Vitamin D 1 Alpha-Hydroxylase Deficiency

National Cancer Institute

Source

National Cancer Institute. Vitamin D 1 Alpha-Hydroxylase Deficiency. NCI Thesaurus. Code C131073.

An autosomal recessive form of rickets caused by inactivating mutation(s) in the CYP27B1 gene, encoding 25-hydroxyvitamin D-1 alpha hydroxylase, the renal enzyme that converts 25-hydroxyvitamin D to 1 alpha,25-dihydroxyvitamin D (calcitriol), the active metabolite of vitamin D (cholecalciferol). The condition is characterized by reduced serum concentrations of 1 alpha,25-hydroxyvitamin D, normal concentrations of 25-hydroxyvitamin D, increased serum alkaline phosphatase, hypocalcemia due to reduced intestinal calcium absorption, hypophosphatemia due to renal phosphate wasting, secondary hyperparathyroidism, rickets, seizures, muscle weakness and failure to thrive.