Errata *for* Nutrition Management of Patients with Inherited Metabolic Disorders

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Corrections highlighted in yellow and red below.

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Table 2-1 Expected Analyte Profiles for MS/MS Newborn Screening Disorders

MS/MS ANALYTES	POSSIBLE DISORDER(S)	ACRONYM	McKUSICK NUMBER
↑Cit	Citrullinemia (argininosuccinate synthetase deficiency)	ASA	215700
	Argininosuccinic aciduria (argininosuccinate lyase deficiency)	CIT	207900
	Citron Defciency	CIT II	603471
↑Met	Homocystinuria (cystathionine β synthetase deficiency)	HCY	236200
	Methionine adenosyltransferase deficiency	MET	250850
↑Phe, Phe/Tyr	Phenylketonuria (phenylalanine hydroxylase deficiency)	PKU	261600
	Biopterin synthesis defects	BIOPT-BS	261630
	Biopterin reg		
↑Tyr	Tyrosinemia: Fumarylacetoacetic acid hydrolase deficiency	TYR Ia *	276700
	Methylacetoacetic acid isomerase deficiency	TYR Ib *	603758
	Tyrosine aminotransferase deficiency	TYR II	276600
	p-OH phenylpyruvatic acid dioxygenase deficiency	TYR III	276710
↑Leu (± Val)	Maple syrup urine disease (branched chain α-keto acid dehydrogenase deficiency)	MSUD	248600
↓ C0	Primary carnitine deficiency (carnitine plasma membrane transporter deficiency/ carnitine uptake deficiency)	CUD	212140
↑C3, C3/C2	Methylmalonic acidemia (methylmalonyl CoA mutase deficiency and cobalamin defects)	MUT	251000
	Cobalamin A	CblA	251100
	Cobalamin B	CblB	251110
	Cobalamin C	CblC	277410
	Cobalamin D	CblD	277400
	Propionic acidemia (propionyl CoA carboxylase deficiency)	PROP	606054
↑C4	Short chain acyl CoA dehydrogenase deficiency	SCAD	201470
	Isobutyryl CoA dehydrogenase deficiency	IBG	611283
↑C5	Isovaleric acidemia (isovaleryl CoA dehyrogenase deficiency)	IVA	243500
	2-methyl butyryl CoA dehydrogenase deficiency/ short branched chain CoA dehydrogenase deficiency	2MBG (SBCAD)**	600006 (600301)
↑C5:1 (±C5OH)	β ketothiolase deficiency (acetoacetyl CoA thiolase deficiency)	BKT	248600
↑C5-DC	Glutaric acidemia type I (glutaryl CoA dehydrogenase deficiency)	GA I	231670
↑C5-OH (± C5:1)	3-methylcrotonyl glycinuria (3 methyl crotonyl CoA carboxylase deficiency)	3MCC	210200
↑C5-OH (± C6-DC)	3-OH-3-methyl-glutaryl-CoA lyase deficiency	HMG	246450
↑C5-OH (± C3)	Multiple carboxylase deficiency	MCD	253260
↑C5-OH	3-methylglutaconyl hydratase deficiency	3MGA	250950
↑C8 and C8/C10 (±C6,C10:1)	Medium chain acyl CoA dehydrogenase deficiency	MCAD	201450
↑C14:1 and ↑C14:1/C12:1 (± C14, C16, C18:1)	Very long chain acyl CoA dehydrogenase deficiency	VLCAD	201475
↑C16, C18:1	Carnitine palmitoyl transferase II deficiency	CPTII	255110
	Carnitine-acylcarnitine translocase deficiency	CACT	212138
↑C16-OH, C18:1-OH	Long chain hydroxy acyl CoA dehydrogenase deficiency	LCHAD	609016
	Trifunctional protein deficiency	TFP	609015
↑C4, C5, C5-DC, C6, C8, C12, C14, C16	Glutaric acidemia type II (multiple acyl CoA dehydrogenase deficiency)	GA II	231680

* TYR 1a and 1b may be missed by MS/MS NBS targeting elevated Tyr – see text

**2MBG and SBCAD are two names for the same disorder

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Figure 2.5 The Carnitine Transport Cycle (correct diagram below)

