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Genetic screening for region genetic counseling protocols

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Purpose: This systematic review explores the global landscape of inherited retinal dystrophies (IRDs), emphasizing prevalence, genetic variations, clinical manifestations, and diagnostic approaches across different countries to understand the heterogeneity in presentation and progression of IRDs.

Methods: Adhering to PRISMA guidelines, we conducted a comprehensive literature search across databases like PubMed, Scopus, Web of Science, and Google Scholar. Our strategy included terms related to IRDs, with filters for human studies and English language publications.

Results:
Our findings reveal significant genetic heterogeneity of inherited retinal dystrophies (IRDs) on a global scale, with notable differences in genetic prevalence across regions. Notably, the PRPH2 gene was identified among the top ten most prevalent genes in studies from Western countries but did not rank within the top ten in any of the studies from Africa and Asia. This highlights the variability of certain genes and underscores the importance of worldwide testing to define and prioritize research on medications that could benefit the largest possible population. Other genetic variations, such as USH2A, RPGR, and ABCA4 mutations, showed varying prevalence across different regions, further emphasizing the complex genetic landscape of IRDs.

Discussion: The study underscores the importance of considering geographical and ethnic backgrounds in the genetic diagnosis and management of IRDs, suggesting a tailored approach to genetic screening and a need for region-specific genetic counseling protocols.