CHAPTER 5

POPULATION GENETICS

(Hours: 1L + 3T)

CHAPTER 5: POPULATION GENETICS

5.1 Gene Pool Concept

5.2 Hardy-Weinberg Law

UPS 2	PSPM 1
3 MCQ	6 marks

LEARNING OUTCOMES

5.1 Gene pool concept

a) Explain population genetics, gene pool, allele frequencies, genotype frequencies and genetic equilibrium. (C2 -LECTURE)

Population Genetics

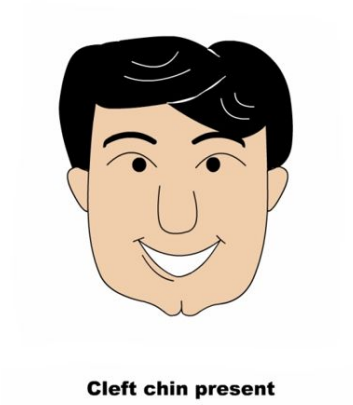
- The study of the genetic variation in a population of a species and the evolutionary forces that act on it.

Population

- A group of individuals of the **same species** living together in the **same place** at the **same time**.
- that can **freely interbreed** with each other and produce **fertile offspring** in nature.



Genetic Variation Examples



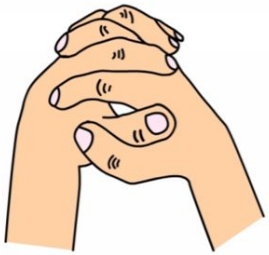
Cleft chin present



Cleft chin absent



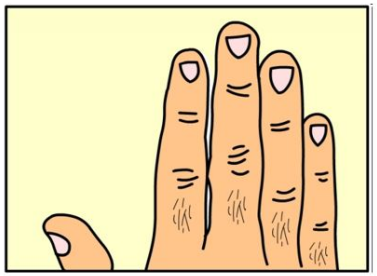
Hand Clasping - Left thumb over right



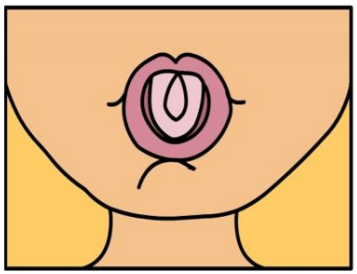
Hand Clasping - Right thumb over left



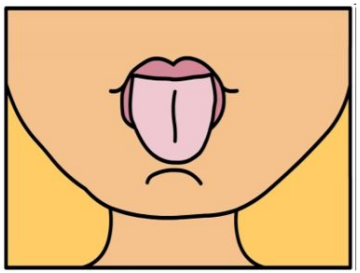
Mid-Digital Hair Present



Mid-Digital Hair Absent



Able to roll tongue



Unable to roll tongue

Figure 1

Gene Pool

- The **total of all alleles for all loci present in a population**.
- Consist of all copies of every type of allele at every locus in all members of the population.

