

Pile|Line

Example #4.

Annotate Genomic Position files with Ensembl genes.

PileLine

Step 1.

Download and uncompress the Genomic Position example files:

Sample Data

Here you can find some files to test PileLine and PileLine GUI. All files are compressed. You need to uncompress them before use.

File	Type	Description
hg18.zip  (929MB)	Genome (.fa and .fai)	Human Genome 18 in FASTA (.fa) and indexed FASTA (.fai). The sequence names are without the 'chr' prefix.
Control1Files.zip (36MB) Case1Files.zip (36MB) Control2Files.zip (38MB) Case2Files.zip (38MB)	Genotype data (.pileup)	Several genotyped samples. each .zip file contains 2 files: a complete and a variants-only pileup. Note: there is only data for the chromosome 10
Hg18_hgnc_ensembl_genes.bed.zip (365KB)	Gene annotation (.bed file)	Ensembl Genes for the Human Genome 18
DbSNP_36.3.bed.bgz.zip (150MB)	SNP annotation (custom in compressed (bgz) BED format)	SNPs from dbSNP version 36.3 for the Human Genome 18

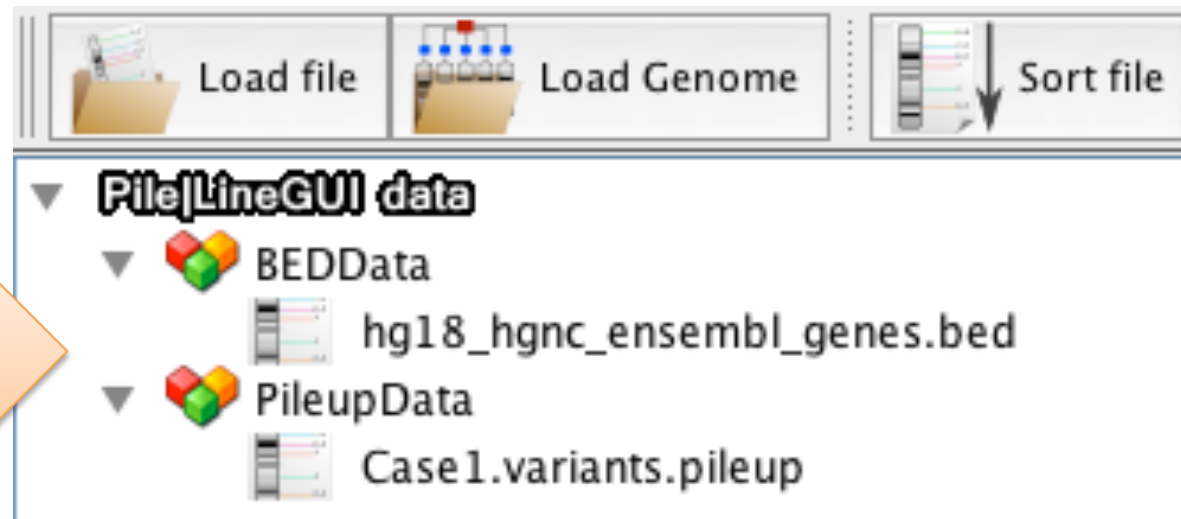
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Step 2.

Load uncompressed Genomic Position files:

Hg18_hgnc_ensembl_genes.bed
Case1.variants.pileup

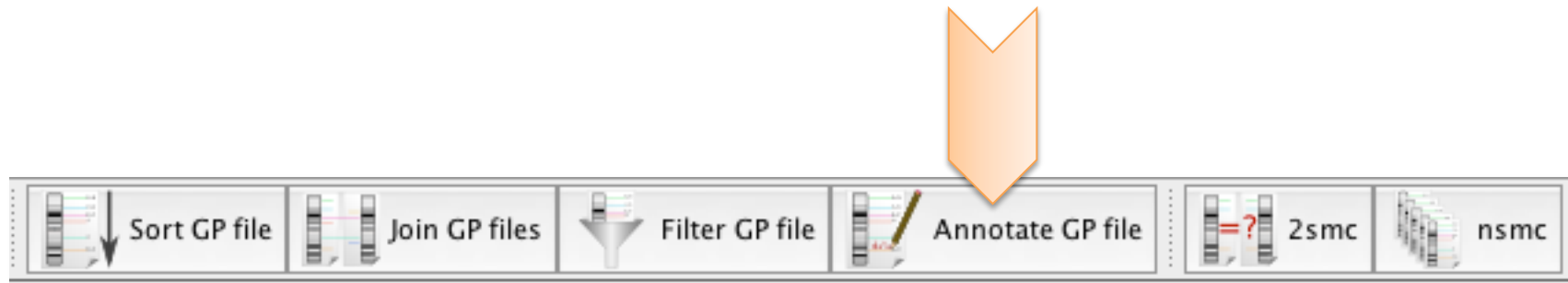
*Find the
loaded
files in the
Data
Repository*



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Step 3.

