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Case Report

KOCHER DEBRE SEMELAIGNE SYNDROME – A CASE REPORT

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ABSTRACT

We report a case of 11 year old female child with Kocher Debre Semelaigne Syndrome (KDSS). It is a rare clinical presentation of Hypothyroidism with muscle pseudo-hypertrophy. There are many myopathies associated with hypothyroidism, KDSS being one and important differential diagnosis. Pseudo muscular hypertrophy of KDSS is an acquired type of myopathy associated with severe and long standing hypothyroidism and is reversible with thyroxine supplement.

Key Words: *Hypothyroidism, Myopathy, Kocher Debre Semelaigne Syndrome, Pseudohypertrophy*

INTRODUCTION

Kocher Debre Semelaigne Syndrome (KDSS) is a rare association of muscular pseudohypertrophy and long standing moderate to severe hypothyroidism in children (Tull *et al.,* 2003). Myopathy associated with hypothyroidism classically presents with fatigue, exertional pain, slow movement, diminished deep tendon reflexes, stiffness, myalgia, myxoedema, proximal weakness and less commonly cramps (Mehrotra *et al.,* 2002). The condition is rare in countries with screening programmes. Among patients with myopathy associated with hypothyroidism less than 10% patients develop pseudomuscular hypertrophy known as KDSS.

CASES

A eleven years old female born of non-consanguineous marriage residing at Saharanpur (U.P. India), presented to the Department of Pediatrics, Maharishi Markandeshwar Medical College and Hospital with history of lethargy, feeling of coldness, constipation, dry skin, clumsiness, hoarseness of voice, decreased sleep and poor social interaction since the age of six years. An increasing swelling in neck region was noticed since two years. Her birth history was normal and a similar family history was lacking.

On examination, pulse rate was 84 beats/min, blood pressure 80/60 mmHg and body temperature 97 degree F. Her height was 109cms (Expected is 144cms) and weight was 22kgs (Expected is 37.4kgs). She was having pallor, swelling was present over neck in the midline which was having smooth surface, was firm in consistency and not moving on swallowing (figure 2).

Her SMR staging was Breast-Stage II and Pubic Hair-Stage I. Hirsutism was present over forehead. There were coarse facial features with protruding tongue, odematous face and depressed nasal bridge and pouting lips (figure 1). Extremities were cold and dryness all over the skin. On interaction with the child hoarseness of voice was also confirmed.

On CNS examination, her movements were slow. There was hypertonia with a power of 4/5 in all four limbs. The sensory system examination was normal. The gait was stiff and wide based with the planter flexors. The deep tendon reflexes were brisk. She had impression of hypertrophy of neck muscles. The muscle had a firm feel. Rest of the systemic examination was normal. A suspicion of hypothyroidism with presentation of hypertrophy of muscles classically visible over the neck region was considered and further evaluated.

Due to monitory constraints only few relevant investigations were carried out which revealed the following: Hemoglobin 9.6 gm/dl, Total leukocyte count 10,800/mm3, MCV 88.2 Fl, MCH 28.9 pg with

normocytic and normochromic red blood cells on peripheral blood smear. Thyroid profile: T3 was 0.40 mg/dl, T4 3.0 mg/dl and TSH was 33.3 uIU/ml. EMG and NCV studies showed demyelinating type of polyneuropathy and myopathic muscles. USG neck revealed hypertrophy of neck muscles and normal position of thyroid gland. Child was treated with thyroxine replacement as 75 microgram once a day (ELTROXIN) and multivitamins and Iron and Calcium. She showed a rapid clinical improvement within few weeks. There was improvement in symptoms and regression in muscle hypertrophy was seen.

