Useful tool: annot-regs

Common problem:

- find overlaps in two sets of genomic regions (e.g. two CNV callsets)
- match records by a common column (e.g. sample name)
- transfer a field 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

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Source file:

#chr	beg	end	smpl	type	qual	length	npı
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 <u>-s src.txt</u> <u>-d dst.txt</u> <u>-c chr,beg,end:2,3,4</u> <u>-m smpl:1</u> <u>-t qual:qual</u>



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

annot-regs usage

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).

All indexes and coordinates are 1-based and inclusive.

Usage: annot-regs [OPTIONS] DST

Options:

--allow-dups Add annotations multiple times -c. --core src:dst Core columns [chr,beg,end:chr,beg,end] -d. --dst-file file Destination file -m. --match src:dst Require match in these columns Minimum required overlap (non-reciprocal, unless -r is given) -o, --overlap float Require reciprocal overlap -r, --reciprocal Source file -s. --src-file 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. --version Print version string and exit

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
```