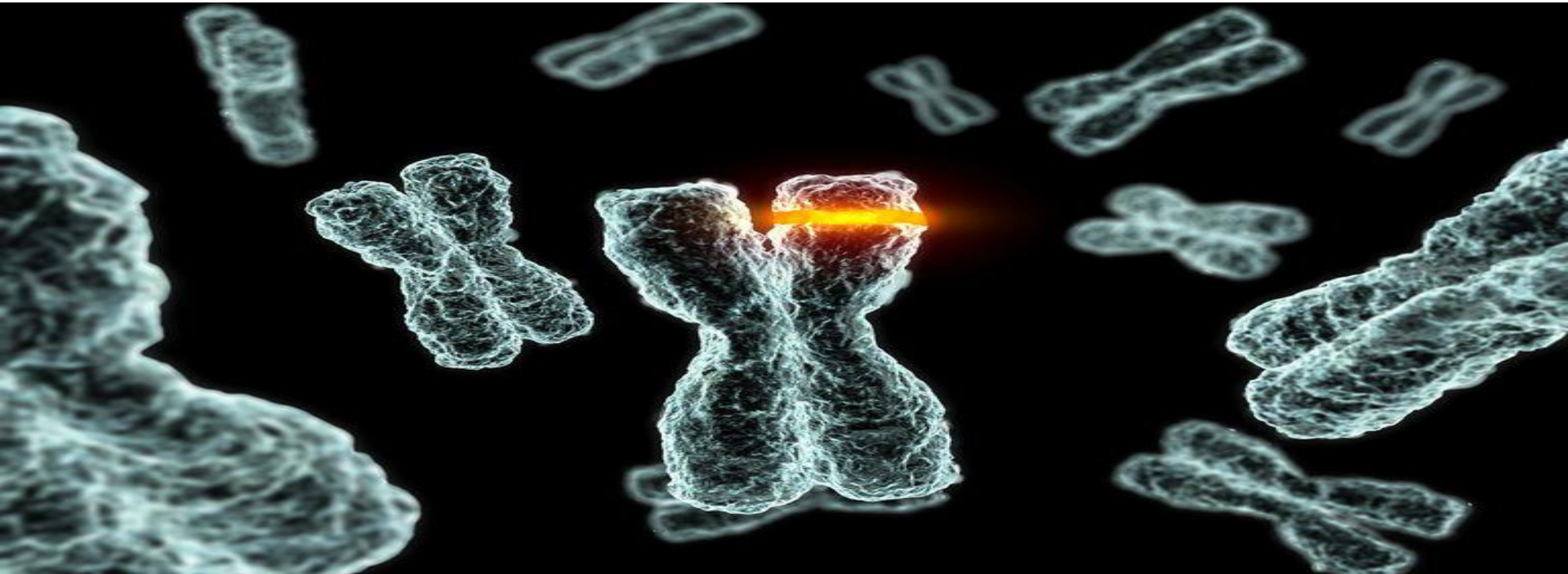
جامعة الملك سعود
King Saud University

Principles of Genetics (Zoo-352)

