Sex Linked Inheritance, Chromosome Mapping & Mutations

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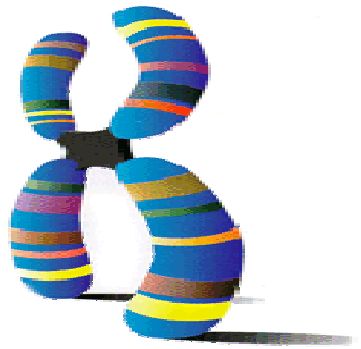
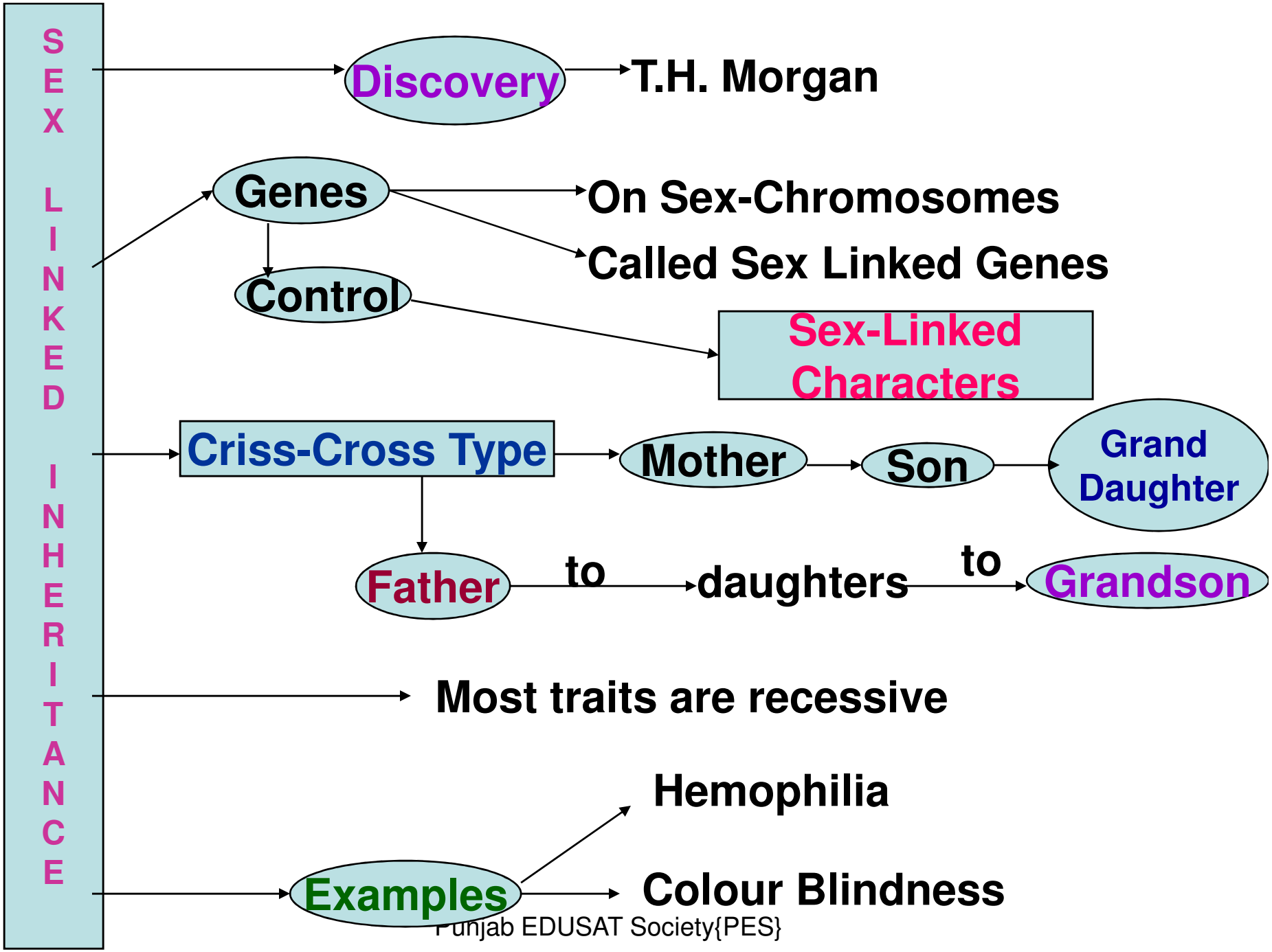



SEX LINKED INHERITANCE
Discovery
T.H. Morgan
Genes
On Sex-Chromosomes
Called Sex Linked Genes
Control
Sex-Linked Characters
Criss-Cross Type
Mother
Son
Grand Daughter
Father
to
daughters
to
Grandson
Most traits are recessive
Examples
Hemophilia
Colour Blindness

