Genetics 1 – From Genes to Proteins

1. Genetics is the study of how inheritable traits such as flower colour, eye colour are passed from one generation to another

a. the proteins made by our cells determine a vast number of characteristics

b. **genes** contain the information needed to make the protein

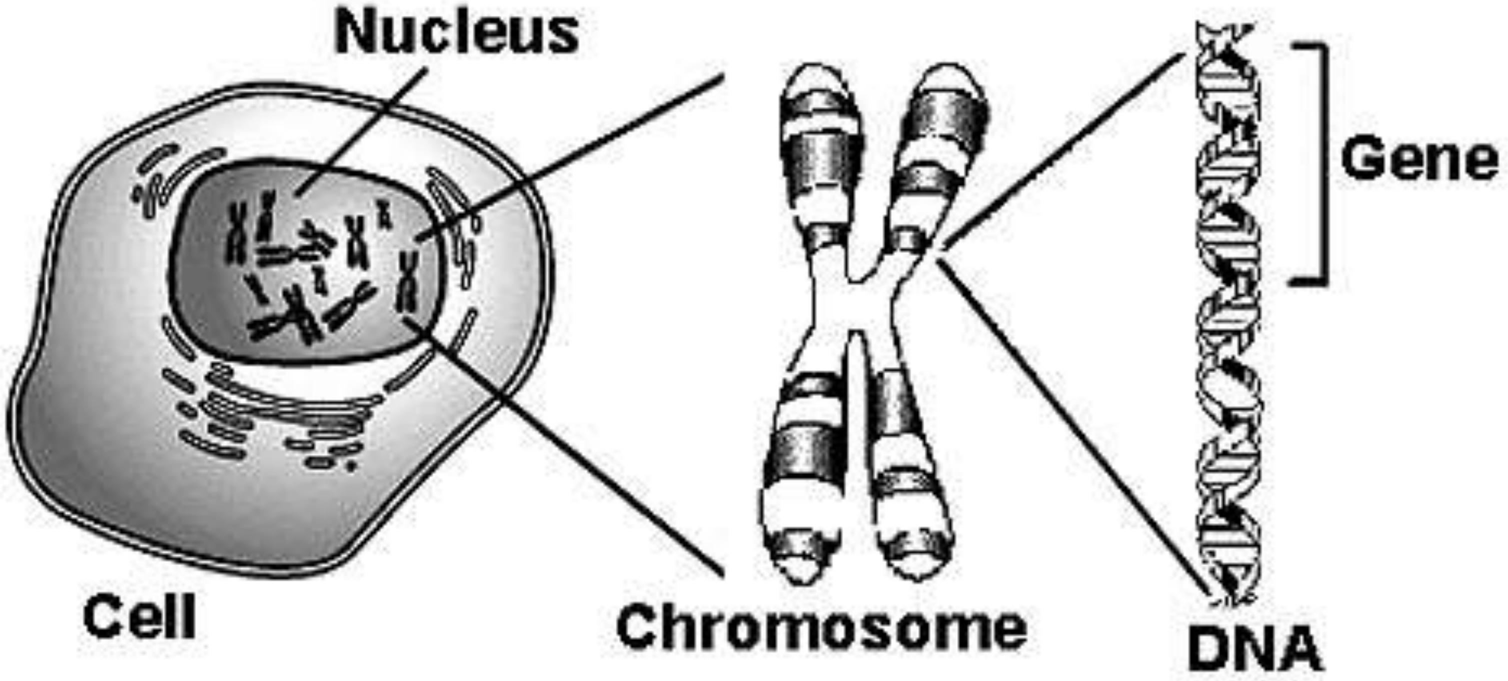
i. genes are what parents pass on to their offspring during reproduction

2. Molecular biologists study how genes work

a. In doing so, they have discovered the chemical language that is used to transmit the genetic information – this is known as the genetic code.

b. A gene is a location on our chromosomes where genetic information is stored

- i. Chromosomes are made of long strands of molecules called DNA

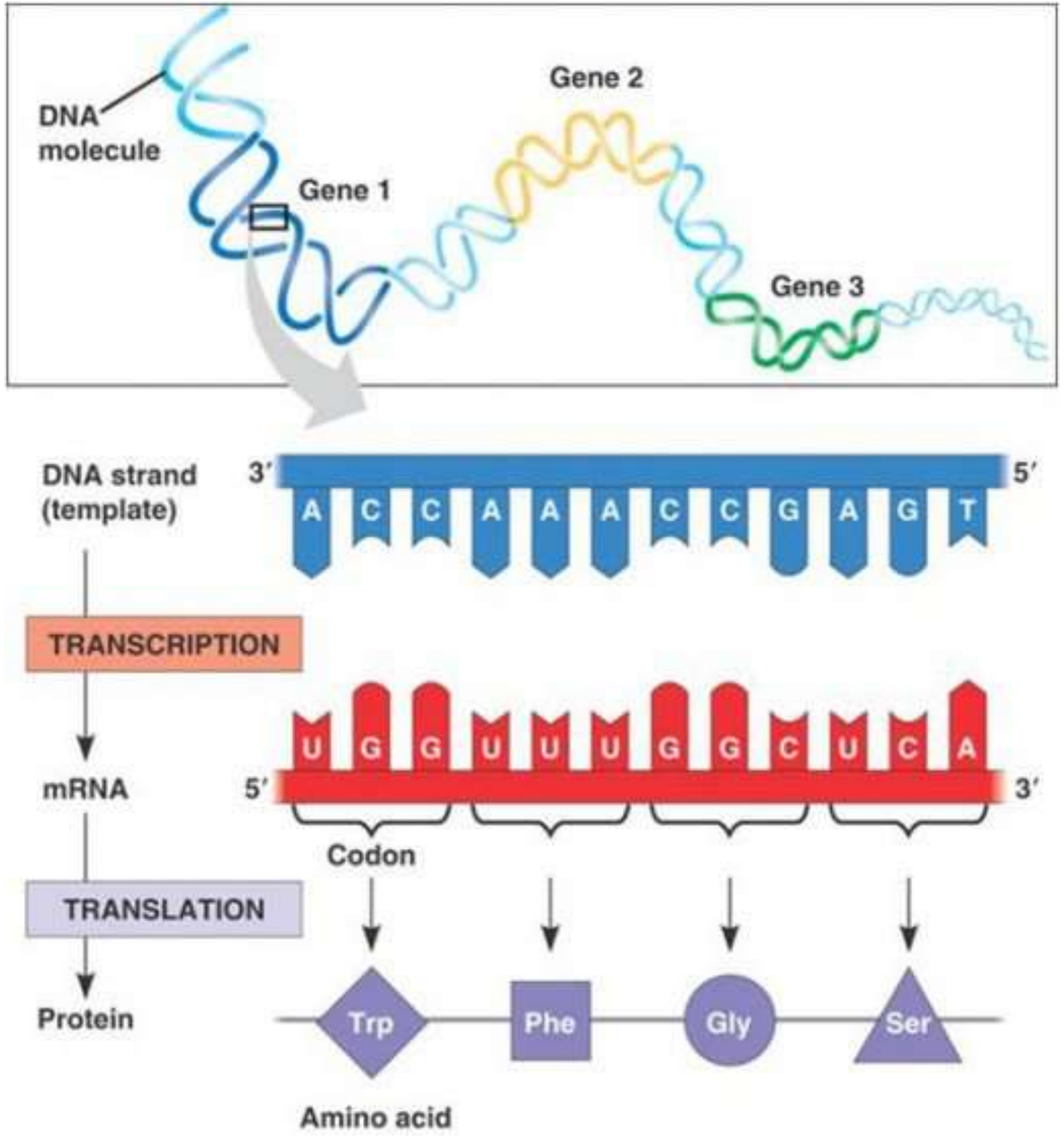


c. The information in a gene is used to determine which of the 20 different amino acids are linked together into a chain

i. The chain of amino acids is called a polypeptide or protein

d. Nearly every cell contains all the genetic information necessary to produce a human being

i. Human DNA contains enough genetic information to assemble about 100 000 different proteins



