

Useful tool: annot-regs

Common task:

- find overlaps in two sets of genomic regions (e.g. two CNV callsets)
- .. while matching records by a common column (e.g. sample name)
- .. and transferring one or more fields from one file to another for matching records

Requirements:

- columns can be named differently and can appear in arbitrary order
- headers may or may not be present
- can require multiple columns to match (e.g. sample name AND variant type)
- can transfer multiple fields

Simple usage

Source file:

#chr	beg	end	smpl	type	qual	length	npr
5	67591246	67591246	DDDP167799	DEL	3.91	19611039	4
7	42006018	42006018	DDDP154344	DUP	64.02	19580982	29
12	14825806	14825807	DDDP123456	DEL	7.25	66506	2
16	30978217	30978217	DDDP345139	DEL	55.76	42248	3
19	42474652	42474652	DDDP114567	DUP	63.27	19581359	28

Destination file:

DDDP345139	16	30978217	30978217
DDDP123456	12	14825806	14825807
DDDP154344	7	42006018	42006018
DDDP114567	19	42474652	42474652
DDDP167799	5	67591246	67591246

annot-regs -s src.txt -d dst.txt -c chr,beg,end:2,3,4 -m smpl:1 -t qual:qual

file with source
annotations

destination
file

the names or
indexes of core
columns
(chr,beg,end) in
source and
destination file

the names or
indexes of
columns required
to be identical

the names or
indexes of
columns to
transfer

Note: this works also for files with a list of positions, not just regions, just use -c chr,pos,pos.

Usage page

About: Annotate regions in DST file with texts from overlapping regions in SRC file.

The transfer of annotations can be conditioned on matching values in one or more columns (-m), multiple columns can be transferred (-t).

In addition to column transfer and adding special annotations, the program can simply print (when neither -t nor -a is given) or drop (-x) matching lines.

All indexes and coordinates are 1-based and inclusive.

Usage: annot-regs [OPTIONS] DST

Options:

--allow-dups	Add annotations multiple times
-a, --annotate list	Add special annotations:
	cnt .. number of overlapping regions
	frac .. fraction of the destination region with an overlap
	nbp .. number of source base pairs in the overlap
-c, --core src:dst	Core columns [chr,beg,end:chr,beg,end]
-d, --dst-file file	Destination file
-H, --ignore-headers	Use numeric indexes, ignore the headers completely
-m, --match src:dst	Require match in these columns
--max-annots int	Adding at most int annotations per column to save time in big regions
-o, --overlap float	Minimum required overlap (non-reciprocal, unless -r is given)
-r, --reciprocal	Require reciprocal overlap
-s, --src-file file	Source file
-t, --transfer src:dst	Columns to transfer. If src column does not exist, interpret as the default value to use. If the dst column does not exist, a new column is created. If the dst column exists, its values are overwritten when overlap is found and left as is otherwise.
--version	Print version string and exit
-x, --drop-overlaps	Drop overlapping regions (precludes -t)

Usage examples

```
# Header is present, match and transfer by column name
annot-regs -s src.txt.gz -d dst.txt.gz -c chr,beg,end:chr,beg,end -m type,sample:type,smpl -t tp/fp:tp/fp

# Header is not present, match and transfer by column index (1-based)
annot-regs -s src.txt.gz -d dst.txt.gz -c 1,2,3:1,2,3 -m 4,5:4,5 -t 6:6

# If the dst part is not given, the program assumes that the src:dst columns are identical
annot-regs -s src.txt.gz -d dst.txt.gz -c chr,beg,end -m type,sample -t tp/fp

# One of source or destination files can be streamed to stdin
gunzip -c src.txt.gz | annot-regs -d dst.txt.gz -c chr,beg,end -m type,sample -t tp/fp
gunzip -c dst.txt.gz | annot-regs -s src.txt.gz -c chr,beg,end -m type,sample -t tp/fp

# Print matching regions as above but without modifying the records
gunzip -c src.txt.gz | annot-regs -d dst.txt.gz -c chr,beg,end -m type,sample
```