

Familial hypophosphatemia is a term that describes a group of rare inherited disorders characterized by impaired kidney conservation of phosphate and in some cases, altered vitamin D metabolism. In contrast, other forms of hypophosphatemia may result from inadequate dietary supply of phosphate, or its poor absorption from the intestines. The chronic hypophosphatemia resulting from these impairments can lead to rickets, a childhood bone disease with characteristic bow deformities of the legs, growth plate abnormalities, and progressive softening of the bone, referred to as osteomalacia. In children, growth rates may be impaired, frequently resulting in short stature. In adults, the growth plate is not present so that osteomalacia is the evident bone problem. Familial hypophosphatemia is most often inherited in an X-linked dominant manner, however, autosomal dominant and recessive forms of familial hypophosphatemia occur.