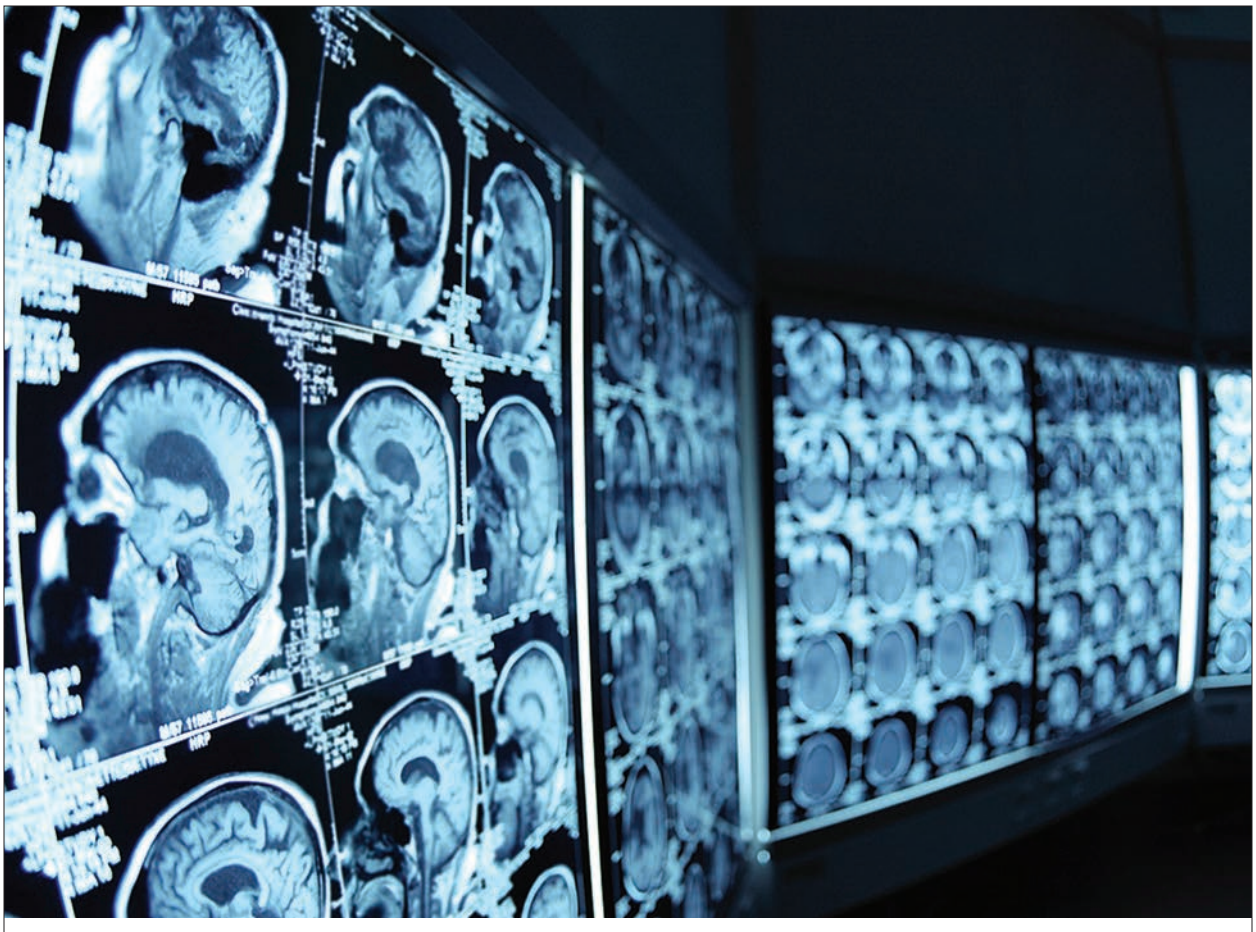

Research Journal of Radiology

ISSN 1819-348X

Academic Journals Inc.

www.academicjournals.com

OPEN ACCESS Research Journal of Radiology

ISSN 1819-348x
DOI: 10.3923/rjr.2017.1.4

Case Report
Hoffmann Syndrome: An Unusual Cause of Proximal Muscle Hypertrophy

Anees Dudekula, Merwyn Fernandes, Ajit Mahale and Hitakshi Sharma

Department of Radiodiagnosis, Kasturba Medical College and Other Affiliated Hospital, Manipal University, Attavar Mangalore, 575 001, Karnataka, India

Abstract
Hoffmann syndrome is a specific and very rare form of hypothyroid myopathy occurring in individuals of long standing hypothyroidism in which proximal muscle weakness, muscle stiffness and pseudohypertrophy is seen. The etiology of pseudo hypertrophy in Hoffman's syndrome remains obscure and myopathy may be only clinical findings in these patients to indicate hypothyroidism. These patients are often misdiagnosed as muscular dystrophies of other etiology, so special awareness about this syndrome can lead to accurate diagnosis as this is an easily treatable condition.

