Study Guide: Human Biology

Terminology

Homology

Homology refers to the existence of shared ancestry between a pair of structures, or genes, in different species. A common example is the limb structure of humans, cats, whales, and bats which, despite serving different functions, share similar bone structures, indicating a common evolutionary origin.

Molecular Homology

Molecular homology involves the comparison of molecules (such as DNA, RNA, or proteins) across different species to identify similarities that suggest a common evolutionary origin. This can help trace the evolutionary history of species and the functions of genes.

Carcinogen

A carcinogen is any substance, radionuclide, or radiation that promotes carcinogenesis, the formation of cancer. This can be by damaging the genome or disrupting cellular metabolic processes. Examples include tobacco smoke, ultraviolet radiation, and certain chemicals.

Cell Cycle Check-point

Cell cycle checkpoints are control mechanisms in the cell cycle that ensure the cell's proper division. They detect and repair DNA damage and ensure the stages of the cell cycle, such as DNA replication and chromosome separation, occur correctly and at the correct times.

Cytokinesis

Cytokinesis is the process during cell division where the cytoplasm of a parental cell is divided into two daughter cells. It occurs after mitosis (the division of the nucleus) and ensures that each daughter cell receives a portion of the cytoplasm.

Mitosis

Mitosis is a part of the cell cycle where replicated chromosomes are separated into two new nuclei, followed by division of the cell's cytoplasm (cytokinesis), resulting in two identical daughter cells. It's crucial for growth, repair, and asexual reproduction in eukaryotic organisms.

Proto-oncogene

A proto-oncogene is a normal gene that can become an oncogene due to mutations or increased expression. Proto-oncogenes code for proteins that help to regulate cell growth and division.

Oncogene

An oncogene is a gene that has the potential to cause cancer. In tumor cells, these genes are often mutated or expressed at high levels. Most oncogenes originate from proto-oncogenes.

Tumor Suppressor Gene

Tumor suppressor genes are involved in controlling cell growth and division. When these genes are mutated or inactivated, cell division can proceed unchecked, leading to the development of tumors.

Benign Tumor

A benign tumor is a mass of cells that lacks the ability to invade neighboring tissue or metastasize. Examples include lipomas (fat tissue tumors) and fibroids (muscle tissue tumors).

Malignant Tumor

A malignant tumor is characterized by the ability to invade neighboring tissues and spread to distant organs (metastasis). Examples include breast cancer, lung cancer, and melanoma.

Metastasis

Metastasis is the process by which cancer cells spread from the place where they first formed to another part of the body. Cancer cells break away from the original (primary) tumor, travel through the blood or lymph system, and form new (secondary) tumors in other organs or tissues.

Allele

An allele is a variant form of a gene. Each individual inherits two alleles for each gene, one from each parent. Different alleles can result in different observable phenotypic traits.

Gene

A gene is a sequence of DNA that contains the information needed to produce a specific protein or RNA molecule. Genes act as instructions to make molecules called proteins.

Chromosome

A chromosome is a long DNA molecule with part or all of the genetic material of an organism. Chromosomes are located in the nucleus of our cells and are key to determining the inheritance of traits.

Haploid Cell

A haploid cell has a single set of unpaired chromosomes, which is half the number of chromosomes of a diploid cell. Gametes (sperm and egg cells) are haploid.

Diploid Cell

A diploid cell contains two complete sets of chromosomes, one from each parent. Most human cells are diploid, containing 46 chromosomes.

Meiosis

Meiosis is a type of cell division that reduces the number of chromosomes in the parent cell by half and produces four gamete cells. This process is required to produce egg and sperm cells for sexual reproduction.

Zygote

A zygote is the initial cell formed when two gamete cells are joined by means of sexual reproduction. It is a diploid cell resulting from the fusion of two haploid gametes.

Heterozygous Chromosome

This term is somewhat incorrect in the context of chromosomes themselves; rather, it's more accurate to say heterozygous at a particular gene locus, meaning having two different alleles of a gene.

Homozygous Chromosome

Again, this term is more accurately applied to a specific gene locus rather than a whole chromosome. Homozygous refers to having two identical alleles of a gene at a particular locus.

Sources of Heritable Variation

Question: Which of the following is[are] source[s] of heritable variation?

- Mutations
- Recombination of alleles during sexual reproduction
- New chromosomes created by crossing over
- All of the above
- Some but not all of the above

Answer: All of the above.

Key Points

- **Mutations** are changes in the DNA sequence. They can be spontaneous or induced by external factors, providing new alleles for evolution.
- **Recombination of alleles** during sexual reproduction mixes the genetic material from two parents, creating diversity in the offspring.
- **Crossing over** during meiosis can result in new chromosomes that combine genes in novel ways, further contributing to genetic diversity.

Relationship Between DNA, Gene, and Chromosome

Question: What is the relationship between DNA, gene, and chromosome?

Key Points 1

- **DNA (Deoxyribonucleic Acid)** ~~is a long molecule that contains our unique genetic code. It's like a blueprint for building and maintaining an organism.~~
- A **gene** is a specific segment of DNA that holds the instructions for producing a particular protein or RNA molecule.
- A **chromosome** is a structure that organizes and condenses DNA, allowing it to fit inside the cell's nucleus. Each chromosome contains many genes.

