2nd Lecture

Population Genetics
(Gene Frequency)

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Population Genetics

Is the study of genetic variation within populations, and involves the examination and modelling of changes in the frequencies of genes and alleles in populations from one generation to the next

Any population contains a large number of breeding groups and is the effect of **gene transfer** from generation to generation on productive performance for each generation.

Population genetics gives us an opportunity to step back and observe the genetic change over time. By comparing populations to each other (and to themselves), we can begin to see how outside factors might spark the evolution of a trait.

Gene Frequency:

In breeding breeders are usually more interested in changes in the genetic characteristics of a population from one generation to another.

Gene frequency which is the ration of the total number of **A** genes to the total number of **all genes** in the population which are at the same locus.

The range of any gene frequency is (zero – one)

- In a population of **N** animals there are **2N** alleles at a particular locus.

- Since each AA (aa) individual has **2A (2a)** alleles while each Aa individual has only **one (for each)**. So the gene frequency of **A (a)** will be:

p = (2AA + Aa) / 2N = the frequency of A Where: N=AA+Aa+aa
While the gene frequency of **a** will be:

q = (2aa + Aa) / 2N = the frequency of a

p+q=1 gene frequency
So: q = 1-p
(p+q)(p+q)=1 genotype frequency
$p^2+2pq+q^2 = 1$

Example:-

Suppose we have a flock short horn cows consist of **16, 48, and 36** individuals with **Red (WW), redden white (Ww),** and **white (ww)** color:

N=16+48+36= 100 individuals in the flock, in other words we have 2N=200 alleles in this population.

$p_W = (2WW + Ww) / 2N$

$= 2*16+48/200 = 80 / 200 = 0.4$ W,

$q_w = (2ww + Ww) / 2N = 120 / 200 = 0.6$ w,

So, (0.4W, 0.6w) is the gametic array (gene frequency)

zygotic or genotypic array (genotype frequency) is

$(0.4W, 0.6w)^2 = (0.16WW + 0.48\,Ww + 0.36ww)$

Example: a flock is composed of 36 **black** (*BB*), 44 **blue** (*Bb*) and 20 **white** (*bb*). Compute the gene and genotypic frequencies.

Genotypic frequencies*:*

- Frequency (*BB*) = p = 36/100 = 0.36

- Frequency (*Bb*) = h = 44/100= 0.44

- Frequency (*bb*) = q = 20/100 = 0.20

- *Gene frequencies:*
- *p* = frequency (*B*) = *(2x36+44)/(2x100) = 116/200= 0.58*
- *q* = frequency (*b*) = *(44+2x20)/(2x100) = 84/200 = 0.42*

- Note that $q = 1-p$

In a population there are 1000 animal (for example), there are 160 recessive homozygous and 480 heterozygous genotypes. Calculate the frequency of the dominant allele.

AA		Aa		aa	
360	+	480	+	160	**= 1000**

Aa = 480/ 2 = 240 so ; 240 for AA, 240 for aa

AA= 360 + 240 = 600; 600/1000 **= 0.6 freq A**

Or; pA=2*360+480/2000 = **0.6**

aa = 240 + 160 = 400; 400/1000 = 0.4 freq a

or freq a = 1- 0.6 = 0.4

Example:

A1A1 15	**A2A2 10**	**A3A3 12**	
A1A2 18	**A1A3 25**	**A2A3 30**	**N=110**

qA1= 2 *15+18+25/2*110= 73/220

qA2= 2*10+ 18+30/2*110= 68/220

qA3= 2*12+25+30/2*110= 79/220

73/220+68/220+79/220=220/220= 1

Random mating: It is a type of mating where each and every individual has equal chance of being mated with other individual.

Hardey-Weinberg Law: Random mating exists when each individual of a particular sex in a population has an equal probability of mating with a certain individual of the other sex. So " In the **absence of selection**, **mutation**, **chance**, and **migration**, the gene frequency remains **constant** from generation to generation under **random mating**, **such a population is said to be in genetic equilibrium?**

frequency does not change from generation to generation

Two aspects of the Hardey-Weinberg law can be stated:

1. The **gene frequencies** are the same in **parents** and **progeny**.
2. The **genotype frequencies** in the **progeny** depend only on the **gene frequency** in the **parents** and not on the **genotypes** frequencies,

Test of Hardey-Weinberg equilibrium:

According to H-W law, the genotype frequency of progeny are determined by the gene frequency in their parents. If the population is in equilibrium, the gene frequency is the **same** in parents and progeny.

