Cureus
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Open Access Case Report

A Possible Pseudociliopathy in a Patient With Retinitis Pigmentosa and Compound Heterozygous GNB3 Variants: A Case Report

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Review began 09/12/2026
Review ended 10/07/2026
Published 10/29/2026

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DOI: 10.7759/cureus.118795

Abstract

Retinitis pigmentosa (RP) comprises a group of inherited retinal disorders in which photoreceptor degeneration results in progressive peripheral visual field loss and, at later stages, impairment of central vision. Biallelic guanine nucleotide-binding protein beta-3 subunit (*GNB3*) variants have previously been associated with autosomal recessive congenital stationary night blindness (CSNB) and childhood-onset retinopathy, with the human phenotypic spectrum described to date differing from the progressive rod-cone degeneration observed in retinitis pigmentosa. We report a 20-year-old male patient with a retinal phenotype consistent with retinitis pigmentosa (RP), compound heterozygous *GNB3* variants of uncertain significance, and a reported diagnosis of Crohn's disease. Ophthalmic evaluation showed best-corrected visual acuity of 20/30+2 in both eyes, bilateral optic disc pallor, attenuation of retinal vessels, peripheral bone-spicule pigmentation, decreased macular volume, marked retinal nerve fiber layer thinning, and severe bilateral visual field loss. Next-generation sequencing (NGS) identified two heterozygous *GNB3* variants of uncertain significance, c.667del (p.Thr223Leufs*39) and c.680C>T (p.Ser227Leu), located in trans, consistent with compound heterozygosity. Although *GNB3* has been investigated in functional gastrointestinal disorders such as irritable bowel syndrome, available evidence does not establish an association between *GNB3* variants and Crohn's disease. To our knowledge, this is the first reported case of RP in a patient with compound heterozygous *GNB3* variants of uncertain significance. Because β3 subunit of the heterotrimeric G protein (Gβ3) functions in G-protein-mediated retinal signal transduction rather than the ciliary trafficking machinery, we further propose the term "pseudociliopathy" as a conceptual framework: a ciliopathy phenocopy in which disruption of intracellular signaling produces a retinal phenotype resembling those observed in true ciliopathies. This case suggests that the phenotypic spectrum of *GNB3*-associated retinal disease may extend beyond the stationary retinal disorders previously described and highlights the need for additional genotype-phenotype correlation studies and functional validation.

Categories: Genetics, Ophthalmology
Keywords: congenital stationary night blindness, genetic variant, gnb3, inherited retinal disease, pseudociliopathy, retinitis pigmentosa

Introduction

Retinitis pigmentosa (RP) comprises a heterogeneous group of inherited retinal disorders and has an estimated worldwide prevalence of approximately 1 in 4,000 individuals [1,2]. These disorders are characterized by the progressive degeneration of retinal photoreceptors. In typical rod-cone RP, rod photoreceptor dysfunction precedes secondary cone degeneration [2]. Clinically, patients often experience nyctalopia during adolescence, followed by gradual constriction of the peripheral visual field and, ultimately, impairment of central vision [1,2]. Retinal findings include pallor of the optic disc, attenuation of retinal arterioles, and bone-spicule pigmentation of the retina [2]. The genetic basis of non-syndromic RP is highly heterogeneous, with pathogenic variants reported across more than 80 genes and inheritance patterns that include autosomal dominant, autosomal recessive, and X-linked forms, underscoring the remarkable genetic complexity of this condition [2]. Despite advances in next-generation sequencing (NGS), a substantial proportion of RP cases remain genetically unsolved, and novel genotype-phenotype correlations continue to emerge as the mutational landscape expands [1].

The guanine nucleotide-binding protein subunit beta-3 gene (*GNB3*) encodes the β3 subunit of the heterotrimeric G protein (Gβ3), a highly conserved protein expressed in cone photoreceptors and optic nerve (ON)-bipolar cells across vertebrate species, where it plays essential roles in cone phototransduction and ON-bipolar cell signaling [3,4]. Biallelic *GNB3* variants were first linked to human retinal disease by Vincent and colleagues, who identified homozygous and compound heterozygous variants in affected individuals from two unrelated families with autosomal recessive congenital stationary night blindness (CSNB) characterized by abnormalities of ON-bipolar cell function and cone sensitivity [5]. Independently, Arno and co-workers reported a patient with childhood-onset retinal dysfunction associated with a homozygous *GNB3* nonsense variant [6]. The patient presented with childhood-onset nystagmus and mild

central macular abnormalities on retinal imaging [6]. Together, these independent reports provide complementary evidence supporting an association between biallelic *GNB3* variants and human retinal dysfunction, while also demonstrating variability in the associated retinal phenotype [5,6].

