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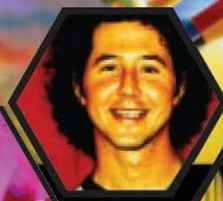
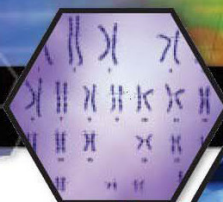
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October University for Modern Sciences and Arts

Faculty of Dentistry
First Year

PRINCIPLES OF GENETICS SGS124



Dr. Tarek Kapiel; 2005



**OCTOBER UNIVERSITY FOR MODERN
SCIENCES AND ARTS**

**Faculty of Dentistry
First Year**

**PRINCIPLES GENETICS
SGS124**

Dr. Tarek Kapiel

2005

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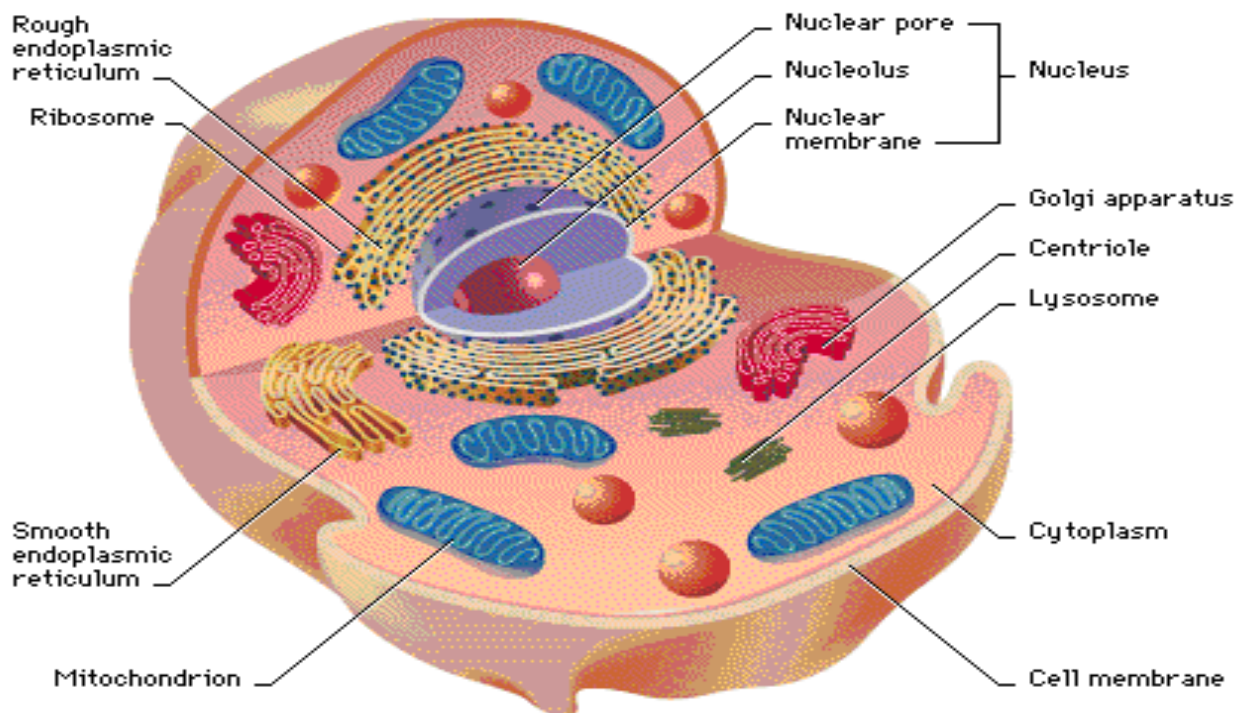
I INTRODUCTION

Genetics *is the scientific study of inherited variation*. Human genetics *is the scientific study of inherited human variation*. This field has been energized in recent years by the *Human Genome Project*. Scientists expect that the project will lead to the development of new drugs targeted to specific genetic disorders. Increasingly, modern genetics involves *genetic engineering*; a technique used to manipulate genes and has produced many advances in medicine.

II PRINCIPLES OF GENETICS

- The site where genes work is the cell.
- Each cell's function within an organism is determined by the genetic information encoded in DNA.
- In eukaryotes (organisms whose cells contain a nucleus), DNA resides within membrane-bound structures in the cell (nucleus, mitochondria, and chloroplasts in plants).
- In prokaryotes (one-celled organisms that lack internal membrane-bound structures), DNA floats freely within the cell body.
- DNA is packaged into structures called chromosomes within a cell.
- Every chromosome in a cell contains many genes, and each gene is located at a particular site, or locus, on the chromosome.
- Chromosomes usually occur in matched pairs called homologues.
- The number of homologous chromosomes in the human body contain 23 pairs of chromosomes.

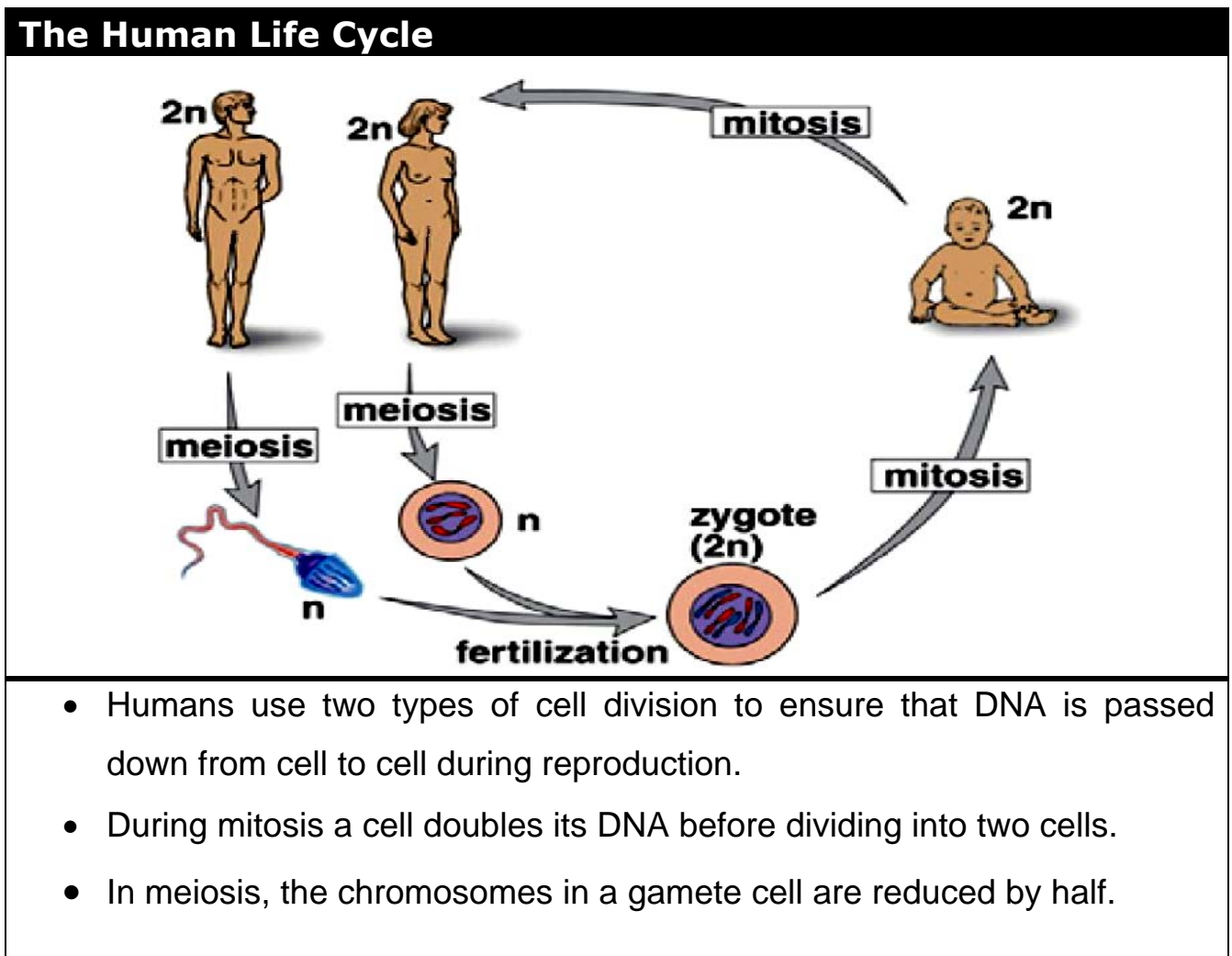
Animal Cell



- An animal cell typically contains several types of membrane-bound organs, or **organelles**.
- The **nucleus** directs activities of the cell and carries genetic information from generation to generation.
- The **mitochondria** generate energy for the cell.
- Proteins are manufactured by **ribosomes**, which are bound to the **rough endoplasmic reticulum** or float free in the cytoplasm.
- The **Golgi** apparatus modifies, packages, and distributes proteins.
- **Lysosomes** store enzymes for digesting food.
- The entire cell is wrapped in a **lipid membrane** that selectively permits materials to pass in and out of the **cytoplasm**.

