Lectures 41 and 42 – Population genetics

I. Population genetics
- population – group of organisms of same species living in area within which likely to find mates
- subpopulation – distinct breeding group within limited area
- gene pool – complete set of genetic info within individuals in a population

II. allele and genotype frequencies
A. sometimes measurable, eg. M, N blood group
   - codominant
   - 2 alleles of single gene
   - 3 possible genotypes: MM, MN and NN
   - British study of 1000 individuals

<table>
<thead>
<tr>
<th>phenotypes</th>
<th>genotypes</th>
</tr>
</thead>
<tbody>
<tr>
<td>298 M</td>
<td>298 MM</td>
</tr>
<tr>
<td>489 MN</td>
<td>489 MN</td>
</tr>
<tr>
<td>213 N</td>
<td>213 NN</td>
</tr>
</tbody>
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- frequencies must add to 1 when all possibilities have been considered
- if allele frequency = 1.0, allele is fixed
- if allele frequency = 0, allele is lost

III. Behavior of alleles in population
A. Hardy-Weinberg principle
   1. applies to random mating, equivalent to random union of gametes
      - consider M and N, present with frequencies of p and q, respectively

   - if only two alleles in population:

   2. can use Hardy-Weinberg to calculate allele or genotype frequencies in population (if population meets assumptions below)
      eg. studying some recessive phenotype present in 16% of population
      - allele frequencies:

      - genotype frequencies:
3. conditions under which Hardy-Weinberg holds true
   a. random mating
   b. allele frequencies same in male and female
   c. no selection
   d. no migration into population
   e. no new mutation
   f. population large enough that allele frequency doesn’t change through generations by chance
4. implications of Hardy-Weinberg
   a. for rare alleles, frequency of heterozygote >> frequency of homozygote
   b. frequencies of alleles stable in population
5. can derive allelic and genotypic frequencies
   a. autosomal recessive
     eg 1: cystic fibrosis 1/1700 individuals are affected
     - double check: frequency of two producing affected child
       P(2 carriers having child) X P(child homozygous)
     eg 2: galactosemia 1/40000 individuals are affected

   - carrier frequency can inform pedigree analysis
     eg. autosomal recessive galactosemia
     What is the probability that the person indicated by question mark will be affected?

   b. autosomal dominant
     eg: familial hypercholesterolemia - affects 1/500
     Note: affected individuals are HH or Hh

   b. autosomal dominant – a simplification
     eg: familial hypercholesterolemia – affects 1/500
     Note: affected individuals are HH or Hh, but if affected is <1/100, then vast majority of affected are Hh
c. X-linked recessive (if allele frequencies same in male and female), males are either mutant or wild type, $q$ or $p$
   eg: colorblindness – affects 0.08 males
   Note: frequency of effected males = $q$

d. X-linked dominant (if allele frequencies same in male and female), males either mutant or wild type, $p$ or $q$, effected females are $2q$
   eg: Coffin-Lowry – affects 1/50,000 males
   Note: frequency of effected males = $q$

IV. mating system affects behavior of alleles in population
   - mating system – norms by which members of population choose mates
     - random – mating pairs form without regard to phenotype
     - assortative – mating pairs form on basis of similarity of phenotype
       - positive
       - negative
     - inbreeding – mating between relatives
     - eg. negative assortative mating

- insects that work at surface of flower, pollinate pin with thrum pollen
- insects that work deep in flower, pollinate thrum with pin pollen
V. Genetics and evolution
- requires deviation from Hardy-Weinberg
- evolution refers to change in gene pool resulting from progressive adaptation
A. 4 processes account for most of the change in allele frequency
1. mutation
2. migration – movement of subpopulations
3. natural selection – favors particular allele or combination of alleles
4. random genetic drift – random change in allele frequency that occurs by chance

B. natural selection – 3 premises
1. more offspring produced than can survive
2. organisms differ in their ability to survive and reproduce
3. genotypes that promote survival are present in excess among reproducing individuals
4. selection in diploids:

C. adaptation permits organism to change in response to changing environment
eg: peppered moth (*Biston betularia*)

- before 1850, 99% were peppered
- by 1959, in some parts of Britain, melanistic >90% of population
- by 1985, melanistic had declined to 50%