

Case Report

From weakness to recovery a case report on Kocher-Debre Semelaigne syndrome

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ABSTRACT

Kocher-Debre-Semelaigne syndrome is a rare hypothyroid myopathy in children, characterized by muscle pseudohypertrophy and associated systemic symptoms. We report the case of a 5-year-old girl who presented with progressive muscle weakness, cold intolerance, dry skin, and constipation over seven months with prominent calf muscle hypertrophy, proximal muscle weakness, and hypotonia. This case underscores the importance of prompt diagnosis and treatment of KDSS to reverse symptoms and avoid misdiagnosis as a primary muscle disorder. Awareness is crucial for timely intervention and improved patient outcomes as hypothyroidism can be treated with thyroxine replacement therapy.

Keywords: Hypothyroidism, Pseudohypertrophy, Kocher-Debre-Semelaigne syndrome, Myopathy

INTRODUCTION

Kocher-Debre-Semelaigne syndrome (KDSS) is an uncommon disorder that causes muscle pseudohypertrophy and sustained mild to severe hypothyroidism in children. Hoffmann syndrome (HF) is a related disorder that affects adults.^{1,2} The length of time and degree of thyroid hormone deprivation are frequently associated with the severity of myopathy. In this uncommon illness, symptoms may reverse with prompt identification and initiation of thyroxine supplementation.³ Although the exact cause of pseudohypertrophy in KDSS is unknown, the lack of thyroid hormone impairs a number of metabolic processes, which causes muscles to accumulate glycogen and increased connective tissue in muscles.

CASE REPORT

A previously asymptomatic 5-year-old girl developed progressive weakness and an inability to bear weight on her knees over 7 months. There were no familial similar

illnesses, and dietary history was insignificant. Weight and height of the patient is normal within 25-50th percentile. General examination revealed cold intolerance, dry skin, and constipation.

The vitals of the patient were within normal limits, she exhibited prominent calf muscles, proximal muscle weakness, and difficulty in climbing stairs. Hypotonia was present, and power was graded at 3 in both limbs. Investigations revealed elevated TSH (7.04), creatine kinase activity also increased by 3120 units/l (Normal up to 195 U/l). Ultra sonogram neck revealed small-sized thyroid gland.

Nerve conduction study showed normal report however Electromyography showed low amplitude and short motor unit potential (MUAPS) in the quadriceps muscle leading to the diagnosis of KDSS. Thyroxine replacement therapy (levothyroxine 25 mcg/day) was initiated, resulting in euthyroidism after 2 months. Although hypothyroid symptoms regressed and creatine kinase

levels decreased after treatment muscular hypertrophy persisted for 9 months before gradually receding.



Figure 1: Left calf muscle hypertrophy.

DISCUSSION

Thyroid hormone deficiency accounts for 50% of acquired myopathies and is treatable, making clinical recognition essential as it can mimic primary muscle disorders.^{3,4} In this case, hypothyroidism, pseudohypertrophy, thyroid ultrasonography findings, and decreasing creatine kinase levels after levothyroxine treatment suggest KDSS. First reported by Kocher in 1892, with further descriptions by Debre and Semelaigne in 1935, KDSS primarily affects children aged 3–10 years age, although rare cases have been documented in neonates and at 15 months. It can result from congenital or acquired hypothyroidism.^{5,6}

The exact mechanisms behind pseudohypertrophy in KDSS remain unclear. A prolonged deficiency of thyroid hormone disrupts various metabolic functions in the body, including those in the musculoskeletal system.⁹ Additionally, impaired carbohydrate metabolism results in glycogen buildup in the muscles, along with increased deposits of connective tissue and mucopolysaccharides, which contribute to the appearance of muscle hypertrophy.¹⁰ The pseudo hypertrophy involves the muscles of the extremities, facial muscles, tongue, limbs, girdle, hands and feet, trunk, calf muscles, but it is more

prominent and first seen in the calf muscles as seen in our case.^{5,6,8}

CONCLUSION

KDSS, a rare hypothyroid myopathy causing muscle hypertrophy, can be mistaken for primary muscle disorders. Early recognition, confirmed by hypothyroidism exclusion, ensures effective treatment with a favorable prognosis. Awareness is vital for proper diagnosis and management, preventing complications.

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