# The Remap Service from NCBI

Mapping genomic coordinates from one genome assembly to another for selective organisms https://www.ncbi.nlm.nih.gov/genome/tools/remap/ National Center for Biotechnology Information • National Library of Medicine • National Institutes of Health • Department of Health and Human Services

### Scope

Updates of genome assemblies or the availability of multiple assemblies for a single organism present an annotation comparison challenge. Performing a *de novo* annotation, especially for some datasets, can be computationally and time intensive. One way to quickly address this issue is to align the assemblies to each other and then 'map' the features from one coordinate system to the other using these alignments as a guide. While not a replacement for *de novo* annotation, this 'remap' process can be useful when looking at individual regions. When performed on annotations across the genome, this process can provide useful information on the degree of difference between the two assemblies. The Remap Service is a tool from NCR



information on the degree of difference between the two assemblies. The Remap Service is a tool from NCBI that makes remapping features/annotations simple and straightforward.

### Access to the service

The Remap Service is accessible online through the genome tools collection at <u>https://www.ncbi.nlm.nih.gov/genome/</u> <u>tools/remap</u>. The Assembly-Assembly Remap Service allows for remapping of features from one assembly to another. Clinical Remap allows for the remapping of features (including HGVS-defined sequence variations) to RefSeqGene sequences, including the underlying transcript and protein sequences. It also maps from the RefSeqGene sequences to an assembly.

## The input form

The Remap Service takes input from a web form (as shown). The type of remap to be performed can be selected using tabs at the top (**A**). For Assembly-Assembly remap, organism and source/ target genomes can be selected using the lists in the "Genome Information" section (**B**). Mapping criteria can be adjusted using parameters in the "Remapping Options" (**C**). Increasing the "maximum ratio of bases" and decreasing the "maximum ratio of difference" improve the stringency of the mapping process. Popups linked from question mark icons (**D**) provide context-specific help.

The actual list of coordinates to be remapped can be uploaded as a text file using the "Choose File" button (E) or pasted directly in the textbox (F). In the example, a set of features from GRCh37 in GFF3 format is pasted in the input box, see page 2 for details. Multiple formats for input/output coordinates are supported with specific formats selectable using the pull-down list (G). A

BI Genome Remapping licates required fields.	3 Service				
ssembly-Assembly Clinical	Remap Alt loci remap 🖌 🗛				
Genome Information—					
Source Organism *					
Homo sapiens					
Start typing to get a list of availa	ble organisms				
Source Assembly *					
Assm Name		Assm SeqID	Date		
GRCh38.p2		GCF_000001405.28			
GRCh38.p1	GCF_000001405.27				
GRCh38 (hg38)	GCF_000001405.26				
GRCh37.p13	GCF_000001405.25	2013/06/28			
GRCh37.p12	GCF_000001405.24	2013/03/26			
Target Assembly *					
Assm Name			Assm SeqID	<ul> <li>Date</li> </ul>	
GRCh38.p8			GCF_000001405.34	2016/06/30	
GRCh38.p7			GCF_000001405.33	2016/03/21	
GRCh38.p6			GCF 000001405.32	2 2015/12/21	
GRCh38.p2		GCF 000001405.28	-		
GRCh38.p1			GCF 000001405.27	-	
	ember 27, 2016, software version: 1.7			-	
	Source Assembly Coverage	Target As:	sembly Coverage	<b>D</b>	
Alignments	(GRCh37.p13, GCF_000001405.25)	(GRCh38.p8,	GCF_000001405.34)		
First Pass(Reciprocal Best Hits)	97.273		94.705	99.997	
Total	99.817		97.587	99.980	
Remapping Options		of the t	the remapping, wha feature that must be afault is 50%.	it is the minimum am able to be remapped	
Maximum ratio for difference b	etween source length and target length: 2		ſ	GFF3 V	
Allow multiple locations to be r	eturned: 🗹 🔞		-	Same as input	
Merge Fragments: 🗹 🔞				HGVS	
				BED GVF	
ata				GFF	
nput format: Best Guess	Output format: GFF3		v results in Ge	GTF	
nput format: Best Guess	Output format: GFF3	<b>(</b>	y results in Ge	GFF3	
Upload a file: Choose File	Vo file chosen		_	Text ASN.1 Binary ASN.1	
				UCSC Region	
R				VCF	
aste data here: #GFF	3 for remap from GRCh37 to GRCh38				
Paste data here: #GFF #COL	S: sid,src(SO:id),var,start,stop,s			. TD=rs14899684	
Paste data here: #GFF #COL Chr1 You can paste chr1	S: sid,src(SO:id),var,start,stop,s . snp 154437613 . snp 169519894	154437613 169519894	· + · +	. ID= <u>rs14899684</u> . ID= <u>rs6015</u>	
You can paste multiple lines into	S: sid,src(S0:id),var,start,stop,s . snp 154437613 . snp 169519894 . snp 10942437	154437613 169519894 10942437	· +	. ID= <u>rs6015</u> . ID= <u>rs15512786</u>	
Vaste data here: #GFF #COL Chr1 70u can paste multiple lines into the text area. Chr6	<pre>S: sid,src(SO:id),var,start,stop,s</pre>	154437613 169519894 10942437 10940942 10941008	· + · + · + · +	<ul> <li>ID=rs6015</li> <li>ID=rs15512786</li> <li>ID=rs15074664</li> <li>ID=rs14993936</li> </ul>	
Vaste data here: #GFF #COL chr1 multiple lines into the faxt area chr6 chr6	<pre>S: sid,src(SO:id),var,start,stop,s</pre>	154437613 169519894 10942437 10940942	. + . + . + . +	. ID= <u>rs6015</u> . ID= <u>rs15512786</u> . ID= <u>rs15074664</u>	
Vaste data here: #GFF #COL Chr1 70u can paste multiple lines into the text area. Chr6	<pre>S: sid,src(SO:id),var,start,stop,s</pre>	154437613 169519894 10942437 10940942 10941008	· + · + · + · +	<ul> <li>ID=rs6015</li> <li>ID=rs15512786</li> <li>ID=rs15074664</li> <li>ID=rs14993936</li> </ul>	

