Title: Health Disparities in Colorectal Cancer: A Comparative Analysis of SNPs in Hispanic and Non-Hispanic White Populations

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Abstract:

Colorectal cancer (CRC) is the second most dangerous cancer in terms of death rate and considered as a significant public health challenge, with disparities in early diagnosis and outcomes observed particularly in Hispanic populations compared to Non-Hispanic Whites. This study aims to address this issue by identifying single nucleotide polymorphisms (SNPs) and differentially expressed genes associated with those SNPs as novel biomarkers for early detection of CRC in Hispanic individuals utilizing RNA sequence data from Hispanics (H) and Non-Hispanic Whites (NHW) CRC tissues and their respective normal tissues adjacent to the tumor (NATs) followed by a comprehensive bioinformatics analysis.

The Genome Analysis Toolkit (GATK) software was used for the identification of SNPs and indels. Additionally, the Snpeff software was employed for annotating and predicting the functional consequences of identified SNPs. The gene expression levels were quantified and subsequently, the DEGs in the mentioned populations were identified employing tools like featureCounts and DESeq2. Potential SNPs in Hispanic CRC were narrowed down by screening the SNP data. 134 unique SNPs and 21 DEGs were identified from the screening step. Afterward, pathway analysis was done to reveal the biological pathways influenced by these DEGs associated SNPs and how these pathways play a vital role in progression of the cancer. From the pathway analysis, we found out that PTCD3 and MRPL28 enriched in the mitochondrial translation pathway and NUP214, a nuclear pore complex protein, enriched in SUMOylation (small ubiquitin-like modifier) pathways are responsible for carcinogenesis in Hispanic CRC.

The outcome of the study is promising since identifying novel biomarkers specific to the Hispanic population through the experiment could be a breakthrough for early detection and personalized treatment of colorectal cancer (CRC) within this population.

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