CHAPTER 11.1 – MONOHYBRID INHERITANCE

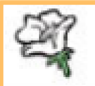
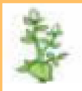
Definition of monohybrid inheritance

- Monohybrid inheritance involves inheritance of one characteristic and contrasting traits controlled by a gene

Mendel experiments

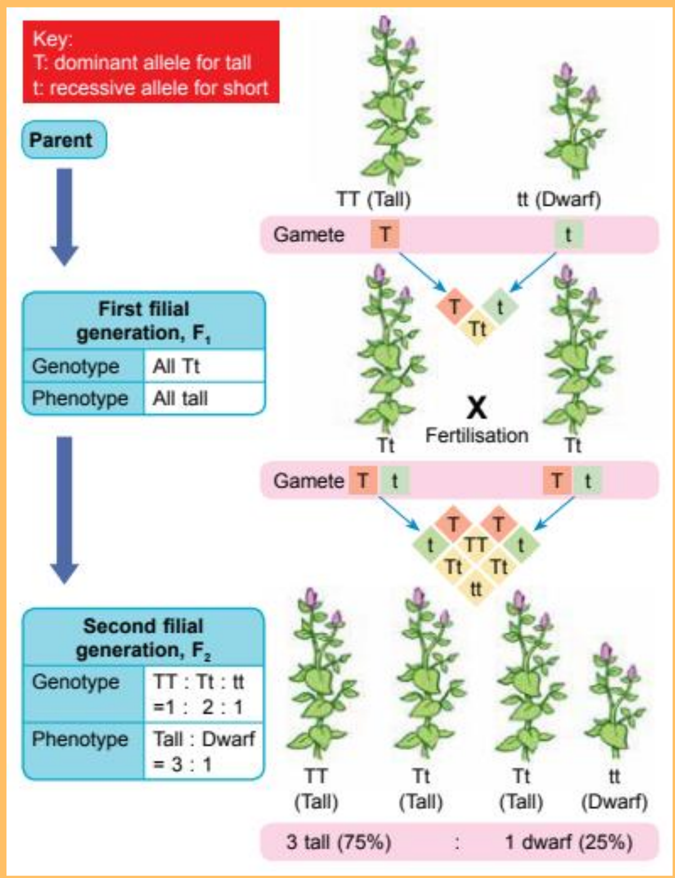
- ✓ Mendel figured that pea plants, *Pisum sativum*, possessed seven different characteristics
- ✓ Pea plants have contrasting traits which are obvious and easy to identify
- ✓ Mendel suggested in his theory that a characteristic in a parent plant is passed down to its next generation through male gametes and female gametes
- ✓ This shows that the nuclei of the male and female gametes carry genetic materials of the inherited characteristic
- ✓ The inheritance factor which determines a characteristic is known as a gene
- ✓ In the monohybrid inheritance experiment, Mendel used purebreed pea plants as the parental generation to study the inheritance of pea plant traits
- ✓ Purebreed pea plants are produced through self-cross plant which have similar traits to that of its parent plant in terms of genetic contents
- ✓ Therefore, self-cross of purebreed tall pea plants produce only tall offsprings
- ✓ In the dominance principle, Mendel explained that in a pea plant with a pair of contrasting traits, its trait is determined by a dominant inheritance factor whereas another contrasting trait is determined by a recessive inheritance factor
- ✓ The dominant inheritance factor suppresses the effect of the recessive inheritance factor
- ✓ Hence, the recessive trait is not visible although its inheritance factor exists together with the dominant inheritance factor in a pea plant

SEVEN CHARACTERISTICS STUDIED BY MENDEL

CHARACTERISTICS	TRAITS	
	DOMINANT	RECESSIVE
Seed shape	 Round	 Constricted
Seed colour	 Yellow	 Green
Pod shape	 Inflated	 Constricted
Pod colour	 Green	 Yellow
Flower colour	 Purple	 White
Flower position	 Axial	 Terminal
Plant height	 Tall	 Dwarf

MONOHYBRID CROSS CARRIED OUT BY MENDEL

- ❑ Mendel crossed a purebreed tall (TT) pea plant with a purebreed dwarf (tt) pea plant
- ❑ The cross product between the two purebreed plants was the F_1 generation which consisted of only tall (Tt) pea plants
- ❑ This shows that the tall trait (T) is dominant whereas the dwarf trait (t) is recessive
- ❑ Mendel then crossed the F_1 generation by self-pollination (Tt × Tt)
- ❑ The ratio of tall plant to dwarf plant in F_2 generation was 3:1



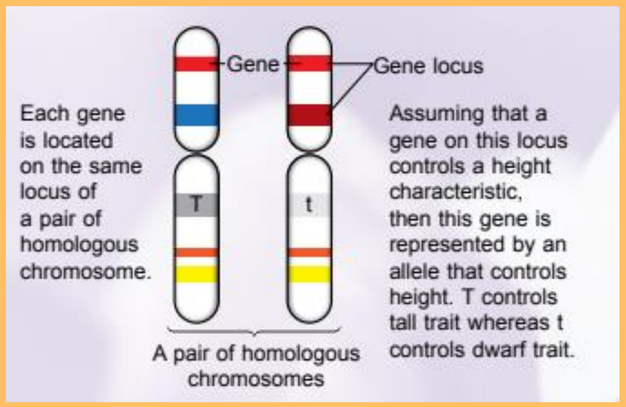
Terms related to inheritance

GENES

- ⊕ A gene is the basic unit of inheritance which consists of a DNA segment located on a specific locus of a chromosome
- ⊕ A gene controls a specific characteristic of an organism

ALLELES

- An allele is an alternative form of a gene for a specific trait that is located on the same locus of a pair of homologous chromosomes



CHARACTERISTICS

- A characteristic is a heritable feature (height, eye colour, blood group and presence of dimples)
- Each characteristic is a feature of an organism

TRAITS

→ A trait is a variation of a specific characteristic

→ Each inherited characteristic consists of a specific trait

→ For example, height is a characteristic whereas tall or dwarf is a trait

CHARACTERISTICS AND TRAITS

CHARACTERISTIC	TRAIT	GENOTYPE	PHENOTYPE
Height	Tall	TT, Tt	Tall
	Dwarf	tt	Dwarf
Colour	Red	RR, Rr	Red
	White	rr	White
Blood group	A	I^AI^A, I^AI^O	A
	B	I^BI^B, I^BI^O	B
	AB	I^AI^B	AB
	O	I^OI^O	O

PHENOTYPES

» Phenotype is the observable characteristic of an organism

» For example, height

GENOTYPES

- Genotype is the genetic composition of an organism that cannot be seen
- For example, TT and Tt (genotypes for tall); tt (genotype for dwarf)

DOMINANT ALLELES

Δ Dominant allele is an allele which always shows its trait when it is present, and suppresses the effect of recessive allele

Δ It is represented by a capital letter

Δ For example, B

RECESSIVE ALLELES

- Recessive allele is an allele which shows its trait when both alleles are recessive allele

- The effect of recessive allele is suppressed by the presence of dominant allele
- It is represented by a small letter
- For example, b

HOMOZYGOTES

- Both alleles at loci of a pair of homologous chromosomes are the same
- For example, BB or bb

