

Case Report



Identification of a Pathogenic Variant in the *Retinitis Pigmentosa 1* Gene in a Sudanese Family with Retinitis Pigmentosa: A Case Report

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Abstract

Introduction

Retinitis Pigmentosa (RP) is a hereditary retinal dystrophy characterized by progressive visual impairment and marked genetic heterogeneity, with more than 90 associated genes reported. Identifying disease-causing variants is important for establishing a molecular diagnosis, understanding inheritance patterns, and guiding genetic counseling and management. This case report aimed to characterize the clinical and molecular findings in individuals with RP and to identify genetic variants potentially associated with their phenotype using next-generation sequencing (NGS).

Case presentation

Two individuals (IV-7 and IV-6) underwent premarital genetic screening, which revealed they are first cousins because their parents are siblings. The female carrier (IV-7) reported gradual vision loss and flashes, while the male carrier (IV-6) showed bone-spicule pigmentation in the retina. Night blindness was noted in earlier generations.

Discussion

Comprehensive genetic analysis identified a pathogenic frameshift variant in the *RP1* gene (*c.4242_4243delTA*), consistent with an autosomal dominant inheritance. A Variant of Uncertain Significance (VUS) was also observed in the *ZNF408* gene (*c.1889G>A*), though its clinical impact is still unknown. These findings highlight the challenges of genetic diagnosis in RP, the need for thorough testing, and the role of genetic counseling, especially in premarital screening. The RP finding provides a molecular explanation for the familial RP phenotype and supports the suspected inheritance pattern, whereas the *ZNF408* VUS requires further evidence before a disease association can be established. These findings demonstrate the value of comprehensive genetic testing in clarifying the molecular diagnosis and inheritance pattern of RP, particularly in the context of premarital genetic testing screening.

Conclusion

RP is a rare disorder driven by autosomal dominant mutations. Identifying *RP1* variant types using next-generation sequencing (NGS)-based testing for accurate diagnosis, and linking them to existing literature, can play a crucial role in managing and counseling RP patients.

Keywords: Retinitis Pigmentosa; *RP1* variants; Inherited Retinal Dystrophy; Molecular diagnosis; Case Report

1. Introduction

Retinal dystrophies are classified by the location of the retina, damaged cells (rods, cones, or both), Retinal Pigment Epithelium (RPE), inner retina, and whether progression is stagnant or progressive [1]. Retinitis Pigmentosa (RP) is a hereditary retinal dystrophy that causes progressive vision loss in over 1.5 million people worldwide. It is classified as a rare disease and a common Inherited Retinal Dystrophy (IRD), with global prevalence estimates of 1 in 4000 [2]. It is marked by progressive photoreceptor degeneration, primarily affecting rods followed by cones, leading to night blindness, visual field constriction, specific fundus changes, and eventual blindness. RP is primarily a monogenic disease, meaning it is caused by mutations in a single gene [1]. In contrast, genetic heterogeneity occurs when different gene mutations (changes) cause the same disease or condition [3]. However, considerable genetic and phenotypic variation has been observed, with more than 90 genes associated with different forms of the disease and its pathogenesis. Recent advances in diagnostic technologies are likely to uncover more variants with pathogenic significance [1].

RP is classified into two types: syndromic and non-syndromic. Non-syndromic RP begins with night blindness, progresses to vision loss, and ultimately leads to loss of peripheral vision. Syndromic RP manifests when RP is accompanied by systemic symptoms affecting other organs. In contrast to isolated eye disease, syndromic cases often present with hearing loss and neurological or metabolic disorders that require multidisciplinary care [1].

RP can follow different inheritance patterns, including autosomal dominant, which often shows variable expression within families, has a later onset, and milder progression than other variants. The X-linked pattern, which primarily affects males and shows complex inheritance with variable carrier effects in females, is often associated with more severe phenotypes and earlier onset. It is rare, affecting 5-15% of cases, and usually involves mutations in the *Retinitis Pigmentosa GTPase Regulator (RPGR)* and *Retinitis Pigmentosa 2 (RP2)* genes. Current RP management is primarily supportive, including low-vision rehabilitation and management of associated ophthalmic complications. Emerging therapeutic strategies include gene augmentation/replacement therapy, RNA-based therapies, and genome -editing approaches. For example, Voretigene neparvovec (Luxturna) is an established gene therapy for eligible patients with biallelic RPE65-associated retinal dystrophy [4]. For many individuals, the illness begins with night blindness and progresses to peripheral vision loss, eventually leading to legal blindness by the age of 40. The impact of RP extends beyond visual impairment, contributing to increased anxiety, depression, and social isolation among affected individuals, all of which have a substantial effect on their daily lives, employment, and overall quality of life. Furthermore, RP is associated with an increased incidence of other eye complications, such as cataracts and Cystoid Macular Edema (CME), which can lead to further visual problems [5].

