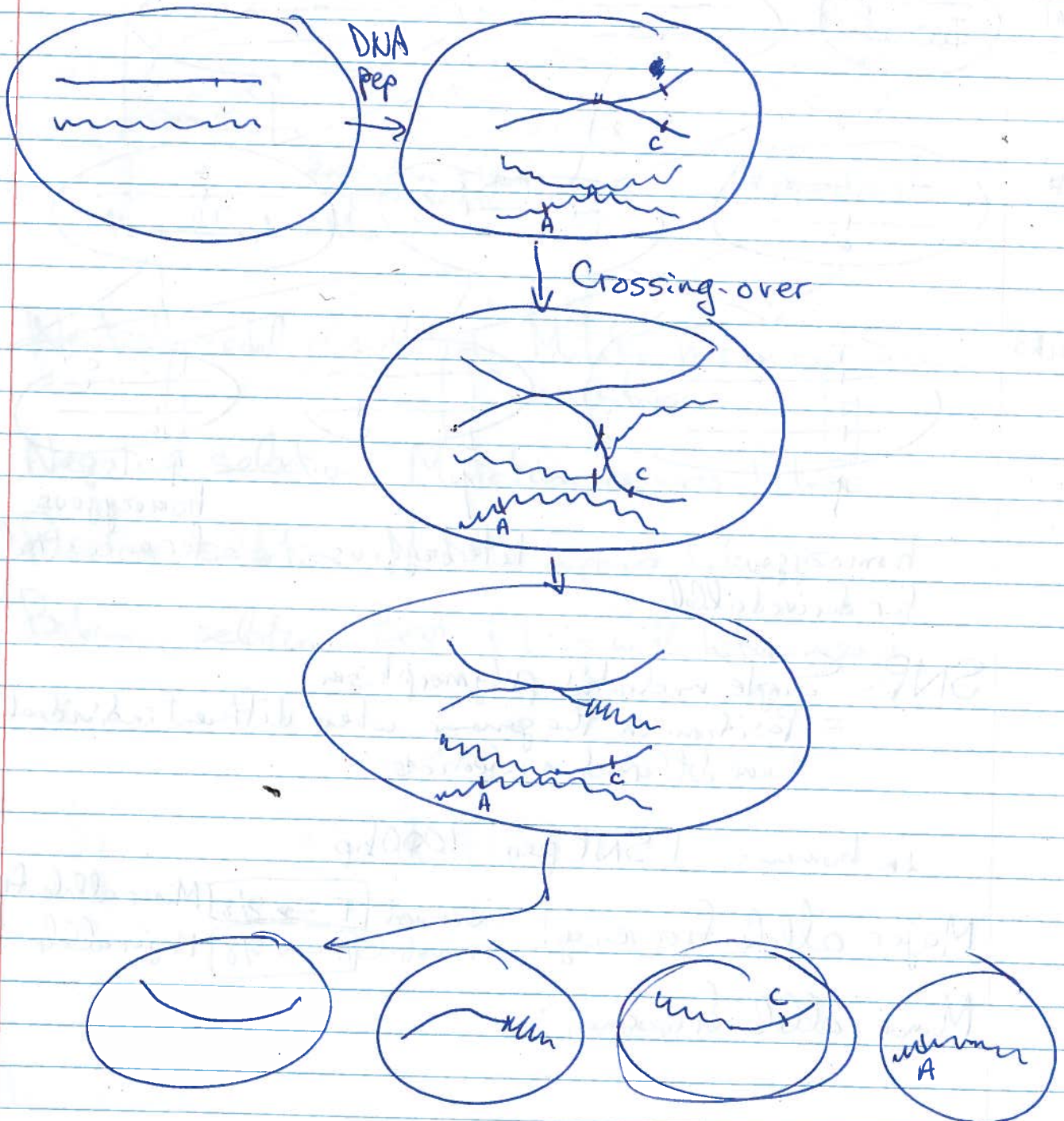
