Part1:Introduction to Human Genetics

Lecturer: Sabha Rabaya, MSc

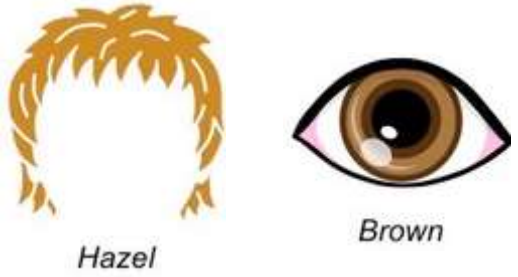
Lecture outcomes

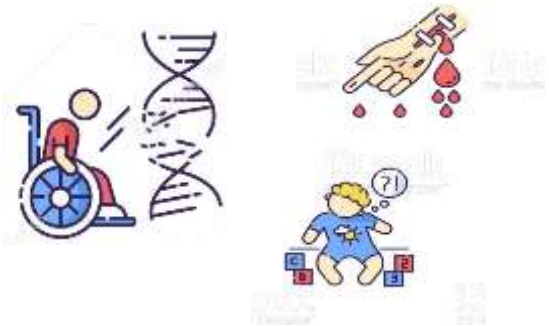
- Understand basic genetic terminology
- Describe the organization of the human genome
- Review the Central Dogma of molecular biology

Traits

- We are all unique
- Traits ⟶ some are inherited , some are shaped by the environment

Physical traits ⟶ Disorders





- Most traits are influenced by genetics and environment factors

What is Genetics?

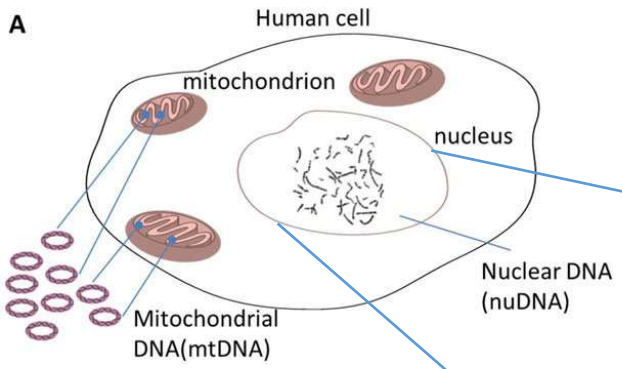
- What does genetics mean in science?

Genetics is the scientific study of genes and heredity—of how certain qualities or traits are passed from parents to offspring as a result of changes in DNA sequence.

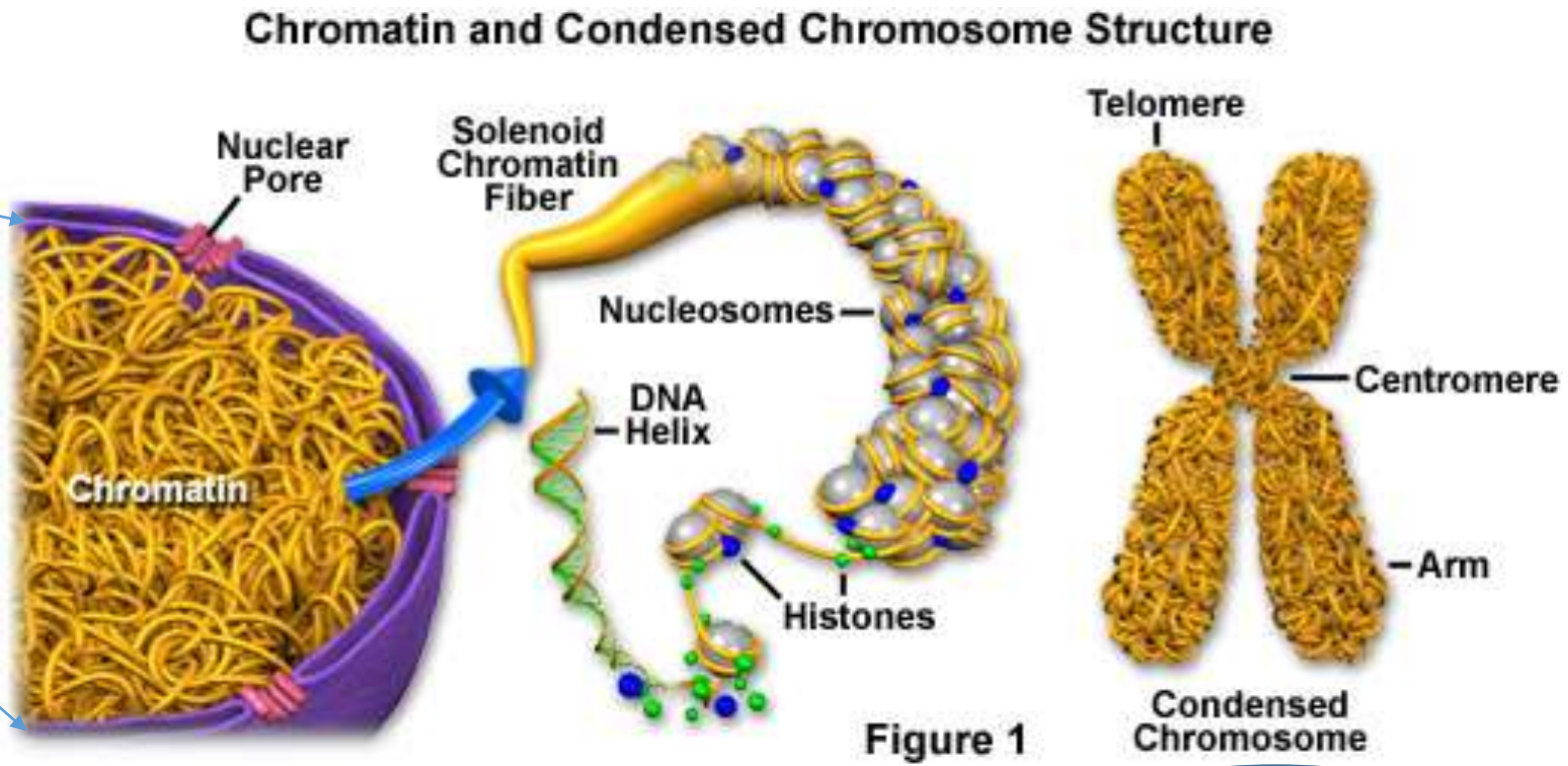
- Heredity is the transmission of traits and biological information between generations

