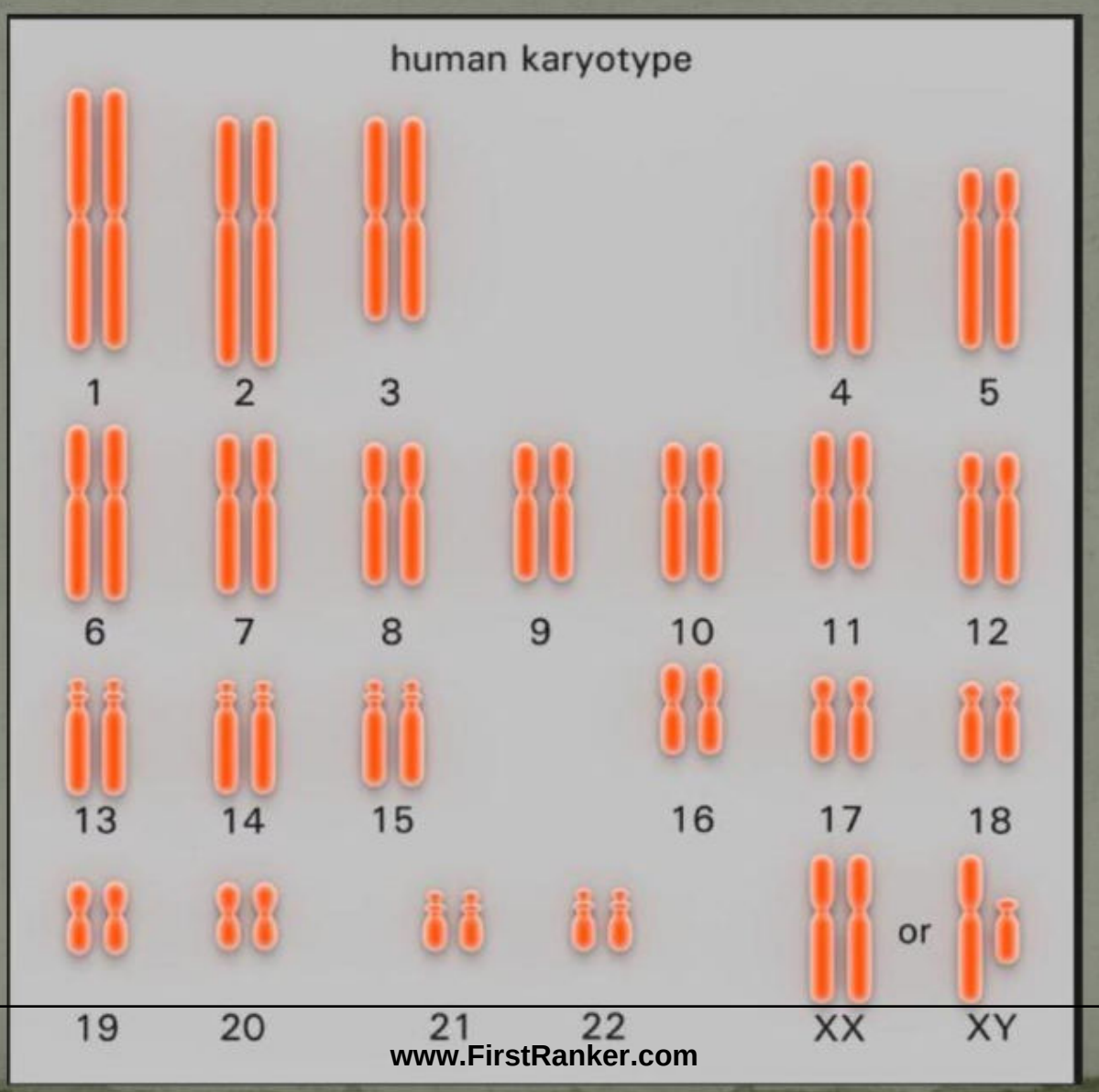
What is Karyotype

- a picture of arranged chromosomes.

The picture is created by photographing the cell during mitosis

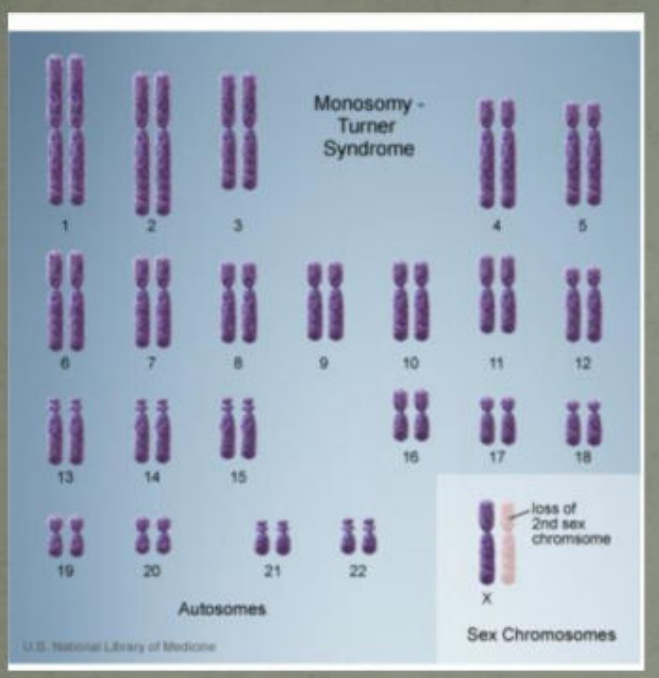
What can be learnt from making a karyotype?



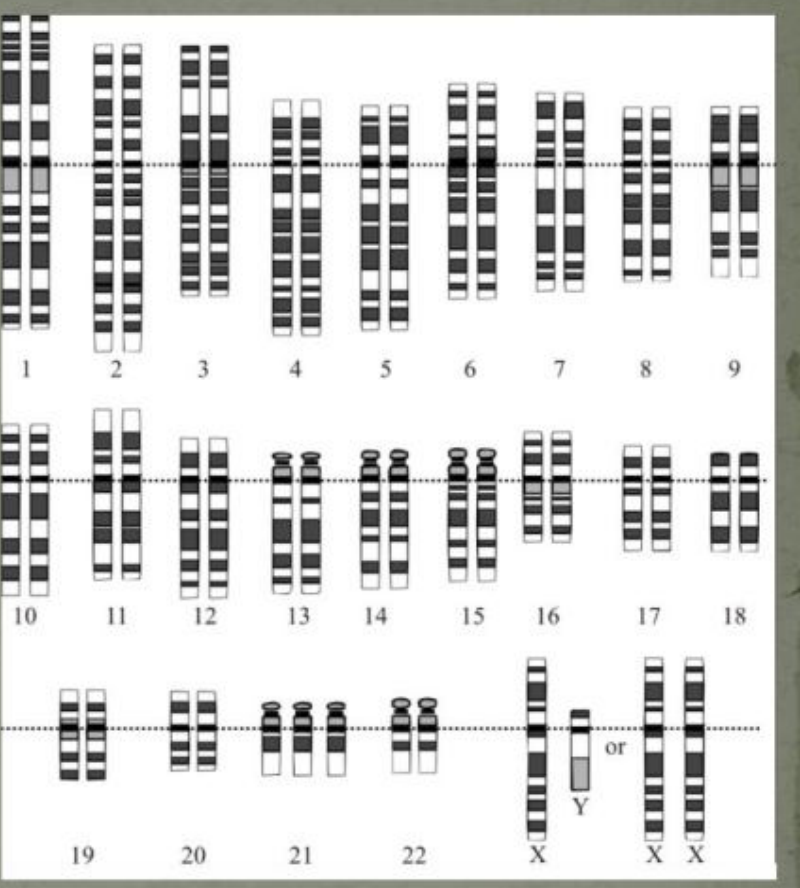
KARYOTYPE

- Refers to the chromosome complement of a cell or a whole organism
- In particular , it shows the number, size, and shape of the chromosomes as seen during metaphase of mitosis
- Chromosomes numbers vary considerably among organisms and may differ markedly between closely related species