Lecture 2 Chromosomes

Department of Zoology, 1438-1439 H

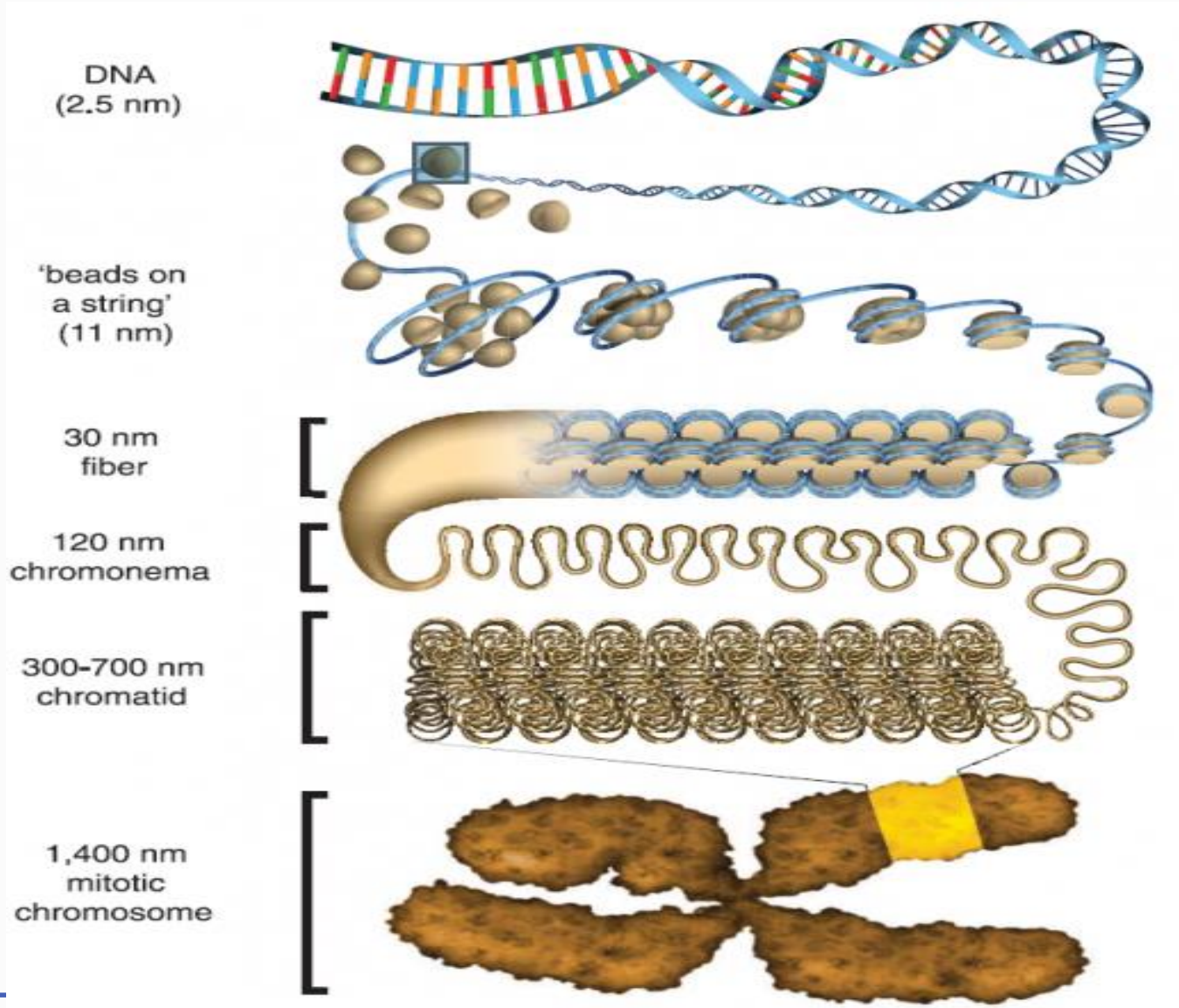


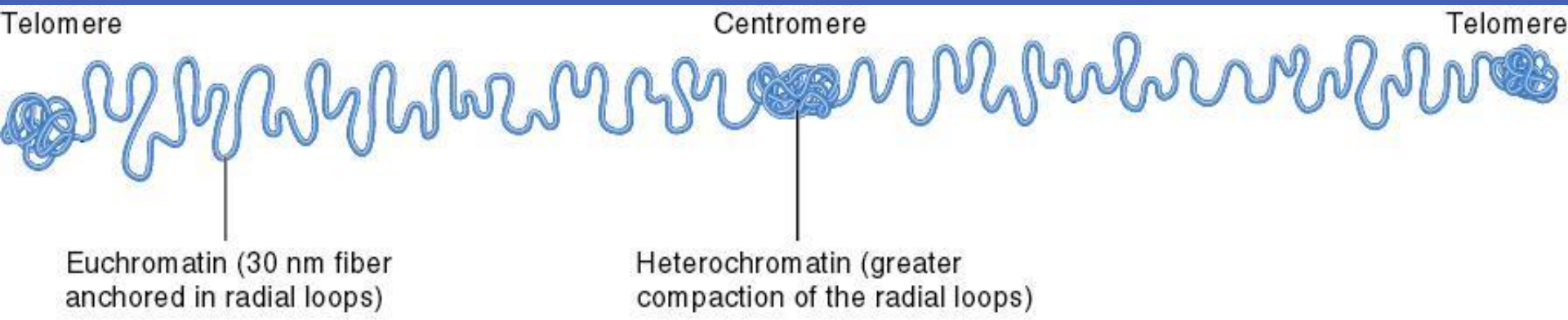
Chromosomes:

- The term chromosome means the colored body.
- Linear eukaryotic chromosomes are composed of a complex of double-stranded DNA and protein, which is referred to collectively as chromatin.
- Chromatin can be found in either:
  - 1.a less condense state termed **euchromatin** or
  - 2.a condensed and readily visible organization termed heterochromatin.

