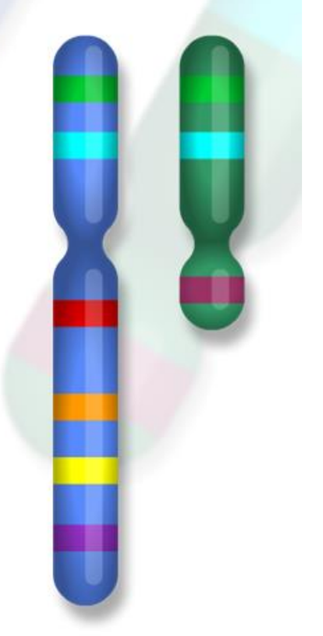
5.5 Genes and patterns of inheritance



Mendel's laws of Inheritance:

1st Law = The law of segregation of factors states that "*when any individual produces gametes, the alleles separate, so that each gamete receives only one allele*". Shown by the separation of homologous chromosomes, carrying the alleles, during anaphase 1 of meiosis.

2nd Law = The law of independent assortment states that *"during gamete formation, the segregation of the alleles of one gene is independent of the segregation of the alleles of another gene"*. Shown by the random arrangement of the homologous pairs on the equator of the spindle at metaphase 1 of meiosis and then their separation during anaphase 1.

5.5.1 Understand the terms genotype and phenotype

- Genetics is the study of **inheritance**.
- Much of our understanding of genetics is due to the work of Gregor **Mendel**, a Czechoslovakian monk, who in the 1850s carried out a vast number of breeding experiments in the garden of his monastery. He concentrated on the garden pea because it had many distinct and easily identifiable characteristics; including flower colour, pea shape and pea colour that varied from plant to plant.

