Journal of Cardiovascular Disease Research
ISSN: 0975-3583, 0976-2833 VOL16, ISSUE 02, 2025

Kocher Debre Semelaigne Syndrome: A Rare Manifestation of Hypothyroidism

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Abstract
Kocher Debre Semelaigne syndrome(KDSS) is an rare condition in which the main characteristic is muscle pseudohypertrophy which was found to be associated with long standing and the cases of severe hypothyroidism which were untreated[1]. Here we report the case of a 14 year old female which previously went to endocrinology clinic because she had a very muscular appearance which was associated growth retardation, classical features of hypothyroidism, which showed improvement upon thyroxine supplementation. Pseudo muscular hypertrophy of KDSS is an acquired type of myopathy which is associated with long standing hypothyroidism and is reversible with thyroxine supplement.

Keywords: Pseudohypertrophy, Kocher Debre Semelaigne Syndrome, Hypothyroidism, Myopathy

Introduction

Overview of Hypothyroidism and Neuromuscular Involvement
Myopathy in hypothyroidism is associated with fatigue, pain on exertion, slow movement, stiffness, myalgia, decreased deep reflexes, proximal weakness and myxoedema. Around 10% of myopathy in hypothyroidism is associated with pseudo muscular hypertrophy known as KDSS in paediatric age group[2]. KDSS is an acquired type among myopathies which is associated with long-standing hypothyroidism irrespective of cause and it is also reversible with thyroxine supplement. We present clinical features of a rare case of KDSS in 14 yr old girl who responded to thyroxine supplements.
Pathophysiology of KDSS

The key mechanisms of KDSS are :
- Mucopolysaccharide accumulation within muscles, causing non-inflammatory enlargement[3].
- Reduced mitochondrial oxidative phosphorylation, leading to muscle fatigue and stiffness[4].
- Decreased myosin ATPase activity, contributing to sluggish reflexes and slow muscle contraction[5].
- Fluid retention, edema, increasing muscle bulk without pseudohypertrophy[5].

Epidemiology and Clinical Relevance
KDSS is extremely rare, with very few reported cases in the medical literature which affects:
- Pediatric patients (2–15 years old) with long-standing untreated congenital or acquired hypothyroidism[6].
- Cases are often missed in settings with inadequate neonatal thyroid screening programs[7].

Early recognition of KDSS is critical because thyroxine replacement therapy can lead to complete resolution, preventing misdiagnosis as a progressive muscular disorder[1].

Case Report

Clinical Presentation

A 14-year-old girl presented to the paediatric with history of progressive swelling over the peri-orbital region along with generalized body weakness, muscle stiffness, and not gaining height for over a year.

Her parents told that she had normal developmental milestones in past but had been experiencing increased weakness over the past four months, along with difficulty in lifting heavy objects and climbing upstairs. Patient gives was no history of exercise intolerance ,muscle pain, and significant weight gain.
She had a good school performance , and there was no history of neuromuscular disorders or thyroid disease in the family.

Physical Examination
General examination revealed normal vitals She had dry scaly skin, pallor, coarse facial features ,cold periphery, and short with broad shoulders with hypertrophy of calf, deltoid and paraspinal muscles. There was hung-up knee reflex. Gait was normal with negative Gower sign. Height was eight < -3 SD, Body Mass Index was 17.9kg/m2. The visual acuity, audiometry and fundus examination were found to be normal. Her Intelligent Quotient(IQ) was 98. Physical examinations revealed that both her thyroid lobes were diffusely enlarged. Rest of the systemic examination were normal

Physical examination of the child revealed that she had pseudohypertrophy of the calf muscle, deltoid muscle , and paraspinal muscles. On testing the reflexes there was hung-up knee reflex, with indicates the delayed relaxation of deep tendon reflexes. The Gower's sign was negative, which ruled out Duchenne Muscular Dystrophy (DMD). Additionally, her shoulders were broad yet short, and she exhibited thickened facial features, further pointing towards an endocrine disorder rather than a primary muscular dystrophy.
Laboratory and Imaging Findings
The following laboratory investigations were conducted to confirm the diagnosis:

Test	Result	Reference Range
TSH	Elevated (14.8 µIU/mL)	0.5–4.5 µIU/mL
Free T4	Low (0.5 ng/dL)	0.8–2.0 ng/dL
Creatine Phosphokinase (CPK)	983 U/L (elevated)	24–195 U/L
Anti-TPO Antibodies	Elevated (82 IU/mL)	<35 IU/mL
Neck Ultrasound	Enlarged thyroid, hypoechoic	-

A significantly elevated TSH with low free T4 confirmed primary hypothyroidism. The markedly raised CPK levels indicated muscle involvement, consistent with KDSS. Anti-TPO antibodies were positive, confirming the autoimmune nature of hypothyroidism (Hashimoto's Thyroiditis). Neck ultrasound findings showed an enlarged, hypoechoic thyroid gland, supporting the diagnosis of Hashimoto Thyroiditis.

Diagnosis
Based on clinical features, laboratory investigations, and imaging studies, the patient was diagnosed with Hashimoto's Thyroiditis leading to KDSS.

Treatment and Outcome
The patient was initiated on Levothyroxine 50 mcg/day, with dosage adjustments based on follow-up thyroid function tests. Within initial two months of treatment initiation, the patient exhibited a marked reduction in muscle hypertrophy, improvement in fatigue, and increased physical endurance. Her facial puffiness and dry skin also showed significant improvement.
By the six-month follow-up, the patient's thyroid function tests had normalized, and her muscle pseudohypertrophy had regressed completely. Her TSH levels reduced to 2.5 µIU/mL, and her free T4 levels normalized to 1.2 ng/dL. She also reported increased height growth of 2.5 cm, indicating improvement in overall endocrine function.

Discussion

Differentiation from Other Myopathies

KDSS presents with muscle hypertrophy, which can be confused with other conditions such as:

Condition	Hypertrophy	Weakness	Reflexes	CPK Levels	Reversible?
KDSS	Present	Mild	Sluggish	Elevated	Yes
Duchenne MD	Present	Progressive	Absent	Markedly high	No
Pompe Disease	Present	Severe	Hyporeflexia	Very high	No

The hallmark feature of KDSS is complete reversibility, which distinguishes it from genetic muscular dystrophies.

Management Strategies

- Thyroid hormone replacement therapy (Levothyroxine) is the primary treatment.
- Regular monitoring of thyroid function and muscle symptoms ensures optimal recovery.
- Supportive measures such as physiotherapy may be needed for cases with prolonged neuromuscular involvement.

Prognosis

Unlike progressive neuromuscular disorders, KDSS has an excellent prognosis when diagnosed early. Delayed treatment may lead to persistent growth failure and intellectual impairment.

Conclusion

KDSS is a rare but fully reversible cause of muscle pseudohypertrophy due to untreated pediatric hypothyroidism. Awareness of KDSS is essential to prevent unnecessary genetic testing and misdiagnosis as a muscular dystrophy. Timely thyroid hormone replacement therapy leads to remarkable clinical recovery.

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