Biology 121-224
Lecture 2

Genetics Terminology – Chromosomes and Genes

Source: Scientific American

Genetics Study Survey

Dear BIOL121 - section 224 students,
you are warmly invited to complete our Genetics Study Survey, which you can access at the following URL: https://ubc.ca1.qualtrics.com/jfe/form/SV_d1ho1T7gwyriC34

- The consent to let us use your responses for the study is at the end of the survey (Q. 39)
- Participation credit is allocated by your instructor for completion regardless of whether you choose to give consent for the study :)
- Your instructor won't know your answers and won't know whether you provided consent for the study (only the project PI and Co-PI will know).
- When entering your name and student ID at the (almost) end of the survey, please ensure the information is accurate!
- The survey will 'expire' on **Thursday, Jan 19th, 11:59 pm** Vancouver time.

THANK YOU!
Questions or concerns, or interested in further study participation?
Please email Pam at: kalas@zoology.ubc.ca
(include "genetics survey" in the subject)

Office hours – now posted

BIOLOGY 121 – Office Hours - Spring 2023

Office hours are in person in BIOL1114 (light green) unless otherwise noted

For Zoom office hours: Join Zoom Meeting (yellow)

https://ubc.zoom.us/j/68482751346?pwd=b2FLbUJIWjJ4ckJWbnhTdnBmQTlJQT09

Meeting ID: 684 8275 1346 Passcode: 796991

Time	Monday	Tuesday	Wednesday	Thursday	Friday
9:00 – 9:30 am				Nadia (9-11)	
9:30 – 10:00 am					
10:00 – 10:30 am	Melissa		Risa	Haley (on Zoom)	Brett
10:30 – 11:00 am					
11:00 – 11:30 am		Brett			
11:30 –12:00 pm					
12:00 – 12:30 pm					
12:30 – 1:00 pm		Rory			
1:00 – 1:30 pm				Kira	
1:30 – 2:00 pm					
2:00 – 2:30 pm			Lynn	Lynn	
2:30 – 3:00 pm					
3:00 – 3:30 pm	Christie		Kung-Ping		Ema
3:30 – 4:00 pm					
4:00 – 4:30 pm		Ema			
4:30 – 5:00 pm					
5:00 – 5:30 pm			Ruby		
5:30 – 6:00 pm					

Any questions before we get started?

Genetics Unit

Genetics is the branch of biology that studies genes, their role in inheritance (i.e. how certain genetic traits or conditions are passed from one generation to the next) and genetic variation

Genomics (beyond the scope of BIOL121) – is a newer term that describes the study of all of an individuals' genes including how genes interact with each other and with the environment. Genomics plays an important in the study of human conditions that are caused by a combination of genetic and environmental factors (e.g. cancer, heart disease).

In BIOL121, we will not be addressing interactions between genes.

Topics that we will be covering in the Genetic Unit:

- **How genetic information flows from parent cells to daughter cells and from one generation to the next:**
  - cell cycle, mitosis, meiosis (including how genetic variation arises)
- **Patterns of inheritance**
  - Mendelian genetics (autosomal dominant and recessive traits)
  - Beyond Mendel (codominance and X-linked traits)
- **How to determine the most likely mode of inheritance for a trait**
  - Genetic crosses (if you can breed many individuals)
  - Pedigrees (humans)

In the Evolution Unit: **Population genetics**

- Hardy-Weinberg Equilibrium

Genetics and medicine

How genetics influence immunity in patients with type 1 diabetes

Date: June 7, 2022

Gene Variation May Be Early Indicator for Gastric Cancer

June 6, 2022 — Researchers are hoping to catch stomach cancer before it develops in at-risk patients. Researchers identified a genetic variation that could help identify when patients with Helicobacter pylori are ...

CRISPR technology improves Huntington's disease symptoms in models

Date: December 12, 2022

In ironic twist, CRISPR system used to befuddle bacteria

Date: November 7, 2022

Combining genetics and brain MRI can aid in predicting chances of Alzheimer's disease

Date: June 29, 2022

Source: Simon Fraser University

Summary: Researchers are studying how a combination of genetics and brain MRIs may be used to predict the chances of developing Alzheimer's disease in the future.

A sample of papers published in the last 6 months.

Genetics may predict bladder cancer immunotherapy response

Date: August 4, 2022

Genetics & Wildlife Conservation

CRISPR helps researchers uncover how corals adjust to warming oceans

Date: December 21, 2020

Elephant genetics guide conservation

Date: November 19, 2020

Source: Penn State

Summary: A large-scale study of African elephant genetics in Tanzania reveals the history of elephant populations, how they interact, and what areas may be critical to conserve in order to preserve genetic diversity of the species.

Saving **wildlife** with forensic **genetics** -- ScienceDaily

ScienceDaily › releases › 2011/06

Jun 10, 2011 **...** Using forensic **genetics** techniques, the University of Arizona's **Conservation Genetics** Lab is working to protect wild animals and catch the ...

Labeled Featured ...

Massive **genetic** study of humpback whales to inform **conservation** ...

ScienceDaily › releases › 2017/01

Date: January 9, 2017; Source: **Wildlife Conservation** Society; Summary: Scientists have published one of the largest **genetic** studies ever conducted on the ...

Labeled Featured ...

Open Access

Select Language

Translator Disclaimer

1 October 2007

Estimation of population size for wolverines *Gulo gulo* at Daring Lake, Northwest Territories, Using DNA based mark-recapture methods

Robert Mulders, John Boulanger, David Paetkau

Author Affiliations +

Wildlife Biology, 13(sp2):38-51 (2007). https://doi.org/10.2981/0909-6396(2007)13[38:EOPSFW]2.0.CO;2

ERIC NIILER SCIENCE 01.10.2022 07:00 AM

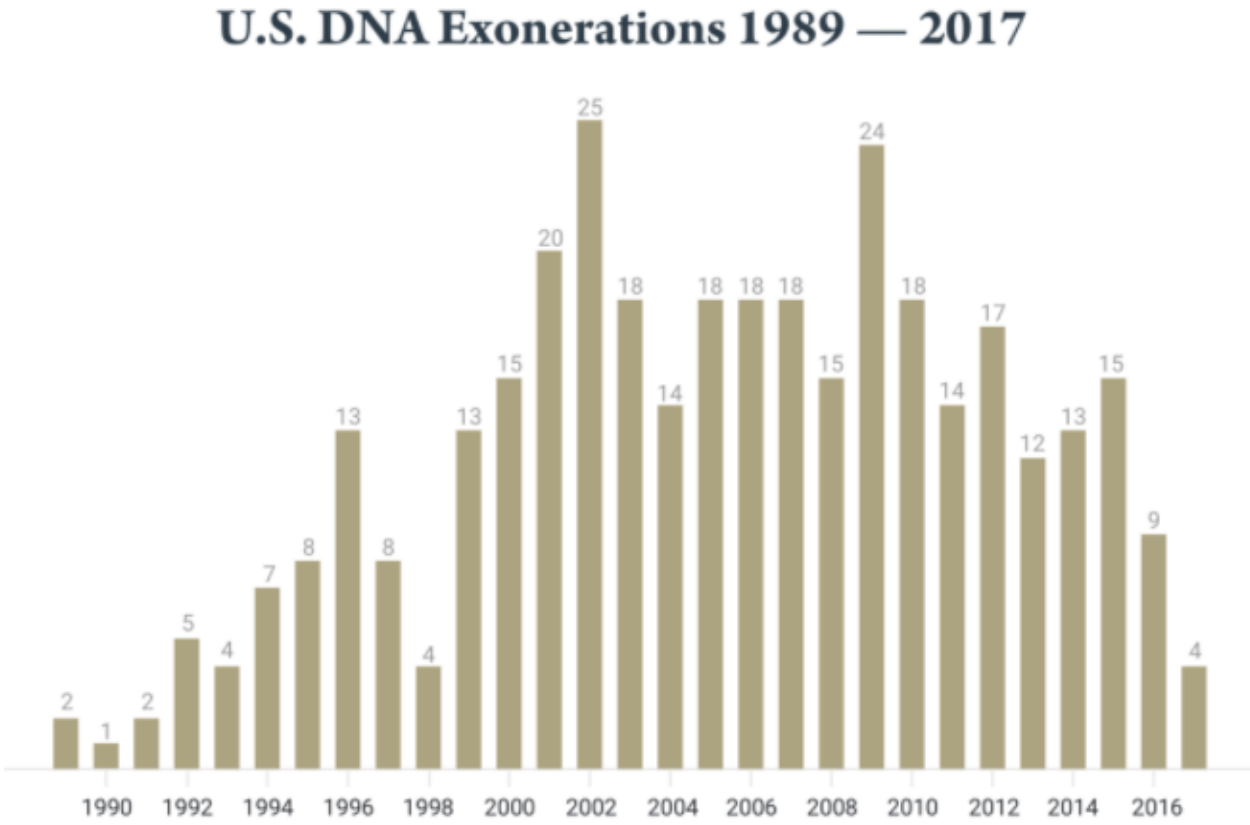
Scientists Capture Airborne Animal DNA for the First Time

Researchers filtered the air around two zoos and identified genetic material from dozens of species, a technique that could help track and conserve wildlife.

DNA evidence used to convict people who are guilt of crimes, and free innocent people that are wrongly convicted

Tanya and Jay. Murdered in 1987.

>30 years later, murdered was identified and convicted thanks to DNA evidence.



Source: Innocence Project

Genetics – joy of discovery

Feline genetics help pinpoint first-ever domestication of cats

Cat genes reveal how invention of agriculture bonded cats with people in ancient Mesopotamia, leading to worldwide feline migration with humans

Date: December 5, 2022

The octopus' brain and the human brain share the same 'jumping genes'

A new study has identified an important molecular analogy that could explain the remarkable intelligence of these invertebrates

Date: June 24, 2022

Ice Age Wolf DNA Reveals Dogs Trace Ancestry to Two Separate Wolf Populations

June 29, 2022 — An international group of geneticists and archaeologists have found that the ancestry of dogs can be traced to at least two populations of ancient wolves. The work moves us a step closer to ...

Here today, gone tomorrow: How humans lost their body hair ...

ScienceDaily › releases › 2023/01

7 days ago ... Amanda Kowalczyk, Maria Chikina, Nathan Clark. Complementary evolution of coding and noncoding sequence underlies mammalian **hairlessness**. eLife, ...

By the end of today's class..

Be familiar with the following terms:

- Chromosomes
- Chromosome structure
  - centromere
  - arm
  - loci
- Replicated vs. unreplicated chromosomes
- Sister chromatids, non-sister chromatids
- Homologous chromosomes
- Genes
- Alleles
- DNA

Autosome vs. Sex Chromosome

Ploidy

Haploid number

Genotype

- heterozygous vs. homozygous

Somatic cells (versus gametes)

iClicker Question – Are you familiar with these terms?

A. Yes, all of these terms
B. Yes, most of these terms
C. About half of these terms
D. No, I only no a few of these terms
E. No, I know none of these terms

Tips & Tricks to Succeed #2

There is a lot of terminology in genetics.

Research from the University of Madison, Wisconsin indicates that to master new terms, biology students need to encounter those term:

- at least 7 different times

- novel situations each time
  (lecture, draw, read text or watch video, quiz, worksheets, +2)

- so, if the language feels overwhelming that is completely understandable.

Tips & Tricks to Succeed #3

If you looking for a way to improve your focus go for a walk (take a notepad).

Great thinkers that would go for strolls to think things through included people such as Darwin, Beethoven, Einstein, V. Woolfe, Socrates, Aristotle, Nietzche.

- When you walk (or exercise) your muscle cells release a protein called Irisin.
- Irisin stimulates the part of the brain involved in memory and learning
- Specifically, it can increase levels of a protein involving in memory and learning (BDNF or Brain-Derived Neurotropic Factor).


Multiple Roles in Neuroprotection for the Exercise Derived Myokine Irisin

Mohammad Jodeiri Farshbaf[1] and Karina Alviña[1,2*]

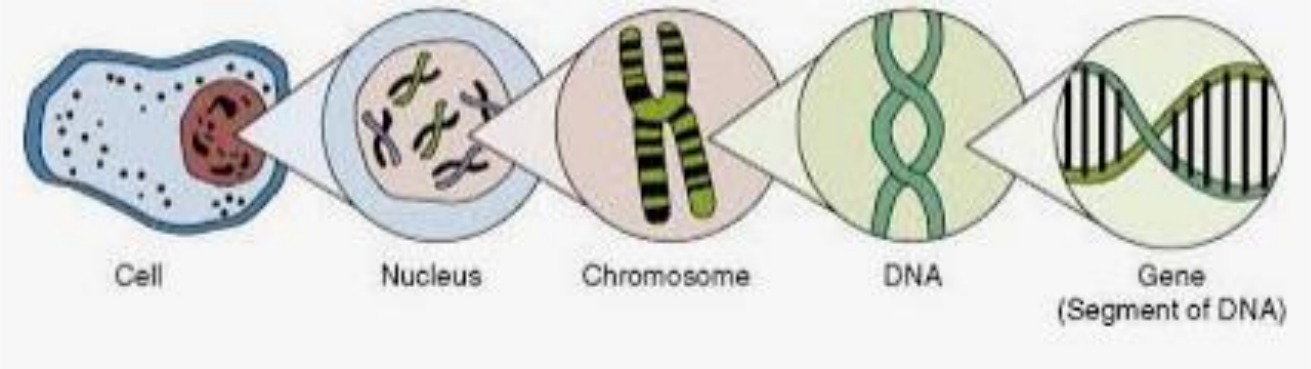

Mol Med Rep. 2019 Feb; 19(2): 1074–1082. PMCID: PM
Published online 2018 Dec 12. doi: 10.3892/mmr.2018.9743 PMID

Irisin regulates the expression of BDNF and glycometabolism in diabetic rats

Lingning Huang,[1,2] Sunjie Yan,[1,2] Li Luo,[3] and Liyong Yang[1,2]

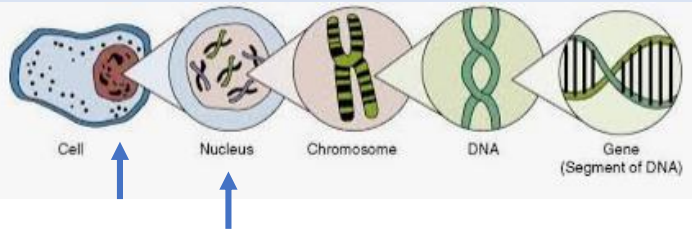
Terminology – Part 1

For this part of the lecture, I will zoom in from a somatic cell to a gene



http://www.cancermoonshotlund.com/index.php/6-what-are-genes-dna-and-proteins/

Somatic cells (versus gametes)