3. All known life forms use the same genetic code to create proteins

a. This is why a human gene can be inserted into a bacteria to produce the protein insulin

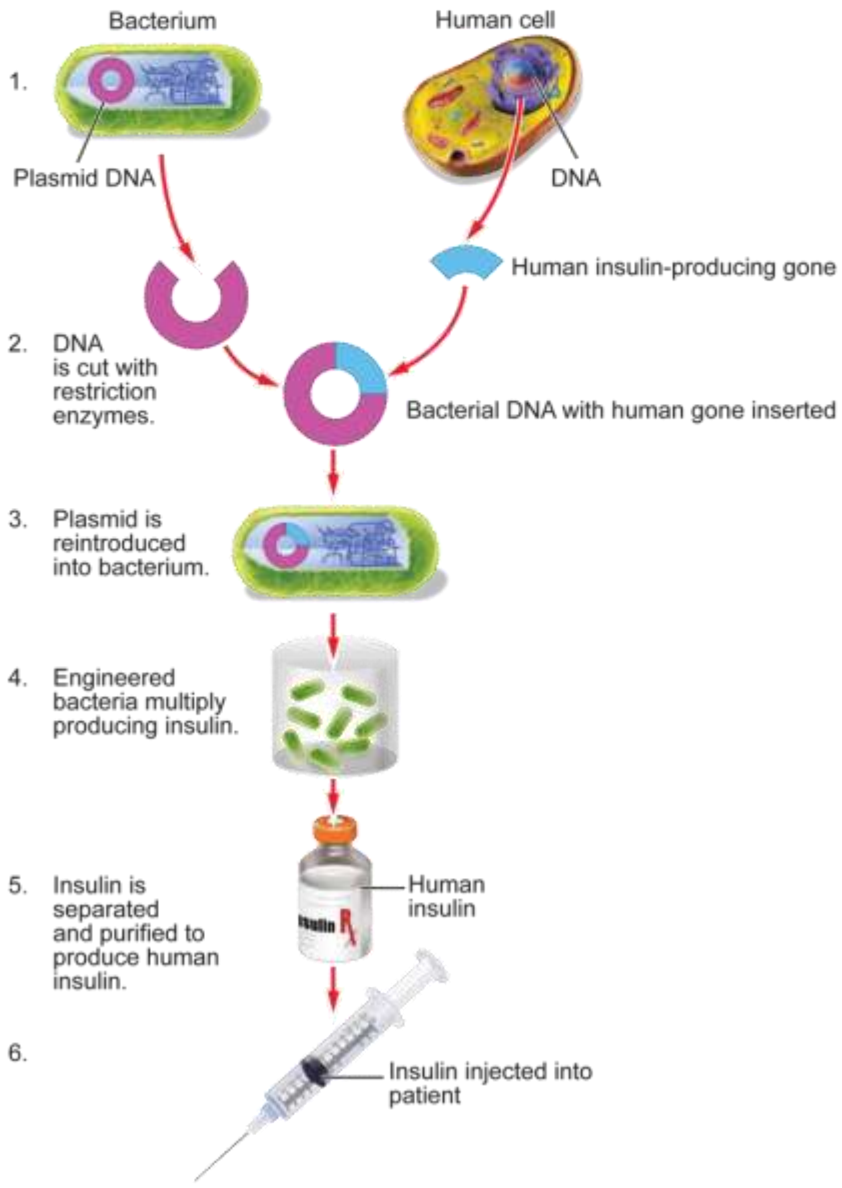
b. We share 98% of the same genes as a chimpanzee

  i. We share 60% similarity with a fruit fly

c. Some viruses attack the cells in our body by substituting their own genes into our cells

  i. Instead of making normal human proteins, the infected cells are altered to make viral proteins instead.

  ii. Because of genetic similarities a flu virus can spread from a duck to a pig to a human



4. Chromosomes are made of a special molecule called DNA that stores genetic information

a. There are 4 special chemicals that can be found in our DNA called bases

i. The bases are represented by the letter at the beginning of the chemical name

ii. Adenine, Thymine, Cytosine, Guanine – A, T, C, G

b. A human strand of DNA can have several million bases joined together

i. Opposite from every C on one strand will always be a G on other strand

ii. Opposite from every A on one strand will always be a T on other strand

d. The two strands wind around each other which is why DNA is sometimes called a double helix

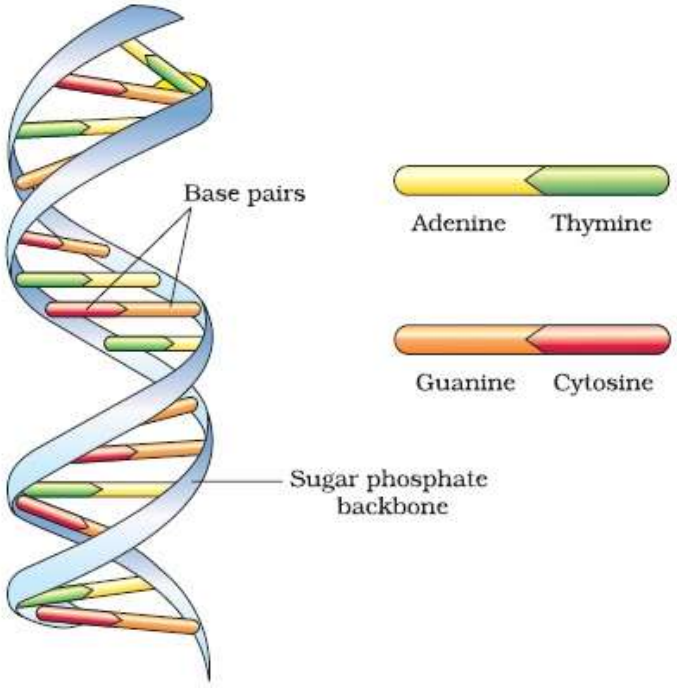


Figure 6.3 DNA double helix

Genetics 2 – Mendel and the Gene Hypothesis

1. **Gregor Mendel** was an Austrian monk whose job it was to cultivate pea plants.

a. He grew more than 50 000 plants over a ten year period

i. This allowed him to make observations over a large sample size

2. The pea plants were a good organism for study for a number of reasons

a. Multiple generations of plants could be grown in one season

b. The flowers of a pea plant have both male and female reproductive parts
   i. If the pollen is transferred between the flowers of the same plant then it has self-pollinated

   ii. If the pollen moves between the flowers of two different pea plants then it has cross-pollinated

c. Pea plants can be made to reproduce either by self-pollinating or cross-pollinating
   i. Male or female parts of the flower can be removed or covered to prevent pollination
   ii. Pollen can be transferred by paintbrush from one flower to another

d. Pea plants have a number of easily observable inherited traits that have only two options

