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

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Editors

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Milan Blaskovics (1928–2013)

The Editors take this opportunity to acknowledge Dr. Milan Blaskovics, one of the founding members of the Editorial team who passed away on 13 December 2013, for his longstanding efforts and contributions to our series Physician's Guide and Laboratory Guide in Metabolic Disease, as well as his longstanding involvement in improving the life of children and adults with inherited metabolic diseases, especially phenylketonuria. In recognition of his constant guidance and valuable contributions, we wish to dedicate this edition of the Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases to his memory.

Nenad Blau
Marinus Duran
K. Michael Gibson
Carlo Dionisi-Vici

Foreword

In a recent study of urea cycle disorders, two of the main findings were that many patients presented as adults, and at all ages there were frequently long delays before the correct diagnosis was made. It is to be hoped that this book, aimed at physicians responsible for all ages, children, adolescents, and adults, will reduce delays in the diagnosis of inborn errors of metabolism. This is vital as early and rapid diagnosis is often the key to a good outcome.

This new book brings up-to-date and combines two earlier volumes, *Physician's Guide to the Treatment and Follow-Up of Metabolic Diseases* and *Physician's Guide to the Laboratory Diagnosis of Metabolic Diseases*. It is not a conventional textbook but rather a book for reference with information in a form that is readily accessible. The format is broadly the same as before with some text and much of the information presented in tables. This single volume is an important step forward as the diagnosis and management of inborn errors are highly dependent on laboratory investigations, so it makes sense to combine the clinical with the laboratory aspects. The clinical chapters cover the full range of inborn errors covering clinical symptoms and signs with management. The chapters on investigations range from the simple to the complex and include imaging as well as laboratory tests.

Most inborn errors are rare so experience is generally spread quite thinly. This volume will undoubtedly be useful for busy physicians faced with difficult clinical problems that need quick decisions.

Oxford, UK — James Leonard

Preface

The editors feel that the present edition of the *Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Metabolic Diseases* takes clinical practice in rare metabolic disorders to the next level. Exactly 100 experts in the field authored 55 chapters dealing with a total of 530 inherited metabolic disorders. The structure of the book and chapters was redesigned, and many new chapters and new disorders were added to the current edition. Additionally, the entire content of this edition is stored in a single database, which will become the foundation for the future knowledgbase of inborn errors of metabolism, IEMBASE.

The major goals of this edition, however, remain comparable to those of earlier editions. The 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 question the significance of laboratory tests and reported values.

Recognized authorities have described each disorder. Based upon their experience, they have created flow charts and diagnostic algorithms and have recommended a variety of confirmatory tests and initial treatment schemes to help those practitioners who do not have extensive experience in inborn errors of metabolism. The second part of each chapter describes the treatment of groups of disorders in more detail. With regard to the latter, the current edition utilizes expanding progress with computer technology to make accessing all of the data in the book seamless. In addition to the hardcover version, an ebook will be available that will allow the user to rapidly locate a disorder utilizing standard searches with keywords. It is the hope of the editors that the readers will find this edition helpful, both now and in the future, for the treatment and care of patients with inborn errors of metabolism.

Heidelberg, Germany — Nenad Blau
Amsterdam, The Netherlands — Marinus Duran
Spokane, WA, USA — K. Michael Gibson
Rome, Italy — Carlo Dionisi-Vici

How to Use This Book

This book is meant to supply clinicians and clinical biochemists with data that should facilitate the 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 highlighted. Subsequently, the current knowledge on the immediate emergency intervention, standard treatment, and experimental options are given. Entry to the book is achieved 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 provided.

Nomenclature
Disorders in each chapter are numbered in accordance with the corresponding OMIM number [1], gene symbols and gene products 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 the most important laboratory tests, in relation to age. In all instances, the signs and symptoms are in the untreated (natural) state. The signs written in **bold** are characteristic feature of the particular disease.

± 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, whichever the case.

In all tables, the test substance, material, compound, metabolite, etc., are listed and the source—(U), (B), (CSF), (P), (RBC), etc.—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
BM	Bone marrow		

Age groups are defined as:

Neonatal	Birth to 1 month
Infancy	1–18 months
Childhood	1.5–11 years
Adolescence	11–16 years
Adulthood	>16 years

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) 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., ↑, ↓) according to the disorder. Values are limited to the analyses which can be performed in a laboratory experienced in selective screening. Data on enzyme studies are not given in most cases, which can be found in the pertinent literature.

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.

Treatment and Follow-Up
This section outlines urgent treatment to consider before a definitive diagnosis is established for each (or each group of) disorder(s). Long-term treatment and alternative therapeutic options are highlighted in this book.

Indices

Three indices are included: (1) disorders, (2) signs and symptoms, and (3) tests and medications. Each entry is linked to the corresponding disorder or page.

Reference

1. OMIM (2013) Online Mendelian Inheritance in Man®, http://www.omim.org

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Abbreviations

17βHSD	17-beta-hydroxysteroid dehydrogenase
17OHP	17-hydroxyprogesterone
1H-MRS	Proton magnetic resonance spectroscopy
2KG	2-ketoglutaric acid
2OGA	2-oxoglutaric acid
3β-HSDH	3-beta-hydroxysteroid dehydrogenase
3HBD	D-3-hydroxy-*n*-butyrate dehydrogenase
3MCC	3-methylcrotonyl-CoA carboxylase; 3-methylcrotonylglycinuria
3MT	3-methoxytyramine
3-OH-GA	3-hydroxyglutaric acid
3OMD	3-*O*-methyldopa
5-ALA	5-aminolevulinate
5HIAA	5-hydroxyindoleacetic acid
5HTP	5-hydroxytryptophan
5-LO	5-lipoxygenase
5-LOAP	5-lipoxygenase-activating protein
5MTHF	5-methyltetrahydrofolate
7DHC	7-dehydrocholesterol
AADC	Aromatic L-amino acid decarboxylase
AAs	Aminoacidopathies
AASA	Alpha-aminoadipic semialdehyde
AASS	2-aminoadipic semialdehyde synthase
ABAT	GABA transaminase gene
ABCA1	ATP-binding cassette, subfamily member A1
ABCC8	ATP-binding cassette, subfamily C; hyperinsulinism of infancy
ABCD1	ALD protein
ABCD4	Lysosomal export of cobalamin
ABL	Abetalipoproteinemia
ABS	Antley-Bixler syndrome
ACAD2	Isovaleryl-CoA dehydrogenase gene
ACAD8	Isobutyryl-CoA dehydrogenase gene
ACAD9	Acyl-CoA dehydrogenase 9 gene
ACADM	Medium-chain acyl-CoA dehydrogenase gene
ACADS	Short-chain acyl-CoA dehydrogenase gene
ACADSB	2-methylbutyryl-CoA dehydrogenase gene
ACADVL	Very-long-chain acyl-CoA dehydrogenase gene
ACAT1	Methylacetoacetyl-CoA thiolase gene
ACAT2	Cytosolic acetoacetyl-CoA thiolase gene
ACC	Acetyl-CoA carboxylase
ACOX1	Peroxisomal acyl-CoA oxidase 1
ACSF3	Acetyl-CoA-synthase family 3
ACT	Aspartate carbamoyltransferase

