Pseudo Muscular Hypertrophy Secondary to Kocher Debre Semelaigne Syndrome

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Clinical Image

We report a case of a 7 year 8-month-old female child brought by parents with concern of hardening of muscles of calf and forearm since few months. On inquiry, she has lethargy, feeling of冷ness, constipation, dry skin, hoarseness of voice and increased sleep. On examination, pulse rate was 68 beats/min, height was 89 cm (expected is 123 cm) and weight was 12.9 kg (expected is 22.5 kg). She had periorbital edema, facial puffiness, coarse facial features, dry skin and hypertrophy of calf and forearm muscles. She did not have obvious goiter. Based on the clinical feature and examination findings, pseudo muscular hypertrophy secondary to Kocher Debre Semelaigne syndrome was suspected. Endocrine workup revealed primary overt hypothyroidism with elevated serum TSH (>150; reference range 0.4–4.0 µIU/ml), low T3 (14.1 ng/dl; reference range 82–179 ng/dl) and low T4 (<0.3; reference range 5.2–12.5 µg/dl). Child was treated with thyroxin replacement as 50 mg once a day. She showed a rapid clinical improvement within few weeks and some regression in muscle hypertrophy was seen (Figure 1).

Figure 1: Clinical picture showing Pseudo muscular hypertrophy of a: Calf and b: Forearm muscles.

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