Retinal Dystrophies: Functional Genomics to Gene Therapy: No. 255 (Novartis Foundation Symposia)

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Retinal dystrophies are the major causes of incurable blindness in the Western world. Our insight into their aetiology has improved remarkably over the past decade and a number of key genes have been identified. Together with a more detailed understanding of disease processes, this knowledge is stimulating new approaches to therapeutic strategies involving gene therapy, growth factors and retinal cell transplantation.

Molecular genetic studies have provided detailed information on the pathogenesis of retinal dystrophies. An important proof of principle that gene therapy holds great promise for the treatment of these conditions was demonstrated in the *rds* mouse: introduction of a functional copy of the *peripherin* gene subretinally resulted in complete rescue of rod outer segment structure. Novel approaches are being developed based on the manipulation of biochemical pathways that previously were not considered relevant to these diseases. For example, renewed interest in retinal dystrophy pathogenesis led to the successful use of high dose vitamin A treatment in Sorsby fundus dystrophy.

This important new book covers all aspects of retinal dystrophies from the molecular and developmental biology of these disorders to possible therapeutic approaches, with special reference to gene therapy. Specific chapters deal with the molecular genetics of gene therapies, clinical genetic studies, molecular and cellular mechanisms of the development of the disease, functional genomics of retinal diseases, animal models of retinal dystrophies, and finally with studies on gene therapeutic approaches to correcting the disorder. With contributions by many of the leading researchers worldwide, this book is likely to be an important milestone in this rapidly developing field.

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