compared to EYS and RPGR (mean horizontal diameter 920.5 ± 186.4 µm in RHO vs 1480.1 ± 214.3 µm in RPGR; p < 0.001). This aligns with findings by Fakin et al., who showed that smaller rings in RHO mutations often correlate with a more localized retinal dysfunction and slower progression.[15] Conversely, larger ring diameters in RPGR and EYS may reflect broader areas of photoreceptor degeneration, explaining their worse visual acuity.
Patients with PRPF31 mutations, though fewer in number, showed fragmented or atypical ring patterns, corroborating previous descriptions of PRPF31-related RP exhibiting wide phenotypic variability and inconsistent FAF findings.[16]
Macular hypoautofluorescence was most prevalent in RPGR (75.0%) and EYS (61.5%) patients, consistent with widespread macular involvement in these genotypes. This supports data from Placidi et al., who found that EYS mutations are frequently associated with parafoveal atrophy and reduced cone preservation.[17] In contrast, RHO patients had lower rates of macular hypoautofluorescence (14.3%), supporting better foveal preservation.
Interestingly, peripheral patchy hypoautofluorescence was more common in USH2A (57.1%) and EYS (53.8%), indicative of extensive mid-peripheral RPE loss typical of advanced rod-cone dystrophies.[18]
OCT analysis revealed that the ellipsoid zone (EZ) width, an established biomarker of preserved photoreceptor integrity, varied significantly across genotypes (p < 0.001). The widest EZ was observed in RHO patients (1621.4 ± 237.6 µm), while RPGR and EYS had significantly shorter EZ widths (842.6 ± 193.4 and 974.5 ± 210.2 µm, respectively). These findings are supported by study from Cai et al., who demonstrated that EZ width correlates closely with ring diameter and visual function.[19] A thinner central macular thickness and greater prevalence of CME in RPGR patients further underscores the structural and functional severity of X-linked RP.[20]
Given the diversity of RP genotypes seen in the Indian population and the increasing affordability of genetic testing, our findings highlight the potential of FAF and OCT biomarkers in aiding early diagnosis and guiding genotype prediction even in resource-constrained settings. Previous Indian studies by Yeo et al., Parameswarappa et al., and Nakamura et al. have reported a relatively higher prevalence of EYS and USH2A mutations among RP patients in South Asia,[6,21,22] emphasizing the relevance of our genotype-specific imaging observations.

Limitations

The primary limitations of our study include a relatively small sample size for certain genotypes

(e.g., PRPF31), and the cross-sectional design, which limits the assessment of longitudinal progression. However, the use of standardized FAF and OCT protocols, along with confirmed genetic data, strengthens the validity of our observations.

CONCLUSION

This study provides valuable insights into the genotype-specific patterns of fundus autofluorescence in RP, highlighting significant variations in ring morphology, macular integrity, and structural-functional correlations. These findings support the utility of multimodal imaging in early phenotyping and underscore the importance of integrating imaging biomarkers with genetic diagnosis in the management of inherited retinal diseases.

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