

## Abbreviations

$\Delta 4A$	Androstenedione
AA	Amino acid
AADC	Aromatic L-amino acid decarboxylase
abn.	Abnormal
ACTH	Adrenocorticotrophic hormone
ADA	Adenosine deaminase
ADDP	$\delta$ -Aminolevulinic acid dehydratase-deficient porphyria
ADE	Adenine
ADP	Adenosine diphosphate
AE	Acrodermatitis enteropathica
AF	Amniotic fluid
AFC	Amniotic fluid cells (Amniocytes)
AGAT	Arginine:glycine amidinotransferase
AGT	Alanine:glyoxylate aminotransferase
AHC	Adrenal hypoplasia congenita
AICA	Aminoimidazole carboxamide
AIP	Acute intermittent porphyria
Ala	Alanine
5-ALA	$\delta$ -Aminolevulinic acid
ALAD	$\delta$ -Aminolevulinic acid dehydratase
ALAT	Alanine aminotransferase
ALD	Adrenoleukodystrophy
Aldo	Aldosterone
ALP	Alkaline phosphatase
a.m.	Before noon
AMPD	Adenosine monophosphate deaminase
AnA	Anthranilic acid
ANT	Adenine nucleotide translocator
apo	Apolipoprotein
APRT	Adenine phosphoribosyltransferase
Arg	Arginine
ARG	Argininemia
ASA	Argininosuccinic aciduria
ASAT	Aspartate aminotransferase

Asn	Asparagine
Asp	Aspartic acid
ATP	Adenosine triphosphate
ATPase	Adenosine triphosphatase
AV	Aortic valve
B	Blood
B/A	3-Hydroxybutyrate/acetoacetate
BAEP	Brainstem acoustic evoked potential
BCKD	Branched-chain $\alpha$ -ketoacid dehydrogenase
BH <sub>4</sub>	Tetrahydrobiopterin
Bio	Biopterin
Br	Brain
BUN	Blood urea nitrogen
bw	Body weight
CAC	Carnitine acylcarnitine carrier
CAD	Coronary artery disease
CAFC	Cultured amniotic fluid cells
CAH	Congenital adrenal hyperplasia
CAT	Computer-assisted tomography
cbl	Cobalamin
CBS	Cystathionine $\beta$ -synthase
ccv	Cultured chorionic villi
CDP	Cytidine diphosphate
CDPC	Cytidine diphosphate choline
CE	Capillary electrophoresis
CETP	Cholesteryl ester transfer protein
CFTR	Cystic fibrosis transmembrane regulator
Chol.	Cholesterol
CITR	Citrullinemia
CK	Creatine kinase
CMP	Cytidine monophosphate
CNS	Central nervous system
CoA	Coenzyme A
CPC	Cetylpyridinium chloride
CPS	Carbamylphosphate synthetase
CPT	Carnitine palmitoyltransferase
Creat	Creatinine
CRTR	Creatine transporter
CSF	Cerebrospinal fluid
CT	Computed tomography
CTH	$\gamma$ -Cystathionase
CTP	Cytidine triphosphate
CTX	Cerebrotendinous xanthomatosis
cv	Chorionic villi