Trait	Option 1		Option 2	
Plant height				
Flower colour				
Flower location				
Seed colour				
Seed skin				
Pea pod colour				
Pea pod structure				

d. Pea plants have a number of easily observable inherited traits that have only two options

Trait	Option 1		Option 2	
Plant height	tall		short	
Flower colour				
Flower location				
Seed colour				
Seed skin				
Pea pod colour				
Pea pod structure				

d. Pea plants have a number of easily observable inherited traits that have only two options

Trait	Option 1		Option 2	
Plant height	tall		short	
Flower colour	Purple		white	
Flower location				
Seed colour				
Seed skin				
Pea pod colour				
Pea pod structure				

d. Pea plants have a number of easily observable inherited traits that have only two options

Trait	Option 1		Option 2	
Plant height	tall		short	
Flower colour	Purple		white	
Flower location	grow in between the leaves on a branch		Grow at the end of a branch	
Seed colour				
Seed skin				
Pea pod colour				
Pea pod structure				

d. Pea plants have a number of easily observable inherited traits that have only two options

Trait	Option 1		Option 2	
Plant height	tall		short	
Flower colour	Purple		white	
Flower location	grow in between the leaves on a branch		Grow at the end of a branch	
Seed colour	Yellow peas		Green peas	
Seed skin				
Pea pod colour				
Pea pod structure				

d. Pea plants have a number of easily observable inherited traits that have only two options

Trait	Option 1		Option 2	
Plant height	tall		short	
Flower colour	Purple		white	
Flower location	grow in between the leaves on a branch		Grow at the end of a branch	
Seed colour	Yellow peas		Green peas	
Seed skin	Smooth skin		Wrinkled skin	
Pea pod colour				
Pea pod structure				

d. Pea plants have a number of easily observable inherited traits that have only two options

Trait	Option 1		Option 2	
Plant height	tall		short	
Flower colour	Purple		white	
Flower location	grow in between the leaves on a branch		Grow at the end of a branch	
Seed colour	Yellow peas		Green peas	
Seed skin	Smooth skin		Wrinkled skin	
Pea pod colour	Green pea pod		Yellow pea pod	
Pea pod structure				

d. Pea plants have a number of easily observable inherited traits that have only two options

Trait	Option 1		Option 2	
Plant height	tall		short	
Flower colour	Purple		white	
Flower location	grow in between the leaves on a branch		Grow at the end of a branch	
Seed colour	Yellow peas		Green peas	
Seed skin	Smooth skin		Wrinkled skin	
Pea pod colour	Green pea pod		Yellow pea pod	
Pea pod structure	Inflated pea pod		Constricted pea pod	

3. At first Mendel would perform a crossbreeding experiment involving only one trait, called a **monohybrid cross**.

a. Mendel would follow the inheritance of just one of the traits for three generations of plants

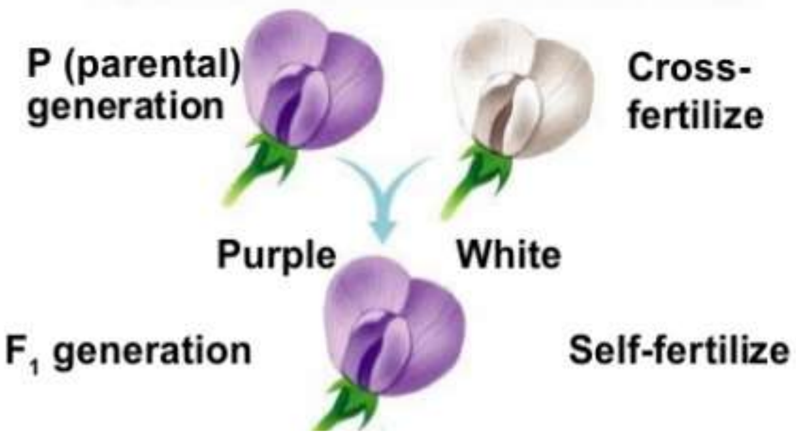
i. He would often start with two purebred plants

b. A purebred plant will always produce offspring plants with that trait 100% of the time

ii. i.e. a purebred purple flower plant always produces offspring plants with purple flowers

4. E.g. flower colour

a. First cross



i. Mendel would begin with two purebred plants with different flower colour and cross breed them together

- He called these plants the P generation (for ”parent” generation)
- i.e. Purple flower x White flower

ii. All of the offspring plants were called the F_1 generation (for “first filial”) and they would only produce purple flowers

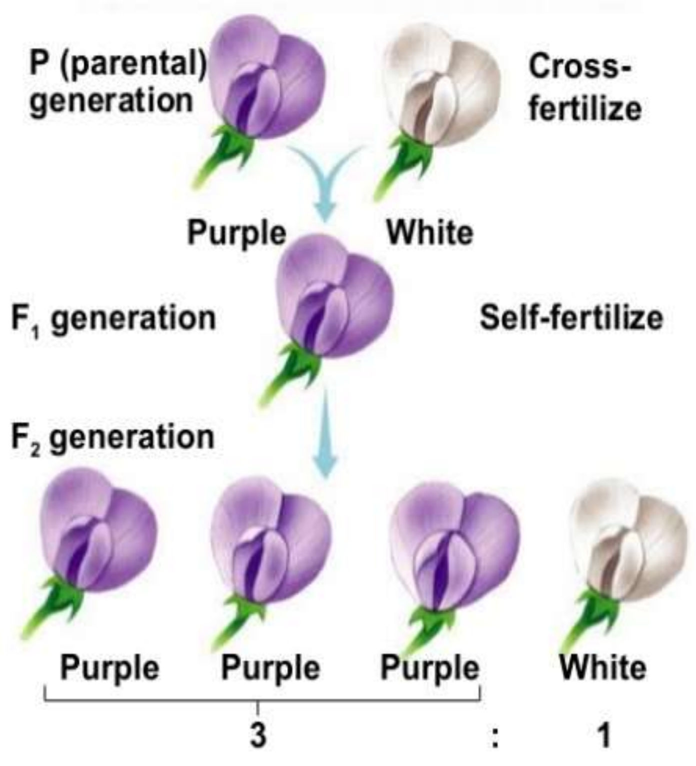
-The white flower trait had seemed to disappear

iii. Mendel called the purple flower a **dominant** trait and the white flower a **recessive** trait

b. Second cross

i. Mendel would take the F_1 plants and crossbreed them with each other or self-pollinate them

- Purple flower x Purple flower
- The resulting offspring plant were called the F_2 generation (for "second filial" generation)



ii. About ¾ of all of the F_2 generation still had purple flowers

- ¼ of the F_2 generation had white flowers
- The F_2 generation showed a ratio of
  3 purple :1 white flower

iii. Mendel determined that the recessive white flower trait had been present in the F_1 generation plants but it had been **masked** by the dominant purple flower trait

iv. Mendel performed the same type of procedure with the different traits of the pea plant and the results would always be the same

v. He determined that certain traits in the pea plant were dominant and the other trait was recessive

vi. He hypothesized that some unknown factor was being transmitted from the parents to the offspring that controlled the characteristics of the plants