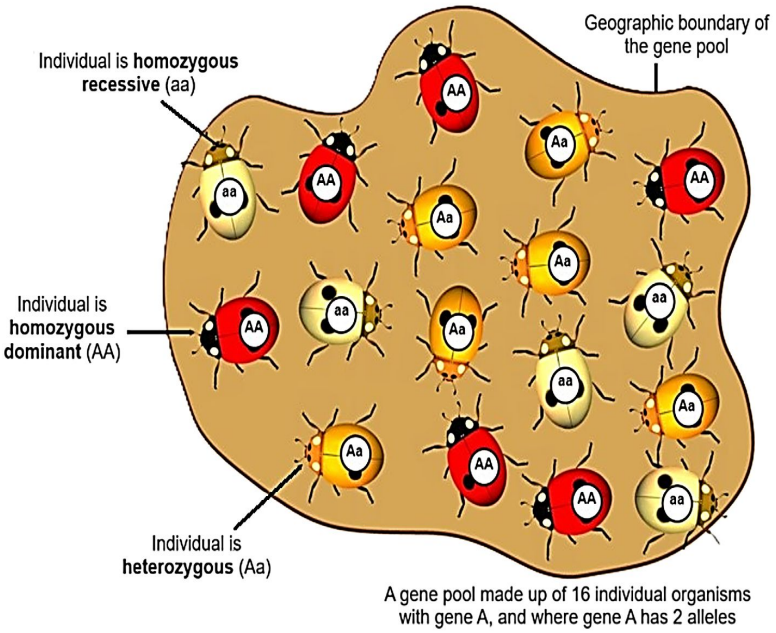


Figure 2

Calculations Involve

Allele frequency

- *Dominant allele frequency*
- *Recessive allele frequency*

Genotype frequency

- *Homozygous dominant genotype frequency*
- *Homozygous recessive genotype frequency*
- *Heterozygous genotype frequency*

Phenotype frequency

- *Dominant phenotype frequency*
- *Recessive phenotype frequency = homozygous recessive*

ATTENTION

**Tips

- Please follow decimal places stated in the question for your calculation and answer.
- If the question doesn't state decimal places, then follow this guide:
  - 1-99 → 1 decimal place
  - 100-999 → 2 decimal places
  - 1000 and above → 3 decimal places

Gene Pool

Example:

- In a population of 80 rabbits, coat colour character is controlled by a gene.
- The 2 alleles involved are *B* & *b*.

40 black rabbits with the genotype *BB*
30 black rabbits with the genotype *Bb*
10 white rabbits with the genotype *bb*

- **Calculate the gene pool**

Total number of alleles = 2 X total number of individuals
= 2 X 80
= 160 alleles

Allele Frequency

The proportion of a specific allele relative to all alleles of one gene in a population.

Example :

Allele frequency of purple flower, R *or* allele frequency of white flower, r

Formula:

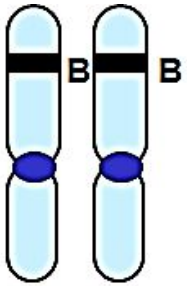
$$\text{Frequency allele of purple flower, } (R) = \frac{\text{total number of allele } R}{\text{total number of alleles}}$$

$$\text{Frequency allele of white flower, } (r) = \frac{\text{total number of allele } r}{\text{total number of alleles}}$$

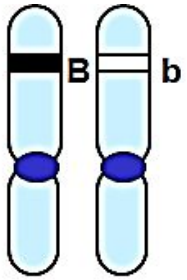
Example 1 : Allele Frequency

In a population of 80 rabbits, there are :

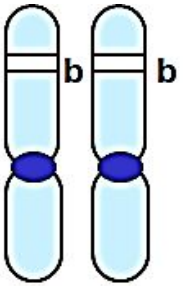
40 black rabbits (genotype *BB*)	• has 80 allele *B*
30 black rabbits (genotype *Bb*)	• has 30 allele *B* and 30 allele *b*
10 white rabbits (genotype *bb*)	• has 20 allele *b*



Each homozygous black rabbit has :
2 allele B



Each heterozygous black rabbit has :
1 allele B
1 allele b



Each homozygous white rabbit has :
2 allele b

Example 1 : Allele Frequency

In a population of 80 rabbits, there are :

40 black rabbits (genotype *BB*)	• has 80 allele *B*
30 black rabbits (genotype *Bb*)	• has 30 allele *B* and 30 allele *b*
10 white rabbits (genotype *bb*)	• has 20 allele *b*

- Total number of alleles = 2 X 80
  = 160 Alleles

Frequency of allele for black fur (B),

$$= \frac{\text{total number of black fur allele}}{\text{total number of alleles}}$$

$$= \frac{(40 \times 2) + 30}{160} = \frac{110}{160}$$

$$= 0.7$$

Example 1 : Allele Frequency

In a population of 80 rabbits, there are :

40 black rabbits (genotype *BB*)	• has 80 allele *B*
30 black rabbits (genotype *Bb*)	• has 30 allele *B* and 30 allele *b*
10 white rabbits (genotype *bb*)	• has 20 allele *b*

