**American Journal of Bioscience and Clinical Integrity**
https://biojournals.us/index.php/AJBCI
**ISSN: 2997-7347**

# Current Perspectives on the Etiology and Epidemiology of Retinitis Pigmentosa

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**Received:** 2025, 15, Aug
**Accepted:** 2025, 21, Sep
**Published:** 2025, 29, Oct



**Annotation:** Retinitis pigmentosa (RP) is a group of inherited retinal dystrophies characterized by progressive photoreceptor degeneration, leading to night blindness, peripheral vision loss, and eventual central vision impairment. RP affects approximately 1 in 4,000 individuals worldwide with significant genetic heterogeneity involving more than 60 identified genes. Current management strategies are largely supportive, including low-vision aids and retinal implants, but emerging gene-based therapies and stem-cell approaches have shown promising results in early clinical trials. This article reviews the epidemiology, pathophysiology, and current therapeutic approaches to RP, emphasizing recent advances in gene therapy and potential future directions.

**Keywords:** Retinitis Pigmentosa; inherited retinal dystrophies; photoreceptor degeneration; gene therapy; stem cell therapy; retinal implants; epidemiology; pathophysiology; visual impairment; molecular genetics.

**Introduction:** Retinitis pigmentosa (RP) is one of the most common forms of inherited retinal dystrophy, characterized by progressive degeneration of rod and cone photoreceptors [1]. The global prevalence of RP is estimated to be approximately 1 in 4,000 individuals, affecting more than 1.5 million people worldwide [2]. Clinically, patients typically present with night blindness during adolescence, followed by progressive constriction of the visual field and eventual central

vision loss in adulthood [1,3].

Genetically, RP is highly heterogeneous, with over 60 causative genes identified to date [3]. These genes are involved in diverse cellular processes, including phototransduction, retinal metabolism, and structural maintenance of photoreceptors. RP can be inherited in autosomal dominant, autosomal recessive, or X-linked patterns, with significant variability in disease onset and severity [4].

Despite decades of research, RP remains incurable. Current management primarily focuses on supportive measures such as low-vision rehabilitation, use of optical aids, and, in selected cases, implantation of retinal prostheses [5]. However, rapid advances in molecular genetics and biotechnology have opened promising therapeutic avenues, particularly gene therapy, stem-cell transplantation, and optogenetic strategies [6]. These developments highlight the urgent need for continued research into novel treatment approaches that may significantly improve patient outcomes.

**Aim of study** is to analyze the current understanding of the etiology and epidemiology of Retinitis Pigmentosa (RP). This paper seeks to summarize the genetic mechanisms underlying RP, examine the global distribution and prevalence of the disease, and highlight recent research findings that contribute to early diagnosis and potential therapeutic strategies.

**Etiology**: Genetic mutations responsible for RP produce biochemical dysfunction that specifically affects rod photoreceptors in the retina.[7] Defects may be associated with multiple injury pathways, including apoptosis, light damage, ciliary transport dysfunction, and endoplasmic reticulum stress. The typical result of all the possible pathways is the death of the rod photoreceptors.[8] Since the rods are responsible for low-light vision, the ever-increasing loss of these cells produces the characteristic night blindness and a gradual diminution of peripheral vision. Eventually, the destruction of large numbers of rods has a deleterious effect on the retinal pigment epithelium (RPE) and affects cone photoreceptors. As cones succumb to the toxic environment created by progressive cell death in the retina, dyschromatopsia, or disturbance of color perception, may develop (rod-cone dystrophy).

RP is genetically heterogeneous, meaning multiple genetic mutations are associated with the condition. The affected genes participate in critical retinal functions, such as phototransduction, the visual cycle, ciliary transport, and photoreceptor structure.[9] To date, over 3100 different mutations have been identified as causing RP [7] and there is phenotypic variability with the same mutation even within the same family.[10] This genetic diversity contributes to the variation in the severity and progression of the disease. Research findings have discovered more than 80 different genes that cause multiple patterns of inheritance and expression for nonsyndromic RP.[11][13] Approximately 5% to 20% of RP cases are autosomal recessive (arRP) [11] and around 15% to 25% of RP is autosomal dominant (adRP), whereas around 5% to 15% of cases are X-linked recessive (xlRP).[11] The remaining cases (around 40%-50%) are termed sporadic or simplex (with a single reported case in the family); no family history or known molecular basis is found.[11] Digenic RP is very rare.[12]

