Key terms

Chromatin: DNA wound on histones; a fine network in a nondividing nucleus.

Chromosome: thread-like body in the nucleus, one long DNA molecule with histones, carrying genes.

Sister chromatids: two identical copies of a chromosome, joined at the centromere.

Homologous chromosomes: pair with the same genes at the same loci; one from each parent.

Karyotype: all a cell's chromosomes in pairs, by size.

Diploid (2n) / haploid (n): two sets / one set.

Heredity: characters pass from parents to offspring.

Variation: differences within a species.

Gene: stretch of DNA for one protein or RNA (Johannsen, 1909).

Locus: a gene's fixed place.

Allele: one form of a gene (T, t).

Dominant / recessive: shows / is hidden when both are present.

Homozygous / heterozygous: TT or tt (pure) / Tt (hybrid).

Genotype / phenotype: the alleles / the character seen.

Formula

$$\text{arm ratio} = \frac{q}{p}$$

q long arm; p short arm (same unit); never below 1
Use: type of chromosome

- 6.0 µm long, centromere 1.2 µm from one end: 4.8 ÷ 1.2 = 4 → acrocentric
- 9 µm, ratio 2: $p + 2p = 9$ → arms 3 µm, 6 µm

3.1 Chromosome and its types

- named by Waldeyer, 1888: *chroma*, color + *soma*, body (take up stains strongly)
- seen clearly at cell division, best at **metaphase**: tightly coiled, lined up in the middle
- DNA of 46 chromosomes: about 2 m; 0.34 nm per base pair
- chromosome 1: 248 million base pairs ≈ 8.4 cm, coiled to about 0.01 mm

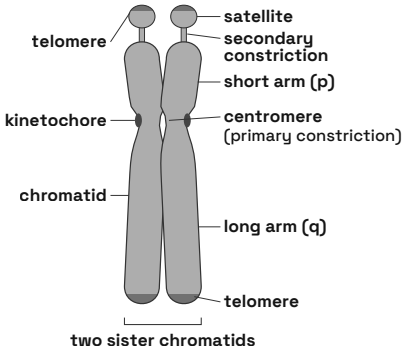
By structure

Type	Centromere	q/p	Anaphase	Human
metacentric	middle	1–1.7	V	1, 3, 16, 19, 20
submetacentric	off the middle	1.7–3	L	2, 4–12, 17, 18, X
acrocentric	near one end	over 3	J	13, 14, 15, 21, 22, Y
telocentric	at the end	no short arm	I (rod)	none (house mouse: nearly all 40)

- Levan's finer scheme (1964): **subtelocentric** ratio 3–7; acrocentric beyond 7; in an exam, the four school types
- by centromeres
  - **monocentric** (one)
  - **dicentric** (two)
  - **acentric** (none): torn or lost
  - **holocentric** (whole length, *C. elegans*)
- bacterium: no nucleus; single DNA ring; not wound on histones as ours is

Parts at metaphase

- **chromatids**: two identical sisters; pulled apart at division
- **centromere** (primary constriction)
  - joins them
  - **kinetochore** discs either side, where spindle fibers attach
  - leads to the pole
- **arms**: short **p**, long **q**
- **telomeres**
  - ends
  - TTAGGG repeated in humans
  - protect, stop sticking
- **secondary constriction**: on a few; many are **nucleolar organizers** (ribosome genes)
- **satellite**
  - knob beyond it
  - **SAT chromosome**
  - human 13, 14, 15, 21, 22
- old names
  - chromonemata
  - matrix
  - pellicle (no skin seen with the electron microscope)



A chromosome at metaphase.

- human 2 = two apes' chromosomes joined (apes 48, we 46): facing telomere repeats, an unused centromere

By function

Autosomes	Sex chromosomes (allosomes, heterosomes)
body's characters	decide sex
22 pairs (44)	1 pair (2)
pair alike	may differ (X, Y)
same in both sexes	XX female, XY male

- woman **44 + XX**; man **44 + XY**
- X: large, submetacentric, about 800 genes; Y: small, acrocentric, a few dozen
- human 2n = 46; egg or sperm n = 23 (22 autosomes + 1 sex chromosome)
- birds: female ZW, male ZZ; most flowering plants: no sex chromosomes
- Painter 48 (1923) → Tjio and Levan 46 (1956): colchicine, weak salt solution
- metaphase of mitosis
  - 46 chromosomes
  - 46 centromeres
  - 92 chromatids

3.2 Introduction to genetics

Genetics: study of heredity and variation (Bateson, 1905–1906).

Hereditary variation	Environmental variation
in the genes	from how one lives
passed on	not passed on
blood group, earlobe	wrestler's arms, tan, scar

- height: both; genes set the range, food and health decide where in it
- **Mendel**: Brno; peas, 1856–1863; published 1866; found again 1900; **father of genetics**
- particles in pairs, one from each parent; don't blend
- TT, Tt: different genotypes, same phenotype (tall)

Why it matters

- farming: crops and animals bred for
  - yield
  - short stems
  - resistance
  - milk
- medicine
  - thalassemia
  - sickle-cell anemia
  - hemophilia
  - tests, advice
- identifying people: DNA fingerprinting
- understanding life
  - variation
  - change
  - relatedness

3.3 Gene and chromosome

- gene = segment of a chromosome's DNA; hundreds to thousands in a line, each at its locus
- homologous pair: same genes at same loci → genes come in pairs (TT, Tt)
- X and Y share only a few genes

Chromosome theory of inheritance (Sutton and Boveri, 1902–1903): genes are carried on chromosomes, and pass from parents to offspring because chromosomes do.

Genes	Chromosomes
in pairs in a body cell	in pairs in a body cell
separate in gametes	separate in meiosis
pairs shared independently	pairs line up independently
one from each parent	one from each parent

- **Morgan** (*Drosophila*, 1910): white-eyed male; white eyes almost only in males, grandfather → grandson through daughters → gene on X; Nobel 1933
- **linkage**: genes on one chromosome inherited together; Sturtevant's first gene map, 1913
- humans
  - about 20,000 genes for proteins
  - chromosome 1 about 2,000
  - under 2% of our DNA
- mitochondria
  - own ring
  - from the mother only
  - 37 genes

Gene	Chromosome
a segment of DNA	a whole DNA molecule + proteins
one protein or RNA	hundreds or thousands of genes
too small for a light microscope	seen at cell division
fixed locus	in the nucleus
about 20,000 kinds	23 kinds, 46 in all

COMMON MISTAKES

✗ arm ratio = short ÷ long
✓ long ÷ short; never below 1.

✗ egg has 23 autosomes
✓ 22 autosomes + X.

✗ a chromosome is made of genes
✓ carries genes, with much other DNA.

✗ homologous chromosomes have the same alleles
✓ same genes; alleles may differ.

✗ metaphase: 92 chromosomes
✓ 46 chromosomes, 92 chromatids.

✗ acquired scars or strong arms are inherited
✓ environmental; not passed on.

✗ telocentric chromosomes in humans
✓ none.