

Case Report

“Kocher Debre Semelaigne Syndrome” – A Case Report

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Abstract:

We report a case of 12 year old male child with Kocher-Debré-Semelaigne syndrome (KDSS) which is a rare clinical presentation of hypothyroidism associated with muscular pseudo-hypertrophy. The main differential diagnosis is Duchenne Muscular Dystrophy. Pseudomuscular hypertrophy of KDSS is an acquired type of myopathy associated with severe and long standing hypothyroidism and is reversible with thyroxine supplement.

Key Words: Hypothyroidism, Myopathy, Kocher Debre Semelaigne syndrome, Pseudo hypertrophy.

Introduction:

Irrespective to the cause of hypothyroidism, many of these patients have peripheral nerve involvement (19%) and metabolic myopathy (30-80%). Myopathy associated with hypothyroidism classically presents with fatigue, exertional pain, slow movement, diminished deep reflexes, stiffness, myalgia, myoedema, proximal weakness, and less commonly, cramps (Mehrotra et al, 2002). Among the patients with myopathy, less than 10% of patients develop pseudo-muscular hypertrophy known as Kocher Debre Semelaigne syndrome (KDSS) in children (Yeshwanth et al, 1987) and Hoffmann syndrome in adults, which are reversible with treatment.

Case report:

A 12 year old boy from middle socio- economic status presented with complaints of generalized weakness, decreased physical activity and facial puffiness for 2 years. On eliciting further, history of snoring, breathlessness on exertion and not gaining height for last 1 year was also obtained. There were no associated complaints of fever, cough, chest pain, swelling over feet, constipation, cold intolerance, blood loss, worm infestation or muscle cramps. He had past history of decreased vision, which improved after treatment with vitamin A.

He was the eldest son among three siblings, born to non- consanguinous marriage with full term normal vaginal delivery and uneventful antenatal, natal

and postnatal periods. His milestones were normal with very good scholastic performance.

General examination revealed - pulse rate 66/minute; blood pressure 80/60 mm of Hg and oral temperature 97 degree F. He had pallor, xerophthalmia stage X1a, dry scaly skin, cold periphery, coarse facial features and short and broad shoulders with athletic look (Fig.I). Systemic examination revealed obvious hypertrophy of calf, deltoid and paraspinal muscles which were firm on palpation (Fig. II, III). There was hung-up ankle reflex with slow relaxation. His gait was normal, he could jump, hop, skip and squat. His anthropometric assessment was: - height 116 cms (expected height for the age is 149 cms), arm span 118 cms, upper segment & lower segment ratio was 1:1 and weight 21.5 Kg (expected weight for age is 40 Kg). Visual acuity, fundus examination and audiometry were normal. His Intelligent Quotient was 95.

Investigations revealed: Hemoglobin 10.2 gm/dl, Total Leucocyte Count 6,600/cu mm, MCV 89.9 fL, MCH 29.5 pg with normocytic and normochromic red blood cells on peripheral smear. His serum cholesterol was 207 mg/dl (normal 130-204 mg/dl), S. LDH, 687 IU/L (normal range upto 450 IU/L) and creatine phosphokinase (CK) 1708 IU/L (normal upto 190 IU/L). Bone age was 6 years only (Fig.IV). Diagnosis of primary hypothyroidism was confirmed by low serum triiodothyronine (T3) 0.26 ng/ml (Normal range 0.59-1.70 ng/ml) and serum thyroxine (T4) 0.5 microgram/dl (Normal range 4.66-9.32 microgram/dl), and high serum thyroid stimulating hormone (TSH) >60.00 mIU/ml (Normal range 0.25-5.0 mIU/ml). Thyroid peroxidase antibody (TPO Ab) as well as anti-thyroglobulin antibody titres were high.

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Fig. I: Photograph showing broad shoulders with atheletic look.



Fig.II: Photograph showing hypertrophied Calf muscle.



Fig.III: Photograph showing prominent hypertrophied paraspinal muscles.



Fig.IV: Photograph of wrist joint showing only 5 carpal bones
(Bone age 6 years)

Electromyography (EMG) and Nerve Conduction Velocity (NCV) studies showed generalized demyelinating type of polyneuropathy of both sensory and motor nerves with myopathic kind of picture of muscle. Ultrasonography (USG) of thyroid gland showed normal size but heterogenous echotexture.