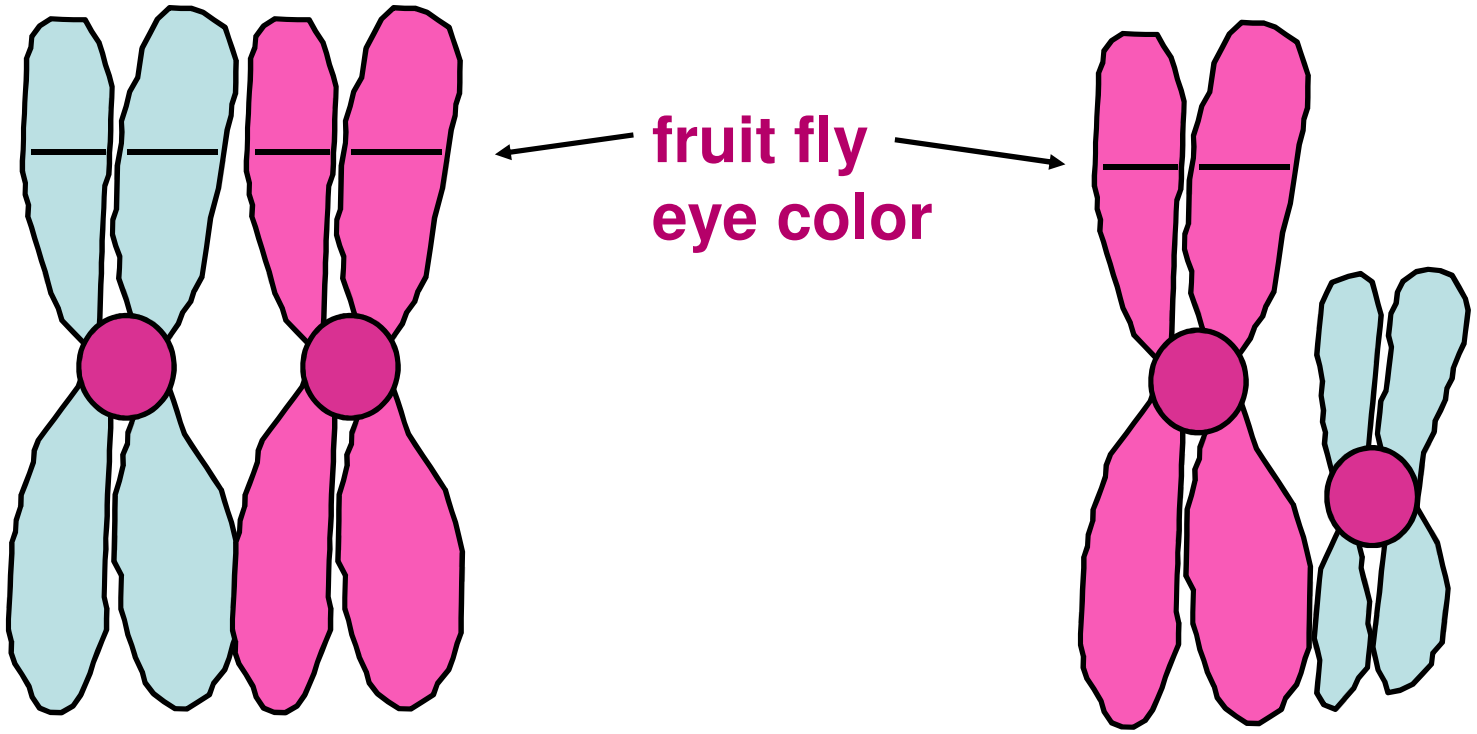
Sex-linked Traits

- Traits (genes) located on the sex chromosomes
- Sex chromosomes are X and Y
- XX genotype for females
- XY genotype for males
- Many sex-linked traits carried on X chromosome

Sex-linked Traits

Example: Eye color in fruit flies

Sex Chromosomes



XX chromosome - female **Xy chromosome - male**

Hemophilia

Parents	Normal Woman	X	Hemophilic Man
Genotypes	XX		X^hY
Gametes	X X		X^h Y

Progeny

	X^h	Y
X	XX^h	XY
X	XX^h	XY

Parents	Hemophilic Man	X	Carrier Woman
Genotypes	X^hY		XX^h

Gametes

Progeny

	X	X^h
X^h	X^hX	X^hX^h
Y	XY	X^hY

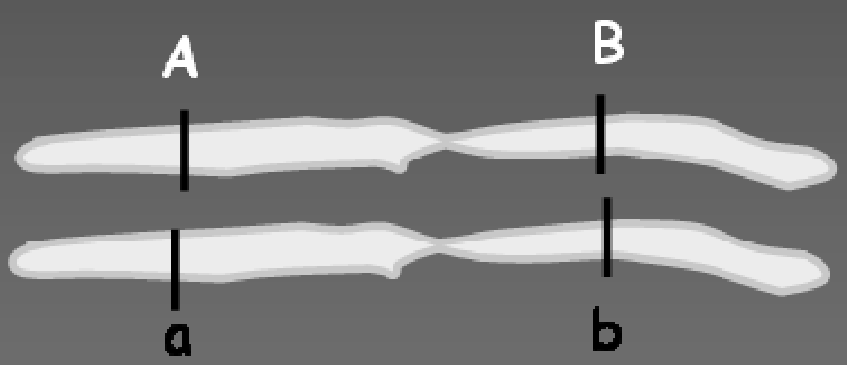
Lethal

Gene Mapping

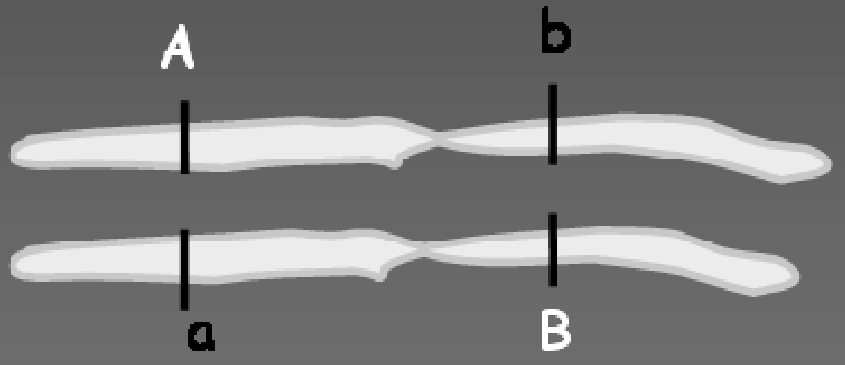
determines order of genes & relative distances between them

1 map unit = 1 cM (centimorgan)

1 locus; 2 loci



A & B are Cis arrangement
(coupled)