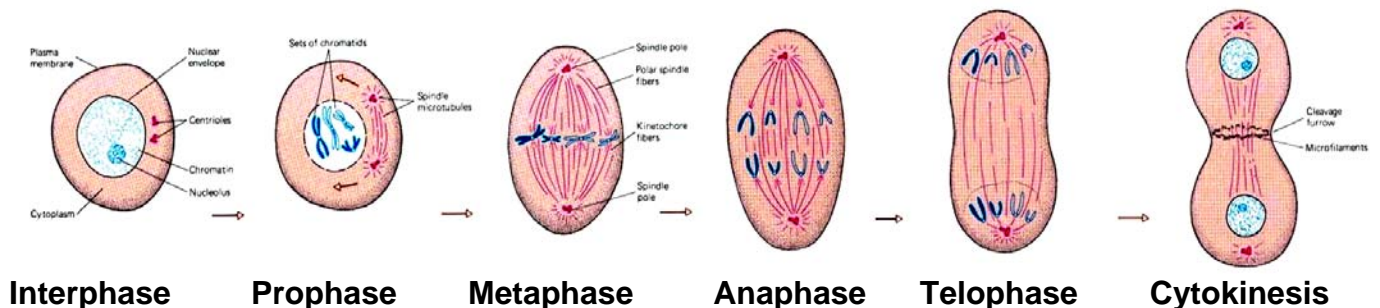
A. Cell Division and Reproduction

- Organisms use two types of cell division to ensure that DNA is passed down from cell to cell during reproduction.
- Simple one-celled organisms reproduce by a process called mitosis. During mitosis a cell doubles its DNA before dividing into two cells and distributing the DNA evenly to each resulting cell.
- Organisms that reproduce sexually produce special cells called gametes, or egg and sperm.
- During sexual reproduction, an egg and sperm unite to form a zygote, in which the full number of chromosomes is restored.



A1 Mitosis

Mitosis

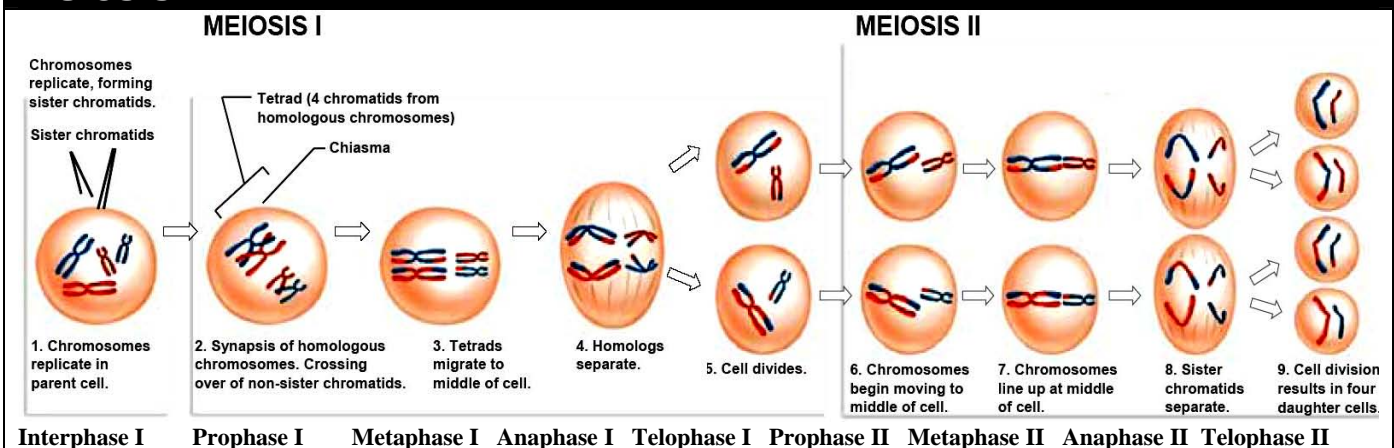


- **Mitosis occurs in five stages:**

1. ***Interphase***: the start of mitosis, the DNA of each chromosome replicates. Each chromosome then reorganizes into paired structures called sister chromatids, with each member of the pair containing a full copy of the DNA sequence.
2. ***Prophase***: the sister chromatids condense, thickening until they appear joined at a single site, known as the centromere.
3. ***Metaphase***: the sister chromatids line up in the middle of the cell.
4. ***Anaphase***: the chromatid pairs split apart at the centromere, and each half of the pair then moves toward opposite poles of the cells.
5. ***Telophase***: the final stage of mitosis, a nuclear membrane forms around the chromosomes at each pole of the cell.
 - Mitosis ends with the formation of two new cells, each with a matching full set of chromosomes.
 - The cytoplasm divides; the cell membrane pinches inward ultimately producing two daughter cells (**Cytokinesis**).

A2 Meiosis

Meiosis



- **Meiosis comprises two successive nuclear divisions.**

- **First division of meiosis:**

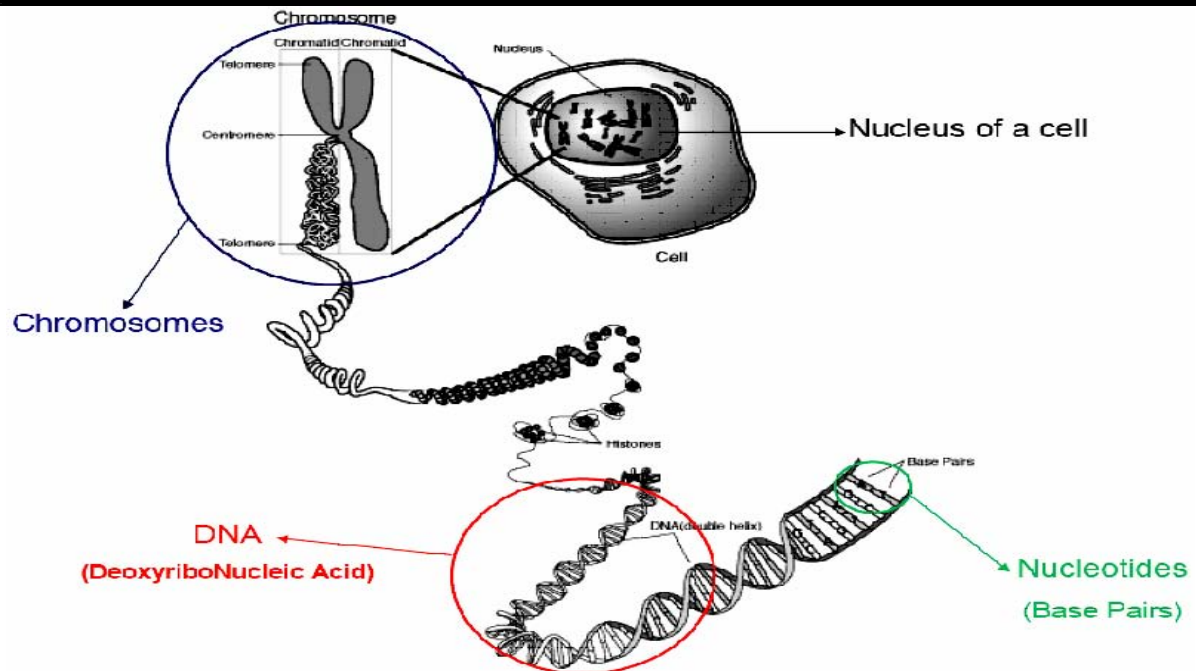
- 1. Prophase I:** Each chromosome duplicates and remains closely associated. These are called sister chromatids.
- 2. Metaphase I:** Homologous chromosomes align at the equatorial plate.
- 3. Anaphase I:** Homologous pairs separate with sister chromatids remaining together.
- 4. Telophase I:** Two daughter cells are formed with each daughter containing only one chromosome of the homologous pair.

- **Second division of meiosis:**

- 1. Prophase II:** DNA does not replicate.
- 2. Metaphase II:** Chromosomes align at the equatorial plate.
- 3. Anaphase II:** Centromeres divide and sister chromatids migrate separately to each pole.
- 4. Telophase II:** Cell division is complete. Four haploid (n) daughter cells are obtained.

Meiosis ensures that reproduction will produce a zygote that has received one set of chromosomes (n) from each parent to form a full set of chromosomes known as the diploid number (2n).

B. Human Chromosomes

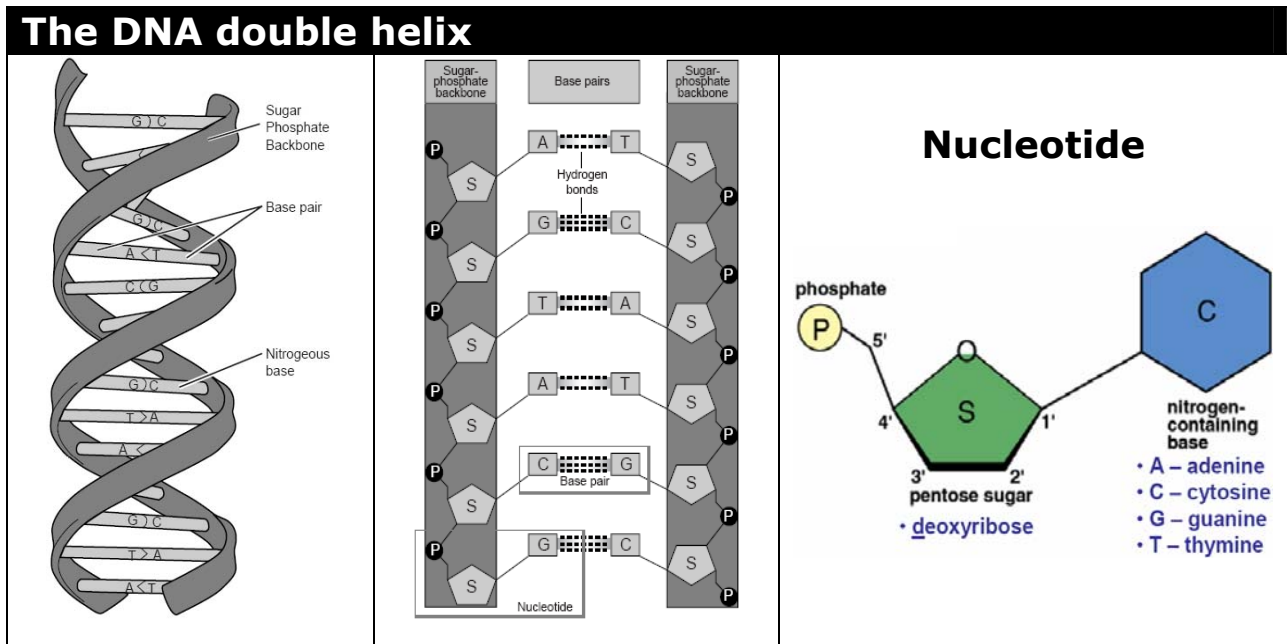


- Humans have 23 pairs of chromosomes. Twenty two pairs, the autosomes, are the same in either sex and are numbered from 1-22. The 23rd chromosome pair is called the sex chromosome, this chromosome pair consists of two X chromosomes in women, and men have one X + one Y chromosomes.