Animal models, however, suggest a broader and potentially more severe phenotypic spectrum associated with *GNB3* dysfunction. The retinopathy globe enlarged (rge) chicken, which harbors a homozygous D153del mutation in *GNB3*, develops progressive retinal degeneration, globe enlargement, and extraocular manifestations, including renal abnormalities with glomerulomegaly and tubulointerstitial inflammation [7,8]. Similarly, Gnb3 knockout mice show cone photoreceptor and ON-bipolar cell dysfunction, with reduced cone transducin levels and diminished light sensitivity [9]. These findings raise the possibility that *GNB3*-associated disease may not be limited to stationary retinal dysfunction and may have broader phenotypic potential, although this has not been clearly established in humans.

This report describes a 20-year-old male patient with a retinal phenotype consistent with retinitis pigmentosa, compound heterozygous *GNB3* variants of uncertain significance (VUS), and a reported diagnosis of Crohn's disease, without the nyctalopia characteristic of CSNB. To our knowledge, this is the first reported case of RP in the setting of compound heterozygous *GNB3* variants of uncertain significance, suggesting a possible expansion of the *GNB3*-associated phenotypic spectrum while underscoring the need for further evidence to clarify the pathogenic significance of these findings.

Case Presentation

A 20-year-old male patient with a reported diagnosis of Crohn's disease presented with blurred vision and visual loss in the left eye (OS). The patient denied nyctalopia, photophobia, difficulty with dark adaptation, color vision abnormalities, or photopsias. The remainder of the review of systems was unremarkable. He had no history of prior ocular surgery or other ocular diseases. Additional historical details, including age at onset of visual symptoms, symptom duration, family history of inherited retinal disease, and parental consanguinity, were not documented in the available medical records.

Upon initial comprehensive ophthalmic evaluation by one of the authors (NJI), best-corrected visual acuity (BCVA) was 20/30+2 in both eyes (OU). Cycloplegic refraction was +0.75 +0.50 x 90° and +1.25 +0.50 x 90° in the right and left eyes, respectively. Intraocular pressure (IOP), measured by Goldmann applanation tonometry, was 10 mmHg and 11 mmHg in the right and left eyes, respectively. No remarkable findings were identified upon external and slit-lamp examination. Upon indirect ophthalmoscopy, the patient had bilateral optic disc pallor, retinal vessel attenuation, and peripheral bone-spicule pigmentation.

Macular optical coherence tomography (OCT) showed decreased macular volume and thickness bilaterally (Figure *1*). Central subfield thickness measured 237 µm and 230 µm in the right and left eyes, respectively. Cube volume was 9.1 mm^3 in the right eye and 8.9 mm^3 in the left eye. Average cube thickness measured 254 µm and 247 µm in the right and left eyes, respectively.