The most commonly mutated genes in arRP include USH2A, ABCA4, CERKL, CRB1, EYS, PDE6A, PDE6B, RPE65, RP1, and SAG[11]. The common genes involved in adRP include *RHO*, *RP1*, *CRX*, *GUCA1B*, *IMPDH1*, *KLHL7*, *NR2E3*, *PRPF8*, *PRPF3*, *PRPF31*, *PRPH2*, *SEMA4A*, *SNRNP200*, and *TOPORS*; mutations in genes *RPGR*, *RP2*, and *OFD1* cause xlRP.[11]

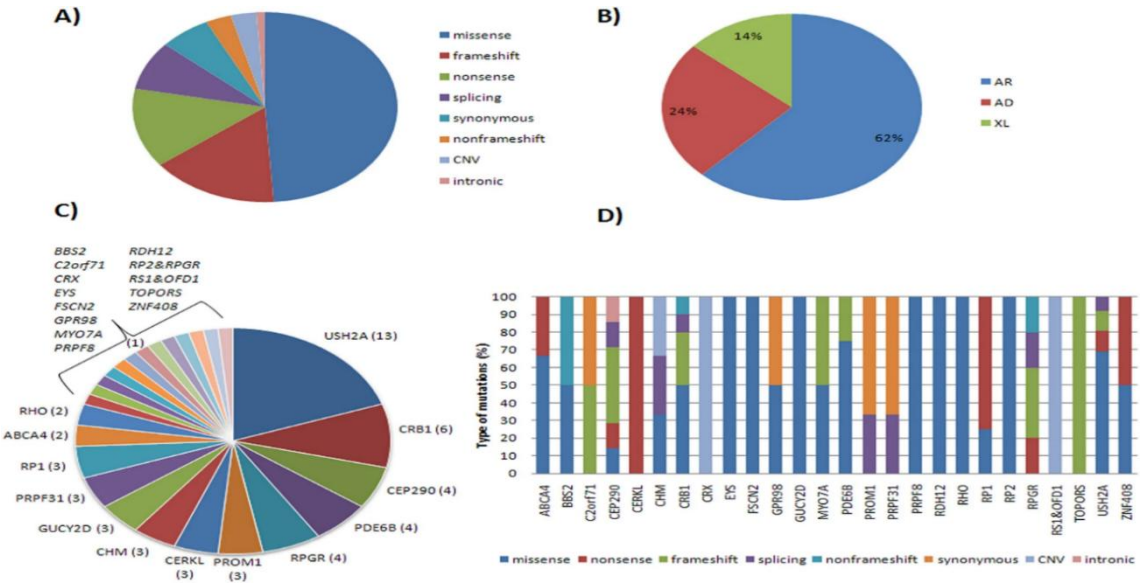


**Fig. 1: Prevalence analyses of solved cases (A) Proportion of each type of mutation. (**B**) Principal detected modes of inheritance. (**C**) Recurrence of all mutated retinal genes. (**D**) Distribution of different types of mutations for each of the identified mutated gene.**

