16- inherited change

16.1 PASSAGE OF INFO FROM PARENT TO OFFSPRING

a) homologous chromosome pairs

- KARYOGRAM - visual profile of chromosomes
  - → chromosomes are arranged into homologous pairs according to size
  - → sex cells are shown last

- HOMOLOGOUS CHROMOSOMES
  - → pair of chromosomes in a DIPLOID CELL (one from mother, the other from father)
  - → have the same structure & genes (although diff versions) at the same LOCI
  - → pair together to form a BIVALENT during the first division of meiosis

- locus - position at which a particular gene is found on a particular chromosome
  - → the same gene is always found at the same locus

- gene - length of DNA that codes for a particular protein or polypeptide

- allele - particular variety of genes

- autosomes - all other chromosomes aside from sex chromosomes

b) haploid & diploid

- HAPLOID (n) cells have 1 set of chromosomes

- DIPLOID (2n) cells contain 2 sets of chromosomes

- **need for reduction division**

① no. of chromosomes doubles every generation

② introduces genetic variation (may also arise from mutation

d) ~~Gametogenesis~~ chromosome behaviour during meiosis

MEIOSIS (2n)
- meiosis I (2 x n) — reduction division resulting in 2 daughter nuclei
- divides again
- meiosis II (4 x n) — like mitosis of 2 haploid cells resulting in 4 haploid daughter cells

VARIATION
- independent assortment of homologous chromosomes
- crossing over between chromatids of homologous chromosomes

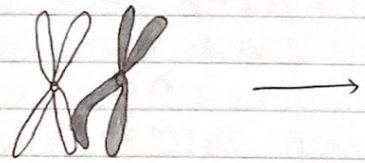
* starting cell is 2n = 4 (diploid)

• MEIOSIS I

1. PROPHASE I

PMAT

EARLY	MIDDLE	LATE
* chromosomes condense	* SYNAPSIS (homologous chromosomes pair up)	* nuclear envelope, nucleolus disappears
* mitotic spindle begins to form (formed at end of PI)	* each pair is called a BIVALENT	* chromatids may cross over
* centrosomes move to opposite poles		



CHIASMA: - point where crossing over occurs
→ one or more chiasmata may occur

2. METAPHASE I

* bivalents line up across equator of spindle, attached by centromeres

3. ANAPHASE I

* homologous chromosomes move ~~separate~~ to opposite ends of the cell (centromeres first, pulled by microtubules)
→ sister chromatids stay together

4. TELOPHASE I

* nucleolus, nuclear envelope reforming } as mitosis
* cytokinesis

0 **MEIOSIS II** ↗ like mitosis for haploid cells

* chromosomes separate as in mitosis

1. **PROPHASE II**

* nuclear envelope & nucleous disperse
* centrosomes & centrioles replicate & move to opposite poles of cell

~~* chromosomes condense~~

2. **METAPHASE II**

* chromosomes line up separately across equator of spindle

3. **ANAPHASE II**

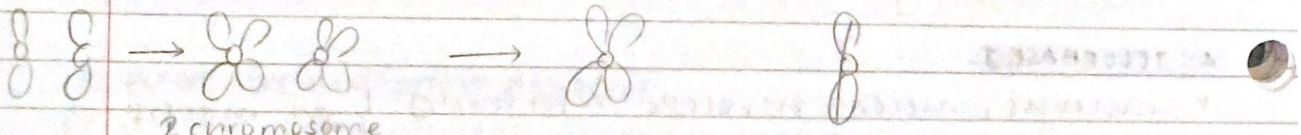
* centromeres divide & spindle microtubules pull the chromatids to opposite poles

4. **TELOPHASE II**

* 4 haploid daughter cells are formed

phase (meiosis I)	# chromoso	# chromatids
PI	46	92
MI	46	92
AI	46	92
TI	46	92
end	23	46

before meiosis in interphase
→ chromosomes are counted by # of centromeres present so 46
→ when they duplicate & get sister chromatids, there's still 46 centromeres so we say there's 46 chromosomes but 92 chromatids