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Cys	Cysteine
DA	Dopamine
dAdo	Deoxyadenosine
dATP	Deoxyadenosine triphosphate
dBs	Dried blood spot
D $\beta$ H	Dopamine $\beta$ -hydroxylase
dCDP	Deoxycytidine diphosphate
dCyK	Deoxycytidine kinase
DD	Developmental delay
def.	Deficiency
DGGE	Denaturing gradient gel electrophoresis
dGuo	Deoxyguanosine
dGTP	Deoxyguanosine triphosphate
DH	Dehydrogenase
2,8-DHA	2,8-Dihydroxyadenine
DHAP	Dihydroxyacetonephosphate
DHAPAT	Dihydroxyacetonephosphate acyltransferase
DHEA	Dehydroepiandrosterone
DHPA	Dihydropyrimidinase
DHPD	Dihydropyrimidine dehydrogenase
DHPR	Dihydropteridine reductase
DHT	Dihydrotestosterone
dIno	Deoxyinosine
DMB	Dimethylmethylene blue
DMD	Duchenne muscular dystrophy
DNA	Deoxyribonucleic acid
DNPH	Dinitrophenylhydrazine
DOC	Deoxycorticosterone
DOCA	Deoxycortisone acetate
DOPA	Dihydroxyphenylalanine
DOPAC	Dihydroxyphenylacetic acid
DOPS	Dihydroxyphenylserine
DRD	DOPA-responsive dystonia
DS	Dermatan sulfate
dUr	Deoxyuridine
E	Epinephrine
EC	Electrochemical
ECG	Electrocardiogram
EDTA	Ethylenediaminetetraacetic acid
EEG	Electroencephalogram
EMG	Electromyogram
ERG	Electroretinogram
ETF	Electron transfer flavoprotein
ETF-DH	Electron transfer flavoprotein dehydrogenase

F	Fluorescence
FA	Fumaric acid
FAAH	Fumarylacetoacetase
FAB	Fast atom bombardment
FADH <sub>2</sub>	Reduced flavin adenine dinucleotide
FB	Fibroblasts
FD	Familial dysbetalipoproteinemia
FDB	Familial defective apolipoprotein B-100
FFA	Free fatty acids
FH	Familial hypercholesterolemia
Figlu	Formiminoglutamic acid
FISH	Fluorescence in situ hybridization
Fru-1-P	Fructose-1-phosphate
FSH	Follicle stimulating hormone
5-FU	5-Fluorouracil
GAA	Guanidinoacetate
GABA	$\gamma$ -Aminobutyric acid
GAD	Glutamic acid decarboxylase
GAGs	Glycosaminoglycans
Gal	Galactose
Gal-1-P	Galactose-1-phosphate
GALK	Galactokinase
GALT	Galactose-1-phosphate uridyltransferase
GAMT	Guanidinoacetate methyltransferase
GC	Gas chromatography
GCMS	Gas chromatography/mass spectrometry
GCS	Glycine cleavage system
general.	Generalized
GFR	Glomerular filtration rate
GI	Gastrointestinal
GK	Glycerol kinase
Glc	Glucose
Gln	Glutamine
Glu	Glutamic acid
Gly	Glycine
G3P	Glycerol-3-phosphate
GSD	Glycogen storage disorders
$\gamma$ GT	$\gamma$ -Glutamyl transpeptidase
GT	GABA transaminase
GTP	Guanosine triphosphate
GTPCH	GTP cyclohydrolase 1
GTT	Glucose tolerance test
GUA	Guanine
Guo	Guanosine

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GVHD	Graft versus host disease
HA	Homogentisic acid
Haw	Hawkinsin
Hb	Hemoglobin
HCS	Holocarboxylase synthetase
HDL	High-density lipoproteins
HDL-C	High-density lipoprotein cholesterol
HFI	Hereditary fructose intolerance
HGA	Hydroxyglutaric acid (D-2, L-2, or 3-)
HHH	Hyperornithinemia-hyperammonemia-homocitrullinuria
5HIAA	5-Hydroxyindoleacetic acid
His	Histidine
HL	Hepatic lipase
HMG-CoA	3-Hydroxy-3-methylglutaryl-CoA
HOGA	Hyperornithinemia gyrate atrophy
HPLC	High-performance liquid chromatography
HPO	Hydroxyproline oxidase
HPRT	Hypoxanthine phosphoribosyltransferase
HS	Heparan sulfate
17 $\beta$ -HSD	17 $\beta$ -Hydroxysteroid dehydrogenase
3 $\beta$ -HSDH	3 $\beta$ -Hydroxy- $\Delta^5$ -C <sub>27</sub> -steroid dehydrogenase
5HT	Serotonin
5HTP	5-Hydroxytryptophan
HVA	Homovanillic acid
HYP	Hypoxanthine
IEF	Isoelectric focusing
Ile	Isoleucine
IM	Intramuscular(ly)
Ino	Inosine
IPyA	Imidazolepyruvic acid
IR	Infrared
IRD	Infantile Refsum disease
ISSD	Intralysosomal sialic acid storage disease
ITP	Idiopathic thrombocytopenic purpura
IV	Intravenous
K	Kidney
2-KGA	2-Ketoglutaric acid
2-KGD	2-Ketoglutarate dehydrogenase
KS	Keratan sulfate
KSS	Kearns-Sayre syndrome
KynA	Kynurenic acid
L	Liver
LCAD	Long-chain acyl-CoA dehydrogenase
LCAT	Lecithin:cholesterol acyltransferase

