

# Basics of human genetics and terminologies

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# Learning Objectives

- Understand the basics of genetics
- Explain the relation of genes to chromosomes
- Learn to draw and analyse pedigrees

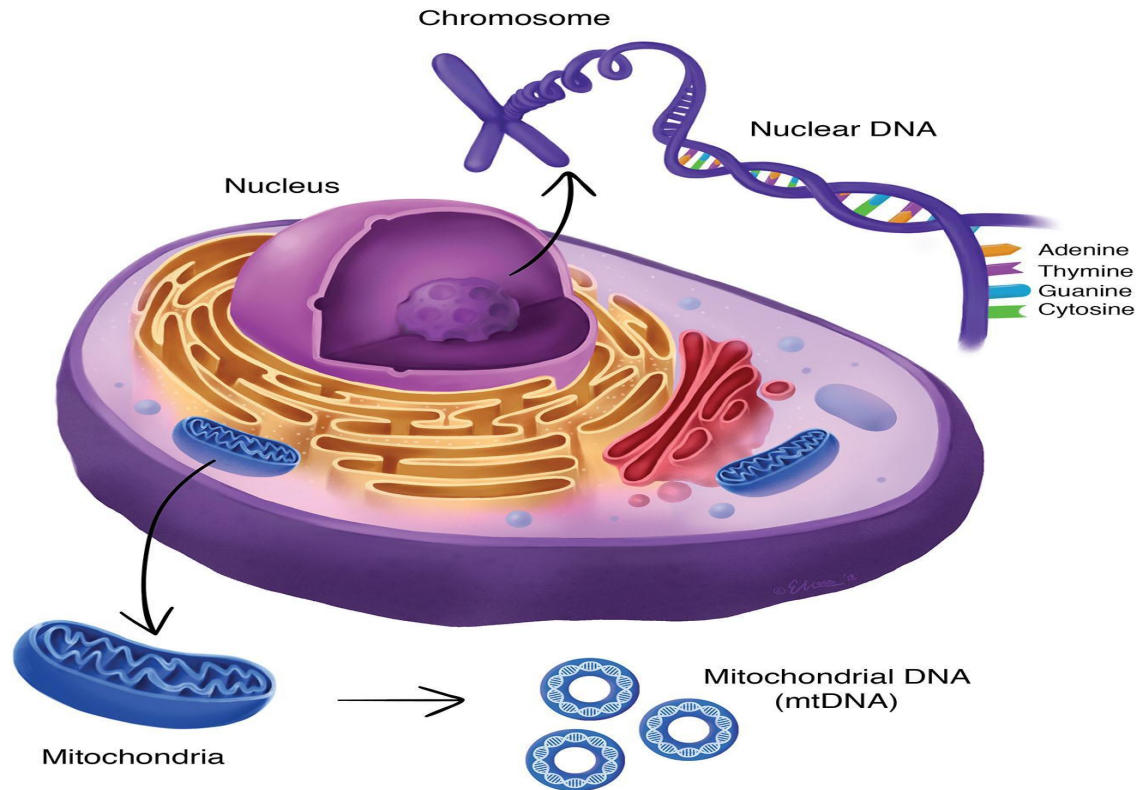
## Definition of basics concept

- **Heredity:** Transmission of traits from one generation to their descendants
- **Traits:** a distinguishing characteristic that determines a person's appearance
- **Genome:** the total DNA or biological information present in a cell or an organism
- **DNA (deoxyribonucleic acid ):** the genetic information responsible for the development and function of an organism
  - is a polymer of four simple nucleic acid units called nucleotides

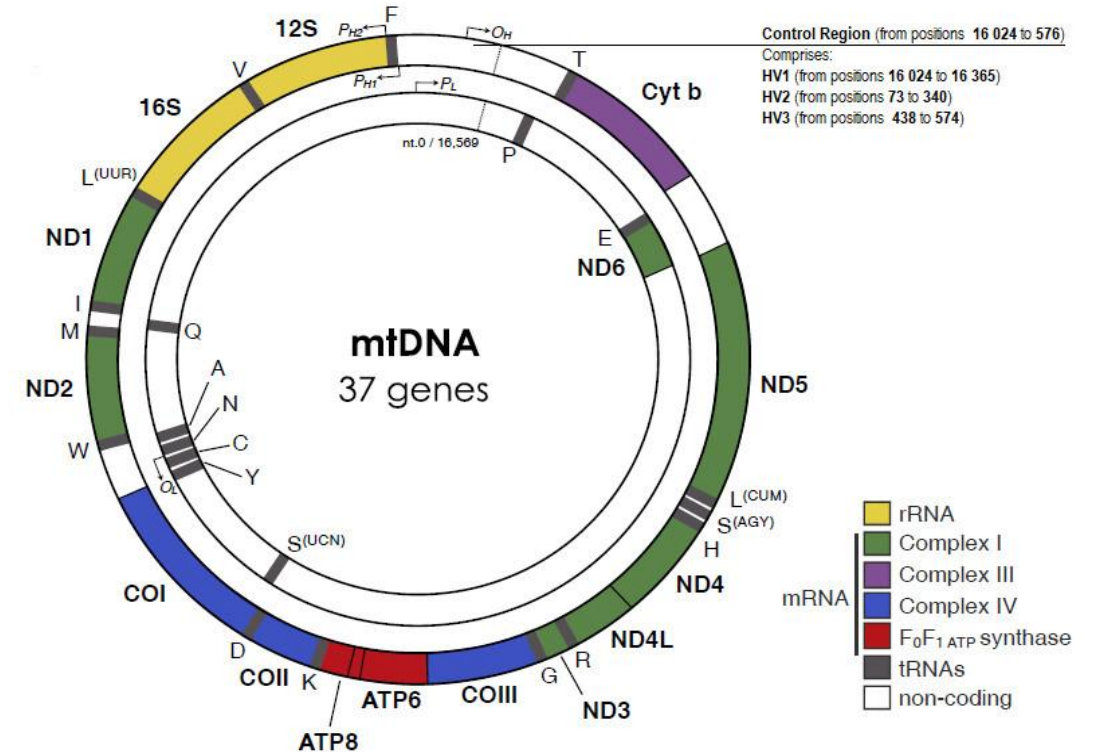
## DNA can be found in:

- Chromosomes in nucleus of eukaryotes
- Mitochondria

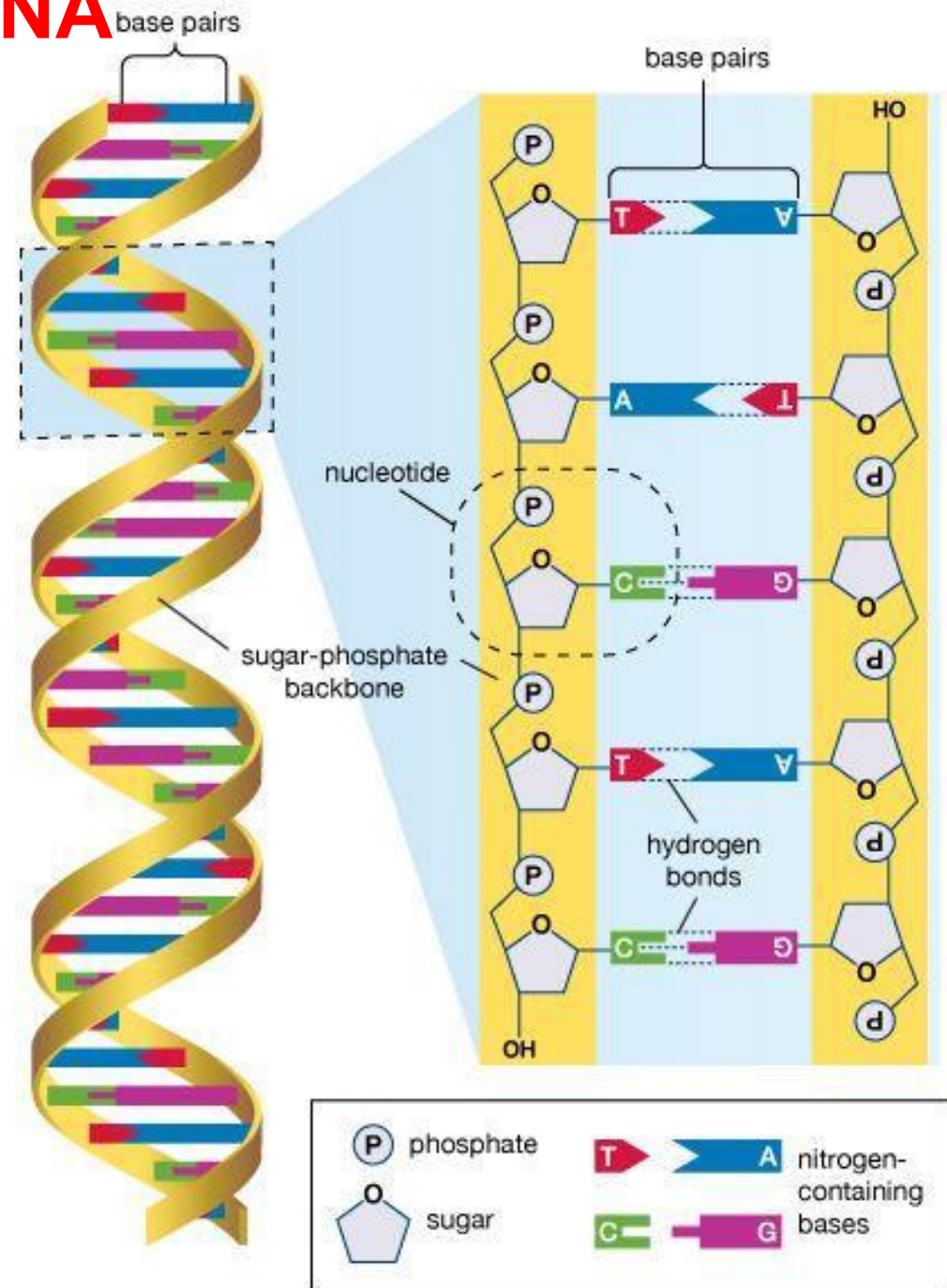
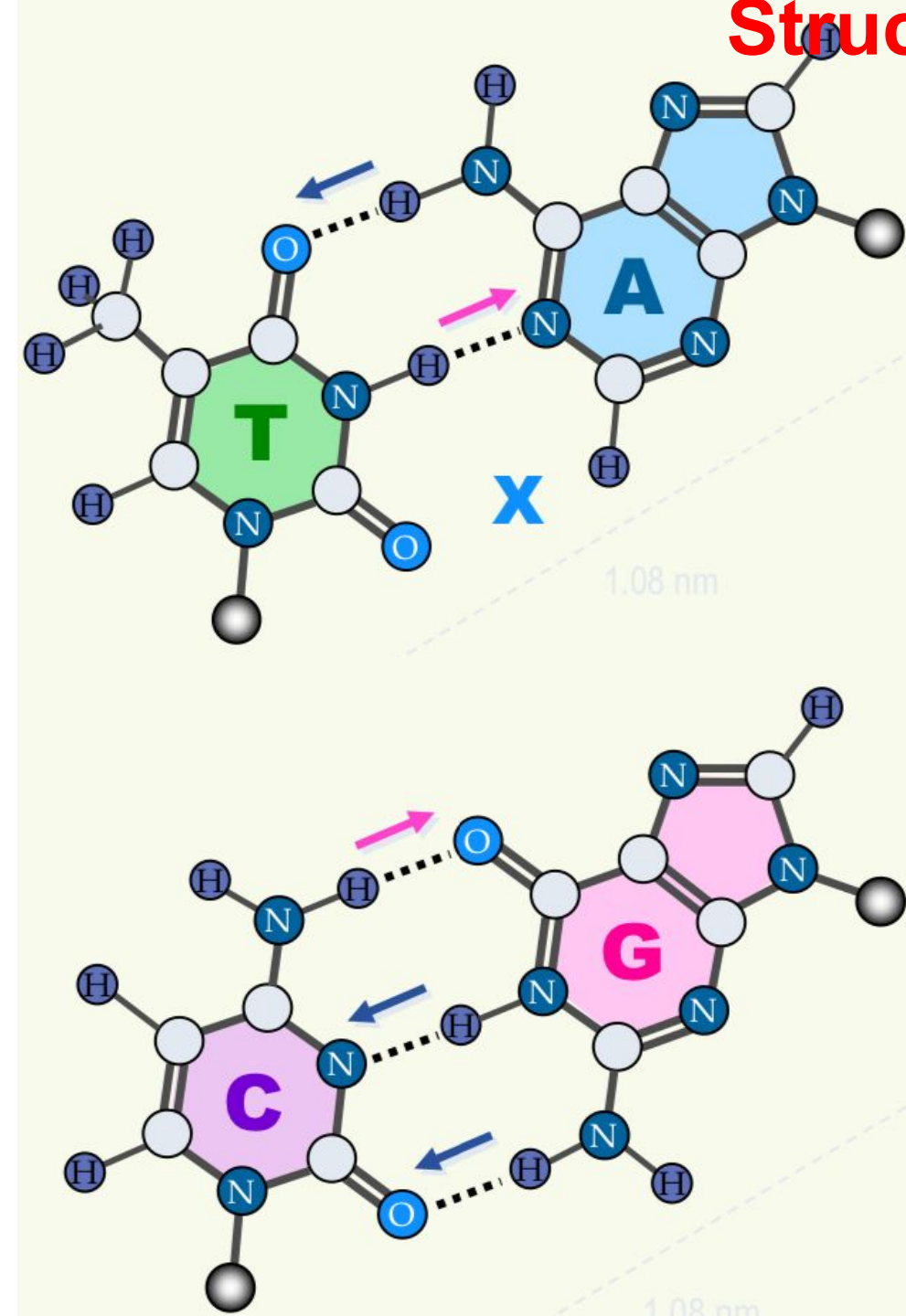
## Nucleus DNA



## Mitochondrial DNA



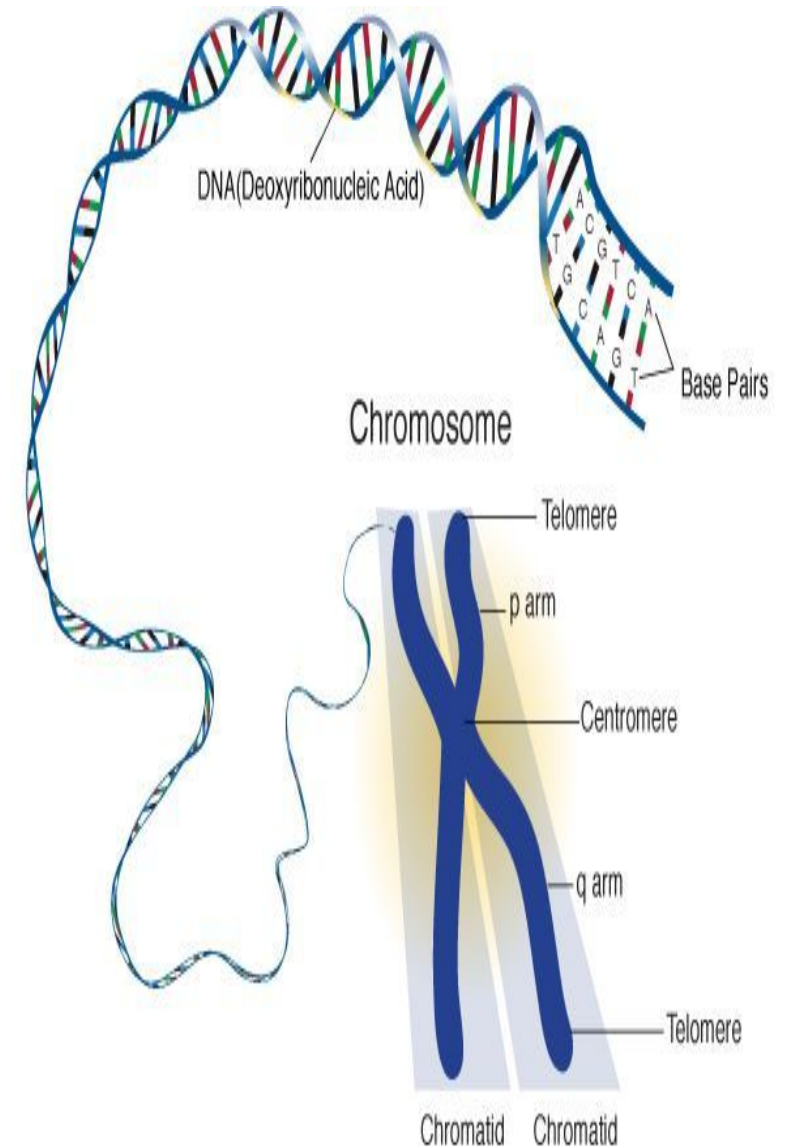
# Structure of DNA





# Definition of basics concept

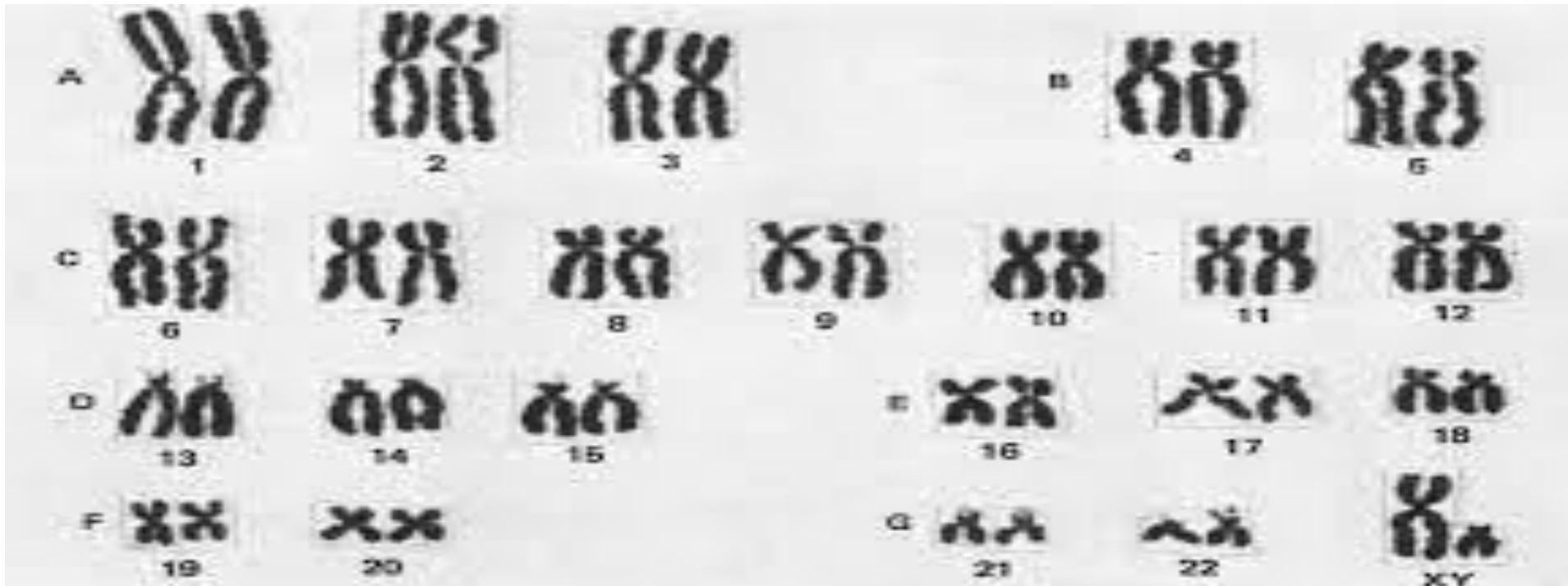
- **Chromosomes:** structure found inside the nucleus of a cell made of several DNA molecules
- A Human cell contains 23 pairs of chromosomes
- Each chromosome has 2 chromatids joint by a centromere
- Each chromatid contains a DNA molecule
- Every cell in our body has the same DNA