DNA: The Molecular Basis of Genetic Information

DNA (Deoxyribonucleic Acid) is a nucleic acid, a macromolecule that serves as the primary carrier of genetic information in all living organisms and many viruses. DNA is composed of two long strands that coil around each other to form a double helix. Each strand is made up of a sequence of four types of nucleotide bases: adenine (A), cytosine (C), guanine (G), and thymine (T). The order of these bases constitutes the genetic instructions used in the development, functioning, growth, and reproduction of all known living organisms and viruses.

Transcription: From DNA to mRNA

Transcription is the first step in gene expression, where a particular segment of DNA is copied into RNA (specifically mRNA) by the enzyme RNA polymerase. This process occurs in the nucleus of eukaryotic cells and involves the following steps:

1. **Initiation:** RNA polymerase binds to a specific sequence of DNA called the promoter, signifying the start of a gene.
2. **Elongation:** The RNA polymerase unwinds the DNA helix and reads the template strand of DNA, synthesizing a single strand of mRNA by adding RNA nucleotides that are complementary to the DNA template. In RNA, uracil (U) is used instead of thymine (T).
3. **Termination:** Once RNA polymerase reaches a termination sequence in the DNA template, the transcription of the mRNA molecule is completed, and the mRNA detaches from the DNA.

Translation: From mRNA to Protein

Translation is the process by which the sequence of bases in mRNA is decoded to produce a specific protein. This occurs in the cytoplasm of the cell, where ribosomes are located. Translation involves:

1. **Initiation:** The ribosome assembles around the target mRNA. The first codon, usually AUG, is recognized by an initiator tRNA molecule that carries the amino acid methionine.
2. **Elongation:** The ribosome moves along the mRNA, reading its bases in groups of three, known as codons. Each codon specifies a particular amino acid (out of 20 possible types), which is brought to the ribosome by a tRNA molecule. The amino acids are then linked together, forming a polypeptide chain.
3. **Termination:** The process continues until the ribosome encounters one of the three stop codons, which do not code for any amino acid and signal the end of protein synthesis. The complete polypeptide chain is then released and folds into its functional three-dimensional structure.

The Role of Proteins

Proteins are fundamental components of all living organisms, performing a vast array of functions including catalyzing metabolic reactions, DNA replication, responding to stimuli, providing structure to cells and organisms, and transporting molecules from one location to another. The sequence of amino acids in a protein determines its structure and function.

Conclusion

Thus, DNA should not be simplistically thought of as a blueprint but rather as the repository of genetic information required for the synthesis of RNA and, subsequently, proteins, which perform essential functions in living organisms. This flow of information from DNA to RNA to protein is known as the **Central Dogma of Molecular Biology**, highlighting the directional and functional flow of genetic information.

Cell Division: How and Why?

Question: How and why do cells divide, and how is cell division regulated?

Key Points 2

- Cells divide for growth, repair, and reproduction. In multicellular organisms, cell division allows for the growth of tissues and the healing of wounds. In unicellular organisms, it's a means of reproduction.
- **Mitosis** is the process by which a cell divides to produce two identical daughter cells, essential for growth and repair.
- **Meiosis** produces gametes (sperm and eggs) with half the number of chromosomes, critical for sexual reproduction.
- Cell division is regulated by **cell cycle checkpoints** to ensure DNA is correctly replicated and divided, preventing errors that could lead to disease, like cancer.

Development of Cancer

Question: What factors contribute to the development of cancer?

Key Points 3

- Mutations in **proto-oncogenes** can convert them into **oncogenes**, leading to uncontrolled cell growth.
- **Tumor suppressor genes**, when mutated, fail to regulate cell division, further contributing to cancer development.
- Hereditary factors can predispose individuals to cancer if mutations are passed from parents to offspring.
- Environmental factors, such as exposure to carcinogens, can induce mutations that lead to cancer.

Recap from Previous Meetings

- Organisms inherit traits from their parents, which include physical features, behaviors, and disease risks.
- Traits are encoded in genes, segments of DNA that are part of chromosomes located in the cell nucleus.
- Humans have 46 chromosomes, organized into 23 pairs. These are homologous chromosomes, except for the sex chromosomes (XY in males), which are heterologous.
- Homologous chromosomes may have different alleles, or versions of the same gene, while heterologous chromosomes carry different genes.
- A karyotype displays an organism's chromosomes, facilitating the identification of genetic abnormalities that may lead to diseases.

Understanding Proto-oncogenes and Tumor Suppressors

- **Proto-oncogenes** are genes that normally help cells grow. When they mutate, they can become **oncogenes**, driving the cancer process.
- **Tumor suppressor genes** are involved in controlling cell growth and division. Mutations can inactivate these genes, leading to cancer.

- The balance between proto-oncogenes and tumor suppressor genes is crucial for healthy cell function. Disruptions in this balance can lead to uncontrolled cell growth and cancer.