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Corneal Dystrophies

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Walter Lisch Hanau
Berthold Seitz Homburg/Saar

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Walter Lisch
Department of Ophthalmology
City Hospital of Hanau
Leimenstrasse 20
DE - 63450 Hanau (Germany)

Berthold Seitz
Department of Ophthalmology
University of Saarland
Kirrbergerstrasse 1, Building 22
DE - 66424 Homburg/Saar (Germany)

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List of Contributors

Anthony J. Aldave
The Jules Stein Eye Institute
David Geffen School of Medicine
The University of California
100 Stein Plaza, Los Angeles, CA 90095 (USA)
E-Mail aldave@jsei.ucla.edu

Satoshi Kawasaki
Department of Ophthalmology
Kyoto Prefectural University of Medicine
465 Kajii-cho, Hirokoji-agaru
Kawaramachi-dori, Kamigyo-ku
Kyoto 602-0841 (Japan)
E-Mail bluenova@koto.kpu-m.ac.jp

Arbi J. Khemichian
Kresge Eye Institute
Wayne State University School of Medicine
4717 St Antoine, Detroit, MI 48201 (USA)
E-Mail arbi.khemichian@utsouthwestern.edu

Shigeru Kinoshita
Department of Ophthalmology
Kyoto Prefectural University of Medicine
465 Kajii-cho, Hirokoji-agaru
Kawaramachi-dori, Kamigyo-ku
Kyoto 602-0841 (Japan)
E-Mail shigeruk@ophth.kpu-m.ac.jp

Walter Lisch
Department of Ophthalmology
City Hospital of Hanau
Leimenstrasse 20, DE - 63450 Hanau (Germany)
E-Mail lisch.hanau@t-online.de

H.U. Møller
Department of Pediatric Ophthalmology
Viborg Hospital
DK - 8800 Viborg (Denmark)
E-Mail hans.ulrik.moeller@sygehusviborg.dk

Somasheila I. Murthy
Cornea and Anterior Segment Service
L. V. Prasad Eye Institute
Kallam Anji Reddy Campus,
L. V. Prasad Marg, Banjara Hills,
Hyderabad, 500 034 (India)
E-Mail smurthy@lvpei.org

Varsha M. Rathi
Cornea and Anterior Segment Service
L. V. Prasad Eye Institute
Kallam Anji Reddy Campus,
L. V. Prasad Marg, Banjara Hills,
Hyderabad, 500 034 (India)
E-Mail varsharathi@lvpei.org

Berthold Seitz
Department of Ophthalmology
University of Saarland
Kirrbergerstrasse 1, Building 22,
DE - 66424 Homburg/Saar (Germany)
E-Mail berthold.seitz@uks.eu

Geeta K. Vemuganti
School of Medical Sciences
University of Hyderabad
Hyderabad, 500046 (India)
E-Mail gkvemuganti@gmail.com

Jayne S. Weiss
Chair of Department of Ophthalmology
Herbert Kaufman, Professor of Ophthalmology
Louisiana State University Health Science Center
2020 Gravier Street
New Orleans, LA 70112 (USA)
E-Mail jayneweiss@aol.com

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.

Walter Lisch, Hanau
Berthold Seitz, Homburg/Saar