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Hoffman's syndrome as initial manifestation of hypothyroidism a rare presentation: a Case report

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Abstract: Hoffman's syndrome is characterized by muscle stiffness and muscle pseudo hypertrophy in adult rarely a early manifestation of hypothyroidism. Here we present a 18 year-old adolescent diagnosed as Hoffman's Syndrome documented by elevated enzyme, clinical manifestations such as generalized weakness, cold intolerance, constipation, and hoarse voice, difficulty in walking and progressive enlargement of muscles of thighs and back with crampy pains for one years and electromyography findings of myopathy pattern.
Keywords: Hoffman's, hypothyroidism, dystrophy, myopathy, weakness

INTRODUCTION

Hoffmann's syndrome, first described by Hoffmann in 1897, is characterized by the presence of hypothyroidism with muscle stiffness and pseudo hypertrophy. Whereas muscle hypertrophy with weakness and slowness of movement in children is known as Kocher Debra Semelaigne syndrome [1, 2]. The common symptoms of myopathy due to hypothyroidism are proximal muscle weakness, muscle cramps, myoedema on percussion, delay in deep tendon reflexes and rarely development of muscle hypertrophy. Severity of myopathy generally correlates with the duration and the degree of thyroid hormone deficiency.3 The muscular hypertrophy with muscle stiffness is reported in less than 10% of hypothyroid patients .4 Here we present a case with Hoffmann's syndrome with characteristic muscle hypertrophy, documented enzyme assays and electromyography findings

CASE REPORT

An 18 year old adolescent boy presented with a history of weakness of all the four limbs for the past one year along with history of muscle cramps and weight gain. The weakness is more of proximal muscles than distal, and there was no history of seizure, fasciculation, sensory involvement, bowel/bladder disturbance. The clinical examination revealed mild periorbital puffiness, swelling of lips, enlarged tongue, dry coarse skin [figure 1a] and hoarse voice. His pulse rate was 58/minute and blood pressure was 128/94 mmHg. Neurological examination showed normal higher mental functions and cranial nerves. Motor system examination revealed muscle power of 4/5 in all four limbs with predominant proximal muscle weakness, normal muscle tone with generalized hypo reflexia and delayed ankle jerk. There was hypertrophy of calf muscles of both the lower limbs [Figure1b]. There was hypertrophy of back muscle, upper limb, and thigh muscle. [Figure 1c]. Sensory system examination and bowel, bladder functions were normal. The systemic examination was normal.

