Medullary Thyroid Carcinoma in Multiple Endocrine Neoplasia 2A
A Therapeutic Challenge

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Introduction
Medullary thyroid carcinoma (MTC) occurs in a hereditary pattern in 25% of cases and accounts for approximately 4% of thyroid cancers. Virtually all patients with multiple endocrine neoplasia 2A (MEN2A) develop MTC. MTC aggressiveness and natural history varies according to the RET mutation. Prophylactic thyroidectomy may cure and/or prevent metastatic disease in most cases, which is of paramount importance due to typical chemo and radioresistance.

Case Report
A 27-year-old man with past history of colostomy at five months of age for Hirschsprung disease and total thyroidectomy at age of 14 years old for the genetic diagnosis of MEN2A (C620R RET). Patient's underwent genetic study when his mother was diagnosed with MTC and a germline mutation in RET gene (C620R) was identified. Patient's post-operative histological diagnosis revealed two MTC foci of 1 cm and 0.3 cm. Patient was lost to follow-up at the age of 16. In the last year he reported low back pain radiated to right thigh.

Initial Evaluation
Low back pain
Iliac bone biopsy: lesion compatible with MTC

Examination of thyroid region
Detected two nodules in the left flank of the neck with 3 cm and 2 cm of diameter, hard consistency and absent mobility.

Thyroid
Left surgical site: Heterogeneous and irregular borders nodule (22X7mm) compatible with local recurrence.
Left para-tracheal region: Two communicating nodules with 37 mm and 25 mm (adenopathy?)

Thyroid Vials
- Na123I PET
- 131I-MIBG Scintigraphy
- 131I-DOTATATE Scintigraphy

Surgery
Bilateral cervical lymph node dissection
Subtotal resection of iliac metastasis

Histology
MTC metastasis

Therapeutic Approach
Post-therapeutic 200 mCi 131I-DOTATATE Scintigraphy

Moderate/high uptake: treatment with 131I-MIBG

Moderate/high uptake: ongoing treatment

Conclusion
• CMT is the first manifestation of MEN2A. As shown in this case the age of prophylactic thyroidectomy is of decisive importance to the prognosis.
• Tumour somatostatin receptors heterogeneity may be responsible for different responses to radionuclides. The treatment of metastatic disease is challenging due to the poor response to systemic therapy and/or radiotherapy.