HUMAN GENETICS

Topic Covered

Introduction to Genetics

Objectives

Review of the Principles of Genetics

Differencing between human Genetics and Medical Genetics, overview of Medical genetics and it diseases in humans, Definitions need to be known in this course

Content

The course is given in English based on Explaining and Discussing in details for all course lectures

Teaching and learning methods

The course includes theoretical parts support with PowerPoint lectures

Assessments

Weekly quizzes , hand writing reports in each course , first and second course exam, mid exam and final exam

Source book

Human Genetics Book Tenth Edition by **Lewis** **McGraw-Hill International Edition**

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INTRODUCATION

Genetics is a field of biology that studies how traits are passed from parents to their offspring. The passing of traits from parents to offspring is known as heredity, therefore, genetics is the study of heredity. This introduction to genetics takes you through the basic components of genetics such as DNA, genes, chromosomes and genetic inheritance.

Genetics is built around molecules called DNA. DNA molecules hold all the genetic information for an organism. It provides cells with the information they need to perform tasks that allow an organism to grow, survive and reproduce. A gene is one particular section of a DNA molecule that tells a cell to perform one specific task.

Heredity is what makes children look like their parents. During reproduction, DNA is replicated and passed from a parent to their offspring. This inheritance of genetic material by offspring influences the appearance and behavior of the offspring. The environment that an organism lives in can also influence how genes are expressed.

Terms and Definitions;

Medical genetics is the branch of medicine that involves the diagnosis and management of hereditary disorders and concerned the variation and its relationship to health and disease. Medical genetics differs from human genetics in that human genetics is a field of scientific research that may or may not apply to medicine, while medical genetics refers to the application of genetics to medical care. For example, research on the causes and inheritance of genetic disorders would be considered within both human genetics and medical genetics, while the diagnosis, management, and counseling people with genetic disorders would be considered part of medical genetics so the medical genetics includes; classical genetics, cytogenetic, molecular genetics, biochemical genetics, genomics, population genetics, developmental genetics, clinical genetics, and genetic counseling.

In contrast, the study of typically non-medical phenotypes such as the genetics of eye color would be considered part of human genetics, but not necessarily relevant to medical genetics (except in situations such as albinism). Genetic medicine is a newer term for medical genetics and incorporates areas such as gene therapy, personalized medicine, and the rapidly emerging new medical specialty, predictive medicine.

Chromosome; a thread (strand)-like structure of nucleic acids and protein found in the nucleus of most living cells, carrying genetic information in the form of genes.
In most cells, humans have 22 pairs of these chromosomes plus the two sex chromosomes (XX in females and XY in males) for a total of 46.

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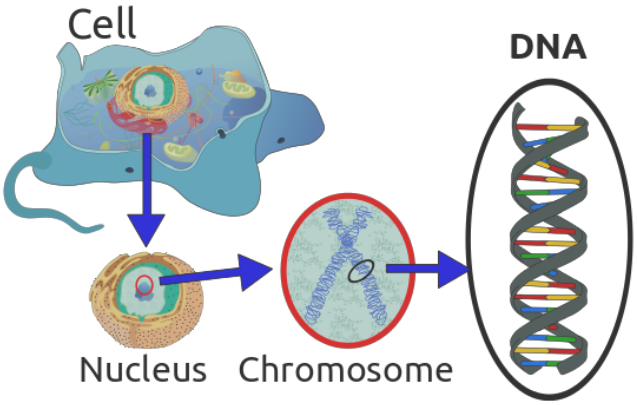
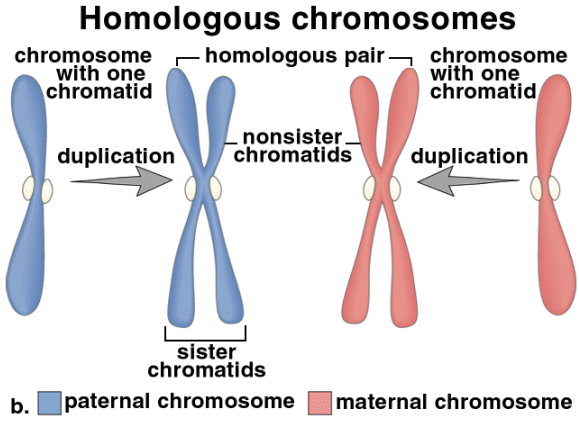


A non-duplicated chromosome is single-stranded and is comprised of a centromere region that connects two arms regions. The short arm region is called the (p) arm and the long arm region called the (q) arm. A duplicated chromosome is comprised of two identical chromosome called "sister chromatids" that connected at the centromere region.



Genes: genes are segment of DNA located on the chromosome. Gene exist in alternative forms called alleles. Genes contain the codes for the production of specific proteins.

The information contained within DNA is not directly converted to proteins, but must first be transcribed in a process called (DNA transcription). This process takes place within the nucleus of our cells. Actual protein production takes place in the cytoplasm of our cells through a process called (translation).

