Case Report

A case report of Kocher Debre Semelaigne syndrome – Congenital hypothyroidism with pseudomuscular hypertrophy

Nidha Fatima[1], Faraaz Adil[2], Habib G Pathan[3]

From [1]Resident, [2]Assistant Professor, Department of Pediatrics, [3]Pediatric Neurologist, Department of Neurology, Princess Esra Hospital, Deccan College of Medical Sciences, Hyderabad, Telangana, India

ABSTRACT

Kocher Debre Semealaigne Syndrome (KDSS) is a rare condition with the manifestation of hypothyroidism associated with pseudomuscular hypertrophy. This syndrome is rare in countries with screening programs for hypothyroidism at birth. We are reporting a rare case of KDSS in which earlier diagnosis and treatment would result in the reversal of many symptoms. The main objective of presenting this case is to enlighten pediatricians about prompt diagnosis and timely management to prevent avoidable complications.

Key words: Hypothyroidism, Kocher Debre Semelaigne syndrome, Pseudomuscular hypertrophy

Kocher Debre Semelaigne Syndrome (KDSS) is a rare presentation of long-standing unidentified or inappropriately treated hypothyroidism [1-3]. It can be presented as early as 18 months of age. The real incidence of KDSS is still not known. Only 10% of cases with myopathic hypothyroidism may present with muscular hypertrophy [4-6]. Pseudohypertrophy of muscles usually mimicking congenital myopathy in the background of hypothyroidism is labeled as KDSS. It is also known as cretinism-muscular hypertrophy, hypothyroid myopathy, or myxoedema-myotonic dystrophy syndrome.

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 [5,6]. Such muscles are remarkably weak. The actual cause of muscle hypertrophy is not clear. A similar variant with pseudohypertrophy of muscles in adults with the existing background of hypothyroidism in children is known as Hoffman syndrome [2,3].

It is a rare disorder in which prompt diagnosis and initiation of thyroxine supplementation will result in the reversal of symptoms [7]. We report this rare case of a 10-year-old child to increase the suspicion and diagnose the condition and to bring awareness about its investigations and treatment. It also puts forth the reversibility of pseudomuscular hypertrophy and the importance of screening in such conditions.

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CASE REPORT

A 10-year-old oblivious girl born of non-consanguineous parentage with uneventful peri-natal and non-contributory family history. She presented to our outpatient department with growth and developmental delay. Parents also noticed progressive weakness of lower limbs with early fatiguability.

On examination, she had coarse facial features, yellowish dry skin, a depressed nasal bridge, a broad forehead, hypertelorism, and puffed lips. She was noted to have prominent calf muscles and a distended abdomen with an umbilical hernia (Fig. 1a and b). There was a substantial deficit in the attainment of height with apparent short stature. Her weight was 16 kg (<3rd centile). Her height was 90 cm (<3rd centile – normal height for age 138.6 cm) and upper segment and lower segment ratio of 1:1. Intraoral examination revealed that the patient had a protruding thick tongue and complete primary dentition.

Laboratory investigations revealed low hemoglobin (7.6 g/dL) with normal total leukocyte counts and platelets but free T3 and T4 levels were remarkably reduced 0.84 pg/mL and 0.15 ng/dL, respectively (Normal levels of free T3 1.3–4.2 ng/dL, T4 1.0–2.1 ng/dL) and TSH was significantly elevated >60 mIU/L (Normal – 0.8–6.0 mIU/L). Creatine kinase activity also increased by 4720 units/L (Normal up to 195 U/L). Ultra sonogram neck revealed small-sized thyroid gland. Orthopantomogram showed primary dentition with multiple impacted teeth.

On account of the presence of myopathy and muscular hypertrophy associated with congenital hypothyroidism, a

Correspondence to: Faraaz Adil, Assistant Professor, Department of Pediatrics, Princess Esra Hospital, Deccan College of Medical Sciences, Hyderabad, Telangana, India. E-mail: faraazadil2@gmail.com

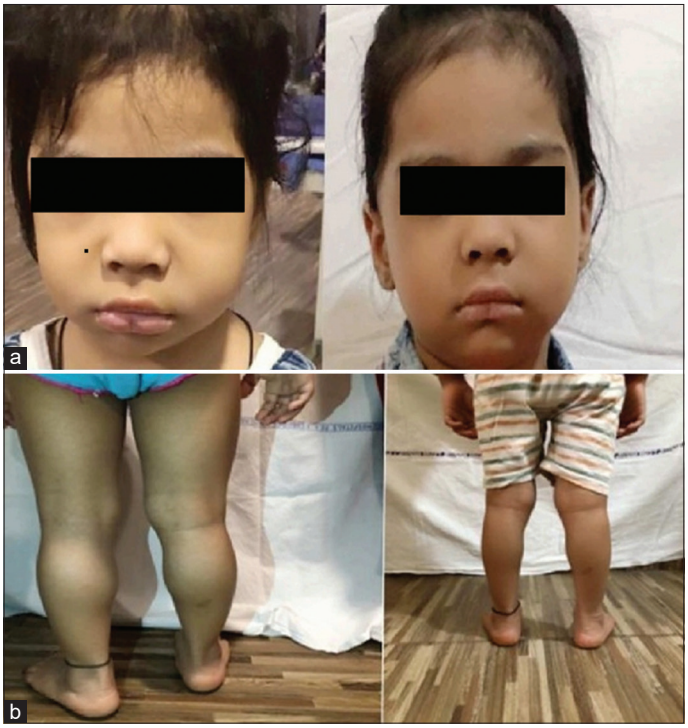

Figure 1: (a) Facial features before and after treatment, (b) Appearance of the calf before and after treatment

diagnosis of KDSS was made. Levothyroxine was started in appropriate doses for hypothyroidism. After 2 months showed improvement in the form of loss of edema, reduced visible muscle mass of calf muscle, and enhanced attentiveness. Repeat creatine kinase levels had dropped down to 247 units/L.

DISCUSSION

KDSS is a rare manifestation associated with severe, untreated, long-standing congenital hypothyroidism, and diffuse muscle pseudohypertrophy [1-3]. The usual age of presentation is usually between 18 months and 10 years [8]. It is associated with various forms of hypothyroidism such as absence of the thyroid gland, defective synthesis of thyroid hormone, or autoimmune causes of hypothyroidism [9].

Clinical recognition of the disorder is important as it may be mistaken for primary muscle disorder. The pathogenesis of pseudohypertrophy is still not clearly understood. It is believed to be one of the following or a combination of one or more of these. It can be attributed to increased glycogen accumulation and increased mucopolysaccharide deposition in muscles as a result of the defective metabolism of carbohydrates [10]. Another hypothesis involves ineffective reuptake of calcium into the sarcoplasmic reticulum causing increased levels of intracellular calcium resulting in a sustained contraction leading to hypertrophy [3,5]. KDSS should be considered as a differential diagnosis of other primary myopathies. Hypothyroidism should be ruled out before making a diagnosis of primary muscle disorder.

Furthermore, severe hypothyroidism can have deleterious effects on tooth development and eruption and lead to prolonged retention of the primary dentition, subnormal growth of the maxilla and mandible with a marked reduction in the dimensions of the facial complex, and a lack of coordination between mandibular growth and dental development [8].

CONCLUSION

The awareness of striking clinical features and the availability of rather uncomplicated management with a good prognosis of the condition is important for all pediatricians. Worldwide newborn screening program helps in early diagnosis and treatment and aids in ascertaining one of the most common preventable causes of intellectual disability.

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