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Ophthalmic Genetic Diseases-Medicine in the 21st Century

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The past few years have witnessed extraordinary advances in molecular genetic technologies and the accumulation of an immense amount of structural genomic information about human and the model organisms including whole genome sequences as well as their variations. The dramatic availability and increase in the amount of such rich genomic information and technologies, when combined with the biologic resources of well-characterized and phenotyped population cohorts, provides unique and unprecedented opportunities to gain insights into the genetic mechanisms and potential risk factors underlying many human genetic diseases. So far, nearly 500 genes that contribute to inherited eye diseases have been identified. Disease-causing mutations are associated with many ocular diseases, including glaucoma, cataracts, strabismus, corneal dystrophies, and a number of forms of retinal degenerations. This remarkable new genetic information highlights the significant in roads that are being made in understanding the molecular basis of human ophthalmic diseases. As a result, gene-based therapies are actively being pursued to ameliorate ophthalmic genetic diseases that were once considered untreatable. Recent advances in molecular genetics of ocular diseases and ongoing efforts to develop strategies for the diagnosis and treatment of inherited eye diseases will be presented.

