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A RARE CASE OF MYOPATHY MIMIC LIMB GIRDLE DISEASE

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Background - Differential diagnosis in muscle disorders of young subjects is always unbalanced towards inherited diseases. Nevertheless acquired diseased are important to be considered since are reversible and easily treatable.

Case Report - An Indian 18 year-old female, born from non consanguineous parents and without a positive family history, was admitted to our Department for a two years progressive gait disorder. The patient complained difficulties in running and climbing stairs and muscle pain in proximal lower limb muscles when standing up. Neurological examination revealed muscle weakness involving upper and lower limb muscles with a waddling gait and a Gowers' sign. Moreover hirsutism and truncal obesity was present. Primitive muscular fiber damage was detected on elettromyography in all muscles examined; no spontaneous activity or myotonic phenomenon was observed. Elettroneurography was normal. No alterations on cardiologic and abdominal examination were registered. Laboratory analyses showed very low levels of calcemia (6.1 mg/dL), undetectable 25-OH Vitamin D (< 10 nmol/L) and high PTH (318 pg/mL). Thyroid function, CK and lactic acid were normal. Low bone mass index was detected, but there was no history of fractures and no signs of osteomalacia. Cushing disease and most common adrenogenital syndromes were excluded. Therapy with calcium, calcitriol and colecalciferol supplementation was made with progressive improvement of muscles weakness. After 8 months of supplementation therapy and normalization of vitamin D, calcium and PTH levels, the patient completely recovered the muscle strength.

Discussion - Vitamin D deficiency is defined as a 25-hydroxy vitamin D level of less than 20 ng/ml and a level between 21 ng and 29 ng/ml is considered as relative insufficiency. Level < 10 ng/ml can cause generalized musculoskeletal pain and increases the risk of falls. In 30% of patients it can present as proximal muscle weakness, mimicking LGMD. Lack of sunlight exposure, higher skin melanin content and dietary insufficiency of vitamin D led to vitamin D deficiency in our patient. Because of the striking improvement after medical therapy, we decided to postpone a muscle biopsy, the indication of which will be re-evaluated in case of clinical worsening despite adequate dosage of vitamin D. Vitamin D deficiency is still under-diagnosed and should be kept as one of the differential diagnosis for muscle weakness as the condition is reversible and easily treatable.

References:
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