He was treated with thyroxine 75 microgram once a day. On follow up at 2 months, he showed marked improvement in clinical features including significant regression of muscle hypertrophy and normal thyroid function test.

Discussion:

Kocher in 1892 and Debre & Semelaigne in 1935 described a syndrome of muscular hypertrophy associated with delayed muscle relaxation in two athyreotic cretins (Kung et al, 1987). Kocher-Debre-Semelaigne syndrome is also labeled as Cretinism-muscular hypertrophy, hypothyroid myopathy, hypothyroidism-large muscle syndrome, myopathy-mixedema syndrome, mixedema-muscular hypertrophy syndrome (Dharaskar et al, 2007).

Kocher Debre Semelaigne Syndrome involves muscles of extremities, limb girdle, trunk, hands and feet, but it is more prominent in muscles of limbs hence giving the athletic or Herculean look to the patient (Tullu et al 2003) as was seen in the present case. It is especially seen in the boys with disease of longer duration and severity of hypothyroidism (LaFranchi, 2004).

It may be mistaken for primary muscular disorder like Duchenne Muscular Dystrophy leading to delay in the specific treatment. But, unlike Duchenne Muscular Dystrophy, KDSS myopathy is reversible and responds favourably to adequate treatment for hypothyroidism.

Though the KDSS is seen more often in 1.5 to 10 year age group, it has been also reported in a neonate (Ghosh,1988).

Cause of hypothyroidism in our case was lymphocytic thyroiditis as was evident by raised auto antibodies. This is the most common cause of thyroid disease in children (>5 years) and adolescent as well as the most common cause of acquired hypothyroidism. The incidence is 1% among school age children (LaFranchi, 2004). Lymphocytic thyroiditis was first described by Dr. Hakaru Hashimoto in 1912 as immunological damage of thyroid gland that leads to impaired function and decreased production of T4. Thyroid autoantibodies like TPO Ab (previously known as antimicrosomal antibody) is demonstrated more often (>90%) as compared to antithyroglobulin antibody (< 50%). If both the tests are used, sensitivity increases to >95%.

The pathogenesis of the pseudohypertrophy in KDSS is not completely understood. Nonspecific histochemical and ultra structural changes seen on muscle biopsy return to normal with treatment.

Lack of thyroid hormone impairs many metabolic functions of the body including musculoskeletal system. Impaired carbohydrate metabolism leads to Glycogen accumulation in the muscle. Increased amount of connective tissue and mucopolysaccharide deposits in the muscle also gives the appearance of hypertrophy of muscles.

In hypothyroidism shift of fast twitch muscle fiber to slow twitch fiber leads to slow muscle contraction and relaxation as seen in KDSS. Muscular hypertrophy is also contributed by prolonged muscular contraction due to delayed Ca^{++} re-uptake by the sarcoplasmic reticulum. These changes improve or disappear by proper treatment refer as was seen in our

case (Mehrotra et al,2002).

The elevation of CK and LDH in KDSS seen in our case has been attributed to the changes in cell membrane permeability leading to the release of these enzymes (Tashko et al, 1999). This is reported in 80% of the hypothyroidism even with the absence of muscle involvement and their level normalize on correction of hypothyroidism.

An unusual case presentation of KDSS with presence of pericardial effusion with complete resolution of the pericardial effusion and clinical improvement in response to thyroxin replacement has also been reported (Dharaskar et al, 2007).

Conclusion:

Hypothyroidism presenting with classic manifestations of lethargy, cold intolerance, hoarseness of voice, dry skin, constipation, bradycardia, and slow relaxation of deep tendon reflexes, is easily recognized and, therefore, easy to diagnose and treat. But occasionally patients may present with atypical and rare manifestations making the diagnosis less apparent thus delaying the treatment. By reporting this case, we intend to bring the awareness about this unusual presentation of hypothyroidism in the form of Kocher-Debre Semelaigne syndrome which is a rare and reversible myopathy. It causes pseudo hypertrophy of muscles specially calf muscles and should be considered in differential diagnosis of Duchenne Muscular Dystrophy.

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