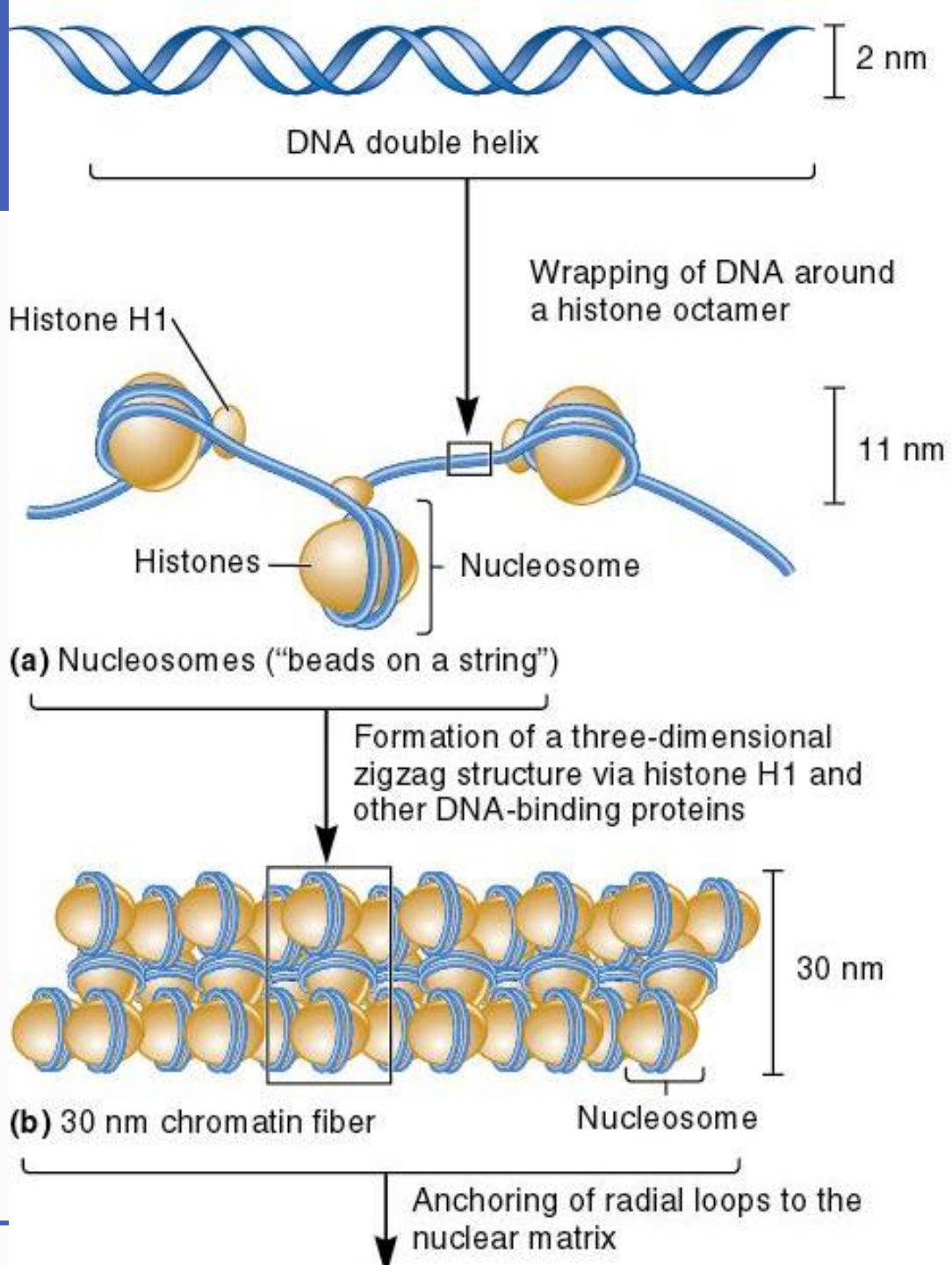
Heterochromatin *vs* Euchromatin

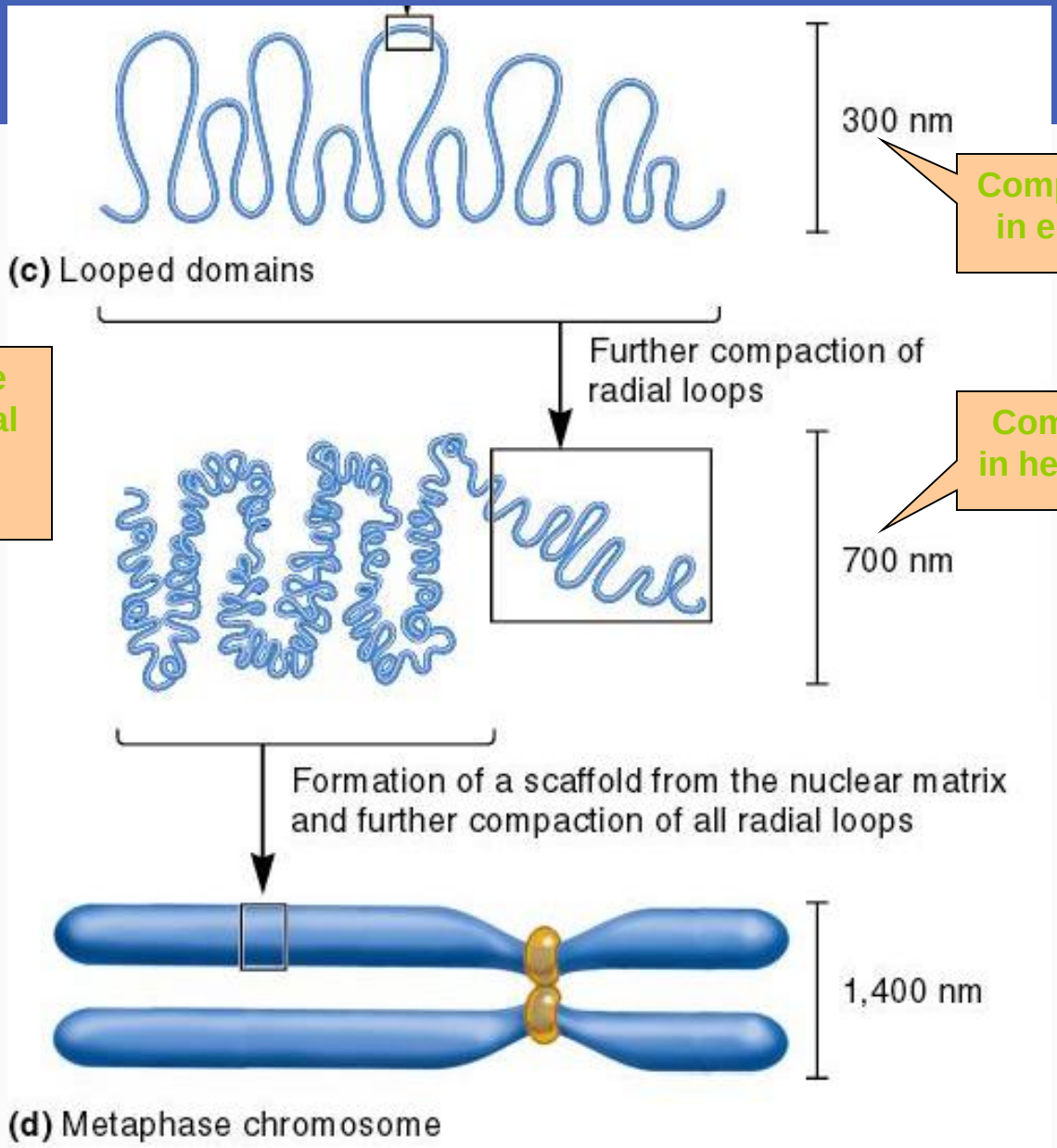
- The compaction level of interphase chromosomes is not completely uniform
  - Euchromatin
    - Less condensed regions of chromosomes
    - Transcriptionally active
    - Regions where 30 nm fiber forms radial loop domains
  - Heterochromatin
    - Tightly compacted regions of chromosomes
    - Transcriptionally inactive (in general)
    - Radial loop domains compacted even further