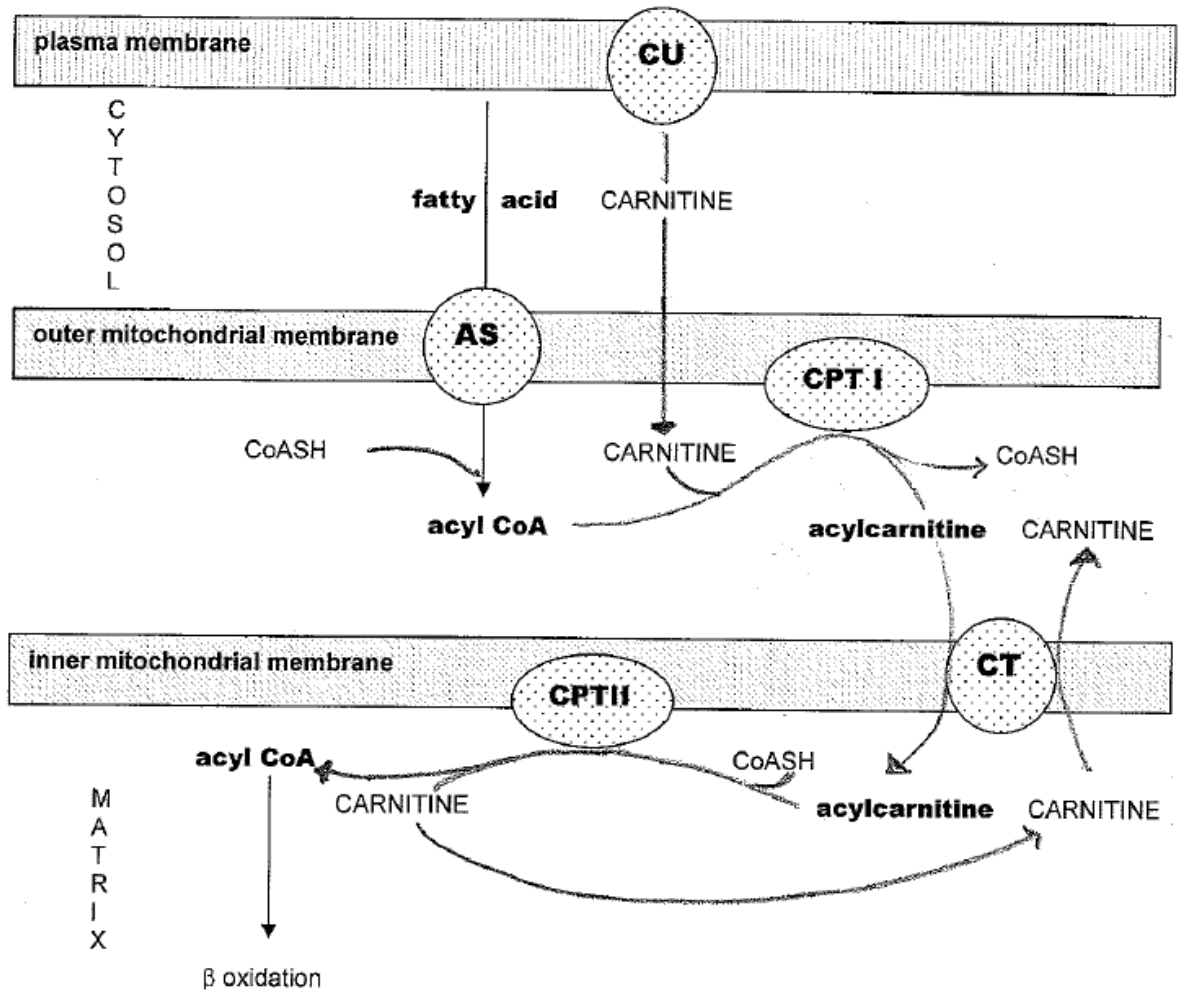

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Table 3-1. Recommended Daily Intakes [RDIs] for Patients Ingesting Elemental Diets

Nutrient	**Recommended Intake at Age***							
	0 <6 mos	**6 < 12 mos**	**1 < 4 yr**	**4 < 7 yr**	**7<11 yr**	**11 < 19 yr**	**Adult**	**Pregnant**
Protein†, g	4.5 - 3.0 /100 kcal		≥ 35	≥ 40	≥ 50	≥ 65	≥ 70	1st trim 70 2nd trim 85 3rd trim 100
Fat, g	6.0 - 3.3 / 100 kcal		> 30	45	55	60	60-70	75

