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Genetic Risk Variants are Common in Native-Born and Immigrant Pediatric Inflammatory Bowel Disease Patients from Minoritized Backgrounds

Introduction: IBD comprised of Crohn's disease (CD) and ulcerative colitis (UC), are traditionally thought of as affecting individuals of European and Ashkenazi Jewish heritage. There is a significant genetic component to disease risk, and genome-wide association studies (GWAS) have identified over 200 risk genes. Majority of IBD patients at our center are of Afro-Caribbean descent, and many are first- or second-generation immigrants. Frequency and significance of many risk alleles is unknown in our population; thus, we aimed to describe the frequency of a limited number of risk alleles.

Methods: Retrospective chart review of individuals aged 0-21 diagnosed with IBD between 2009-2024 was done. Data collected from the electronic medical record (EMR) included demographics, including ethnic background and immigration status, clinical characteristics, disease phenotype, and laboratory results, including the Prometheus Laboratories SGI panel, which reports genotypes for known IBD risk variants in ATG16L1, ECM1, NKX2-3, and STAT3.

Results: 19 patients met inclusion criteria and had sufficient data available for analysis. 4 were native-born Americans (21.1%), 3 first-generation immigrants (15.8%) and 12 second-generation immigrants (63.2%). Majority were African American (10, 52.6%), 2 Hispanics (10.5%), and 7 from other ethnic backgrounds (36.8%). We noted a high frequency of disease associated variants in ATG16L1 and STAT3, with less frequency in ECM1 and NKX2-3. Second-generation immigrant exhibited the highest genotypic diversity across all genes, particularly with variants in ATG16L1 and STAT3. African Americans showed the highest genetic diversity, predominantly within the second generation immigrant group, with significant representation of both ATG16L1 and STAT3.

Discussion: Our data reveals that IBD risk alleles, well-documented in Caucasians, are also common in Afro-Caribbean patients, though their effect remains unclear, highlighting the need for further study in diverse populations.