Case Report
Masseter Muscle Hypertrophy and Pericardial Effusion in Kocher-Debre-Semelaigne Syndrome Child

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Citation

Abstract: Muscular pseudohypertrophy associated with severe congenital hypothyroidism has been described as Kocher Debre Semelaigne syndrome, which is a rare disorder. We report a case of 9 year old female child with hypothyroidism, limb muscular pseudo-hypertrophy with involvement of masseter muscle along with pericardial effusion in Kocher-Debré-Semelaigne syndrome.

Key Words: Hypothyroidism, Kocher Debre Semelaigne Syndrome, Pseudohypertrophy, Pericardial effusion

Introduction:
Kocher-Debre-Semelaigne syndrome (KDS) is a rare association of muscular pseudohypertrophy and myopathy in long-standing severe hypothyroidism in the paediatric age group. Hypothyroidism is common in countries where the neonatal screening is not advised for early detection of congenital thyroid deficiency.(1-2) It occurs more commonly in males, and has been reported in children who are the products of consanguineous marriages. The manifestations vary from general symptoms like loss of interest, loss of appetite, constipation to growth failure and mental retardation. Pericardial effusion is known in hypothyroidism but rarely reported with KDSS. Here we present case of KDSS with hypertrophy of the masseter muscle and pericardial effusion.

Case Report
A 10 years old female child product of a non-consanguineous marriage presented with swelling in both parotid regions and not gaining height for last 1 year. On asking detailed history, the child was not playful, take little interest in the surrounding. She had poor appetite, constipation, passing hard stools, less physical activity, and cold intolerance. Her milestones were normal with very good scholastic performance. Parents noticed that her weight and height is less as compared to the other children of same age group. The swelling in the both parotid regions seemed to be growing gradually over a period of one year. There was no history of fever, any local pain and tenderness, difficulty inswallowing, recurrent respiratory infection, hard of hearing, breathlessness and pain in chewing movements.

On general examination, pulse rate was 64 beats/min, blood pressure 80/60 mmHg and body temperature 97.4 degree F. Her weight was 19 kg (less than 3rd percentile) and height was 114 centimetres (less than 3rd percentile) with upper segment and lower segment ratio of 0.8. She had paller, hypertelorism, depressed nasal bridge, thick lips, hypertrophied tongue, and dry rough skin, short neck and umbilical hernia. There was no tender swelling at both angles of mandible, extending to arch of mandible and angle of lips (Figure 1).

On cardiovascular system examination, the apex beat was in the left 5th intercostal space in the midclavicular line. Heart sounds were muffled and there was no murmur. On central nervous system examination, the child had normal sensorium, the tone was normal with prominent calf muscles (Figure 2) with proximal muscle weakness in the lower limbs (muscle power being 4/5, distal muscle power being normal). The deep tendon reflexes were diminished and the plantar responses were flexor. The sensory examination was normal. Other systemic examination was normal. In presence of features of hypothyroidism, the pseudohypertrophy of muscles suggested a diagnosis of KDSS.
The hemogram revealed that the hemoglobin level was low at 9 gm/dl. A peripheral smear showed normocytic, normochromic red blood cells, 48% neutrophils, 38% lymphocytes, 14% monocytes. RBC count: 2.87 million/µl, hematocrit: 26%, MCV: 91 µm², MCH: 29 pg, MCHC: 30%, RDW: 48%, WBC count: 5,900/µl, platelet count: 175,000/µl. Liver function test and renal function test were normal. Serum cholesterol was 320 mg/dl, serum phosphate 4.5mg%, serum calcium 8gm% and serum amylase was 84U/L. Thyroid function tests showed total T3 < 0.26 ng/ml (normal 0.60-1.51ng/ml), total T4 <0.59 microgm /dl (normal 4.66-9.33 microgm/dl), TSH >60 microIU/ml (normal 0.25-5.0 microIU/ml), which confirmed the diagnosis of hypothyroidism.
Bone age found to be 6 years on wrist x-ray. Creatine kinase activity was also normal. The electromyography findings were suggestive of myopathy. Ophthalmology evaluation revealed no evidence of nystagmus. X-ray chest shows cardiomegaly (CT ratio 0.6 with normal pulmonary vasculature. Electrocardiography showed low voltage with sinus bradycardia. Transthoracic echocardiography was suggestive of moderate pericardial effusion (Figure 3) without evidence of cardiac tamponade. Ultrasonography office and neck revealed hypertrophy of masseter muscle and normal thyroid gland. A diagnosis of KDSS with atypical finding was made on the basis of the above mentioned findings. The child was started on levothyroxine supplementation, 75µg/day.

