

# MultiMuC

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MultiMuC is a **somatic mutation caller for multi-regional tumors**.

This method can use the stochastic modeling of the state-of-the-art mutation calling methods for single-regional tumors, e.g., Strelka2, NeuSomatic, through the probability-based outputs. Furthermore, this method can avoid a case of performance degradation ("No-TP" case defined in our paper).

## How to build

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This script is implemented in Julia language. We checked the result in Julia v1.0.1. Install julia language before using our script.

## Instantiate package

Before running the script, please instantiate the package and install dependencies.

```
% cd ./src
% julia
julia> ]
(v1.0) pkg>
(v1.0) pkg> activate .
(src) pkg> instantiate
```

## How to run

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Note. At the first execution, package building will be conducted, and simultaneous run (running in multiple processes) should be conducted after the first time of running.

To execute, type as follows.

```
julia --project ./src/main.jl MAP \\  
--seed ${seed} -n ${iter} -b ${burnin} \\  
--pdata=0.02 --pevidence=0.2 --pcon=0.999 --theta=0.5 --rho=0.1 \\  
-o ${output_dir} ${BF_L} ${BF_H}
```

where

$\backslash\{\text{seed}\}$ : a random seed.

$\backslash\{\text{iter}\}$ : number of sampling after burnin.

$\backslash\{\text{burnin}\}$ : number of sampling in burnin of MCMC.

$\backslash\{\text{BF\_L}\}$ : a tsv file for  $\backslash\log_{10}L_{i,j}$ .

$\backslash\{\text{BF\_H}\}$ : a tsv file for  $\backslash\log_{10}H_{i,j}$ .

The following shows an example setting of  $L_{i,j}$ ,  $H_{i,j}$ .

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$L_{i,j} = \begin{cases}$

$\text{BF}_{i,j} \cdot 10^a$  &  $\{\text{Bayes factor is available}\} \backslash$

$10^{-2}$  &  $\{\text{Otherwise}\} \backslash$

$\end{cases}, \backslash$

$H_{i,j} = \begin{cases}$

$\text{BF}_{i,j} \cdot 10^{-1.5}$  &  $\{\text{Bayes factor is available}\} \backslash$

$10^{-2}$  &  $\{\text{Otherwise}\} \backslash$

$\end{cases},$

\$\$

where

$i$ : the sample index

$j$ : the genomic position

$\text{BF}_{i,j}$ : the original Bayes factor from a state-of-the-art mutation calling method.

$\frac{1}{10^a}$ : threshold of original Bayes factor to detect mutation, e.g.,  $a=0.0$  or  $a=-0.5$ .

## Input format

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#Comments 1

gene\_1  $\backslash\log_{10}L_{1,1}$   $\backslash\log_{10}L_{2,1}$   $\backslash\log_{10}L_{3,1}$

gene\_2  $\backslash\log_{10}L_{1,2}$   $\backslash\log_{10}L_{2,2}$   $\backslash\log_{10}L_{3,2}$

gene\_3  $\backslash\log_{10}L_{1,3}$   $\backslash\log_{10}L_{2,3}$   $\backslash\log_{10}L_{3,3}$

#Comments 2

gene\_1  $\backslash\log_{10}H_{1,1}$   $\backslash\log_{10}H_{2,1}$   $\backslash\log_{10}H_{3,1}$

gene\_2  $\backslash\log_{10}H_{1,2}$   $\backslash\log_{10}H_{2,2}$   $\backslash\log_{10}H_{3,2}$

gene\_3  $\backslash\log_{10}H_{1,3}$   $\backslash\log_{10}H_{2,3}$   $\backslash\log_{10}H_{3,3}$

## Output format

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#Comments 1 are copied

gene\_1  $Y_{1,1}$   $Y_{2,1}$   $Y_{3,1}$

gene\_2  $Y_{1,2}$   $Y_{2,2}$   $Y_{3,2}$

gene\_3    $Y_{1,3}$     $Y_{2,3}$     $Y_{3,3}$

where  $Y_{i,j}$  is the inferred state of the mutation by our method in  $i$ -th sample at  $j$ -th genomic position.

## Publication

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The result will appear at the proceedings of ISMCO2019 (<http://ismco.net/>) in Springer LNCS format.

## License

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