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Structural variant analysis of a cancer reference cell line sample using multiple sequencing technologies

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Abstract

Background: The cancer genome is commonly altered with thousands of structural rearrangements including insertions, deletions, translocation, inversions, duplications, and copy number variations. Thus, structural variant (SV) characterization plays a paramount role in cancer target identification, oncology diagnostics, and personalized medicine. As part of the SEQC2 Consortium effort, the present study established and evaluated a consensus SV call set using a breast cancer reference cell line and matched normal control derived from the same donor, which were used in our companion benchmarking studies as reference samples.

Results: We systematically investigated somatic SVs in the reference cancer cell line by comparing to a matched normal cell line using multiple NGS platforms including Illumina short-read, 10X Genomics linked reads, PacBio long reads, Oxford Nanopore long reads, and high-throughput chromosome conformation capture (Hi-C). We established a consensus SV call set of a total of 1788 SVs including 717 deletions, 230 duplications, 551 insertions, 133 inversions, 146 translocations, and 11 breakends for the reference cancer cell line. To independently evaluate and cross-validate the accuracy of our consensus SV call set, we used orthogonal methods including PCR-based validation, Affymetrix arrays, Bionano optical mapping, and identification of fusion genes detected from RNA-seq. We evaluated the strengths and weaknesses of each NGS technology for SV determination, and our findings provide an actionable guide to improve cancer genome SV detection sensitivity and accuracy.



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Cancer Reference

National Cancer Institute (U.S.)



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Oxford Desk Reference: Oncology

Thankamma Ajithkumar, Sarah Jane Jefferies, 2021 This easy to read practical guide distils and compiles all the disparate literature on cancer into one succinct volume It includes the essential evidence based clinical guidelines needed for the safe and effective management of patients with cancer and has a clear layout to allow for quick reference whilst on the ward All aspects of cancer and its management are covered including prevention screening diagnosis and treatment The text begins by outlining the clinical approach to suspected cancer and the principles of multidisciplinary prevention and management It then progresses through site specific cancer management including head and neck CNS thoracic breast gastrointestinal system genitourinary system female genital system skin musculoskeletal system haemopoietic system and endocrine Later chapters cover oncological emergencies and acute oncology and special situations such as cancer in younger and older people and pregnancy and fertility The guide also offers information about coping with the lifestyle and social issues that may arise with a diagnosis of cancer such as insurance travel and support and includes a chapter dedicated to palliative care for the cancer patient A unique appendix of clinical management flowcharts assists fast appropriate decision making

Encyclopedic Reference of Cancer Manfred Schwab, 2001-10-15 This comprehensive encyclopedic reference provides rapid and focused information about topics of cancer research for the clinical and basic scientist students and informed laymen It will be readily accessible both electronically and in print such that it will be of value to both the scientific community and the public

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