GE02 Exercises day 1

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- Consider a biallelic SNP with alleles T and G. The SNP is located at X-chromosome. Males have only 1 X-chromosome and thus could have genotype T (which cannot be distinguished from TT) or G (which cannot be distinguished from GG). Assuming that population consist of 50% men and 50% women, what genotypic proportions are expected?

- P(T) = 0.20

	Female			Male	
True	TT = 0.04	GT = 0.32	GG = 0.64	TY = 0.20	GY = 0.80
Observed	TT	GT	GG	TT	GG

- 1 : 1 mixture
- In mixed population: TT = 0.12 GT = 0.16 GG = 0.72

- A study population is a 3 : 1 mixture of population A and population B. Frequency of the allele of interest is 0.1 in pop A and 0.2 in pop B. Both populations are under HWE.
- 1.What is allelic frequency in the mixed population?
- Pa(A) * ¾ + Pb (A) * ¼
- = 0.1 * ¾ + 0.2 * ¼
- = 0.125

- 2.What is genotypic distribution in the mixed population?

	AA	Aa	aa
Pop A	0.01	0.18	0.81
Pop B	0.04	0.32	0.64
3:1 mixture	0.017	0.215	0.767

- What genotypic frequencies would be expected under HWE? Is it likely that the deviation from HWE due to Wahlund's effect will be detected?

- P(A) = 0.125

- Expected AA = 0.015 Aa = 0.218 aa = 0.765

- In a large population, average inbreeding is 0.03. For a variant with frequency of 0.01, compute HWE frequencies with and without assumption of inbreeding. Is it likely that the deviations from HWE due to inbreeding can be detected?

- F = 0.03
- P(a) = 0.01

Without	AA = 0.9801	Aa = 0.0198	aa = 0.0001
With	$Q^2 + pqF$	$2pq (1 – F)$	$P^2 + pqF$
	AA = 0.980397	Aa = 0.019206	aa = 0.000397