Exam 4 Material

Thursday, April 23, 2020 1:35 PM

Lecture 19 Part I - Population Genetics

What is population genetics?

- Population genetics is defined as the genetic makeup of groups of individuals
  - Whereas mendelian genetics focuses on the genetics of an individual, population genetics focuses on the more macroscopic scale of a group of people.
  - Population genetics may be categorized two main ways:
    - Changes in genetic makeup of groups under different times or different conditions
    - Changes within groups or changes between groups.
- Population genetics is highly dependent on mathematical models.

Gene Pools

- The total set of alleles in a population
- Described by allele frequency
- One individual can carry 2 alleles at a single locus, but a population gene pool can include any number of alleles at a single locus.

A Mathematical Description of Genetic Structure of a Population

- In population genetics, the genetic structure of a population is described by its frequency of certain alleles, known as the "genotypic frequency" of a population
  - Recall that there are 3 possible genotypes, AA, Aa, and aa. The alleles are A and a respectively.
  - For example, in a population of 100 people, if 10 people have AA alleles, then the percentage of AA would be ten percent.
- Calculating genotypic frequencies
  - Simply the frequency of AA, Aa, or aa in a population.
  - f(AA) = #AA individuals / N, where f = frequency of each genotype, and N = total # individuals.
  - The sum of all phenotypic frequencies should equal to 1
- Calculating allelic frequencies
  - Defined as the number of copies of an allele (A,a) in a population divided by the total alleles.
  - f(A) = (2(AA) + (Aa)) / 2N, where N = total # individuals
  - f(a) = (2(aa) + (Aa)) / 2N, where N = total # individuals
  - Note also that this means that f(A) + f(a) = 1.
  - In population genetics, f(A) and f(a) are assigned to p and q, respectively.
  - In a population with multiple alleles (A1, A2, A3, A4 etc.), the same methodology applies:
    - Example with 3 alleles:

      p = f(A1) = (2(A1A1) + (A1A2) + (A1A3)) / 2N
      q = f(A2) = (2(A2A2) + (A1A2) + (A2A3)) / 2N
      r = f(A3) = (2(A3A3) + (A1A3) + (A2A3)) / 2N

  - For the calculation of X-linked loci, calculation separately includes males and females, as females have 2 X chromosomes, and males only have one.
    - Example:

      p = f(X1) = (2(X1X1) + (X1X2) + (X1Y)) / 2N
      q = f(X2) = (2(X2X2) + (X1X2) + (X2Y)) / 2N

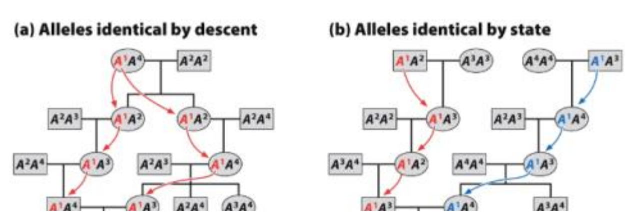
      where N is the total of both male and female populations.

Hardy Weinberg Law

- How do segregation of alleles in gamete formation and combining of alleles at fertilization influence the gene pool?
- The Hardy Weinberg law is a mathematical model of how reproduction impacts the genotypic and allele frequencies of a population.
  - p^2 represents the homozygous dominant allele pair frequency
  - q^2 represents the homozygous recessive allele pair frequency
  - 2pq represents the heterozygous allele pair frequency.
- Assumptions in order for Hardy Weinberg Law to work:
  - The population must be large
  - Mating within the population must be random
  - The population is not affected by mutation, migration, or natural selection.
  - If these assumptions are met, then two predictions can be drawn:
    - The allelic frequencies of the population do not change
    - The genotypic frequencies of the population stabilize
    - That is, after one generation of random mating, allelic and genotypic frequencies are not altered, and allelic frequencies determine genotypic frequencies.
- These assumptions and their results demonstrated that reproduction alone will not result in evolution because the genetic structure of a population will not change only with reproduction.
- Hardy Weinberg Equilibrium
  - The allelic and genotypic frequencies in a population will remain constant from generation to generation in the absence of other evolutionary influences.
  - $1 = p^2 + 2pq + q^2$
  - If the population deviates from this equilibrium, this indicates that other evolutionary influences are at work.
- The implications of the Hardy Weinberg Law
  - When the frequency of one allele is high, most of the individuals in the population are homozygous for that allele.
  - The greatest frequency possible for heterozygous traits is 50%, or when p = q = 0.5

