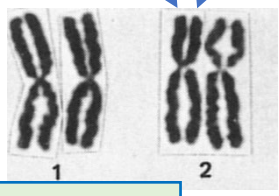
Sister Chromatids
Exactly identical copies
Same alleles for every gene

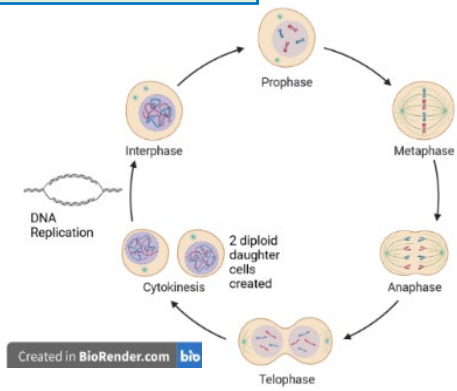


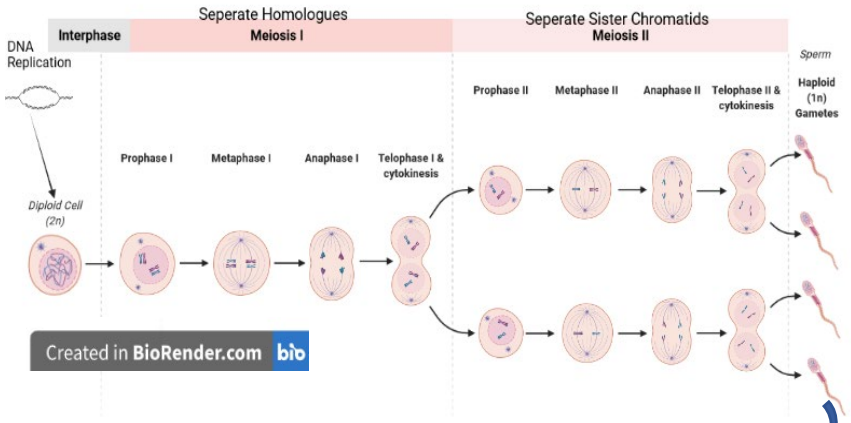
Meiosis:

1 round of replication followed by 2 rounds of cell division

4 Gamete (sperm or egg) cells are produced. These cells are haploid (1n)

Mitosis:





Fertilization:

Egg

Haploid (1n)

1 copy of every chromosome

+

Sperm

Haploid (1n)

1 copy of every chromosome

=

Zygote

Diploid (2n)

1 copy of every chromosome from Mom
+
1 copy of every chromosome from Dad
=
2 total copies of every chromosome

Chromosomally typical humans have 23 unique chromosomes so n=23. 2n=46 total chromosomes

Euploidy (normal number of chromosomes):

2n: XX XX XX XX

Atypical chromosome number:

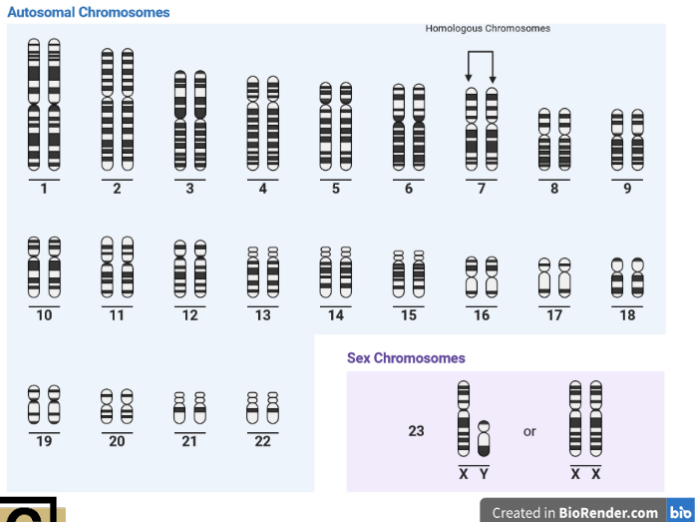
This can be a result of non-disjunction (incorrect chromosome splitting in meiosis):

Aberrant Euploidy: Full extra or missing set of all chromosomes
4n:
XX XX XX XX
XX XX XX XX

Aneuploidy: one or a few extra or missing chromosomes
2n+1:
XXX XX XX XX

Karyotype:

A visual representation of all chromosomes in an individual. *Typically shown directly following mitosis (cell division) and so it only has one copy of each chromosome from each parent.