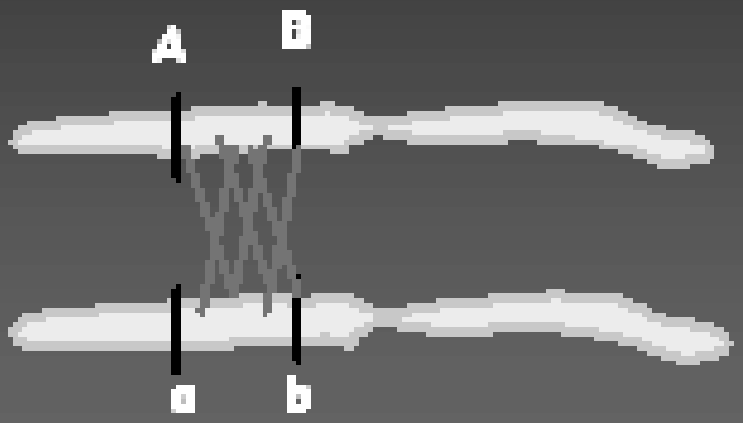


Trans arrangement
(repulsed)

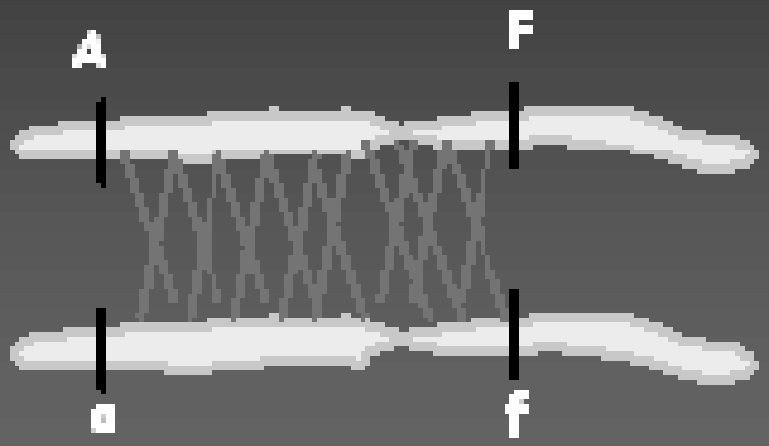
Gene Distance

recombination frequencies between alleles:
- determine relative distance between them
- proportional to their distance apart

1% recombination = 1 map unit = 1 cM



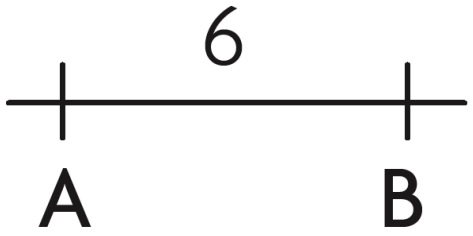
10% recombination



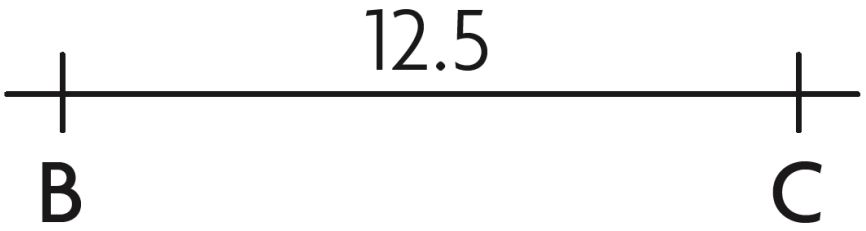
45% recombination

Cross-over frequencies can be converted into map units.

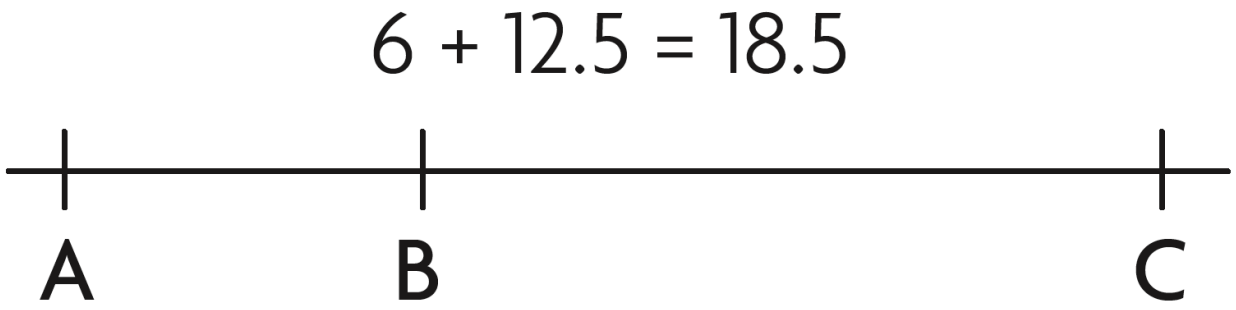
– gene A and gene B cross over 6.0 percent of the time



– gene B and gene C cross over 12.5 percent of the time



– gene A and gene C cross over 18.5 percent of the time



Linkage maps estimate distances between genes.

- The closer together two genes are, the more likely they will be inherited together.
- Cross-over frequencies are related to distances between genes.
- Linkage maps show the relative locations of genes.

MUTATIONS

Mutations can be defined as sudden, stable, discontinuous and inheritable variations which appear due to permanent change in the genotype of an organism .

