



### RESEARCH ARTICLE

## MYOPATHYAS AN UNUSUAL PRESENTATION OF OSTEOMALASIA IN ADOLESCENCE, CASE REPORT.

**Alaa Qasim Alawadi.**

FIMC of pediatrics, Karbala Teaching Hospital for children, Karbala, Iraq.

### Manuscript Info

#### Manuscript History

Received: 10 November 2018

Final Accepted: 12 December 2018

Published: January 2019

#### Key words:-

Proximal myopathy, osteomalacia in adolescence, vitamin D deficiency.

### Abstract

Reporting an adolescent patient with osteomalacia presented initially with complete inability to walk and clinical features consistent with proximal myopathy which is one of the uncommon presentation for osteomalacia and vitamin D deficiency.

After excluding other causes leading to myopathy by clinical, laboratory, graphical, and imaging methods and confirming the diagnosis of osteomalacia by lab. And radiological methods. Remarkable improvement of the myopathy and other signs and symptoms of vitamin D deficiency after starting the treatment with vitamin D and calcium supplement was noted.

This report illustrates the importance of considering osteomalacia as a possible and treatable cause of proximal myopathy in adolescents and young adults.

Copy Right, IJAR, 2017,. All rights reserved.

### Introduction:-

Osteomalacia is a disorder of decreased mineralization of newly formed osteoid at sites of bone turnover, whereas rickets is a disorder of defective mineralization of cartilage in the epiphyseal growth plates of children. Osteomalacia and rickets can occur together in children (open growth plates), but only osteomalacia occurs in adults (fused growth plates). Several different disorders cause osteomalacia [1]. Populations at risk include the homebound who have little sun exposure and insufficient dietary calcium and vitamin D, patients with malabsorption related to gastrointestinal bypass surgery or celiac disease, and women who wear traditional veils or dresses which prevent sun exposure [2,3].

Hereditary forms of vitamin D deficiency and resistance, which are identified in childhood, are associated with osteomalacia in adults, but these disorders are less common.

In developing countries, vitamin D deficiency rickets is still considered as a major community health problem in infants and children causing, while osteomalacia due to vitamin D deficiency is reported with increasing frequency among adolescents [2-3].

Proximal myopathy occurs in association with several muscle diseases. However, myopathy associated with vitamin D deficiency is an unusual cause of myopathy [4-6]. This causative association can easily be overlooked although the condition is readily treatable.

**Corresponding Author:-Alaa Qasim Alawadi.**

Address:-FIMC of pediatrics, Karbala Teaching Hospital for children, Karbala, Iraq.

In this case report, we present our experience with an adolescent patient with osteomalacia who presented with proximal myopathy. The patient responded very well to vitamin D therapy and calcium supplement.

### Methods & Results:-

A 13-year-old female presented to us in the general pediatric ward, in a wheel chair, unable to walk, complaining of pain in her extremities and back with a history of progressive weakness, fatigability, lethargy and difficulty in climbing stairs and standing up from sitting position. Her condition worsened in the last 6 months.

Her diet consisted of no milk or milk product intake. She reported minimal sun exposure in her daily life. She had no symptoms to suggest malabsorption, liver and renal diseases or history of fracture.

On examination the patient was fully conscious, depressed looking, well nourished, white skin (Caucasian), alert girl with no dysmorphic features. Stable vital signs. Growth assessment indicated that her weight was 50 kg at 50<sup>th</sup> centile, and her height was 147 cm at 25<sup>th</sup> centile. She had carpopedal spasm (noted when we took her blood pressure). Cardiovascular, chest and abdomen examination was normal. Examination of the central nervous system revealed cranial nerves normal. Unstable gait, unable to rise from squatting position with proximal muscle weakness which was demonstrated by failure to overcome mild resistance at lower limbs and by moderate resistance at upper limbs. There was no evidence of muscular atrophy or tenderness either in the upper or lower limbs. Deep tendon reflexes and sensation were intact. Laboratory investigations showed normal complete blood count (CBC), Random blood sugar (RBS) 126 mg/dl, serum calcium was 4.2 mg/dl, normal serum phosphate, normal other electrolytes including serum potassium level. Alkaline phosphatase 447.4 U/L (N= 40-150), normal liver function, renal function & blood gas, CPK 53.26 u/dl (N = 29-

200), normal celiac screening and thyroid function, and iPTH was 515.7 pg/mg (N=15-68), 25 hydroxyvitamin D was very low 1.6 ng/ml (N=30-40). X-ray of pelvis AP showed severe generalized osteopenia with no fractures. EMG was normal. DEXA scan (bone density measuring scan) not available in our hospital.