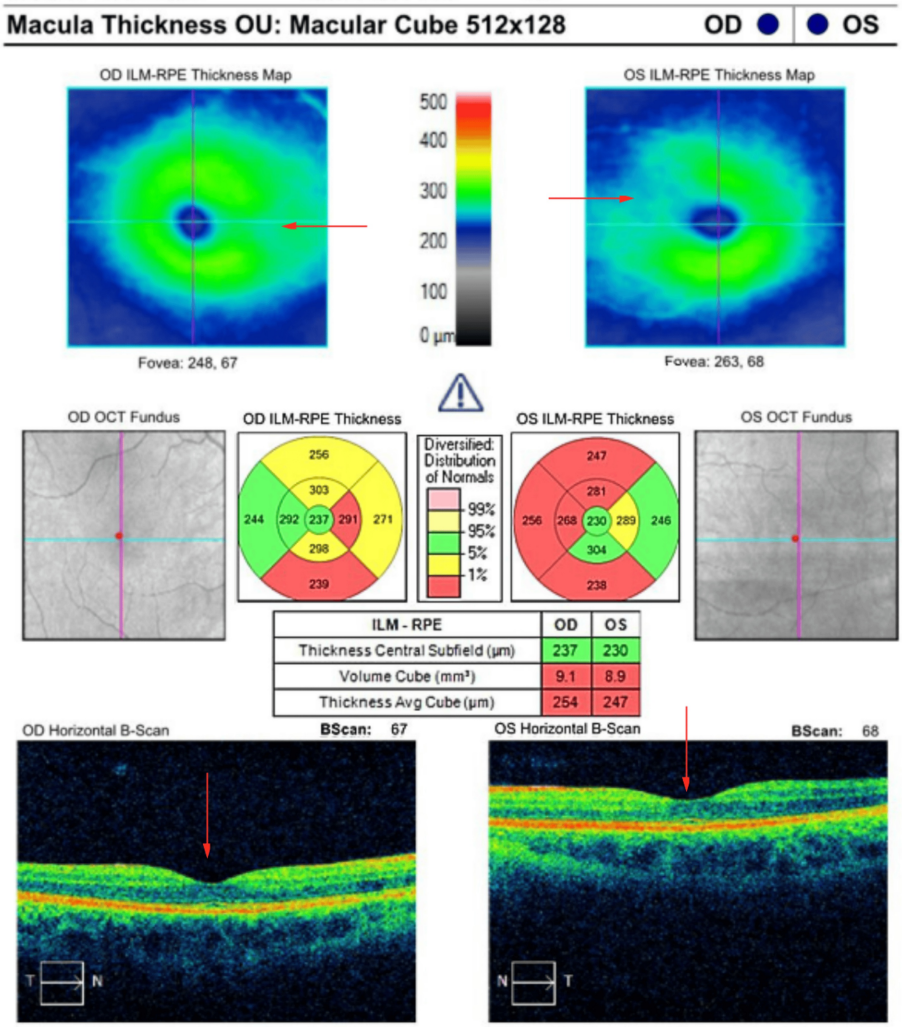


FIGURE 1: Macular optical coherence tomography showing bilateral macular thinning

OCT: optical coherence tomography; OD: right eye (oculus dexter); OS: left eye (oculus sinister); OU: both eyes (oculus uterque); ILM: internal limiting membrane; RPE: retinal pigment epithelium; ILM-RPE: retinal thickness measured from the internal limiting membrane to the retinal pigment epithelium. The arrows highlight areas of macular thinning observed on optical coherence tomography (OCT).

Optic nerve OCT showed marked bilateral retinal nerve fiber layer (RNFL) thinning, with average RNFL thicknesses of 46 µm and 47 µm in the right and left eyes, respectively (Figure *2*). Minimal optic nerve cupping was observed, with average cup-to-disc ratios of 0.06 bilaterally.