HETEROZYGOTES

- Alleles at loci of a pair of homologous chromosomes are different
- For example, Bb

PARENTAL GENERATIONS

- Parental generation refers to the first generation of two individuals which are mated to predict or analyse genotypes of their offsprings

FILIAL GENERATIONS

- Filial generation refers to a successive generation as a result of mating between individuals of purebreed parental generation

DOMINANT TRAITS

- Dominant trait is expressed when both alleles are dominant alleles or one dominant allele is paired with a recessive allele
- For example, BB or Bb

RECESSIVE TRAITS

- Recessive trait is expressed if a recessive allele is paired with another recessive allele
- For example, bb

PUREBREEDS

- Purebreed refers to individual which carries two identical alleles for a trait
- Self-cross always produces offsprings with the same characteristics in every generation

HYBRIDS

- Hybrid is the product of mating between two purebreed varieties

Inheritance of flower colour

- If allele for the dominant purple trait is labelled B (capital letter), then the allele for recessive white trait is labelled b (small letter)
- Therefore, each purebreed parent has two identical alleles, either BB for purple flower or bb for white flower
- During formation of gametes, homologous chromosomes separate during meiosis and produce gametes that carry one B allele from purple flower and one b allele from white flower
- Fertilisation between a gamete which carries B allele and a gamete which carries b allele produces offsprings with genotype Bb in the first filial generation (F_1)

- Since B allele is dominant, the Bb genotype combination expresses only phenotype with purple flower
- The effect of recessive allele is suppressed by the presence of the dominant allele
- If the first filial generation is self-crossed, the second filial generation (F_2) will have offsprings with BB, Bb and bb genotypes
- BB and Bb genotypes express phenotype with purple flower whereas bb genotype expresses phenotype with white flower
- The trait that is not observed in the F_1 generation (white flower colour) reappears in the F_2 generation
- The above explanation for inheritance of flower colour can be explained in the form of a schematic diagram of inheritance

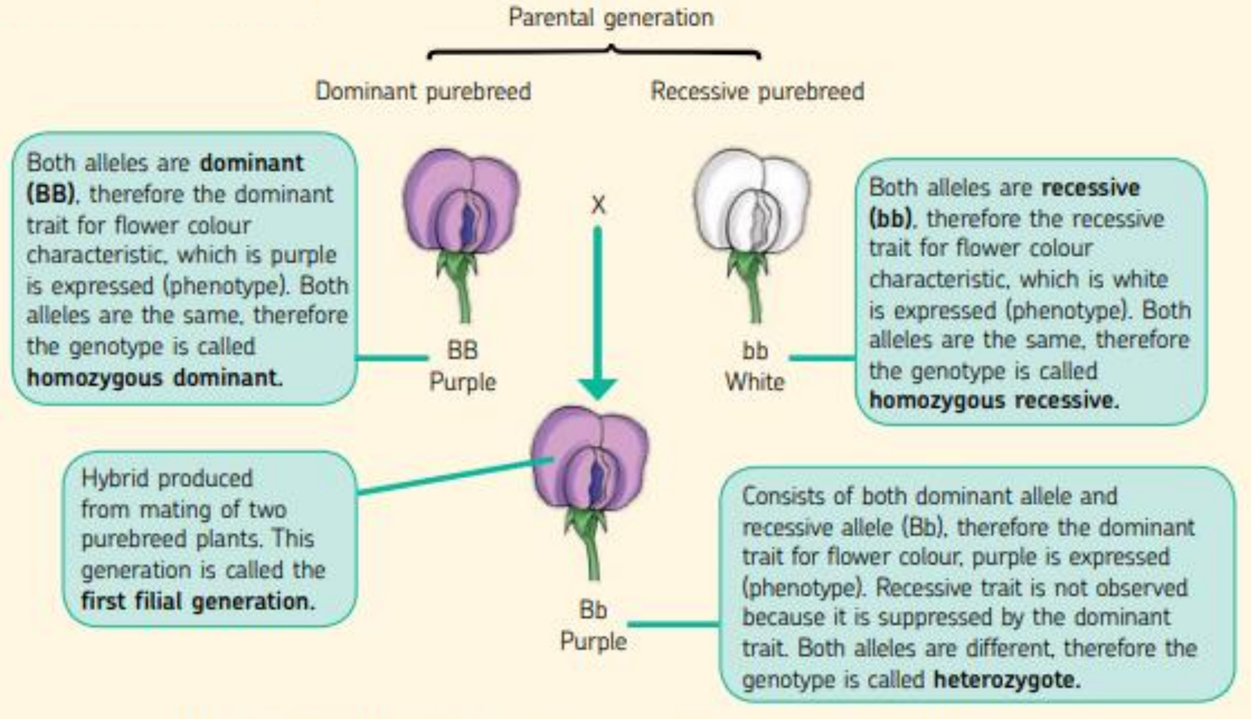
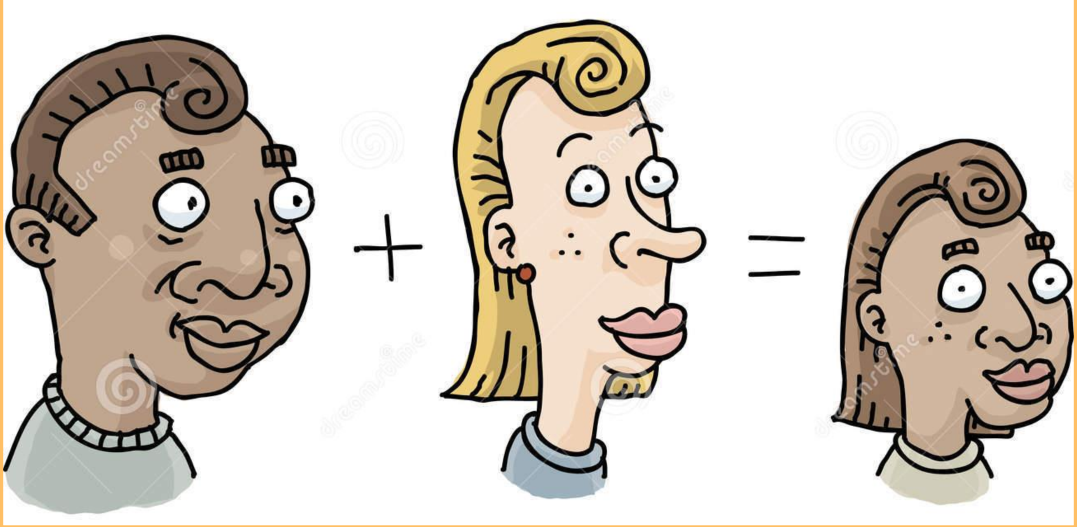
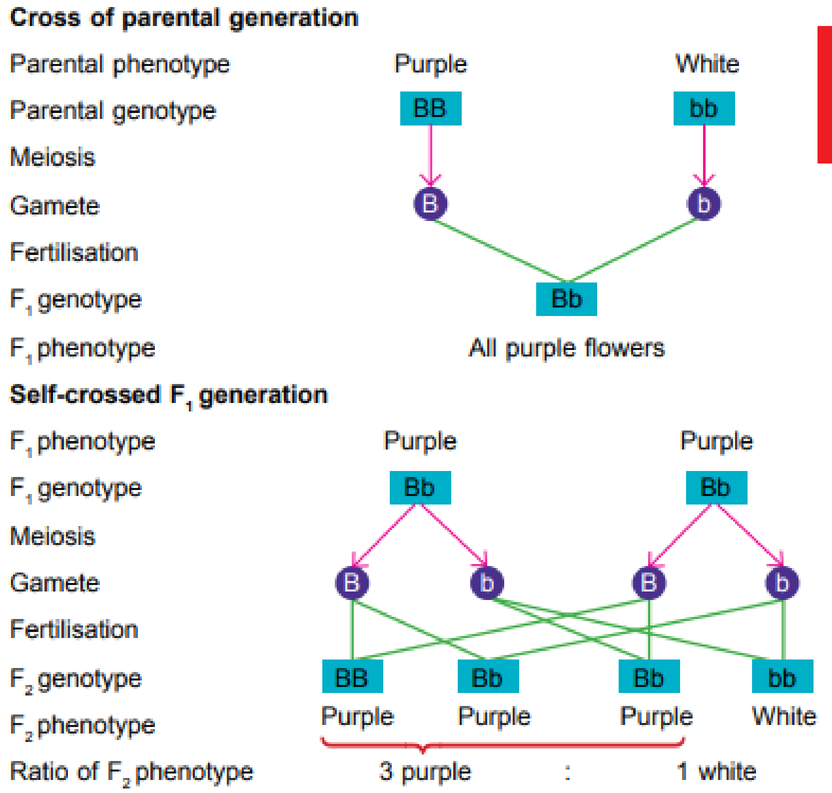


