J Gastroenterol Hepatol. 2016 Apr;31(4):795-801. doi: 10.1111/jgh.13221.

Association of vitamin D receptor gene polymorphisms and serum 25-hydroxyvitamin D levels with Crohn's disease in Chinese patients.

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Abstract

BACKGROUND AND AIM: The vitamin D receptor (VDR) regulates immune responses and inflammation through binding with 1,25-dihydroxyvitamin D, the active form of vitamin D. The serum 25-hydroxyvitamin D (25(OH)D) level clinically reflects vitamin D status in the human body. We investigated the association of VDR polymorphisms and 25(OH)D levels in Chinese patients with Crohn's disease (CD).

METHODS: Vitamin D receptor polymorphisms (FokI, BsmI, ApaI, and TaqI) were genotyped by SNaPshot. Serum 25(OH)D levels were measured by electro-chemiluminescence immunoassay.

RESULTS: A total of 297 patients with CD and 446 controls were recruited. Compared with controls, mutant alleles and genotypes of BsmI and TaqI were less prevalent in patients with CD (all $P < 0.05/4 = 0.0125$). The AAC haplotype formed by BsmI, ApaI, and TaqI was also less prevalent in patients with CD ($P = 0.004$). Furthermore, 124 patients and 188 controls were randomly selected for measurements of 25(OH)D levels. Average 25(OH)D level was lower in patients with CD than in controls (15.46 ± 8.11 vs 21.64 ± 9.45 ng/mL, $P < 0.001$) and negatively linked to CD activity index ($\beta = -0.829$, $P < 0.001$), platelet count ($\beta = -0.253$, $P < 0.001$) and neutrophil percentage ($\beta = -0.136$, $P = 0.005$) in patients with CD. The ApaI mutant genotype and vitamin D deficiency (<20 ng/mL) were independently associated with CD ($P = 0.009$, $P < 0.001$, respectively). In patients with CD, vitamin D deficiency interacted with FokI, ApaI, and TaqI mutant genotypes ($P = 0.027$, $P = 0.024$, and $P = 0.040$, respectively).

CONCLUSIONS: Vitamin D receptor (BsmI, ApaI, and TaqI) mutations and lower 25(OH)D levels are associated with CD in Chinese patients. Moreover, VDR (FokI, ApaI, and TaqI) mutations and vitamin D deficiency may have a combined impact on CD.

KEYWORDS: 25-hydroxyvitamin D; Crohn's disease; polymorphism; vitamin D receptor

PMID: 26513524 DOI: 10.1111/jgh.13221