## POPULATION GENETICS

GENETIC STRUCTURE OR CONSTITUTION OF POPULATIONS

# GENETIC CONSTITUTION OF POPULATIONS 

$>$ Genes come in diifferent variants (alleles). >Individuals can have one or two alleles for any gene:
Two different alleles = heterozygous Two of the same allele = homozygous
$\rightarrow$ Dominant alleles are expressed whether there is one copy or two.
$>$ Recessive alleles must be present in two copies to be expressed.

## GENETIC CONSTITUTION OF POPULATIONS

$>$ So if there are N individuals Total number of genes or alleles at locus $=\mathbf{2 N}$
$>$ Allele : altermative forms of a gene.
$>$ Two alleles at a locus $\longrightarrow$ simple case or common.
$>$ More than two alleles at locus

Example. ABO blood group system in human.

## GENETIC CONSTITUTION OF POPULATIONS

Allele for purple flowers


## Gene frequency or allelic frequency or gametic array

# Gene frequency or allelic frequency or gametic array 

Allele frequency is the proportion of that allele relative to all alleles at a locus in the population
$>$ Frequency of dominant allele A
$>$ Frequency of recessive allele a


$$
\begin{gathered}
>p+q=1 \\
>p=1-q \\
>q=1-p
\end{gathered}
$$

$>\mathrm{p}+\mathrm{q}+\mathrm{r}=1$ ( if there are three different alleles at a locus).

## Calculation of allele frequency

## Suppose that there are N individuals having the following genotypes

| Phenotype | Red | Roan | White | Total |
| :---: | :---: | :---: | :---: | :---: |
| Genotype | AA | Aa | aa |  |
| No. of individuals | K | L | M | N |
| No. of alleles A | $\mathbf{2 K}$ | L | - | $\mathbf{2 K}+\mathbf{L}$ |
| No. of alleles a | - | L | $\mathbf{2 M}$ | $\mathbf{2 M}+\mathbf{L}$ |
| Genotypic frequency | D | $\mathbf{H}$ | $\mathbf{R}$ | $\mathbf{1}$ |

$2 K+L+2 M+L=2(K+L+M)=2 N$

## Methods of calculation

## 1. From number of phenotypes

(No.of indivi. homozygous for that allele) $+\left(\frac{1}{2} \mathrm{~N}\right.$.of indivi. heterozygous for the same allele)
Freq. of a gene =
Total number of individals
$\therefore P(A)=\frac{K+\frac{1}{2} L}{N} \quad$ also $\quad q(a)=\frac{M+\frac{1}{2} L}{N}$

## Methods of calculation

## 2. From genotypic frequency

Freq. of a gene $=$ freq. of indivi. homozyous for that allele $+\frac{1}{2}$| $\begin{array}{l}\text { Freq. of individual } \\ \text { Heterozygous for } \\ \text { the same allele }\end{array}$ |
| :--- | :--- |

$$
P_{(A)}=D+\frac{1}{2} H
$$

$$
q_{(a)}=R+\frac{1}{2} H
$$

## Methods of calculation

## 3. From number of alleles

(2No.of indivi. homozygous for that allele) + (No.of indivi. heterozygous
for the same allele)
2 Total number of individals
$\therefore \mathrm{P}(\mathrm{A})=\frac{2 \mathrm{~K}+\mathrm{L}}{2 \mathrm{~N}}$
also

$$
q(a)=\frac{2 M+L}{2 N}
$$

## Genotypic frequency or zygotic array or genotypic array

## Genotypic frequency or zygotic array or genotypic array

Genotypic frequency: the relative frequency of a particular genotype in a population.

| phenotype | Red | Roan | White | Total |
| :--- | :---: | :---: | :---: | :---: |
| Genotype | AA | Aa | aa |  |
| No. of indivi. | K | L | M | N |
| Genotype <br> Frequency | D | H | R | $\mathbf{1}$ |

## Calculation of genotypic frequency

Genotypic frequencies $=\frac{\text { The No.of individuals of a particular genotype }}{\text { The total No.of individuals in the population }}$

$$
D=\frac{K}{N} \quad H=\frac{L}{N} \quad R=\frac{M}{N}
$$

## Example

$>$ In shorthorn cattle, 3 coat colors are Red, Roan and White. In a sample of 1000 shorthorn assume the number of animals with each colour is:

| phenotype | Red | Roan | White | Total |
| :--- | :---: | :---: | :---: | :---: |
| Genotype | $\mathbf{R R}$ | $\mathbf{R r}$ | $\mathbf{r r}$ |  |
| No. of indivi. | $\mathbf{3 6 0}$ | $\mathbf{4 8 0}$ | $\mathbf{1 6 0}$ | $\mathbf{1 0 0 0}$ |

a. What are the gene frequency.
b. What are the genotypic frequency.