SOMATIC CELLS

- are all of the cells in your body other than eggs and sperm.
- skin cells, liver cells, kidney cells are examples of somatic cells

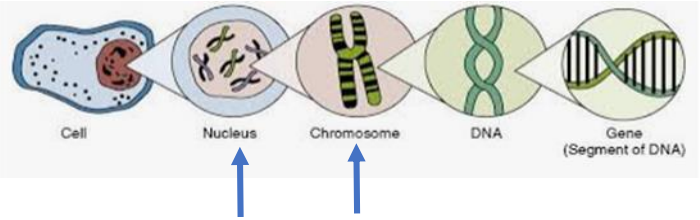
http://www.cancermoonshotlund.com/index.php/6-what-are-genes-dna-and-proteins/

Human cheek cells (Wikicommons, Mulletsrokk)

- Stained and magnified about 400X magnification

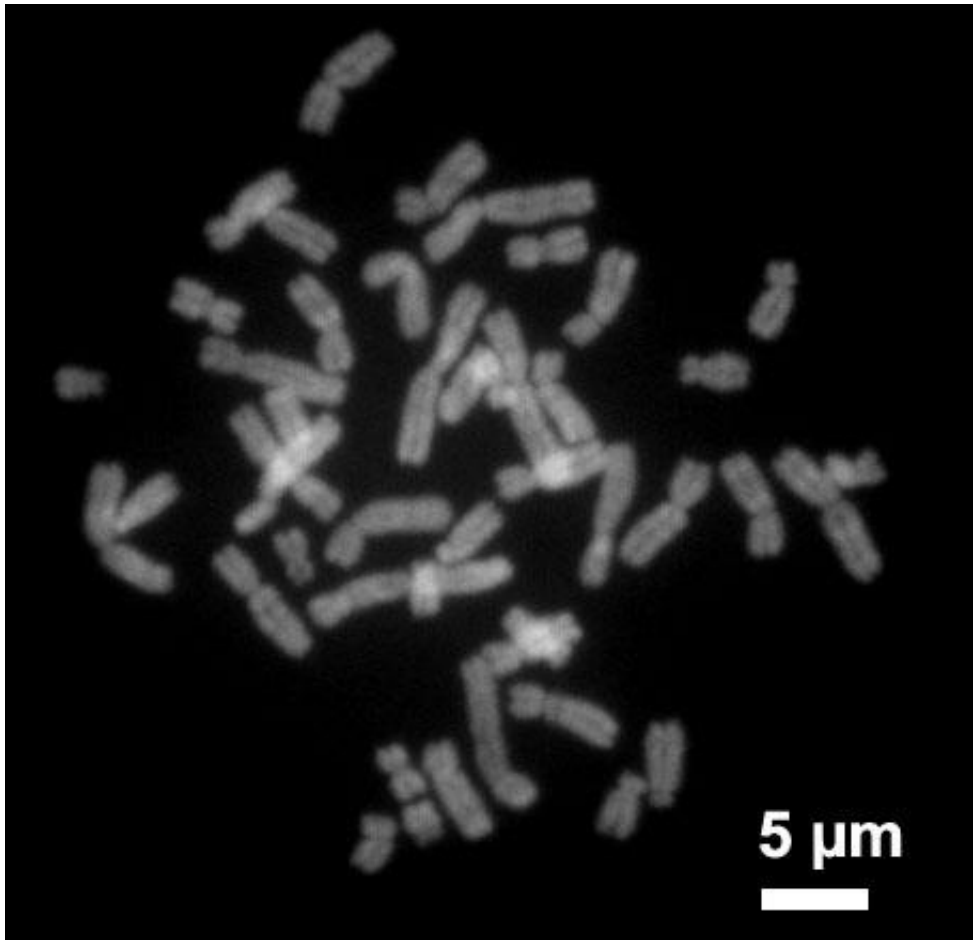
Dark oval is the NUCLEUS

CHROMOSOMES



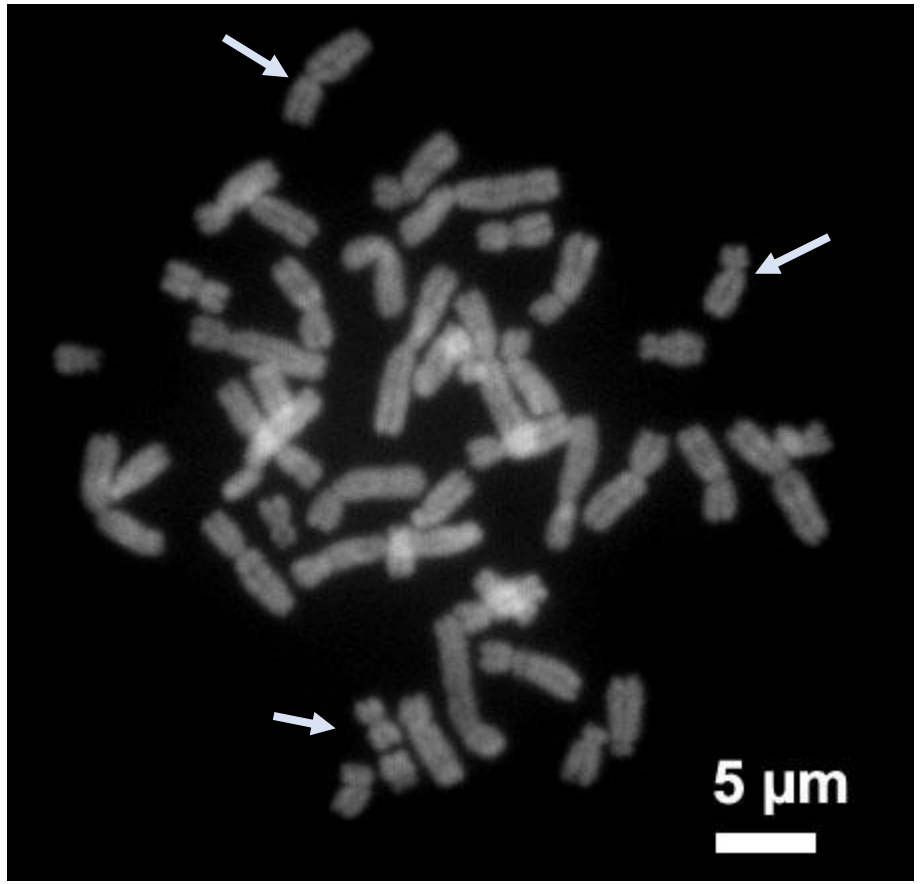
If a cell is undergoing cell division, and you had access to a light microscope, you would be able to see structures called chromosomes.

Chromosomes carry genetic information in the form of genes



https://evolutionnews.org/2016/05/human_chromosom/

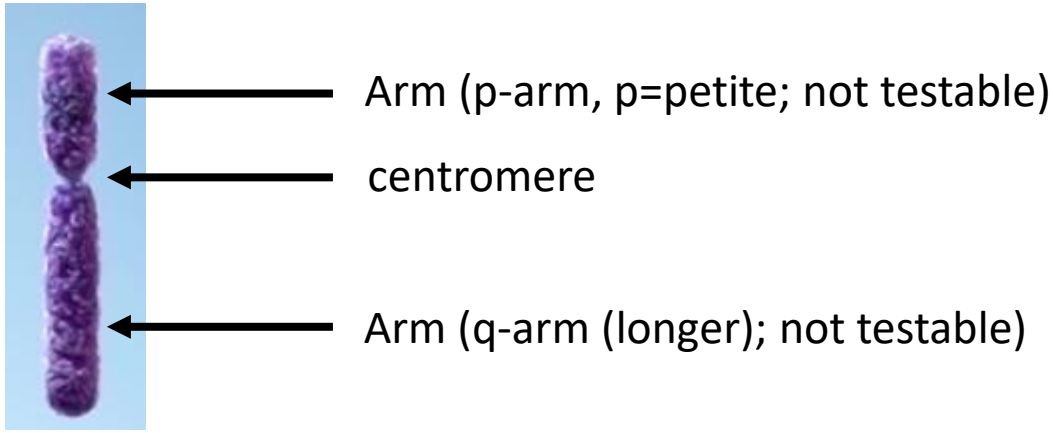
Terminology - Chromosome Structures



https://evolutionnews.org/2016/05/human_chromosom/

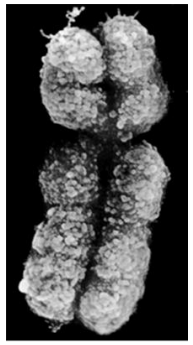
Each chromosome has a constriction point called a CENTROMERE
- specialized region that is an attachment for microtubules during mitosis and meiosis.

The centromere divides the chromosome into two sections or ARMS

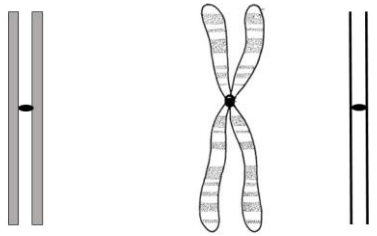


Chromosome structures

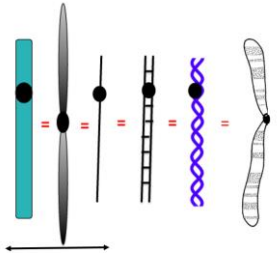
In this image the chromosomes are replicated for the purposes of cell division (X-shape)

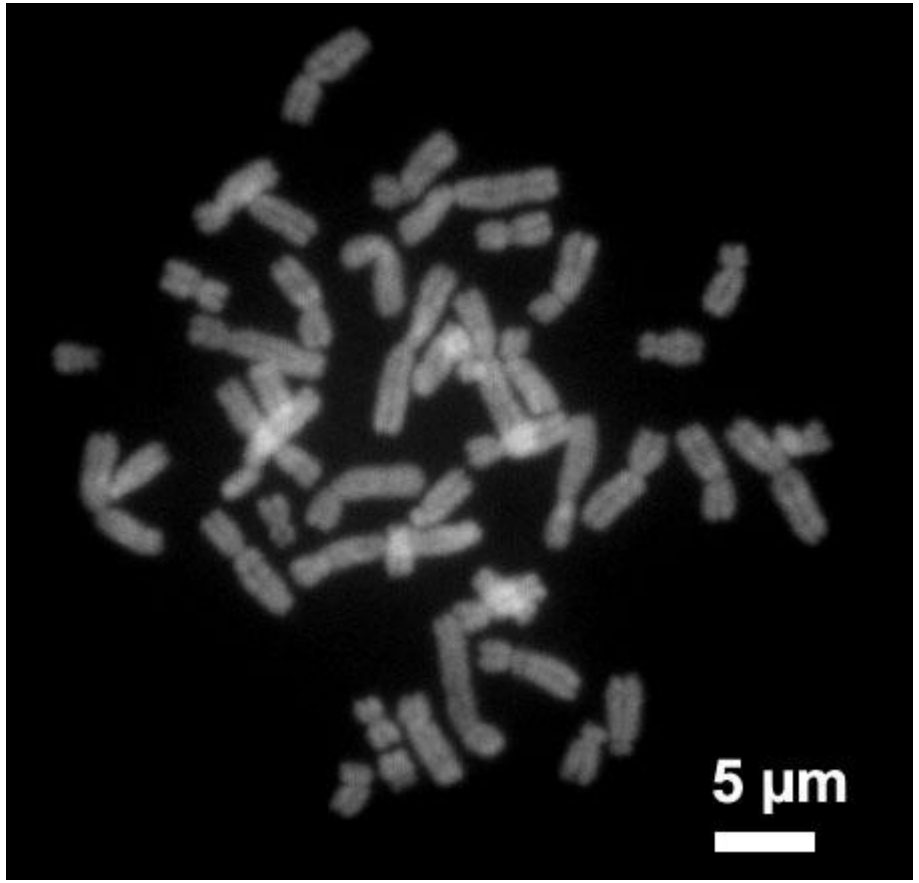


Replicated chromosome



Unreplicated chromosome

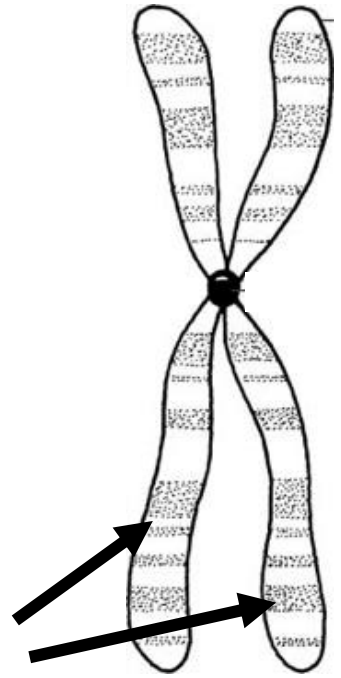




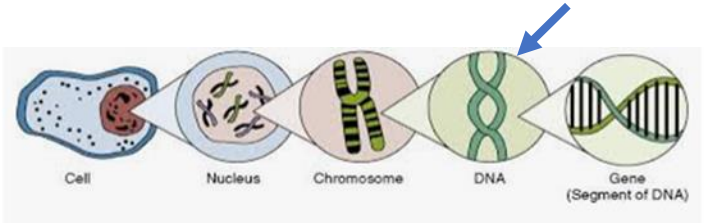
https://evolutionnews.org/2016/05/human_chromosom/

Sister Chromatids

- A replicated chromosome is composed of two SISTER CHROMATIDS that are connected at the centromere region.
- Ultimately sister chromatids will become chromosomes but not until they separate in mitosis or meiosis II.
- While attached at centromere = one chromosome.



