LS1101
Introduction to Biology

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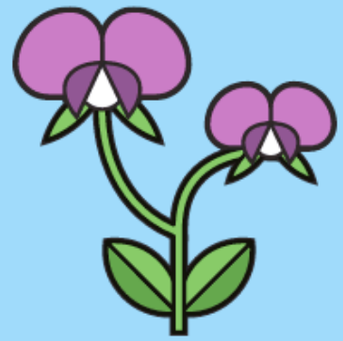
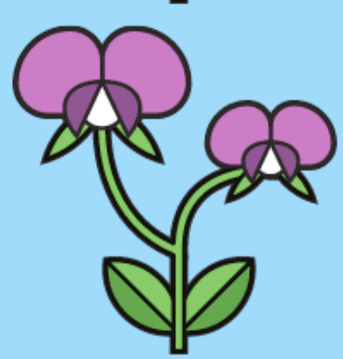
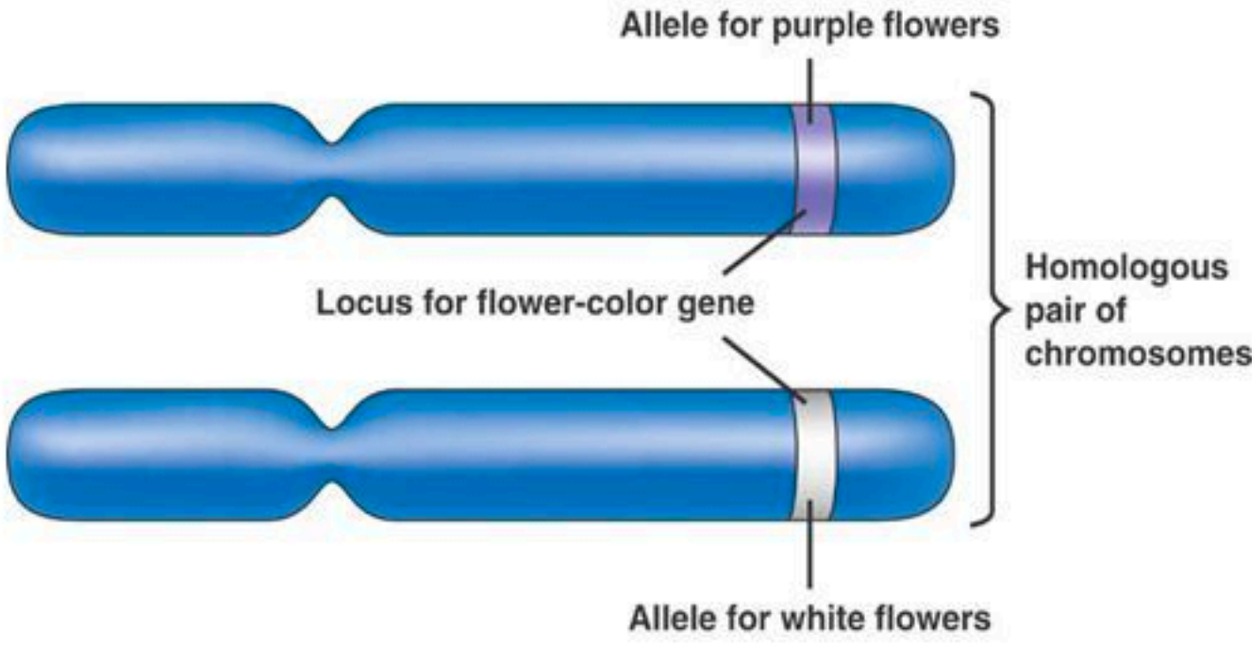
Genotype vs Phenotype

GENOTYPE	PHENOTYPE
The genotype is an organism's genetic information.	The phenotype is the set of observable physical traits.
BB homozygous dominant	purple 
Bb heterozygous	purple 
bb homozygous recessive	white 