- Human genetics: The scientific study of genes and heredity in humans

Basics of Human Genetics



DNA



Chromatin

Genome

Chromosome

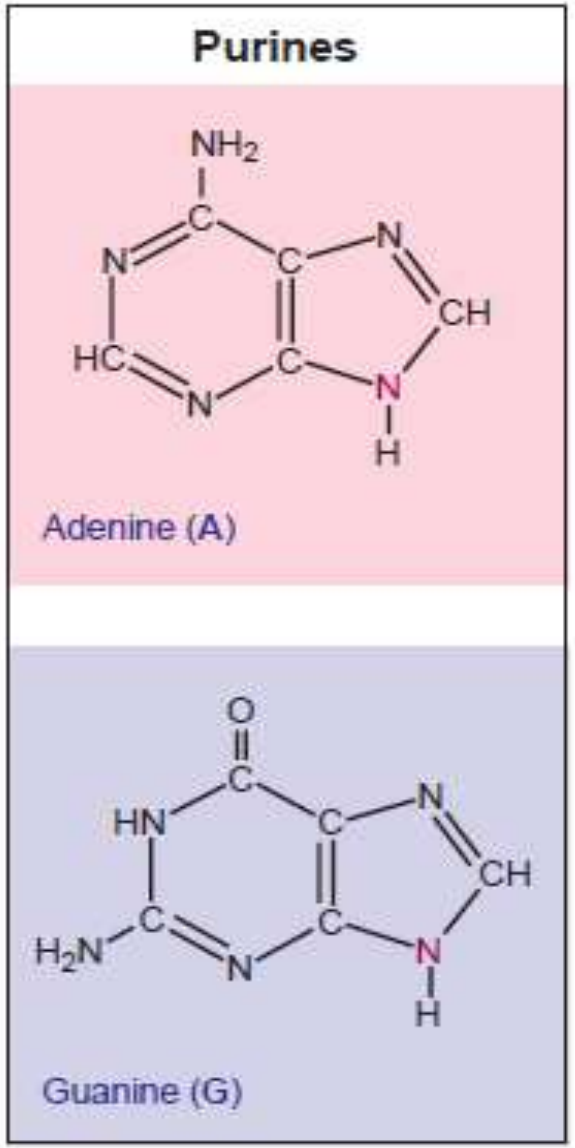
Gene

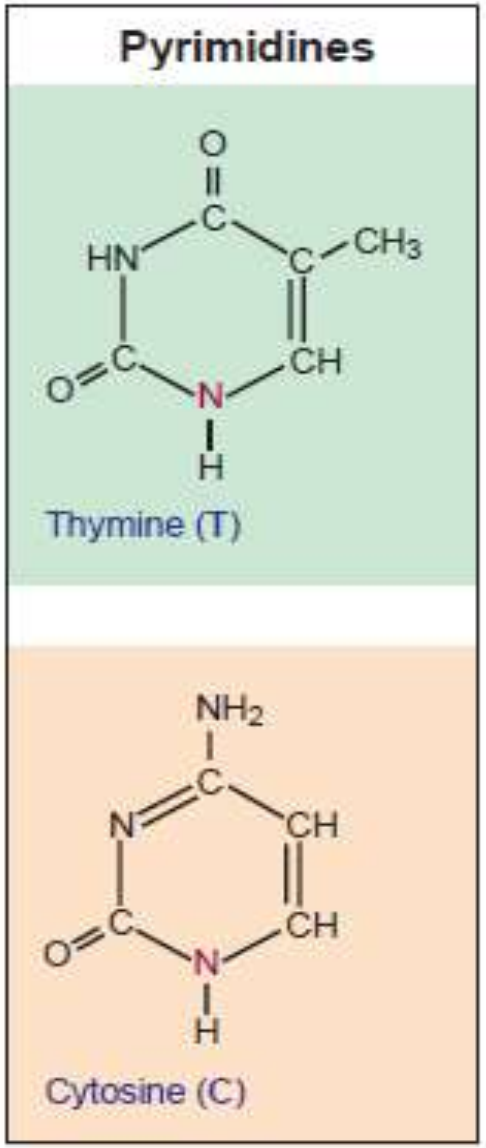
Chromatid

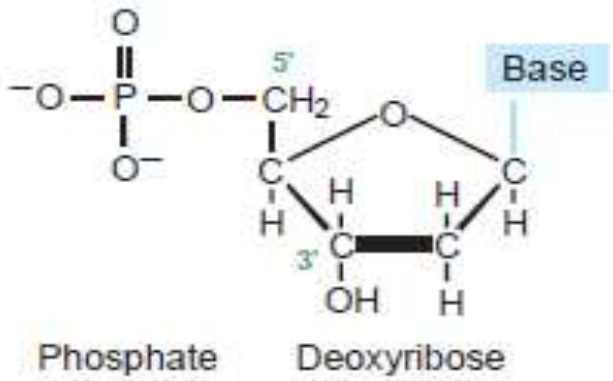
Basics of Human Genetics: DNA

- DNA: The hereditary material :
  - Nucleic acid
  - Deoxyribonucleic Acid
  - DNA: Polynucleotide chain
  - Building block: Nucleotides
  - Stores genetic information