DNA & Histone Proteins

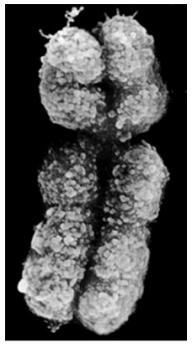


- If you could Zoom in farther, and take a closer look at a chromosome, you would see that…

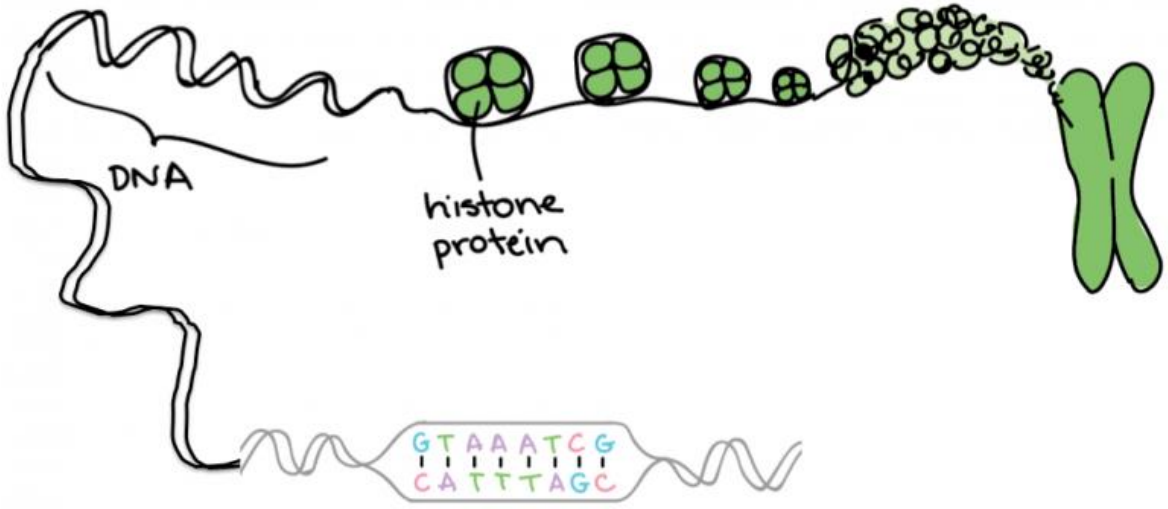
- …an UNREPLICATED chromosome is one condensed molecule of DNA, plus organizing histone proteins.

Organization is important. Your nucleus (10 um in diameter) contains about 2m of DNA

One condensed DNA molecule/proteins

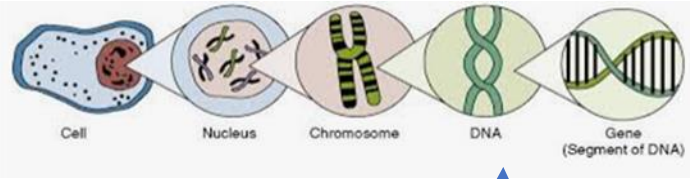


Two condensed DNA molecules/proteins



https://bio1220.biosci.gatech.edu/sex-01/2-06-dna-and-genes/

DNA (Deoxyribonucleic acid)



DNA is a double-stranded molecule that looks like a steep spiral staircase

https://www.nature.com/scitable/topicpage/dna-is-a-structure-that-encodes-biological-6493050/

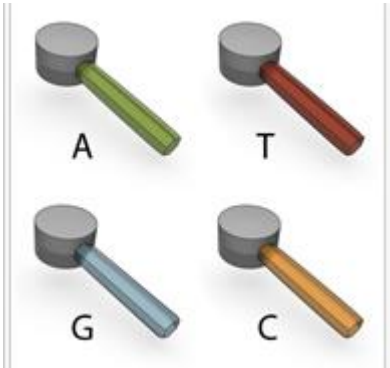
DNA structure not testable, but may help you understand alleles and mutations, which are testable

DNA is a composed of smaller molecules called nucleotides

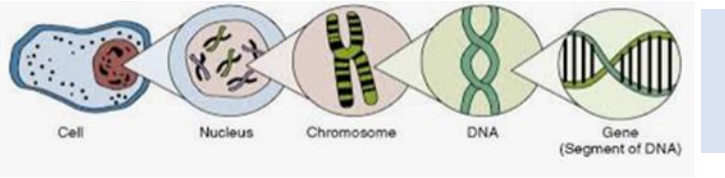
A nucleotide has 3 components:
Nitrogenous base + sugar + phosphate group

Sugar + phosphate group = (grey) handrails of DNA molecule

Two nitrogenous bases = steps of DNA molecule
4 nitrogenous bases:
A (Adenine)
T (Thymine)
C (Cytosine)
G (Guanine)



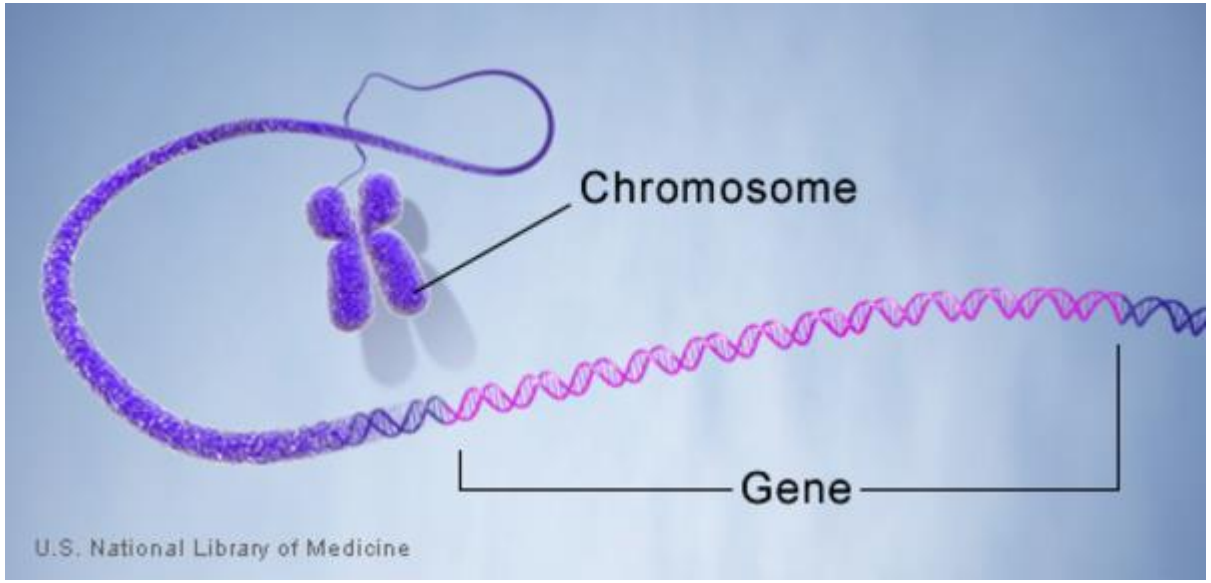
Genes & Loci



For purposes of 121:
A GENE is a segment of DNA (or sequence of nucleotides) that codes for a particular protein.

Human genome projects estimates that we have between 20,000 to 25,000 protein coding genes in our cells.

One chromosome can contain 100's to 1,000's of genes.
Genes have a specific LOCI or address on a chromosome.



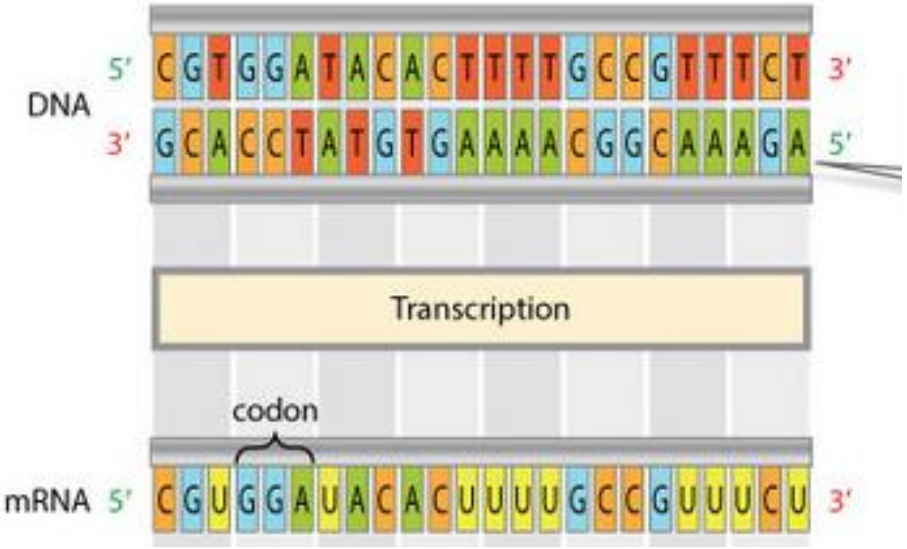
From gene to a protein (not testable)

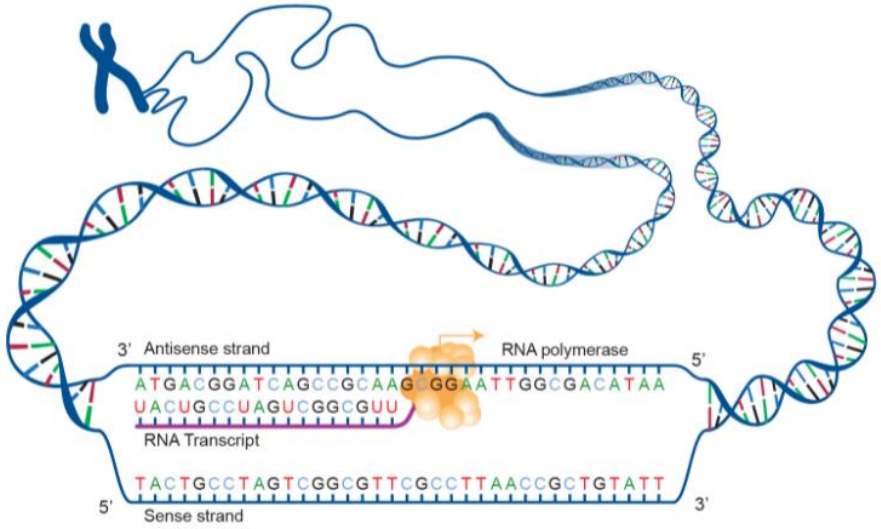
Several steps

Step 1. Transcription (DNA to RNA)

DNA is partially “unzipped” (remember it is double-stranded)

One strand of the DNA of gene serves as a template for the synthesis of an mRNA strand/molecule





ν.nature.com/scitable/topicpage/what-is-a-gene-colinearity-and-transcription-430/
https://www.genome.gov/genetics-glossary/Transcription

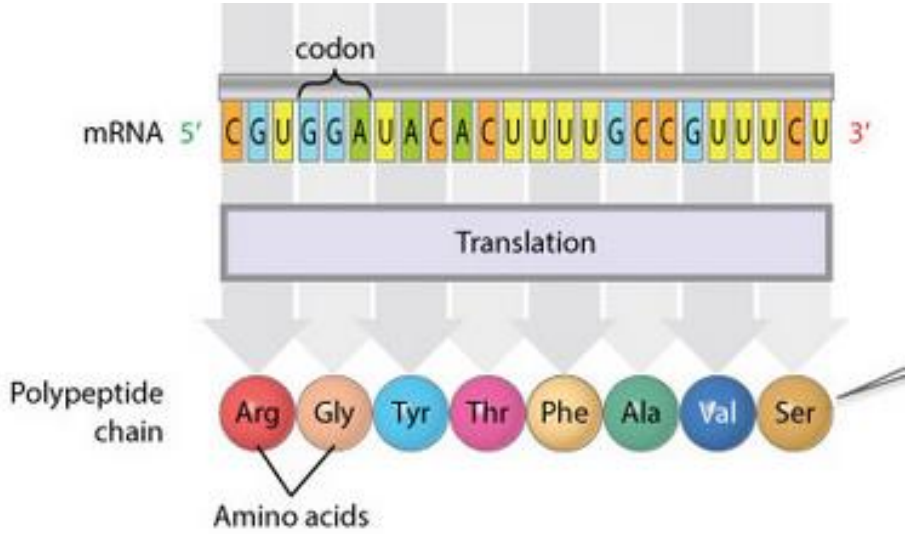
To get from a gene to the protein (not testable)

Step 2. Translation (mRNA to Protein)

- the mRNA molecule is read in-3 letter blocks (codon)

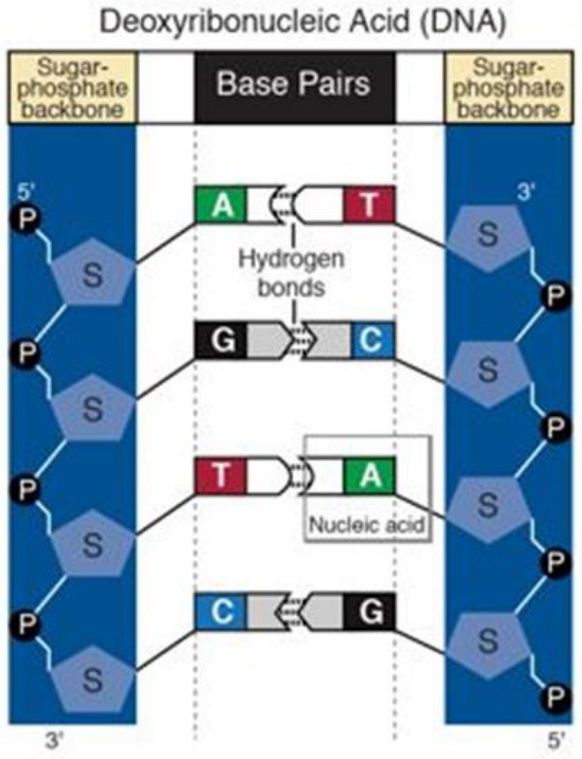
- each 3-letter block produces a specific amino acid

- amino acids join together to form a protein



https://www.nature.com/scitable/topicpage/what-is-a-gene-colinearity-and-transcription-430/

The sequence of the nucleotides in a coding region of DNA is important…



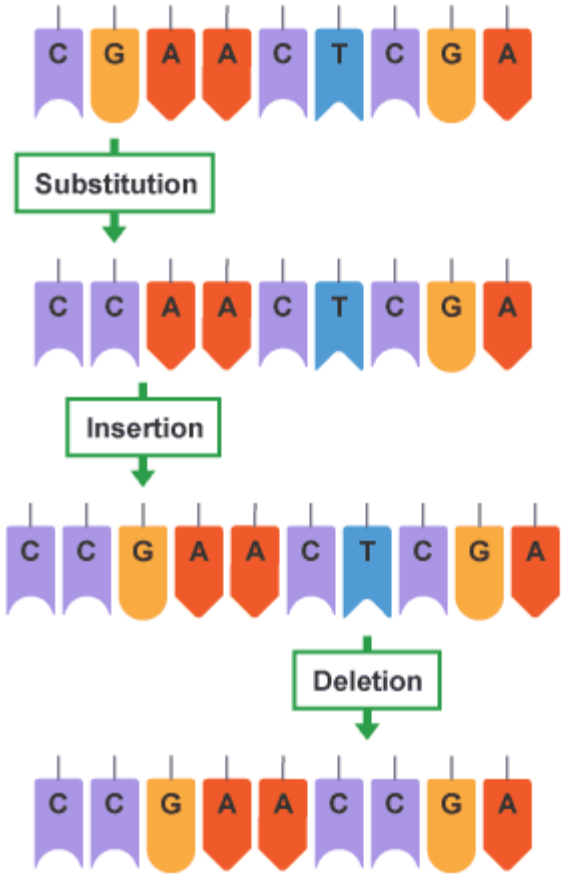
..because the sequence of steps encodes the instructions for making proteins.

