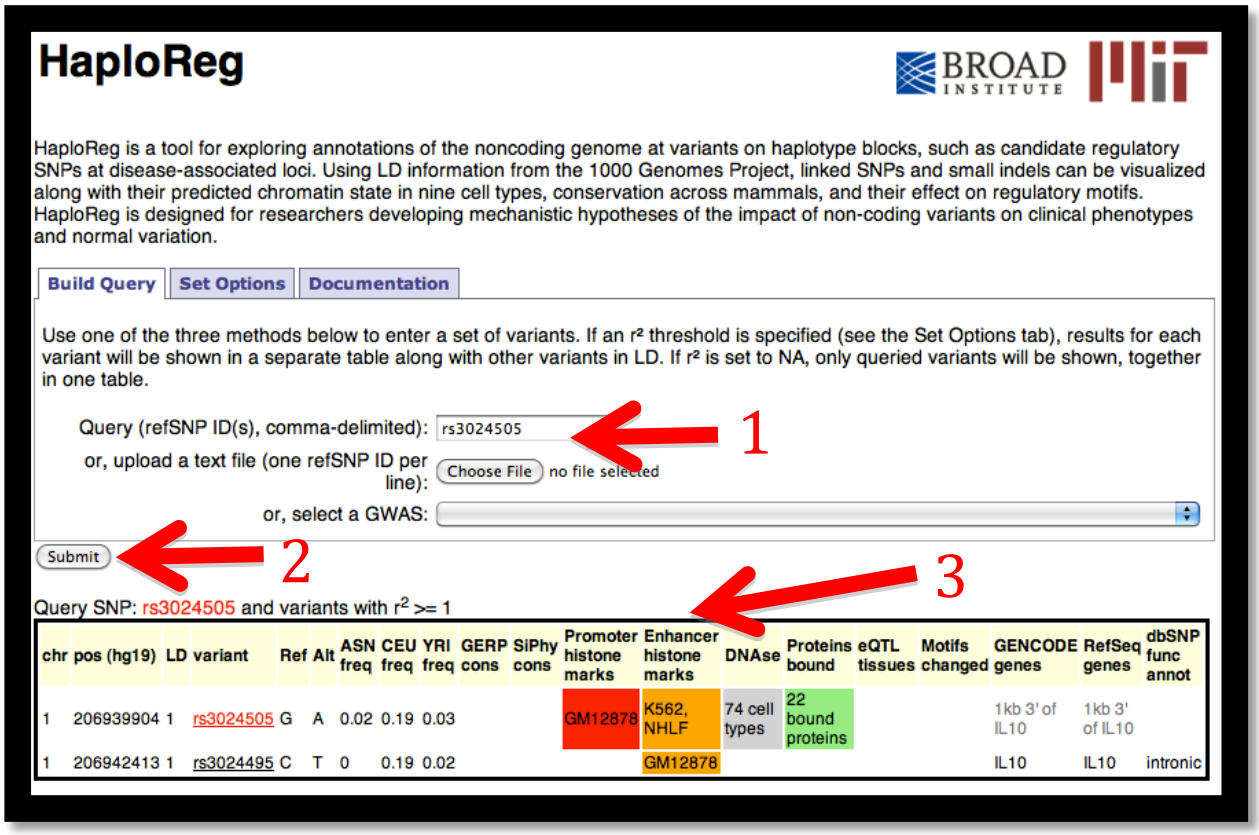
Using HaploReg and RegulomeDB to mine ENCODE data:

(Updated 16 November 2012, Mike Pazin)

HaploReg and RegulomeDB are ENCODE-funded tools described in recent publications that retrieve ENCODE annotations at SNPs of interest. Use of HaploReg is briefly described on this page, and RegulomeDB is described on the following page.

HaploReg:

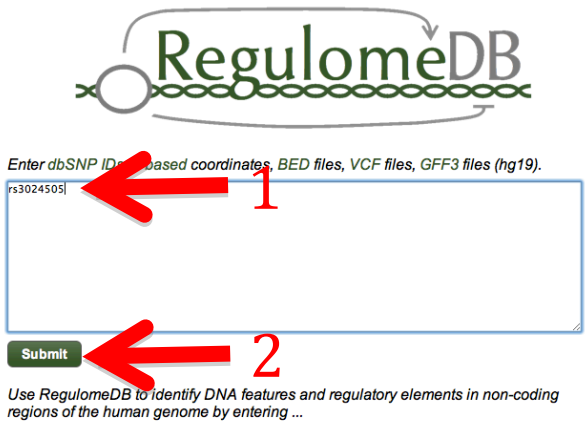
Go to the HaploReg site, and enter the name of the SNP of interest (Arrow 1).