C. DNA Structure

- The structure of DNA encodes all the information every cell needs.
- DNA molecules form chains of building blocks called nucleotides.
- Each nucleotide consists of a sugar molecule called deoxyribose that bonds to a phosphate molecule and to a base.
- DNA uses four bases in its structure: Adenine (A), Cytosine (C), Guanine (G), and Thymine (T).
- The pairing of bases in the DNA double helix is highly specific (A always joins with T, and G always links to C). These base combinations, known as complementary base pairing.
- Genes line up in a row along the length of a DNA molecule.

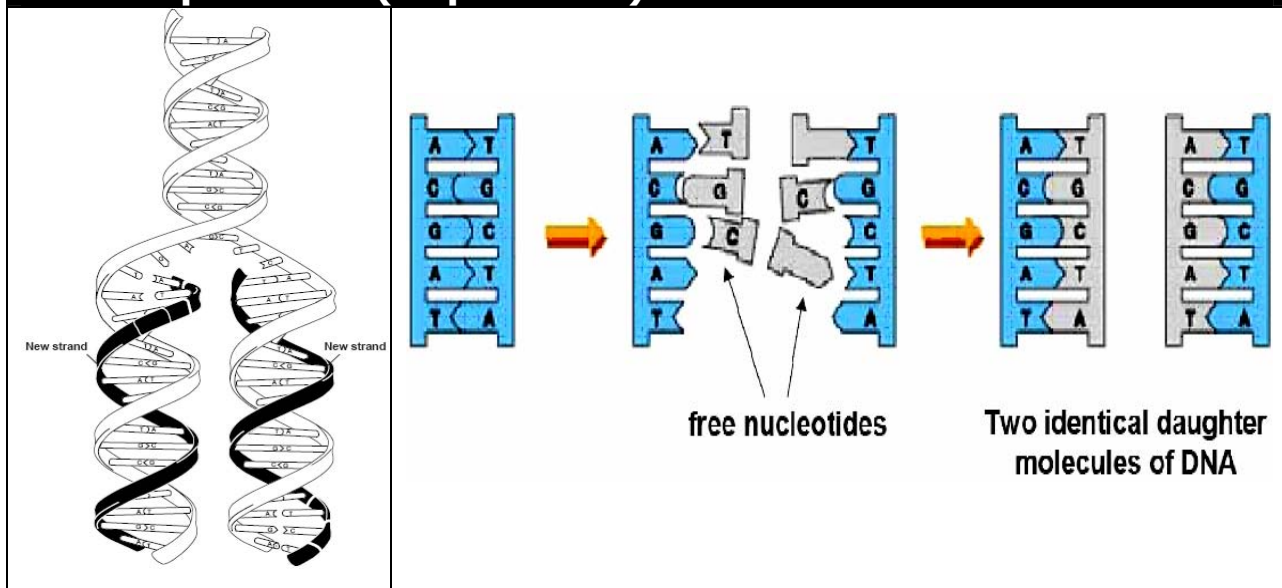
- In humans a single gene can vary in length from 100 to over 1,000,000 bases.
- Genes make up less than 2% of the length of a DNA molecule. The rest of the DNA molecule is made up of long, highly repetitive nucleotide sequences "junk" DNA".



D. DNA Replication (duplication)

- During replication of DNA, the DNA double helix unwinds and bonds joining the base pairs break, separating the DNA molecule into two separate strands.
- Each strand of DNA directs the synthesis of another complementary strand.
- The unpaired bases of each DNA strand attach to bases floating within the cell.
- The complementary bases then link to each other, forming a new DNA double-helix molecule. Thus the original DNA molecule replicates into two DNA molecules that are exact duplicates.

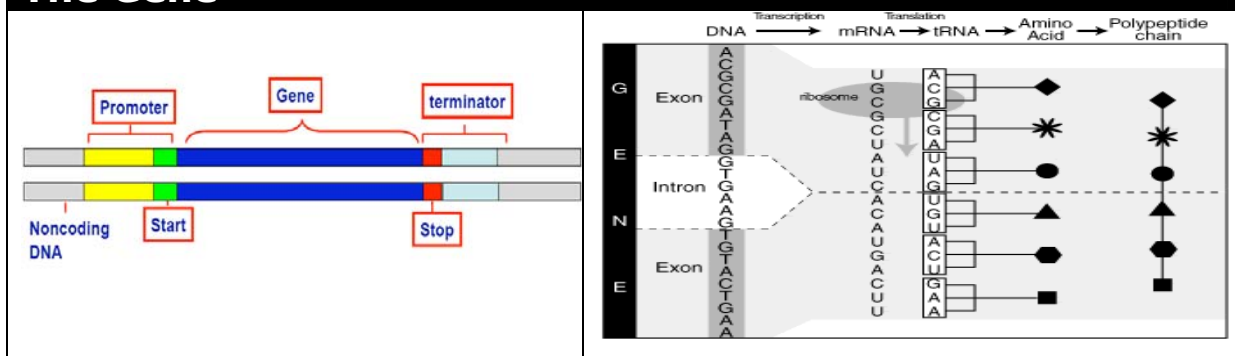
DNA Replication (duplication)



E. The Genetic Code

- A gene is a segment of DNA.
- The biochemical instructions found within most genes, known as the genetic code, specify the chemical structure of a particular protein.
- A gene consists of a promoter (regulate the initiation of transcription), the codons for a protein (protein-encoding sequence) and a stop codon (terminate the transcription).
- Many of the genes are duplicates.
- The DNA structure of a gene determines the arrangement of amino acids in a protein.

The Gene



F. Protein Synthesis

- Proteins are composed of long chains of amino acids.
- The process of utilizing the genetic code to create proteins, known as protein synthesis, has two steps: transcription and translation.

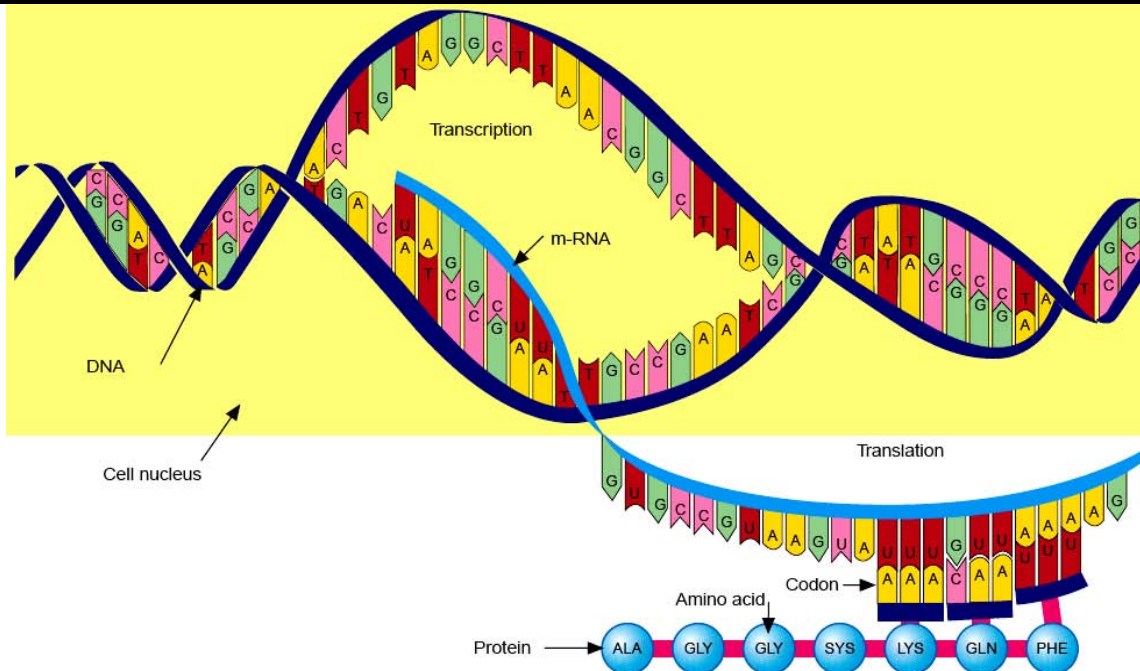
F1. Transcription

- Transcription transfers the genetic code from a molecule of DNA to an intermediary molecule called ribonucleic acid (RNA).
- The two compounds have three critical differences:
 - 1) The structure of RNA incorporates the sugar ribose rather than deoxyribose in DNA.
 - 2) RNA uses the base Uracil (U) instead of Thymine (T).
 - 3) RNA usually exists as a single strand, unlike the double-helix structure that normally characterizes DNA.
- Transcription involves the production of a messenger RNA (mRNA).
- The process begins when the two strands of a DNA molecule separate, a task directed by the enzyme RNA polymerase.
- After the double helix splits apart, one of the strands serves as a template, for the formation of a complementary mRNA molecule.
- Free-floating individual bases within the cell bind to the bases on the DNA template to form a strand of mRNA.
- In eukaryotes, the mRNA strand consists of coding regions called exons (link together to form an mRNA strand) separated by regions called introns (do not contribute to protein synthesis).