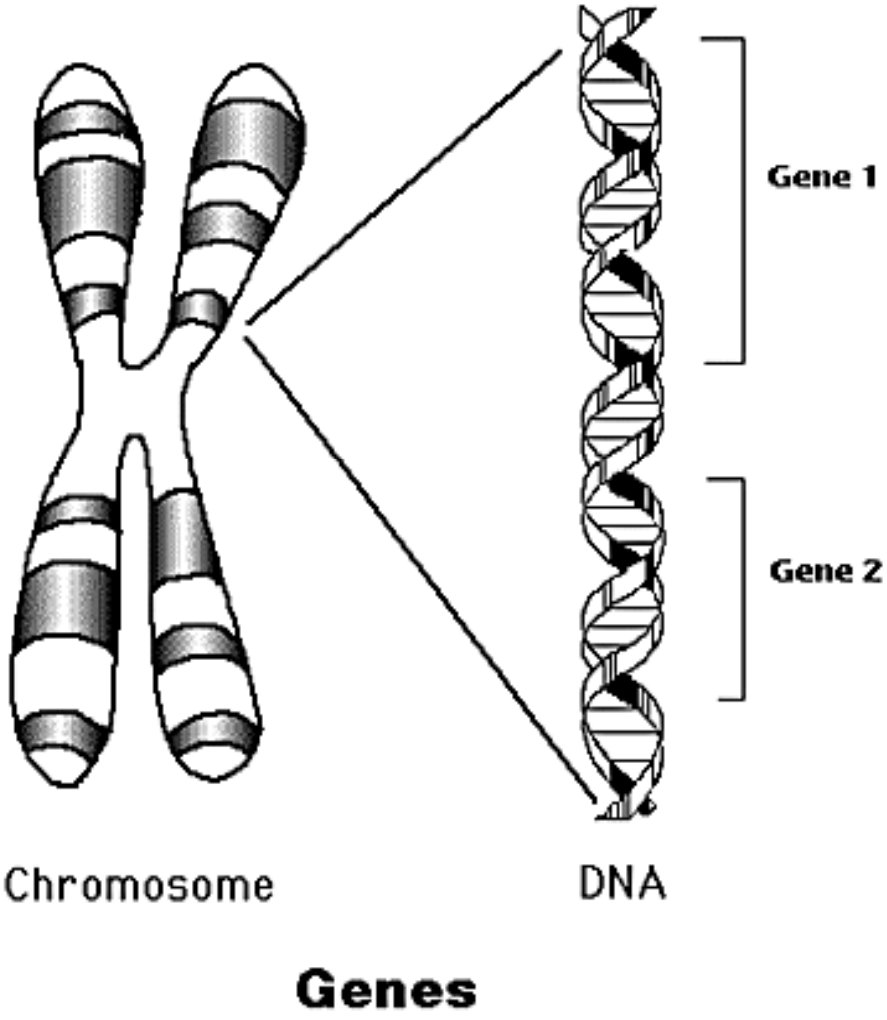
They are responsible for the origin of new traits and therefore mutations are the source of all variations .

Somatic vs Germ Mutations

Some people may have mutations in their skin cells or hair. Such mutations are termed Somatic. Germ mutations occur only in the sex cells.

These mutations are more threatening because they can be passed to offspring (forever).

Types of mutations



Chromosomal: affecting whole or a part of a chromosome

Gene: changes to the bases in the DNA of one gene

Chromosomal Mutations

1.Morphological modification of chromosomes

- Deletion
- Inversion

} Intra-chromosomal modification

- Translocation
- Duplication

} Inter-chromosomal modification

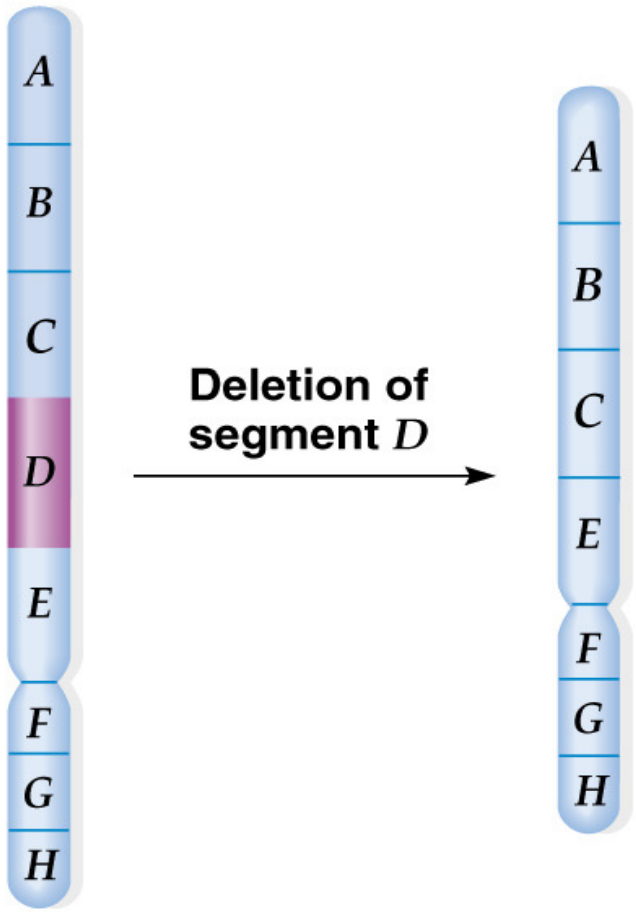
2. Numerical changes in chromosome

- Aneuploidy
- Euploidy

Deletion

1. In a deletion, part of a chromosome is missing
   a. Deletions start with chromosomal breaks induced by:
      i. Heat or radiation (especially ionizing).
      ii. Viruses.
      iii. Chemicals.
      v. Errors in recombination.
   b.Deletions do not revert, because the DNA is missing.
2. The effect of a deletion depends on what was deleted.