If there is a change of the sequence of nucleotides, it can potentially change the amino acid produced.

This may affect the protein's function (e.g. by changing shape).

MUTATIONS in the DNA

A MUTATION is a change in the sequence of nucleotides in the DNA



Source: www.bbc.co.uk

A mutation may be:

- as small as a change in one nucleotide (SNiP or single nucleotide polymorphism); see figure to left.

Most mutations happen during DNA replication.

Environmental factors (e.g. U.V. light) can also damage DNA and result in mutations.

Fun Fact – according to John Hopkins Medicine

- You have an estimated 37 trillion cells in your body. Every time the DNA in one of your cells replicates
  - It is estimated that there are 3 random DNA copying mistakes
- So, we are all mutants ☺

ALLELES

- ALLELES are different versions of the same gene (that arise due to mutations/changes in the sequence of nucleotides).
- Alleles are responsible for variation in inherited traits (or variation in PHENOTYPE – observable characteristic of an organism).
- For example, according to the findings of the Human Genome Project we are 99.9% genetically identical in our genetic make-up (not testable); but we don't look like clones, right?

(https://www.genome.gov/about-genomics/fact-sheets/Genetics-vs-Genomics#:~:text=All%20human%20beings%20are%2099.9,about%20the%20causes%20of%20diseases.)

ALLELES

- The 0.1% differences are responsible for the phenotypic differences between us.
- e.g. we carry different alleles that affect our height, eye colour, susceptibility to disease, etc.

New note: As we will discuss in the genetics unit, most mutations have no effect on the individual, i.e. they may occur in a non-coding region (so no new allele produced), or there may be no change in amino acid.

But, some mutations can be harmful (e.g. sickle cell – see next slide). And some mutations can be beneficial (e.g. Kermode bear's white fur).

ALLELES – Sickle Cell Disease

- For example, sickle cell disease is caused by a SNiP in the ß-globin gene.
- The 6th amino acid in the chain is valine instead of glutamic acid affecting the shape (and function) of the hemoglobin protein (oxygen-carrying protein).
- Outcome is red blood cells that are sickle-shaped.
- Negative affects on oxygen carrying capacity.

https://www.nature.com/scitable/topicpage/genetic-mutation-441/

The white coat of the Kermode bear caused by a SNiP

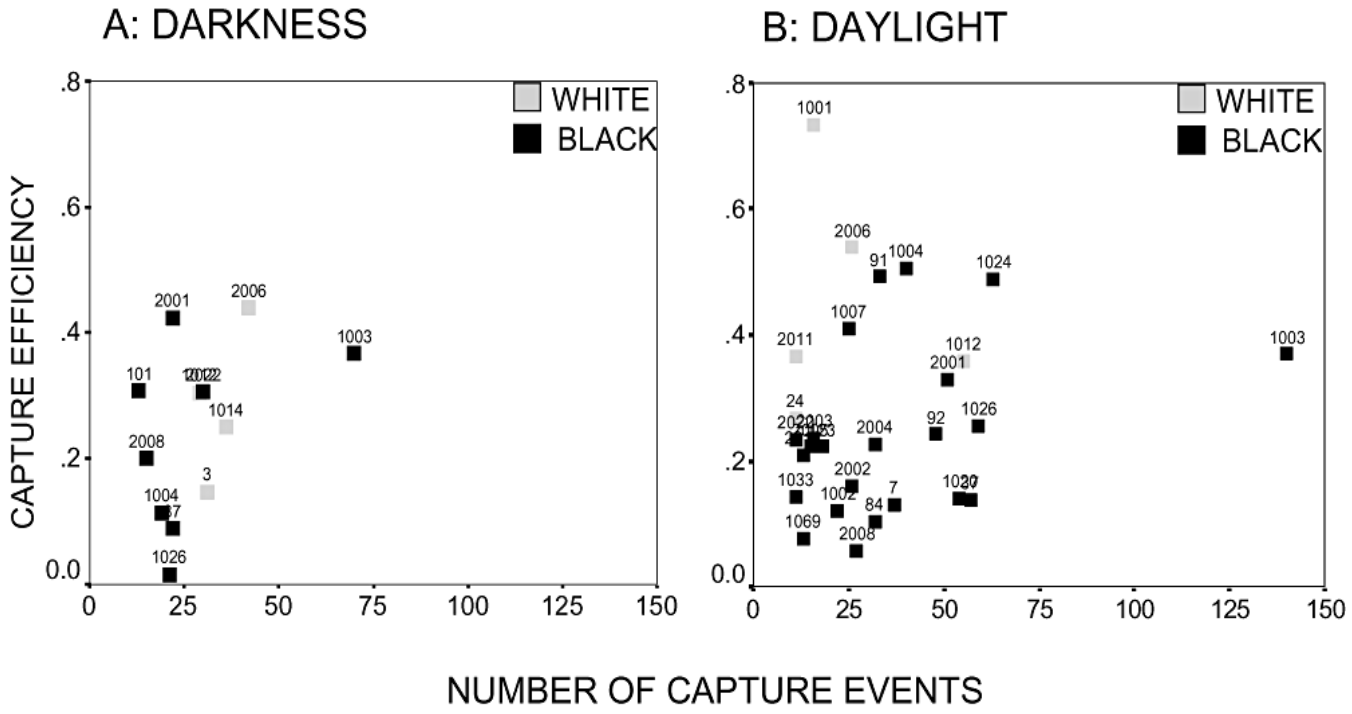
There was a single nucleotide **substitution** of Adenine (A) for Guanine (G) in the *mc1r* gene.

The protein produced by this new allele was non-functional. Melanin not produced.

Resulted in white fur instead of black fur.

Source: unknown

Research by Dr. Tom Reimchen, University of Victoria



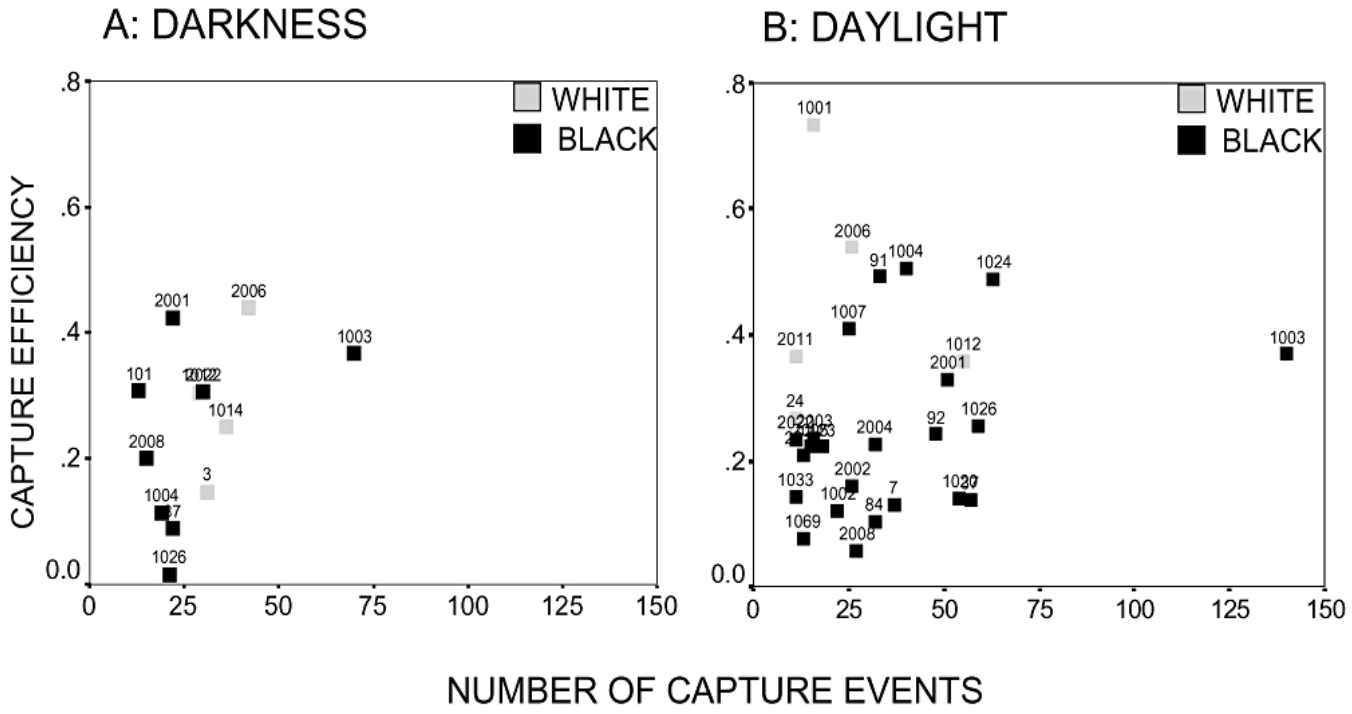
iClicker Question: Is there a significant difference in salmon capture efficiency of Kermode bears and black bears?

A. No
B. Yes, black bears are more efficient at capturing salmon both at night and during the day.
C. Yes, Kermode bears are more efficient at capturing salmon both at night and during the day.
D. Yes, Kermode bears are more efficient at capturing salmon during the day, but not at night.
E. Not sure

Figure 2. Salmon capture efficiency by individual black and white bears within darkness, and daylight on Gribbell Island, coastal British Columbia during the fall of 2000–2002.
A, darkness. Salmon capture efficiency during darkness ($F = 0.12$, d.f. = 1, $P = 0.74$).
B, daylight. Salmon capture efficiency during daylight ($F = 10.9$, d.f. = 1, $P = 0.003$).

Biol J Linn Soc, Volume 98, Issue 3, November 2009, Pages 479–488, https://doi.org/10.1111/j.1095-8312.2009.01306.x

Research by Dr. Tom Reimchen, University of Victoria



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E. Not sure

Figure 2. Salmon capture efficiency by individual black and white bears within darkness, and daylight on Gribbell Island, coastal British Columbia during the fall of 2000–2002.
A, darkness. Salmon capture efficiency during darkness ($F = 0.12$, d.f. = 1, $P = 0.74$).
B, daylight. Salmon capture efficiency during daylight ($F = 10.9$, d.f. = 1, $P = 0.003$).

Biol J Linn Soc, Volume 98, Issue 3, November 2009, Pages 479–488, https://doi.org/10.1111/j.1095-8312.2009.01306.x

iClicker Question

In Kermode bears, the mutant *mc1r* gene would be found in which of the following cells?

A. Gametes (sperm and egg cells)

B. Melanocytes (cells that make fur and skin pigment)

C. Kidney cells

D. Gametes and melanocytes

E. Gametes, melanocytes, and kidney cells.

Answer

In Kermode bears, the mutant *mc1r* gene would be found in which of the following cells?

A. Gametes (sperm and egg cells)

B. Melanocytes (cells that make fur and skin pigment)

C. Kidney cells

D. Gametes and melanocytes

E. Gametes, melanocytes, and kidney cells.

Every cell in the bear's body, and every cell in the approximately 10^{13} cells in the adult human body has its own copy or copies of chromosomes. The only exceptions are those few cell types, such as mature red blood cells or keratinized cells (e.g. hair), which lack a nucleus. What differs amongst cells is which genes get expressed. Genes related to liver function will be expressed in liver cells, not in the skin

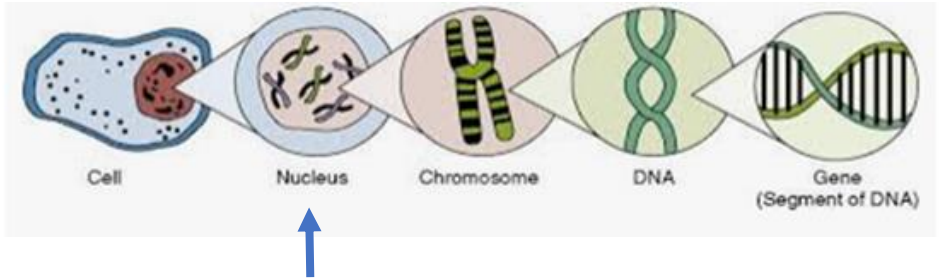
End of Part 1 of terminology

Somatic cells, chromosomes, DNA, genes, alleles

4-minute break

Flying bird or bunny on skis?

Terminology – Part II

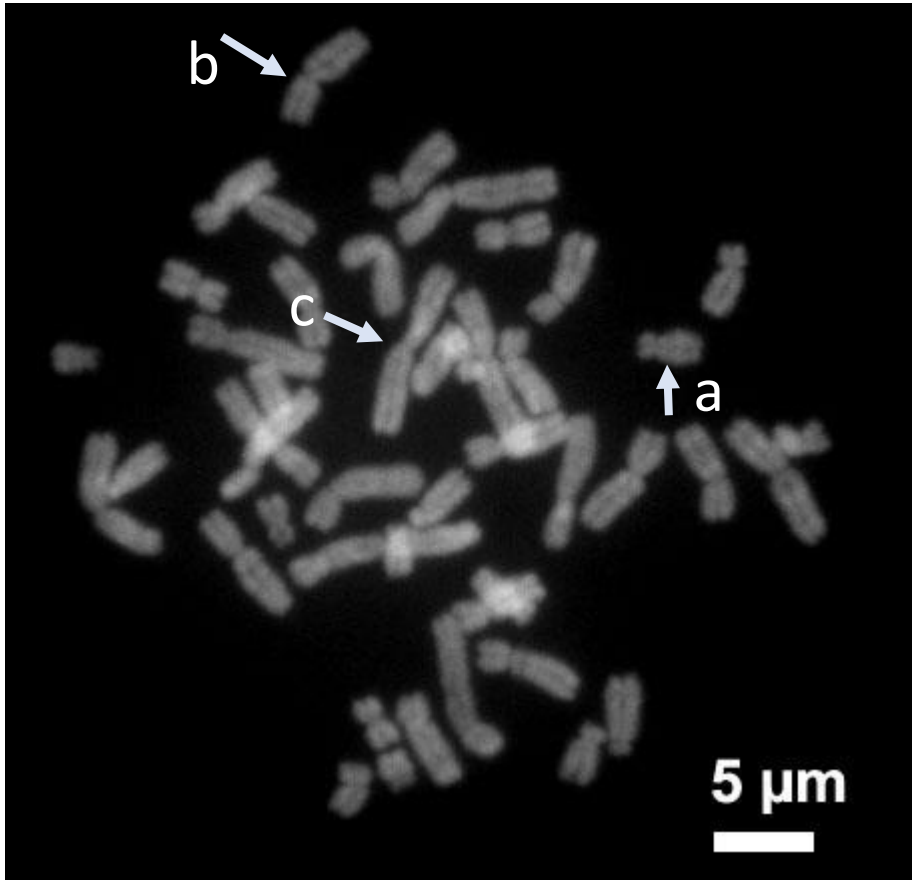


We have 46 chromosomes (in total) in our nucleus.