Organism	Chromosome number (2n)
Drosophila	8
Honey bee	32 or 16
Goldfish	94
Rat	42
Rabbit	44
Cat	38
Dog	78
Gorilla	48
Chimpanzee	48
Human	46

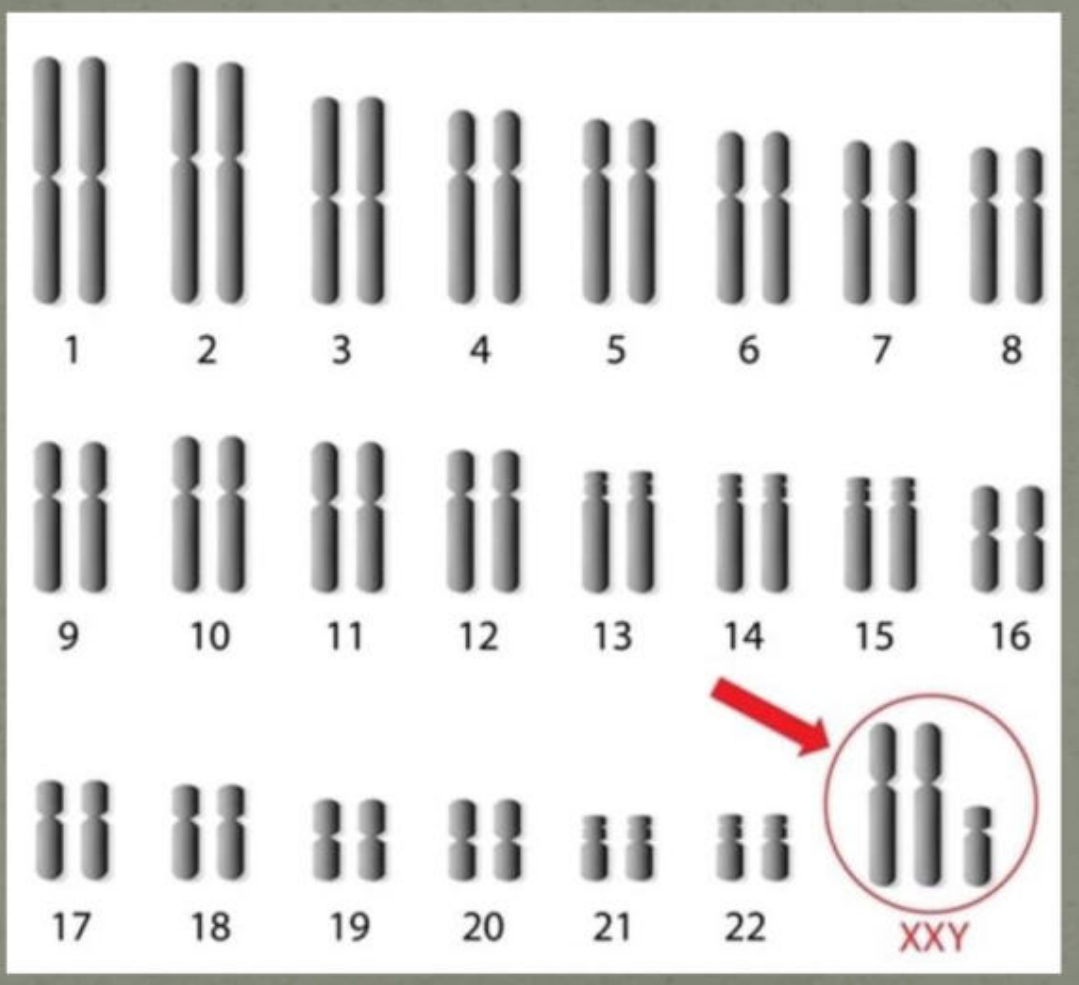


Monosomy (Turner syndrome)