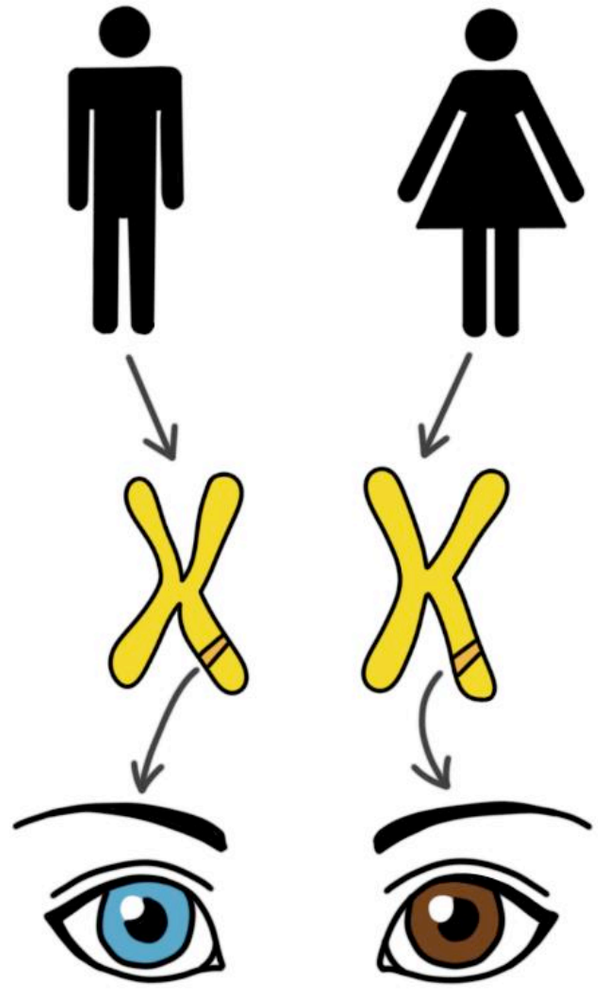
Genes and Alleles

- Gene: a heritable factor that controls a specific characteristic
  - Located in specific places on chromosomes
  - Humans have roughly 20,000-25,000 genes.
- Alleles: alternative forms of gene
- Example:
  - Gene: eye color
  - Alleles: blue vs. brown



humans get 2 copies of every gene, one copy from each parent

the 2 copies don't have to be identical



each variation of a gene is called an allele

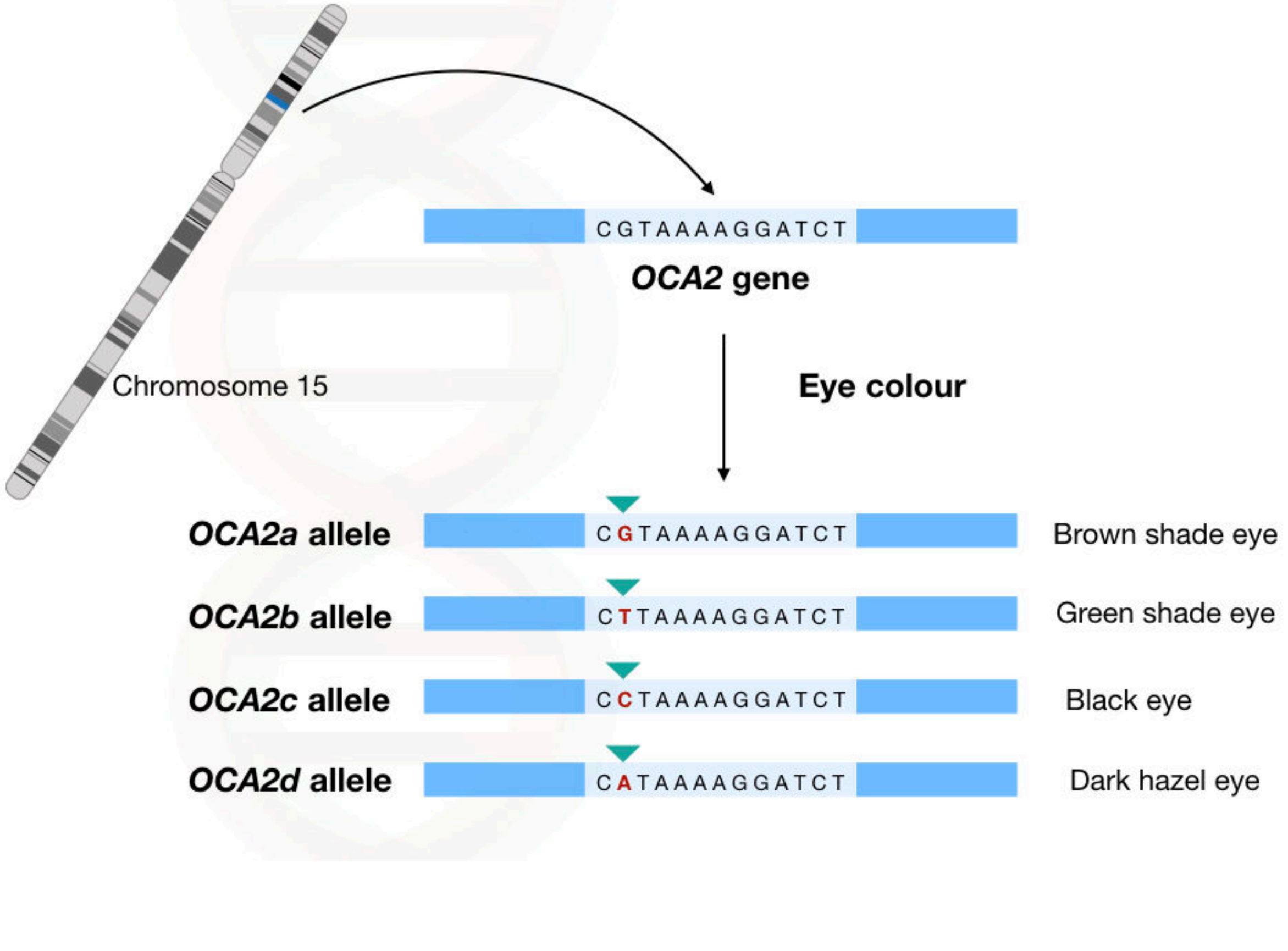
For example: if a gene contains information for eye color, one allele might code for blue eyes and another might code for brown eyes

