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## How To Use This Book

This book is meant to supply clinicians and clinical biochemists with data to facilitate the rapid diagnosis of an inherited metabolic disease. No information about detailed laboratory methods is given, rather, the relationship between laboratory data and clinical signs and symptoms is stressed. Entry to the book is by scanning either of the indices, i.e. the signs and symptoms index, the tests index or the disorders index. Due to the great clinical variability of inherited metabolic diseases one should not restrict oneself to one disorder when observing a given symptom or sign.

Most chapters have a uniform layout as given below. In a few chapters, however, this was not possible and information is given for the entire related group of disorders in the chapter.

### Introduction

The introduction gives a brief overview of the clinical conditions described in the chapter and relates them to the biochemical abnormalities. Key references for further reading are given here.

### Nomenclature

Disorders in each chapter are numbered in accordance with the corresponding MIM number [1] and chromosomal localization if known.

### Metabolic Pathway

Disorders are identified by corresponding reference numbers at the step where the defect is localized. Pathological metabolites ('markers') are given in most chapters.

### Signs and Symptoms

The tables describe most, if not all, of the signs and symptoms for each disorder, including its reference number, and most important laboratory tests, in relation to age. In all instances, the signs and symptoms are in the untreated (natural) state.

± indicates that a sign or symptom *may* occur but is not inevitably present.

- + indicates that a sign or symptom is always or nearly always present. If there are significant clinical signs and symptoms which exceed the usual, or if changes occur, this is indicated with + to + + +, etc.
- n (normal) is used only when it is significant and may be useful in distinguishing one condition from another.

Relative increases or decreases of substances, compounds, metabolites etc. are indicated with the use of arrows; for example, metabolite X ↑ to ↓↓↓. Where metabolite X *may* change, it would be indicated by n-↑ for a possible increase or ↓-n for a possible decrease, whatever the case.

In all tables, the test substance, material, compound or metabolite, etc., is listed and the source – (U), (B), (CSF), (P), or (RBC) – is given in parentheses, with an arrow or arrows indicating increase/decrease or relative increase/decrease.

Body fluids, cells, tissues, etc. are defined as:

P	plasma	CV	chorionic villi
S	serum	AF	amniotic fluid
B	blood	AFC	amniocytes
U	urine	CCV	cultured chorionic villi
CSF	cerebrospinal fluid	PLT	platelets
RBC	red blood cells	WBC	white blood cells
LYM	lymphocytes	Hb	hemoglobin
FB	fibroblasts	creat	creatinine

Age groups are defined as:

Neonatal	birth to 1 month
Infancy	1–18 months
Childhood	1.5–11 yr
Adolescence	11–16 yr
Adulthood	>16 yr

### Normal Values/Pathological Values/Differential Diagnosis

Reference and pathological values are listed for all parameters relevant to the diagnosis according to the specimen (e.g. P, U, CSF, etc.) and age. For some parameters, normal values depend on methodology and may differ from chapter to chapter. Methods are specified where necessary. Pathological values are listed either as absolute values or with symbols (e.g. ↑, ↓, etc.) according to the disorder. Values are limited to the analyses which can be performed in a well-equipped laboratory for selective screening. Data on enzyme studies are not given in most cases.

### Loading Tests

There is a brief description of the tests, with a table or figure to illustrate the interpretation.

### Diagnostic Flow Chart

The flow charts use simple yes/no algorithms to demonstrate the sequence for differential diagnosis; starting with clinical symptoms or general tests and proceeding to specific tests and a final diagnosis.

### Specimen Collection

This table lists preconditions, material, handling and pitfalls for each parameter used in the diagnosis.

### Prenatal Diagnosis

This table lists the tissue or specimen, timing and pitfalls for each disorder.

### DNA Analysis

This table lists the tissue or specimen and methodology for each disorder.

### Initial Treatment

This section outlines briefly urgent treatment to consider before a definitive diagnosis is established for each (or each group of) disorder(s). Long-term treatment is not considered in this book; other books cover this topic.

### Indices

Three indices are included: (1) Disorders; (2) Signs and Symptoms, and (3) Tests. Each entry is linked to the corresponding disorder or page.

### Reference

1. OMIM, Online Mendelian Inheritance in Man,  
<http://www3.ncbi.nlm.nih.gov/omim/>, 2002