

Resource Summary Report

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mutationSeq

RRID:SCR_006815

Type: Tool

Proper Citation

mutationSeq (RRID:SCR_006815)

Resource Information

URL: <http://compbio.bccrc.ca/software/mutationseq/>

Proper Citation: mutationSeq (RRID:SCR_006815)

Description: A software suite using feature-based classifiers for somatic mutation prediction from paired tumour/normal next-generation sequencing data. mutationSeq has the advantages of integrating different features (e.g., base qualities, mapping qualities, strand bias, and tailed distance features), and validated somatic mutations to make predictions. Given paired normal/tumour bam files, mutationSeq will output the probability of each candidate site being somatic.

Abbreviations: mutationSeq

Resource Type: software resource

Defining Citation: [PMID:22084253](#)

Keywords: next-generation sequencing, somatic mutation, tumor, normal

Related Condition: Tumor, Normal

Resource Name: mutationSeq

Resource ID: SCR_006815

Alternate IDs: OMICS_00086

Record Creation Time: 20220129T080238+0000

Record Last Update: 20260801T190317+0000

Ratings and Alerts

No rating or validation information has been found for mutationSeq.

No alerts have been found for mutationSeq.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 24 mentions in open access literature.

Listed below are recent publications. The full list is available at [ASWG](#) .

Zeng PYF, et al. (2024) The genomic and evolutionary landscapes of anaplastic thyroid carcinoma. Cell reports, 43(3), 113826.

Deyell RJ, et al. (2024) Whole genome and transcriptome integrated analyses guide clinical care of pediatric poor prognosis cancers. Nature communications, 15(1), 4165.

Darbandsari A, et al. (2024) AI-based histopathology image analysis reveals a distinct subset of endometrial cancers. Nature communications, 15(1), 4973.

Titmuss E, et al. (2024) Exploration of Germline Correlates and Risk of Immune-Related Adverse Events in Advanced Cancer Patients Treated with Immune Checkpoint Inhibitors. Current oncology (Toronto, Ont.), 31(4), 1865.

Titmuss E, et al. (2023) Immune Activation following Irbesartan Treatment in a Colorectal Cancer Patient: A Case Study. International journal of molecular sciences, 24(6).

Vázquez-García I, et al. (2022) Ovarian cancer mutational processes drive site-specific immune evasion. Nature, 612(7941), 778.

Reisle C, et al. (2022) A platform for oncogenomic reporting and interpretation. Nature communications, 13(1), 756.

Tessier-Cloutier B, et al. (2022) The impact of whole genome and transcriptome analysis (WGTA) on predictive biomarker discovery and diagnostic accuracy of advanced malignancies. The journal of pathology. Clinical research, 8(4), 395.

Pilsworth JA, et al. (2021) Adult-type granulosa cell tumor of the ovary: a FOXL2-centric disease. The journal of pathology. Clinical research, 7(3), 243.

Liu LY, et al. (2020) Quantifying the influence of mutation detection on tumour subclonal reconstruction. Nature communications, 11(1), 6247.

Salcedo A, et al. (2020) A community effort to create standards for evaluating tumor subclonal reconstruction. Nature biotechnology, 38(1), 97.

Williamson LM, et al. (2019) Genomic characterization of a well-differentiated grade 3 pancreatic neuroendocrine tumor. Cold Spring Harbor molecular case studies, 5(3).

Chun HE, et al. (2019) Identification and Analyses of Extra-Cranial and Cranial Rhabdoid Tumor Molecular Subgroups Reveal Tumors with Cytotoxic T Cell Infiltration. Cell reports, 29(8), 2338.

Thibodeau ML, et al. (2018) Whole genome and whole transcriptome genomic profiling of a metastatic eccrine porocarcinoma. NPJ precision oncology, 2(1), 8.

Echeverria GV, et al. (2018) High-resolution clonal mapping of multi-organ metastasis in triple negative breast cancer. Nature communications, 9(1), 5079.

Ko JJ, et al. (2018) Whole-genome and transcriptome profiling of a metastatic thyroid-like follicular renal cell carcinoma. Cold Spring Harbor molecular case studies, 4(6).

Prentice LM, et al. (2018) Formalin fixation increases deamination mutation signature but should not lead to false positive mutations in clinical practice. PloS one, 13(4), e0196434.

Grewal JK, et al. (2017) Detection and genomic characterization of a mammary-like adenocarcinoma. Cold Spring Harbor molecular case studies, 3(6).

Thibodeau ML, et al. (2017) Genomic profiling of pelvic genital type leiomyosarcoma in a woman with a germline CHEK2:c.1100delC mutation and a concomitant diagnosis of metastatic invasive ductal breast carcinoma. Cold Spring Harbor molecular case studies, 3(5).

Parker JD, et al. (2016) Molecular etiology of an indolent lymphoproliferative disorder determined by whole-genome sequencing. Cold Spring Harbor molecular case studies, 2(2), a000679.