Figure 11.4 Relationship among a few important terms in genetics





Key:
B: dominant allele for purple trait
b: recessive allele for white trait

Bio Exploration

Based on monohybrid cross, Mendel obtained a phenotypic ratio of 2.9:1 in F_2 generation which is close to the ratio of 3:1 if the number of pea plants used was big. Small sample size causes the ratio to be less accurate. In a natural environment, the ratio obtained is dependent on chance and probability. Therefore, the actual ratio of offsprings produced is probably different from the expected ratio.

Figure 11.5 Monohybrid schematic diagram for flower colour characteristic

- For monohybrid inheritance, the genotypic ratio in F_2 generation is 1 BB : 2 Bb : 1 bb; whereas the phenotypic ratio in F_2 generation is 3 purple : 1 white
- Punnett square can also be used to predict the ratios and probabilities of genotype and phenotype in the offsprings produced. Boxes in the Punnett square represent different allelic combinations of zygote or offsprings that are produced

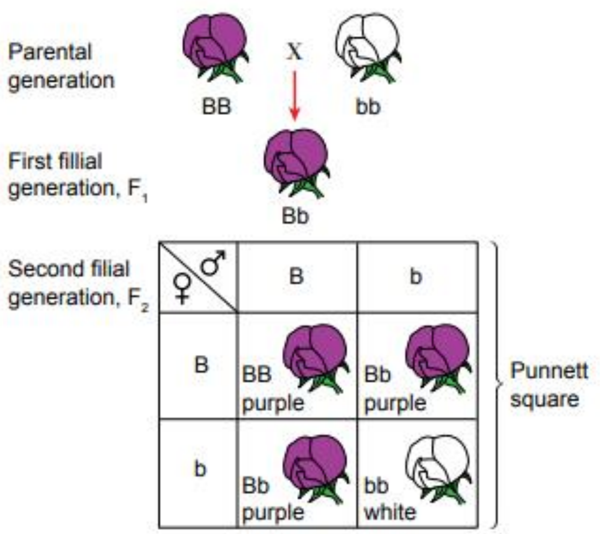


Figure 11.6 Monohybrid cross using Punnett square

Mendel's First Law

→ Law of Segregation or Mendel's First Law states that a characteristic of an organism is controlled by a pair of alleles, and only one of the allelic pair is inherited in a gamete