ACTH	Adrenocorticotropic hormone
ADA	Adenosine deaminase
ADCK3	AARF domain-containing kinase 3
ADK	Adenosine kinase
ADKD	Adenosine kinase deficiency
AdoCbl	Adenosylcobalamin
AdoHcy	Adenosylhomocysteine
AdoMet	Adenosylmethionine
ADSL	Adenylosuccinate lyase
AEZ	Acrodermatitis enteropathica
AFC	Amniotic fluid cells
AGA	Glycosylasparaginase
αGalA	α-Galactosidase A
AGAT	Arginine:glycine amidinotransferase
AGAT	L-arginine:glycine amidinotransferase
AGC1	Global cerebral hypomyelination due to AGC1 defect
AGL	Amylo-1,6-glucosidase
AGPS	Alkyl-DHAP synthase
AGT	Alanine-glyoxylate aminotransferase
AGU	Aspartylglucosaminuria
AGXT	Alanine-glyoxylate aminotransferase
AHCY	S-adenosylhomocysteine hydrolase
AICAR	5′-phosphoribosyl-5-aminoimidazole-4-carboxamide
AICART	5′-phosphoribosyl-5-aminoimidazole-4-carboxamide transformylase
AIP	Acute intermittent porphyria
AIS	Androgen insensitivity syndrome
AKR1C	3α-hydroxysteroid dehydrogenase deficiency
AKR1D1	Delta(4)-3-oxosteroid-5 beta-reductase
AKU	Alkaptonuria
ALA	5-aminolevulinic acid
ALAD	5-aminolevulinate dehydratase
ALAS	5-aminolevulinate synthase
ALAS2	5-aminolevulinate synthase 2
ALD	Adrenoleukodystrophy
ALDH18A1	Pyrroline-5-carboxylate synthetase gene
ALDH4A1	Pyrroline-5-carboxylate dehydrogenase gene
ALDH5A1	Succinic semialdehyde dehydrogenase gene
ALDH6A1	Methylmalonate semialdehyde dehydrogenase gene
ALDH7A1	Alpha-aminoadipic semialdehyde dehydrogenase gene
Aldo	Aldosterone
ALDOA	Aldolase A
ALDOA-D	Glycogen storage disease type XII
ALDOB	Fructose-1-phosphate aldolase
ALG 12	Mannosyltransferase 8
ALG 2	Mannosyltransferase 2
ALG 8	Glucosyltransferase 2
ALG1	Mannosyltransferase 1
ALG1	Mannosyltransferase 7-9
ALG11	Mannosyltransferase 4-5
ALG3	Mannosyltransferase 6
ALG6	Glucosyltransferase 1
ALP	Alkaline phosphatase
ALSDP	Doss porphyria

AMACR	2-methylacyl-CoA racemase
AME	Apparent mineralocorticoid excess
AMN	Adrenomyeloneuropathy; amnionless
AMP	Adenosine monophosphate
AMPD	Adenosine monophosphate deaminase deficiency
AMPD1	Adenosine monophosphate deaminase
AMPDA	Adenosine-5′-monophosphate deaminase
AMPK-A	Constitutional AMP-activated protein kinase activation
AMRF	Action myoclonus-renal failure syndrome
α-NAGA	α-N-acetylgalactosaminidase
ANCL	Adult neuronal ceroid lipofuscinoses
ANGPTL3	Angiopoietin-like protein 3
AOA1	Ataxia oculomotor apraxia 1 (AOA1)
AP1S1	Adaptor-related complex protein 1; Mednik syndrome
APOA1	Apolipoprotein A-I
APOB	Apolipoprotein B
APOC2	Apolipoprotein C-II
APOE	Apolipoprotein E
APRT	Adenine phosphoribosyltransferase
APTX	Aprataxin
AR	Aldose reductase; androgen receptor
ARC	Arthrogryposis, renal dysfunction, and cholestasis
ARG1	Arginase 1
ARH1	Autosomal recessive hypercholesterolemia (ARH)
ARO	Aromatase deficiency
ARSA	Arylsulfatase
ARSB	N-acetylgalactosamine-4-sulfatase
ASA	Arylsulfatase A; argininosuccinic acid
ASAH1	Acid ceramidase
ASL	Adenylosuccinate lyase; argininosuccinic aciduria
ASPA	Aspartoacylase (aminoacylase 2)
ASS1	Argininosuccinate synthetase
AThDP	Adenosine thiamine diphosphate
AThTP	Adenosine thiamine triphosphate
ATIC	AICAR transformylase/IMP cyclohydrolase
ATP	Adenosine triphosphate
ATP13A2	Lysosomal type 5 P-type ATPase gene
ATP6V0A2	Vesicular H(+)-ATPase subunit a2 gene
ATP6V0A2	ATPase H + transporting V0 subunit 2 gene
ATP7A	Copper-transporting P-type ATPase gene
ATP8B1	ATP8B1 (type-4 P-type ATPase) gene
AUH	3-methylglutaconyl-CoA hydratase
B	Corticosterone
B^0AT	Sodium-dependent neutral amino acid transporter
B3GALTL	O-fucose-specific beta-1,3-N-glucosyltransferase gene
B4GALT1	Beta-1,4-galactosyltransferase 1 gene
B4GALT7	Beta-1,4-galactosyltransferase 7 gene
BA CoA LD	Bile acid-CoA ligase deficiency
BAAT	Bile acid-CoA amino acid N-acyltransferase
BAT	BCAA aminotransferase
BCAA	Branched-chain amino acids
BCAT	BCAA aminotransferase deficiency
BCAT1	Branched-chain amino acids transporter

BCAT2	Branched-chain amino acids transporter
BCKDC	Branched-chain a-keto acid dehydrogenase complex
BCKDHA	Branched-chain alpha-keto acid dehydrogenase complex
BH2	7,8-dihydrobiopterin
BH4	Tetrahydrobiopterin
BHMT	Betaine-homocysteine methyltransferase
Bio	Biopterin
BKT	Alpha-methylacetoacetic aciduria
BMT	Betaine-homocysteine methyltransferase
BSEP	Bile salt export pump
BTD	Biotinidase
BVVLS	Brown-Vialetto-Van Laere syndrome
BWS	Beckwith-Wiedemann syndrome
C27-3β-HSD	3β-hydroxy-Δ5-C27-steroid dehydrogenase/isomerase
C5-DC	Glutarylcarnitine
C7orf10	Glutaryl-CoA oxidase
CACT	Carnitine acylcarnitine translocase
CAH	21-hydroxylase deficiency
CAH	11-Beta-hydroxylase type I deficiency
CAH	Congenital adrenal hyperplasia
CaOx	Calcium oxalate
CARN	Carnosinemia and homocarnosinosis
Cbl	Vitamin B_{12}
cblA	Adenosylcobalamin synthesis defect-cbl A
cblB	Adenosylcobalamin synthesis defect-cbl B
cblC	Adenosylcobalamin and methylcobalamin synthesis defect-cblC
cblD-HC	Methylcobalamin synthesis defect-cblD-HC
cblD-MMA	Adenosylcobalamin synthesis defect-cblD-MMA
cblD-MMA/HC	Combined MMAuria and homocystinuria
cblE	Methionine synthase reductase deficiency-cblE
cblF	Adenosylcobalamin and methylcobalamin synthesis defect-cblF
cblG	Methionine synthase deficiency-cblG
cblJ	Adenosylcobalamin and methylcobalamin synthesis defect-cblJ
Cbl-TC	Holotranscobalamin
CBS	Cystathionine beta-synthase
CCHLND	SLC33A1 deficiency with low serum copper and ceruloplasmin
CD	Canavan disease
CD	Collecting duct
CD320	CD32 receptor
CDG	Congenital defects of glycosylation
CDG Ie	GDP-Man:Dol-P mannosyltransferase subunit 1 deficiency-CDG Ie
CDG-Id	Mannosyltransferase 6 deficiency-CDG-Id
CDG-If	Dol-P-Man utilization 1 deficiency
CDG-Ih	Glucosyltransferase 2 deficiency-CDG-Ih
CDG-Ii	Mannosyltransferase 2 deficiency-CDG-Ii
CDG-Ij	UDP-GlcNAc:Dol-P-GlcNac-P transferase deficiency-CDG-Ij
CDG-Il	Mannosyltransferase 7-9 deficiency-CDG-IL
CDO	Cysteine dioxygenase
CDPX2	3β-hydroxysteroid-delta8, delta7-isomerase deficiency; chondrodysplasia punctata 2
CEP	Congenital erythropoietic porphyria
CETP	Cholesteryl ester transfer protein
CFD	Cerebral folate deficiency

