Introduced to Chromosome

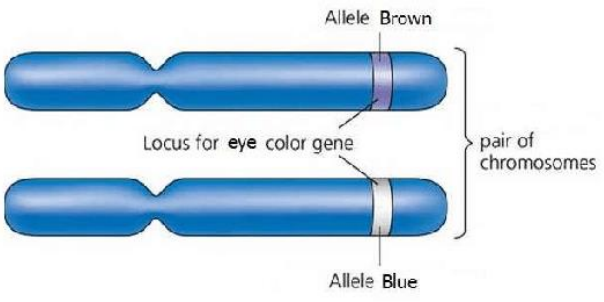
- A **chromosome** is a single DNA molecule together with other molecules (proteins and RNA) needed to support and read the DNA.
- The word **chromosome** comes from the Greek (chroma, **"colour"**) and (soma, **"body"**), describing their strong staining by particular dyes.

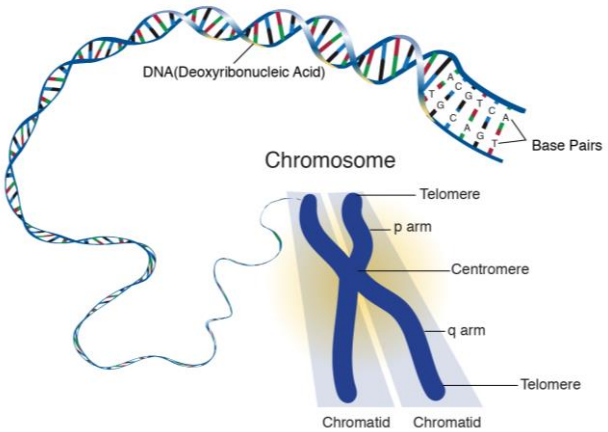
- The term **chromosome** was coined by the German anatomist **Heinrich Wilhelm Waldeyer**, referring to the term **chromatin**, which was introduced by **Walther Flemming**, the discoverer of cell division.

- Some of the **early karyological** terms have become outdated. For example, **Chromatin** (Flemming 1880) and **Chromosome** (Waldeyer 1888), both ascribe color to a non-colored state.
- Chromosome: The genetic material of the cell, complexes with protein and organized into a number of linear structures. It literally **means "colored body"** because the threadlike structures are visible under the microscope only after they are attained with dyes.
- **Chromatin:** The piece of **DNA-protein complex** that is studied and analyzed. Each chromatin fragment reflects the general features of chromosomes **but not** the specifics of any individual chromosome.
- The term ***gene*** is a word that **Mendel** never knew. It was not coined until 1909, when Danish geneticist **Wilhelm Johannsen** first used it. Additionally, he is best known for coining the terms **phenotype** and **genotype**, and for his 1903 **"pure line" experiments** in genetics.

- The definition of gene varies with the context of its use, so its definition will change as we explore different aspects of heredity. For our present use in the context of genetic crosses, we define a gene as an inherited factor that determines a characteristic.
- ***Genes*** are often described as units of inheritance (inherited factor) that helps determine a **trait (the appearance or manifestation of a characteristic)**; but **what exactly is a gene**?
- At the molecular level **A gene** is as a DNA sequence of nucleotides that provides cells with the instructions to synthesize a specific protein or a particular type of RNA.