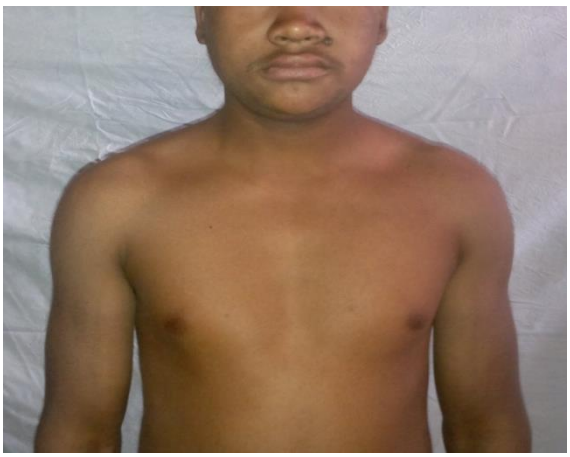

Fig 1a: Mild periorbital puffiness, swelling of lips

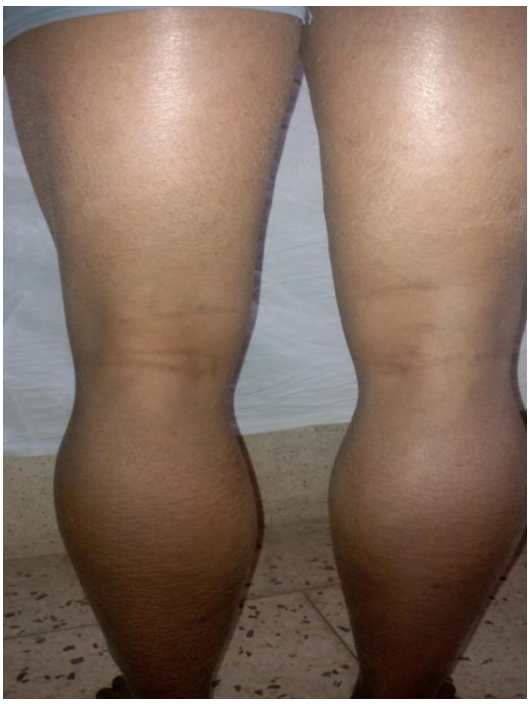

Fig 1b: Hypertrophy of thigh and calf muscle

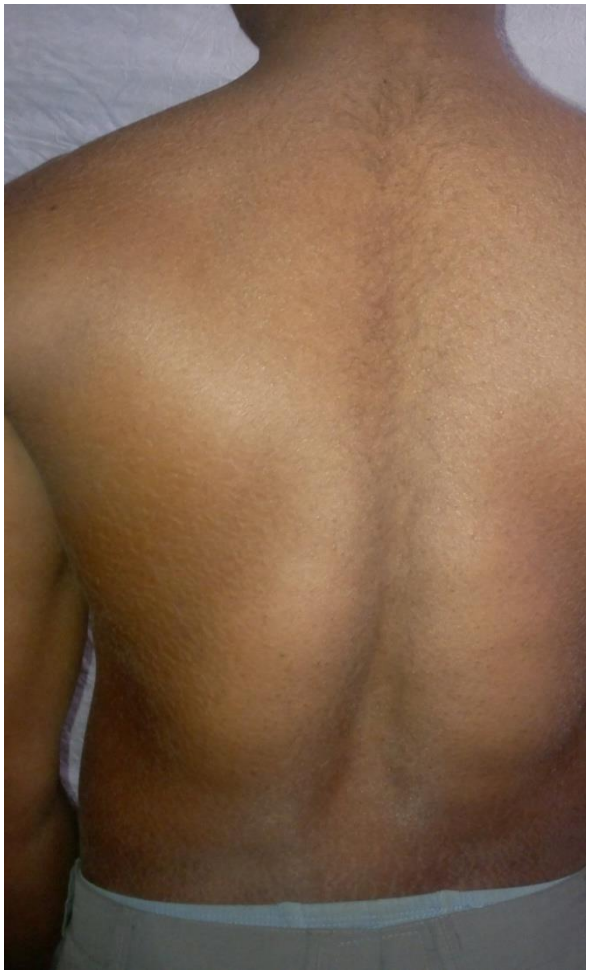

Fig-1c: Hypertrophy of back muscle

The laboratory investigation showed CPK of 3028 units/L (Normal <140u/l), Thyroid profile revealed T3 of 23 ng/dl (normal 60-200 ng/dl), T4 of 1.7 mg/dl (normal 4.5-12 mg/dl), TSH of 131.64mU/ml (Normal: 0.35- 5.50). Serology for anti-microsomal antibody was positive in high titres. The ultrasound imaging showed thyroiditis. Other pertaining investigation was within normal limit.

Electromyographic (EMG) evaluation showed mild spontaneous activity, polyphasic MUAPs with small amplitude and short duration in proximal muscles. The patient was treated with thyroid hormone replacement therapy. In follow up in OPD after 6 months after the treatment, the clinical and laboratory improvement of the patient seen. The enlargement of the muscle remarkably become normal [figure 2a] . There were no more complaints of fatigue, cramps, stiffness of the muscles and edema. [Figure 2b]. Patient did not give consent for biopsy.

