Biology Class 06

Previous Class Topic

- Discussion on assertion-reason type questions, focused on diseases such as **diabetes insipidus**.
- Introduction to genetics—structure of **DNA**, chromosomes, and genes within the cell nucleus.

Gene Expression

Nature and Function of Genes

- **DNA**, a nucleic acid in each cell's nucleus, serves as the repository of genetic information.
- **Genes**are segments of DNA carrying information that controls bodily functions and traits, such as hair and eye color.
- The physical attributes and functions of an organism are determined by the specific proteins produced in its body.
- **Gene expression**refers to the process by which the genetic information in a gene is converted into a functional product, typically a protein.

Steps and Locations in Gene Expression

- **Transcription**converts a gene's DNA sequence into messenger RNA (*mRNA*) within the nucleus.
- **Translation**involves the production of specific proteins from mRNA, occurring at ribosomes in the cytoplasm.
- The presence, absence, and interaction of proteins within cells define the organism’s characteristics.

Protein Synthesis Mechanism

- Genes, residing in the nucleus, cannot directly reach the ribosomes in the cytoplasm due to the size and structure of DNA.
- DNA sequences for required proteins are transcribed into*mRNA*, a smaller molecule capable of exiting the nucleus.
- *mRNA*carries the genetic recipe from the DNA to the ribosomes, where the recipe is translated into protein.
- The sequential order of genetic information flow:**DNA → mRNA (transcription) → Protein (translation)**.

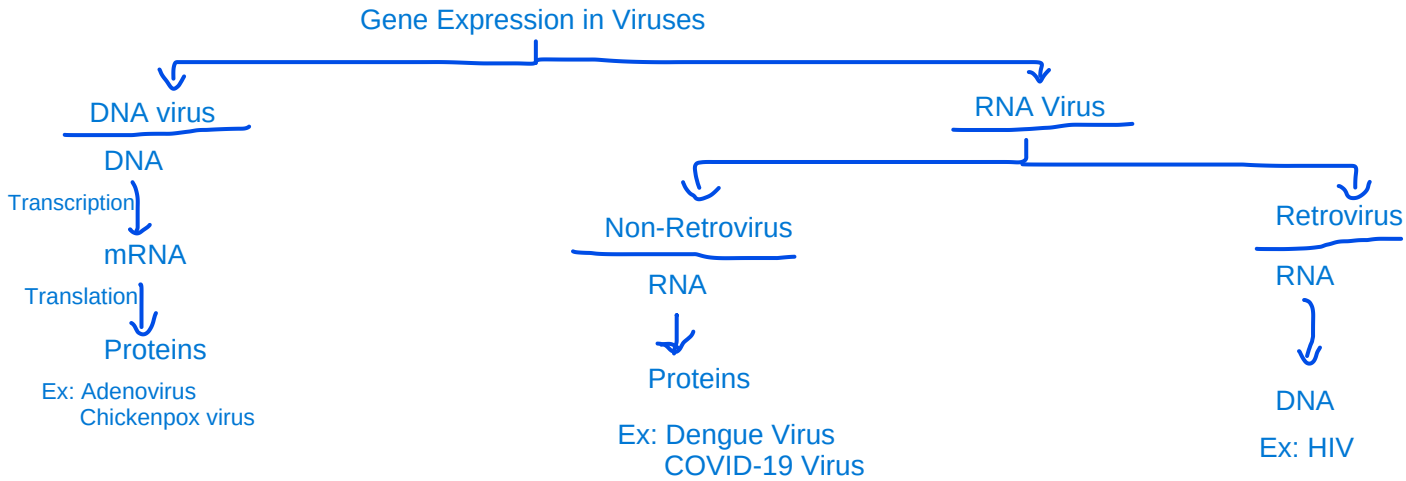
Summary Table: Processes in Gene Expression

Process	Starting Material	Product	Location
Transcription	DNA (Gene segment)	mRNA	Nucleus
Translation	mRNA	Protein	Ribosome/Cytoplasm

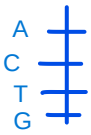
Gene Expression in Viruses

Classification of Viruses by Genetic Material

- Viruses can be**DNA viruses**(genetic material is DNA) or**RNA viruses**(genetic material is RNA).
- DNA viruses follow the typical flow: DNA → mRNA → Protein.
- RNA viruses possess RNA as their only genetic material.



