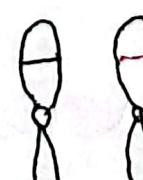


Chapt 16

Inheritance

- Q) What is **haploid (n)** and **diploid ($2n$)**?
- Haploid : one complete set of chromosomes.
 - Diploid : two complete sets of chromosomes.
- # Homologous pair of chromosomes :
- => two chromosomes - that carry the same genes in the same position.
 - => Same shape
 - => Same length
 - => one from male & another from female.
- 

Explain the need for a reduction division

during meiosis in the production of gametes

→ During fertilisation, nuclei of gametes fuse together to form the zygote.

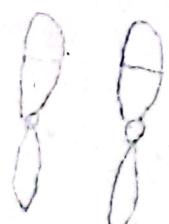
→ Gametes must be haploid for the zygote to survive after $n+n=2n$.

∴ So the first step of meiosis is REDUCTION Division.

It is a division that reduces the chromosome number to half.

To make gametes haploid.

So fertilisation restores diploid number.



Diploid



Haploid

Behaviour of chromosomes during Meiosis

Meiosis

Prophase I
Metaphase I
Anaphase I
Telophase I

prophase II
metaphase II
anaphase II
telophase II

Prophase I

- ⇒ Chromosomes start to coil and condense after some rearrangement
- ⇒ homologous chromosomes pair up and so bivalent formed (synapsis)
- ⇒ Nuclear envelope breaks and nucleolus may disappear
- ⇒ Crossing over of chromatids may occur, thus formation of chiasmata



Metaphase I

- ⇒ Bivalents line up on equator
- ⇒ independent assortment of spindle.
- ⇒ attached by centromeres.

Anaphase I

- ⇒ chromosomes move to poles
- ⇒ bivalents separate
- ⇒ by the shortening of microtubules
- ⇒ centromeres pull
- ⇒ whole chromosome move towards the opposite ends of spindle.
- ⇒ centromeres first pulled by microtubules

Telophase

- ⇒ nuclear envelope reforming.
- ⇒ nucleolus reforming.
- ⇒ chromatin formation.
- ⇒ cytokinesis

Reduction division

⇒ Dicccide \rightarrow haploid

Miosis II

Prophase II

- ⇒ Nuclear envelope and nucleus **disperse**.
- ⇒ Centrosomes and centrioles ~~rePLICATE~~ move to opposite poles of the cell.

Metaphase II

- ⇒ Chromosomes line up separately across ~~the~~ equator of spindle.

Anaphase II

→ Centromere divide and spindle microtubules pull the chromatids to opposite poles.

Telophase II

⇒ cytokinetic to form 4 haploid cell

Production of genetic variation

Crossing over

→ During prophase I, homologous chromosomes pair up and exchange genetic material.

⇒ Non-sister chromatids form chiasmata where crossing over occurs. ($X + X \rightarrow XX$)

- 3) Linkage groups broken
⇒ new combination of alleles.
- random orientation (independent assortment),
- During metaphase I, random arrangement of bivalent or homologous chromosomes occurs at the equator of spindle.

results in independent assortment

of chromosome at Anaphase I, II, and metaphase II

⇒ Produces many different chromosome combinations

Thus, reshuffling of genes during crossing over and independent assortment produce genetic variation produced by an individual.

So produce genetically different gametes

~~for random mating~~

⇒ Random fusion of gametes at fertilisation produces genetically different individuals.

Transmission, mutation & recombination result in descendants replaced by hybrid form which is refractory to environment.

~~Random mating leads to following results:~~

~~Role of genes in transmission:~~

⇒ Gene: A sequence of nucleotides that forms a section of DNA molecule that codes for a particular and specific polypeptide.

→ Each file contains many information.

⇒ Allele: Different form of a gene
which controls variation of
Same trait,
occupying some locus on
homologous chromosomes.

2) Locus: the position of a gene
on a chromosome.

⇒ Dominant: • An allele that always expresses itself
in the phenotype when present
• It influences the phenotype
even in the presence of an
alternate allele.

