GENETICS

Sheet no. ?.?

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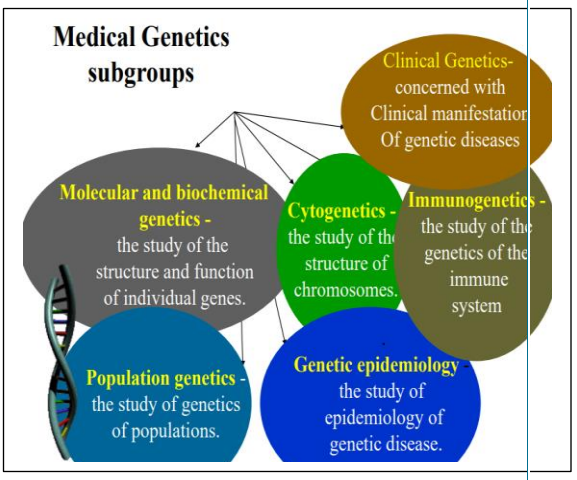
The slides were written in black And the doctor's interpretation was written in blue

A family portrait with members of four generations:

Why do some of the children look like only one of the parents, while some of the other children look more like the great, great grandparents? This is buzz they different from each other in genetics.

All of us have the same genetic material, but we are different in genetic variations and the modification that occurs on genetic material.

You should read the subtype of genetics.



• GENETICS – a branch of biology that deals with heredity and variation of organisms.

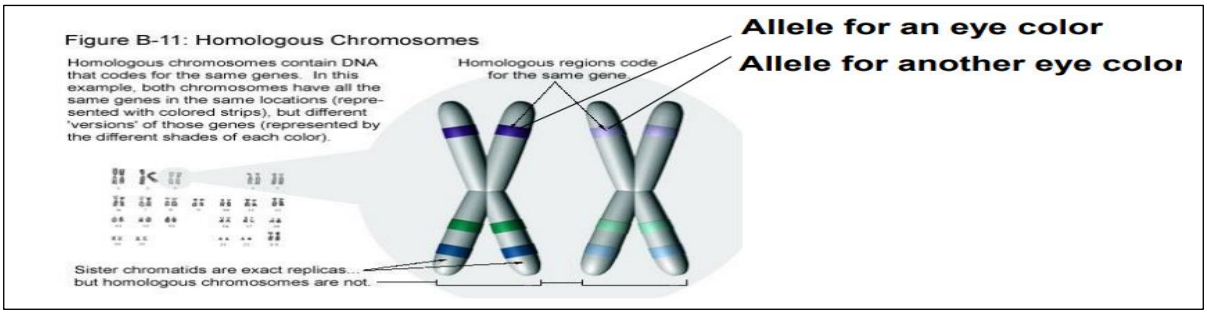
Chromosomes carry the hereditary information (genes) rod-like structure consists from two parts one comes from the mother called maternal and the anther one comes from the father and is called the paternal.

• Arrangement of nucleotides in ,DNA • DNA → RNA → Proteins

Chromosomes is 23 pairs one half from mother and second half from father and they exist in any cell.

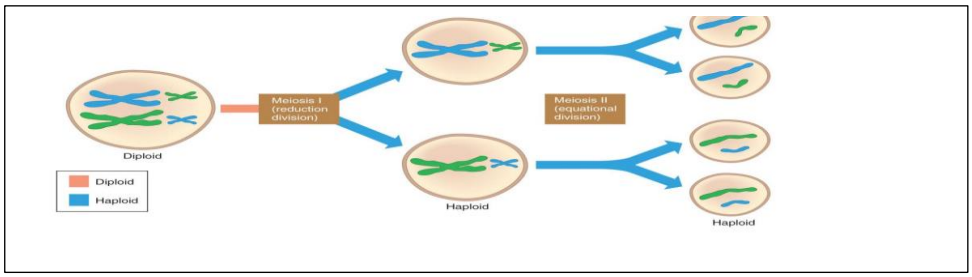
• Chromosomes (and genes) occur in pairs Homologous Chromosomes
• New combinations of genes occur in sexual reproduction

In this picture chromosomes differ from each other in the color of bands and the band represent to allele (the alternative form of allele called gene). And we have four alleles due to duplication of chromosome in S phase of mitosis to form sister chromatid.



Meiosis

Meiosis one in prophase there the meeting of two chromosomes and exchange of the genetic material that the source of genetic variation



The complete genetic composition of a cell or organism is called a genome. We have more than one genome and not in all cells just the cells that have the nucleus and there are many type of genomes for example mitochondrial genome is the smallest and mitochondria isn't existed as a single copy in each cell, cells differ in the number of mitochondria depends on the cell need.

NOTE: not all mitochondrial diseases are caused by mitochondrial genome.

