Chapter 3
Genetics and Evolution

“Life is a DNA software system”

Craig Venter

Overview

To fully understand the science of ecology, one must first be able to grasp basic evolutionary concepts. Genetics underpins life itself, as genes are the building blocks of all organisms. Genes are passed down through generations, and variation in genetic material gradually accumulates, forming the basis for evolutionary change. Natural selection is just one of many factors influencing evolutionary change, but it is the most important in providing a link to the ecological context within which organisms exist and interact.

Contents

3.1

From molecules to meiosis

DNA and genes

To go right back to basics, all life on Earth is built from four different types of carbon-containing molecules:

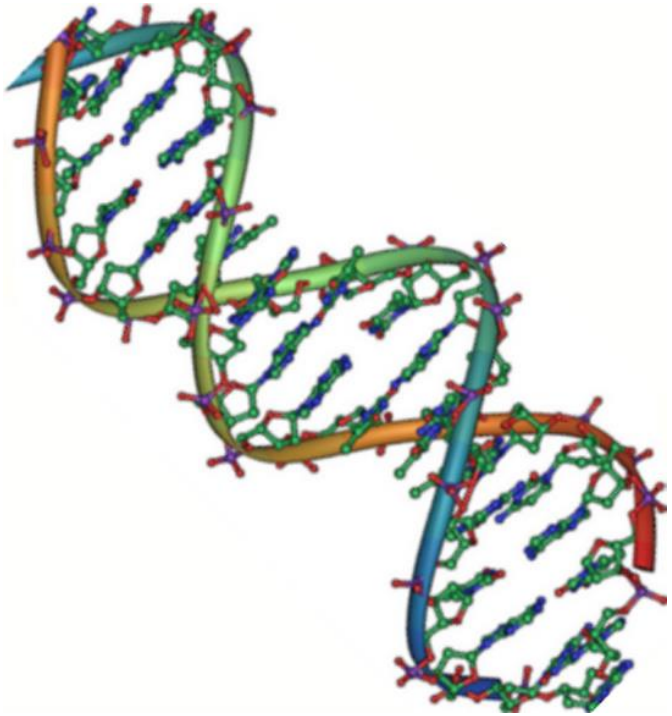
- Carbohydrates
- Lipids
- Proteins
- Nucleic acids

Nucleic acids are the essential biomolecules within all life-forms that contain the genetic code for organisms.

DNA (deoxyribose nucleic acid) and RNA (ribose nucleic acid) are the 2 closely related types needed to transmit genetic information from parent to offspring.

DNA and genes

DNA and RNA are composed of chains of many repeating units of smaller units called **nucleotide**s. Nucleotides are assembled into helical structures. DNA is double stranded (double-helix) and RNA is single-stranded.



DNA and genes

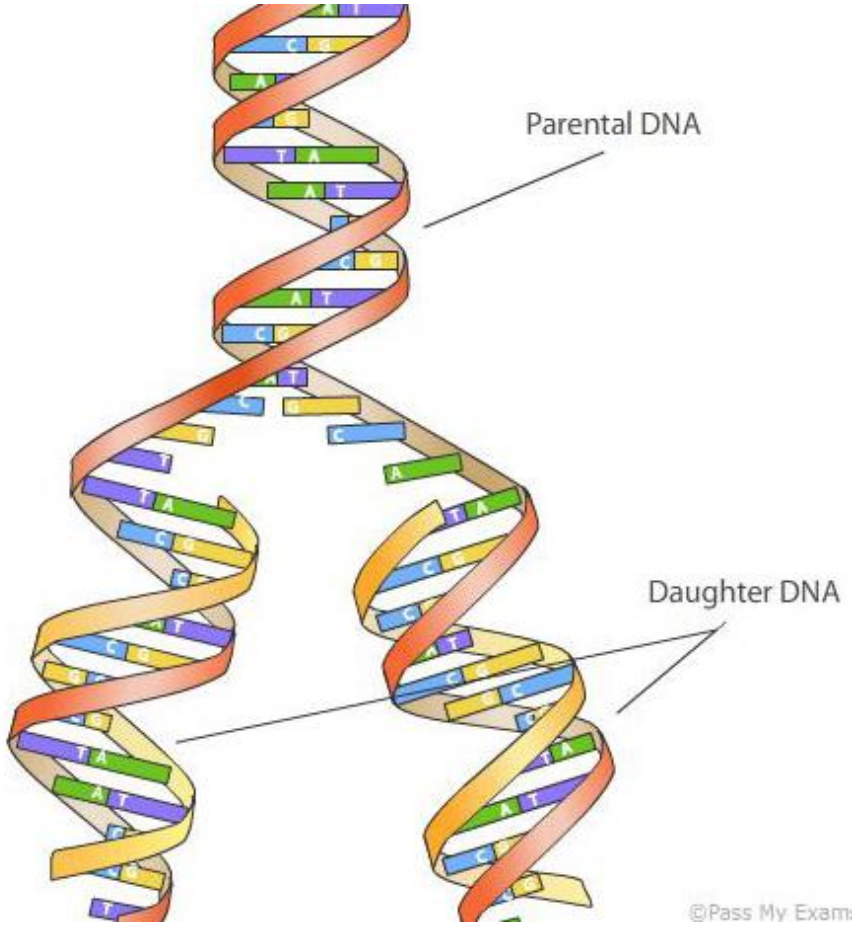
Nucleotides themselves contain nitrogen -containing molecules called **nucleotide bases**. There are 4 nucleotide bases in DNA: Adenine (A), Cytosine (C), Guanine (G) and Thymine (T). In RNA, Uracil replaces Thymine.

Bases on different strands of nucleotides bind together to form **base-pairs.** Base-pairing contributes to the helical structure of DNA which resembles a spiral staircase.

