Hoffmann syndrome is a specific and rare form of hypothyroid myopathy in adults characterized by presence of muscle stiffness, proximal weakness and pseudohypertrophy. When this occurs in a child with cretinism it is known as Kocher-Debré-Sémélaigne syndrome. Patients with more severe or longstanding untreated hypothyroidism are more likely to develop clinically significant muscle disease. Serum muscle enzyme levels as CK, myoglobin and lactate dehydrogenase are frequently elevated. Although this increase is usually mild (CK <1000 IU/L), reports of rhabdomyolysis do exist in the literature.

**Case presentation**

A 9 year old boy presented with hoarse voice, pallor, weakness and tiredness of 6 months duration. He associated poor height gain in the last year and muscular hypertrophy in the last months. He maintained adequate school performance without other associated symptoms.

**Physical examination** revealed short stature (height 121.6 cm, -2.4 SD; BMI 18.6 kg/m2, 0.2 SD), bradycardia (47 bpm), palpebral edema, generalized muscle hypertrophy with proximal limb weakness and dry skin, without goiter.

**Laboratory studies** showed a severe primary hypothyroidism: TSH 894.84 µU/ml [0.57-5.92], free-T4 0.036 ng/dl [0.72-2.0], and positive thyroid autoimmunity (anti-TPO antibodies > 600 IU/ml, anti-TG antibodies > 4000 IU/ml). He also presented elevated CK 3172 U/l [1-175], Cholesterol 265 mg/dl, LDL 155 mg/dl, ALT 76.2 U/l [5-26], AST 123.6 U/l [5-37] and LDH 411 U/l [120-300].

The ultrasound demonstrated a heterogeneous and enlarged thyroid gland and the brain MRI a hyperplasia of the pituitary gland. With these findings the patient was started on levothyroxine 2.7 µg/kg/day.

**Conclusion**

We expose a case of Hoffmann syndrome in a child, presented with typical symptoms of muscular pseudohypertrophy caused by long-standing untreated hypothyroidism. This clinical picture has been very rarely reported in children. Although thyroid hormone deficiency is the underlying etiology of acquired myopathies in a small proportion of cases, all patients with an acquired myopathy and pseudohypertrophy should be screened to rule out hypothyroidism.

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