CHI	Congenital hyperinsulinism
CHILD	Congenital hemidysplasia, ichthyosis, and limb defects
CHP	Chronic hepatic porphyria
CHSY1	Chondroitin sulfate synthase 1
CHSY1-CDG	Chondroitin sulfate synthase 1 deficiency
CI	Complex I deficiency
CII	Complex II deficiency
CIII	Complex III deficiency
CIV	Complex IV deficiency
CKS	CK syndrome
CLN1	Ceroid lipofuscinosis, neuronal, 1
CLN10	Ceroid lipofuscinosis, neuronal, 10
CLN12/ PARK9	Ceroid lipofuscinosis, neuronal, 12
CLN13	Ceroid lipofuscinosis, neuronal, 13
CLN14/ EPM3	Ceroid lipofuscinosis, neuronal, 14
CLN2	Ceroid lipofuscinosis, neuronal, 2
CLN3	Ceroid lipofuscinosis, neuronal, 3
CLN3	Lysosomal transmembrane CLN3 protein
CLN4A	Ceroid lipofuscinosis, neuronal, 4A
CLN4B	Ceroid lipofuscinosis, neuronal, 4B
CLN5	Ceroid lipofuscinosis, neuronal, 5
CLN6	Ceroid lipofuscinosis, neuronal, 6
CLN7	Ceroid lipofuscinosis, neuronal, 7
CLN8	Ceroid lipofuscinosis, neuronal, 8
CLN8-EPMR	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant
CM	Chylomicron
CMAMMA	Combined malonic and methylmalonic aciduria
CMO	Corticosterone methyl oxidase deficiency
CNCL	Congenital neuronal ceroid lipofuscinoses
CNDP1	Carnosinase
COG1	COG complex 1
COG5	COG complex 5
COG6	COG complex 6
COG7	COG complex 7
COG8	COG complex 8
COMT	Catecholamine methyltransferase
CoQ	Coenzyme Q
CoQ10	Coenzyme Q10; ubiquinone
COQ2	4-hydroxybenzoate-polyprenyltransferase
COQ6	CoQ6 monooxygenase
COQ9	Coenzyme Q10
COX	Cytochrome C oxidase
COXPD1	Combined oxidative phosphorylation defect 1
COXPD2	Combined oxidative phosphorylation defect 2
COXPD3	Combined oxidative phosphorylation defect 3
COXPD4	Combined oxidative phosphorylation defect 4
COXPD5	Combined oxidative phosphorylation defect 5
COXPD6	Combined oxidative phosphorylation defect 6
COXPD7	Combined oxidative phosphorylation defect 7
CP	Ceruloplasmin
CPOX	Coproporphyrinogen oxidase
CPS1	Carbamoyl phosphate synthetase I
CPS2	Carbamoyl phosphate synthetase 2

CPT I	Carnitine palmitoyltransferase I
CPT II	Carnitine palmitoyltransferase II
CPT1	Carnitine palmitoyltransferase
CPT2	Carnitine palmitoyltransferase 2
CR	Carbonyl reductase
CRD	Cortisone reductase deficiency
CRD/ARD	Refsum disease (classic, adult)
CrT	Creatine transporter
CSAT	Cysteine sulfinate α-oxoglutarate aminotransferase
CSD	Cysteine sulfinate decarboxylase
CSE	Cystathionine γ-lyase
CT	Cytosolic acetoacetyl-CoA thiolase
CTH	Cystathionine gamma-lyase; cystathionase
CTLN1	Citrullinemia type I
CTLN2	Citrullinemia type II
CTNS	Cystinosin
CTNS	Cystinosin
CTSA	Protective protein/cathepsin A
CTSD	Cathepsin D
CTSF	Cathepsin F
CTX	Sterol 27-hydroxylase deficiency
CUBN	Cubilin
CV	Chorionic villi
CVD	Cardiovascular disease
CyD	Cysteine dioxygenase
CYP11A1	P450 side-chain cleavage
CYP11B1	P450 11 beta-hydroxylase type 1
CYP11B1/B2	11-beta-hydroxylase I/II
CYP11B2	Corticosterone methyl oxidase
CYP17A1	Cytochrome P450 17 alpha-hydroxylase
CYP19A1	P450 aromatase
CYP21A2	P450 21-hydroxylase
CYP27A1	Sterol 27-hydroxylase
CYP51	Lanosterol demethylase deficiency
CYP7A1	Cholesterol 7α-hydroxylase
CYP7B1	Oxysterol 7α-hydroxylase
CySD	Cysteine sulfinate decarboxylase
CYSTA	Cystathioninuria
D2HG	D-2-hydroxyglutaric acid
D2HGA	D-2-hydroxyglutaric aciduria
D2HGA I	D-2-hydroxyglutaric aciduria type I
D2HGA II	D-2-hydroxyglutaric aciduria type II
D2HGDH	D-2-hydroxyglutarate dehydrogenase
D4A	Androstenedione
DA	Dicarboxylic aminoaciduria
DAT	Dopamine transporter
DBH	Dopamine beta-hydroxylase
DBL	Dysbetalipoproteinemia
DBP	D-bifunctional protein deficiency
DBS	Dried blood spot
DCT	Distal convoluted tubule
DDC	Aromatic L-amino acid decarboxylase gene
dGUOK	Deoxyguanosine kinase

DHCA	Dihydroxycholestanoic acid
DHCR14	3β-hydroxysteroid-delta14-reductase
DHCR24	3β-hydroxysteroid-delta24-reductase; desmosterolosis
DHCR7	7-dehydrocholesterol reductase
DHEA	Dehydroepiandrosterone
DHF	Dihydrofolate
DHFR	Dihydrofolate reductase
DHO	Dihydroorotase
DHODH	Dihydroorotate dehydrogenase
DHP	Dihydropyrimidinase
DHPR	Dihydropteridine reductase deficiency
DHT	Dihydrotestosterone
DK1	Dolichol kinase
DK1-CDG	Dolichol kinase deficiency
DLD	Dihydrolipoyl dehydrogenase
DLP1	Dynamin-like protein 1
DMGDH	Dimethylglycine dehydrogenase
DMGLY	Dimethylglycinuria
DNPH	2,4-dinitrophenylhydrazine
DOC	Deoxycorticosterone
DOPS	Dihydroxyphenylserine
DPAGT1	UDP-GlcNAc:Dol-P-GlcNac-P transferase
DPD	Dihydropyrimidine dehydrogenase
DPM1	GDP-Man:Dol-P mannosyltransferase subunit 1
DPM3	GDP-Man:Dol-P mannosyltransferase 3
DPYD	Dihydropyrimidine dehydrogenase
DPYS	Dihydropyrimidinase
DRD	Dopa-responsive dystonia
DS	Dermatan sulfate
DSD	Defects of salt-water homeostasis and sexual development
dTMP	Deoxythymidine monophosphate
dUMP	Deoxyuridine monophosphate
DWI	Diffusion-weighted images
E	Epinephrine
E2	Estradiol
E3BP	Pyruvate dehydrogenase complex deficiency E3 X
EA6	Episodic ataxia due to EAAT1 glutamate transporter defect
EAAT3	Excitatory amino acid transporter 3
EBP	3β-hydroxysteroid-delta8, delta7-isomerase
EE	Ethylmalonic encephalopathy
EFEMP2	Fibulin 4
EIEE3	Early infantile epileptic encephalopathy 3; neonatal myoclonic epilepsy due to mitochondrial glutamate carrier GC1 defect
EIHI	Exercise-induced hyperinsulinism
ELN	Elastin
EMA	Ethylmalonic acids
ENO3	Beta-enolase
EPP	Erythropoietic protoporphyria
ER	Endoplasmic reticulum
ERT	Enzyme replacement therapy
ESR1	Estrogen resistance
ESRF	End stage renal failure
ETF	Multiple acyl-CoA dehydrogenase deficiency