DNA
(2.5 nm)
'beads on
a string'
(11 nm)
30 nm
fiber
120 nm
chromonema
300-700 nm
chromatid
1,400 nm
mitotic
chromosome



- There are two types of heterochromatin
  - Constitutive heterochromatin
    - Regions that are always heterochromatic
    - Permanently inactive with regard to transcription
  - Facultative heterochromatin
    - Regions that can interconvert between euchromatin and heterochromatin
    - Example: **Barr body**


2 nm
DNA double helix
Wrapping of DNA around a histone octamer
Histone H1
11 nm
Histones
Nucleosome
(a) Nucleosomes ("beads on a string")
Formation of a three-dimensional zigzag structure via histone H1 and other DNA-binding proteins
30 nm
(b) 30 nm chromatin fiber
Nucleosome
Anchoring of radial loops to the nuclear matrix



Compaction level in euchromatin

During interphase most chromosomal regions are euchromatic

Compaction level in heterochromatin

Classification of chromosomes:

- Chromosomes are classified by the location of their **spindle attachment point**, which have distinct positions.
- The attachment point occurs at a construction in the chromosome termed the centromere (figure 1).
- Centromere is composed of several specific DNA sequences.
- The kinetochore is the proteinaceous structure on the surface of the centromere to which the spindle microtubules attach.
- The centromere and the kinetochore are structural units that are essential for mitosis and meiosis.
- Chromosomes can be classified according to the location of the centromere as bellow:
- **1.Metacentric chromosome:** The centromere is in the middle of the chromosome (Figure 2).