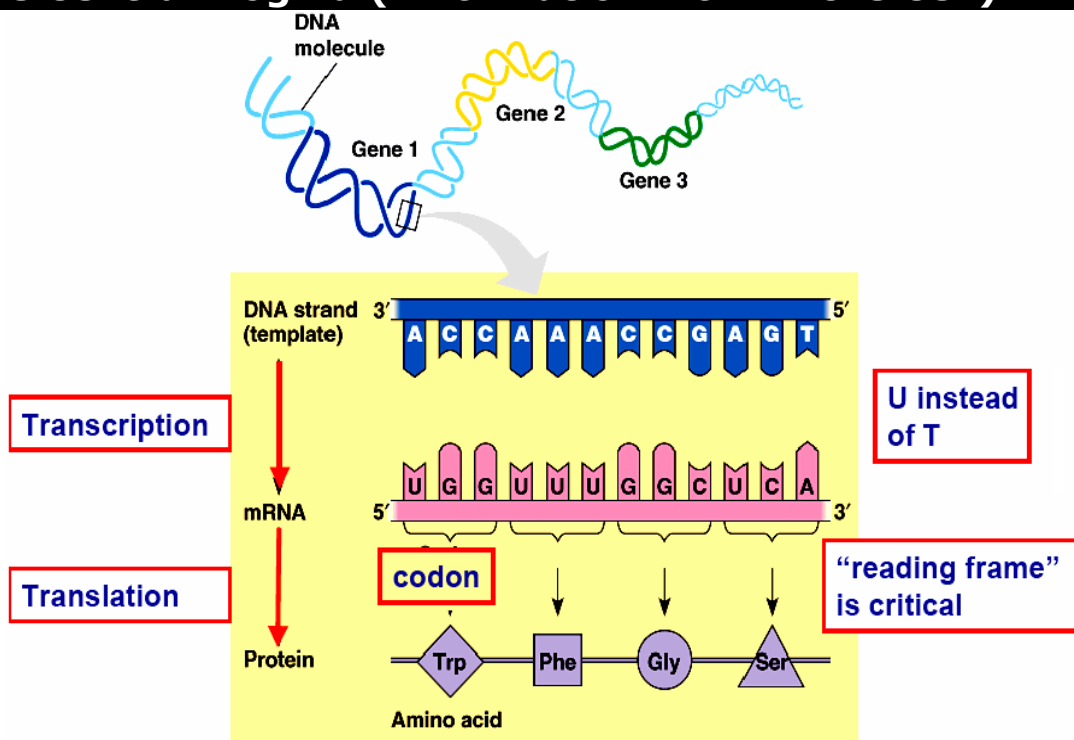
F2. Translation

- Translation takes place in ribosomes. The information coded in the four bases found in mRNA is translated into the instructions encoded by the 20 amino acids used in the formation of proteins.
- In eukaryotes, mRNA travels out of the nucleus into the cytoplasm to attach to a ribosome.
- Another form of RNA called transfer RNA (tRNA) is found in the cytoplasm of the cell. There are many different types of tRNA, and each type binds with one of the 20 amino acids used in protein formation.
- One end of a tRNA binds with a specific amino acid. The other end carries three bases, known as an anticodon.
- The anticodon of the tRNA undergoes complementary base pairing with a series of three bases on the mRNA, known as the codon.
- The mRNA codon codes for the type of amino acid carried by the tRNA.
- A second tRNA bonds with the next codon on the mRNA. The resident tRNA transfers its amino acid to the amino acid of the incoming tRNA and then leaves the ribosome.
- This process continues repeatedly until the formation of a polypeptide chain (a chain of amino acids).
- The process ends when the entire sequence of mRNA has been translated.
- The polypeptide chain falls away from the ribosome as a newly formed protein, ready to go to work in the cell.

Conversion Of Genetic Information



The Central Dogma (Information Flow in the Cell)



G. Mutations

- Any alteration in the structure of a gene results in a mutation.
- The altered genes continue to replicate in their changed form unless another mutation occurs.
- Most mutations harm an organism and the implications can be significant. **For example**, the amino acid sequence distinguishing normal hemoglobin from the altered form of hemoglobin responsible for sickle-cell anemia differs by only a single amino acid.
- In a **Substitution** (**point mutation**), a single nucleotide replaces another nucleotide.
- **Deletion** or **Insertion** even a single base from a normal sequence during transcription can disrupt translation by shifting the “**reading frame**” of every subsequent codon.
- **For example**, in **frameshift** mutation an mRNA strand may include two codons in the following sequence: AUG UGA. The addition of a Cytosine base at the beginning of this sequence shifts the “**spelling**” of these codons so that they read: CAU GUG. This may result in an incorrect amino acid sequence during translation, or the protein may be truncated. This type of alteration could result in the production of a protein with no real function or one with a harmful effect.
- In **transposition** mutation, parts of DNA (containing one or more genes) move from one chromosome to another (jumping genes or transposons) which change the type of amino acids in protein.
- Mutations can occur spontaneously, or can be caused by exposure to physical or chemical agents in the environment called mutagens.

- Common mutagens include ultraviolet rays and various chemicals, such as nitrous acid and high-energy radiation (such as X rays) which cause damage and disrupting the function of many genes.
- The cell has highly effective self-repair mechanisms that can correct the harmful changes made by mutations.

Types of Mutations	
<ul style="list-style-type: none"> • In <u>Substitution</u> (point mutation), a <u>single nucleotide</u> replaces <u>another nucleotide</u>. • <u>Deletion</u> (subtracting) or <u>Insertion</u> (Adding) of a <u>single base</u> from a normal sequence can disrupt <u>translation</u> by shifting the “<u>reading frame</u>” of every subsequent <u>codon</u>. 	<p>The diagram illustrates four types of DNA mutations using a sequence of six base pairs: A-T, T-A, G-C, G-C, C-G, T-A.</p> <ul style="list-style-type: none"> Original DNA strand: Shows the original sequence of base pairs: A-T, T-A, G-C, G-C, C-G, T-A. Substitution: Shows a single base pair (T-A) being replaced by another (A-T). The sequence becomes: A-T, T-A, A-T, G-C, C-G, T-A. Deletion: Shows a single base pair (T-A) being removed. The sequence becomes: A-T, T-A, G-C, G-C, T-A. A red arrow points to the missing T-A pair. Insertion: Shows a single base pair (A-T) being added. The sequence becomes: A-T, T-A, A-T, G-C, G-C, C-G, T-A.

H Gene Regulation

- Different cells within an organism share the same set of chromosomes.
- In each cell some genes are active while others are not.
- Each cell produces different proteins according to its needs so that it does not waste energy by producing proteins that will not be used.
- A variety of mechanisms regulate gene activity in cells. One method involves turning on or off gene transcription, sometimes by blocking the action of RNA polymerase (an enzyme that initiates transcription).

- Gene regulation may also involve mechanisms that slow or speed the rate of transcription, using specialized regulatory proteins that bind to DNA. A region of DNA known as an operon controls this gene regulation process.
- Gene regulation in eukaryotes is more complex than in prokaryotes.