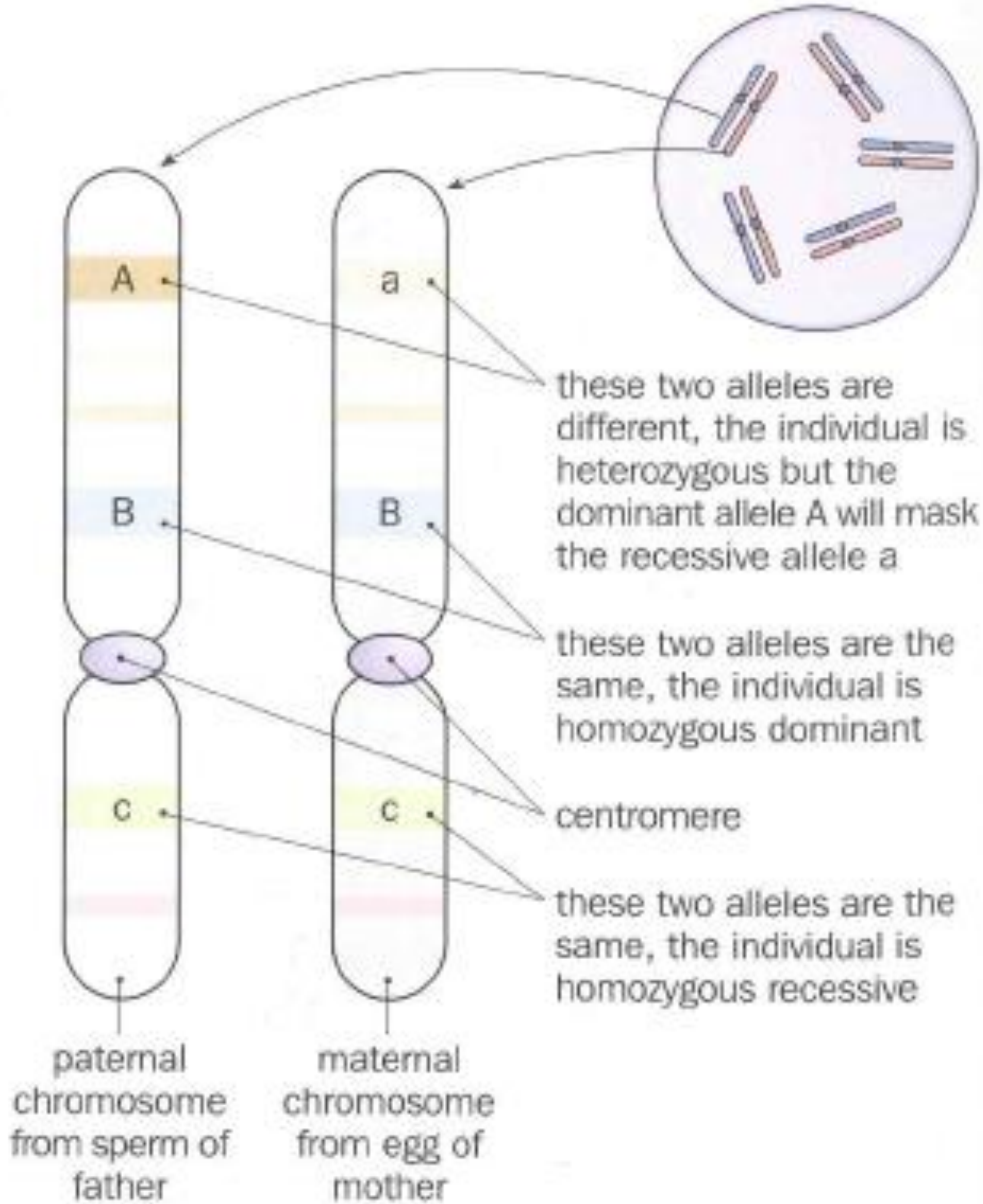
Figure 3.7 Round and wrinkled peas

- Mendel carried out a range of breeding experiments in which he crossed plants carrying particular characteristics that he was interested in. By careful observation of the offspring, he was able to draw conclusions about the nature of inheritance.
- Mendel completed his work without being aware of the existence of chromosomes or genes. Although he published his work in 1866, it was largely ignored and it was only at the start of the 20th Century when chromosomes were discovered under the microscope that the significance of his findings was appreciated. Although additional research and knowledge has increased our understanding of genetics in recent times, it is important to note that this knowledge has built on Mendel’s findings, not contradicted them.
- In recognition of his work, the genetics that we study is called **Mendelian** Genetics.

Useful Definitions

- **Genes** are lengths of DNA which code for a particular polypeptide or trait. They carried on the chromosomes and as chromosomes are found in pairs, the genes are also found in pairs. For any characteristic there are alternative or different forms of the gene and these are called **alleles**.
- **Alleles** occur in the same position on the chromosome called the **locus**, many genes have two alternative alleles but some may have more.
- An allele that is always expressed (shows its effect) is said to be **dominant** – given with a capital letter.

- An allele that is only expressed when no dominant is present is said to be **recessive** – given with a lower case letter.
- If the individual has two alleles the same they are **homozygous** for that characteristic (either dominant or recessive).
- If the individual has two different alleles they are **heterozygous** for that characteristic.
- The genetic make up of an individual is known as its **genotype**.
- The expression of the genotype (the characteristics we can see) is called its **phenotype**. This may of course also be influenced by the **environment** (diet, sunlight etc.)


A
a
B
B
c
c
these two alleles are different, the individual is heterozygous but the dominant allele A will mask the recessive allele a
these two alleles are the same, the individual is homozygous dominant
centromere
these two alleles are the same, the individual is homozygous recessive
paternal chromosome from sperm of father
maternal chromosome from egg of mother

