

?

- X1: GATGCAT...
- X2: GCTGCAT...
- X3: GATAAAT...
- X4: CATGCAT...

There's a SNP for that

An introduction to personal genomics

- Sample direct-to-consumer genetics reports
- There are even direct-to-pets genetics reports:
<http://www.wisdompanel.com/>

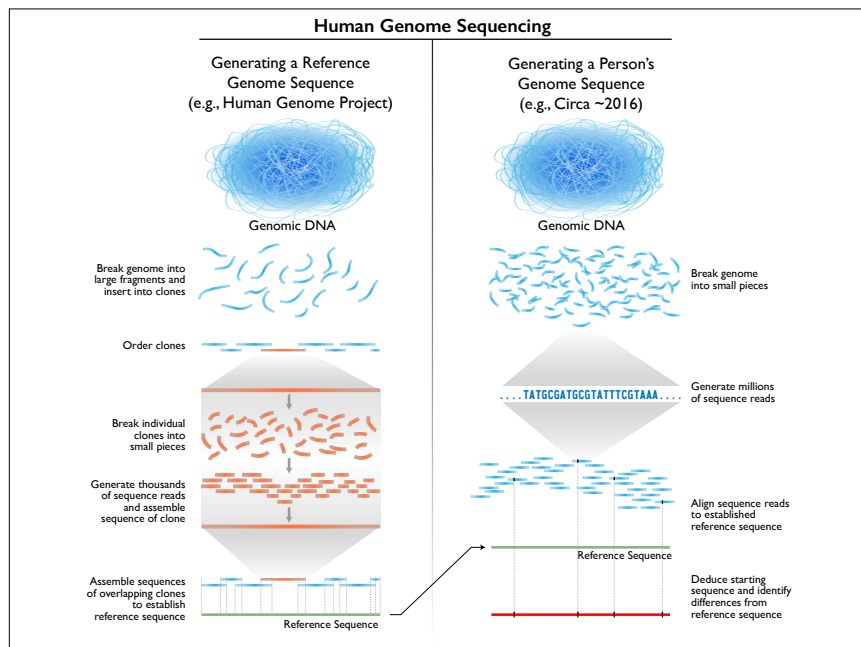
SNPedia.org

How did we get here?

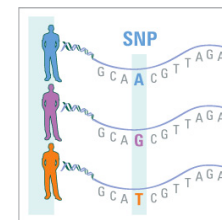
A million base pairs isn't cool, you know what's cool?

A billion base pairs

HIV genome	Human genome
10	3 x 10
10 genes	10,000s of genes
typical difference of 1-2% in gag, pro, pol from founder strain in single infection	typical difference of ~0.2% from ref genome
~10	~10



SNP: single nucleotide polymorphism

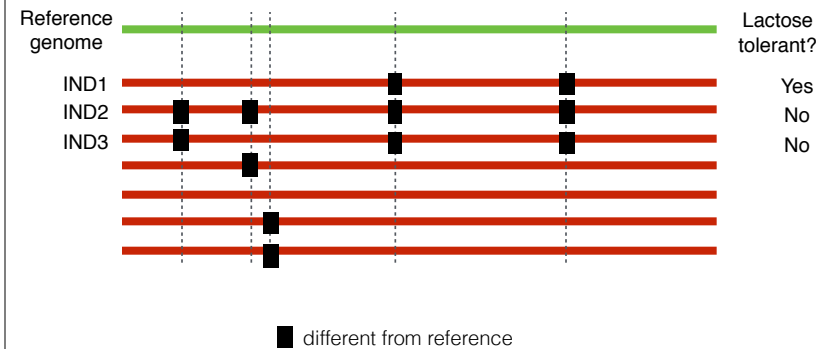


...GCA^ACGTTAGA...

...GCA^GCGTTAGA...

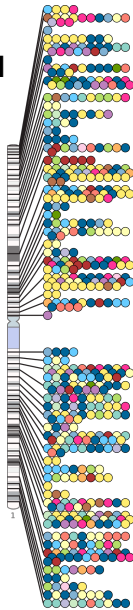
Estimated to number in the millions in the human genome.

A SNP is not a gene



Genome-wide association studies (GWAS) test whether SNPs are associated with traits

Chromosome 1



- SNP-trait associations with p-value less than 5×10^{-8} as reported in the GWAS catalog: <http://www.ebi.ac.uk/gwas/diagram#>
- traits are color-coded, e.g. cardiovascular diseases are in dark red

Some sources of genotype-phenotype data

- <http://www.personalgenomes.org/>
- opensnp.org

We are going to do our own mini-GWAS

- collect and process phenotype data
- test for association between phenotype and genotype
- discuss the ethical concerns of direct-to-consumer genetic testing