- Total number of alleles = 2 X 80
  = 160 alleles

- Frequency of allele for white fur (b) $= \dfrac{\text{total number of white fur allele}}{\text{total number of alleles}}$

$= \dfrac{(10 \times 2) + 30}{160} = \dfrac{50}{160}$

$= 0.3$

Example 2 : Allele Frequency

In a population of 500 wildflowers, allele A is dominant allele for red flower and allele a is recessive allele for white flower. There are 320 homozygous dominant plant, 160 heterozygous and 20 white flowers. Calculate:-

a) Frequency of dominant allele (A)

$$= \frac{(320 \times 2) + 160}{500 \times 2} = \frac{800}{1000}$$

$$= 0.80$$

b) Frequency of recessive allele (a)

$$= \frac{(20 \times 2) + 160}{500 \times 2} = \frac{200}{1000}$$

$$= 0.20$$

Genotype Frequency

The proportion of a particular genotype relative to all genotypes of the gene in a population.

Example:

- Frequency of homozygous dominant genotype
- Frequency of homozygous recessive genotype
- Frequency of heterozygous genotype

Genotype Frequency

Formula:

$$\text{Frequency of homozygous dominant genotype} = \frac{\text{Total number of homozygous dominant genotype}}{\text{Total number of individuals}}$$

$$\text{Frequency of homozygous recessive genotype} = \frac{\text{Total number of homozygous recessive genotype}}{\text{Total number of individuals}}$$

$$\text{Frequency of heterozygous genotype} = \frac{\text{Total number of heterozygous genotype}}{\text{Total number of individuals}}$$

This formula is applied when:

- The number of organisms for all three genotypes are given.
- The population does not meet the Hardy-Weinberg condition

Example 1 : Genotype Frequency

In a population of 80 rabbits, there are :

40 black rabbits (genotype *BB*)
30 black rabbits (genotype *Bb*)
10 white rabbits (genotype *bb*)

- **Frequency of homozygous dominant genotype (BB),**

$$= \frac{\text{number of homozygous dominant individuals}}{\text{total number of individuals}} = \frac{40}{80} = 0.5$$

- **Frequency of heterozygous genotype (Bb),**

$$= \frac{\text{number of heterozygous individuals}}{\text{total number of individuals}} = \frac{30}{80} = 0.4$$

- **Frequency of homozygous recessive genotype (bb),**

$$= \frac{\text{number of homozygous recessive individuals}}{\text{total number of individuals}} = \frac{10}{80} = 0.1$$

Example 2 : Genotype Frequency

- In a population of 500 wildflowers, allele A is dominant allele for red flower and allele a is recessive allele for white flower. There are 320 homozygous dominant plant, 160 heterozygous and 20 white flowers. Calculate:-

a) Frequency of homozygous dominant genotype (AA) $= \frac{320}{500} = 0.64$

b) Frequency of heterozygous genotype (Aa) $= \frac{160}{500} = 0.32$

c) Frequency of homozygous recessive genotype (aa) $= \frac{20}{500} = 0.04$

Phenotype Frequency

- The proportion of a particular phenotype relative to all phenotypes in the population.
- E.g: Dominant phenotype frequency (sum of homozygous dominant & heterozygous individuals)
- Formula:

Dominant phenotype frequency:

$$= \frac{\text{total number of dominant phenotype}}{\text{total number of individuals}}$$

$$= \frac{\text{total number of (homozygous dominant + heterozygous)}}{\text{total number of individuals}}$$

Example 1 : Phenotype Frequency

- The proportion of a particular phenotype relative to total number of individuals in the population

a) Genotype *BB* = 40 rabbits (Black)

b) Genotype *Bb* = 30 rabbits (Black)

c) Genotype *bb* = 10 rabbits (White)

E.g:

- Phenotype frequency of black rabbit = (40 + 30) / 80 = 0.9
- Phenotype frequency of white rabbit

= 10 / 80 = 0.1

Example 2 : Phenotype Frequency

- In a population of 500 wildflowers, allele A is dominant allele for red flower and allele a is recessive allele for white flower. There are 320 homozygous dominant plant, 160 heterozygous and 20 white flowers.