L R C

Mutations:

*Hint: Only say something is a mutation if you are told it is. Many alleles in present day came from a mutation, but you would not tell that person they are a mutant. *

Haplosufficiency: This refers to a gene!

Haplosufficent: One wildtype allele is enough to produce the wildtype phenotype.

- The WT allele is dominant to the Mut allele.
- The mut allele is recessive.
- If a gene is haplosufficent, a heterozygous individual will hae a wildtype phenotype.

Haploinsufficent: One wildtype allele is NOT enough to produce the wildtype phenotype.

- The mut allele is dominant to the WT allele.
- The WT allele is recessive.

Silent mutation: changes in nucleotide sequence does not result in a change in amino acid sequence and thus the same protein is made.

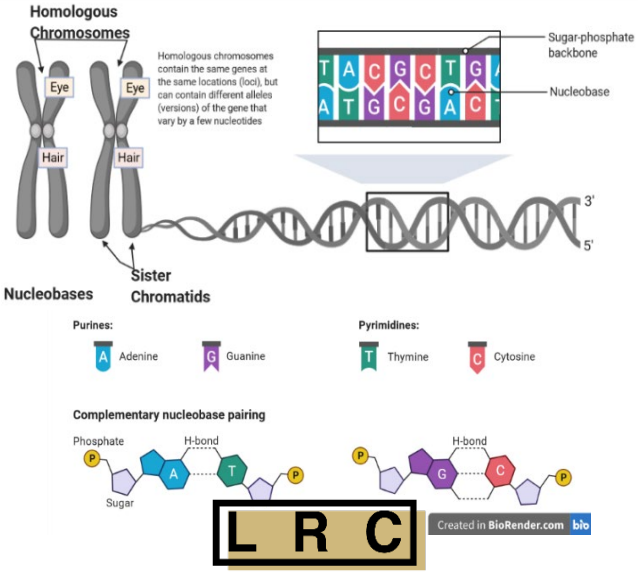
Missense Mutation: Changes in nucleotide sequence results in a change of one/a few amino acids. This can result in a small change in protein function (a leaky mutation).

Nonsense mutation: Changes in nucleotide sequence results in a large change in amino acid sequence and protein length due to a pre-mature stop codon. These result in null mutations which are characterized by the protein having a completely different function. Frameshift mutations (from insertions and deletions) and changes to promoter and terminator regions often cause null mutations

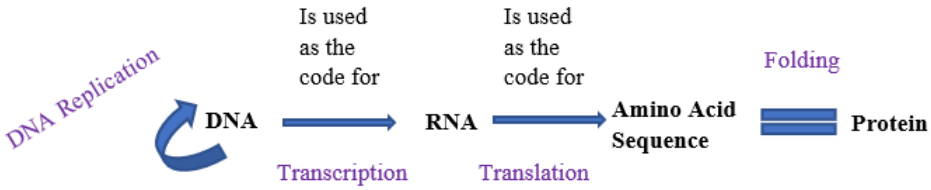
Each chromosome is comprised of two complementary strands of DNA running antiparallel to each other intertwined in a double helix.

SNP (Single nucleotide polymorphism): A change in DNA sequence where there is a change in one base for another base.

A different allele has a different (yet similar) nucleotide sequence to a wildtype allele. Small changes *CAN* code for a different protein and thus a different phenotype because it could be in a transcribed region of the DNA, the translated region of the mRNA transcript and could result in a codon that codes for a different amino acid.



Central Dogma of Biology:



A change in the DNA sequence COULD change the mRNA transcript.

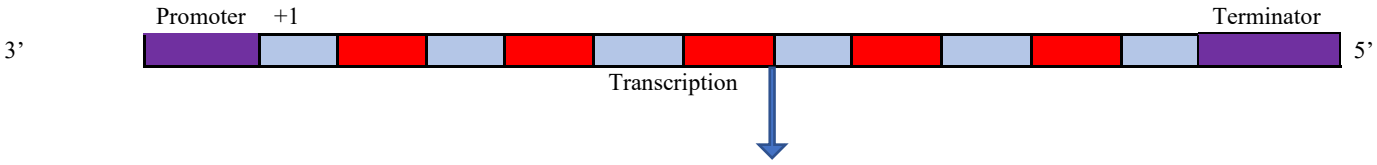
A change in the mRNA transcript COULD change the amino acid sequence.