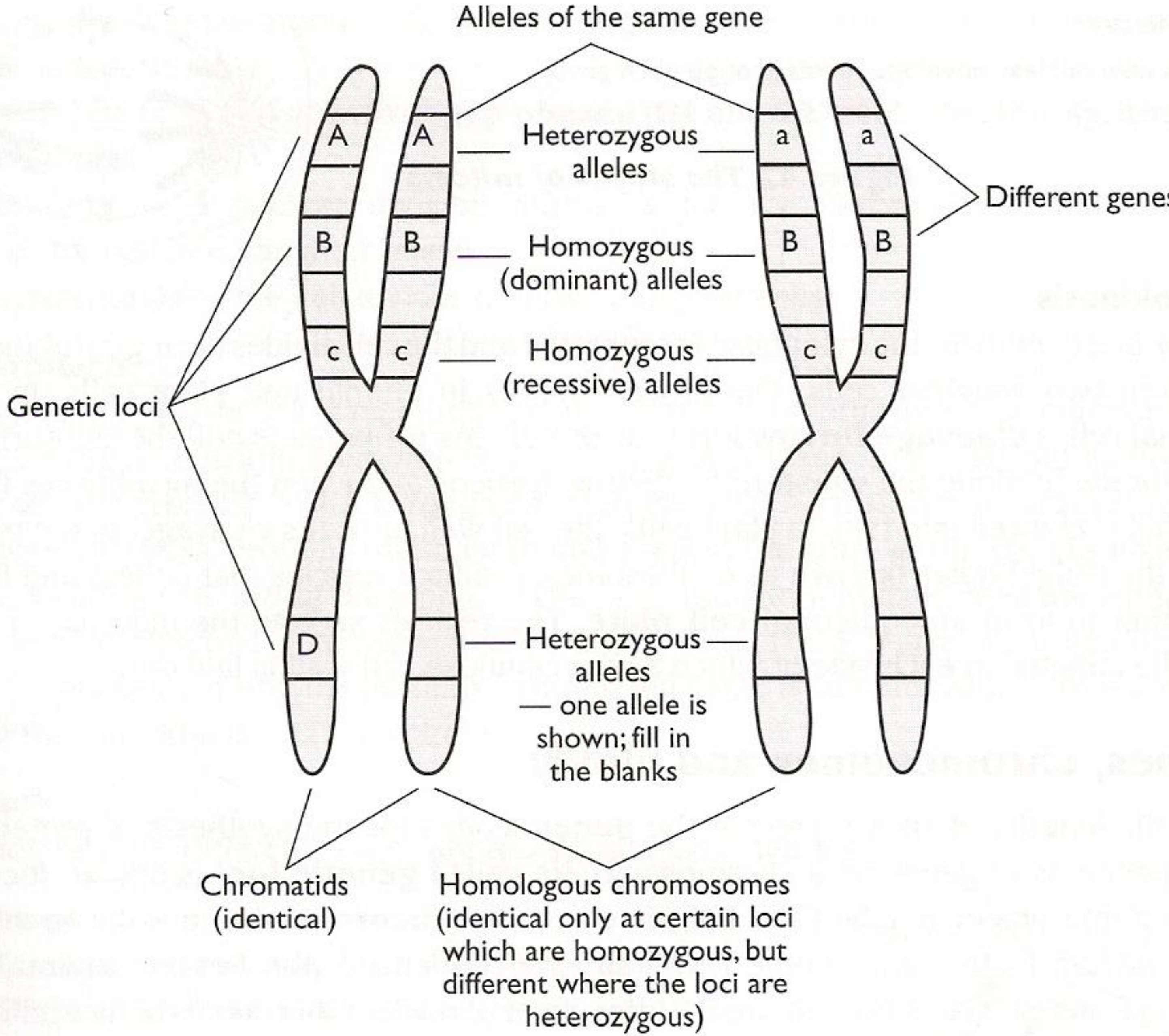


Figure 43 Genes, alleles, chromatids and homologous chromosomes

5.5.2 Understand the relationship between chromosomes, genes & alleles

- Many people find the concept of alleles and genes confusing. A **gene** is a short length of DNA on a chromosome coding for a specific polypeptide. An individual gene may have more than one form that differs slightly from the others in the sequence of nucleotide bases that make it up. These different forms of the gene are known as **alleles**. It is the nature of these alternatives that some are more likely to express themselves than others (dominant) and that our **genotype** is the allele combination that we possess.

- The phenotype which is ultimately expressed depends upon which allele is dominant. A **dominant** allele has its instructions followed and so its effect is produced in the heterozygous condition. The recessive allele does not have its instruction followed in the heterozygous condition.

Representing alleles

A gene can be represented using a letter: upper case for the dominant allele, and lower case for the recessive allele.

For example, the allele for wing length in *Drosophila* can be either long (L) or short (l).

