

## Genetic Markers Associated With Inflammatory Bowel Disease - Detection of NOD1, 2 Mutations In Inflammatory Bowel Disease Among Sudanese Patients

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### Abstract

Inflammatory Bowel Disease (IBD) comprising Ulcerative Colitis (UC) and Crohn's Disease (CD) is debilitating chronic immune disorder of the intestinal mucosa, multigenic in nature, resulting from dysfunctional interactions between the intestinal immune system and its micro flora, influenced by host genetic susceptibility as suggested in linkage, epidemiologic, racial, familial aggregation and twin studies. It has led to the discovery of mutations in nucleotide-binding oligomerization domain containing protein 2 (NOD2) also known as caspase recruitment domain containing protein 15 (CARD15) or inflammatory bowel disease protein 1 (IBD1) which located in chromosome 16q12 and associated with ileal CD, also numerous other genes' mutations have been found to be associated with IBD susceptibility such as NOD1/CARD4 mutations in chromosome 7p14.3 in UC patients. Nod-like receptor family plays a key role in realization of innate and adaptive immune response. Their polymorphisms may shift balance between pro- and antiinflammatory cytokines, modulating the risk of chronic inflammation. The rational and prevalence of IBD is dependent on geographic location and racial background. The study met its objective in determining whether genetic, environment and socio-economical components contribute to the development of IBD in Sudanese population and furthermore, whether the race and dietary intake associated. Different groups' joined this study were from Sudanese population. They had been divided into two; patients with colonoscopy diagnosed IBD and individuals who don't have IBD as controls. Each group was classified according to gender, age and race. Blood samples (Biospecimen Retention) from the included participants at the Gastroenterology Department in Ibn-Sina Hospital were collected and sent to Al-Neelain Medical Research Centre for DNA purification and isolation, Polymerase Chain Reaction (PCR) amplification, establishment of sequence analysis and product identification of genetic polymorphisms associated with IBD. Finally genotyping was the end

point. Also, a questionnaire was designed to collect data in such a way to achieve objectives of the study. The collected data was tabulated and analyzed using Statistical Package for Social Sciences (SPSS 14.0) computer program. The study revealed that Shaigeia and Ga'aleia (Northern tribes) were found to be more affected by Ulcerative Colitis than other tribes (13.3%). Thus, suggesting possible link with ethnic origin. Also, it resulted that ulcerative colitis was associated with the economic status of the patient. Subjects with low economic status were found to be more likely to develop it (60%). The most important result after the laboratory experiments was the obvious association of ulcerative colitis with the allelic mutation in NOD1 (70%). While Crohn's disease was not found to be associated with (-/C) allele insertion.

**Biography:** Asmaa AlFadil Hassan Khalifa has completed her BSc from University of Gezira School of Pharmacy and MSc studies from University of Medical Sciences and Technology, Sudan. In 2018, she has completed a Diploma in IV Compounding Techniques from Notting Hill College, Manchester, United Kingdom. She is a Clinical Pharmacy Specialist and a Member of Sudan Medical Council. She is licensed as Clinical Pharmacist in Department of Health, Abu Dhabi, United Arab Emirates also a BLS Provider in American Heart Association. She did a research in Injection Safe Disposal: Safe Disposal Policy in Compliance with WHO recommendations in 2010 for her BSc thesis sponsored by WHO. Late in 2013 she presented her second research: Genetics Markers associated with Inflammatory Bowel Disease: Detection of NOD1,2 Mutations in Inflammatory Bowel Disease among Sudanese Patients, in completion of her thesis for MSc degree. Through her professional journey she worked as Community Pharmacist, House-Officer Pharmacist, Hospital In-Charge Pharmacist and Clinical Pharmacist. She joined many facilities while she was in Sudan which include: Paediatric Hospital, Oncology Institute, Khartoum Teaching Hospital and Police Hospital. In UAE she worked in Tawam Hospital as a Trainee and lately worked for Canadian Medical Centre.