A always pairs with T, and G always pairs with C, so that the two bound strands are always complementary.

Each strand acts as a template to ensure an exact duplicate is passed on to its daughter cell when cells divide.

The linear sequence of nucleotide bases along a single strand is what constitutes the **genetic code** for all life on earth.

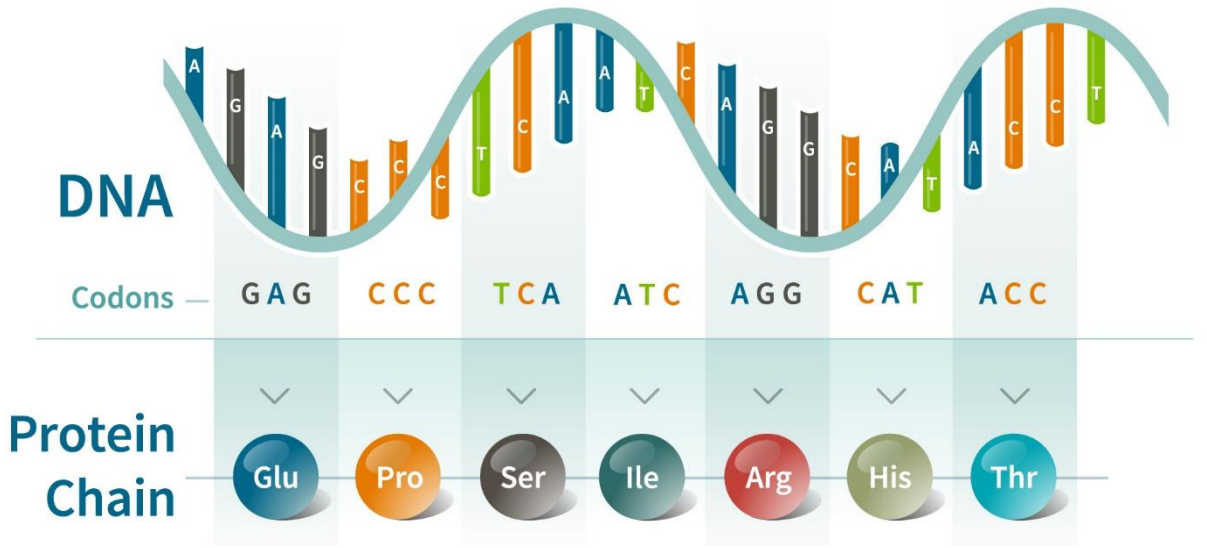


DNA and genes

The 4-letter 'alphabet' of ATCG in DNA forms 'words' of 3 letters, called **codons.** Codons code for **amino acids**, which are the building blocks of **proteins.**

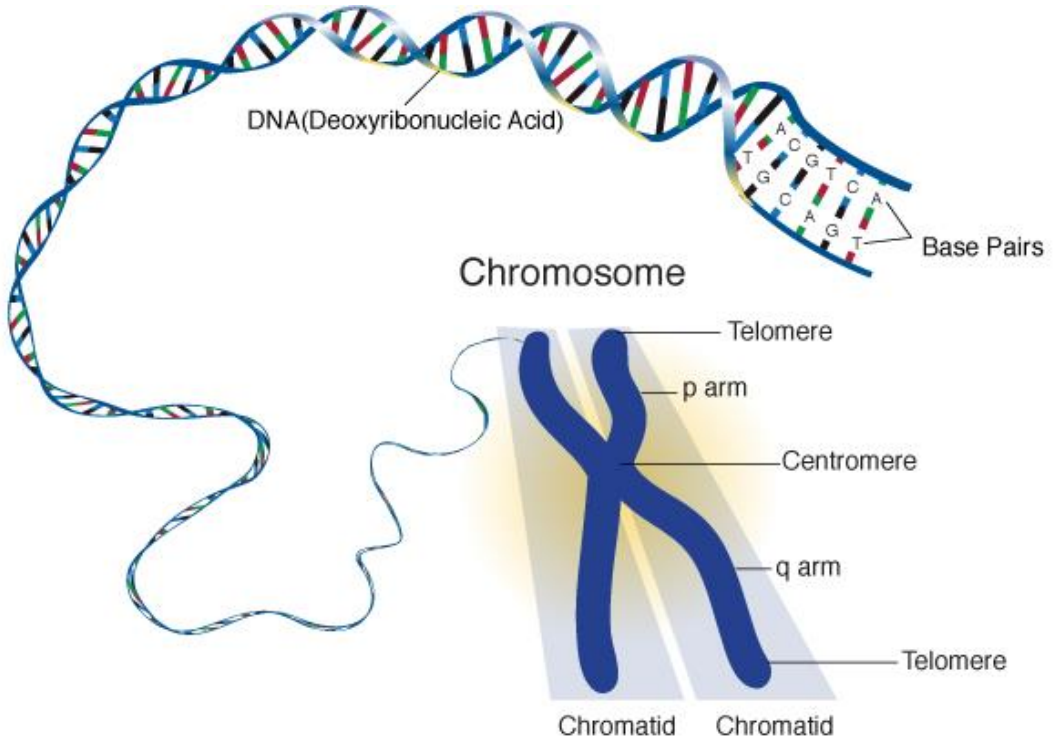
A gene is therefore a sequence of nucleotides along a DNA strand that specifies the sequence of amino acids that link together to form proteins.