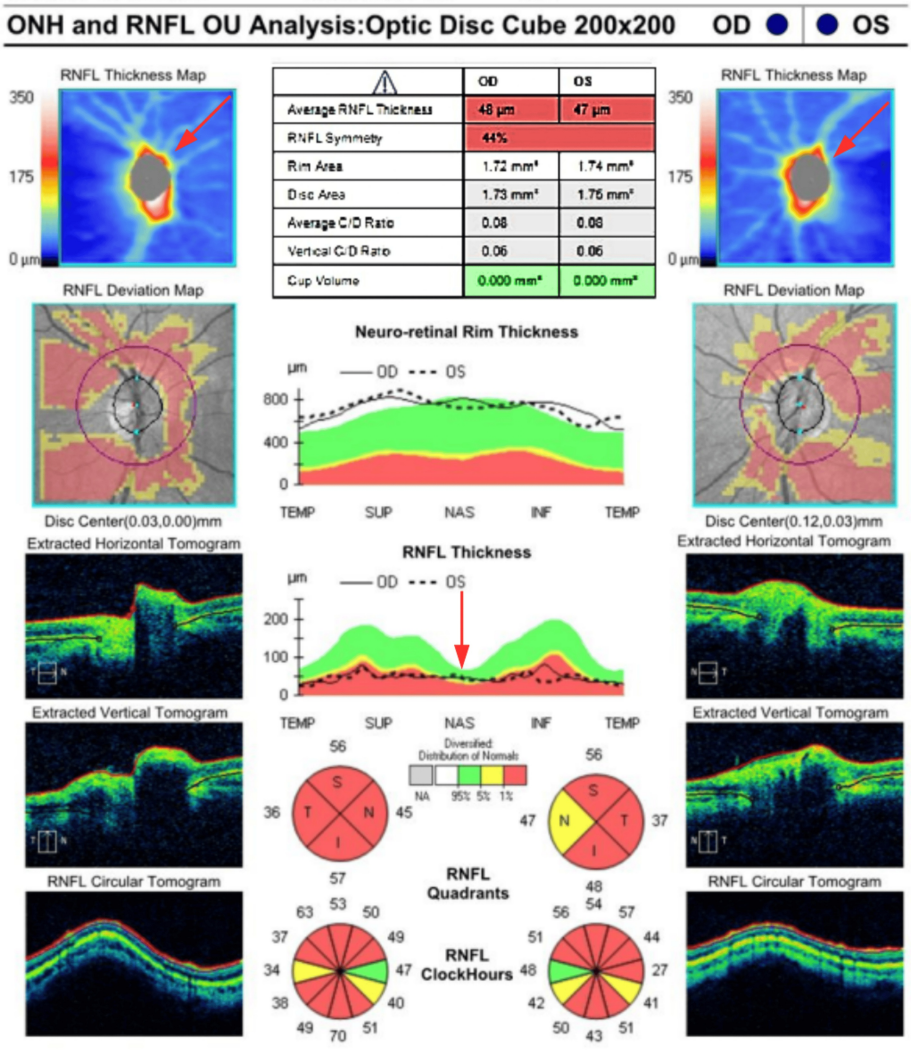


FIGURE 2: Optic nerve head and retinal nerve fiber layer optical coherence tomography showing bilateral RNFL thinning

ONH: optic nerve head; RNFL: retinal nerve fiber layer; OD: right eye (oculus dexter); OS: left eye (oculus sinister); OU: both eyes (oculus uterque); R/D: rim-to-disc ratio; SUP: superior; NAS: nasal; INF: inferior; S: superior; T: temporal; I: inferior; N: nasal. The arrows indicate areas of retinal nerve fiber layer (RNFL) thinning.

Humphrey visual field (HVF) testing was done using the 30-2 threshold strategy. In the right eye, the glaucoma hemifield test was outside normal limits, with a visual field index (VFI) of 52%, mean deviation (MD) of -22.10 dB, and pattern standard deviation (PSD) of 14.37 dB (Figure *3*). The left-eye test was flagged as having low reliability; nevertheless, the glaucoma hemifield test was outside normal limits, with a VFI of 35%, MD of -26.19 dB, and PSD of 12.74 dB (Figure *4*).

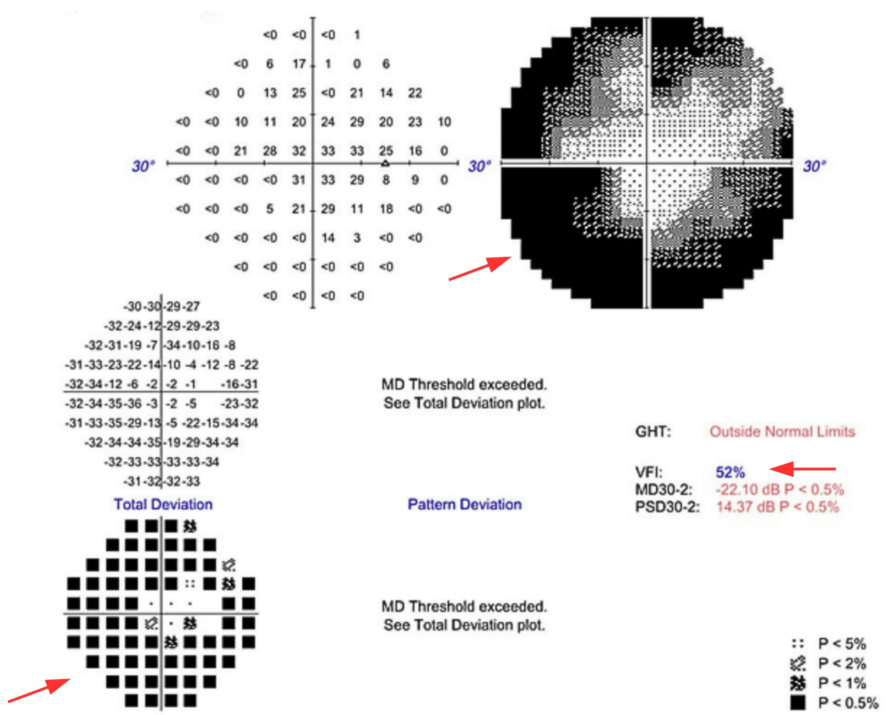


FIGURE 3: Humphrey visual field 30-2 testing showing severe visual field loss in the right eye

GHT: glaucoma hemifield test; VFI: visual field index; MD: mean deviation; PSD: pattern standard deviation.