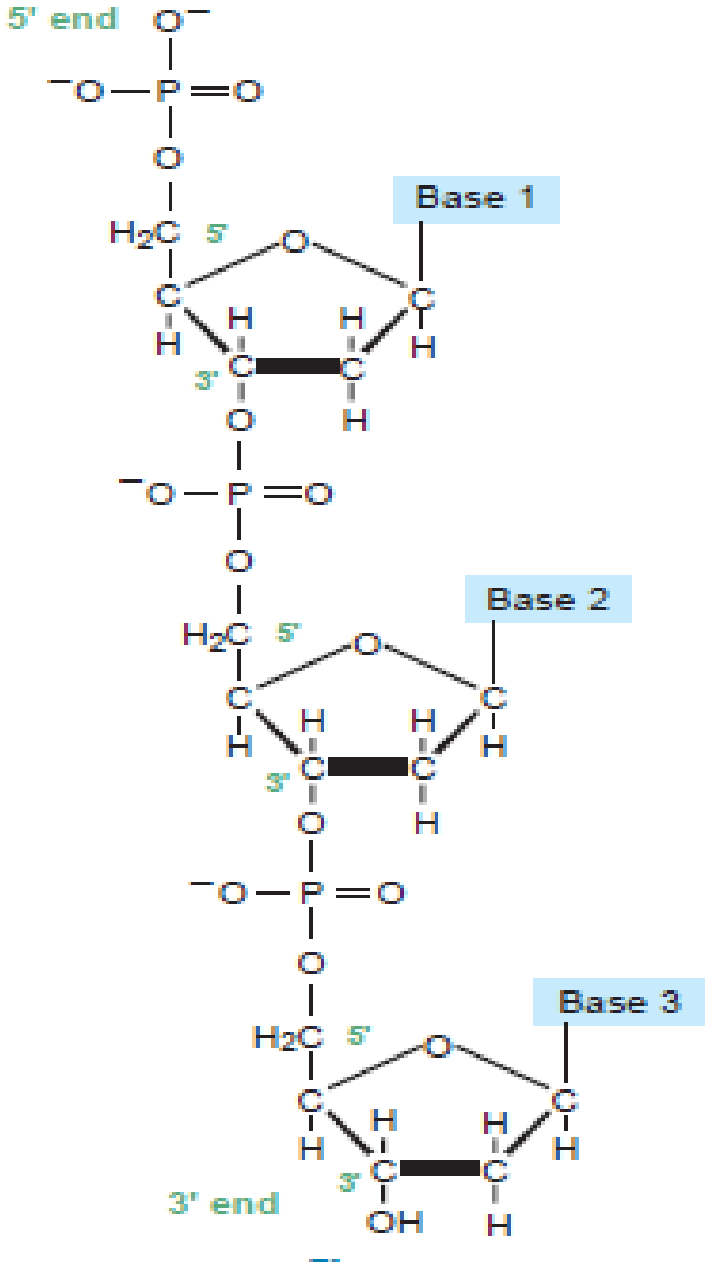
Basics of Human Genetics: Nucleotides





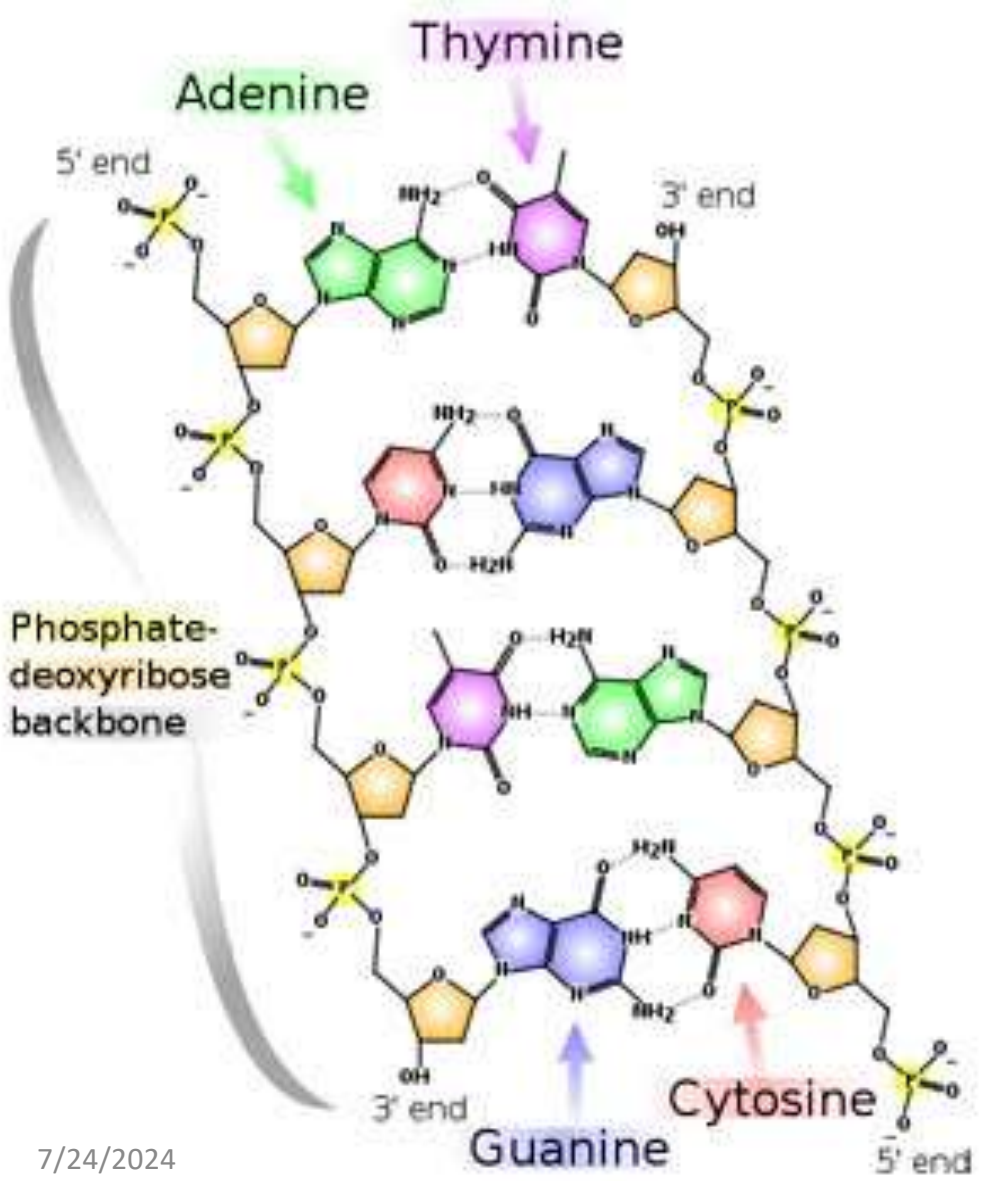


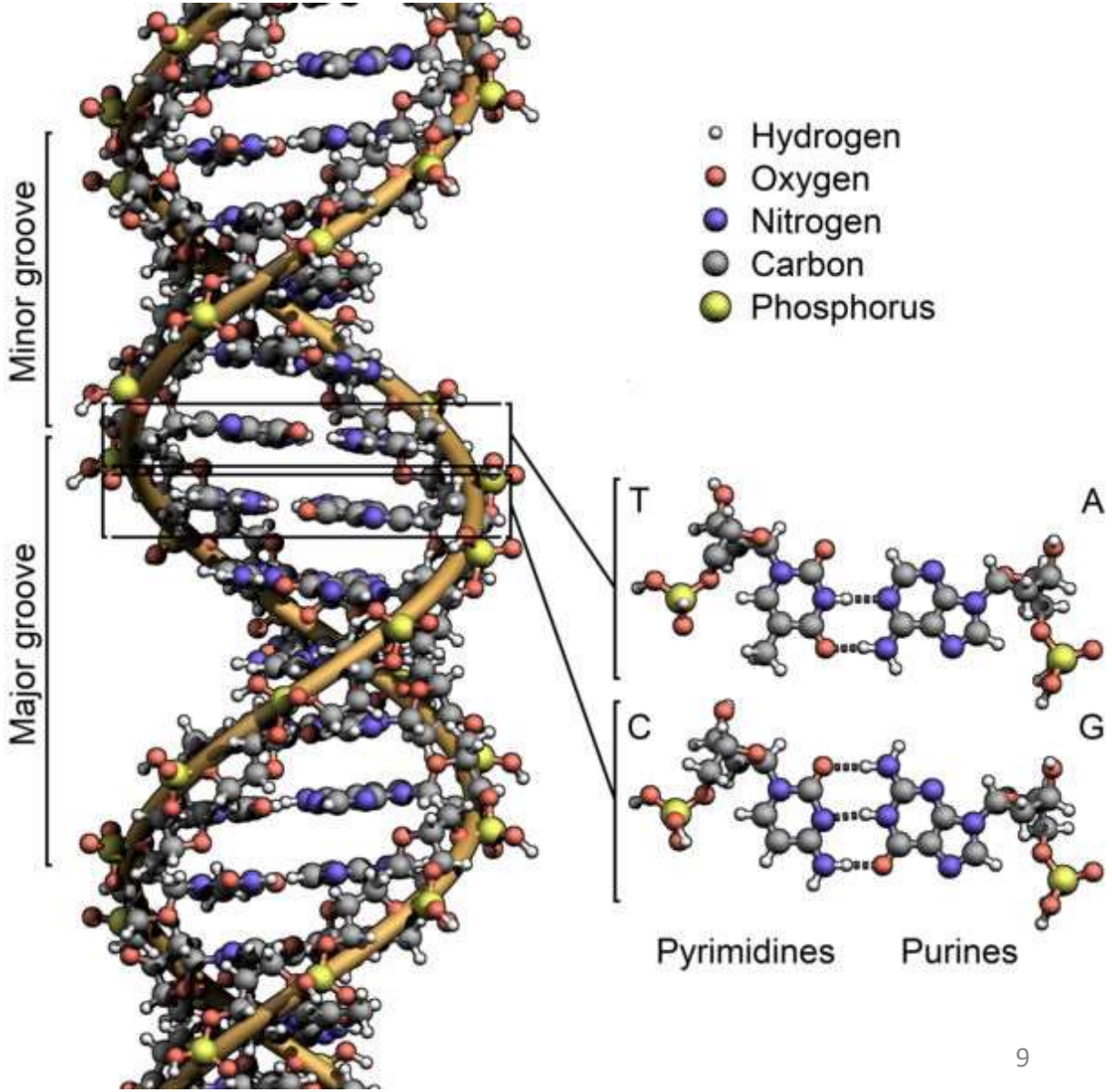
Basics of Human Genetics: DNA Polynucleotide



Basics of Human Genetics: DNA, Double Helix

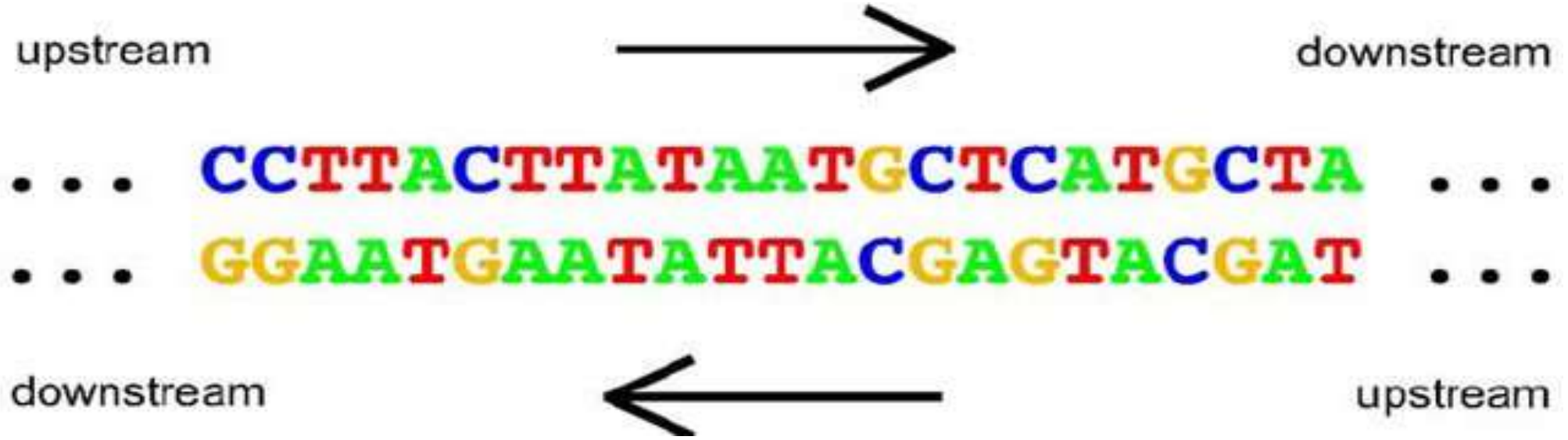
Video





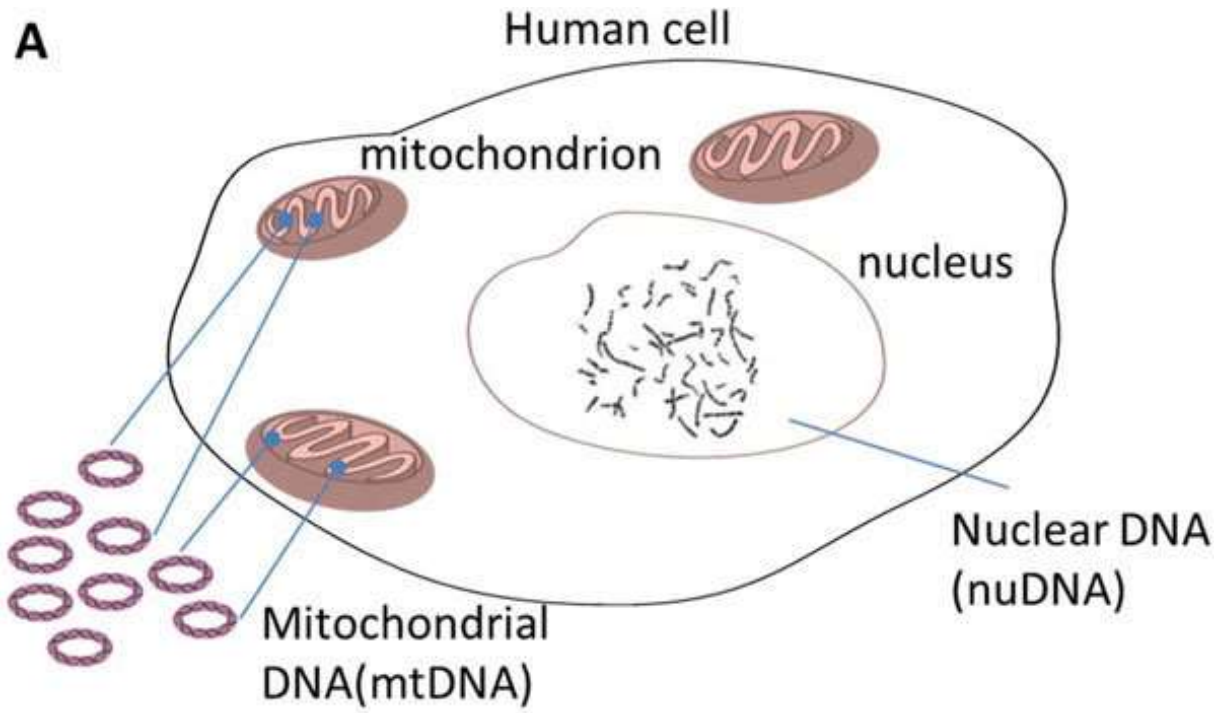
Basics of Human Genetics: DNA

- 5‘ to 3‘ direction
- Megabase (Mb) : unit of length for DNA fragments equal to 1 million nucleotides and roughly equal to 1 cm.



Basics of Human Genetics: DNA, Human Genome

- The human genome, encoded on both nuclear and mitochondrial chromosomes.



