Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome

National Cancer Institute

Source

National Cancer Institute. Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome. NCI Thesaurus. Code C129029.

An autosomal recessive disorder caused by mutation(s) in the SLC25A15 gene, encoding mitochondrial ornithine transporter 1. The condition is characterized by failure to thrive, liver dysfunction, psychomotor retardation, encephalopathy and seizures.