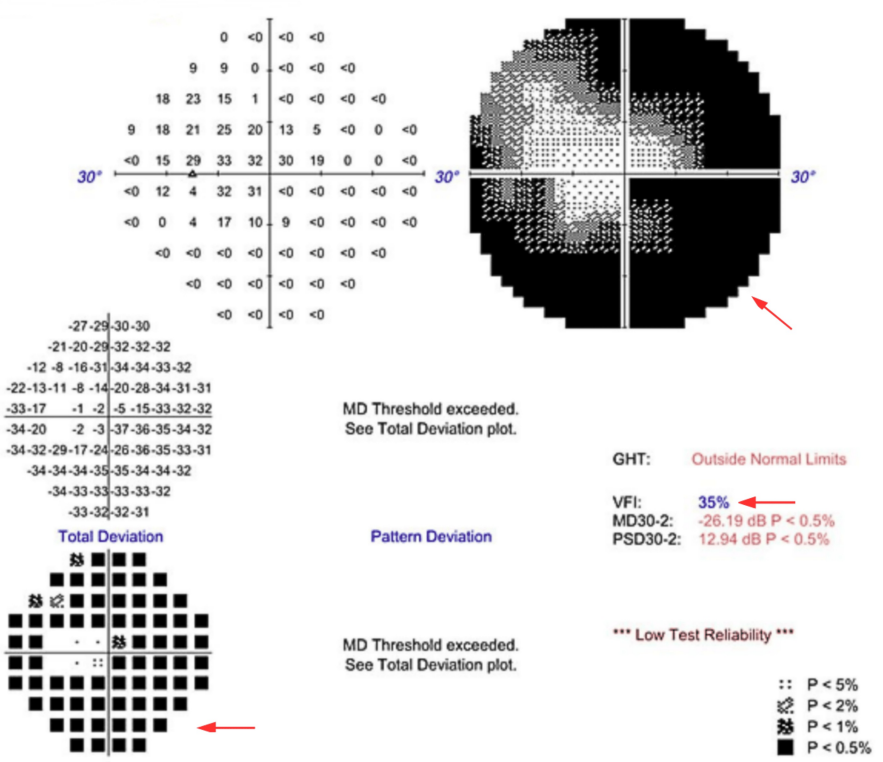


FIGURE 4: Humphrey visual field 30-2 testing showing severe visual field loss in the left eye

GHT: glaucoma hemifield test; VFI: visual field index; MD: mean deviation; PSD: pattern standard deviation.

Genetic testing using the Invitae (Invitae Corporation, San Francisco, California, United States) Inherited Retinal Disorders Panel, including sequence and deletion/duplication analysis, identified two heterozygous *GNB3* VUS: c.667del (p.Thr223Leufs*39) and c.680C>T (p.Ser227Leu). The c.667del variant introduces a frameshift predicted to cause premature termination and consequent loss or disruption of the encoded protein. The c.680C>T variant is a missense alteration that replaces serine with leucine at amino acid position 227. The variants were reported to be in trans (on opposite chromosomes). Written informed consent was obtained from the patient for publication of this case report and accompanying clinical images.

Discussion

Biallelic *GNB3* variants have previously been associated with autosomal recessive CSNB and childhood-onset retinopathy [5,6]. Vincent and coworkers described both homozygous and compound heterozygous biallelic *GNB3* variants associated with an autosomal recessive form of CSNB characterized by ON-bipolar cell dysfunction, reduced cone sensitivity, and nyctalopia [5]. Our patient had two different *GNB3* variants in trans, consistent with a compound heterozygous biallelic configuration. In contrast to the CSNB phenotype described by Vincent and coworkers, our patient denied nyctalopia and showed findings consistent with rod-cone degeneration, including visual field constriction, marked retinal nerve fiber layer (RNFL) thinning, and decreased macular volume (Figures *1*-*4*). These findings differ from the stationary phenotype previously reported and raise the hypothesis that the phenotypic spectrum associated with *GNB3* may extend beyond stationary retinal dysfunction [5].