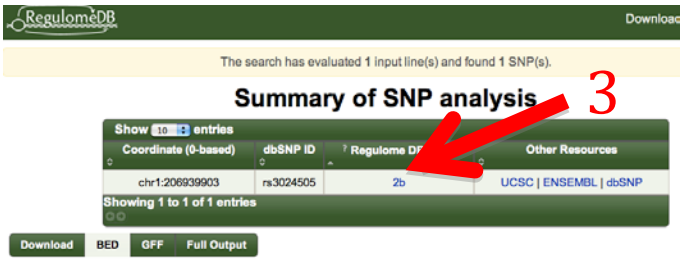
Click on the submit button (Arrow 2).
HaploReg retrieves the ENCODE annotation for the selected SNP, as well as other SNPs in LD (arrow 3).
Using the "Set Options" tab, the user can configure values such as the LD threshold and the population used from 1000 Genomes data used to calculate LD.

RegulomeDB:

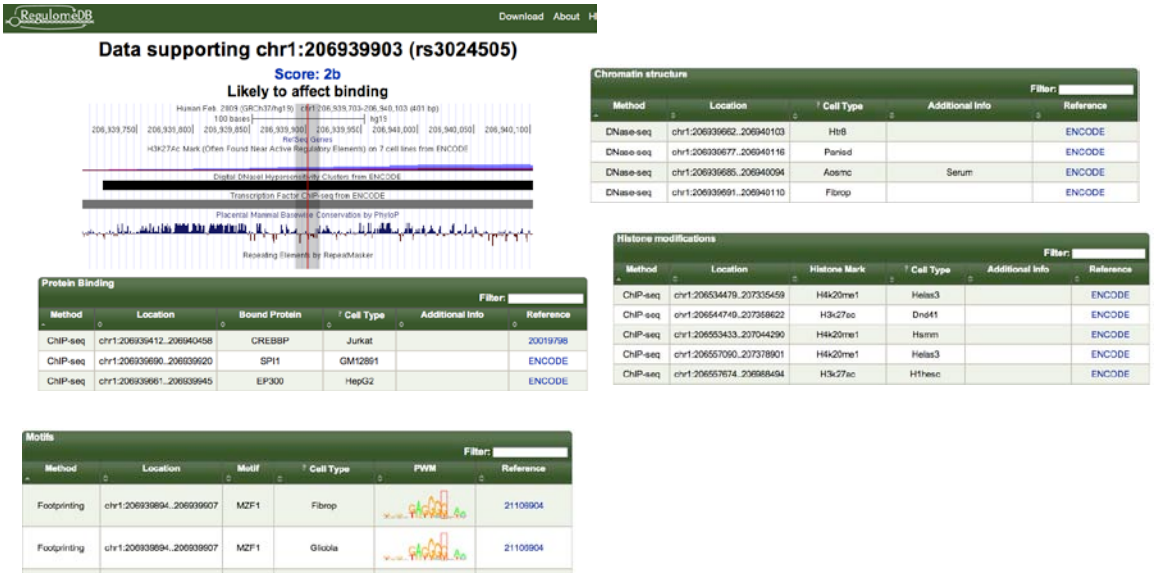
Go to the RegulomeDB site and enter the name of the SNP of interest (Arrow 1). Click on the submit button (Arrow 2).



RegulomeDB calculates a score for the regulatory potential of this region.



Clicking on the score retrieves the ENCODE annotation for the region (arrow 3).



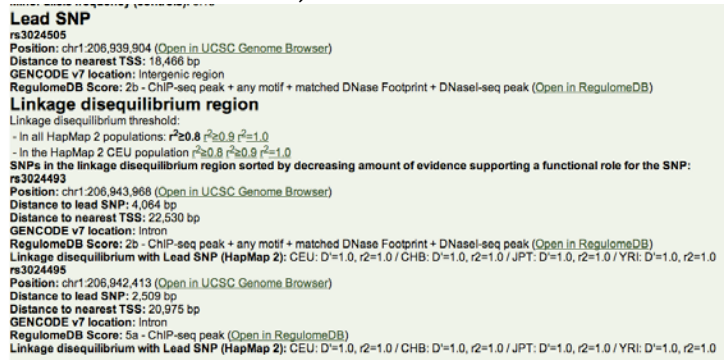
RegulomeDB also has a database of predicted functional SNPs, by disease/trait and by SNP, available at: http://regulome.stanford.edu/GWAS

There is a list of over 4700 SNPs associated with human traits and disease (arrow 1), as well as a list of over 470 human traits and diseases (arrow 2).

Clicking on a trait/disease returns a list of SNPs that have been associated with that trait or disease:

Clicking on a SNP (red arrow) returns the evidence for the association:

As well as the annotation for the lead SNP, and other SNPs in LD that, based on functional annotation, are candidates for the functional variant:



One can follow the links to view the genomic annotation of these SNPs in the genome browser.