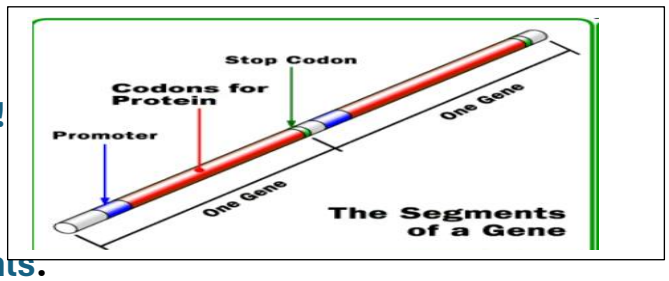
Mitochondria has 13 genes are responsible for 13 diseases, but we have other genes produce mitochondrial diseases, but they are not exist in mitochondria.

The genome encodes all the proteins a cell or organism can make.

- **Many key discoveries in genetics are related to the study of genes and genomes.**
- **Medical genetics is the science of human biological variation as it relates to health and disease.**

A gene, the basic unit of inheritance; it is a segment within a very long strand of DNA with specific instruction to produce one specific protein.

Gene consists from exons and introns, exons are important in an encoding gene, but introns are not important in gene coding



Defect in genes not totally corrected!

Some of these defects encoded and

produce Variation between individuals.

- Genes located on a chromosome on it's place or locus

Transcription occurs in nucleus and translation occurs in cytoplasm. And when the genetic material goes to the cytoplasm it should be protected from the environment and the orientation of DNA sequence is highly important.

AA is a homozygous genome, but Aa is an heterozygous genome. If adenine converted to cytosine the adenine called wide type allele and cytosine called mutant allele.

- Alleles: are the individual copies of a particular gene.
- Genotype: genetic constitution of any individual with respect to a particular trait.
- Phenotype: physical manifestation of a trait.
- Trait can be transmitted as autosomal, or sex linked and as Dominant or Recessive traits.
- Dominant allele: allele that mask or hides the expression of its alternative.
- Recessive allele: the allele which is masked or hidden.
- Sex-Linked Genes: Genes located on a sex chromosome.

• **Mendel – basic laws of inheritance – 1865 (who is the father of genetics buzz who is the first one discovered the processes of inherence)**

• **Watson and Crick – structure of DNA – 1953.**

• **Human Genome Project and Celera – sequence and annotation of human genome – 2003.**

• **Mendel inferred laws of genetics that allowed predictions about which traits would appear, disappear, and then reappear.**

• **Mendel's paper "Experiments in plant hybrids" was published in 1866 and became the cornerstone of modern genetics.**

• **Mendel's law of segregation: The two alleles for each trait separate (segregate) during gamete formation, and then unite at random, one from each parent, at fertilization.**

Alleles of different genes assorts independently of each other. And defect of the genes determines the phenotype. For example, even if the patients have the same disease the response of drugs may be different due to genetic variation between them.

• **Mendel's law of Independent Assortment: The alleles of different genes assort independently of each other.**

Genes underlie the formation of every heritable trait, e.g. cleft chin, hair loss, color of hair, skin, and eyes.

• **Some traits are causes by a single change in a single gene, e.g. sickle-cell anemia**

• **Some traits are caused by complex interactions between many genes, e.g. facial features.**

Organization of genetic information in cells

Chromosomes are organelles that package and manage the storage, duplication, and expression of DNA.

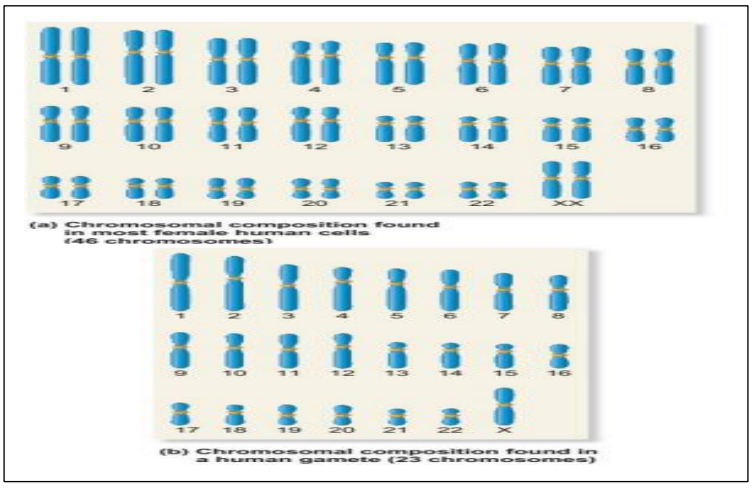
Genomes are the entire collection of chromosomes in each cell of an organism (24 kinds of chromosomes).

– The haploid human genome contains approximately 3 billion base pairs of DNA packaged into 23 chromosomes.

– Of course, most cells in the body (except for female ova and male sperm) are diploid, with 23 pairs of chromosomes. That makes a total of 6 billion base pairs of DNA per cell.