chromosome

# Definition of basics concept

**Karyotype:** Number and appearance of chromosomes in the nucleus of a eukaryotic cell



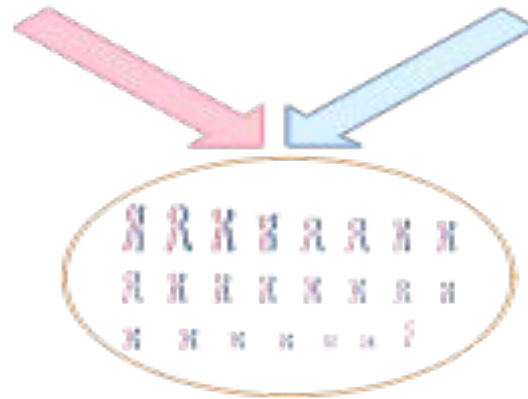
Karyotypes

# Chromosomes structure and Human genome

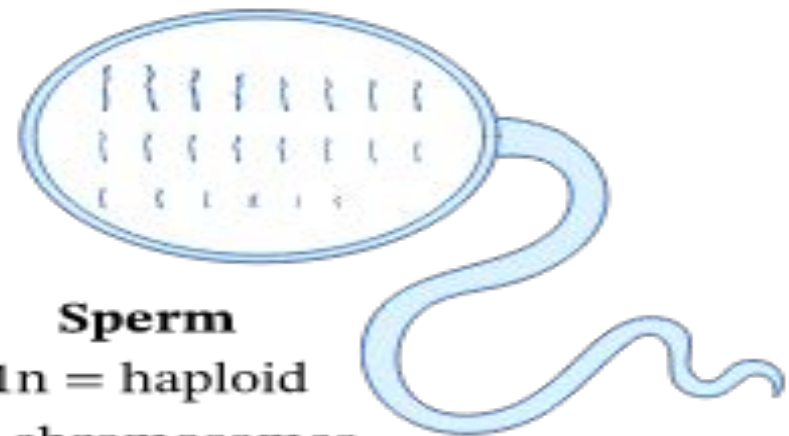
- Each chromosome pair contain two similar DNA molecules, one from each parent



**Ovum**  
 $1n = \text{haploid}$   
23 chromosomes



**Zygote**  
 $2n = \text{diploid}$   
46 chromosomes



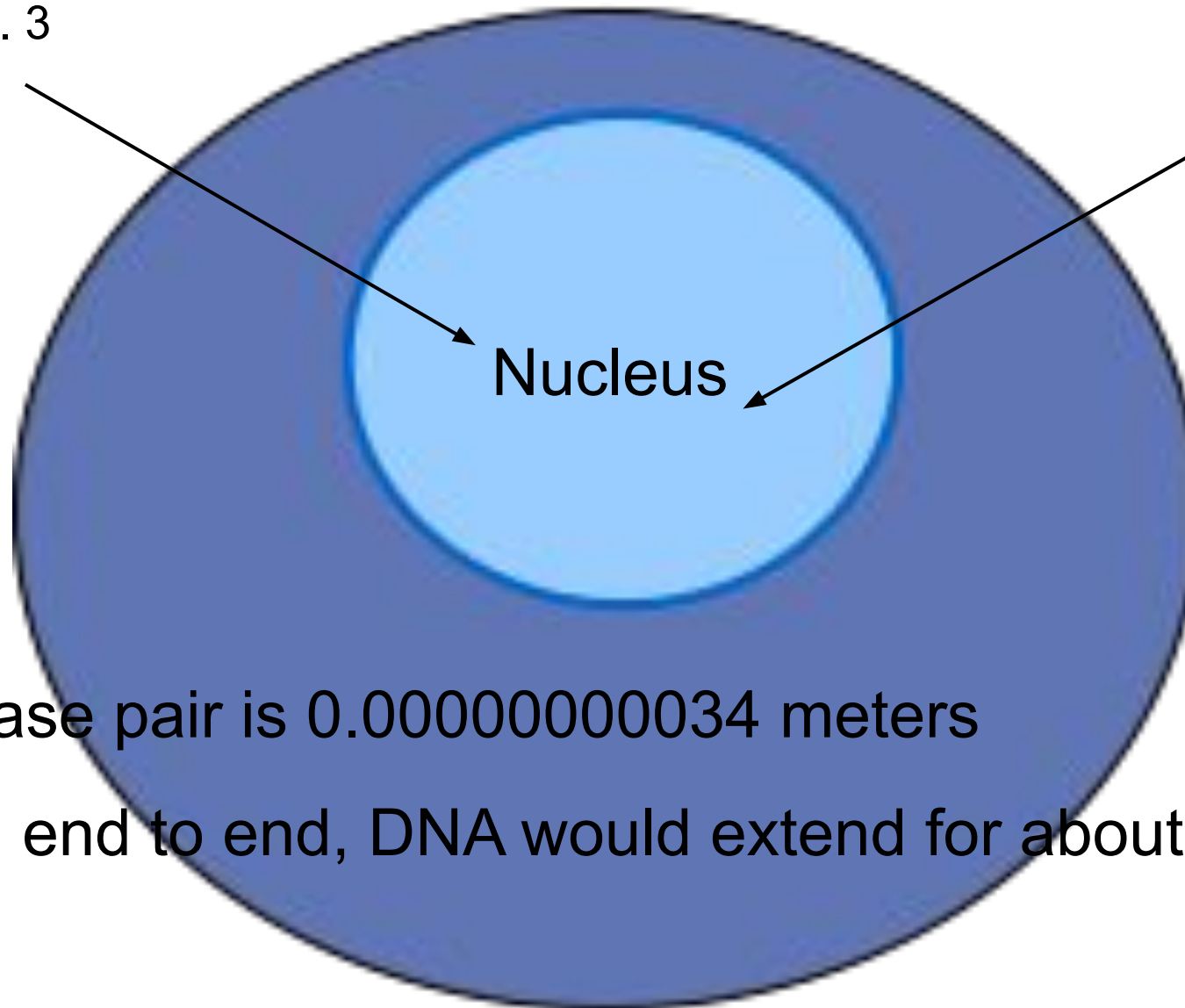
**Sperm**  
 $1n = \text{haploid}$   
23 chromosomes



# Chromosome structure and the Human genome

Genome = approx. 3  
billion base pairs

Human Genome = 46  
chromosomes



Nucleus

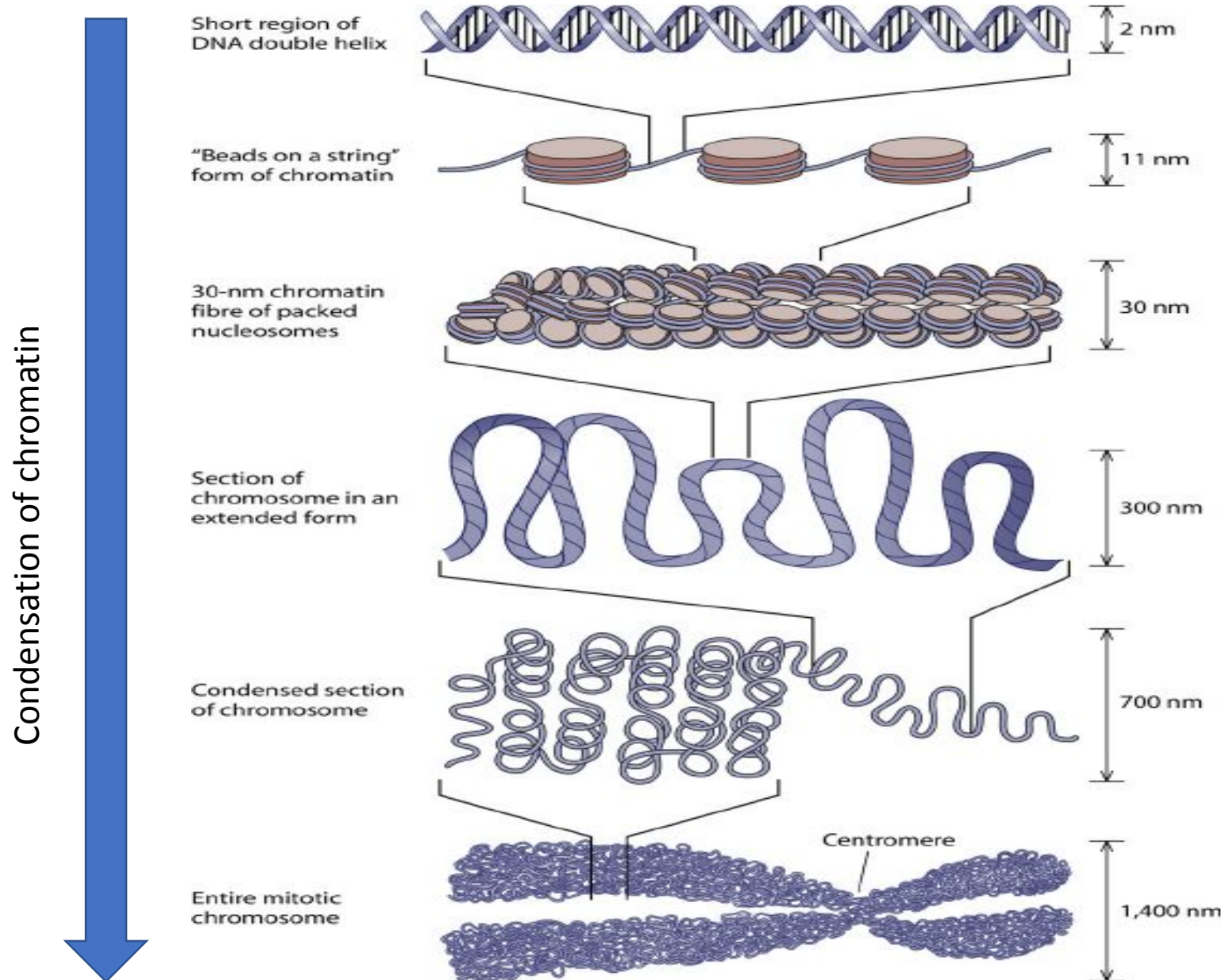
- One base pair is 0.00000000034 meters
- placed end to end, DNA would extend for about one meter long.