Key words: Hypothyroid myopathy, pseudohypertrophy of muscle, hypothyroid

Received: July 06, 2015 Accepted: October 12, 2015 Published: December 15, 2016

Citation: Anees Dudekula, Merwyn Fernandes, Ajit Mahale and Hitakshi Sharma, 2017. Hoffmann syndrome: An unusual cause of proximal muscle hypertrophy. Res. J. Radiol., 4: 1-4.

Corresponding Author: Anees Dudekula, Department of Radiodiagnosis, Kasturba Medical College and Other Affiliated Hospital, Manipal University, Attavar Mangalore, 575 001, Karnataka, India

Copyright: © 2017 Anees Dudekula *et al.* This is an open access article distributed under the terms of the creative commons attribution License, which permits unrestricted use, distribution and reproduction in any medium, provided the original author and source are credited.

Competing Interest: The authors have declared that no competing interest exists.

Data Availability: All relevant data are within the paper and its supporting information files.

INTRODUCTION

Hoffmann syndrome is a specific and rare form of hypothyroid myopathy in which proximal muscle weakness, muscle stiffness and pseudohypertrophy occurs in adults[1]. Clinically evidence of hypothyroid myopathy has been reported in 20-88% of patients. The commonly observed symptoms are proximal weakness, cramps, painful muscles, myoedema on percussion, delay in deep tendon reflexes and development of muscle hypertrophy[2]. Johann Hoffmann first desribed this entity in 1897[3]. Various forms of hypothyroid myopathy have been described chiefly atrophic form, Myasthenic Syndrome, Hoffmann's Syndrome, polymyositis-like Syndrome and Kocher-Debre-Semelaigne Syndrome that is described in infants with cretinism[4,5]. Clinically significant muscle disease develops in patients with severe and longstanding untreated hypothyroidism. Myopathy may be only clinical findings in a few patients suffering from hypothyroidism. Pseudohypertrophy and muscle stiffness is seen in less than 10% hypothyroid patients[6]. These patients are often misdiagnosed as muscular dystrophies of other etiology, so special awareness about this syndrome can lead to accurate diagnosis as this is an easily treatable condition.

Clinical scenario: A 39 year old male patient presented to the medicine department with history of swelling of bilateral proximal calf since 1 month. It was associated with cramp like pain. The patient was a quarry worker and the pain was severe enough to incapacitate him from his work. Patient had no history of hoarseness of voice, constipation or difficulty in walking. No significant illness in the past. No family history of similar complaints.

Patient was referred to the department of radiodiagnosis for evaluation of bilateral leg swelling with initial clinical suspicion of chronic deep vein thrombosis.

Radiographs anteroposterior and lateral views of the calf muscles shower no evidence of bony neoplasm. There was no cortical thickening of the bone nor any erosions. There was soft tissue enlargement in the posterior compartment of the proximal leg with smooth distal tapering. No areas of altered radio density. No blurring of fat planes.

Ultrasonography with doppler examination revealed symmetrical enlargement of the bilateral proximal calf muscles (Fig. 1a-c). No evidence of increased vascularity. No evidence of altered echogenic areas within the muscles to suggest mass lesions. Doppler examination revealed no thrombosis of the deep and superficial veins in the entire bilateral lower limbs.

Computed tomography of the bilateral legs revealed symmetrical hypertrophy of the gastrocnemius muscles (Fig. 2). No altered density within the muscles, no mass lesions and no altered densities in the visualized bones (Fig. 3). No abnormalities in rest of the muscles and neurovascular bundles.

Correlation with lab investigations revealed altered thyroid function. Thyroid Stimulating Hormone (TSH) was markedly elevated with values of 83 μL U mL^{-1}

