# 1 src/hotspot.R

## 1.1 依赖:

optparse, readxl, magrittr, stringr, tidyr, Biostrings, foreach包

## 1.2 用法:

首次可运行 Rscript hotsport.R -h自动安装依赖的包

显示如下结果表示依赖安装完成:

optparse readxl magrittr stringr tidyr Biostrings foreach   
 TRUE TRUE TRUE TRUE TRUE TRUE TRUE   
Usage: src/hotspot.R [options]  
Options:  
 -d DIR, --dir=DIR  
 mutation\_hotspot Excel file dir  
 -v VCF, --vcf=VCF  
 -o OUTDIR, --outdir=OUTDIR  
 -h, --help  
 Show this help message and exit

使用示例:  
Rscript src/hotspot.R \

-d ctDNA\_output\_filter/ \

-v test/Ct18100047\_1121\_BB18100007.snvIndel.raw.vcf \

-o ./

## 1.3 输入/输出文件

* 输入: **xxx.snvIndel.raw.vcf**
* 输出: **xxx.hotspot.xls**

## 1.4 输出文件格式

#chr pos\_start pos\_end ref alt Gene Type CDSChange exon sequence AA X\_\_1 chr pos ref.1 alt.1 d  
7 55249071 55249071 C T EGFR nonSynonymous\_Substitution NM\_005228.3 exon20 c.2369C>T p.T790M NA 7 55249071 C T DP=4;I16=1,0,1,0,69,4761,71,5041,50,2500,42,1764,12,144,25,625;QS=0.543478,0.456522;SGB=-0.379885;RPB=1;MQB=1;BQB=1;MQ0F=0

# 2 src/Somtic\_RemoveSite.R

## 2.1 依赖:

optparse, readxl, magrittr, stringr, tidyr, Biostrings包

## 2.2 用法:

首次可运行 Rscript Somtic\_RemoveSite.R -h自动安装依赖的包  
显示如下结果表示依赖安装完成:

optparse readxl magrittr stringr tidyr Biostrings   
 TRUE TRUE TRUE TRUE TRUE TRUE   
Usage: src/Somtic\_RemoveSite.R [options]  
Options:  
 -f FILE, --file=FILE  
 -s HOTSPOT, --hotspot=HOTSPOT  
 -d DIR, --dir=DIR  
 -o OUTDIR, --outdir=OUTDIR  
 -h, --help  
 Show this help message and exit

## 2.3 输入/输出文件

* 输入: **xxx.hotspot.xls** , **xxx.annotate.merge.filter.xls**
* 输出: **xxx.annotate.merge.filter.final.xls**

## 2.4 输出文件格式

#Sample Chromosome Start End Ref Alt cosmic\_id cosmic\_FATHMM\_prediction cosmic\_FATHMM\_prediction\_score Gene Region Classification CDS\_change Protein\_change Exon\_number Ensembl\_transcriptID Mutant\_frequency Effective\_depth Effective\_mutant\_depth Confidence HotSpot Oncokb\_Mutation\_Effect Oncokb\_Oncogenicity Oncokb\_PMIDs\_for\_Mutation\_Effect CKB\_Protein\_effect CKB\_Variant\_description 1000G\_all Esp6500\_all ExAC\_EAS CytoBand GenomicSuperDups CLINSIG CLNDBN CLNACC CLNDSDB CLNDSDBID SIFT\_prediction Polyphen2\_prediction MutationAssessor\_prediction  
Ct1809\_1113\_NCCL1817\_B 1 65312344 65312344 G T . . . JAK1 exonic nonsynonymous SNV c.C1975A p.R659S exon14 ENST00000342505 0.0029 699 2No No . . . . . . . . 1p31.3 . . . . . . T;0.260 P;0.616 L;0.288  
Ct1809\_1113\_NCCL1817\_B 1 156843598 156843598 C T . . . NTRK1 exonic nonsynonymous SNV c.C1024T p.R342W exon8 ENST00000524377 0.0024 12503No No 激活突变 可能致病 1695324, 11313867, 23636398, 22460905 . . . 7.7e-05 . 1q23.1 . . . . . . T;0.240 P;0.624 L;0.380  
Ct1809\_1113\_NCCL1817\_B 1 156845877 156845877 C T . . . NTRK1 exonic nonsynonymous SNV c.C1507T p.H503Y exon13 ENST00000524377 0.0019 10312No No . . . . . . . . 1q23.1 . . . . . . D;0.721 B;0.425 L;0.450  
Ct1809\_1113\_NCCL1817\_B 1 156849086 156849086 C G . . . NTRK1 exonic nonsynonymous SNV c.C1978G p.Q660E exon15 ENST00000524377 0.0017 11542No No . . . . . . . . 1q23.1 . . . . . . T;0.010 B;0.104 N;0.017  
Ct1809\_1113\_NCCL1817\_B 3 10183646 10183646 G A COSM14370;OCCURENCE=4(kidney) . . VHL exonic nonsynonymous SNV c.G115A p.G39S exon1 ENST00000256474 0.0014 1414 2 No No . . . . . . . . 3p25.3 . . . . . . T;0.287 B;0.112 N;0.181  
Ct1809\_1113\_NCCL1817\_B 3 10183766 10183766 C A . . . VHL exonic nonsynonymous SNV c.C235A p.R79S exon1 ENST00000256474 0.0014 1401 2 No No . . . . . . . . 3p25.3 . . . . . . D;0.555 P;0.651 L;0.453  
Ct1809\_1113\_NCCL1817\_B 3 10188287 10188287 G A COSM1035874;OCCURENCE=1(peritoneum),1(endometrium) . . VHL exonic nonsynonymous SNV c.G430A p.G144R exon2 ENST00000256474 0.0016 1229 2 No No . . . . . . . . 3p25.3 . . . . . . .;. D;0.916 M;0.649  
Ct1809\_1113\_NCCL1817\_B 4 1806143 1806143 C A . . . FGFR3 exonic nonsynonymous SNV c.C1162A p.L390M exon9 ENST00000440486 0.0027 752 2 No No . . . . . . . . 4p16.3 . . . . . . T;0.241 B;0.348 L;0.246

# 3 RemoveSite\_and\_db/

* Somtic\_File\_RemoveSite.xlsx 需要移除的位点
* Oncokb-annotate.xlsx Oncokb注释信息
* CKB-anV1.xlsx CKB注释信息