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Assessment of Genetic Variability in an Inflammatory Bowel Disease Patients Population by a Clinical Exome Survey

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ABSTRACT

Inflammatory bowel disease (IBDs) is complex, multifactorial disorder that comprise Crohn's disease (CD) and ulcerative colitis (UC). Recent discoveries have brought much attention to the genetic predisposition of patients with IBD. Considering that IBDs are genetics and multi-factorial diseases, we embarked here in assessing genetic diversity in four (4) IBDs pediatric patients (patient 1, 2, 3 and 4) previously submitted to a clinical exome sequencing. Genomic sequences obtained from that analysis were aligned on hg19 human genome by bowtie2 package in Galaxy platform. Next, we performed variant features calling (VCF) analysis. Focusing on genetic variants covered by at least 20 reads, we selected and quantified genomic functions due to exonic and intronic mutations in each IBDs patients. Multivariate statistical analysis as well as analysis of variance (ANOVA) from R package, applied to that quantitative data do not support any variance difference between IBD patients ($p>0.05$). The same surveys, by applying "fitcon" parameter (fitcon ≥ 0.6 for mutation in exon and $0.3 \leq$ fitcon ≤ 0.4 for mutation in intron), indicating the probability that a given mutation significantly impacts the phenotype, attributed genetic variability in the IBD population to intronic mutations ($p=0.20$), contrary to exonics mutations ($p=0.97$). Pair-wise statistical test applying the same fitcon parameters confirmed intron mutations as favoring genetic variability. The same statistical test shown a relative difference between IBD patient 2 and IBD patients 1 ($p=0.24$), 3 ($p=0.31$) and 4 ($p=0.34$) respectively, confirming Crohn's phenotype of the latter by contrast to IBD patient 2 ulcerative colitis phenotypes. Our survey suggested the integration between biostatistics and next generation sequencing, as a valid support in characterizing genetic diversity in an IBD population.

Keywords: Inflammatory bowel disease, Crohn disease, Exome sequencing, Biostatistics

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