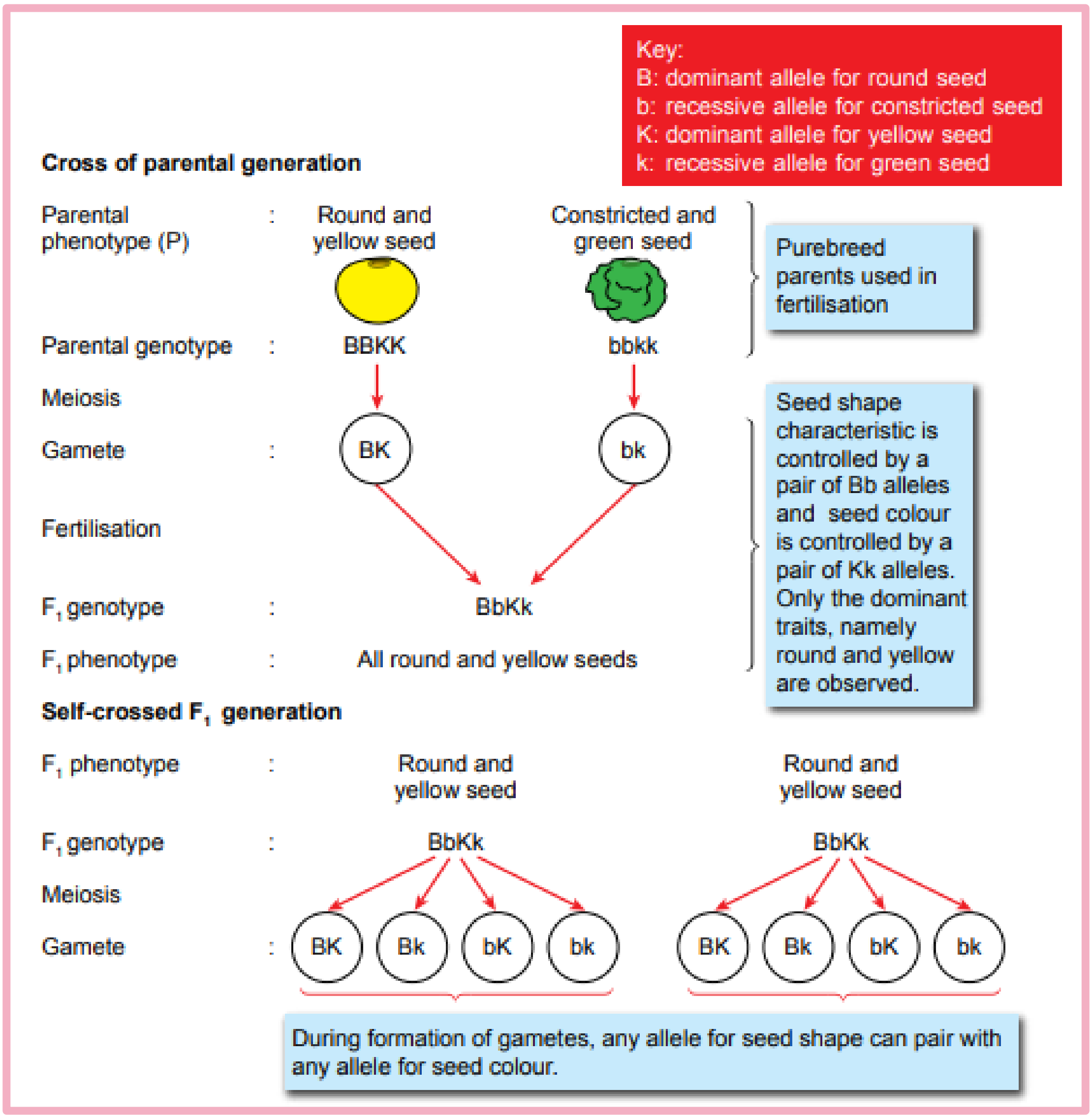
SUMMARY OF MENDEL MONOHYBRID EXPERIMENT

- » A characteristic inherited from a parent to the next generation is determined by a genetic factor, which is now known as gene
- » A trait is controlled by a pair of genetic factors known as allele
- » A pair of alleles segregates (separates) randomly during formation of gametes and only one allele from the pair is found in a single gamete
- » During fertilisation, a zygote formed possesses two alleles (one allele from each parent) for a specific characteristic
- » Fertilisation is random
- » Genotypic combination which is homozygous dominant shows dominant trait whereas, homozygous recessive shows recessive trait
- » Heterozygous genotype (combination of one dominant allele and one recessive allele) shows dominant trait

CHAPTER 11.2 – DIHYBRID INHERITANCE

Definition of dihybrid cross

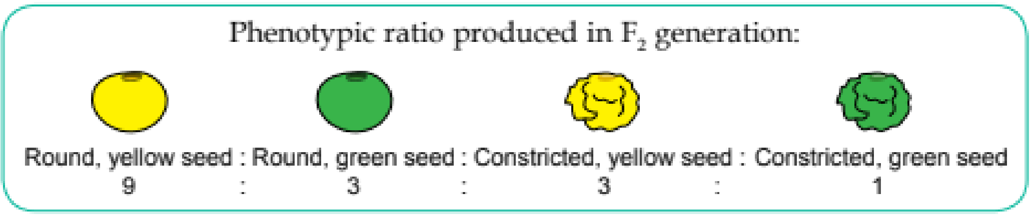
- Dihybrid inheritance involves inheritance of two characteristics, each characteristic is controlled by a different gene located at a different locus
- A schematic diagram below of a dihybrid inheritance between two purebreed parents to study the characteristics of pea, namely seed colour and seed shape. The two purebreed parents either have a pair of homozygous dominant alleles or a pair of homozygous recessive alleles
- Fertilisation between the two purebreed parents produce offsprings in F_1 generation with the dominant traits of round and yellow seeds



- When the F_1 generation produced from dihybrid cross was self-crossed, 16 F_2 genotype combinations are produced
- The cross shows four phenotypic characteristics in F_2 generation
- Outcome of the dihybrid cross is presented in a Punnett square

Female gamete \ Male gamete	BK	Bk	bK	bk
BK	BBKK Round, yellow	BBKk Round, yellow	BbKK Round, yellow	BbKk Round, yellow
Bk	BBKk Round, yellow	BBkk Round, green	BbKk Round, yellow	Bbkk Round, green
bK	BbKK Round, yellow	BbKk Round, yellow	bbKK Constricted, yellow	bbKk Constricted, yellow
bk	BbKk Round, yellow	Bbkk Round, green	bbKk Constricted, yellow	bbkk Constricted, green

Figure 11.8 Dihybrid cross of F_1 generation using Punnett square



Mendel's second law

- Law of Independent Assortment or Mendel's Second Law states that during gamete formation, each allele from a pair of alleles can combine randomly with any allele from another pair of alleles

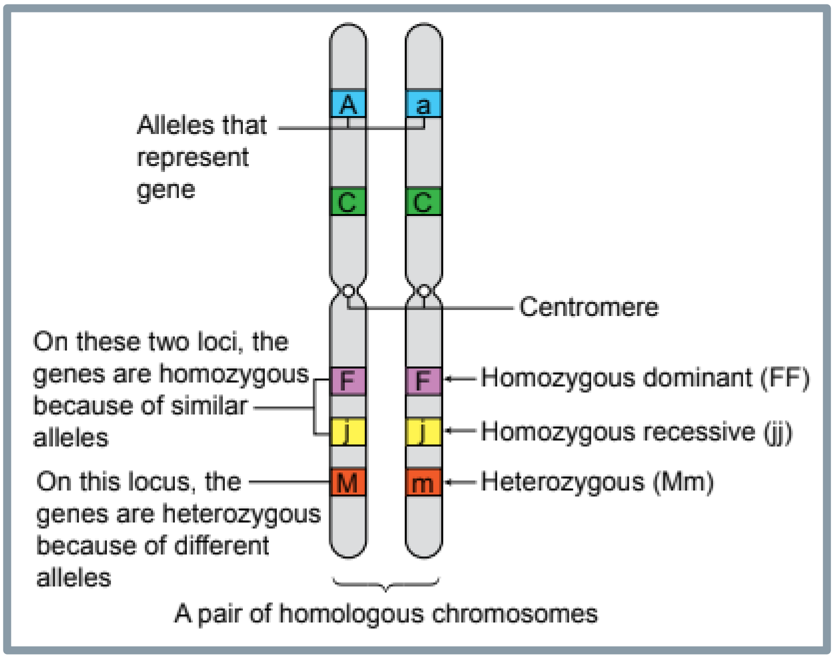
SUMMARY OF MENDEL DIHYBRID EXPERIMENT

- New combinations of characteristics are produced in the F_2 generation namely constricted yellow seed and round green seed
- Two characteristics (seed shape and colour) are combined in F_1 generation but later they separate and react freely in F_2 generation

CHAPTER 11.3 – GENES AND ALLELES

Definition of locus

- Locus is a specific location of a gene in a chromosome
- Each chromosome carries many genes
- In human, the number of genes that code for proteins in a set of haploid chromosomes is estimated to be 25 000
- Allele which represents a gene is located at the same locus as the gene
- Below diagram shows five genes with their respective alleles on specific loci of a pair of homologous chromosomes



CHAPTER 11.4 – INHERITANCE IN HUMANS

Human chromosomes

- There are two types of human chromosomes, namely autosomes and sex chromosomes
- Human somatic cell consists of 44 autosomes and 2 sex chromosomes
- Autosomes vary in terms of size and length

The types of human chromosomes

AUTOSOME

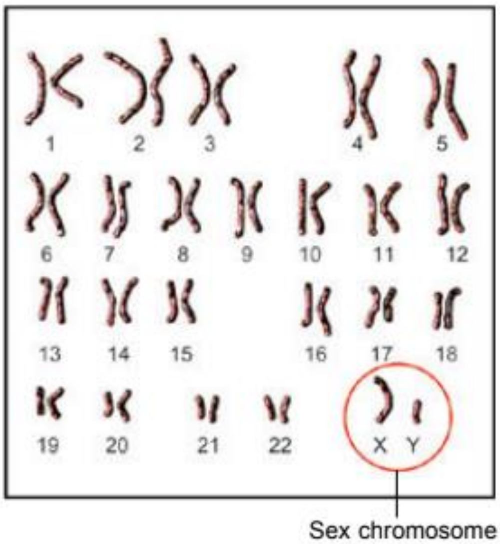
- Consists of chromosome pairs from number 1 to 22
- The function is to controls all characteristics of somatic cells
- Examples:
  1) Types of blood group
  2) Height
  3) Skin colour

