endocrines

Special Issue Reprint

Update on X-linked Hypophosphatemia

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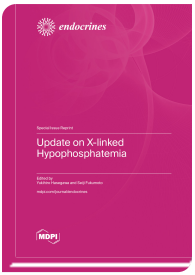

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Rickets and osteomalacia are associated with impaired mineralization in growth plate cartilage and the bone osteoid. Vitamin D deficiency has been one of the major causes for rickets and osteomalacia. In addition, it has been also known that there are diseases called vitamin D-resistant rickets/osteomalacia which cannot be cured by native vitamin D. Since the cloning of *FGF23* in 2000, it has become clear that most cases of vitamin D-resistant rickets/osteomalacia are caused by excessive actions of FGF23. X-linked hypophosphatemia (XLH) is the most common cause of inherited FGF23-reltated hypophosphatemic rickets. Patients with XLH suffer from life-long morbidity of bone, cartilage, ligament, joint, tooth and muscle which significantly affects quality of life. Since FGF23 was shown to be the humoral factor causing hypophosphatemia in patients with XLH, anti-FGF23 antibody that blocks the actions of FGF23 has been developed and become clinically available in several countries. The introduction of this new drug together with advanced knowledge concerning physiological and pathophysiological significance of FGF23 has further stimulated research and promoted dissemination of fruits of the studies in this field. From these backgrounds, a variety of both basic and clinical topics concerning XLH are covered in this special issue to provide up-to-date summaries of the current understanding.

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