# Chromosomes structure and Human genome

- The total DNA of these chromosomes is called **human genome**.
- Most human cells are **diploid** and each cell contains a total of 2m of DNA.
- The long DNA interact with **nucleoproteins** to form **nucleosomes**
- A group of nucleosomes (about six) are further supercoiled to form a more condensed structure called **chromatin fibres**

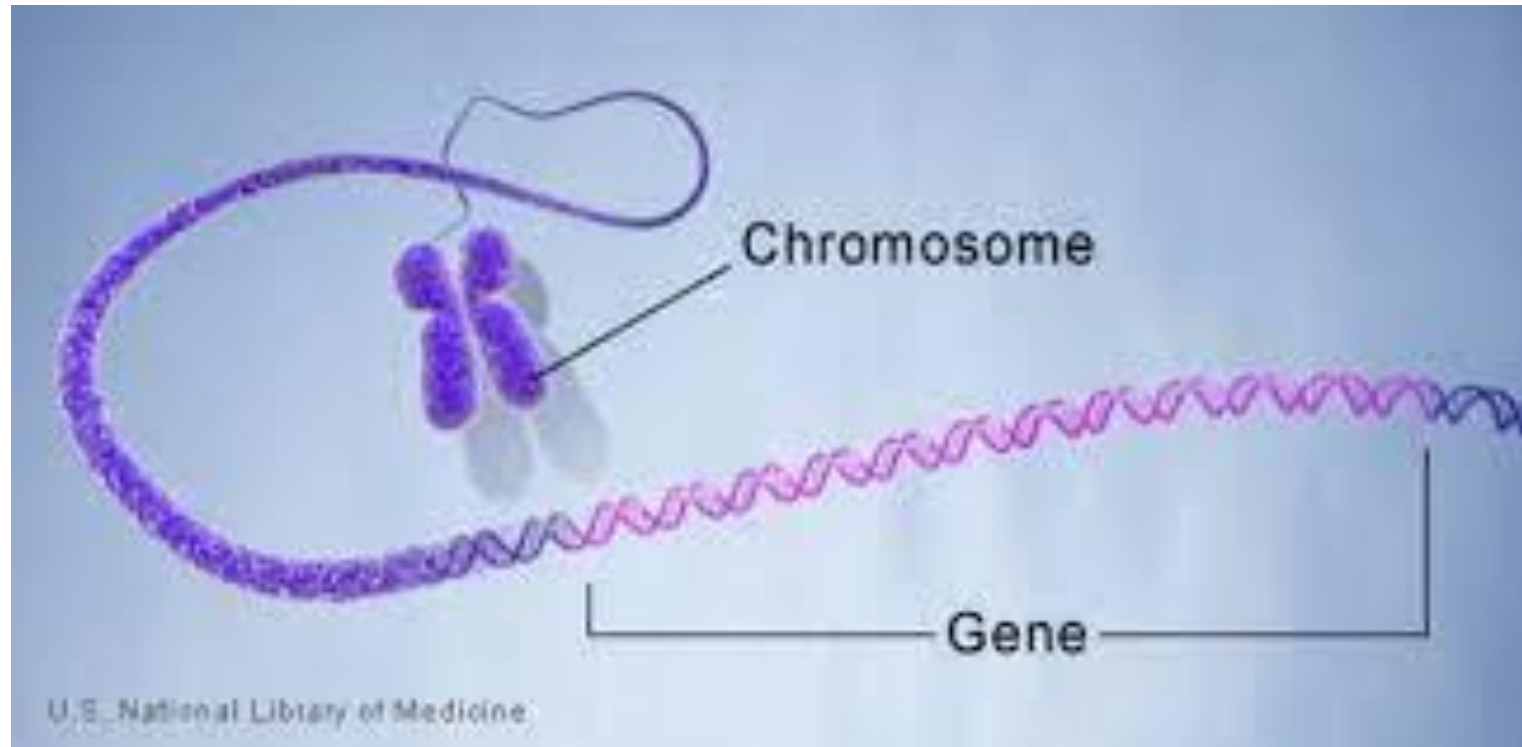
# Chromosomes structure and Human genome

- The chromatin fibres are further supercoiled to form loops and condensed to form **chromosomes.**



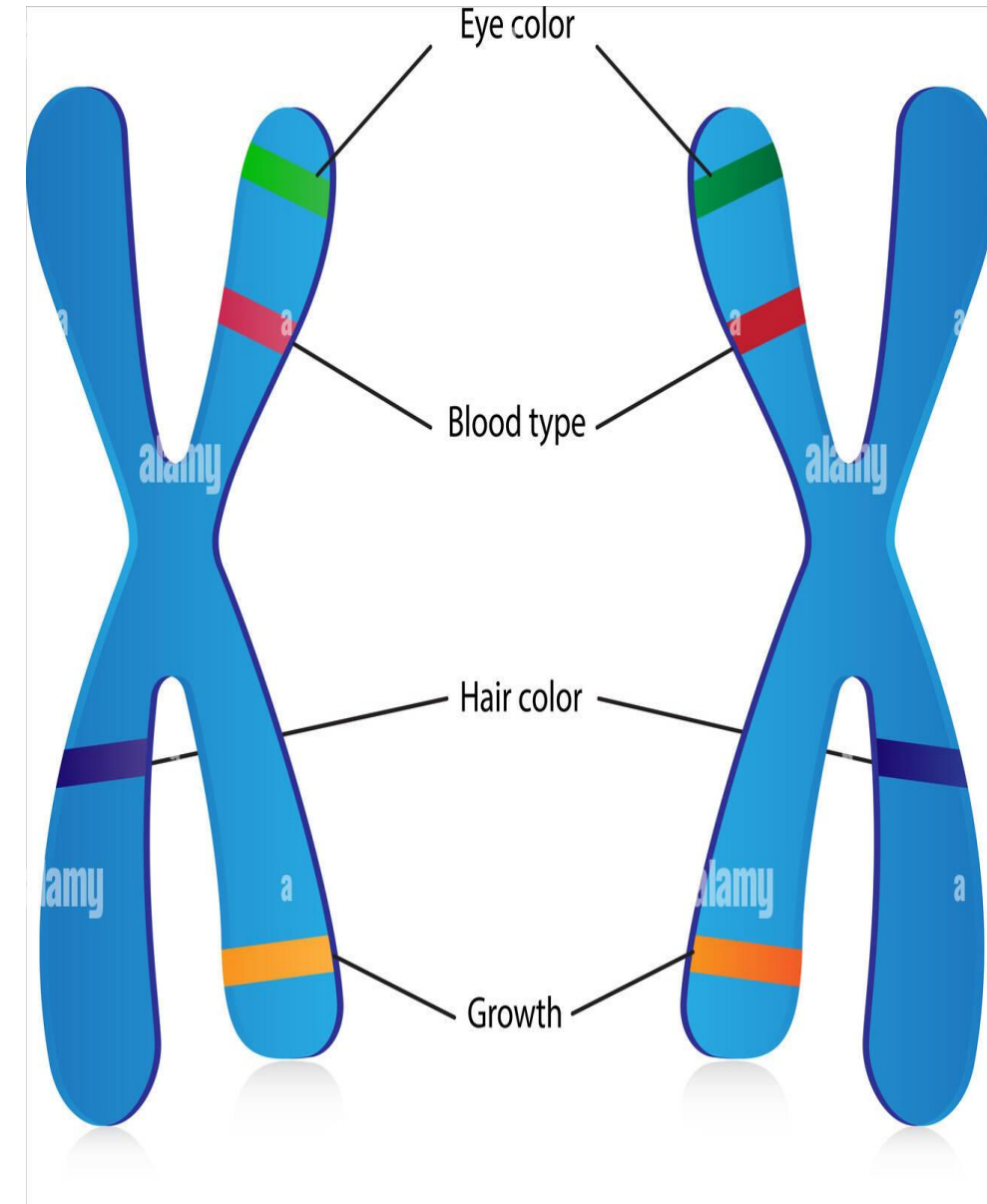
# Definition of basics concept

- **Gene** is the basic physical and functional unit of heredity
- Some encode proteins that form the shape and characters of the organism.
- Gene is present on the chromosome in a site called **Locus**



