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Commentary on mutations in interleukin-10 receptor in inflammatory bowel disease in Iranian IBD cohort

Razieh Khoshnevisan and **Roya Sherkat**
Isfahan University of Medical Sciences, Iran

Introduction: Early-onset inflammatory bowel disease 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. To generally determine prevalence of IL10R mutation in IBD patients in Iran-Isfahan, we performed sequencing of all exons in IL10RA and IL10RB in a cohort of IBD patients and healthy control.

Materials & Methods: Total DNA content of each patient was extracted from whole blood with and PCR amplification was done.

Results & Discussion: 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) and P.(S563S) were not reported variant in IBD variants. The most common mutations were p.(A153A) and p.(R361G) which found 63.1% (48/76) patients. Like as 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.

Biography

Razieh Khoshnevisan born on September 5th, 1985 in Qom, Iran. She is currently PhD Student in medical immunology at the Medical University of Esfahan, Iran under the supervision of Prof. Roya Sherkat and Prof. Abbas Rezaie. and pass around two years internship in lab of prof christoph klein-munich -Germany. She will be involved in the recruitment of children with primary immunodeficiencies and very early onset inflammatory bowel disease from Iran. Our collaboration will provide Iranian patients with rare diseases access to modern genetic diagnostics and therapies.

razikhosh@yahoo.com

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