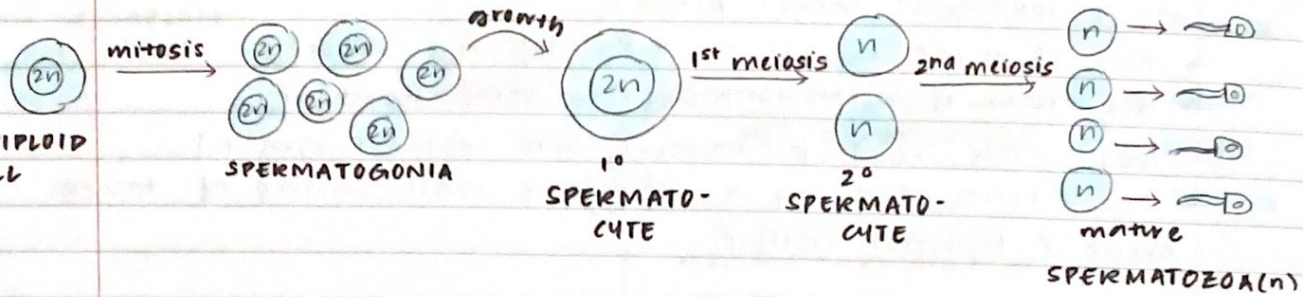
2 chromosome
~~w/ sister~~
chromatids
4 chromatids

2 chromo some

c) gametogenesis

- SPERMATOGENESIS
  - > formation of male gametes (sperm)
  - takes place inside SEMINIFEROUS TUBULES of the testes

① diploid cells divide by mitosis to produce diploid SPERMATOGONIA
② Spermatogonia grow to form diploid PRIMARY SPERMATOCYTES
③ Primary spermatocyte undergoes 1st meiosis forming two haploid SECONDARY SPERMATOCYTES
④ 2nd meiosis division produce haploid SPERMATIDS
⑤ Spermatids mature into spermatozoa



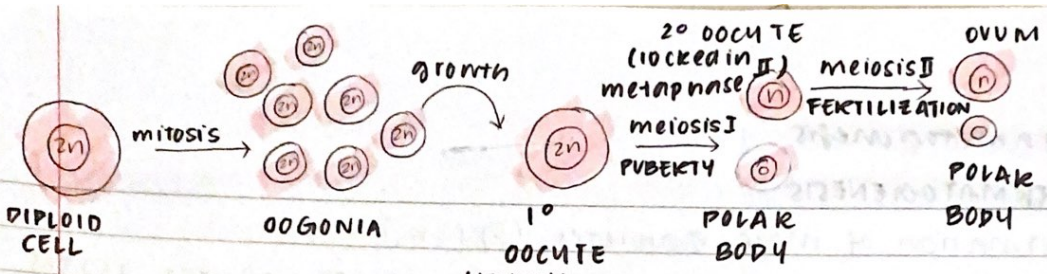
- OOGENESIS
  - > production of female gametes (ova)
  - takes place inside ovaries

① diploid cells divide via mitosis to form OOGONIA during foetal development
② oogonia undergo growth till they're large enough to undergo meiosis becoming 1° OOCYTES
* 1° oocytes begin meiosis but are stopped at prophase I (so still diploid)
* then remain in prophase I till puberty
③ FSH triggers division of 1° oocytes every month (post/during puberty)
* then complete meiosis I forming 2 haploid cells

2° OOCYTE (gets most cyto-plasm) ↙ ↘ POLAR BODY

④ 2° oocyte starts meiosis II but is stopped at metaphase II and released into oviduct
⑤ If it's fertilized, it completes meiosis II and is called an ovum ↙ ↘ polar body