Independently, Arno and coworkers reported a patient with a homozygous *GNB3* nonsense variant presenting with childhood-onset retinopathy, nystagmus, and mild macular abnormalities [6]. In contrast, our patient showed bilateral retinal degeneration consistent with RP and macular abnormalities (Figure *1*), but did not exhibit nystagmus. Although both cases involved biallelic *GNB3* variants, the phenotype observed in our patient differs substantially from the previously reported childhood-onset retinopathy.

The two *GNB3* variants identified in our patient, c.667del (p.Thr223Leufs*39) and c.680C>T (p.Ser227Leu), were classified as VUS by Invitae. The c.667del variant causes a frameshift predicted to introduce an early termination codon, potentially resulting in loss of normal protein function, whereas c.680C>T is a missense variant that replaces serine with leucine at codon 227 [10]. Under the American College of Medical Genetics and Genomics (ACMG) variant-interpretation standards, classification of a variant as a VUS indicates that

the available evidence is currently insufficient to establish either pathogenicity or benignity; therefore, these variants cannot be regarded as definitive molecular evidence of disease [11,12]. Nevertheless, the presence of two variants in trans (opposite chromosomes) within a gene known to be associated with retinal disease, together with the patient's RP phenotype, raises the possibility of a genotype-phenotype association. Further evidence, including segregation studies, functional assays, and identification of unrelated affected individuals carrying the same variants, will be necessary to determine whether these variants should ultimately be reclassified.

Animal models suggest that *GNB3* deficiency may produce a more severe and progressive retinal phenotype than described in humans. The retinopathy globe enlarged chicken, which carries a homozygous *GNB3* mutation, develops progressive retinal degeneration, globe enlargement, and renal abnormalities [7,8]. Similarly, Gnb3 knockout mice exhibit impaired cone photoreceptor and ON-bipolar cell function with markedly reduced retinal light responses [9]. Our patient showed a degenerative retinal phenotype consistent with RP rather than the stationary phenotype previously reported in humans. Although animal models cannot establish causality in humans, they provide biological support for the possibility that *GNB3* dysfunction may contribute to a degenerative retinal phenotype.

Previous studies have investigated associations between *GNB3* polymorphisms and functional gastrointestinal disorders [13-15]. These studies have primarily evaluated the common C825T polymorphism because of its potential effects on gastrointestinal sensory and motor function [13,14]. However, reported associations between the *GNB3* C825T polymorphism and functional gastrointestinal disorders have been inconsistent across populations, and meta-analytic evidence has not demonstrated a significant overall association with irritable bowel syndrome (IBS) [14,15]. In contrast, Crohn's disease has a distinct genetic architecture involving immune- and autophagy-related susceptibility loci [16]. In addition to the retinal phenotype, our patient had a reported diagnosis of Crohn's disease. Given the potential overlap of gastrointestinal symptoms between functional and inflammatory bowel disorders, and in the absence of detailed gastroenterologic data, it is not possible to determine whether concomitant functional gastrointestinal symptoms contributed to the clinical presentation. Based on the available evidence, no causal relationship between the identified *GNB3* variants and Crohn's disease can be established.

Ciliopathies comprise a heterogeneous group of inherited disorders caused by defects in the assembly, maintenance, or function of primary cilia or their basal bodies, resulting in impaired intracellular signaling and multisystem disease involving organs such as the retina, kidneys, liver, brain, and skeleton [17,18]. In the retina, ciliopathies characteristically disrupt intraflagellar transport and protein trafficking across the photoreceptor connecting cilium, the obligate structural conduit through which all outer segment proteins are trafficked [18]. Failure of this trafficking pathway can lead to progressive photoreceptor degeneration and inherited retinal dystrophies, including RP, as exemplified by Bardet-Biedl syndrome and related syndromic ciliopathies [18].

Disease associated with *GNB3* is mechanistically distinct from this paradigm. Gβ3 functions as a component of heterotrimeric G-protein signaling downstream of G-protein-coupled receptors (GPCRs), in which Gβ and Gγ form a tightly associated complex that interacts with the Gα subunit and modulates downstream signaling [19,20]. In the retina, Gβ3 is expressed in cone photoreceptors and is strongly localized to cone outer segments, supporting its proposed role as the β subunit of cone transducin [19,20]. *GNB3* dysfunction is therefore mechanistically distinct from disruption of the ciliary trafficking machinery and instead affects G-protein-mediated retinal signal transduction [9,19,20].

