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Corneal Dystrophies

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Walter Lisch Hanau

Berthold Seitz Homburg/Saar

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Preface

The cornea, basically composed of the epithelium, stroma and endothelium, is the major refractive organ of the optic system in addition to serving as a mechanical barrier. The corneal epithelium is the most regular arrangement of stratified epithelium in the whole human body. The cells, composed of 6–7 different layers, are tightly and orderly arranged without intercellular spaces. We know that some corneal dystrophies are only characterized by the occurrence of epithelial opacities. The contact lens-induced regression of opacities in epithelial corneal dystrophies can be interpreted as a contact lens-induced reduction of epithelial layers. As in other connective tissues, the major portion of the corneal stroma is composed of extracellular matrix macromolecules which are responsible for the strength and transparency of this tissue. Some corneal dystrophies are thought to result in part from abnormalities in corneal stromal cell function. Corneal stromal cells synthesize and degrade matrix materials during corneal morphogenesis and proper metabolism of such materials is essential. Stromal corneal dystrophies recur after decades on the graft due to the long-term transformation of transplant keratocytes into pathological host keratocytes. The corneal endothelium is a monolayer of hexagonal cells that forms the posterior corneal surface. An intact monolayer of endothelial cells is essential for the functional endothelial barrier to preserve a relative dehydration of the stroma and a prerequisite to corneal transparency. If the integrity of the monolayer is breached, corneal edema rapidly develops as we can see in some endothelial corneal dystrophies. The replacement of the posterior cornea, called Descemet's stripping endothelial keratoplasty, represents a modern and sophisticated surgical procedure in the treatment of endothelial corneal dystrophies.

With the revolution in molecular genetics, our understanding of corneal dystrophies has changed in the last 15 years as disorders have been mapped and the genes responsible have been identified. Today we know that phenotypic heterogeneity – the same gene causing different forms of corneal dystrophies – and genotypic heterogeneity – different genes causing a phenotypically identical corneal dystrophy – do exist. Research continues to uncover important knowledge on corneal dystrophies. However, the identification of the gene and mutations in corneal dystrophies can only be interpreted as a start in the mosaic puzzle for uncovering the complex relationships

in the pathophysiological molecular mechanisms. In general, further molecular physiological examinations and the evaluation of animal models are necessary to precisely define the essential protein defect in the different types of corneal dystrophy. The development of a causal therapy for corneal dystrophies must be the big scientific challenge in the future.

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