• Encodes 20,000 – 30,000 genes (that give proteins)

• Human also have a small amount of DNA in their mitochondria, which has also been sequenced



Eukaryotic genome

• Each base pair is ~ 0.34 nanometers long (a nanometer is 1/ 109 of a meter)

• Each diploid cell => contains ~ 2 meters of DNA [(0.34 × 10-9) × (6 × 109)]

• Our body contains about >37 trillion cells, which works out to 100 trillion meters of DNA per human.

• Now, consider the fact that the Sun is 150 billion meters from Earth. We enough DNA to go from here to the Sun and back more than 300 times!

– It needs to squeeze into 5-10 µm nucleus reducing its length as much as 10,000-fold

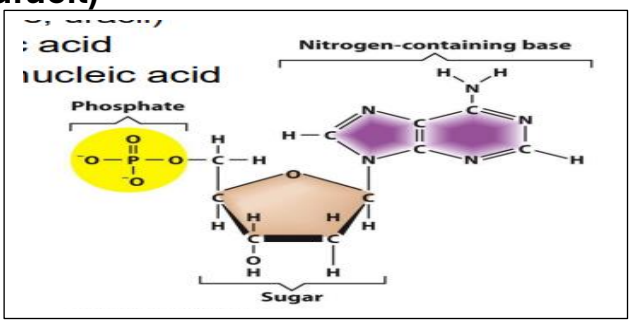
• How is this possible? Due to DNA condensation makes DNA able to fits and organized inside the nucleus.

This is accomplished by the packing of linear DNA molecules into chromatin (DNA with its associated proteins).

DNA/RNA?

• Genes are made of nucleic acids • Nucleic acids are made of building blocks called nucleotides

• Nucleotides have three components – Sugar molecule (ribose or deoxyribose) – Phosphate molecule – Nitrogen-containing molecule (adenine, guanine, cytosine, thymine, uracil)



• RNA is ribonucleic acid

• DNA is deoxyribonucleic acid

There is no O2 on the carbon

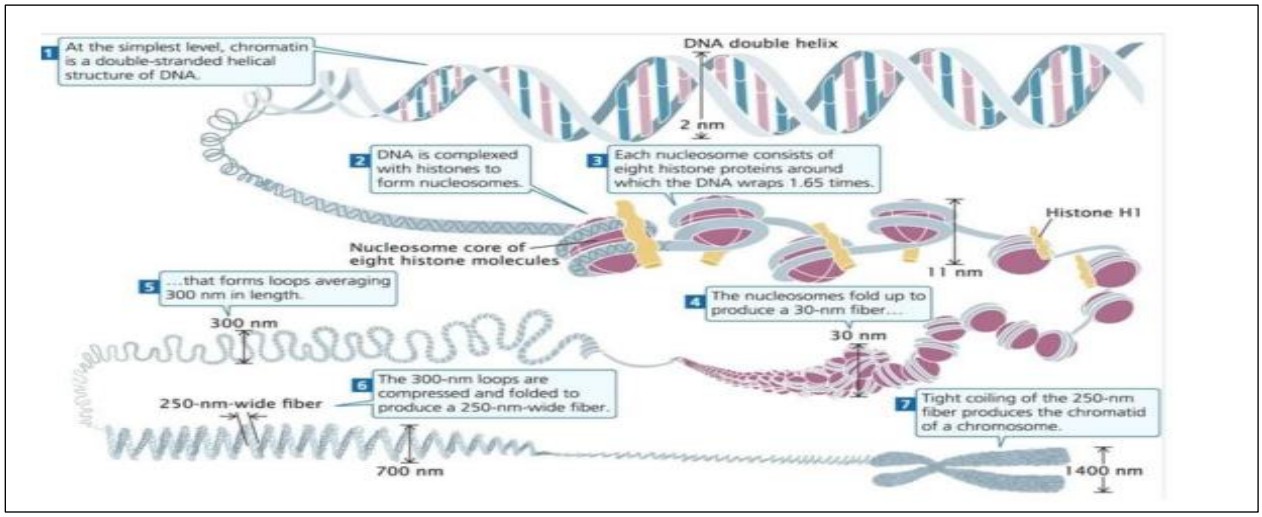
Number two in DNA

Carbon number 3 has hydroxyl group.

Bond between bases called hydrogen bond. (is the weakest) to makes DNA breaking easily. And to decease the energy that needed to break the DNA.

Bond between nucleotide to gather called phosphodiester bond.

Bonds between sugar and bases called glycosidic bond.



core proteins called histones (octamer) buzz it is consists form 8 proteins that encircled by DNA .DNA encircled each protein (histone) two times.

To form nucleosome= DNA + protein.

In bacteria there is no histone so it will make looping to decrease the length of DNA.

Chromosome appears compacted in metaphase in mitosis (two sister chromatids connected by each other) to stop the growth of cell at this level.

Colchicine is a drug inhibits cell division.