Character	Contrasting traits		F_1 results	F_2 results	F_2 ratio
Seed shape	round/wrinkled		all round	5474 round 1850 wrinkled	2.96:1
Seed color	yellow/green		all yellow	6022 yellow 2001 green	3.01:1
Pod shape	full/constricted		all full	882 full 299 constricted	2.95:1
Pod color	green/yellow		all green	428 green 152 yellow	2.82:1
Flower color	violet/white		all violet	705 violet 224 white	3.15:1
Flower position	axial/terminal		all axial	651 axial 207 terminal	3.14:1
Stem height	tall/dwarf		all tall	787 tall 277 dwarf	2.84:1

Human Traits – earlobes

Attached earlobe

Unattached earlobe

Human Traits – tongue rolling

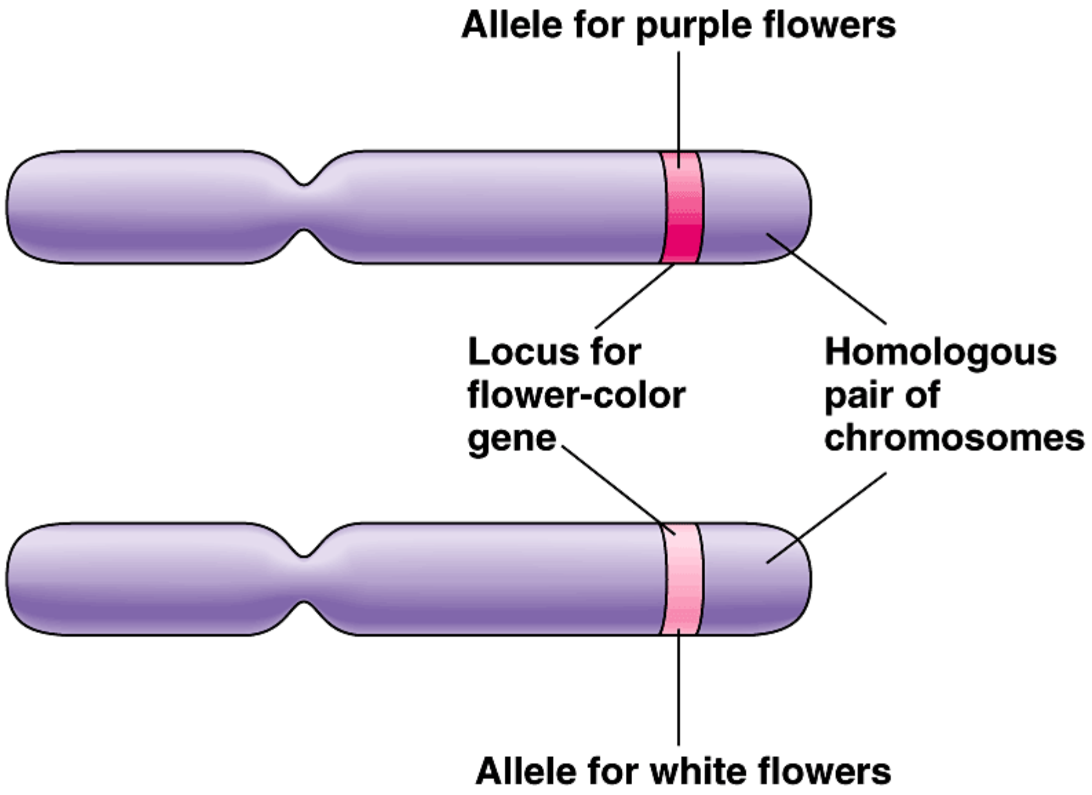
Genetics 3 – Mendel's Gene Hypothesis

1. Without any knowledge of chromosomes or DNA, Mendel generated the following ideas

a. A hereditary unit of information called a **gene** is passed form parent to offspring

b. An **allele** is one of the possible versions of the gene

i. E.g. the flower colour gene has two alleles, one for purple flowers, and one allele for white flowers



2. Mendel took his ideas about genes and alleles and made the following hypotheses

a. The presence of different alleles is responsible for the variation in the appearance of an organism

b. An organism always has two genes present for each characteristic
   i. Each parent donates one gene to the offspring

c. If the alleles on both of the genes are the same, then the organism is **purebred**
   i. E.g. a plant's genes for flower colour are both the purple allele

d. If the two alleles are different, then the organism is a **hybrid** for the characteristic
   i. One trait will be dominant and the other trait will be recessive, and only the dominant trait is expressed in the organism

   ii. Eg. one of a plant's genes for flower colour is purple, the other gene is white → The plant only produces purple flowers

3. Mendel studied seven different traits in pea plants and sometimes followed more than one trait at a time, e.g. short, purple flower plant crossed with tall, white flower plant

a. He found that none of the traits were affecting the other
   i. i.e. the purple flower trait did not affect the height of the plant

b. this is known as **his law of independent assortment**

4. We can look at the alleles in the generations created by crossing two pure plants for flower colour

a. We use letters to represent the two alleles for flower colour

i. F = the purple flower allele (capital letter for dominant)

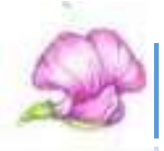
ii. f = the white flower allele (lowercase letter for recessive)

b. each plant has two genes for the flower colour trait and they are both purebred

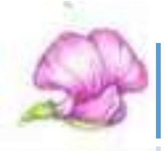
i. FF = purebred purple flower plant

ii. ff = purebred white flower plant

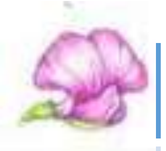
c. **First cross**

	Purebred purple flower plant 		
Purebred white flower plant 			

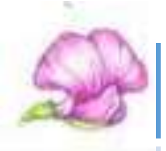
c. First cross

	Purebred purple flower plant 		
Purebred white flower plant 		F	F

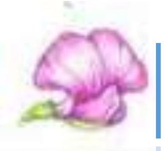
c. First cross

	Purebred purple flower plant 		
Purebred white flower plant 		F	F
	f		
	f		

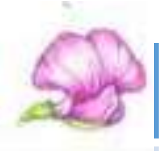
c. First cross

	Purebred purple flower plant 		
Purebred white flower plant 		F	F
	f	Ff 	
	f		

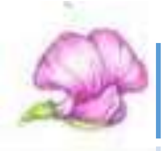
c. First cross

	Purebred purple flower plant 		
Purebred white flower plant 		F	F
	f	Ff	Ff 
	f		

c. First cross

	Purebred purple flower plant 		
Purebred white flower plant 		F	F
	f	Ff	Ff
	f	Ff 	

c. First cross

	Purebred purple flower plant 		
Purebred white flower plant 		F	F
	f	Ff 	Ff 
	f	Ff 	Ff 