Discussion:
In 1892, the KDSS was first reported by Emil Theodore Kocher. In 1935 Robert Debre and Georges Semelaigne reported association of muscular pseudo hypertrophy (1). It is also known as hypothyroid myopathy, cretinism-muscular hypertrophy, myopathy-myxedema syndrome, hypothyroidism-large muscles syndrome, hypothyrotic muscular hypertrophy in children, or myxedema-muscular hypertrophy syndrome (2-3). The condition is usually seen in the age group 3 to 10 years, however, there have been rare reports of cases presenting as early as in the neonatal period (4-5). KDSS is commonly seen in males and presented in children with consanguineous parents which has been suggestive of an autosomal recessive inheritance (1). However, the present case was a female and her parents had a non-consanguineous marriage, thus labelling this case very rarer. The underlying thyroid effect may vary from both congenital (Athyreosis, enzyme synthesis defects) or acquired (autoimmune) forms of hypothyroidism (1,6). The histology is not contributory. The light microscopic features are variation of fibre size and hyalinisation of muscle fibres (2-3).

The pathogenesis of the pseudo hypertrophy in KDSS is not well understood. While the lack of thyroid hormone for a prolonged period of time impairs metabolic functions of the body including musculoskeletal system, impaired carbohydrate metabolism leads to accumulation of glycogen in the muscle and increased connective tissue amount and deposits mucopolysaccharide in the muscle gives the muscle hypertrophy appearance. Also, there is shift of fast twitch muscle fibre to slow twitch fibre which leads to slow muscle contraction and relaxation. Prolonged muscular contraction due to delayed calcium re-uptake by the sarcoplasmic reticulum is another factor responsible for muscular hypertrophy (3,7). The pathogenesis of pseudo hypertrophy in our case could be the result of long standing hypothyroidism. The child usually has florid symptoms of hypothyroidism, severity and duration usually correlates directly with the muscle pseudohypertrophy. It involves muscles of extremities, limb girdle, trunk, hands and feet, and tongue but it is more prominent in muscles of limbs hence giving the athletic look to the patient (2-4), similar look was also present in our case. The other striking feature was the hypertrophy of the masseter muscle of the face which is very uncommon.

In myxedema, pericardial effusion may be a common manifestation in, but is a rare association of hypothyroidism in an early mild stage (8). The increased capillary permeability and impaired lymphatic drainage with subsequent leakage of protein into the interstitial space, resulting in pericardial effusion which is related with hypothyroidism (9). The pericardial effusion in hypothyroidism appeared to be dependent on the severity of the disease. The gradual accumulation of fluid and the remarkable distensibility of the pericardium suggestive of the rarity of cardiac tamponade in pericardial effusion patient (9). Kabadi et al (8) mentioned that the incidence of pericardial effusion in hypothyroidism was only 6%. Dharaskar et al (10) reported an unusual case of KDSS with presence of pericardial effusion with complete resolution and clinical improvement in response to thyroxin replacement. This is a very rare report of pericardial effusion and KDSS together. The common differential diagnosis of hypertrophied muscle includes Duchene muscular dystrophy. The elevation of creatinine phosphokinase level (CPK) levels is high in the proportion in muscular dystrophies (7), whereas the CPK level was normal in our case. The clinical feature of hypothyroidism, the muscular pseudohypertrophy as well as pericardial effusion revert back to normal in due course of time after thyroxine supplementation (3,7), except that the height may still be short.

Conclusion
The classic presentation of hypothyroidism is easily recognised. But rarely patients may present with rare manifestations making the diagnosis difficult thus delaying the management. By reporting this case we try to bring the awareness about rare presentation of hypothyroidism of the masseter muscle and pericardial effusion in KDSS in a female child who was a product of non-consanguineous marriage.

References