The gene is the basic unit of heredity



Chromosomes and ploidy

In organisms with more than one cell (**eukaryotes**) the nucleus of each cell DNA is supercoiled and packaged into thread-like structures called **chromosomes.**



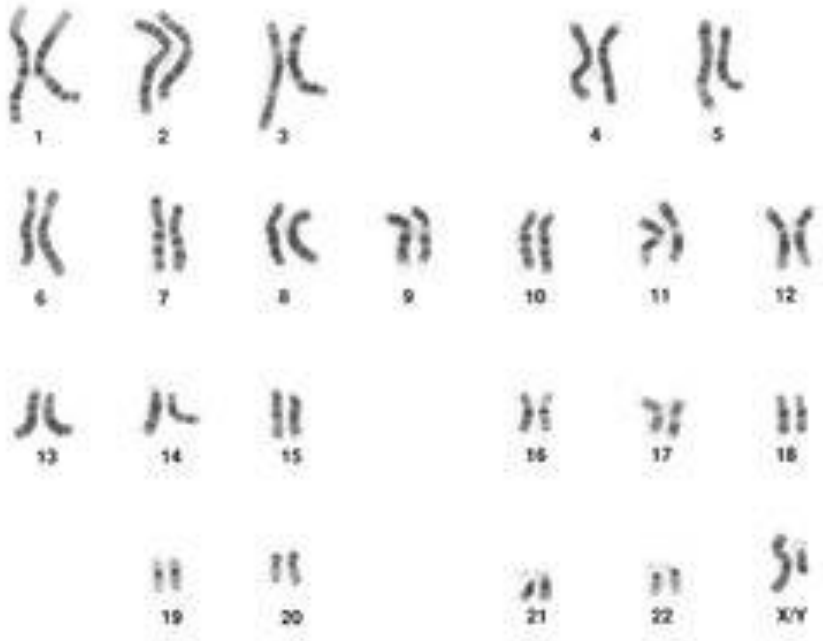
Chromosomes and ploidy

Different organisms have different numbers of chromosomes- these range enormously between taxa. Organisms also have different numbers of sets of chromosomes – this is called the **ploidy** level.

Most mammals- such as humans- are **diploid organisms**, which means they have 2 complete sets of chromosomes, one inherited from each parent.

Humans, therefore have 2 homologous sets of 23 chromosomes (22 pairs of autosomes and 1 pair of allosomes, or sex chromosomes), making a total of 46.

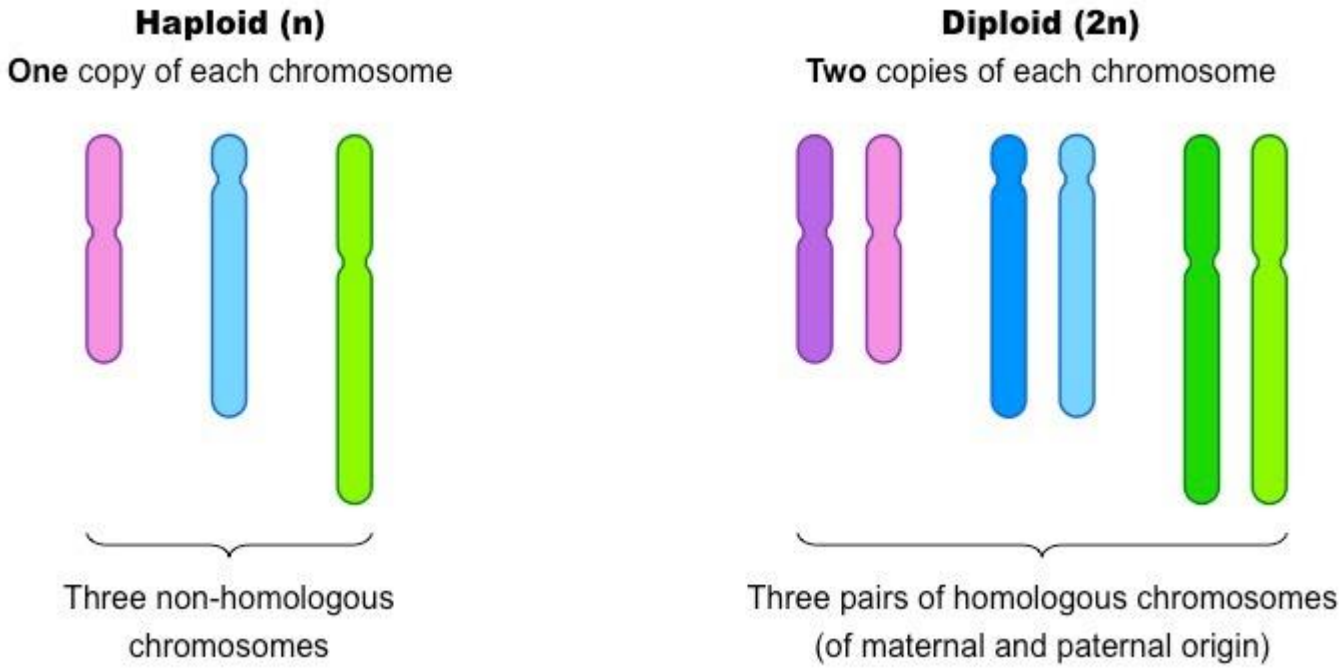
Other organisms however, such as male ants, live as **haploid** organisms throughout their life cycle. This means they have only 1 complete set of chromosomes. Some species, such as algae, go through a phase where their cells will be haploid.



Chromosomes and ploidy

The haploid number is denoted as **n**. Most mammalian cells (the somatic cells, or non-reproductive cells) are diploid (**2n**) and contain both sets of chromosomes- one set inherited from each parent.

However, reproductive cells- or gametes- are haploid (n)- they have only one set of chromosomes.



Chromosomes and ploidy

While mammals are diploid, the ploidy level of organisms differs across taxa. The number of chromosomes found in a single complete set of chromosomes is called the **monoploid number** (x). Many organisms are **polyploid**, i.e. they have more than 2 sets of chromosomes.