Recessive allele: only affect the pheno type if no dominant allele is present.

Co-dominant:

Each allele affects the phenotype iff present.

Phenotype

Observable features of an organism,

affected by genes and environment.

Genotype:

genetic constitution or allele present in an organism.

Heterozygous : two different alleles of a gene

Homozygous : two ^{identical} alleles of a gene.

F_1 generation: the offspring formed from the cross between two homozygous recessive and homozygous dominant genotype individuals.

$\text{TT} \times \text{tt}$ $\xrightarrow{\text{P1}}$ first generation of offspring \Rightarrow parents are homozygous (pure breeds)

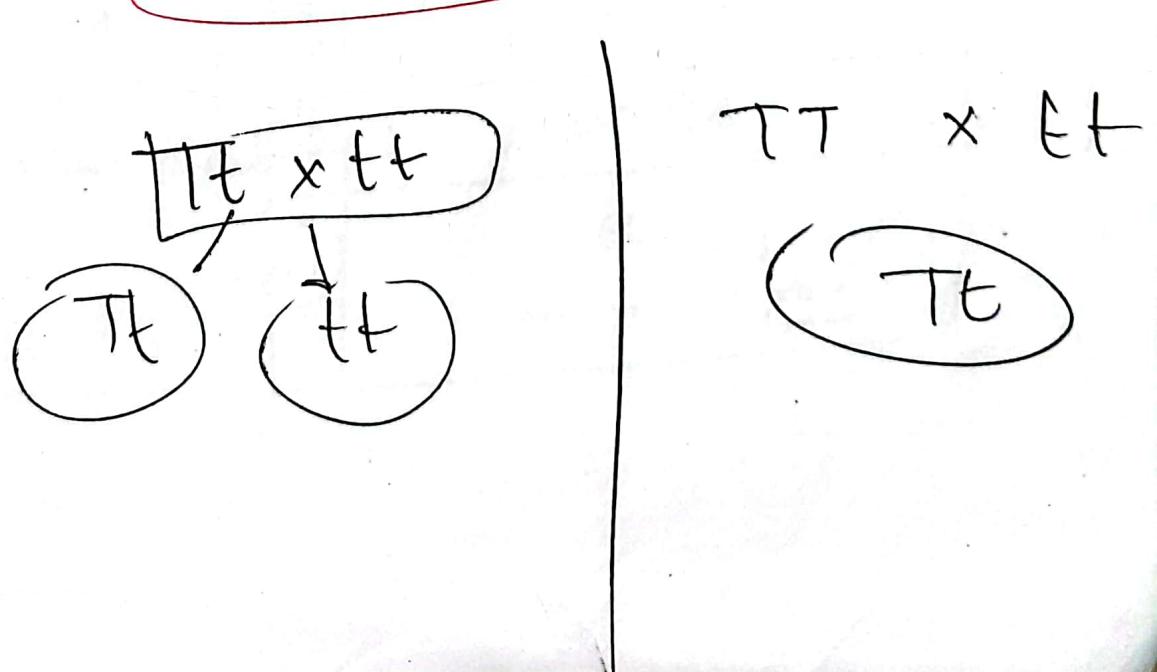
$\Rightarrow F_1$ generation will be heterozygous.

F_2 generation: the offspring formed from the cross between two F_1 individuals.

Test Cross: Cross between Dominant character individual and homozygous recessive organism.

