

(Part-1)

UNIT-III B.Tech., F.Y.G.I QUESTION PAPER ①

Terminology used in Genetics:

① Dominant gene / trait: is that which expresses itself in heterozygous / hybrid condition.

② Recessive gene / trait: is that which is not able to express itself in hybrid condition & expresses only in homozygous condition.

③ Allele: Alternative form of the same gene.

④ Homozygous trait / Pure character: organism Trait having identical genes controlling it.

⑤ Heterozygous / Hybrid: Trait having different alleles controlling it (unlike).

⑥ Genotype: Genetic makeup of an individual.

⑦ Phenotype: Expression or observable structural & functional traits produced due to interaction of genes & environment.

Genotype → Phenotype.

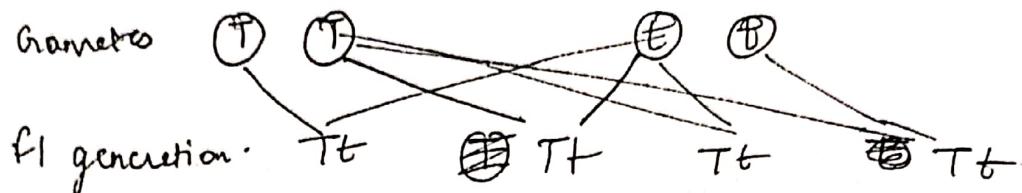
MENDEL'S LAWS OF INHERITANCE

Father of Genetics: Mendel performed experiments on garden pea, 'Pisum sativum' and chose 7 pairs of contrasting characters in it. He proposed 'laws of inheritance' based on his experiments.

Experiments consist of Monohybrid & Dihybrid cross.

① Monohybrid cross: Mating experiment where a single pair of character is used to determine the role of inheritance.

Parents TT X tt
 Tall dwarf



Grametes

Fl cross

\textcircled{P}	\textcircled{T}	\textcircled{T}
\textcircled{T}	TT	Tt
\textcircled{D}	Tt	tt

f₂ generation - TT : Tt : tt
 Cisnotypic ratio = 1 : 2 : 1

Phenotypic ratio = 3 : 1
Tall : dwarf

Dihybrid cross: Mating experiment where ^{two} pairs of contrasting characters are used to determine the role of inheritance; (on next page)

Parents $RRYY$
Round yellow seeds

$rryy$
wrinkled green seeds

Granules

RY

RY

ry

ry

F₁ generation $RrYy$ $RrYy$

$RrYy$

$RrYy$

Granules :

RY

Ry

rY

ry

F₂ generation →

RY

Ry

rY

ry

RY

$RRYY$

$RRYy$

$RrYY$

$RrYy$

~~Counting~~

F₂ Phenotypic ratio — 9 : 3 : 3 : 1

Round
yellow

wrinkled
yellow

Round
green

wrinkled
green

Mendel's laws of inheritance:

① Law of Unit Factor: States that the various hereditary characters are controlled by factors (genes) & there is a pair of factors controlling each character.

② Law of Dominance: States that out of a pair of contrasting characters, only one expresses itself in the hybrid. The one that expresses itself is said to be dominant over other which remains suppressed (unexpressed) & is called recessive.

③ Law of Segregation: When the factors that determine a pair of contrasting characters are brought together in a hybrid, they do not intermix. During gamete formation, the factors of a pair become segregated such that each gamete receives only one factor for a character. Hence, this law is also known as Law of purity of gametes. It is based on Monohybrid cross.

④ Law of Independent Assortment: States that in the inheritance of more than one pair of characters, the factors responsible for each pair of characters are distributed independently to the gametes i.e. assorted independent of those of other pair. This law is based on dihybrid cross.

CENTRAL DOGMA OF MOLECULAR BIOLOGY:

- Central dogma of Molecular Biology: was first enunciated by Francis Crick in 1958.
- It means the flow of information from DNA to mRNA and then decoding the information present in mRNA to form polypeptide/protein.

