

# Vitamin D deficiency presenting like hypophosphatemic osteomalacia

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### ABSTRACT

**Introduction:** Osteomalacia is one of the most common osteometabolic diseases. It is typically caused by lack of vitamin D and is characterized by mineralization deficiency of the osteoid matrix in the cortical and trabecular bone. Indians are at particularly high risk for developing osteomalacia, because of their traditional clothing style and skin color. This condition is frequently misdiagnosed and it can present with bone pain and muscle weakness. **Objective:** We report a case of osteomalacia with predominant hypophosphatemia. **Case report:** A 41-year-old male presented with gradually progressive quadriparesis for past 6 months. Patient had low phosphorous (1.9 mg/dl) with calcium being 8.1 mg/dl and 25(OH)Vitamin D3 levels being 8.12 ng/ml. The patient was thought to have concomitant Vitamin D deficiency with possibility of tumor-induced osteomalacia. Further evaluation for the same was planned. However, patient was given a trial of Vitamin D supplementation before further investigations and had dramatic improvement in the form that muscle power improved gradually over next 6 weeks and was able to climb stairs after 2 months. Retrospectively patient gave a history of being at home for many months and there was no exposure to sunlight at all. The biochemical parameters normalized within 4 weeks of starting Vitamin D and serum alkaline phosphatase showed drastic reduction after 6 weeks. All his family members were screened and found to have Vitamin D deficiency including recent born twins having rachitic changes. This was a case of vitamin D deficiency, in 41-year-old male, which presented more like hypophosphatemic osteomalacia but characteristic history of lack of sunlight exposure and dramatic improvement with only vitamin D3 supplementation confirmed the diagnosis. **Conclusion:** The presentation of vitamin D deficiency can be varied and high index of suspicion is important for treatment of this common but frequently misdiagnosed condition.

**Key words:** Vitamin D deficiency, hypophosphatemia, osteomalacia

## INTRODUCTION

Osteomalacia is one of the most common osteometabolic diseases. It is typically caused by lack of vitamin D and is characterized by mineralization deficiency of the osteoid matrix in the cortical and trabecular bone. Adequate exposure to sunlight and fortification of dairy products with vitamin D has decreased the prevalence of vitamin D deficiency. Indians are at particularly high risk for developing

osteomalacia, because of their traditional clothing style, social customs and dark skin color. The factors like poor diet, lack of sun exposure, and the age-related decline in the dermal synthesis of 7-dehydrocholesterol predispose to vitamin D deficiency and consequent bone disease.<sup>[1]</sup> Here, we present a case of severe osteomalacia, with predominant hypophosphatemia, presenting with progressive weakness, to the extent, enough to make him bed bound secondary to poor diet, and a lack of exposure to sunlight.

## CASE REPORT

A 41-year-old male presented with gradually progressive quadriparesis for past 6 months. Patient had low phosphorous (1.9 mg/dl) with calcium being 8.1 mg/dl and 25(OH)Vitamin D3 levels being 8.12 ng/ml. The

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patient was thought to have concomitant vitamin D deficiency with possibility of tumor-induced osteomalacia. Further evaluation for the same was planned. However, the patient was given a trial of vitamin D supplementation before further investigations were done and he had dramatic improvement in the muscle power, which improved gradually over next 6 weeks and he was able to climb stairs after 2 months. Retrospectively patient gave a history of being at home for many months and there was no exposure to sunlight at all. The calcium and phosphorus levels normalized within 4 weeks of starting vitamin D supplementation and serum alkaline phosphatase showed drastic reduction after 6 weeks. All his family members were screened and found to have vitamin D deficiency including twins aged 1 year who had rachitic changes.

## DISCUSSION

This was a case of vitamin D deficiency, in 41-year-old male, which presented more like hypophosphatemic osteomalacia but characteristic history of lack of sunlight exposure and dramatic improvement with only vitamin D3 supplementation confirmed the diagnosis. The biochemical profile of our case was typical of osteomalacia. Osteomalacia is a condition of adults characterized by softening of bones because of accumulation of osteoid matter, which is protein matrix, mainly comprising of collagen, which fails to mineralize. The most common cause is vitamin D deficiency. The main biological effect of vitamin D is to increase calcium absorption by the gut and mobilize mineral from the bone. The effects on renal tubules are inhibition of calcium resorption and encouraging phosphate resorption. However, vitamin D deficiency leading on to secondary hyperparathyroidism can have decreased phosphorus because of its phosphaturic effect.

Osteomalacia when presents classically manifests with symptoms and signs that can be collectively referred to as osteomalacic syndrome. Some symptoms are so vague, nonspecific, and sometimes misleading that it can easily escape the attention of the clinician, whereas others are highly specific and often diagnostic.<sup>[2]</sup> In this case too, patient presented with progressive quadriparesis, which made him visit physician and neurologist for many a times before he was referred to us on strong suspicion of metabolic bone disease, based on one single report of low phosphorus. A high degree of suspicion in the right clinical context is necessary to diagnose osteomalacia especially in the early stages.

Varied clinical manifestations of osteomalacia are based on different underlying pathogenetic mechanisms, though still it is poorly understood. The classic symptoms are bone pain and tenderness, muscle weakness, and difficulty in walking, all of which can often be vague and unremitting. However, history of decrease in severity might be there during summer months. Consequently, patients are misdiagnosed as fibromyalgia, severe myopathy, unusual pain syndrome, or neurologic disorders of unknown cause. Proximal limb muscle weakness is characteristic of osteomalacia. The severity varies from a subtle abnormality found only on careful physical examination to severe disability like complete paralysis.<sup>[3]</sup>

In its classic presentation, hypocalcemia, hypophosphatemia, and increased serum alkaline phosphatase level are the classic biochemical triad of osteomalacia and increased serum alkaline phosphatase level is the most frequent and the earliest biochemical manifestation.<sup>[4]</sup>

Radiographic findings show generalized demineralization with loss of transverse trabeculae and persistent transverse loosers zone. The loosers zone or pseudofractures typically extend across the bone. These indicate incomplete fractures that have healed by callus consisting of osteoid tissue devoid of mineralization. It typically occurs at neck of femur, ramii of ischium, pubis, and axillary edge of scapula just below glenoid.<sup>[2]</sup> In spine, the vertebra has typical biconcave appearance leading to cord fish appearance.

Treatment can be in the form of vitamin D replenishment. Depending upon etiology, based on patient's absorption capacity, vitamin D can be supplemented orally or given intramuscular. Careful and close follow-up is necessary during the first few months (1–3 months) of therapy to avoid therapy-related problems, although it is extremely uncommon to see hypercalcemia or renal dysfunction. Increased risk of long-bone and hip fractures should be kept in mind as the patient's muscle weakness improves and bone pain disappears before bone mass and strength is restored.

## CONCLUSION

Hypophosphatemia is commonly missed due to nonspecific signs and symptoms, but it causes considerable morbidity and can contribute to mortality. Proper diagnosis requires a thorough medication history, family history, examination, and assessment of renal tubular phosphate handling to identify the cause.<sup>[5]</sup> The presentation of vitamin D deficiency can be varied and high index of suspicion is important for treatment of this common but frequently misdiagnosed condition.

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