This highlights the importance of a multidisciplinary approach to diagnosing and managing RP, emphasizing the role of genetic counseling and NGS to ensure an accurate diagnosis. Currently, a variety of RP management strategies are available, including genetic and psychiatric counseling as well as interventions for RP-related complications, given the significant emotional and psychosocial burden associated with progressive vision loss. While these strategies are primarily supportive, they help alleviate the physical, psychological, and socio-emotional burdens patients may encounter. In this context, research on RP is crucial because of its impact on quality of life and the potential for advances in gene therapies and other therapeutic approaches [4]. RP epidemiology in Africa, including Sudan, remains poorly studied because of limited genetic studies, underdiagnosis, and healthcare system challenges. Consequently, inherited retinal diseases such as RP are often considered rare conditions, with many cases remaining undetected or diagnosed late [6].

This case study details autosomal dominant *RP1* mutations in a family with consanguineous marriages. Utilizing genomic testing highlights the importance of molecular diagnostics in improving disease diagnosis, prognosis, and genetic counseling. In the present case, we performed molecular genetic analysis using complementary sequencing approaches. We analyzed the female individual's genomic DNA using the ClinExomeSure test (DNA Lab India), which covers more than 5,200 genes. In-solution hybridization was used to capture coding exons and flanking intronic regions, followed by paired-end NGS using the Illumina NextSeq system. For the male individual, we amplified the target gene by PCR and sequenced the product bidirectionally by Sanger sequencing. This information can help clinicians offer patients better diagnostic and treatment options, ultimately guiding management of their condition. The rarity of genetically confirmed RP cases reported from Sudan highlights the importance of documenting such cases to expand the understanding of the disease within underrepresented populations.

2. Case Presentation

A couple (IV-7 and IV-6), in Sudan, not yet married, sought genetic evaluation as part of a routine premarital screening. During molecular testing, we identified two probands. Pedigree analysis revealed a consanguineous relationship between probands IV-7 and IV-6 as first cousins. Specifically, the parents of probands II-3 and II-2, designated as Parent III-3 and Parent III-5 in generation III, were siblings, and both were offspring of individuals II-4 and II-3 in generation II. As a result, we deduced that the probands (IV-7 and IV-6) are children of siblings, confirming their first-cousin relationship. **Figure 1.**

2.1. Clinical Findings

In the carrier male and female (IV-7 and IV-6), a gradual decrease in vision and redness of the eyes was reported. Both probands presented with a history of night blindness, and prior to molecular testing, both received an ophthalmological

examination. Clinical examination of both probands showed gradual loss of vision and development of nyctalopia (night blindness). Eye examination of both probands yielded funduscopy findings of bone spicules in the retina and waxy pallor of the optic disc characteristic of RP, as well as gradual visual field constriction compatible with rod-cone retinal dystrophy. While only the female carrier (IV-7) reported seeing flashes or shimmering lights, bone spicule pigmentation in the retina was observed in the male carrier (IV-6) only. There was a family history of night blindness with progressive peripheral vision loss. Specifically, night blindness was exclusive to previous generations. However, they had not been tested for RP, and therefore, once IV-7 and IV-6 were diagnosed, other family members were also tested for *RHO* variants since *RHO* was the gene suspected on previous testing that the family went through in 2011, prior to presenting to the clinic in 2019, where *RHO* testing was re-conducted. Moreover, for two members, normal molecular results were observed, even though they were showing some features associated with RP, such as difficulty with dark adaptation, nyctalopia, and color vision defects.

