Vitamin D 25-Hydroxylase Deficiency

National Cancer Institute

Source

National Cancer Institute. Vitamin D 25-Hydroxylase Deficiency. NCI Thesaurus. Code C131074.

An autosomal recessive form of rickets caused by inactivating mutation(s) in the CYP2R1 gene, encoding vitamin D 25-hydroxylase, the hepatic enzyme that converts vitamin D to 25-hydroxyvitamin D, the precursor of 1,25-dihydroxyvitamin D (calcitriol). The condition is characterized by reduced serum concentrations of 25-hydroxyvitamin D, hypophosphatemia, hypocalcemia with secondary hyperparathyroidism and elevated serum alkaline phosphatase, and by failure to thrive, seizures, muscle weakness, and rickets.