### **BS-SNPer User Guide**

version 1.0, 2015/06/11

### 1. Introduction

BS-SNPer is an ultrafast and memory-efficient package, a program for BS-Seq variation detection from alignments in standard BAM/SAM format (Li, et al., 2009). The purpose of this program is to call SNP using bisulfite sequencing data.

# 2. System requirement

BS-SNPer works on Unix (Linux, Ubuntu, MacOSX, etc) based systems.

#### Hardware requirements

One computing node equipped with at least 8 GB Memory

### Software requirements

Linux 64-bit version GCC 4.6.0 or higher

Perl 5.16.3 or higher

# 3. Getting started

#### **Install**

Download BS-Snper from https://github.com/hellbelly/BS-Snper. After extracting the downloaded package, execute the command ./BS-Snper.sh. Make sure the executable file rrbsSnp is generated.

#### Run

You can run BS-Snper in Linux or MAC OS, using the command like:

perl BS-Snper.pl --interval hg19.len --fa hg19.fa --input merge.sort.bam --output SNP.candidate --methoutput Meth.out --minhetfreq 0.1 --minhomfreq 0.85 --minquali 15 --mincover 10 --maxcover 1000 --minread2 2 --errorate 0.02 --mapvalue 20 >SNP.out2 2>SNP.log

# 4. Input file

Any alignments in standard BAM/SAM format.

## 5. Output files

Meanings of the fields of the output SNP information are:

- 1. CHROM: Chromosome
- 2. POS: Coordinate
- 3. ID: Just like vcf format, but in this version, we didn't open this function. The user can get the information according to the DBSNP dataset easily.
  - 4. Ref: reference base(s): Each base must be one of A,C,G,T (case insensitive).
  - 5. ALT: alternate base(s).
  - 6. QUAL: quality: Phred-scaled quality score.
  - 7. FILTER: filter status: PASS if this position has passed all filters, i.e. a call is made at this position.
  - 8. GENOTYPE: genotype of this position.
  - 9. FREQUENCY: allele frequency
  - 10. Number\_of\_watson: the number of A,T,C,G covered in Watson strand.
  - 11. Number\_of\_crick: the number of A,T,C,G covered in Crick strand.
  - 12. Mean\_Quality\_of\_Watson: the mean base quality of A,T,C,G covered in Watson strand.
  - 13. Mean\_Quality\_of\_Crick: the mean base quality of A,T,C,G covered in Crick strand.

PS: Please refer to the format of VCF from 1st to 7th column.

## 6. Contact information

If you have any problem please do not hesitate to contact us!

gaoshengjie@genomics.org.cn