Packaging of DNA

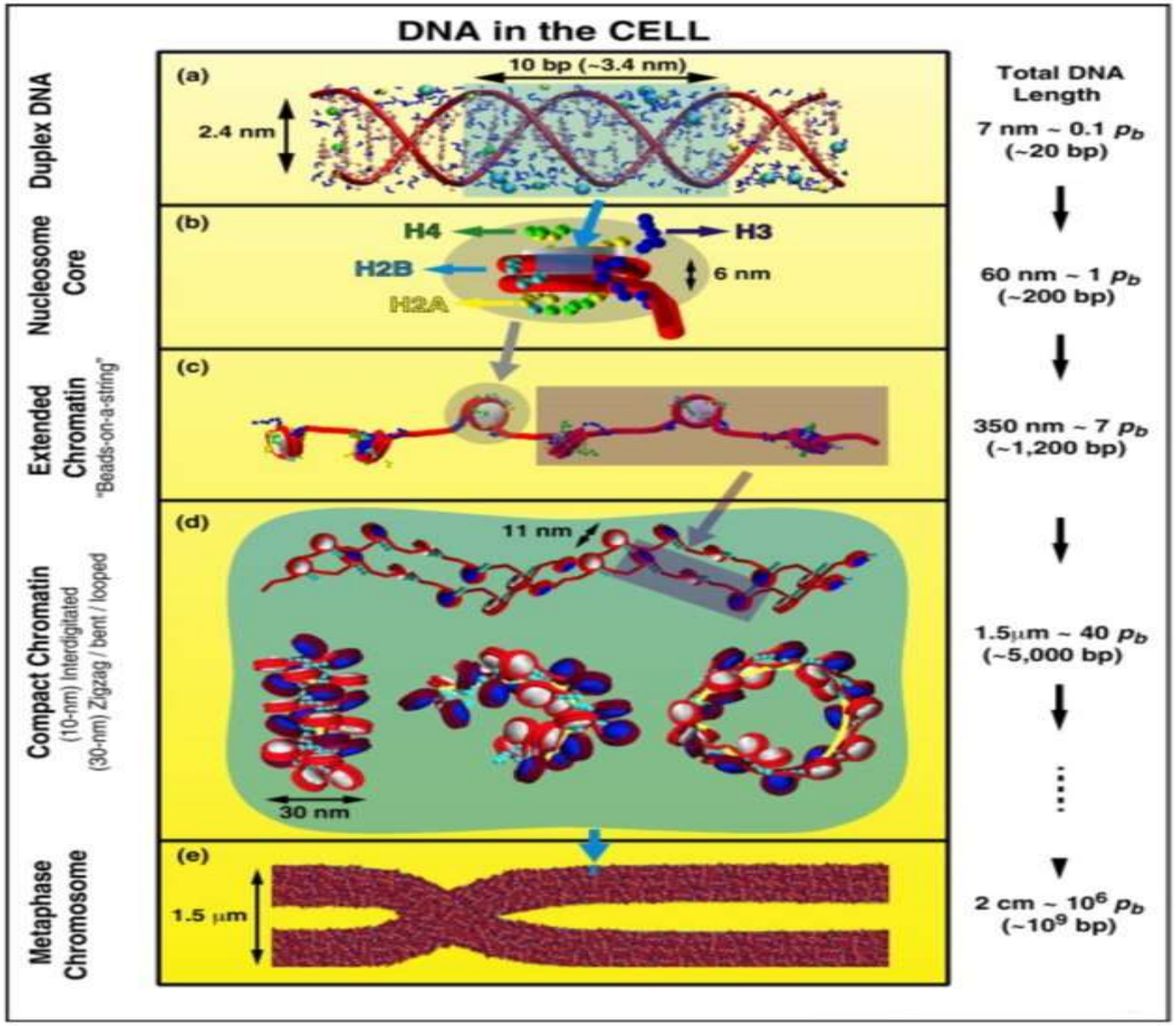
- The genome is packaged as chromatin: DNA+Proteins

  functions of these proteins ???

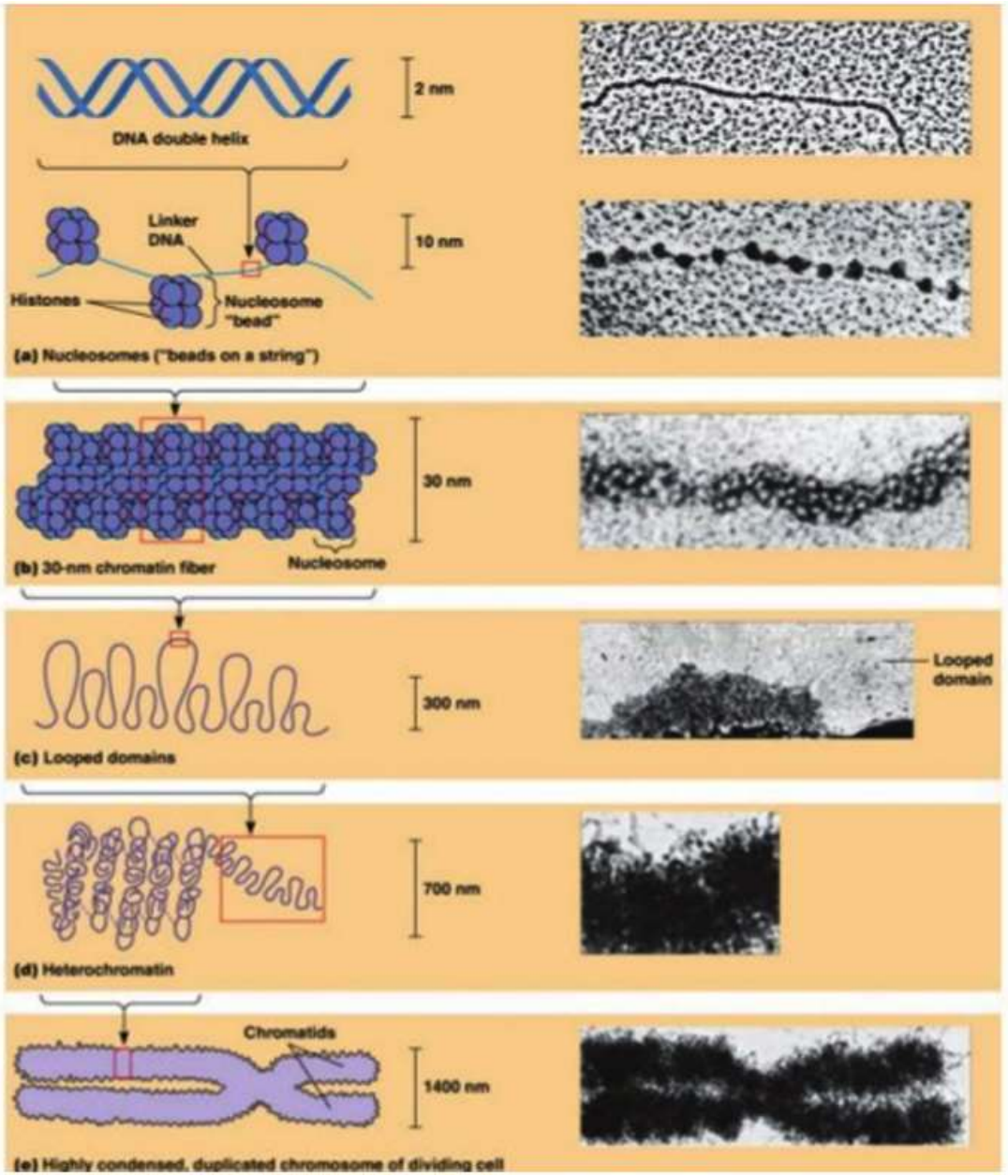
- Except during cell division, chromatin is distributed throughout the nucleus and is relatively homogeneous in appearance under the microscope.

- During cell division: chromatin condenses to appear as microscopically visible chromosomes. Chromosomes are thus visible as discrete structures only in dividing cells