SEX CHROMOSOME

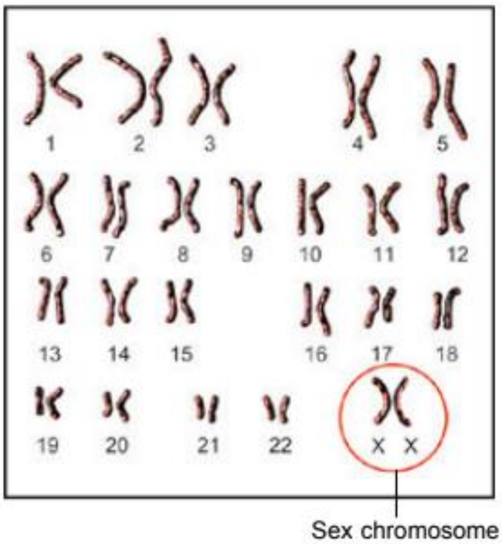
- Consists of one chromosome pair, that is number 23
- Consists of genes which determine gender
- Examples:
  1) Male has XY chromosomes
  2) Female has XX chromosomes

Human karyotype

- The number and structure of chromosomes present in a cell nucleus is known as karyotype
- Chromosomes are arranged in pairs, based on homologous chromosomes in terms of their sizes, centromere locations and banding pattern of chromosomes



Male karyotype (44 + XY)



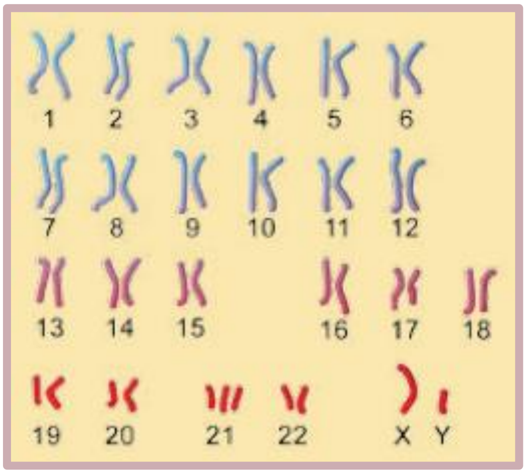
Female karyotype (44 + XX)

Nondisjunction

- Changes in number of chromosomes can occur due to failure of homologous chromosomes to separate during anaphase I or failure of sister chromatids to separate during anaphase II
- This disorder is known as nondisjunction which can occur in some chromosomes
- When nondisjunction occurs in humans, either male gamete (sperm) or female gamete (ovum) can possess chromosome number of less than 23, that is 22 or more than 23, which is 24
- Therefore, fertilisation that involves the abnormal gamete with a normal gamete produces a zygote with 45 chromosomes or 47 chromosomes
- Examples of genetic diseases caused by nondisjunction are Down syndrome, Turner syndrome and Klinefelter syndrome

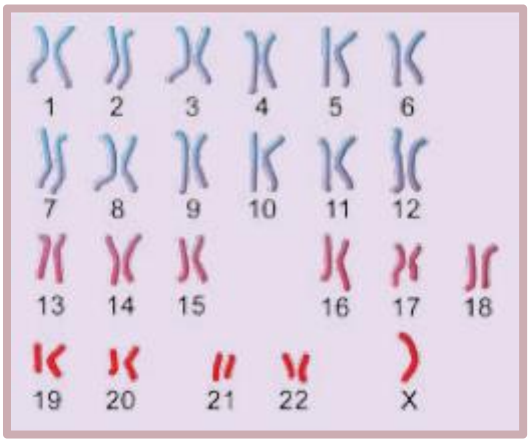
DOWN SYNDROME

- Total chromosome number is 47, which is 45 + XY
- There is an extra chromosome for chromosome pair number 21
- Down syndrome is also known as trisomy 21
- Down syndrome can occur in both males and females



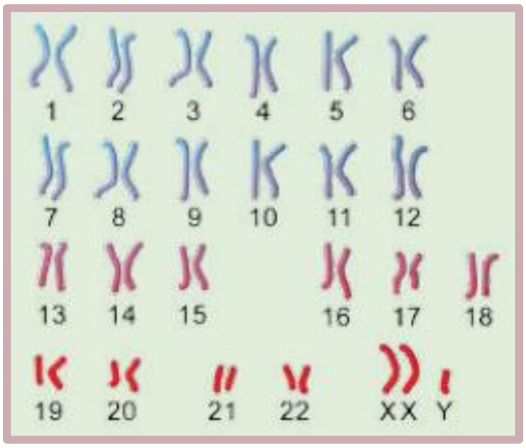
TURNER SYNDROME

- In Turner syndrome, total number of chromosomes is 45, which is 44 + XO
- There is a missing X chromosome in the pair of sex chromosomes
- The gender of individual with Turner syndrome is a female



KLINEFELTER SYNDROME

- Karyotype of Klinefelter syndrome has a total of 47 chromosomes, that is 44 + XXY
- There is an extra X chromosome in the pair of sex chromosomes
- The gender of individual with Klinefelter syndrome is male
- However, his secondary sex characteristics are not well-developed



Human inheritance

ABO blood groups

- ABO blood group in humans is an example of multiple alleles
- Blood group is controlled by a gene which consists of three different alleles, namely allele I^A, I^B and I^O
- These alleles determine the types of antigens present on the surface membrane of red blood cells
- However, a person only possesses two alleles to determine his/her blood group
- Both I^A and I^B are dominant alleles whereas I^O is recessive allele
- Therefore, a combination of I^A and I^O (I^AI^O) alleles expresses a group A blood phenotype whereas I^BI^O expresses a group B blood phenotype
- I^A and I^B alleles are codominant to one another
- When these two alleles are present together, effects of both alleles show
- A combination of both alleles gives an AB blood group phenotype

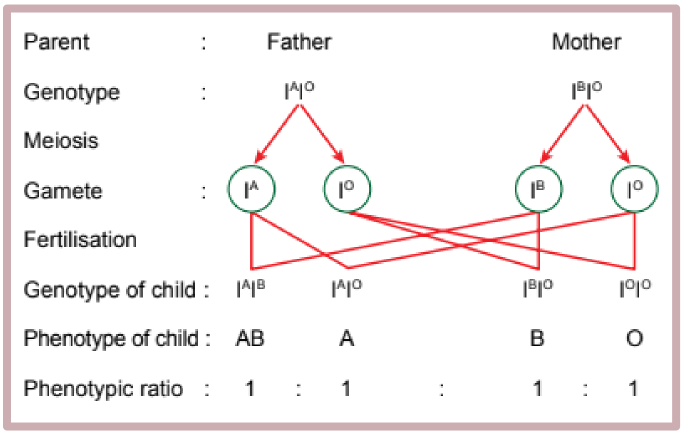
PHENOTYPE AND GENOTYPE OF HUMAN BEING BLOOD GROUP