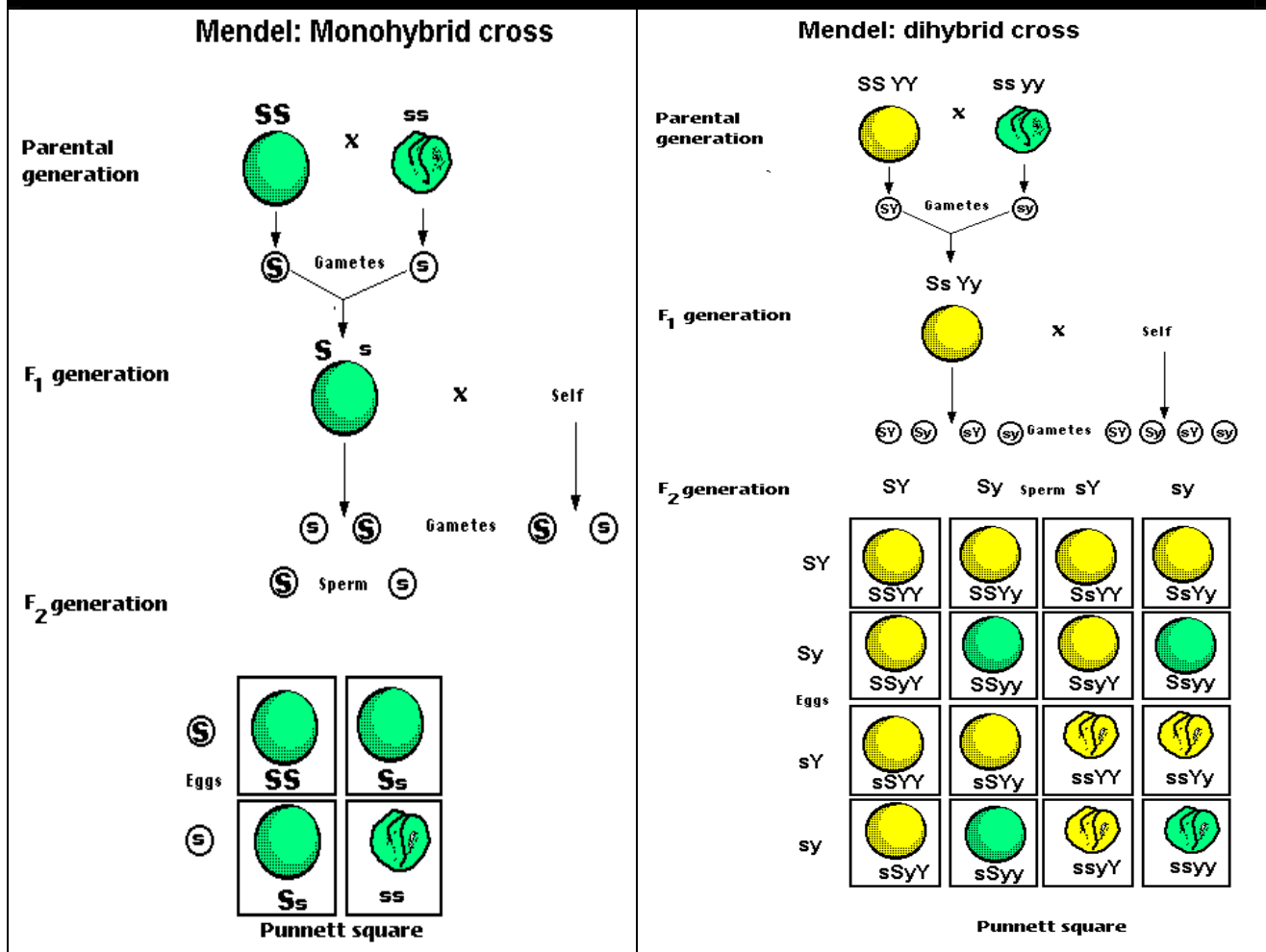
III. MENDELIAN INHERITANCE

- Gregor Mendel was the first scientist to observe that characteristics were inherited as separate units (genes), each of which was inherited independently of the others.
- Mendel suggested that each parent has pairs genes but contributes only one of each pair to offspring.
- Mendel recognized that a gene can exist in different forms (alleles), and he concluded that when an organism has two different alleles, one of the two may be **dominant** (represented by a **CAPITAL** letter) and the other is said to be **recessive** (represented by a **small** letter).
- Mendel also demonstrated that the patterns of inheritance observed in his experiments with single traits also apply to cases involving more complex gene combinations.

A. Mendel's Laws:

1. **Law of segregation:** states that alleles brought together in the F1 generation can be segregated in the F2 generation.
2. **Law of Independent Assortment:** states that most of the characters of parents can appear in any combination in their offspring.

● Mendel's Laws



B. Exceptions to Mendel's Rules

- The genetic principles that Mendel first discovered in plants apply to humans as well, but sometimes genes do not easily conform to the so-called Mendelian patterns of inheritance.

B1. Incomplete Dominance

- In cases of incomplete dominance, the inheritance of a dominant and a recessive allele results in production of intermediate characteristics. **For example**, four-o'clock paint plants may have red, white, or pink flowers. Plants with red flowers have two copies of the dominant allele **R** for red flower color (**RR**). Plants with white flowers have two copies

of the recessive allele r for white flower color (rr). Pink flowers result in plants with one copy of each allele (Rr).

B2. Quantitative Inheritance

- Traits such as skin color differ from the ones Mendel studied because they are determined by more than one pair of genes.
- In this form of inheritance, known as quantitative inheritance, each pair of genes has only a slight effect on the trait, while the cumulative effect of all the genes determines the physical characteristics of the trait.
- At least four pairs of genes control human skin color.

B3. Multiple Alleles

- Certain traits are controlled by multiple alleles that have complex rules of dominance.
- In humans, for example, the gene for blood type has three alleles: I_A , I_B , and i , with three alternatives for each member of a gene pair, there are six possible combinations of these genes ($I_A I_A$, $I_B I_B$, ii , $I_A i$, $I_B i$, $I_A I_B$).
- Although there are six possible combinations, humans have only four major blood types: **A**, **B**, **AB**, and **O**. This results because both I_A and I_B dominate over i , but not over each other, so a person with a gene combination of $I_A I_A$ or $I_A i$ has blood type **A**.
- The gene combinations $I_B I_B$ and $I_B i$ both produce blood type **B**.
- $I_A I_B$ results in a blood type **AB**, and ii results in blood type **O**.

B4. Gene Linkage

- Today scientists understand that independent assortment occurs when the genes affecting the phenotypes are found on different chromosomes.
- When genes occur on the same chromosome, they are inherited as a single unit. Genes inherited in this way are said to be linked.

B5. Sex-Linked Traits

- Most chromosome pairs consist of identical (homologous) partners.
- In humans, there is one pair of chromosomes in which the partners noticeably differ from each other. These are called the sex chromosomes because they determine the differences between males and females.
- In human females, the sex chromosomes consist of two X chromosomes, while males have an X chromosome and a shorter Y chromosome with many fewer genes.
- A male's X chromosome may contain a recessive allele associated with a genetic disorder, such as hemophilia and red-green color blindness in humans.
- Males do not have a normal second copy of the gene on the Y chromosome to mask the effects of the recessive gene, and disease typically results in the above cases.

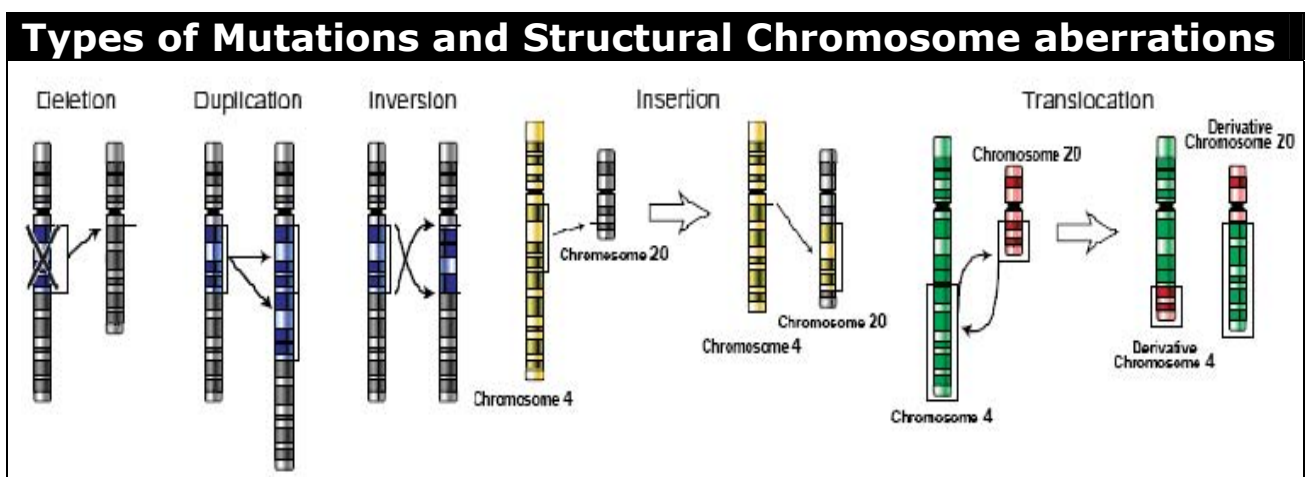
IV. GENES AND DISEASES

- The bases for hereditary diseases are the changes in genes which are passed on from one generation to the next via the germ line cells.
- About **5%** of all newborn babies have inherited disorders which are the **5th** most frequent cause of death.
- Most deaths result from inherited heart disorders followed by anomalies of the central nervous system as well as urogenital anomalies and gastrointestinal anomalies (digestive organs).

A. Genetic characteristics of inherited diseases

A1. Chromosome aberrations:

- Changes in the chromosomes themselves are either expressed by a change in the number of chromosomes (**numerical**, +) or by changes in the chromosomal structure (**structural** e.g. **translocation**: transfer of chromosome segments to make a **non-homologous** chromosome).
- Advanced age in the mother is one risk factor for numerical chromosome aberrations.



A2. Monogenic diseases:

- In monogenic diseases, only a single gene is altered (**mutant**) with the consequence that the pattern for a specific protein is imperfect, which in turn leads to the development (manifestation) of a disease.
- Monogenic diseases are often rare and cause severe illnesses. **Sickle cell disease** is an example of this.
- At present, over 6,000 genes are known whose mutations lead to various monogenic disorders. Currently, a molecular genetics analysis can be made on 1,000 of these diseases.