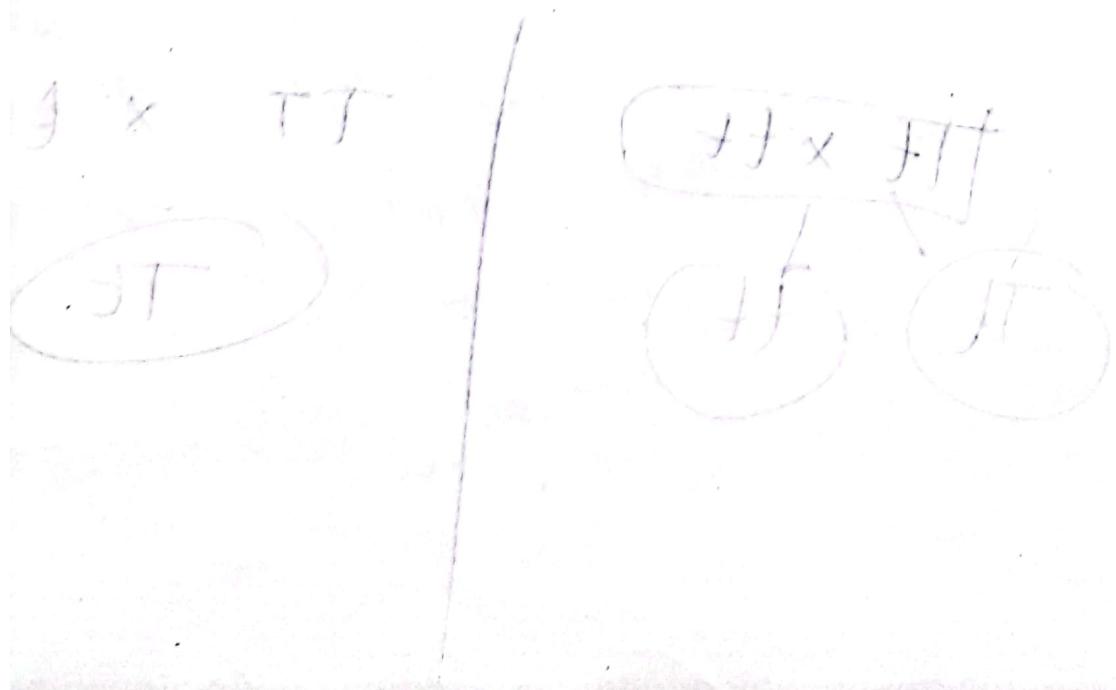
$\text{TT} \times \text{tt}$

↓
to see whether
the original
organism is
homozygous or
heterozygous



Sex-Linkage

- \Rightarrow Genes located in X-chromosome
are called Sex-linked.
- \Rightarrow Females have two copies of X-chromosome, controlling the same trait
- \Rightarrow Males have only one copy of the allele



Dihybrid Cross

Heterozygous dominant x homozygous recessive

A = allele for purple stem

a = allele for green stem.

A = dominant

a ~ recessive

D = allele for cut leaves

\perp = allele for prototrophic leaves^r.