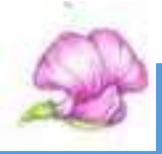
i. All of the F_1 generation are hybrids, i.e. they have both alleles (Ff)

ii. Since purple is dominant over white, the flower colour is always purple

d. **Second cross** – two hybrid parents

	Hybrid purple flower plant 		
Hybrid purple flower plant 			

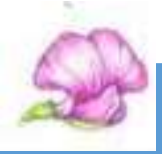
d. Second cross – two hybrid parents

	Hybrid purple flower plant 		
Hybrid purple flower plant 		F	f
	F		
	f		

d. Second cross – two hybrid parents

	Hybrid purple flower plant 		
Hybrid purple flower plant 		F	f
	F	FF 	
	f		

d. Second cross – two hybrid parents

	Hybrid purple flower plant 		
Hybrid purple flower plant 		F	f
	F	FF	Ff 
	f		

d. Second cross – two hybrid parents

	Hybrid purple flower plant 		
Hybrid purple flower plant		F	f
	F	FF	Ff
	f	Ff 	

d. Second cross – two hybrid parents

	Hybrid purple flower plant 		
Hybrid purple flower plant 		F	f
	F	FF 	Ff 
	f	Ff 	ff 

i. In the F_2 generation

- Only 1 out of the four possible combinations will be white flower (ff)
- Three out of the four possible combinations are purple flower (FF and Ff)
- But even if they have purple flower they may still be genetically a hybrid and carry the recessive allele

5. **Phenotype** refers to the appearance of a characteristic in an organism

a. You can observe the phenotype of an organism
   i. E.g. The colour of the flower is the phenotype in the plant

b. The phenotypic ratio compares the number of each phenotype that is expresses in the offspring
   i. E.g. if in a particular cross, there are 30 tall offspring plants and 10 short offspring plants, the phenotypic ratio is 3: 1

6. **Genotype** refers to the genetic makeup of the organisms

a. You cannot easily observe the genotype of the organism
   - i. E.g. a purple flower plant may be FF or Ff
   - ii. E.g. a white flower plant must be ff however since it is the only way that the recessive white allele can be expressed

b. Organisms that have matching alleles for a trait are said to be homozygous for the trait
   - i. E.g. FF would be homozygous dominant while or ff would be homozygous recessive

c. Hybrid organisms have non-matching alleles and are said to be heterozygous for the trait

i. E.g. Ff is a heterozygous genotype

d. The genotypic ratio compares the number of each genotype in the offspring

i. If the results of the pea height example were 10 homozygous dominant (TT), 20 (Tt) heterozygous and 10 homozygous recessive (tt), the genotypic ratio would be 1:2:1

Genetics 4 – Other Patterns of Inheritance

1. All of the traits that Mendel observed in his pea plants exhibited **complete dominance**

a. The dominant allele completely masked the recessive allele

b. There were only two versions of the trait, i.e. white flower or purple flower

   i. There was no blending of the two traits , i.e. light purple

	Purebred purple flower plant		
Purebred white flower plant		F	F
	f	Ff 	Ff 
	f	Ff 	Ff 

2. In **incomplete dominance**, the hybrid expresses a mixture of the traits

a. Each trait is present in the offspring but is only partly expressed
b. The hybrid expresses an intermediate phenotype.
c. E.g. Red carnation flowers (RR) x white carnation flowers (rr)

	Red Carnation flower plant		
White Carnation flower plant			

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c. E.g. Red carnation flowers (RR) x white carnation flowers (rr)

	Red Carnation flower plant		
White Carnation flower plant			
	r		
	r		

i.

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	Red Carnation flower plant		
White Carnation flower plant		R	R
	r		
	r		

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	Red Carnation flower plant		
White Carnation flower plant		R	R
	r	Rr	
	r		

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	Red Carnation flower plant		
White Carnation flower plant		R	R
	r	Rr	
	r	Rr	

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	Red Carnation flower plant		
White Carnation flower plant		R	R
	r	Rr	Rr
	r	Rr	

2. In incomplete dominance, the hybrid expresses a mixture of the traits

a. Each trait is present in the offspring but is only partly expressed
b. The hybrid expresses an intermediate phenotype.
c. E.g. Red carnation flowers (RR) x white carnation flowers (rr)

	Red Carnation flower plant		
White Carnation flower plant		R	R
	r	Rr	Rr
	r	Rr	Rr

i. All of the F1 generation are heterozygous and produce pink flowers
ii. The hybrid can only produce half as much red pigment so the flowers are pink

d. If we cross two pink carnation plants

   i. Pink carnation flowers (Rr) x pink carnation flowers (Rr)

	Pink Carnation flower plant		
Pink Carnation flower plant			

d. If we cross two pink carnation plants

   i. Pink carnation flowers (Rr) x pink carnation flowers (Rr)

	Pink Carnation flower plant		
Pink Carnation flower plant		R	r
	R	RR	
	r		

d. If we cross two pink carnation plants

i. Pink carnation flowers (Rr) x pink carnation flowers (Rr)

	Pink Carnation flower plant		
Pink Carnation flower plant		R	r
	R	RR	
	r	Rr	

d. If we cross two pink carnation plants

i. Pink carnation flowers (Rr) x pink carnation flowers (Rr)

	Pink Carnation flower plant		
Pink Carnation flower plant		R	r
	R	RR	Rr
	r	Rr	

d. If we cross two pink carnation plants

i. Pink carnation flowers (Rr) x pink carnation flowers (Rr)

	Pink Carnation flower plant		
Pink Carnation flower plant		R	r
	R	RR	Rr
	r	Rr	rr

ii. In the F2 generation

- The phenotypic ratio is **1 red: 2 pink : 1 white**
- The genotypic ratio is **1 RR : 2Rr: 1 rr**

3. In codominance, both traits for a characteristic are completely expressed in the hybrid

a. Human A and B blood types display codominance

i. A person with type A blood produces the A protein on the outside of their red blood cells

- A person with type B blood produces the B protein
- An O type person would not produce any proteins

b. E.g. A person with homozygous for type A blood has a child with someone who is homozygous for B type blood

	Homozygous A type parent		
Homozygous B type parent			

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	Homozygous A type parent	
Homozygous B type parent	A	A

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	Homozygous A type parent		
Homozygous B type parent		A	A
	B		
	B		

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	Homozygous A type parent		
Homozygous B type parent		A	A
	B	AB	
	B		

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	Homozygous A type parent		
Homozygous B type parent		A	A
	B	AB	
	B	AB	

i

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	Homozygous A type parent		
Homozygous B type parent		A	A
	B	AB	AB
	B	AB	

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	Homozygous A type parent		
Homozygous B type parent		A	A
	B	AB	AB
	B	AB	AB

i. Note that both A and B are dominant alleles and are both capitalized

ii. The child will produce both proteins on the red blood cells and be classified as type AB

c. E.g. A person with AB blood has a child with someone who has AB blood

	AB type parent		
AB type parent			

c. E.g. A person with AB blood has a child with someone who has AB blood

	AB type parent		
AB type parent		A	B

c. E.g. A person with AB blood has a child with someone who has AB blood

	AB type parent		
AB type parent		A	B
	A		
	B		

c. E.g. A person with AB blood has a child with someone who has AB blood

	AB type parent		
AB type parent		A	B
	A	AA	
	B		

c. E.g. A person with AB blood has a child with someone who has AB blood

	AB type parent		
AB type parent		A	B
	A	AA	
	B	AB	

c. E.g. A person with AB blood has a child with someone who has AB blood

	AB type parent		
AB type parent		A	B
	A	AA	AB
	B	AB	

c. E.g. A person with AB blood has a child with someone who has AB blood

	AB type parent		
AB type parent		A	B
	A	AA	AB
	B	AB	BB

i. There is a 50% chance that the child will have type AB blood

ii. There is a 25% chance that the child will have type A blood

iii. There is a 25% chance that the child will have type B blood

d. E.g. a person with type O blood has a child with someone who has AB blood

i. A type O person carries a recessive allele and does not produce any protein on their red blood cells