Trisomy 21 Down syndrome

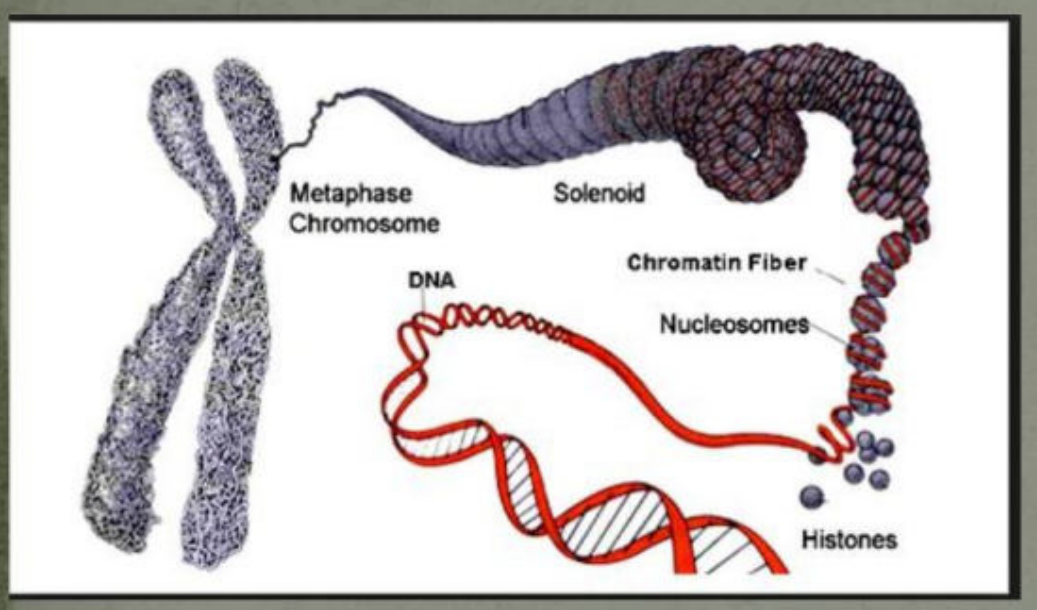
Klinefelter syndrome



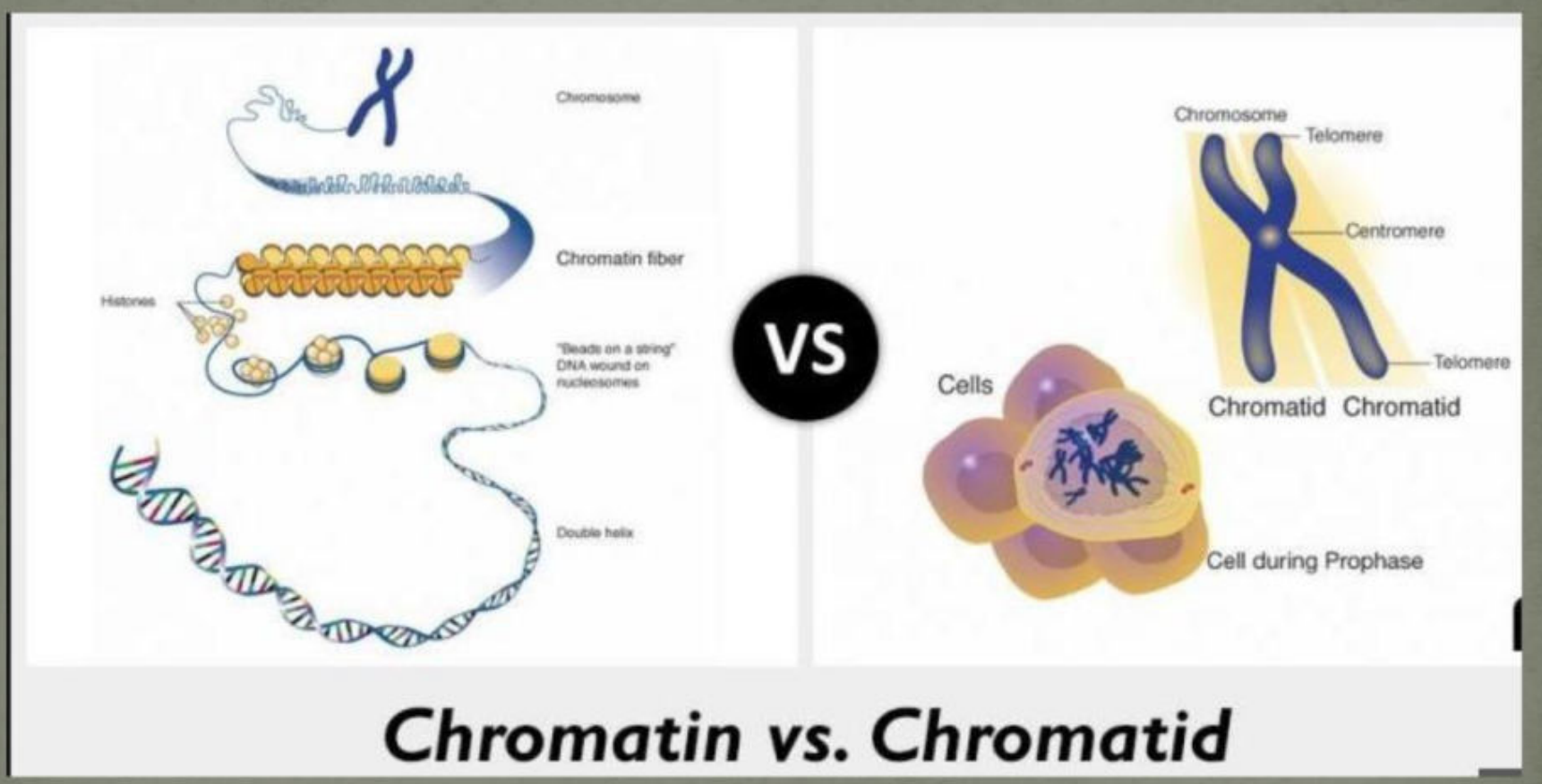
Chromatin/Chromosome/Chromatid

(Euchromatin and Heterochromatin)

Chromatin : Euchromatin and Heterochromatin



Chromatin is the complex of DNA and proteins found in the eukaryotic nucleus

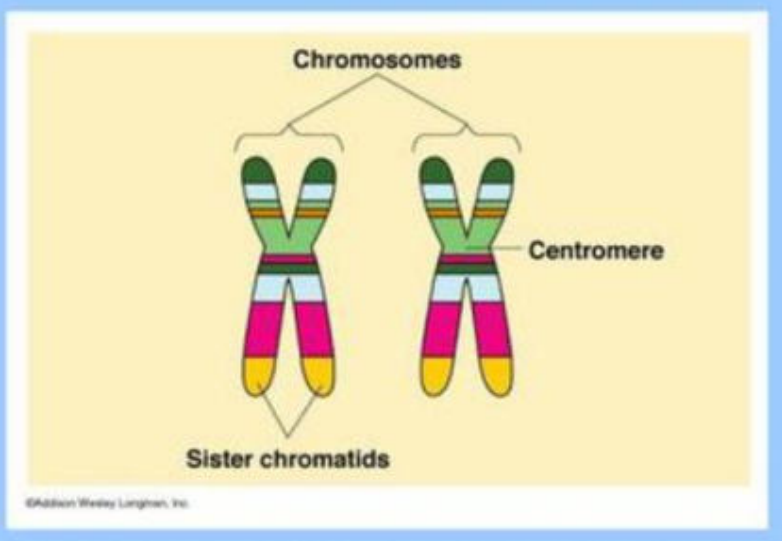


Chromatin vs. Chromatid

Chromosomes vs. Chromatin

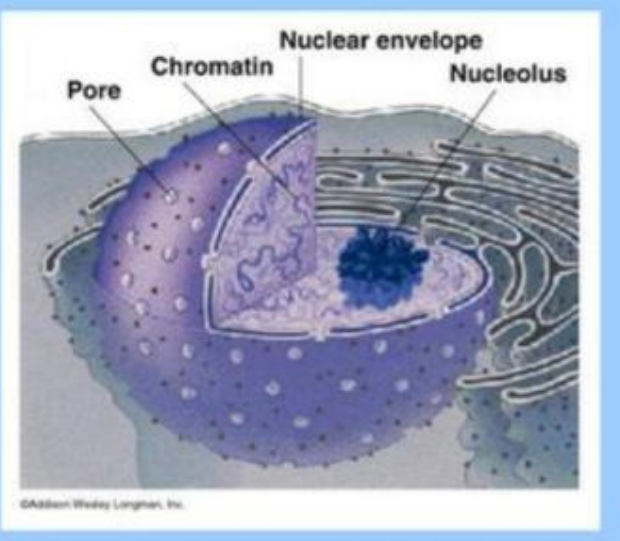
Chromosomes

- Tightly packaged DNA
- Found only during cell division
- DNA is not being used for macromolecule synthesis

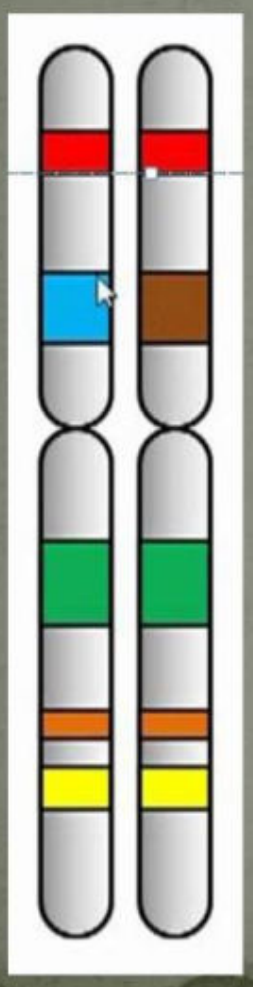


Chromatin

- Unwound DNA
- Found throughout Interphase
- DNA *is* being used for macromolecule synthesis



Gene/ Allele/ Locus



Gene:

Portion of a chromosome that serves as the basic unit of heredity

Allele:

Pair of genes that code for polypeptide s that express the same physical trait are called alleles