alleles create diversity

An **allele** is one of two or more versions of a gene. An individual inherits two alleles for **each gene**, one from each parent. Each pair of alleles represent the genotype of that specific gene.

If the two alleles are the same, the individual is homozygous for that gene.

Alleles contribute to phenotype of the organism.

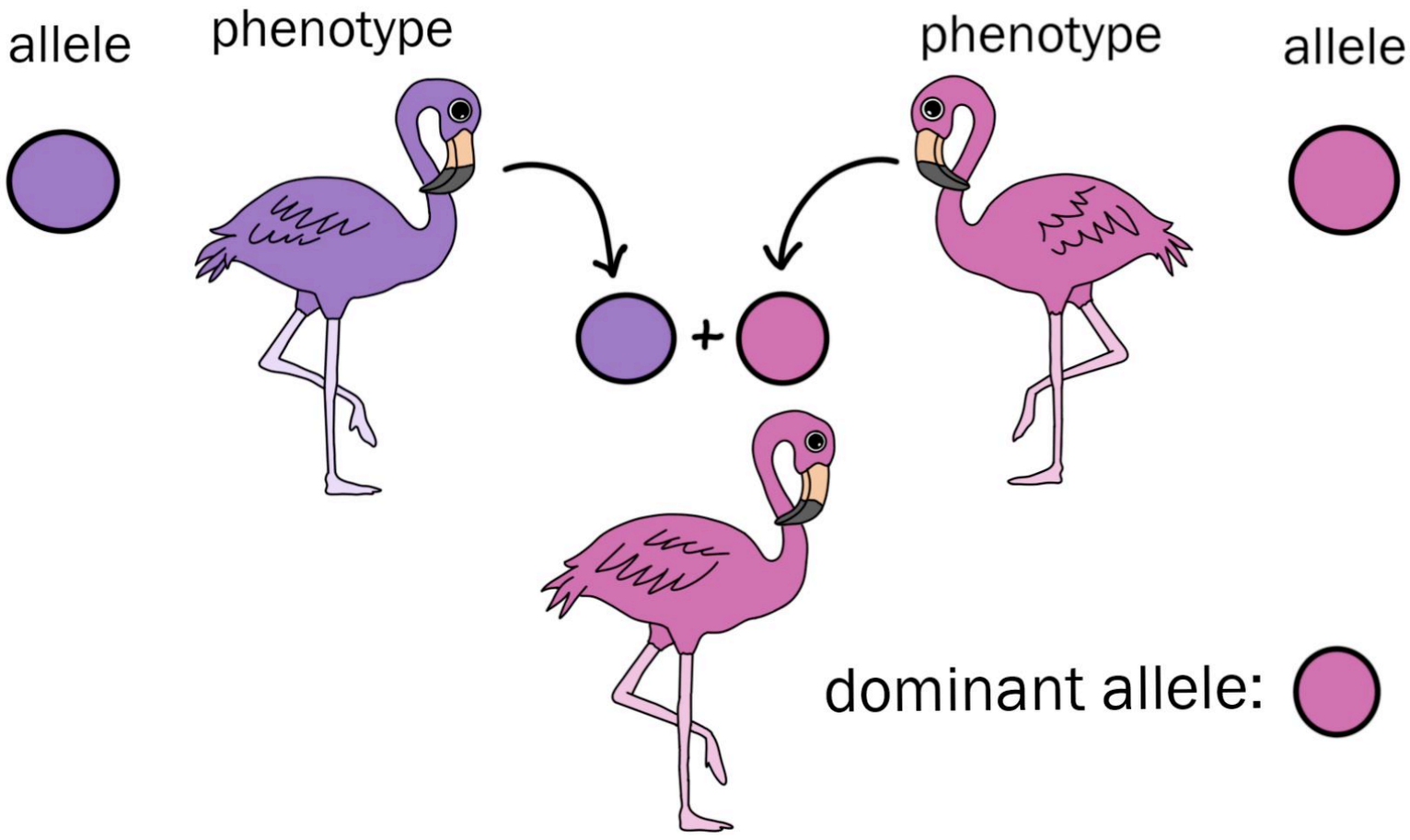

Chromosome 15
CGTAAAAGGATCT
OCA2 gene
Eye colour
OCA2a allele
CGTAAAAGGATCT
Brown shade eye
OCA2b allele
CTTAAAAGGATCT
Green shade eye
OCA2c allele