Linoleic acid, g	4.4	4.6	7.0	10.0	12.0	16.0	17.0	14.0
□-linolenic acid, g	0.5	0.5	0.7	0.9	1.2	1.6	1.6	1.5
Energy‡, kcal	120/kg	110/kg	900-1800	1300-2300	1650-3300	1500-1900	2000-3300	1700-2700
Fluid, mL	1.5/kcal	1.5-1.0/kcal	900-1800	1300-2300	1650-3300	1500-1900	2000-3300	1700-2700
Minerals								
Calcium, mg	400	600	800	800	1300	1300	1200	1300
Chloride								
mg	55-150/100 kcal		1500	1900	2300	2300	2300	2300
mEq	1.55-4.2/100 kcal		42.3	54	65	65	65	65
Chromium, µg	0.2	5.5	11	15	25	35	30	3.0
Copper, mg	0.60	0.80	1.5	2.0	2.5	3.0	3.0	3.5
Iodine, µg	25-75 / 100 kcal		90	90	120	150	150	220
Iron§,¶, mg	10	15	15	15	15	18	18	48
Magnesium, mg	50	75	150	200	250	420	420	430
Manganese, mg	0.3	0.6	1.5	2.0	2.0	2.5	2.5	3.0
Molybdenum, µg	2.0	3.0	17	22	34	45	45	50
Phosphorus, mg	350	500	800	800	1250	1250	1000	1250
Potassium,								
mg	80-200/100 kcal		117	149	176	184	184	184
mEq	2.0-5.1/100 kcal		3.0	3.8	4.5	4.7	4.7	4.7
Selenium, µg	20	25	30	40	50	65	65	70
Sodium								
mg	20-60/100 kcal		1000	1200	1500	1500	1500	1500
mEq	0.87-2.61/100 kcal		43	52	65	65	65	65
Zinc, mg	5	5	10	10	15	15	15	20
Vitamins								
A								
IU	250-750 / 100 kcal		400	500	700	900	900	1000
µg RAE	75 - 225 / 100 kcal		120	150	210	270	270	300
D								
IU	40-100/100 kcal		400	400	400	400	400	480
µg	1.0-2.5/100 kcal		10	10	10	10	10	12

E, mg	6	7	6	7	11	15	15	12
K, µg	5	10	30	55	60	75	90	90
Ascorbic acid, mg	40	50	45	45	45	75	60	60
Biotin, µg	35	50	50	50	50	50	50	50
B_6, mg	0.3	0.6	0.9	1.3	1.6	1.8	2.0	2.2
B_{12}, µg	0.6-0.9	0.9-1.8	2.0	2.5	3.0	3.0	3.0	3.0
Choline, mg	125	150	200	250	375	550	550	450
Folate, µg	65	80	150	200	300	400	400	800
Inositol, mg	4/100 kcal		60	80	100	120	130	140-200
Niacin Equivalent**, ††, mg	6	8	9	11	16	18	19	20
Pantothenic acid, mg	2.0	3.0	2.0	3.0	4.0	5.0	5.0	6.0
Riboflavin, mg	0.4	0.6	0.8	1.0	1.4	1.6	1.7	1.7
Thiamin, mg	0.3	0.5	0.7	0.9	1.2	1.4	1.5	1.6