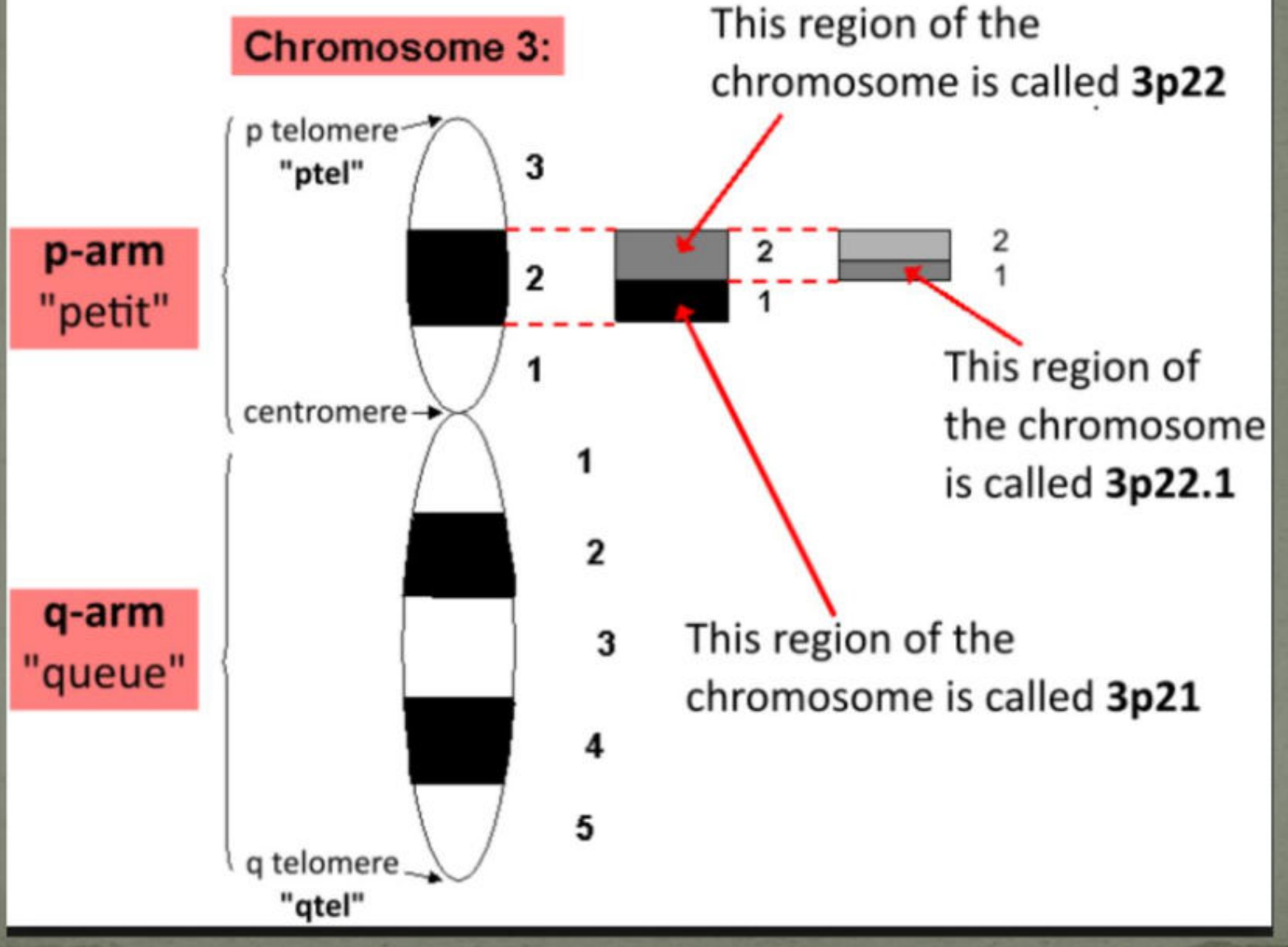
Locus:

Specific position of a gene on chromosome

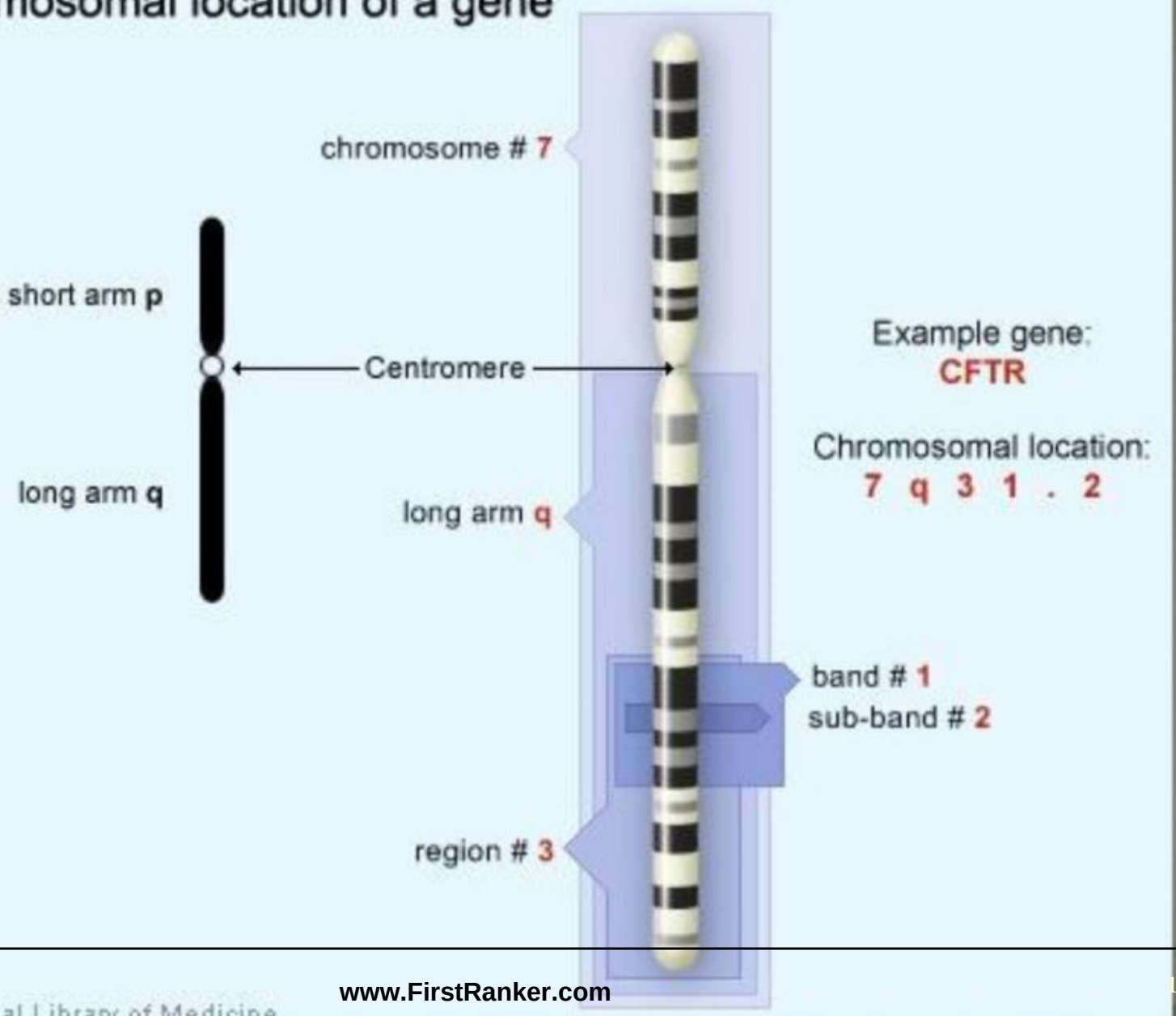
3p22.1

Cytogenetic location

Cytogenetic Banding Nomenclature



Chromosomal location of a gene



Genotype:

Combination of alleles of a gene carried by an organism

Phenotype:

Expression of alleles of a gene carried by an organism

Homozygous:

Having two similar alleles

Homozygous dominant: Having two copies of the same dominant allele

Homozygous recessive: Having two copies of the same recessive allele

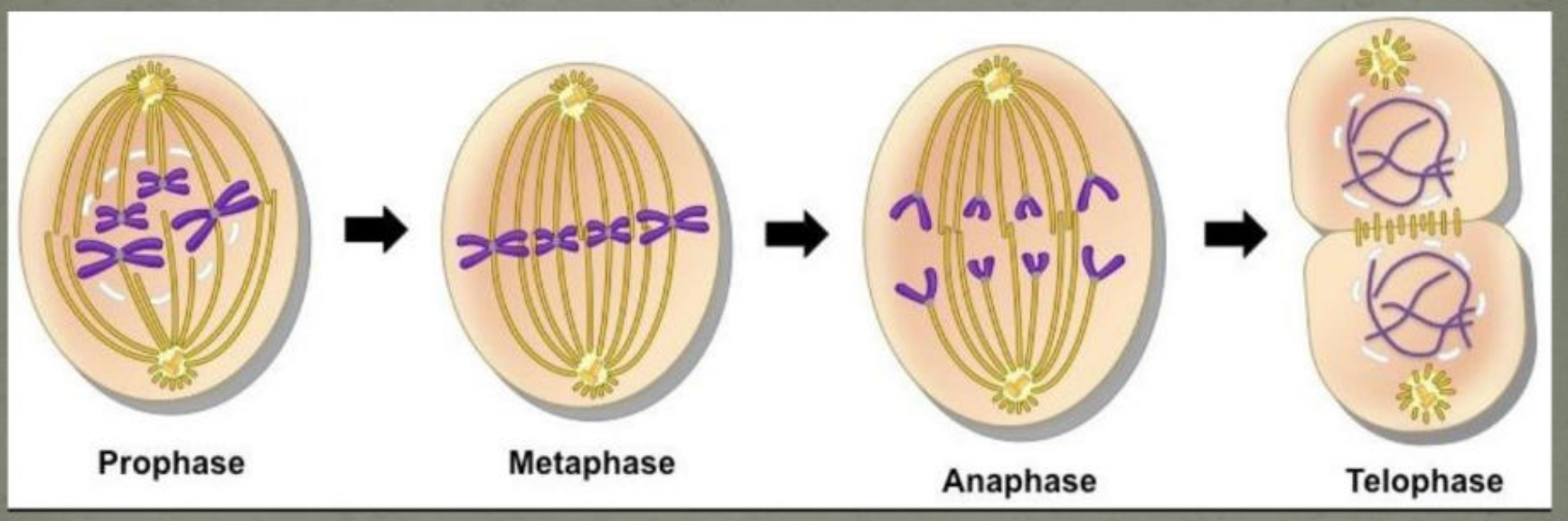
Heterozygous:

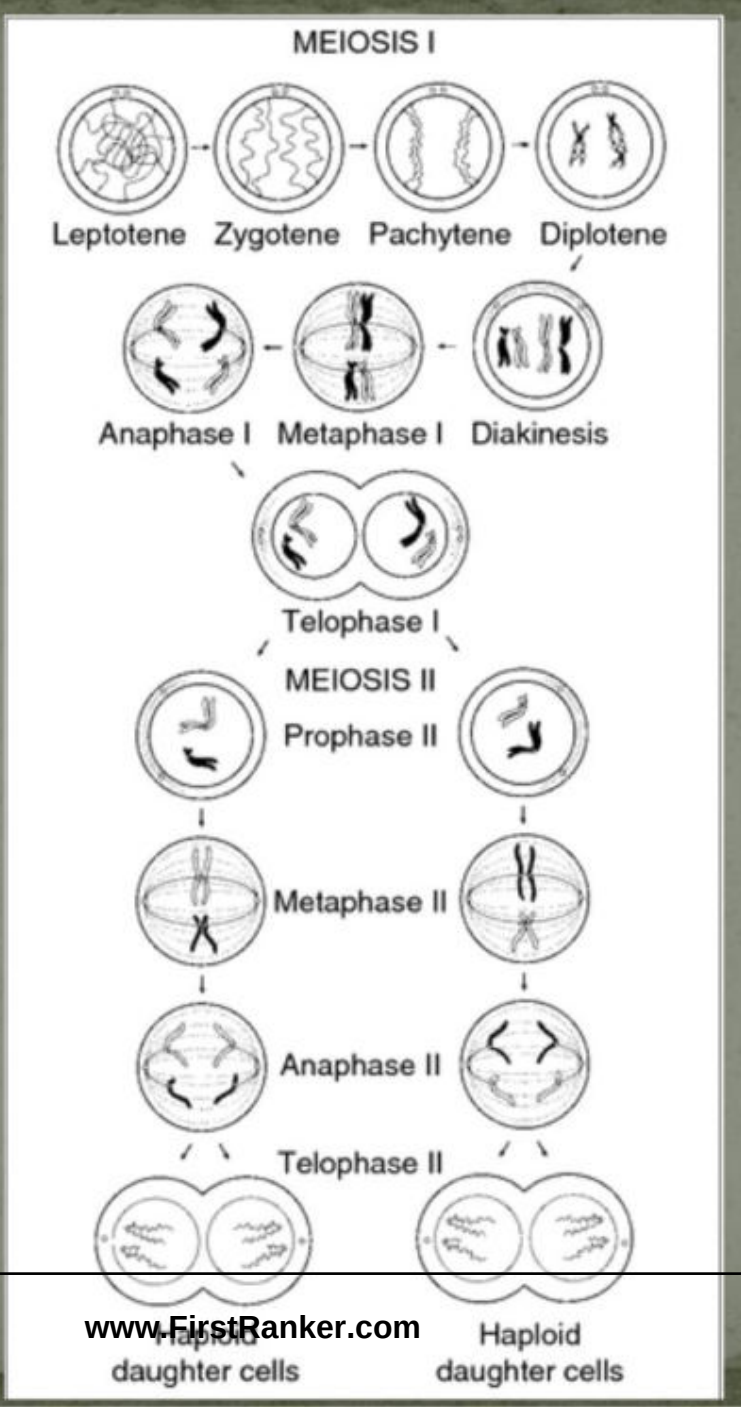
Having two different alleles, the dominant allele is expressed

Carrier:

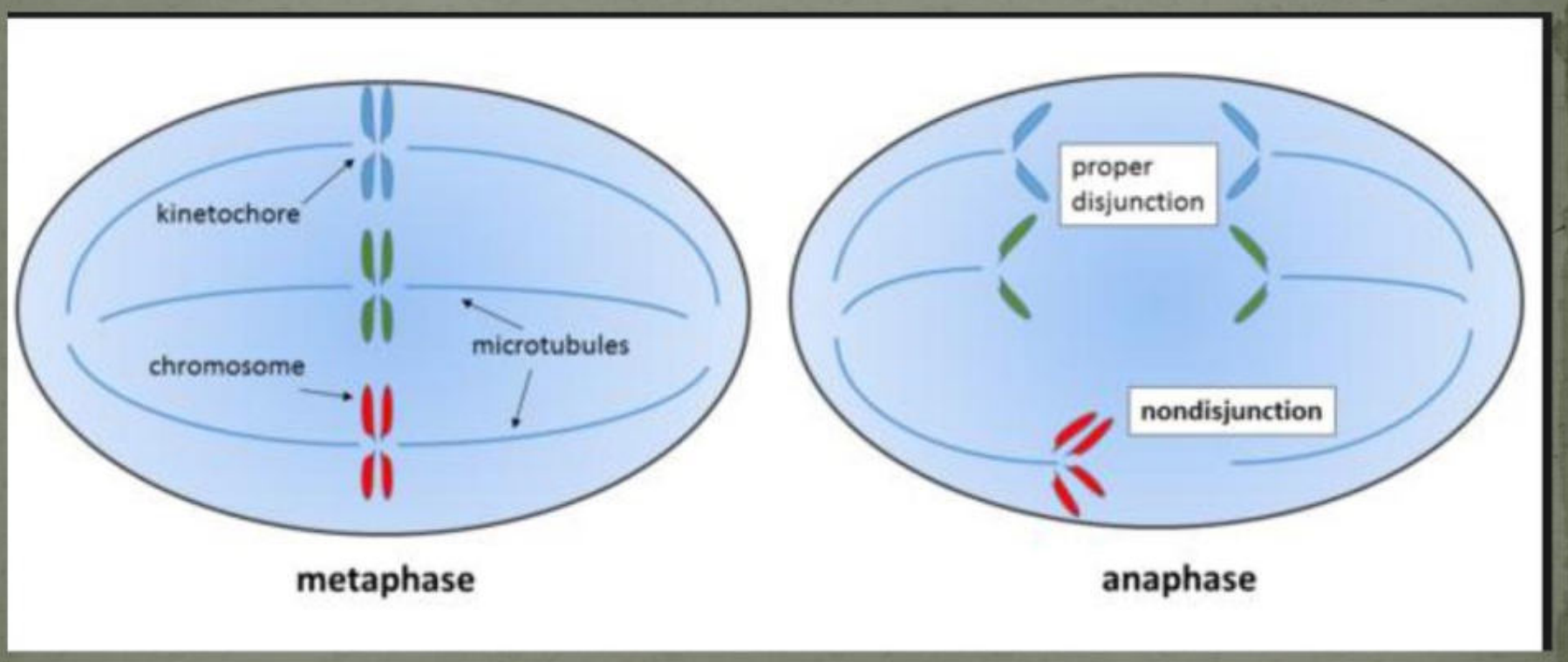
Heterozygous carrier of a recessive disease causing allele