CCTAAAAGGATCT
Black eye
OCA2d allele
CATAAAAGGATCT
Dark hazel eye

Recessive

Refers to a trait that is expressed only when genotype is homozygous; a trait that tends to be masked by other inherited traits, yet persists in a population among heterozygous genotypes

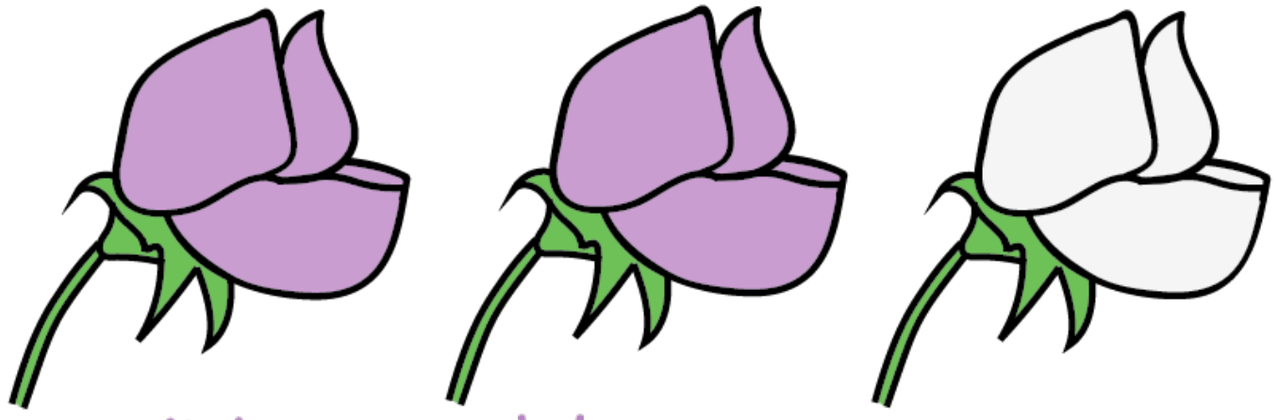
Dominant

An allele that produces the same phenotype whether its paired allele is identical or different. Refers to a trait that appears more frequently than another trait, resulting from interactions between gene alleles.