No. sets of chromosomes	Ploidy Level	Monoploid number	Example	Chromosome number
1	Haploid	x	male honeybees	16
2	Diploid	2x	Asian elephant	56
3	Triploid	3x	Banana	33
4	Tetraploid	4x	Peanut	40
5	Pentaploid	5x	Bread wheat	
6	Hexaploid	6x	Domestic oat	42
7	Septaploid	7x		
8	Octoploid	8x	Common strawberry	56

Polyploidy

There are two general categories of polyploidy:

Autopolyploidy : where chromosome sets are derived from the same species.

Allopolyploidy : where chromosome sets are derived from different species. Allopolyploidy is the most common form of polyploidy.

Polyploidy is very common in plants, but rare in animals. In animals polyploidy is found in a variety of invertebrates, such as leeches and flatworms; some fishes, for example, salmon and trout which are tetraploids; and it is found in salamanders, lizards and frogs.

Polypoloidy is believed to be an important driver of plant speciation: it is estimated that between 30-80% living plant species are polyploid. Polyploidy has also played an important role in the domestication of plant crops: many common fruits, vegetables and house plants are polyploid, such as apples, banana, ginger (triploid), cotton, pototo, leek, peanut (tetraploid), chrysanthemum, oat, kiwi (hexaploid), strawberry, sugarcane (octaploid). It is thought that polyploids have greater ecological tolerance and colonization ability than diploids and that it may also facilitate species invasion.

Haplodiploidy

Haplodiploidy is an important concept in Behavioural Ecology because it underpins understanding of the evolution of **eusociality**- the highest level of social organisation commonly found in arthropods (see Chapter 6).

Haplodiploidy is a sex-determination system in which males develop from unfertilized eggs and are **haploid**, and females develop from fertilized eggs and are **diploid**.

Haplodiploidy determines the sex in all members of the insect orders Hymenoptera (bees, ants, and wasps) and Thysanoptera ('thrips').

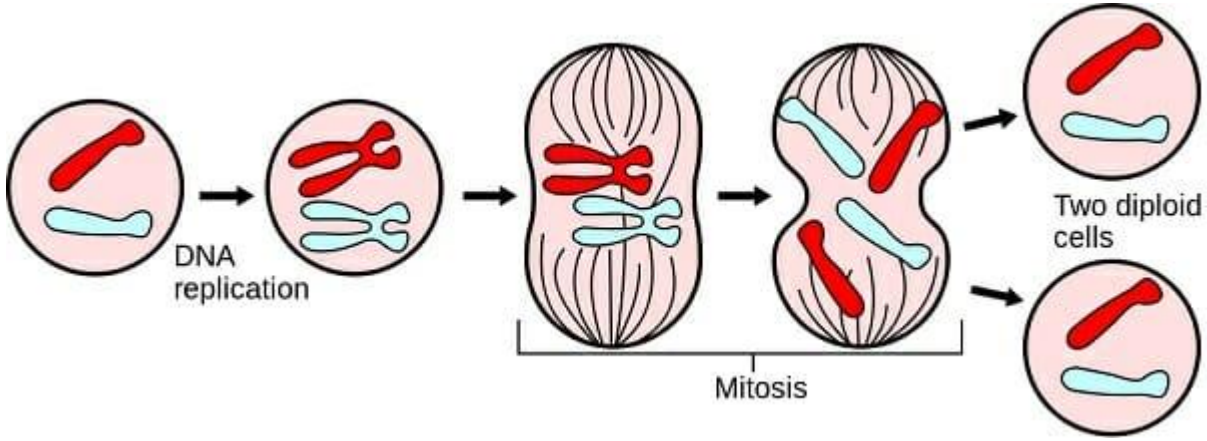
In this system, sex is determined by the number of sets of chromosomes an individual receives. An offspring formed from the union of a sperm and an egg develops as a female, and an unfertilized egg develops as a male.

This system affects the relatedness between different caste members. For example, diploid female workers in single-queen colonies are significantly more related to each other (0.75) than in other systems, such as mammals, where siblings which are usually related by 0.5. It is this point which drives the kin selection theory of how eusociality evolved.