LCHAD	Long-chain 3-hydroxyacyl-CoA dehydrogenase
LCT	Long-chain triglyceride
LDH	Lactate dehydrogenase
LDL	Low-density lipoproteins
LDLR	Low-density lipoprotein receptor
Leu	Leucine
LH	Luteinizing hormone
LHON	Leber hereditary optic neuropathy
L/P	Lactate/pyruvate
LPI	Lysinuric protein intolerance
LPL	Lipoprotein lipase
LSIMS	Liquid secondary ion mass spectrometry
LYM	Lymphocytes
Lys	Lysine
M	Muscle
MA	Malonic acid
MAD	Multiple acyl-CoA dehydrogenation defects
MAO	Monoamine oxidase
MAT	Methionine adenosyltransferase
MCAD	Medium-chain acyl-CoA dehydrogenase
MCT	Medium-chain triglyceride
MD	Menkes disease
MELAS	Mitochondrial encephalopathy, lactic acidosis, stroke-like episodes
MERRF	Myoclonus epilepsy, ragged-red fibers
Met	Methionine
Met-Hb	Methemoglobin
MHPG	3-Methoxy-4-hydroxyphenylglycol
MI	Myocardial infarction
MITEM	Mitochondrial energy metabolism
MMA	Methylmalonic acid
MNGIE	Mitochondrial neurogastrointestinal encephalomyopathy
MPS	Mucopolysaccharidoses
MR	Mental retardation
MR/DD	Mental retardation/developmental delay
MRI	Magnetic resonance imaging
MRS	Magnetic resonance spectroscopy
MSUD	Maple syrup urine disease
3MT	3-Methoxytyramine
mtDNA	Mitochondrial DNA
MTHF	5-Methyltetrahydrofolate
MTHFR	5,10-Methylenetetrahydrofolate reductase
MTP	Microsomal transfer protein
MU	Methylene units

n	Normal
N/A	Not applicable
NADH	Reduced nicotinamide adenine dinucleotide
NAGA	$\alpha$ -N-acetylgalactosaminidase
NAGS	N-acetylglutamate synthase
NALD	Neonatal adrenal leukodystrophy
NARP	Neurogenic muscle weakness, ataxia, retinitis pigmentosa
NCV	Nerve conduction velocity
n.d.	Not detectable
NE	Norepinephrine
Neo	Neopterin
NKH	Nonketotic hyperglycinemia
NMN	Normetanephrine
NMR	Nuclear magnetic resonance
OAT	Ornithine aminotransferase
ODC	Orotidine monophosphate decarboxylase
3OHAnA	3-Hydroxyanthranilic acid
3OH-Kyn	3-Hydroxykynurenine
17OHP	17-Hydroxyprogesterone
OHS	Occipital horn syndrome
3OMD	3-O-methyldopa
OPRT	Orotate phosphoribosyltransferase
Orn	Ornithine
OTC	Ornithine transcarbamylase
P	Plasma
PAA	Plasma amino acids
PAH	Phenylalanine-4-hydroxylase
PBG	Porphobilinogen
PBGD	Porphobilinogen deaminase
P5C	Pyrroline-5-carboxylic acid
PCD	Pterin-4 $\alpha$ -carbinolamine dehydratase
P5CDH	Pyrroline-5-carboxylate dehydrogenase
PCR	Polymerase chain reaction
PCRE	Plasma creatinine
PEG	Polyethylene glycol
PEO	Progressive external ophthalmoplegia
PH	Primary hyperoxaluria
Phe	Phenylalanine
PHYT	Phytanic acid
PKU	Phenylketonuria
PLT	Platelets
p.m.	Afternoon
PNP	Purine nucleoside phosphorylase
p.o.	Orally

