CASE REPORT

# A rare complication of untreated congenital hypothyroidism in a Sudanese child

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## ABSTRACT

The clinical features and management of a 7-year-old boy who presented with Kocher–Debre–Semelaigne syndrome (KDSS) are described. KDSS is a rare complication of the long-standing, untreated congenital hypothyroidism. It can be encountered in clinical practice in countries where neonatal screening program of hypothyroidism is not yet applied. The condition could clinically mimic muscular dystrophy, and should be recognized, as it is a reversible condition upon simple and straightforward treatment by thyroxin.

## KEYWORDS

Hypothyroidism; Kocher–Debre–Semelaigne syndrome; Muscular dystrophy; Child; Sudan.

## INTRODUCTION

Kocher–Debre–Semelaigne syndrome (KDSS) is a rare complication of the long-standing, untreated congenital hypothyroidism associated with generalized muscular hypertrophy, mainly of the extremities, giving the child an athletic appearance or Herculean look. Age of presentation varies from 18 months to 10 years; however, there are anecdotal reports on KDSS in the neonatal period [1]. Though KDSS affects both sexes, it is more common in male children [2] and may easily be confused with primary muscle disorders unless the physician is aware of the condition. KDSS is rare in countries with neonatal screening program (NSP), as congenital hypothyroidism is diagnosed and treated early [3,4]. As regular NSP is not activated in Sudan, the condition can be easily missed or confused with muscular dystrophy. However, owing to the striking clinical features, availability of a simple treatment, and a good prognosis, pediatricians should be aware of it and recognize this syndrome.

## CASE REPORT

A 7-year-old boy was brought by his mother due to inability to walk, an increasing neck swelling, and delayed speech. Examination revealed the full-blown picture of severe hypothyroidism with large diffuse goiter, dull, waxy, pale, coarse face, with swollen, baggy eye lids, and thick pouting lips (Figure 1A), sitting on the mother lap with paucity of movement, disinterest, and non-interactive with the surroundings. The child was small for age with a weight of 16.5 kg and height 91 cm (both were below the 3rd centile for age) (Figure 1B). His upper to lower segment ratio was 1.4 (corresponding to age 2.4–3 years). His skin was dry, cold, and scaly over the lower 3rd of his legs. His muscle tone was decreased, but he had hypertrophied calf muscles and brisk tendon reflexes with delayed relaxation

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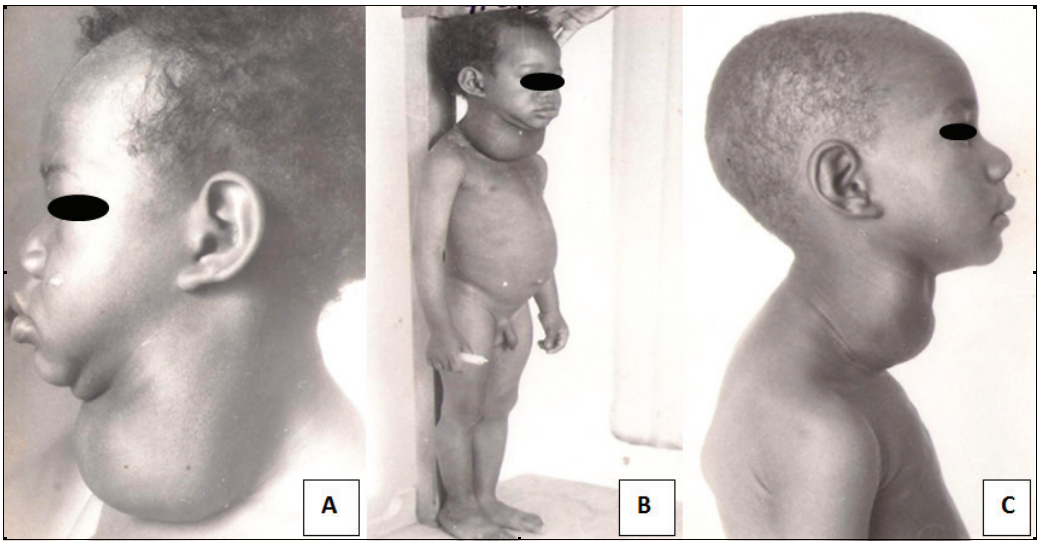


**Figure 1.** A child with Kocher–Debre–Semelaigne syndrome, a rare complication of untreated congenital hypothyroidism. (A) Characteristic faces and large goiter, (B) small stature and masculine habitus, and (C) dramatic response to L. thyroxin with disappearance of the facial swelling and regression of goiter size.

on the knee-jerk. There was no hypertrophy of the shoulder girdle muscles. His muscle power was decreased to 4/5 in the upper limbs and to 2/5 in the lower limbs, with mainly proximal weakness. He was able to stand alone but could not walk even few steps. His voice was so hoarse, deep, and the words were not clear, and the speech was not articulated in sentences. Investigations revealed thyroid hormones deficiency [T3 = 2.57 pmol/l (Reference range: 12–30), T4 = 21.88 nmol/l (Reference range: 71–160), and markedly elevated TSH (18.3 mU/l) (Reference range for age and sex: 0.73–8.45 mU/l)]. The thyroid enzymes test was not available that time. He had delayed bone age by 2.5 years and his IQ was 46 (moderate mental retardation). The child responded dramatically to L. thyroxin, the facial swelling disappeared, the goiter decreased in size (Figure 1C), words became clearer but not in sentences. The lower limb masculinization regressed.

Consent was taken from the mother to report on this case for educational purposes.

## DISCUSSION

The mechanism of weakness in hypothyroidism is different from that in muscular dystrophy. In severe hypothyroidism, accumulation of connective tissue and mucopolysaccharides occur in the muscles causing muscle hypertrophy. It is also known that thyroid hormones are necessary for the expression of fast myofibril proteins in the muscles and their deficiency lead to marked accumulation of the slow myofibril proteins, explaining the difficulty and slowness of movement, associated with hypothyroidism [4,5].

There were no other systems involvement in our case, such as in the eyes (nystagmus), the heart, oral/facial, or other systems like those seen in other very rare reported cases [6].

KDSS can be encountered in our country, yet it is uncommon. In 80 cases of children with thyroid problems studied as partial fulfillment of the University of Khartoum Clinical MD in Pediatrics and Child Health, attained by the author in February 1996, only one boy (1.25%) was found to have this complication [7]. Our case has got primary congenital hypothyroidism mostly due to dyshormonogenesis. Unlike most reported cases, hypertrophy was mainly in the calves and did not involve the shoulder muscles [1,2]. In agreement with the literature, the case demonstrated sound response to thyroxin treatment [5].

Since the neonatal screening program is not officially applied in our country, many cases of congenital hypothyroidism could be missed; hence, KDSS can be encountered in clinical practice. By reporting on this case, I aimed to increase the awareness of my colleagues on the presence of KDSS in our country.

## CONCLUSION

Since NSP is not officially applied as yet in Sudan, KDSS can be encountered in Sudanese children and can even be the heralding presentation of congenital hypothyroidism. It is a treatable condition and should not be confused with muscular dystrophy.

## ACKNOWLEDGMENT

I am deeply indebted to the mother of this child as she gave the consent to take photos of her child and to report the case. This case is part of a thesis in partial fulfillment of the University of Khartoum Clinical MD in Pediatrics and Child Health, attained by the author in February 1996.

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