BASHKËSHOQËRIMI I KARCINOMËS PAPILLARE TË TIROIDES ME POLIPOZËN ADENOMATOZË FAMILJARE

FRANCESCO CETTA, PAOLO TOTI, LUTFI ALIA, ARMANDO DHAMO*

Summary

ASSOCIATION BETWEEN THYROID PAPILLAR CARCINOMA AND FAMILIAL ADENOMATOUS POLYPOSIS

Thyroid carcinoma is present in about 1–2 % of patients with familial adenomatous polyposis (FAP). Less than 100 cases have been reported in details. Familial papillary thyroid carcinoma has been described as an isolated defect, but has also been associated with non-polyposis colon cancer syndrome (HNPCC). The follicular variant of papillary thyroid carcinoma is a common subtype of papillary thyroid carcinoma and are usually sporadic tumours. We have investigated the suggestion that FAP associated thyroid carcinoma is significantly different morphologically from both papillary and follicular types, and can be considered as a separate entity.

Methods and results. Specimens from three patients with FAP associated thyroid tumours, all but one having single nodules, have been analysed. All three patients belonged to an extended kindred (23 siblings in four generations) who bad genetic analysis and intensive screening for thyroid nodules. Seven patients had the same APC mutation at codon 1061. Pathological examination revealed a typical papillary carcinoma, encapsulated variant, in all patients with follicular areas in one case. Immunohistochemistry was carried out using the following monoclonal antibodies against thyroglobulin, chromogranin A, carcinomebryonic antigen and cytokeratin AE1/AE3. Colour was developed using the APAAP method. The thyroid specimens, were also specifically studied for activation of the RET – PTC oncogen, that seems to be restricted to papillary thyroid carcinoma. Research for activation of RET-PTC was performed using immunohistochemistry. This method based on the fact that RET proto-oncogen is not expressed in thyroid follicular cells unless its expression is driven by activating sequences replacing its 5' portion. Two of three patients had RET – PTC activation (PTC1 isoform).

Conclusions. These findings suggest that the tumours were certainly papillary, at last in the present kindred. Further studies in different families are required for a better understanding of this peculiar tumour and of its biological behaviour. Further studies are required for a deeper insight into FAP associated thyroid carcinoma. They could be of importance not only for nosologic, genetic and pathogenetic purpose, but also for early diagnosis and proper treatment.

---

* Dërguar në Redaksi në Janar 2007, miratuar për botim Shtator 2007

Nga Instituti i Kirurgjisë, Universiteti i Sienës, Itali.
Adresa për letërkëmbim: Cetta F., Instituti i Kirurgjisë, Universiteti i Sienës, Itali