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

Useful tool: annot-regs

Source file: Destination file:

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

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

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 -a. --annotate list Add special annotations: frac .. fraction of the destination region with an overlap nbp .. number of source base pairs in the overlap Core columns [chr.beg.end:chr.beg.end] -c, --core src:dst -d. --dst-file file Destination file Use numeric indexes, ignore the headers completely -H. --ignore-headers -m, --match src:dst Require match in these columns Adding at most int annotations per column to save time in big regions --max-annots int -o, --overlap float Minimum required overlap (non-reciprocal, unless -r is given) 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 # 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