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Hereditary Folate Malabsorption

National Cancer Institute

Source

National Cancer Institute. *Hereditary Folate Malabsorption*. NCI Thesaurus. Code C156424.

An autosomal recessive condition caused by mutation(s) in the SCL46A1 gene, encoding proton-coupled folate transporter. It is characterized by low concentrations of folate resulting in megaloblastic anemia, immune deficiency, and neurologic deficits.