W - dominant purple allele

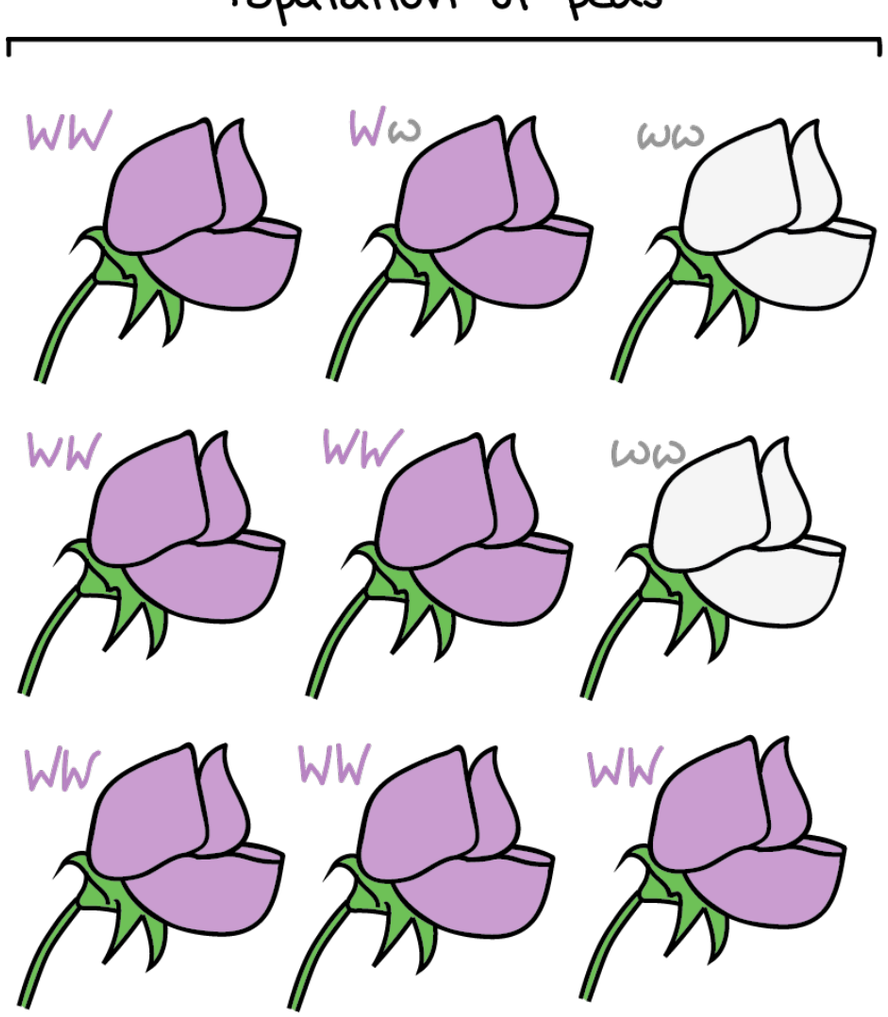
w - recessive white allele



Phenotype — flower color

Genotype — pair of alleles

Allele frequency **refers to how frequently a particular allele appears in a population.**