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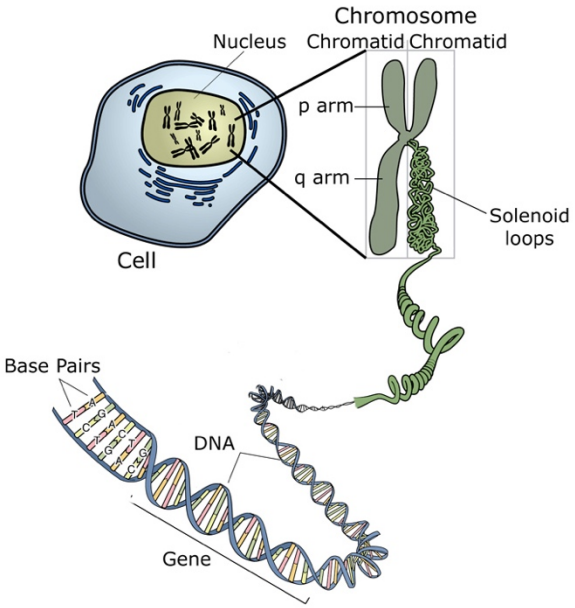
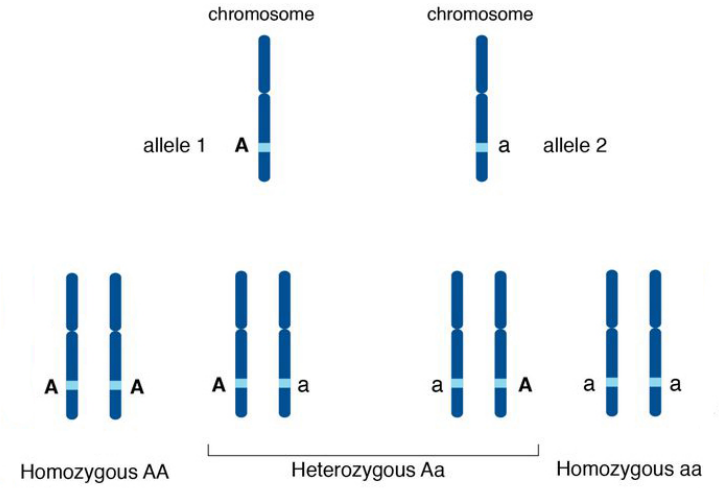


Allele; an allele is an alternative form of a gene (one member of pair) that is located at specific position on a specific chromosome. These DNA coding determine distinct traits that can be passed on from parents to offspring.



the process by which alleles are transmitted was discovered by Gregor mendal and formulated in what is known as Mendel's law of segregation.

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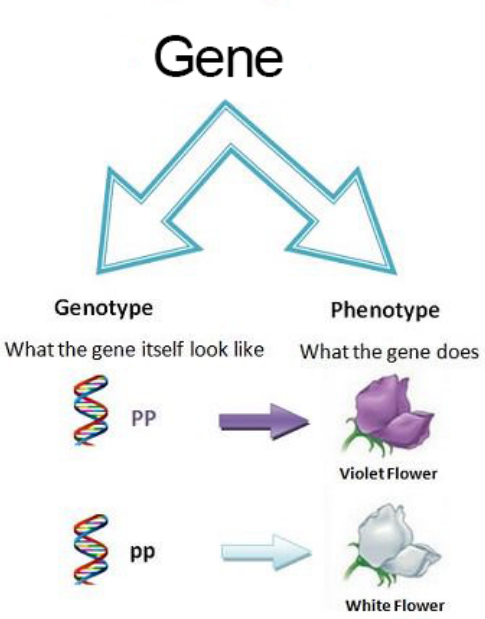
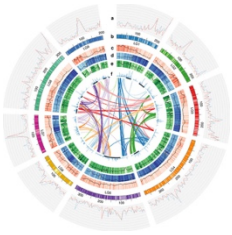
Genotype; the genotype of an organism is the inherited instruction it carries within its genetic code (i.e. the genetic makeup of an organism).



Phenotype; A phenotype is the composite of an organism's observable characteristics of traits, such as is Morphology, Development, Biochemical and Physiological properties. Phenotype result from the expression of an organism's genes as well as the influence of environmental factors and the interactions between the two.



Genome; A genome is an organism's complete set of DNA, including all of its genes. Each genome contains all of the information needed to build and maintain that organism. In humans, a copy of the entire genome—more than 3 billion DNA base pairs—is contained in all cells that have a nucleus.

Mendel's Law of Segregation;
Definition: The principles that govern heredity were discovered by Gregor Mendel in the 1860's. One of these principles, now called Mendel's law of segregation, states that allele pairs separate or segregate during gamete formation, and randomly unite at fertilization.

There are four main concepts related to this principle. They are as follows:
- A gene can exist in more than one form or allele.
- Organisms inherit two alleles for each trait.
  - When sex cells are produced (by meiosis), allele pairs separate leaving each cell with a single allele for each trait.
  - When the two alleles of a pair are different, one is dominant and the other is recessive.

Example: The gene for seed color in pea plants exists in two forms. There is one form or allele for yellow seed color (Y) and another for green seed color (y). In this example, the allele for Yellow seed color is dominant and the allele for green seed color is recessive. When the alleles of a pair are different (heterozygous), the dominant allele trait is expressed and the recessive allele trait is masked. Seeds with the genotype of (YY) or (Yy) are yellow, while seeds that are (yy) are green.