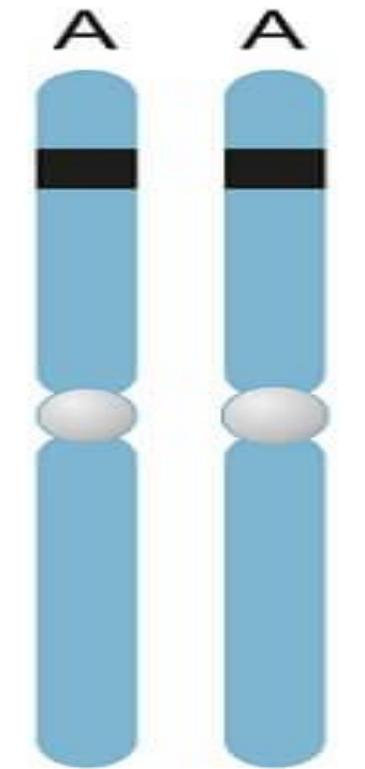
# Alleles

- Each pair of genes carries the code for a certain character
- As chromosomes are present in pairs, each gene is also present twice, each one is called an **allele**
- The two **alleles** although carry the same character, however, may be different forms of this character

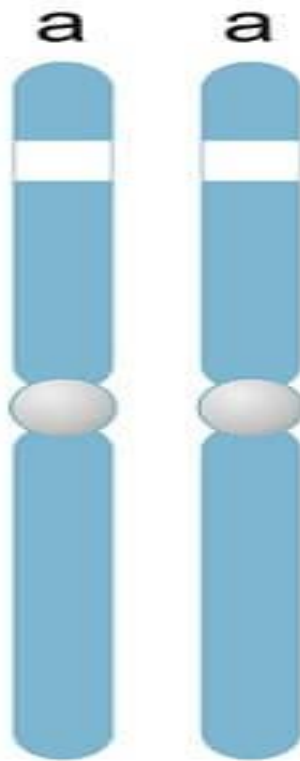


## Definition of basics concept

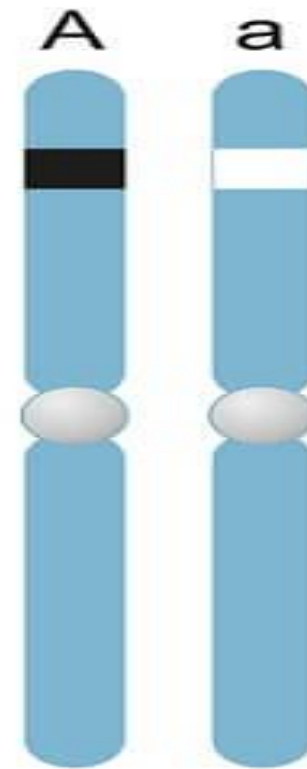
- **Homozygote:** The presence of two identical alleles at a particular gene locus
- **Heterozygote:** when the two alleles different



Homozygous  
organism



Homozygous  
organism



Heterozygous  
organism



# Definition of basics concept

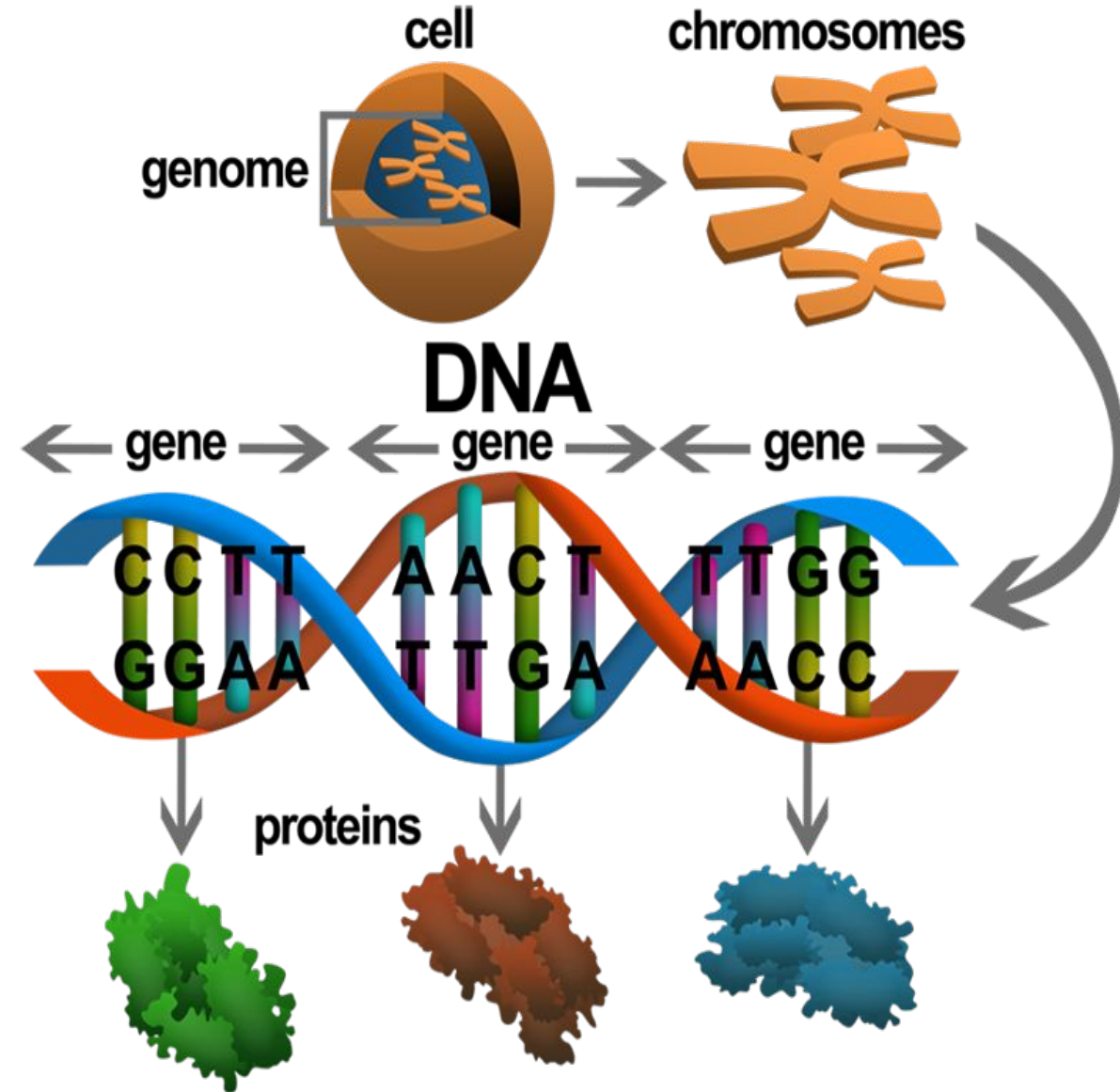
- **Phenotype:** an individual's observable traits: height, skin/eye color...
- **Genotype:** genetic contribution to the phenotype