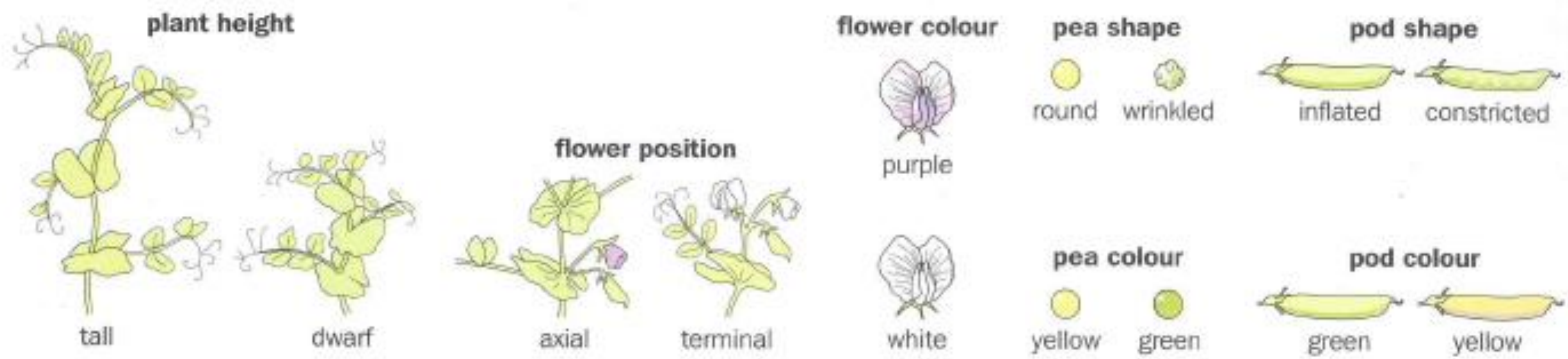
Genotype		Phenotype
LL	homozygous dominant	long wings
Ll	heterozygous	long wings
ll	homozygous recessive	short wings

For example, humans possess a gene that determines the ability to taste phenylthiocarbamide (PTC). The PTC gene, TAS2R38, was discovered in 2003 as a consequence of work carried out during the Human Genome Project. There are two alleles: one (designated by the symbol **T**) is the tasting allele; while, the other (designated **t**) is the non-taster allele. The tasting alle (**T**) codes for a bitter taste receptor protein to which PTC can bind. The non-tasting allele (**t**) codes for a non-functional protein. **T** is dominant over **t**, since a heterozygote, **Tt**, possesses the allele **T** and produces the taste receptor protein. Things are never so simple, and environmental factors can affect PTC tasting ability – for example having a dry mouth may make it more difficult to taste PTC and what is eaten or drunk beforehand may also affect tasting ability.

Practical – who has the PTC "taster" allele?

5.5.3 Understand the inheritance of traits showing discontinuous variation

- **Monohybrid Inheritance**
- **Heredity** is the transfer of genetic factors from one generation to the next, i.e. from parents to their offspring. The first breeding experiments carried out by Mendel were on **monohybrid inheritance**. This involves the inheritance of the alleles of a **single gene.** Mendel used pea plants for his experiments because they had a number of clearly identifiable characteristics such as height, flower colour and pea colour and shape (see below). More importantly, peas self-pollinate (they are true breeding). Mendel cross bred the peas by removing the anthers of one type of plant so that it could not self pollinate and then used feathers to artificially pollinate it with pollen from another type of plant.


plant height
tall
dwarf
flower position
axial
terminal
flower colour
purple
white
pea shape
round
wrinkled
pea colour
yellow
green
pod shape
inflated
constricted
pod colour
green
yellow

- To begin with he crossed tall pea plants with short ones, he expected medium plants in the first generation but to his surprise they were all tall, when he allowed these first generation plants to self-pollinate, the second generation was ¾ tall and ¼ short.
- Tall x short (parental type P)
- All tall (selfed) (first generation F1)
- Tall (787) Short (277) (second generation F2)

Genetic crosses: a history

One of the first people to study genetics was an Austrian monk called Gregor Mendel in the 1850s and 1860s.

He experimented with thousands of pea plants and established the basic foundations of inheritance, such as dominant and recessive characteristics.

Mendel had no knowledge of DNA or genes but he did identify that inheritance is particulate, i.e. it depends on the transfer of separate (discrete) factors from parents to offspring.

