

Exercises for the SeattleSNPs Resource (Version 1, 2006)

You can choose to do these using the questions as your only guide—or see the following pages for the step-by-step checklist to finding these answers.

1) Use SeattleSNPs to locate a gene and examine the details of SNPs that have been genotyped for this gene using the gene structure graphical representation.

Skills: Accessing genes; exploring data; changing views

2) Explore the genotyping data details for a gene, in graphical and text forms.

Skills: accessing gene data; examining graphical representations

3) Use the Visual Haplotype software to examine the theoretical haplotypes along the ERMAP gene. Cluster by “Site and Sample” to see if blocks that are probably inherited together become more apparent.


Skills: Visual Haplotype interface and display

SeattleSNPs Exercises, version 1.0. Correspond to the data available in October 2006

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Step-by-Step checklist/instructions for the SeattleSNPs introduction exercises

1. Use SeattleSNPs to locate a gene and examine the details of SNPs that have been genotyped for this gene using the gene structure graphical representation.

Step	Action	✓
1	Go to the SeattleSNPs homepage: http://pga.gs.washington.edu/	
2	From the navigation links on the upper left side of the page, find the Sequencing Resources area. Click the link for Sequenced Genes.	
3	Scroll down the page to find the ERMAP gene, Erythroblast membrane-associated protein. Examine the data on this row.	
4	Which genomic DNA panel was used in the analysis of this gene, p1 or p2? _____ Which populations were examined in this analysis? (<i>Click the panel link to discover this.</i>) _____ and _____	
5	Click the back button on your browser to return to the gene list page.	
6	Go to the gene page by clicking the HUGO gene symbol ERMAP.	
7	Examine the gene structure diagram. How many exons are indicated for this gene? _____ Does this gene display untranslated regions (UTR)? _____	
8	Now, let's focus on an interesting exon. Exon 2 appears to have several SNPs, but it is hard to distinguish them in the Full gene view. We will move exon 2 to the center and then zoom in. <i>Note: Exons are labeled with numbers, but the appearance varies slightly on different browsers. Exon 2 should have a number 2 above the structure. May require a close look.</i> <ul style="list-style-type: none"> • Click the left-pointing Scroll Direction arrow  in the Image Controls area to move that exon to the near the center arrow ↓. It will take about 5 clicks with the default setting of moving by 1000 bp. • Use the <u>Image Controls View menu</u> to change the image to show 20% of Gene. <p>You should have a better view of the exon and the SNPs.</p>	
9	How many SNPs are visible in exon 2? _____ Do these represent different classes of SNPs? _____ Which classes? _____, _____ and _____	

10	Some of the Exon 2 SNPs are lower frequencies, as judged by the length of the SNP lines. Change the Frequency menu to “Low Freq” to see them better. Note the scale on the right changes so you can more clearly determine approximate frequencies.	
11	Change the SNP Views menu to say: coding. Only those SNPs in coding regions should be left on the graphic.	
12	To learn more about those SNPs, move down to the data table area and click the link for cSNPs in the Mapping Data section. You should be able to see the details of these SNPs in text format; they are listed as NON-SYN and SYN in this table. Scroll over to the right to see the observed amino acid changes.	

2) Explore the genotyping data details for a gene, in graphical and text forms.

Step	Action	✓
1	From the ERMAP page in exercise 1, scroll down to the data table area, below the “gene specific links” area.	
2	In the Genotyping Data row, click either the blue/yellow/red box graphic or the link for Genotyping Data in the first column. You should find the documentation that describes the data in the area. Don't forget this documentation is always at your fingertips! Use the back button to return to the ERMAP page.	
3	On the ERMAP page, in the Genotyping Data section click the link for SNP Alleles. A new web page will open with text about each SNP. The nucleotide site is indicated, followed by information about Allele 1 and its occurrence in the African Descent (AD) population and in the European Descent (ED) population.	
4	Examine the data in the first row, for site 000463. Allele 1 is a G, found in 0 African and 5 European chromosomes. Allele 2 is A, and found in 46 African and 41 European chromosomes. <i>Keep this in mind as we move to the next information.</i>	
5	Use the back button to return to the ERMAP page. In the Genotyping Data section, click the link that says Visual Genotypes. A new web page will open with the “prettybase” graphical view of the genotyping data. <i>On some computers you will need to hover over the image and click with the magnifying glass to make it full size.</i>	
6	Note the individuals listed on the y-axis; DY are African Yoruba samples, E are European samples. Note the nucleotide sites from 5' to 3' across the top. Note the color key at the bottom of the page.	
7	Remember our information about site 000463 from the text page? There were 5 European chromosomes present at that site. Look through the column for the site 463. Can you find 5 individuals who have a variation at this site? _____, _____, _____, _____, and _____ Are they homozygous or heterozygous for this allele? _____	
8	Look at the 696 site column. Are there individuals who are homozygous for the rare allele at this site? _____, _____ Which population are these individuals from? _____	
9	Click the back button to return to the ERMAP page.	

3. Use the Visual Haplotype software to examine the theoretical haplotypes along the ERMAP gene. Cluster by “Site and Sample” to see if blocks that are probably inherited together become more apparent.

Step	Action	✓
1	From the ERMAP page, move to the Haplotyping Data area.	
2	In the Haplotyping Data area, click the link for Visual Haplotype.	
3	A new web page will open with the interface for the Visual Haplotype software. Before moving on, examine the page. You will see information about the color codes, and helpful details about how to format and upload your own data later.	
4	To examine the theoretical haplotype data for ERMAP, find the pulldown menu called: PGA Finished Gene Phasebase Input File: Use the menu to select the ERMAP gene.	
5	Leaving all other settings as default, click the button that says “Run VH1 on the Web!”	
6	A new window and web page will open with the haplotype data. As before in Visual Genotype, note the individuals listed on the y-axis; DY are African Yoruba samples, E are European samples. <i>However, the data here will show 2 entries for each person, one for each copy of the gene on each autosome.</i> Note the nucleotide sites from 5’ to 3’ across the top. Note the color key at the bottom of the page. How many Europeans carry the rare allele at site 463? _____	
7	Although some patterns can be detected from the data with this default view, let’s go back to the VH1 software and make a change so that the “blocks” or segments that may be inherited together become more apparent. Close the haplotype diagram window page.	
8	Back on the Visual Haplotypes interface, change the Cluster by: menu to say “SITE AND SAMPLE”.	
9	Leaving all other settings as default, click the button that says “Run VH1 on the Web!”	
10	Your new Visual Haplotypes diagram should have grouped the data differently, and blocks or segments along this gene that may be more likely to be inherited together over time are more apparent. Selecting “tag” SNPs in these regions may be more effective for determining haplotypes in other samples.	

Answers to questions within exercises

(as of October 2006, results may change over time)

- 1.4** panel = p2. This represents DNA from the HapMap project, including Yoruba (YRI) from Nigeria and European ancestry Utah residents (CEU).
- 1.7** 11 exons. Yes, this gene has green UTRs on both 5' and 3' locations.
- 1.9** 4 SNPs are visible in exon 2. Yes, the SNPs are UTR, non-synonymous, and synonymous classes.
- 2.7** Individuals with variation at site 463 are: E103, E109, E113, E121, and E123. These individuals are heterozygous at this allele.
- 2.8** Individuals homozygous for the rare allele at 696 are DY10, DY22. They are from the Yoruba population.
- 3.6** Five Europeans carry the rare allele at site 463.