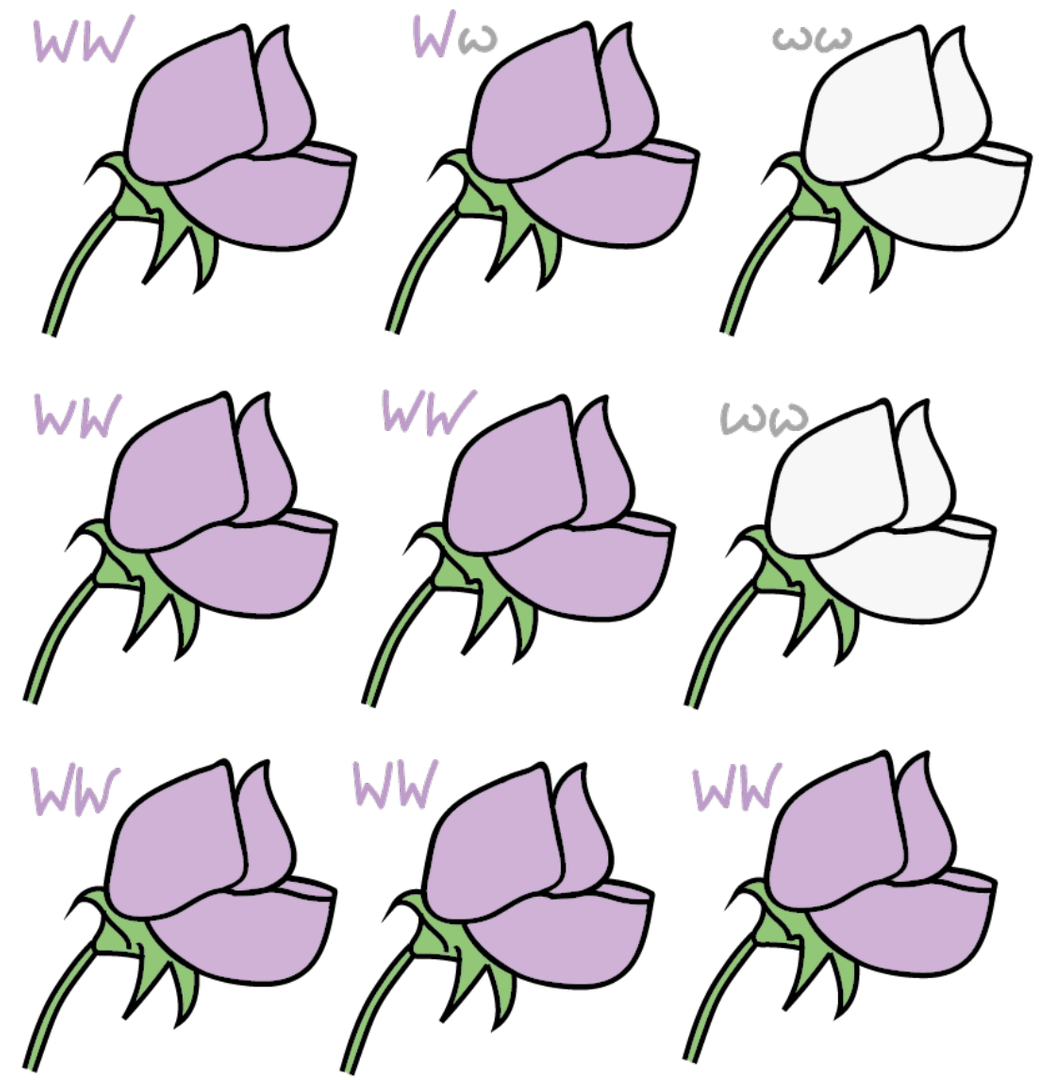


p = frequency of W = 13/18 = 0.72 or 72%

q = frequency of w = 5/18 = 0.28 or 28%

Allele Frequency

Population of peas



GENOTYPE FREQUENCY: → How often we see each allele combo

WW, Ww, or ww

Freq. of WW = 6/9 = 0.67

Freq. of Ww = 1/9 = 0.11

Freq. of ww = 2/9 = 0.22

PHENOTYPE FREQUENCY: → How often we see white vs. purple

Freq. of purple = 7/9 = 0.78

Freq. of white = 2/9 = 0.22

ALLELE FREQUENCY: → How often we see each allele

W or w

p = Freq. of W = 13/18 = 0.72

q = Freq. of w = 5/18 = 0.28

Number of copies of a particular allele in a population

Total number of all alleles for that gene in a population

Hardy-Weinberg Principle

$$p^2 + 2pq + q^2 = 1$$

p^2 = frequency of homozygous dominant genotype
q^2= frequency of homozygous recessive genotype

Problem:

A certain allele within moths causes them to be white while all others are brown. Only a moth with two recessive alleles will be white. When observe a population, suppose there are 84 brown and 16 white moths.

White (q^2): 16; Brown (p^2) : 84

q= 0.4

Practise problem 1

16% of a population cannot digest a certain food, FD. These individuals are recessive for this trait.

$q^2 = 0.16$

$q = 0.4; p = 0.6$

Hint!

Allele frequency: p; q
individuals, people: p^2, q^2

What percentage of individuals can digest FD?

