International Journal of Science and Research (IJSR)
ISSN: 2319-7064
SJIF (2020): 7.803

Kocher-Debry Semelaigne Syndrome: An Infrequent Encounter in Hypothyroidism - A Report of Two Cases

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Abstract: ***Kocher-Debre-Semelaigne syndrome (KDSS) is a rare association of muscular pseudohypertrophy and long-standing moderate-to-severe hypothyroidism in the paediatric age group. The striking clinical features, availability of a simple treatment and a good prognosis for the condition makes it worthwhile to report such two cases so that all practitioners be aware of the condition and its management.***

Keywords: Hypothyroidism, KDSS, Kocher Debre Semelaigne syndrome, Pseudo muscular hypertrophy, Short stature

1. Introduction

Kocher Debre Semelaigne syndrome [KDSS] is a rare condition of muscular pseudo hypertrophy and long standing moderate to severe hypothyroidism in children and Hoffmann syndrome [HF] in adults [1-3]. Though thesyndrome was initially reported by Emil Theodar Kocher [1892], the association of hypothyroidism with pseudohypertrophy of muscles was emphasized by Robert Debre and George Semelaigne [1935] [1, 4-6]. The overall reported incidence of KDSS is less than 10% [7-9].

Severity of myopathy generally correlates with the duration and the degree of thyroid hormone deficiency [7, 10]. It most commonly occurs in males, and has been reported in children as the products of consanguineous marriage [11].

The pathogenesis of the pseudohypertrophy in KDSS is not completely understood. The lack of thyroid hormone impairs many metabolic functions of the body including musculoskeletal system. Impaired carbohydrate metabolism leads to glycogen accumulation in muscles; while increased amounts of connective tissue and mucopolysaccharide deposits in the muscles also give the appearance of hypertrophy of muscles [12].

2. Case Report 1

A7years old female child presented to our institute with history of generalized swelling of the body for the past 1 month which was insidious in onset and gradually progressive in nature. There was no history of any perinatal insult or history suggestive of any cause of anasarca. Birth weight and length were according as per gestational age. On examination the girl was lethargic with puffiness of face, potbelly, pedal edema with dry and scaly skin and enlargement of the calf muscles. There were no dysmorphic features. The anthropometric measurements showed weight of 15.5 kg and height of 100 cm. Her height and weight for age was below 3rd percentile [figure 1]. It was grade 3 Stunting as per Waterlow classification. The mid parental height was 147 cm, the upper segment to lower segment ratio and arm span was suggestive of Proportionate short stature [figure2]. Secondary sexual characters were not developed with sexual maturity rating [SMR] of of P1 b1.Systemic examination was within normal limits. Investigations showed no abnormality in routine haemogram, liver function and renal function tests, stool and urine examination including spot urine protein creatinine ratio and 24- hour urinary protein.2D echo showed pericardial effusion. Her bone age was approximately 5 years and her height age was approximately 4 years. [Height age<Bone age<Chronologic age].Her Thyroid function tests revealed Free T3 of 0.39 pg/ml [normal 2.5-7 pg/ml], Free T4 was 0.038 ng/dl [normal 0.8 -2.2 ng/dl] and TSH >1000IU/ml [normal 0.5 -5.5 IU/ml]. Antibody workup was done and Anti TPO antibody titre was543.39 IU/ml [normal <5.61 IU/ml] and Anti TGA antibody titre was 244.69 IU/ml [normal< 4.11 IU/ml].USG abdomen revealed minimal ascites and USD neck showed thyroid gland of normal size and echotexture. A diagnosis of Autoimmune Thyroid Disease was made and further investigations like hba1c, Anti ttg and Anti endomysial antibody were done to rule out other associated autoimmune disorders which were within normal limits. The child was started on Tablet Levothyroxine at 10 mcg/kg/day OD, 3 months following which the child became euthyroid and symptoms of hypothyroidism had regressed with resolution of pericardial effusion but muscular hypertrophy is still persistent even after 3 months of thyroxine supplementation.