DNA $\xrightarrow{\text{Transcription}}$ mRNA $\xrightarrow{\text{Translation}}$ Polypeptide (Protein)

- The first step in this flow of genetic information is transcription i.e. formation of complementary mRNA from a DNA strand.
- Transcription is followed by translation: i.e. formation of polypeptide in a particular sequence of amino acid as directed by mRNA.
- Four letter code language of DNA is transcribed into four letter language of mRNA which is then translated into twenty letter code of protein.

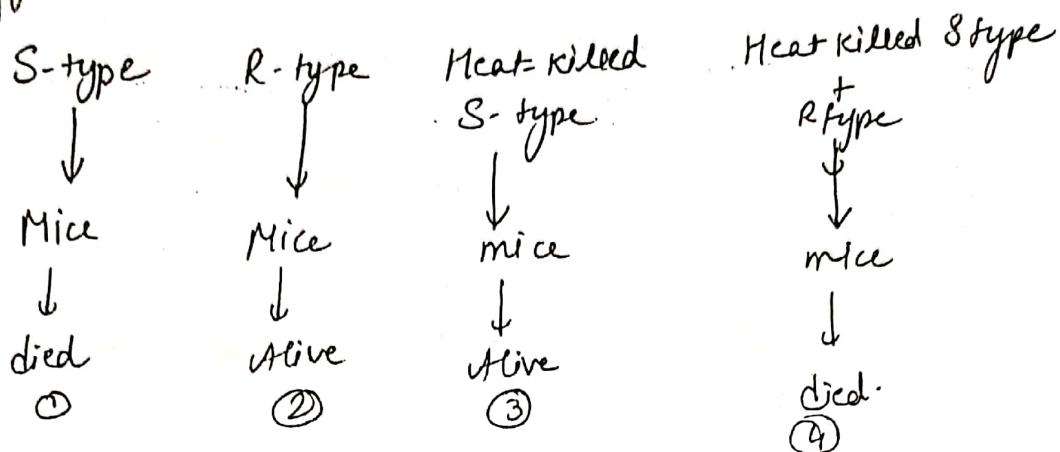
However in Retrovirus (RNA virus), central dogma reverse operates, where RNA first synthesizes DNA & then ~~the~~ the information flows as usual

DNA $\xrightarrow{\text{Transcription}}$ mRNA $\xrightarrow{\text{Translation}}$ Polypeptide
Reverse transcription.

EXPERIMENTS TO PROVE DNA IS THE GENETIC MATERIAL (NUCLEIC ACID)

In 1928, Griffith experimented on Diplococcus pneumoniae, a bacteria that causes pneumonia and suggested a transforming substance. Diplococcus pneumoniae has two strains — one with polysaccharide capsule is smooth

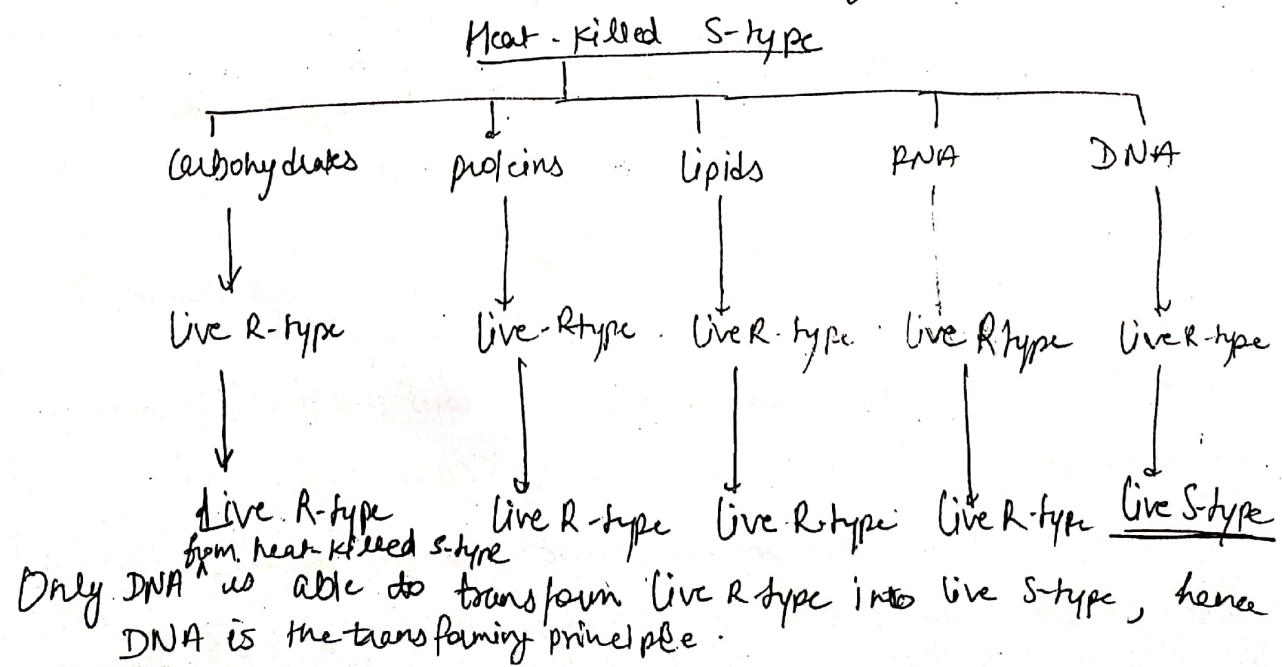
strain (S-strain) and one without capsule is rough strain (R-strain). The S-type is virulent (causes disease) & R-type is non-virulent.
 Griffith conducted the experiment with 4 cases as follows:



The bacteria isolated from died mice in ④th case were S-type. So it was concluded that there was a transforming principle that transformed live-R-type into live S-type in ④th case.

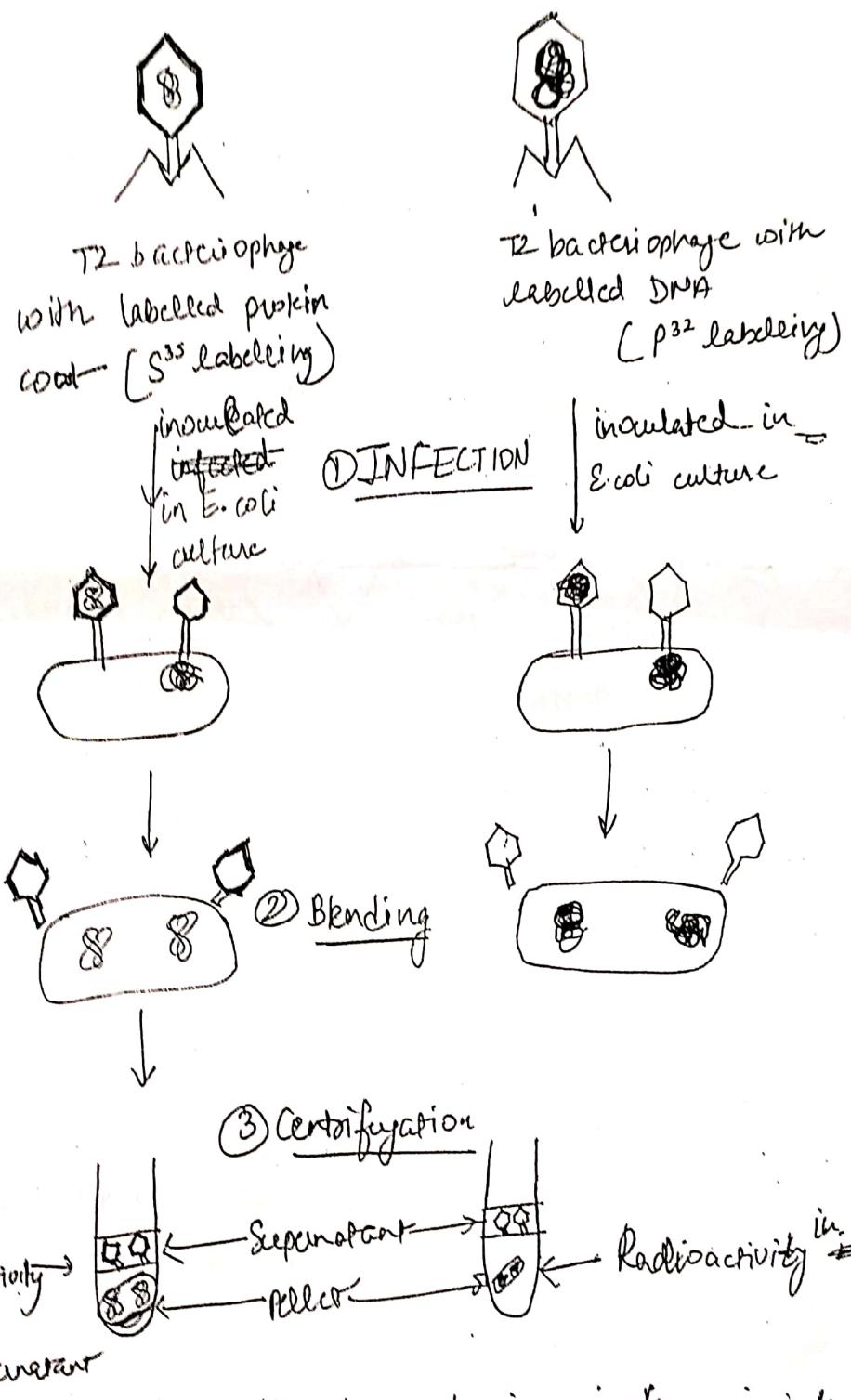
However, the nature of "transforming" principle was not known.

