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Background and objectives

Medullary thyroid carcinoma is a form of significantly aggressive carcinoma originating from parafollicular (C) thyroid cells. Most sporadic medullary carcinomas arise following a somatic mutation of the RET proto-oncogene. To date, the protocols for carrying out RET mutational tests on circulating tumor DNA extracted from peripheral blood are absent and there is no clinical validation study of this type of analysis in patients with medullary thyroid carcinoma. The aim of our study is the validation of the mutation analysis of the RET gene performed on peripheral blood ctDNA, through the use of technologies based on Real Time PCR and digital PCR, in patients with medullary thyroid carcinoma at various stages of disease.

Study design and population

This is a multicenter, cross-sectional study, which includes the patients undergoing total thyroidectomy surgery with subsequent histological diagnosis of sporadic thyroid medullary carcinoma.

In particular, 10 subjects will be selected with non-metastatic medullary thyroid carcinoma and 10 metastatic medullary thyroid carcinoma. Each individual will be made a blood sample to be able to perform the analysis of mutations on the RET gene on free circulating tumor DNA either by Real Time PCR technique or by digital PCR analysis. The mutational investigations will be carried out at the IFO (Digital PCR) and at the CBM (Real Time) to evaluate the concordance and reliability of the method.

Expected Results

The results of this study could allow us to demonstrate that the liquid biopsy is applicable with diagnostic and prognostic benefit to patients with sporadic thyroid medullary carcinoma. As already highlighted in other types of solid neoplasia, the enormous benefit for patients could be that of using this method to monitor follow-up and evaluate the degree of response to treatments.