



3. GENETICALLY DETERMINED PIGMENTARY RETINOPATHY (PR) IN SYSTEMIC DISEASES

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Introduction. The term "pigmentary retinopathy" broadly refers to a widespread alteration of the retina and pigment epithelium preventing normal vision. The condition is clinically diverse and can be inherited in various ways. It can be associated with different genetic syndromes and can be indicative of systemic disease

Aim of study. To analyze the relationship between pigmentary retinopathy and systemic diseases, shedding light on the underlying genetic factors contributing to this condition.

Methods and materials. Databases - PubMed, Scopus, and Web of Science - selected articles according to the keywords. Out of 280 articles only 42 of these studies met our strict inclusion criteria and were included in our analysis

Results. Pigmentary retinopathy can have different symptoms and severity levels among individuals. Genetic polymorphism has been extensively studied and has revealed genes and variations that increase the risk of developing the condition. Some hereditary pigmentary retinopathies are part of syndromes that involve multiple organ systems. They can be inherited in different modes of genetic transmission, including through autosomal recessive, autosomal dominant, X-linked recessive, digenic, or mitochondrial transmission.

Conclusion. Understanding the genetic basis of pigmentary retinopathy and its relation to systemic diseases, is crucial for accurate diagnosis and predicting the visual prognosis.