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Papillary thyroid carcinoma as first and isolated tumor in a young woman with MLH1 gene mutation

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The hereditary non-polyposis colorectal cancer(HNPCC), is an autosomal dominant disorder characterized by a strongly increasing risk of developing colorectal cancer and several extra-colonic malignancies, such as carcinomas of endometrium, ovary, ureter, stomach and small intestine. Lynch syndrome is an HNCPP caused by germline mutations in mismatch repair genes (MMR). It has been associated to germline mutations in one of the DNA MMR genes, most frequently MLH1 and MSH2, rarely MSH6 and PMS2. Some cancers, rare in this syndrome, are associated to MMR deficiency, when incidentally diagnosed in one of the components of a Lynch syndrome family. Here we report the case of unusual presentation of papillary classical thyroid carcinoma in a young woman carrying c545+3>G mutation of MLH1gene. A 36 yrs. old woman, was referred to our endocrine unit in 2017 because of the incidental finding of anti-thyroperoxidase autoantibodies presence. History revealed that she carried a c545+3>G mutation of MLH1 gene as component of a Lynch Syndrome family: her father underwent surgery for colorectal cancer before 50 yrs. and after for squamous cell carcinoma of the skin. His sister also underwent surgery for colorectal cancer and endometrial cancer before 50 yrs. For this reason they were submitted to genetic analysis, which confirmed the presence of the same germinal mutation c545+3>G of MLH1 gene. 5/8 first degree relatives had the same germinal mutation, and also patient 'sister carried the same mutation and developed both endometrial cancer and colorectal cancer before 50 years old. She was euthyroid at the first observation and ultrasound imaging of the neck showed an hypoechoic nodule (5x6x11mm) in the left lobe of thyroid gland. Fine needle aspiration biopsy (FNAB) of this nodule was performed and cytology indicated the presence of thyroid carcinoma (TIR5). Pathology confirmed the presence of a classical variant of papillary thyroid carcinoma, with thyroid capsule infiltration (T1a,N0,Mx).Genetic analysis was than performed on the neoplastic thyroid tissue and DNA panel showed a BRAF mutation p V600E in 10% and TERT mutation C228T in 16% of extracted DNA . Radioiodine ablation (1850 MBq), after 0.9 mg of rhTSH was performed and whole body scan(WBS) post- 131I administration showed thyroid bed up-take, without other sites of pathological up-take. Stimulated thyroglobulin (Tg) was 0.6 ng/mL (cut-off<0.1ng/ml), Tg autoantibodies (TgAb) were 145 UI/mL (0-60 UI/mL). Actually, Tg is<0.1 ng/mL, TgAb 15UI/mL. Ultrasound imaging of the neck is negative for the presence of suspicious lymph nodes. No other neoplasms have been diagnosed during four years follow-up. Probably it could be due to an incidental association or to a common genetic background yet to be discovered, which could explain a lower aggressiveness of the syndrome. This case, as the others rare cases of thyroid carcinoma reported in Lynch syndrome, raise the importance of considering individual cancer genetics in familial cancer syndromes, particularly when other cancers do not fit the described syndrome. In fact appropriate genetic testing could be useful to understand the genetic alterations that may explain apparently unrelated cancers.

Biography

Antonella Carbone MD has completed her studies at The Vanvitelli University of Naples. She is actually the chief of the Endocrine Unite of Asm-Matera in the South of Italy. The main field of clinical and scientific interest in differentiated thyroid carcinoma. She has published more than 25 papers in reputated journal and is a member of the Italian Thyroid Cancer Observatory (ITCO).