Calcium was given to correct the acute symptoms & signs of hypocalcaemia. As osteomalacia was confirmed biochemically and radiologically, the patient received 1,25-dihydroxyvitamin D<sub>3</sub> (0.05 microgram/kg) calcium intake of at least 1000 mg per day. All patients should maintain a calcium intake of in the range of 1000 to 1500 for 2 months, since inadequate intake of calcium may contribute to the development of osteomalacia [18-19].

The patient's myopathy improved remarkably within the first 4 days & responded very well to treatment in terms of feeling of well-being, mobility, gait, and muscle power. She was discharged after a week of hospitalization to complete the line of therapy at home with instructions regarding diet, sport, and sun exposure. After 2 months of treatment symptoms and signs of myopathy recovered completely and symptoms of fatigue, depression disappeared, the vitamin D level was in normal range.

### Discussion:-

There is a growing prevalence of vitamin D deficiency in many countries, which when severe (25-hydroxyvitamin D <10 ng/mL [25 nmol/L]) and prolonged, results in: hypocalcemia, secondary hyperparathyroidism, secondary hypophosphatemia, and osteomalacia [8].

The main cause of osteomalacia worldwide is vitamin D deficiency. The well-known causes are low intake and limited sun exposure [9], gastrointestinal diseases as malabsorption. Other causes include Vitamin D resistance, hereditary or acquired disorders of phosphate wasting, type 2 renal tubular acidosis, and excessive exposure to inhibitors of bone mineralization (eg, aluminum toxicity, endemic fluorosis).

No study showing the prevalence of osteomalacia in Karbala nor in Iraq was found. But studies from Saudi Arabia [6-7] showed that vitamin D levels tend to be low among the general population especially children and females. The conservative environment in the holy city of Karbala is so much similar to that of Saudi Arabia regarding women who wear traditional veils or dresses.

(thick black rather sacred clothing) .one study done in Department of Pediatrics, College of Medicine and King Khalid University Hospital, King Saud University, Riyadh [6],got to the same conclusion of our study .in the study mentioned they had 3 cases which were all females which had myopathy as a presenting feature of thereosteomalasia. This out weightthe explanationof the clothing habits..

Osteomalacia may be asymptomatic and present radiologically as osteopenia. It can also produce characteristic symptoms, independently of the etiology, including diffuse bone pain, polyarthralgias, muscle weakness, and difficulty walking [10-12]. Few previous studies reported the association of myopathy with vitamin D deficiency . In a report[13], 17 patients with osteomalacia on bone biopsy, the following findings were observed: (Bone pain and muscle weakness in 84 percent ,Bone tenderness in 84 percent,Fracture in 76 percent.Difficulty walking and waddling gait in 24 percent .This diversity in the presentationof osteomalasia and limited number of studies addressing the problem may explain the lack of attention of doctors to osteomalasia as a cause of myopathy in practice.famous academic books didn't mention the myopathy as a finding in osteomalasia[22].

The actual scientific explanation of myopathy related to osteomalasia have been poorly elicited ,Some studies implicate it to direct effect of vit .D on skeletal muscles,others to the effect of secondary hyperparathyroidism causing atrophy in muscle fibers type 2 , [9,14,15]

No study was found regarding theimprovement timeneeded ,thehealing of the myopathy- in our patient - was remarkable, started to be obvious within the first 4 daysof starting the treatment, patient was walking on discharge,(10 days later) .follow up 1 month later ,still on therapy, normal gait and muscle power , can climb stairs , vit. D reading was 19.3ng/dl and two months later 22ng/dl her mother then reported her daughter less depressed more active participating in sports .she was changed to vit.D3 50u/wk to maintain the results and prevent recurrence.

Healing of osteomalaciaand bone densityhealing may take many months to a year and varies with the degree and duration of the deficiencyin one study bone density may improve within three to six months but it starts by when there are increases in urinary calcium excretion[17]. This will be reported with future follow up of the patient.

DEXA scan not available in our governmental hospitals in karbala.to confirm osteomalacia.Several studies have demonstrated markedly reduced spine, hip, and forearm bone density in patients with osteomalacia related to vitamin D deficiency [20 -21].

It is not a common clinical practice to consider osteomalasia and vitamin D deficiency in the differential diagnosis of muscle weakness and proximal myopathy, Normal CPK in the presence of clinical evidence of myopathy may draw the attention to the possibility of osteomalasia& vitamin D deficiency as the underlying cause.