ovum



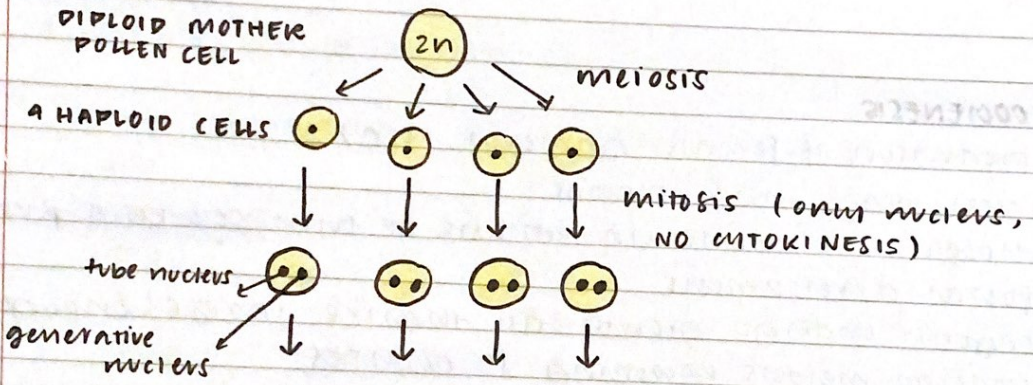
- GAMETOGENESIS IN FLOWERING PLANTS

→ **male gametes (pollen)**

① inside the anthers, diploid mother cells divide by meiosis to form 4 haploid cells

② Nucleus of each haploid cell divides by mitosis, no cytokinesis takes place
→ one of these nuclei is the tube nucleus
→ the other is called a generative nucleus

③ These cells develop / mature into pollen grains
→ each surrounded by a protective wall made of tougher exine & thinner intine



→ **female gametes (egg cell)**

① inside each ovule, a large DIPLOID SPORE MOTHER CELL develops
→ it divides by meiosis & produces 4 HAPLOID CELLS
→ 3 of them degenerate leaving one

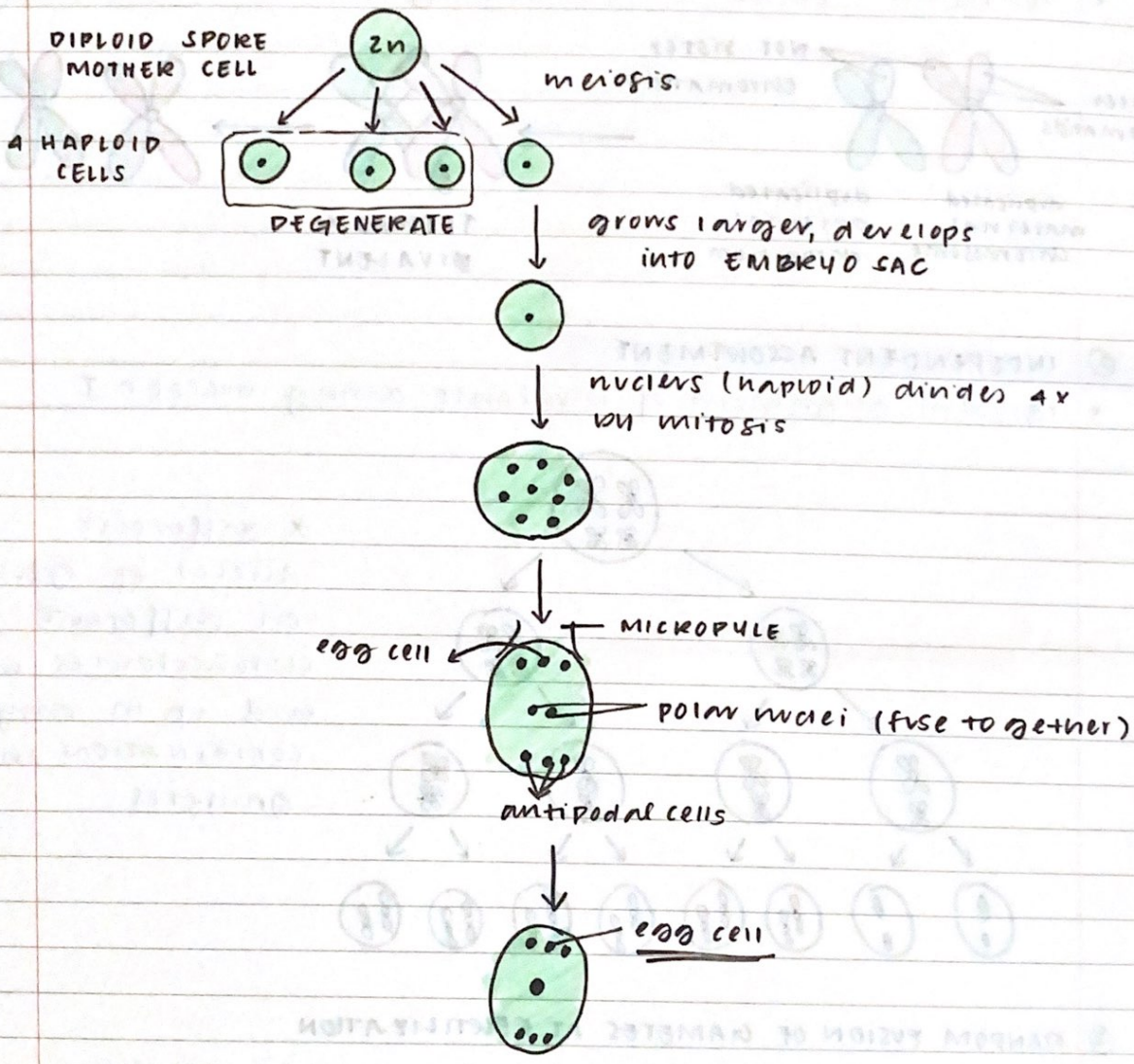
② Surviving haploid cell develops into EMBRYO SAC
→ embryo sac grows larger
→ then its nuclei divide by mitosis 4x to form 8 haploid nuclei

