(correction after step 5, shown in green)

My goal is to get a file with five columns organized as follows:

Chr Start End N. overlaps Patient IDs whose CNV regions overlaps of the overlapping regions

And then sorted this table according to the n. of overlaps to get a kind of ranking (prioritization according to the number of overlaps).

- 1 Load the file with the original dataset
- 2 Click on "Sort data in ascending or descending order" and sort the original dataset according to the chromosomes (necessary for step 3).
- 3 Click on "Create a BedGraph of genome coverage".

In the output file you can see: chr (column1), start (c2), end (c3), number of overlaps (c4) but <u>not the patient IDs</u>.

chr1	833831	4061509	1
chr1	4795388	5967499	1
chr1	5967499	6023558	2
chr1	6023558	17364849	1
chr1	23689659	25570112	1
chr1	25616336	25657021	1

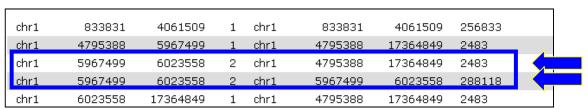
Example with five regions

4 – Select "Join the intervals of two datasets side-by-side" and join dataset '3' and '1'.

In the output file you can see:

- chr (c1), start (c2), end (c3) of the **overlapping regions**
- number of overlaps in that regions (c4)
- chr (c5), start (c6), end (c7) of the **original regions** (I mean the regions with CNV of the patients)
- patient ID (c8)

Note: the regions that overlap are split in two rows(see blue arrows)... need to group the data!



Example with five regions

5 – Group on dataset '4' by column c4 (number of overlaps) and concatenate on column c8 (patient IDs). WRONG!

If I do this, Galaxy pools together all the IDs and the regions... (see the output file down below).

In this case: column1 contains the number of overlaps; column 2 contains all the IDs of the patients whose CNV regions overlap 1 time (or 2 times for the second row, etc...).

- 1 289515,2541(3/48c),248354,248354,268350(1/2),268350(2/2),276232,276232,250369(1/3),2541(4/48c),
- 2 248354,288279(1/2),248354,268350(1/2),268350(2/2),276232,276232,267222,276232,261505(1/2),2762
-) 076000 056500 067000 076000 067000 061505(1/0) 076000 061505(1/0) 000011 076000 061505(1/0) 0

POSSIBLE SOLUTION (that works!)

- 5 Group on dataset '4' by column c2 (start of the overlapping regions) with two operations: concatenate distinct on c4 (num of overlaps) and concatenate on c8 (patient IDs).
- 6 Join datasets '3' (BEDGraph), using c2, and '5', using c1.

 In the output file there are two columns repeated (those columns I used before for grouping).

chr1	833831	4061509	1	833831	1	256833
chr1	4795388	5967499	1	4795388	1	2483
chr1	5967499	6023558	2	5967499	2	2483,288118
chr1	6023558	17364849	1	6023558	1	2483

7 – Click on "Text Manipulation" and then on "Cut columns from a table", to eliminate columns c5 and c6. Cut columns: - c1 (chr)

- 01 (0111)
 - c2 (start)
 - c3 (end)
 - c4 (num of overlaps)
 - c7 (patient IDs whose CNV regions overlap)

This is the correct output file:

chr	start	end n. o	verla	ps IDs
chr1	833831	4061509	1	256833
chr1	4795388	5967499	1	2483
chr1	5967499	6023558	2	2483,288118
chr1	6023558	17364849	1	2483

But the output file entries are still sorted according to the chromosomes and not to the number of overlaps... so I need to sort the dataset again!

8 – Sort according to the num. of overlaps to get a ranking (a kind of prioritization), using dataset '7' on column c4 (num. o f overlaps).

The resulting output file it's exactly what I want (chr / start / end / n. of overlaps / IDs):

chr1	5967499	6023558	2	2483,288118
chr1	4795388	5967499	1	2483
chr1	833831	4061509	1	256833