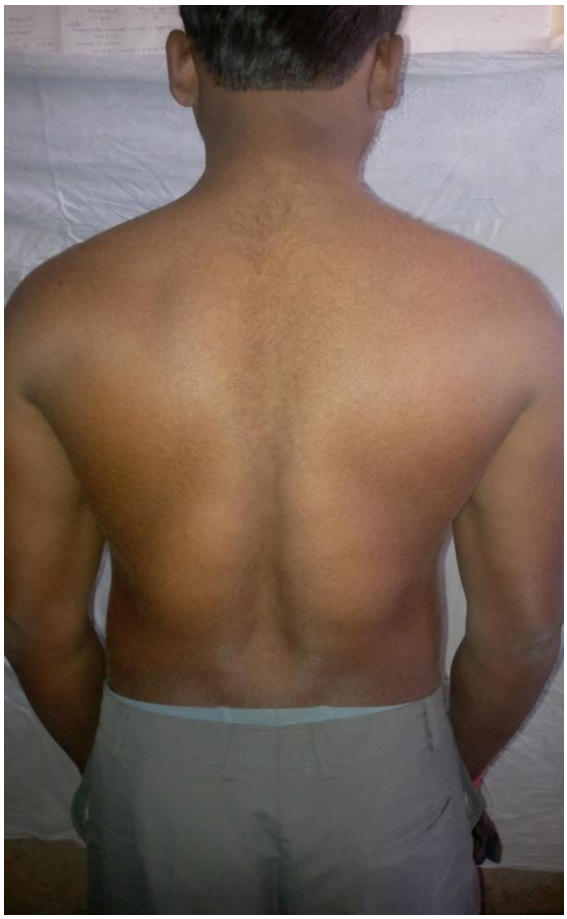

Fig-2a: Back muscle after treatment

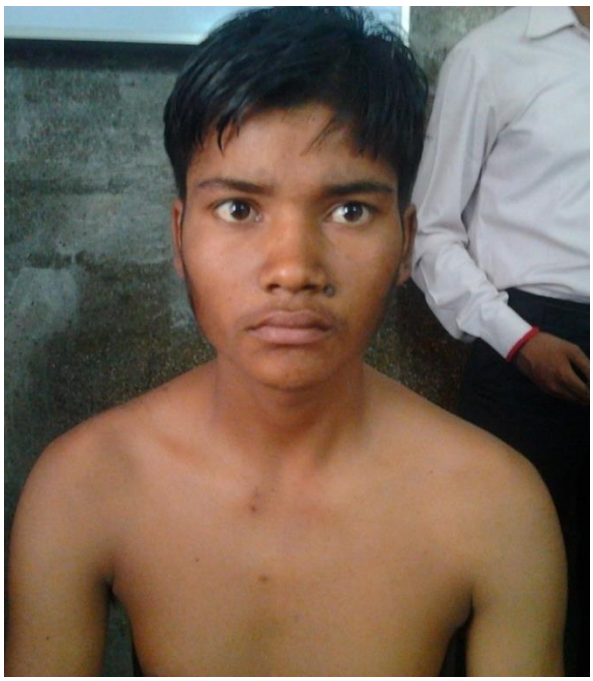

Fig-2b: fatigue, cramps, stiffness of the muscles and edema become normal

DISCUSSION

Neuromuscular symptoms present in 30-80% of patients with hypothyroidism. Patients may complain of muscle cramping, proximal symmetrical muscle weakness, muscle stiffness, and exercise intolerance [5]. The first presentation with myopathy and pseudo hypertrophy of muscles is very rare and unusual to see [6]. Muscle enlargement, stiffness, and cramping are a constellation of findings seen in individuals with hypothyroidism. In adults, these findings are known as Hoffman syndrome. In children, they are called Kocher-Debré-Sémélaigne syndrome. Hypothyroidism can impair mitochondrial metabolism, resulting in decreased muscle energy production. A low thyroid hormone level is the main causative factor [7]. The creatine kinase level can be very high (10-100 times greater than the normal level) in some patients, but it has no correlation with weakness but best biochemical marker of myopathies. The fall of the enzyme levels with the treatment occurs slowly form weeks to year [8].

The main etiology of primary hypothyroidism is Hashimoto's thyroiditis and Primary hypothyroidism accounts for 95% of the cases of thyroid insufficiency [4]. The etiology of pseudo hypertrophy in Hoffman's syndrome remains elusive and it has been postulated that the mechanisms involved could include an increase in connective tissue, increase in size and the number of muscular fibers also change in muscle fibers from fast twitching type II to slow twitching type-I fibers, abnormalities in oxidative enzymatic activity and hypertrophy due to accumulation of glycosaminoglycans. [9, 10, 11]. The muscle groups commonly involved are the tongue, arm and leg muscles. The electrophysiological study in hypothyroid myopathy may show findings compatible with neurogenic, myogenic, or a mix of those patterns. The EMG findings compatible with myogenic pattern are diminished duration, and amplitude of motor unit potentials [11]. The muscle hypertrophy and muscle weakness will disappear after treatment with thyroid hormones.

CONCLUSION

Hoffmann's syndrome is very rare condition and its initial presentation with myopathic manifestations is a diagnostic challenge as calf muscle hypertrophy can present in other diseases in this age group such as Duchenne and Becker's muscular dystrophy, Therefore clinician should cautious about this syndrome will lead to the correct and early diagnosis. It is also important to encultate that both the symptoms and serum levels of muscle enzymes return to normal with hormone replacement therapy.

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