Deletion in Chromosome



Inversion

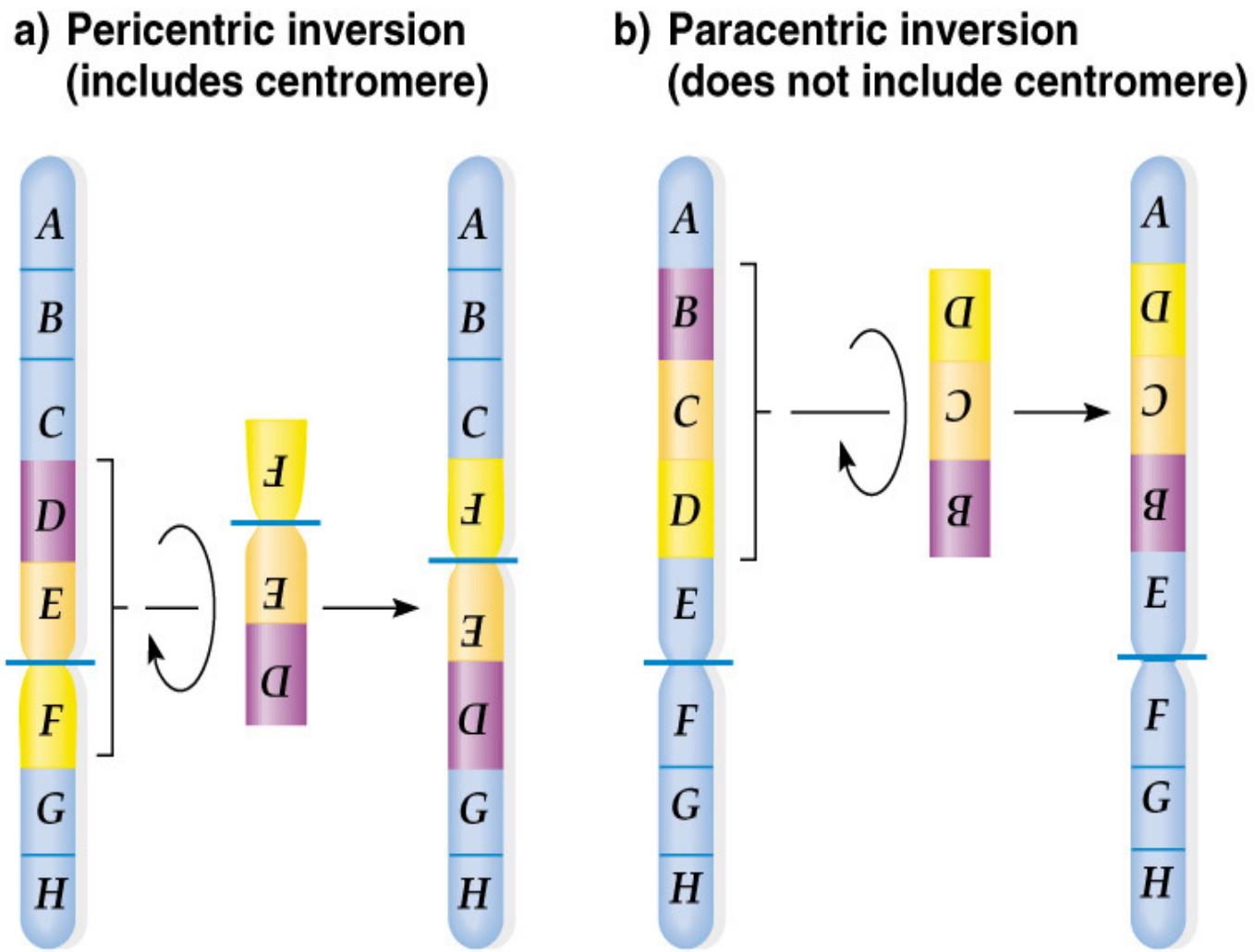
1. Inversion results when a chromosome segment excises and reintegrates oriented 180^0 from the original orientation. There are two types:

a. Pericentric inversions include the centromere.

b. Paracentric inversions do not include the centromere.

2. Inversions generally do not result in lost DNA, but phenotypes can arise if the breakpoints are in genes or regulatory regions.

INVERSIONS


a) Pericentric inversion
(includes centromere)
b) Paracentric inversion
(does not include centromere)
A
B
C
D
E
F
G
H

Duplication

Duplications result from doubling of chromosomal segments, and occur in a range of sizes and locations.

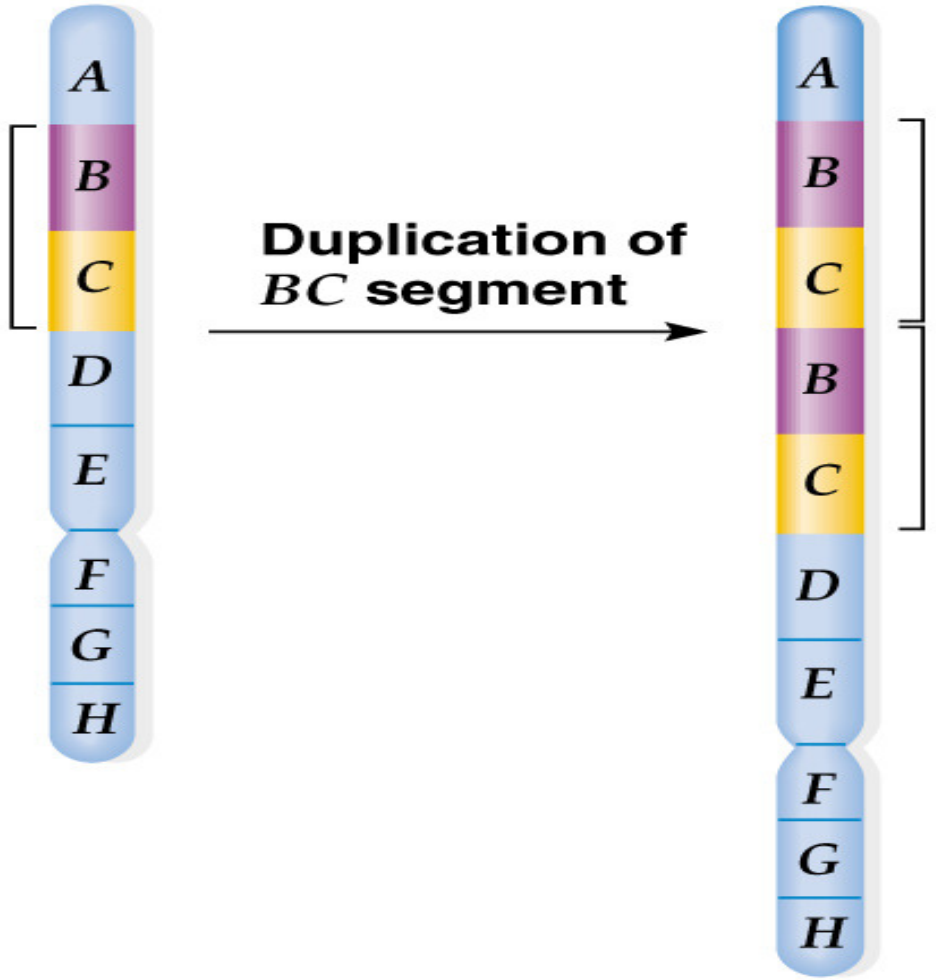
a. Tandem duplications are adjacent to each other.

b. Reverse tandem duplications result in genes arranged in the opposite order of the original.