2.2. Family History

Genetic testing was performed across multiple family members; the two probands (IV-7 and IV-6) were tested along with three other family members in generation IV: one carrier female, one affected female, and one affected male. The affected male and female had started experiencing gradual vision loss between the ages of 17 and 19, respectively, and by the age of 21, both had completely lost vision. This is consistent with progressive RP, where vision deteriorates over time. Individuals from previous generations showed no pathogenic variants associated with RP on initial screening, suggesting either a de novo event or incomplete detection by earlier gene-specific testing.

2.3. Diagnostic Workup

2.3.1 Genetic testing with Next Generation Sequencing

The cases were referred to the Al Neelain Institute for Medical Research, Faculty of Medicine, Al Neelain University, Khartoum, Sudan. The facility received the case in June 2019. However, because high-end molecular technologies required for research were unavailable at the time, the samples were sent to an accredited lab through a laboratory-to-laboratory partnership. Isolated genomic DNA was subjected to in-solution hybridization targeting the coding exons and flanking intronic regions of genes. Initial assessments revealed no pathogenic variants in the *RHO* gene. Illumina NextSeq was used to perform paired-end sequencing. Low-coverage areas, if present, accounted for fewer than 2-5% of the targeted exons or nucleotides.

2.3.2 Bioinformatics Analysis

Sequencing reads were aligned to the human reference genome (GRCh37/hg19). Variant calling was conducted using the Burrows-Wheeler Aligner and Genome Analysis Toolkit (BWA-GATK), followed by annotation through Eurofins Clinical Genetics India (ECGI's) in-house bioinformatics pipeline known as ECGIVarSure, incorporating internal and external databases. The analysis included coding exons and ± 10 bp of flanking intronic regions, with selected deep intronic regions also interrogated.

Variant interpretation was done based on the American College of Medical Genetics and Genomics (ACMG) guidelines. Identified variants were annotated using several publicly available and curated databases including Clinical Variation Database (ClinVar), Online Mendelian Inheritance in Man (OMIM), Human Gene Mutation Database (HGMD), Genome-Wide Association Studies (GWAS), SwissVar, database of Single Nucleotide Polymorphisms (dbSNP), the 1000 Genomes Project, the National Heart, Lung, and Blood Institute (NHLBI) Exome Sequencing Project (ESP), Exome Aggregation Consortium (ExAC), the Genome Aggregation Database (gnomAD), and the Eurofins Clinical Genetics India Database (ECGIdb). In silico prediction tools such as Polymorphism Phenotyping version 2 (PolyPhen-2), Sorting Intolerant From Tolerant (SIFT), and MutationTaster were used to assist variant classification.

2.3.3 Sanger Sequencing

The male proband initially underwent NGS-based screening for autosomal recessive RP. After identifying the candidate variant, Sanger sequencing confirmed the heterozygous c.1889G>A (p.R630Q) variant of uncertain significance (VUS) in the *Zinc Finger Protein 408* (*ZNF408*) gene. For targeted analysis of the *ZNF408* gene, high-fidelity polymerase chain reaction (PCR) was used to amplify the region of interest from genomic DNA. Bidirectional Sanger sequencing was performed, and chromatograms were analyzed using ABI Sequence Analysis software (version 6). FASTA sequences were aligned using Codon Code Aligner (version 7.0.1), with final variant quality assessment performed using ChromasLite (version 2.6.4).

3. Molecular analysis of Probands (IV-7 and IV-6)

3.1. *RHO* Gene Analysis

No pathogenic variants were identified in the *RHO* gene in both probands. Although this finding reduced the likelihood of *RHO*-associated RP, it did not exclude undetected variants outside the analyzed regions.

3.2. RP1 Gene Analysis

NGS analysis of the proband identified a heterozygous frameshift variant in exon 4, *c.4242_4243delTA* (p.H1414Qfs*5) in the *RP1* gene for proband IV-7. This variant, classified as likely pathogenic, is consistent with an autosomal dominant form of RP.

3.3. ZNF408 Gene Analysis

Targeted sequencing detected a heterozygous missense variant *c.1889G>A* (p.R630Q) in exon 5 of the *ZNF408* gene. Comprehensive annotation using ClinVar, OMIM, HGMD, GWAS, SwissVar, dbSNP, the 1000 Genomes Project, ESP, ExAC, gnomAD, and ECGIdb found no earlier published reports indicating that this variant is disease-causing or pathogenic. Following the ACMG criteria, this variant was classified as a VUS, and its clinical significance remains undetermined.