ETF	Electron transfer flavoprotein
ETFA	Electron transfer flavoprotein A
ETFB	Electron transfer flavoprotein B
ETFDH	ETF-ubiquinone oxidoreductase, ETF dehydrogenase; electron transfer flavoprotein dehydrogenase; myopathic form of CoQ10 deficiency
ETF-DH	Electron transfer flavoprotein dehydrogenase
ETF-DH	Multiple acyl-CoA dehydrogenase deficiency DH
ETHE1	Ethylmalonic encephalopathy
EXT1	Exostosin 1
EXT2	Exostosin 2
F	Cortisol
FA	Fumaric acid
FAD	Flavin adenine dinucleotide
FAH	Fumarylacetoacetase
FAICAR	Formyl-5′-phosphoribosyl-5-aminoimidazole-4-carboxamide
FAO	Fatty acid oxidation
FAODs	Fatty acid oxidation disorders
FAOs	Fatty acids oxidation disorders
FB	Fibroblasts
FBP1	Fructose-1,6-bisphosphatase
FBS	Fanconi-Bickel syndrome
FCH	Familial combined hypolipidemia (ANGPTL3)
FECH	Ferrochelatase
FED	Familial LCAT deficiency (partial)
FEV	Forced expiratory volume
FFA	Free fatty acids
FGAR	Formyl-5′-phosphoribosylglycinamide
FGE	Cα-formylglycine-generating enzyme
FH1	Fumarase deficiency
FID	Flame-ionization detector
FIGLU	Glutamate formiminotransferase
FITHFCH	Formimino-THF cyclodeaminase
FK-D	Essential fructosuria; fructokinase deficiency
FMN	Flavin mononucleotide
FMO3	Flavin-containing monooxygenase
FOLR1	Folate receptor alpha
FP	False-positive
FRα	Folate receptor alpha
FSH	Follicle-stimulating hormone
FTCD	Formiminotransferase
FTHFDH	Formyl-THF dehydrogenase
FTHFI	Formyl-THF isomerase
FTHFS	10-formyl-THF synthase
FUCA1	Alpha-L-fucosidase
FUCO	Fucosidosis
FUM	Fumarase
G6PC	Glucose-6-phosphatase
G6PT1 (SLC37A4)	Glucose-6-phosphate translocase
GA	Glutaric acid
GA3	Glutaric aciduria type 3
GAA	Guanidinoacetate
GABAT	GABA transaminase deficiency

GAGs	Glycosaminoglycans
GA-I	Glutaric aciduria type I
GALC	Galactocerebrosidase
GALE	Uridine diphosphate galactose-4-epimerase
GALE	Galactose-1-phosphate uridyltransferase
GALK	Galactokinase
GALK-D	Galactokinase deficiency
GALNS	N-acetylgalactosamine-6-sulfatase
GALNT3	Polypeptide N-acetylgalactosaminyltransferase 3
GALT	Galactose-1-phosphate uridyltransferase
GALT-D	Galactosemia
GAMT	Guanidinoacetate methyltransferase; arginine:glycine amidinotransferase
GAR	5′-phosphoribosylglycinamide
GARTF	5′-phosphoribosylglycinamide transformylase
GBA	Glucocerebrosidase
GBE1	Glycogen branching enzyme
GCCR	Glucocorticoid resistance
GCDH	Glutaryl-CoA dehydrogenase
GCH	Global cerebral hypomyelination
GCH1	GTP cyclohydrolase I gene
GCK	Glucokinase (hexokinase-4)
GCLC	Gamma-glutamylcysteine synthetase
GCMS	Gas chromatography/mass spectrometry
GCS	γ-Glutamylcysteine synthetase
GCS1	Glucosidase 1
GDH	Glutamate dehydrogenase
GEPH	Gephyrin
GGCS	Gamma-glutamylcysteine synthetase deficiency
GGCT	γ-Glutamyl cyclotransferase
GGM	Intestinal glucose-galactose malabsorption
GGT1	Gamma-glutamyl transpeptidase
GGUOK	Mitochondrial deoxyguanosine kinase
GIF	Transcobalamin III
GK	Glycerol kinase
GKD	Glycerol kinase deficiency, isolated
GLA	Alpha-galactosidase
GLB1	Beta-galactosidase
GlcNAc	N-acetylglucosamine conjugate
GLD	Krabbe disease
GLDC; AMT; GCSH	P protein; T protein; H protein
Gluc	Glucuronide
GLUD1	Glutamate dehydrogenase-1
GLUL	Glutamine synthetase
GLUT1	Glucose transporter-1
GLUT10	Glucose transporter-10
GLUT1-D	Glucose transporter-1 deficiency
GLUT2	Glucose transporter-2
GLYCTK	Glycerate kinase
GLYCTK-D	Glycerate kinase deficiency
GLYT2	Neuronal glycine transporter
GMAP210	Golgi-microtubule-associated protein
GNE	UDP-GlcNAc epimerase/kinase

GNMT	Glycine N-methyltransferase
GNPAT	Dihydroxyacetone phosphate acyltransferase
GNPTAB	Alpha-/beta-subunit of N-acetylglucosamine-1-phosphotransferase
GNPTG	Gamma-subunit of N-acetylglucosamine-1-phosphotransferase
GNS	N-acetylglucosamine-6-sulfatase
GORAB	SCYL1 binding protein
GRA	Glucocorticoid suppressible hyperaldosteronism
GRACILE	GRACILE syndrome
GRHPR	D-glycerate dehydrogenase
GRHPR	D-glycerate dehydrogenase and hydroxypyruvate reductase
GRN	Progranulin
GS	Glutathione synthetase
GSD	Glycogen storage disorder
GSD-0a	Glycogen storage disease type 0 a
GSD-0b	Glycogen storage disease type 0 b
GSD-I non-a	Glycogen storage disease type I non-a
GSD-Ia	Glycogen storage disease type I a
GSD-IIa	Glycogen storage disease type II a
GSD-IIb	Glycogen storage disease type II b
GSD-III	Glycogen storage disease type III
GSD-IV	Glycogen storage disease type IV
GSD-IXa-c	Glycogen storage disease type IX a-c
GSD-IXd	Glycogen storage disease type IX d
GSD-V	Glycogen storage disease type V
GSD-VI	Glycogen storage disease type VI
GSD-VII	Glycogen storage disease type VII
GSD-X	Glycogen storage disease type X
GSD-XIII	Glycogen storage disease type XIII
GSD-XIV	Glycogen storage disease type XIV
GSD-XV	Glycogen storage disease type XV
GSH	Glutathione
GSL	Galactosialidosis
GSS	Glutathione synthetase
GUSB	Beta-glucuronidase
GYG1	Glycogenin-1
GYS1	Muscle glycogen synthase
GYS2	Liver glycogen synthase
HADH	Short-chain L-3-hydroxyacyl-CoA dehydrogenase
HADH	Hydroxyacyl-Coenzyme A dehydrogenase
HADHA	Long-chain 3 hydroxyacyl-CoA dehydrogenase
HADHB	3-oxothiolase; long-chain 3-ketoacyl-CoA thiolase
HAL	Histidase
HAMP	Hepcidin
HAT	Heteromeric amino acid transporters
HAWK	Hawkinsinuria
HBL	Hypobetalipoproteinemia (APOB)
HC	Hereditary coproporphyria
HC	Haptocorrin; transcobalamin 1
HCD	Haptocorrin deficiency
HCHOLA2	Familial defective apolipoprotein B (APOB)
HCHOLA3	Autosomal dominant hypercholesterolemia
HCP	Hereditary coproporphyria
HCSD	Holocarboxylase synthetase deficiency