PO	Proline oxidase
pp	Postprandial
PPribP	Phosphoribosylpyrophosphate
PRA	Plasma renin activity
PRIS	Pristanic acid
Pro	Proline
Prot.	Protein
PRPP	Phosphoribosylpyrophosphate
PRPS	Phosphoribosylpyrophosphate synthetase
PTPS	6-Pyruvoyltetrahydropterin synthase
R-5-P	Ribose-5-phosphate
RBC	Red blood cells
RCDP	Rhizomelic chondrodysplasia punctata
RNA	Ribonucleic acid
r.p.m.	Revolutions/minute
rRNA	Ribosomal RNA
RT	Room temperature
S	Serum or Sepiapterin
SA	Succinylacetone
SAA	Succinylacetoacetate
SAdo	Succinyladenosine
SAICAr	Succinyl-aminimidazole carboxamide riboside
SAM	S-Adenosyl methionine
s.c.	Subcutaneously
SCAD	Short-chain acyl-CoA dehydrogenase
SCHAD	Short-chain 3-hydroxyacyl-CoA dehydrogenase
SCID	Severe combined immunodeficiency
SD	Standard deviation
SDH	Sarcosine dehydrogenase
Ser	Serine
SGA	Small for gestational age
SLE	Systemic lupus erythematosus
SO	Sulfite oxidase
SSD	Succinic semialdehyde dehydrogenase
STAR	Steroidogenic acute response protein
subst.	Substance
T	Testosterone
TAA	Tubular amino acid reabsorption
TAT	Tyrosine aminotransferase
TG	Triglycerides
TH	Tyrosine hydroxylase
THAM	Tris(hydroxyaminomethyl)buffer
THCA	Trihydroxycholestanoic acid
THC	Tetrahydrocortisone



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THF	Tetrahydrofolic acid or Tetrahydrocortisol
Thr	Threonine
TIA/CVA	Transient ischemic attack/cerebrovascular accident
TID	Three times/day
TLC	Thin-layer chromatography
TMS	Tandem mass spectrometry
tRNA	Transfer RNA
Trp	Tryptophan
Tyr	Tyrosine
U	Urine
UAA	Urinary amino acids
UCRE	Urine creatinine
UDP	Uridine diphosphate
UMP	Uridine monophosphate
UMPH	Uridine monophosphate hydrolase
UMPS	Uridine monophosphate synthase
UroA	Urocanic acid
UTI	Urinary tract infection
UTP	Uridine triphosphate
UV	Ultraviolet
Val	Valine
VDAC	Voltage-dependent anion channel
VEP	Visual evoked potential
VLA	Vanillactic acid
VLCAD	Very-long-chain acyl-CoA dehydrogenase
VLCFA	Very-long-chain fatty acid(s)
VLDL	Very-low-density lipoproteins
VMA	Vanillylmandelic acid
WBC	White blood cells
WD	Wilson disease
XA	Xanthurenic acid
X-ALD	X-linked adrenoleukodystrophy
Xan	Xanthine
XDH	Xanthine oxidase/dehydrogenase
ZS	Zellweger syndrome