



Hereditary Retinal Degeneration (Acta Anatomica)

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Special Topic Issue: Cells Tissues Organs 1998, Vol. 162, No. 2-3 Retinal hereditary dystrophies are a heterogeneous group of disorders. They involve both the neuroretina and the retinal pigment epithelium. Although molecular genetics has in many cases revealed the origin of these disorders, the mechanisms leading to progressive degeneration are largely unknown. Recent progress in modern techniques, however, has provided novel insights into the basic pathophysiological mechanisms underlying these dystrophies. Major aspects covered in this special issue include the search for genes with molecular biological methods, mapping and cloning of candidate genes, and correlation of mutations to the clinical phenotypes. Animal models of the various types of degenerative diseases, especially RCS rats, help identify ultrastructural and biochemical correlates of photoreceptor degeneration. They also provide useful systems for studying functional deficits and investigating the immunology of pigment epithelium transplantation. The findings gathered from animal models together with imaging studies and non-invasive function tests performed in patients contribute to a better understanding of the degenerative mechanisms. The ultimate goal is a contribution to the development of treatments for the hundreds of thousands of people world-wide who suffer from such disorders.

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