Parental phenotypes

parental genotypes

~~parental~~ gametes

A and

(A)P Ad ad (A)l

aa dd

ad

Gametes
from the
other parent

game for from one parent

1

~~AaDd~~ AaDd

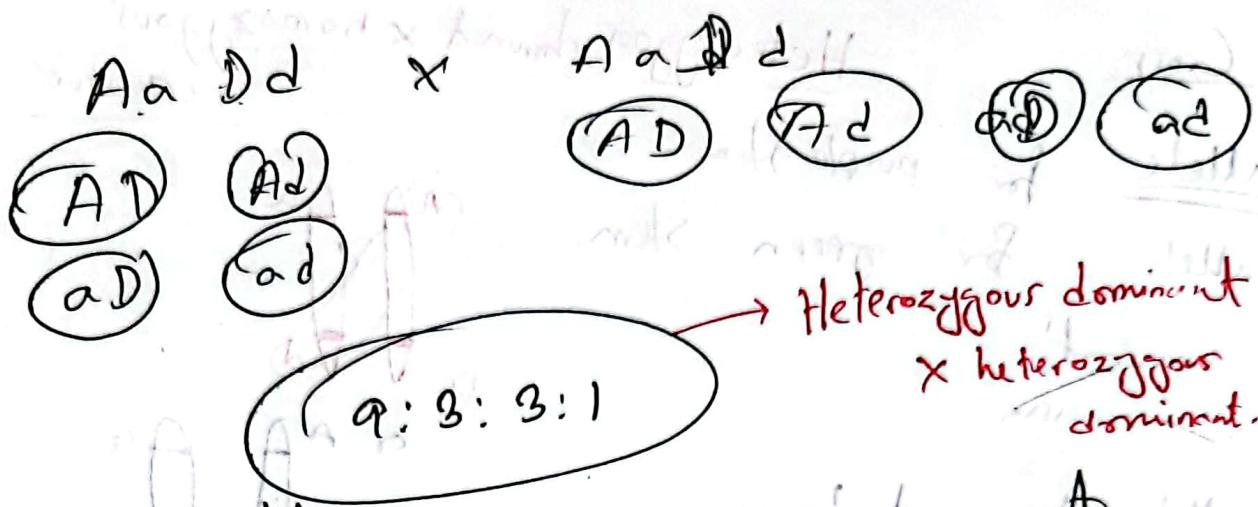
A a d d

add

58 d d

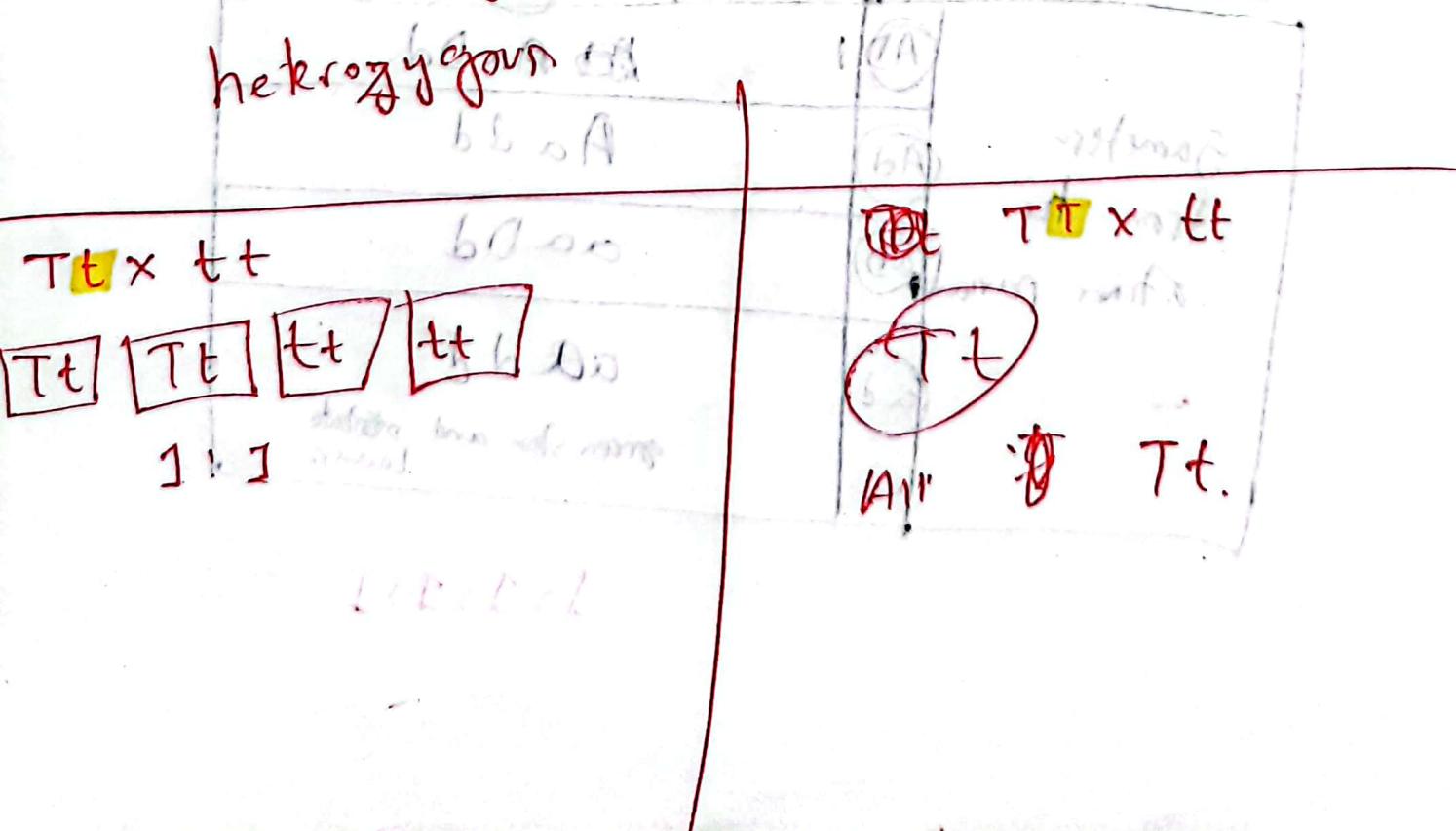
green stem and ^{purple} leaves.