From the figure to the right, notice:

- The chromosomes differ in size.
- The chromosomes also differ in centromere location

If you organized these chromosomes by size, etc., you would find that…

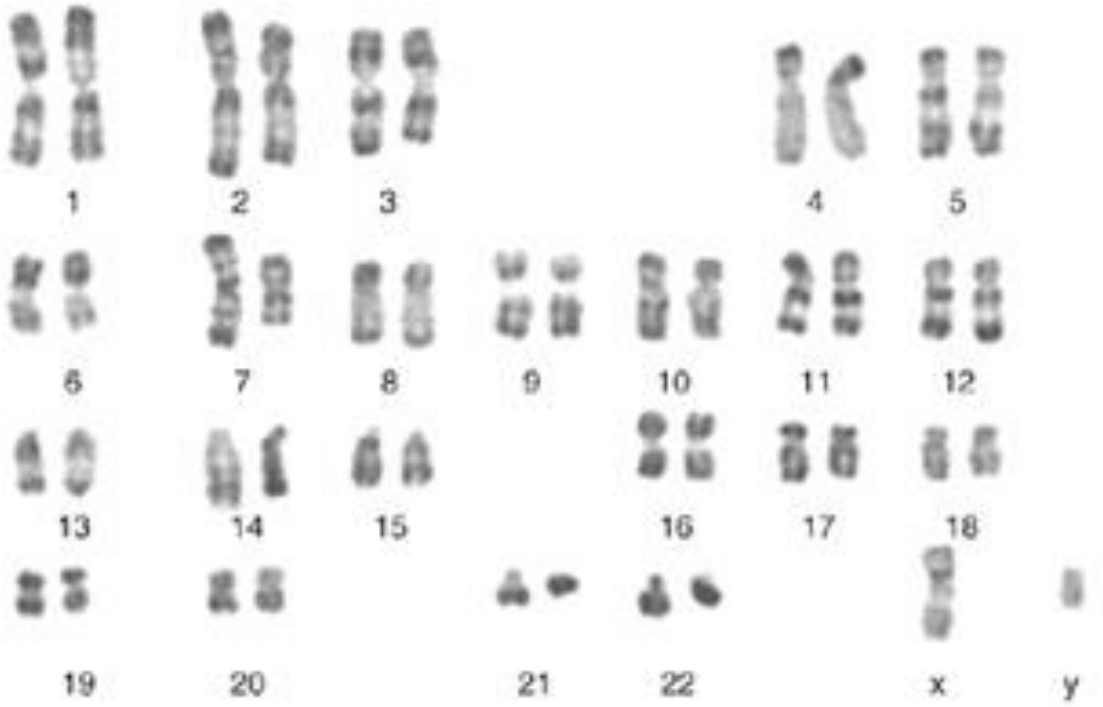


https://evolutionnews.org/2016/05/human_chromosom/

23 pairs of chromosomes, including…

22 pairs of AUTOSOMES – non-sex chromosomes (numbered 1-22, roughly based on size from biggest to smallest); and

1 pair of SEX CHROMOSOMES – not numbered, given letters (in this case an X and a Y chromosome)



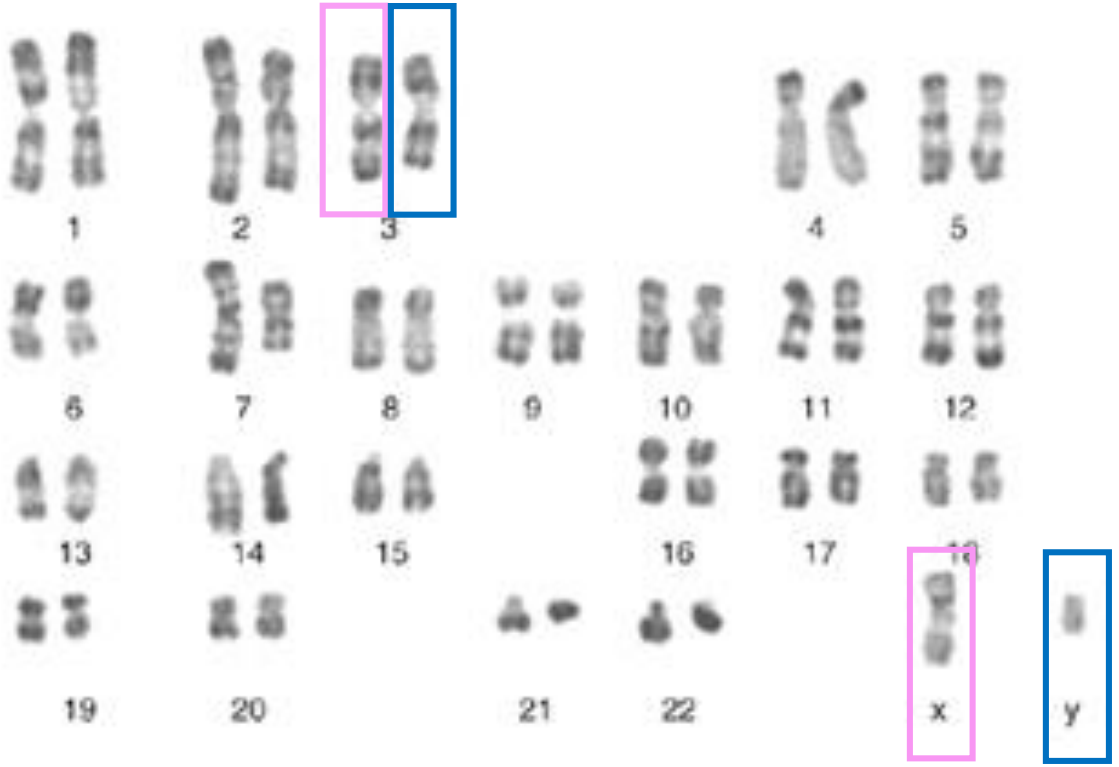
https://www.nature.com/scitable/content/a-karyotype-of-human-chromosomes-6873458/

Why you have 23 pairs of chromosomes…

You inherited one set of 23 chromosomes (22 autosomes + 1 sex chromosome from one parent).

You inherited the other set of 23 chromosomes from your other parent.

https://bio1220.biosci.gatech.edu/sex-01/2-06-dna-and-genes/



https://www.nature.com/scitable/content/a-karyotype-of-human-chromosomes-6873458/

Ploidy = number of sets of chromosomes in a cell

Because your somatic cells contain two sets of chromosomes; your somatic cells are classified as

DIPLOID

Di = 2

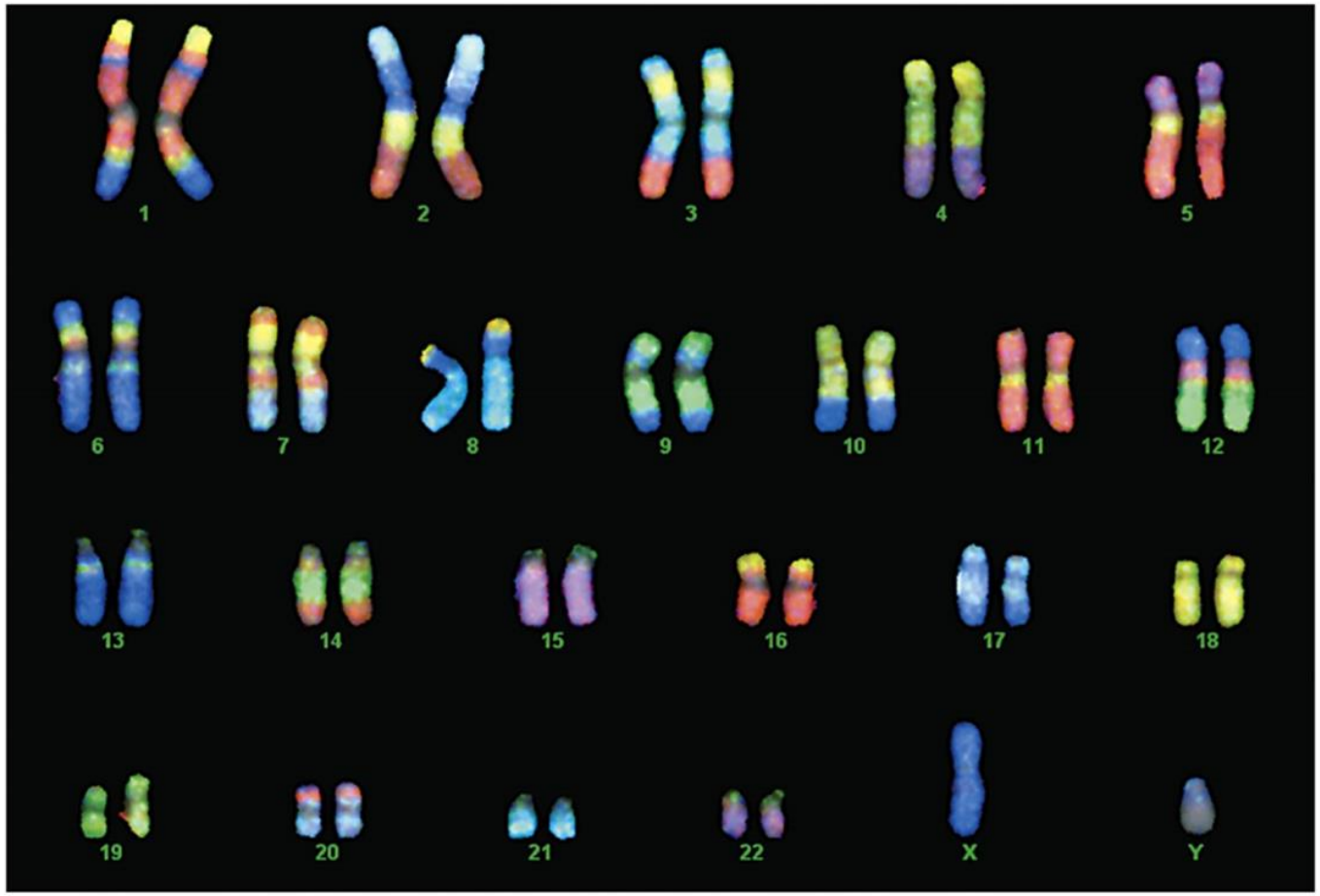
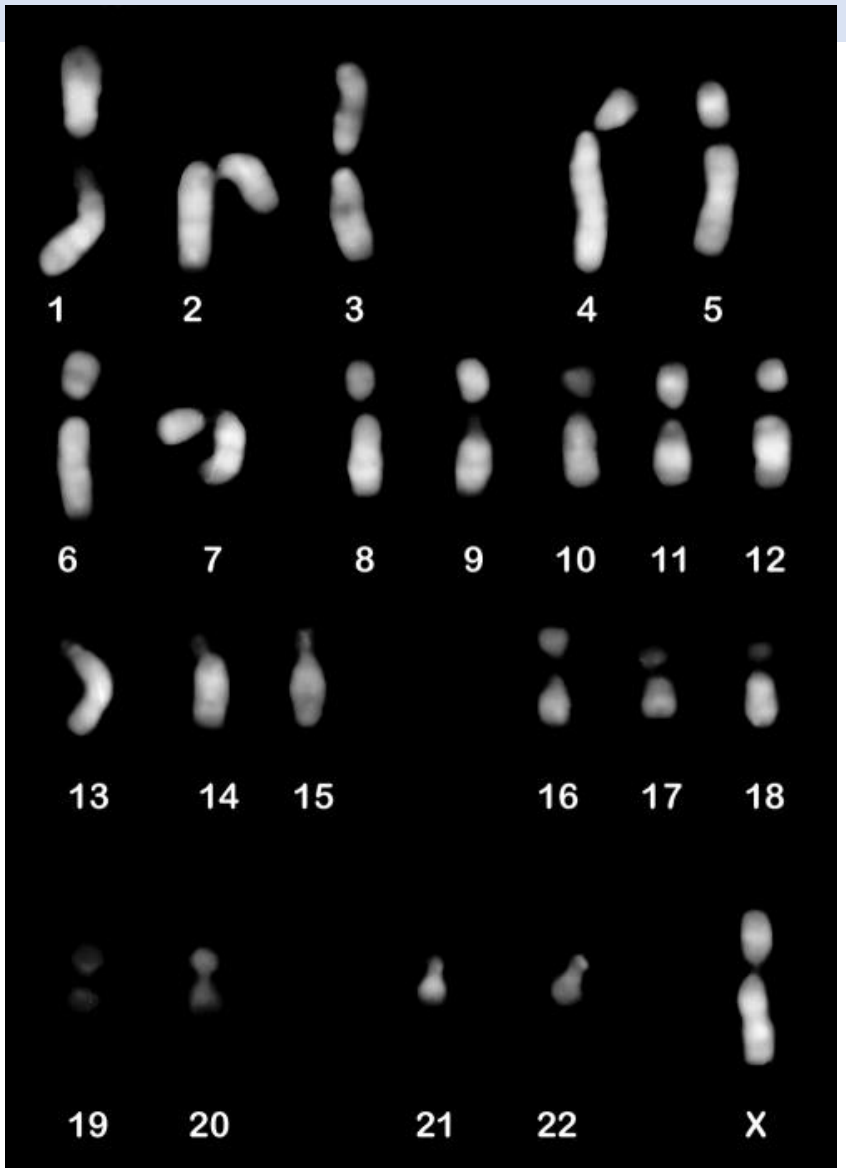


Figure 12-2a Biological Science, 2/e

In contrast, your eggs or sperm are HAPLOID, ha=1

Because they each have one set of chromosomes.

