

# Human genetics in the 21st century: Using bioinformatics to link genotype and phenotype

High School Student Program 2015

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## Introduction and preliminaries

- 1 - Create a name for your *alter ego*.
- 2 – Describe the ethnicity of your *alter ego*.
- 3 – Find the reference population below which most closely matches the ethnicity of your *alter ego*.

African ancestry in Southwest USA	ASW
Utah residents with Northern and Western European ancestry from the CEPH collection	CEU
Han Chinese in Beijing, China	CHB
Chinese in Metropolitan Denver, Colorado	CHD
Gujarati Indians in Houston, Texas	GIH
Japanese in Tokyo, Japan	JPT
Luhya in Webuye, Kenya	LWK
Maasai in Kinyawa, Kenya	MKK
Mexican ancestry in Los Angeles, California	MEX
Tuscan in Italy	TSI
Yoruban in Ibadan, Nigeria	YRI

Each SNP is identified with an “rs” ID. You will use online resources to find information about the SNP and determine the trait (i.e., phenotype) that is associated with each of your *alter ego*'s genotypes.

### Exercise: Demo2

Open two web pages:

- Go to the HapMap project ([hapmap.ncbi.nlm.nih.gov/](http://hapmap.ncbi.nlm.nih.gov/)) and, under “Project Data”, click on the first genome browser.
- Go to SNPedia (<http://www.snpedia.com>)

For a row in the table, go to the HapMap browser

1. Under “Landmark or Region”, type the SNP rs ID.
2. Note the chromosome on which this SNP is located.
3. Look for the “Entrez Genes” track on the browser and note any gene that overlaps the SNP.
4. Click on the graph under “Genotyped SNPs”.
5. Look for your reference population and your genotype and note the frequency of that genotype.

Go to SNPedia

6. Search with your SNP rs ID.
7. On the right side of the SNP page, find your genotype and write down at least part of the “Summary” describing the phenotype (trait(s)) associated with your genotype. The genotype listed on SNPedia may have a different orientation (eg. plus or minus).

**Websites/Resources:**

SNPedia: <http://www.snpedia.com>

HapMap Project: <http://hapmap.ncbi.nlm.nih.gov>

1000 Genomes Browser: <http://browser.1000genomes.org>