1-1-1-1



Parents with heterozygous at
 both gene loci.

Test cross is used to identify whether
 the original organism is Homozygous or
 heterozygous.



How genotype of original parent can be determined.

⇒ The dominant character individual is

⇒ Test cross

with ^{homozygous} recessive allele - pure individual (genotype)

⇒ if all offspring showing dominant

characteristic, must be homozygous

⇒ If not all (1:1) offspring show dominance
must be heterozygous.

Dihybrid cross involve:

⇒ Dominance (Aa or AA)

⇒ codominance (Aa or aa)

⇒ multiple alleles ($AaBb$)

⇒ sex linkage (X^fX^f)

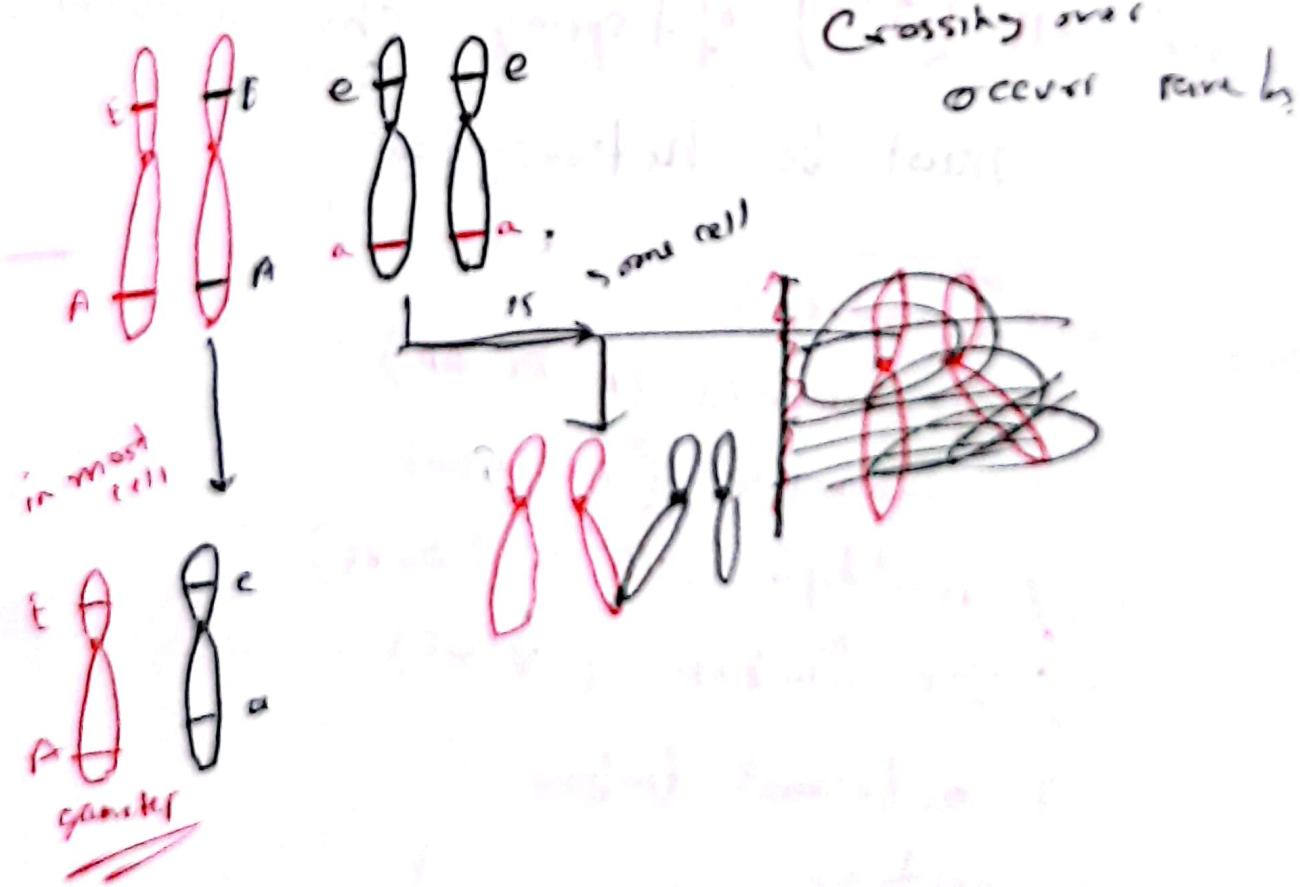
⇒ autosomal linkage

⇒ epistasis.

Autosomal Linkage

autosome: ~~any chromosome other than~~
sex chromosome

→ The presence of two genes on the same autosome, so that they tend to be inherited together and do not assort independently.



-) If the genes are on the same homologous pair of chromosomes -
-) genes would be inherited together
-) No independent assortment.
-) rare crossing over event.
ratio is different

~~TA x Aa~~

~~TAa~~

Epistasis : interaction of two genes at different chromosomes and different loci where one gene affects the expression of the other.

~~Expo~~

Investigation

The χ^2 test

It is used to test the significance of differences between observed and expected results.

Null hypothesis, H_0 : The difference between observed and expected values is insignificant.

$$\chi^2 = \sum \frac{(O-E)^2}{E}$$

degree of freedom:
 $df = n - 1$

n = number of categories

Identify critical χ^2 value at 0.05 probability.