### **Conclusion:-**

proximal myopathy is an uncommon butimportant feature of osteomalacia in adolescence . in this study we highlight the importance of considering osteomalacia as one of the differential diagnosis of anypatient who presents with proximal muscle weakness.

### **Abbreviations:-**

PTHParathyroid Hormone

CPKCreatinine Phosphokinase

ALP Alkaline Phosphatase

Vit.D Vitamin D

EMGElectromyogram

DEXADual -Energy X-ray Absorptiometry

**References:-**

1. CJ Menkes, MD. Osteomalacia up to datefile:///F:/UpToDate19.3/UpToDate19.3/contents/mobipreview.htm?37/34/38446#H3169360
2. Al-Shoha A, Qiu S, Palnitkar S, Rao DS. Osteomalacia with bone marrow fibrosis due to severe vitamin D deficiency after a gastrointestinal bypass operation for severe obesity. *EndocrPract* 2009; 15:528.
3. Kennel KA, Drake MT, Hurley DL. Vitamin D deficiency in adults: when to test and how to treat. *Mayo ClinProc* 2010; 85:752.
4. Pfeifer M, Begerow B, Minne HW. Vitamin D and muscle function. *Osteoporosis Int* 2002; 13: 187-94
5. . Glerup H, Mikkelsen K, Poulsen L, et al. Hypovitaminosis D myopathy without biochemical signs of osteomalacicbone involvement. *Calcif Tissue Int* 2000; 66: 419-424.
6. Hessah M. AL-Otaibi, Nasir A.M AL-Jurayyan, Sarar Mohamed, Mustafa A. M. SalihOsteomalacia in adolescents presenting as proximal myopathy
7. Department of Pediatrics, College of Medicine and King Khalid University Hospital, King Saud University, Riyadh, Saudi Arabia.*CurrPediatr Res* 2012; 16 (1): 57-60
8. Abdullah MA, Salhi HS, Bakry LA et al .adolescent rickets in Saudi Arabia: a rich and sunny country. *J PediatrEndocrinolMetab* 2002; 15: 1017-1025
9. shwell M, Stone EM, Stolte H, et al. UK Food Standards Agency Workshop Report: an investigation of the relative contributions of diet and sunlight to vitamin D status. *Br J Nutr* 2010; 104:603
10. Glerup H, Mikkelsen K, Poulsen L, et al. Hypovitaminosis D myopathy without biochemical signs of osteomalacic bone involvement. *Calcif Tissue Int* 2000; 66: 419-424
11. Lips, P, van Schoor, NM, Bravenboer, N. Vitamin D-related disorders. In: ``Primer on the metabolic bone diseases and disorders of mineral metabolism" Seventh edition, Rosen, CJ (Eds), American Society of Bone and Mineral Research, 2008. p.329.
12. Gifre L, Peris P, Monegal A, et al. Osteomalaciarevisited : a report on 28 cases. *ClinRheumatol* 2011; 30:639.
13. Bhan A, Rao AD, Rao DS. Osteomalacia as a result of vitamin D deficiency. *EndocrinolMetabClin North Am* 2010; 39:321.
14. Basha B, Rao DS, Han ZH, Parfitt AM. Osteomalacia due to vitamin D depletion: a neglected consequence of intestinal malabsorption. *Am J Med* 2000; 108:296.
15. Ceglia L. Vitamin D and skeletal muscle tissue andfunction, *Molecular Aspects of medicine* 2008; 29:407-414.
16. Russell JA, Ostemalacic Myopathy. *Muscle Nerve* 1994; 17: 578-580
17. Bhan A., Rao AD, Rao AS. Osteomalacia as a result of vitamin D deficiency. *Endocrinology and Metabolismclinic* 2010; 39 (2): 321-331
18. Allen SC, Raut S. Biochemical recovery time scales in elderly patients with osteomalacia. *J R Soc Med* 2004; 97:527
19. Thacher TD, Fischer PR, Pettifor JM, et al. A comparison of calcium, vitamin D, or both for nutritional rickets in Nigerian children. *N Engl J Med* 1999; 341:563.
20. Bishop N. Rickets today--children still need milk and sunshine. *N Engl J Med* 1999; 341:602.Basha B, Rao DS, Han ZH, Parfitt AM. Osteomalacia due to vitamin D depletion: a neglected consequence of intestinal malabsorption. *Am J Med* 2000; 108:296.
21. Bhambri R, Naik V, Malhotra N, et al. Changes in bone mineral density following treatment of osteomalacia. *J ClinDensitom* 2006; 9:120.
22. Robert M. Kliegman, MD. Nelson text book of pediatrics 20 editionPhiladelphia : ELSEVIER;2016.