CASE REPORT

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Diffuse Symmetric Lower Limb Pseudohypertrophy and Muscle Stiffness as the Initial Manifestation of Hypothyroidism

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Abstract

Hypothyroidism is a common disorder that can be accompanied by various neurologic manifestations like mental status changes, seizures, myopathy and neuropathy. Asymptomatic elevations of creative Kinase are more common than symptomatic myopathy. The clinical features of hypothyroid myopathy include proximal weakness, cramps, aching or painful muscles, sluggish reflexes, myoedema and Hoffman's syndrome. We report an unusual case of hypothyroid myopathy in which lower limb pseudohypertrophy and muscle stiffness with markedly elevated CK and TSH levels was the initial and sole manifestation of hypothyroidism.

Introduction

Myopathy can occur in patients with either severe or longstanding untreated hypothyroidsm. Although more than 60% of patients may have an elevated creatine kinase level, symptomatic muscle disease is uncommon[1]. We report an extremely rare case of a young adult male with diffuse symmetric lower limb pseudohypertrophy and muscle stiffness as the initial and sole manifestation of hypothyroidism.

Case report

A 26-year-old male, presented to us with a 2 months history of progressive prominence and bulkiness of the thighs and calves accompanied by stiffness and a mild dull aching pain in both the lower limbs. It was not associated with any limb weakness or difficulty in walking or rising up from the squatting position. Upper limbs were normal. He denied any history of weakness of the trunk, neck or cranial musculature. There was no history of cramps, sensory, bowel-bladder or systemic disturbances. Past and family history were insignificant.

On examination vitals were maintained. General and systemic examination was normal. On motor examination, size, tone, power and reflexes were normal in the upper limbs. Lower limb examination revealed diffuse bilateral, symmetric hypertrophy of both the thigh and calf muscles with marked muscle stiffness. On palpation muscles were firm but not doughy and there was no evidence of tenderness, oedema or percussion myotonia. Deep tendon reflexes were normal.

In view of the history of lower limb muscle hypertrophy and stiffness of recent onset and a negative family history, a possibility of an acquired myopathy was entertained. On laboratory evaluation, haemogram, blood counts and ESR were normal. Routine blood chemistry including blood sugar, KFT, serum electrolytes and lactate were in the normal range. LFT showed mildly elevated liver enzymes (AST/ALT/ALP- 322/111/79 U/L). Total CPK levels were significantly raised (14241 IU/L). Serum calcium, phosphorus, alkaline phosphatase and vitamin D levels were in the normal range. Blood examination was negative for HIV, hepatitis B surface antigen (HbsAg), antibodies to hepatitis C virus (anti HCV). Workup for vasculitic profile was negative for ANA, ds-DNA, p-ANCA, c-ANCA, RA factor. TFT revealed a grossly elevated TSH (816.5 mIU/ml) with decreased levels of FT3 (0.77pg/ml, FT4 (0.02 ng/dl) and markedly elevated titres of anti TPO antibody (2240 IU/ml) suggestive of hypothyroidism. ECG and X-ray chest were normal. Ultrasonography of the thigh and leg muscles revealed mild oedema but MRI did not reveal any significant pathology. Muscle biopsy showed some amount of individual muscle fibre necrosis and hyaline degeneration but no evidence of inflammation or muscular dystrophy. Electromyogram (EMG) showed no evidence of myotonia. The grossly elevated CPK and TSH levels confirmed the diagnosis of a hypothyroid myopathy. Presentation with pseudohypertrophy and stiffness of the lower limbs in the absence of any overt muscle weakness or any other systemic features of hypothyroidism was the odd feature in our case.

Discussion

Hypothyroidism is a common disorder with a prevalence of about 0.8 - 5% in the general population[2]. Clinical features of hypothyroidism are highly variable and depend upon the duration and severity of thyroid hormone deficiency besides the age of disease onset. It is frequently accompanied by neurologic manifestations including mental status changes, seizures, myopathy and neuropathy.

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Myopathy is usually common in patients with either severe or longstanding untreated disease. Although more than 60% of patients may have an elevated creatine kinase level, symptomatic muscle diseaseis uncommon[1].