-> Because of HIV virus AIDS happens and as we can see this comes under Retrovirus which forms its own DNA from RNA and mix with DNA of cell which then starts damaging the cell.



In case of DNA replication the same sequence of nitrogenous base should be formed and later Proof Reading also happens which ensure same sequential arrangement but in case of RNA replication this process of Proof Reading does not happens so different variant of RNA can also form which we see in COVID-19 virus.

Subtypes of RNA Viruses

Non-Retrovirus RNA Viruses

- Contain only RNA, from which proteins are synthesized directly.
- Examples include*Dengue*and*COVID-19 viruses*.

Retroviruses

- Retroviruses have RNA as their genetic material but undergo**reverse transcription**to create DNA from RNA.
- This newly formed DNA is integrated into the host cell’s DNA, altering the host's transcriptional processes.
- The process is termed*reverse transcription*and is unique to retroviruses.
- Example:*Human Immunodeficiency Virus (HIV)*, which causes AIDS.

Mutation Rate and Variability in RNA Viruses

- DNA replication involves proofreading mechanisms to correct copying errors, reducing mutation rates.
- RNA does not possess proofreading systems, resulting in higher rates of mutations and rapid variant emergence, as seen during the COVID-19 pandemic.
- This high variability in RNA viruses explains the frequent appearance of new viral strains.

Applications of Gene Expression: Vaccine Technology

Principles of Vaccination

- Vaccines introduce antigens to stimulate B-cells, creating immunological memory and protection.
- Antigens can be administered as live organisms, dead organisms, or subunits like specific proteins.

Nucleic Acid Vaccines

- Utilize DNA or mRNA encoding the antigen rather than the antigenic protein itself.
- **DNA Vaccines**: A DNA segment from the pathogen coding for the target antigen is introduced, resulting in the in vivo production of antigenic protein inside the vaccinated individual.
- **mRNA Vaccines**: mRNA sequences specifying the antigen are introduced directly, leading to protein production in the cytoplasm without incorporation into cell DNA.
- Direct injection into the nucleus or integration into the host DNA is strictly avoided to prevent interference with native genetic material.
- Proper delivery mechanisms have enabled stable mRNA vaccine development, particularly during the COVID-19 pandemic.

Examples

Vaccine Type	Mechanism	Example(s)
DNA Vaccine	Introduces DNA coding for antigen; protein synthesized in body	Zykov-D (COVID-19)
mRNA Vaccine	Introduces mRNA coding for antigen; protein synthesized in body	Pfizer, Moderna (COVID-19)

Advantages

- Fast and flexible vaccine development for pandemic-level threats.
- Simplified manufacturing due to small mRNA sequence size.

Inheritance of Genetic Material

Basics of Heredity

- Genetic material, encoded as DNA and organized into chromosomes, is transferred from parents to offspring.
- The reproductive cells (*gametes*) responsible for this transfer are eggs (from mother) and sperms (from father).
- Fertilization unites an egg and sperm, forming a**zygote**— the initial cell of a new organism.
- The zygote undergoes successive divisions to form an embryo, which differentiates into all body organs.

