

CONTENTS

Dedication
Preface: Credits and Acknowledgments

Chapter 1: Introduction

- Classification of the Ichthyoses (Disorders of Cornification) by Vinzenz Oji
- Synopsis of Normal Stratum Corneum Structure and Function
- Historic Pathogenic Concepts
- Function-Driven Pathogenesis of the Ichthyoses
- Permeability Barrier Dysfunction as the 'Driver' of Disease Expression
- Basis for Inflammation in the Ichthyoses
- Basis for Abnormal Desquamation in the Ichthyoses
- Systemic Consequences of Barrier Abnormalities in the DOC
- Utility of Ultrastructure in the Differential Diagnosis of the Ichthyoses
- References

Chapter 2: Inherited Disorders of Lipid Metabolism

- Disorders of Fatty Acid Metabolism (Nonsyndromic)
- Multisystem Diseases of Fatty Acid Metabolism
- Multisystem Diseases of Cholesterol Metabolism
- Multisystem Diseases of Sphingolipid Metabolism
- Defective Lipid Transporters
- References

Chapter 3: Inherited Disorders of Accelerated Desquamation

- Netherton Syndrome (NS)
- Relationship of NS to Atopic Dermatitis
- Peeling Skin Syndrome
- References

Chapter 4: Inherited Disorders of Corneocyte Proteins

- The Keratinopathic Ichthyoses
- Disorders of the Corneocyte Envelope
- Ichthyosis En Confettis
- References

Chapter 5: Appendices

- Appendix 1: Ultrastructural and Histochemical Methods
- Appendix 2: Glossary of Terminology
- Appendix 3: Molecular Diagnostic Resources

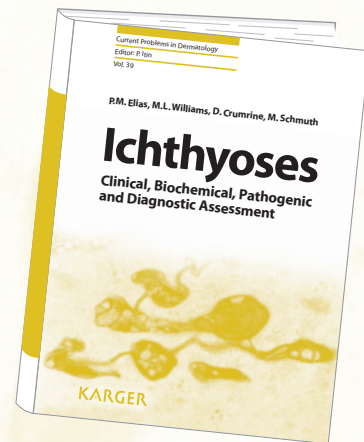
Fields of Interest: Dermatology; Genetics; Metabolism; Pediatrics, Histology,

From molecular genetics to clinical features

P.M. Elias, M.L. Williams, D. Crumrine, M. Schmuth

Ichthyoses

Clinical, Biochemical, Pathogenic and Diagnostic Assessment



The various manifestations of ichthyoses are classified either by their appearance or their molecular genetics. This volume focuses on generalized, inherited disorders of cornification, which constitute an ever-enlarging group of monogenic diseases caused by a large number of genes that affect a broad array of cellular functions.

The authors' overview reflects their unique perspective that the clinical phenotype in the inherited ichthyoses mirrors a 'best attempt' by a metabolically compromised epidermis to maintain a barrier sufficiently impermeable for survival in a desiccating external environment.

The basis for threats to survival is illuminated, and the systemic problems, including growth failure, also reflect a compromised barrier. A new consensus classification of these disorders is provided, and the distinguishing clinical features of each disorder are described. Further, the latest molecular genetic information is succinctly reviewed with up-to-date and comprehensive references. Yet, the major emphases of this volume are on disease pathogenesis and on the identification of key ultrastructural features.

This publication will prove an invaluable aid to dermatologists, pediatric dermatologists and pediatricians dealing with patients with inherited ichthyoses. In addition, clinical geneticists and dermatopathologists will find it interesting reading.

Current Problems in Dermatology, Vol. 39
Elias, P.M.; Williams, M.L.; Crumrine, D.
(San Francisco, Calif.);
Schmuth, M. (Innsbruck)

Ichthyoses
Clinical, Biochemical, Pathogenic and
Diagnostic Assessment
X + 146 p., 89 fig., 15 in color, 9 tab.,
hard cover, 2010
CHF 192.- / EUR 142.- / USD 192.00
ISBN 978-3-8055-9394-6
e-ISBN 978-3-8055-9395-3

www.karger.com/cupde

KARGER

Well-referenced and timely reports from the most active areas of dermatology and skin research

Current Problems in Dermatology

Series Editor: **Itin, P. (Basel)**

ISSN 1421-5721 / e-ISSN 1662-2944

This series presents a selection of new ideas and information in the field of clinical and investigative dermatology. Crossing the borders of skin diseases it explores interdisciplinary strategies, linking dermatology with immunology, allergology and other research areas. Each volume is individually designed to provide an up-to-date presentation of new and interesting topics in dermatology and skin research. The contributing authors are acknowledged experts in their fields. The wide range of topics discussed includes normal and pathological skin conditions, the development of new equipment, drugs and analytical techniques, and recent advances in the treatments that can be offered to patients.