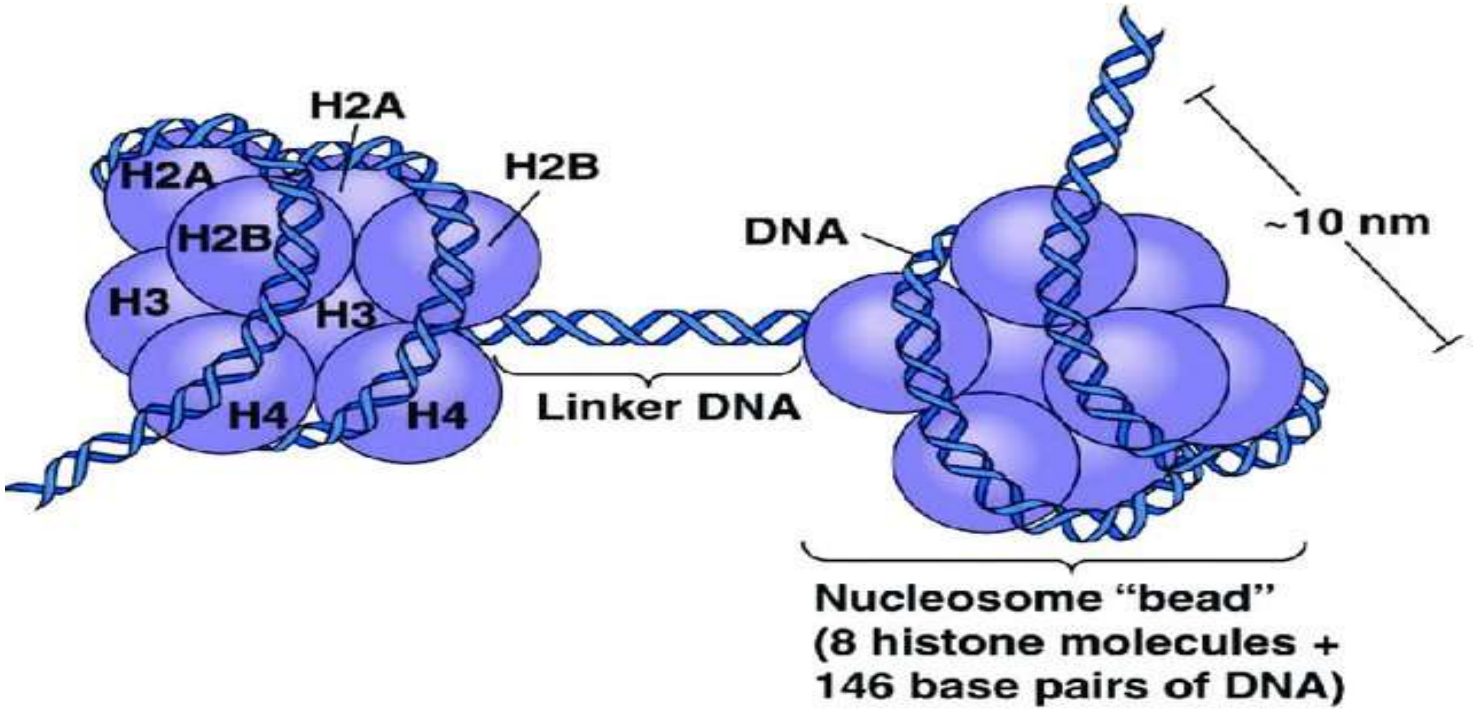
Packaging of DNA

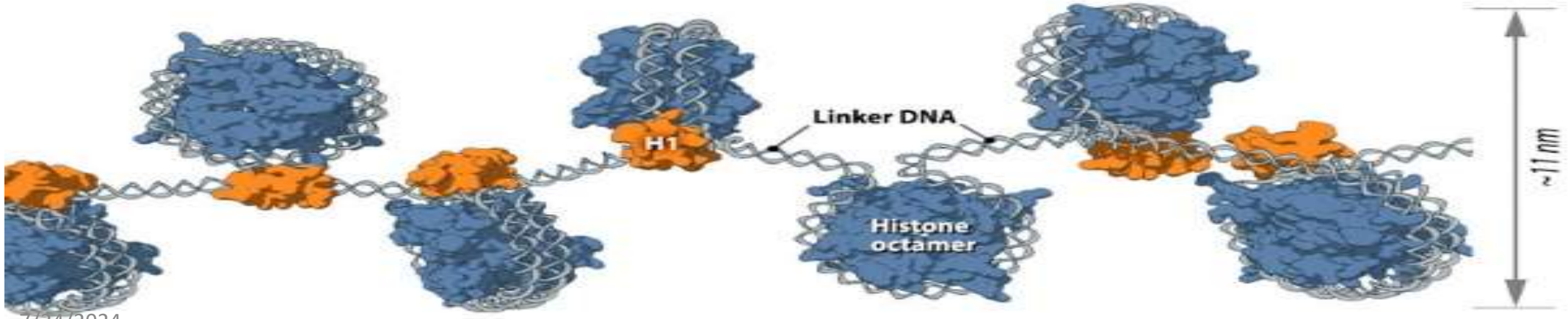


Packaging of DNA

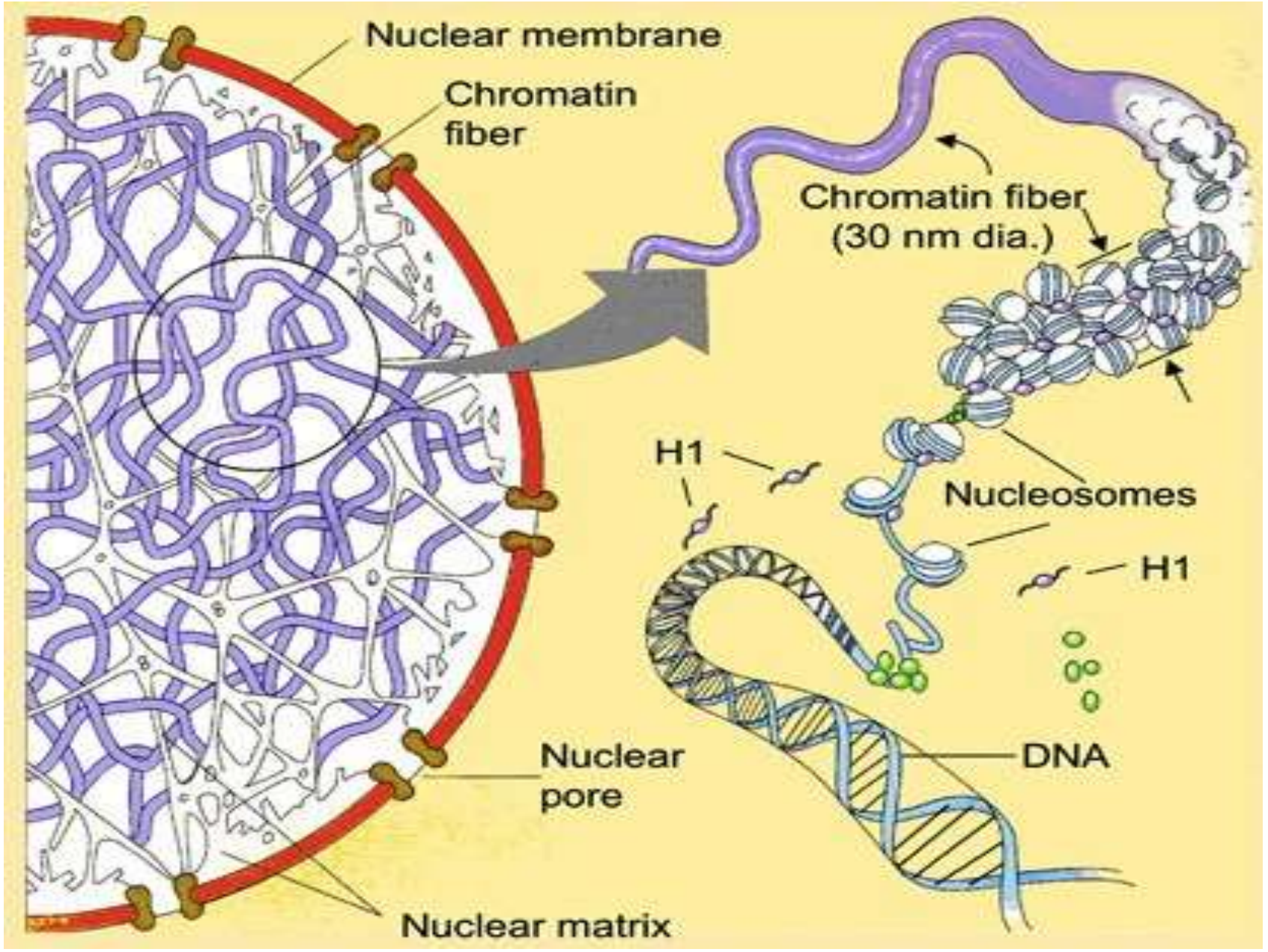


Packaging of DNA: Nucleosomes





Packaging of DNA: Nucleosomes



Chromatin

- Chromatin: Chromatin is a substance made up of DNA or RNA and proteins, such as histones. It condenses during cell division (mitosis or meiosis) and becomes a chromosome. Chromatins are “unwound” condensed structures whereas chromosomes are highly packaged and more condensed than chromatins

- There are two forms of chromatin:

(1) Euchromatin

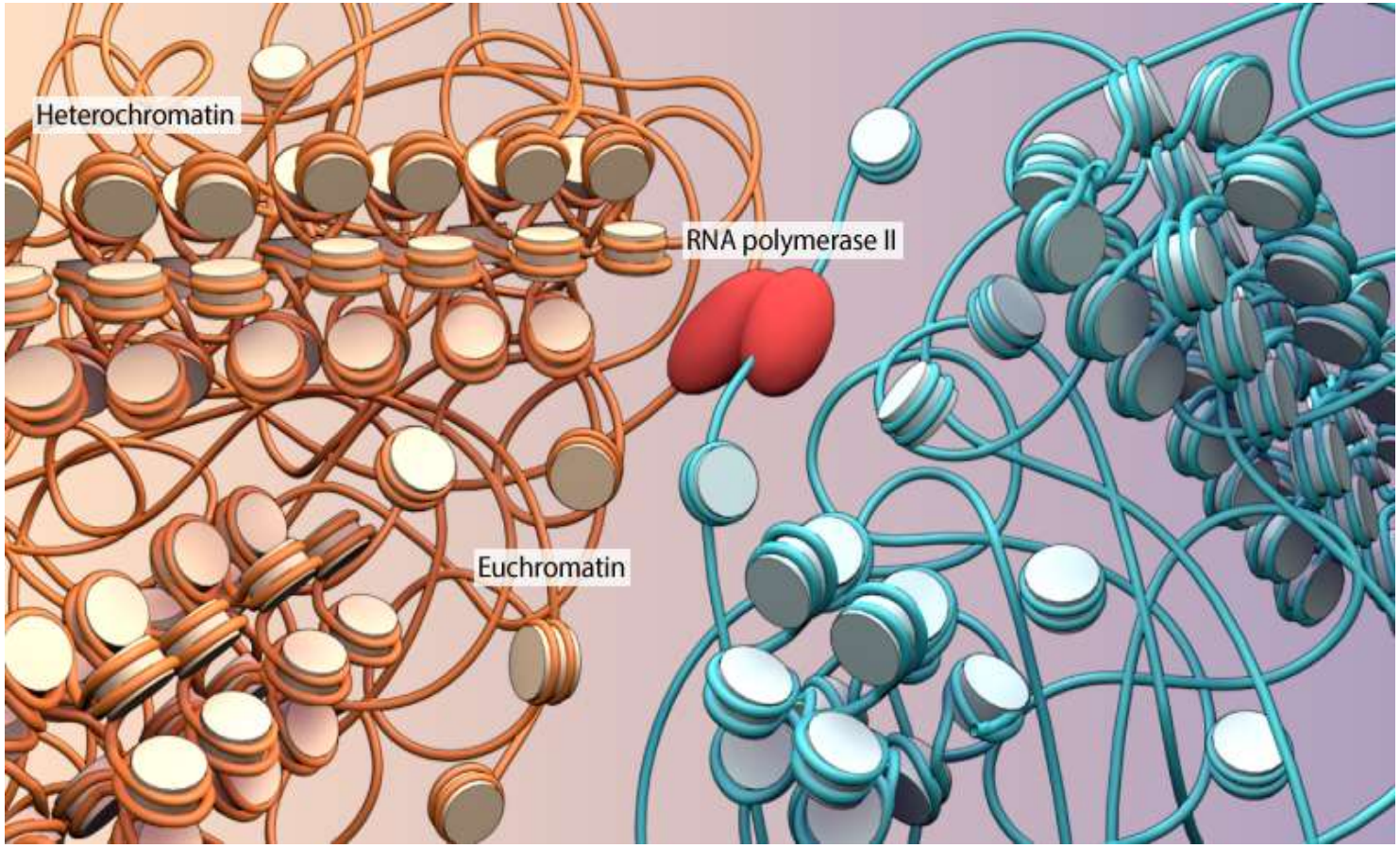
(2) Heterochromatin.

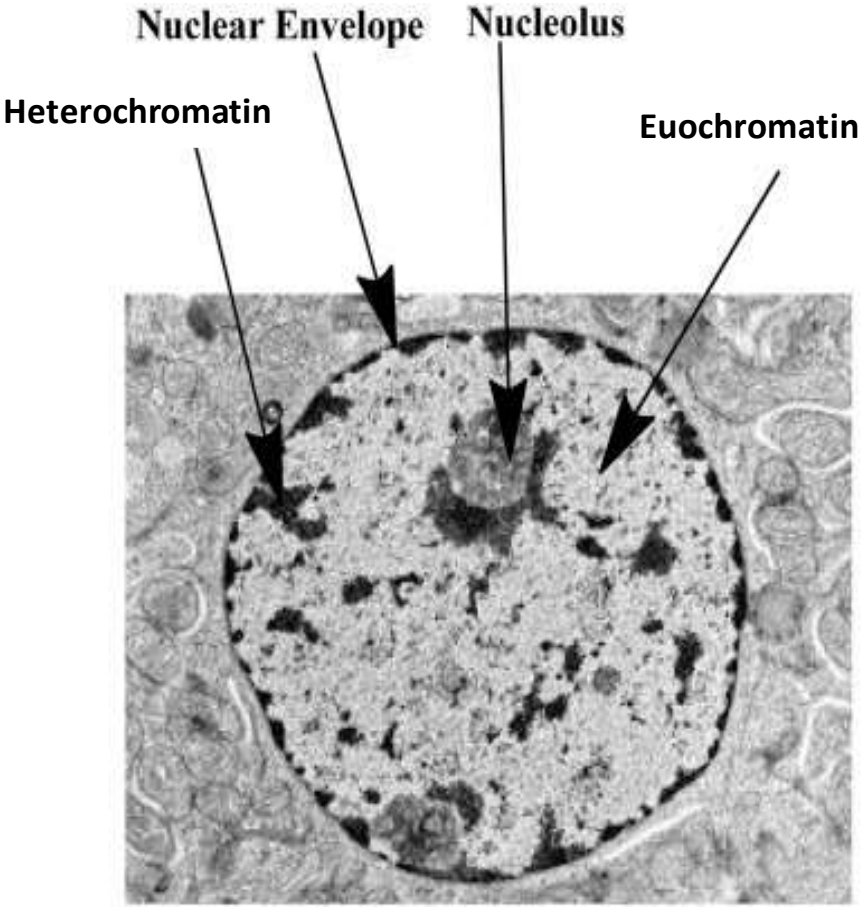
Euchromatin is less condensed in interphase and can be transcribed (active)

Heterochromatin is highly condensed in interphase and cannot typically be transcribed (not active).

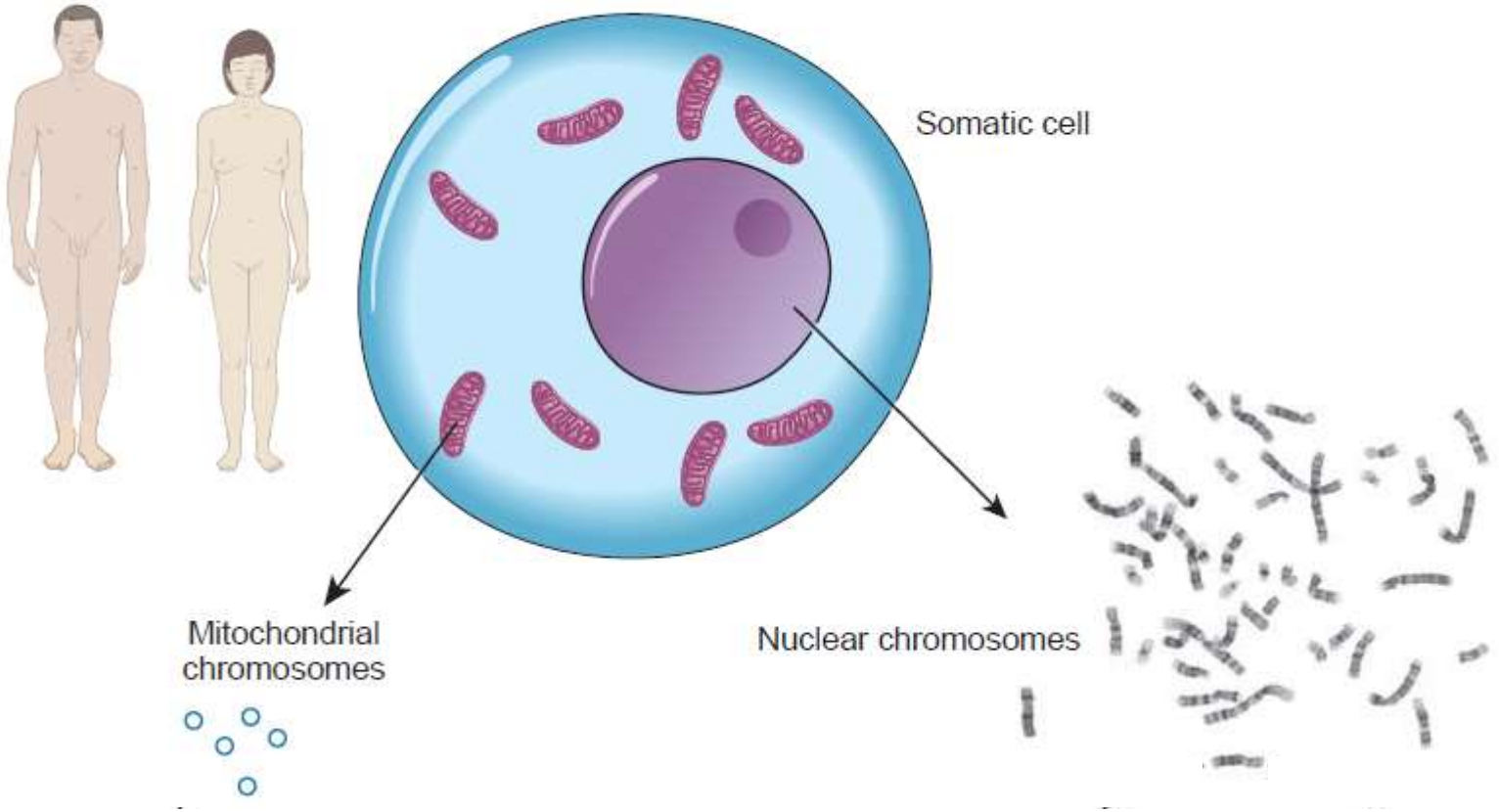
- Heterochromatin has long stretches of repeat sequences of DNA called Satellite DNA

