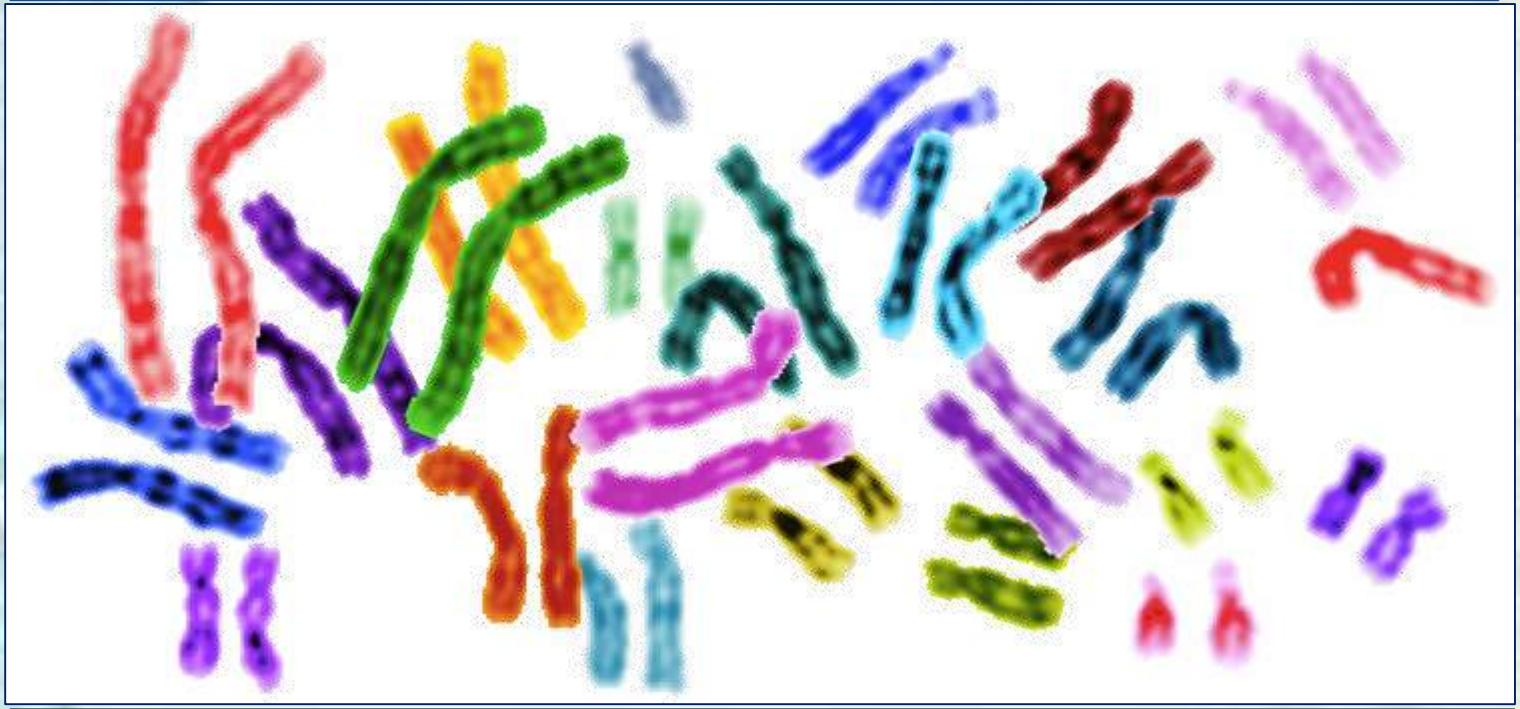
Numerical Chromosomal Variations



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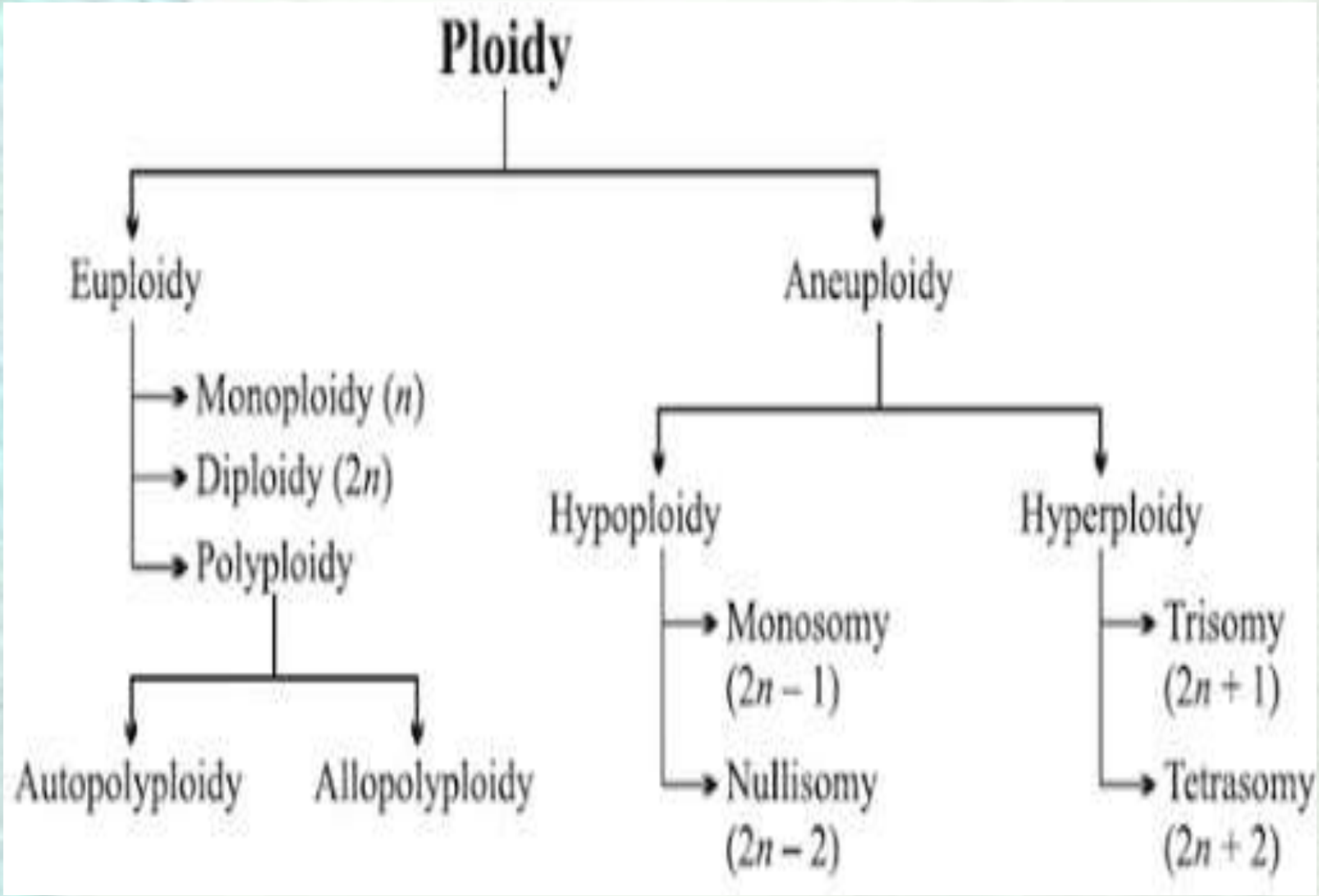
Introduction

✓Each species has a characteristic chromosome number, such as 46 chromosomes for a typical human body cell.

✓ In organisms with two full chromosomes sets, such as humans, this number is given the name 2n.

✓When an organism or cell contains 2n chromosomes (or some other multiple of n), it is said to be **euploid**, meaning that it contains chromosomes correctly organized into complete sets (*eu*- = good).

✓These numbers represent what is known as our ploidy number, which is the number of sets of chromosomes a species has.


Ploidy
Euploidy
Aneuploidy
Monoploidy (n)
Diploidy (2n)
Polyploidy
Autopolyploidy
Allopolyploidy
Hypoploidy
Hyperploidy
Monosomy
(2n – 1)
Nullisomy
(2n – 2)
Trisomy
(2n + 1)
Tetrasomy
(2n + 2)

✓**Aneuploidy** - the abnormal condition were one or more chromosomes of a normal set of chromosomes are missing or present in more than their usual number of copies

✓**Monoploidy** - the loss of an entire set of chromosomes

✓**Euploidy** - an entire set of chromosomes is duplicated once or several times

The different conditions of aneuploidy are:

✓**Nullisomy** - the loss of both pairs of homologous chromosomes; individuals are called nullisomics and their chromosomal composition is 2N-2

