# CÁNCER COLORECTAL

#### Microsatellite instability in patients with diagnostic of colorectal cancer.

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Rev. gastroenterol. Perú, Lima, v. 36, n. 1, enero 2016.

### Abstract

OBJECTIVE: To determine the presence of microsatellite instability in patients with colorectal cancer using the molecular panel Bethesda and discuss its significance in patients with suspected hereditary nonpolyposis colorectal cancer (HNPCC) or Lynch Syndrome. MATERIALS AND METHODS: We worked with samples of peripheral blood and tumor tissue of 28 patients diagnosed with colorectal cancer referred to the Laboratory of Molecular Biology of the Instituto Nacional de Enfermedades Neoplasicas (INEN), Lima, with suspected of Lynch syndrome. DNA was extracted using kits of nucleic acid extraction of peripheral blood and paraffin-embedded tumor tissue. Five microsatellite markers of Bethesda panel were amplified: BAT25, BAT26, D2S123, D5S346 and D17S250, by polymerase chain reaction. IMS analysis was performed by electrophoresis on chip in the Bioanalyzer Agilent 2100. RESULTS: Of the patients studied, 11 had high IMS(IMS-H) and one could not be fully ranked, staying as MSI-H / IMS-L. In all cases of IMS-H both BAT26 and BAT25 were unstable. The IMS-H in these patients indicates high probability of HNPCC or Lynch syndrome; it must be contrasted with the genetic analysis of MMR genes. CONCLUSION: The technique allowed determine which patients have to continue with the study of system mismatch repair genes, for establish whether we facing to HNPCC or sporadic colorectal cancer.

# Cáncer colorrectal en los jóvenes: factores pronósticos y características clínico patológicas en un instituto del cáncer de Perú.

Ruiz, R., Taxa, L., Ruiz, E. F., Mantilla, R., Casanova, L. & Montenegro, P.

Rev Gastroenterol Peru. 2016 Jan-Mar;36(1):35-42.

### <u>Abstract</u>

OBJECTIVE: To determine clinicopathological features and prognostic factors among young colorectal cancer (CRC) patients in a Peruvian Cancer Institute. METHODS: Data of patients 40 years or younger, admitted between January 2005 and December 2010, were analyzed. RESULTS: During the study period, 196 young patients with CRC were admitted. The tumor was located in the rectum, left colon and right colon in 45.9%, 28.6% and 25.5% of cases. Family history of CRC was found in 13.2% and an autosomal pattern of inheritance, in 8.6% of the cases. The most common symptoms were pain (67.9%) and bleeding (67.3%). The majority (63.1%) of colon cancer cases and more than a third (34.4%) of rectal cancer cases were diagnosed in stage III or IV. The histologic type was tubular, mucinous and signet ring cell adenocarcinoma in 73.5%, 14.8% and 8.6%, respectively. The depth of invasion was T3 in 21.4% and T4 in 53%. Nodal involvement was detected in 44.5%. Fiveyear overall survival (OS) was 44.3%. In the multivariate analysis, only the stage resulted an independent prognostic factor for survival. CONCLUSIONS: CRC in Peruvian young patients is mostly sporadic. It presents more often in the distal colon or rectum and at advanced stages of the disease. Mucinous and signet ring cell carcinoma were requent histological types. Five-year OS stage by stage is similar to that reported in the literature for older patients. Stage was the only independent prognostic factor for survival.

# Lynch syndrome, Muir Torre variant: 2 cases.

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Rev Gastroenterol Peru. 2016 Jan-Mar;36(1):81-5.

# <u>Abstract</u>

Lynch syndrome (LS) is an autosomal-dominant inherited cancer predisposition syndrome caused by germline mutations in DNA mismatch repair genes (MLH1, MSH2, MSH6 or PMS2). Muir-Torre syndrome (MTS) is a phenotypic variant of LS that includes a predisposition to sebaceous glands tumors and keratoacanthomas. We report two patients with MTS, with more than one LS-related cancer, skin lesions, family history of cancer andmicrosatellite instability and immunohistochemistry analysis.