Chromosome Structure and Transmission

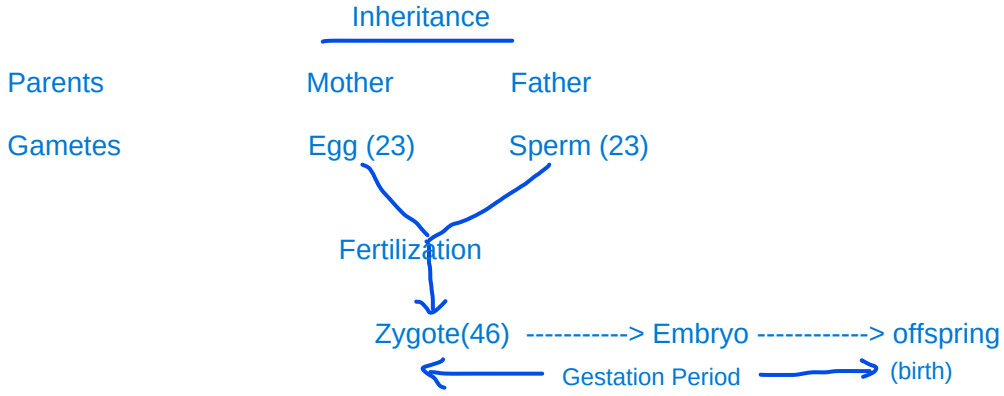
- DNA exists in chromosomes within the nucleus; each species maintains a fixed chromosome number.
- Humans have 46 chromosomes (23 pairs), with 23 inherited from each parent.
- To maintain species-specific chromosome numbers, gametes are haploid (N=23), and somatic cells are diploid (2N=46).

Terms for Chromosome Numbers

Term	Description	Example (Human)
Haploid (N)	One set of unpaired chromosomes (gametes)	23 (egg/sperm)
Diploid (2N)	Two sets of paired chromosomes (somatic cells)	46 (body cells)

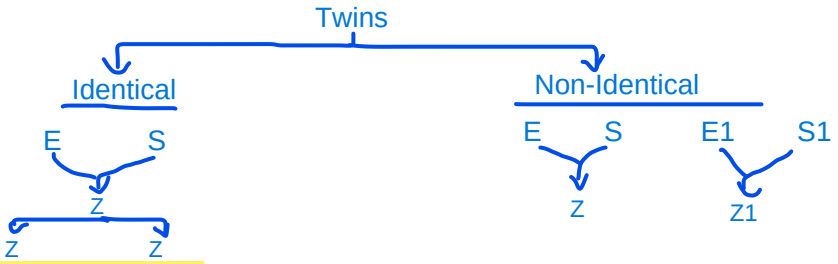
Chromosome Pairing

- Chromosomes appear in homologous pairs, having similar structure, length, and gene positions derived separately from each parent.
- This arrangement ensures paired information for each trait.



-> In both mother and father DNA is present in the form of chromosomes and there exist 46 chromosomes in each of them and during fertilization 23 from each mother and father comes together and forms zygote which then have 46 chromosomes.

Definition



Homologous Chromosomes: Pairs that are alike in shape, size, and gene position, each paralleling a chromosome from one parent.

Patterns of Inheritance: Mendelian Principles

Mendel's Experiments and Discoveries

- Traits are governed by pairs of genes, which may exist in contrasting forms (*alleles*).
- Experiments in peas revealed traits like pod color, plant height, and flower color exist in only two forms.

Main Experimental Insights

- Crosses between organisms with contrasting traits (like green vs. yellow pods) yield offspring expressing only the dominant trait.
- In subsequent generations, recessive traits reappear, demonstrating the inheritance of masked genetic factors.

Law of Dominance

- Among two contrasting alleles, the dominant allele expresses itself in the organism, while the recessive one is masked in the heterozygote.
- Recessive traits reappear only when both alleles are recessive in progeny.

Phenotype and Genotype

- **Phenotype**: Observable trait (e.g., tall, dwarf, green, yellow).
- **Genotype**: Underlying genetic constitution (e.g., GG, GY, YY).

Cross Generation Example

Parental Genotype	Possible Gametes	Offspring Genotypes	Offspring Phenotypes
GG (Green) x YY (Yellow)	G, Y	GY	Green (dominant expressed)
GY x GY	G, Y	GG, GY, GY, YY	Green:Yellow :: 3:1 (Phenotypic Ratio)

- **Homozygous**: Two identical alleles (GG or YY).
- **Heterozygous**: Two different alleles (GY).

Genotype Ratio
GG : GY : YY
1 : 2 : 1

Inheritance of Human Blood Groups

ABO Blood Group Inheritance

- Four phenotypes exist: A, B, AB, and O.
- The gene for blood group exists as three alleles: A, B, and O.

Genetic Basis

Phenotype	Genotype(s)	Description
A	AA or AO	A is dominant over O
B	BB or BO	B is dominant over O
AB	AB	A and B are co-dominant, both expressed
O	OO	Both O alleles are recessive; O appears only when both are present

Concept: Co-Dominance

In the**AB group**, both A and B alleles are expressed equally, exemplifying co-dominance.