hCys	Homocysteine
HD	Hartnup disorder
HD	Hemodialysis
HDL	High-density lipoprotein
HE	Hyperekplexia due to Gly transporter GLYT2 defect
HeFH	Familial hypercholesterolemia heterozygous (LDLR)
HEMD	Greenberg skeletal dysplasia
HEXA	Alpha-subunit of hexosaminidase
HEXA	β-Hexosaminidase A subunit
HEXB	Beta-subunit of hexosaminidase
HF	Hemofiltration
HFE	Hereditary hemochromatosis
HFE2A	Hereditary hemochromatosis (type 2a)
HFE2B	Hereditary hemochromatosis (type 2b)
HFE3	Hereditary hemochromatosis (type 3)
HFI	Hereditary fructose intolerance
HFM	Hereditary folate malabsorption
HGD	Homogentisate 1,2-dioxygenase
HGPRT	Hypoxanthine guanine phosphoribosyltransferase
HHF2	Hyperinsulinism
HHH	HHH syndrome
HI	Hyperinsulinism
HI/HA	Hyperinsulinism and hyperammonemia
HIBCH	3-hydroxyisobutyryl-CoA deacylase
HIDS	Hyper Ig D syndrome
HJV	Hemojuvelin
HJV	Hemojuvelin
HLCS	Holocarboxylase synthetase
HLP	Hyperlipoproteinemia
HLP1	Lipoprotein lipase deficiency (LPL)
HMBS	Hydroxymethylbilane synthase
HMGA	3-hydroxy-3-methylglutaryl
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase
HMG-CoA	3-hydroxy-3-methylglutaryl-CoA
HMG-CoAL	3-hydroxy-3-methylglutaryl-CoA lyase
HMG-CoAS	3-hydroxy-3-methylglutaryl-CoA synthase
HMGCS2	3-hydroxy-3-methylglutaryl-CoA synthase 2
HMGL	3-hydroxy-3-methylglutaryl-coenzyme A lyase
HML	Hereditary myopathy with lactic acidosis
HNF1A	Hepatocyte nuclear factor 1 alpha
HNF4A	Hepatocyte nuclear factor 4 alpha
HoFH	Familial hypercholesterolemia homozygous
HOGA1	4-hydroxy-2-oxoglutarate aldolase
HOPS	Congenital hypophosphatasia
HOT	Hydroxyacid:oxoacid transhydrogenase
HP II	Hyperprolinemia type II
HP1	Hyperprolinemia type I
HPA	Hyperphenylalaninemia
HPD	4-hydroxyphenylpyruvate hydroxylase
HPRT	Hypoxanthine guanine phosphoribosyltransferase
HRSA	Health Resources and Service Administration
HS	Heparan sulfate
HSCT	Hematopoietic stem cell transplantation

HSD10	2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency
HSD10	17beta-hydroxysteroid dehydrogenase type 10
HSD11B1/H6PDH	11beta-hydrosteroid dehydrogenase type 1
HSD11B2	11beta-hydroxysteroid dehydrogenase type 2
HSD17B10	17beta-hydroxysteroid dehydrogenase type 10
HSD17B3	17beta-hydroxysteroid dehydrogenase type 3
HSD17B4	D-bifunctional protein
HSD3B2	3β-hydroxysteroid dehydrogenase type II
HSD3B7	3β-hydroxy-Δ5-C27-steroid dehydrogenase/isomerase
HSP	Hereditary spastic paraplegia
HTG	Hypertriglyceridemia
hTHTR1	Human thiamine transporter 1
hTHTR2	Human thiamine transporter 2
HTOx	Hypotaurine:NAD; oxidoreductase
HVA	Homovanillic acid
HYAL1	Hyaluronidase
HYPO	Hydroxyprolinemia
IBD	Isobutyryl-CoA dehydrogenase deficiency
IBDH	Isobutyryl-CoA dehydrogenase
ICD	Implanted cardiac defibrillator
IDH2	Isocitrate dehydrogenase 2
IDL	Intermediate-density lipoprotein
IDS	Iduronate 2-sulfatase
IDUA	Alpha-iduroanidase
IFD	Intrinsic factor deficiency
IG	Iminoglycinuria
IGFBP-1	Insulin growth factor binding protein 1
IGS	Cubilin deficiency
IGS	Amnionless deficiency
IGS	Imerslund-Gräsbeck syndrome
IMM	Inner mitochondrial membrane
IMPDH	Inosine-5′-monophosphate dehydrogenase
IMPDH1	Inosine monophosphate dehydrogenase
INCL	Infantile neuronal ceroid lipofuscinoses
IP	Intestinal alkaline phosphatases
IRD	Infantile Refsum disease
ITPA	Inosine-5′-triphosphate pyrophosphohydrolase
IVA	Isovaleric acidemia
JNCL	Juvenile neuronal ceroid lipofuscinoses
KAA	2-ketoadipic and 2-aminoadipic academia
KCNJ11	Kir6.2 subunit of the inwardly rectifying potassium channel
KCTD7	Potassium channel tetramerization domain-containing protein 7
KHK	Fructokinase
KS	Keratan sulfate
KSS	Kearns-Sayre syndrome
KYN	Hydroxykynureninuria
KYNU	Kynureninase
L2HG	L-2-hydroxyglutaric acid
L2HGA	L-2-hydroxyglutaric aciduria
L2HGDH	L-2-hydroxyglutarate dehydrogenase
L2HGDH	L-2-hydroxyglutaric acid dehydrogenase
LA	Lactic acid
LAD2	GDP-fucose transporter deficiency