message box at the bottom (not shown) identifies fields needing input data. It disappears when all requirements have been satisfied. The "Submit" button (H) will then become available.

#### Example input format

The Remap Service supports several input formats. The example shown below is a list of SNPs from GRCh37p13 (hg19) in GFF3 format, which can be used as test input to the Remap Service to map them forward to GRCh38p2. For testing purposes, source and target assemblies can be switched.

148996840
\$6015
1551278630
150746645
149939366
\$6781

## Displaying and downloading remap results

A completed remap request is displayed in a summary format with four main sections: Summary Data, Mapping Report, Annotation Data, and Genome Workbench Files.

The Summary section (A) provides a synopsis of the remapped features grouped by chromosome. Complete summary is downloadable using the disk icon. Up to ten remapped entries are shown in the Mapping Report section (B), with the complete set in the downloadable file marked by the disk icon (**C**). Clicking the icon activates a download window (D) from where the result can be saved or opened in Excel (under Firefox, other browsers may behave differently).

#### All features are in this report

whether they successfully remapped provides interval-by-interval compar and the target assemblies. Lastly, the dividing the length of the feature in the feature in the source assembly. A value of "1.00000" is perfect, while a value of less than 1 indicates a deletion in the target assembly and a value of greater than 1 indicates an insertion in the target assembly. This sort of metric does not assess single base changes within the region.

	ap Resul apping in		from GRCh37.	o13 (G	CF_0000014	05.25	i) to GR	Ch38.p8 (G	CF_000001405.34)
Summary	/ Data—		A						7
Download S	iummary Dat	ta 💾							
ID	Source Features		Remapped Features		Source Intervals		Remapped Intervals		
chr1	2		2		2		2		Links in page —
chr6	3		3			3			Summary Data
chr9	1		1		1		1		Annotation Data
	Report (s		B	<mark></mark>					Genome Workbench Fil
Feature	Src Intervals	Remap Intervals	Src Location	Src Length	Map Location		Map Length	Coverage	What is NCBI Remap?
rs6781	1	1	chr9:130698043	1	chr9:1279	85764	1	1.00000	About our alignment
rs15512	1	1	chr6:10942437	1	chr6:1094	2204	1	1.00000	
rs15074	1	1	chr6:10940942	1	chr6:1094	0709	1	1.00000	API Documentation
rs14993	1	1	chr6:10941008	1	chr6:1094	0775	1	1.00000	About Genome Workbe
rs14899	1	1	chr1:154437613	1	chr1:15	37	1	1.00000	Support Center
rs6015	1	1	chr1:169519894	1	chr1:16	56	1	1.00000	
Annotatio	on Data –	ata 🗎	E		Opening re	_	_		ea.xls
	Workben Genome Work	ch Files-	<b>F</b>		which	rt_ro	emap_i LS file	n: <b>textarea.</b> : .ncbi.nlm.ni	
arison o the cov	of the fe erage s	atures of score is	this report on the sourc calculated b oy the length	y	• <u>O</u> p	ıld Fir en wi ve File	th Mi	with this file	

The link in the Annotation Data section (E) provides the remap data for the target assembly only. The link in the Genome Workbench Files section (F) provides the data in a format that can be readily imported into the standalone Genome Workbench tool (https://www.ncbi.nlm.nih.gov/projects/gbench/) from NCBI. Online links to help documents are also available from the result page (G) to provide more detailed explanation on the file format. Additional requests for tech-

## nical assistance and feature additions to this tool can be emailed to NCBI using the "Write to the Help Desk" link (H).

### API access

The Remap Service also provides API access. An example Perl script along with example command line are also provided. Detailed explanation is available in the online document:

https://www.ncbi.nlm.nih.gov/genome/tools/remap/docs/api?page=result

Do this automatically for files like this from now on.

OK

Cancel