inactive compounds and/or causing resistance to its action. The more marginal the vitamin D intake in the diet, the more likely that anticonvulsant therapy will lead to abnormal mineral and bone metabolism.

Vitamin D–Dependent Rickets Type I Vitamin D–dependent rickets type I, previously termed *pseudo-vitamin D–resistant rickets*, is caused by homozygous or compound heterozygous mutations in the gene encoding 25(OH)D-1α-hydroxylase. It differs from true vitamin D–resistant rickets (vitamin D–dependent rickets type II, see below) in that it is typically less severe and the biochemical and radiographic abnormalities can be readily reversed with physiologic doses of the vitamin's active metabolite, $1,25(OH)_2D$ **(Chap. 409)**. Clinical features include hypocalcemia, often with tetany or convulsions; hypophosphatemia due to secondary hyperparathyroidism; and thus, osteomalacia and increased levels of alkaline phosphatase.

Vitamin D–Dependent Rickets Type II Vitamin D–dependent rickets type II results from end-organ resistance to the active metabolite $1,25(OH)_2D$. The clinical features resemble those of the type I disorder and include hypocalcemia, hypophosphatemia, secondary hyperparathyroidism, and rickets but also partial or total alopecia. Plasma levels of $1,25(OH)_2D$ are elevated, in keeping with the refractoriness of the end-organs. This disorder is caused by homozygous or compound heterozygous mutations in the gene encoding the vitamin D receptor; treatment requires regular, usually nocturnal calcium infusions, which normalize PTH levels, thus reducing urinary phosphate excretion and thereby improving rickets and thus growth, but do not restore hair growth **(Chap. 409)**.

PSEUDOHYPOPARATHYROIDISM PHP refers to a group of distinct inherited disorders. Patients affected by PHP type Ia (PHP1A) develop symptoms and signs of hypocalcemia in association with distinctive skeletal and developmental defects, referred to as Albright's hereditary osteodystrophy (AHO). The hypocalcemia is due to a deficient PTH response in the proximal renal tubules, probably leading to insufficient $1,25(OH)_2D$ production and thus impaired intestinal calcium absorption. Furthermore, PTH resistance