

---

N. Blau · M. Duran  
M.E. Blaskovics · K.M. Gibson  
(Eds.)

# **Physician's Guide to the Laboratory Diagnosis of Metabolic Diseases**

---

Second Edition

Foreword by C.R. Sriver

With 100 Figures and 270 Tables

 Springer

---

Nenad Blau  
Division of Clinical Chemistry  
and Biochemistry  
University Children's Hospital  
Steinwiesstrasse 75  
8032 Zürich  
Switzerland  
e-mail: [nenad.blau@kispi.unizh.ch](mailto:nenad.blau@kispi.unizh.ch)

Marinus Duran  
Academic Medical Center  
Dept. of Pediatrics  
and Clinical Chemistry  
Meibergdreef 9  
1105 AZ Amsterdam  
The Netherlands  
e-mail: [m.duran@amc.uva.nl](mailto:m.duran@amc.uva.nl)

Milan E. Blaskovics  
3639 Amesbury Road  
Los Angeles, CA 90027  
USA  
e-mail: [blaskk@earthlink.net](mailto:blaskk@earthlink.net)

K. Michael Gibson  
Biochemical Genetics Laboratory  
Dept. of Molecular and Med. Genetics  
Oregon Health & Science University  
2525 SW 3rd Avenue, Suite 350  
Portland, Oregon 97201, USA  
e-mail: [gibsonm@ohsu.edu](mailto:gibsonm@ohsu.edu)

ISBN 3-540-42542-X 2nd Edition Springer Berlin Heidelberg New York  
2nd Edition 2003. Corr. 2nd printing 2005

Title of the 1st Edition:

N. Blau, M. Duran, M.E. Blaskovics

Physician's Guide to the Laboratory Diagnosis of Metabolic Diseases

© 1996 Chapman & Hall

Library of Congress Cataloging-in-Publication Data

Physician's guide to the laboratory diagnosis of metabolic diseases / [edited by] N. Blau

.. [et al.]. – 2nd ed. p.; cm. Includes bibliographical references and index.

ISBN 3-540-42542-X (hd.: alk.paper)

1. Metabolism – Disorders – Diagnosis – Handbooks, manuals, etc. 2. Metabolism,

Inborn errors of – Diagnosis – Handbooks, manuals, etc. 3. Diagnosis,

Laboratory – Handbooks, manuals, etc. I. Blau, N. (Nenad), 1946 –

[DNLM: 1. Metabolism, Inborn Errors – diagnosis. 2. Diagnosis, Differential.

3. Laboratory Techniques and Procedures. WD 205 P578 2002]

RB147.P476 2002 616.3'9075–dc21 2002021642

This work is subject to copyright. All rights are reserved, whether the whole or part of the material is concerned, specifically the rights of translation, reprinting, reuse of illustrations, recitation, broadcasting, reproduction on microfilm or in any other way, and storage in data banks. Duplication of this publication or parts thereof is permitted only under the provisions of the German Copyright Law of September 9, 1965, in its current version, and permission for use must always be obtained from Springer-Verlag. Violations are liable for prosecution under the German Copyright Law.

Springer is a part of Springer Science+Business Media

[springeronline.com](http://springeronline.com)

© Springer-Verlag Berlin Heidelberg 2003

Printed in Germany

The use of general descriptive names, registered names, trademarks, etc. in this publication does not imply, even in the absence of a specific statement, that such names are exempt from the relevant protective laws and regulations and therefore free for general use. Product liability: The publishers cannot guarantee the accuracy of any information about the application of operative techniques and medications contained in this book. In every individual case the user must check such information by consulting the relevant literature.

Graphics: Günther Hippmann, Nürnberg  
Cover-Design: Erich Kirchner, Heidelberg

Typesetting: K + V Fotosatz, Beerfelden  
Printing and bookbinding: Stürtz AG, Würzburg

Printed on acid-free paper

SPIN 11319092

24/3150 PF

5 4 3 2 1 0