- 22 autosomes
- 1 sex chromosome (either an X or a Y chromosome)

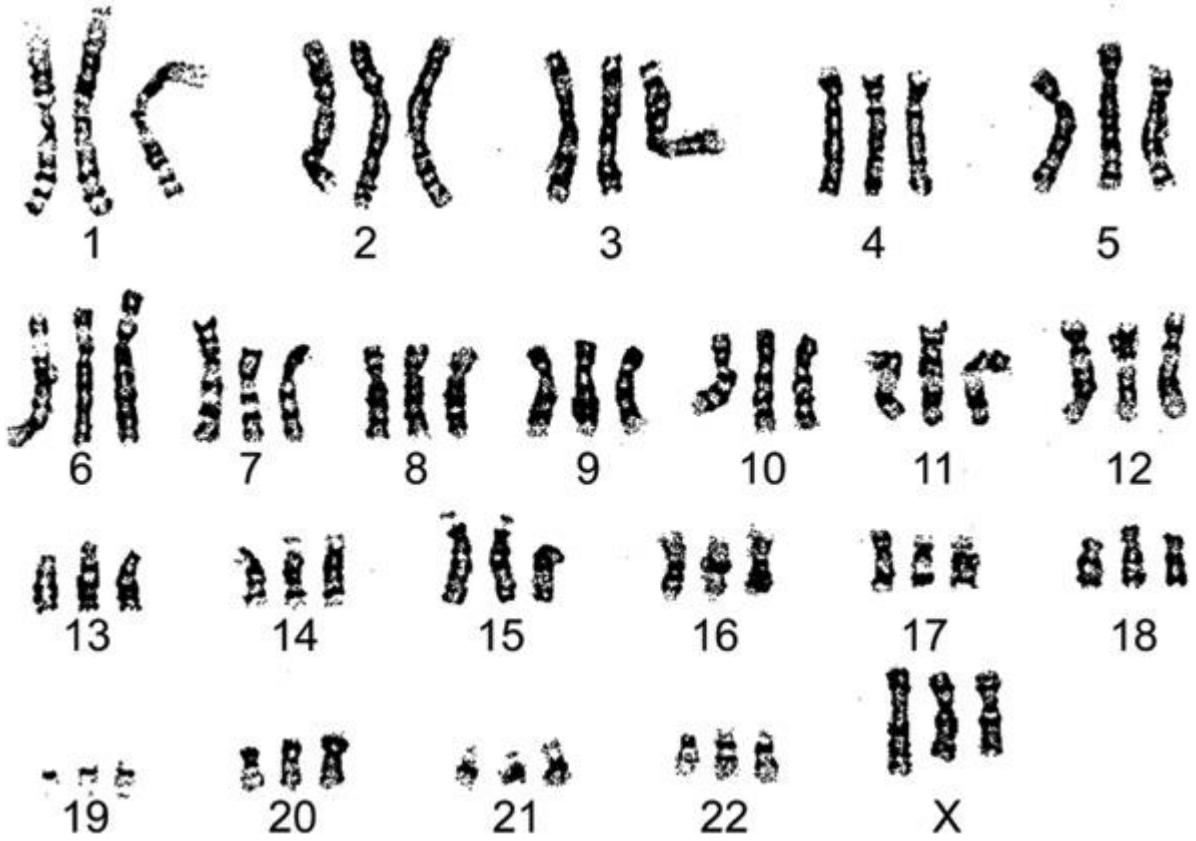


Karyotype of a sperm cell

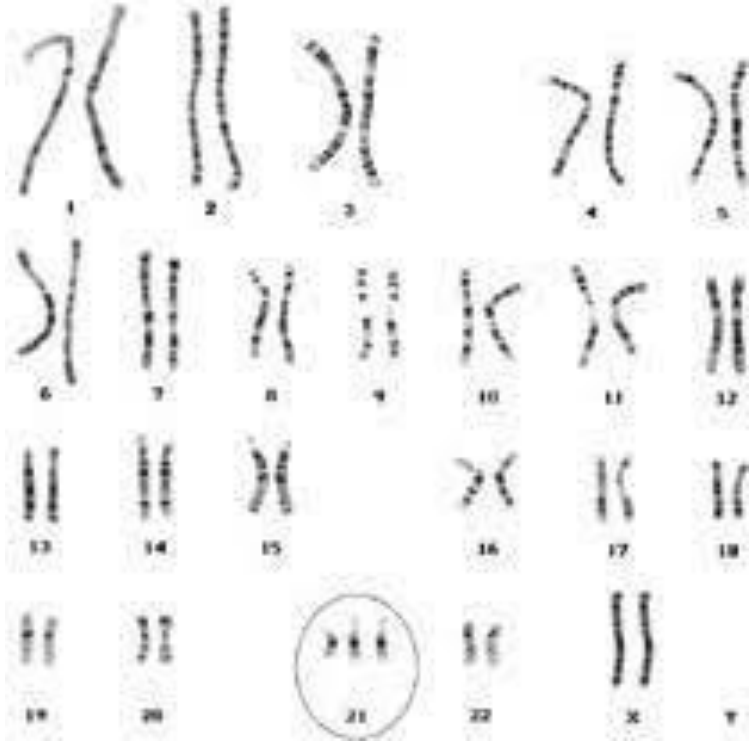
Martin, 2008

Polyploidy (meaning more >2 sets of chromosomes) - can occur in humans; but person may have medical or developmental problems – not testable

This individual did not survive

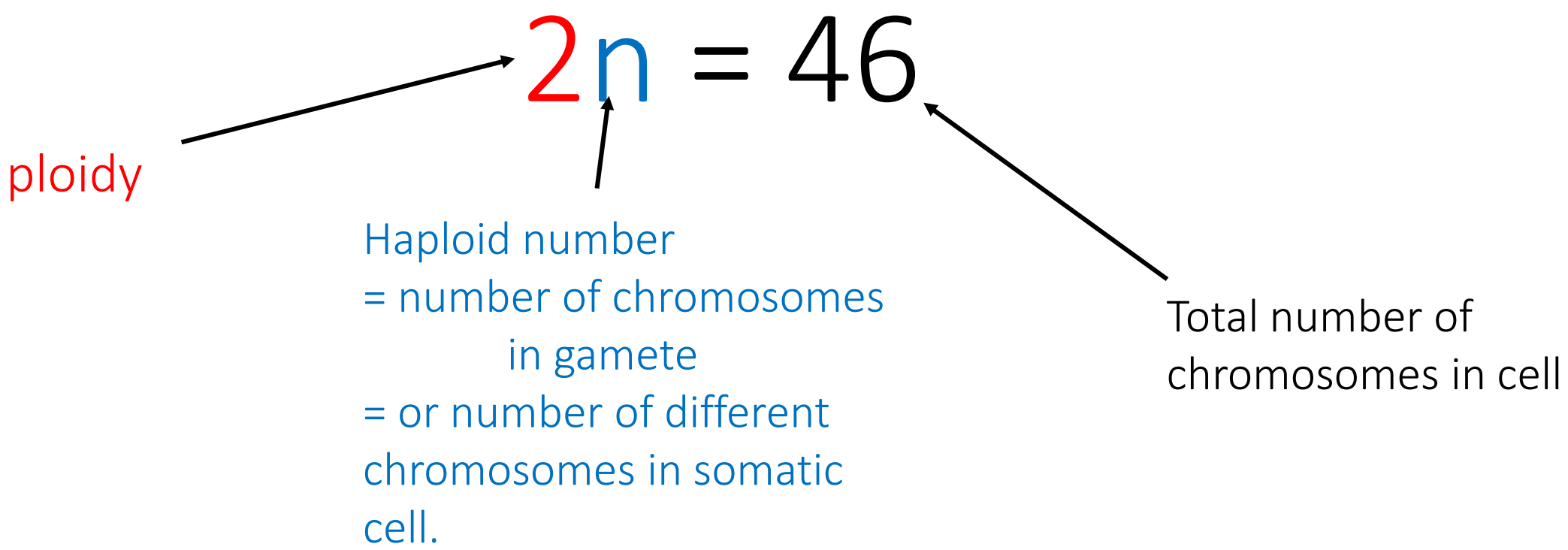


Trisomy 21. People have Down's Syndrome



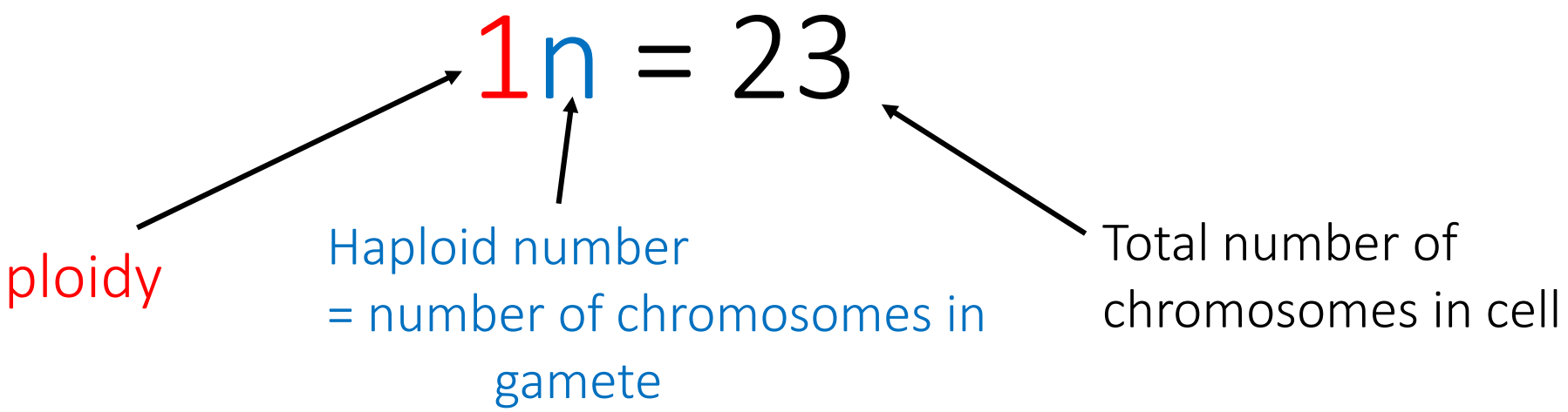
https://www.nature.com/scitable/content/trisomy-21-karyotype-4324/

Equation that describes chromosomes in the nucleus of your somatic cells



If asked to draw the chromosomes in a cell, e.g. a 2n=4 cell, where n=2, keep in mind that the different chromosomes should be different sizes and have different centromere locations)

Chromosome equation for human gamete cells



For a haploid cell, the “1” is often dropped from the equation, e.g. n=23

Homologous chromosomes (or homologs)

HOMOLOGOUS CHROMOSOMES* OR HOMOLOGS

Refers to a set of one pair of chromosomes (one inherited from each parent)

*An important term to know for meiosis.

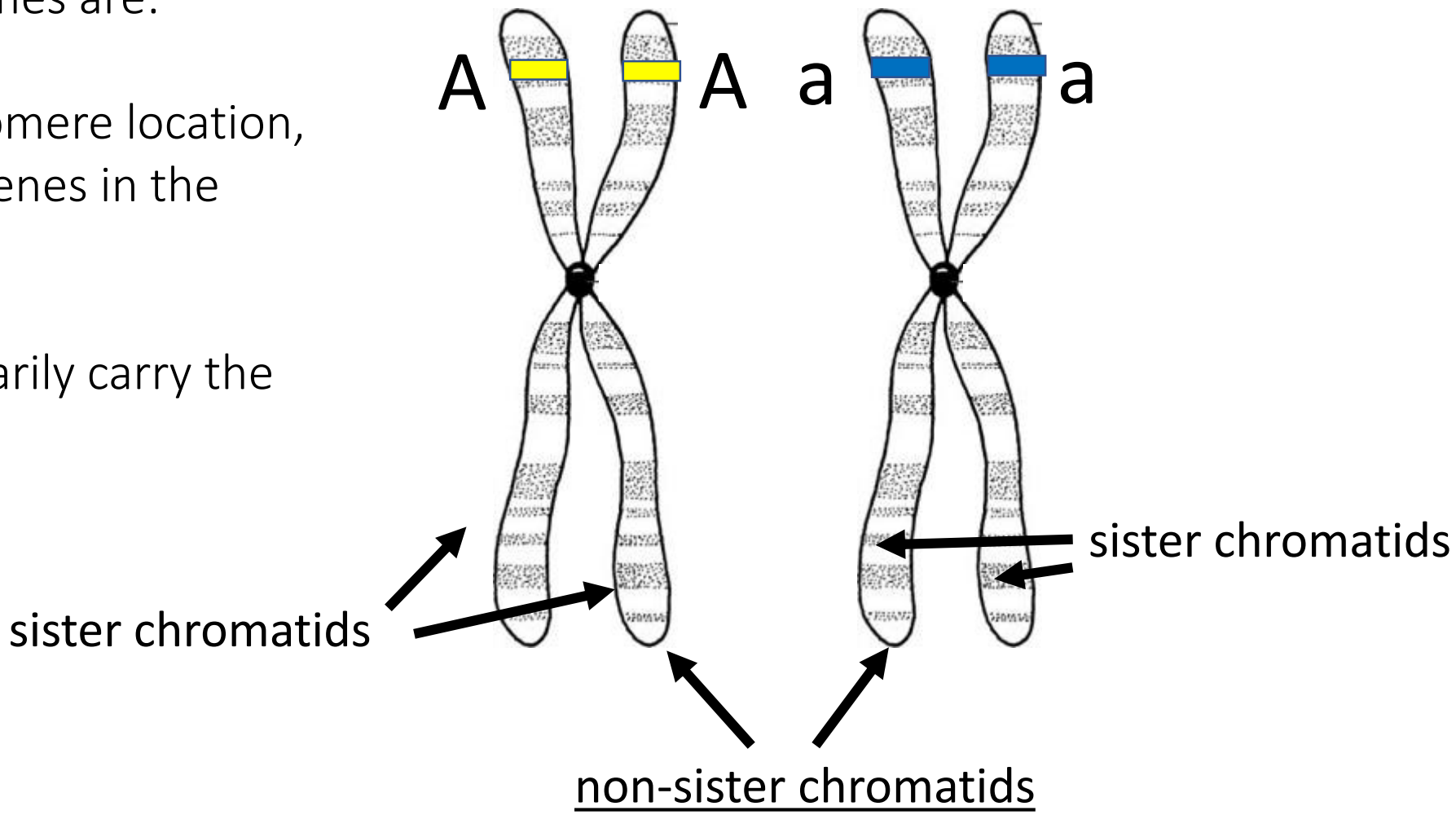
Figure 12-2a Biological Science, 2/e

: Homologous chromosomes

Homologous chromosomes are:

- the same size
- have the same centromere location,
- and carry the same genes in the same locus.

BUT they do not necessarily carry the same same alleles.