We have two methods to test H.W. equilibrium:

1. By comparing numbers of **observed** genotypes with numbers of **expected** genotypes, in other words we compare **calculated (chi-square) X^2** by **tabulated** one.
2. Comparing the **heterozygotes** genotypes portion by the value **2pq**.

	P										
DF	0.995	0.975	0.2	0.1	0.05	0.025	0.02	0.01	0.005	0.002	0.001
1	.0004	.00016	1.642	2.706	3.841	5.024	5.412	6.635	7.879	9.55	10.828
2	0.01	0.0506	3.219	4.605	5.991	7.378	7.824	9.21	10.597	12.429	13.816
3	0.0717	0.216	4.642	6.251	7.815	9.348	9.837	11.345	12.838	14.796	16.266
4	0.207	0.484	5.989	7.779	9.488	11.143	11.668	13.277	14.86	16.924	18.467
5	0.412	0.831	7.289	9.236	11.07	12.833	13.388	15.086	16.75	18.907	20.515
6	0.676	1.237	8.558	10.645	12.592	14.449	15.033	16.812	18.548	20.791	22.458
7	0.989	1.69	9.803	12.017	14.067	16.013	16.622	18.475	20.278	22.601	24.322
8	1.344	2.18	11.03	13.362	15.507	17.535	18.168	20.09	21.955	24.352	26.124
9	1.735	2.7	12.242	14.684	16.919	19.023	19.679	21.666	23.589	26.056	27.877
10	2.156	3.247	13.442	15.987	18.307	20.483	21.161	23.209	25.188	27.722	29.588
11	2.603	3.816	14.631	17.275	19.675	21.92	22.618	24.725	26.757	29.354	31.264
12	3.074	4.404	15.812	18.549	21.026	23.337	24.054	26.217	28.3	30.957	32.909
13	3.565	5.009	16.985	19.812	22.362	24.736	25.472	27.688	29.819	32.535	34.528
14	4.075	5.629	18.151	21.064	23.685	26.119	26.873	29.141	31.319	34.091	36.123
15	4.601	6.262	19.311	22.307	24.996	27.488	28.259	30.578	32.801	35.628	37.697
16	5.142	6.908	20.465	23.542	26.296	28.845	29.633	32	34.267	37.146	39.252
17	5.697	7.564	21.615	24.769	27.587	30.191	30.995	33.409	35.718	38.648	40.79
18	6.265	8.231	22.76	25.989	28.869	31.526	32.346	34.805	37.156	40.136	42.312
19	6.844	8.907	23.9	27.204	30.144	32.852	33.687	36.191	38.582	41.61	43.82
20	7.434	9.591	25.038	28.412	31.41	34.17	35.02	37.566	39.997	43.072	45.315

Assuming H W equilibrium

if p = frequency of A1 allele

if q = frequency of A2 allele

Genotype frequencies are

	A1 (p)	**A2 (q)**
A1 (p)	A1A1 *pxp* $=p^2$	A1A2 *pxq*
A2 (q)	A2A1 *qxp*	A2A2 *qxq* $=q^2$

p^2 A1A1

2pq A1A2

q^2 A2A2

In the previous example, test if the flock is in H.W. equilibrium?

p = 0.4, q = 0.6,

$(0.4W, 0.6w)^2$ = (0.16WW+ 0.48 Ww + 0.36ww)

so 2pq = 2(0.4)(0.6) = 0.48

This means that this flock is in H.W. equilibrium.

