Genetic polymorphisms of vitamin D metabolism genes and serum level of vitamin D in colorectal cancer

Abstract
Background: The metabolism of vitamin D is complex, its receptor (VDR) and proteins encoded by the genes CYP27B2 and CYP24A1 can influence vitamin D serum levels. The aim of this study was to investigate the relationship of the polymorphisms of VDR (ApaI and BsmI), CYP27B1 and CYP24A1 with serum vitamin D levels in both forms, 25(OH)D-3 (circulating form) and 1,25(OH)(2)D-3 (active form), in colorectal cancer (CRC) patients. Methods: One hundred fifty-two CRC patients and 321 controls were included. DNA was extracted from peripheral blood. Polymorphisms of Bsml and Apal were identified by PCR-RFLP. Those of CYP24A1 (rs6013897, rs158552 and rs17217119) and CYP27B1 (rs10877012) were determined by gene sequencing. Results: The median serum levels of circulating vitamin D were not different between CRC patients and controls; however, the percentage of those with deficient vitamin D was higher in patients with cancer. The active form of the vitamin D was higher in CRC patients. VDR, CYP27B1 and CYP24A1 polymorphic genotypes had no influence on serum levels of circulating vitamin D. The correlation between circulating and active vitamin D forms was lower among patients with CRC, regardless of the presence or absence of any genetic polymorphism. The mean serum levels of active vitamin D were higher among patients with polymorphic genotype variants of Apa1 or Bsm1. Conclusions: CRC patients had a higher frequency of insufficient vitamin D and a higher concentration of active vitamin D. These concentration were higher between patients with polymorphic genotypes variants of Apal and Bsml, CYP24A1 and CYP27B1. Polymorphic genotypes cause a lower correlation between the forms of vitamin D. (AU)