Calculate:-

a) Dominant phenotype frequency $= \frac{320 + 160}{500} = 0.96$

b) Recessive genotype frequency $= \frac{20}{500} = 0.04$

LEARNING OUTCOMES

5.2 Hardy-Weinberg Law

a) State the Hardy-Weinberg Law (C1- LECTURE)

b) Explain the five assumptions of Hardy-Weinberg Law for genetic equilibrium (C3-TUTORIAL)

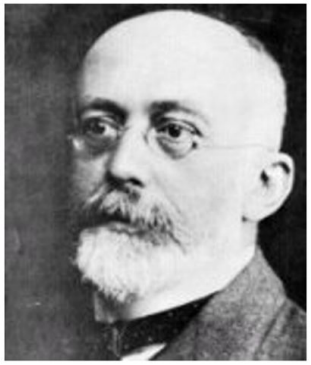
   i. Large population size
   ii. Random mating
   iii. No mutation
   iv. No migration and,
   v. No natural selection

c) Calculate allele and genotype frequencies (C3-TUTORIAL)

HARDY-WEINBERG LAW



Godfrey Hardy
(1877-1947)



Wilhelm Weinberg
(1862-1937)

Figure 3

- in 1908, G. H. Hardy (an English mathemathician) and W. Weinberg (a German physician) independently identified a mathematical relationship between alleles and genotypes in populations.

- This relationship has been called: The Hardy-Weinberg Equilibrium (The Hardy-Weinberg Law) and it concerns allele frequency

HARDY-WEINBERG LAW

In a population that exists in genetic equilibrium, the alleles and genotypes frequencies will remain constant from one generation to next generation under certain conditions are met.

HARDY-WEINBERG LAW

Assumptions of Hardy-Weinberg Law

A population is said to be in genetic equilibrium if these five conditions are met.

5 conditions for Hardy-Weinberg Equilibrium.

i. **Large population size**
ii. **Random mating**
iii. **No mutation**
iv. **No migration**
v. **No natural selection**

HARDY-WEINBERG LAW

i. Large population size

- A large population produces a large sample of successful gametes.
- So that chance in fluctuation in the gene that can cause phenotype frequencies to change over time can be avoided (genetic drift can be avoided).

HARDY-WEINBERG LAW

ii. Random mating

- Each individual in a population has an equal chance of mating with any individual of the opposite sex.
- Mate at random & does not select their mate.
- If individuals preferentially **choose** mates with certain genotypes, including close relatives (inbreeding), random mixing of gametes does not occur; causing changes in genotype frequencies.

HARDY-WEINBERG LAW

iii. No mutation

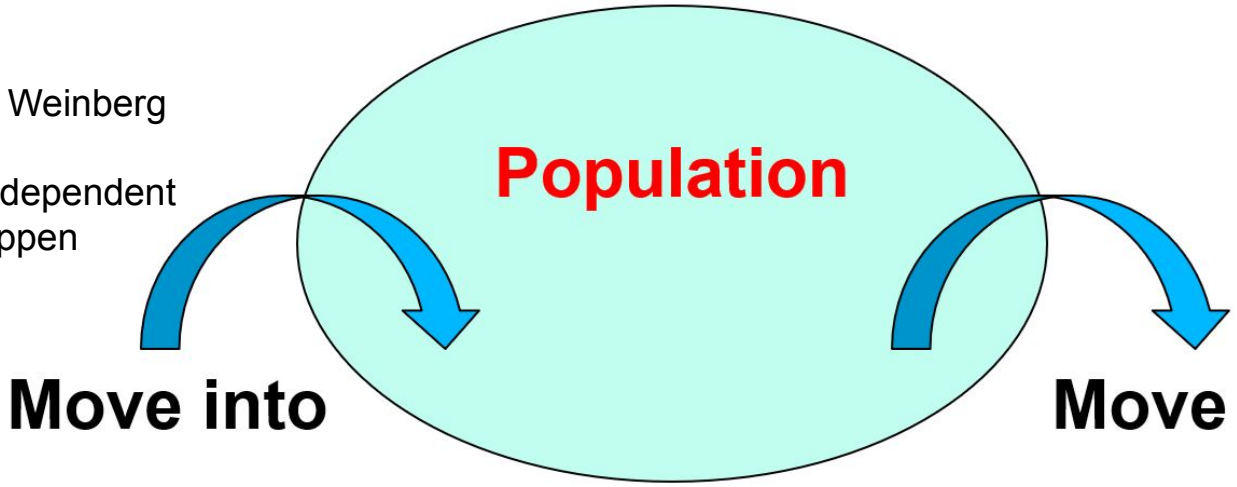
- There must be no net mutations.
- Allele and genotype frequencies may change through the loss or addition of alleles through mutation.
- The gene pool is modified if mutations occur or if entire genes are deleted or duplicated.