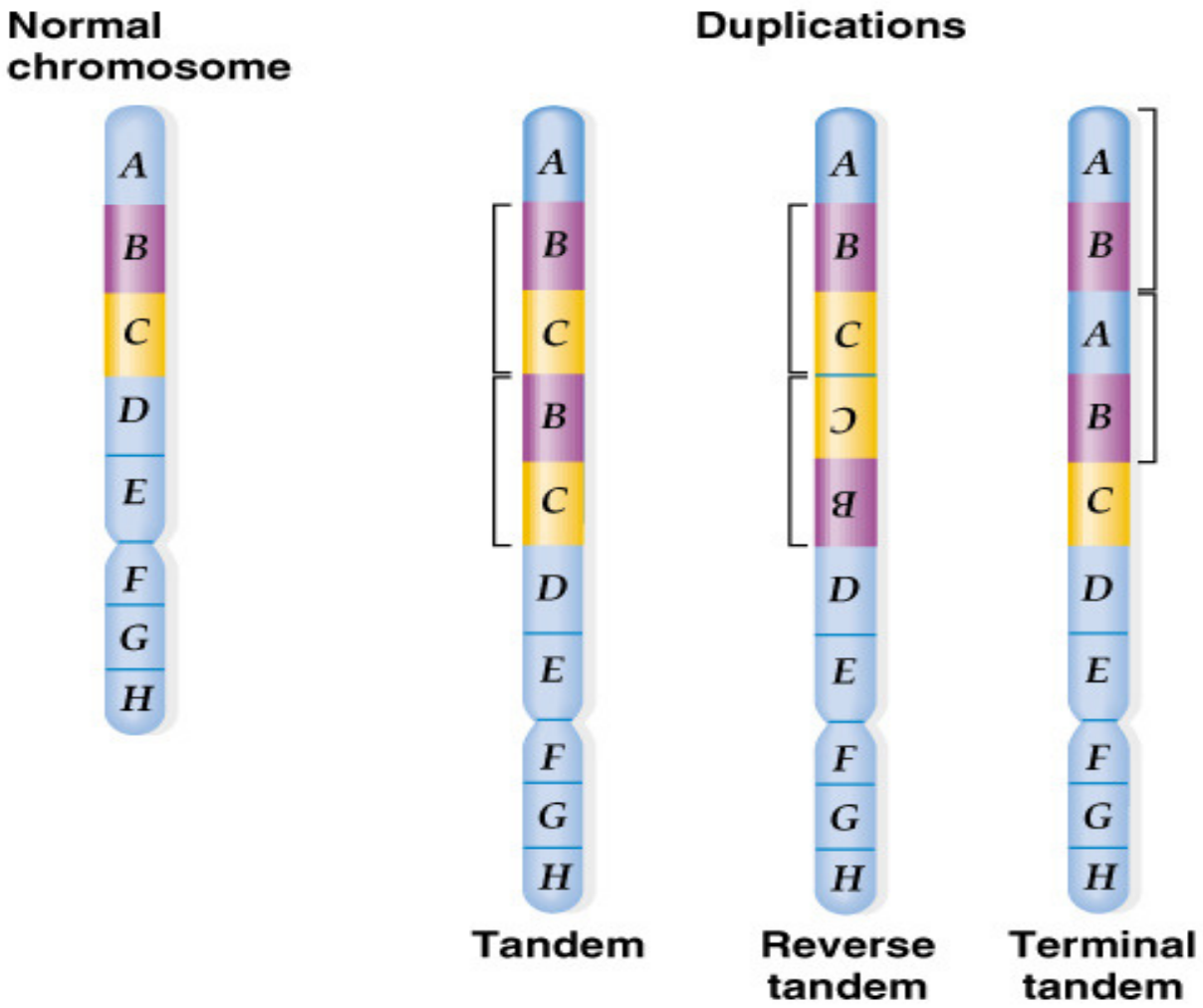
c. Tandem duplication at the end of a chromosome is a terminal tandem duplication.

Duplication

(a chromosome segment repeated)



Forms of chromosome duplications are tandem, reverse tandem and terminal tandem duplications

