World Academy of Science, Engineering and Technology
International Journal of Biotechnology and Bioengineering
Vol:18, No:12, 2024

Computational Pipeline for Lynch Syndrome Detection: Integrating Alignment, Variant Calling, and Annotations

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Abstract : Lynch Syndrome is an inherited genetic condition associated with an increased risk of colorectal and other cancers. Detecting Lynch Syndrome in individuals is crucial for early intervention and preventive measures. This study proposes a computational pipeline for Lynch Syndrome detection by integrating alignment, variant calling, and annotation. The pipeline leverages popular tools such as FastQC, Trimmomatic, BWA, bcftools, and ANNOVAR to process the input FASTQ file, perform quality trimming, align reads to the reference genome, call variants, and annotate them. It is believed that the computational pipeline was applied to a dataset of Lynch Syndrome cases, and its performance was evaluated. It is believed that the quality check step ensured the integrity of the sequencing data, while the trimming process is thought to have removed low-quality bases and adaptors. In the alignment step, it is believed that the reads were accurately mapped to the reference genome, and the subsequent variant calling step is believed to have identified potential genetic variants. The annotation step is believed to have provided functional insights into the detected variants, including their effects on known Lynch Syndrome-associated genes. The results obtained from the pipeline revealed Lynch Syndrome-related positions in the genome, providing valuable information for further investigation and clinical decision-making. The pipeline's effectiveness was demonstrated through its ability to streamline the analysis workflow and identify potential genetic markers associated with Lynch Syndrome. It is believed that the computational pipeline presents a comprehensive and efficient approach to Lynch Syndrome detection, contributing to early diagnosis and intervention. The modularity and flexibility of the pipeline are believed to enable customization and adaptation to various datasets and research settings. Further optimization and validation are believed to be necessary to enhance performance and applicability across diverse populations.

Keywords : Lynch Syndrome, computational pipeline, alignment, variant calling, annotation, genetic markers
Conference Title : ICBCMB 2024 : International Conference on Bioinformatics and Computational Molecular Biology
Conference Location : Cairo, Egypt
Conference Dates : December 16-17, 2024