Mutations can sometimes (not always) result in a change in protein, protein function, and phenotype.

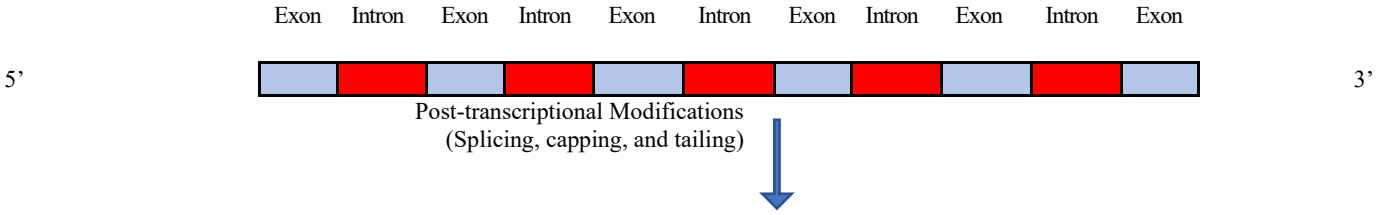
*DNA is read 3' to 5' to synthesize a daughter strand of DNA or transcribe a mRNA transcript that is 5' to 3'.

mRNA is read 5' to 3' to translate an amino acid sequence N-terminus to C-terminus

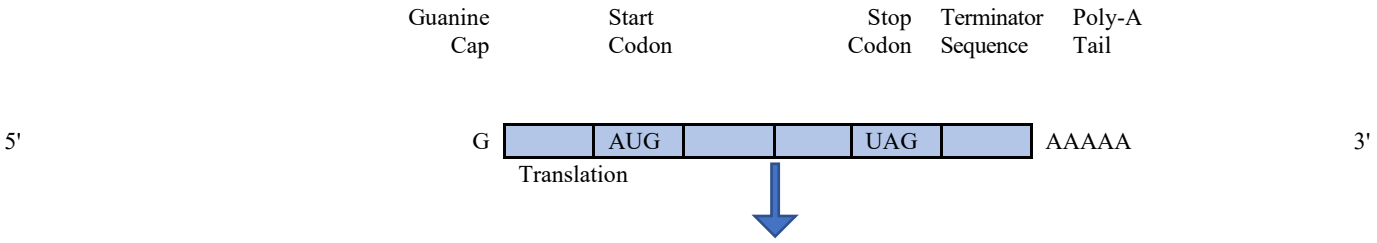
DNA



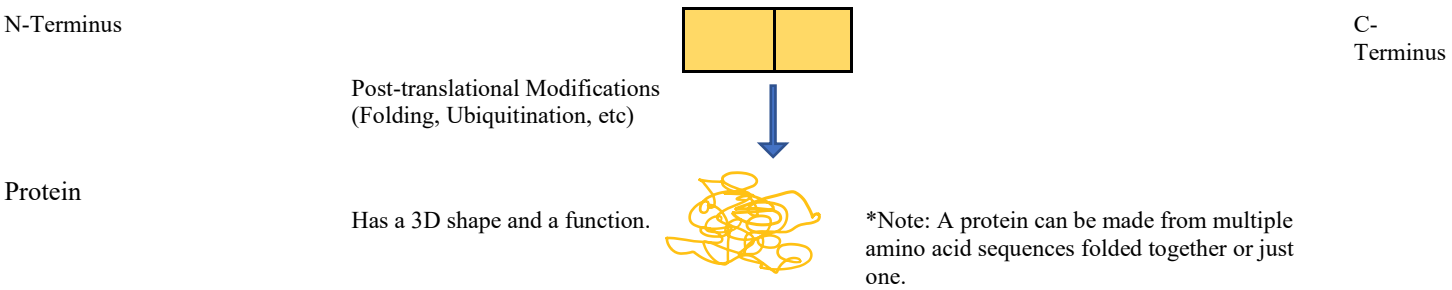
Pre-RNA (Immature RNA)