Complete the diagram:

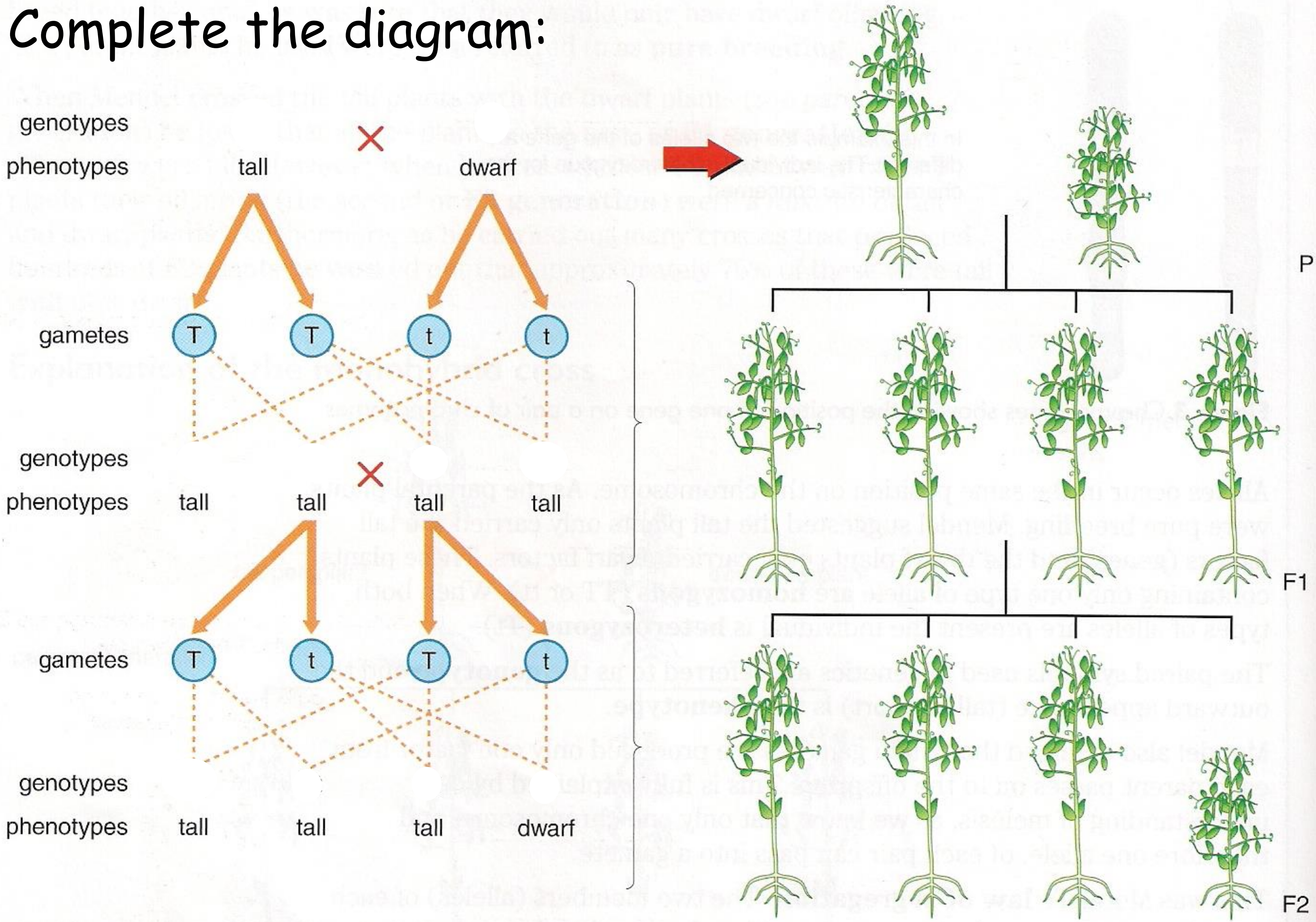


Figure 4 Explaining Mendel's results

Conclusion 1

- Note that in all his breeding experiments Mendel never got plants that showed an intermediate form (medium height in the example above). From this we can conclude that inheritance is not a process in which the features of the two parents are blended but rather a process in which **chromosomes**, carrying the alleles for characteristics (which may or may not show themselves) are transmitted from parents to offspring.

Conclusion 2

- There were no short plants in the F1 generation although they did reappear in the F2. From this we can conclude that although the F1 plants are tall, they do possess an allele for shortness that remains 'hidden' in the heterozygote F1 plants, confirming that **two** alleles are at work.

