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.

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.

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).

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.

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.