A3. Polygenic diseases:

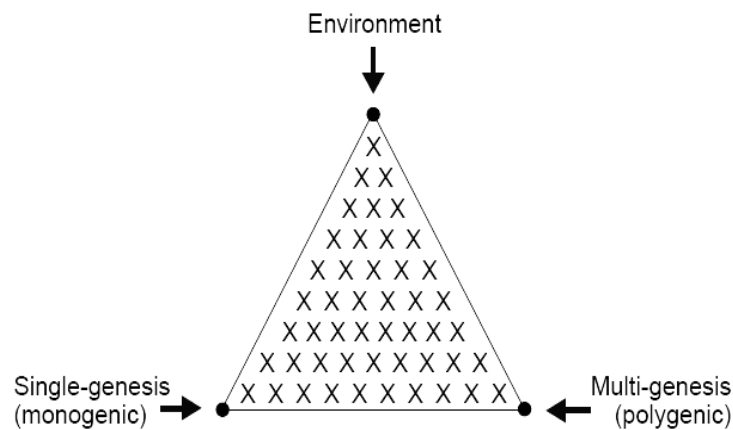
- It is the interaction of several gene alterations (**mutations**) which leads to the development of an illness.
- Polygenic diseases are very common in the population. Environmental factors (e.g. nutrition, exercise) usually contribute to the manifestation of these diseases (**multifactorial diseases**).
- Examples of diseases for which a combination of several genes and environmental factors are thought to be responsible is **Short sightedness**, **Diabetes** and **High blood pressure**.

B. Inheritance rules for human genetic diseases

B1. Inheritance process in monogenic diseases

- These inheritance patterns were already observed by Gregor Mendel in his famous cross-breeding experiments with peas, which form the basis for today's comprehensive family tree analyses.

Inheritance rules for genetic diseases

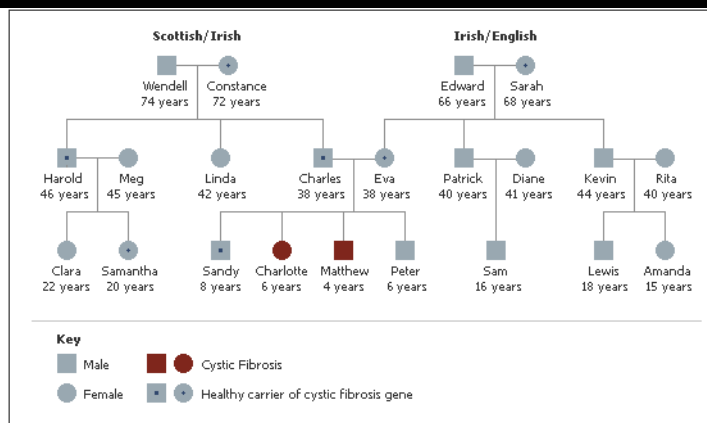


- **Genes** can be represented as more or less strongly influencing factors for the **development** of an illness.
- Every illness (X) lies within the **control pattern triangle**, none lie on an edge or a corner and predominate factors depends on where the illness is located inside the triangle.

B1.1. Family Pedigree

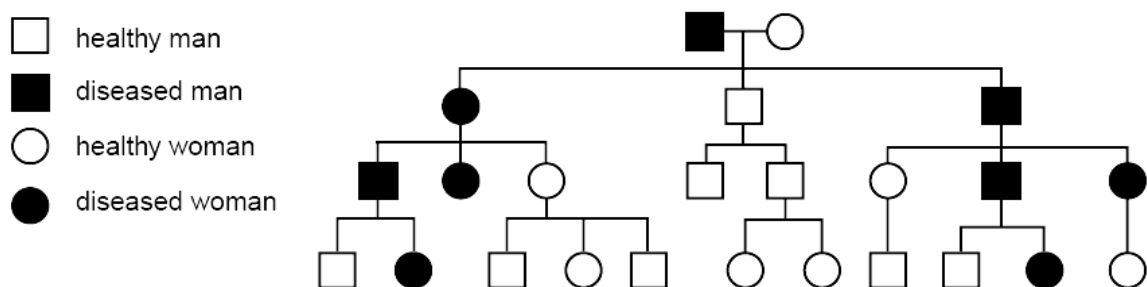
- Family pedigrees trace specific genetic characteristics through three or more generations.
- Pedigrees which illustrate the inheritance of a gene associated with cystic fibrosis help genetic counselors to identify which individuals in a family are at risk of either inheriting a genetic disorder or being a carrier for a disorder.

Family Pedigree



B1.2. Autosomal dominant inheritance process

- A dominant condition is transmitted in unbroken descent from each generation to the next.
- Only one of the two homologous genes is mutated and although another normal gene is present (**heterozygosity**), the illness still appears (**dominant gene effect**). If, therefore, one of the parents carries this gene, there is a 50% probability that it will be transmitted to each child. Both men and women can be affected by this.
- This inheritance pattern accounts for over 60% of monogenic diseases, representing by far the most common inheritance process.
- A typical pedigree might look like this:

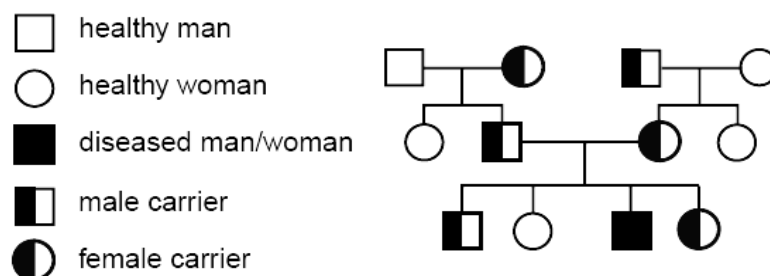


- Examples include *Tuberous sclerosis* and *neurofibromatosis*.

B1.3. Autosomal recessive inheritance process

- A recessive trait will only manifest itself when both **homologous genes** are mutated (**homozygosity**) in order to produce an illness in the affected person.
- Both sexes can be affected and individuals, who only receive one version of the mutated gene, are called **carriers** (**heterozygotes**).

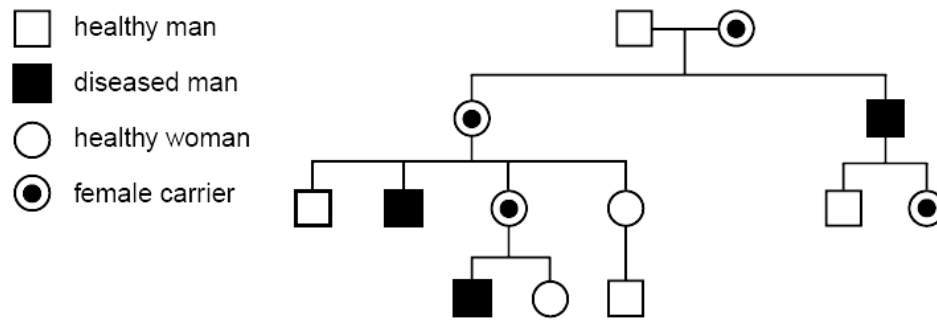
- If, both parents are carriers, there is a **25%** chance that the child will receive both mutated genes and so develop the illness.
- If it is a severe condition the **homozygotes** will not live, thus most occurrences of the condition will be in matings between two (or **carriers**).
- A typical autosomal recessive pedigree might look like this:



- Examples include many metabolic diseases (e.g. *cystic fibrosis*, and *phenylketonuria*).

B1.4. X chromosome inheritance (sex-linked inheritance)

- Women have two **X chromosomes**. If they have a recessively acting mutated gene on one **X chromosome**, they are carriers for the corresponding illness.
- Men have only one **X chromosome**, since the other sex chromosome is a **Y chromosome**. If they have the mutated gene on the **X chromosome**, they will develop the illness as a rule.
- If a woman is a carrier for the illness inherited by the **X chromosome**, there is a 50% chance that she will pass on this illness to her son. Her daughters have a 50% chance of becoming a carrier for this illness.
- A few X-chromosomal genes have a dominant action; in such cases women can also develop the illness.



B2. Inheritance process in polygenic diseases

- There are always several genes involved in causing polygenic diseases with different types of interaction possible.
- One or more environmental factors may contribute to the manifestation of the corresponding illness.
- In general, there is a transition from the healthy state to the pathological state in polygenic diseases.

V. IDENTIFICATION OF INHERITED DISEASES

A. Conventional genetic tests

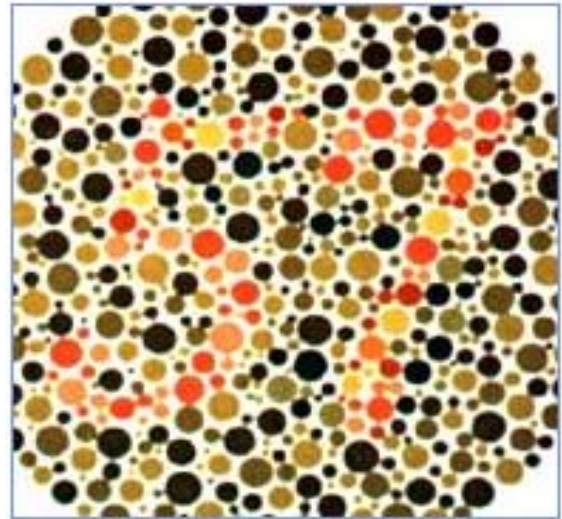
A1. Phenotype analysis

- The simplest and the most common type of genetic analysis.
- The analysis involves proteins, the end product of the genes. These proteins determine the way an individual's body looks.
- In this type of investigation, a certain body characteristics of the individual (*a phenotype*) is examined and compared with members of a normal population.
- A phenotype analysis is facilitated by conventional clinical and chemical investigation methods, such as a physical examination to

determine the *red-green blindness* phenotype, ultrasound or a blood analysis to determine the *blood group*.