3.4. Gene and Variant Interpretation

RP1 is a microtubule-associated protein specific to photoreceptors, primarily tasked with maintaining the structural integrity and optimal function of the outer segments of the photoreceptors (rods and cones). The identified *RP1* mutation in this family, *c.4242_4243delTA* (p.H1414Qfs*5), is a frameshift mutation in the fourth exon of *RP1*. This mutation creates a premature stop codon, yielding a truncated protein consistent with the retinal phenotype observed in affected individuals. Mutations of this nature in the *RP1* gene are a recognized cause of autosomal dominant RP [7] **Table 1**. *ZNF408* encodes a zinc-finger transcription factor implicated in retinal vascular and developmental anomalies. Whole-exome sequencing identified a VUS in *ZNF408* in the proband (IV-6). This novel VUS has been linked to autosomal recessive RP vitreal alterations in some studies and has been associated with the generation of a missense variant, *c.1889G>A* (p.R630Q) in exon 5, resulting in the substitution of arginine to glutamine at codon 630 [8]. **Table 1**. Although a VUS in the *ZNF408* gene was detected, insufficient data exist to conclude whether it is benign or pathogenic; therefore, its role in the phenotypic changes in this case remains uncertain [9]. Therefore, although documented, the *ZNF408* variant was not deemed causative of the retinal phenotype in this case. For any VUS, standard practice is to periodically reassess as new evidence emerges in the literature.

Table 1: Genetic variants identified in the probands

Gene	Nucleotide Change	Protein Change	Exon No.	Inheritance patterns/ genotype	ACMG Classification
<i>RP1</i> (IV-7)	<i>c.4242_4243delTA</i>	p.H1414Qfs*5	4	Autosomal dominant/ heterozygous	Pathogenic
<i>ZNF408</i> (IV-6)	<i>c.1889G>A</i>	p.R630Q	5	Autosomal Recessive/ heterozygous	Variant of Uncertain Significance

4. Pedigree analysis and family testing

As shown in **Figure 1**, a multigenerational pedigree was constructed based on available family history and genetic testing results. The pedigree included four generations, with the proband IV-7 and IV-6, aged 25 and 29, identified in the most recent generation. Earlier generations (Generations I and II) underwent targeted genetic evaluation and showed no detectable pathogenic variants associated with RP, indicating an unaffected status. Within Generation III, the family members who were presumed to be carriers of retinal disease-associated variant(s) were identified based on the family history provided verbally during genetic counseling sessions. Family members reported having night blindness across Generations I- III, but no molecular testing or clinical testing was performed to confirm whether or not specific individuals in Generation III were carriers. The proband (IV-7) carried a pathogenic variant in the *RP1* gene, while IV-6 carried a VUS in the *ZNF408* gene. These findings suggest variable genetic contribution within the family and support an autosomal dominant inheritance pattern with possible reduced penetrance or variable expressivity. The proband (IV-7) was heterozygous for the likely pathogenic *RP1* frameshift variant *c.4242_4243delTA* (p.H1414Qfs*5), consistent with the observed clinical phenotype of RP. In addition to these probands, genetic testing for the *RP1* pathogenic variant was performed in three additional Generation IV relatives: one affected male (IV-1), one affected female (IV-3), and one carrier female (IV-4). The data on the specific variant gene names were not available, as both probands provided verbal information about the family history during the counseling session. Segregation analysis of this variant within the family is recommended to confirm inheritance and to better define genotype–phenotype correlation. Pedigree analysis emphasizes the importance of family-based genetic testing in inherited retinal disorders, helping distinguish affected individuals, carriers, and unaffected members, and guiding accurate genetic counseling.

