

Preface

The euphoria we experienced in 1989, in localizing ‘the’ gene for autosomal dominant retinitis pigmentosa to the long arm of chromosome 3, was an exuberance that was exceedingly short-lived. It immediately became apparent, through lack of evidence for genetic linkage in a second family, that ‘one more’ gene remained to be identified. The reality today, is that for this one disease, we are dealing with an immensely complex set of molecular pathologies—considering all hereditary forms of retinopathy, of which RP is the most common, loci for well over two hundred genes have been revealed, and the chances are that the same number again still remain to be identified. It is a demanding enough endeavor to develop a gene-based medicine for a disease in which only a single gene has been implicated, but in considering hereditary degenerative retinopathies, notwithstanding the triumphant successes such as those achieved for restoration of vision in those forms of Leber congenital amaurosis with mutations within the RPE65 gene, the question is: just how logistically and economically feasible is this? If our armament resided only in gene therapy, we have embarked upon a protracted journey. However, in spite of the immense genetic complexity facing us, commonalities are evident in the molecular mechanisms through which vision is lost in these diseases, and much evidence is now available to encourage us to believe that a combination of gene-based and molecular medicines will, inevitably, result in the development of effective therapies for many of these conditions within a realistic timeframe.

Hereditary Retinopathies

Progress in Development of Genetic and Molecular
Therapies

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