Hereditary Hypophosphatemic Rickets with Hypercalciuria

National Cancer Institute

Source

National Cancer Institute. Hereditary Hypophosphatemic Rickets with Hypercalciuria. NCI Thesaurus. Code C131450.

An autosomal recessive form of hypophosphatemic rickets caused by inactivating mutation(s) in the SLC34A3 gene, encoding sodium-dependent phosphate transport protein 2C, a protein involved in maintenance of inorganic phosphate concentration in the kidney. The condition is characterized by elevated 1,25-dihydroxyvitamin D (calcitriol) concentrations, resulting in increased intestinal calcium absorption and hypercalciuria. This form of hypophosphatemic rickets is also distinguished by the lack of elevated fibroblast growth factor 23 (FGF23) concentrations.