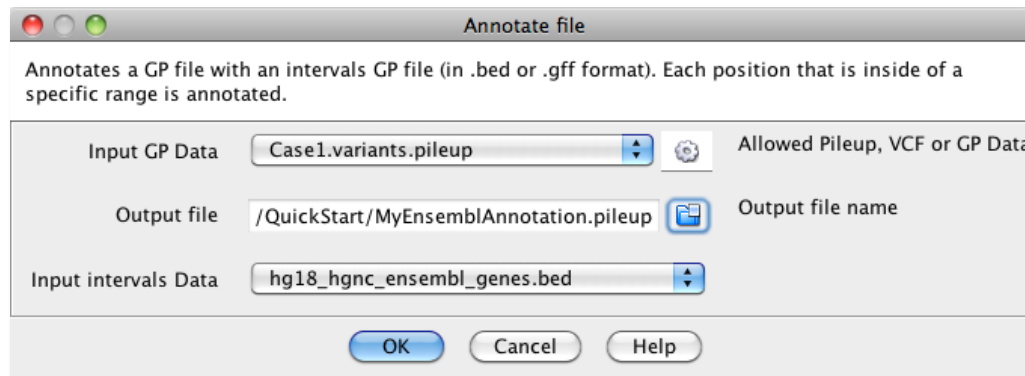
- a) Click on 'Annotate GP file' button in the upper toolbar.



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Step 4.

a) Choose your annotation files.



Allowed Pileup, VCF or GP Data

Output file name

File to annotate

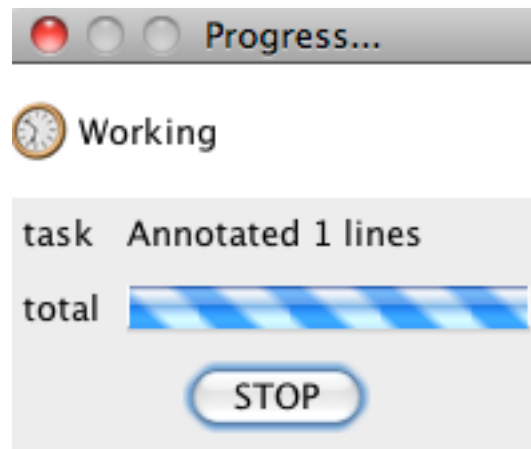
Output name

Ensembl file

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Step 5.

Now take it easy, your file is being annotated in PileLineGUI...



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Step 6.

Check results output in the Data Repository!!

10	121337	A	G	123	123	36	32	GggGggg...]a`Ub`[aa...	ENSG00000217176 IL9RP2
10	121636	T	C	114	114	36	29	ccCcccCCc... bb]aaa[aX...	ENSG00000217176 IL9RP2
10	121716	C	Y	84	132	31	29	tt,,ttttt,t,t,... aaZ]W[ba...	ENSG00000217176 IL9RP2
10	123402	G	S	49	49	30	10	..CCC.CCC. aa]R_aab_]...	NULL
10	123416	A	R	34	34	33	16	.GGG.GG... ^[a^[ab_]...	NULL
10	6034700	A	M	228	228	35	56	.\$C,,c,cc...]^aaa`ba...	ENSG00000134470 IL15RA
10	6035058	C	Y	228	228	37	47	,t\$t,t,t,t,t... b^aa^)`a...	ENSG00000134470 IL15RA
10	6042267	G	R	104	104	37	27	A\$...A..AA...]`b_a`b`...	ENSG00000134470 IL15RA
10	6101787	T	C	90	90	36	21	C\$C\$CCC... Z[YZ^[__...	ENSG00000134460 IL2RA
10	6103325	c	Y	95	95	37	12	..T.T.TTT.T.][^V`_[a...	ENSG00000134460 IL2RA
10	6103389	T	C	78	78	36	17	CCCCCCC... B]]^a^Ua`...	ENSG00000134460 IL2RA
10	6103680	T	C	105	105	37	26	c\$c\$ccccc... aaaabaaa...	ENSG00000134460 IL2RA
10	6106206	C	S	228	228	37	57	g\$G...,g..G... a^^ab[`^...	ENSG00000134460 IL2RA

Your input file

*Ensembl
annotation*



For further info see PileLineGUI Documentation at:

<http://sing.ei.uvigo.es/pileline/pilelineguihelp/>