In 1944, Avery, MacLeod & McCarty repeated Griffith's experiments to find out the transforming principle.



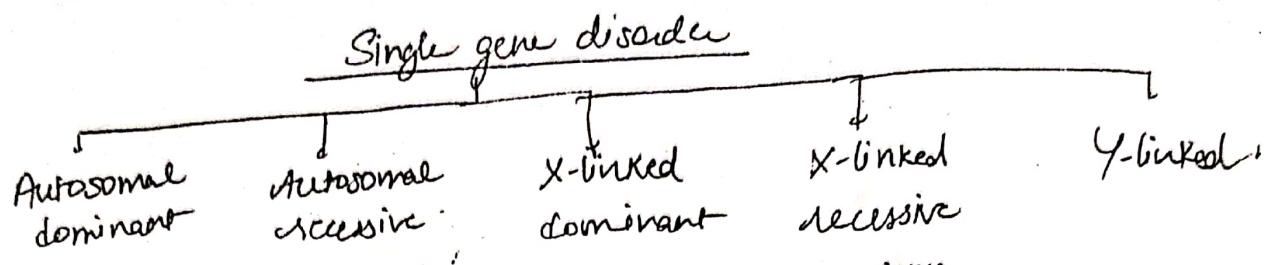
Only DNA is able to transform live R-type into live S-type, hence DNA is the transforming principle.

Experiment 3 : Hershey & Chase, in 1952, is based on the fact (4)
 DNA contain Phosphorus (hence can be labelled with P^{32})
 and no sulphur ; proteins contain Sulphur but no phosphorus
 (hence can be labelled with S^{35}).



DNA is the transforming principle principle which gets transferred to *E. coli* from T2 bacteriophage.

SINGLE GENE DISORDERS : Also known as Mendelian disorders : These occur when changes / mutations occur in only a single gene

