TABLE 434e-1 INHERITED DISORDERS OF AMINO ACID METABOLISM (CONTINUED)

Amino Acid(s)	Condition	Enzyme Defect	Clinical Findings	Inheritance
Lysine	Hyperlysinemia, saccharopinuria	α-Aminoadipic semialdehyde synthase	Benign	AR
	Pyridoxine-dependent seizures	L-Δ^1-Piperideine-6-carboxilate dehydrogenase	Seizures	AR
Lysine, tryptophan	α-Ketoadipic acidemia	α-Ketoadipic acid dehydrogenase	Benign	?
	Glutaric acidemia type I	Glutaryl-CoA dehydrogenase	Progressive severe dystonia and athetosis, motor delays	AR
	Glutaric acidemia type II	Electron transfer flavoproteins (ETF) or ETF:ubiquinone oxidoreductase	Hypoglycemia, metabolic acidosis, "sweaty feet" odor, hypotonia, cardiomyopathy	AR
Ornithine	Gyrate atrophy of the choroid and retina	Ornithine-δ-aminotransferase	Myopia, night blindness, loss of peripheral vision, cataracts, chorioretinal degeneration	AR
Urea cycle	Carbamoylphosphate synthase-1 deficiency	Carbamoylphosphate synthase-1	Lethargy progressing to coma, protein aversion, intellectual disability, hyperammonemia	AR
	N-Acetylglutamate synthase deficiency	*N*-Acetylglutamate synthase	Lethargy progressing to coma, protein aversion, intellectual disability, hyperammonemia	AR
	Ornithine transcarbamylase deficiency	Ornithine transcarbamylase	Lethargy progressing to coma, protein aversion, intellectual disability, hyperammonemia	XL
	Citrullinemia type I	Argininosuccinate synthase	Lethargy progressing to coma, protein aversion, intellectual disability, hyperammonemia	AR
	Argininosuccinic acidemia	Argininosuccinate lyase	Lethargy progressing to coma, protein aversion, intellectual disability, hyperammonemia, trichorrhexis nodosa	AR
	Arginase deficiency	Arginase	Spastic tetraparesis, intellectual disability, mild hyperammonemia	AR
	Hyperornithinemia, hyperammonemia, homocitrullinuria	Mitochondrial ornithine carrier ORNT1	Vomiting, lethargy, failure to thrive, intellectual disability, episodic confusion, hyperammonemia, protein intolerance	AR
	Citrullinemia type 2	Mitochondrial aspartate/glutamate carrier CTLN2	Neonatal intrahepatic cholestasis, adult presentation with sudden behavioral changes and stupor, coma, hyperammonemia	AR
Proline, ornithine, arginine	Δ^1-Pyrroline-5-carboxylate synthase deficiency	Δ^1-Pyrroline-5-carboxylate synthase	Hypotonia, seizures, neurodegeneration, peripheral neuropathy, joint laxity, skin hyperelasticity, subcapsular cataracts, hyperammonemia	AR
Glutamine	Glutamine synthase deficiency	Glutamine synthase	Brain malformations, pachygyria, seizures, hypotonia, dysmorphic features	AR
Valine	Hypervalinemia	Valine aminotransferase	Vomiting, fever, failure to thrive, hypotonia	AR
	Isobutyryl-CoA dehydrogenase deficiency	Isobutyryl-CoA dehydrogenase	Failure to thrive, anemia, and dilated cardiomyopathy(?)	AR
Leucine, isoleucine	Hyperleucine-isoleucinemia	Leucine-isoleucine aminotransferase	Seizures, failure to thrive, intellectual disability	?
Valine, leucine, isoleucine	Maple syrup urine disease	Branched chain ketoacid dehydrogenase (E1α, E1β, E2, E3 deficiency)	Lethargy, vomiting, encephalopathy, seizures, intellectual disability, "maple syrup" odor, protein intolerance	AR
Leucine	Isovaleric acidemia	Isovaleryl-CoA dehydrogenase	Acidosis, ketosis, vomiting, coma, hyperammonemia, "sweaty feet" odor, protein intolerance	AR
	3-Methylcrotonyl glycinuria	3-Methylcrotonyl-CoA carboxylase	Stress-induced metabolic acidosis, hypotonia, hypoglycemia, "cat's urine" odor	AR
	3-Methylglutaconic aciduria type I	3-Methylglutaconyl-CoA hydratase deficiency	Stress-induced acidosis, leukodystrophy, hypotonia, hepatomegaly	AR
	3-Hydroxy-3-methylglutaric aciduria	3-Hydroxy-3-methylglutaryl-CoA lyase	Stress-induced hypoketotic hypoglycemia and acidosis, encephalopathy, hyperammonemia	AR
Isoleucine	2-Methylbutyryl-glycinuria	2-Methylbutyryl-CoA dehydrogenase	Fasting-induced metabolic acidosis/hypoglycemia	AR
	2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency	2-Methyl-3-hydroxybutyryl-CoA dehydrogenase	Developmental regression, seizures, and rigidity sometimes triggered by illnesses	XL
	3-Oxothiolase deficiency	3-Oxothiolase	Fasting-induced acidosis and ketosis, vomiting, lethargy	AR
Valine, isoleucine, methionine, threonine	Propionic acidemia (pccA, -B, -C)	Propionyl-CoA carboxylase	Metabolic ketoacidosis, hyperammonemia, hypotonia, lethargy, coma, protein intolerance, intellectual disability, hyperglycinemia	AR
	Multiple carboxylase/biotinidase deficiency	Holocarboxylase synthase or biotinidase	Metabolic ketoacidosis, diffuse rash, alopecia, seizures, intellectual disability	AR
	Methylmalonic acidemia (mutase, cblA, -B, racemase)	Methylmalonyl-CoA mutase/racemase or cobalamin reductase/adenosyltransferase	Metabolic ketoacidosis, hyperammonemia, hypertonia, lethargy, coma, protein intolerance, intellectual disability, hyperglycinemia	AR

Abbreviations: AD, autosomal dominant; AR, autosomal recessive; Cbl, cobalamin; DOPA, dihydroxyphenylalanine; GABA, γ-aminobutyric acid; GTP, guanosine 5′-triphosphate; XL, X-linked.