A plausible mechanism supports the possibility that *GNB3* dysfunction could contribute to retinal degeneration rather than solely stationary retinal dysfunction. In Gnb3 knockout mice, cone transducin subunit levels are markedly reduced, cone sensitivity is diminished, and Gβ3 is additionally required for synaptic maintenance in ON-bipolar pathways [9]. In the rge chicken, the D153del mutation destabilizes the Gβ3 protein, leading to dysregulation of cyclic nucleotide (cyclic guanosine monophosphate (cGMP)/cyclic adenosine monophosphate (cAMP)) levels and downstream phosphorylation cascades in the retina. Consequently, these birds develop progressive retinal degeneration [7,8]. Disruption of phototransduction and cyclic nucleotide homeostasis can contribute to photoreceptor death in monogenic retinal dystrophies caused by other phototransduction genes [4]. Together, these data provide a biologically plausible mechanism by which impaired Gβ3 function could contribute to the rod-cone degeneration observed in our patient.

Despite these fundamentally different mechanisms, the resulting phenotypes may converge. Our patient exhibited rod-cone degeneration resembling ciliary retinal dystrophies, while the *GNB3* rge chicken model shows retinal degeneration accompanied by renal abnormalities, a retina-kidney pattern also observed in ciliopathies [7,8,17]. Based on this phenotypic convergence, we hypothesize that *GNB3*-associated retinal degeneration could be considered within a conceptual framework of "pseudociliopathy": a ciliopathy phenocopy in which disruption of intracellular signal transduction produces a ciliopathy-like retinal phenotype without an established primary ciliary defect. Further studies, including functional characterization of *GNB3* variants, evaluation of downstream signaling pathways, and longitudinal phenotyping of additional patients with biallelic GNB3 variants, will be necessary to determine whether retinal degeneration is a consistent feature of *GNB3*-associated disease and whether this conceptual

framework is supported by additional clinical and molecular evidence.

This report has several limitations. As a single case, this report describes the co-occurrence of this phenotype and biallelic *GNB3* variants but cannot establish their association, frequency, or broader genotype-phenotype relationship. Additionally, the left-eye Humphrey visual field was flagged as having low test reliability, limiting interpretation of its quantitative parameters. Moreover, both *GNB3* variants were classified as VUS and were not functionally validated; therefore, a definitive genotype-phenotype correlation cannot be established. Electroretinography, which could have further characterized ON-bipolar cell and cone system function, was not available. Finally, although the retinal phenotype in our patient resembles retinal degeneration described in animal models of *GNB3* deficiency, interspecies differences limit the strength of this comparison.

Conclusions

Our patient showed retinitis pigmentosa in the setting of compound heterozygous *GNB3* variants of uncertain significance, a phenotype that differs from the stationary retinal disorders previously reported in humans. Because Gβ3 acts within G-protein-mediated retinal signal transduction rather than the ciliary trafficking machinery, we propose that this presentation may be conceptualized as a pseudociliopathy, a ciliopathy phenocopy potentially arising from disrupted intracellular signaling. However, the pathogenicity of the identified variants has not been established, and the findings are limited by the single-patient nature of this report and the absence of functional validation. Therefore, this case raises the hypothesis that the phenotypic spectrum of *GNB3*-associated retinal disease may extend beyond the stationary retinal disorders described to date. Confirmation will require additional genotype-phenotype correlation and functional studies, including functional characterization of the identified variants.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

Concept and design: Adriana I. Ramirez Gandia, Maria Garcia, Natalio Izquierdo

Acquisition, analysis, or interpretation of data: Adriana I. Ramirez Gandia, Maria Garcia

Drafting of the manuscript: Adriana I. Ramirez Gandia, Maria Garcia, Natalio Izquierdo

Critical review of the manuscript for important intellectual content: Adriana I. Ramirez Gandia, Maria Garcia, Natalio Izquierdo

Supervision: Natalio Izquierdo

Disclosures

Human subjects: Informed consent for treatment and open access publication was obtained or waived by all participants in this study. **Conflicts of interest:** In compliance with the ICMJE uniform disclosure form, all authors declare the following: **Payment/services info:** All authors have declared that no financial support was received from any organization for the submitted work. **Financial relationships:** All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might have an interest in the submitted work. **Other relationships:** All authors have declared that there are no other relationships or activities that could appear to have influenced the submitted work.

Acknowledgements

Adriana I. Ramírez Gandía and Maria Garcia contributed equally to the work and should be considered co-first authors.

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