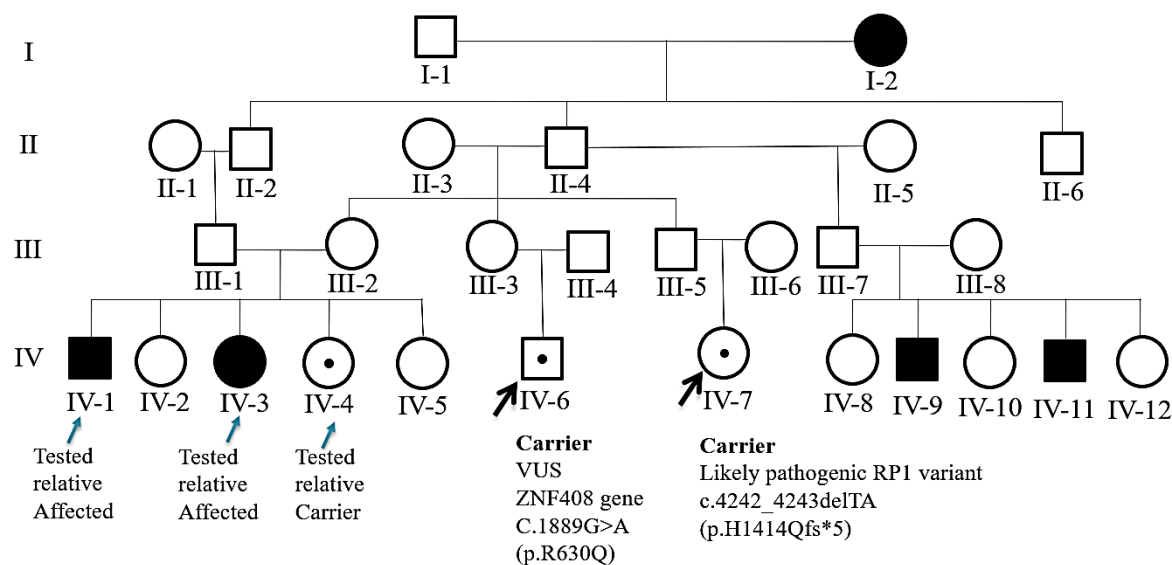


Figure 1: Pedigree of a family with RP associated with an *RP1* likely pathogenic variant. Squares represent males and circles represent females. Filled symbols indicate clinically affected individuals with RP, while open symbols indicate unaffected individuals. Half-filled symbols denote asymptomatic carriers. Blue arrows indicate the probands. Generations are labeled using Roman numerals (I–IV), and individuals within each generation are numbered sequentially. The heterozygous *RP1* frameshift variant c.4242_4243delTA (p.H1414Qfs*5) is indicated in one of the probands and carrier family members. A heterozygous *ZNF408* VUS (c.1889G>A; p.R630Q) is shown in one carrier. Unavailable genetic data is indicated where applicable. The pedigree pattern is consistent with autosomal dominant inheritance with possible variable penetrance. Previous generations (e.g., the proband's great-grandmother, individual I-2) did not undergo genetic testing as it was not available at that time (approximately the 1930s). The family reports that individual I-2 had loss of vision with night vision difficulties. Additionally, the family history for Generation III was provided verbally; therefore, the exact members of Generation III who carried retinal disease-associated variants are unknown.

5. Discussion

The most common Inherited Retinal Dystrophy (IRD) is RP, affecting more than 1.5 million people worldwide. RP is a degenerative disorder characterized by early loss of night vision, followed by decreased visual acuity and progressive narrowing of the visual field. It is a hereditary retinal dystrophy caused by photoreceptor loss, distinguished by retinal pigment deposits visible on fundus examination. The most common type of RP is rod-cone dystrophy, characterized first by nyctalopia, followed by progressive loss of peripheral vision during daylight, and eventually leading to blindness over many decades [10]. This case study describes autosomal dominant *RP1* mutations in a family with consanguineous marriages.

In this case study, as shown in **Table 1**, we identified a heterozygous frameshift variant in exon 4 of the *RP1* gene, c.4242_4243delTA (p.H1414Qfs*5), in the proband (IV-7). The deletion of two nucleotides (TA) at codon positions 4242–4243 shifts the reading frame starting at codon 1414, leading to a histidine-to-glutamine substitution and a premature termination codon five residues downstream, predicted to produce a truncated protein. A heterozygous missense variant in exon 5 of the *ZNF408* gene, c.1889G>A (p.R630Q), was found in the proband (IV-6). This nucleotide substitution replaces arginine with glutamine at amino acid position 630.