HARDY-WEINBERG LAW

iv. No migration

- There can be no migration between individuals into or out of the population.
- So, no exchange of alleles with other populations that might have different allele frequencies.

Tips from LKC:
If its not in Hardy Weinberg Equilibrium
Reason must be dependent
e.g. migration happen



HARDY-WEINBERG LAW

v. No Natural Selection

- No individuals will have a reproductive advantage over another individual.
- All individuals have the same potential to reproduce sexually.
- If natural selection occurs, certain phenotypes are favored over the others
- Certain genotypes has greater fitness / more advantage to survive; this will cause changes in allele frequencies

HARDY-WEINBERG EQUATION

1) Allele frequencies $p + q = 1$

p = frequency of dominant allele

q = frequency of recessive allele

1) Genotype frequencies $p^2 + 2pq + q^2 = 1$

p^2 = frequency of homozygous dominant genotype

2pq = frequency of heterozygous genotype

q^2 = frequency of homozygous recessive genotype

SUMMARY

- These frequencies can be calculated by using these formulae :

Allele frequency	$p + q = 1$
Genotype frequency	$p^2 + 2pq + q^2 = 1$
Dominant phenotype frequency	$p^2 + 2pq$
Recessive phenotype frequency	q^2

Tips from LKC:
Must write the Hardy-Weinberg equation completely

HARDY-WEINBERG EQUATION

**Tips

Determine the type of information. They may be in the form of:

- ✔ number of individuals
- ✔ percentage
- ✔ ratio
- ✔ probability
- ✔ fractional
- ✔ total number of allele

Tips from LKC:
The number of individuals must be rounded according to the rounding concept in mathematics.
(e.g.: 18.49 - 18 individuals;
18.51 - 19 individuals)

Tips from LKC:
Probability must be in simplest form of a fraction. (e.g.: ½, ¼, 1)

HARDY-WEINBERG EQUATION

****Tips**

Identify any information of recessive traits, we always **begin** calculations by **calculating the frequency of homozygous recessive genotype (q^2)**.

NEVER START calculation with information of dominant trait.

If **allele frequency is required**, answer should be either in the form of:

- p (dominant)
- q (recessive)

HARDY-WEINBERG EQUATION

**Tips

If **genotype frequency is required**, answer should be either in the form of:

- p^2
- q^2
- 2pq

If **number of heterozygous individual or the carrier is required**, answer should be:

- 2pq x number of individual

Rounding number rules applied:

•If the next digit is less than 5, keep the rounding digit the same.

•If the next digit is 5 or more, increase the rounding digit by 1.

HARDY-WEINBERG EQUATION

**Tips

If **number of individual with dominant trait** is required, answer should be:

- $(p^2 + 2pq)$ x number of individual

If **heterozygous or carrier frequencies is required**, answer should be :

- 2pq

If **percentage of heterozygous or carrier is required**, answer should be :

- 2pq x 100

HARDY-WEINBERG EQUATION

**Tips

- Must label p, q, p^2, 2pq, and q^2
- Please follow decimal places stated in the question for your calculation and answer.
- If the question doesn't state decimal places, then follow this guide:
  - Tens – 1 decimal place
  - Hundreds – 2 decimal places
  - Thousands and above 3 decimal places

HARDY-WEINBERG EQUATION

**Tips

- Must label p, q, p^2, 2pq, and q^2
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  - Thousands and above 3 decimal places

EXAMPLE 1

In a population of 300 plants, the total number of recessive allele is 60. Calculate the recessive and dominant allele frequency.