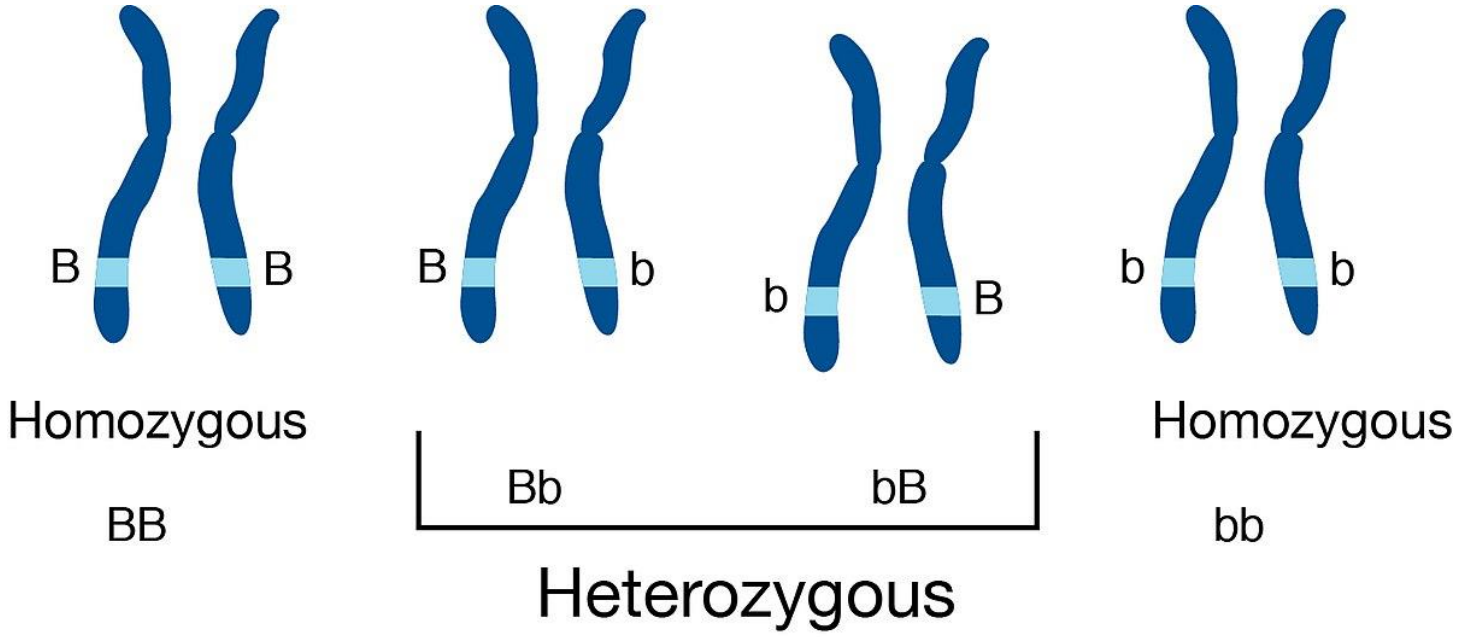


Homozygous & Heterozygous Genotypes

GENOTYPE (narrow definition) refers to the set of alleles present at one or more specific loci

Two homologs (or homologous chromosomes) that carry the same alleles at a specific locus (e.g. BB or bb) have a HOMOZYGOUS GENOTYPE

Two homologs that carry two different alleles (e.g. Bb) at the same locus have a HETEROZYGOUS GENOTYPE



Genome

Multiple definitions. For our purposes:

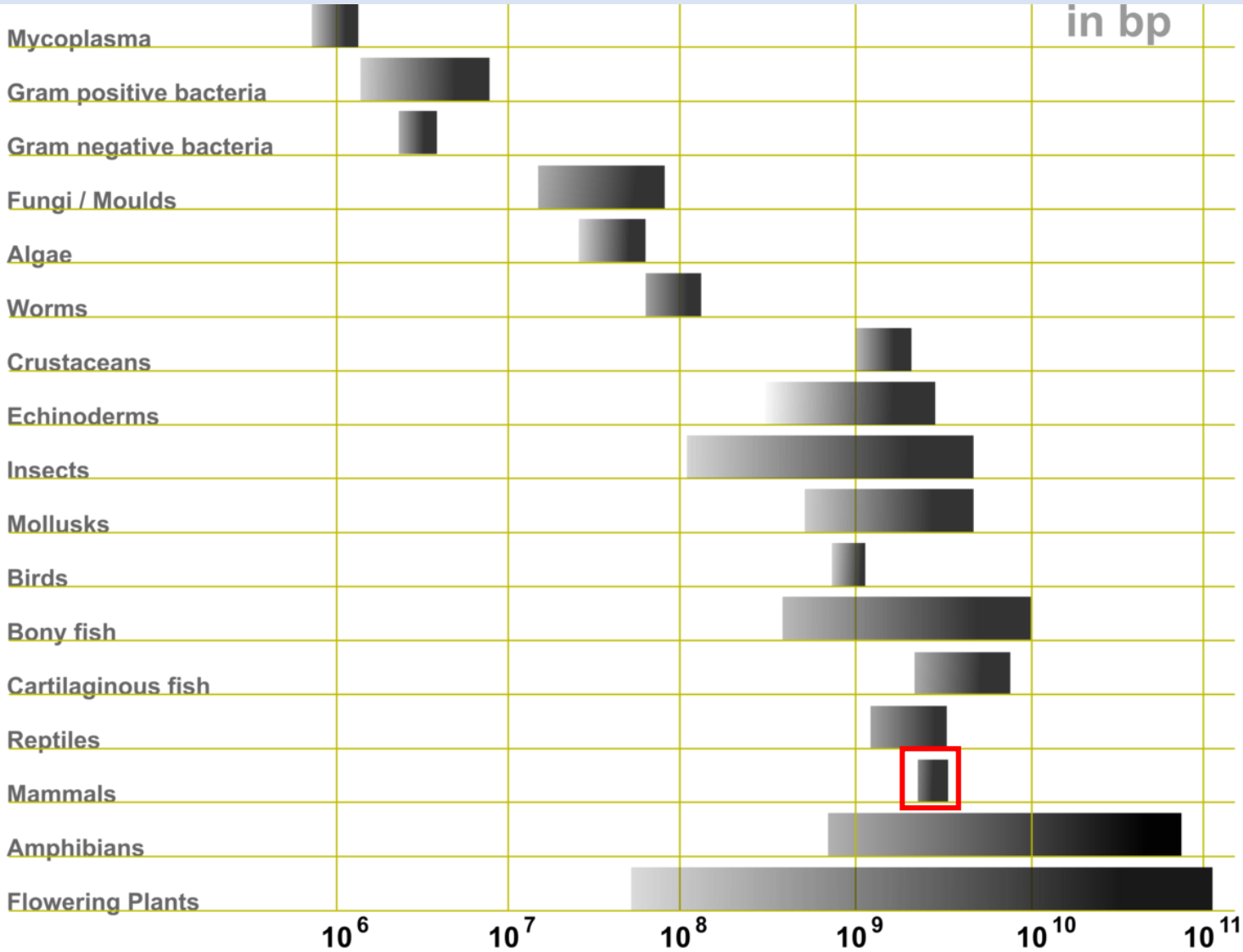
A GENOME is the complete set of genetic information found in an organism's cell.

It includes an organism's complete set of DNA and its genes.

Typically measured in number of bases or base-pairs (bp or Mbp).

The human nuclear genome comprises approximately 3 200 000 000 nucleotides of DNA or 1600 Mbp (https://www.ncbi.nlm.nih.gov/books/NBK21134; not testable)

Variation in genome size



Take home point – genome size not necessarily a measure of complexity.

Humans – about 1600Mbp

By Abizar at English Wikipedia, CC BY-SA 3.0, https://commons.wikimedia.org/w/index.php?curid=19537795

Total number of chromosomes in a somatic cell varies

- Great White Shark: 82
- Pineapple: 50
- Chimpanzees & Gorillas: 48*
- Human: 46
- Earthworms: 36
- Mosquitoes: 6
- Jack Jumper Ant: 2

*Fun Fact: Chromosome 2 in humans shows evidence of fusion

- IJdo, J.W, Baldini, A, Ward, D.C, Reeders, S.T, Wells, R.A. (1991) Origin of human chromosome 2: an ancestral telomere-telomere fusion *Proc Natl Acad Sci U S A.*, **88** 9051-9055.[pdf

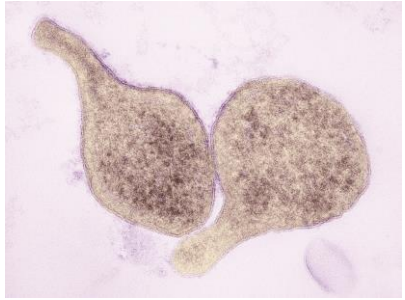
Variation in number of genes amongst organisms (not testable)

- Humans are estimated to have 20,000-25,000 protein coding genes.
- Pineapples have 29,412 genes

https://www.nigms.nih.gov/education/fact-sheets/Pages/genetics.aspx

- Rice (*Oryza sativa*) has 50,000 protein coding genes.

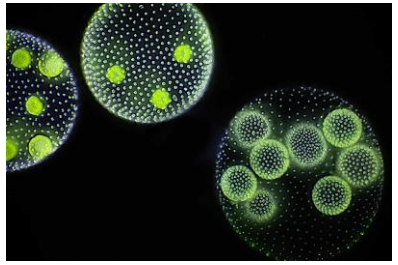
- *Mycoplasma genitalium (bacteria)*~256 genes



wikicommons

Take home point – number of genes is not necessarily a measure of complexity

Ploidy varies amongst taxa – haploid (n), diploid (2n), polyploid (>2n)



Volvox (*n*)

Mushrooms (*n*)

Mammals (*2n*)

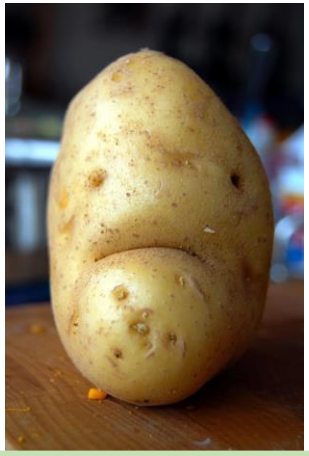
Wheat (7*x*)

Male bees, ants (*n*)
Females (2n)

Yeast (*n* or 2*n*)

Genetically engineered salmon & store bought bananas (3*n*)

Pineapple 2n to 4n



Potato 2n to 5n

iClicker Question

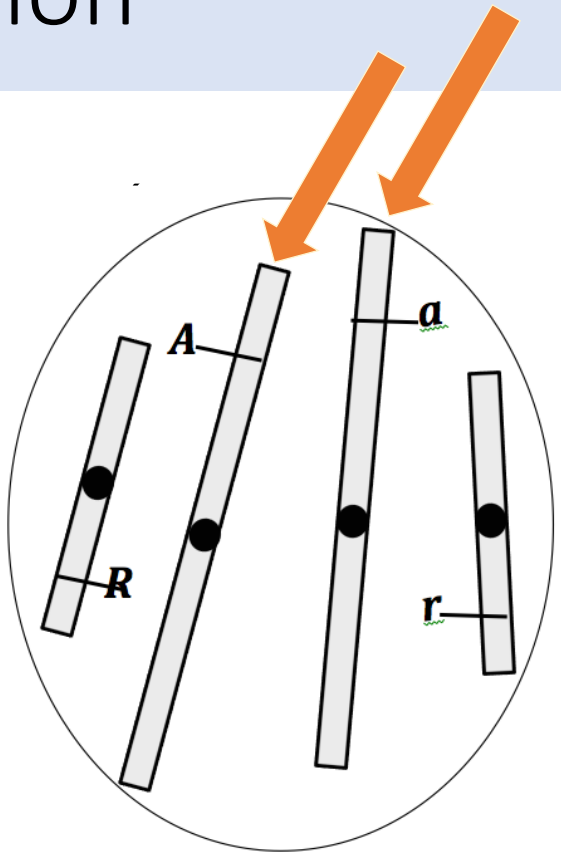
What are the arrows pointing to?

A. Sister chromatids

B. Homologous chromosomes

C. Non-homologous chromosomes

D. Homologous sister chromatids



Answer

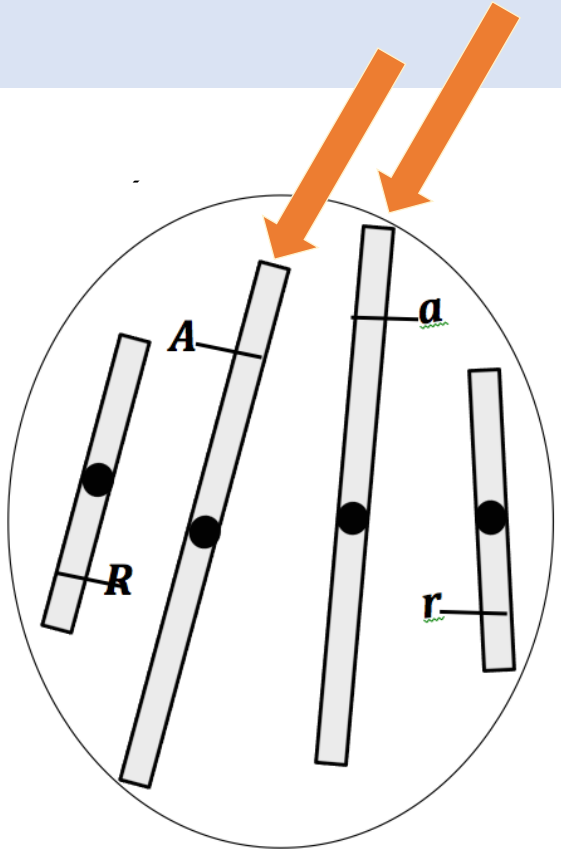
What are the arrows pointing to?