- **Genes** reside at specific locations, known as **loci** (plural) or **locus** (singular), on a particular chromosome.
- The form of a **gene** at a given **locus** is an allele; thus, each **locus** has **two alleles** (**one per chromosome**).
- Genes frequently come in different versions called allele. These gene variants still code for the same trait (*i.e.*, hair color), but they differ in how the trait is expressed (*i.e.*, brown vs blonde hair). Different versions of the same gene are called alleles.
- The ***genotype*** is the set of **alleles** that an individual organism possesses. A **diploid** organism with a ***genotype*** consisting of **two identical alleles** is ***homozygous*** at that locus. One that has a ***genotype*** consisting of **two different alleles** is ***heterozygous*** at the locus.
- **Heterozygous:** Having two different alleles at a locus.
- **Homozygous:** Having two identical alleles at a locus.
- **Genotype:** The set of alleles possessed by an individual organism and/or (The complete genetic makeup of an organism.
- A **phenotype** is the manifestation or appearance of a characteristic (The observable properties of an organism (that are produced by the genotype and its interaction with the environment).
- The **genotype** is the genetic constitution of an organism. The **phenotype** is the observable manifestation of the genetic traits. The genes give the potential for development of characteristic; this potential often is affected by interactions with other genes and with the environment.
- ***Thus, individuals with the same phenotypes may have different genotypes***. ***And individuals with the same genotype can have different phenotypes, how?***
- The **genotype** is essentially a fixed character of an individual organisms; the genotype remains constant throughout life and is essentially unchanged by environmental effects. Most phenotypes change continually throughout the life of an organism as its genes interact with a sequence of environments.





- **EUKARYOTIC CHROMOSOMES**: Each eukaryotic species has a ***characteristic number of chromosomes per cell***: potatoes have 48 chromosomes, fruit flies have 8, and humans have 46. There appears to be no special relation between the complexity of an organism and its number of chromosomes per cell.
- In most eukaryotic cells, there are two sets of chromosomes. The presence of two sets is a consequence of ***sexual reproduction***: one set is inherited from the male parent and the other from the female parent.
- Each chromosome in one set has a ***corresponding*** chromosome in the other set; together, the *two chromosomes* constitute a ***homologous pair*** (See **Figure 1**). Human cells, for example, have 46 chromosomes, constituting ***23 homologous pairs***.
- Human somatic cells contain 46 chromosomes (two copies of each of the autosomes, chromosomes 1 to 22, and two sex chromosomes, either XX [female] or XY [male]).
- Germ cells (eggs and sperm) are produced by **meiosis** and contain a haploid number of chromosomes (23) that consists of one copy each of the autosomes, and one sex chromosome.

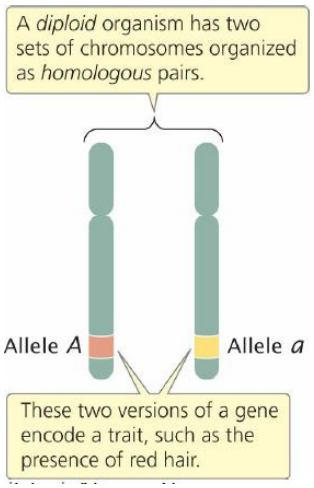


Figure 1: Diploid eukaryotic cells have two sets of chromosomes.

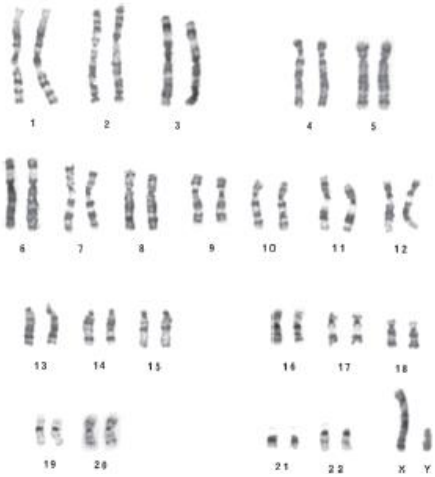


Figure 2: An example of a male karyotype.

- A **karyotype** displays all of the chromosomes in a cell, obtained from a metaphase spread (See **Figure 2)**.
- The *two chromosomes* of a ***homologous pair*** are usually alike in ***structure*** and ***size***, and ***each carries genetic information*** for the same set of hereditary characteristics.
- For example, if a gene on a particular chromosome encodes a characteristic such as hair color, another copy of the gene (each copy is called an allele) at the same position on that chromosome's homolog also encodes hair color. However, these two alleles need not be identical: one might encode brown hair and the other might encode blond hair.