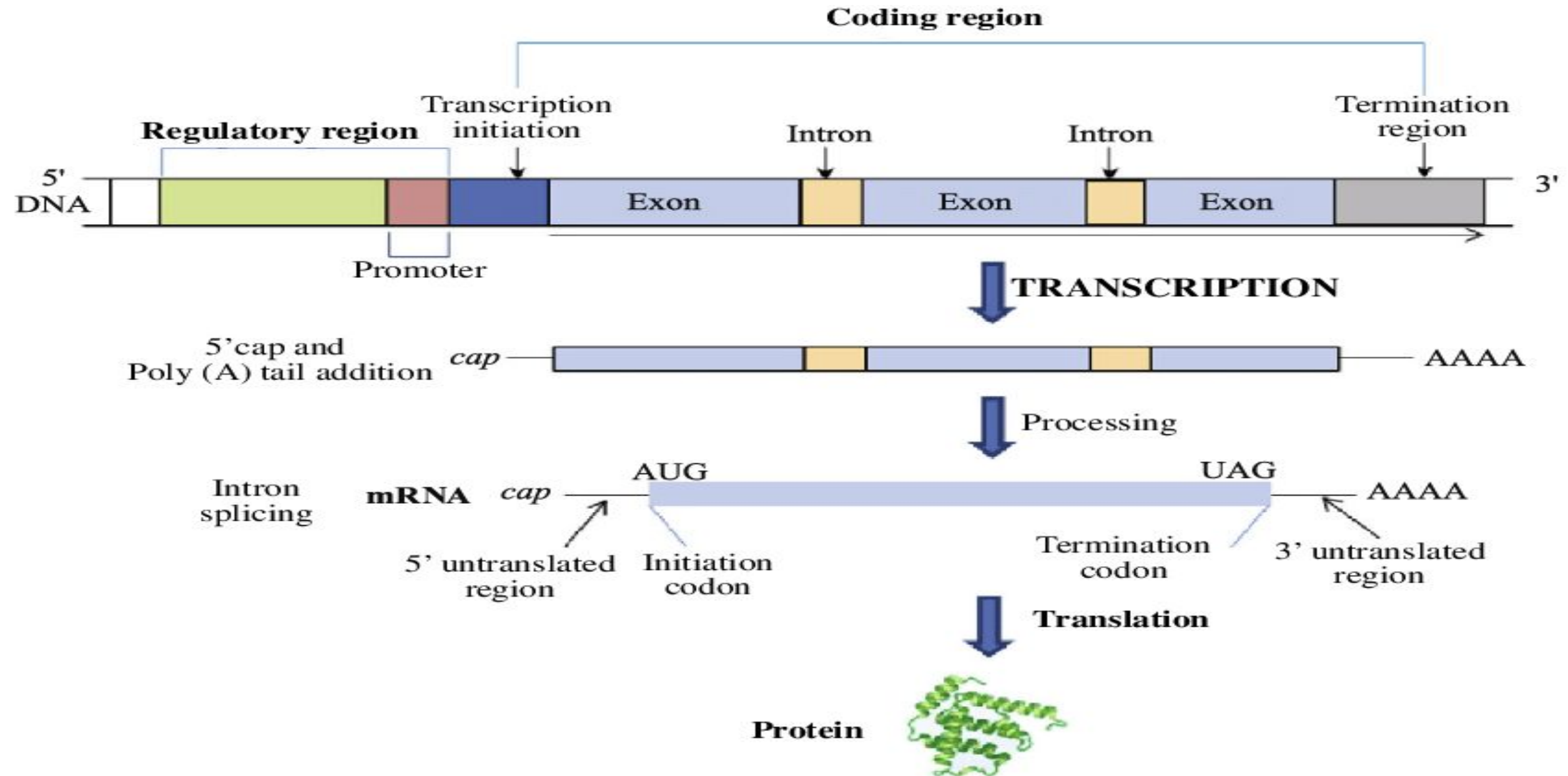
# Definition of basics concept

- **Dominant** : is always expressed when present
- **Recessive**: is only expressed when no dominant genes are present.
  - dominant allele + dominant allele = **dominant phenotype**
  - dominant allele + recessive allele = **dominant phenotype**
  - recessive allele + recessive allele = **recessive phenotype**

- The expression of a **gene** is a series of chemical syntheses and reactions leading to the production of a **protein**:
- First step, synthesis of an **mRNA**: this is **TRANSCRIPTION**.
- Second stage, the mRNA is "read" allowing the **synthesis of the protein**: this is **TRANSLATION**.



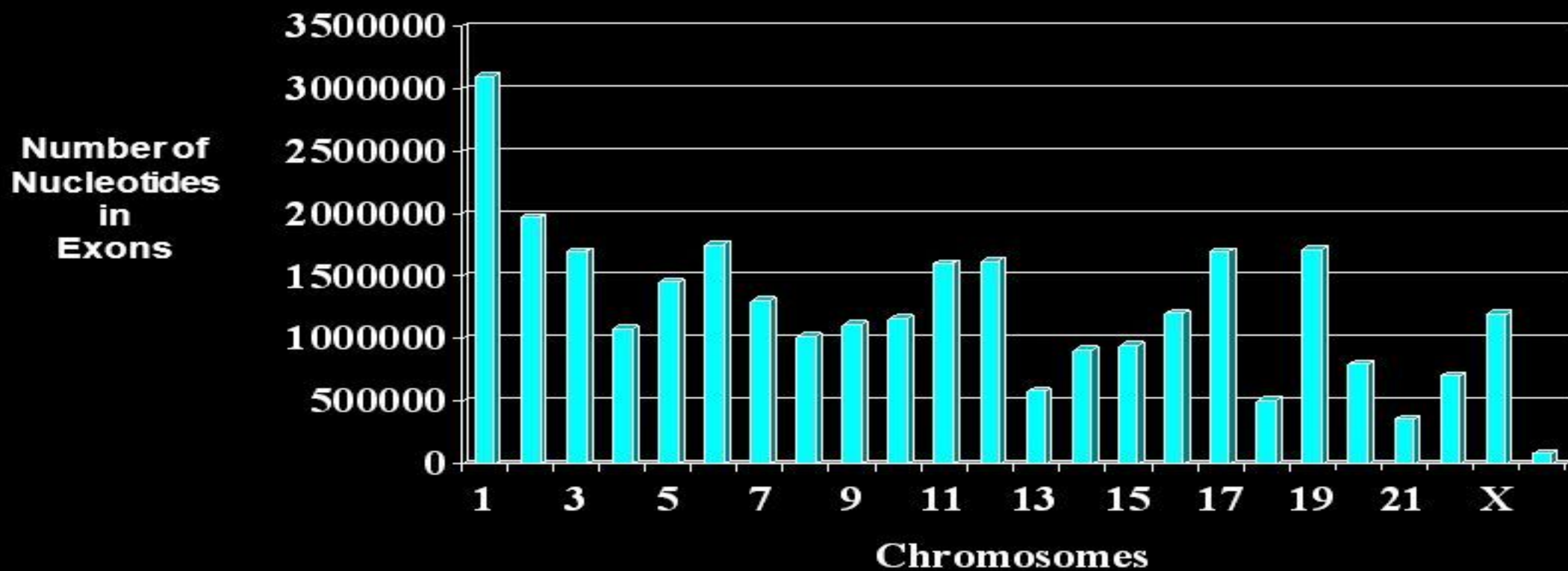
# Structure of a gene



# Definition of basics concept

- **Exons:** are coding sequences of a gene that can be translated to protein
- **Introns:** are non-coding nucleic acid within a gene that separate exons and does not code for proteins

# Some chromosomes are richer in genes than others





- DNA sequence in any two people is **99.9%** identical
- Only **0.1%** is **unique!**



# What makes the genome unique?

A change in the usual DNA sequence at a particular gene locus (**Mutation**) can occur and change the outcome.

Original sequence:

5'-GCC ATT TCA ACT GCC TGC AGC 3'



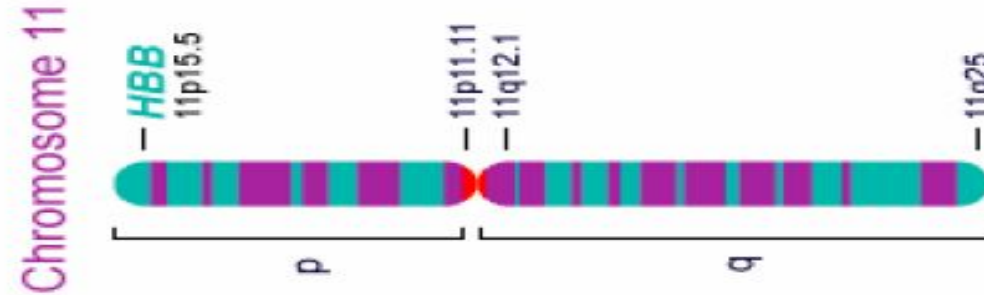
MUTATION

5'-GCC ATT TCG AGC CTG CAC TAG C 3' insertion

shifting the reading frame

5'-GCC ATT TCG CCT GCA CTA GC 3' loss

# Example



Sequence for normal adult hemoglobin:

Nucleotide	CTG	ACT	CCT	GAG	GAG	AAG	TCT



Wild-type Hemoglobin Protein



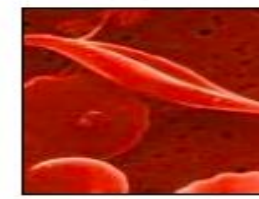
Normal Red Blood Cell

Sequence for mutant hemoglobin:

Nucleotide	CTG	ACT	CCT	GTG	GAG	AAG	TCT



Mutant Protein



Abnormal Red Blood Cell

# What is a Pedigree

A family tree is a genealogical chart showing the ancestry, descent, and relationship of all members of a family

- Provides a clear record of genetic information in a family
- Provides medical data
- Demonstrates biological relationships
- A tool for establishing the pattern of inheritance
- Used to calculate risks
- Identifies at-risk family members
- Can be used as a psychosocial tool

