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Mouse Models of Inherited Retinal Degeneration with Photoreceptor Cell Loss.

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Inherited retinal degeneration (RD) leads to the impairment or loss of vision in millions of individuals worldwide, most frequently due to the loss of photoreceptor (PR) cells. Animal models, particularly the laboratory mouse, have been used to understand the pathogenic mechanisms that underlie PR cell loss and to explore therapies that may prevent, delay, or reverse RD. Here, we reviewed entries in the Mouse Genome Informatics and PubMed databases to compile a comprehensive list of monogenic mouse models in which PR cell loss is demonstrated. The progression of PR cell loss with postnatal age was documented in mutant alleles of genes grouped by biological function. As anticipated, a wide range in the onset and rate of cell loss was observed among the reported models. The analysis underscored relationships between RD genes and ciliary function, transcription-coupled DNA damage repair, and cellular chloride homeostasis. Comparing the mouse gene list to human RD genes identified in the RetNet database revealed that mouse models are available for 40% of the known human diseases, suggesting opportunities for future research. This work may provide insight into the molecular players and pathways through which PR degenerative disease occurs and may be useful for planning translational studies.

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