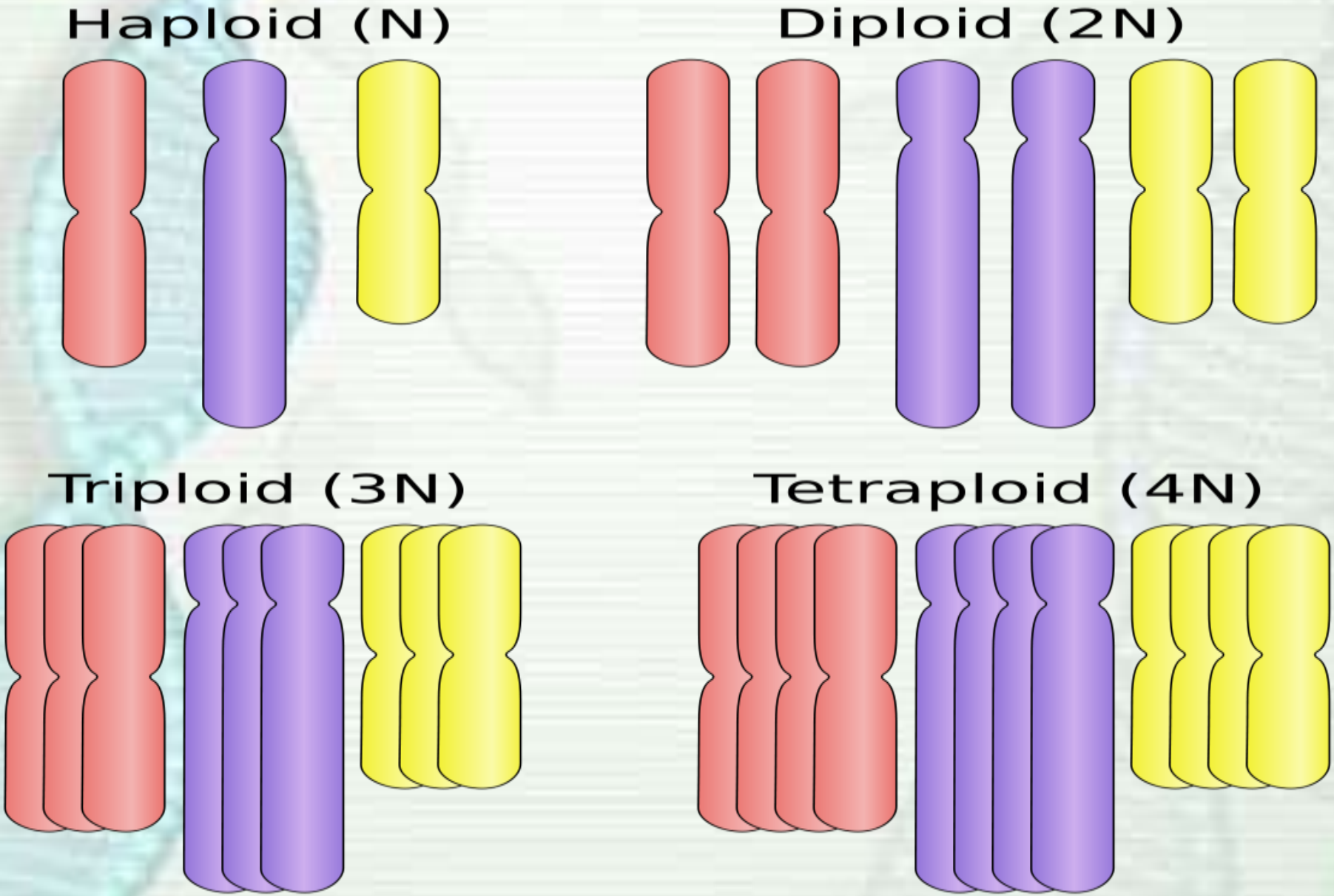
✓**Monosomy** - the loss of a single chromosome; individuals are called monosomics and their chromosomal composition is 2N-1

✓**Trisomy** - the gain of an extra copy of a chromosome; individuals are called trisomics and their chromosomal composition is 2N+1

✓**Tetrasomic** - the gain of an extra pair of homologous chromosomes; individuals are called tetrasomics and their chromosomal composition is 2N+2

✓In addition to these conditions, more than one pair of homologous chromosomes may be involved.

✓For example, a **double monosomic** is missing one chromosome from each of two pair of homologous chromosome (designated 2N-1-1), and a **double tetrasomic** contains an extra pair of two pairs of homologous chromosomes (2N+2+2).


Haploid (N)
Diploid (2N)
Triploid (3N)
Tetraploid (4N)

