

# PROXIMAL MYOPATHY: A DIAGNOSTIC DILEMMA

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## INTRODUCTION

Vitamin D deficiency with secondary hyperparathyroidism is common in South-east Asia. In contrast, primary hyperparathyroidism is relatively rare. We present a case of severe proximal myopathy with significant diagnostic delay.

## CASE REPORT

A 23-year-old lady presented with 2yrs history of lower back pain, radiating to both groins and upper thighs associated with recurrent falls. Her pain and weakness progressed insidiously leading to difficulty in standing or walking independently. She had been admitted under orthopedic and neurology departments at different hospitals. MRI spine/thigh and nerve conduction studies were done which were both normal, but serum calcium was low. She was diagnosed with 'lumbago' and sciatica. She was asked to drink four glasses of milk/day and treated with ultrasonic massage and physiotherapy.

Examination revealed proximal muscle weakness of the limbs, waddling gait, generalized bony tenderness, and bilateral genu valgus. Investigations showed results as per Table no 1.

**TABLE 1:**

SL. NO	INVESTIGATION	OBSERVED VALUE	NORMAL RANGE
1.	S. 25 OH VITAMIN D3	<4	<10ng/ml- deficient
2.	PTH	898	15-65 pg/ml
3.	PHOSPHORUS	1.5	2.5-4.9 mg/dl
4.	MAGNESIUM	1.9	1.8-2.4mg/dl
5.	CALCIUM	10.0	8.5-10.1mg/dl
6.	CREATININE KINASE	20	21-215 IU/L
7.	ALKALINE PHOSPHATASE	1959	50-136 IU/L
8.	24HRS URINARY CALCIUM	264	mg/day
9.	24HRS URINARY PHOSPHORUS	0.4	0.4-1.3gm/24hrs

She was commenced on Vitamin D and Phosphate. At follow-up, there was significant improvement in symptoms, particularly bony pain, ALP improved but serum calcium and PTH increased (Table 2).

She was diagnosed with myopathy secondary to osteomalacia, primary hyperparathyroidism and hypophosphatemia. Neck USS and Sestamibi scan elucidated a 2.0x1.3x1.1cms right inferior parathyroid adenoma.

BMD revealed severe osteoporosis (Z-score L2-4: -4.7, femoral neck -3.9, forearm -5.7). She underwent Right inferior parathyroidectomy.

Recent biochemistry shows normal PTH and calcium (Table 2) with ergocalciferol 2000IU/day and Calcium 1000mg/day. Her symptoms subsided, except genu valgus, and she is independent and working.

## DISCUSSION

Myopathy has a wide spectrum of aetiological factors. Our patient had severe osteomalacia, hypophosphatemia and primary hyperparathyroidism. All of these conditions cause myopathy of variable severity. In our patient, it's difficult to determine the predominant aetiological factor. Myopathy due to metabolic causes is treatable and requires prompt diagnosis.

**TABLE 2:**

SL. NO	INVESTIGATION	OBSERVED VALUES			NORMAL RANGE
		BEFORE VITAMIN D REPLACEMENT	AFTER VITAMIN D REPLACEMENT	AFTER PARATHYROIDECTOMY	
1.	CALCIUM	10.0	10.9	8.8	8.5-10.1mg/dl
2.	PTH	897.4	1161	38.44	15-65 pg/ml
3.	PHOSPHORUS	1.5	2.6	3.6	2.5-4.9mg/dl
4.	ALKALINE PHOSPHATASE	1959	1251	696	50-136IU/L
5.	24HRS URINARY CALCIUM	264	198	28.8	mg/day