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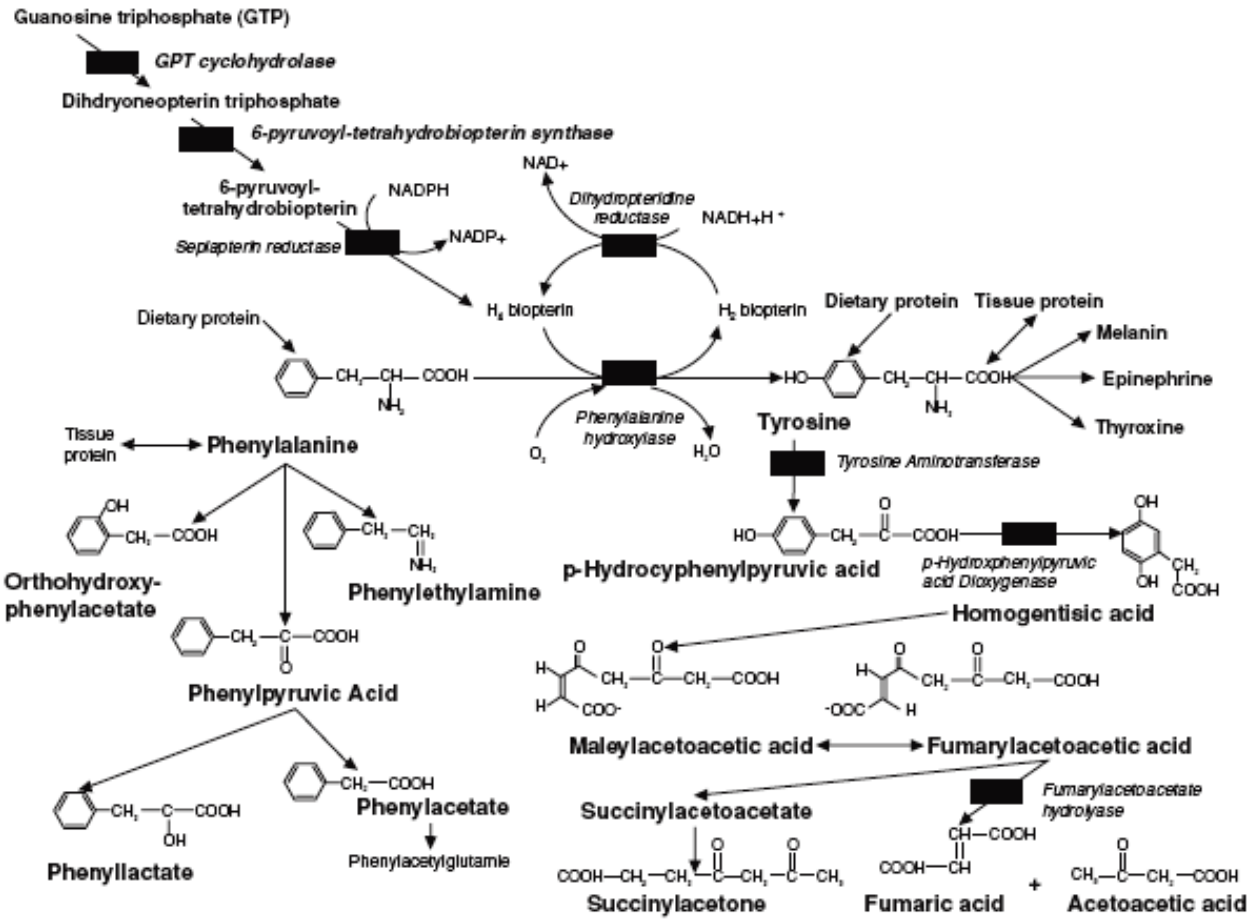
Figure 5-1 Synthesis of Tetrahydrobiopterin and Metabolism of Aromatic Amino Acids (correct diagram below)



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After 10 years of age, plasma PHE concentrations could be increased to > 600 umol/L and at 20 years to > 1200 umol/L.

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The disorders for which nutrition management are outlined in this chapter are MSUD, isovaleric acidemia (IVA), β-methylcrotonyl-CoA carboxylase deficiency (βMCC), β-hydroxy-β-methylglutaryl- CoA (HMG-CoA) lyase deficiency, and mitochondrial acetoacetyl- CoA thiolase deficiency, commonly known as β-ketothiolase (βKT) deficiency.

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Figure 6.1 Metabolism of Branched-Chain Amino Acids (correct diagram below)