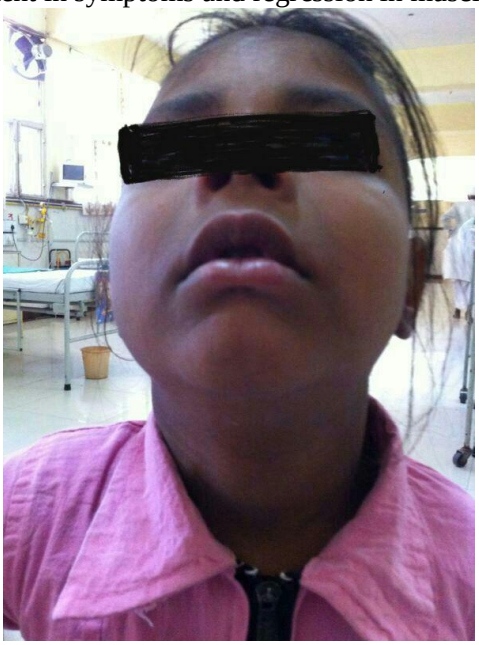

Figure 1: Photograph showing the typical facial features

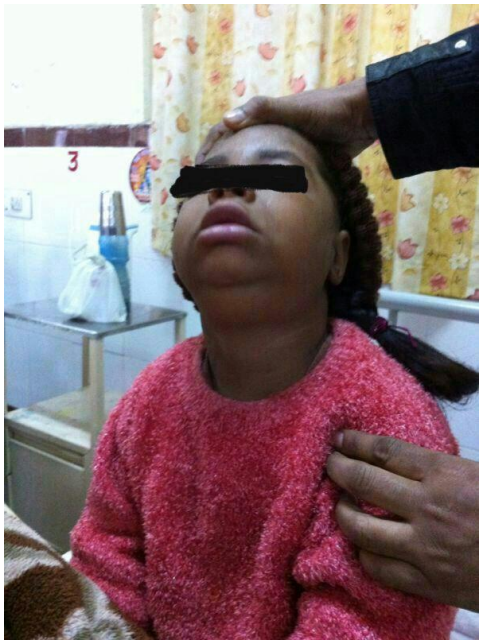

Figure 2: Photograph showing the midline neck swelling and hypertrophy of neck muscles

DISCUSSION

Kocher in 1892 and Debre and Semelaigne in 1935 described a syndrome of muscular hypertrophy associated with delayed muscle relaxation (Kung *et al.*, 1987).

The pathogenesis of muscular pseudohypertrophy remains obscure and is believed to be due to the long standing effects of hypothyroidism on the muscle fibres. Lack of thyroid hormone impairs many metabolic functions of the body including musculoskeletal system. Impaired carbohydrate metabolism lead to Glycogen accumulation in the muscle. Increased amount of connective tissue and mucopolysaccharide deposits in the muscle also gives the appearance of hypertrophy of muscles. In hypothyroidism shift of fast twitch muscle fiber to slow twitch fiber leads to slow muscle contraction and relaxation as seen in KDSS.

KDSS involves muscles of extremeties, limb girdle, trunk, hands and feet, being more prominent in muscles of limbs hence giving the athletic look to the patient (Tull *et al.*, 2003). It may be mistaken for Duchenne Muscular Dystrophy leading to delay in the treatment. KDSS is usually seen the age group of 18 months to 10 years. The usual features are of hypothyroidism and the degree of short stature and severity of hypothyroidism is variable and correlates directly with the severity of the muscle pseudohypertrophy. The underlying thyroid effect may vary from both congenital and acquired forms of hypothryoidsm.

Cause of hypothyroidism in our case could not be found for reasons known but clinical analysis and relation of the pseuhypertrophy with positive response to the valid treatment lead to the confirmation of the diagnosis. The pseudohypertrophy is most striking in neck muscles in this case reported by us. The signs and symptoms of hypothyroidism as well as the muscular pseudohypertrophy revert back to normal in due course of time after initiation of thyroxine supplementation. A well planned and graded physiotherapy programme may be beneficial in getting rid of the muscle stiffness and achieving full potential for the muscle strength.

An unusual case presentation of KDSS with presence of pericardial effusion with complete resolution of the pericardial effusion and clinical improvement in response to thyroxine replacement has also been reported (Dharaskar *et al.*, 2007).

Conclusion

Hypothyroidism presenting with classic manifestations of lethargy, cold intolerance, hoarseness of voice, dry skin, and constipation is easily recognized and therefore easy to diagnose and treat. But occasionally patients may present with atypical and rare manifestations making the diagnosis less apparent thus delaying treatment. By reporting this case we intend to bring the awareness about this unusual presentation of hypothyroidism in the form of Kocher-Debre-Semelaigne syndrome which is a rare and a reversible condition.

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