- Frequency of recessive allele, $q = \frac{\text{total number of recessive allele}}{\text{total number of alleles}}$
- Frequency of recessive allele, $q = \frac{60}{600} = 0.10$

$p + q = 1$

- Frequency of dominant allele, $p = 1 - 0.1 = 0.90$

EXAMPLE 2

A wildflower population with 2 varieties contrasting in flower colour. An allele for pink flowers (*A*) are completely dominant to an allele for white flowers, (*a*). Frequency of recessive allele, *a* is 20%. Find the frequency of dominant allele, *A*.

- Frequency of recessive allele, q = 20% = 0.20
- $p + q = 1$;
- Frequency of dominant allele, p = 0.80

EXAMPLE 3

- In a gene pool, there is 60% allele H. Calculate:-

  - Frequency of dominant allele, p $= \frac{60}{100}$

    $= 0.60$

  - Frequency of recessive allele, q $= 1 - 0.6$

    $= 0.40$

EXAMPLE 3

- In a gene pool, there is 60% allele H. Calculate:-
  - The percentage of heterozygote in that population?

  Frequency of dominant allele, p = 0.60
  $\Rightarrow p + q = 1$

  Frequency of recessive allele, q = 1 – 0.6 = 0.40

  Frequency of heterozygous genotype, 2pq = 2(0.6) (0.4)
  = 0.48

  Percentage of heterozygous genotype = 0.48 X 100
  = 48 %

Example 4

- In a population, 84% have the ability to roll their tongue. Ability to roll the tongue is determined by a dominant allele, *L*. The inability to roll the tongue is due to recessive allele, *l*.

  Find the allele frequency for *L* and *l*.

  Find the genotype frequency for homozygous dominant, heterozygous & homozygous recessive.

  - The percentage of individuals that cannot roll their tongue :
    = 100 % - 84 %
    = 16 %
  - If there are 100 person, 16 individuals cannot roll their tongue
    $= \frac{16}{100} = 0.16$

Example 4

- In a population, 84% have the ability to roll their tongue. Ability to roll the tongue is determined by a dominant allele, *L*. The inability to roll the tongue is due to recessive allele, *l*.

- Frequency of homozygous recessive genotype, $q^2 = \frac{16}{100} = 0.16$
  - Frequency of recessive allele, q $= \sqrt{0.16} = 0.40$

    $p + q = 1$
  - Frequency of dominant allele, p $= 1 - 0.4 = 0.60$

- Frequency of homozygous dominant genotype, $p^2 = (0.6)^2 = 0.36$
- Frequency of heterozygous genotype, 2pq $= 2\ (0.6)\ (0.4) = 0.48$

Example 5

In Kuala Lumpur, one out of 10,000 babies were born with albino disease. What is the frequency of albino and normal individual in KL? How many people are actually a carrier to the albino recessive allele? Show your calculation and answer in **four decimal places**.

- Frequency of homozygous recessive genotype, $q^2 = \frac{1}{10,000} = 0.0001$
- Frequency of recessive allele, q. $= \sqrt{0.0001} = 0.0100 \text{ // } 0.01$

p+q=1,

- Frequency of dominant allele, $p = 1 - q = 1 - 0.0100$
  $= 0.9900 \text{ // } 0.99$
- Frequency of normal individual, $p^2 + 2pq = (0.9900)^2 + 0.0198$
  $= 0.9801 + 0.0198$
  $= 0.9999$

Example 5

In Kuala Lumpur, one out of 10,000 babies were born with albino disease. What is the frequency of albino and normal individual in KL? How many people are actually a carrier to the albino recessive allele? Show your calculation and answer in **four decimal places**.