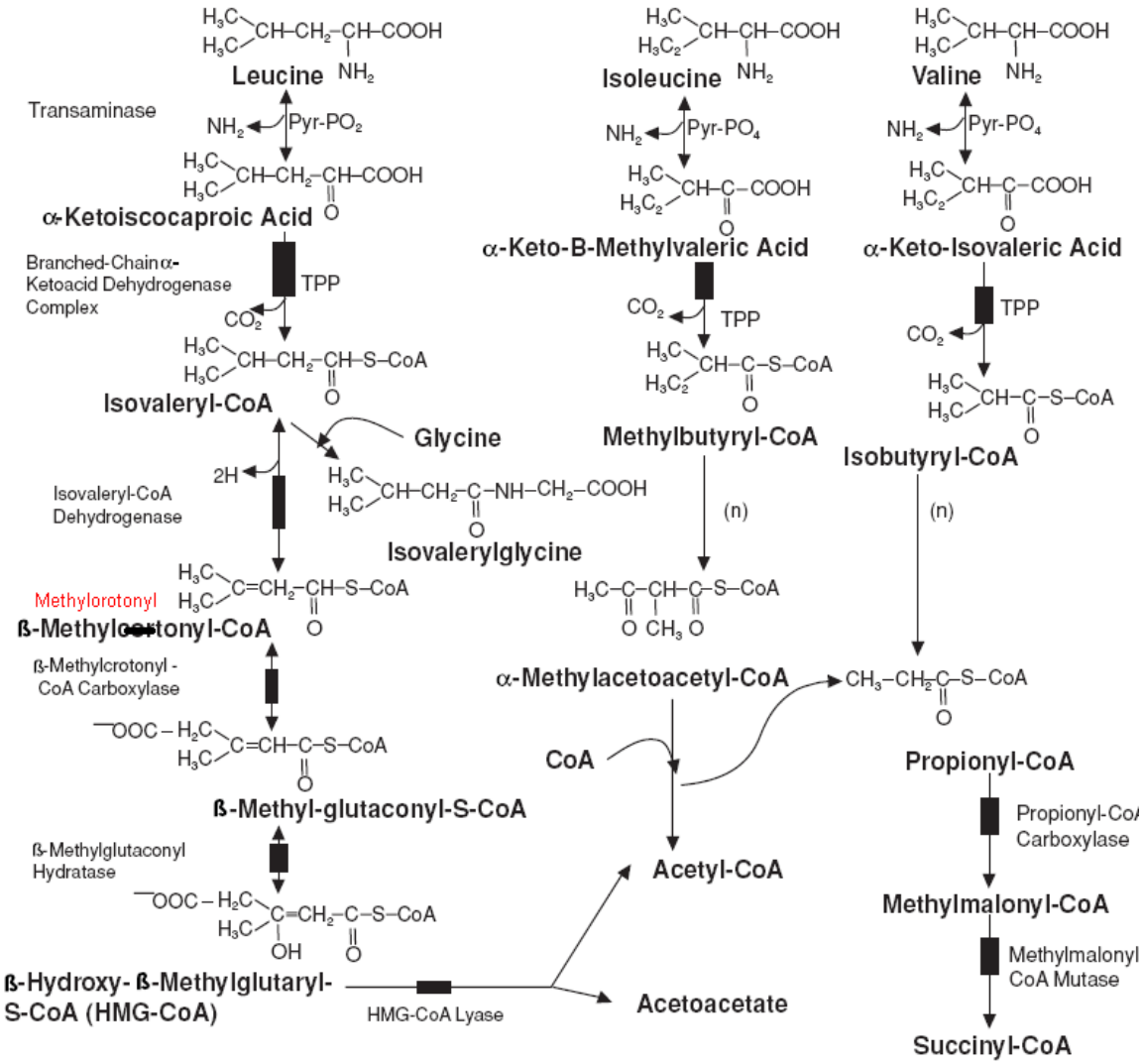

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Table 6-6 Nutrition Composition and Sources of LEU-Free Medical Foods (continued)

Medical Foods	*Modified[a] Nutrient(s) (mg/100 g)*	*Protein Equiv[b] (g/100 g, source)*	*Fat (g/100 g, source)*	*Carbohydrate (g/100 g, source)*	*Energy (kcal/100 g/kJ/100 g)*	*Linoleic Acid/ α-Linolenic Acid (mg/100 g)*
			Nutricia[f]			
XLeu Analog™	Glycine 2050 L-carnitine 10 Taurine 20	13 Amino acids[c]	20.9 High oleic safflower, coconut, and soy oils	59 Corn syrup solids, galactose	475/1985	3025/NA
XLeu Maxamaid®	Glycine 3990 L-carnitine 20 Taurine 140	25 Amino acids[c]	<0.1	56 Sugar, corn syrup solids	324/1754	0
XLeu Maxamum®	Glycine 6300 L-carnitine 39 Taurine 140	40 Amino acids[c]	<1	34 Sugar, corn syrup solids	305/1275	0

Notes:
NA = not available.
Values listed, although accurate at time of publication, are subject to change. For current information, refer to product labels.
[a]LEU-free.
[b]Nitrogen, g × 6.25 = g protein.
[c]Abbott Nutrition, 3300 Stelzer Road, Columbus, Ohio, 43219. 800-551-5838.
[d]All except glycine are in the L-form.
[e]Mead Johnson Nutritionals, 2400 West Lloyd Expressway, Evansville, Indiana 47721. 800-457-3550.
[f]Nutricia North America, PO Box 117, Gaithersburg, Maryland 20884. 800-365-7354.