This case underscores the complexity of genetic diagnosis in RP. Initial single-gene *RHO* testing was insufficient to identify the molecular cause, whereas an expanded NGS-based panel revealed a likely pathogenic *RP1* variant. Frameshift mutations in *RP1* are well documented in autosomal dominant RP and often correlate with progressive disease. The additional detection of a *ZNF408* VUS highlights the need for cautious interpretation and further functional or familial studies.

The detection of a pathogenic variation in the *RP1* gene in this study is consistent with previous studies identifying *RP1* as a prominent cause of RP, especially in autosomal dominant cases (**Table 2**). A study conducted in 2024 by Mustafa Neissi et al. revealed a novel deletion mutation c.5326delC (p. D1777Ifs*32) in exon 4 of the *RP1* gene. This mutation was discovered in a 19-year-old Iranian lady diagnosed with RP, who had a 15-year history of progressive vision loss and sought medical attention. The clinical examination revealed night visual field impairments, moderate myopia, and a family history of consanguinity. Exome sequencing detected an *RP1* gene mutation, which Sanger sequencing confirmed, showing homozygosity in the proband and heterozygosity in the parents. The mutation caused a frameshift, resulting in a truncated *RP1* protein and suggesting its association with RP-1 progression. Integrating advanced genetic sequencing with comprehensive clinical evaluations has

deepened understanding of the molecular intricacies linked to *RP1* mutations [11].

Another 2025 study by Wei Wu et al. included an index patient, a 56-year-old male with night blindness since age 10. This person reported experiencing blurred vision for five years, with progressive exacerbation during the last year. Interestingly, their results identified a novel truncating mutation that has not been reported before. The novel 4 bp deletion in exon 4, c.2015_2018del (p. Lys672Argfs*9), caused a frameshift in the *RP1* gene, resulting in a truncated protein referred to as p. Lys672Argfs*9 [12].

Although we classified the *ZNF408* variant in our case study as a VUS, previous studies have shown a potential association between *ZNF408* mutations and RP, suggesting a potential role in retinal disease. A 2021 study by Jennifer B. Nadelmann et al. reported a 70-year-old male with pigmentary retinopathy, and genetic assessment identified two variants in the *ZNF408* gene. A next-generation sequencing panel identified biallelic variants (c.1699C > T; p. Arg567Ter; and c.1697 T > A: p. Leu566His) in the *ZNF408* gene, confirming that *ZNF408*-associated retinal dystrophies may exhibit retinal findings and should be considered in the differential diagnosis of RP [13].

Another study conducted by Almudena Avila-Fernandez et al. identified a homozygous mutation (c.358_359delGT; p. Ala122Leufs*2) in the *ZNF408* gene by homozygosity mapping and whole exome sequencing in a Spanish family with autosomal recessive RP. A screening conducted in 217 unrelated families showed another homozygous mutation (c.1621C>T; p. Arg541Cys) in one isolated case of RP. The *ZNF408* gene is a potential novel candidate gene for autosomal recessive RP because it has been previously described as involved in vascular development and is expressed in the human retina [8].

The case reports indicate the critical role of molecular diagnosis in identifying candidate gene variants in RP and related retinal dystrophies.

Table 2: Previously reported *RP1* and *ZNF408* variants associated with RP and related retinal dystrophies.

Study ref	Gene and variant /Amino Acid Change	Patient age/gender	Clinical Presentation	Key findings
Wu et al., 2025 [12]	<i>RP1</i> /c.2015_2018del (p. Lys672Argfs*9)	56/ male	Night blindness since age 10; progressively worsening blurred vision	Novel truncating frameshift variant resulting in a shortened RP1 protein and associated with RP
Neissi et al., 2024 [11]	<i>RP1</i> /c.5326delC (p.Asp1777Ilefs*32)	19/female	Progressive vision loss for 15 years; night blindness; visual field defects; moderate myopia; family history of consanguinity	Novel homozygous frameshift variant causing protein truncation; confirmed by exome and Sanger sequencing
Avila-Fernandez et al.2024 [8]	<i>ZNF408</i> /c.358_359delGT (p.Ala122Leufs*2)	Spanish family with autosomal recessive RP	Retinitis pigmentosa	Homozygous frameshift mutation identified by homozygosity mapping and whole-exome sequencing, suggesting <i>ZNF408</i> as a candidate RP gene
Nadelmann et al., 2021 [13]	<i>ZNF408</i> /c.1699C>T (p.Arg567Ter) and c.1697T>A (p.Leu566His)	70/male	Pigmentary retinopathy	Bi-allelic variants identified by NGS; findings support the association of <i>ZNF408</i> with retinal dystrophy and RP-like phenotypes