Mature RNA



Amino Acid Sequence

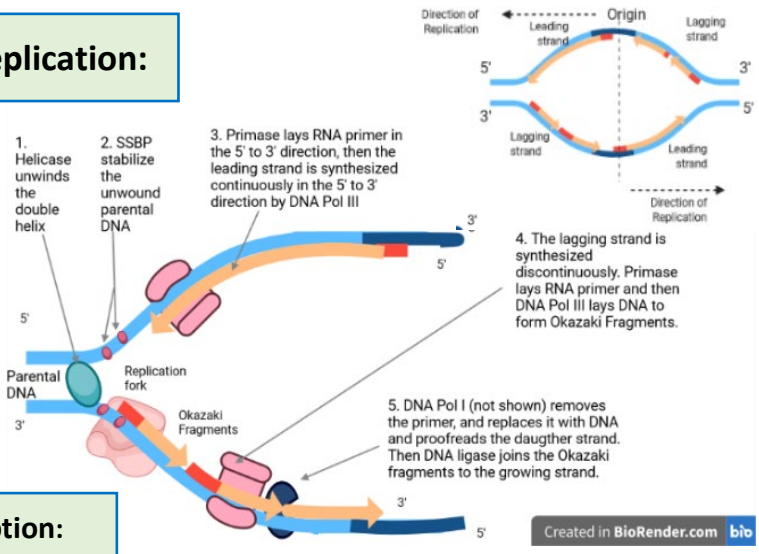


DNA Replication:

Leading strand: Continuous, made in the same direction as the fork opens.

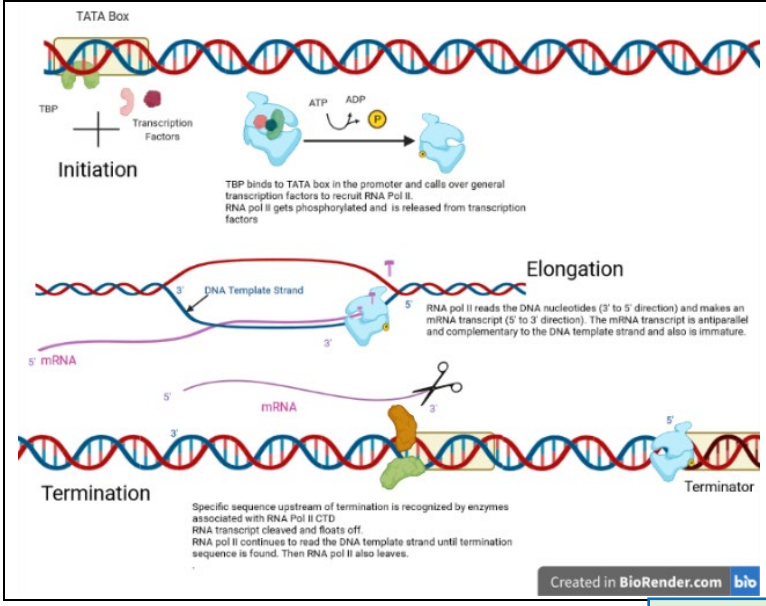
Lagging strand: Made in small pieces (Okazaki fragments) because it is made in the opposite direction as the fork opens.

Every new daughter strand of DNA has a leading and lagging strand

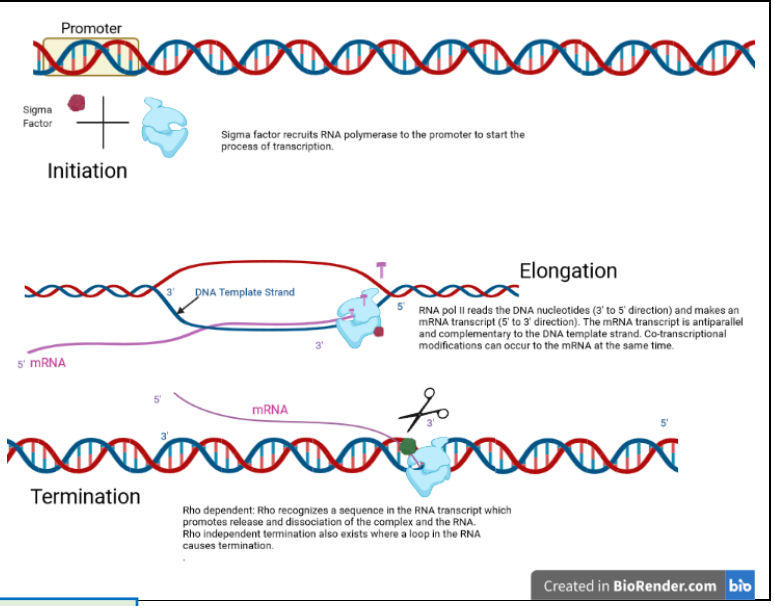


Transcription:

Eukaryotes

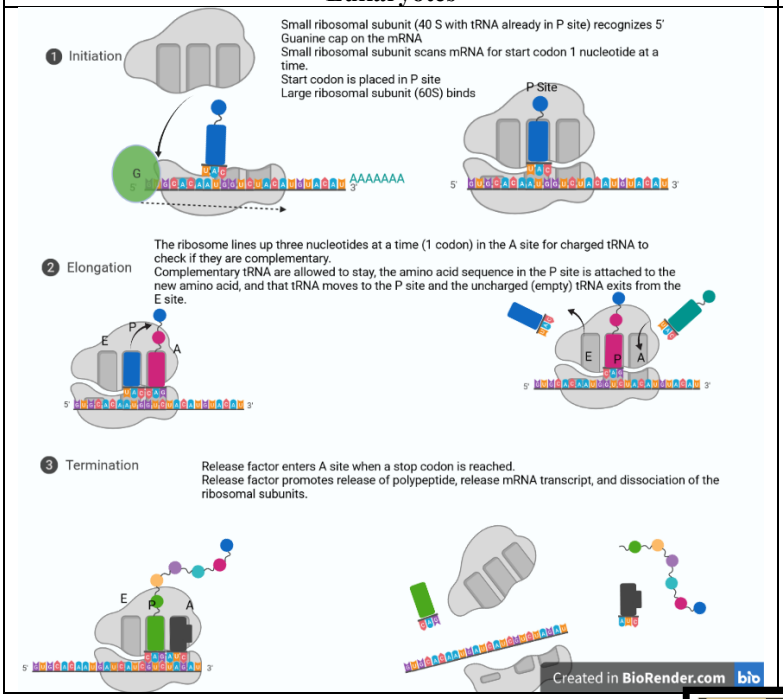


Prokaryotes

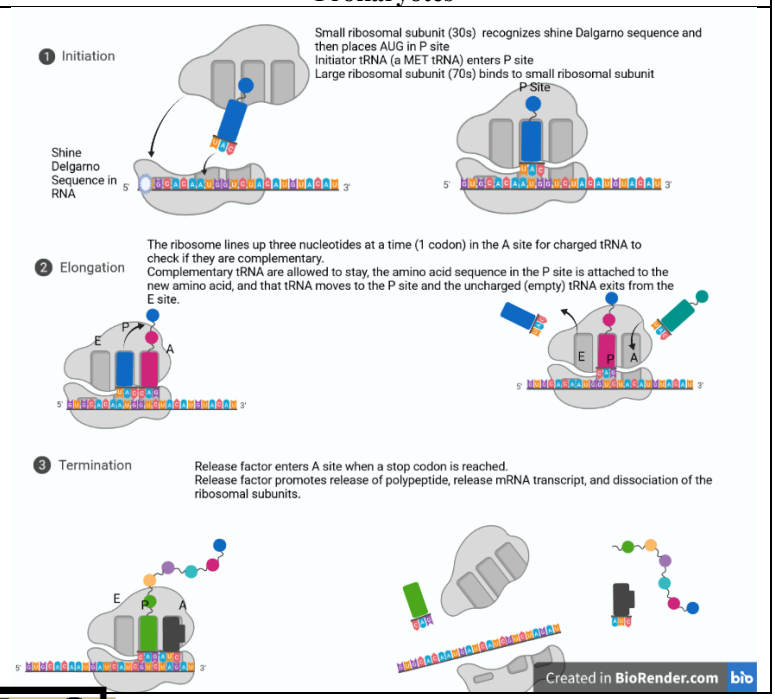


Translation:

Eukaryotes

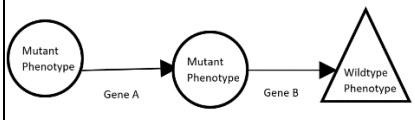
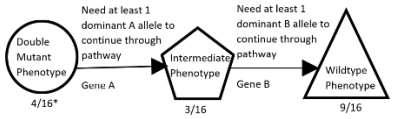
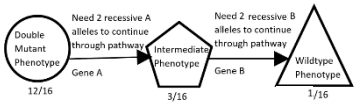


Prokaryotes



L R C

Ratios:

Parents: **AaBb x AaBb** * Take note of the collapsed phenotypic classes. *							
Dihybrid Cross Unlinked, un-interacting genes		Complementation		Recessive Epistasis		Dominant Epistasis	
		WT=Dom; Mut=Rec.		WT=Dom; Mut=Rec.		WT=Rec; Mut=Dom.	
4 phenotypic classes		2 phenotypic classes		3 phenotypic classes		3 phenotypic classes	
						
A/- B/-	9	A/- B/-	9	A/- B/-	9	A/- B/-	9* 12*
A/- b/b	3	A/- b/b	3*	A/- b/b	3	A/- b/b	3*
a/a B/-	3	a/a B/-	3* 7*	a/a B/-	3* 4*	a/a B/-	3
a/a b/b	1	a/a b/b	1*	a/a b/b	1*	a/a b/b	1
		Must have at least one wildtype A and B gene to have a wildtype phenotype.		Any dominant wildtype alleles will allow continuation through the pathway		Any dominant mutant alleles will result in not continuing past that step in the enzyme pathway.	

To be allelic means to be at the same loci.

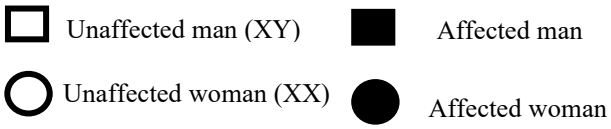
Non-allelic genes are complementary to each other and when crossed produce a wildtype phenotype.

For epistasis, pay attention to each step of the pathway and which genotype is needed to continue through pathway.

Parents: **AaBb x aabb**	
Dihybrid Test-Cross Unlinked un-interacting genes	
4 phenotypic classes	
AaBb	1
Aabb	1
aaBb	1
aabb	1

Pedigrees & Modes of Inheritance:

*Assume that anyone mating into the family is homozygous wildtype. *



Steps for determining mode of inheritance:

1. Is the mode of inheritance mitochondrial?
   - All affected mothers always pass down the disorder to all offspring regardless of sex.
   - Only mothers can pass down the disorder.
   - No carriers or skipping of generations
2. Is there a sex-bias?
   - If yes, proceed to sex-linked.
   - If no, proceed to autosomal.

*** You must state what type of autosomal or sex-linked mode of inheritance it is. Simply stating "sex-linked" is not a full answer for a mode of inheritance. ***

Autosomal:

Autosomal dominant:

- Many affected individuals.
- No skipping of generations.
- No carriers.

Autosomal recessive:

- Fewer affected individuals
- Skipping of generations and carriers
- Only homozygous recessive (homozygous mutant) individuals have a mutant phenotype

Sex-linked:

X-linked dominant:

- More females than males affected
- No carriers or skipping of generations
- Mothers with a mutant phenotype *COULD* pass the mutation to offspring of either sex
- Fathers with a mutant phenotype *WILL ALWAYS* pass the mutation to all (and any) daughters.

X-linked recessive:

- More males than females affected.
  - Females need 2 mutant alleles to show the mutant phenotype.
- Carriers and skipping of generations possible.
- Mothers with a mutant phenotype *COULD* pass the mutation to offspring of either sex. Sons of mutant phenotypic mothers will always have a mutant phenotype.
- Mothers with a mutant phenotype *WILL ALWAYS* pass a mutant allele to all (and any) daughters, but these daughters may have a wildtype allele from mom and be carriers.

Y-linked:

- Only males can have this disorder.
- Dads with a mutant phenotype will always pass the disorder to all sons and all sons then will be affected.
- No carriers or skipping of generations

Horizontal Gene Transfer:

Name of the type of horizontal gene transfer	What happens	Requirements	Important information & distinguishing factors for this type of horizontal gene transfer
Conjugation	A bacterial cell donates genetic material to another bacterial cell through a sex pilus.	The donor must have a fertility factor. The fertility factor in donor can be in two places: • F^+ plasmid (more easily shared) • In the chromosome (called Hfr).	Interrupted mating experiments use conjugation. An exconjugant is the recipient after it incorporates new DNA into its chromosome or a plasmid. DNA enters as a single linear strand through the sex pilus. The sex pilus could break at any time and conjugation ends. Genes close to the origin of transfer are more likely to be received by a recipient than genes further away.
Transformation	A competent bacterial cell picks up DNA from the environment. This DNA could have come from a live or dead bacterial cell (or from a scientist putting it there).	The bacterial cell must encounter DNA in its environment. The recipient must be competent.	DNA can only enter as ssDNA because of the size of the pores. A competent bacterial cell has a membrane that is permeable to the DNA (so it can enter the cell). A competent bacterium is needed for transformation. A bacteria can be made competent in a lab.
Transduction	A bacteriophage picks up bacterial DNA in its capsid along with viral DNA. When it infects a bacterial cell, this recipient bacterial cell receives new DNA.	A bacteriophage (the virus) must infect a bacterial cell, pick up bacterial DNA, and then infect another bacterial cell. Note: While the DNA originally came from a bacterial cell, the donor (vector) is said to be the virus because it is where the DNA came from right before it enters the recipient.	Transduction can be specialized or generalized. Specialized transduction moves only genes near the attachment site while generalized transduction can move any genes regardless of their location. Virulent viruses are in the lytic cycle. Temperate viruses are in a lysogenic cycle. (This is when a virus hides dormant in DNA. Eventually the virus will enter the lytic cycle to replicate itself.)

*All newly received DNA enters single stranded and must be incorporated into a plasmid or the chromosome and replicated in order for the recipient cell to keep it. Incorporation into a chromosome requires two recombination events. *

Plating:

Toxins:
R-Resistant (Can live with the toxin)
S-Susceptible (Dies in presence of toxin)

Nutrients:
+: Functional gene can make nutrient and thus can live even if the nutrient is not available on the plate.
-: Non-functional gene cannot make nutrient and thus needs it in the environment to survive

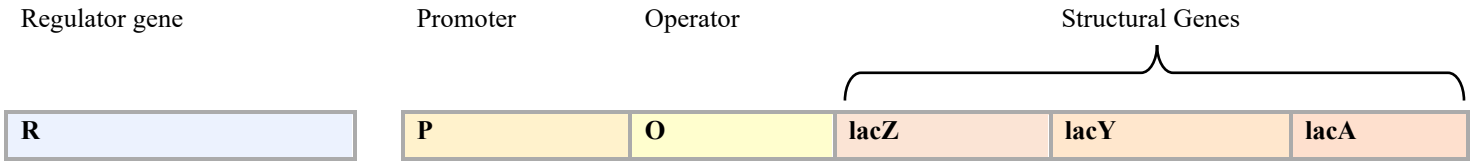
Minimal media: No nutrients or toxins on the plate.
Full media: All nutrients are supplied on the plate for the bacteria to use

A bacterium needs all amino acids in order to make proteins. If a bacterium does not have a functional copy of the gene to make each amino acid, it must have those nutrients it cannot make supplemented on the plate to survive. If a toxin/poison is in the environment, a bacteria must be resistant to it in order to survive.

Steps to figure out if growth of colonies occurs:

1. State which nutrients are on the plate.
2. Determine which nutrients are absent from the plate.
3. Understand genotypes. A bacteria will only survive if it can make those nutrients that are missing from the plate. (This means it must have a functional gene for these).
4. If there is a toxin/poison on the plate, the bacteria must have a resistance gene to that toxin in order to survive.

Lac Operon:



Codes for repressor protein

*Note: The regulator gene is a separate gene and has its own promoter and terminator

RNA Polymerase binding spot

Repressor binding spot

Codes for lactose digesting enzymes

Each of these genes code for a different protein, but they are all controlled together.

Steps for Lactase Production Genotype Questions

1. Check the promoter.

P - (Non-functional): RNA polymerase can't bind and begin transcription on this strand regardless of the other genes.

2. Check the operator.

O^c (Operator is constitutively on); Lactase proteins will always be made, even with a super repressor or in the absence of lactose as long as there are functional structural genes. This is because the repressor protein cannot bind even when lactose levels are low.

3. Check the repressor gene.

If I^s (Super repressor): There will be no expression of the gene (unless there is an O^c)

If I^- (Mutated repressor): The genes are always expressed regardless of if lactose is present.

4. Check the structural genes.

Z^-, Y^-, or A^-: These do not code for a functional protein, so there will be no lactose metabolism.

The structural genes explain what will be made. Only wildtype structural genes code for a functional protein. If there are any mutant structural genes, lactose metabolism will not occur.

*Repressors are trans-acting. They can repress the strand that they are on or another strand. All other portions of the lac operon are cis-acting. *