Disorder	Enzyme defect	Clinical Symptoms (if unmanaged)	Biochemical	Management
Tyrosinemia type I (HT-1)	Fumarylacetoacetate hydrolase (FAH)	Severe liver failure, vomiting, bleeding, neuropathy, neurological crises; cirrhosis; hepatocarcinoma	↑ urine and blood succinylacetone (diagnostic) ↑ TYR; ↑ MET (when liver disease has progressed)	Nitisinone (NTBC); PHE- and TYR-restricted diet (liver transplantation for select patients)
Tyrosinemia type II	Tyrosine aminotransferase (TAT)	Corneal lesions, hyperkeratosis Neurological complications	↑ ↑ TYR (blood and urine), ↑ increased PHE, ↑ urinary phenolic acids,	PHE- and TYR-restricted diet
Tyrosinemia type III and neonatal tyrosinemia	4-hydroxy phenylpyruvic acid dioxygenase (4-HPPD)	Impaired mental function Uncertain clinical relevance	↑ TYR; ↑ urinary 4-HPPA	PHE- and TYR- restricted diet; short term protein restriction in neonatal tyrosinemia
Hawkinsinuria	4-hydroxyphenylpyruvic acid dioxygenase (4-HPPS); autosomal dominant mutation	Failure to thrive, acidosis; doubtful clinical relevance	↑ urinary hawkinsin acetic acid (2-L-cystein-S-yl, 4-dihydroxycyclohex-5-en-1-yl)	PHE- and TYR restricted diet + Vit.C supplementation in infancy
Alkaptonuria	Homogentisate oxygenase (HGD)	Arthritis, cardiac valve disease	↑ homogentisic acid	Low protein diet, possibly NTBC