Retrospective study in patients with simultaneous occurrence of medullary and papillary thyroid carcinoma

M. Appetecchia1, R. Elisei2, M. G. Castagna3, R.Lauretta1, A. Barnabei1, I. Terrenato4, Mian5, S. Mariotti6, L. Fugazzola7, F. Orlandi8, E. Cavedon5, C. Romei2, F. Pani6, M. Calanchini9, P. Loli10, P. Limone11, E. Seregni12, C. Durante13, A. M. Isidori14, D. Van Doorne15, Fabbri9, S. Filetti13, F. Pacini3, A. Lenzi14

1Endocrinology Unit-Istituto Nazionale Tumori Regina Elena Roma, 2Dip.to di Medicina Clinica e Sperimentale, Univ. degli studi di Pisa, 3UOC Endocrinologia, Policlinico S.M. alle Scotte Siena, 4 Biostatistical Unit-Istituto Nazionale Tumori Regina Elena Roma, 5Dip.to di Medicina, Università degli studi di Padova, 6Dip.to Scienze Mediche Internistiche M. Aresu, Az. Osp.-Univ. di Monserrato, Cagliari, 7Dip.to di Scienze Cliniche e di Comunità, Univ. degli Studi di Milano,

800C Medicina Interna, Presidio Osp. Gradenigo Torino, 900C Endocrinologia e Diabetologia, Osp. S. Eugenio e CTO A. Alesini Roma, 10UO Endocrinologia e Malattie del Ricambio, Osp. Niguarda Milano, 11UO Endocrinologia, Diabetologia e Malattie del Metabolismo, Osp. Mauriziano Torino, 12UO Terapia Medico Nucleare ed Endocrinologia, INT Milano, 13Dip.to di Medicina Interna e Specialità Mediche, Sapienza Università di Roma, 14Dip.to di Medicina Sperimentale, Sapienza Università di Roma, 15Endocrinologia Roma.

Baackground

The simultaneous occurrence of medullary carcinoma (MTC) and papillary carcinoma (PTC) in the same thyroid gland is a rather rare phenomenon. In fact, the presence of two synchronous carcinomas in the organ, one of the medullary type and the other papillary is a quite unique phenomenon, deriving these tumors from two different embryological cell lines: the first from the C cells, parafollicular thyroid, the second, also differentiated, from the common follicular cells much more widely represented in the gland. The percentage of association between MTC and PTC (starting from a screening of the MTC population) is about 10%. The cases reported in the literature show the presence, in the same patient, of wellseparated pathologies of MTC and PTC, with a high tendency of cases of papillary carcinoma of a size less than or equal to one centimeter (micro papillary carcinoma). The hypothesis of a common pathogenesis has always been very appealing especially considering that in both tumors alterations of the same oncogenes come into play. It is in fact known that the RET oncogene is mutated in the MTC (activating point mutations) but also in the PTC, although with another mechanism (RET / PTC gene rearrangement). Recently it has been shown that the RAS oncogene, already known to be involved in the pathogenesis of PTC, can also be mutated in MTC. From previous studies, the mutational RET analyzes performed to verify if the prevalence of mutational events were higher in the MTC / PTC group vs the MTC group, showed no differences. On these data it was speculated that the presence of RET mutations in MTC does not increase the risk of developing a PTC. Despite this, it can not yet be ruled out that there is a common pathogenetic mechanism. We also consider that from 2004 to today the methods of analysis have been improved and there is the possibility to analyze more mutations of different oncogenes. Increasing attention has also been paid by the scientific community to the study of microRNAs, which intervening on gene transcription could however play a role in the pathogenesis of these tumors.

Study design and methods

Observational, retrospective, multicentre, non-profit study.

It will collected and evaluated the epidemiological data of patients with MTC and PTC in the same gland (MTC / PTC), in order to perform an analysis of their characteristics and the course of their follow up, compared to patients with only PTC or only MTC. Data collection will be carried out by filling out a form in which the entries relating to the patient's clinicalepidemiological information will be reported. The data will then be inserted into a database for the final statistical evaluation.

PRIMARY OBJECTIVE

Identification of specific epidemiological and pathological characteristics of MTC / PTC cases

SECONDARY OBJECTIVE

If the collection of epidemiological data identifies a consistent series, we will proceed with molecular analysis, to evaluate the pattern of specific molecular alterations of MTC / PTC cases. In this case, it will be necessary to have the availability of paraffin sections of the patient's operating piece.





