Wolcott-Rallison Syndrome

National Cancer Institute

Source

National Cancer Institute. Wolcott-Rallison Syndrome. NCI Thesaurus. Code C131007.

A rare, autosomal recessive condition caused by mutation(s) in the EIF2AK3 gene, which encodes translation initiation factor 2-alpha kinase-3. The condition is characterized by the following: permanent insulin-dependent diabetes, with onset in the neonatal period or infancy; epiphyseal dysplasia; deficient bone mineralization, diagnosed in the first year or two of life; and liver dysfunction, occurring in early childhood. Other features may include intellectual deficit, hypothyroidism, renal dysfunction, neutropenia, and thyroid dysfunction. The manifestations and clinical course are variable.