

## Vitamin D deficiency induced Myopathy in Children and Adolescents: A Cross sectional study

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### **Abstract:**

**Aim:** To assess the response of Vitamin D therapy in Vitamin D deficiency induced Myopathy in Children and Adolescents

**Study Design:** Cross sectional study

Place and duration: This study was conducted in Pakistan Institute of Medical Sciences Islamabad, Pakistan From February 2018 to March 2020

**Methodology:** The demographics, clinical presentation, laboratory findings, and treatment response of children and adolescents with osteomalacic myopathy were studied. Clinical, biochemical, and radiographic evidence was used to diagnose vitamin D deficiency. Vitamin D deficiency was identified as the source of myopathic symptoms after therapy with vitamin D.

**Results:** In the final analysis, the children ranged in age from 21 months to 18 years. Eight children (53.33%) were under the age of six, three (26.66.5%) were between the ages of seven and twelve, and fifteen (20%) were between the ages of thirteen and eighteen. Difficulty climbing stairs was reported by 100 % of participants (n=15), followed by difficulty walking, which was reported by 93.33 % of participants (n=14). In 53.33 % of the participants (n=08), abnormal gait was seen. From 4 weeks to 6 months, patients' responses to treatment was recorded.

**Conclusion:** To summarize, vitamin D deficiency in children can result in myopathic symptoms. Vitamin D therapy has had a fantastic clinical response. All the study participants responded to vitamin D therapy

**Keywords:** Children, myopathy, osteomalacia, rickets, Vitamin-D deficiency

### **Introduction:-**

In children and young adults, rickets is a very well-known clinical vitamin-D deficiency (VDD) condition. Secondary to hypocalcemia, children with VDD may experience neurological symptoms such as irritability, seizures, tetany, or paresthesia. [1-3] Osteomalacic myopathy is a

well-known but rare symptom of VDD in children, with a high prevalence in teenagers.[3, 4] Persistent walking difficulty, waddling gait, difficulty in ascending stairs, trouble getting up, and eventually, loss of ambulation are all common side effects of osteomalacic myopathy. A typical complication is diffuse musculoskeletal discomfort. On clinical examination, rickets symptoms might be seen, especially in younger children. [5-7]

Vitamin D is a fat-soluble vitamin that belongs to the secosteroids family. It aids in the absorption of calcium, magnesium, iron, phosphate, and zinc in the intestine. Vitamin D (particularly cholecalciferol) is mostly synthesized in the skin, and only a few foods are high in vitamin D. The dermal synthesis of vitamin D from bodily cholesterol is dependent on the amount of time spent in the sun, primarily UVB light. Many people who live in sunny climate locations are discovered to be vitamin D deficient. This is likely due to their clothing and/or cultural behaviours. [8, 9]

Despite being a well-known manifestation of VDD, osteomalacic myopathy is frequently overlooked in the differential diagnosis of muscle weakness in children, resulting in wasteful referrals, investigations, and treatment delays. [10, 11]

Our research aims to report our experience with osteomalacic myopathy in children and adolescents, emphasizing its importance in neurology practice. We also monitored the response of vitamin D therapy in vitamin D deficiency induced myopathy.

### **Methodology:-**

We analyzed 15 children and adolescents diagnosed with vitamin-D deficiency-associated myopathy, commonly known as rachitic or osteomalacic myopathy, who were seen at a tertiary care hospital in Pakistan Institute of Medical Sciences Islamabad, Pakistan between February 2018 and March 2020. Permission was taken from the ethical review committee of the institute.

Clinical symptoms of growing, walking difficulty, difficulty moving upstairs, difficulty rising from a sitting position, or capacity to rise from a bed were used to diagnose myopathy, as was a neurological test that revealed mostly proximal weakness. A combination of clinical (existence of indicators of rickets), biochemical (increased alkaline phosphatase, low serum phosphate, and low/normal serum calcium), and radiographic evidence of rickets and/or osteomalacia were used to diagnose vitamin-D deficiency. Although serum 25-hydroxyvitamin D and parathormone (PTH) were not required for diagnosis, they were recorded whenever they were available. Children having the muscle weakness other than the vitamin D deficiency were excluded from the study.

To attribute myopathy to vitamin D deficiency, a positive response to treatment with a reversal of symptoms was required. All of the youngsters were given 600,000 units of vitamin D, divided into 60,000 units per week for ten weeks, as well as oral calcium supplementation. Unless an underlying condition demanded ongoing follow-ups, children were clinically observed until their symptoms resolved.

**Results:**

The final analysis comprised 15 children, ranging from 21 months to 18 years. Total 08 (53.33%) of the children were under the age of Six, 03 (26.66%) were between the ages of seven and twelve, and 15 (20%) were between the ages of thirteen to eighteen. **[As seen in Table 1]**. Total 07 (50%) participants had recognized vitamin-D insufficiency risk factors. Eight children had additional nutritional deficits, including macrocytic anaemia in five, iron deficiency anaemia in three, and scurvy in two.

