A case of Kocher-Debre-Semelaigne syndrome and ichthyosis secondary to acquired hypothyroidism

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Introduction

Kocher-Debre-Semelaigne (KDSS) syndrome is a form of muscular pseudo-hypertrophy due to hypothyroidism. Kocher first described it in 1892, but Debre and Semelaigne linked hypothyroidism and muscular hypertrophy in 1935[1]. KDSS has a 2:1 male preponderance and is commoner in offspring of consanguineous parents[2]. Ichthyosis is a rare association of longstanding hypothyroidism[3]. KDSS due to congenital hypothyroidism has already been published in Sri Lanka[4]. However, no paediatric cases of KDSS secondary to acquired hypothyroidism have been reported in Sri Lanka.

Case report

An 8-year-old boy presented with a history of dry skin and constipation of 3 years duration. He was the first child born to consanguineous parents at term following normal vaginal delivery with a birth weight of 2.2 kg. He had a normal newborn hypothyroidism screening test. He was well until five years of age when he developed dry, scaly skin, for which he was followed up at a dermatology clinic. He had age-appropriate development in early childhood. However, his mother noted a significant deterioration in his school performance. In addition, he has been having constipation for the last two years, for which he was on laxatives from time to time. There was significant easy fatiguability, and he got calf pain with minimal exertion. He found it difficult to stand from a squatting position and had difficulty climbing staircases.

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On examination, the child had short stature with an upper to lower segment ratio of 1.2:1.0. His height was below the mid parental height. His weight and head circumference were at the 50th and 75th centile respectively. There was generalized dry, scaly skin and icthyosis mainly over the extremities (Figure 1). Muscular athletic built with a mild degree of calf hypertrophy was noted. His pulse rate was 74 beats/minute, and his blood pressure was 90/60 mmHg. The rest of his cardiovascular system was normal. He had a protruded abdomen with palpable faecal masses without hepatomegaly or splenomegaly. Neurological examination showed lower limb power of 4/5 with slow relaxing ankle jerk bilaterally. There were no focal neurological signs. Mild homogeneous enlargement was noted over the thyroid region.

His basic haematological investigations were normal. The blood picture showed macrocytosis without hypersegmented neutrophils. Serum thyroid stimulating hormone was >100 mIU/L (reference range 0.55–5.31 mIU/L), and thyroxine level was 0.8ng/dL (reference range 6.0 to 12.8ng/dL). The neck ultrasound scan revealed an enlarged thyroid gland with homogeneous echogenicity with increased vascularity suggestive of thyroiditis. His aspartate transaminase level was 53U/L (Reference range 0-45U/L), and the serum creatine phosphokinase level was 1161 U/L (Reference range 0-171 U/L). His bone age was compatible with four years (Figure 2).

There were no abnormalities detected in his renal and liver functions. Two dimenstional echocardiogram (2D Echo) did not show any pericardial effusions. Thyroid peroxidase antibodies were not done due to financial constraints. A diagnosis of acquired hypothyroidism complicated by hypothyroid muscular pseudohypertrophy was made. Subsequently, he was commenced on 75µg of Levothyroxine and limb physiotherapy. He became clinically and biochemically euthyroid within four weeks of treatment. His follow-up CPK level dropped to the normal range within 2 months. A marked improvement of the ichthyosis was noted after one month of treatment.