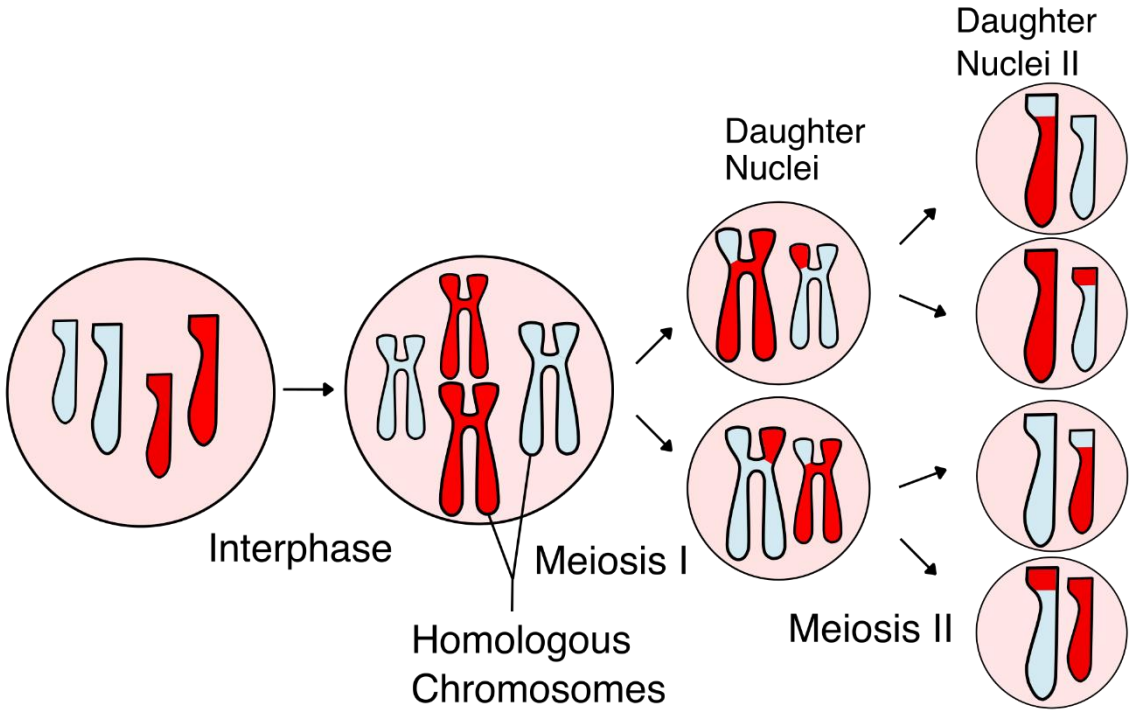

Mitosis

When our body needs to produce new cells for growth or repair, it does so by a process called **mitosis**, where the chromosomes are replicated and the cells divide into two to produce two identical daughter cells. The main purpose of mitosis is for growth and cell replacement. Mitosis occurs in somatic (non-reproductive) cells and is essential for tissue growth and repair, and also forms the basis of asexual reproduction. When errors in mitosis occur, mutations can arise. This can lead to cell cycle disruption and uncontrolled cell division. Cancer is essentially the result of uncontrolled mitosis.



Meiosis

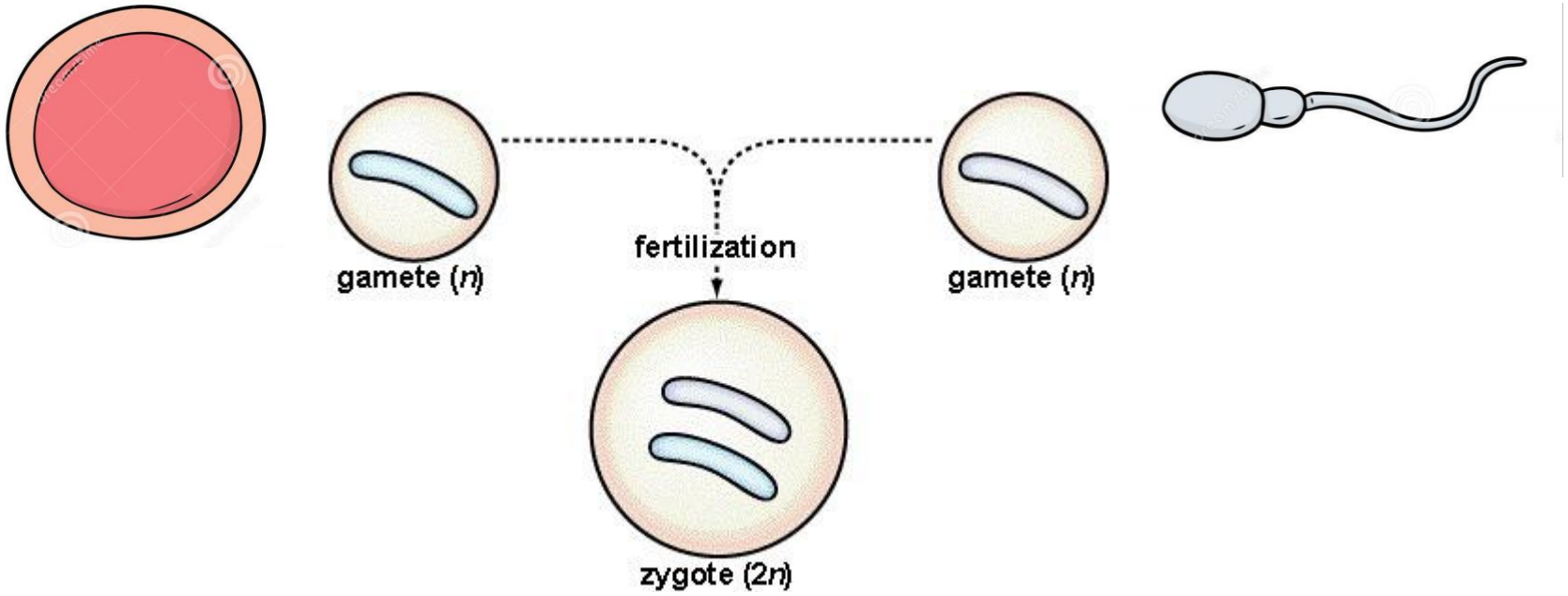
Meiosis is the process whereby a single cell divides twice to form 4 daughter cells each with half the number of chromosomes as the original parent cell. Chromosomes replicate, and then genetic information is shared between homologous chromosomes in a process called genetic recombination. The daughter cells that are formed are **gametes** (sperm, ova, pollen), each containing a unique combination of genes shuffled from the parent chromosomes.



Meiosis

Meiosis occurs in reproductive cells and enables sexual reproduction- during fertilisation gametes fuse together to form zygotes with the full number of chromosomes.

Meiosis occurs in all animals and plants and is unique to eukaryotes. It is the process by which genetic information is passed from one generation to the next- it is the mechanism for **heredity**. Because genes are shuffled into unique combinations, it increases genetic variation in a population and explains why every individual is unique.



3.2

Inheritance and variation

Alleles

Genes come in variant forms – each variant is known as an **allele.** A gene may have many different alleles in a population, or hardly any at all. In diploid organisms, each individual carries at least two alleles for each gene, one inherited from each parent. Each copy may be the same, or they may be different. Different alleles can cause variations in the proteins that are produced, or they can change protein expression: when, where, and how much protein is made. Proteins affect traits, so variations in protein activity or expression can produce different phenotypes.

Alleles are what cause the variation in the different traits we see (e.g. eye colour, height, hair and skin colour, shell colouration etc).

Genotypes

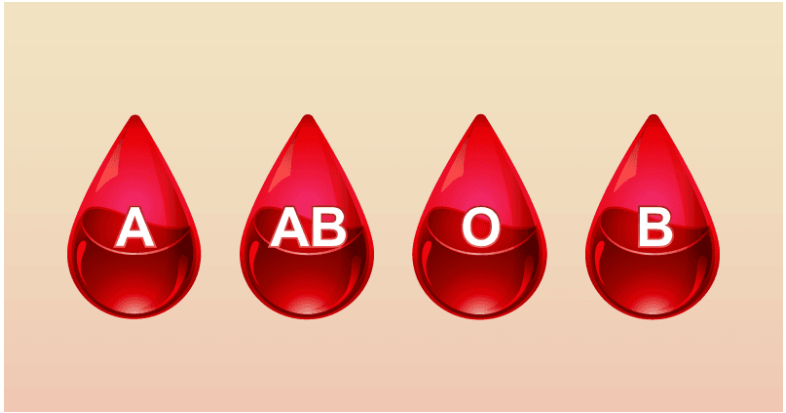
The exact fixed position on the chromosome that contains a particular gene and its different alleles is known as a **locus**. An individual's **genotype** can be said to be it's genetic constitution at a particular locus. For a diploid organism, this will be the combination of alleles on homologous chromosomes, each inherited from one parent.

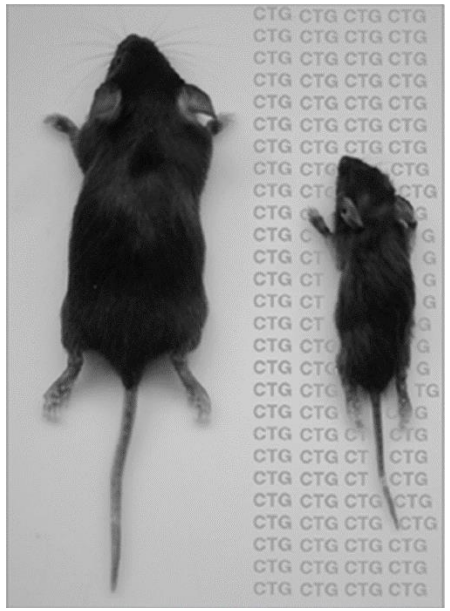
For example, if an individual has inherited two different alleles ***A*** and ***a***, we can say that at this particular locus it's genotype is ***Aa***.

Phenotypes

Phenotypes are the observable characteristics of an organism– size, colour, mass, behaviour. Phenotypes are the result of varying degrees of the physical expressions of genes, the influence of the environment, or a combination of both.

Blood type, for example, is a phenotype resulting only from gene expression. Blood types are inherited and do not change as result of environmental influences in life.





Body size, however, is an example of a phenotype resulting from the combination of gene expression and environmental factors. Variation in body size is regulated by genes but affected by environmental factors such as diet. For example, mice fed a rich diet will attain a larger body size than those fed a poor diet, but none will attain the size of a horse.

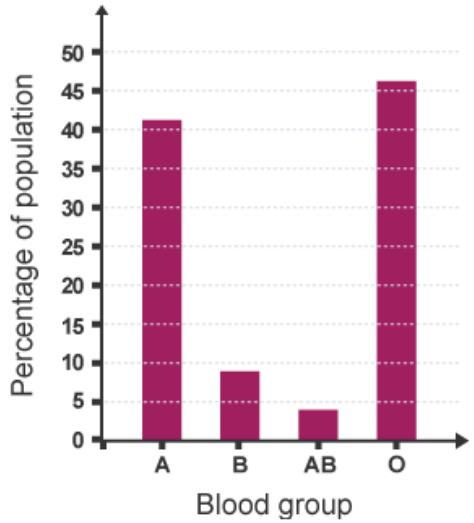
Phenotypes

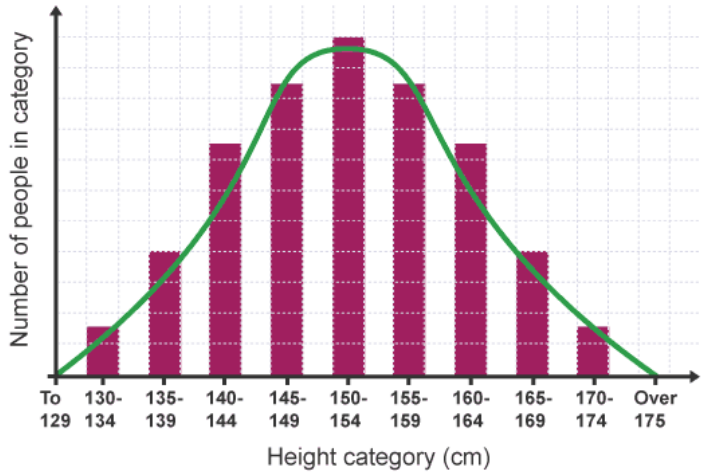
Phenotypic variation is in itself discrete (discontinuous) or continuous.

Discontinous variation describes traits where there are only one or a few distinct possibilities. Blood group is discontinous as it comes in only a few distinct possibilities. There are no 'in-between's'.

Other examples include flower colour in pea plants (purple or white- see Mendel's famous study), sex (male or female), number of fingers. Such phenotypes are regulated by just one or a few genes, and are not (usually) influenced by the environment over time.

Continous variation describes traits that come in a range or continuum. Examples of continuous (or quantitative) traits include hair and skin colour, IQ, stem height, clam shell coloration, body weight. Such traits are controlled by multiple genes and are affected by the environment to varying degrees.





Dominant and Recessive alleles

The terms dominant and recessive describe the inheritance patterns of certain traits: how likely it is for a certain phenotype to pass from parent offspring.

A **dominant** allele produces a dominant phenotype in individuals who have at least one copy of the allele, which can come from either parent.

For a **recessive** allele to produce a recessive phenotype, the individual must have two copies, one inherited from each parent.

An individual who carries **one dominant** and **one recessive** allele for a gene will have the **dominant** phenotype. They are generally considered "carriers" of the recessive allele: the recessive allele is there, but the recessive phenotype is not.

Dominant and recessive are important concepts, but they are often over-emphasized. Most traits have complex, unpredictable inheritance patterns, arising from the expression of multiple genes.

Also, it is important to remember that the **mode of inheritance** has nothing to do with whether an allele **benefits** an individual or not.

Inheritance

Imagine a trait that is coded for by a single gene with two alleles: a dominant allele ***A*** and recessive allele ***a***. (In reality only a minority of traits are controlled in this way. Some examples in humans include free-hanging earlobes, widow's peaks and colour blindness).

An individual therefore may have the following genotypes:

A A : where two dominant alleles are inherited from each parent.

A a : where one dominant and one recessive allele is inherited from each parent

a a : where two recessive alleles are inherited from each parent.

In ***A A*** or ***a a***, the genotypes are known as **homozygous,** because in both cases the individual carries identical copies of the same allele.

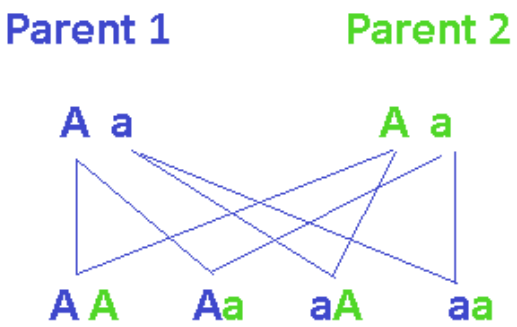
In ***A a***, the genotype is **heterozygous**, because the individual carries different alleles**.**

In ***A A*** and ***A a***, the dominant allele will express the phenotype ***A***. Phenotype ***a*** will only be expressed in the double-recessive genotype ***a a*****.**

So here, we can see that phenotype and genotype are different: because of dominance in genes, individuals may have the same phenotype but different genotypes.

Monohybrid cross

Punnet squares are often used to work out the probability of particular genotypes being passed from one generation to the next. If we assume that two heterozygous individuals with the genotype ***A a*** reproduce, we can draw out or use a Punnet square to work out the probability of their offspring having a particular genotype. This is known as a monohybrid cross.



	A	a
A	A A	a A
a	A a	a a

In this example the probability of the offspring having the genotype ***AA*** is 25%, ***Aa*** is 50%, and ***aa*** is 25%. Genotypes ***AA*** and ***Aa*** will express phenotype ***A***, whereas only genotype ***aa*** will express phenotype ***a***. The ratio of the phenotypes is therefore 3:1.

The 3:1 ratio is well-described: Gregor Mendel used punnet squares in his famous pea-plant experiments to predict the outcome of any genetic cross. British mathemeticians Hardy and Weinberg applied a similar approach to predict the outcome of mating for an entire population. This relationship, known as Hardy-Weinberg equilibrium is a fundamental concept in population genetics and is described in Chapter 5.

3.3

Relatedness

Coefficient of relatedness

Understanding the level of relatedness between individuals is important to behavioural ecologists, for example when studying the evolution of cooperation and conflict, since that may affect the costs and benefits of the actions of individual organisms; and also in understanding mating strategies and patterns of dispersal and distribution.

The **coefficient of relatedness** is a measure of the degree of consanguinity or biological relatedness between two individuals. It is also used to express degrees of kinship between humans. It refers to the probability that two related individuals have inherited a particular allele of a single gene from their common ancestor. This allele is termed IBD (identical by descent). For diploid organisms the coefficient is calculated as follows, where n represents the number of generations separating in the geneology and $\sum$ the sum of the different possible pathways:

$$r = \sum \left(\frac{1}{2}\right)^{\mathrm{n}}$$

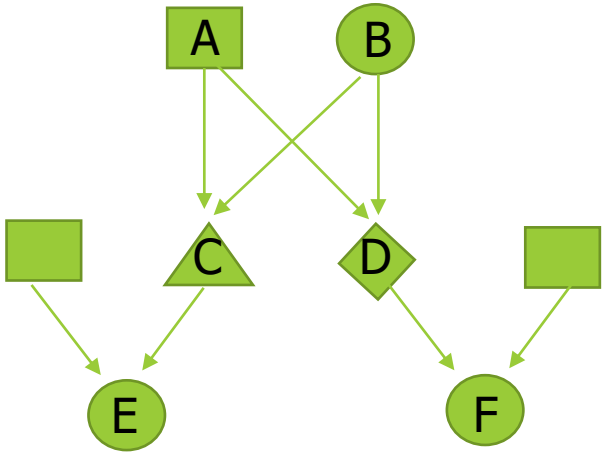
r ranges from a minimum of 0 (unrelated) to 1 (clones or identical twins).

Coefficient of relatedness

So, for example, the coefficient of relatedness (r) between a parent (A) and child (C) is $(1/2)^1 = 0.5$, since there is just 1 genealogical step between them. For two siblings (C and D) there are 2 pathways of two steps relating them via common ancestors A and B (CAD and CBD), so $r = (1/2)^2 + (1/2)^2 = 0.5$.

For aunt and nephew C and F, the common ancestors are A and B, with two pathways of 3 steps CADF and CBDF, so $r = (1/2)^3 + (1/2)^3 = 0.125 + 0.125 = 0.25$.

For first cousins E and F, $r = (1/2)^4 + (1/2)^4 = 0.0625 + 0.0625 = 0.125$ and so on.



3.4

Evolution

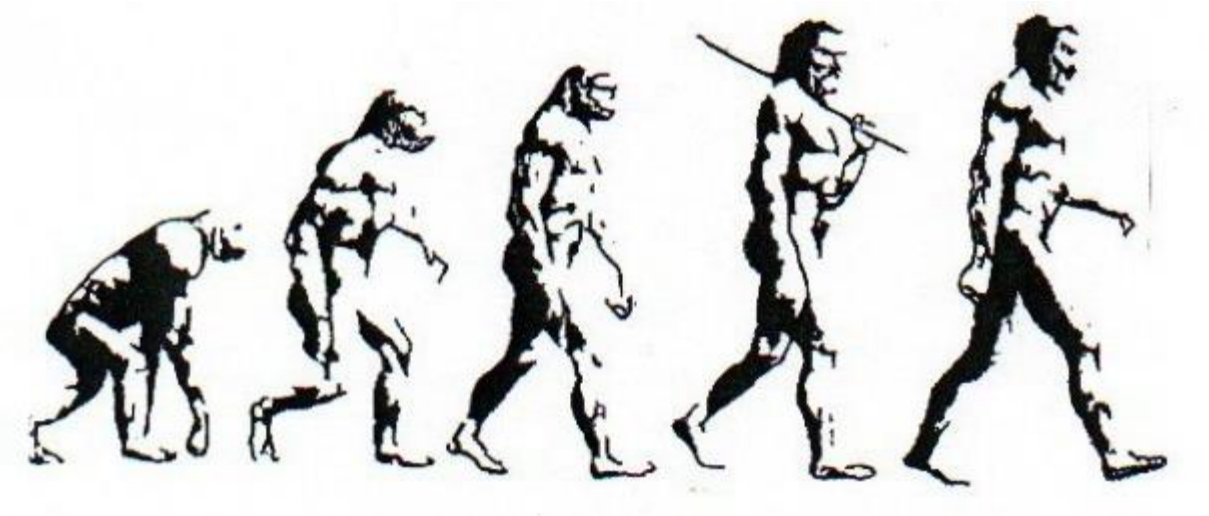
Genes and evolution

Organisms are vehicles for genes.

Organisms are mortal, but because genes are passed on to successive generations, they are potentially immortal.

Because genes are shuffled into unique combinations, it increases genetic variation in a population and explains why every individual is unique.

Evolution is the processes that results in gradual changes in heritable traits of biological populations over successive generations.



Genes and evolution

Evolution is a ‘blind’ process.

Species do not set out to ‘become’ more evolved, and DNA is not programmed to turn species into ‘higher’ life forms.

Evolution is really the product of chance events that result in an accumulation of genetic change over time.

Biological complexity is not achieved by pure chance alone, however, but by **random variation** combined with **cumulative selection**

The basic mechanisms of evolution are known as **natural selection, mutation, migration**, and **genetic drift**

Natural Selection

This is often referred to as ‘survival of the fittest’

In any population of organisms there is variation. Some of that variation will be beneficial, some will be detrimental and some will be neutral and won’t have any effect

Organisms who have a slight adaptation that is beneficial to them will have a better chance of survival.

Because they have a better chance of surviving, their beneficial alleles also have a better chance of being passed down to the next generation.

Over time and many generations, the most desirable characteristics will replace the detrimental ones, and have been **selected for.**

Mutation

Sometimes changes occur to the genetic material itself- either to the DNA or to whole chromosomes. This is known as **mutation**.

Mutations can be caused by errors in meiosis, damage by radiation, chemical poisoning, or it can happen purely by chance.

Mutations may be bad for the organism, neutral, or beneficial

Sometimes mutations are lethal. In this case, the organism dies and the mutation is deleted from the population.

When mutations are not lethal, the changes are heritable- passed to the next generation.

Mutations are the ultimate source of variation, upon which natural selection acts

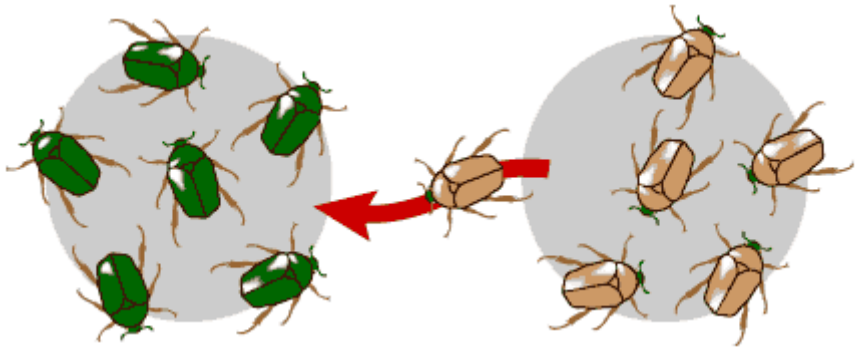
Migration

This is also referred to as ‘gene flow’.

Genetic variation is transferred from one population to another when organisms move, migrate or disperse and reproduce within the other population

It could be pollen being blown over a mountain top, or people moving to new areas.

The loss or addition of alleles through gene flow can easily change gene pool frequencies and is an important driver of evolution



Genetic drift

Genetic drift is the **changes** in the frequency of alleles in a population due to random chance (either in meiosis or whether an organism survives and reproduces).

When there are few copies of an allele in a population (a rare allele), the effects of genetic drift are larger

When there are many copies of an allele (common), the effects are smaller.

3.5

Take-home message

Take-home message

- The gene is the basic unit of heredity.
- Meiosis is the mechanism for heredity.
- Evolution is really the product of chance events that result in an accumulation of genetic change over time.
- The basic mechanisms of evolution are natural selection, mutation, migration, and genetic drift.
- Mutations are the ultimate source of variation, upon which natural selection acts.
- Natural selection is the most important in providing a link to the ecological context within which organisms exist and interact.

3.7

Resources

Suggested Reading

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