PHENOTYPE (blood group)	GENOTYPE
A	I^AI^A or I^AI^O
B	I^BI^B or I^BI^O
AB	I^AI^B
O	I^OI^O

Key:
I^A and I^B: dominant allele
I^O: recessive allele

EXAMPLE OF BLOOD GROUP INHERINTANCE IN HUMANS

- A man with A blood group married a woman with B blood group
- There is a probability of the couple in getting a child with O blood group
- Both mother and father are heterozygous for A blood group and B blood group



Rhesus factor

→ Besides antigen A and antigen B on the surface of human red blood cell, there is another antigen called antigen D which is known as Rhesus factor (Rh)

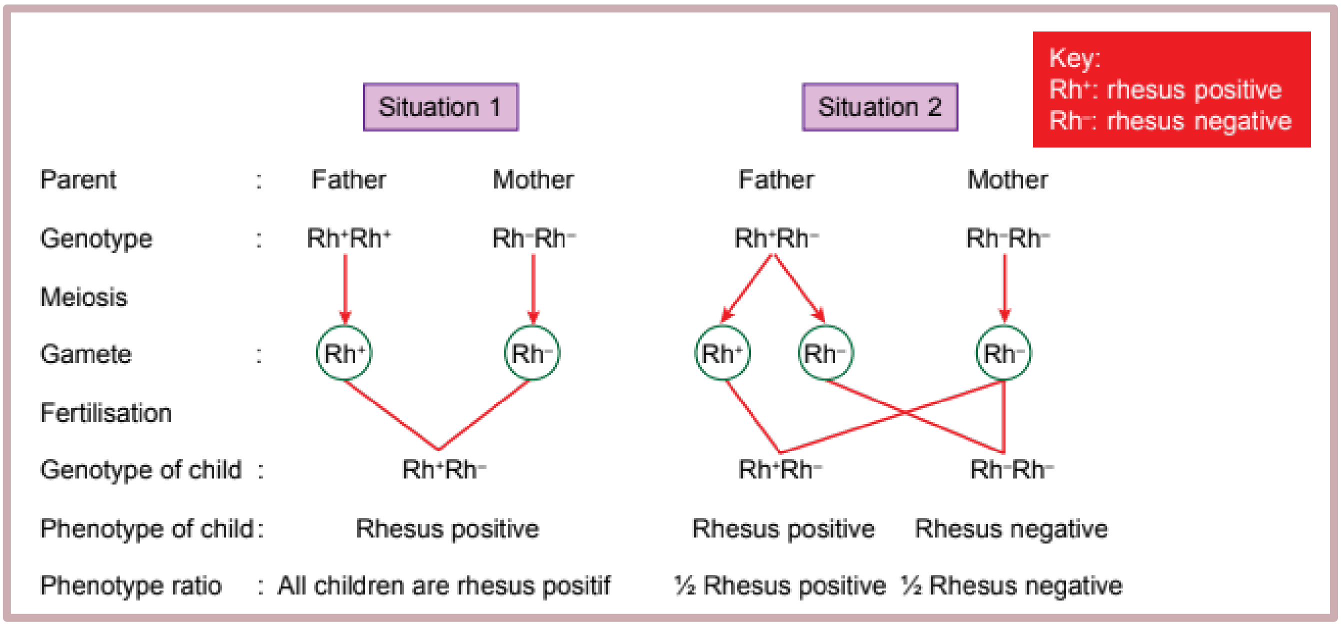
→ An individual whose red blood cell has Rhesus factor is said to be Rhesus positive (Rh^+) whereas an individual without the Rhesus factor is said to be Rhesus negative (Rh^-)

→ Inheritance of Rhesus factor from parents to children is based on principles of Mendel's Law

→ Rhesus factor is controlled by genes which consists of a pair of alleles, namely Rh^+ dominant and Rh^- recessive

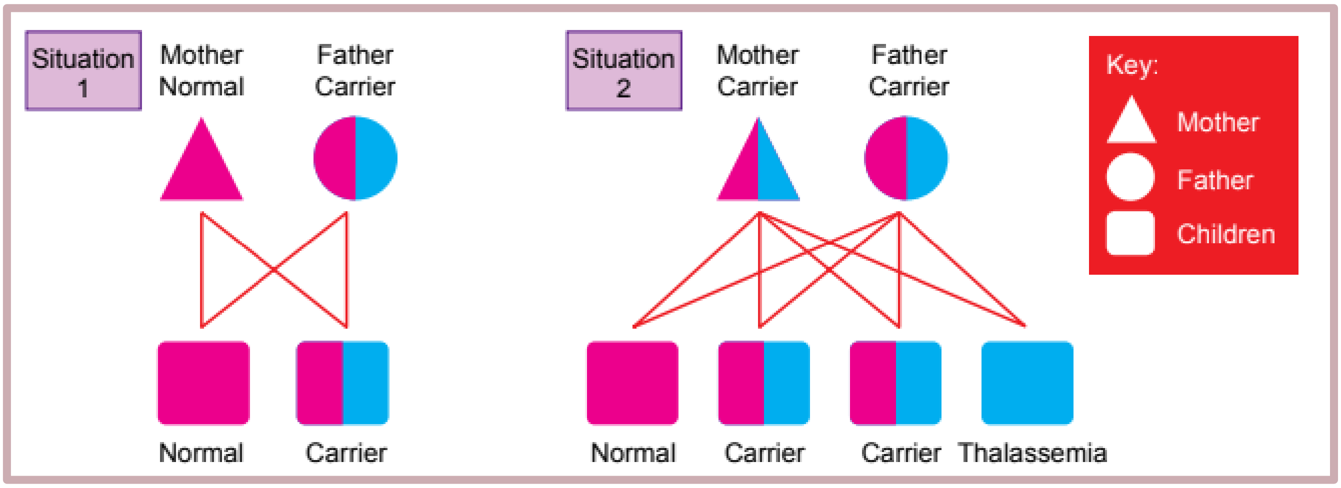
→ Genotype of a Rh-positive individual is either homozygous dominant (Rh^+Rh^+) or heterozygous (Rh^+Rh^-)

→ Rh negative individual is homozygous recessive (Rh^-Rh^-)



