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Hartnup Disease

National Cancer Institute

Source

National Cancer Institute. *Hartnup Disease*. NCI Thesaurus. Code C84748.

An autosomal recessive inherited metabolic disorder caused by mutations in the SLC6A19 gene. It is characterized by defective absorption of neutral amino acids. Signs and symptoms include skin eruptions reminiscent of pellagra, aminoaciduria, and cerebellar ataxia.