Classification of chromosomes

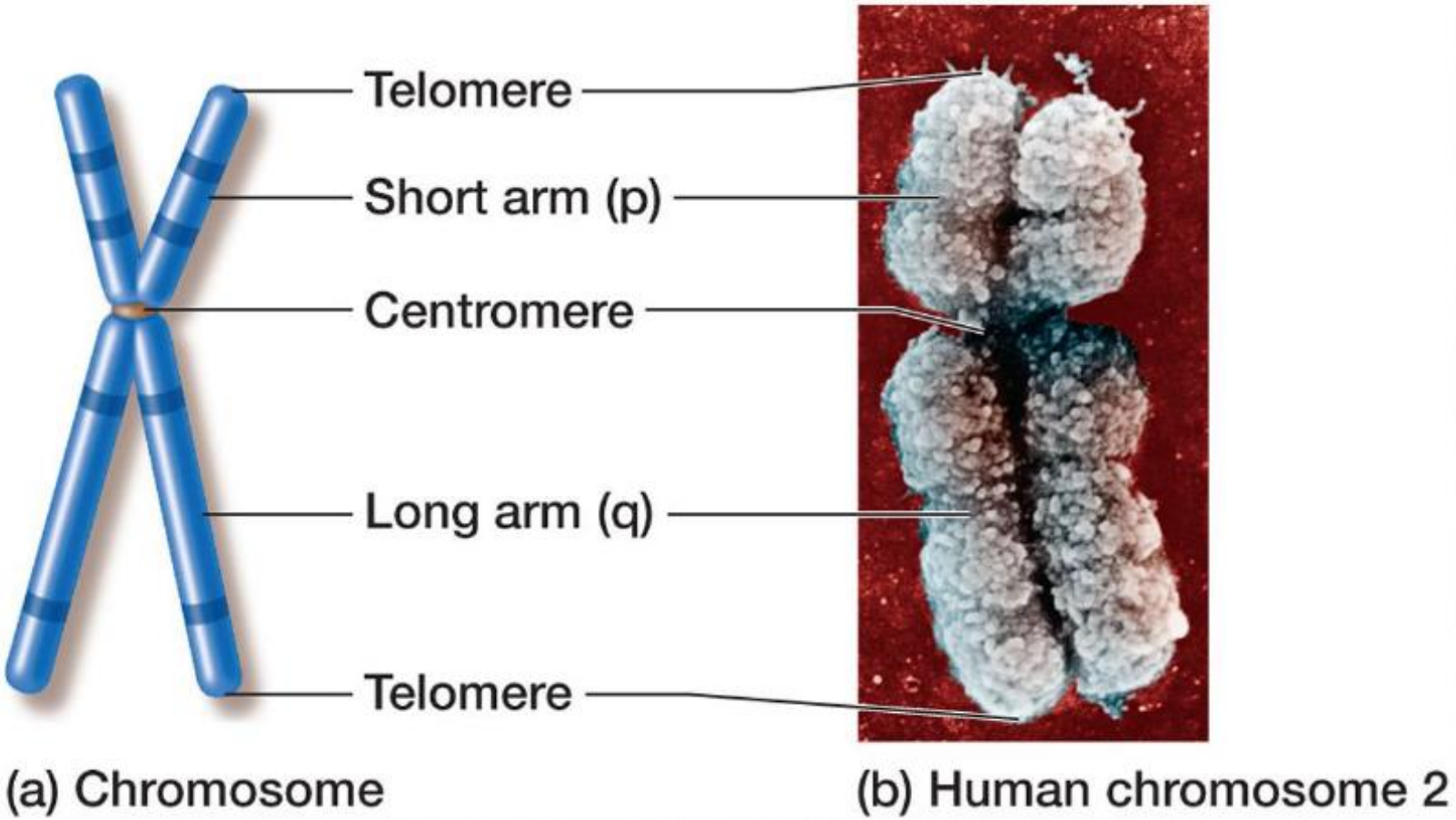
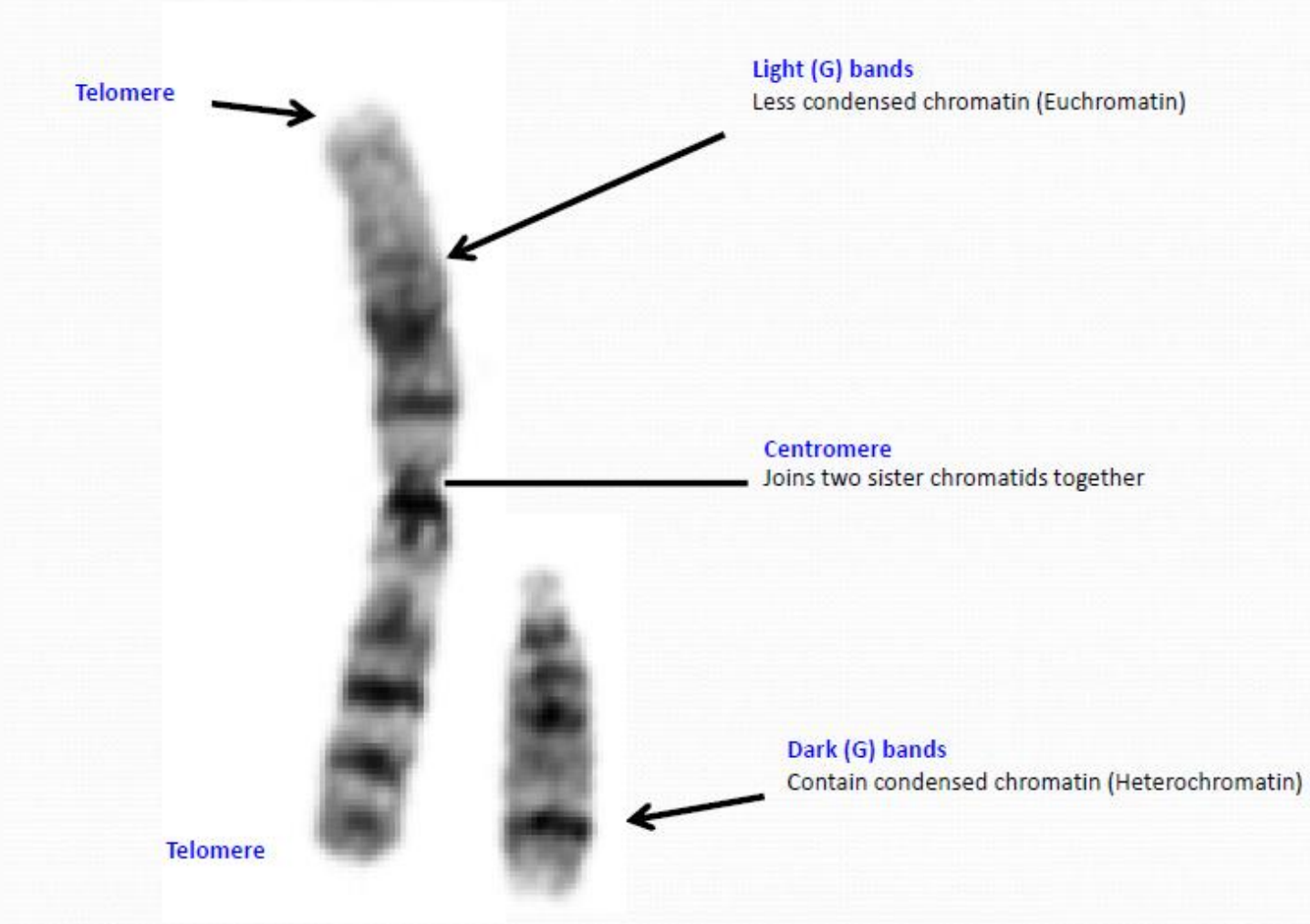


Figure 1: Schematic of Submetacentric chromosome (a) and an electron micrograph of human chromosome 2 (b).