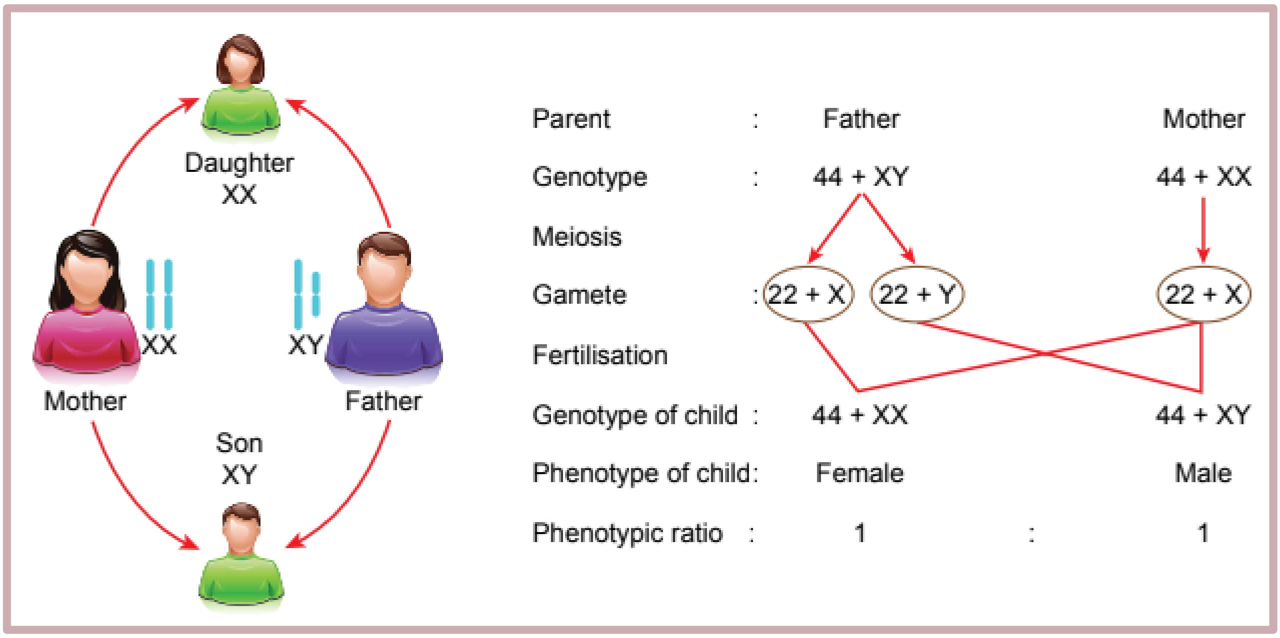
Thalassemia

- Thalassemia is an inherited disease
- The disease can be passed down from generation to generation
- Thalassemia is due to gene mutation on an autosome, that is on chromosome 11 or 16
- Thalassemia is due to the abnormality and low number of haemoglobins
- The red blood cell is smaller and paler
- Thalassemia carrier is said to have a thalassemia minor condition in which the individual possesses recessive allele of thalassemia but the individual does not show any symptoms of the disease
- Detection of thalassemia can only be confirmed by a blood test
- A thalassemia patient is said to have thalassemia major when the individual has both the recessive alleles
- A thalassemia patient shows symptoms such as tiredness, paleness, breathing difficulty and changes in facial bone formation from the age of 3 to 18 months



Sex determination

- ∆ A male has 44 + XY chromosomes and a female has 44 + XX
- ∆ Sperms produced in the testis are haploid, and each sperm has either 22 + X or 22 + Y chromosomes
- ∆ Secondary oocytes produced in the ovary are also haploid and each secondary oocyte has only one set of chromosomes, namely 22 + X chromosomes
- ∆ Sex or gender of a child is determined during fertilisation



Sex-linked inheritance

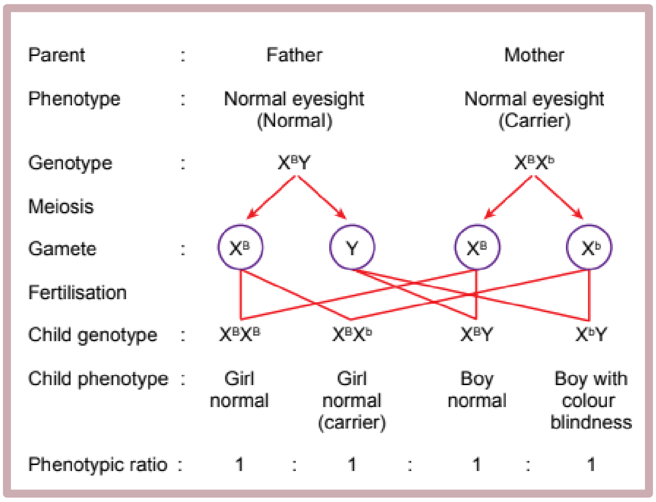
- Genes located on sex chromosomes which control specific characteristics but are not involved in sex determination are known as sex-linked genes
- Genes of colour blindness and haemophilia are located in the X chromosome
- These genes are called sex-linked genes
- Characteristics of colour blindness and haemophilia are caused by

recessive genes linked to X chromosome

- Y chromosome is shorter than X chromosome and does not contain as many alleles as X chromosome
- Therefore, any traits in males caused by either the dominant allele or recessive allele on chromosome X is observed

COLOUR BLINDNESS

- Colour blindness is a condition in which a person cannot differentiate some specific colours such as red and green
- Colour blindness is caused by the recessive allele found in the X chromosome and most people with colour blindness are males
- In sex-linked inheritance research, X and Y chromosomes must be shown when writing the genotypes
- Dominant allele is represented by a capital letter whereas recessive allele is represented by a small letter on the X chromosome



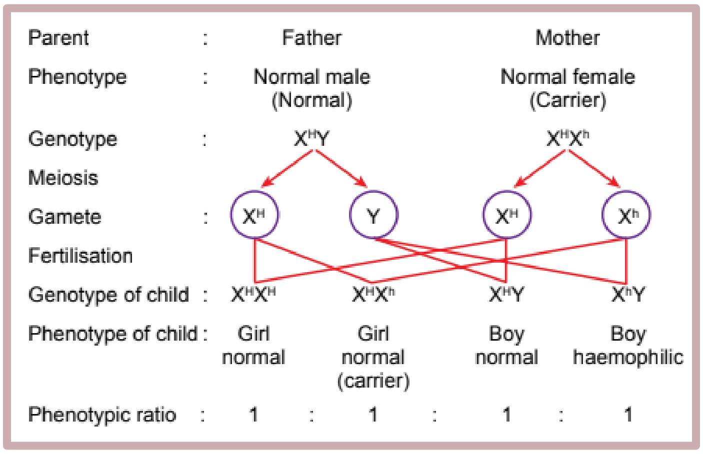
Genotype and phenotype of colour blindness inheritance

PHENOTYPE	GENOTYPE	
	MALE	FEMALE
Normal	X^BY	X^BX^B
Carrier	-	X^BX^b
Colour blind	X^bY	X^bX^b

Key:
X^B: dominant allele
X^b: recessive allele

HAEMOPHILIA

- Haemophilia is a condition in which blood cannot clot in normal circumstances due to the lack of blood clotting factor
- This can result in excessive internal or external bleeding which may be fatal
- Haemophilia is due to the presence of the recessive allele in the X chromosome, which causes the male to be haemophilic
- The female will only be haemophilic if both recessive alleles are present on both X chromosomes



Genotypes and phenotypes of haemophilic inheritance

PHENOTYPE	GENOTYPE	
	MALE	FEMALE
Normal	X^HY	X^HX^H
Carrier	-	X^HX^h
Haemophilic	X^hY	X^hX^h

