

Number of Chromosomes

Introduction:

During the cell cycle, numerous issues can arise. Some such mistakes will cause only some genes to be affected while others will alter the total number of chromosomes in a cell. If the resulting change in chromosome number is not noticed during a checkpoint in the cell cycle, the cell can continue to exist and replicate. This will result in more cells with this issue.

Humans are diploid organisms. This means they have two full copies of every chromosome and thus two copies of every gene. The number of chromosomes can be denoted as $2n$, where 2 stands for how many copies of each chromosome there are and n is the number of unique chromosomes. Humans have twenty-three unique chromosomes (the n number) and typically two copies of each, so most humans have 46 total chromosomes.

Euploidy:

Euploidy means having a typical chromosome number. For humans, this means having exactly two copies of every chromosome because we are diploid. Because not all organisms are diploid, not all organisms will be considered to have euploidy if they have two copies of every chromosome. Some frogs, for instance, have more than two sets of every chromosome. Bacteria without plasmids have one chromosome and only one copy of every gene.

Aberrant Euploidy:

In some cases, an organism may survive with an extra full set of chromosomes. In this case, the organism would have an extra copy of every chromosome. This is rare in human cells but fairly common in plants such as wheat and fruit-bearing plants. Aberrant euploidy is the term for the complete loss or gain of a full set of chromosomes. To write the number of chromosomes of an organism with aberrant euploidy, the coefficient in front of the n changes. For example, if an organism has three copies of every chromosome, $3n$ could be used to represent how many chromosomes they have.

Aneuploidy:

Not all chromosome mutations affect the full set of chromosomes. Aneuploidy is when there is a loss or gain of just one or a few chromosomes rather than a full set of chromosomes. Trisomy is an example of aneuploidy; an individual who has trisomy has one extra copy of only a single chromosome. In humans, the most common case of aneuploidy is trisomy 21, a condition where an individual has an extra copy of the 21st chromosome. An individual with this condition has three copies of chromosome 21 and the typical two copies of all of the other chromosomes. These chromosomes can be expressed as $2n+1$ where the 2 represents the person having two copies of each chromosome and the +1 representing that they have 1 extra chromosome.

Practice:

Complete the following chart to test your knowledge of euploidy, aberrant euploidy, and aneuploidy.

Image or Example	Description	n	Name
XX XX XX XX	Typical number of chromosomes		Euploidy
XX XX XX XX <u>XX</u> XX XX XX			Aberrant euploidy
<u>XXX</u> XX XX XX			Aneuploidy
A human has exactly one extra copy of chromosome number 21.			
Typically, cherry trees have two copies of every chromosome, but this cherry tree has three.			
Challenge: A human man makes a diploid sperm cell. It fertilizes a typical egg. Complete this chart about the zygote that is produced.			
Your example (written or drawn):		2n-3	

Non-disjunction:

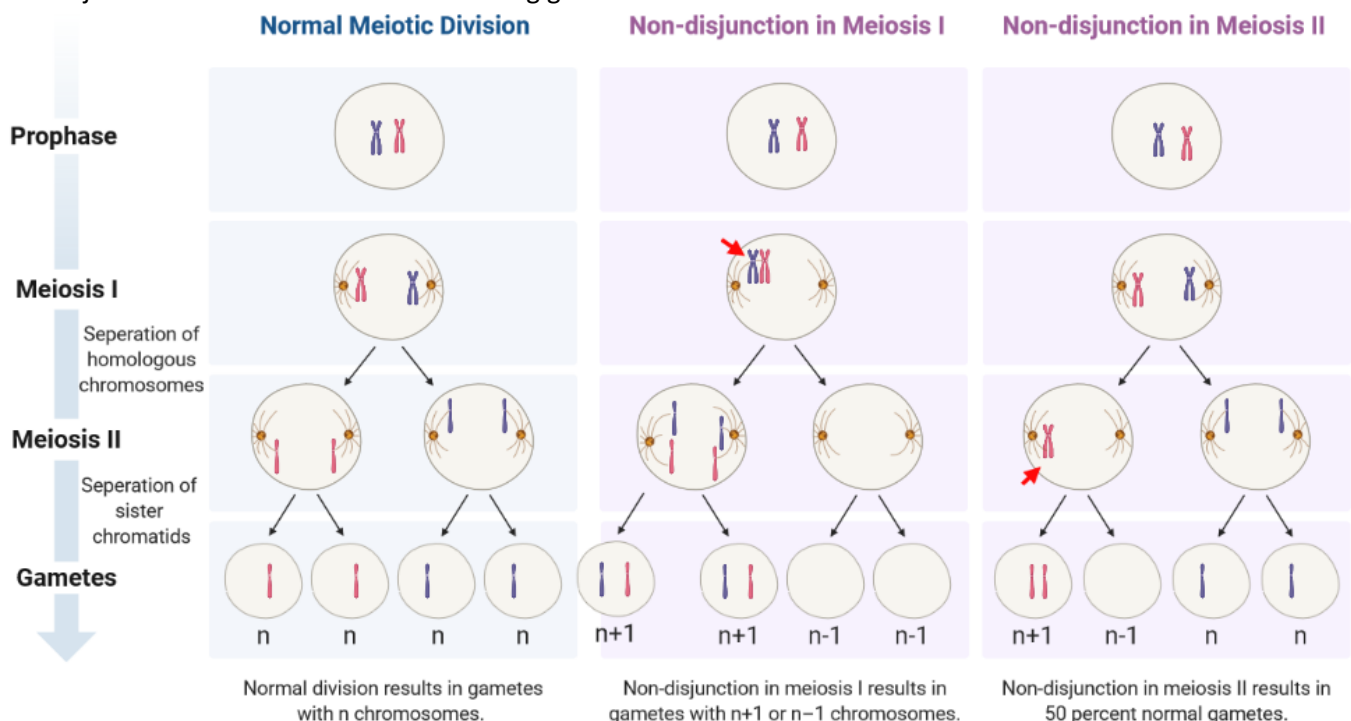
Non-disjunction is a process that can result in aneuploidy. During the process of meiosis, homologous chromosomes are supposed to line up on the metaphase plates. During the first cell division called meiosis I, the homologous chromosomes separate into separate cells leaving the two resulting cells each with one of each chromosome. The second cell division, called meiosis II, occurs on these newly formed cells. In this division, sister chromatids separate resulting in four haploid daughter cells each with exactly one copy of every gene.

Sometimes, these separations do not occur. The process of the homologues or sister chromatids not separating correctly is called non-disjunction. This can alter chromosome number by having one more or less chromosome in some of the resulting gametes. When fertilization occurs, the zygote will have aneuploidy.

Non-disjunction in meiosis I results in exactly half of the produced gametes having a lack of one chromosome and the other half having two similar (homologous) copies of a chromosome. In the first division, one cell has two homologous chromosomes and the other lacks any copies of that chromosome. When both of these cells undergo the second round of division, two out of four gametes have one copy of the affected chromosome from the individual's parents (two chromatids that are not sisters but instead homologues) while the other two gametes lack any copies of the affected chromosome.

Non-disjunction in meiosis II results in two normal gametes containing one copy of the chromosome (n), one gamete containing zero copies of the chromosome, and one gamete containing two copies of the same chromosome (n+1). The first round of cell division goes normally; the two homologues separate into separate cells. The second round of cell division is where the mistake occurs. The sister chromatids do not separate and thus the resulting gametes have aneuploidy.

The figure below illustrates both non-disjunction in meiosis I and meiosis II. Please compare where non-disjunction occurs and what the resulting gametes are.



Practice:

State whether each example of aneuploidy is due to non-disjunction in meiosis I or meiosis II. Then, write or draw two examples of your own. For reference, please use the following:

C_M1- Chromosome 1 from Mom's DNA; **C_D1- Chromosome 1 from Dad's DNA**

Description or image	Non-disjunction in Meiosis 1 or 2?
Homologous chromosomes did not separate	
C _M 1 C _M 2 C _M 3 C _M 4C _M 4	
C _M 1 C _M 2 C _M 3 C _M 4C _D 4	
C _M 1 C _D 2 C _M 3C _M 3 C _D 4	
Sister chromatids did not separate	
There are two copies of chromosome 5 from mom in a sperm	
Mom and Dad's chromosome 2 stuck together when an egg was made	
A sperm lacks a chromosome # 4.	
Your example:	1
Your example:	2