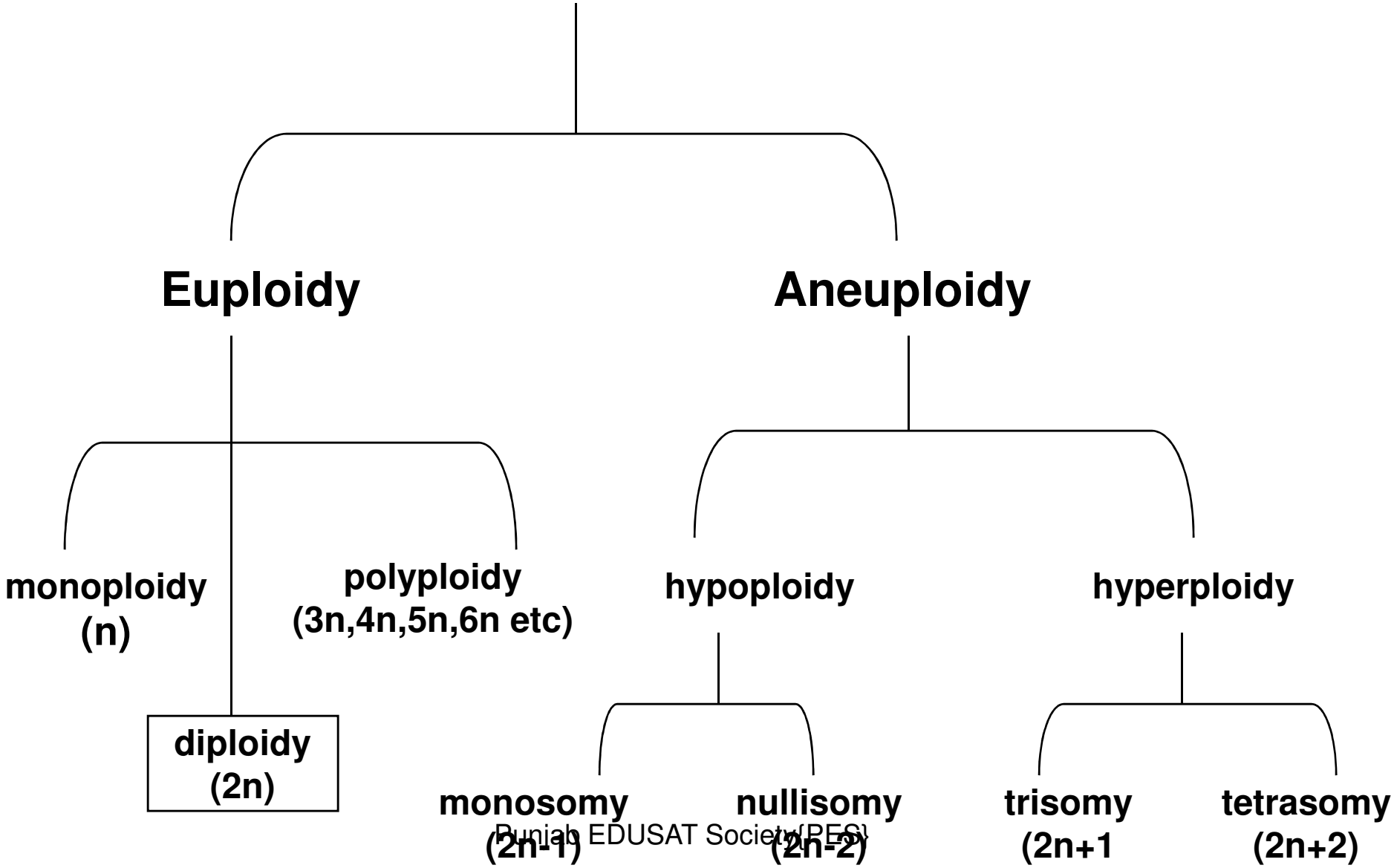


Variations in Chromosome Number

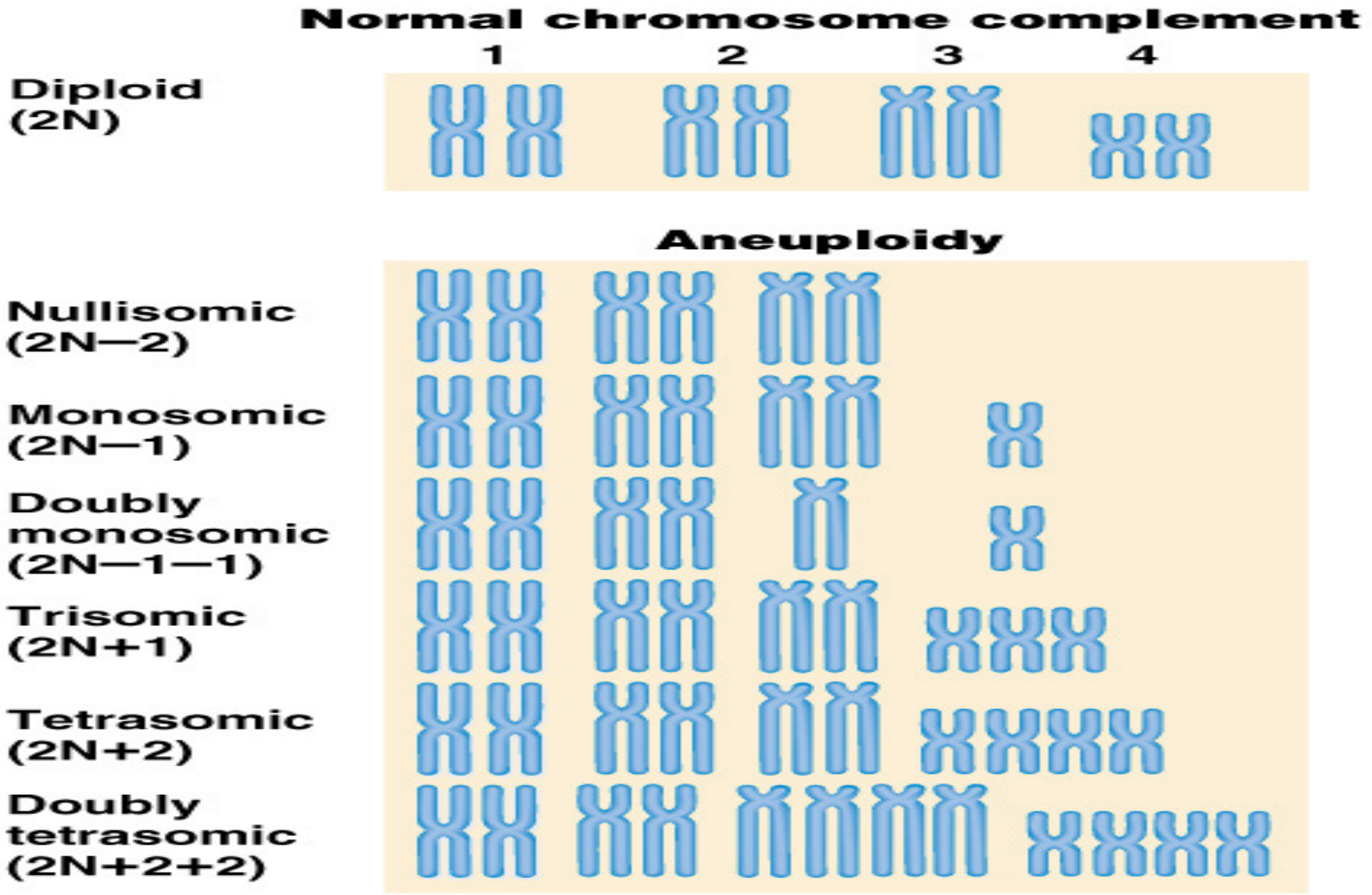
1. An organism or cell is **euploid** when it has one complete set of chromosomes, or exact multiples of complete sets. Eukaryotes that are normally haploid or diploid are euploid, as are organisms with variable numbers of chromosome sets.