NORMAL CHROMOSOME #

$2n = 46$

EUPLOID

MONOSOMY

missing chromosome

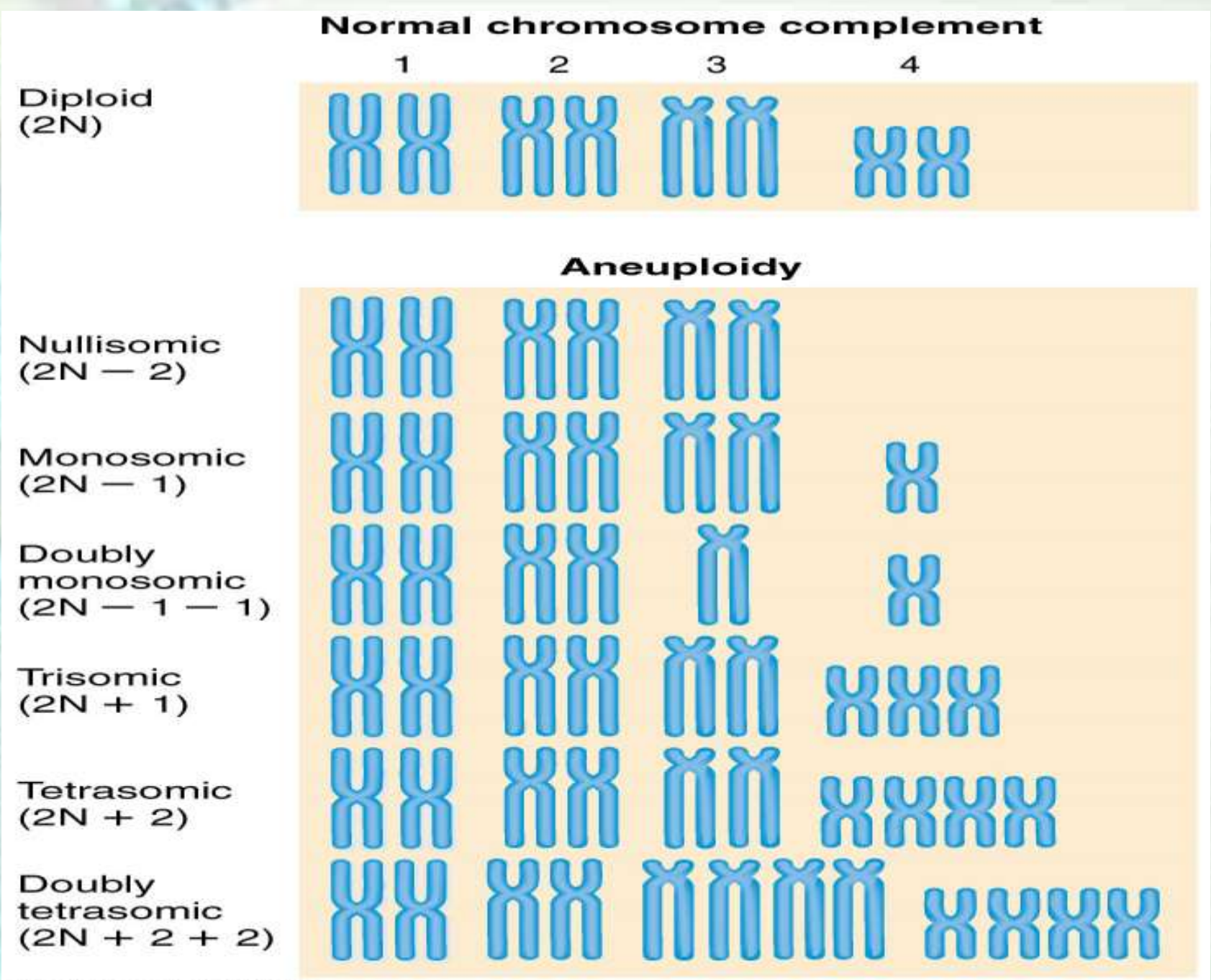
$2n - 1 = 45$

TRISOMY

extra chromosome

$2n + 1 = 47$

ANEUPLOID



✓Changes in chromosome number can occur by the addition of all or part of a chromosome (aneuploidy), the loss of an entire set of chromosomes (monoploidy) or the gain of one or more complete sets of chromosomes (euploidy).

✓Each of these conditions is a variation on the normal diploid number of chromosomes.

