Tyrosinemia type 1

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Tyrosinemia type 1. ORPHA:882

Tyrosinemia type 1 (HTI) is an inborn error of tyrosine catabolism caused by defective activity of fumarylacetoacetate hydrolase (FAH) and is characterized by progressive liver disease, renal tubular dysfunction, porphyria-like crises and a dramatic improvement in prognosis following treatment with nitisinone.