

Reading: Chap. 4 pp. 117-119

Lecture Outline:

1. Allele frequencies in populations
2. Hardy Weinberg equilibrium
3. Disease prediction/Genetic counseling

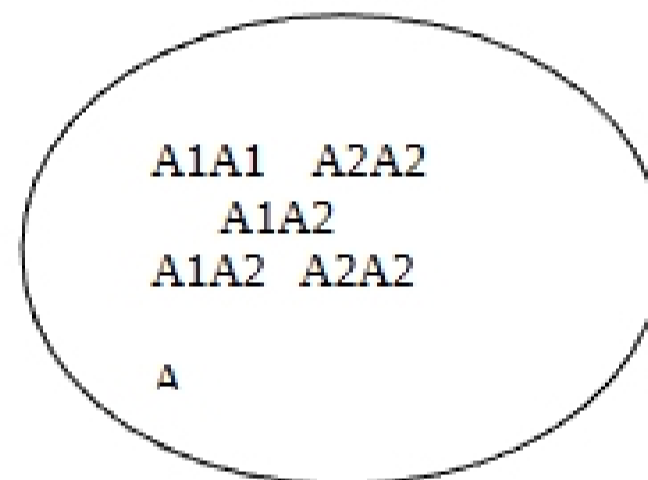
Lecture

1. Allele frequencies

Population: Group of interbreeding individuals.

Gene Pool: All the alleles for a particular gene in a population.

Gene frequency=allele frequency (more precise phrase)=proportion of all alleles for gene A of one type (such as A1)



Pick an allele at random from the gene pool

Chance= p that allele is A1
q that allele is A2

Pick a second allele at random

Chance= p that allele is A1
q that allele is A2

Chance that both alleles were A1 = $p \times p = p^2$

Chance that both alleles were A2 = $q \times q = q^2$

Chance that first allele was A1, second was A2 = pq

Chance that first allele was A2, second was A1 = qp
Overall chance of one A1 and one A2 allele is $2pq$.

2. Hardy Weinberg equilibrium

Gene frequencies and genotypes of a population can be predicted using probabilities.

Autosomal locus

Genotype	Frequency
A1A1	p^2
A1A2	$2pq$
A2A2	q^2

X-linked locus

Females	Frequency	Males	Frequency
A1A1	p^2	A1	p
A1A2	$2pq$	A2	q
A2A2	q^2		

Assumptions:

- Random mating
 - Inbreeding=non-random mating decreases heterozygosity; and increases homozygosity

3. Disease prediction/Genetic counseling

See Box 4.6

a. Calculate carrier frequency

An autosomal recessive condition affects 1 newborn in 10,000. What is the expected frequency of carriers?

	unaffected	unaffected	affected
phenotypes	AA	Aa	aa
frequencies	p^2	$2pq$	q^2

$$\text{Affected individuals} = q^2 = 1/10,000$$

$$\sqrt{q^2} = q = 1/100$$

1 in 100 alleles at the A locus are a

99 in 100 are A

The carrier frequency, $2pq = 2 (99/100)(1/100) = 0.02$

If a parent of an affected child remarries, what is the risk of producing an affected child in the new marriage?

Apply the product rule

Probability that parent that had affected child is a carrier		Risk of new spouse being carrier	probability of affected child
1	x	0.02	x ¼ = .005

X-linked recessive red-green color blindness affects 1 in 12 British males. What proportion of females will be carriers? What proportion will be affected?

Males A=normal
a=colorblind

Females AA or Aa = normal
aa= colorblind

Females	Frequency	Males	Frequency
AA	p^2	A	p
Aa	$2pq$	a	$q = 1/12$
aa	q^2		

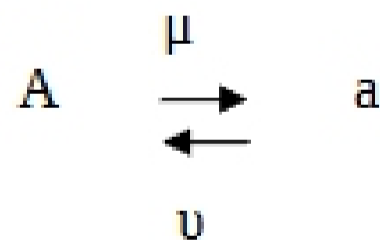
From males, $q=1/12$ therefore $p=11/12$.

Female carriers = $2pq = 2 \times 11/12 \times 1/12 = 22/144$

Females affected = $q^2 = 1/12 \times 1/12 = 1/144$

Other forces that affect allele frequencies

a. mutation



μ = mutation rate/gene/generation

u = back mutation rate/gene/generation

b. Natural selection

Can act on dominant or recessive trait

Can have heterozygote superiority $Aa > AA$ or aa