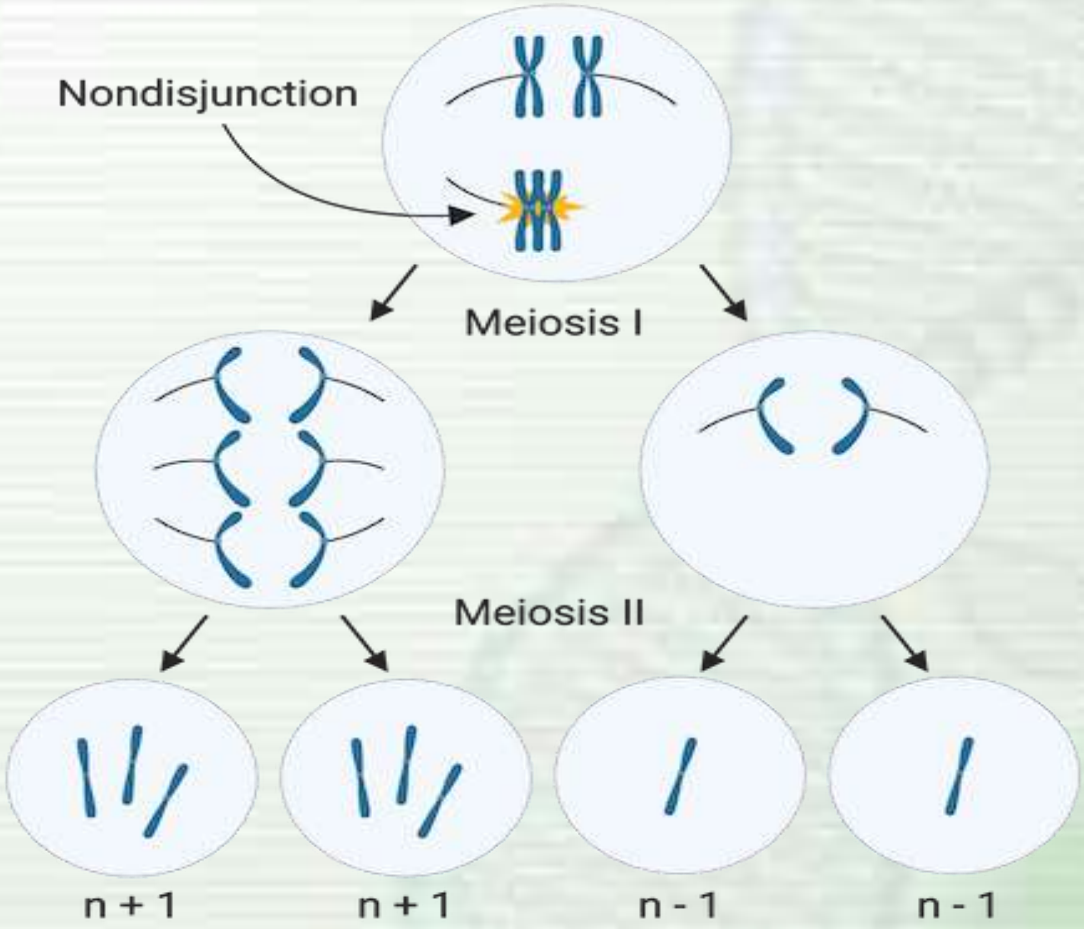
✓As you would expect each of these can have drastic effects on phenotypic expression.

Nondisjunction of chromosomes

✓Disorders of chromosome number are caused by **nondisjunction**, which occurs when pairs of homologous chromosomes or sister chromatids fail to separate during meiosis I or II (or during mitosis).

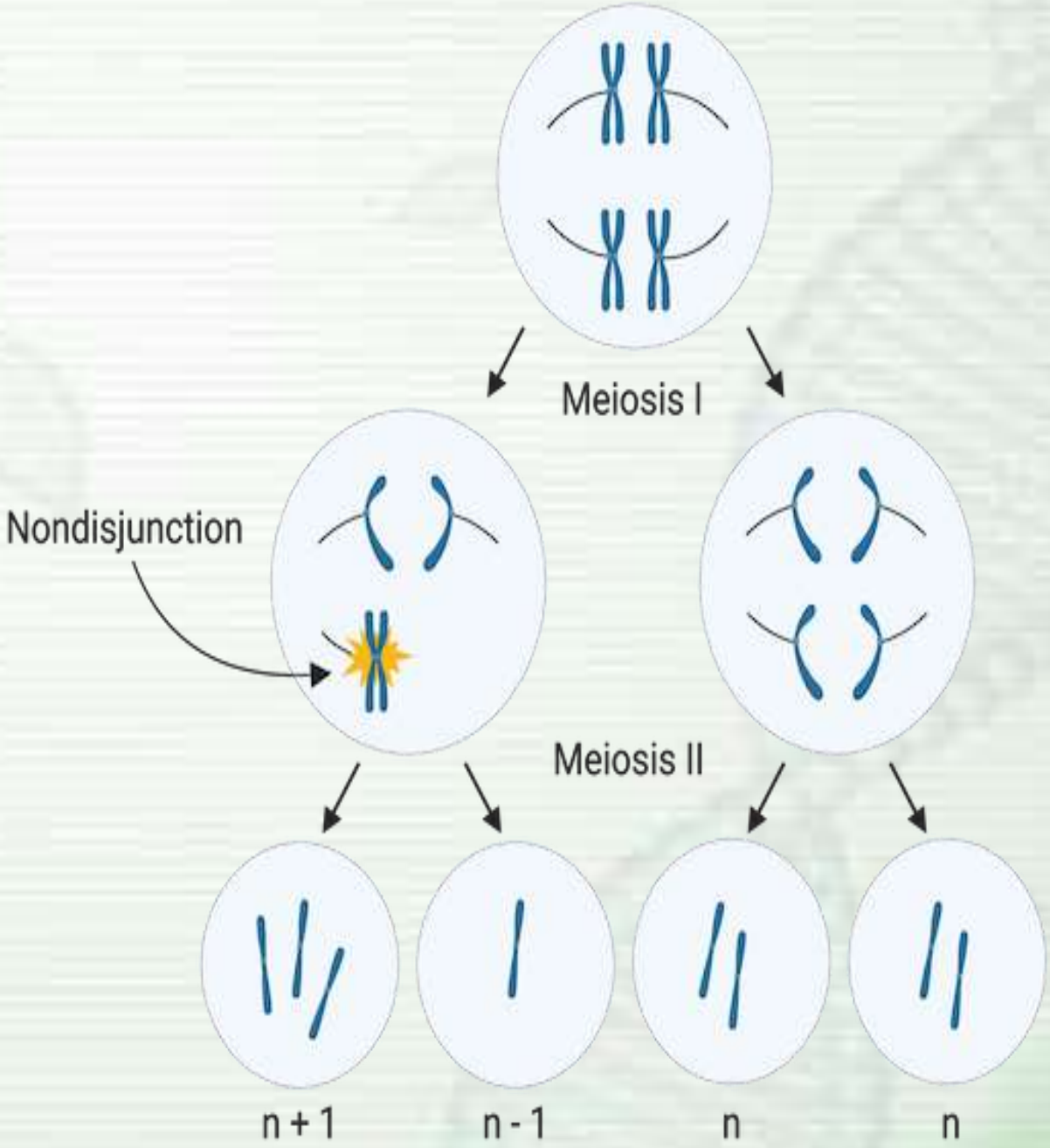
Meiosis I.