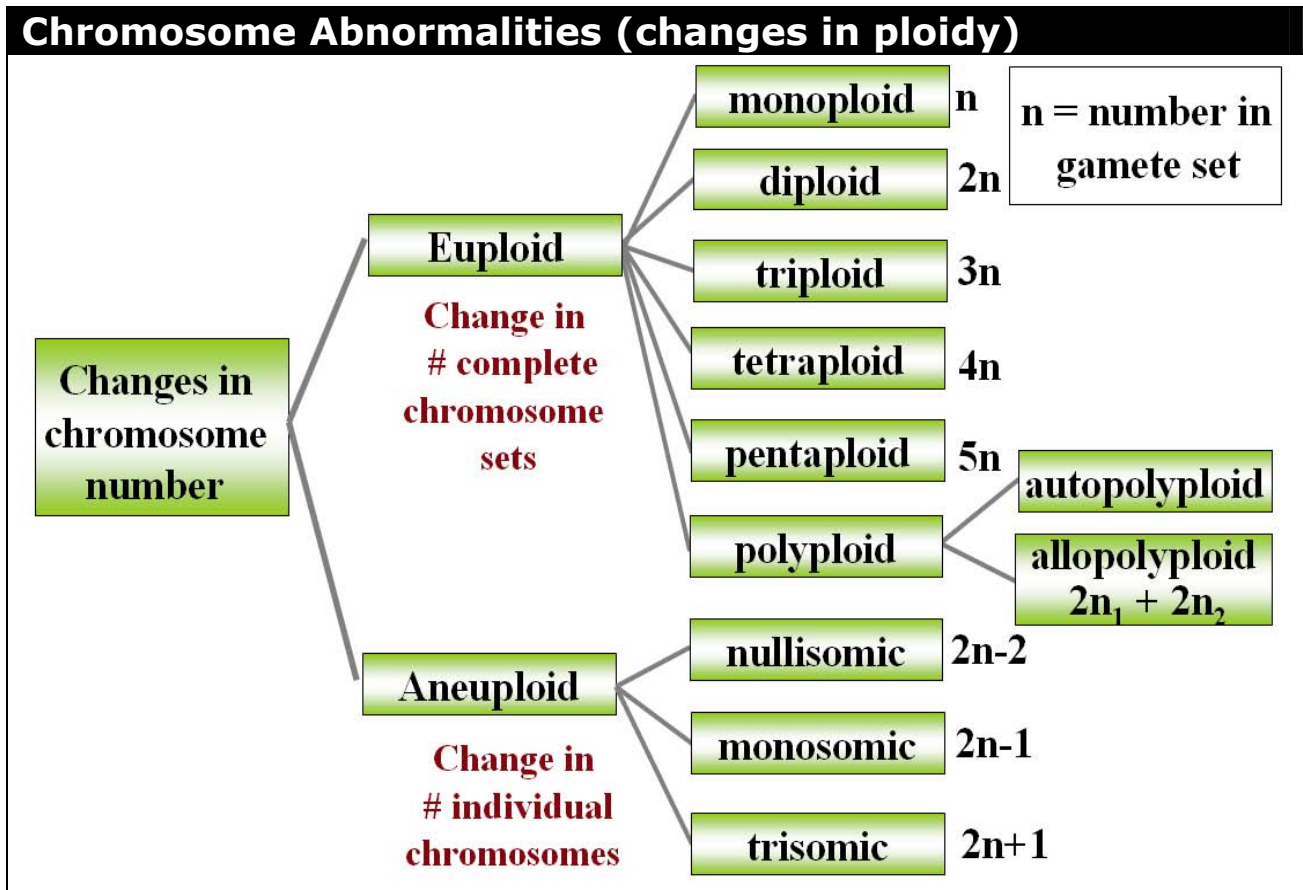
Red-Green Color Blindness Test

- Individuals with normal color vision will see the number 57, while those with red-green deficiencies will see the number 35.
- Color blindness, an inability to distinguish between red and green and sometimes between blue and yellow, is caused by a defect in one of the three color-sensitive cells in the retina.



A2. Chromosome analysis (cytogenetic investigations)

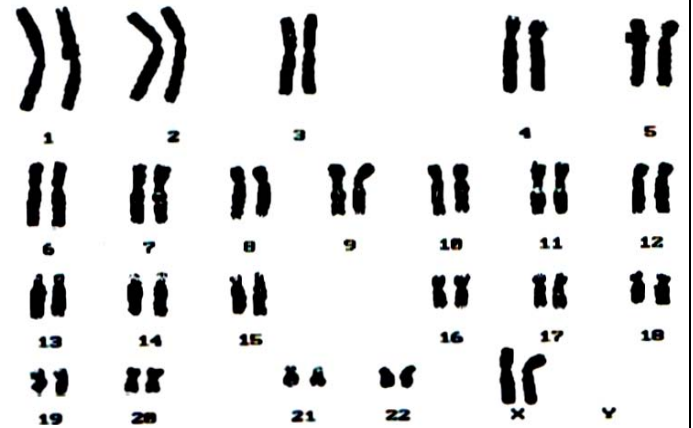
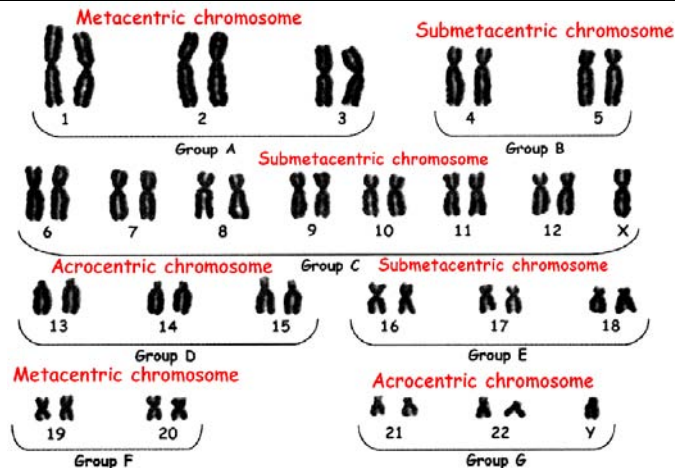
- Cytogeneticists study chromosomes microscopically.
- This includes microscope examinations to investigate chromosome alterations in terms of number and in terms of structure.
- There is no detailed investigation of individual genes in such cases, however, there is a new technique which uses a combination of cytogenetic and molecular genetics methods (molecular cytogenetics), in which a fluorescence-labelled DNA sequences are often used as diagnostic "probes", e.g. "FISH" (Fluorescence *in situ* hybridization)
- Examples of chromosome aberrations:
 - *Klinefelter's syndrome*: 47, XXY.
 - *Edward's Syndrome* (Trisomy 18): 47, XY +18.
 - *Down's syndrome* (mongolism): 47, XY, +21; or 47, XX, +21.



A3. Karyotype:

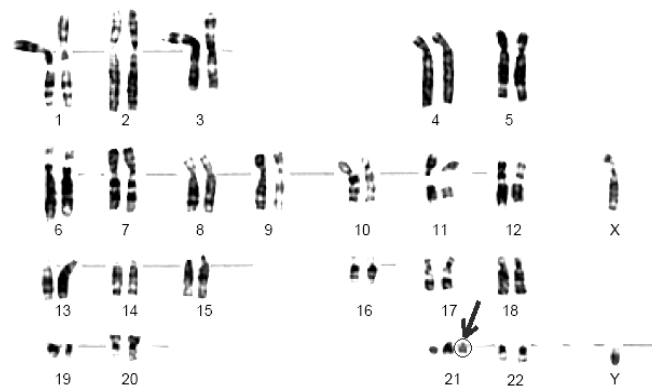
- A karyotype is a photographic image that shows the sum of all the chromosome information in an individual cell.
- Images are rearranged so that the chromosomes are lined up in pairs, typically beginning with the autosomes (Chromosomes 1-22 and ending with the sex chromosomes - normally XX or XY).
- A complete karyotype helps doctors determine if a person has extra chromosomes or missing chromosomes, or chromosomes that have attached to one another in unusual ways.