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	AB type parent	
O type parent		
	A	B
o	Ao	Bo
o	Ao	Bo

- There is a 50% chance that the child will have type A blood
- There is a 50% chance that the child will have type B blood

4. Sex-linked traits

a. In animals such as us, the combination of X and Y chromosomes determines the sex of the organism

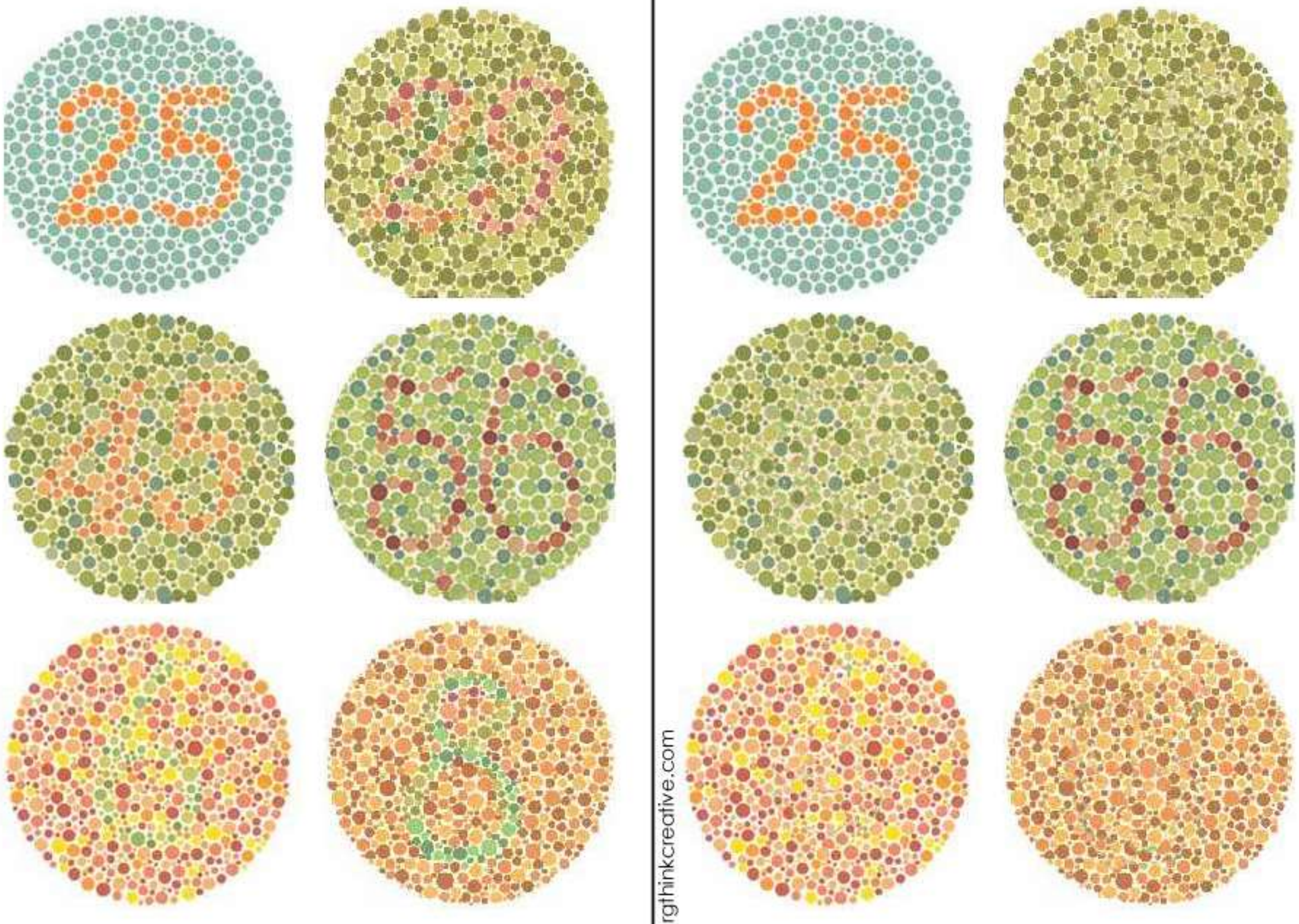
  a. XY is male
  b. XX is female

b. In the traits so far, it has not made a difference which parent had which genotype

  i. Some traits show an inheritance pattern that depends upon the sex of the organism
  ii. E.g. red-green colour blindness is more common in males than in females

Ishihara Test For Color Blindness

What People With Regular Vision See | **What Red-Green Color Blind People See**



c. A sex-linked gene is located on a sex chromosome

   i. E.g. The colour-blindness gene is recessive and is located on the X chromosome

      - The Y chromosome does not have a matching gene for this characteristic

   ii. The following genotypes would represent

      - a male that is colour blind X^bY
      - a regular vision male X^BY
      - a regular vision female X^BX^B
      - a regular vision female that carries the colour blind gene X^BX^b

d. E.g. A colour blind male has a child with a regular vision female

	Regular vision female		
Colour blind male			

i

d. E.g. A colour blind male has a child with a regular vision female

	Regular vision female		
Colour blind male		X^B	X^B

d. E.g. A colour blind male has a child with a regular vision female

	Regular vision female		
Colour blind male		X^B	X^B
	X^b		
	Y		

i.

d. E.g. A colour blind male has a child with a regular vision female

		Regular vision female	
Colour blind male		X^B	X^B
	X^b	X^BX^b	
	Y		

d. E.g. A colour blind male has a child with a regular vision female

	Regular vision female		
Colour blind male		X^B	X^B
	X^b	X^BX^b	
	Y	$X^B Y$	

d. E.g. A colour blind male has a child with a regular vision female

	Regular vision female		
Colour blind male		X^B	X^B
	X^b	$X^B X^b$	$X^B X^b$
	Y	X^B Y	

d. E.g. A colour blind male has a child with a regular vision female

	Regular vision female		
Colour blind male		X^B	X^B
	X^b	X^BX^b	X^BX^b
	Y	X^B Y	X^B Y

i. Note that there is a 50% chance of the child being a boy or a girl

ii. There is a 100% chance that a female child will be a carrier for colour blindness

iii. There is a 100% chance that a male child will have regular vision

Genetics 5 – Mutations and Their Effects

1. A mutation is a change in the DNA of a gene

a. Since DNA contains the information about the assembly of proteins, a mutation can affect the structure or function of a protein and therefore affect the structure or function of a cell

b. The organism that carries the mutation is called a mutant

c. Mutations can be classified as

i. **Silent or neutral** – the effect is not seen in the organism

- E.g. a gene responsible for producing a red pigment is still able to do so even after a mutation and change to the amino acid sequence

ii. **Negative** - it has a damaging effect on the organism's ability to survive or reproduce successfully

- E.g. a fruit fly with a single gene mutation can grow an extra pair of wings and be unable to fly

iii. **Positive** – it produces an advantageous characteristic that enhances an organism's ability to survive or reproduce successfully

- A bacteria cell mutates to become resistant to an antibiotic

d. Mutations are responsible for **genetic diversity** in a population

i. A population whose members are genetically different is better able to deal with to deal with new diseases or changes to the environment

2. Negative mutations may persist in a population indefinitely

a. **Albinism** is caused by a mutation in one of several genes

i. An albino is an individual who does not have the ability to produce melanin which is a pigment that gives skin its colour.

