TUMOR MARKERS IN MYOTONIC DISTROPHY TYPE 1: ARE THEY REALLY USEFUL?

A. Pulcini[1], A. Plutino [1], A. Lorusso [1], S. Lattanzi [1], C. Cagnetti [1], M. Silvestrini [1], L. Provinciali [1]

[1]Neurological Clinic, Department of Experimental and Clinical Medicine, Marche Polytechnic University, Ancona

BACKGROUND

Myotonic dystrophy type 1 (MD1) is an autosomal dominant disease caused by the multiple repeat of CTG on chromosome 19. It is a multisystemic and progressive disorder with a wide phenotypic variability: it involves many tissues and organs and is characterized by myotonia, muscles weakness, early-onset cataracts, baldness, and skeletal, cardiac and endocrine disorders. The life expectancy is mostly reduced by lung and cardiac complications, but recent studies have also provided evidence of an increased risk to develop cancer.

The aim of this study was to evaluate the predictive role of the tumor markers (TMs) in patients affected by MD1.

MATERIALS AND METHODS

We retrospectively identified consecutive patients diagnosed with MD1 followed at the Day Hospital of the Neurological Clinic, Ancona, Italy. Data about demographics, medical history, neurological and clinical examination and laboratory findings including the TMs (CEA, CYFRA, NSE, CA 19-9, CA 72-4, alpha-fetoprotein, PSA) were abstracted from medical records. The main study outcome was the occurrence of any cancer during a 10-year follow-up. (table 1)

TMs	TUMOR	HEALTHY
Positive value	12 (17%)	21 (29,5 %)
Negative value	11 (15,5%)	27 (38%)

Table 1: 71 patients with DM1

RESULTS

Seventy-one patients were included in the study, whose 34 (47.8%.) were women. The mean age of the study cohort was 45± 13 years (range 18-66). The TMs resulted abnormally increased in most of the patients (46.5%) whose 12 (17%) was found to suffer from any cancer.
Eleven patients (15.5%) had tumor without any TM alteration (figure 1).

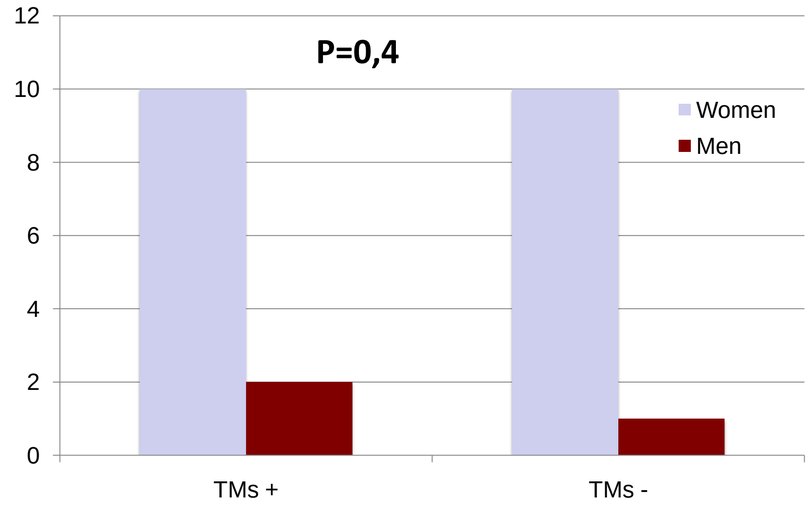

Figure 1: patients with tumor

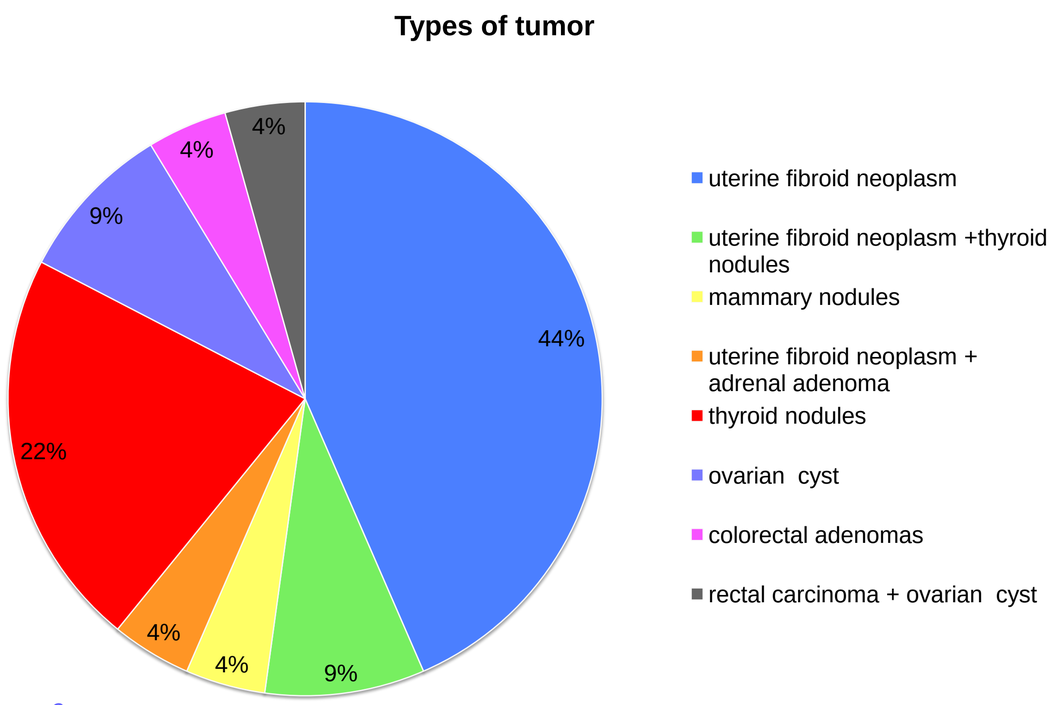

Figure 2

The Neural Enolase Specific (NSE) resulted beyond the upper limit of normal ranges in 22 (66,6%) patients and turned out to be the most frequent altered TM. Overall, near to one third of the study subjects developed tumor; uterine fibroid neoplasm was the most frequent. (figure 2)

CONCLUSIONS

The risk to develop neoplasia wasn't influenced from the TMs levels whose increasing in DM1 patients could be reasonably related to the disease itself. There is no evidence to use the TMs to early diagnosis tumors; instead, their indiscriminate use may lead to inappropriate investigations.

BIBLIOGRAPHY

Das M, Moxley RT 3rd, Hilbert JE, Martens WB, Letren L, Greene MH, Gadalla SM. Correlates of tumor development in patients with myotonic dystrophy. J Neurol. 2012;259:2161-6.
Win AK, Perattur PG, Pulido JS, Pulido CM, Lindor NM. Increased cancer risks in myotonic dystrophy. Mayo Clin Proc. 2012;87:130-5.