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Priorização de variantes

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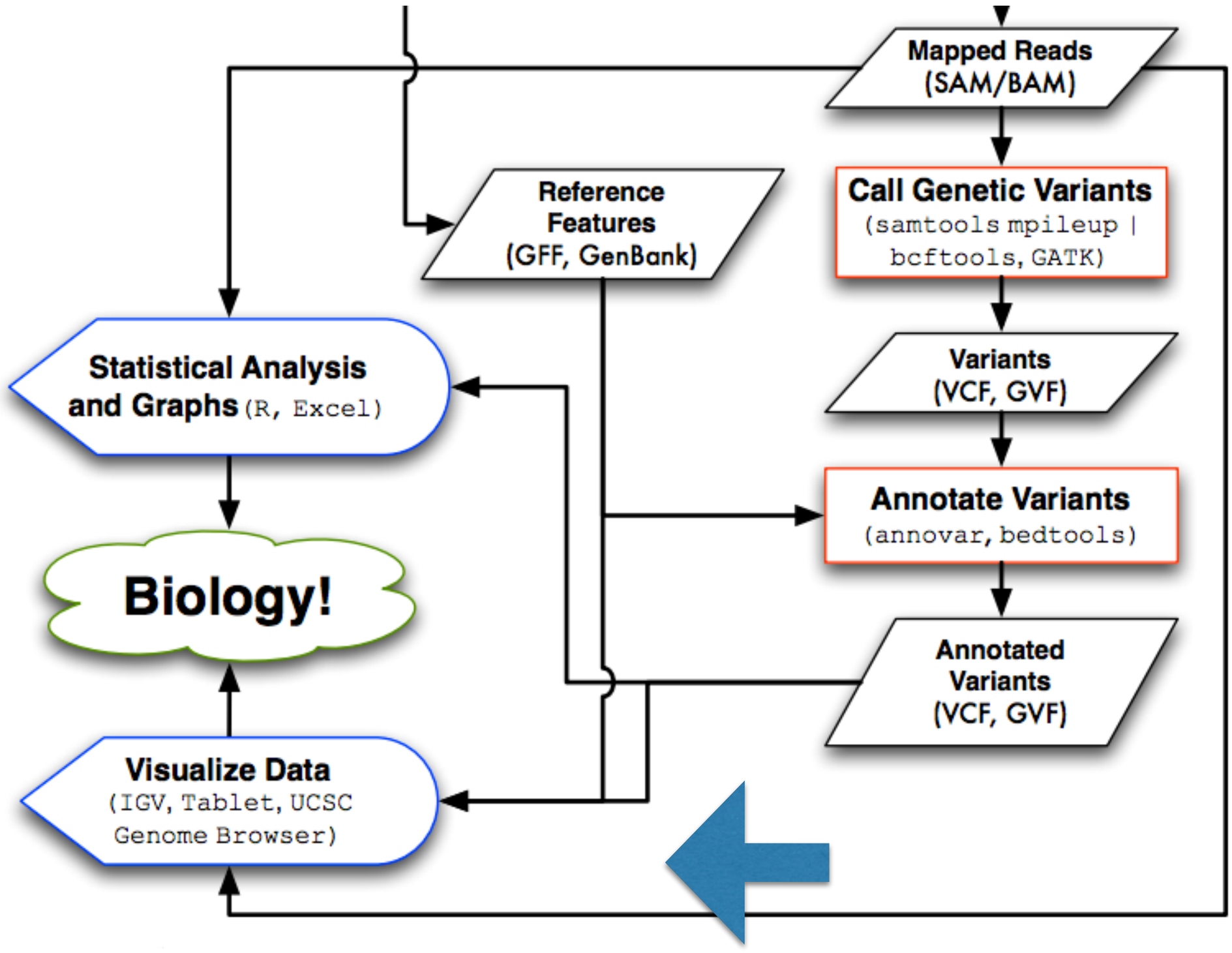
Pipeline



