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UNUSUAL MANIFESTATION OF PERICARDIAL EFFUSION IN KOCHER - DEBRE - SEMELAIGNE SYNDROME - A CASE REPORT

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Abstract

Kocher – Debre – Semelaigne (KDS) syndrome is known by various names like hypothyroid myopathy, hypothyroidism – large muscle syndrome, myopathy-myxedema syndrome, myxedema muscular hypertrophy syndrome and cretinism muscular hypertrophy etc. It is a rare disorder characterised by pseudohypertrophy of calf muscles and non pitting edema of legs due to development of long standing hypothyroidism1. Pericardial effusion (PE) can occur in hypothyroidism but it is very rare in KDS syndrome.

CASE REPORT

A 13 year old school drop out girl presented with difficulty in walking and getting up from sitting posture since 8 months , fatiguabilty and breathlessness since 3 months. There was no history of palpation, joint pain, oliguria and chest pain.Menstrual history revealed cessation of menses. On examination, she was conscious, with coarse facies and loss of eyebows laterally. Vitals BP 90/60 mmHg, Pulse 62 beats/min, Respiratory rate 22/min, oedema feet present, jugular venous pressure elevated by 10 cm. There was no diastolic collapse. Her height was 114 cm and weight 20 kg (both low). Neck examination revealed no thyromegaly. Tongue was normal. Examination of both calf muscles revealed hypertrophy. Cardiovascular examination revealed muffled heart sounds without any murmur and pericardial rub. Central nervous system examination showed muscle weakness with muscle power of 4/5 in proximal group of muscles and normal power in distal group of muscles. Deep tendon reflexes were diminshed with plantar response flexor. Sensory, motor and cerebellar system examination was normal. Laboratory investigations revealed Hemoglobin 8.8 %, Total leucocyte count 6500 /mm^3, Differential cell count P 75, L 25, E 0, B 0. Liver, renal, lipid and, electrolyte profile were normal. ECG revealed low voltage graph and bradycardia, X-ray chest showed cardiomegaly. 2 D Echocardiography and CT scan chest revealed pericardial effusion. Bone age was 11 years(delayed). Creatinine phosphokinase levels increased, thyroid function tests revealed T3 30 ng/dl (N 70-200), T4 1.2 ug/dl (N 5.5-13.5), TSH 90 uIU/ml (N 0.2 – 5) which confirmed the diagnosis of hypothyroidism. Antimicrobial antibodies were positive. Radioactive Iodine uptake was poor. Based on the history, clinical examination and investigations, a diagnosis of Kocher-Debre-Semelaigne syndrome with Pericardial effusion(PE) was made. She was started on 25 ug L thyrotoxine which was gradually increased to 100 ug. She responded dramatically with marked clinical improvement and resolution of PE within 4 weeks. She was discharged in a stable condition but was lost on follow up. Patient refused muscle biopsy of calf muscle.

DISCUSSION

Hypothyroidism is responsible for 5% cases of acquired myopathies[2]. The association of hypothyroidism and muscle pseudohypertrophy is called KBS syndrome in infants and children while this association in adults is called Hoffman's syndrome. KDS syndrome usually presents between 18 months to 10 years of age [1,2,3]. But cases have been described even at early age[1]. Our patient reported at 13 years of age. These patients have clinical features of hypothyroidism along with muscle pseudohypertrophy[1]. The pathogenesis of muscle pseudo hypertrophy is not clear but it is thought to be due to long standing hypothyroidism[1] or defective synthesis of thyroid hormone or absence of thyroid gland[2]. The etiology in our case could be auto immune thyroiditis as documented by positive antimicrobial antibodies.

Regarding histological findings , they include type 2 fibre hypertrophy or atrophy, myofibre necrosis and regeneration and permanent core like areas containing amorphous granulo-filamentous material[4]. The presence of cores correlated with the severity of hypothyroidism, muscle hypertrophy, cramps and duration of hypothyroid state[5]. The muscle biopsy shows hypertrophy of muscle fibres with increased nuclei, few necrotic fibres and mycord deposits at places.[4]

Kabadi et al[6] reported the incidence of PE in hypothyroidism as 6% (2 out of 30 cases). PE is common in myxedema (30-50%) which is advanced severe stage of hypothyroidism (overt hypothyroidism) but it is very rare in early mild cases of hypothyroidism[6]. The pathogenesis of PE in myxedema includes increased capillary permeability and impaired lymphatic drainage resulting in protein leakage into the interstitial space[4,6]. PE in hypothyroidism can rarely cause cardiac temponade[7]. Clinical features of hypothyroidism and associated PE usually respond to thyroxine therapy[4,6,]. Our patient responded well to thyroxine supplementation resulting in marked clinical improvement and disappearance of PE. Very few cases of KDS with PE have been reported in the literature. Hence the case report.

CONCLUSION

Kocher – Debre – Semelaigne syndrome is a rare disorder usually occuring in children below 10 years age. PE is common in overt hypothyroidism but it is extremely uncommon in childhood hypothyroidism

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