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Increased concentrations of 3-hydroxyisovaleric acid and β- methylcrotonylglycine are found in urine with the absence of methylcitrate, and α-methyl-β-hydroxybutyrate ~~is~~ found in combined carboxylase deficiency.[128]

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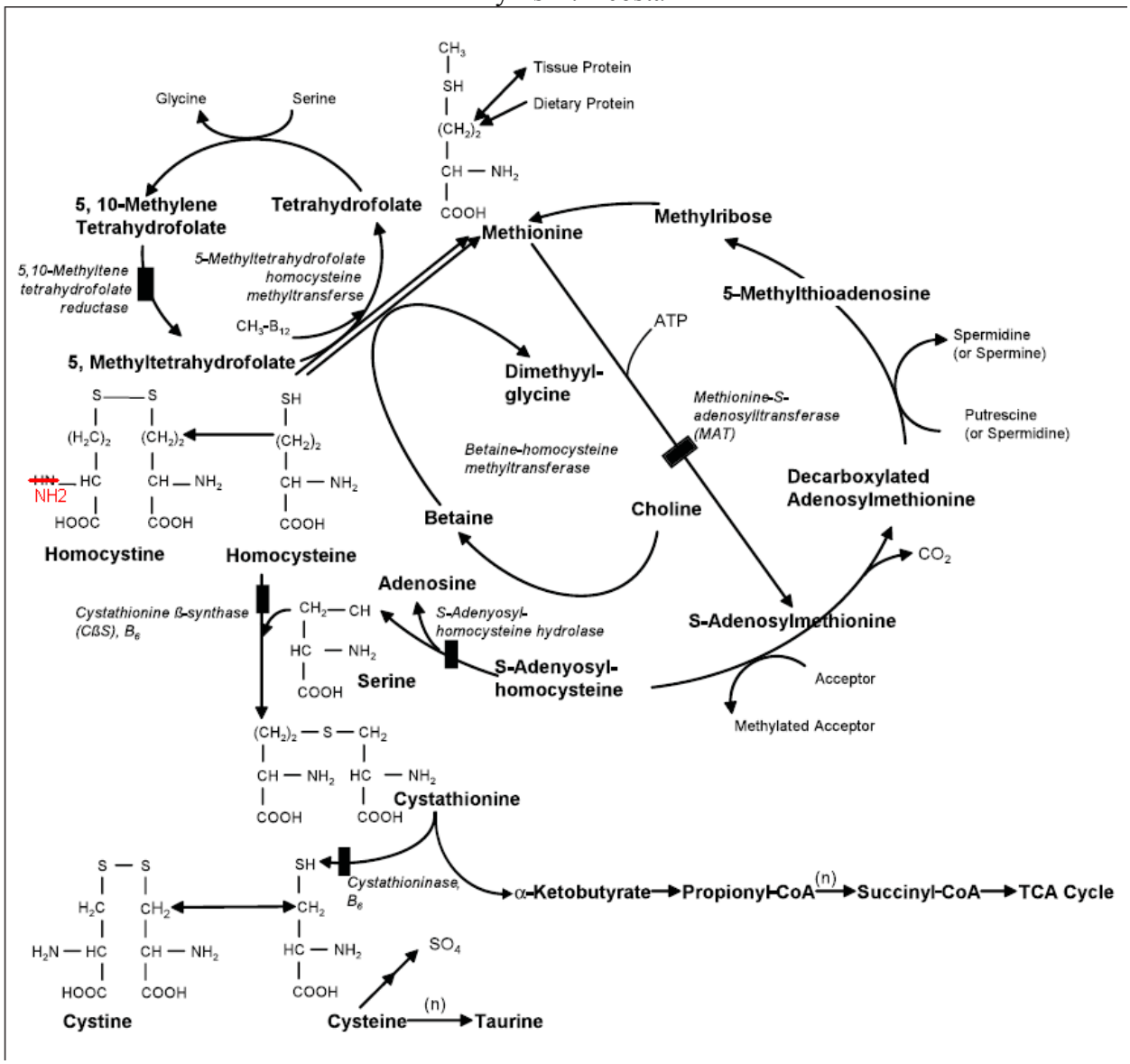
CYS is used as an abbreviation for both cystine and cysteine since interconversion in the body is nonenzymatic.

Figure 7.1



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Use of ratios, such as MET to leucine (LEU) and isoleucine (MET:LEU + ILE), can also help reduce false detection of hypermethioninemia.

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More recently, Picker and Levy suggested the following protocol:

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However, using current diagnostic criteria, review of these studies did not verify this diagnosis.[67]

In addition, long-term biochemical control is necessary to maintain ocular health in patients with CβS deficiency.

