TABLE 434e-1 INHERITED DISORDERS OF AMINO ACID METABOLISM

Amino Acid(s)	Condition	Enzyme Defect	Clinical Findings	Inheritance
Phenylalanine	Phenylketonuria	Phenylalanine hydroxylase	Intellectual disability, microcephaly, hypopigmented skin and hairs, eczema, "mousy" odor	AR
	DHPR deficiency	Dihydropteridine reductase	Intellectual disability, hypotonia, spasticity, myoclonus	AR
	PTPS deficiency	6-Pyruvoyl-tetrahydropterin synthase	Dystonia, neurologic deterioration, seizures, intellectual disability	AR
	GTP cyclohydrolase I deficiency	GTP cyclohydrolase I	Intellectual disability, seizures, dystonia, temperature instability	AR
	Carbinolamine dehydratase deficiency	Pterin-4α-carbinolamine dehydratase	Transient hyperphenylalaninemia (benign)	AR
Tyrosine	Tyrosinemia type I (hepatorenal)	Fumarylacetoacetate hydrolase	Liver failure, cirrhosis, rickets, failure to thrive, peripheral neuropathy, "boiled cabbage" odor	AR
	Tyrosinemia type II (oculocutaneous)	Tyrosine transaminase	Palmoplantar keratosis, painful corneal erosions with photophobia, intellectual disability (?)	AR
	Tyrosinemia type III	4-Hydroxyphenylpyruvate dioxygenase	Hypertyrosinemia with normal liver function, occasional mental delay	AR
	Hawkinsinuria	4-Hydroxyphenylpyruvate dioxygenase	Transient failure to thrive, metabolic acidosis in infancy	AD
	Alkaptonuria	Homogentisic acid oxidase	Ochronosis, arthritis, cardiac valve involvement, coronary artery calcification	AR
	Albinism (oculocutaneous)	Tyrosinase	Hypopigmentation of hair, skin, and optic fundus; visual loss; photophobia	AR
	Albinism (ocular)	Different enzymes or transporters	Hypopigmentation of optic fundus, visual loss	AR, XL
	DOPA-responsive dystonia	Tyrosine hydroxylase	Rigidity, truncal hypotonia, tremor, intellectual disability	AR
GABA	4-Hydroxybutyric aciduria	Succinic semialdehyde dehydrogenase	Seizures, intellectual disability, ataxia	AR
Tryptophan	Tryptophanuria	Unknown	Intellectual disability, ataxia, skin photosensitivity	AR
	Hydroxykynureninuria	Kynureninase	Intellectual disability, spasticity	AR
Histidine	Histidinemia	Histidine-ammonia lyase	Benign	AR
	Urocanic aciduria	Urocanase	Benign	AR
	Formiminoglutamic aciduria	Formiminotransferase	Occasional intellectual disability	AR
Glycine	Glycine encephalopathy	Glycine cleavage (4 enzymes)	Infantile seizures, lethargy, apnea, profound intellectual disability	AR
	Sarcosinemia	Sarcosine dehydrogenase	Benign	AR
	Hyperoxaluria type I	Alanine:glyoxylate aminotransferase	Calcium oxalate nephrolithiasis, renal failure	AR
	Hyperoxaluria type II	D-Glyceric acid dehydrogenase/ glyoxylate reductase	Calcium oxalate nephrolithiasis, renal failure	AR
Serine	Phosphoglycerate dehydrogenase deficiency	Phosphoglycerate dehydrogenase	Seizures, microcephaly, intellectual disability	AR
Proline	Hyperprolinemia type I	Proline oxidase	Benign	AR
	Hyperprolinemia type II	Δ^1-Pyrroline-5-carboxylate dehydrogenase	Febrile seizures, intellectual disability	AR
	Hyperhydroxyprolinemia	Hydroxyproline oxidase	Benign	AR
	Prolidase deficiency	Prolidase	Mild intellectual disability, chronic dermatitis	AR
Methionine	Hypermethioninemia	Methionine adenosyltransferase	Usually benign	AR
	S-Adenosylhomocysteine hydrolase deficiency	S-Adenosylhomocysteine hydrolase	Hypotonia, intellectual disability, absent tendon reflexes, delayed myelination	AR
	Glycine N-methyltransferase deficiency	Glycine N-methyltransferase	Elevated liver transaminases	AR
	Adenosine kinase deficiency	Adenosine kinase	Intellectual disability, seizures, liver dysfunction	AR
Homocystine	Homocystinuria	Cystathionine β-synthase	Lens dislocation, thrombotic vascular disease, intellectual disability, osteoporosis	AR
	Homocystinuria	5,10-Methylenetetrahydrofolate reductase	Intellectual disability, gait and psychiatric abnormalities, recurrent strokes	AR
	Homocystinuria	Methionine synthase (cblE, -G)	Intellectual disability, hypotonia, seizures, megaloblastic anemia	AR
	Homocystinuria and methylmalonic acidemia	Vitamin B_{12} lysosomal efflux and metabolism (cblC, -D, -F, -J, -X)	Intellectual disability, lethargy, failure to thrive, hypotonia, seizures, megaloblastic anemia	AR
Cystathionine	Cystathioninuria	β-Cystathioninase	Benign	AR
Cystine	Cystinosis	Cystinosin CTNS (lysosomal efflux)	Renal Fanconi's syndrome, rickets, photophobia, hypotonia, renal failure	AR
S-Sulfo-L-cysteine	Sulfocysteinuria	Sulfate oxidase or molybdenum cofactor deficiency	Seizures, intellectual disability, dislocated lenses	AR

(*Continued*)