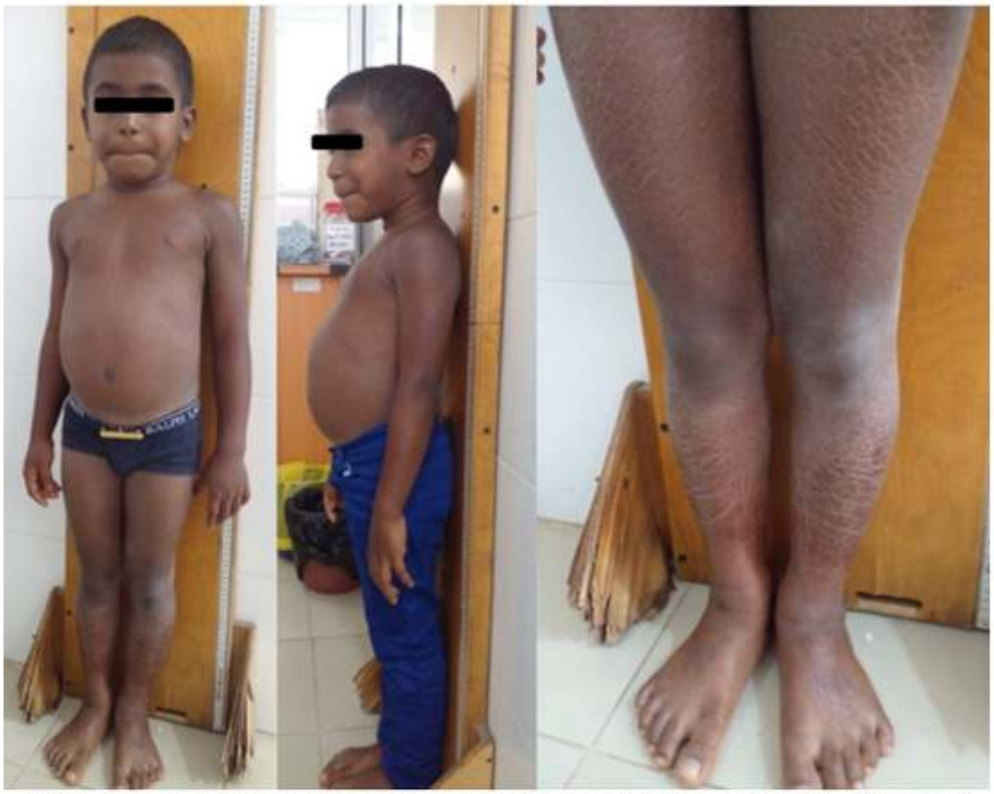

Figure 1: ***Muscular built, short stature and ichthyosis of lower limbs***
Permission given by parents to publish photograph

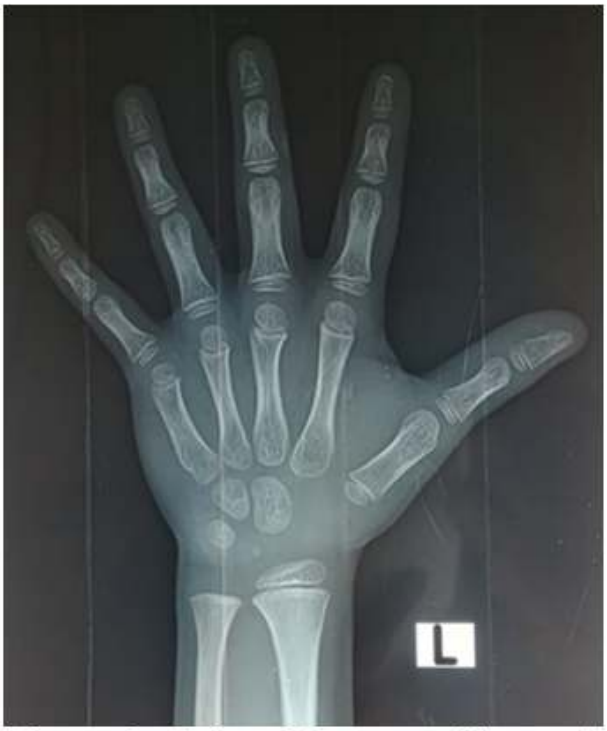

Figure 2: ***Delayed bone age (4 years)***

Discussion

Thyroxine stimulates glycogenolysis, degradation of glycosaminoglycans (GAG) and mitochondrial oxidative phosphorylation. When there is hypothyroidism, glycogen and GAG accumulate inside the muscles, causing muscular hypertrophy and weakness[5]. Elevated CPK is evident in 57-90% of patients with hypothyroidism. The value could range from 10-100 times the upper limit. However, it does not correlate with the severity of muscle symptoms[5]. In this child, CPK level was about ten times the upper normal limit, and it dropped to normal range with thyroxine therapy. KDDS has become rare due to newborn screening programmes. However, it could occur secondary to acquired causes as in this child. It is common among children born to consanguineous parents[2]. In this case, parents were first cousins. Pericardial effusion and nystagmus are some associations of KDDS[5] but in this case, neither was present. Electromyographic (EMG) features of KDSS include low amplitude short motor unit potential[5]. EMG was not done in this case since the diagnosis was evident clinically and there was a marked response to thyroxine treatment.

In addition to KDSS, this child had most of the classical clinical features of hypothyroidism, and the skin manifestation was one of the most worrying concerns for parents. Dry, scaly skin with acquired ichthyosis is a rare complication of prolonged untreated hypothyroidism[3]. Thyroxine is the main treatment and limb physiotherapy is beneficial in preventing muscle stiffness and achieving full muscle potential[5]. This child was started on both treatments and he remained clinically and biochemically euthyroid after 8 weeks.

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