A manual for hot_scan

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What it is hot_scan?	

hot_scan is a free software to detect genomic regions unusually rich (hotspot) in a given pattern via scan statistics.

Requirements

hot_scan is designed to run on linux-based operating system. It is implemented in $Perl^1$ and R^2 languages. There are only few non-standard Perl module required to run hot_scan:

- 1. Math::GSL::SF
- 2. AnyEvent::ForkManager
- 3. File::Path

These modules can be installed via The Comprehensive Perl Archive Network (CPAN) at http://search.cpan.org. Make sure these modules are properly installed if there are no error messages.

Availability

The source code is available at https://github.com/aholanda/psa

¹http://www.perl.org

²www.r-project.org

Configuring hot_scan

After download the compressed folder **hot_scan.tar.gz** to an appropriate directory, extract it using following command: **tar-zxvf hot_scan.tar.gz**. Where,

- BY.R: a R script;
- events/: a directory to put the events file in BED format as decribed in following section;
- test/: a directory with some test data files;
- cs/: a directory with mappable chromosome size file for *Mus musculus* (mm9 and mm10) and *Homo sapiens*(hg19);
- hot_scan: the main program.

Running hot_scan

Before running hot_scan, you need to first provide a file in BED³ format within **events/** directory. Only the first three fields are required, where:

- 1. **chrom**: the name of the chromosome (e.g. chr1, chr2, chrM);
- 2. **chromStart**: the start position of the event in the chromosome;
- 3. **chromEnd**: the end position of the event in the chromosome.

Parameters

The main script is hot_scan and the essential parameters needed to run it are as follows:

- **-m**: window of width *m* to scan on chromosome;
- -c: file with the name and mappable chromosome size;
- **-e**: events file name;
- -o: output directory name;
- **-p**: max parallel forking count (default: 8);
- -s: significant level (default: 0.05);
- -a: adjust *p-values* using Benjamini & Yekutieli (2001) (default: no).

http://genome.ucsc.edu/FAQ/FAQformat.html

Notes:

(1) The chromosome size file should be tab delimited and structured as follows: <chromName><TAB><chromSize>

For example, Human (hg19):

chr1 225280621 chr2 238204518

•••

- (2) The chromosome size files for *Mus musculus* (mm9 and mm10) and *Homo sapiens* (hg19) are in **cs/** folder.
- (3) The events file should be a file in BED format as defined before.

Output files

The output files are put in the directory name specified by user (-o option). In the output folder, there will be two folders named as $scan_out_c\prime$ and $tracks_c\prime$. The log files are in the $scan_out_c\prime$. A BED file having the name of the corresponding events file is created in $tracks_c\prime$ folder with all hotspot information. Thus, for each hotspot, there will be one row having the chromosome, start, end and p-value with the corresponding hotspot id. The BED file in $tracks_c\prime$ folder can be upload into the Genome Browser on the Add Custom Tracks page⁴.

Example of running hot_scan on test data

Change the working directory to that which contains the hot_scan package:

\$ cd hot_scan

Copy events files from test directory to events directory:

\$ cp test/cMyc_AIDrv.bed events/

Execute hot_scan using the following command:

\$ perl hot_scan.pl -m 500 -c cs/mm9.txt -e cMyc_AIDrv.bed -p 15 -s 0.05 -o cMyc_AIDrv

The command line option for hot_scan above takes six parameters: i) a window width size (specified by -m); ii) chromosome size file (-c); name of the file with events (-e); iv) number of fork to launch (-p); v) significant level (-s) and vi) name of the directory (-o) in which are reported all logs and hotspots informations in BED format file as described in former section.

⁴http://genome.ucsc.edu/cgi-bin/hgCustom

⁵the file must be inside of the **events/** directory

How To Cite

SIlva IT, Rosales RA, Holanda AJ, Jankovic M and Nussenzweig MC. Identification of chromosomal translocation hotspots via *scan statistics*.

If you find a bug, please send an email to isilva@rockefeller.edu or rrosales@usp.br.

License and Copyright

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⁶http://www.gnu.org/copyleft/gpl.html