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CASE REPORT

Kocher-Debri-Semelaigne Syndrome: A Case Report

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Introduction

Kocher-Debri-Semelaigne Syndrome (KDSS) is a rare disease with association of muscular hypertrophy and prolong untreated moderate to severe hypothyroidism in pediatric age group. It was first discovered by Theodor Kocher in 1892 but its association with pseudohypertrophy of muscle was emphasized by Robert Debri & George Semelaigne in 1935.[1-t] Though the overall reported incidence of KDSS is less than 10%,u ¯w identification is very important because hormone replacement therapy can fully reverse the clinical pictures which is potentially serious.

The pathogenesis of this disease is not clearly understood. Deficiency of Thyroid hormone impairs many metabolic functions of the body in different organ system including musculoskeletal system. Abnormal carbohydrate metabolism cause glycogen deposition in muscle; while increased amount of connective tissue and mucopolysaccharide deposits in the muscles give the appearance of hypertrophy of muscles.

Case Report

A 5 year 9 month old female child of low socio-economic background hailing from a village of Nilfamari, Bangladesh was referred to Department of Endocrinology & Metabolic Disorder of Bangladesh Shishu (Children) Hospital & Institute, Dhaka due to not yet achieved walking and speech, not growing well in comparison to other peers and constipation since birth. She was diagnosed as a patient of Congenital Hypothyroidism at 2 years of her age which was grossly delayed and situation further deteriorated with poor drug compliance.

Fig.-1 *Photograph of the patient showing facial features.*

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On examination, she was found to be severely short statured [ht - 80cm against expected of 115 cm] in infantile proportion [US : LS = 1.4 : 1] with coarse facies, depressed nasal bridge (Fig.-1), dry texture of hair and skin, umbilical hernia (Fig.-2) with prominent muscular build at biceps and calf muscles (Fig.-3). Her systemic examination revealed low intelligence quotient with psychomotor retardation, generalized hypotonia and slow relaxation of knee jerk.

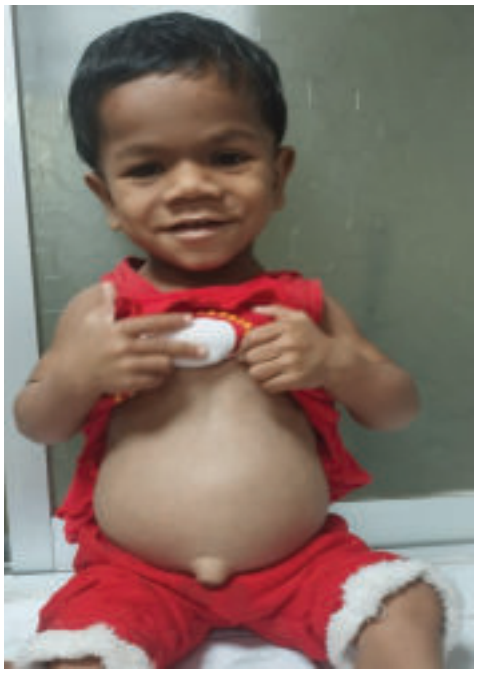

Fig.-2 *Profile photograph of the patient showing dismorphic face & umbilical hernia*

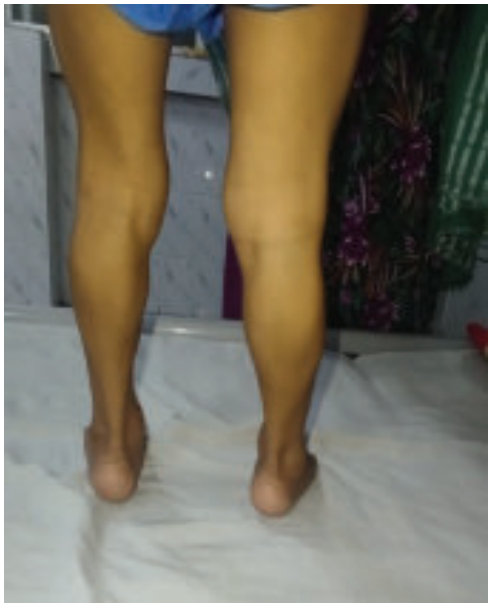

Fig.-3 *Photograph of the patients limbs showing calf muscles*

On investigations, she was found to have primary hypothyroidism with TSH >100 mIU/ml and FT_4 0.4 ng/dl, X-ray wrist revealed bone age <6 months; thyroid aplasia was found at USG of gland. Creatinine Phosphokinase (CPK) was elevated and Electromyogram revealed non inflammatory myopathy.

On the basis of myopathy and muscular hypertrophy associated with congenital hypothyroidism a diagnosis of Kocher-Debri-Semelaigne Syndrome was made. Treatment of hypothyroidism was started with Tab. Levothyroxine half of total dose (100 ugm/m² BSA) for 2 weeks and then increased up to full dose (50 ugm/day OD) to avoid side effects of sudden increase in metabolic rate due to thyroxin supplementation. Patient was discharged 1 week after establishment of full dose with nutritional & regular follow up advice.

Discussion

KDSS usually occurs between 18 months to 10 years of age with no sex differences.y ,u There is a wide range of clinical symptoms and signs mainly related to hypothyroidism, lethargy and insomnia, facial myxoedema, macroglossia, open fontanels, prolong neonatal jaundice, constipation, dry skin, growth delay and muscle hypertrophy, preferentially involving the trunk and all four limbs and causing a muscular appearance while our patient had most of the features. Despite of this muscular appearance however patients usually report muscle weakness which sometimes causes difficulty in sitting and in controlling their head position[10,11] although our patient didn't have such gross muscle weakness. The occurrence of disseminated intravascular coagulation syndrome[10] and even arrhythmogenic cardiomyopathies has been reported in most severe cases.[11]

The pathogenesis of pseudohypertrophy in our case could be the result of long standing hypothyroidism which impaired metabolic functions of the body that lead to glycogen and mucopolysaccharides deposits in the muscles give appearance of muscle hypertrophy.[12] The pseudohypertrophy may involves the muscles of extremities, limb girdle, trunk, hands, feet and the facial muscles but it is more prominent in the muscles of the limbs (calf).[13-15] The muscular hypertrophy and muscle weakness were observed mainly on the lower limbs on physical examination in our patient. Another report has shown that a child with KDSS with deleterious effect on tooth

development and eruption along with subnormal growth of maxilla and mandible[12] but our patient had no tooth anomaly. The signs and symptoms of hypothyroidism along with muscular hypertrophy revertv back to normal with time after initiation of thyroxine supplementation, but the fact that final height may still be short. A well planned and graded physiotherapy programme may be beneficial in getting rid of the muscle stiffness & achieving full muscle strength.[1]

Conclusion

KDSS is a rare case that may be confused with primary muscular disorders lest one is cautious enough to investigate for hypothyroidism. The striking clinical features, avaibility of simple treatment and good prognosis of the condition makes it worthwhile to report the case so that all practitioners can be aware of the condition and its management.

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