Calculated χ^2 > Critical χ^2 value
 $p < 0.05$
null hypothesis is rejected

Calculated χ^2 < Critical χ^2 value
 $p > 0.05$
Null hypothesis accepted

If Null ~~rejected~~ accepted:

- ⇒ No significant deviation from expected
- ⇒ Difference between observed and expected values is insignificant
- ⇒ Difference is due to chance.

Why?
⇒ There is significant deviation difference
⇒ environmental effects
⇒ mutation ⇒ chance deviation

Genes, proteins and phenotype

③ H₂H
(no melanin)

① TYR gene; tyrosinase and albinism.

2) TYR gene Tyrosine \rightarrow DOPA \rightarrow Dopachinone \rightarrow melanin.

\Rightarrow normal gene product is tyrosinase

\Rightarrow converts Tyrosine to DOPA to Dopachinone to melanin.

\Rightarrow in ~~normal~~ melanin melanocytes

\Rightarrow mutant allele is recessive

\Rightarrow tyrosinase not produced or inactive

\Rightarrow affects hair, skin, iris

\Rightarrow only in homozygous recessive people.

HBB gene → haemoglobin, and sickle cell anaemia.

→ HBB gene codes for amino acids

Sequence in β -globin polypeptide

polypeptide in haemoglobin

→ Altered β polypeptide by mutation

causes amino acid sequence (Glu to Val)

A to T to A

by base substitution (~~A to T~~)

C TT → C AT.

stability of protein

→ Valine is non-polar

→ makes haemoglobin less

soluble.

→ causes ~~change~~ change

in tertiary structure,

- ⇒ Hb has lower affinity for O_2
- ⇒ RBC carry less O_2
- ⇒ becomes sickle shaped
- ⇒ Block blood flow as RBCs get stuck in capillaries causes pain.
- ⇒ becomes mutant allele homozygous

F8 gene, factor VIII, and haemophilia?