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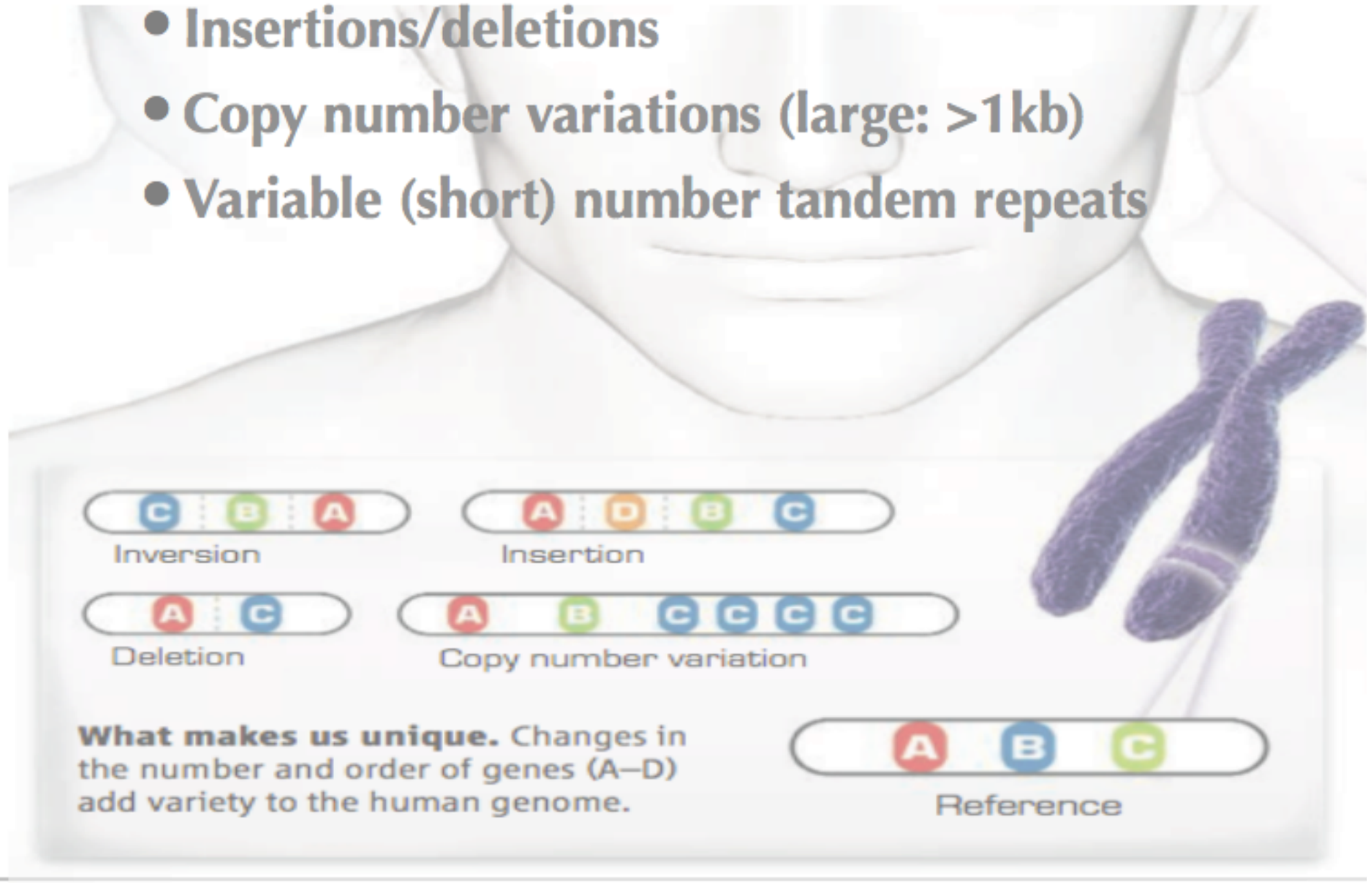