Answer: 84% (100- 16)

What is the frequency of dominant and recessive allele?

Answer: p = 0.6 ; q = 0.4

What percentage of population are heterozygous for this trait?

Answer: 2pq = 0.48 or 48% (p = 0.6 ; q = 0.4)

Practise problem 2

The delta-32 mutation, a recessive gene, gives humans protection from HIV infection. The allele frequency in a town in Sweden is 20%

q = 0.2
p = 0.8

Hint!

Allele frequency: p; q
individuals, people: p^2, q^2

What percentage of the population have two copies of the gene and immune to HIV?

Answer: 4%

What percentage of the population are less susceptible to the disease because they are heterozygous?

Answer: 2 (0.2) (0.8) = 0.32 or 32%

Alleles encode for the phenotypic polymorphisms of a particular trait and may be beneficial, detrimental or neutral:

- Beneficial alleles will better equip the organism to survive and hence produce more offspring (encodes beneficial adaptations)
- Detrimental alleles will harm the survival prospects of an organism, leading to fewer viable offspring
- Neutral alleles will not affect the organisms survival prospects

Due to natural selection, the proportion of different alleles will change across generations (evolution)

- As beneficial alleles improve reproductive prospects (more offspring), they are more likely to be passed on to future generations
- Conversely, detrimental alleles result in fewer offspring and hence are less likely to be present in future generations

If environmental conditions change, what constitutes a beneficial or detrimental trait may change, and thus the allele frequencies in a population are constantly evolving

First generation

p (**A** allele frequency) = 0.5

q (**a** allele frequency) = 0.5

AA aa

AA aa

AA aa

AA aa

AA aa

Targeted by predators

Second generation

p (**A** allele frequency) = 0.7

q (**a** allele frequency) = 0.3

AA AA

AA Aa

AA Aa

AA aa

AA aa

Targeted by predators

Third generation

p (**A** allele frequency) = 0.9

q (**a** allele frequency) = 0.1

AA AA

AA AA

AA AA

AA Aa

AA Aa

Key:

A = light brown fur *(good camouflage)*

a = dark fur (black) *(poor camouflage)*

The Hardy-Weinberg formula allow scientists to determine whether evolution has occurred. Any changes in the gene frequencies in the population over time can be detected. The law essentially states that if no evolution is occurring, then an equilibrium of allele frequencies will remain in effect in each succeeding generation of sexually reproducing individuals. In order for equilibrium to remain in effect (i.e. that no evolution is occurring) then the following five conditions must be met:

1. No mutations must occur so that new alleles do not enter the population.

2. No gene flow can occur (i.e. no migration of individuals into, or out of, the population).

3. Random mating must occur (i.e. individuals must pair by chance)

4. The population must be large so that no genetic drift (random chance) can cause the allele frequencies to change.

5. No selection can occur so that certain alleles are not selected for, or against.

REVISION POINTS

- Evolution is a unifying concept in biology.
- Darwin defined evolution as "descent with modification," the idea that species change over time, give rise to new species, and share a common ancestor.
- Natural Selection
- Evidences for evolution
- Patterns of evolution
- Speciation concepts and patterns, reproductive isolation
- Inheritance concepts (Mendel genetics)
- Chromosome theory of inheritance, genes, alleles, Hardy-Weinberg principle.

1859 Darwin
Natural selection

1865 Mendel
Heredity factors are transmitted

1902 Sutton and Boveri
chromosome theory

1911 Morgan
Chromosomes carry genes

1927 Muller shows X-rays induce mutations

1941 Beadle and Tatum
Genes direct proteins

1944 Avery, MacLeod, McCarty
DNA transforms cells

1953 Watson and Crick
DNA structure

2003 Human Genome project

94 years after Darwin, the DNA structure was solved!

what makes a great SCIENTIST?

@by-addyspicer

CONFIDENCE
Not afraid to question the status quo

PERSERVERANCE
Allow themselves to fail, but always try again

COMPASSION
Support their peers and credit the work of others

CREATIVITY
creative problem solver, think outside the box

SCHOLAR
Never stop learning

CONSCIENTIOUS
consider the larger impact of their actions

Evolution

"Nothing in biology
makes sense except in
the light of evolution"

Theodosius Dobzhansky
(1900-1975)