- Frequency of heterozygous genotype, 2pq = 2(0.9900) (0.0100)
  = 0.0198

- Number of carrier individuals = 0.0198 X 10,000 = 198

EXAMPLE 6

The allele y occurs with a frequency of 0.8 in a population of clams. Give the frequencies of the genotypes YY, Yy and yy. Show your calculation.

Frequency of recessive allele, q = 0.8

p + q = 1,

Frequency of dominant allele, p = 1 - q
= 1 – 0.8
= 0.2

EXAMPLE 6

Now, use the other equation to get the genotype frequencies

Frequency of homozygous dominant genotype, $p^2 = (0.2)^2$
$= 0.04$

Frequency heterozygous genotype, $2pq = 2(0.2)(0.8)$
$= 0.32$

Frequency of homozygous recessive genotype, $q^2 = (0.8)^2$
$= 0.64$

QUESTION 7

In a population of 500 wildflowers, 20 are white flowers or having recessive phenotype and the rest have dominant phenotype.

What are the genotype frequency of *Aa* individuals, frequency of dominant phenotype and frequency of recessive phenotype individuals ?

QUESTION 7

Solution

Frequency of homozygous recessive genotype, $q^2 = 20/500$

$=$

Frequency of allele, $q = 0.20$

$p + q = 1$

Frequency of dominant allele, $p = 1 - q$

$= 1 - 0.20$

$= 0.80$

QUESTION 7

Solution

Frequency of homozygous dominant genotype, $p^2 = (0.80)^2$
$= 0.64$

Frequency of heterozygous genotype, $2pq = 2\ (0.80)(0.20)$
$= 0.32$

Frequency of dominant phenotype $= p^2 + 2pq$
$= 0.64 + 0.32$
$= 0.96$

QUESTION 8

Albinism in mice is a recessive allele due to a mutant gene. In a population of mice, 16% are albino. Assume that the population in the Hardy-Weinberg equilibrium, what is the frequency of carrier?

Frequency of homozygous recessive genotype, $q^2 = 0.16$

$= \sqrt{0.16}$

Frequency of recessive allele, $q = 0.40$

$p + q = 1$

Frequency of dominant allele, $p = 1 - p$

$= 1 - 0.40$

$= 0.60$

So, frequency of the carrier, $2pq$

$= 2\ (0.60)(0.40)$

$= 0.48$

QUESTION 9

In a population of 400 people who dwell on a Pacific island, there are 16 people with homozygous recessive trait. What is the number of heterozygous individual?

Frequency of homozygous recessive genotype, $q^2 = 16/400$

$= \sqrt{0.04}$

Frequency of recessive allele, $q = 0.20$

$p + q = 1$

Frequency of dominant allele, $p = 1 - q$

$= 0.80$

Number of heterozygous people in the population

EXTRA QUESTION 1

Thalassemia is an inherited autosomal recessive blood disease in human. Thalassemia major is a severe form of anaemia due to homozygous recessive condition, while thalassemia minor is a mild form of anaemia shown in individuals with heterozygous genotype. In a population of 12 750, two individuals are suffering from thalassemia major.

a) Determine the frequencies of the dominant and recessive alleles in the population. (*Calculate up to five decimal places*).

b) How many individuals would be suffering from thalassemia minor in the population?

c) If 1000 normal individuals migrated out of the population, what would be the new frequencies of the dominant and recessive alleles?

EXTRA QUESTION 1

a) Determine the frequencies of the dominant and recessive alleles in the population. (*Calculate up to five decimal places*).

Frequency of homozygous recessive genotype, $q^2 = 2/12750$

$= \sqrt{0.00016}$

Frequency of recessive allele, $q = 0.01265$

$p + q = 1$

Frequency of dominant allele, $p = 1 - q$

$= 1 - 0.01265$

$= 0.98735$

EXTRA QUESTION 1

b) How many individuals would be suffering from thalassemia minor in the population?