# Pedigree Rules

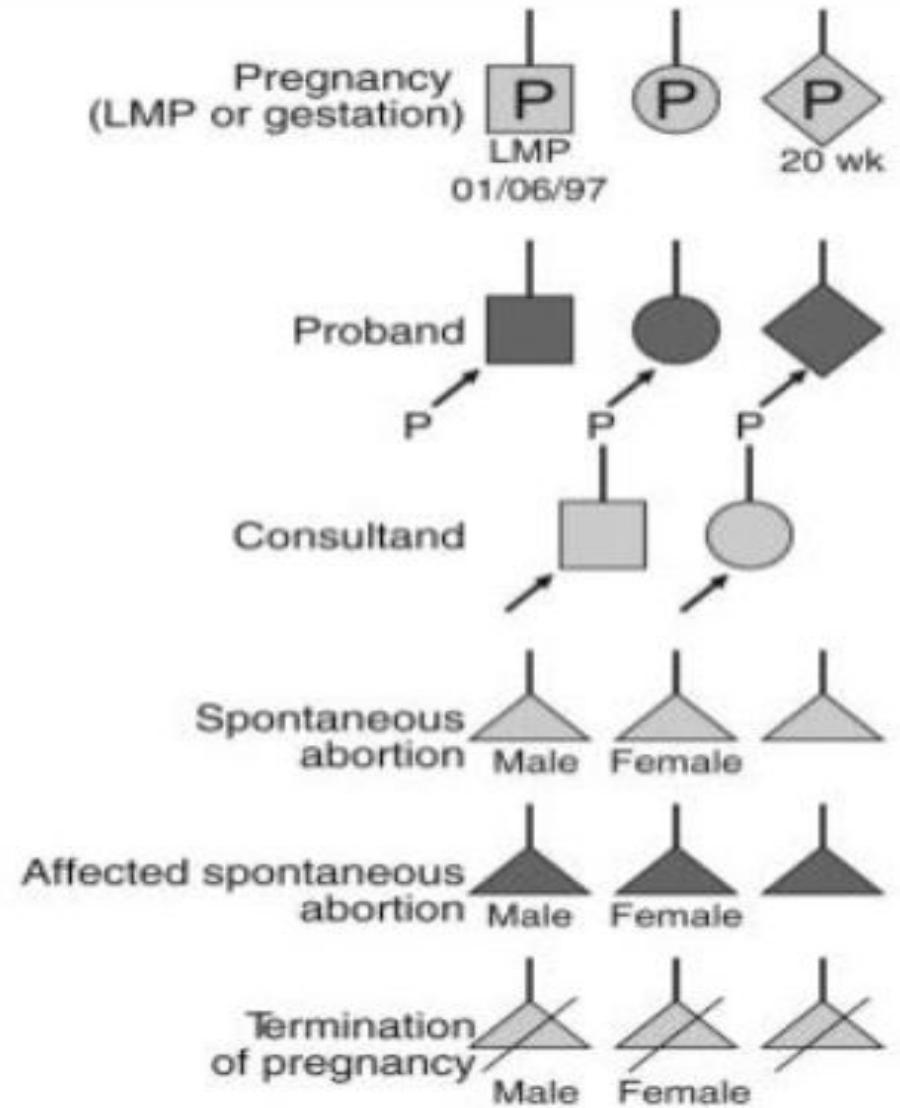
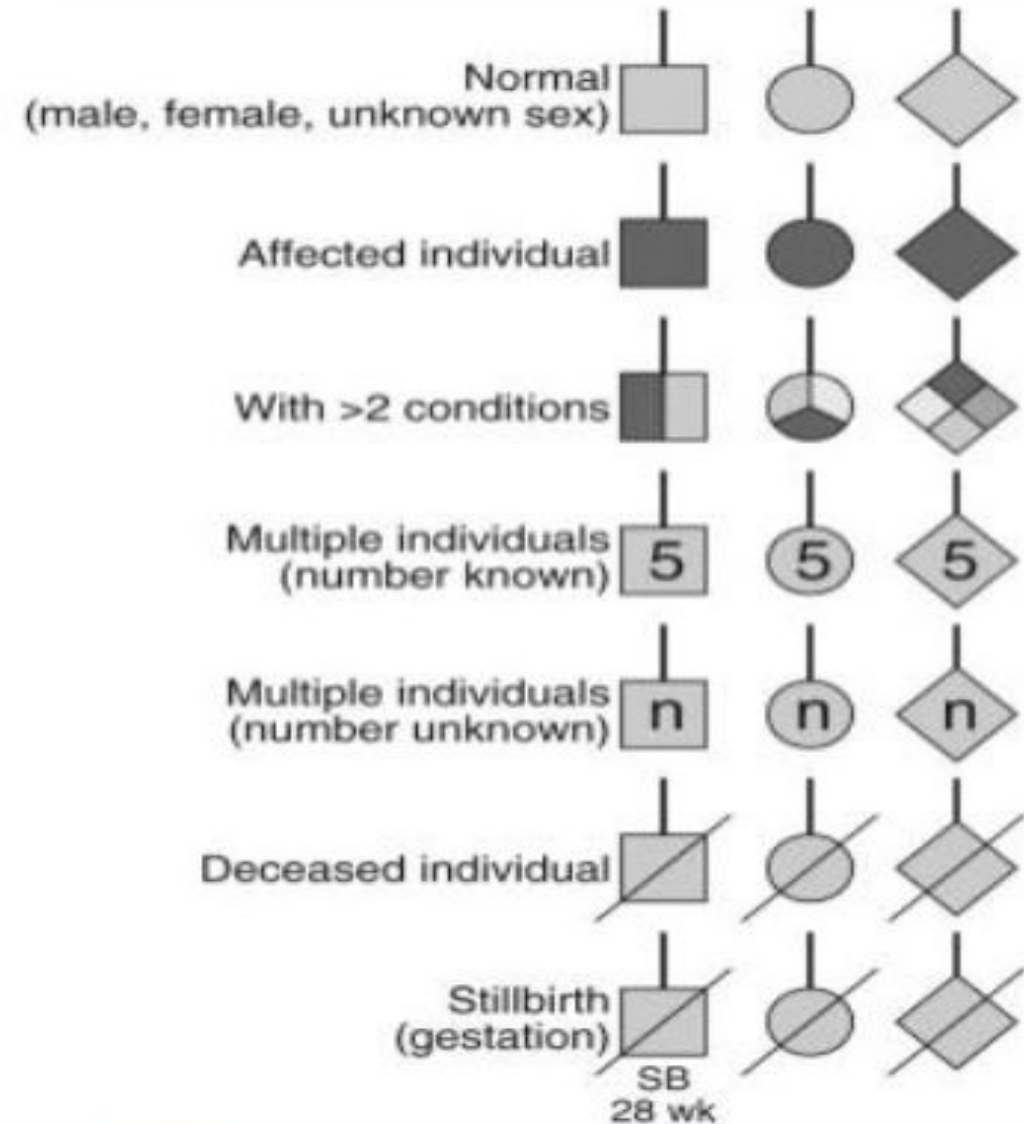
- Must be easy to read
  - Must use standardized symbols
  - Indicate the proband/index person with an arrow
  - Indicate when a person is diseased
  - 3 generations
  - Multiple miscarriages can be indicative of genetic disorders
  - If possible add age of onset of condition
  - Be vigilant of the way you ask questions (E.g. Marriage)
- 
- For research – label all individuals who you have taken a sample for

Can someone else understand your pedigree??



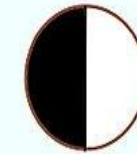
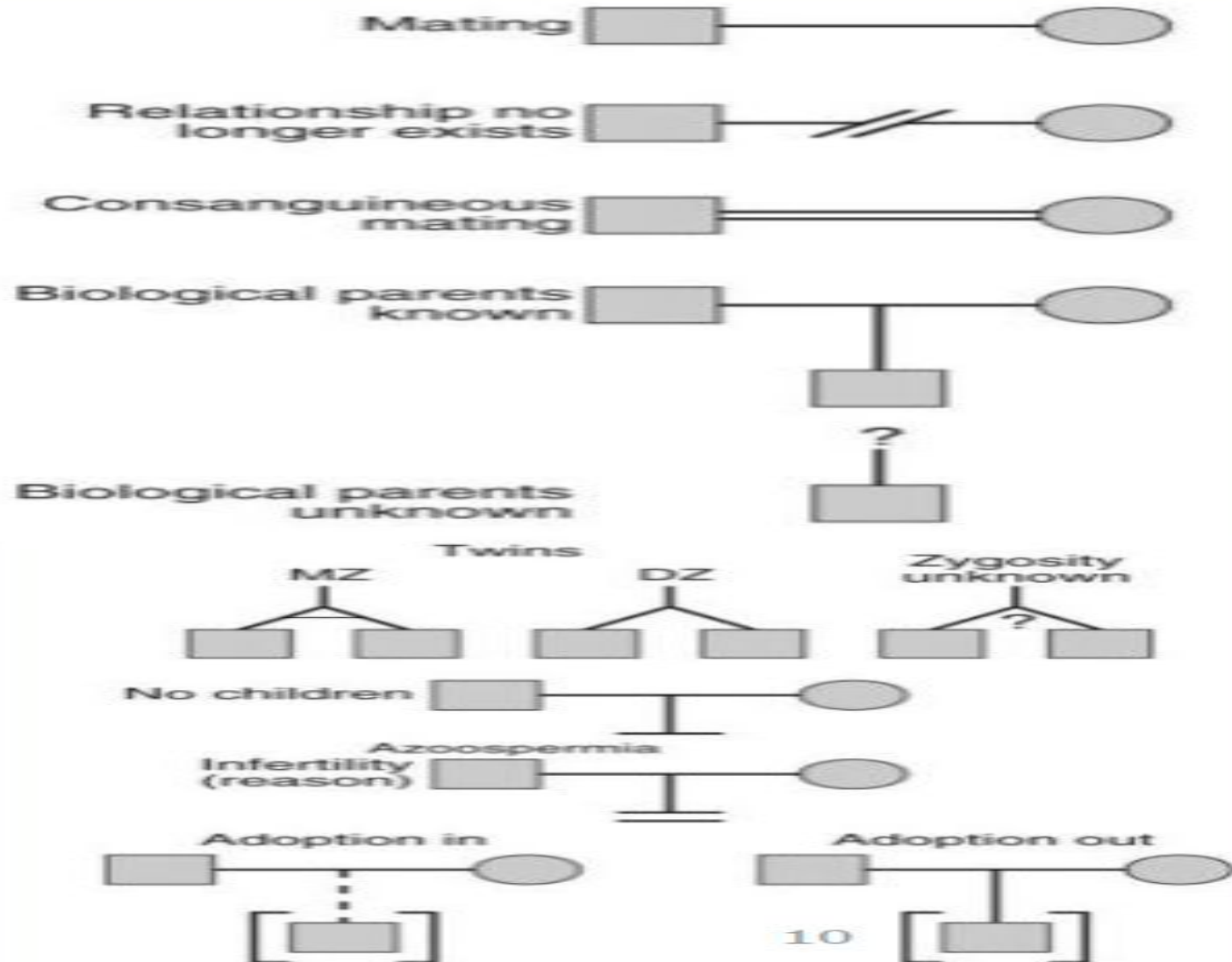
# Pedigree Key

