

XXXVII COMU 2018 – Congresso Médico Universitário da FMUSP

Researches Classified – Oswaldo Cruz Awards – Case Report

Intussusception reveals MUTYH-related polyposis syndrome and colorectal cancer: a case report

Academic Authors: **Fernanda Nii, Kayo Augusto de Almeida Medeiros, Bárbara Justo Carvalho,
Diego Ramos Martines, Gustavo Heluani Antunes de Mesquita**

Academic Advisors: **Daniel Reis Waisberg, Alberto Meyer**

Faculdade de Medicina da Universidade de São Paulo - FMUSP

We are reporting a rare case of MUTYH-associated polyposis, a colorectal cancer hereditary syndrome, diagnosed after an intussusception. Colorectal cancer is an important cause of cancer related mortality that can be manifested by an intussusception, a rare occurrence in adults and almost always related to tumors. Approximately 5% of colorectal cancers can be attributed to syndromes known to cause hereditary colorectal cancer, such as MUTYH-associated polyposis, autosomal genetic syndrome associated with this disease. We present the case of a 44 years old male, that sought medical consultation with a complaint of abdominal discomfort, that after five days changed its characteristics. The patient was sent to the emergency department where a CT-scan revealed intestinal sub-occlusion by ileocolic invagination. Right colectomy was carried out. The anatomic-pathological examination revealed a moderately differentiated mucinous adenocarcinoma and multiple sessile polyps, which led to the suspicion of a genetic syndrome. In the genetics analysis two mutations were observed in the MUTYH gene, and MUTYH-associated polyposis was diagnosed. This case demonstrates the importance of meticulous analysis of the patient examination results to identify possible discrete alterations that can lead to improved understanding of disease.

Keywords: MUTYH-associated polyposis; Intussusception; Hereditary colorectal cancer.