Arakawa Syndrome II

National Cancer Institute

Source

National Cancer Institute. Arakawa Syndrome II. NCI Thesaurus. Code C99081.

A rare autosomal dominant inherited metabolic disorder characterized by deficiency of the enzyme tetrahydrofolate-methyltransferase. It results in the abnormal metabolism of methylcobalamin. Signs and symptoms include mental retardation, megaloblastic anemia, hypotonia, epilepsy, and hepatosplenomegaly.