Frequency of individuals with thalassemia minor, 2pq

= 2 (0.98735)(0.01265)

= 0.02498

Number of individuals with thalassemia minor;

= 0.02498 X 12 750

= 318 individuals

EXTRA QUESTION 1

c) If 1000 normal individuals migrated out of the population, what would be the new frequencies of the dominant and recessive alleles?

New population =12 750 – 1000 = 11 750

$$\text{Frequency of recessive allele} = \frac{\text{Total recessive allele}}{\text{Total of all alleles}}$$

$$= \frac{(2\text{x}2) + 318}{11\,750 \text{ x } 2}$$

$$= \frac{322}{23\,500}$$

$$= 0.014$$

EXTRA QUESTION 1

c) If 1000 normal individuals migrated out of the population, what would be the new frequencies of the dominant and recessive alleles?

New population =12 750 – 1000 = 11 750

$$\text{Frequency of dominant allele} = \frac{\text{Total dominant allele}}{\text{Total of all alleles}}$$

$$= \frac{(11\,430 \times 2) + 318}{11\,750 \times 2}$$

$$= \frac{23\,178}{23\,500}$$

$$= 0.986$$

EXTRA QUESTION 2

In a population, 1101 samples were MM, 1496 were MN and 503 were NN. (*Calculate up to four decimal places*).

a) Calculate the allele frequencies of M & N

b) Calculate the genotype frequencies of MM, MN and NN

a) Calculate the allele frequencies of M and N

$$\text{Frequency of allele M} = \frac{(1101 \times 2) + 1496}{3100 \times 2} = \frac{3698}{6200} = 0.5965$$

$$\text{Frequency of allele N} = \frac{(503 \times 2) + 1496}{3100 \times 2} = \frac{2502}{6200} = 0.4035$$

EXTRA QUESTION 2

b) Calculate the genotype frequencies of MM, MN and NN

Frequency of genotype MM = 1101/3100
= 0.3552

Frequency of genotype MN = 1496/3100
= 0.4826

Frequency of genotype NN = 503/3100
= 0.1623

EXTRA QUESTION 3

In a population of 1800 birds, a sampling shows that 1200 birds have the dominant phenotype.

a) What is the genotype frequency for heterozygous individuals.

b) Individuals of the population randomly mate over generations. If in a new generation the number of this population increases to 2300, how many would be expected to have homozygous dominant genotype?

EXTRA QUESTION 3

a) What is the genotype frequency for heterozygous individuals.

Number of recessive phenotype = 1800 -1200 = 600

Frequency of homozygous recessive genotype, q^2 = 600 / 1800

= $\sqrt{0.333}$

Frequency of recessive allele, q = 0.577

p + q = 1

Frequency of dominant allele, p = 1 - q

= 1 - 0.577

= 0.423

Frequency of heterozygous genotype, 2pq = 2(0.577)(0.423)

= 0.488

EXTRA QUESTION 3

b) Individuals of the population randomly mate over generations. If in a new generation the number of this population increases to 2300, how many would be expected to have homozygous dominant genotype?

Frequency of homozygous dominant genotype, $p^2 = (0.423)^2$
$= 0.179$

Number of birds with homozygous dominant genotype
$= 0.179 \times 2300$
$= 411.7$
$= 412$ birds

REFERENCES

- Campbell N.A , Urry L.A,Cain M.L, Wasserman S.A, Minorsky P.V, Reece J.B Biology, 12th ed. (2021), Pearson Education, Inc.
- Solomon E.P & Martin C.E., Martin C.W., Berg, L.R, Biology, 11th ed. (2019) Thomson Learning, Inc.

** *Notes: All reference is referred to hardcopy books (including pages)*
A slight difference between paging in e-book and hardcopies

REFERENCES (FIGURE)

- Figure 1 - https://www.quia.com/jg/2543185list.html
- Figure 2 - https://www.gktoday.in/major-gene-pool-centres/
- Figure 3 - https://www.palomar.edu/anthro/synthetic/synth_2.htm
- Figure 4 - Adapted from *Campbell*,11th ed., page 488