Chromatin


Heterochromatin
RNA polymerase II
Euchromatin

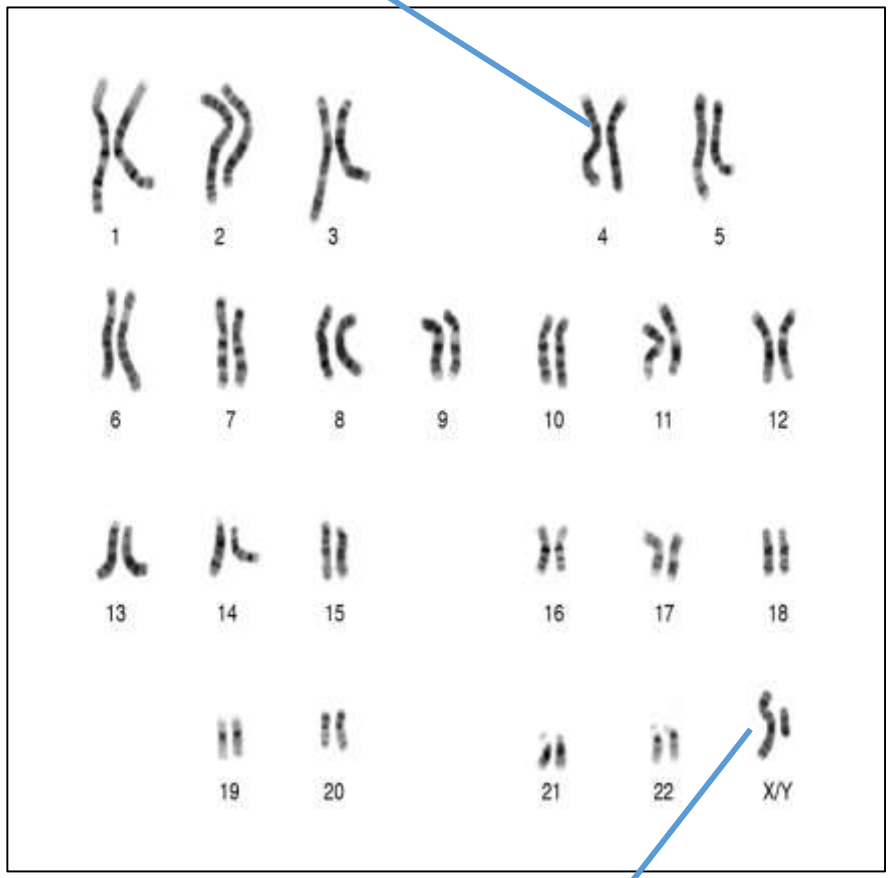

Nuclear Envelope
Nucleolus
Heterochromatin
Euochromatin

Chromosomes



Human Karyotype: 46,XY

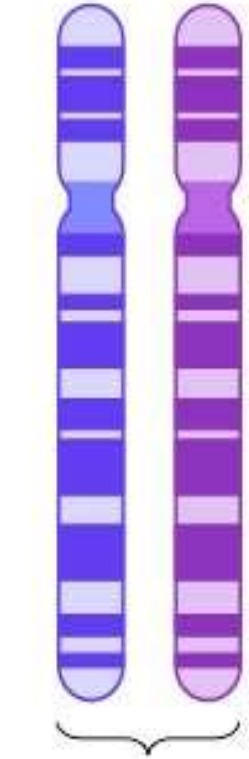
Autosomes



Sex chromosomes

Chromosomes

- 46 chromosomes (Diploid cells)
- Human cells are diploid
- 23 pairs : 22 pairs autosomes, 1 pair sex chromosomes (XY in males, XX in females)
- Members of a pair of chromosomes: homologous chromosomes or homologues: the order of genes on both chromosomes is the same
- Gene density
- Chromatid
- Locus (pleural Loci): a segment of DNA occupying a particular position or location on a chromosome



Homologous pair
(chromosome 1)

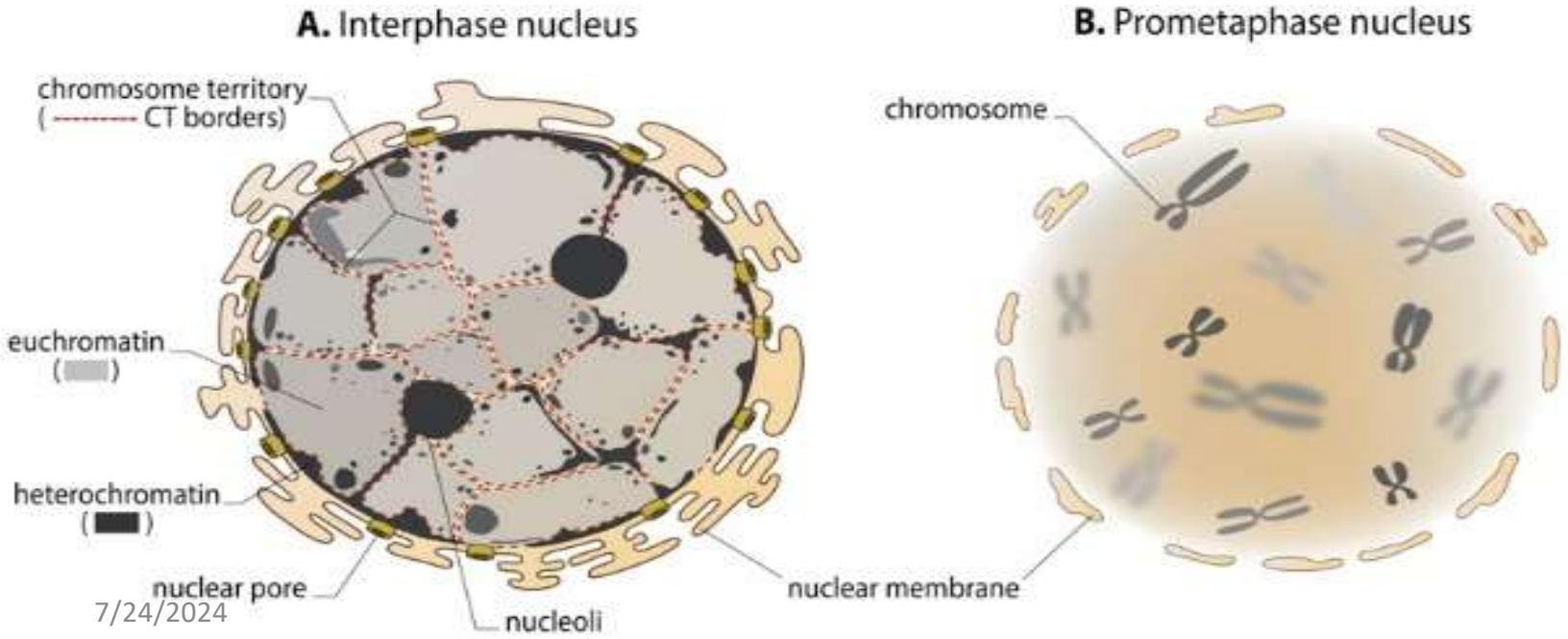
Chromosomes

- Chromosomes range in size from

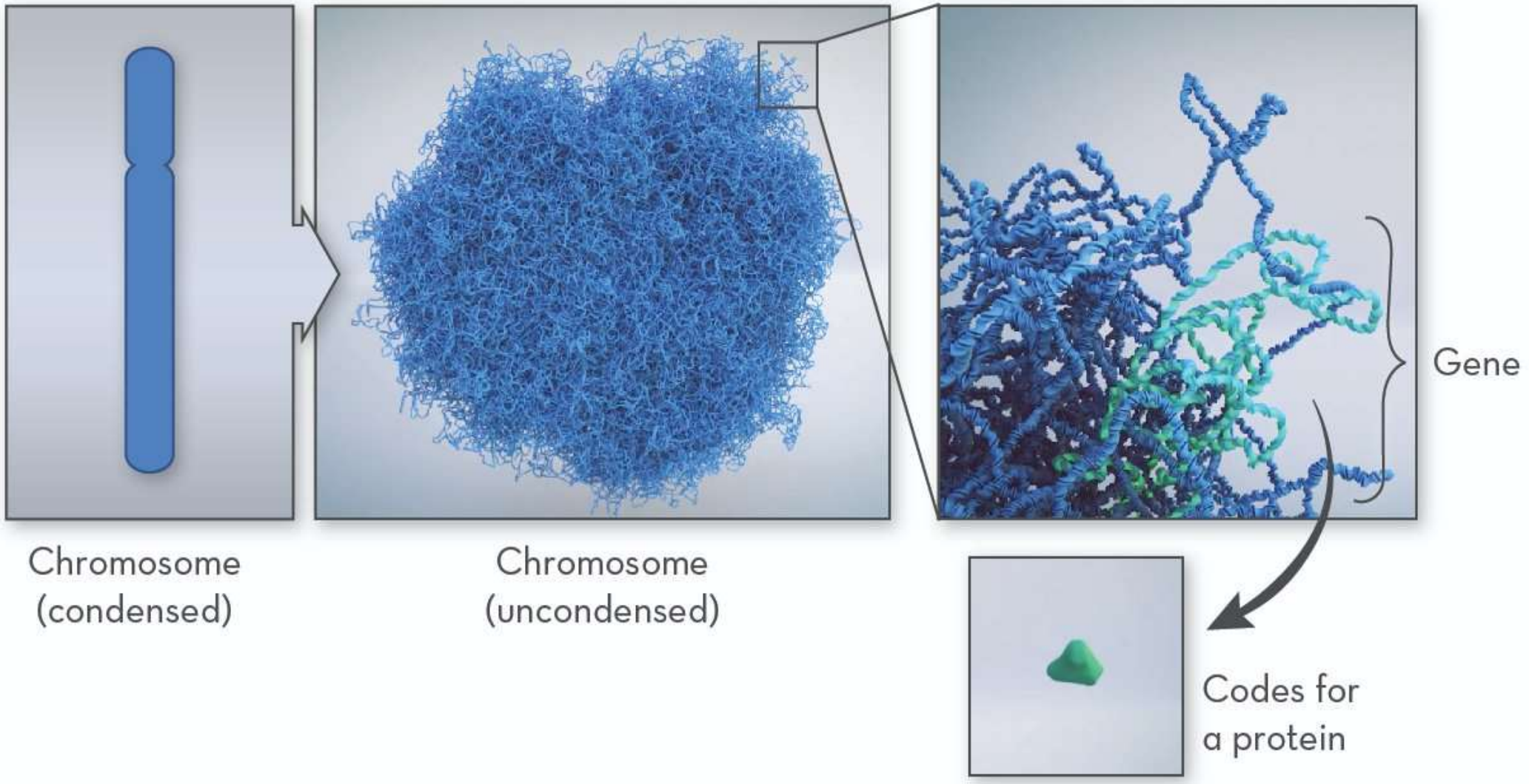
50 million to 300 million base pairs of DNA