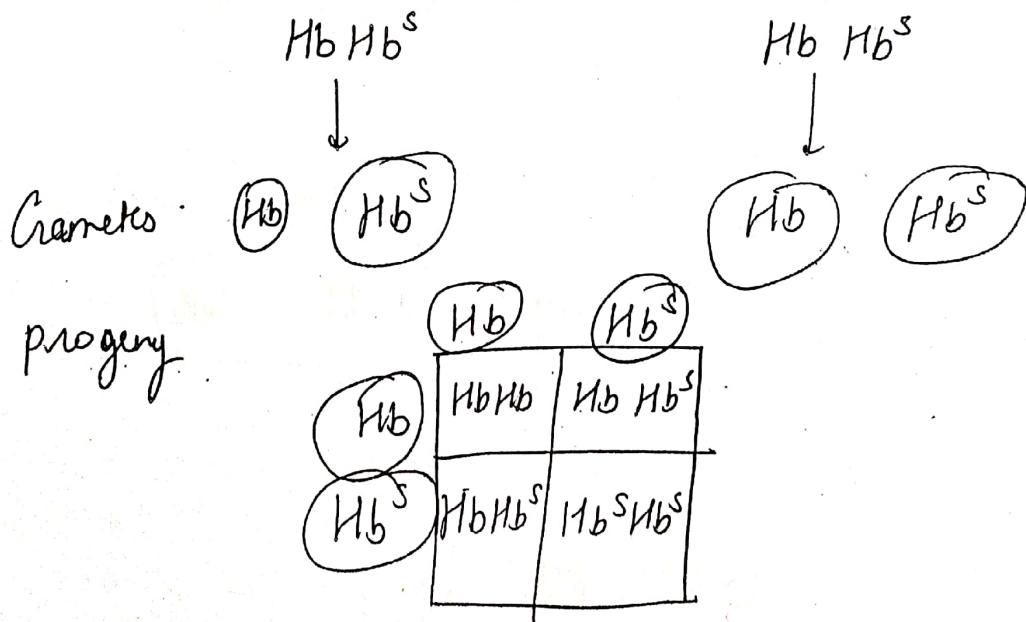


① Autosomal dominant disorder occurs when ^{even} only one mutated copy of the gene that is ~~located~~ on the autosome is present.

Eg. Marfan syndrome

② Autosomal recessive disorder occurs when both the copies of the gene are mutated / defective & those genes are present on autosome. For e.g. sickle cell anaemia.

CARRIER father \times CARRIER mother



Probability of having normal child (Hb⁺Hb) = $\frac{1}{4} \approx 25\%$

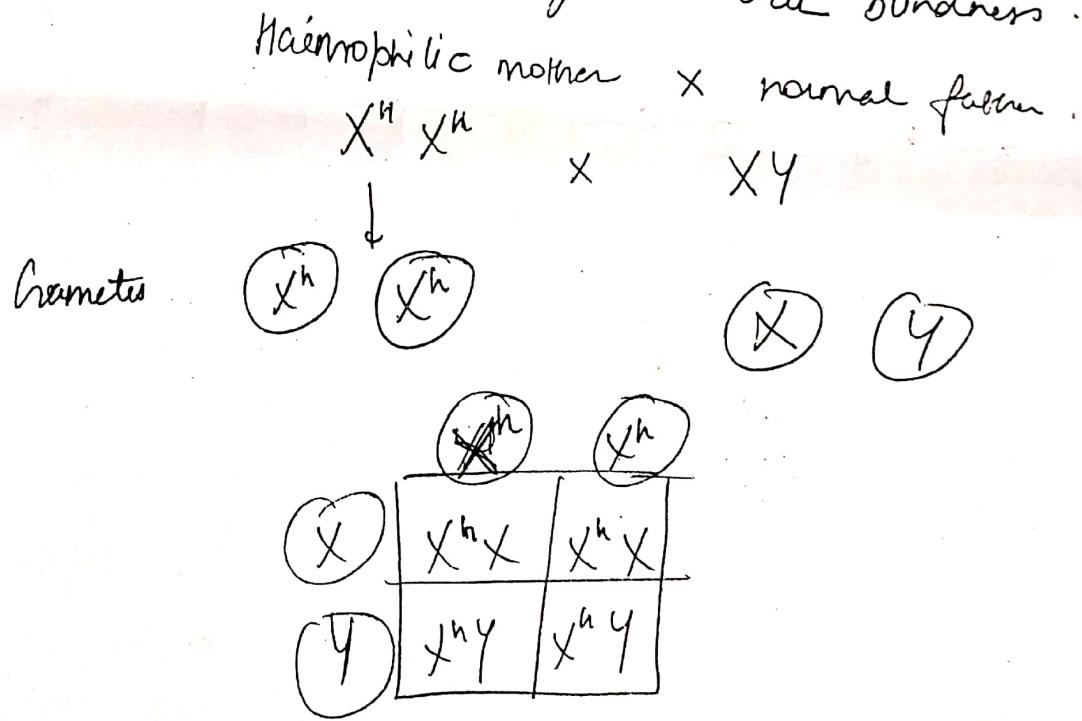
" " " carrier = $\frac{1}{2} \approx 50\%$

" " " affected = $\frac{1}{4} \approx 25\%$

(3) X-linked dominant disorder: occur when mutated gene is present on X-chromosome and even a single defective gene is able to cause disease. Eg. Rett's syndrome. However, since males only have one copy of genes on X-chromosome, they...

