



Foreword to 2nd Edition

The first edition of this book, published in 1996, was a success (at least in one way – it is sold out) because it provides useful information and thus enhances the care of patients with hereditary metabolic diseases. One message in the original Foreword is having a particular impact; the present edition of the Book will have a CD-ROM format, allowing authors to provide pictures, chromatograms or even short videos. All of the books serving our field are moving in this direction.

Significant developments have occurred over the past six years between editions of the *Physician's Guide*. For example, a draft sequence of the Human Genome is now available. As genes are identified there has been a proliferation of locus specific mutation databases (<http://ariel.ucsf.edu.au:80/~cotton/mdi.htm>), many of them linked to OMIM (<http://www.ncbi.nlm.nih.gov/Omim/>). These databases and much other valuable information are also accessible through the Human Genome Sequencing Consortium and Human Genome Central (<http://www.ensembl.org/genome/central>). The relevance of mutation analysis at loci associated with disease and its practical role in medicine have become significant concerns and they will form a leitmotif in this Book.

Meantime, the number of so-called orphan genetic diseases, among them the hereditary metabolic diseases, increases. One can identify many of them through OMIM, the “genomic catalogue” of human phenotypic variation. The fact of this increase means that the *Physician's Guide* is ever challenged; it will always be attempting to keep up with what is new. When it is on-line and becomes a continuously expanding *Physician's Guide*, it will be able to do that; it will become a complementary resource to those many others serving genetic medicine.

Tandem mass spectrometry has entered the diagnostic arena; this powerful technology is now in place at referral centers across the world. As an important resource in laboratory diagnosis of metabolic diseases, it is expanding the repertoire of diagnoses and problems to address. Here is another reason for continuing relevance of the *Physician's Guide*.

The range of resources for patients and for those who care for them also grows. For example, GeneTests (<http://www.genetests.org>) is a reliable curated knowledge base in which many inborn errors of metabolism ap-

pear with links to other resources. Yet another for readers of this book, is Metab-L (<http://www.franken.de/lists/metab-l/>) a listserve acting as a site for information exchange. It is not hard to predict a long and useful life for the *Physician's Guide to the Laboratory Diagnosis of Metabolic Diseases*, in its present form, its transformations and its links.

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