③ 6 move to the sides (3 to each side)

→ the ones ~~near the~~ opposite the micropyle are the ANTIPODAL CELLS

→ the ones at the top, out of the 3, one becomes the egg cell → called POLAR NUCLEI

→ 2 nuclei are present at the middle of the embryo sac & they fuse together



→ **double fertilization (extra info)**

① pollen released into embryo sac

② one of 2 nuclei fertilizes the egg cell

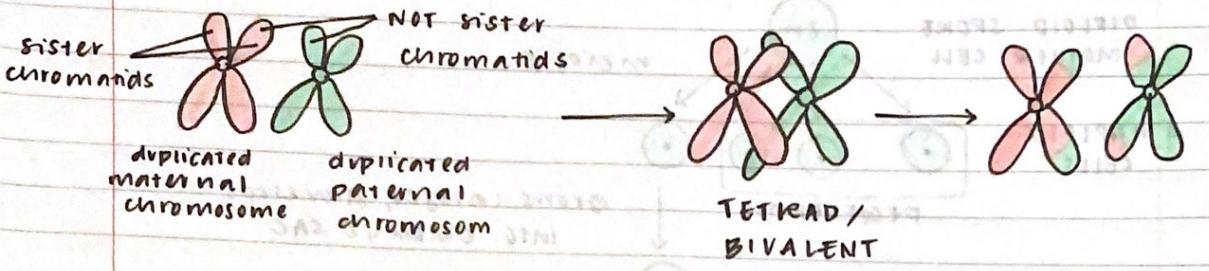
→ other one fuses with central nuclei forming a triploid cell

③ Triploid cell develops into endosperm & is the embryo's food supply during early development

d) genetic variation as a result of meiosis

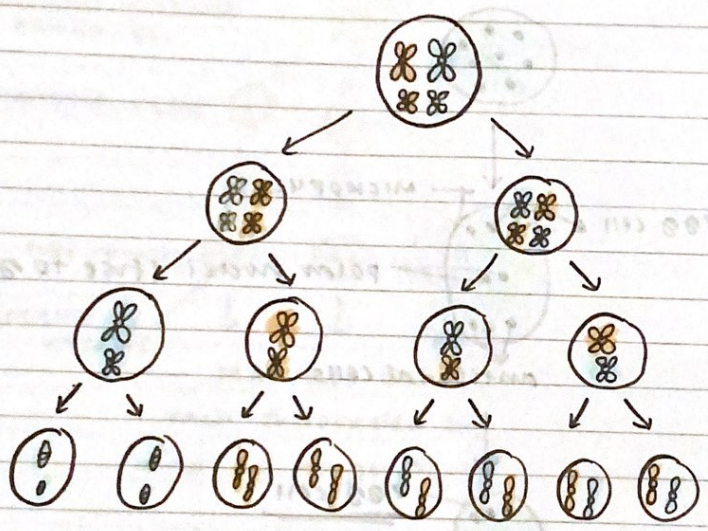
① CROSSING OVER

* takes place during prophase I
* chromatids of 2 homologous chromosomes break & rejoin
→ part of 1 chromatid swaps places with the same part (exchange of gene loci between maternal & paternal chromatid)
* chiasma - point where crossing over occurs