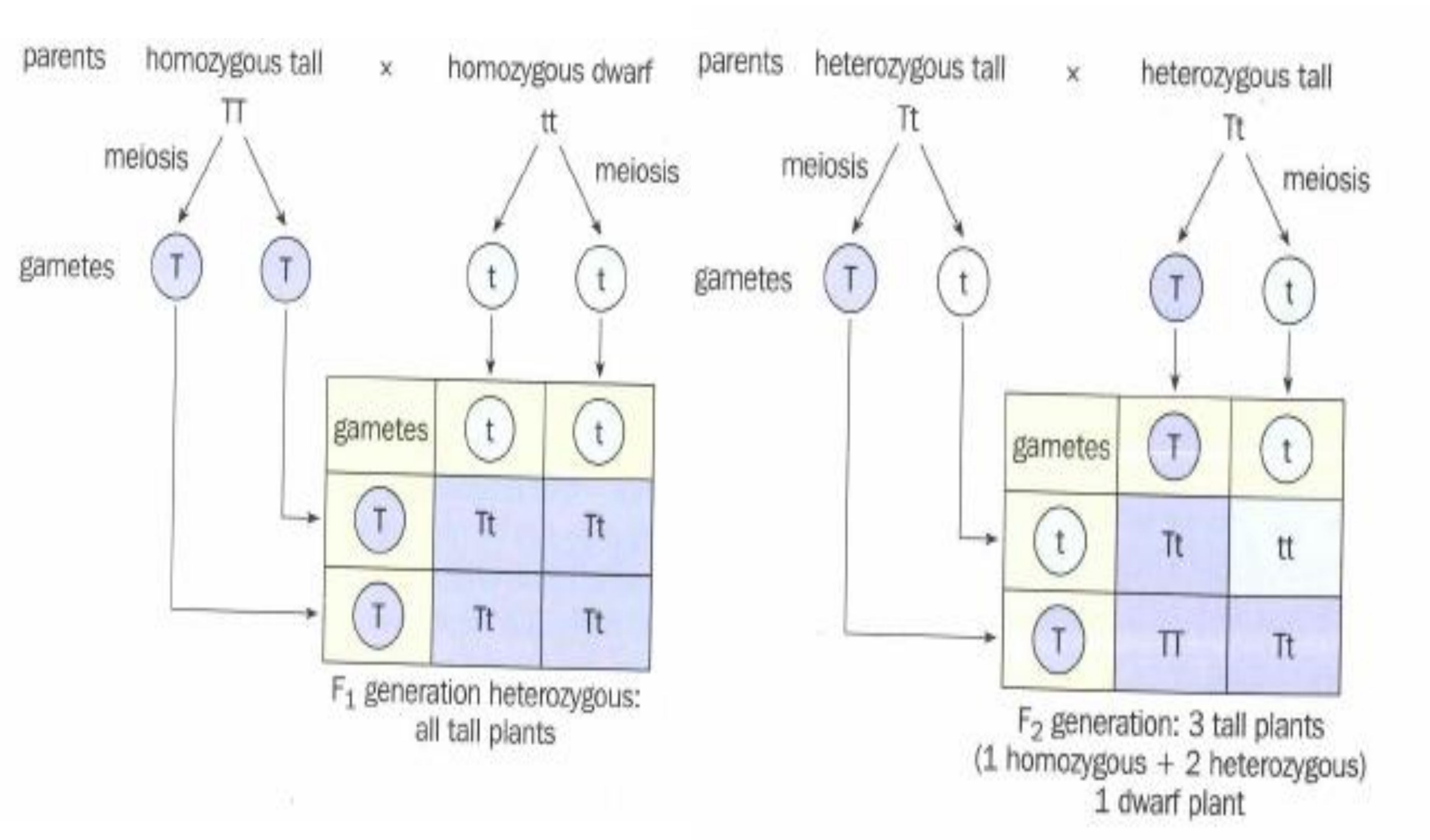
Conclusion 3

- As all the plants in the F1 generation were tall we can conclude that the allele for shortness was somehow swamped by the allele for tallness. The tallness is the **dominant** characteristic and the shortness is described as **recessive** because it does not exert its effects in the heterozygote.

Mendel's first law of inheritance – Law of Segregation

Alleles segregate so that only one of a pair of alleles is transmitted via each gamete.