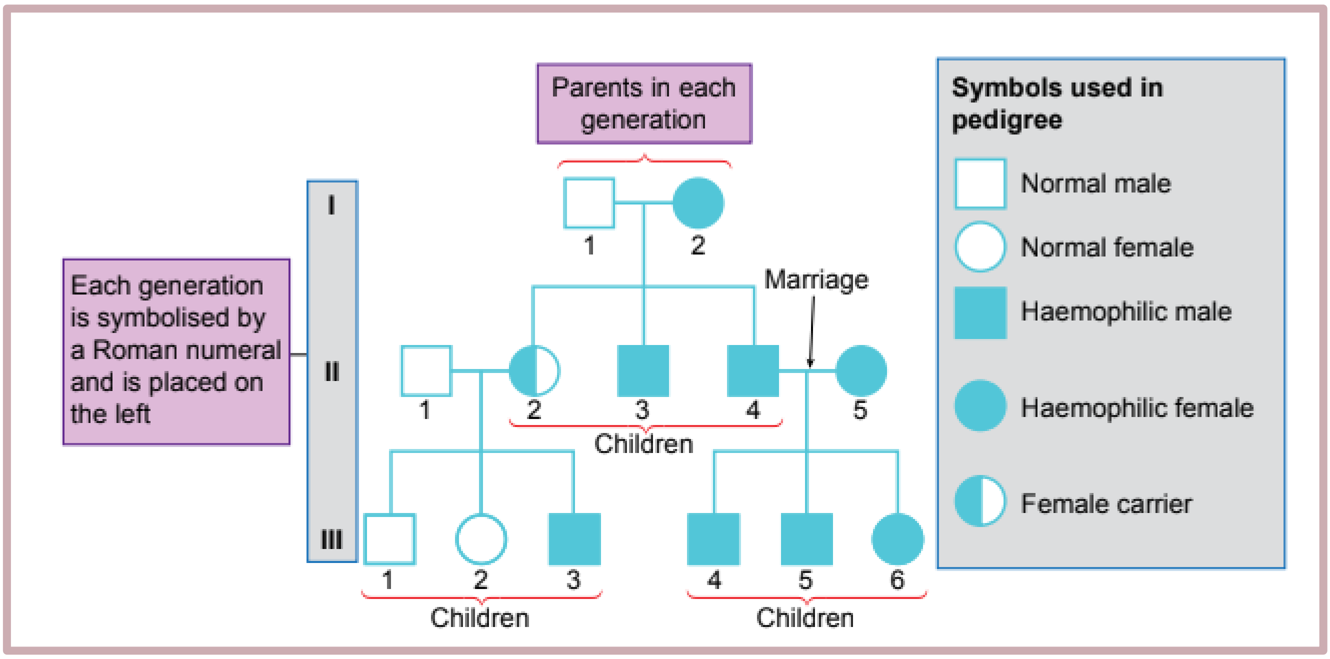
Key:
X^H: dominant allele
X^h: recessive allele

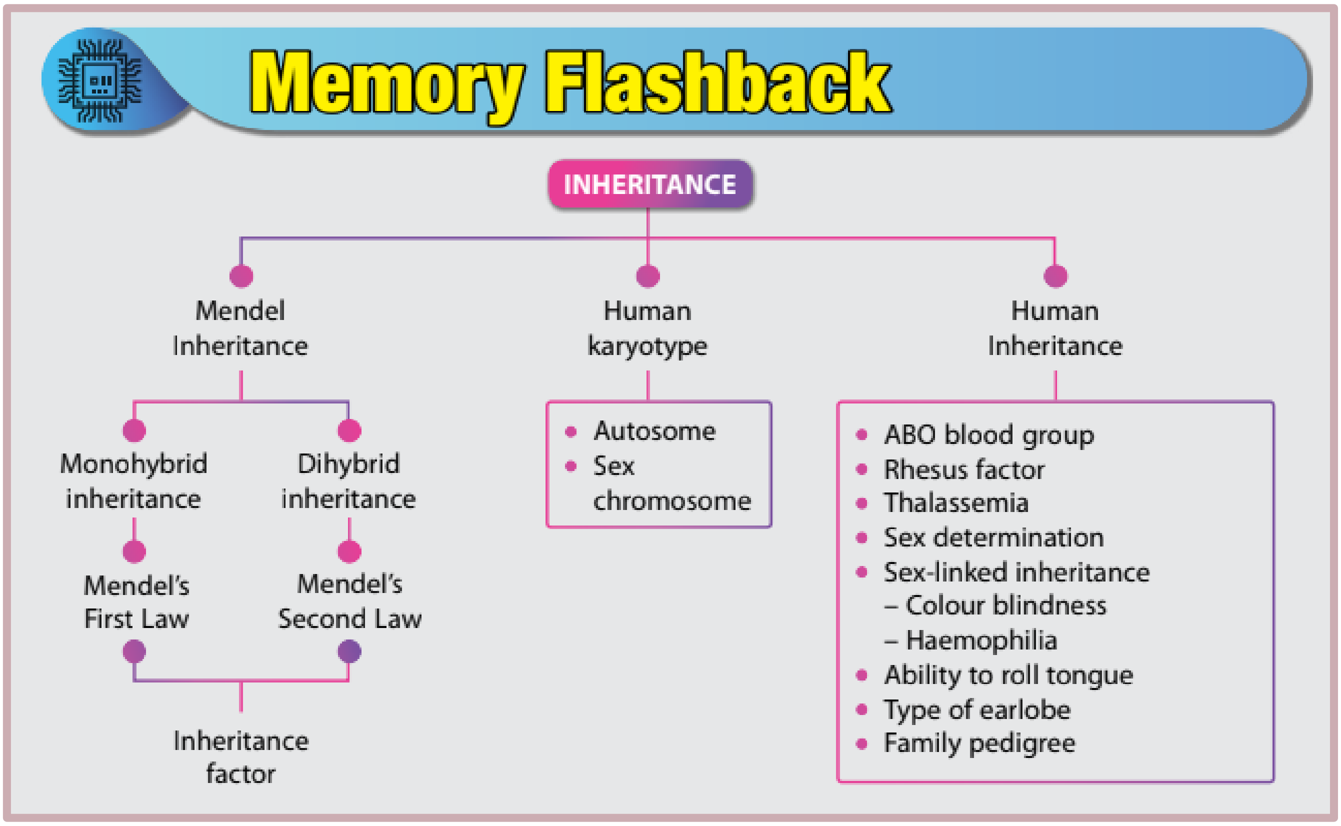
Ability to roll tongue and types of earlobes

- The ability to roll tongue and the types of earlobes are two characteristics that can be inherited from parents to children according to Mendel's Law
- Ability to roll tongue is a dominant trait
- Free earlobe is a dominant trait whereas attached earlobe is a recessive trait

Family pedigree

- Family pedigree or lineage can be analysed to investigate inheritance of human characteristics
- Family pedigree is a flowchart through a few generations to show ancestral relationship and inheritance of characteristics from ancestors to individuals in the present generation
- Analysis of family pedigree enables the geneticist to predict an inherited characteristic of interest and also to identify the features of dominant or recessive gene
- Normally a dominant gene appears in every generation whereas a recessive gene is probably hidden in certain generations




Memory Flashback
INHERITANCE
Mendel Inheritance
Human karyotype
Human Inheritance
Monohybrid inheritance
Dihybrid inheritance
Mendel's First Law
Mendel's Second Law
Inheritance factor
• Autosome
• Sex chromosome
• ABO blood group
• Rhesus factor
• Thalassemia
• Sex determination
• Sex-linked inheritance
– Colour blindness
– Haemophilia
• Ability to roll tongue
• Type of earlobe
• Family pedigree