② INDEPENDENT ASSORTMENT

* random alignment of bivalents during meiosis I



* different alleles of genes on different chromosomes may end up in any COMBINATION in gametes

③ RANDOM FUSION OF GAMETES AT FERTILIZATION

* including the expression of rare, recessive alleles.

16.2 THE ROLES OF GENES IN DETERMINING PHENOTYPE

a) terms for human genetics

- GENE :- length of DNA that codes for a particular protein, polypeptide
- LOCUS :- position at which a particular gene is found on a particular chromosome (same gene on same locus)
- ALLELE :- particular variant of a gene
- DOMINANT :- an allele whose effect on the phenotype of a heterozygote is identical to its effect on a homozygote
- RECESSIVE :- the allele that is expressed when no dominant allele is present
- CODOMINANT :- alleles that both have an effect on the phenotype of a heterozygous organism
- LINKAGE :- the presence of 2 genes on the same chromosom so that them tend to be inherited together & do not assort independently
- TEST CROSS :- genetic cross in which an organism showing a characteristic caused by a dominant allele is crossed with an organism that's homozygous recessive: the phenotypes of the offspring can be a guide to whether the first organism is homozygous or heterozygo us
- F1 :- offspring resulting from a cross between an organis with a homozygous dominant genotype & one with a homozygous recessive genotype

- F2 :- the offspring resulting from a cross between two F1 (heterozygous) organisms

- PHENOTYPE :- organisms' characteristics, often resulting from an interaction between its genotype & its environment

- GENOTYPE :- the alleles possessed by an organisms

- HOMOZYGOUS :- having 2 identical alleles of a gene

- HETEROZYGOUS :- having 2 different alleles of a gene

b) Genetic diagrams

- in sexual reproduction, haploid gametes are made following meiosis
  → each gamete contains 1 pair of chromosomes = 1 gene

① MONOHYBRID INHERITENCE

› inheritence of 1 gene

	♀	♂
PARENT'S PHENOTYPE	brown	brown
PARENT'S GENOTYPE	Bb	Bb
GAMETES	(B) or (b)	(B) or (b)

F2 GENOTYPE

	B	b
B	BB	Bb
b	Bb	bb

F2 PHENOTYPE 3:1 → Brown : Blue

② CODOMINANCE

> alleles both have an effect on the phenotype of a heterozygous organism

genotype	phenotype
$C^R C^R$	red
$C^R C^W$	pink
$C^W C^W$	white

parental phenotype — red × white

parental genotypes — $C^R C^R$ × $C^W C^W$

gametes — (C^R) (C^R) (C^W) (C^W)

offspring genotype

	C^W	C^W
C^R	$C^R C^W$	$C^R C^W$
C^R	$C^R C^W$	$C^R C^W$

offspring phenotype — pink

③ MULTIPLE ALLELES

> when a gene has more than one allele

parental phenotype — group A × group B

parental genotype — $I^A I^O$ × $I^B I^O$

gametes — (I^A) or (I^O) × (I^B) or (I^O)

F2 genotypes

	I^A	I^O
I^B	$I^A I^B$	$I^B I^O$
I^O	$I^A I^O$	$I^O I^O$

F2 genotypes — AB, A, B, O

• sex inheritence

- sex chromosomes differ from the autosomes in that the 2 sex chromosomes are not always alike
- they don't have genes in the same positions so are not always homologous
- this is due to 2 different (X, Y) of chromosomes