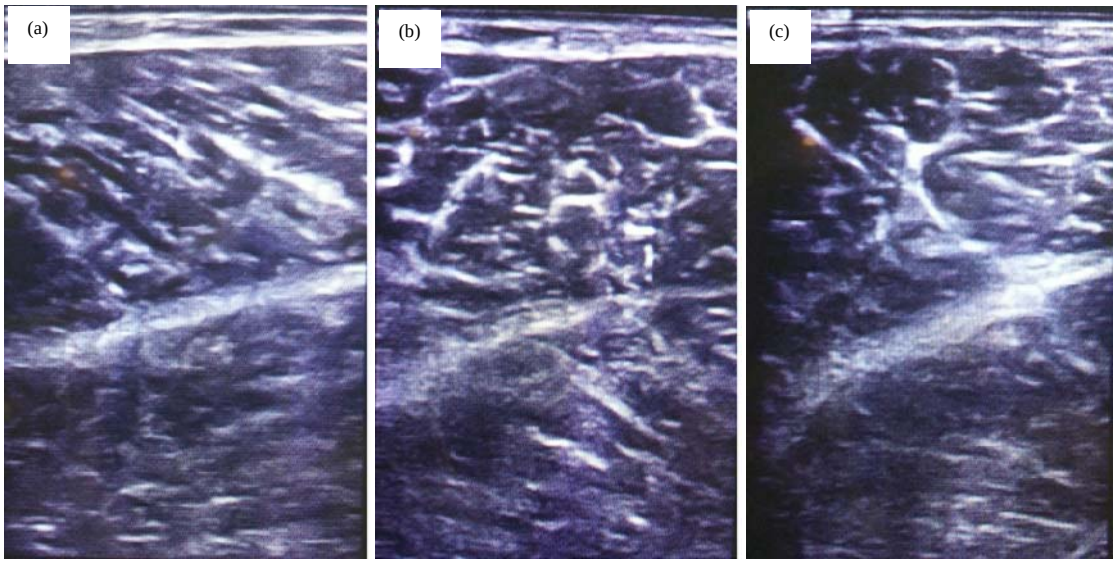

Fig. 1(a-c): Enlargement of the (a) Right calf muscles and (b and c) Left calf muscles with no altered echogenic areas

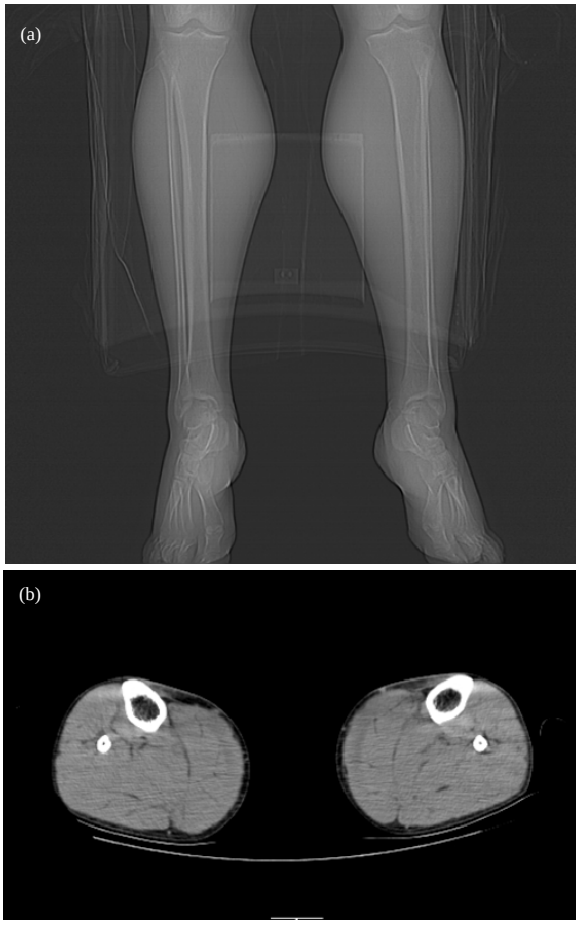

Fig. 2(a-b): Computed tomography scanogram and soft tissue window showing symmetrical enlargement of bilateral gastrocnemius muscle with no areas of altered density

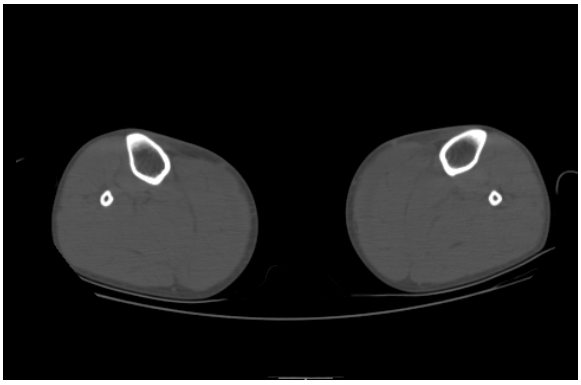

Fig. 3: Computed tomography one window showing no areas of altered density in the bilateral tibia and fibula. no evidence of cortical breach or mass lesion

(normal-0.27-4.2), T3 and T4 levels were low. Taking the factors of clinical hypothyroidism and no mass lesions or deep vein thrombosis a diagnosis of hypothyroid myopathy suggestive of Hoffmann syndrome was made. Patient was started on 150 µg thyroxin. After 3 months of follow up there was decrease in the patient symptoms and calf muscle hypertrophy. Repeat investigations showed drop in the levels of TSH as compared to pre treatment levels.