## Solution

a. Gene frequency

$$
p_{(R)}=\frac{360+\frac{1}{2} 480}{1000}=0.6 \quad q_{r)}=\frac{160+\frac{1}{2} 480}{1000}=0.4
$$

## b. Genotype frequency

$$
\mathrm{D}_{\mathrm{RR})}=\frac{360}{1000}=0.36 \quad H_{R r)}=\frac{480}{1000}=0.48
$$

$$
R_{(r)}=\frac{160}{1000}=0.16
$$

## The Hardy-Weinberg Law

## Table 24.1 Possible Combinations of $A$ and a Gametes from Gametic Pools for a Population

| $\begin{aligned} & \mathscr{U} \\ & \text { U } \\ & \text { Hy } \end{aligned}$ | $A(p)$ | Male gametes |  |
| :---: | :---: | :---: | :---: |
|  |  | $A(p)$ | $a(q)$ |
|  |  | $\begin{aligned} & A A \\ & \left(p^{2}\right) \end{aligned}$ | $\begin{gathered} A a \\ (p q) \end{gathered}$ |
| 菏 | $a(q)$ | $\begin{gathered} A a \\ (p q) \end{gathered}$ | $\begin{gathered} a a \\ \left(q^{2}\right) \end{gathered}$ |

In sum, $p^{2} A A+2 p q A a+q^{2} a a=1.00$

## Test Hardy-Weinberg Law

## Two equations can be used:

## $1 \quad H=2 \sqrt{D . R}$

H: frequency of heterozygote.
D: frequency of homozygous dominant.
R: frequency of homozygous recessive.
2 $\mathbf{H}=\mathbf{2 p q}$
p : frequency of dominant allele.
q : frequency of recessive allele.

## Example

We sampled 2020 Guinea pigs that have one of three phenotypes. Long, Short, or Intermediate hair. The gene for hair length is governed by incomplete dominance, where Long hair is HH, Intermediate hair is Hh, and Short hair is hh. We find that 1322 Guinea pigs have long hair, 450 have Intermediate hair, and 248 have short hair.

1. Determine the genotypic frequencies
2. Determine the allelic frequencies
3. Decide if this population is in Hardy-Weinberg equilibrium or not?

## Test Hardy-Weinberg Law

| phenotype | Long | Intermediate | Short | Total |
| :--- | :---: | :---: | :---: | :---: |
| Genotype | $\mathbf{H H}$ | $\mathbf{H h}$ | hh |  |
| No. of indivi. | $\mathbf{1 3 2 2}$ | $\mathbf{4 5 0}$ | $\mathbf{2 4 8}$ | $\mathbf{2 0 2 0}$ |

$>$ Genotypic frequencies

$$
\mathrm{D}_{(H H)}=\frac{1322}{2020}=0.65 \quad H_{H r)}=\frac{450}{2020}=0.22
$$

$$
R_{(h h)}=\frac{248}{2020}=0.12
$$

## Test Hardy-Weinberg Law

$>$ Allelic frequencies

$$
p_{(H)}=0.65+\frac{1}{2} 0.22=0.76
$$

$$
\mathbf{q}_{(\mathrm{h})}=0.12+\frac{1}{2} 0.22=0.23
$$

> Test of H \& W law

$$
\mathbf{H}=2 \sqrt{D \cdot \mathbf{R}}
$$

$$
0.22=2 \sqrt{0.65 \times 0.12}
$$

$$
0.22 \neq 0.558
$$

Population not in H \& W law

$$
\begin{array}{ll}
\text { Or } & H=2 p q \\
& 0.22=2 \times 0.76 \times 0.23 \\
0.22 \neq 0.34
\end{array}
$$

## Example

One hundred person from a small town in Egypt were tested for $\mathbf{M m}$ blood types the genotypic data are: $\mathbf{M M}=$ $41, \mathbf{M N}=38$, and $\mathrm{NN}=21$. Is the population in HardyWeinberg proportions?

Solution:

$$
\text { A. } \frac{H}{\sqrt{D \cdot R}}=2
$$

| Blood Types | MM | MN | NN | Total |
| :--- | :---: | :---: | :---: | :---: |
| Numbers | 41 | 38 | 21 | 100 |
| Genotype frequency | D | H | R |  |
|  | $(0.41)$ | $(0.38)$ | $(0.21)$ |  |

## Example

$$
\therefore \frac{0.38}{\sqrt{(0.41)(0.21)}}=\frac{0.38}{0.29}=1.30
$$

The equation is not equal 2 , then the population is not in
Hardy-Weinberg equilibrium.

$$
\begin{aligned}
& \text { B. } \mathbf{H}=\mathbf{2 p q} \\
& P(M)=[(41 x 2)+38] / 200=.06 \\
& q(N)=[(21 x 2)+38] / 200=0.4 \\
& H=2 p q \\
& 0.38=2 x 0.6 \times 0.4 \\
& \therefore H \neq 2 p q
\end{aligned}
$$

Then, the population is not in Hardy-Weinberg equilibrium.

## Thanks for your attention