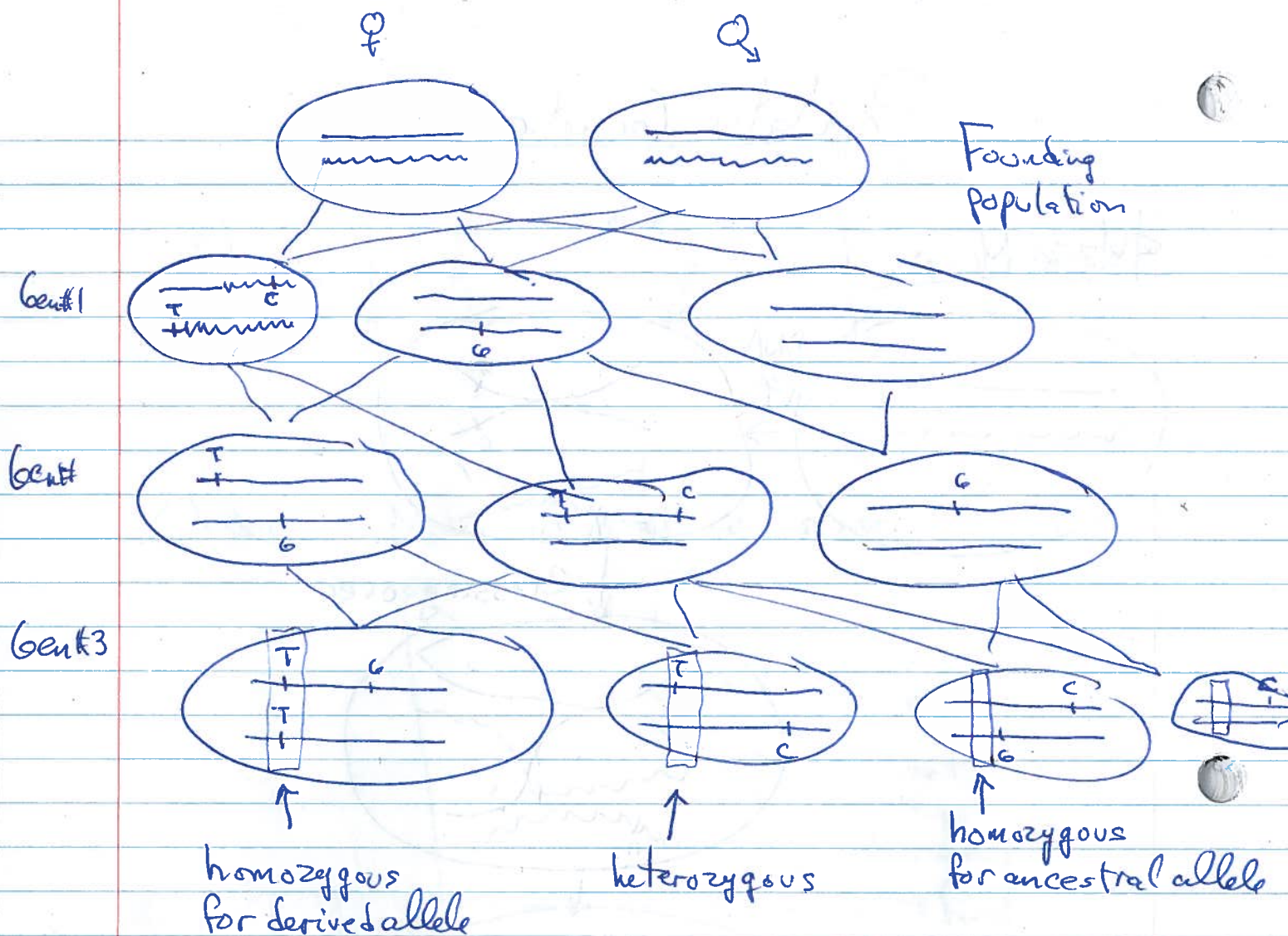