- Cells that carry *two sets* of genetic information are **diploid** (**2n**).
- In general, the **ploidy** of the cell indicates how many sets of genetic information it possesses.
- The reproductive cells of eukaryotes (such as eggs, sperm, and spores), and even the non-reproductive cells of some eukaryotic organisms, contain a single set of chromosomes and are **haploid** (**1n**).
- The cells of some other eukaryotes contain more than two sets of genetic information and are therefore called **polyploidy.**
- A **functional chromosome** has three essential elements: **a centromere, a pair of telomeres,** and **origins of replication**.
- The **centromere** appears as a constricted region on the chromosome (See **Figure 3**). It serves as the attachment point for spindle microtubules—the filaments responsible for moving chromosomes in cell division. Before cell division, a multi-protein complex called the **kinetochore** assembles on the centromere; later, spindle microtubules attach to the kinetochore.
- On the basis of the **location of the centromere**, chromosomes are classified into four types: **metacentric, submetacentric, acrocentric, and telocentric** (**Figure 4**).
- **Telomeres** are the specific DNA sequences and associated proteins located at the tips of whole linear chromosomes. Telomeres protect and stabilize the chromosome ends.
- **Origins of replication** are the sites where DNA synthesis begins; unlike centromeres and telomeres, they are not easily observed by microscopy.

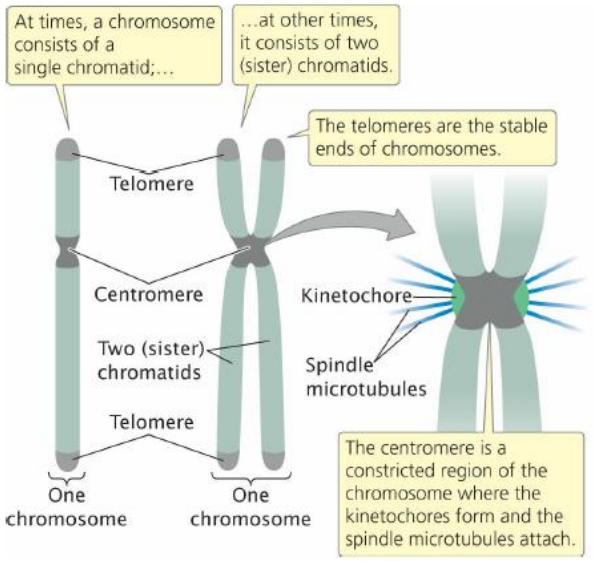


Figure 3: Each eukaryotic chromosome has a **centromere** and **telomeres**.

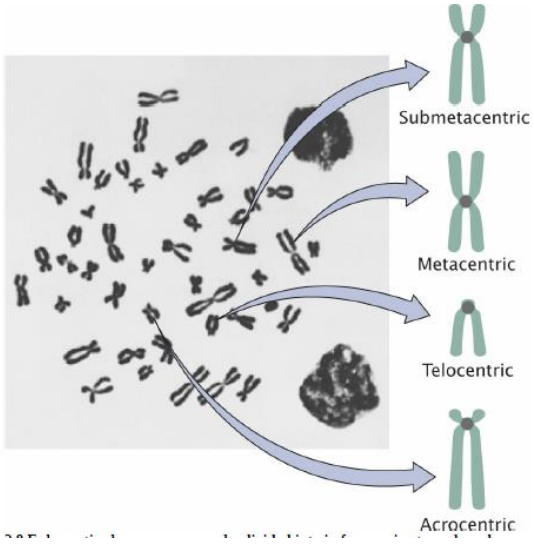
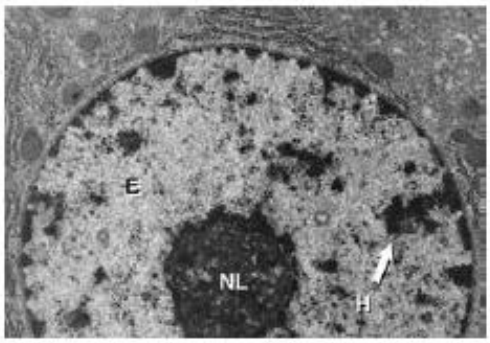


