Title: Uncommon association of hypoparathyroidism and Rendu-Osler syndrome

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Introduction

• Rendu-Osler-Weber syndrome (also called Hereditary Haemorrhagic Teleangectasia) is an autosomal dominant disorder that results from multisystem vascular dysplasia.
• HHT syndrome has been described in association with autoimmune disorders, such as Hashimoto thyroiditis, lupus erythematosus, vitiligo, anti-phospholipidic syndrome and pernicious anemia.

Case report

• We present the case of a 6 year old girl with Rendu-Osler-Weber syndrome who was referred for endocrinological evaluation for moderate hypocalcemia discovered during a biochemical screening.
• The patient complained of paresthesia of the extremities without a history of carpal-pedal spasm, seizures or laryngospasm.
• Physical exam showed:
  - obesity (BMI 22.7 kg/m², above 97th percentile)
  - no bone deformities
  - positive Chvosteck sign
  - dental dystrophia.

Endocrinological evaluation

• During the initial endocrinological evaluation, blood tests showed low serum calcium (8.1 mg/dl and 7.1 mg/dl in two different occasions), serum albumin (5.1 g/dl) and corrected calcium (6.8 mg/dl), high phosphorus (7.4 mg/dl), low urine calcium (5 mg/500 ml, under the reserve of improper collection) and normal 25 hydroxyvitamin D levels (35.19 ng/ml).
• The parathyroid hormone concentration was inappropriately low (24.96 pg/ml).
• Cerebral CT scan showed no calcifications.
• Hand X ray showed normal length of the metacarpal bones and no particular findings were found on electrocardiogram.

• Based on the clinical and biochemical evaluation the diagnosis of hypoparathyroidism was established.
• She was given alphacalcidolum in doses of 0.25 ug and 500 UI colecalciferol, and put on a low phosphorus diet.
• She was advised to have an adequate intake of calcium. Under treatment, at one month evaluation the calcium corrected (serum calcium 8.4 mg/dl).

Discussions

• We present this case because of the rare association between HHT and hypoparathyroidism.
• To our knowledge, there is only one clinical case report of two brothers with clinical manifestations of HHT in addition to severe hypocalcemia associated to hypoparathyroidism.
• One possible explanation for this rare association can be the autoimmune etiology of hypoparathyroidism since other autoimmune disorders were reported to be found in HHT.
• However, the mechanism underlying the connection between HHT and autoimmune disorders is not yet clarified.
• Nowadays, there are no sufficient data to justify the coexistence of these two rare diseases.