(4) X-linked recessive disorder: occurs in females when both the copies of the gene present on X-chromosome are mutated / defective. However, in males, since only one X-chromosome is present, if the gene present is defective, males will be affected. In other words, sons of unaffected mothers will always be diseased.

For ex: Haemophilia, Red green colour blindness.



100% sons \rightarrow haemophilic

100% daughters \rightarrow carriers.

Y-linked disorders: are caused by defective gene present on Y-chromosomes. Only males can get them & all the sons of affected father are affected.

GENETICS OF BLOOD GROUP

(5)

- The 'A BO' Blood group system is controlled by three alleles I^A , I^B & i . I^A encodes for A antigen, i.e. blood group shows multiple allelism.
- I^B encodes for B antigen & 'i' does not encode for any antigen.
- I^A & I^B are codominant & each is dominant over 'i'
- The antigens encoded are present on surface of RBCs.

	Allele from Parent 1	Allele from Parent 2	Genotype	Blood group	Antigen	Antibody
①	I^A	I^A	$I^A I^A$	A	A	anti-B Antibody
②	I^A	i	$I^A i$	A	A	anti-B
③	I^B	I^B	$I^B I^B$	B	B	anti-A
④	I^B	i	$I^B i$	B	B	anti-A
⑤	I^A	I^B	$I^A I^B$	AB	A, B	No Antibody
⑥	i	i	ii	O	No Antigen	anti-A, anti-B

Genotypes = (6)

Phenotypes = 4 (A, B, AB, O)

Rh Blood group system :- If the individual possess

- Rh+ antigen, he is Rh+ & if Rhesus antigen is absent, he is Rh-.

Universal donor : That individual that does not have any antigen on its RBC. i.e. O-.

Universal acceptor : That posses all the antigens on RBCs i.e. AB+ (A antigen, B antigen, ABO antigen)

Type I diabetes : is also called 'juvenile diabetes' or 'Insulin-dependent diabetes mellitus' (IDDM). It is an autoimmune disorder characterized by loss of insulin producing β -cells of pancreas leading to deficiency of insulin.

- Also called "juvenile diabetes" : it represents majority of diabetes cases in children.

Signs & symptoms : Frequent urination (polyuria), unusual thirst (polydipsia), extreme hunger, unusual weight loss, blurred vision, extreme fatigue & irritability.

- If not treated properly with insulin, they may develop fatal ketoacidosis.

Type II diabetes : Non-insulin dependent diabetes mellitus (NIDDM). It develops from insulin resistance & is the most common type. The defective responsiveness of body tissues to insulin involves insulin receptor in the ~~membrane~~ cell membrane.

- It mainly occurs in middle aged & elderly persons.
- Obesity, stress & other environmental factors play important role in etiology of Type-II diabetes.
- In early stages reduced insulin sensitivity results in elevated levels of insulin in the blood. At this stage, hyperglycemia (\uparrow level of glucose in blood) can be reversed by a variety of measures & medications that improve insulin sensitivity.

Symptoms : include any type-I symptoms (except weight loss), frequent infections, blurred vision, cuts/

bruises slow to heal, numbness in hands / feet & recurring skin, gum / bladder infections.

Prevention / Delaying the onset of diabetes :-

- Patient education, exercise, self-monitoring of blood glucose can keep blood glucose level within limits
- Vit.D supplements, antibody to beta cell proteins can lower the incidence of Type-1 diabetes.
- Making changes in diet & increase physical activity can reduce the risk of Type-1 diabetes.
- Maintaining a healthy weight

Treatment :-

- delivery of artificial insulin via injection is the principal treatment of Type-1 diabetes.
- use of insulin pumps.