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Commentary on Mutations in Interleukin-10 Receptor in Inflammatory Bowel Disease in Iranian IBD cohort

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Introduction: Early-onset inflammatory bowel disease (IBD) is a diagnosis of Crohn's disease, ulcerative colitis and inflammatory bowel disease unclassified which runs a chronic, relapsing course and can result in substantial long-term morbidity. IBD is a multifactorial disorder with genetic susceptibility, immunological predisposition and environmental triggers.

Aim: The aim of the study is to determine prevalence of IL10R mutation in IBD patients from Isfahan, Iran.

Materials & Methodology: Total DNA content of each patient was extracted from whole blood with and PCR amplification was done as previously described. We performed sequencing of all exons in IL10RA and IL10RB in cohort of IBD patients and healty control.

Results & Discussions: Overall detection rate of IL-10RA mutations was 69.3% (53/76) and IL10-RB 3.9(3/76) in total patients. Identified IL-10RA mutations were P.(I224V), P.(A153V), P.(A153A), P.(S159G), P.(R263Q), P.(R284C), P.(R351Q), P.(Q376Q), P.(T416I), P.(A493V), P.(A511A) and P.(S563S)and IL10RB mutation was P.(K47E). Of them, P.(A153V), P.(A153A), P.(R284C), P.(T416I), P.(A493V), P.(A511A), P.(S563S) were not reported variant with IBD variants. The most common mutations were P.(A153A) and P (R361G) found in 48 out of 76 patients (63.1%). Like all studies which demonstrate relation between IL10R mutation and IBD our results also confirmed that early-onset IBD could be attributed to a synergistic effect of several variant alleles of the genes encoding IL10 receptors. These variants, alone, could only give rise to a sub-clinical manifestation of the IBD.

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