Cancer Genomics: Chapter 9. Bioinformatics for Cancer Genomics

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Advances in high-throughput sequencing technologies have enabled cost-effective sequencing of a single human genome at an unprecedented rate, facilitating scientific endeavours never imagined possible before. These improvements have transformed the field of cancer genomics, allowing the complete molecular characterization of individual cancer genomes. However, the promise of unveiling the complexity of cancer has lent itself to yet another level of complexity, the task of managing and integrating the massive amount of data that is generated as part of such experiments. There is a need to manage and store large sequence datasets such that they can be accessed and shared readily but, more importantly, there is a need for their thorough and efficient analysis. Developments and improvements in computer hardware and processing power have eliminated the data storage and access issues. Additionally, bioinformatic algorithms and software, designed specifically for the analysis of cancer genomic data, are now able comprehensively to profile the mutations in a cancer sample, to provide a probability score for their role as disease drivers and to identify potential actionable targets. Although the functional validation of putative driver mutations will remain a necessity, continued improvements in sequencing technologies and analysis tools promise to provide increasingly reliable computational analysis of cancer genomes.

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