Population Genetics

~~Meiosis~~ Meiosis



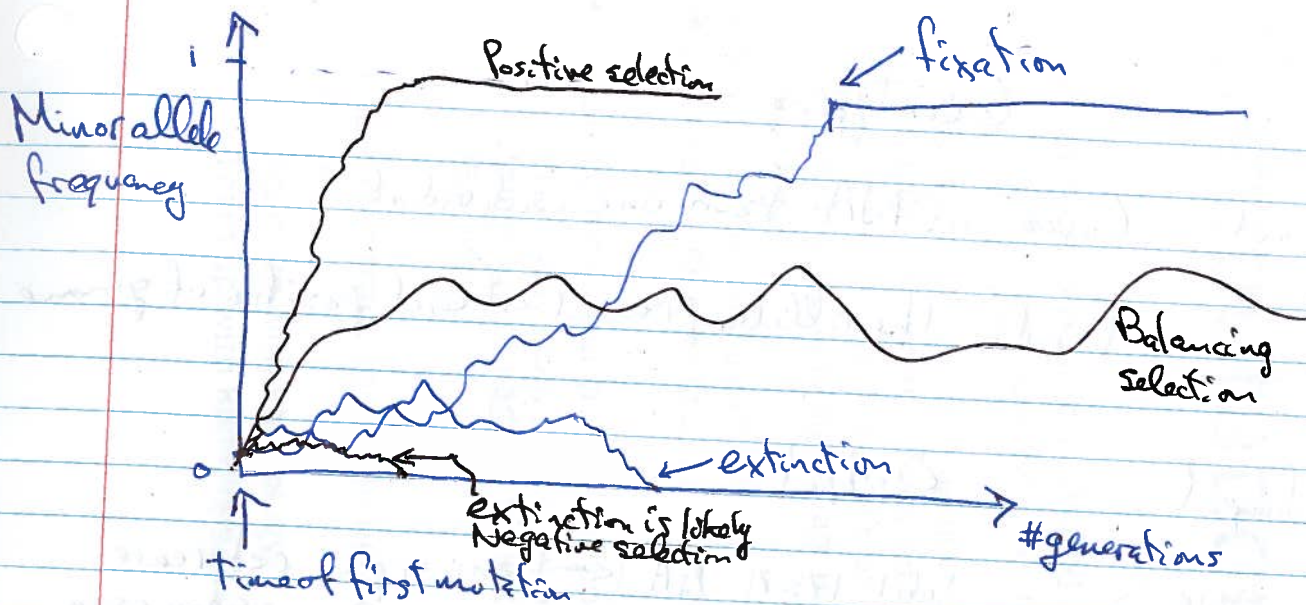


SNP: Single nucleotide polymorphism
 = Position in the genome where different individuals have different nucleotides

In human: 1 SNP per 1000bp

Major allele frequency: derived: $T \rightarrow 3/8$ Minor allele freq: 0.375
 ancestral: $A \rightarrow 5/8$ Major allele: 0.625

Minor allele frequency:



— Neutral model of selection: Mutation has no consequences on fitness

— Negative selection: Mutation reduces fitness

Positive selection: Mutation improves fitness

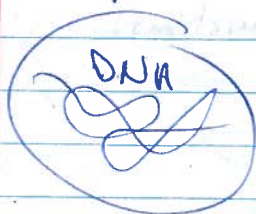
Balancing selection: Best fit is with heterozygous

Genotyping

Goal: Given DNA from one individual

Find: The alleles present at each position of genome

Input



Output

chr1	173271	AA	← homozygous reference
chr2	173471	AC	← heterozygous
chr7	...	TT	← homozygous non-reference
:	:	:	

= same as in reference human genome

~~Genotyping~~ Genotyping arrays

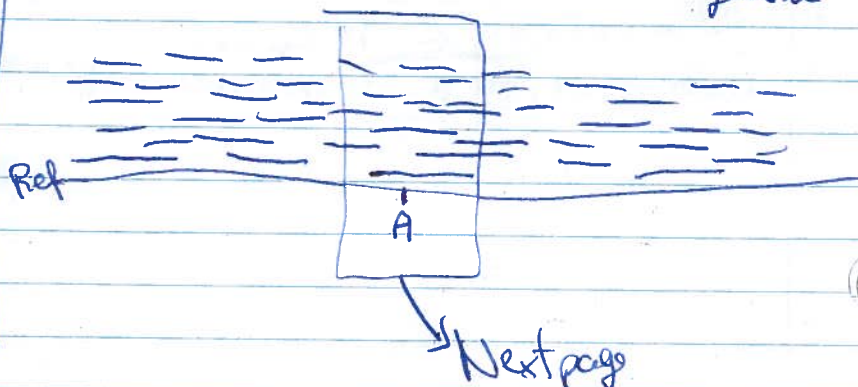
Genotyping by sequencing

① Extract + fragment + sequence DNA

Fastq file

ACZAAAC...
ATTACACCA

② Align (map) each read to reference human genome



C	A	T
A	A	T
A	A	T
C	A	T
C	A	T
C	T	T
A	A	T
T	T	T
A		

↓

4C

3A

⇓

Heterozygous
A/C

↓

1SA

1T

↓

Homozygous
non-reference

↓

Homozygous
reference