6. Limitations

This case report has several limitations. First, it describes the clinical presentations and investigations of a single family; therefore, the findings cannot be generalized to the entire Sudanese population. Secondly, it was performed in a resource-limited setting with limited access to specialized ophthalmic and genetic services. Sudan currently lacks advanced molecular diagnostic facilities and access to genomic testing, so next-generation sequencing analysis required international laboratory-to-laboratory collaboration. Third, family history was only available verbally for some relatives, and complete genetic data were unavailable for several individuals, which could have affected genotype-phenotype interpretation. Despite these limitations, this report

offers important insight into the utility of molecular diagnostics in diagnosing and managing inherited retinal disorders and highlights the need to improve local access to genetic testing facilities and specialized services in low-resource settings.

7. Future Perspectives

Therapeutic approaches designed to rescue photoreceptors, such as gene therapies, require non-functional target cells; however, alternative treatments, such as cell therapies, do not depend on the availability of viable photoreceptors. Gene therapies may be attractive because their fundamental goal is vision restoration; however, cell therapies are expected to have broader applications and do not depend on genetic testing, which can delay therapy [14]. Other recommendations include improved patient-provider communication, connecting patients with integrated care teams, increasing awareness of genetic testing and future treatment options, and increasing access to centers of excellence for genetic ophthalmology and low vision services. The existing RP care pathway is hampered by limited symptom detection, inadequate testing protocols, and insufficient resources. Addressing these barriers through a multidisciplinary approach is essential to ensure patients with RP receive optimal care and support before, during, and after treatment [15].

8. Conclusion

The case study explored the genetic underpinnings of RP in two first-cousin probands and their family. These cases highlight the complexities of genetic diagnosis in RP, where NGS-based genetic testing identified a pathogenic *RP1* variant associated with the observed RP phenotype and clarified the family's likely autosomal dominant inheritance pattern. The additional *ZNF408* VUS highlights the challenges of variant interpretation in genetically heterogeneous disorders. Comprehensive genetic testing and appropriate variant interpretation therefore emphasize the need for accurate diagnosis, genetic counseling, and informed reproductive decision-making in individuals and families affected by RP.

9. Declarations

Ethics Approval and Consent to Participate

Ethical approval was waived because this is an educational case report with no patient identifiers. Written informed consent was obtained from the patient for participation.

Consent for Publication

The authors obtained written informed consent for publication of the case report.

Data Availability Statement

Not applicable

Conflicts of Interest

The authors declare no conflict of interest.

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Author Contributions

Y.A. contributed to conceptualization, methodology, validation, formal analysis, investigation, and data curation; Y.A., O.I., and O.A. prepared the original draft and reviewed and edited the final draft; O.I. and O.A. contributed to illustration and visualization; Y.A. supervised the work. All authors have read and approved the final manuscript.

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Use of Artificial Intelligence

The authors confirm that no content in this manuscript was generated using artificial intelligence (AI) tools, and the authors take full responsibility for the accuracy and integrity of the work.

10. Abbreviations

The following abbreviations are used in this manuscript:

RP	Retinitis Pigmentosa
NGS	Next Generation Sequencing
IRD	Inherited Retinal Dystrophy
RPE	Retinal Pigment Epithelium
RHO	Rhodopsin
RP1	Retinitis Pigmentosa 1
PRPH2	Peripherin-2
USH2A	Usherin
PDE6A	Phosphodiesterase 6A
PDE6B	Phosphodiesterase 6B
RPGR	Retinitis Pigmentosa GTPase Regulator
RP2	Retinitis Pigmentosa 2
ZNF408	Zinc Finger Protein 408
CME	Cystoid Macular Edema
BWA-GATK	Burrows-Wheeler Aligner and Genome Analysis Toolkit
PCR	Polymerase Chain Reaction

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