Page 249

Another recent study suggested that abnormal HCY metabolites replace dehydroascorbic acid in connective tissue metabolism.[87]

Page 260

The edema resolved when therapy was discontinued.[133]

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Table 7.4 *Diet Plan for a Neonate Who Weighs 3.5 kg Diagnosed with Vitamin B_6 Nonresponsive Homocystinuria*

Food	*Amount*	*MET (mg)*	*CYS (mg)*	*Protein (g)*	*Energy (kcal)*
Similac® Advance® infant formula with iron, powder	26 g	71	42	2.8	137
Hominex®-1 powder	63 g	0	284	9.5	302
Cystine solution (13 mg/100 mL)[a]	600 mL	0	78	0.0	0
Water to make[b]					~~615~~
Total		71	404	12.3	~~615~~ 439
Per kg body weight		20	115	3.5	125

[a]L-CYS has a solubility of ~13 mg/100 g water at room temperature. *Source:* Ajinomoto. *Amino Acid Handbook*. Ajinomoto, Japan. 2004.
[b]Add water to yield a total 615 mL (~21 fl oz @ 20 kcal/ fl oz).

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However, severe MATI/III deficiency can cause demyelination with reduced cognitive function later in childhood.[165,168]

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Methionine Synthase (CBlG) Deficiency (OMIM 250940)[13] and Methionine Synthease Reductase (CBlE) Deficiency (OMIM 236270)[13]

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Figure 8.1 Metabolic Pathways for Isoleucine, Valine, Threonine, Methionine, and Odd-Chain Fatty Acids (correct diagram below)

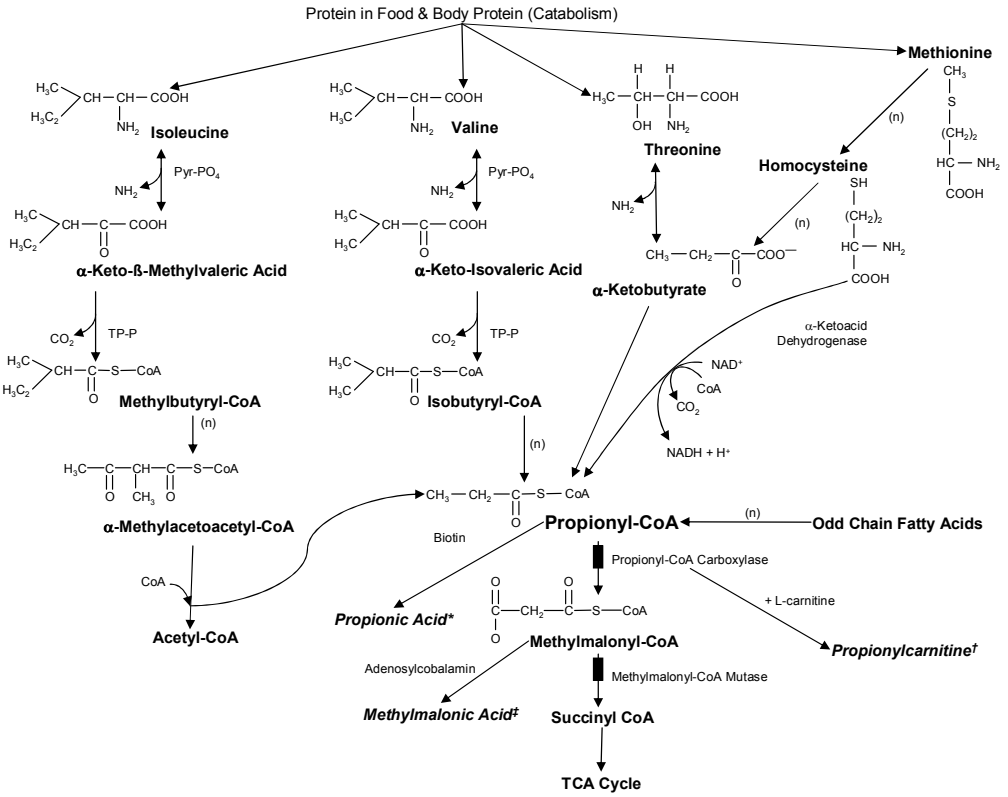

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Table 8.3 *Formulation, Nutrient Composition, and Sources of Medical Foods for Patients with Propionic Acidemia or Methylmalonic Acidemia (per 100 g powder), Continued*

Medical Foods	*Modified Nutrient(s) (mg/100 g)*	*Protein Equiv[a] (g/100 g, source)*	*Fat (g/100 g, source)*	*Carbohydrate (g/100 g, source)*	*Energy (kcal/100 g/kJ/100 g)*	*Linoleic Acid/ α-Linolenic Acid (mg/100 g)*
			Nutricia North America			
XMTVI Analog®	MET 0, VAL 0 ILE trace, THR 0 L-carnitine 10 Taurine 20	13.0 Amino acids[c]	20.9 High oleic safflower, coconut, soy oils	59.0 Corn syrup solids, ~~galactose~~	475/1985	3025/ND
XMTVI Maxamaid®	MET 0, VAL 0 ILE trace, THR 0 L-carnitine 20 Taurine ~~40~~ 140	25.0 Amino acids[c]	<0.1	56.0 Sugar, corn syrup solids	324/1354	0/0
XMTVI Maxamum®	MET 0, VAL 0 ILE trace, THR 0 L-carnitine 39 Taurine 140	40.0 Amino acids[c]	<1.0	34.0 Sugar, corn syrup solids	305/1275	0/0
Milupa OS2®	MET 0, VAL 0 ILE 0, THR 0 L-carnitine 0 Taurine 0	56.0 Amino acids[c]	0.0	18.9 Sugar	300/1254	0/0