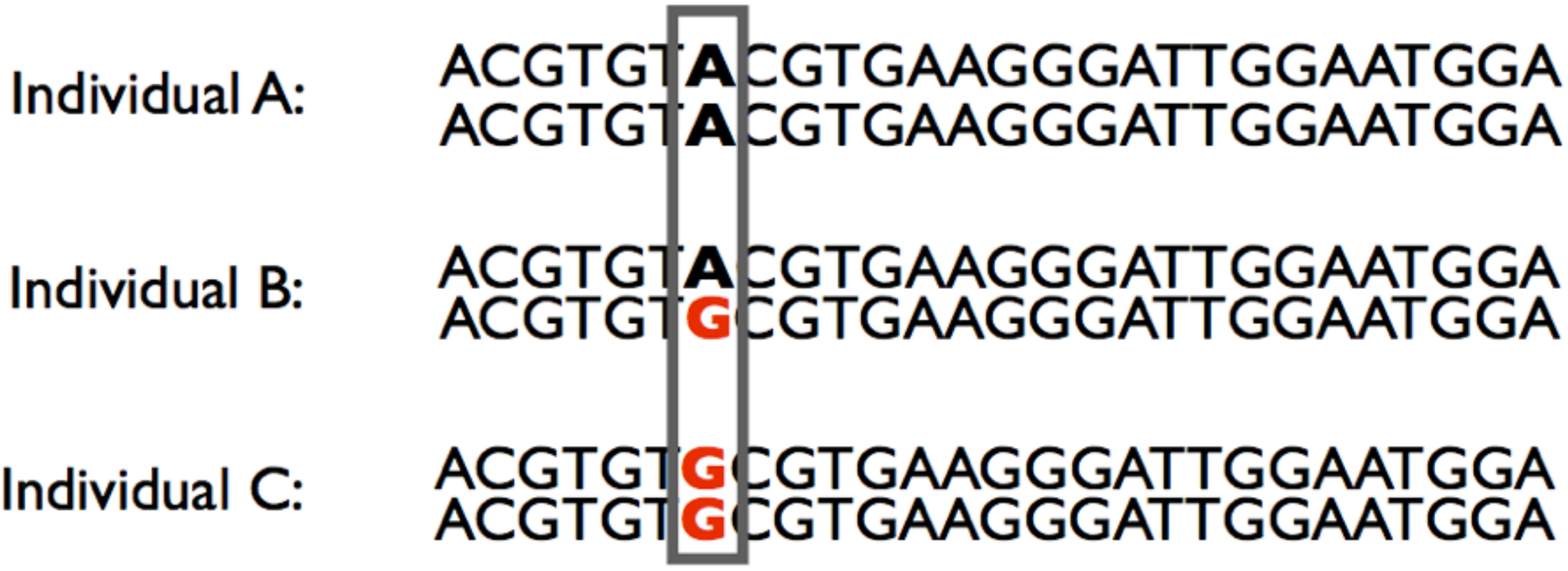
You are here!!

