## Estimation of Mutation in Quantitative Traits:

- Mutation: the source of population genetic variation on which natural or aritifical selection acts (to produce genetic changes leading to adaptive evolution)
- Number of rounds of replication from zygote to the formation of gametes varies between species and is why rates of mutations vary between species
- Men and women: the number of rounds of replication in ova stay at 22 while it continues to grow for spermatozoa in men
- However, chromosomal mutations increase in women because of cytoplasmic deterioration in oocytes over time
- Given the difficulty of identifying genotypes, it is only possible to obtain average estimates of the effect of mutations and their degree of dominance
- Mutational variance  $V_M$  is the increase in additive genetic variance per generation due to mutation
- And thus mutational heritability is the ratio of  $V_M/V_E$
- Probability of fixation of a mutation:
  - In finite populations, genetic drift may cause beneficial alleles to be lost and deleterious alleles to be fixed
  - If the mutational selection coefficient is small  $(N_e s/N < 1)$  and  $N_e s$  is high, then the probability of fixation can be approximated by the selective advantage s, so that most beneficial mutations of small effect are lost in a population of finite size
  - Neutral mutations have a  $P_f$  equal to their initial frequency 1/2N
  - Deleterious mutations are destined to be eliminated by selection
  - Mutations where absolute value of  $N_e$  is substantially smaller than 1, the probability of fixation is the same for that of a neutral mutation. When mutation is advantageous or disadvantageous, the action of drift is more intense than selection and so the fate of the mutation is dependent on chance. This is referred to as quasi-neutral mutations.
  - Mutations can be beneficial, neutral or deleterious based on effective population size
- Estimating the rate of mutation and mutational effects
  - Balanced chromosome technique: cross an individual with another with a balanced chromosome (carrying inversions that inhibit recombinations along with visible markers for identification)

## **Problems**

(1) The aim is to carry out an experiment to estimate the heritability of a trait using pairs of parents and offspring. It is expected that the estimate of heritability will be around 0.6. It is intended to evaluate only one offspring per couple and a single parent or both. How many pairs of data would have to be evaluated in each case to obtain an estimate of heritability with a standard error equal to or less than 0.05?

$$SE(h^2) = \frac{2}{\sqrt{n}}$$

$$n = (\frac{2}{SE(h^2)})^2 = (\frac{2}{0.05})^2 = 1600$$

$$SE(h^2) = \sqrt{\frac{2}{n}}$$

$$n = (\frac{\sqrt{2}}{SE(h^2)})^2 = (\frac{\sqrt{2}}{0.05})^2 = 800$$

- (1) The estimate of heritability obtained from the regression of offspring values on that of their parents will necessarily be biased if the latter are not a random sample of the population.
  - False, if the group of parents were selected, the regerssion estimates would not be affected if the bias would occur to the same extent in the numerator and the denominator of the equation  $b_{OP} = \frac{cov(O,P)}{\sigma_P^2}$