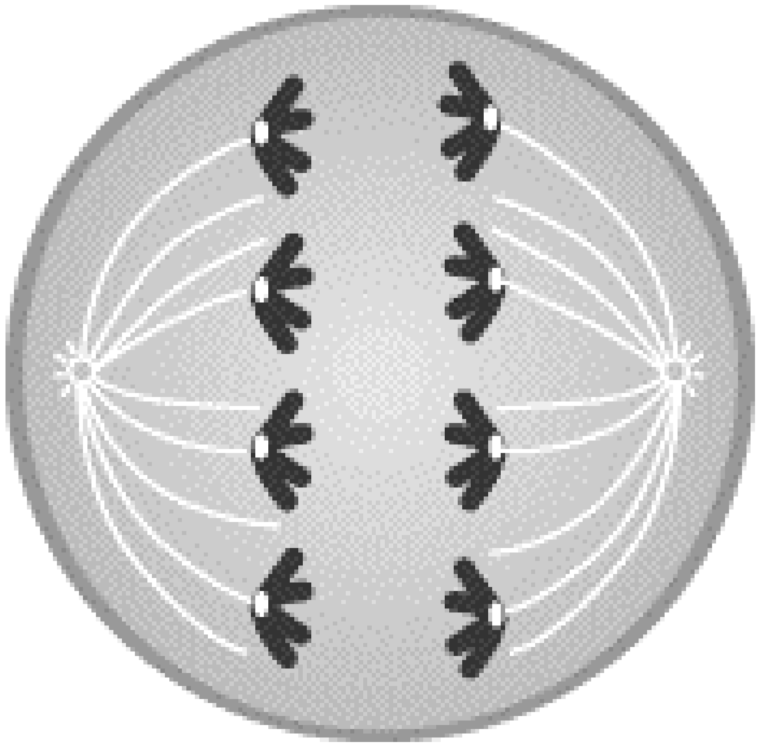
Mendel’s monohybrid cross



Law of segregation further explained

- In sexual reproduction, new individuals develop from a zygote produced by the fusion of male and female gametes. Since gametes are haploid (possess only one set of chromosomes) they only contain one allele of each gene. Thus, while an organism has a pair of alleles for any genetic condition in the somatic cells, only one allele of the pair is passed on via one gamete. If an individual is homozygous, (for example TT in tall plants as shown above) then there can only be one type of gamete produced. i.e "T". If the individual was homozygous tt, then only "t" gametes could be produced. If an individual is heterozygous e.g. Tt, then half the gametes produced will contain one type of allele (T) and half will contain the other (t). This segregation can be explained by the separation of homologous chromosomes, carrying the alleles, during anaphase 1 of meiosis.

- During fertilisation, the gametes combine to form a zygote and the individual alleles are restored to a pair: one from female parent, the other from the male parent. Fertilisation is a random event as any of the gametes from the female parent can combine with any of the gametes from the male.



Alternative patterns of inheritance

1. Dominance

- Mendel's experiments on pea plants revealed the principles of inheritance. Regardless of what feature he was testing for, he bred plants through two generations and in large numbers so that reliable ratios could be calculated. One experiment crossed white-flowered and purple-flowered plants. He found that the **F1** (First generation offspring) were all purple (we now know that this was due to a dominant allele) but that when the F1 were interbred the **F2** (second generation) showed a mixture of purple and white-flowered plants in a ratio of 3:1.

- If **P** codes for the production of purple pigment and **p** coding for no pigment, then fill out the following table with the possible genotypes and phenotypes of flower colour in pea plants.

Genotypes	**Phenotypes**
PP	Purple flowers
Pp	Purple flowers
pp	White flowers

Using these combinations Mendel carried out the following experiment to give an understanding of Dominance.

Parental phenotype	Purple flowers	White flowers	Plants from pure-breeding lines are cross-pollinated
Parental genotype			Both are homozygous
Parental gametes	(P)	(p)	Gametes are haploid, so contain only one allele from a pair — only one type of gamete from each parent
F_1 genotype	Pp		All F_1 plants are heterozygous with purple flowers — purple allele is dominant
F_1 gametes	(P) (p)	(P) (p)	All F_1 plants can produce two types of gamete — half with the purple flower allele and half with the white flower allele (two sets are shown to represent male and female gametes)
Fertilisations possible and F_2 genotypes			This is the standard way of showing all the possible fertilisations and possible combinations of alleles in the F_2 generation
F_2 phenotypes			In this instance, three of the possible four combinations contain at least one dominant allele and so have purple flowers, while only one of the four has two recessive alleles to give white flowers