④ SEX LINKAGE

> a sex-linked gene is a gene that is present on the X chromosome and not the Y chromosome

parental phenotypes — normal man — carrier woman

parental genotypes — X^H or Y — X^H or X^h

offspring genotypes

	X^H	X^h
X^H	$X^H X^H$	$X^H X^h$ — carrier
Y	$X^H Y$	$X^h Y$ — haemophilic male

normal ↙

⑤ DIHUBRID INHERITENCE

> dihybrid crosses look at the inheritence of 2 genes at once

→ dihybrid inheritence involves the inheritence of 2 different characteristics, determined by 2 different genes located on different chromosomes

→ dihybrid crosses are therefore used to investigate the simultaneous inheritence of 2 different characteristics (such as eye colour & height)

→ any one of the two alleles for eye colour can combine with any one of the two genes for height

F1 homozygous cross — YYRR × yyrr

gametes — YR YR YR YR — yr yr yr yr

	YR	YR	YR	YR
yr	YyRr			
yr				
yr				
yr				

100% heterozygous

F_1 = hetero cross

Yy Rr × Yy Rr

(YR) (yR) (Yr) (yr)

transfer into punnet square

e.g. - A - purple stem (DOMINANT)
a - green stem (RECESSIVE)

D - cut leaves (DOMINANT)
d - potato leaves (RECESSIVE)

① dihybrid cross between a heterozygous organism & a homozygous recessive organism

parental genotype: A a D d

GAMETES FROM PURPLE, CUT PLANT: AD, Ad, aD, ad

GAMETES FROM GREEN, POTATO PLANT: ad — all gametes are this so write once

AD	AaDd
Ad	Aadd
aD	aaDd
ad	aadd

Ratio - 1:1:1:1

→ this ratio is typical of a dihybrid cross between a heterozygous & homozygous recessive organism

② dihybrid cross between 2 heterozygous organisms

	AD	Ad	aD	ad
AD	AADD	AADd	AaDD	AaDd
Ad	AADd	AAdd	AaDd	Aadd
aD	AaDD	AaDd	aaDD	aaDd
ad	AaDd	Aadd	aaDd	aadd

Ratio - 9:3:3:1

→ 9 purple, cut ; 3 purple, potato ; 3 green, cut ; 1 green, potato

→ this ratio is typical of a dihybrid cross between 2 heterozygous organisms

→ Epistasis :- the interaction between 2 non-linked genes which causes one gene to mask the expression of the other phenotype

- **INTERACTIONS BETWEEN LOCI**
  - cases where different loci interact to affect one phenotypic characteres
  - e.g. alleles on 2 separate loci both affect colour of feathers on a bird

- **AUTOSOMAL LINKAGE**
  - genes are linked when 2 or more genes loci are present on the same chromosome
  - then tend to be inherited together
  - then do not assort independently

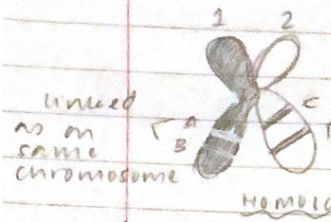
e.g. Drosophila colour gene & shape gene

BODY COLOUR
E - striped
e - ebony

ANTENNAL SHAPE
A - normal
A - weird

* genotype of striped, normal :- (EA)(EA)

EAEA indicates they are not on the same chromosome



linked as on same chromosome

A & C are NOT linked as not on same chromosome

HOMOLOGOUS PAIR

d) χ^2 (chi-squared) test

- chi-squared tests are used to determine whether the difference between an observed & expected ~~test~~ result is statistically significant

$$\left[\chi^2 = \sum \frac{(O-E)^2}{E}\right]$$

① work out expected result (E)
② work out observed result (O) [given]
③ $\frac{(O-E)^2}{E}$
④ Add all answers

to test if there's linkage or epistasis

- degree of freedom
* = [number of classes - 1]

* If χ_2 > critical value at $p = 0.05$
→ more than 95% confident that there's a significant difference between observed & expected frequencies
→ ∴ linkage / epistasis occured

