“Clinical Bioinformatics Methods to Study Genomic Mutational Profiles in Cancer Disease”

Abstract:
Advances in next generation sequencing (NGS) technologies make it possible to comprehensively identify and characterize genetic variation in population samples creating a foundation for understanding human disease. Many bioinformatics steps are required to translate the raw NGS reads into high-quality variant calls. Conceptually, steps can include (1) pre-alignment quality assessment; (2) alignment of reads to a reference; (3) post-alignment quality assessment; (4) variant calling; (5) variant filtering, annotation and prioritization. These types of bioinformatics approaches are presented using colon cancer as a case study. The genomic mutational profiles for a clinical subset of colon cancer patients are presented. The case study illustrates how cutting-edge clinical bioinformatics can be successfully applied to the study of colon cancer disease with the overall aim of broadening the path for novel and improved developments in precision health and personalized theranostics for colon cancer.