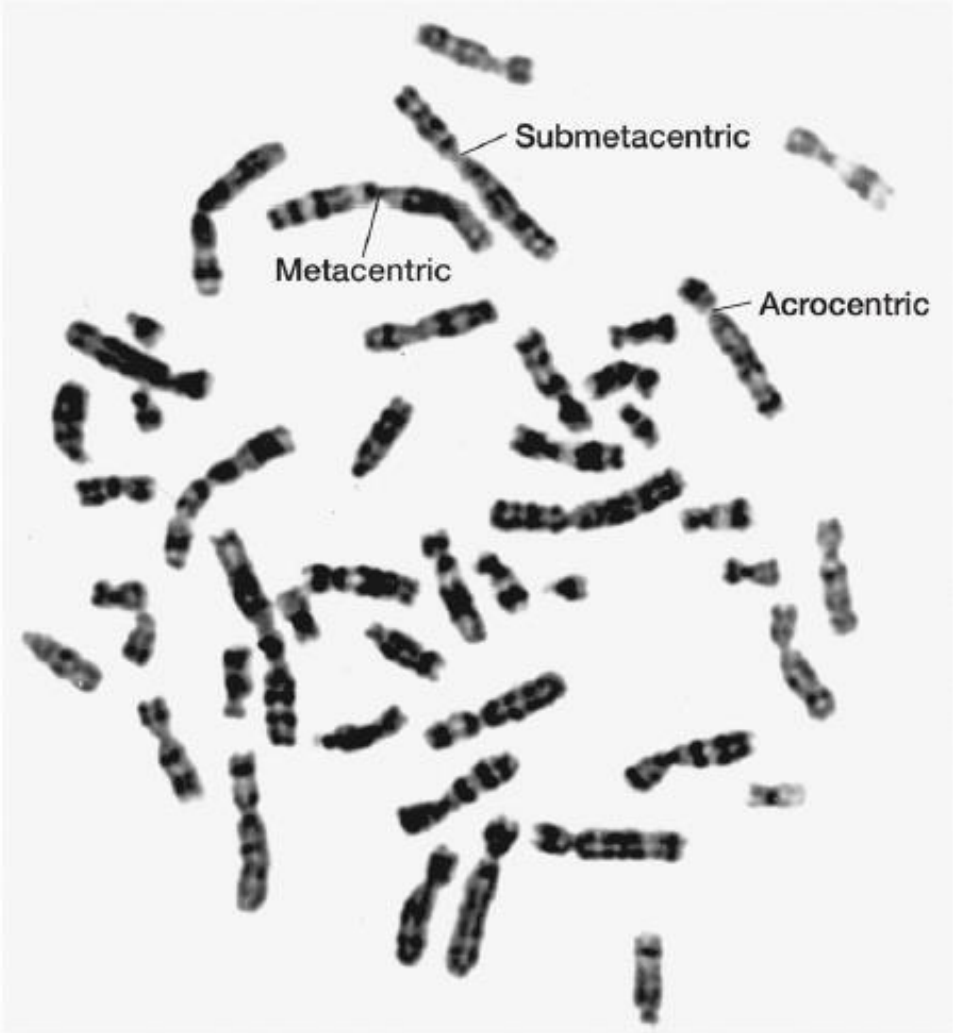
Classification of chromosomes

- **2. Submetacentric chromosome:** The centromere is located between the middle and the end of the chromosome (Figures 1 and 2).
- **3. Telocentric chromosome:** The centromere is located at the end of the chromosome.
- **4. Acrocentric chromosome:** The centromere is near to the end of the chromosome (Figure 2).
- The location of the centromere often divides the chromosome into two parts that are referred to as the short arm (**p arm** for petite) and the long arm (**q arm**).
- A **telomere** is a region of repetitive nucleotide sequences at each end of a chromatid, which protects the end of the chromosome from degradation or from fusion with neighboring chromosomes.

Classification of chromosomes



Classification of chromosomes



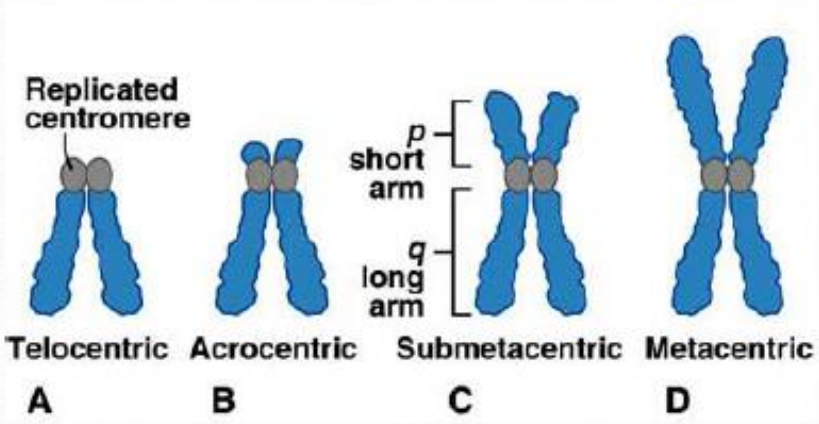


Figure 2: Metacentric, Submetacentric, and acrocentric chromosomes

Chromosome complement:

- Most cells of eukaryotic organisms are **diploid**; that contain two sets of chromosomes. In the diploid state, members of the same chromosome pair are referred to as **homologous chromosome**, or homologs. One member of each pair comes from each parent.
- Humans have **23 homologous** chromosome pairs, which is often expressed as **2n=46**. This expression indicate that humans are diploid (2n) and have a total of 46 chromosomes. The diploid chromosome numbers of several species appear in table 1.
- **Haploid cells**, which include some eukaryotic organisms and the reproductive cells (**gametes**), have only one set of chromosomes.
- In humans, Chromosome pair 1-22 are called **autosomes**, determine body trait. The 23rd pair is called **sex chromosomes** (XX is female, XY is male), determine sex.
- Genes are arranged in the linear order on chromosome (Figure 3).

Species	2*n*
Human being (*Homo sapiens*)	46
Garden pea (*Pisum sativum*)	14
Fruit fly (*Drosophila melanogaster*)	8
House mouse (*Mus musculus*)	40
Roundworm (*Ascaris* sp.)	2
Pigeon (*Columba livia*)	80