Ex.: In the following frequencies are given as follows:

	MM	MN	NN	Total
Observed number:	233	385	129	747

p_M = (2MM + MN) / 2N = 2×233 + 385 / 2×747 = 0.5696,

q_N = 2×129 + 385 / 2×747 = 0.4304,

or: q_N = 1- p_M = 1- 0.5696 = 0.4304,

So, (0.5696M, 0.4304N) is the gametic array,

Zygotic (genotypic) array = $(0.5696M, 0.4304N)^2$

= (0.32MM+ 0.49 MN + 0.19NN),

MM = P^2 * N = 0.32*747 = 239

MN = 2pq * N = 2*0.5696*0.4304*747 = 366

NN = q^2 * N = 0.19*747= 142

	MM	MN	NN	Total
Expected Number:	239	366	142	747

$X^2 = \Sigma$ (observed – expected)^2 / expected

$= \Sigma (Oi – Ei)^2 / Ei$

$$= \frac{(233-239)^2}{239} + \frac{(385-366)^2}{366} + \frac{(129-142)^2}{142} = 0.15 + 0.98 + 1.19 = \mathbf{2.32}$$

It is shown that calculated $\mathbf{X^2}$ **(2.32)** less than tabulated $\mathbf{X^2}$ **(5.99**),

then null hypothesis

will accepted, and we can concluded that the observed values does not differ significantly from those expected one.

So, this flock is in H.W. equilibrium

Example: - A flock of 1000 individuals is composed of the following genotypes at a given locus: 300 BB, 400 Bb, and 300 bb. Is the flock in Hardy- Weinberg equilibrium at this locus?

p = frequency (B) = (300 (2) + 400)/2000 = 0.5

q = frequency (b) = 1-p = 0.5

The expected numbers:

BB ➔ p2 (total number) = (0.5)2 (1000) = 250

Bb ➔ 2pq (total number) = 2(0.5) (0.5) (1000) = 500

bb ➔ q2 (total number) = (0.5)2 (1000) = 250

- To test if the population is in H-W equilibrium we compare the observed numbers with the expected numbers by computing the following test:

$$\chi^2 = \sum \frac{(O-E)^2}{E} = \frac{(300-250)^2}{250} + \frac{(400-500)^2}{500} + \frac{(300-250)^2}{250} = \mathbf{40}$$

(Note: $c^2_{1, 0.05}$ = 3.84)

The calculated value is larger than the tabulated value of 3.84. Therefore, we reject the hypothesis that the population is in H-W equilibrium and **conclude that the population is not in H-W equilibrium**.

Example:-

If male take from the population zygotic array is aa**0.64** Aa**0.32** AA**0.04**

While females took from the population zygotic array is aa**0.36** Aa**0.48** **AA0.16**

What is gametic array and zygotic array of population resulting of mating randomly?

♂ qa=√0.64 =0.8 ♀ qa=√0.36 =0.6

qA=√0.04 =0.2 qA=√0.16 =0.4

♂ / ♀	qA=0.2	1-qA=0.8
qA=0.4	AA 0.08	Aa 0.32
1-qA=0.6	Aa 0.12	aa 0.48

aa 0.48+ Aa 0.44 +AA 0.08 $(aa0.7+AA0.3)^2$

= aa0.49 +Aa0.42 +AA0.09

Or 2q (1-q) = 2×0.7×0.3=0.42

0.42 ≠ 0.44 ∴ **the population is not in H-W equilibrium**.

In a population that is in equilibrium, the proportion of individuals showing the dominant trait at a given locus having 2 alleles is 84%. What is the frequency of recessive allele in the population?

Dominant freq + Recessive freq= 1

Dominant freq allele AA = proportion -84% = 0.84

Freq Recessive aa = 1- 0.84 = 0.16

(q2) a2 = 0.16

(q) a= √0.16 = 0.4

- If 16% of a flock (sheep) (had a recessive trait) and 50% were eaten by wolves before they could mate, how many would have this condition in the next generation?

- In an autosomal recessive inherited condition in animal, if (Proposed) its frequency in newborns is 1/10000 in nature. What is the frequency of the carriers?

- Assume a population in which 36% of the population are homozygous for a certain recessive allele (a) assume the population is at equilibrium.

- **1. what is the freq. recessive allele (a)**
- **2. what is the freq. dominant allele (A)**
- **3. what percentage of the population are homozygous for (A)?**
- **4. what percentage of the population are heterozygous?**
- **5. why do we have to start the problem with the percentage of the homozygous recessive in the population?**