Genetic variation

- **Single nucleotide polymorphisms (SNP)**
- **Insertions/deletions**
- **Copy number variations (large: >1kb)**
- **Variable (short) number tandem repeats**



SNPs



SNP: One nucleotide difference occuring in at least 1% of a population

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How many SNPs differ on average between you and you?

Human: 3 billion base-pairs

1. 0.01% (eg. 300k)
2. 0.1% (eg. 3 million)
3. 1% (eg. 30 million)
4. 10% (eg. 300 million)

How many SNPs differ on average between you and you?

Human: 3 billion base-pairs

~ one base pair out of every 1,000 will be different between any two individuals

1. 0.01% (eg. 300k)
2. 0.1% (eg. 3 million)
3. 1% (eg. 30 million)
4. 10% (eg. 300 million)

SNP and genotype

Human are diploid (we have 2 homologous copies of each chromosome)

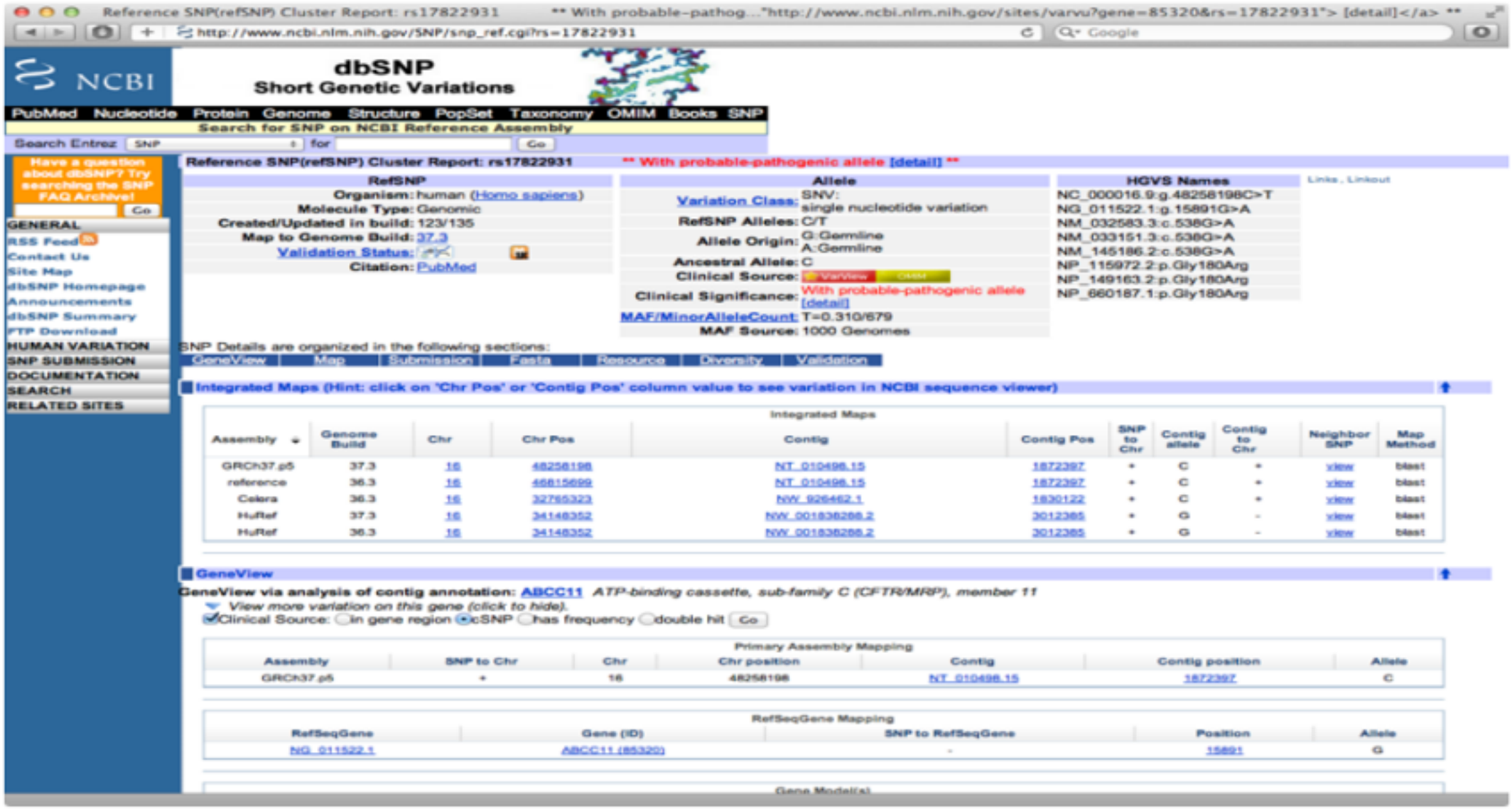
At each SNP there are 3 possible genotypes:

- homozygous reference
- heterozygous
- homozygous non-reference

Homozygous = 2 identical alleles at given locus, eg. AA
Heterozygous = 2 different alleles at given locus, eg. AC

SNP information

- rs number
- location (chr:position)



