Tyrosinemia

National Cancer Institute

Source

National Cancer Institute. Tyrosinemia. NCI Thesaurus. Code C98640.

An autosomal recessive inherited metabolic disorder caused by mutations in the FAH, HPD, and TAT genes. It is characterized by deficiency of one of the enzymes that are involved in the metabolism of tyrosine. It results in elevated blood tyrosine levels and accumulation of tyrosine and its byproducts in the liver, kidney, nervous system and other organs.