Vol. 38

Management of Psoriasis

Editor: Yawalkar, N. (Bern)

VIII + 196 p., 30 fig. in color, 11 tab., hard cover, 2009

CHF 154.- / EUR 110.- / USD 154.00

ISBN 978-3-8055-9151-5 / e-ISBN 978-3-8055-9152-2

Vol. 37

Lyme Borreliosis

Biological and Clinical Aspects

Editors: Lipsker, D.; Jaulhac, B. (Strasbourg)

VI + 212 p., 13 fig., 8 in color, 11 tab., hard cover, 2009

CHF 215.- / EUR 153.50 / USD 215.00

ISBN 978-3-8055-9114-0 / e-ISBN 978-3-8055-9115-7

Vol. 36

Safety Assessment of Cosmetics in Europe

Editors: Pauwels, M.; Rogiers, V. (Brussels)

XX + 214 p., 17 fig., 47 tab., hard cover, 2008

CHF 168.- / EUR 120.- / USD 168.00

ISBN 978-3-8055-8655-9 / e-ISBN 978-3-8055-8656-6

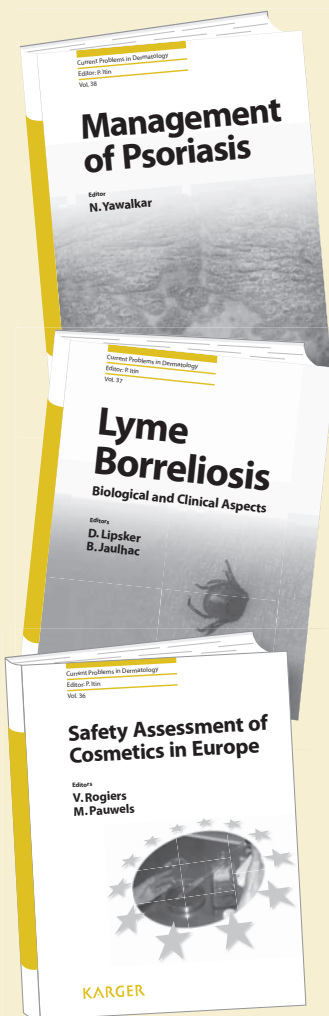
Vol. 1-35 available, please ask for details

Prices subject to change

EUR price for Germany, USD price for USA only

www.karger.com/cupde

KARGER



ORDER FORM

Current Problems in Dermatology

Please send:

copy/ies: **Vol. 39: Ichthyoses**
Clinical, Biochemical, Pathogenic
and Diagnostic Assessment
CHF 192.- / EUR 142.- / USD 192.00
ISBN 978-3-8055-9394-6

the book series **Current Problems
in Dermatology** on continuation
beginning with vol.

information on **vols. 1-35**

All Karger series are available on continuation.
Each volume will be sent upon publication until
order is countermanded.

For easy ordering or further information about the
series log on to: www.karger.com/cupde

Payment

Postage and handling free with prepayment

Please charge this order to my credit card

American Express Diners
 Visa Eurocard MasterCard

Card No.:

Exp. date:

(3 digits in the signature field on the back of Visa and MasterCard)

Check enclosed Please bill me

Name/Address (please print):

E-Mail:

Date:

Signature:

Orders can be placed at agencies, bookstores, directly with the
publisher, or with any Karger distributor.

S. Karger AG, P.O. Box, 4009 Basel (Switzerland)
Fax +41 61 306 12 34, E-Mail orders@karger.ch

USA: S. Karger Publishers, Inc., 26 West Avon Road, P.O. Box 529,
Unionville, CT 06085 (USA), Toll free: 1-800-828-5479

Germany: S. Karger GmbH, 79098 Freiburg
France: Librairie Médi-Sciences, 75007 Paris

Gulf Council Countries, Iran, Middle East, North Africa, Turkey:
Trans Middle East Internat. Distribution Co. Ltd., KaSha, Amman
11953 (Jordan)

Japan: Karger Japan, Inc., Tokyo 113-0034

South East Asia, China and Taiwan: Karger Regional Office, Kuala
Lumpur 50470 (Malaysia)

For details and further representatives and agencies see
www.karger.com/worldwide

To be ordered through: