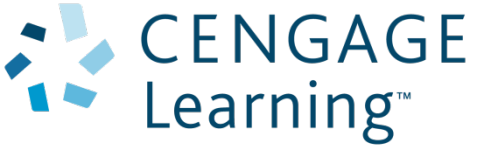
Chapter 4: Variation and mutation



Phenotypic expression of genes

Shutterstock.com/Vinai rasemeejun

Genes carry a set of instructions for how to make a protein. When the gene is read, transcribed and translated into a protein, the gene is said to be **expressed**. By coding for proteins, genes determine important facets of biological structure and function. These observable traits are known as the **phenotype**.

Phenotypic expression of genes

Genes cannot dictate the structure of an organism by themselves. The other crucial component in the formula is the environment. Twins may have the same gene and protein for a trait, but the trait may look different in the two individuals because the environment can influence the final phenotype. There is a diversity of genetic and phenotypic traits within and between populations known as **variation**.

iStock.com/ampueroleonardo

Note the variation in fur covering between these individual little penguins.

Variation terminology

Phenotype

An observable trait produced by the actions of one or more gene-encoded proteins; influenced by the genotype and the effects of the environment

Genotype

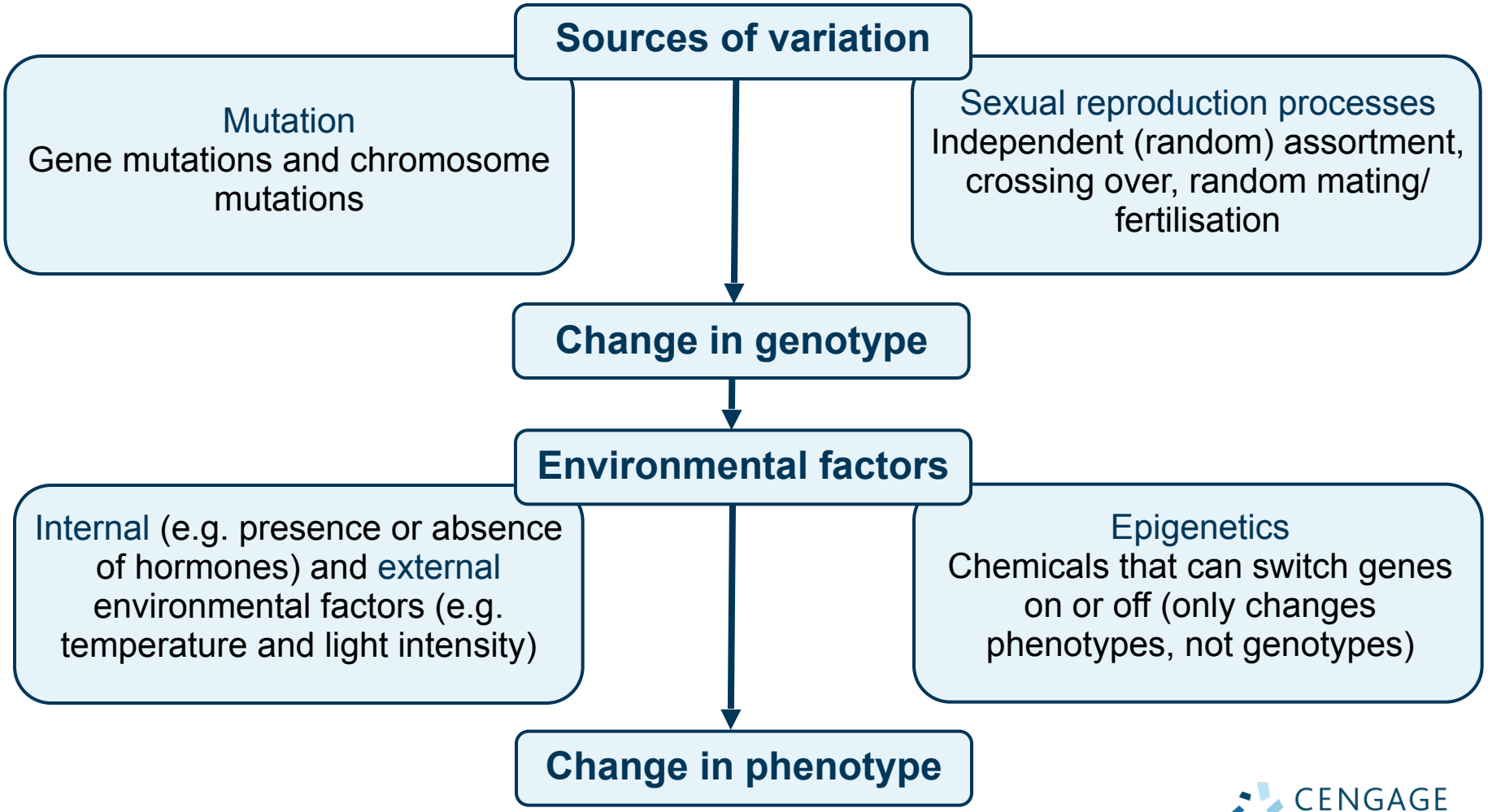
The genetic composition of an organism for a particular trait; the set of alleles that an organism has for a particular trait

Allele

A form of a gene; different alleles code for the same trait but different versions of the trait

Phenotypic expression of genes

Main mechanisms for genotypic and phenotypic variation



Traits influenced by the environment

Some traits that have a range of phenotypes given the same genotype:

- Height
- Size
- Skin colour
- Flower colour

Note: There are some traits that are strictly defined by genotype, such as the ABO blood group system.

Environmental factors that influence a phenotype

Environmental factors that can influence a phenotype:

- Temperature
- pH
- Availability of food
- Light exposure
- Wind exposure

Environmental factors

Examples

A turtle embryo can become either a male or a female depending on the temperatures it experiences while in the egg. Sex for these animals is determined by the environment. Sea turtle eggs incubated at low temperatures produce male turtles, and eggs incubated at higher temperatures produce female turtles.

Shutterstock.com/Jao Cuyos

Environmental factors

Shutterstock.com/Marina Andrejchenko

The range of colours in hydrangea flowers can be traced back to the pH in the soil. A more acidic soil is conducive to blue flowers. The pH does not change the genome because the genotype does not change. It is the interaction of the environment with either the gene or the protein that determines the flower colour in hydrangeas.

Epigenetics

Epigenetics is the study of heritable, but reversible, changes caused by the layer of chemicals above DNA via activation and deactivation of genes. Epigenetics can alter gene expression and therefore affect phenotype

When the chemical layer above DNA, the **epigenome**, is altered, it does not alter the DNA sequence and therefore it is not a mutation, because mutations are permanent changes to the DNA.

Some chemicals, such as acetyl groups can ‘turn genes on’ and some chemicals, such as methyl groups, can ‘turn genes off’.

Because epigenetics affects gene expression, it can affect the growth and development of organisms. For example, a smoking mother can cause epigenetic factors to be modified in her unborn child, causing the child to be more at risk of obesity.

Mutation: the ultimate source of new variation

What is a mutation?

A **mutation** is a permanent change to an organism's DNA sequence of nucleotides.

How do mutations occur?

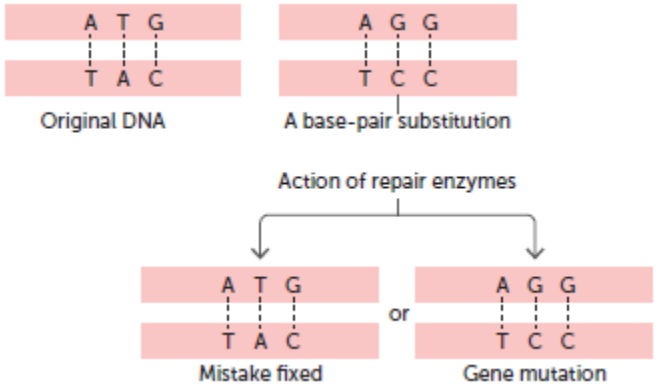
Mutations may arise spontaneously during DNA replication or cell division, or they may be induced by physical or chemical environmental factors called **mutagens**, or through the action of biological agents.

Where do mutations occur?

Mutations can occur in non-reproductive (body, or **somatic**) cells or in the reproductive (**germ-line**) cells that produce **gametes** (sex cells) at the conclusion of meiosis.

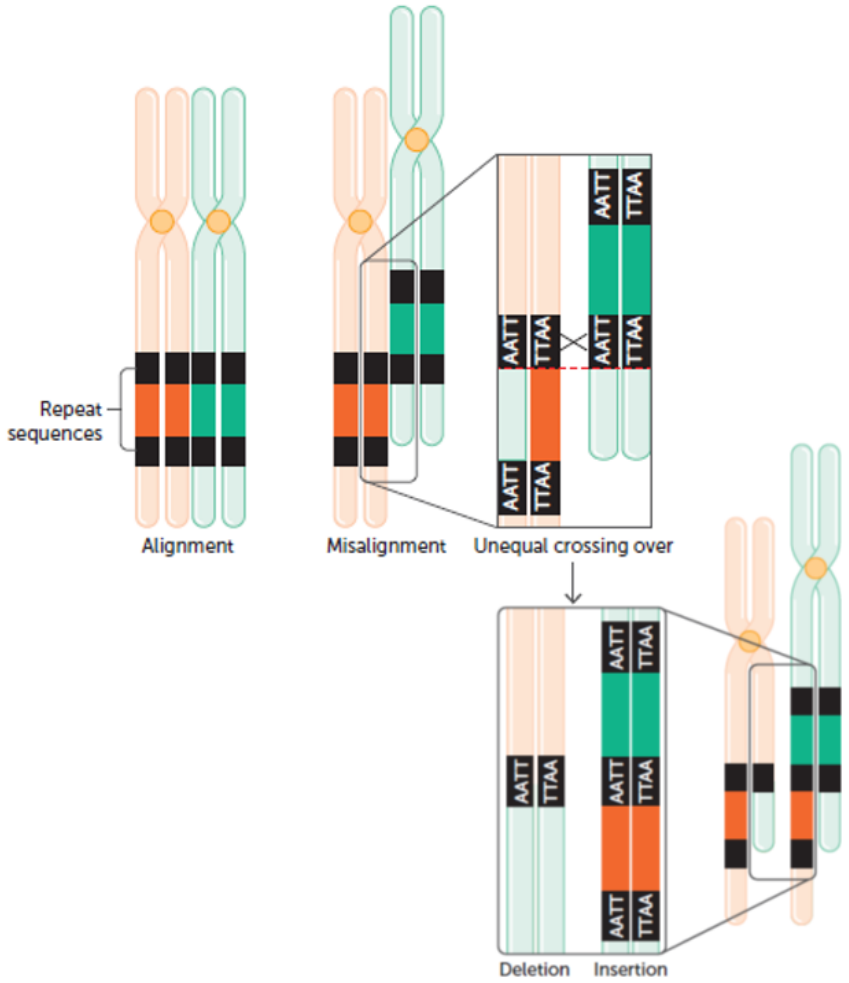
Causes: errors in DNA replication

Base-pair substitution results in either a mistake being fixed or a mutation.



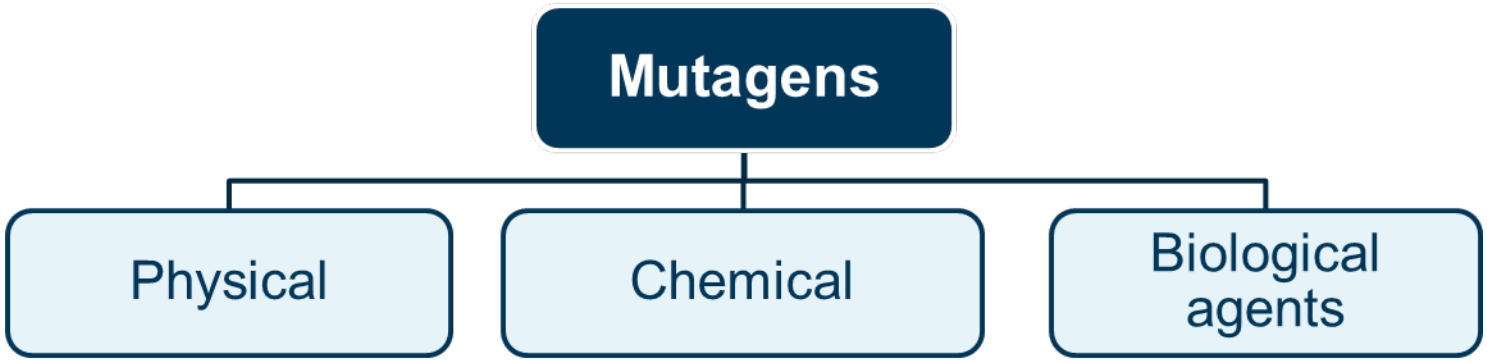
Causes: errors in cell division (mitosis and meiosis)

When homologous chromosomes misalign during meiosis, unequal crossing-over occurs. The result is the deletion of a DNA sequence in one chromosome, and the insertion of a DNA sequence in the other chromosome.



Benjamin Pierce, Genetics: A Conceptual Approach, 2nd Edition, 2004.

Causes: mutagens



Causes: physical mutagens

Physical mutagen	Effect
UV light	Structural distortion by cross-linking neighbouring nucleotides
X-rays	Gene and chromosome aberrations
Nuclear radiation	Breaks in DNA strands

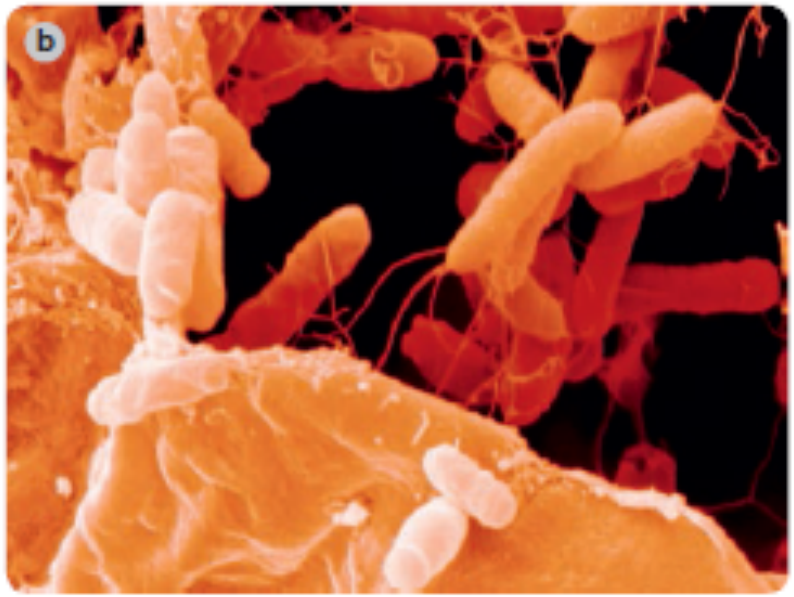
Causes: chemical mutagens

Chemical mutagen	Effect
Mustard gas (sulfur mustard)	Mustard gas affects the base guanine; causes a substitution mutation
2-aminopurine, 5-bromouracil	Nucleotide substitution
Colchicine	Prevents spindle formation in mitosis; doubles chromosome number
Nitric acid	Adenine in DNA is deaminated; it behaves like guanine

Causes: biological agents

Bacteria of the genus *Agrobacterium* cause crown gall disease in the stems of plants. The cell of the host plant becomes modified by **horizontal gene transfer**.

Nature Picture Library/Visuals Unlimited



Alamy/Custom Medical Stock Photo

Types of mutations in genes and chromosomes

Point mutation

The simplest form of mutation is a **point mutation** − a single nucleotide within the original DNA sequence is affected by a substitution, addition or deletion.

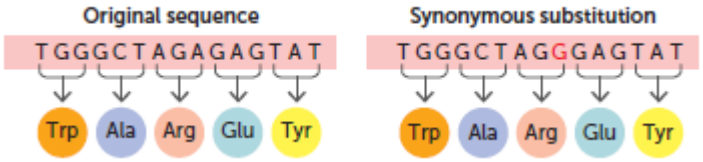
Chromosome mutation

Chromosome mutations can affect many genes simultaneously. Some of the variations that occur with chromosomes, such as chromosome number, are quite natural in certain situations and are therefore integral to the functioning and continuity of the species. Others arise because of anomalies that occur during the formation of the gametes.

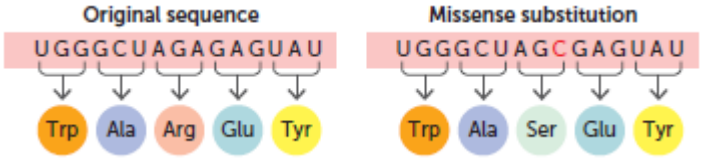
Point mutations: substitution

Substitution point mutations can result in either:

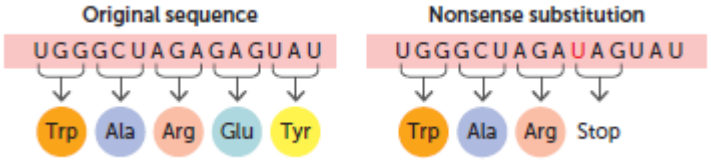
synonymous,



missense

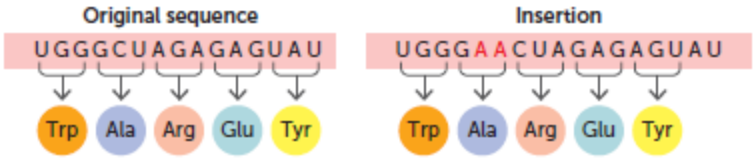


or nonsense mutations.



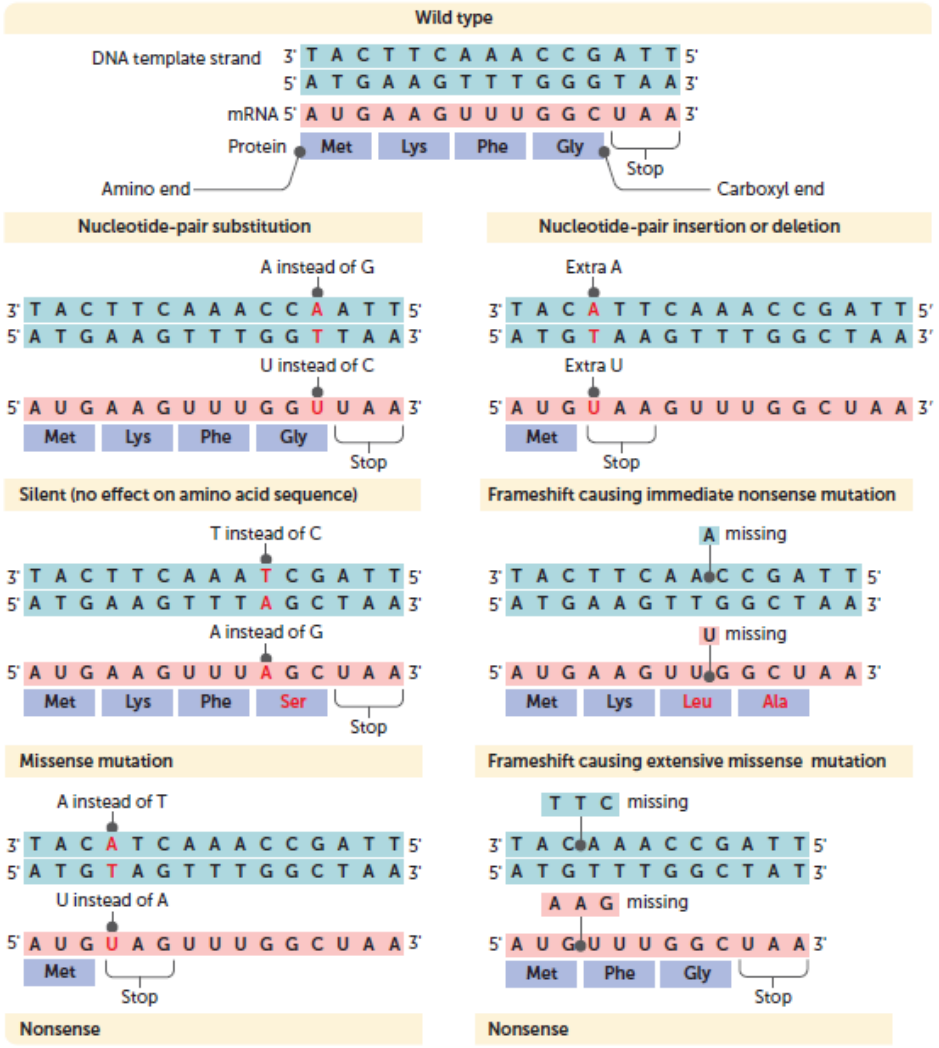
Point mutations: insertion and deletion

Insertion and deletion point mutations can result in a frameshift and, additionally, either synonymous, missense or nonsense mutations.



Mutations

Point mutations: summary of effects



CENGAGE Learning

Chromosome mutations

Aneuploidy

Aneuploidy is the presence of a chromosome number that is different from the simple multiple of the basic chromosome number. Aneuploidy could be the result of either a loss of one or more chromosomes or due to an addition of one or more chromosomes.

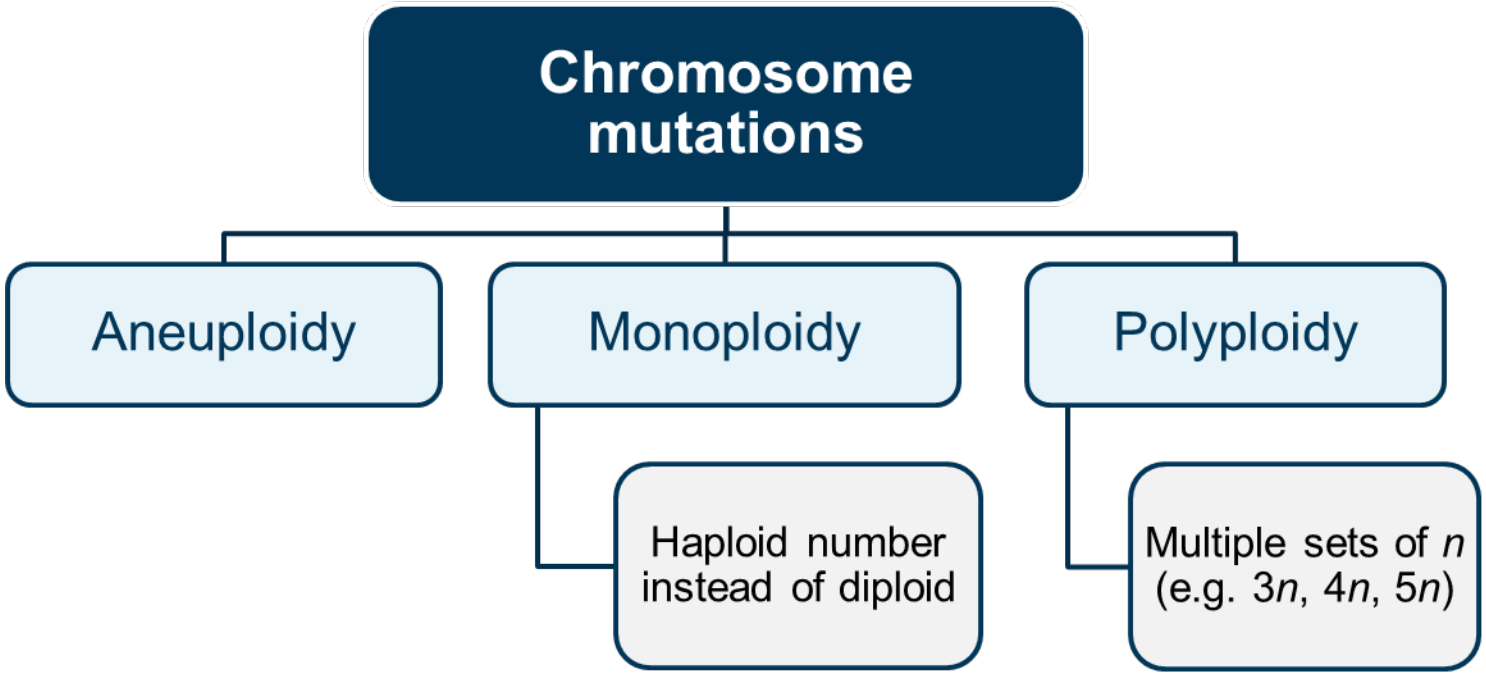
Monoploidy

Monoploidy involves a haploid number of chromosomes that act as a complete set, instead of diploid.

Polyploidy

Polyploidy occurs when an organism acquires one or more complete extra sets of chromosomes.

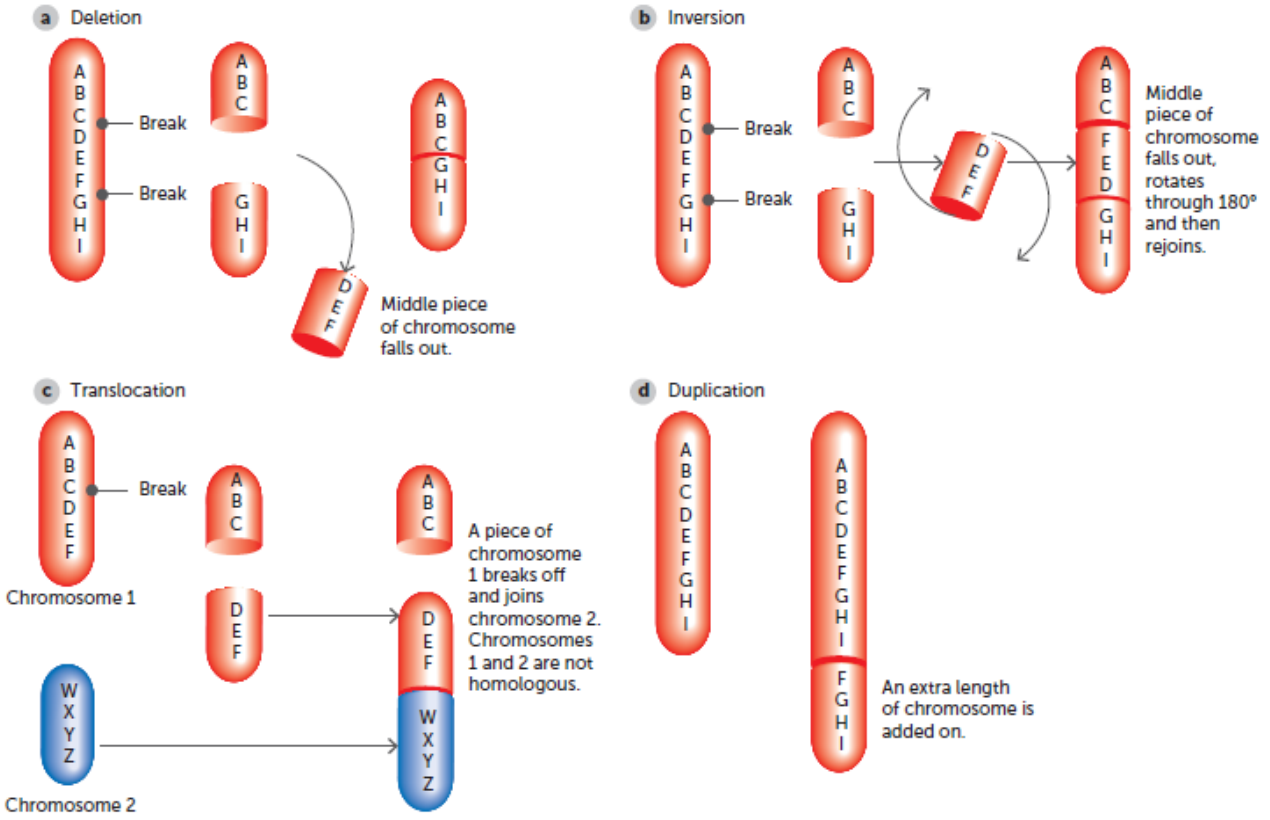
Mutations



Chromosome mutations: variation in chromosome structure

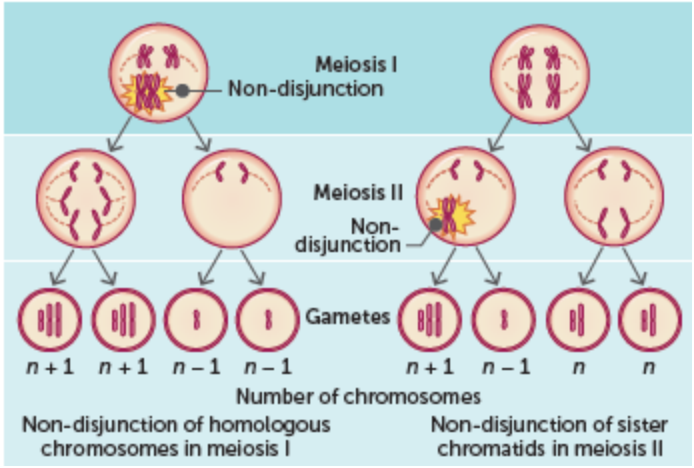
Abnormalities caused by chromosomal mutations may arise by

- deletion,
- inversion,
- translocation
- duplication.

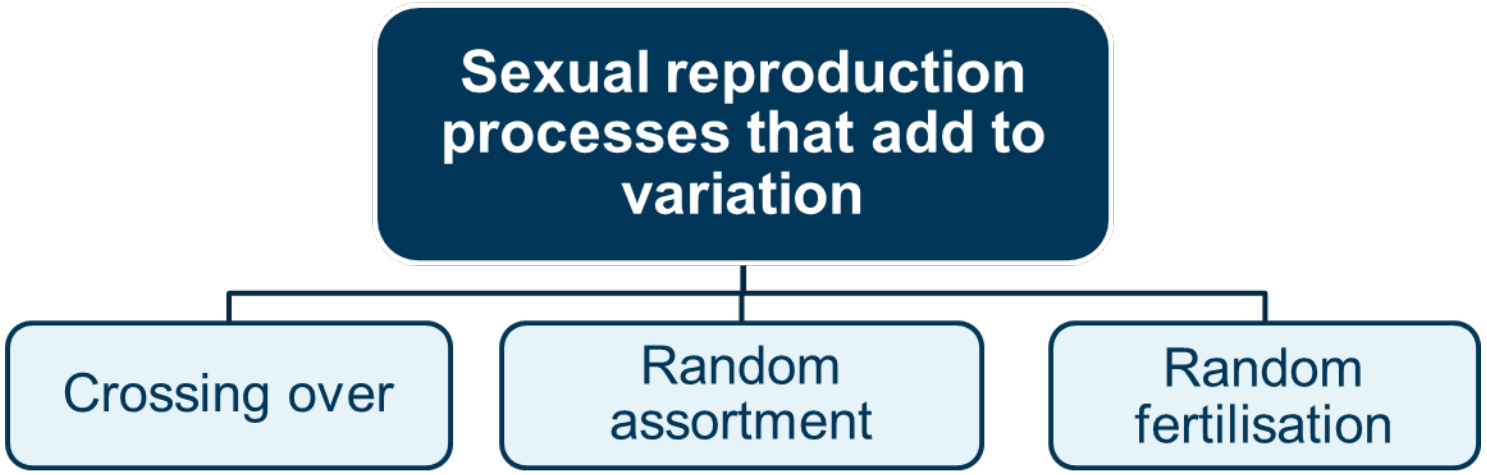


Aneuploidy: mutation via meiosis non-disjunction

In meiosis, identical chromosomes come together and then segregate into separate cells, so that the gametes finish up with only one of each pair of chromosomes. Occasionally, the two identical chromosomes, instead of separating, go into the same cell. This is known as **non-disjunction**.



Variation due to sexual reproduction processes



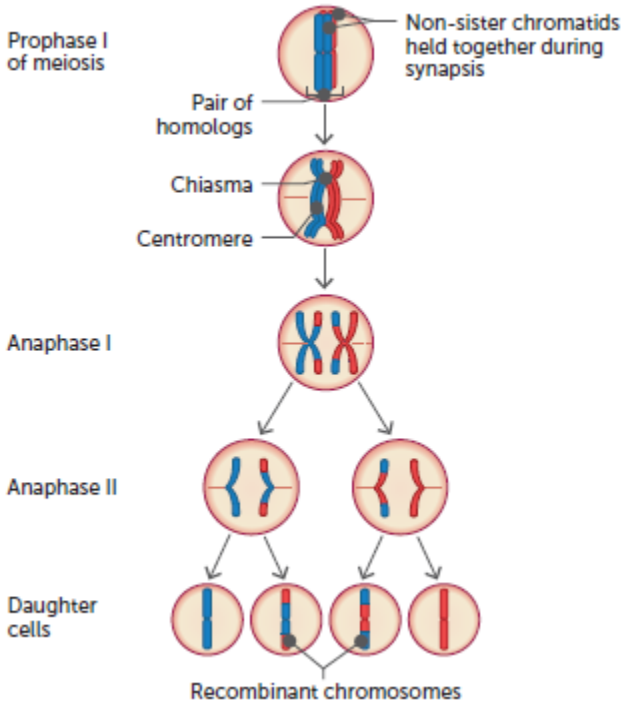
Variation due to sexual reproduction processes

Crossing over

Crossing over is the swapping of alleles (genetic material) that occurs in the germ line during prophase I only.

Crossing over is important for genetic variation, because it allows the exchange of alleles (genetic material) between the maternal and paternal homologous chromosomes (non-sister chromatids).

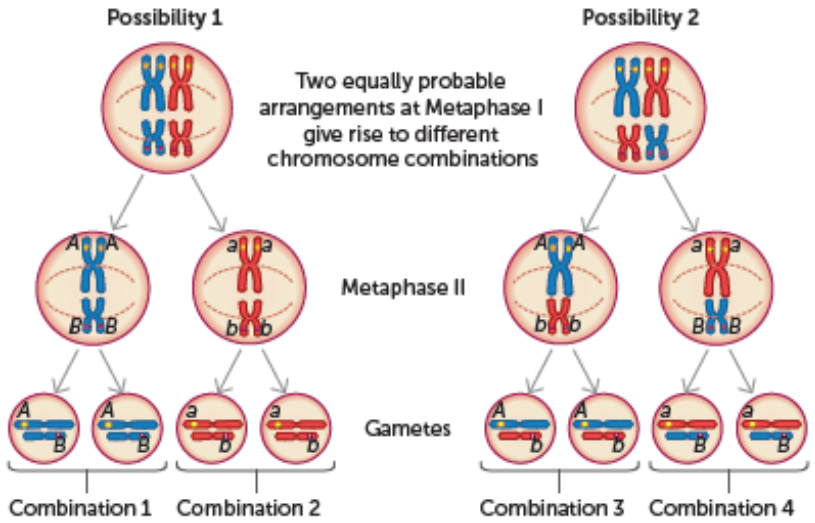
At the end of meiosis, there are four possible gametes. The four haploid cells contain chromosomes that are genetically different from the parent cells and from one another.



Variation due to sexual reproduction processes

Independent assortment and random segregation

The Law of **Independent Assortment** refers to the random orientation of maternal and paternal homologous chromosomes at the equator during metaphase I. The orientation of each homologous pair is randomly left to right, and each pair is unaffected by the orientation of any other homologous pair.



Each gamete ends up with a random selection of maternal and paternal chromosomes. The chromosomes have segregated (separated) randomly into the four gametes.

Random fertilisation and random mating

The random union of gametes to form a zygote is known as **random fertilisation**.

Fertilisation promotes variation because there is a random fusion of gametes at fertilisation, bringing together chromosomes from two different parents, creating new combinations of alleles in the offspring.

Adding to the variation is the fact that the selection of the male and female gametes is random.

Further to this is the random selection of a mate. The offspring produced in sexual reproduction are genetically different to one another and to their parents. This process results in variation within a population because it involves the mixing of genetic information.

Variation due to sexual reproduction processes

Summary

Variation in a population can be due to mutations in genes or chromosomes, or to sexual reproduction processes.

The environment can influence the variation observed in traits.

Alamy Stock Photo/Nature Picture Library