- Visualized in metaphase of mitosis

when DNA is condensed



Chromosomes



Genes

- What is a gene?

Simply it is a sequence of DNA that encodes the information necessary to make a protein (or a functional RNA).

- Each chromosome carries a different subset of genes that are arranged linearly along its DNA. E.g. Chromosome 1 contains ~2000 gene

- Two general types of gene, those whose product is a protein and those whose product is a functional RNA.
  - Protein-coding genes : 20,000-25,000
  - RNA-coding genes (noncoding genes): 20,000-25,000

Genes

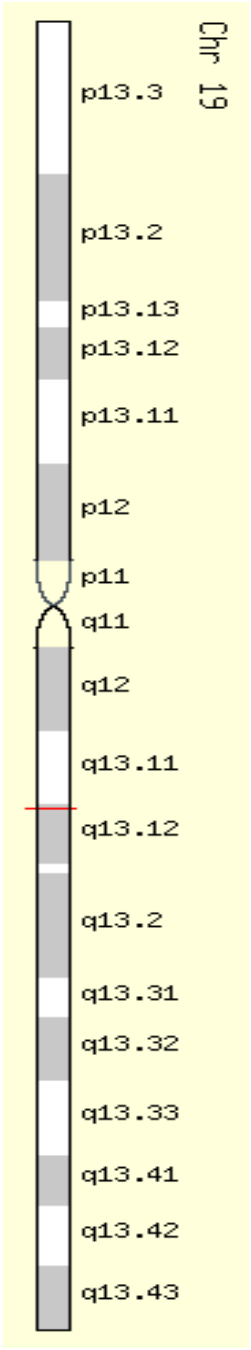
- Each gene has a specific location on one of the 24 chromosomes (gene locus)

- Alternative versions of the DNA sequence at a locus are called **alleles**.

- Genotype and Phenotype

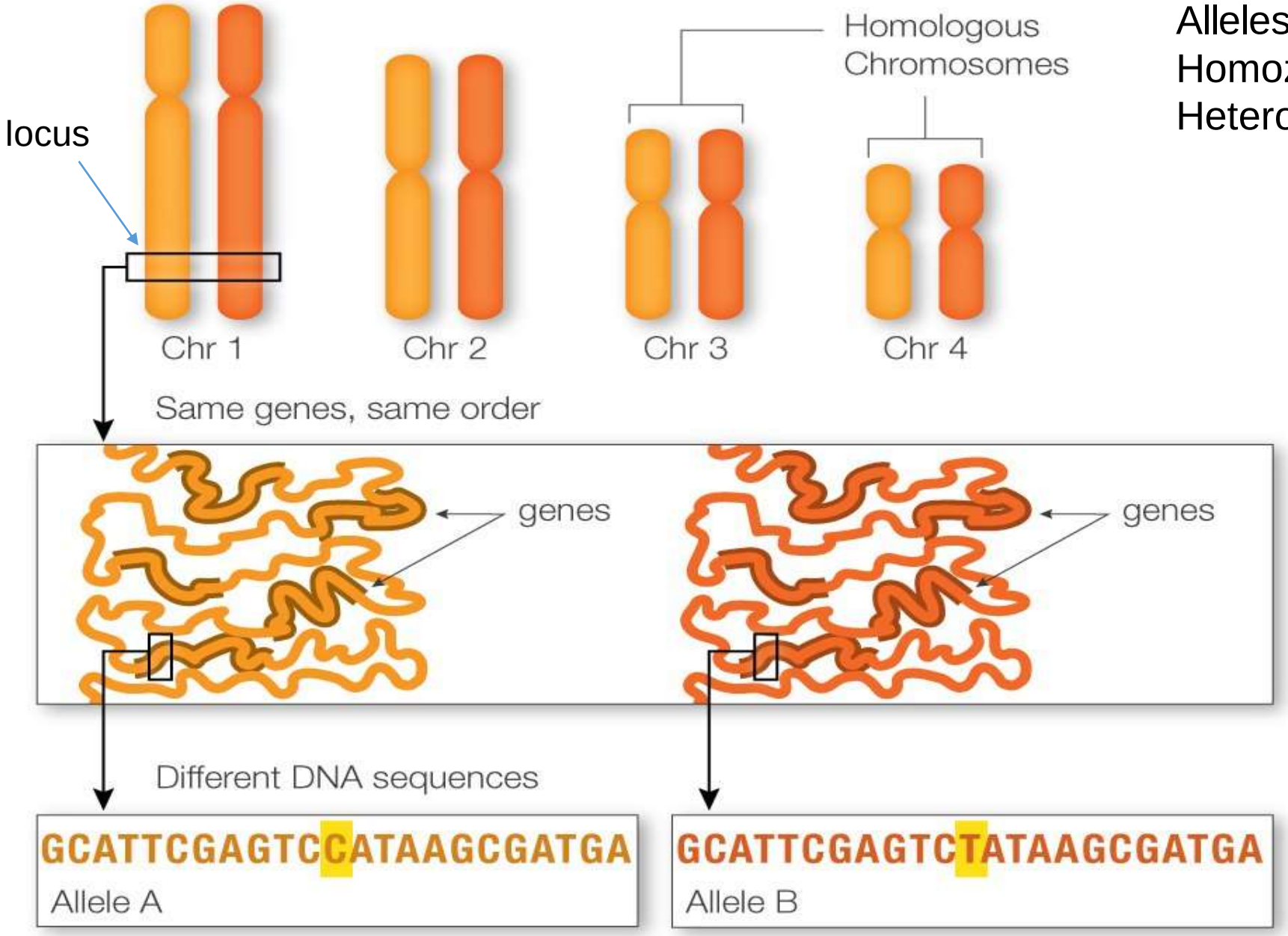
  Genotype: allele combination in an individual that causes a specific trait

  Phenotype: the trait expressed from the genotype



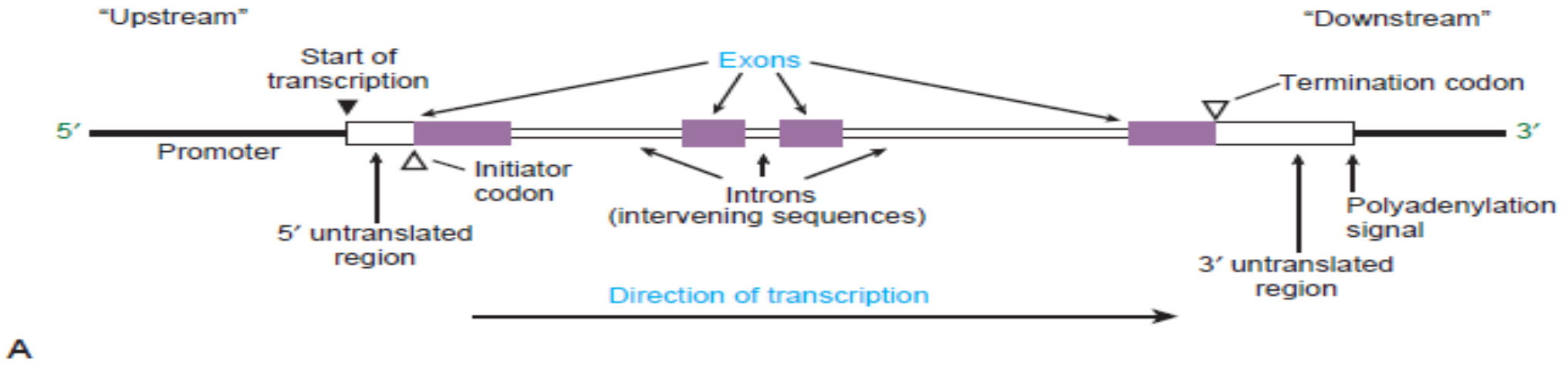
HAMP gene location: 19q13.12

Genes



Alleles :
Homozygous alleles
Heterozygous alleles

Gene Organization and Structure



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