Karyotype

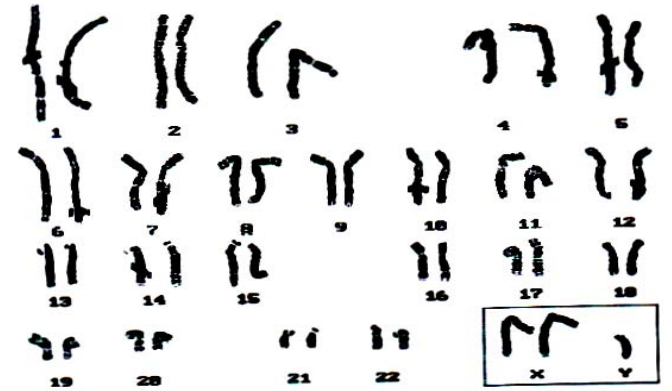


Normal Human Male Karyotype: 46, XY

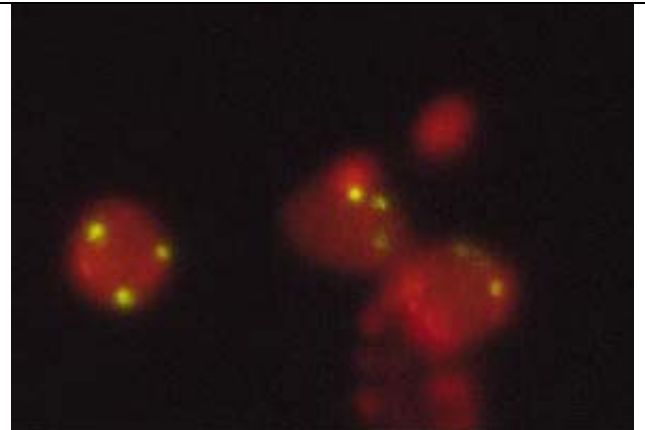
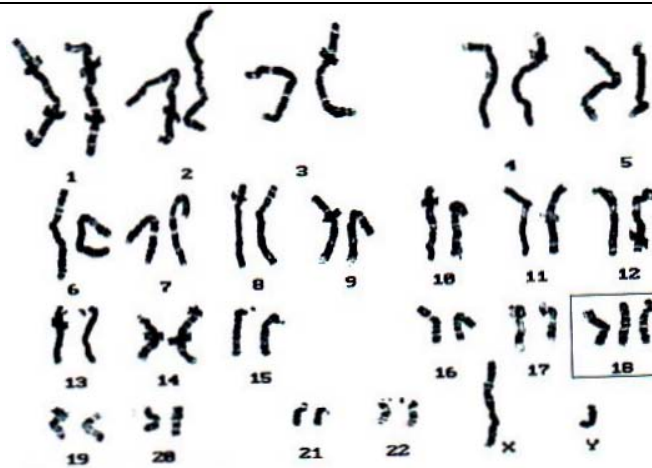
Normal Human Female Karyotype: 46, XX



Chromosome set for trisomy 21 (male)



Klinefelter's syndrome: 47, XXY.



Cytogenetic

Edward's Syndrome (Trisomy 18): 47, XY +18.

Molecular cytogenetics

Fluorescence in situ hybridization ("FISH") of an interphase cell nucleus showing *Edward's Syndrome* using a DNA probe specific for chromosome 18.

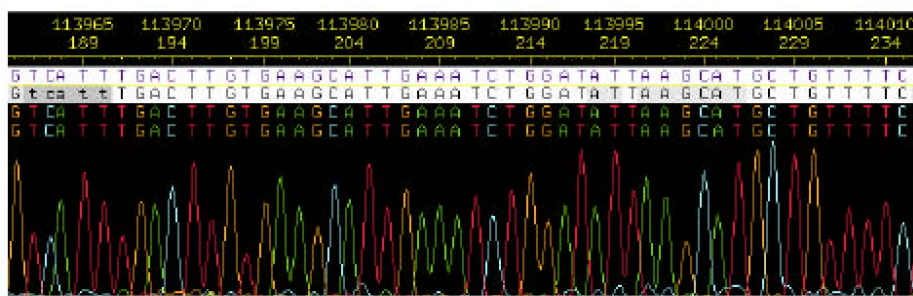
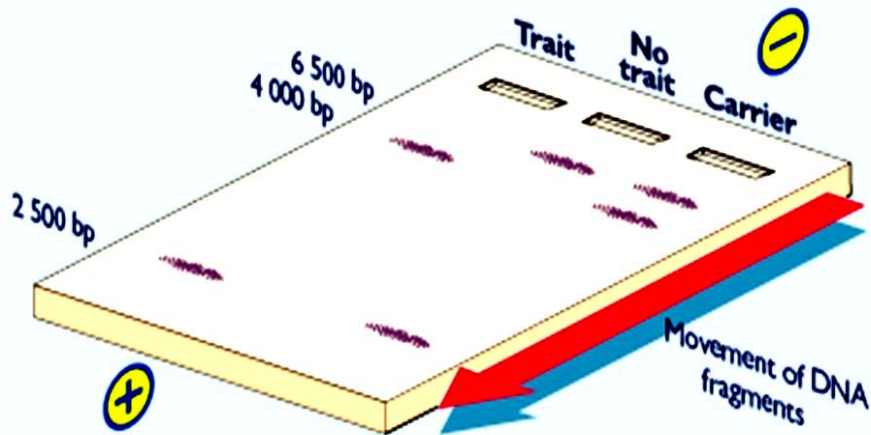
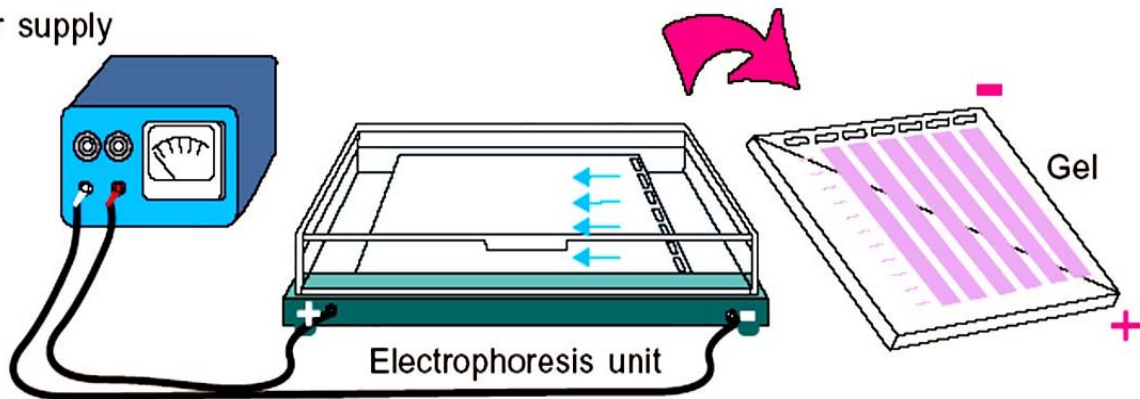
B. Molecular genetics testing (DNA and genome analysis)

- This provides evidence of a gene mutation responsible for producing the illness. Here it is determined whether the sequence of the DNA bases (nucleotide sequence) has changed within the affected gene.
- This is done as confirmation of preliminary clinical diagnoses, evidence of carriers or in predictive (including prenatal) investigations.
- Molecular genetics tests rely on revealing short segments of DNA sequence information and make use of the complementary nature of the strands of DNA in order to do this.
- There is different techniques available for revealing specific DNA double-stranded molecules which are all variations of the same basic principle:
 - In **DNA sequencing** the units of a specific sequence are read off one after the other;
 - In the **polymerase chain reaction (PCR)** one or several identical starting sequences are used to generate so many copies that they can be seen with the naked eye;
 - In the **Southern blot** procedure a mixture of sequences is sorted electrophoretically according to size before a specific sequence is made visible in an autoradiograph using, a radioactively labelled probe.
- With these techniques any deviation of a gene from the “normal state” can be revealed, whether it is a deletion, an insertion or an exchange of individual units (*point mutation*).

- The term *direct genetic test* is used if the alteration which is found is the direct cause of an illness.
- In the *indirect genetic test* (also known as *coupling or segregation analysis*), use is made of the fact that scattered all over the human genome there are sequence segments in which the two homologous chromosomes can be different.
- These differences in the sequence are in no way pathological; they are called *polymorphisms* and are passed on from generation to generation.
- *Polymorphisms* can lie inside or outside the genes, but they always occupy the same chromosomal position (*locus*).
- If it has been shown that a polymorphism lies close to a gene responsible for a disease then it can be used as a disease marker.
- Nevertheless, in view of the growing impact of genetics in medicine (“genetization”), it must be assumed that, in the future, genetic parameters will replace many conventional diagnostic investigative procedures as standard practice because of their objectivity.

Molecular Genetics Testing

Power supply



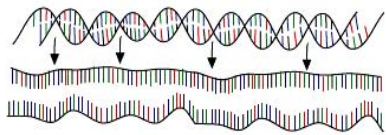
DNA sequence chromatogram from a cystic fibrosis gene

- **Nucleic acid electrophoresis** is a method used to separate DNA fragments to allow their visualization and/or identification.
- **DNA sequencing** is used to read off the units of a specific sequence one after the other.

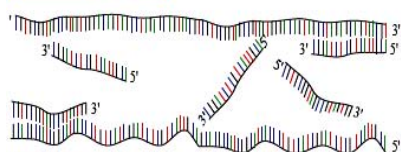
Polymerase Chain Reaction (PCR)

30 - 40 cycles of 3 steps :

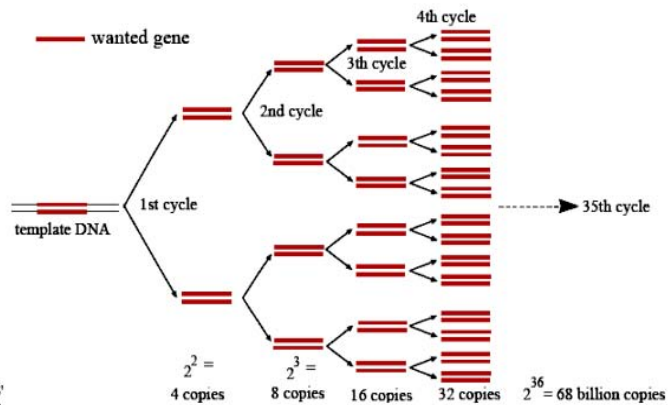
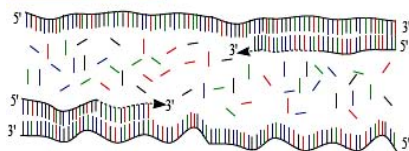
Step 1 : denaturation 1 minut 94 °C



Step 2 : annealing 45 seconds 54 °C



Step 3 : extension 2 minutes 72 °C



Southern Blotting: Gel Transfer

