Introduction: Medullary thyroid carcinoma (MTC) is a rare malignant tumor originating from parafollicular C cells of the thyroid. Calcitonin and carcinoembryonic antigen (CEA) are widely used as MTC prognostic markers. Recent studies have suggested elevated serum levels of carbohydrate antigen 19.9 (CA 19.9), a well-established tumor marker in pancreatic neoplasms, as a marker of aggressiveness and mortality in advanced MTC. The mechanism by which MTC cells secrete Ca 19.9 has not yet been elucidated, but c-cell dedifferentiation has been suggested as a potential explanation. Objective: To evaluate CA 19.9 expression in MTC samples and correlate it with a cellular dedifferentiation, as well as clinical and laboratory data.

Methods: MTC tumor samples from patients attending the Thyroid Division of a tertiary, University-based Hospital were evaluated for CA 19.9 and CD-133 expression by immunohistochemistry using specific antibodies. The slide reading was performed by a pathologist and the quantification by the h-score method.

Results: MTC tumor specimens from 65 patients were evaluated (43.1% hereditary and 56.9% sporadic). The mean age at diagnosis was 36.02 ± 16.37 years, and 56.9% were female. The median levels of calcitonin and CEA were 527 pg/ml (42.5-1168.7) and 16.1 ng/ml (3-49.65), respectively. At diagnosis, 53.8% of the patients had local, and 21.5% had distant metastases. Some level of CA 19.9 expression was observed in 86.2% of samples, and the median of the h-score was 13 (2-30). CD-133 expression was seen in ~90% of samples analyzed (h-score = 40; 3-120). Of note, despite the positivity of both markers in most of the samples, the h-score values were not correlated (p = 0.78). There were no difference in the expression of CA 19.9 on age, sex, calcitonin and CEA values, calcitonin tissue expression or tumor staging (All P > 0.05). However, interestingly, we observed a significant difference between the h-scores in the hereditary vs. sporadic form (23.5 vs. 3; P = 0.017).

Conclusion: Our results demonstrate CA 19.9 and CD-133 expression in the vast majority of CMT samples, including small tumors in early stages of the disease. Higher levels of CA 19.9 expression were observed in hereditary MTC as compared with those with sporadic disease. However, CA 19.9 expression was not associated with cell dedifferentiation nor advanced MTC disease.