**Table 2** shows the participants clinical conditions. Difficulty in Climbing Stairs has been observed in 100% (n=15) participants, followed by difficulty in walking, reported in 93.33% (n=14) participants. Abnormal gait was shown in 53.33% (n= 08) of the participant. Other observed symptoms are bowling of legs, inability to get up from sitting, waddling gait, goiter, body aches, and knee pain were also present. On physical examination, all of the children had a proximal muscular weakness. After the treatment, complete or partial recovery was observed after a different period of time **(As shown in Table 3)**

**Table 1: Demographic Characteristics of the study participants (n=15)**

<b>Variable</b>	<b>Number</b>	<b>Percentage</b>
<b>Gender</b>		
Male	6	40
Female	9	60
<b>Age Group</b>		
Up to 24 months	1	6.66
2 to 6 years	7	46.66
7 to 12 years	4	26.66
13 to 18 years	3	20.00

**Table 2: Clinical presentation and Findings**

<b>Clinical presentation and Findings</b>	<b>Number</b>	<b>Percentage</b>
Difficulty in Walking	14	93.33
Abnormal Gait	08	53.33
Proximal Weakness	15	100.00
Rickets	13	86.66
Difficulty in Climbing Stairs	15	100
Progressive Walking difficulty	13	86.66
Loss of Ambulation	12	80.00
Difficulty Jumping	13	86.66

**Table 3: Duration of Response to Treatment**

<b>Duration</b>	<b>Number</b>	<b>Percentage</b>
4 weeks	3	20
6 weeks	2	12.33
8 weeks	3	20
3 months	1	6.66
4 Months	1	6.66
5 month	2	13.33
6 month	3	20

**Discussion:-**

Vitamin-D deficiency-associated myopathy, also known as osteomalacia (or rachitic) myopathy, causes gradual walking difficulty, waddling gait, difficulty rising from supine or sitting, and difficulty going upstairs, just like other primary muscle illnesses. Patients with severe muscle weakness may need to use a wheelchair. [12, 13] The proximal muscles and lower limbs are the most affected. Osteomalacia is frequently accompanied by body discomfort. Even though it has been recognized for a long time, osteomalacic myopathy is still misdiagnosed as hereditary or inflammatory muscle disease, or even as a psychiatric problem.[12] [8],[14],[15] This is due to the fact that osteomalacic myopathy has no distinguishing features that would lead to the diagnosis. As a result, diagnosing rachitic/osteomalacic myopathy necessitates a high level of clinical suspicion. When a diagnosis is suspected, suitable laboratory and radiographic examinations can quickly confirm it. The remission of myopathic symptoms after vitamin D treatment, however, provides the definitive confirmation. [14][10], [17]

Osteomalacic myopathy is most commonly seen in adolescents, which may be due to an increased need for vitamin D as a result of the physical growth and puberty spurt. [12, 13, 15] At this age, rickets symptoms may not be evident, making clinical testing of VDD even more challenging. [19] Misdiagnosis of rheumatological disorders, primary muscle illness, or even psychiatric disorders, on the other hand, is possible. [16]

In our study, most of the participants were under the age of ten at the time of presentation. Although four of the patients presented with hypocalcemic seizures, a documented but uncommon symptom of VDD in teenagers, they all exhibited proximal myopathy. [1] Myopathy was severe enough in many children to cause full ambulation loss, as others have documented. Diffuse musculoskeletal pain associated with osteomalacia was found in 13 teenagers. [12, 16]. All other



teenagers in our cohort were referred from a different department or outside with suspicion of primary muscle illness.

In addition to teens, our investigation discovered vitamin-D deficiency-related myopathy in younger children. VDD-related myopathy in young children is rarely documented. VDD can cause motor delay in newborns who are still developing motor abilities, which can appear as delays in head control, rolling over, sitting, pulling to sit, standing, or walking, depending on the age of beginning of VDD. [5, 17, 18]

VDD causes regression of previously acquired motor milestones in older children who already have a specific range of motor skills. Due to a lack of awareness, the diagnosis of VDD-associated myopathy may be overlooked, leading to investigations for other hereditary, metabolic, or inflammatory neuromuscular illnesses. [11, 18] In this investigation, eight children aged 21 months to 5 years old presented with symptoms consistent with proximal myopathy.

Muscle-specific tests, such as serum creatine kinase, nerve conduction/electromyographic examinations, and muscle biopsy are ineffective in detecting VDD-associated myopathy since the results are either normal or nonspecific. [11]

Aside from clinical indications of rickets, the existence of radiographic signs of rickets or an aberrant biochemical metabolic bone profile might help to confirm the diagnosis. Before symptoms may be linked to VDD, muscle weakness must be resolved with vitamin D medication. [19]. Only one child in our study exhibited significantly high serum kinase, which could have been caused by muscle injury as a result of seizures and bilateral femoral neck fractures. We did not undertake nerve conduction investigations, electromyography, or muscle biopsy on any of the children because VDD was suspected only at the time of presentation. The therapeutic reaction aided in the discovery of a causal link between VDD and muscle weakness.

Vitamin D deficiency is most common in children at two ages: the first peak occurs throughout childhood and early adolescence, and the second peak occurs during adolescence and is ascribed to an increased requirement for vitamin D due to a physiological development spurt at this age. Furthermore, factors such as nutrition, sun exposure, and skin tone may have contributed to our children's vitamin D insufficiency. In previous investigations, a similar pattern of risk factors was discovered. [12] During follow-up, biochemical and radiological monitoring were not possible due to financial constraints. [20].

**Conclusion:-**

To summarize, vitamin D deficiency can cause myopathic symptoms in children. A high index of suspicion and a thorough clinical examination are required for the diagnosis. Biochemical and radiological tests can be used to confirm the diagnosis if it has been suspected. The clinical response to vitamin D treatment is great. All the study participants responded to vitamin D therapy

**Conflict of interest:**

None

**Funding Source:**

None

**Permission:**

It was taken from the ethical review committee of the institute

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