

BOOK REVIEWS

A book with a comprehensive look at metabolic disorders

Paving the path for better understanding of IEM

Inborn Errors of Metabolism: From Neonatal Screening to Metabolic Pathways,
Edited by: B Lee and F Scaglia
Published by: Oxford University Press
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The book *'Inborn Errors of Metabolism: From Neonatal Screening to Metabolic Pathways'* edited by two well-known specialists in the area of inborn errors of metabolism (IEMs), Brendan Lee and Fernando Scaglia, gives us a different vision in this field.

It was a pleasure to review this book, to be able to browse through it and appreciate it. This is not a common textbook, neither is it a regular book on IEMs. It is a 'guide' into the field of metabolic diseases; it is a comprehensive and updated history of a rapidly evolving field. This is an interesting and original work that will certainly be relevant for students as well as specialists.

The book is cleverly divided into three major sections and organized into 16 chapters covering various areas of IEMs and written by world experts in the respective fields. The three major sections cover newborn screening, metabolic pathways and therapeutic approaches. In this book, information is gathered with the help of comprehensive tables, illustrations and schemes. The illustrations are elegant and easy

to understand, the book itself is easy to follow as it provides well-written up-to-date reviews organized by topics.

This book provides a concise yet detailed coverage of the field of IEMs by addressing the pathway organization and its malfunctions. The presentation of a natural history overview provides a new perspective and highlights the fact that IEMs are a never-ending area rich in complexity. Methods of diagnosis and their underlying principles, as well as present and potential treatment strategies, are thoroughly discussed in the various chapters.

Although lysosomal diseases are not addressed in a single chapter; special attention is given to these organelle disorders under the focus on recent advances in therapeutics.

Written by world-renowned specialists this is a compact, yet very complete, guide into the world of IEMs. This book provides a fresh look at metabolic diseases; the importance of the pathway idea goes beyond the individual diseases and gives an integrated vision of the defects in relation to the mechanisms involved. Undoubtedly, the pathway approach makes this book particularly original in an era of translational research.

This is a most valuable contribution to the field of IEMs and will become an ideal companion for students and clinicians. I found the hard cover version particularly resistant and adequate for frequent use.

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Potentiating growth in loss: dementia as a beginning?

Beyond Loss: Dementia, Identity, Personhood
Edited by: Lars-Christer Hyden, Hilde Lindemann and Jens Brockmeier
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I approached this book with considerable interest professionally as I am a Clinical Geneticist by trade, and for 18 months I have held the position of Director of Mental Health Services in my University Health Board with responsibilities for delivery of services to people affected by dementia. I also find myself at that time of realisation when

one recognises that the greater part of one's life has been lived and thoughts of what lies ahead often creep, unbidden, into conscious thought.

Anyone who has lived or worked with individuals and families affected by dementia, in all its presentations and aetiologies, will recognise many of the lived experiences, vignettes and challenges explored in this collection of essays. Every chapter presents different facets of the condition and examines the impact of dementia on the individual and on all those with whom they interact, from immediate family and friends through to wider social issues.

This book is well researched and extensively referenced. It is divided into three sections: *Persons, Personhood and Dignity; Identity, Agency, Embodiment; Communication, Family and Institutions*. There is a detailed exploration of what it is to be human and how dementia has an impact on that. This exploration includes examination of

notions of dignity, the meanings of love and an analysis of aspects of independence, couplehood and identity. These are issues that are often a complex, frightening struggle for people diagnosed with dementia and for their families. In particular, the loss of 'self' for the individual affected, and for their life partner who changes from being spouse, lover, partner in everything, to being carer with all the associated challenges, is an area that is particularly painful for the families I encounter.

Perhaps inevitably, some of the content feels uncomfortable, such as the description by one carer of being 'chained to a corpse'. As a person who shares their life with a classical musician with a close family history of early-onset dementia, I found the exploration of *musical embodiment, selfhood and dementia* especially thought-provoking, both as a consideration of what it feels like to 'lose' parts of your loved one to dementia, and the more optimistic description of music remaining an avenue of communication with some individuals who otherwise cannot be reached by the verbal and physical exchanges they might once have been able to take part in.

Personally, I do not have psychological theorems and language easily at my fingertips, which meant that parts of this book were difficult to negotiate. The book is not an easy read nor do I feel that it could be described as a practical 'aid memoire'. However, as a detailed reference book that uses psychological and philosophical approaches to extensively explore the effects of dementia, based on evidence of considerable experience and research, this is an erudite work of breadth and detail.

In summary, this book examines the impact, positive and negative, dementia can have on the lives of those affected and those around them, and it is a fantastic reference book from which valuable insights and information can be gleaned about dementia, identity and personhood.

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