Table 1: Chromosome number for selected species

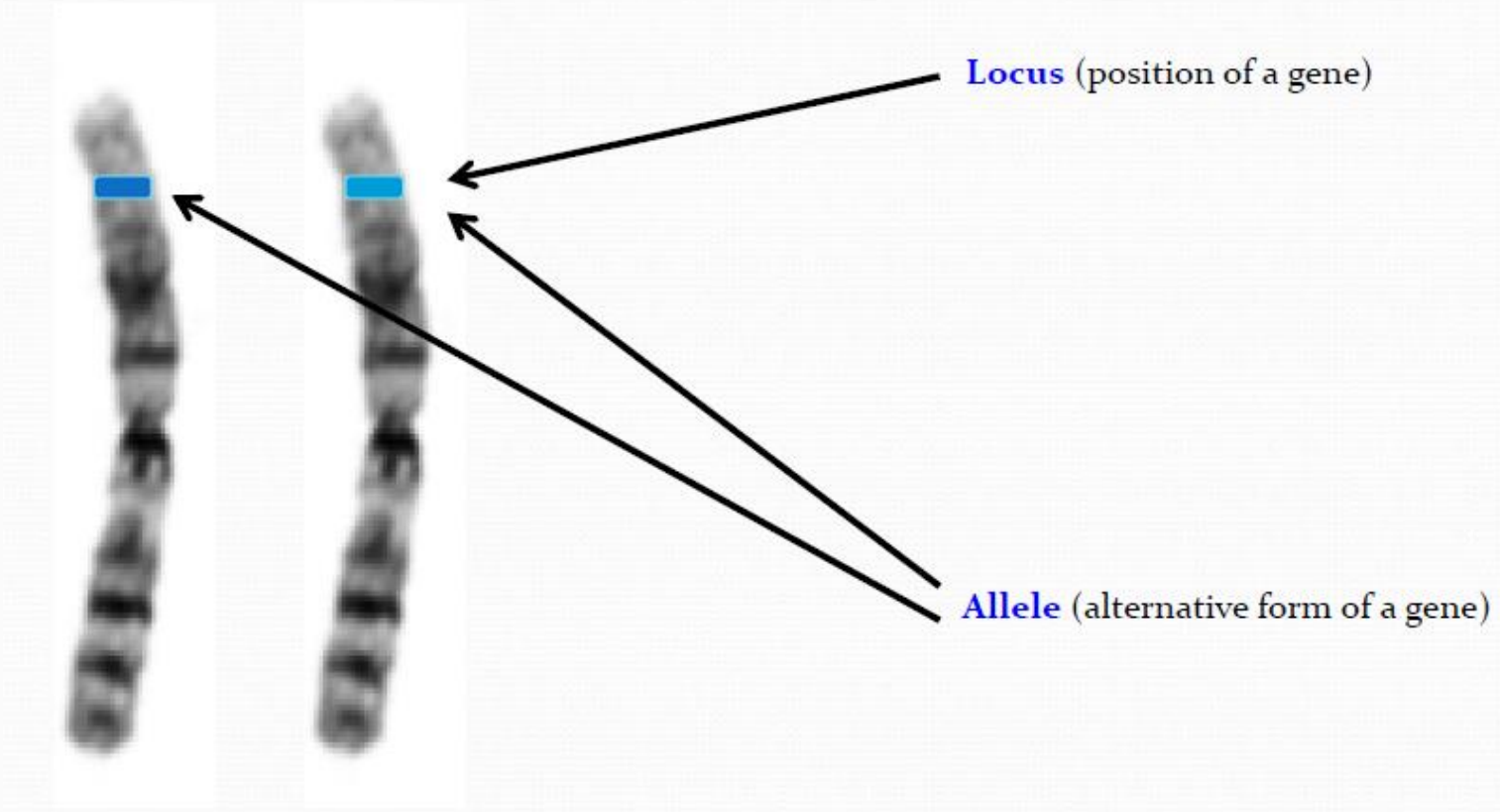


Figure 3: A pair of homologous chromosome 1 in humans

karyotype

- The total human's chromosomes can be photographed during mitosis and rearranged in pairs to make a picture called a karyotype.
- From karyotype, it is possible to see whether the chromosome have any abnormalities and to identify the sex of the individual.
- The chromosome in humans are grouped into categories (A-G, X, Y) based on (1) their length (size), (2) centromere position (location) and (3) the pattern of dark and light G (Giemsa) bands (Figure 4).