LAMAN	Mannosidosis
LAMM	Labyrinthine aplasia, microtia and microdontia
LAMP2	Lysosome-associated membrane protein-2
LBMAN	Beta-mannosidosis
LBSL	Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation
LCAT	Lecithin cholesterol acyl transferase
LCHAD	Isolated deficiency of long-chain 3-hydroxyacyl-CoA dehydrogenase
LDH	Lactate dehydrogenase
LDHA-D	Glycogen storage disease type XI
LDL	Low-density lipoprotein
LDLR	Low-density lipoprotein receptor
LDLRAP1	Low-density lipoprotein receptor associated protein 1
LFNG	O-fucose-specific beta-1,3-N-acetylglucosaminyltransferase
LH	Luteinizing hormone
LHON	Leber Hereditary Optic Neuropathy, LHON
LIMM	Lethal Infantile Mitochondrial Myopathy
LINCL	Late-infantile neuronal ceroid lipofuscinoses
LIPA	Acid lipase
LIPC	Hepatic triglyceride lipase
LKAT	Long-chain 3 ketoacyl-CoA thiolase
LLO	Lipid-linked oligosaccharides
LMBRD1	Lysosomal export of cobalamin?
LN11	Ceroid lipofuscinosis, neuronal, 11
LPA	Apolipoprotein(a) moiety of Lp(a)
LPI	Lysinuric protein intolerance
LPL	Lipoprotein lipase
LS	Leigh syndrome
LSFC	Leigh syndrome with French-Canadian Ethnicity
LT	Leukotriene
LTA4H	Leukotriene A4 hydrolase
LTC4D	Cysteinyl leukotriene synthase 4 deficiency
LTC4S	Cysteinyl leukotriene synthase 4
MA	Malonic aciduria
MADD	Multiple acyl-CoA dehydrogenation defect
MAGT1	Magnesium transporter 1
MAN2B1	Alpha-mannosidase B
MANBA	Beta-mannosidase
MAO	Monoamine oxidase
MAOA	Monoamine oxidase A
MAT	Methionine S-adenosyltransferase
MAT	Methylacetoacetyl-CoA thiolase; β-ketothiolase
MAT I/III	Methionine adenosyltransferase I/III deficiency
MBCD	2-methylbutyrylglycinuria
MBD	2-methylbutyryl-CoA dehydrogenase
MBDD	Membrane-bound dipeptidase deficiency
MCAD	Medium-chain acyl-CoA dehydrogenase
MCCC1 or 2	Methylcrotonyl-CoA carboxylase
MCCD	3-methylcrotonyl-CoA carboxylase deficiency
MCD	Multiple carboxylase defect
MCEE	Methylmalonyl-CoA epimerase
MCM	Methylmalonyl-CoA mutase
MCSU	Molybdenum cofactor sulfurylase

MCT-1	Monocarboxylate transporter
MDH	Malate dehydrogenase
MDR3	Multidrug resistance protein 3
MeCbl	Methylcobalamin
MEGDEL	MEGDEL syndrome
MELAS	Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes
MEN1	Multiple endocrine neoplasia syndrome type 1
MERRF	Myoclonic epilepsy associated with ragged red fibers
MET	Metanephrine
MFSD8	Major facilitator superfamily domain-containing protein-8 (MFSD8)
MGA	3-methylglutaconic aciduria
MGA1	Methylglutaconic aciduria type I
MGA2	Barth syndrome gene
MGA3	Costeff syndrome gene
MGA4	Methylglutaconic aciduria type IV
MGAT2	N-acetylglucosaminyltransferase 2
MGAT2-CDG	N-acetylglucosaminyltransferase 2 deficiency-CDG-IIa
MHBD	2-methyl-3-hydroxybutyryl-CoA dehydrogenase
mHMGS	Mitochondrial 3-hydroxy-3-methylglutaryl-coenzyme A synthase
MHPG	3-methoxy-4-hydroxyphenylglycol
MIDD	Maternally inherited deafness and diabetes
MKD	Mevalonate kinase deficiency
MLASA	Myopathy Lactic Acidosis and Sideroblastic Anemia
MLD	Metachromatic leukodystrophy
MLYCD	Malonyl-CoA decarboxylase
MMA	Methylmalonic acidemia
MMA/MA	Combined MMA and MA
MMAB	Adenosyltransferase
MMAE	Methylmalonyl-CoA epimerase deficiency
MMSDH	Methylmalonate semialdehyde dehydrogenase deficiency
MNGIE	Thymidine phosphorylase deficiency
MNK (MK)	Menkes disease
MoCD	Molybdenum cofactor deficiency
MOCD-A	Molybdenum cofactor deficiency A
MOCD-B	Molybdenum cofactor deficiency B
MOCD-C	Molybdenum cofactor deficiency C
MPDU1	Dol-P-Man utilization 1
MPI	Phosphomannose isomerase
MPI-CDG	Phosphomannose isomerase deficiency-CDG-Ib
MPS I	Hurler, Scheie disease
MPS II	Hunter disease
MPS IIIA	Sanfilippo A disease
MPS IIIB	Sanfilippo B disease
MPS IIIC	Sanfilippo C disease
MPS IIID	Sanfilippo D disease
MPS IVA	Morquio A disease
MPS IVB	Morquio B disease
MPS IX	Hyaluronidase deficiency
MPS VI	Maroteaux-Lamy disease
MPS VII	Sly disease
MPS3C	Acetyl-CoA alpha-glucosaminide acetyltransferase
MPST	3-mercaptopyruvate sulfurtransferase

MRI	Magnetic resonance imaging
MSD	Multiple sulfatase deficiency
MSD	Multiple sulfatase deficiency
MSPT	3-mercaptopyruvate sulfurtransferase
MSUD	Maple syrup urine disease
MT	Methyltransferase
mtDNA	Mitochondrial DNA
MTDP8A	Mitochondrial ribonucleotide reductase subunit 2 deficiency
MTDP8B	Mitochondrial ribonucleotide reductase subunit 2 deficiency
MTDPS1	Mitochondrial depletion syndrome 1
MTDPS2	Thymidine kinase 2 deficiency
MTDPS2	Mitochondrial depletion syndrome 2
MTDPS3	Deoxyguanosine kinase deficiency
MTDPS3	Mitochondrial depletion syndrome 3
MTDPS4A	Mitochondrial depletion syndrome 4A
MTDPS5	Mitochondrial depletion syndrome 5
MTDPS8A	Mitochondrial depletion syndrome 8A
MTDPS9	Mitochondrial depletion syndrome 9
mTFP	Mitochondrial trifunctional protein complex
mTFP, MTP	Mitochondrial trifunctional protein deficiency
MTHF	5-methyltetrahydrofolate
MTHFCH	Methenyl-THF cyclohydrolase
MTHFD1	5,10-Methylenetetrahydrofolate dehydrogenase/5,10-ethenyltetrahydrofolate cyclohydrolase/formyltetrahydrofolate synthetase
MTHFR	5,10-methylenetetrahydrofolate reductase
MTHFS	5,10-methenyltetrahydrofolate synthetase
MTP	Microsomal triglyceride transfer protein
MTP	Mitochondrial trifunctional protein
MTR	Methionine synthase, 5-methyltetrahydrofolate-homocysteine methyltransferase
MTRR	Methionine synthase reductase
MTS	Mohr-Tranebjaerg syndrome
MUT	Methylmalonyl-CoA mutase
MVK	Mevalonate kinase
N/A	Apolipoprotein A-I deficiency (APOA1)
NAA	N-acetylaspartic acid
NAGA	Schindler disease type I
NAGA	Kanzaki disease
NAGA	Schindler disease type III
NAGA	α-N-acetylgalactosaminidase
NAGS	N-acetylglutamate synthase
NAGU	N-acetyl-α-D-glucosaminidase
NALD	Neonatal adrenoleukodystrophy
NARP	Neuropathy ataxia and retinitis pigmentosa
NBIA1	Neurodegeneration with brain iron accumulation 1
NBS	Newborn screening
NCL	Neuronal ceroid lipofuscinoses
nDNA	Nuclear DNA
NE	Norepinephrine
NEFA	Nonesterified fatty acids
Neo	Neopterin
NEU	Sialidosis
NEU1	Alpha-neuraminidase