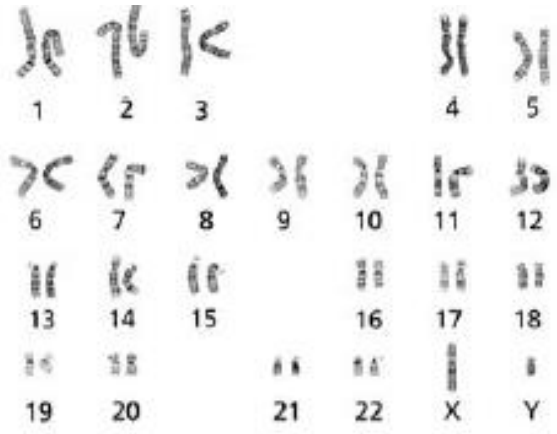
Figure 4: Eukaryotic chromosomes can be divided into in **four major types** based on the **position of the centromere**

A. Studying Human Chromosomes (Figure 1-5)

- **MITOTIC CHROMOSOMES** are fairly easy to study because they can be observed in any cell undergoing mitosis.
- **MEIOTIC CHROMOSOMES** are much more difficult to study because they can be observed only in ovarian or testicular samples. In the female, meiosis is especially difficult because meiosis occurs during fetal development. In the male, meiotic chromosomes can be studied only in a testicular biopsy of an adult male.
- **Blood** is the most convenient source of human cells for **karyotype analysis**. Blood cells are cultured and a **mitogen** is added to the culture media to stimulate the mitosis of lymphocytes. Subsequently, **colchicine** is added to the media which arrests the lymphocytes in metaphase. It is often preferable to use **prometaphase chromosomes** because they are less condensed and therefore show more detail. The **lymphocytes** are then concentrated and treated with a **hypotonic solution** to lyse the lymphocytes and aid in spreading the chromosomes. The cell preparation is then spread on a microscope slide, fixed, and stained by a variety of methods. The separate **metaphase chromosomes** are then identified and photographed. The photos of all the chromosomes are then cut out and arranged in a standard pattern called **the karyotype**. Figure 1-5 shows the **G-banding pattern of metaphase chromosomes** arranged in a karyotype.



● Figure 1-4 Heterochromatin and Euchromatin.



• Figure 1-5 Human Karyotype.

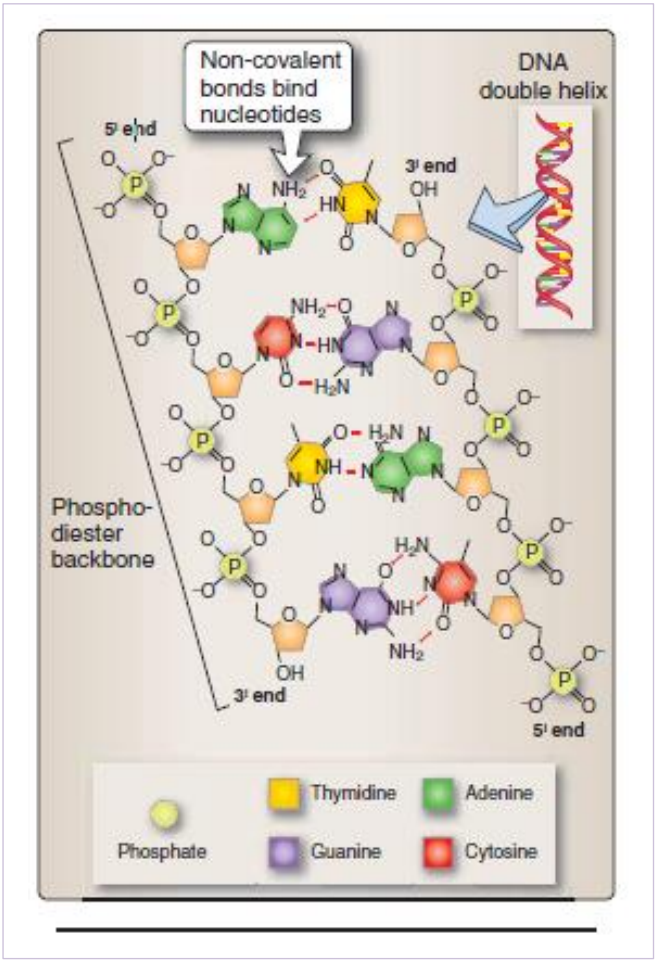
Chromosome Morphology (GENERAL FEATURES)