- ⇒ F8 gene codes for factor VIII
- ⇒ important for blood clotting.
- ⇒ synthesised in liver cells (Homozygous recessive mutant allele)
- ⇒ F8 gene is found on ~~X-chromosome~~ X-chromosomes.
- ⇒ So, it is a sex-linked gene.
- ⇒ mutant allele produces less or no factor VIII.
- ⇒ $X^f Y$ male with haemophilia
- ⇒ $X^f X^f$ abnormal female with normal factor VIII

H T T gene, huntingtin and Huntington disease

16. 2. 6

⇒ Mutant allele on chromosome 4.

⇒ If it is dominant

⇒ Normal recessive allele has 10-35 repeats of CAG.

) Mutant dominant allele has one more extra repeat of CAG.

Mutant allele produces abnormal

huntingtin protein.

↓
Used for the development of many diseases and brain

- ⇒ Neurological condition and brain problem.
- ⇒ Involuntary movements.
- ⇒ Cognitive ~~mot~~ mood changes.
- ⇒ mis-folded protein huntingtin.
- ⇒ larger number of repeats gives earlier onset.
- ⇒ Usual onset after 28 and before 65.

Le gene, gibberellin and stem elongation

- ⇒) Gibberellin synthesis controlled by gene Le / le.
- ⇒) Dominant Allele Le gives functional enzymes.
- ⇒) enzyme converts inactive to active gibberellin GAI.
- ⇒) GAI is a hormone causes cell division and cell elongation.
- ⇒) Recessive allele le forms non-functional enzyme.

→ The enzyme has one nucleotide different to dominant allele

- ~~a single amino acid~~
- ~~cause substitution in the primary structure of the enzyme~~
- ~~so changes the shape of active site.~~
- ⇒ No enzyme, no GA,

So plants unable to grow tall.

Explain

What is Gibberellin? (Q. 16.2.7)

- ⇒ It is a plant growth regulator or hormone
- ⇒ Stimulates cell division
- ⇒ cell elongation
- ⇒ changes plasticity of wall/cell wall.
- ⇒ plant grows tall
- ⇒ Dominant allele (Le) causes synthesis of enzyme.
- ⇒ enzyme catalyses the production of active form gibberellin from inactive form

2) recessive allele has only inactive form of gibberellin.

2) dwarf plants lack active form.

How gibberellin activates gene? (16. 3. 4)

1) GA binds to receptor complex

2) enzyme causes DELLA protein destruction
(see book diagram)

3) PIF and RNA polymerase binds to DNA as DELLA no longer binds

4) growth genes are switched on and transcribed

5) causes cell elongation PIF is the transcription factor

6) cell division

7) loosens cell walls

8) so cells can expand when water enters

Gene control

Structural genes

- ⇒ codes for non-regulatory / structural proteins.
- ⇒ functions within a cell.
- ⇒ Structural gene: λ lac Z; λ lac Y; λ lac A.
 - λ lac Z → β -galactosidase
 - λ lac Y → lactose permease
 - λ lac A → transacetylase
- ⇒ Protein synthesis occurs at ribosome where rRNA, mRNA and tRNA are associated.
- ⇒ Protein such as enzyme: β -galactosidase.

Regulatory genes:

- ⇒ code for regulatory / non-structural protein "repressor"
- ⇒ Control the message of expression of other genes.

2) Gene coding for repressor protein

are Lac I or gene for

transcription factor. operator and over binding to promoter sites

⇒ Repressor coded for binds to promoter sites binding of RNA polymerase

⇒ Regulatory genes switch on and off transcription and thus gene expression

⇒ ~~bind to inducer molecules~~
~~bind to DNA binding sites~~
Repressible vs inducible enzymes