DISCUSSION

Hoffmann's syndrome was first described by Hoffman in 1897 in an adult who had difficulty in relaxation of muscles and developed muscle stiffness after thyroidectomy. First presentation of hypothyroidism as muscle hypertrophy is very rare. Our case showed only myopathy with no systemic manifestations of hypothyroidism[1].

Hoffman's disease is a rare form of hypothyroid myopathy in which because of deposition of connective tissue there is proximal weakness and pseudohypertrophy of muscles. It seems most likely that thyroid deficiency affects muscle cells directly. Primary hypothyroidism accounts for 95% of cases of thyroid insufficiency. The main etiology is Hassimoto's thyroiditis which is an auto immune chronic thyroiditis characterized by high levels of thyroid peroxidase antibodies (TPOAb) and thyroglobulin antibodies (TgAb)[7]. The etiology of pseudo hypertrophy in Hoffman's syndrome remains obscure and is a matter of discussion. Less than 10% of the patients with hypothyroidism present with muscular hypertrophy and muscle stiffness. Various theories have been postulated to explain myopathy seen in hypothyroidism which includes an autoimmune reaction affecting the muscle, infiltration by "Myxedema", or a disorder of muscle membrane. Another assumed mechanism states increase deposition of glycosaminoglycans, increase in muscle fiber size and number as the reason for muscle hypertrophy. It is generally found to be associated with change in muscle fiber type from fast twitch type II to slow twitch type I along with oxidative muscle enzyme activity alteration and decreased activity of calcium ATPase. It is observed that creatinine phosphokinase (CPK) levels are elevated in approximately 70-90% of the cases of thyroid myopathy, few cases show 10-100 times increase than the normal levels, however, no correlation between the CPK levels and degree of weakness was established. Thyroxine has significant well known effects on the cell membranes. In hypothyroidism the serum enzymes may rise, presumable due to thyroid deficiency as that permits leakage across muscle membranes and actual muscle necrosis[8].

Hoffman's disease is a specific, rare form of hypothyroid myopathy that occurs due to connective tissue deposition resulting in proximal weakness and pseudohypertrophy of muscles. Various theories have been postulated to explain myopathy seen in hypothyroidism such as an autoimmune reaction affecting the muscle, infiltration by "Myxedema", or a disorder of muscle membrane. The muscle cells are affected directly by the thyroid deficiency. Less than 10% of the patients with hypothyroidism present with muscular hypertrophy and muscle stiffness[9].

REFERENCES

1. Tuncel, D., A. Cetinkaya, B. Kaya and M. Gokce, 2008. Hoffmann's syndrome: A case report. Med. Principles Pract., 17: 346-348.
2. Oztekin, F., N. Oztekin, O. Yılmaz and E. Unlu, 2005. A case report of Hoffmann's syndrome. J. Neurol. Sci. (Turk.), 22: 340-343.
3. Qureshi, W., G. Hassan, G.Q. Khan, S.M. Kadri and M. Kak *et al*., 2005. Hoffmann's syndrome: A case report. GMS German Med. Sci., Vol. 3.
4. Shams, N., F. Niaz and M.H. Osmani, 2013. Hoffman syndrome: Rare presentation of hypothyroidism. J. Liaquat Univ. Med. Health Sci., 12: 214-217.
5. McKeran, R.O., G. Slavin, P. Ward, E. Paul and W.G.P. Mair, 1980. Hypothyroid myopathy. A clinical and pathological study. J. Pathol., 132: 35-54.
6. Senanayake, H.M., A.D. Dedigama, R.P. De Alwis and K. Thirumavalavan, 2014. Hoffmann syndrome: A case report. Int. Arch. Med., Vol. 7. 10.1186/1755-7682-7-2
7. Praveen, K., S. Aslam and T.K. Dutta, 2011. HoffMann's syndrome: A rare neurological presentation of hypothyroidism. Int. J. Nutr. Pharmacol. Neurol. Dis., 1: 201-203.
8. Uddin, M.N., A. Uddin, M.A.U. Chy and T. Khan, 2014. Hoffmann's syndrome a presenting manifestation of hypothyroidism. Am. J. Med. Case Rep., 2: 75-77.
9. Sundarachari, N.V., A. Sridhar and V.P. Lakshmi, 2013. Rare yet treatable: Hypothyroid myopathy (Hoffman's syndrome). J. Dr. NTR Univ. Health Sci., 2: 203-204.