NH	Neonatal hemochromatosis
NH2TP	Dihydroneopterin triphosphate
NKH	Nonketotic hyperglycinemia
NME	Neonatal mitochondrial encephalocardiomyopathy
NMN	Normetanephrine
NMR	Nuclear magnetic resonance
NPC1	Niemann-Pick disease type C1
NPC1	Niemann-Pick type C
NPC2	Niemann-Pick disease type C2
NR3C1	Glucocorticoid receptor
NR3C2	Mineralocorticoid receptor
NSDHL	3-Beta-hydroxysteroid dehydrogenase
OAs	Organic acidurias
OAT	Ornithine aminotransferase
OAT	Ornithine aminotransferase deficiency
OAT	Ornithine aminotransferase
OAT	Ornithine aminotransferase
OCTN2	Organic cation carnitine transporter 2
OGDH	2-ketoglutarate dehydrogenase
OGDH	2-oxoglutarate dehydrogenase
OHS	Occipital horn syndrome
OMIM	Online Mendelian Inheritance in Man
OMPDC	OMP decarboxylase
OPA1	Childhood-onset autosomal dominant optic atrophy
OPLAH	Oxoprolinase
OPRT	Orotate phosphoribosyltransferase
ORNT1	HHH syndrome gene
ORNT1	Mitochondrial ornithine transporter
OTC	Ornithine transcarbamylase
OXCT1	Succinyl-CoA:3-oxoacid-CoA transferase
OXPHOS	Oxidative phosphorylation system
P450c17	17-α-hydroxylase gene
P450scc	Cholesterol side-chain cleavage
P5CDH	Hyperprolinemia type II
P5CS	Pyrroline-5-carboxylate synthetase
P6C	L-Δ^1-piperideine-6-carboxylate
PA	Propionic acidemia
PA	Propionic acidemia
PAH	Phenylalanine hydroxylase
PANK2	Pantothenate kinase
PAP	Pulmonary alveolar proteinosis
PBD	Peroxisome biogenesis disorders
PBG	Porphobilinogen
PC	Pyruvate carboxylase
PC	Phosphatidylcholine
PCBD	Pterin carbinolamine-4a-dehydratase gene
PCC	Propionyl-CoA carboxylase
PCCA or B	Propionyl-CoA-carboxylase deficiency
PCD	Pterin carbinolamine-4a-dehydratase deficiency
PCD	Pyruvate carboxylase deficiency
PCSK9	Proprotein convertase subtilisin/kexin type 9
PCSK9D	PCSK9 deficiency with low LDL
PCT	Proximal convoluted tubule

PCT	Porphyria cutanea tarda
PDE	Pyridoxine-dependent epilepsy
PDH	Pyruvate dehydrogenase complex
PDHA1	Pyruvate dehydrogenase E1α subunit
PDHB	Pyruvate dehydrogenase E1β subunit
PDHC E2	Pyruvate dehydrogenase complex deficiency E2
PDHC E3	Pyruvate dehydrogenase complex deficiency E3
PDHDD	Dihydrolipoyl transacetylase
PDHPD	Pyruvate dehydrogenase complex deficiency PDHP
PDHX	Pyruvate dehydrogenase E3 binding protein
PDP1	Pyruvate dehydrogenase phosphatase
PDSS1	Prenyl diphosphate synthase
PDSS2	Prenyl diphosphate synthase
PEO	Progressive external ophthalmoplegia
PEPD	Peptidase D
PEX	Different proteins
PFIC	Progressive familial intrahepatic cholestasis
PFIC 2	ABCB11 deficiency
PFIC1	ATP8B1 deficiency
PFKM	Phosphofructokinase
PGAM2	Phosphoglycerate mutase-2
PGK1	Phosphoglycerate kinase
PGR	Progesterone resistance
PH	Primary hyperoxaluria
PH 1	Primary hyperoxaluria type I
PH 2	Primary hyperoxaluria type II
PH 3	Primary hyperoxaluria type III
PHA1	Pseudohypoaldosteronism
PHGDH	Phosphoglycerate dehydrogenase
PHHI	Persistent hyperinsulinemic hypoglycemia of infancy
PHKA1	Muscle phosphorylase kinase
PHKA2	Liver phosphorylase kinase
PHYH	Phytanoyl-CoA hydroxylase
PIGM	Phosphatidylinositol glycan, class M
PIGV	Phosphatidylinositol glycan, class V
PK	Phosphate kinase
PKU	Phenylketonuria
PL	Phospholipid
PLP	Pyridoxal 5′-phosphate
PMM2	Phosphomannomutase 2
PNP	Purine nucleoside phosphorylase deficiency
PNP	Purine nucleoside phosphorylase
PNP	Purine nucleoside phosphorylase
PNPO	Pyridox(am)ine 5′-phosphate oxidase
POADS	Dihydroorotate dehydrogenase deficiency
POLG	Polymerase gamma
POMGNT1	O-mannose beta-1,2-N-acetyglucosaminyltransferase
POMT1	O-mannosyltransferase 1
POMT2	O-mannosyltransferase 2
POR	P450 oxidoreductase
POX	Hydroxyproline oxidase
PPOX	Protoporphyrinogen oxidase
PPP	Pentose phosphate pathway

PPT1	Lysosomal palmitoyl protein thioesterase-1
Pri	Primapterin
PRKAG2	AMP-activated protein kinase
PRODH	Proline dehydrogenase
PRPPS	Phosphoribosyl pyrophosphate synthetase
PRPS1	Phosphoribosyl pyrophosphate synthetase 1
PSAP	Saposin A-D
PSAPD	Combined saposin deficiency
PSAT	Phosphoserine aminotransferase deficiency
PSAT1	Phosphoserine aminotransferase
PSPH	Phosphoserine phosphatase
PSPHD	Phosphoserine phosphatase deficiency
PST	Proximal straight tubule
PTP	6-pyruvoy-tetrahydroptrin
PTPS	6-pyruvoyl-tetrahydropterin synthase
PTS	6-pyruvoyl-tetrahydropterin synthase gene
PTS	Peroxisome targeting signals
PV	Porphyria variegata
PYCS	Pyrroline-5-carboxylate synthase
PYGL	Liver glycogen phosphorylase
PYGM	Muscle glycogen phosphorylase
q-BH2	Quinoid-dihydrobiopterin
QDPR	Dihydropteridine reductase gene
RBC	Red blood cells
RCDP	Rhizomelic chondrodysplasia punctata
RCDP1	Rhizomelic chondrodysplasia punctata type 1
RCDP2	Rhizomelic chondrodysplasia punctata type 2
RCDP3	Rhizomelic chondrodysplasia punctata type 3
RFC1	Reduced folate carrier 1
RFT1	Flippase of Man5GlcNAc2-PP-Dol
RFT1-CDG	Flippase of Man5GlcNAc2-PP-Dol deficiency-CDG-In
RPI	Ribose-5-phosphate isomerase
RPIA	Ribose-5-phosphate isomerase deficiency
RR	Ribonucleotide reductase
RRM2B	Mitochondrial ribonucleotide reductase
SA	Succinic acid
SAHH	S-adenosylhomocysteine hydrolase
SAM	S-adenosylmethionine
SANDO	Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis
SARDH	Sarcosine dehydrogenase
SBCAD	2-methylbutyrylglycinuria, benign
SBCAD	Short-/branched-chain acyl-CoA dehydrogenase
SC4MOL	Sterol C4-methyloxidase
SC5D	Lathosterolosis; sterol C5-desaturase
SCAD	Short-chain acyl-CoA dehydrogenase deficiency
SCARB1	Scavenger receptor B1
SCARB2/LIMP-2	Scavenger receptor class B, member 2/Limp-2
SCHAD	Short-chain 3-hydroxyacyl-CoA dehydrogenase
SCOT	Succinyl-CoA:3-oxoacid-CoA transferase
SCOX	Straight-chain acyl-CoA oxidase
SCP	Sterol carrier protein
SCS	Succinyl-CoA synthetase
SDD	Sarcosinemia