The diagram below shows how nondisjunction can take place during meiosis I if homologous chromosomes don't separate, and how this can lead to the production of aneuploid gametes



Meiosis II

Nondisjunction can also happen in meiosis II, with sister chromatids (instead of homologous chromosomes) failing to separate. Again, some gametes contain extra or missing chromosomes:



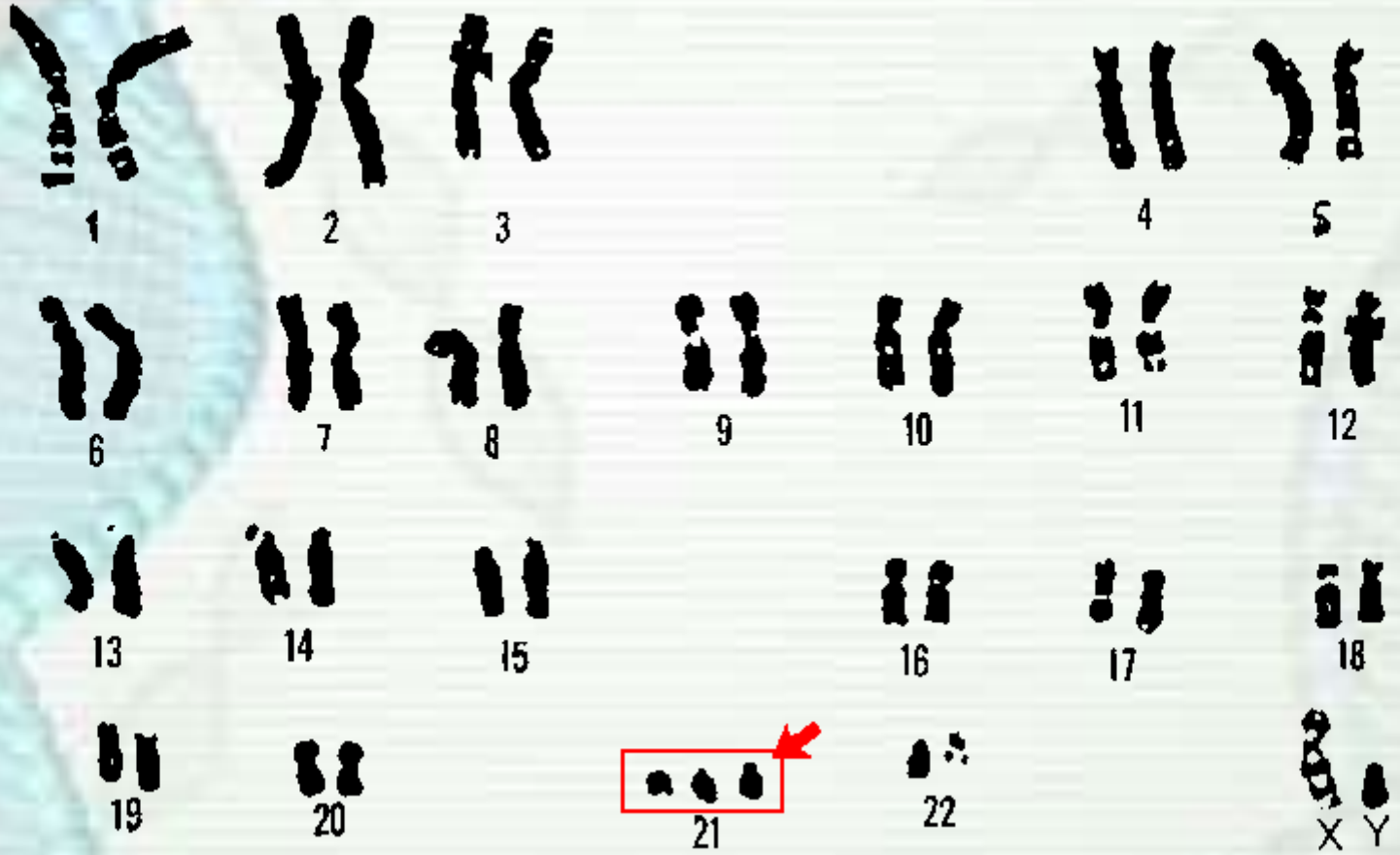
Genetic disorders caused by aneuploidy