karyotype

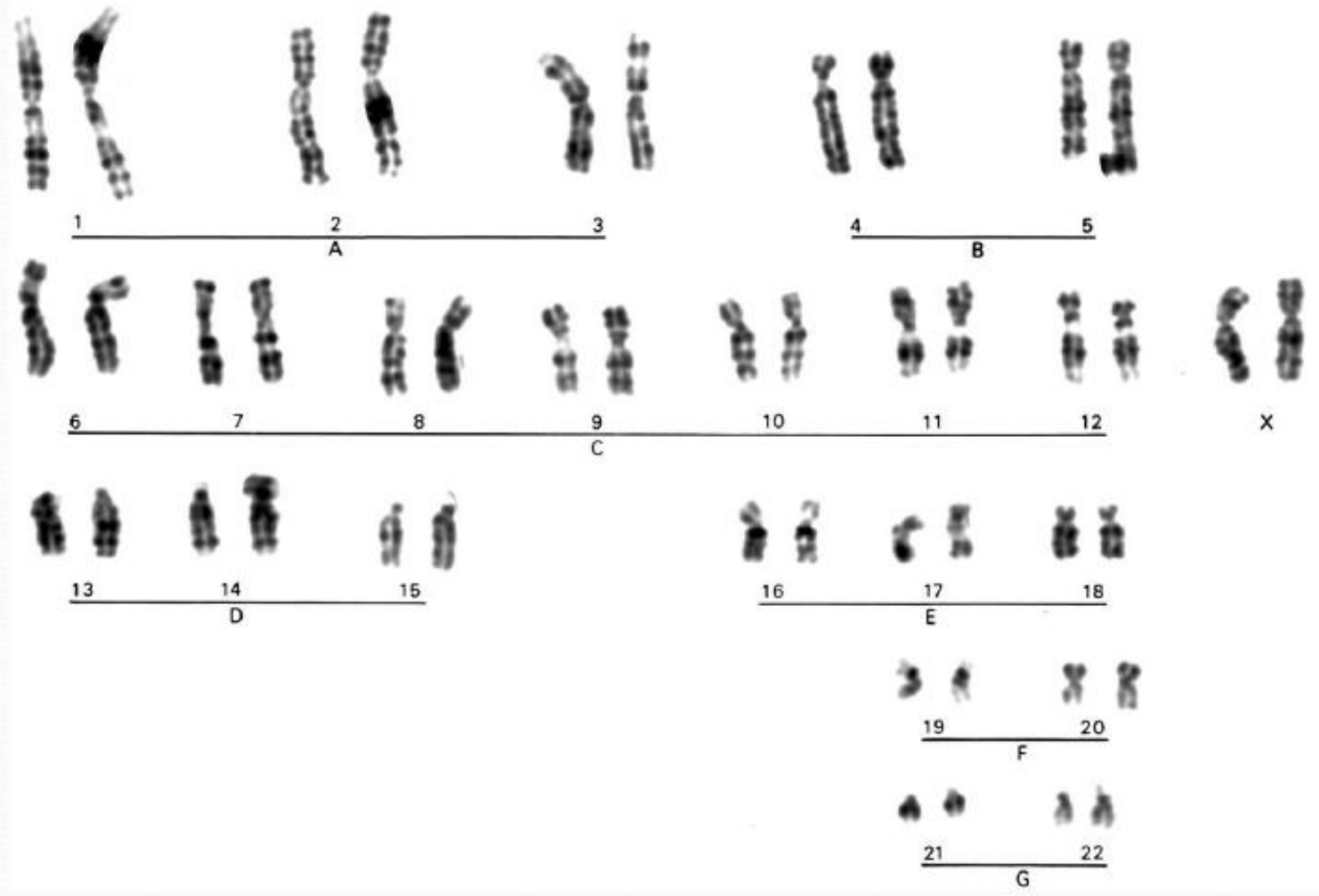


Figure 4: Karyotype of a human female (two X chromosomes, no Y chromosome)

karyotype

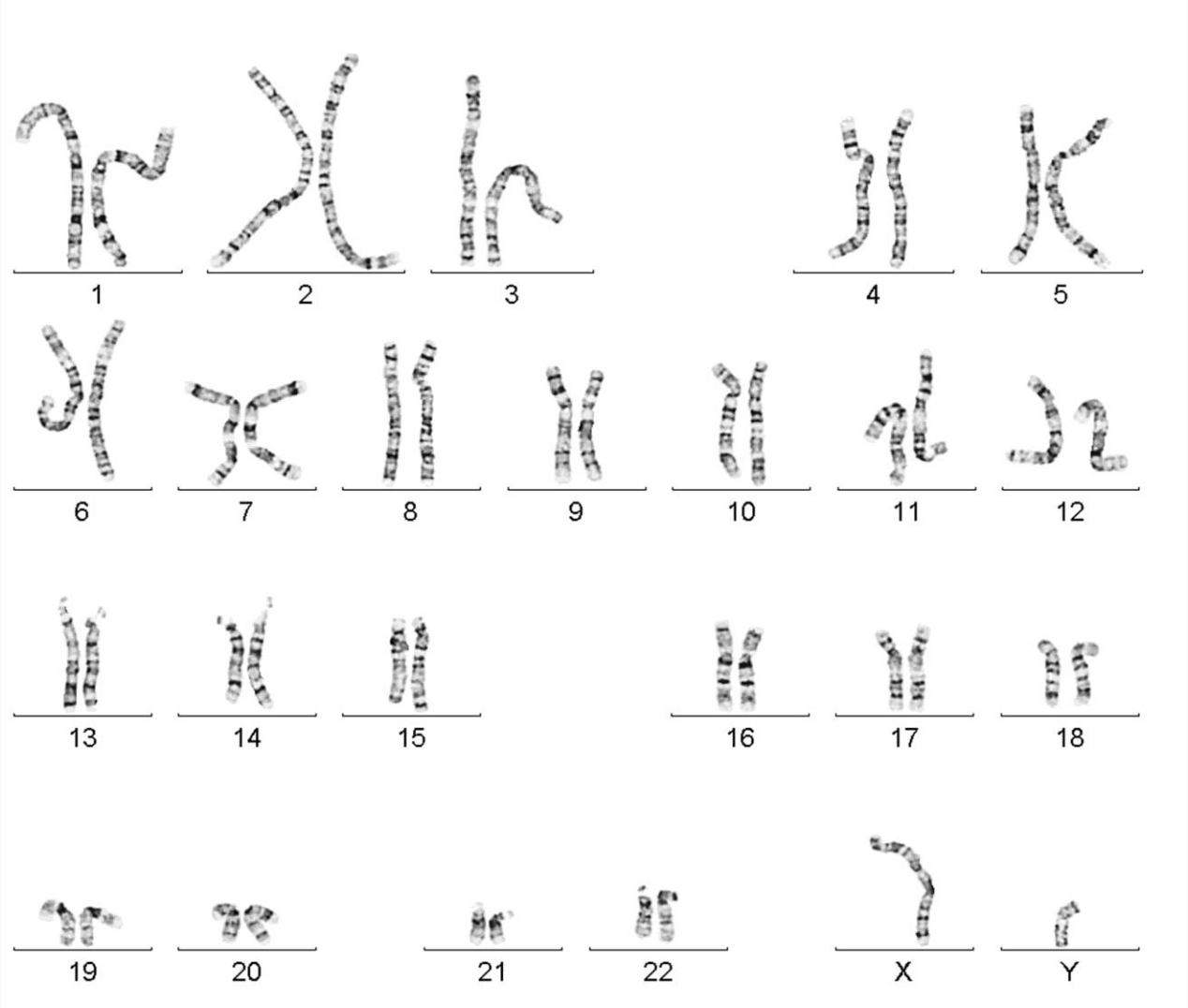


Figure 4: Karyotype of a human male