2. **Aneuploidy** results from variations in the number of individual chromosomes (not sets), so that the chromosome number is not an exact multiple of the haploid set of chromosomes.

Numerical changes in chromosomes



Normal theoretical set of metaphase chromosomes in a diploid (2N) organism top and example of aneuploidy (bottom)



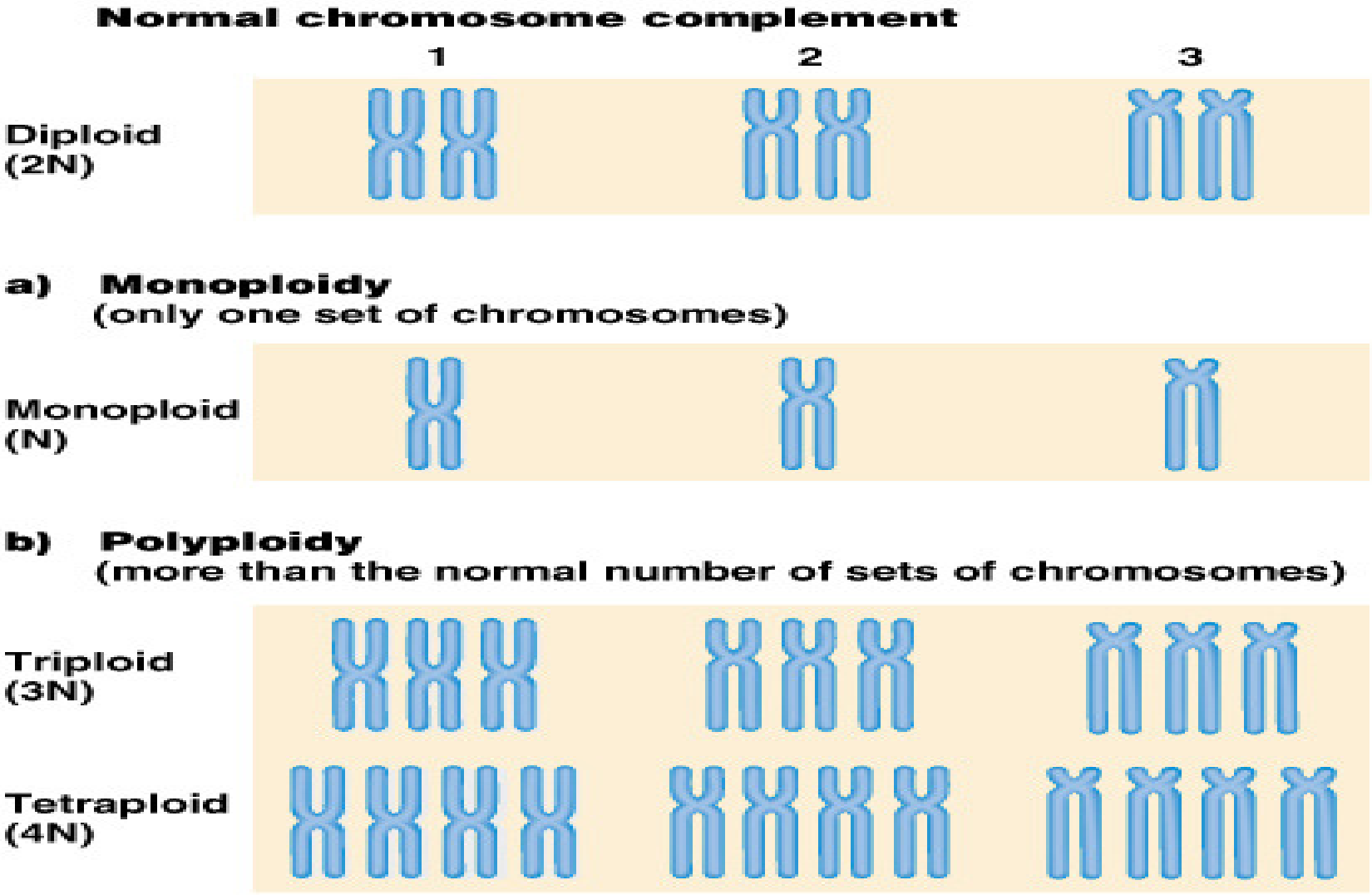
Changes in Complete Sets of Chromosomes

Monoploidy and **polyploidy** involve complete sets of chromosomes, and so both are cases of euploidy.

Euploidy is lethal in most animal species, but often tolerated in plants, where it has played a role in speciation and diversification.

Monoploidy and polyploidy can result when either round of meiotic division lacks cytokinesis, or when meiotic nondisjunction occurs for all chromosomes.

Variations in number of complete Chromosome sets



Plant polyploidy occurs in two types:

i. Autopolyploidy results when all sets of chromosomes are from the same species, usually due to meiotic error. Fusion of a diploid gamete with a haploid one produces a triploid organism. Examples include:

"Seedless" fruits like bananas, grapes and watermelons.

ii. Allopolyploidy results when the chromosomes are from two different organisms, typically from the fusion of haploid gametes followed by chromosome doubling. For example:

Fusion of haploid gametes from plant 1 and plant 2 produces an $N_1 + N_2$ hybrid plant.