## Individuals





# Pedigree Key



=

**Carrier  
of the trait**



Female heterozygous for recessive allele



Male heterozygous for recessive allele

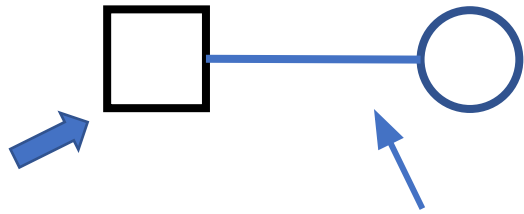


# Drawing a Pedigree

**Step 1: Start with the proband/consultant  
(male or female)**

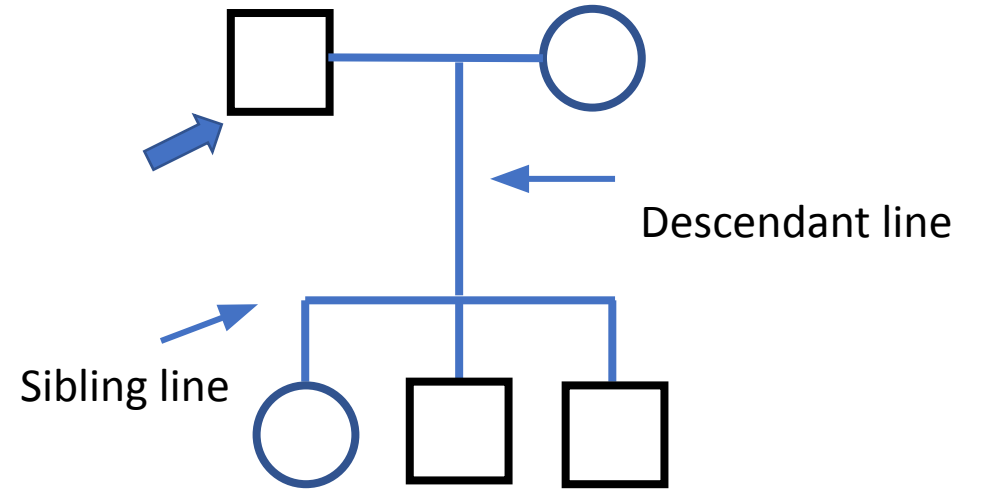


**Step 2: If proband/consultant is an adult  
draw their partner**



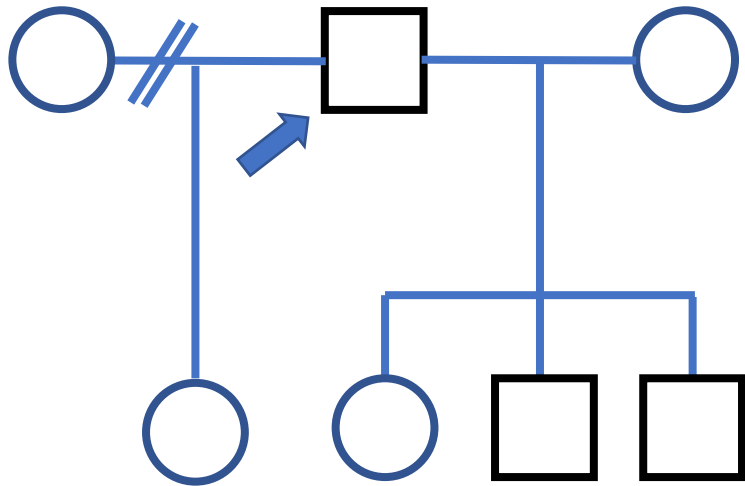
Relationship line

**Step 3: Draw the children, miscarriages or  
stillbirths of the proband**

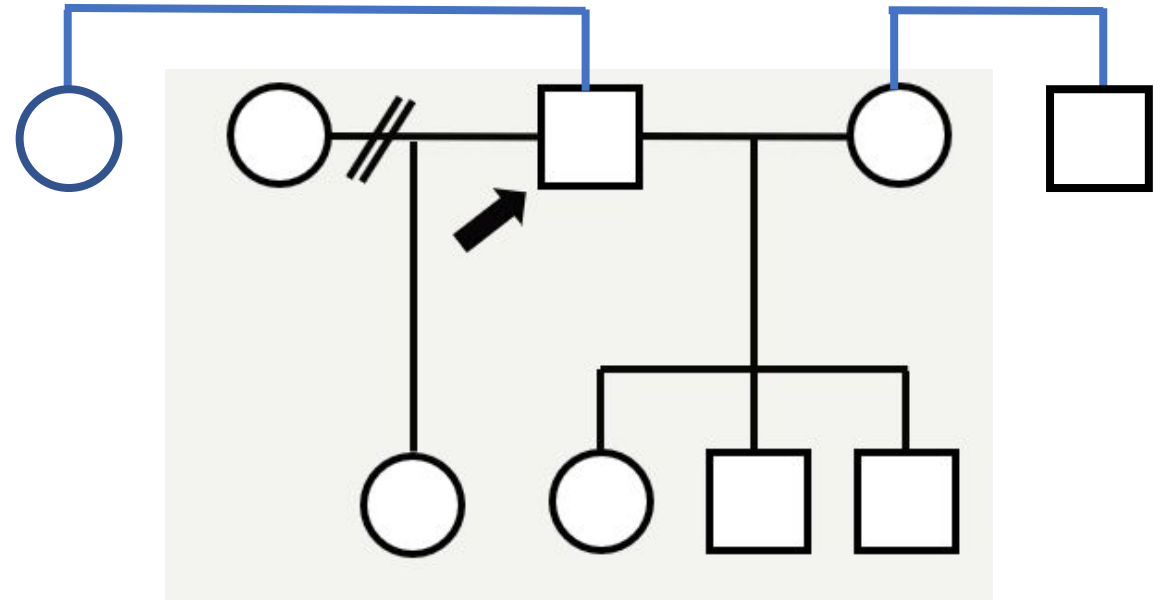


# Drawing a Pedigree

**Step 4: Draw children from previous relationships**



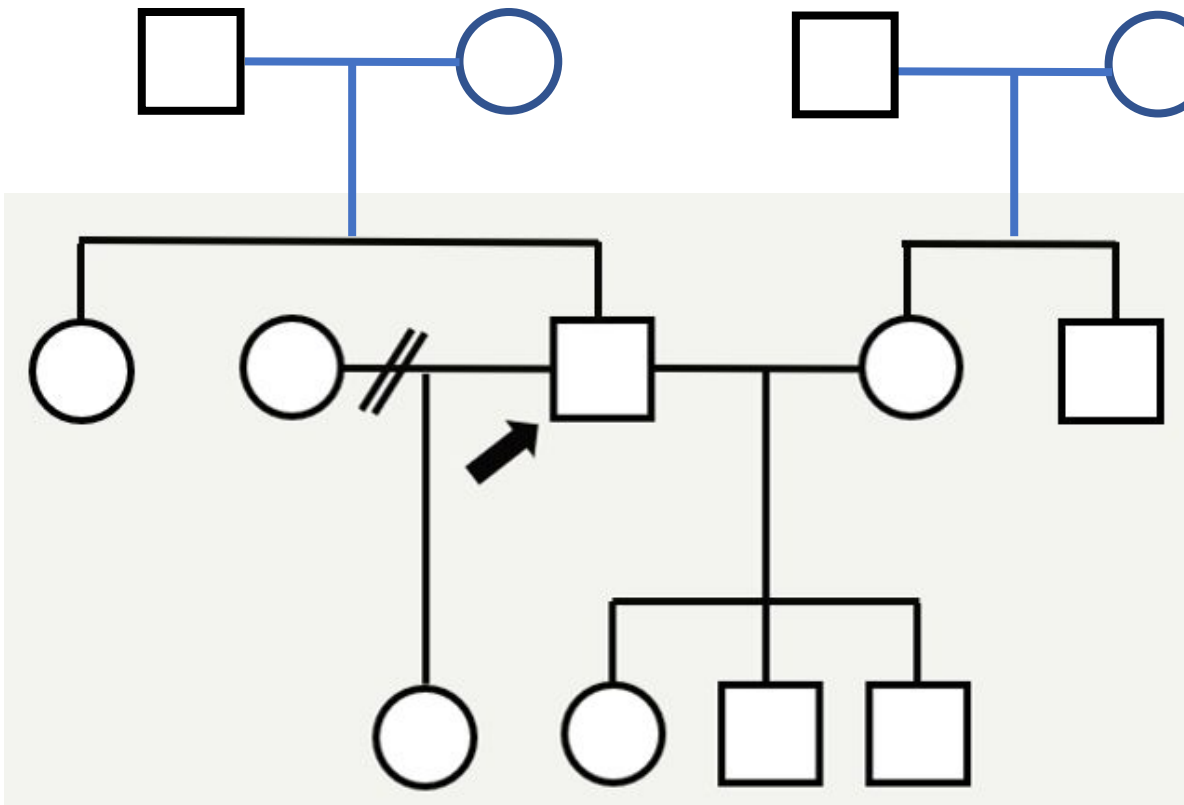
**Step 5: Draw the siblings & half siblings of the proband/consultant and of their partner**



# Drawing a Pedigree

**Step 6: Draw the parents of the proband/consultant and partner**

**Remember to add all the important information (eg: death, medical conditions)**



**NOTE:** If the proband is a child, you will draw their siblings, parents and half siblings.  
Then you will draw uncles and aunts and grandparents

**Merci**