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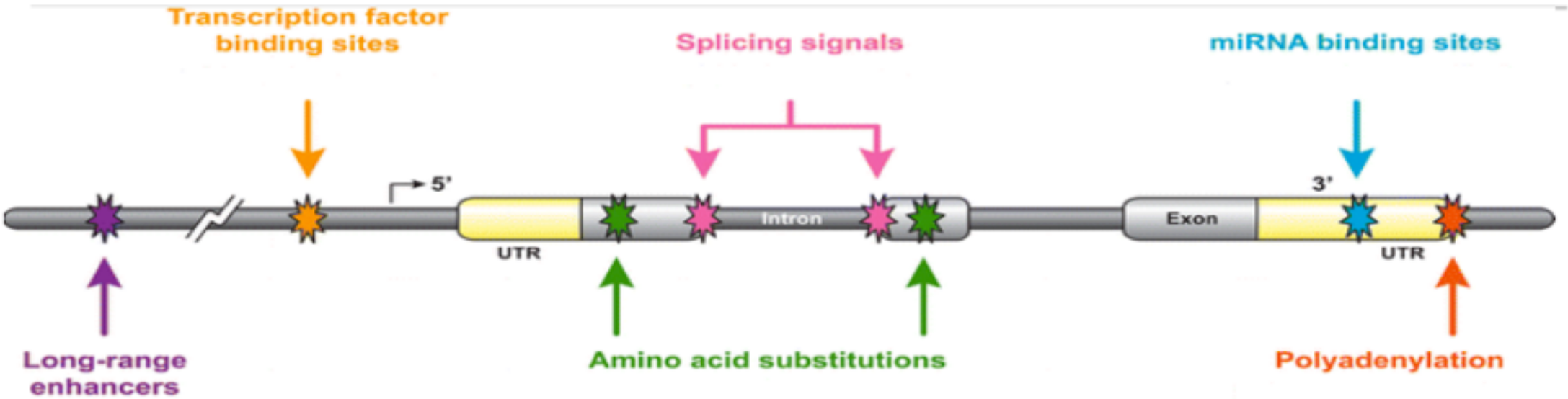
SNP consequence



Coding SNPs:

- non-synonymous coding (missense) – change of an amino acid
- synonymous coding (silent mutation) – no change in protein sequence
- stop codon (nonsense) - premature stop codon
- frame-shift coding – insertion/ deletion resulting in frame shift

Non-coding SNPs:

Mostly non-functional, but may affect:

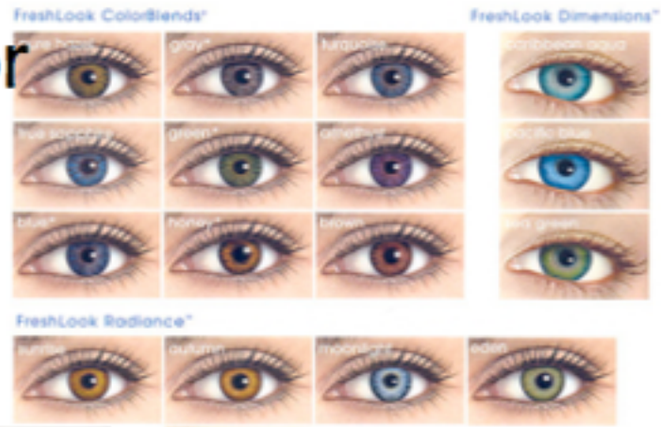
- regulatory region
- splice site
- transcription factor binding
- messenger RNA degradation
- sequence of non-coding RNA

Phenotype

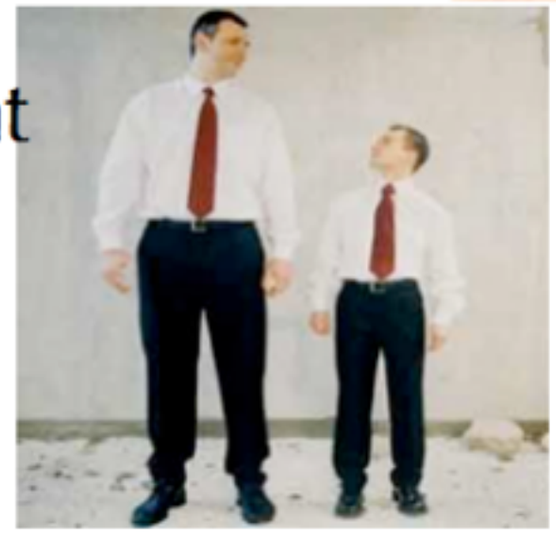
Phenotype = organism's observable characteristic or trait

obesity

eye color



height



An organism's genotype is a major influencing factor in the development of its phenotype

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