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Functional Annotation of Cancer Variants: Tools And Techniques

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Abstract: *Cancer is a complex genetic disease driven by somatic and germline mutations that alter critical cellular functions. With the advancement of Next-Generation Sequencing (NGS), researchers can now identify these mutations at high throughput; however, interpreting their biological and clinical relevance requires robust functional annotation. This study investigates the mutational landscape of prostate, breast, and pancreatic cancers using public RNA-seq datasets and an open-source bioinformatics pipeline. Key oncogenic drivers and tumor suppressor genes, including BRCA1, BRCA2, TP53, and KRAS, were analyzed using tools such as FastQC, Bowtie2, Samtools, FreeBayes, and SnpEff. Our findings highlight the predominance of missense mutations and frequent transition substitutions such as C→T and G→A, with silent and nonsense mutations also contributing to disease mechanisms. These insights emphasize the utility of integrative computational tools in variant annotation and their potential to enhance cancer diagnostics, prognostics, and targeted therapy selection in precision oncology.*

Keywords: Functional annotation, BRCA1, BRCA2, Prostate cancer, Breast cancer, Pancreatic cancer, NGS, Precision oncology

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