- Melanin helps to protect the skin from UV light

ii. An albino individual will be very sensitive to sunlight

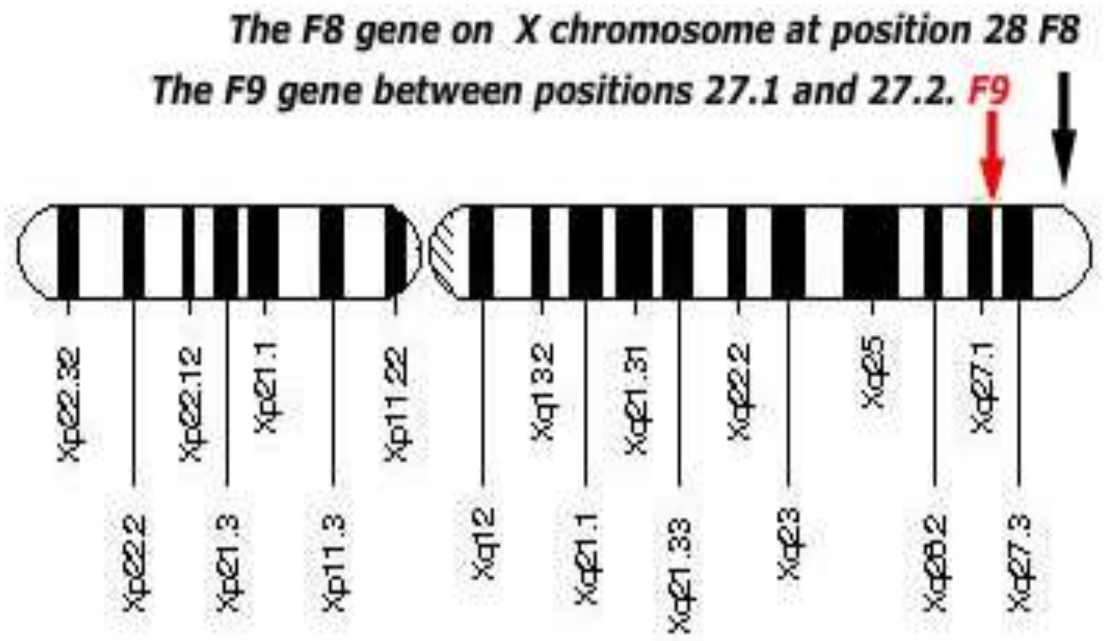
iii. Albinism is a recessive trait, so the albinism gene must be inherited from both parents

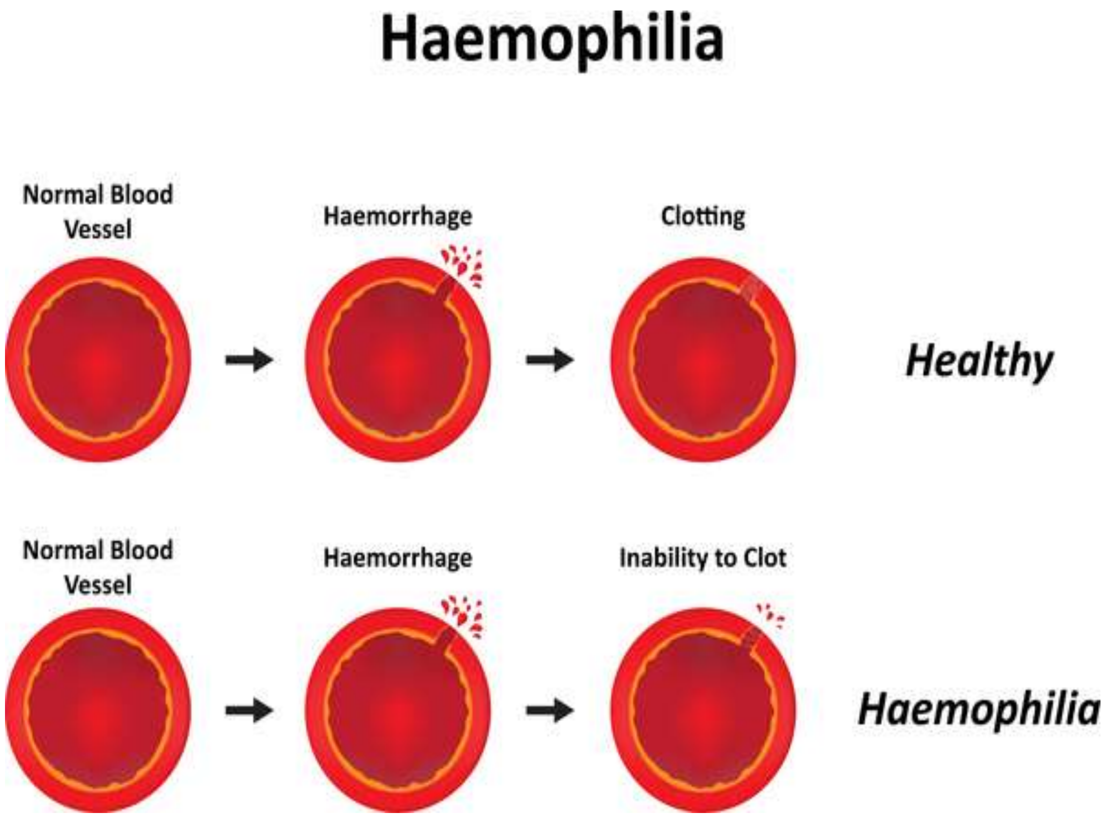
b. **Hemophilia** is genetic disease which causes an individual's blood to not clot properly

i. Hemophilia is caused by a mutation to one of two genes (F8 or F9)

- Once it appears in a population, it can be passes on to succeeding generations

ii. Hemophilia is a sex-linked disorder and is more common in males than in females