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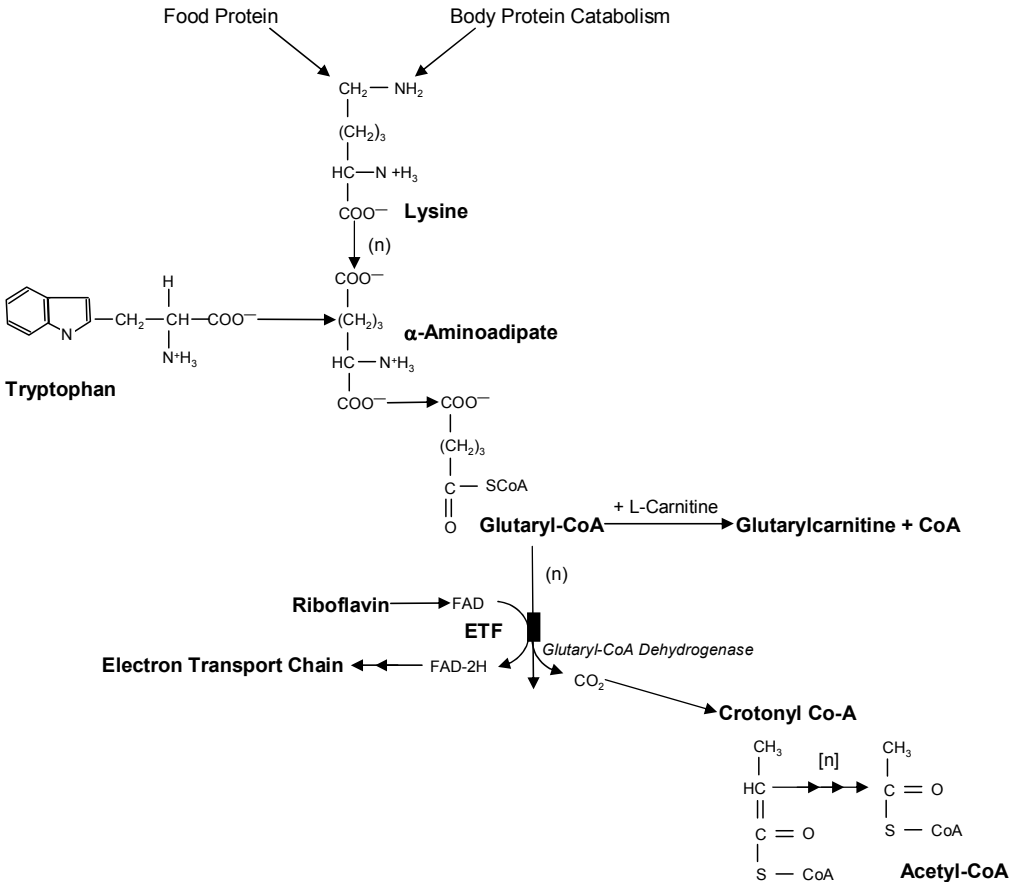
Figure 8.5 Metabolism of Lysine and Tryptophan (correct diagram below)



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Kolker et al.[153] recommended energy intakes at approximately 120% DRIs.

Page 322

Deficient trace mineral and vitamin intakes and plasma concentrations have been reported in children consuming elemental formulas.[56,180]

Page 323

Researchers have reported nearnormal development in many patients treated neonatally.[158,163,173]

Page 350

Proprietary infant formulas made with soy protein isolate contain fat and essential fatty acids (see Appendix F).

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Examples of an emergency letter can be found on the FAOD parent support website at http://www.fodsupport.org.

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For VLCAD, a sum of the long-chain C14:0, C14:1, C16:0, C16:1, C18:0, C18:1, and C18:2 plasma acylcarnitine esters can be calculated.

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Figure 11.1Inherited Metabolic Disorders in the Urea Cycle and Nutrition Approaches to Their Management (correct diagram below)