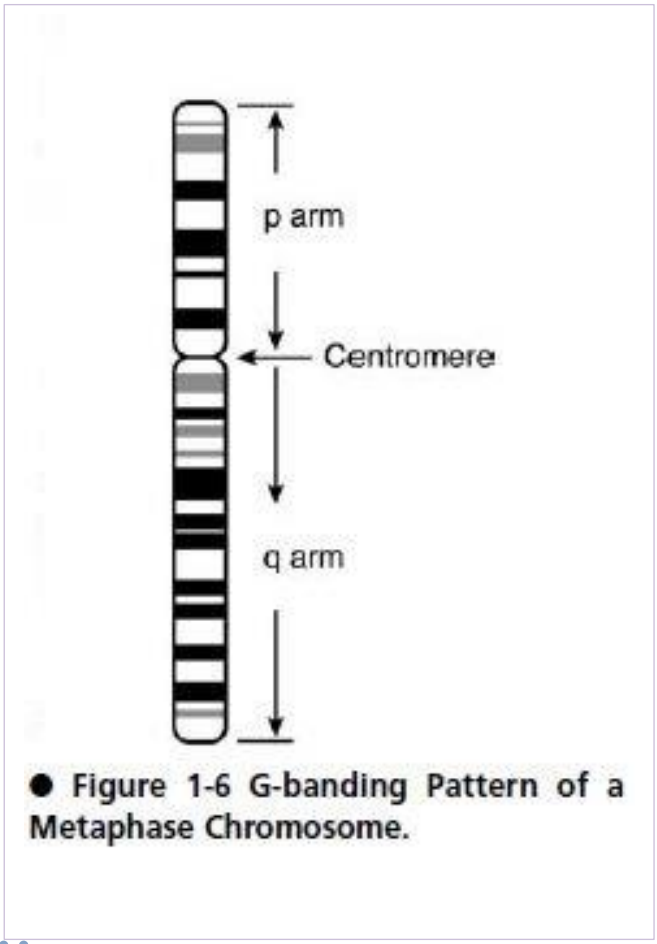
1. The appearance of chromosomal DNA can vary considerably in a normal resting cell (e.g., **degree of packaging**, **euchromatin**, and **heterochromatin**) and a **dividing cell** (e.g., mitosis and meiosis).
2. The pictures of chromosomes seen in karyotype analysis are chromosomal DNA at a particular point in time, that is, arrested at metaphase (or prometaphase) of mitosis. Early metaphase **karyograms** showed chromosomes as **X shaped** because the chromosomes were at a point in mitosis when the protein cohesion no longer bound the sister chromatids together, but the centromeres had not yet separated.

3. Modern metaphase karyograms show chromosome as **I shaped** because the chromosomes are at a point in mitosis when the protein **cohesin** still binds the sister chromatids together and the centromeres are not separated. Additonally, many modern karyograms are prometaphase karyograms where the chromosomes are **I shaped**.

CHROMOSOME NOMENCLATURE (Figure 1-6)

1. A chromosome consists of two characteristic parts called **arms**. The short arm is called the **p (petit) arm** and the long arm is called the **q (queue) arm**.
2. The **arms** can be subdivided into **regions** (counting outward from the centromere), **subregions (bands)**, **subbands** (noted by the addition of a decimal point), and **sub subbands**.
3. For example, **6p21.34** is read as the short arm of chromosome 6, **region 2**, and **sub-region (band) 1**, **sub-band 3**, and **sub-sub-band 4**. This is NOT read as the short arm of chromosome 6, twenty-one point thirty-four.
4. Besides, locations on an arm can be referred to in anatomical terms: proximal is closer to the centromere and distal is farther from the centromere.
5. Figure 1-6 shows the **G-banding pattern of a metaphase chromosome** along with the centromere, p arm, and q arm.





● Figure 1-6 G-banding Pattern of a Metaphase Chromosome.