3. Some mutations can have **both** a negative and a positive effect

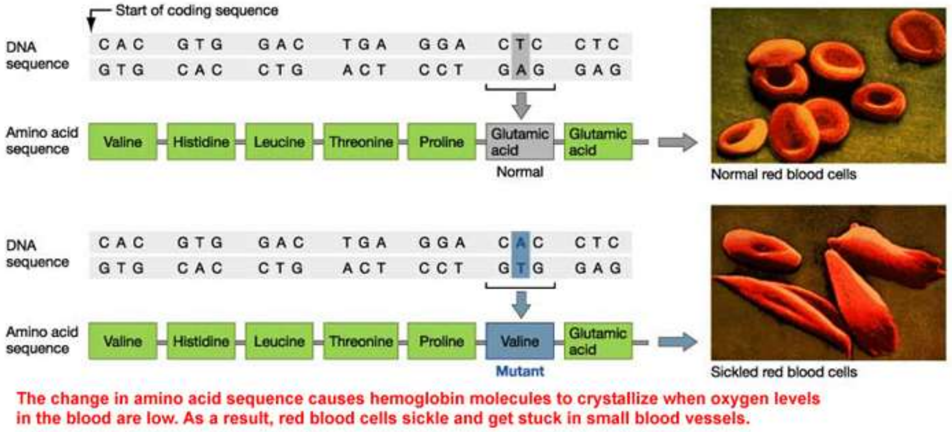
a. **Sickle cell anemia** is caused by various mutations in the HBB gene which is responsible for the one of the proteins (beta-globin) which makes up the hemoglobin molecule

  i. Hemoglobin is found on the red blood cell and binds to oxygen

  ii. It turns red when it binds to the oxygen

b. The mutated gene produces an abnormal version of hemoglobin which causes them to stick together which distorts the shape of the red blood cells into a sickle shape

  i. The elongated red blood cell is relatively inefficient at carrying oxygen



c. The negative effect is that a person with two alleles for the defective hemoglobin gene may develop a painful and life-shortening condition

d. The positive effect of the mutation is that a person who is recessive for the condition (i.e carries both a normal and a mutated hemoglobin allele) may have an immunity to the disease malaria

i. **Malaria** is caused by small organism called Plasmodium vivax

- Plasmodium vivax will infect red blood cells and cause them to burst

ii. The sickle shaped cells prevent the Plasmodium vivax from infecting the mutated red blood cells

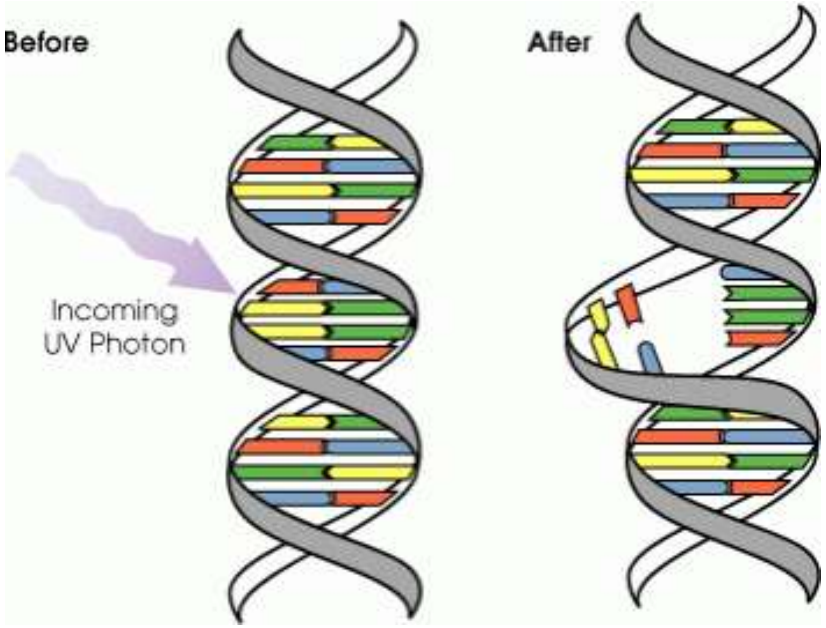
- Many people in central Africa have the sickle cell mutation where it is an advantage

4. Mutations are caused by a **mutagen**

a. Mutagens can be physical, chemical or biological in nature

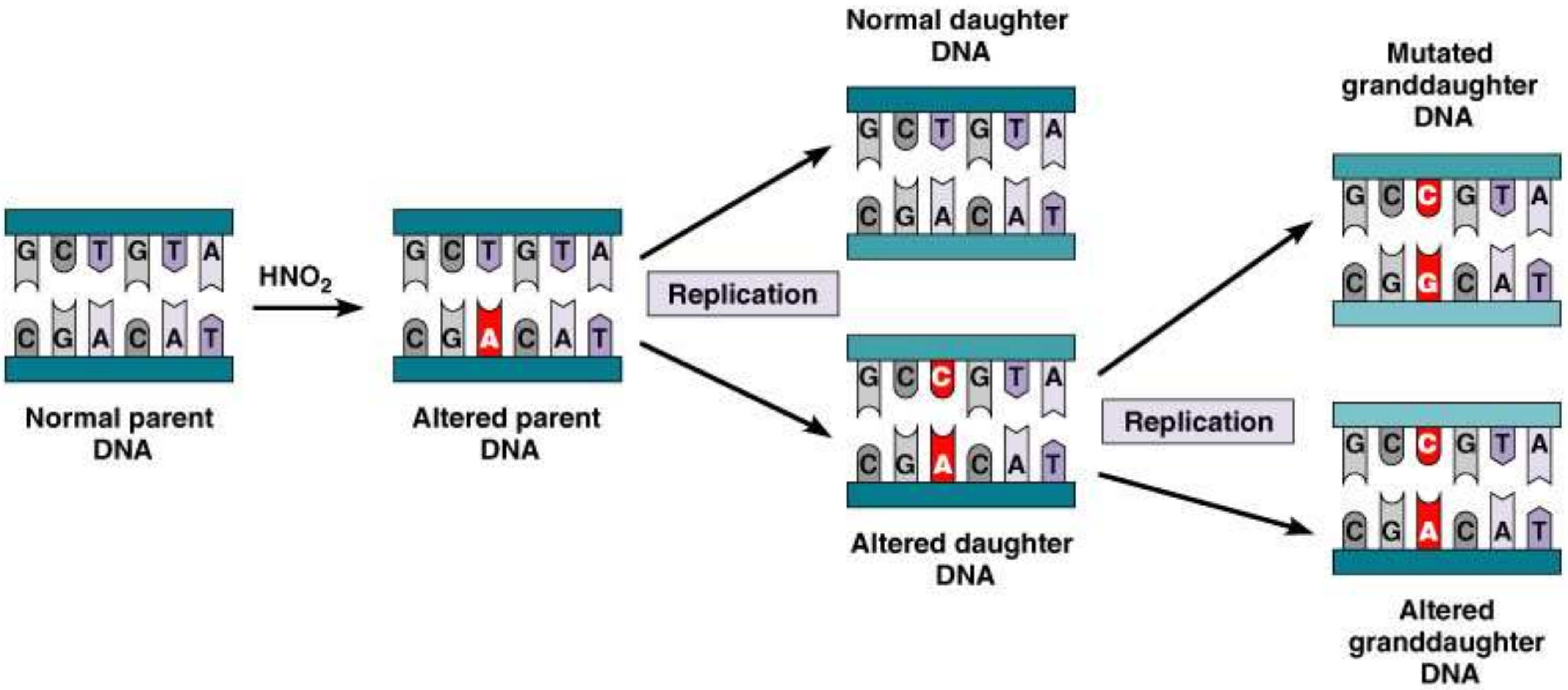
b. A physical mutagen would be radiation such as ultraviolet radiation or X-rays

- i. The ionizing radiation can causes electron to be bumped off of atoms
- ii. This can result in a crooked DNA molecule



c. Many chemicals found in pesticides, cigarette smoke, environmental pollutants can cause mutations

i. Nitrous acid (HNO_2) can cause a chemical reaction in the DNA adenine bases to switch to thymine



d. Certain **viruses** (e.g. HIV) will invade a host cell and insert their DNA into the hosts' DNA
    i. The host will now produce viral proteins instead of their own
    ii. The change to the DNA is considered a mutation