✓Human embryos that are missing a copy of any autosome (non-sex chromosome) fail to develop to birth. In other words, human autosomal monosomies are always lethal.

✓That's because the embryos have too low a "dosage" of the proteins and other gene products that are encoded by genes on the missing chromosome.

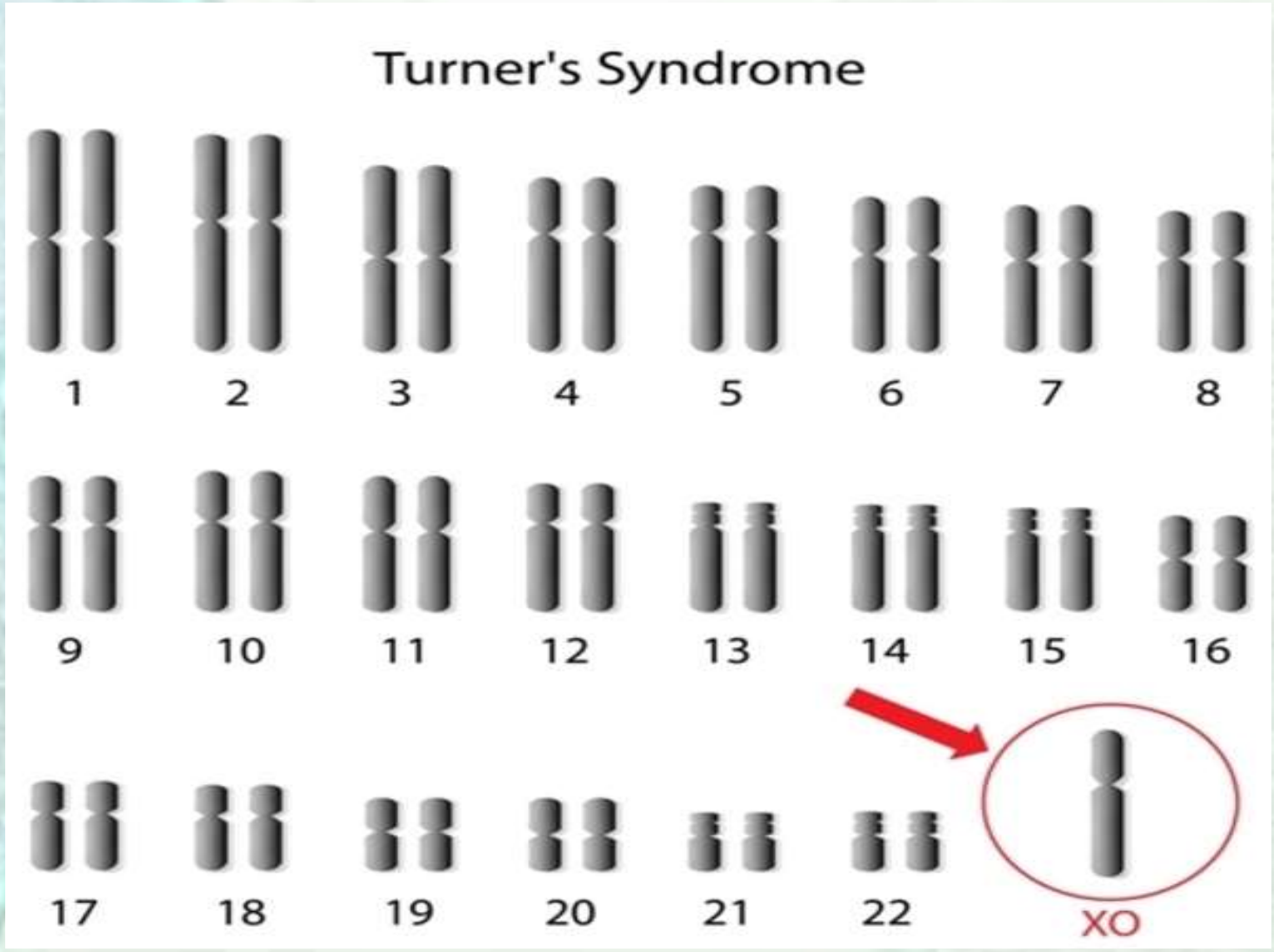
✓The most common trisomy among embryos that survive to birth is **Down syndrome**, or **trisomy 21**.

