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Case Report

Kocher Debre Semilaigne syndrome: a case report

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ABSTRACT

Kocher Debre Semilaigne syndrome is a rare clinical disorder wherein clinical hypothyroidism is associated with pseudo hypertrophy of muscles resulting in a herculean appearance to the affected patient. It is clinical disorder characterized by prompt reversal of hypothyroidism and pseudo hypertrophy following use of thyroid hormone supplementation. We present a case of Kocher Debre Semilaigne syndrome that presented with delayed milestones and had characteristic features, which reversed following thyroxine supplementation.

Keywords: Kocher Debre Semilaigne syndrome, Hypothyroidism, Pseudo hypertrophy, Thyroxine

INTRODUCTION

Kocher Debre Semilaigne syndrome (KDSS) is a disorder in which hypothyroidism is associated with myopathy and pseudo hypertrophy of various group of muscles.[1] Also termed as hypothyroid myopathy and hypothyroidism-large muscle syndrome, the onset is in infancy or in childhood.[2,3] It is a rare disorder in which prompt diagnosis and initiation of thyroxine supplementation will result in reversal of symptoms.[4,5] There are very few reported cases in literature and we present one such case.

CASE REPORT

The index case is a 3 year old boy who presented to our secondary care center affiliated to a medical college at Berhampur, Odisha with complaints of chronic constipation and lethargy. He was first order child born out of non-consanguineous marriage. Mother noticed the delayed milestones and shorter stature in comparison to the second child. The child was only able to speak bisyllables, was able to walk few steps only and was corresponding to chronological age of one year. Clinical examination revealed increased bulk of both thigh and calf muscles and muscles of arms and forearms (Figure 1). This feature coupled with clinical findings of coarse facies, short neck, dry skin, protuberant abdomen (Figure 2) and global developmental delay prompted a work up for hypothyroidism and it was conclusively proven by thyroid function tests.

TSH was 130 ng/L (0.2-6.0 IU/L), total T4 was 4.26 µg/dL (5-12.5 µg/dL) and T3 was 48.4 ng/dL (60–200 ng/dL). Serum creatinine kinase was 275 IU/L (60-175 IU/L). EMG showed slow conduction and low amplitude motor units. Pseudo hypertrophy of muscles with biochemically proven hypothyroidism lead to the diagnosis of Kocher Debre Semilaigne syndrome. USG neck did not reveal any thyroid dysgenesis. Further work up was not possible in our set up. Endocrinologist's opinion was sought and the child was started on 150 mcg of L-thyroxine which was gradually tapered to 50 mcg over 4 weeks. Considerable reduction in muscle size and marked increase in attentiveness and alertness was noted after 4 weeks of therapy, further proving our diagnosis of Kocher Debre Semilaigne syndrome.

DISCUSSION

Described initially by Kocher in 1898 and elaborated in detail in 1935 by the duo of Debre and Semilaigne, Kocher Debre Semilaigne syndrome (KDSS) is a conglomerate disorder of myopathy consequent to hypothyroidism and limb muscle pseudo hypertrophy in infancy or childhood.[1-3] KDSS is a rare disorder usually

seen in congenital hypothyroidism, but may also be associated with acquired hypothyroidism.[6-8] Also known as cretinism-muscular hypertrophy, hypothyroid myopathy, hypothyroidism-large muscle syndrome, hypothyrotic muscular hypertrophy in children, myopathy-myxedema syndrome, or myxedema-muscular hypertrophy syndrome, it is a disease with onset usually between 18 months and 10 years of age, but the condition has also been diagnosed in infants and neonates.[2,3,6-8]

Figure 1: Clinical examination revealed increased bulk of both thigh and calf muscles and muscles of arms and forearms.

The pseudohypertrophy involves the muscles of the extremities, limb girdle, trunk, hands and feet, but it is more prominent in the muscles of the limbs giving a Herculean appearance to the affected child.[2,3] The pathogenesis of such muscle changes have not been elucidated in detail, but is believed to be due to hypothyroidism related changes in glycogen metabolism.[2,3] This condition has a male preponderance, and has been reported in children with consanguineous parents and an autosomal recessive inheritance has been suggested.[4-6] Gross changes in muscles as seen in myopathy are absent in KDSS and electromyography usually reveals a myogenic lesion with low muscle action potential.[9]

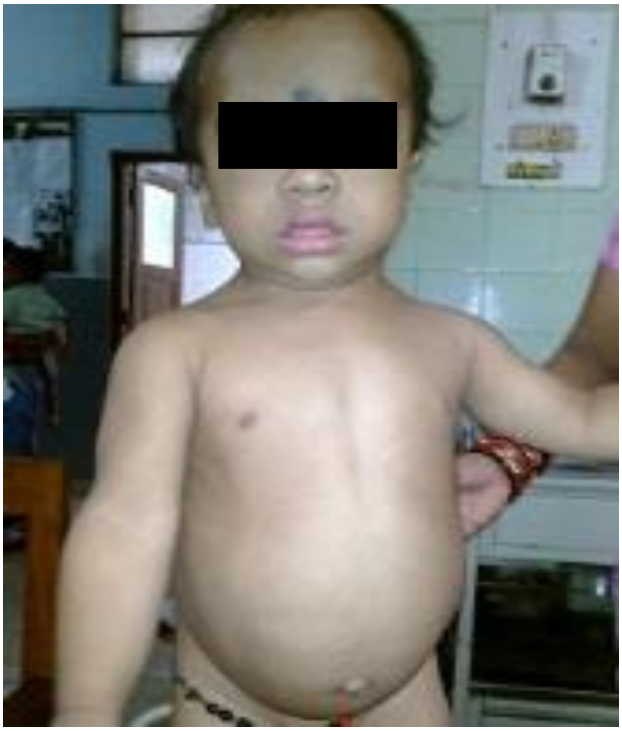

Figure 2: Feature coupled with clinical findings of coarse facies, short neck, dry skin, protuberant abdomen.

KDSS has to be clearly differentiated from primary myopathies like Duchene muscular dystrophy(DMD), which can also present with pseudohypertrophy of muscles with intellectual and learning impairment. But characteristic progression of the disease, pronounced myopathic changes on electromyography and biopsy and a normal thyroid function can help differentiate DMD from KDSS.[8] Though KDSS presents usually as a benign disease, easily manageable by thyroxine supplementation, unusual manifestations like pericardial effusion and cardiomyopathy have been documented in the literature.[2,10] Thyroid hormone replacement is curative and complete recovery is possible, with reversal of muscular and psychomotor changes.[5]

CONCLUSION

Characteristic features and their easy and prompt reversal with thyroid hormone supplementation makes Kocher Debre Semilaigne syndrome a unique condition and should be part of any diagnostic work up in a child presenting with lethargy, delayed milestones and impaired learning ability with increased muscle bulk. We bring to light this case to heighten awareness about KDSS in the medical community, so that such easily manageable and reversible clinical condition should not

be missed and prompt diagnostic workup and therapy should be initiated.

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