Erdheim-Chester disease and papillary thyroid carcinoma: case report of a common association in a rare disease

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Clinical case reports - Thyroid/Others
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Erdheim-Chester disease

- Rare
- ~550 published cases
- Multisystemic form of Non-langerhans cell histiocytosis CD68+
- Clinical signs:
  - Protein and encompass bone pain
  - Diabetes insipidus
  - Neurologic and constitutional symptoms
  - Retroperitoneal, cutaneous, cardiovascular and pulmonary involvement may also be present

Papillary carcinoma

- The most common thyroid cancer

Both diseases may be associated with BRAF V600E mutation

If BRAF V600E+ on histiocytes, a specific pharmacological treatment is available ( vemurafenib), with better results

BRAF V600E is the most common mutation

Figure 1:
Cervical CT: <1cm hypodense thyroid nodule with no suspicious signs (arrow)

69 years old
Previous diagnosis of BRAF V600E negative Erdheim-Chester disease

Follow up

Thyroid nodule follow up: 6 months later

6 years old
Previous diagnosis of BRAF V600E negative Erdheim-Chester disease

Figure 2:
Thyroid ultrasound: 2cm hypoechoic solid nodule with irregular margins and 12mm partial cystic nodule on the right lobe (TIRADS 4c)

Figure 3:
Specimen of total thyroidectomy

Discharged at day 4
No complications

Papillary Carcinoma

FNA

Histological analysis:
- T1b R0
- BRAF V600E + in papillary cells

Radioactive iodine therapy

1 year follow up
No recurrence

Conclusion:

Erdheim-Chester disease + thyroid nodule

Thyroid nodule should be biopsied, even if its clinical aspect is not suspicious

If papillary carcinoma cells are BRAF V600E+

Does not mean that the Erdheim-Chester histiocytes are BRAF V600E+

If a papillary carcinoma is present

BRAF V600E should not be searched on papillary cells, because this does not change therapy

Introduction of the new armamentarium should be based on BRAF V600E positivity in the histiocytes cells

Bibliography: