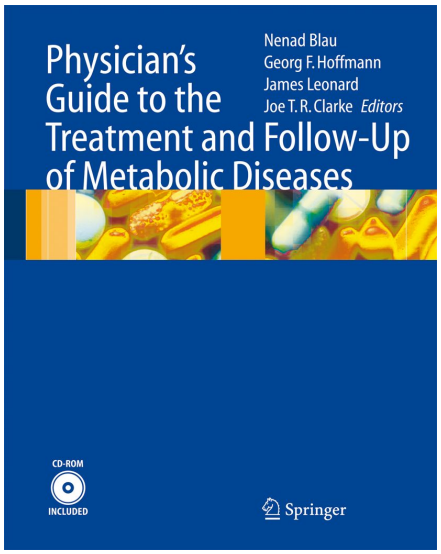

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N. Blau, G.F. Hoffmann, J.V. Leonard, J.T.R. Clarke (Eds.)

Physician's Guide to the Treatment and Follow-Up of Metabolic Diseases

- **Indispensable and concise guide to inherited metabolic diseases for the clinician**
- **World experts give practical advice and guidance for daily practice**

The greatest dif?culty in life is to make knowledge effective, to convert it into practical wisdom. Sir William Osler. The inborn errors of metabolism, as a group of metabolic diseases, are re- tively rare and are sometimes called "orphan diseases. " As a group, they account for about 1 in 2,500 births (Applegarth et al. 2000) and, as a cumulative group reaching 20 years of age, their prevalence is about 40 cases per 100,000 popu- tion. In terms of patient days of continuous supervision and care, hundreds of thousands of such days are involved per generation of these patients. Although experience with these diseases as a class may be small and people expert in their management may be relatively few, in the years to come many caregivers will become involved. This book offers help to them. Until the mid-twentieth century, hereditary metabolic and other genetic diseases were considered to be purely "genetic" problems. Destiny would take its course, treatment did not exist, and genetic counseling about recurrence risks was virtually all that could be offered. Phenylketonuria (PKU) was then shown to be a treatable genetic disease in which early diagnosis and effective treatment prevented the disease (mental retardation) in PKU. Other genetic diseases for which an environmental experience was an essential component of cause (e. g. , exposure to a dietary component or a drug) were then seen to yield to treatment.