Repressible enzyme info 20 162-166

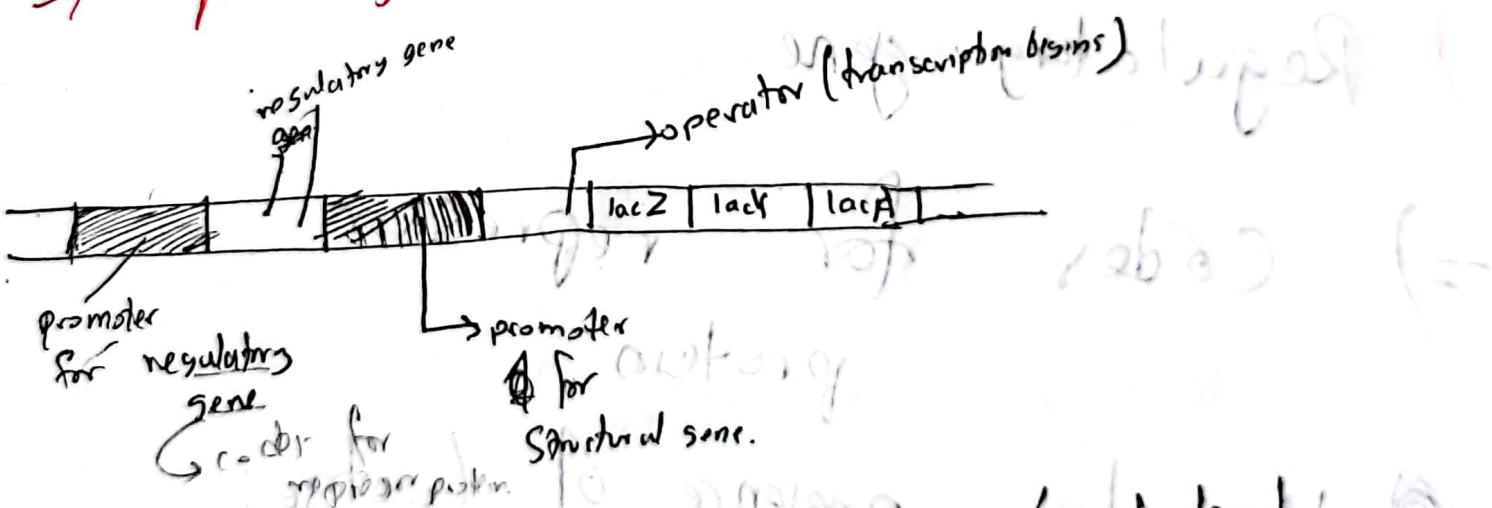
⇒ Generally produced continuously

⇒ Synthesis can be prevented by binding to repressor

ratio of protein to specific site / promoter / operator.

Inducible enzyme

- ⇒ synthesis only occurs when substrate is present.
- ⇒ Transcription of the gene only occurs when substrate binds to repressor protein.
- ⇒ β -galactosidase.



β -galactosidase: hydrolyses lactose to glucose and galactose

⇒ Promoter: Region where RNA polymerase binds.

⇒ Operator and repressor protein binds.

Explain the genetic control of protein production in prokaryote using the lac operon.

- ⇒ In bacteria, cluster of genes called **lac operon** is discovered which works in coordination.
- ⇒ Regulatory gene.
- ⇒ Coder for repressor protein.
- ⇒ ~~Off~~ presence of lactose:
- ⇒ Lactose binds to repressor protein.
- ⇒ Repressor protein changes shape

- no longer binds to operator at DNA
- Transcription continues to protein from three structural genes.

absence of lacZ

- repressor protein blocks promoter
- RNA polymerase cannot bind to promoter
- mRNA is not synthesised
- So enzyme like β -galactosidase is not synthesized.
- so no waste of amino acid/ATP

⇒ Transcription factors are

proteins that bind to DNA and are involved in the control of gene expression.

of gene expression in eukaryotes

by decreasing or increasing the rate of transcription.

How?

- ⇒ TF can form part of protein complex.
- ⇒ by binding to DNA or promoter
- ⇒ So RNA polymerase binds to promoter.
- ⇒ Transcription starts and mRNA is synthesized so gene switched on

or TF binds to DNA no transcription occur so gene switching off

2) can activate genes in correct order or time.

couple this activate

naturally has to

do this (it's probably needed)

that couple things

and it's the binding

ways to switch

switching can make several ways

of turning on/off

and when and when

and when