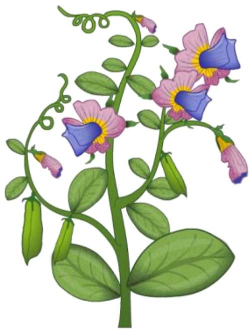
	(P)	(p)
(P)	PP	Pp
(p)	Pp	

Figure 25 A cross between pure-breeding white-flowered and pure-breeding purple-flowered pea plants

Test cross

To determine whether an organism showing the dominant characteristic of a trait is homozygous or heterozygous, a **test cross** can be performed. This involves crossing the organism with another that is homozygous recessive for the trait.

TT or Tt?



×

tt

if TT

	T	T
t	Tt	Tt
t	Tt	Tt

all tall

if Tt

	T	t
t	Tt	tt
t	Tt	tt

2 tall, 2 short

If any of the offspring show the homozygous recessive trait in the phenotype, the parent must have been heterozygous.

<u>**Test Crosses**</u>

- Sometimes it is difficult to know what the genotype of a plant may be by simply looking at it. For example in Mendel's experiments the purple flowers which he used may have had the genotype PP or Pp. To ascertain what the genotype is, a **test cross** has to be carried out. This is simply crossing with the homozygous recessive individual and observing the offspring.

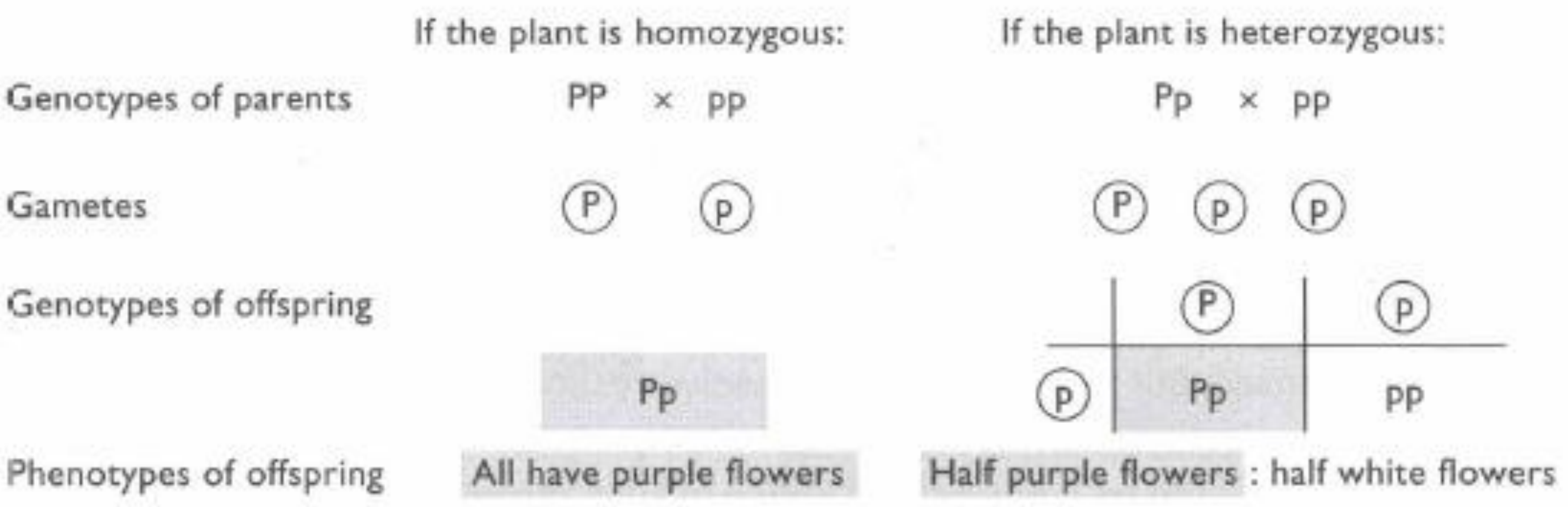


Figure 26 The test cross (testing the genotype of a purple-flowered plant)

- As can be seen from the results above if the offspring produce no variation, then the parent genotype is **homozygous dominant** (e.g PP). If there is variation within the offspring produced then the genotype of the parent is **heterozygous** (e.g Pp)