The clinical features of hypothyroid myopathy include proximal weakness, cramps, aching or painful muscles, sluggish or hung up reflexes and myoedema. Muscle pain, particularly during and after exertion,is a prominent feature[3]. Increased muscle bulk, firmness and mounding (sustained focal contraction of skeletal muscles on striking with a reflex hammer) may also be seen. Our case presented with diffuse symmetric lower limb pseudohypertrophy and muscle stiffness in the absence of any weakness, exertional pain or overt systemic manifestations of hypothyroidism. Muscular hypertrophy with stiffness is seen in less than 10% cases of hypothyroidism[4] and there are only stray case reports of such cases in the Indian literature[5-8]. Sundarachari *et al*, reported a young male with proximal muscle weakness and hypertrophy[8]. Hypertrophy of almost all muscles of body, especially the calf and arm muscles has been reported by Chopra *et al*[7]. However, muscle hypertrophy and stiffness without any muscle weakness as the first presentation of hypothyroidism, as was observed in our case, is quite rare[4].

Hoffman's syndrome, a rare form of hypothyroid myopathy in adults, manifests with severe muscle stiffness, weakness, pseudohypetrophy and elevated levels of creatinine kinase, while Kocher-Debre-Semelaigne syndrome is the childhood equivalent of the same. Our patient had some features of the Hoffman syndrome but absence of proximal muscle weakness and dramatically raised CK levels (nearly 100 times the normal) were the odd points. Although, there are some reports of a polymyositis-like illness or rhabdomyolysis with significant elevations in CK levels, mild elevation (CK < 1000 IU/L) is more common in hypothyroid myopathy[9,10,11]. A grossly elevated level of CK (14241 IU/L) and TSH (816.5 mIU/ml) was observed in our case in the absence of any muscle weakness. Although correlation between elevated CK and TSH levels has been observed, these elevated levels do not necessarily correlate with the degree of muscle weakness, as was seen in our case also[1].

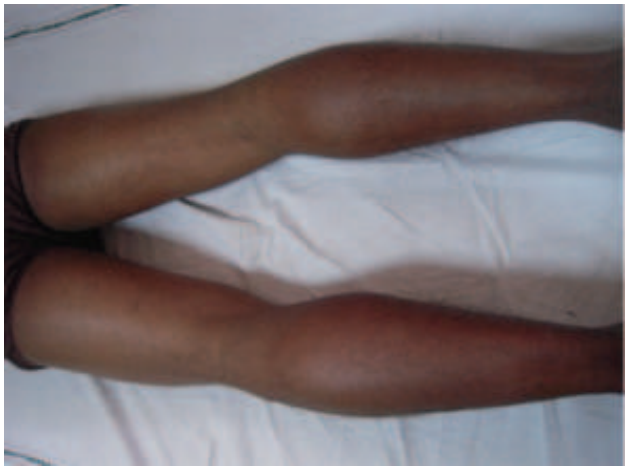

Fig. 1: *Bilateral symmetrical pseudohypertrophy of calf and thigh muscles.*

The pathogenesis of muscle involvement in hypothyroidism is not clearly defined. Deficiency of thyroid hormones probably affects the cellular function and energy metabolism. Abnormal glycogenolysis, mitochondrial oxidative metabolism and triglyceride turnover in muscles also play a role. These effects are reflected in selective atrophy of glycolytic type II fibres and compensatory hypertrophy of oxidative type I fibres. With severe or prolonged oxidative damage, muscle cell injury and rhabdomyolysis may occur, leading to very high CPK values. Symptoms of hypothyroid myopathy usually respond well to thyroid hormone replacement therapy. CPK levels fall within weeks, much before the normalisation of TSH levels, while clinical improvement can take upto 6 months.

In conclusion, muscle pseudohypertrophy and stiffness can be the sole presentation of hypothyroid myopathy. Thus, all patients with a myopathic disorder with or without systemic features of hypothyroidism, should be screened by thyroid function tests. A very high index of suspicion is required to rule-out this rare and treatable presentation of hypothyroid myopathy which can otherwise be misdiagnosed as polymyositis or muscular dystrophy.

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