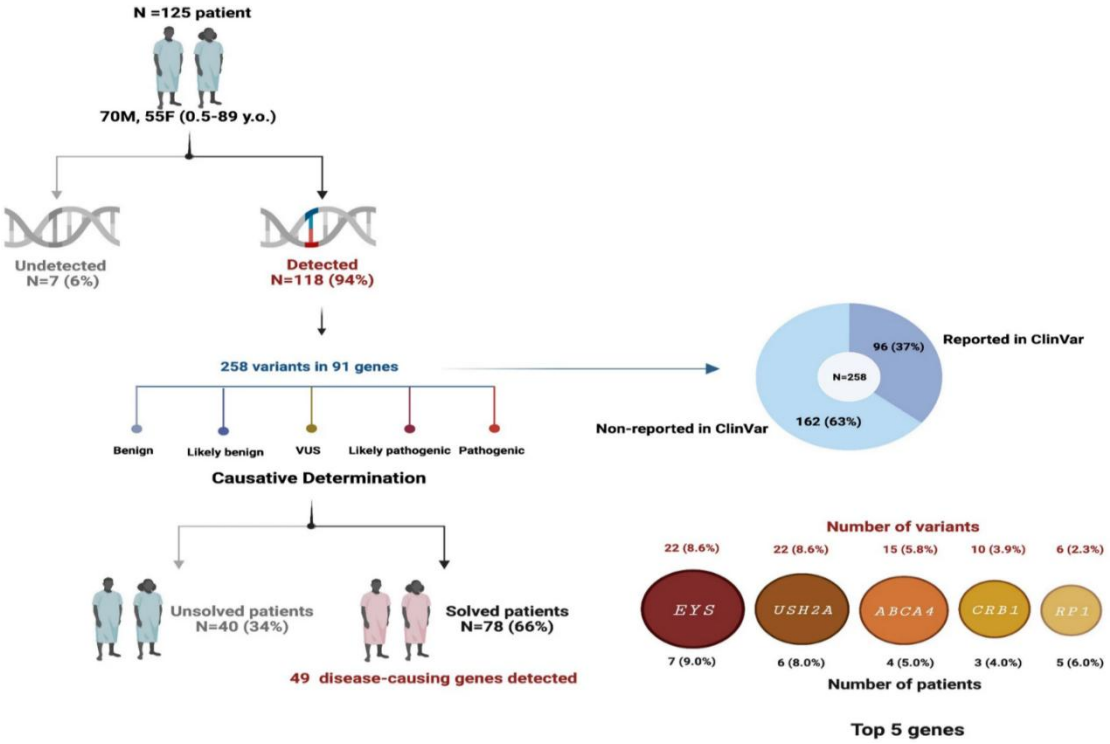


**Fig. 2. WES data of 125 Thai patients with IRD in 2014**

Variants were detected in 94% of the patients. Based on variant classification, 49 genes were determined to cause diseases in 66% of the patients, with the *EYS*, *USH2A*, *ABCA4*, *CRB1*, and *RP1* showing the highest prevalence.

**Epidemiology**: Nonsyndromic RP has a worldwide prevalence of about 1 in 5000 individuals.[13] RP constitutes around 50% of the cases of inherited retinal diseases, and [9] affects more than 1.5 million people worldwide [14] with varying prevalence. Estimates in prevalence range from 1 in 3026 in Denmark [15] to 1 in 4869 in Birmingham, UK,[16] and as high as 1 in 372 in rural India.[17] Some of this variation may be due to differences in methodology and case definitions across studies; furthermore, the prevalence may be higher in some populations with more consanguineous marriages, as seen in certain Middle Eastern and

South Asian countries.[17][18]

Men are affected slightly more often than women due to the X-linked form being expressed more frequently in males. Syndromic RP is much less common, with estimates for Usher syndrome ranging from 4 to 17 cases per 100,000 individuals.[19]

The average age of symptom onset is dependent on the genetic type involved. The autosomal recessive form will develop symptoms in the early adolescent years, but those affected with autosomal dominant RP will likely not have symptoms until well into their 20s. More than three-quarters of individuals with RP will be symptomatic and present for clinical evaluation and diagnosis of the disease by the time they are 30 years.[14] In a study conducted in Japan, the average age of diagnosis was 35.1 years (median age 36.5 years).[20]

**Conclusion:** Retinitis Pigmentosa remains one of the most genetically diverse and clinically variable inherited retinal disorders. Despite significant advances in identifying more than 80 causative genes, RP continues to pose major diagnostic and therapeutic challenges. Epidemiological data show variability in prevalence depending on geographic and genetic factors, with higher rates in regions of increased consanguinity. While no definitive cure currently exists, the rapid development of gene therapy, stem-cell transplantation, and retinal prosthetic devices offers promising directions for future treatment. Continued genetic research and international collaboration are essential for translating molecular discoveries into effective clinical interventions that can preserve or restore vision in affected individuals.

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