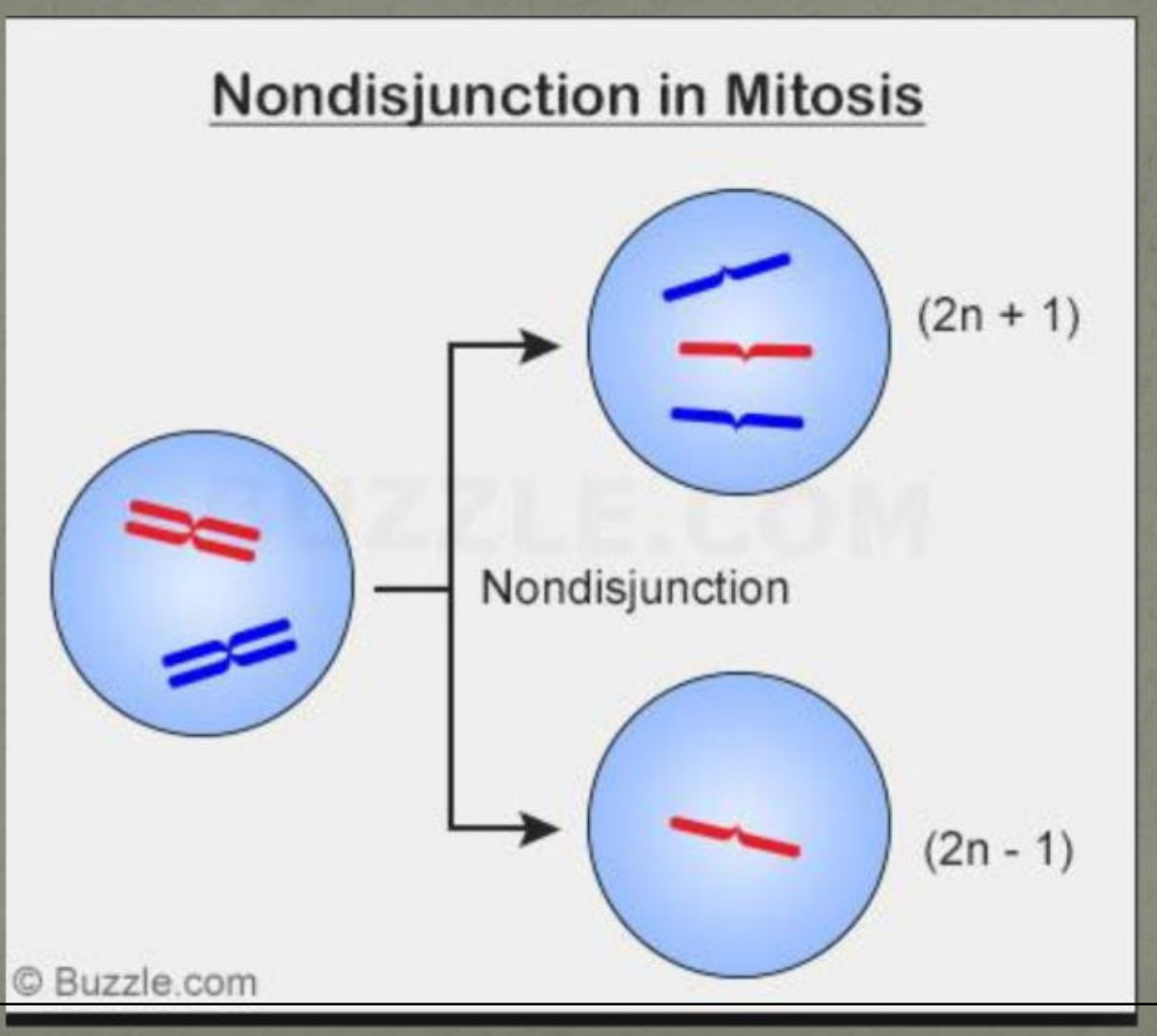
Cell Division :Mitosis and Meiosis

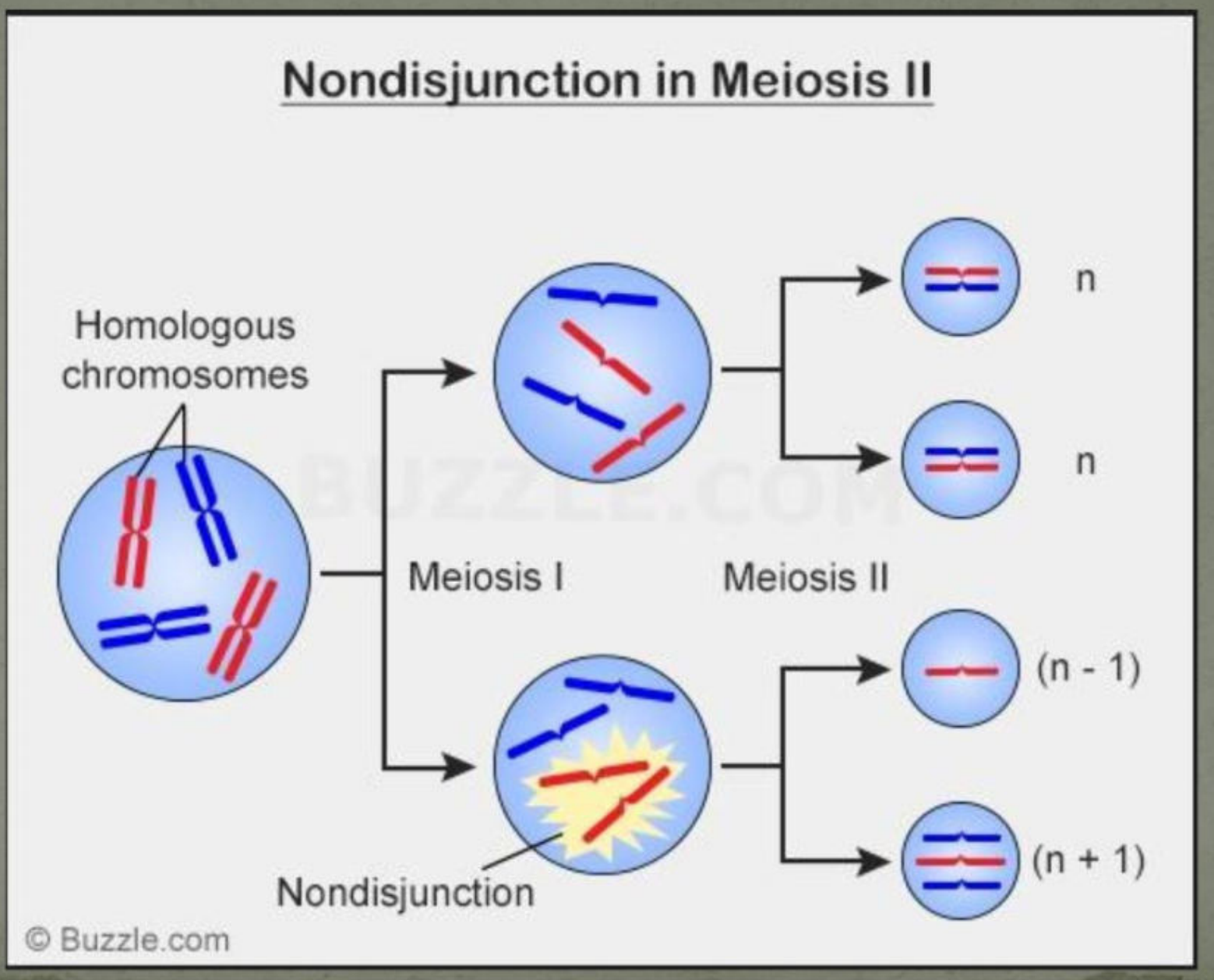


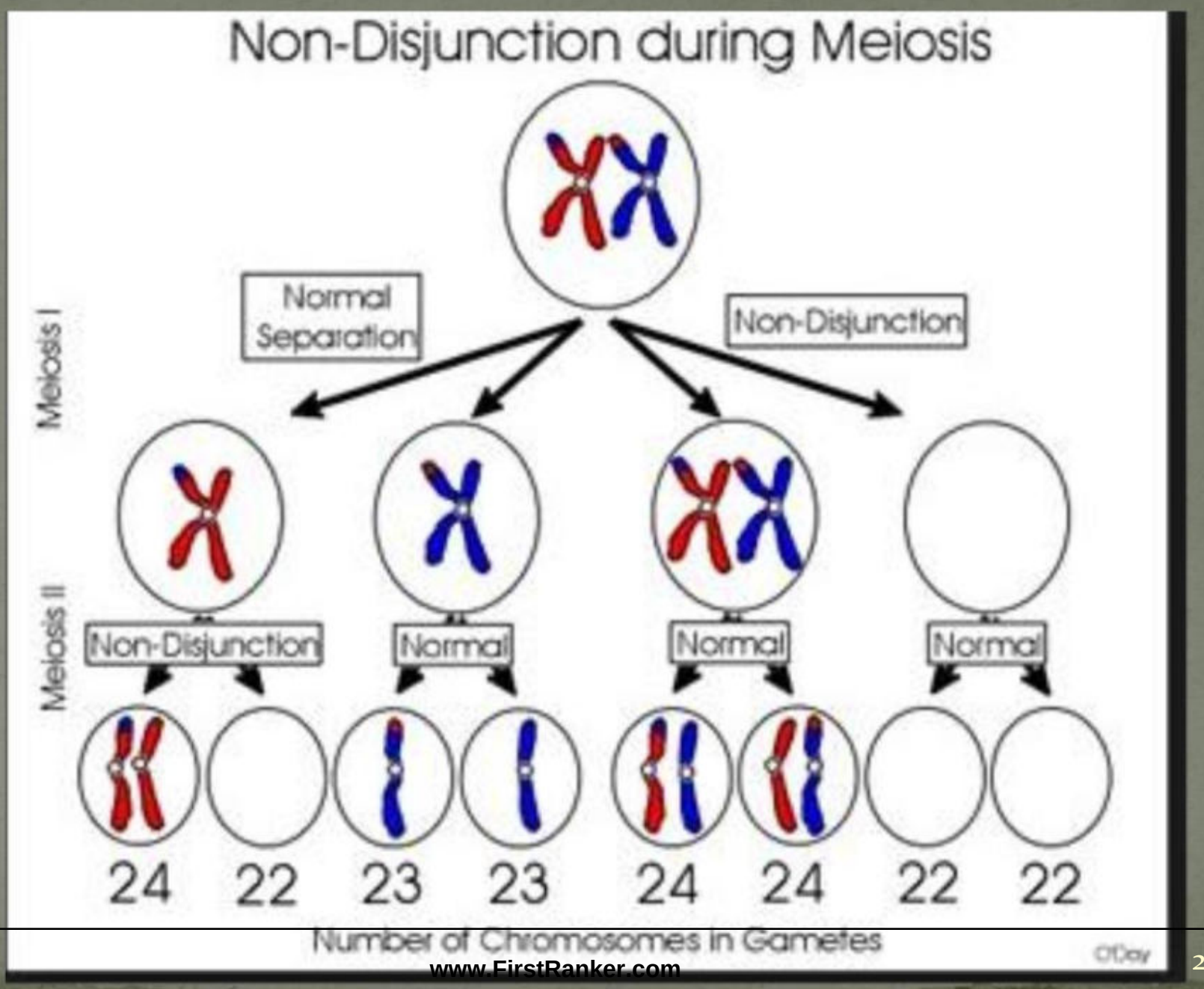


Nondisjunction

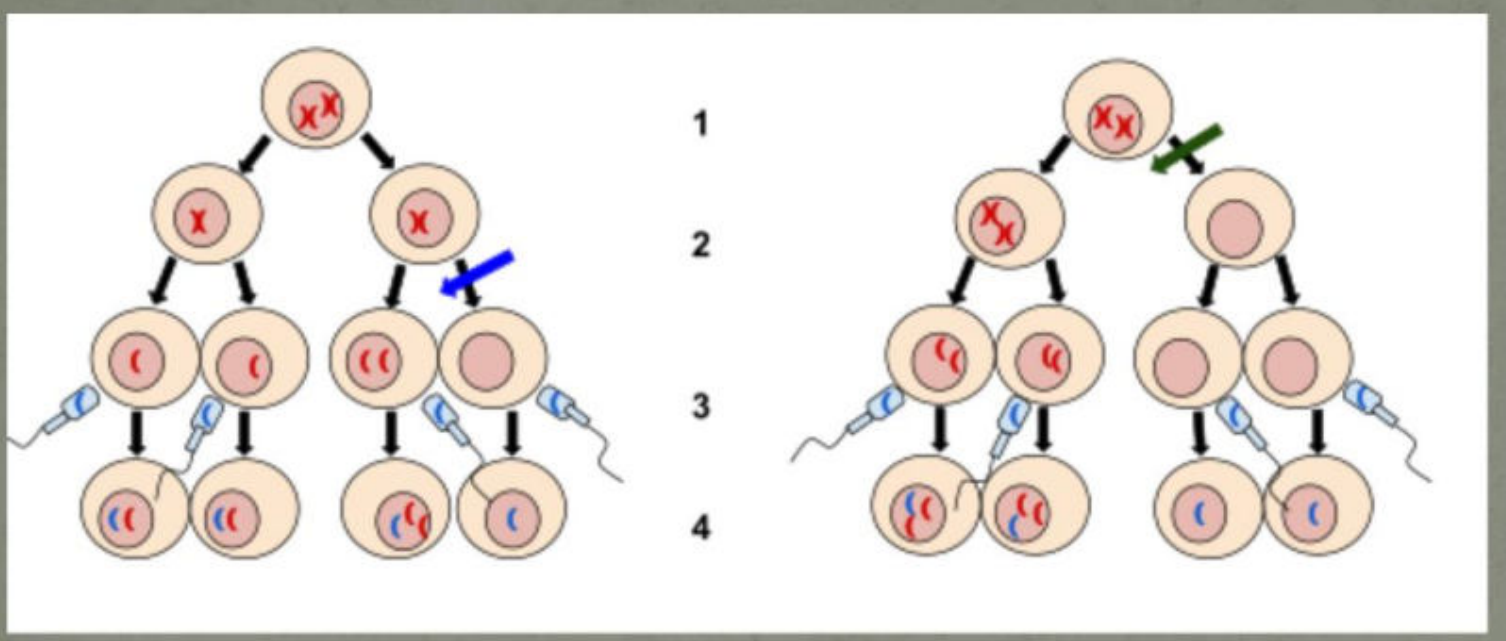








Aneuploidy and Polyploidy



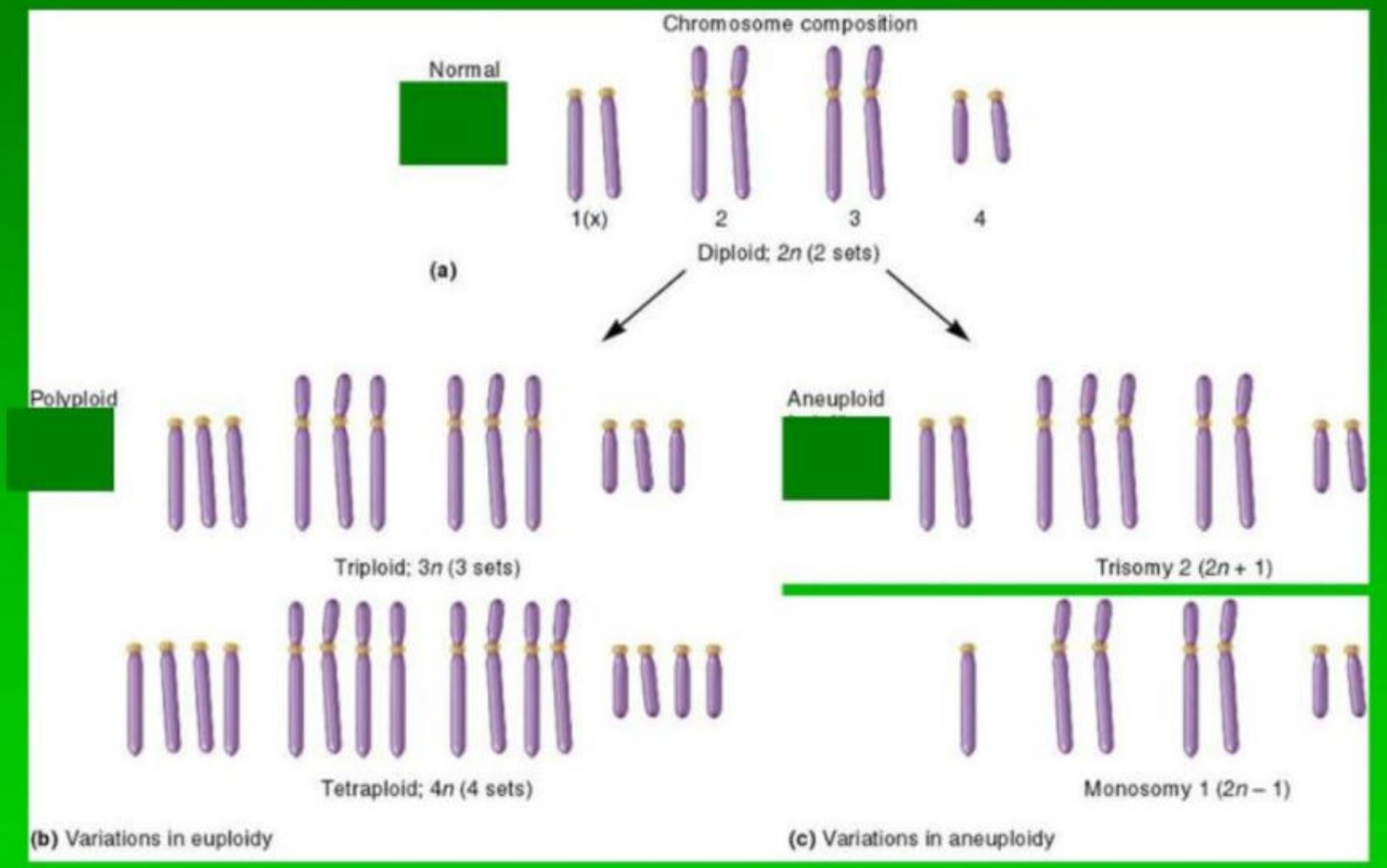
Aneuploidy

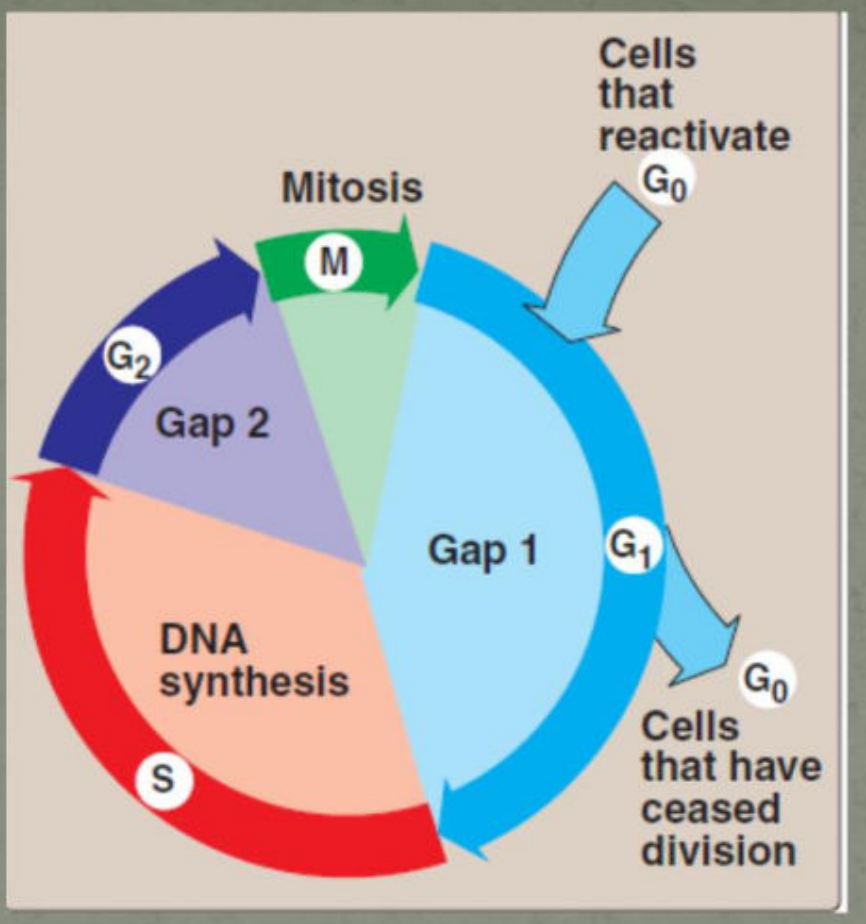
2n+1= Trisomy

2n-1= Monosomy

2n-2= Nullisomy

Polyploidy v Aneuploidy

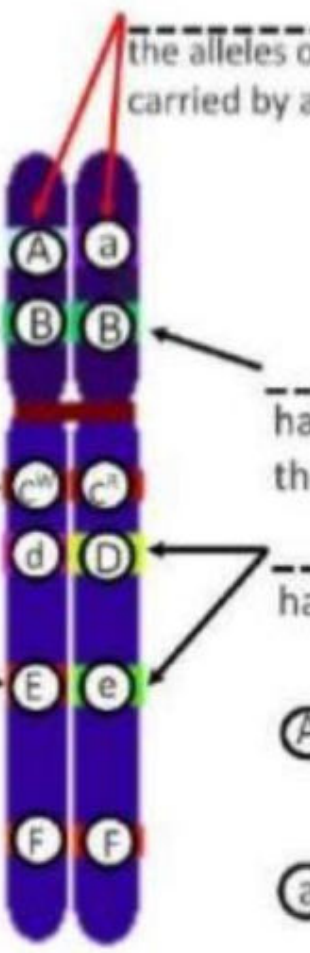




The eukaryotic cell cycle

Definitions of Theoretical Genetics:

What are the terms to match the labels and the definitions?
(get them from your syllabus)



_____ the alleles of a gene carried by an organism

_____ the expression of the gene (e.g. the characteristic - blue/brown eyes, protein on/off etc.)

_____ the specific position of a gene on a chromosome

_____ having two copies of the same allele

_____ pairs of alleles which are both expressed when present

_____ having two different alleles

_____ if an allele codes for a genetic disorder, a person with one recessive faulty allele is known as a carrier - they carry the trait but do not express the disorder.

A _____ expressed whether heterozygous (AA) or homozygous (Aa)

a _____ expressed only when in the homozygous state (aa)

Word Bank

genotype, phenotype, gene locus, codominant, homozygous, heterozygous, dominant allele, recessive allele, carrier