Uncommon association of hypoparathyroidism and Rendu-Osler syndrome

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Introduction

- Rendu-Osler-Weber syndrome (also called Hereditary Haemorrhagic Teleangiectasia) 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 anaemia.

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 ocasions), serum albumin (5.1 g/dl) and corrected calcemia (6.8 mg/dl), high phosphorus (7.4 ng/dl), low urine calcium (5 mg/500 ml, under the reserve of improper collection) and normal 25 hidroxyvitamin 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).

SERUM CALCIUM	**7,1 mg/dl**	**(8.4 – 10.2 mg/dl)**
SERUM ALBUMIN	**5.1 g/dl**	**(3.2 – 5.2 g/dl)**
CORRECTED CALCEMIA	**6.8 mg/dl**	
SERUM PHOSPHORUS	**7.4 ng/dl**	**(2.5 – 4.5 ng/dl)**
URINE CALCIUM	**5 mg/500 ml**	**(5 – 300 mg/24 h)**
SERUM 25 HIDROXYVITAMIN D	**35.19 ng/ml**	**(30 – 100 ng/ml)**
SERUM PTH	**24.96 pg/ml**	**(15 – 65 pg/ml)**

Diagnosis and treatment

- 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.

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