SDH	Hyperlysinemia and saccharopinuria
SDO	Sulfur dioxygenase
SEC23B	COPII component SEC23B
SEC23B-CDG, CDA II	COPII component SEC23B deficiency
SERC1	MEGDEL
SGLT	Sodium-dependent glucose transporter
SGLT2-D	Renal glucosuria
SGSH	Heparan-N-sulfatase
SLC16A1	Monocarboxylate transporter gain-of-function
SLC16A1	Monocarboxylate transporter 1
SLC17A5	Salla disease
SLC17A5	Sialin
SLC18A2	Vesicular monoamine transporter 2
SLC19A2	THTR1 transporter
SLC19A3	Wernicke-like encephalopathy and BRBG
SLC19A3	Reduced folate family of micronutrient transporter
SLC1A1	Neuronal/epithelial high affinity glutamate transporter, excitatory amino acid transporter 3
SLC1A3	Glutamate/aspartate transporter (GLAST), excitatory amino acid transporter 3
SLC22A5	Organic cation carnitine transporter 2
SLC25A12	Neuronal- and muscle-specific mitochondrial aspartate/glutamate transporter 1
SLC25A13	Aspartate glutamate carrier
SLC25A19	Bilateral striatal necrosis
SLC25A19	Amish microcephaly
SLC25A20	Carnitine acylcarnitine translocase
SLC25A22	Mitochondrial glutamate/H + symporter 1
SLC27A5	Bile acid-CoA ligase
SLC2A1	Glucose transporter-1
SLC2A10	Glucose transporter-10
SLC2A2	Glucose transporter-2
SLC35A1	CMP-sialic acid transporter
SLC35A1-CDG	CMP-sialic acid transporter deficiency
SLC35C1	GDP-fucose transporter
SLC35D1	UDP-glucuronic acid/UDP-N-acetylgalactosamine dual transporter
SLC35D1-CDG	UDP-glucuronic acid/UDP-N-acetylgalactosamine dual transporter deficiency
SLC36A2	Amino acid transporter
SLC3A1	Amino acid transport system
SLC46A1	SLC46A1 transporter
SLC52A3	Riboflavin transporter 2
SLC6A19	Sodium-dependent neutral amino acid transporter
SLC6A5	Neuronal glycine transporter GLYT2
SLC6A8	CrT, creatine transporter
SLC7A7	Amino acid transport system (LAT1)
SLOS	Smith-Lemli-Opitz syndrome
SMAX3	X-linked distal spinal muscular atrophy
SMPD1	Acid sphingomyelinase (ASM)
SO	Sulfite oxidase
SOX	Sulfite oxidase
SPR	Sepiapterin reductase gene
SQR	Sulfide CoQ reductase

SR	Sepiapterin reductase
SRB1D	Scavenger receptor B1 deficiency (SCARB1)
SRD5A2	5-alpha-reductase type II
SRD5A3	Steroid 5 alpha-reductase 3
SRD5B1	Δ4-3-Oxosteroid-5β-reductase
SRT	Substrate reduction therapy
SSADH	Succinic semialdehyde dehydrogenase
SSC	S-sulfocysteine
ST	Sulfurtransferase
ST3GAL5	Lactosylceramide alpha-2,3-sialyltransferase
ST3GAL5-CDG	Lactosylceramide alpha-2,3-sialyltransferase
StAR	Steroidogenic acute response protein
StAR	Lipoid adrenal hyperplasia gene
SUCLA2	Succinate-CoA ligase beta-subunit
SUCLG1	Succinate-CoA ligase alpha-subunit
SUMF1	Formylglycine-Generating Enzyme
SUOX	Sulfite oxidase
SUR1	Sulfonylurea receptor 1
TALDO	Transaldolase
TAT	Tyrosine aminotransferase
TC II	Transcobalamin II
TCD	Transcobalamin deficiency
TCN1	B12-binding alpha-globulin
TCN2	Vitamin B12-binding protein 2
TCR	Transcobalamin receptor defect
TD, FHA	Tangier disease (ABCA1)
TF	Transferrin, atransferrinemia
TFR2	Transferrin receptor 2
TG	Triglyceride
TH	Tyrosine-3-hydroxylase
THAN	Transient hyperammonemia of the newborn
THCA	Trihydroxycholestanoic acid
tHcy	Total homocysteine
THF	Tetrahydrofolate
THF	Tetrahydrofolate
ThTP	Thiamine triphosphate
THTR1	Thiamine-responsive megaloblastic anemia syndrome (SLC19A2)
TKS	Thymidine kinase 2
TLC	Thin-layer chromatography
TMA	Trimethylaminuria
TMA	Trimethylamine
TMAO	Trimethylamine N-oxide
TMEM70	Complex V assembly protein
TMS	Tandem mass spectrometry
TNSALP	Tissue nonspecific alkaline phosphatase
TP	Thymidine phosphorylase
TPK	Thiamine pyrophosphokinase
TPMT	Thiopurine S-methyltransferase
TPP	Thiamine pyrophosphate
TPP tr	Thiamine triphosphate transporter
TPP1	Lysosomal tripeptidyl-peptidase-1
TRMA	Thiamine-responsive megaloblastic anemia
TRPV1	Transient receptor potential channel vanilloid subfamily member 1

TS	Thymidylate synthase
TSD	Tay-Sachs disease
TUSC3	Oligosaccharyltransferase subunit tusc 3
TUSC3-CDG	Oligosaccharyltransferase subunit tusc 3 deficiency
TYMP	Thymidine phosphorylase
TYR1	Tyrosinemia type I
TYR2	Tyrosinemia type II
TYR3	Tyrosinemia type III
UCD	Urea cycle disorders
UCP	Uncoupling protein deficiency
UCP2	Uncoupling protein 2 deficiency
UCP2	Uncoupling protein 2
UCP2	Mitochondrial uncoupling protein 2
UMPH1	Pyrimidine-5′-nucleotidase I
UMPH1	Uridine-5′-monophosphate hydrolase
UMPS	Uridine monophosphate synthase
UP	ß-Ureidopropionase
UPB1	Beta-ureidopropionase
UPD	Uniparental disomy
UPLC	Ultra performance liquid chromatography
UROC1	Urocanase
UROD	Uroporphyrinogen decarboxylase
UROS	Uroporphyrinogen III synthase
USF1	Upstream stimulatory factor
VLA	Vanillactic acid
VLCAD	Very-long-chain acyl-CoA dehydrogenase
VLCFA	Very-long-chain fatty acids
VLCS	Very-long-chain acyl-CoA synthase
VLDL	Very low-density lipoprotein
VMA	Vanillylmandelic acid
VMAT2	Dopamine-serotonin vesicular transport
VP	Variegate porphyria
WHO	World Health Organization
WND (WD)	Wilson disease
X-ALD and AMN	X-linked adrenoleukodystrophy and adrenomyeloneuropathy
XDH	Xanthine dehydrogenase (oxidase)
XDH/AO	Combined xanthine oxidase and aldehyde oxidase
XLDPP	X-linked dominant protoporphyria
XLPP	X-linked protoporphyria
XLSA	X-linked sideroblastic anemia
X-MT	S-adenosylmethionine-dependent transmethylation reactions
ZSD	Zellweger spectrum disorders