* If χ_2 < critical value at $p = 0.05$
→ LESS THAN 95% confident that there's a significant difference between observed & expected frequencies (i.e. same as what we expected to see)
→ no linkage / epistasis

e) gene mutations

› mutation :- unpredictable change in the structure of a DNA molecule

› gene mutation :- change in the structure of DNA molecule, producing a different allele of a gene

› mutagen :- a substance that increases the chances of mutation occuring is said to be a mutagen
- ~~can be radio~~ random
- environmental factors (ionising radiation, uv radiation, chemicals)

› chromosome mutation - changes in the structure or no of whole chromosomes in a cell

- TYPES OF MUTATIONS -

① base substitution } most m have no effect
② base addition } * cause FRAMESHIFTS in the code → protein made may be useless
③ base deletion } * may introduce a STOP codon, so complete protein never made

- SILENT MUTATION - A mutation that has no apparent effect on an organism
  → base substitutions are often silent mutations as many amino acids have more than 1 triplet code
  → base subst however can have large effects, especially if a 'stop' triplet was introduced

f) g) effects of mutant alleles on phenotype

1. SICKLE CELL ANAEMIA

* base substitution occurs in the gene coding for ~~glutamic acid~~ amino acid and sequence in β-globin

  CTT (glutamic acid) → CAT (valine)

  $Hb^A \longrightarrow Hb^S$

- EFFECTS -

① Hb becomes less soluble
② Molecules stick together and form long fibres inside RBC
③ RBC become sickle-shaped
④ distorted cells cannot transport oxygen, get stuck in small capillaries and block unaffected cells from passing

2. ALBINISM

- 'classic' form - autosomal recessive mutation
  → individuals that are homozygous for the recessive allele show the mutation

- other form - sex-linked and affects eyes only

* mutation occurs in the gene for enzyme TYROSINASE resulting in absence of tyrosinase or inactive tyrosinase in cells that produce melanin (MELANOCYTES)

tyrosine → DOPA → dopaquinone → melanin
(DOPA → dopaquinone: TYROSINASE)

- tirosinase is a trans-membrane protein found in organelles called MELANOSOMES in the melanocytes
- its an oxidase & has 2 ~~xxxxxxx~~ Cu atoms in its active site which bind an oxygen
- occurs in plants as well as animal tissues

- EFFECTS -

1. melanin totally / partially missing from eyes, skin & hair
2. eye pupils appear red
3. poor vision by rapid, jerken eye movement & by a tendency to avoid bright light

16.3 GENE CONTROL

a) genes & enzymes

x STRUCTURAL AND REGULATORY GENES

→ Structural genes code for proteins needed by a cell
- They may form part of the cellular structure or have different roles such as behaving as an enzyme

→ Regulatory genes code for proteins that regulate the expression of other genes
- The proteins that are produced bind to regulatory regions of a gene & increase / decrease level of transcription

x REPRESSIBLE & INDUCIBLE ENZYMES

→ synthesis of repressible enzymes can be prevented by binding a repressor protein to a specific site (OPERATOR) on a bacterium's DNA
→ an enzyme whose creation is inhibited when its reaction product is plentiful

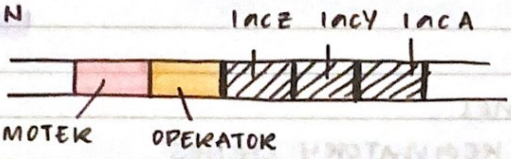
→ synthesis of an inducible enzyme occurs only when its substrate is present
→ Transcription of the gene occurs as a result of the inducer (the enzyme's substrate) interacting w/ the protein produced by the regulatory gene

- **transcription factors are**
  - proteins that bind to a specific DNA sequence
  - control of the formation of mRNA (control of the flow of info from DNA to RNA)

b) prokaryotic lac operon

- OPERON – a length of DNA making up a unit of gene expression in a bacterium
  - → consists of 1 or more regulatory genes &
  - → control regions of DNA that are recognised by the products of regulatory genes

THE LAC OPERON



- lac Z – codes for β-galactosidase (hydrolyses lactose to glucose + galactose)
- lac Y – codes for permease (allows lactose to enter cell)
- lac A – codes for transacetylase

→ close to the promoter, although not part of the operon is the regulatory gene for the lac operon