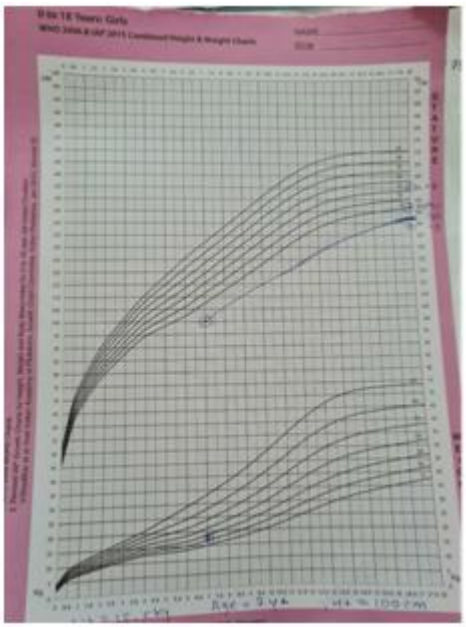

Figure 1: Growth chart showing height for age below 3rd percentile

Figure 2: Short stature in a girl of age 7 years

3. Case Report 2

An eight years old boy of non-consanguineous parents presented with scholastic backwardness, short stature and constipation. Mother informed that her son is dull and slow compared to his peers. On examination, he was having athletic built, coarse facies [figure 3], large protruding tongue [figure 4], dry mottled skin [figure 5]and abdominal distension. There was no evidence of malnutrition, hernia or goiter. Delayed osseous maturation was evident by open anterior fontanel. Height and weight were below third percentile. He had evidence of generalized muscular hypertrophy which was more prominent in girdle and limbs. Investigations showed normal hemogram. Thyroid function tests showed Total T3 <40 ng/dl [normal 70-200 ng/dl], total T4 <1 mcg /dl [normal 5.5-13.5 mcg/dl], TSH >95 mIU/ml [normal 0.2-5.0 mIU/ml], which confirmed the diagnosis of hypothyroidism. Creatine kinase activity was also increased [212 units/L]..Ultrasound studies for thyroid revealed absent thyroid gland in its normal location without any evidence of ectopic thyroid tissue in the neck. Hewas started on L-thyroxine at the dose of 4mcg/kg/day and he has shown marked increase in alertness and activity after 4 weeks of treatment though the muscular pseudo hypertrophy is still persistant after 6 months of thyroxine supplementation. The child was euthyroid after 3 months.

Figure 3: Coarse facies in a child of hypothyroidism

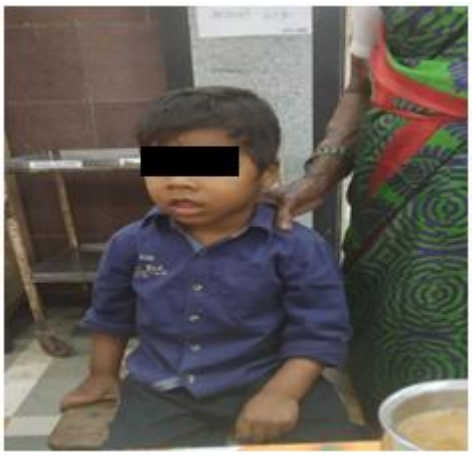

Figure 4: Large protruding tongue

Figure 5: Dry mottled skin

Both the children are under regular followup to monitor height and assessing euthyroid status.

4. Discussion

Kocher-Debre-Semelaigne syndrome involves muscles of extremities, limb girdle, trunk, hands and feet but it is more prominent in muscles of limbs [figure 6]; hence the athletic or Herculean look [1]. Pseudo hypertrophy of muscles comprised 18.7% of cases with a 2:1 female to male ratio, and it was found in various forms of hypothyroidism. Thyroid dysgenesis was the most common etiology [80.9%] [13], Calf muscles involvement is a rare clinical presentation of hypothyroidism. Its pathogenesis is unknown. Nonspecific histochemical and ultra structural changes seen on muscle biopsy return to normal with treatment. Boys are more prone. Siblings of consanguineous marriage affected children are more prone for pseudo hypertrophy of calf muscles. They usually have hypothyroidism of longer durationand severity [14]. Kocher-Debre-Semelaigne syndrome with presence of pericardial effusion an unusual presentation has been reported. The response to thyroxin replacement was excellent, with resolution of the pericardial effusion and clinical improvement [15]. Similar improvement was seen in our patient.

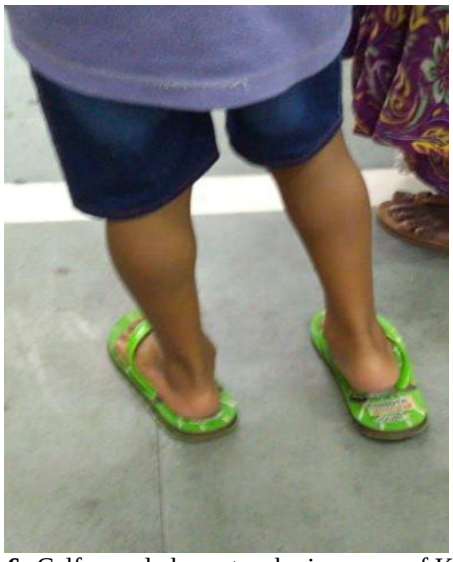

Figure 6: Calf muscle hypertrophy in a case of Kocher-Debre-Semelaigne syndrome

5. Conflict of Interest

We the authors report no competing interests and no funding was received for this study.

6. Ethical Consideration

Informed consent taken from the parents of the patients regarding publishing the data

7. Conclusion

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

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