# Pseudohypertrophic Myopathy Presenting As Initial Manifestation of Hypothyroidism – A Case Report

Talib S.H<sup>1</sup>, Kharche Jyoti<sup>2</sup>, Sainani Rahul<sup>3</sup>, Deshpande Shreyas<sup>4</sup>

<sup>1</sup>Professor & Head, <sup>2</sup>Associate Professor, <sup>3</sup>Chief Resident, <sup>4</sup>Senior Resident Department of Medicine, Mahatma Gandhi Missions Medical College & Hospital, Aurangabad. 431003

**Abstract:** We report a case with infrequent cause of myopathy related to hypothyroidism who presented to us with pseudohypertrophic mass on left calf with serological findings consistent with diagnosis of hypothyroidism. The syndrome of pseudohypertrophic etiology (Hoffman's syndrome) remains obscure. The case is discussed and reviewed briefly.

Keywords: Pseudohypertrophy, myopathy, hypothyroidism.

# I. Case Report:

A 42 years old male patient was hospitalized for diagnostic evaluation of muscle mass on left calf and myalgia for past 6 months, he also complained of difficulty in standing from sitting position and generalized weakness for the said duration.

On investigation patient had generalized hypotonia and muscle power of 4/5, proximal group of muscle affected more than the distal group, deep tendon reflexes were normal except ankle jerk which demonstrated delayed relaxation. Sensations were intact. The calf mass measured 15 x 10 cms, oval in shape, soft, non tender and the overlying skin was normal (Fig.1). He was pale, Hb was 9.5 gm%, peripheral smear was normocytic normochromic. The total counts were within normal limits, platelets were adequate, serum CPK was 979 IU/L (normal 15-130 IU/L), Patient had no apparent thyroid swelling. The thyroid profile revealed FT3 2.5 pg/ml (normal 1.5-5 pg/ml), FT4 level 0.2 ng/dl (normal 0.6-2 ng/dl) and TSH level 126 IU/ml (0.5-4.6 IU/ml), serology for anti TPO Ab was 28 IU/ml (normal <35 IU/ml). His lipid profile was within normal limits. Roentgenography of chest was normal and ECG revealed low voltage complexes with sinus bradycardia. Cardic 2-D ECHO with Doppler did not reveal any abnormality, the ejection fraction was 60%. USG and MRI of the left leg revealed oedematous medial head of 3 gastrocnemius with hypertrophy (Fig.2). Electromyography (EMG) revealed diminished recruiting of motor units, consistent with hypothyroid status. Patient denied consent for the muscle biopsy. Patient kept on thyroid hormone therapy (T.Eltroxin 100 µg) after 3 weeks of thyroid hormone therapy the patient experienced improvement of muscular cramps, 3½ months later the size of mass regressed considerably. Progressive decline in the levels of muscular enzymes were recorded. The presence of pseudohypertrophic form and hypothyroidism in the case with increased muscle enzymes established the diagnosis of Hoffman's syndrome in the case.

### II. Discussion:

Hoffman's syndrome is an entity where in patient is having muscle hypertrophy, muscle weakness complicating with hypothyroidism. The muscular symptoms are common in hypothyroid and may range from 30-80%.1 The major symptoms related are myalgia, weakness and cramps in majority of patients. Muscular hypertrophy with muscle weakness reported in <10% of patients.1,2 Hoffman's syndrome is considered as a rare specific form of hypothyroid myopathy. Calf muscle hypertrophy accompanies wide variety of diseases like Duchenne and Becker's muscular dystrophy, sarcoid granuloma, amyloid deposits etc. In hypothyroidism changes in muscle fibres of type-2 due to accumulation of glycosaminoglycans are typically seen in gastrocnemius which is

almost always involved in majority of such patients as in our case.

The thigh, arm, forearm muscles may also be involved producing pseudomyotonic reflexes (delayed muscle relaxation), the calcium ATPase activity of fast twitching type-2 muscle fibres is decreased in hypothyroidism resulting in pseudomyotonic reflex i.e. delayed relaxation. Further hypothyroidism should always be considered as a differential diagnosis of creatinine kinase elevation. Ono et al in 1987, postulated that in Hoffman's syndrome, the mechanisms involved also included increase in muscle tissue, size and number of muscle fibre.4

The myopathy associated with hypothyroidism could be of four types. Kocher-deber-senelaigne (KDS syndrome), Hoffman's syndrome, atrophic form, myasthenic syndrome.5 KDS syndrome is reported in infants

with cretinism, with hypertrophic muscular weakness. The other less frequent presentations are atrophic myopathy and myasthenic syndrome. Myopathy in some may be recorded as the initial manifestation of hypothyroidism as noted in this case.

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## **FIGURES:**



Figure-1: Showing calf swelling left leg.



Figure-2: MRI of left leg showing oedematous medial head of gastrocnemius with hypertrophy