✓People with this inherited disorder have short stature and digits, facial distinctions including a broad skull and large tongue, and developmental delays.

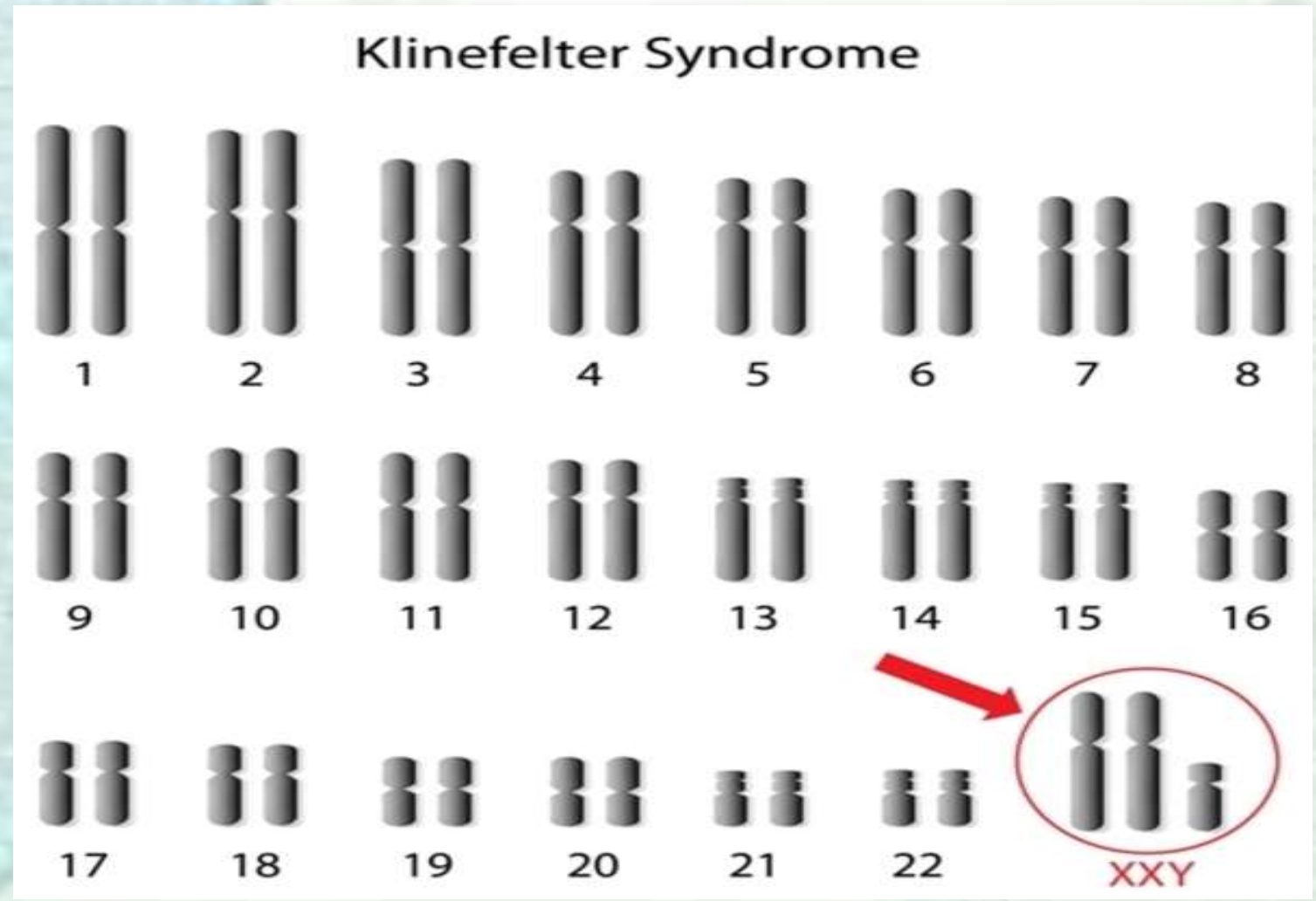


Karyotype, of the chromosomes, from a person with Down syndrome, showing the characteristic three copies of chromosome 21

Turner syndrome: XO; female (Monosomy)



Klinefelter syndrome: XXY, or XXXY, or XXXXY male (Trisomy/tetrasomy)



THANK YOU