Resource Summary Report

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Breakpointer

RRID:SCR_005254
Type: Tool

Proper Citation

Breakpointer (RRID:SCR_005254)

Resource Information

URL: https://github.com/ruping/Breakpointer

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Description: A fast tool for locating sequence breakpoints from the alignment of single end reads (SE) produced by next generation sequencing (NGS). It adopts a heuristic method in searching for local mapping signatures created by insertion/deletions (indels) or more complex structural variants(SVs). With current NGS single-end sequencing data, the output regions by Breakpoint mainly contain the approximate breakpoints of indels and a limited number of large SVs. Notably, Breakpointer can uncover breakpoints of insertions which are longer than the read length. Breakpointer also can find breakpoints of many variants located in repetitive regions. The regions can be used not only as a extra support for SV predictions by other tools (such as by split-read method), but also can serve as a database for searching variants which might be missed by other tools. Breakpointer is a command line tool that runs under linux system. Breakpointer takes advanage of two local mapping features of single-end reads as a consequence of indel/SVs: 1) non-uniform read distribution (depth skewness) and 2) misalignments at the boundaries of indel/SVs. These features are summarized as breakpoint signature. Breakpointer proceeds in three stages in capturing this signature. It is implemented in C++ and perl. Input is the file or files containing alignments of single-end reads against a reference genome (in .BAM format). Output is the predicted regions containing potential breakpoints of SVs (in .GFF format). To be able to read in .BAM files, Breakpointer requires bamtools API, which users should install beforehand.

Abbreviations: Breakpointer

Resource Type: software resource

Keywords: next-generation sequencing, c++, perl, bio.tools

Funding:

Availability: GNU General Public License

Resource Name: Breakpointer

Resource ID: SCR_005254

Alternate IDs: biotools:breakpointer, OMICS_00308

Alternate URLs: https://bio.tools/breakpointer

Record Creation Time: 20220129T080229+0000

Record Last Update: 20250420T014247+0000

Ratings and Alerts

No rating or validation information has been found for Breakpointer.

No alerts have been found for Breakpointer.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 9 mentions in open access literature.

Listed below are recent publications. The full list is available at RRID.

Dalin S, et al. (2023) Double-strand break repair-associated intragenic deletions and tandem duplications suggest the architecture of the repair replication fork. bioRxiv : the preprint server for biology.

Dentro SC, et al. (2021) Characterizing genetic intra-tumor heterogeneity across 2,658 human cancer genomes. Cell, 184(8), 2239.

Cameron DL, et al. (2019) Comprehensive evaluation and characterisation of short read general-purpose structural variant calling software. Nature communications, 10(1), 3240.

Mei Y, et al. (2017) Genomic profile of human meningioma cell lines. PloS one, 12(5), e0178322.

Shang X, et al. (2017) Rapid Targeted Next-Generation Sequencing Platform for Molecular

Screening and Clinical Genotyping in Subjects with Hemoglobinopathies. EBioMedicine, 23, 150.

Kasar S, et al. (2015) Whole-genome sequencing reveals activation-induced cytidine deaminase signatures during indolent chronic lymphocytic leukaemia evolution. Nature communications, 6, 8866.

Shankar GM, et al. (2014) Sporadic hemangioblastomas are characterized by cryptic VHL inactivation. Acta neuropathologica communications, 2, 167.

Drier Y, et al. (2013) Somatic rearrangements across cancer reveal classes of samples with distinct patterns of DNA breakage and rearrangement-induced hypermutability. Genome research, 23(2), 228.

Williams LJ, et al. (2012) Paired-end sequencing of Fosmid libraries by Illumina. Genome research, 22(11), 2241.