



Preface to 2nd Edition

The premise upon which the first edition of this book was based has not changed. Specialization in medicine has decreased, more generalists are encouraged, and governments have reduced programs for postdoctoral training. As a consequence, the little that is taught in Medical schools about inborn errors of metabolism is, if anything, less than before.

The area of molecular genetics dealing with the mechanisms and the basic fault in an inborn error of metabolism, on the other hand, has increased almost exponentially. Unfortunately, this area of science can only operate to deduce the problem after a fault has been recognized.

It is the recognition of the patient with a fault, the so-called “inherited” defects that this book is about. The book is therefore targeted towards physicians and other individuals dealing with diagnosing these relatively rare disorders. Recognizing an individual with these disorders is often difficult because of the fact that they “blend in” with many common medical conditions. The signs and symptoms often do not stand out so that a patient may live for years with recurring problems and the condition never correctly diagnosed.

This book presents the signs and symptoms of most of the recognized inborn errors of metabolism in relation to age. There is a chronological sequence of signs and symptoms from infancy through childhood, adolescence and adulthood, and in addition, normal and pathological values are provided for each of the disorders so that one does not have to wonder about the significance of laboratory tests and reported values.

Recognized authorities describe each disorder. Based upon their experience, they have created flow charts and diagnostic algorithms and recommend confirmatory tests and initial treatment schemes to help the uninitiated.

Since the first edition was published in 1996, 43 new disorders have been recognized and are described in this edition. There is little doubt that more new disorders will be recognized, so that this current effort is really a “work in progress”.

To that end, this edition is utilizing the progress made with computer technology to make accessing the data a little easier. A CD-ROM is in-

cluded with this edition that will allow the user to find a disorder and search via a program which uses key words.

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