Blood Group Cross Analysis Example

- Parents with genotypes AB and OO will have children with possible genotypes AO or BO, corresponding to blood groups A or B.
- The presence of an O group child when both parents are AB/O is impossible, aiding in parentage determination.

Determination of Sex: Chromosomal Basis

Human Sex Chromosomes

- The 23rd chromosome pair (sex chromosomes) determines biological sex—XX for female and XY for male.
- The other 22 pairs are autosomes.
- Chromosomal combinations:
- *XX*in offspring results in a female child.
- *XY*in offspring results in a male child.

Mechanism of Sex Inheritance

Parent	Chromosomes	Gametes Produced
Mother	XX	X
Father	XY	X or Y

- The egg always contributes an X chromosome, and the sperm determines sex by contributing either X (daughter) or Y (son).
- Each outcome (XX or XY) carries a 50% probability.

Chromosomal Abnormalities

Defects in Chromosome Number

- Any deviation from the norm of 46 chromosomes results in genetic disorders.

Sex Chromosome Abnormalities

- **Klinefelter's Syndrome**: Male with XXY chromosomes (total 47); displays both male and some female characteristics, and is sterile.
- **Turner's Syndrome**: Female with only one X chromosome (XO, total 45); sterile and underdeveloped reproductive system.

Autosomal Abnormalities

- **Down's Syndrome**(Trisomy 21): Three copies of chromosome 21 (total 47); characterized by intellectual disability, unique facial features, and normal reproductive ability.

Genetic Diseases: Single Gene Defects

Autosomal Disorders

Sickle Cell Anemia

- Caused by a mutation in the gene controlling hemoglobin structure.
- Normal hemoglobin gene:*HB*; Mutant gene causing sickle cell:*HBS*.
- Homozygous normal (*HB*, *HB*): normal phenotype.
- Homozygous mutant (*HBS*, *HBS*): sickle cell anemia.
- Heterozygous (*HB*, *HBS*): appears normal but is a carrier.
- The defective gene (*HBS*) is recessive; the normal*HB*allele is dominant.

Carriers and Disease Transmission

- Carriers (heterozygotes) can pass the defective gene to offspring, especially when both parents are carriers, increasing disease risk in children.
- Endogamy (marrying within the same bloodline or community) increases the likelihood of genetic disorders due to a higher chance of both parents being carriers.

Sickle Cell Anemia and Malaria Resistance

- Sickle cell anemia is prevalent in malaria-prone regions as the altered red blood cell shape provides resistance against the malaria parasite.

Prevention Strategies

- Genetic counseling, premarital testing, and awareness are critical for disease control in communities with high carrier frequencies.

Sex-Linked Disorders

X-Linked Recessive Disorders

- The X chromosome is larger, containing more genes and thus more potential for genetic defects than the Y chromosome.
- **Hemophilia**: Impaired blood clotting due to a defective gene on the X chromosome.
- **Color Blindness**: Defect in color discrimination genes carried on the X chromosome.

Inheritance Patterns

- Females with two defective alleles exhibit the disease; those with one normal and one defective allele are carriers.
- Males with a single defective X exhibit the disease, as the Y chromosome lacks a compensating normal gene.

XH = X^h

Sex	Genotype	Phenotype
Female	XX	Normal
Female	XXH	Carrier (Normal appearance)
Female	XH XH	Affected
Male	XY	Normal
Male	XH Y	Affected

Example Cross

- A colorblind woman (XH XH) and a normal man (XY) will have:
- *Daughters*: all carriers (XXH).
- *Sons*: all colorblind (XH Y).

Gene Editing and Legal/Ethical Concerns

- Technology now exists to remove defective genes in eggs and sperm and insert normal genes.
- Its application in practice is limited by legal restrictions; few countries permit editing genes in reproductive cells.

Topic to be Discussed in the Next Class

- Applications of biotechnology—gene editing and stem cells.
- Further analysis of inheritance patterns using biotechnology, with a focus on current advancements and practical uses.