Background: Molecular markers in colon cancer are needed for a precise classification and personalized treatment. Almost 20% of pts with sporadic colon cancer present MSI, due to defects in the mismatch repair (MMR) system which is associated with a better prognosis. BRAF mutation is observed approximately in 10% of the colonic tumours and its prognosis correlation is recently being studied. The aim of our study was to determine the mutational status of BRAF in patients with MSI-high (MSI-H) tumours and its effects on clinical outcomes.

Methods: In a consecutive series of 1677 patients with colon cancer diagnosed at the S. Chiara Hospital of Trento from 2004 to 2012 we evaluated the MSI status; later on, in the group with MSI-H tumours, we analysed the V600E BRAF mutation.

Results: MSI-H was observed in 151 cases (~10%); they were 40 Stage I disease (27%), 76 stage II (50%), and 35 stage III (23%). Only 57 pts (38%) have <70 years. BRAF was determinated in 115 of these pts and was mutated in 62 cases (54%): 20/40 stage I, 25/76 stage II and 17/35 stage III; moreover, BRAF mutation was observed in 59% of well-differentiated tumours, in 85% of right side tumours, in 70% of lymph node negative and in 17% of T4 tumours. The median DFS was 27.5 months (range 1 – 71 months) in the BRAF mutated patients and 23 months (range 0.3 – 69.6 months) in the wild type group. At a median follow-up of 29 months (range 0.4 – 72 months), 8 patients (12%) died due to disease in the BRAF mutated group and 9 patients (16%) in the BRAF wild type group.

Conclusion: From our results it appears that BRAF mutation is a quite frequent event in MSI-H colorectal tumors, but it does not seem related to a higher frequency of recurrence and death. These data could suggest that the good prognosis of patients with MSI-H tumours may not be influenced by BRAF mutation.