Lecture 19 Part II - Population Genetics Part II

- Nonrandom mating affects the genotypic frequencies of a population - within a population, nonrandom mating changes the way in which existing alleles combine, which alters frequencies.
  - Positive assortative mating - a tendency for like individuals to mate
  - Negative assortative mating - a tendency of unlike individuals to mate
- Inbreeding - the preferential mating between related individuals
  - Increases the proportion of homozygotes while decreasing the amount of heterozygotes in a population.
  - Causes a departure from Hardy-Weinberg equilibrium



        - Negative assortative mating - a tendency of unlike individuals to mate
- Inbreeding - the preferential mating between related individuals
    - Increases the proportion of homozygotes while decreasing the amount of heterozygotes in a population.
    - Causes a departure from Hardy-Weinberg equilibrium
    - Measurements of inbreeding:
        - F = inbreeding coefficient
        - F = probability that alleles are the same by descent. Therefore:
            - 0 = random mating, 1 = 1 all alleles are identical by descent
        - Inbreeding increases the homozygotic population at the cost of heterozygotes:
            - $f(AA) = p^2 + Fpq$
            - $f(Aa) = 2pq - Fpq$
            - $f(aa) = q^2 + Fpq$
    - Within hermaphrodite species, selfing is the most extreme example of inbreeding, as they are only breeding within themselves.
        - In the table to the right, we see that as generations -> infinity, the amount of Aa genotypes approaches 0.
        - This is genetically unfavorable, as populations that exclusively breed through selfing will ultimately die off.
        - C elegans is a popular model organism that displays this effect.
    - Inbreeding Depression
        - The increase in frequency of homozygotes for recessive alleles can sometimes result in detrimental alleles being prop
        - In crops, inbreeding tends to have deleterious effects
        - Under selection, however, homozygosity may also weed out harmful alleles, but also preserve beneficial alleles.
            - However, this may leave the species unprepared for changes in environmental conditions that may rely on a diverse set of alleles - a lack of "cryptic variation"
- Outcrossing - the preferential mating between unrelated individuals
- Individuals may be homozygous by descent or by state. In other words, their two alleles may have come from the same individual or different individuals. (Refer to diagram)

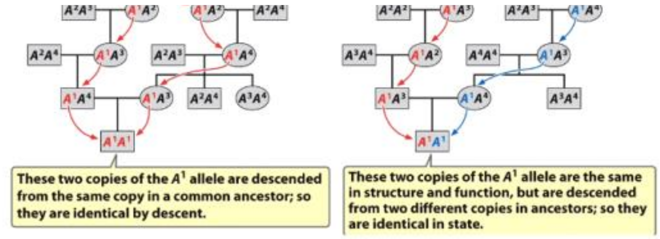


TABLE 25.2 Generational increase in frequency of homozygotes in a self-fertilizing population starting with $p = q = 0.5$

Generation	Genotypic Frequencies		
	AA	*Aa*	*aa*
1	1/4	1/2	1/4
2	1/4 + 1/8 = 3/8	1/4	1/4 + 1/8 = 3/8
3	3/8 + 1/16 = 7/16	1/8	3/8 + 1/16 = 7/16
4	7/16 + 1/32 = 15/32	1/16	7/16 + 1/32 = 15/32
n	$(1 - (1/2)^n)/2$	$(1/2)^n$	$(1 - (1/2)^n)/2$
∞	1/2	0	1/2