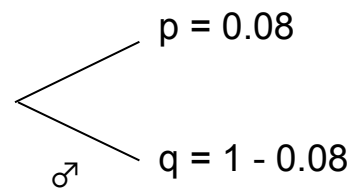
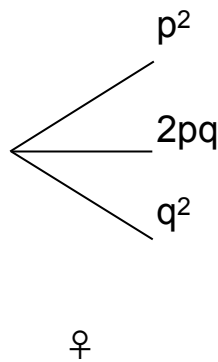


### Exam

1 → x linked in men ⇒ 8%

- 0.64% females affected
- freq allele == 0.08
- 1/7 women == carriers



2 → pop not in HWE:

- 1 of the assumptions is not met (diploid / sexual repr / same  $\sigma^7$   $\varphi$  / no mutmigel)
- 1 evolution mechanism is acting on pop → changing allele freq
- gen freq cannot be calculated using  $p^2$   $2pq$   $q^2$

3 → Allele freq in pop

- counting alleles from genotype freqs
- assuming HWE in case of 2 alleles with dominance

4 → Not nat sel

- Result of nat sel → depends on w of genotypes
- nat sel is the only mechanism to get adaptation to environment
- mutation-sel balance explains observed freq of many deleterious alleles
- **false: nat sel always fixes an allele** [take into account heteros & malaria]

5 → A & a are found in pop → A = 0.7 → environmental change:

rel w → AA=1 / Aa=1 / aa=0.43

- a is recessive detrimental
- after 1st round of sel → Aa = 0.516a and freq of A = 0.737
- when new eq is reached → allele freq: A = 1 / a = 0
- **false:** after 1st round of selection → average w = 0.948700 [it's before sel]

9 → allele with freq 0.1 in pop

- more likely to be fixed than lost by genetic drift
- will have same allele frequency in next gen
- **has a prob of fixation of 0.1**

13 → Linkage disequilibrium between alleles of 2 variants in the same chromosome:

- will decrease over time until reaching linkage eq with faster decrease in regions with low recomb rate
- will increase over time until reaching eq
- will increase over time until reaching val of  $r^2 = 1$  corresponding to existence of only 2 of the 4 possible combinations of alleles
- **will decrease over time until reaching linkage eq with faster decrease in regions with higher recomb rate**

14 → folded site freq spectrum

- requires info from outgroup
- requires info from genealogy (phylogeny)
- compares n° of monomorphic & polymorphic positions
- **all false**

15 → Tajima's D

- compares intraspecific nucleotide variations from 2 or more genes
- requires info from outgroup (single gene)
- requires info on the n° of syn & non syn changes of single gene
- **compares 2 estimators of the heterozygosity from single gene**

16 → MK test compares:

- folded & unfolded site freq spectrums
- n° of syn & non syn changes across  $\geq 2$  genes
- heterozygosity levels across  $\geq 2$  genes
- **n° syn & non syn changes from single gene**

17 → HKA test → appropriate neutrality test to

- determine whether there is a relationship between polymorphism and divergence across syn & non syn changes
- determine whether the levels of intraspecific variation btw 2 regions are as expected under neutral model
- **determine whether there is a relationship between polymorphism and divergence across  $\geq 2$  regions**

18 → mol clock hypothesis was proposed after observing:

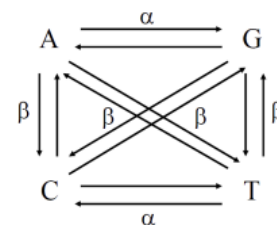
- evolutionary rate across different pros is constant
- all DNA seq have the same evolutionary rate
- the proportion of corrected aa differences for a given pro is very similar across different species
- **evolutionary rate is constant for a given pro**

19 → according to the mol clock hypothesis:

- the higher the n° of aa changes → lower subs rate
- higher n° of corrected nucleotide subs → lower subs rate
- higher length of pro → lower subs rate
- **higher the functional constraint → lower subs rate**

20 → kimura 2 parameter: subs probability from C to T is

- the same as from C to G
- the same as from C to A
- different than T to C, K2P is not reversible
- **the same as from A to G**



21 → k parameter (n° of subs by site btw 2 seq) corrected by jukes and cantor

- range: (0, 1)
- range: (0, 0.75)
- range not defined
- **range: (0,  $\infty$ )**

23 → comparison of the DNA seq of the GPM gene btw 2 species of birds

→ estimation of  $k = 0.05$  (Jukes & Cantor)

- sequences differ by more than 50 differences
- transition rate == transversion rate
- **sequences differ by less than 50** → correction shows top expected

24 → Phylogenetic tree (A — B) — C

B & C are

- monophyletic
- a polytomy
- an outgroup of A
- **paraphyletic**

25 → tree with 5 taxa: formula for rooted & unrooted trees:

- 4 unrooted trees
- 15 rooted trees
- 105 unrooted trees
- **15 unrooted trees**

26 → codon subs model

- $\text{Syn} == \Omega$
- $\text{Non syn} == K$
- Non syn depends on  $\Omega$
- **None are correct**

27 → estimation of  $\Omega$  in pro coding gene to estimate n° of syn and non syn changes ignoring codon usage bias:  $\Omega = 1.8$

- $dN < dS$
- $K_s$  was overestimated
- N° of syn sites was underestimated
- **if codon usage bias is important →  $\Omega$  is overestimated**

28 → n° syn & non nys subs btw 2 species in pro coding gene == 23

- positive selection evolution
- neutral evolution
- impossible to infer conclusion
- **negative selection**

29 → study on intron seq of orthologous genes (positive selection?)

- McDonald & Kreitman test
- Use codon subs models if genes are independent
- Use codon subs models because they take into account transition / transversion biases
- **Cannot use codon subs models**

30 → ultrametric tree

- time axis reflects amount of change
- branch lengths are proportional to the n° of changes
- time axis means nothing
- **tree is unscaled (used in mol clock where the rate of mutation is the same across all lineages of the tree)**