A. Sister chromatids

B. Homologous chromosomes

C. Non-homologous chromosomes

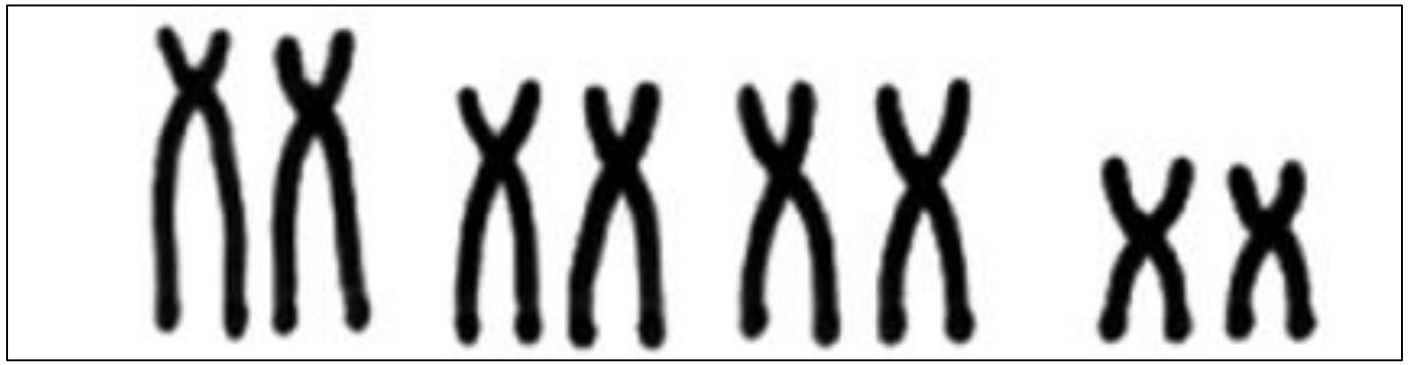
D. Homologous sister chromatids



iClicker Question

How many chromosomes are shown in the figure below?

A. 2
B. 4
C. 8
D. 16

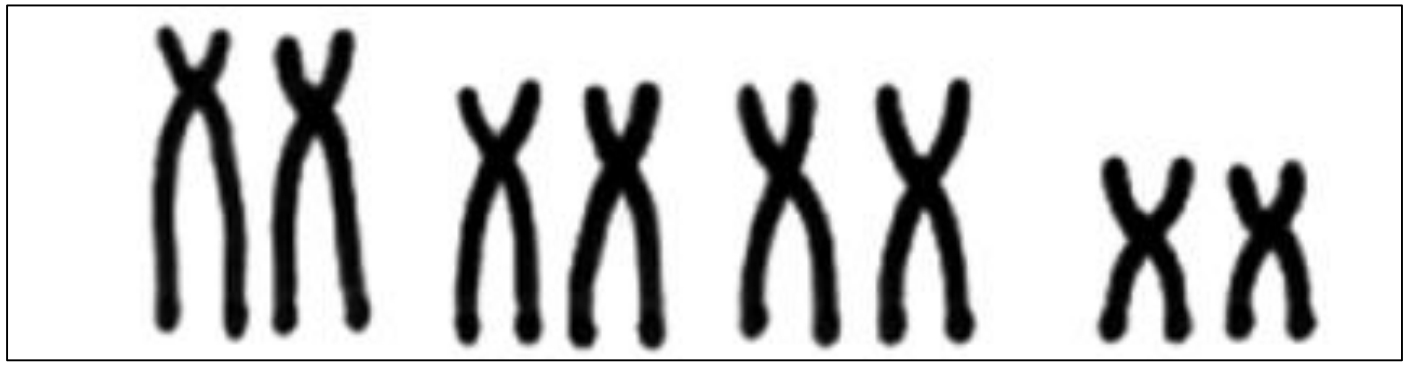


Hint - Count the number of centromeres

Answer

How many chromosomes are shown in the figure below?

A. 2
B. 4
C. 8
D. 16

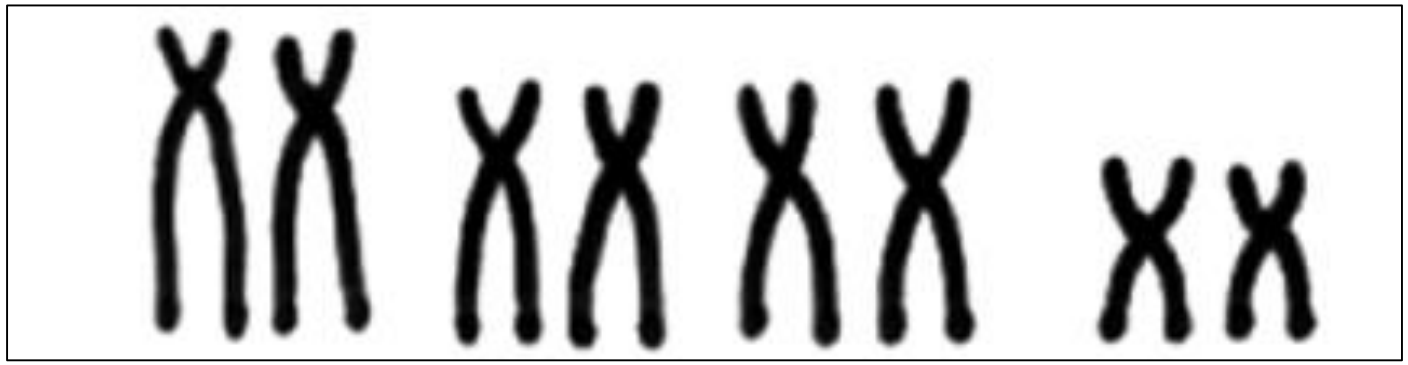


Hint - Count the number of centromeres

iClicker Question

How many DNA molecules are shown in this figure?

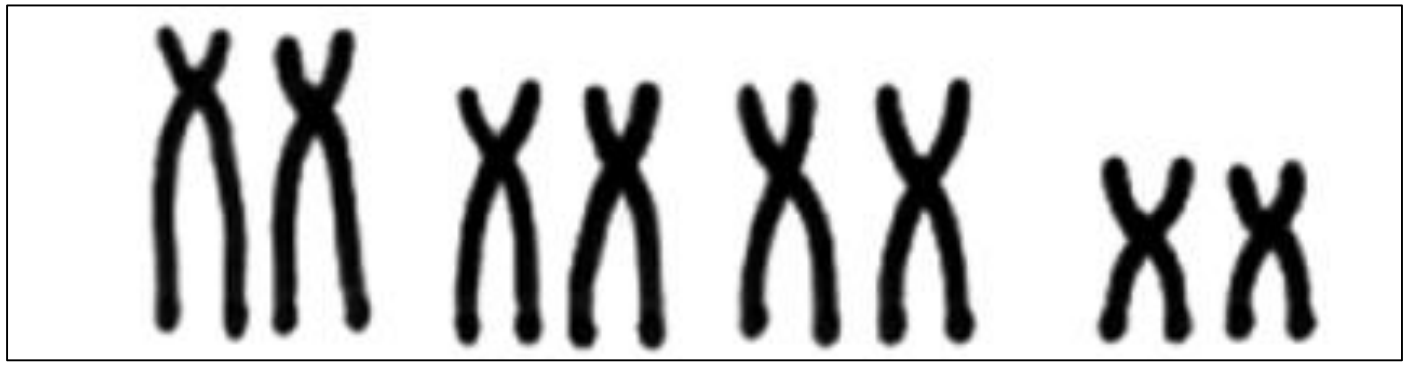
A. 2
B. 4
C. 8
D. 16



Answer

How many DNA molecules are shown in this figure?

A. 2
B. 4
C. 8
D. 16



iClicker Question

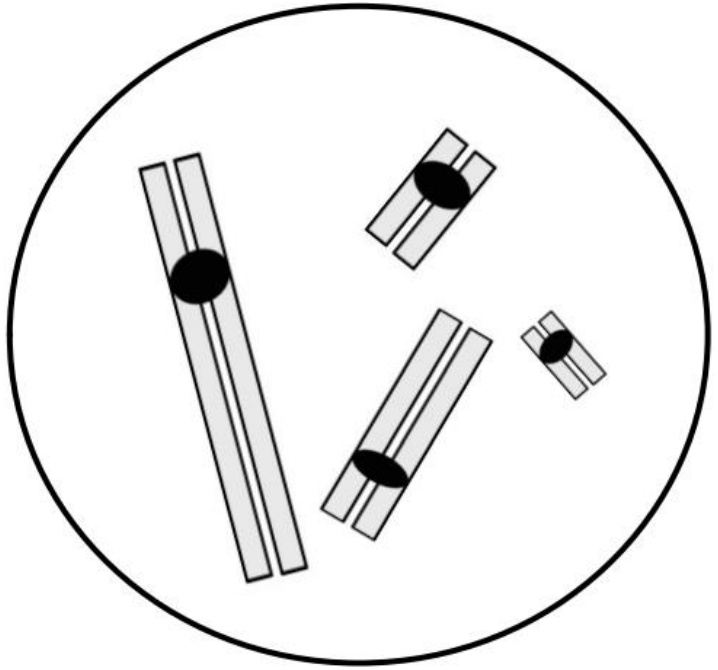
How many chromosomes are there in this cell?

A. 2

B. 4

C. 8

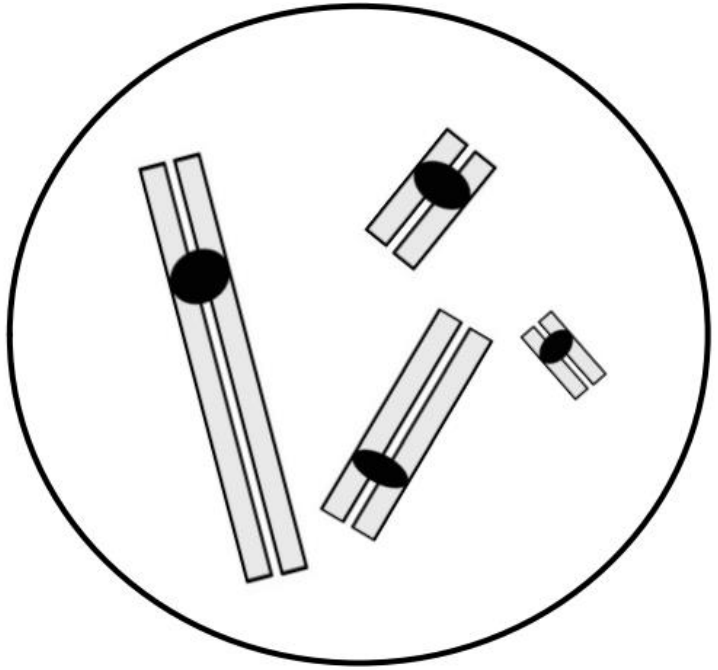
D. 16



- Helpful hint: count the centromeres!

Answer

How many chromosomes are there in this cell?



A. 2

B. 4

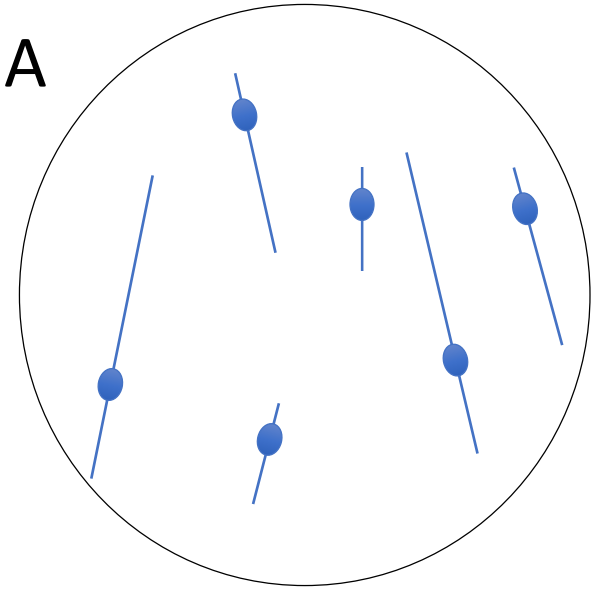
C. 8

D. 16

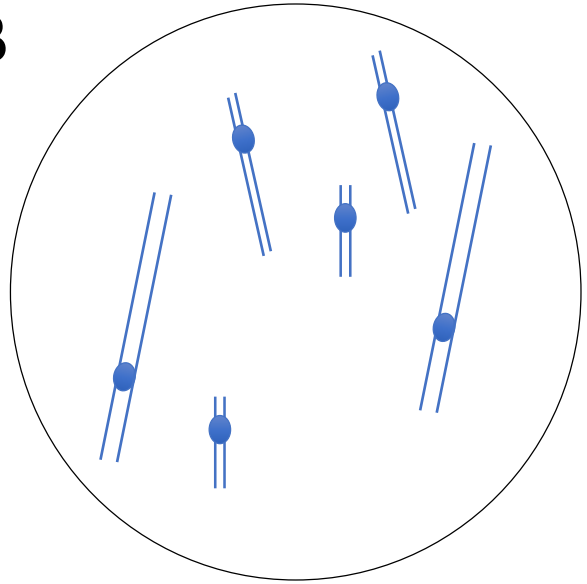
- Helpful hint: count the centromeres!

iClicker Q. - Which cell(s) are 2n=6?

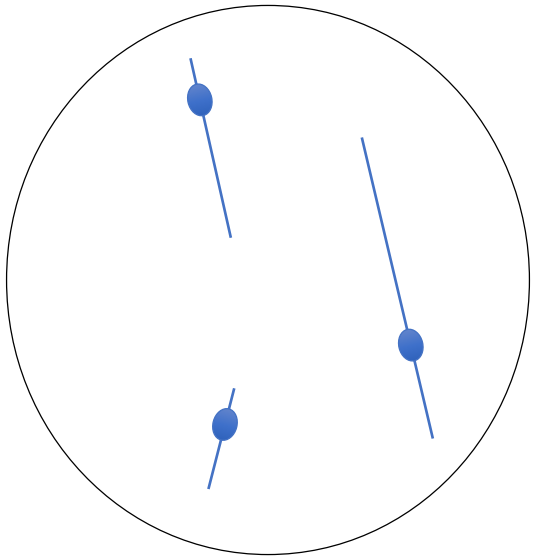
A



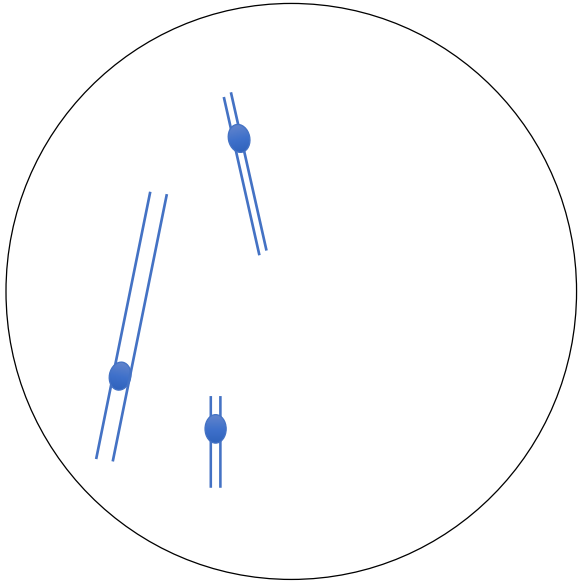
B



C



D



A. A

B. B

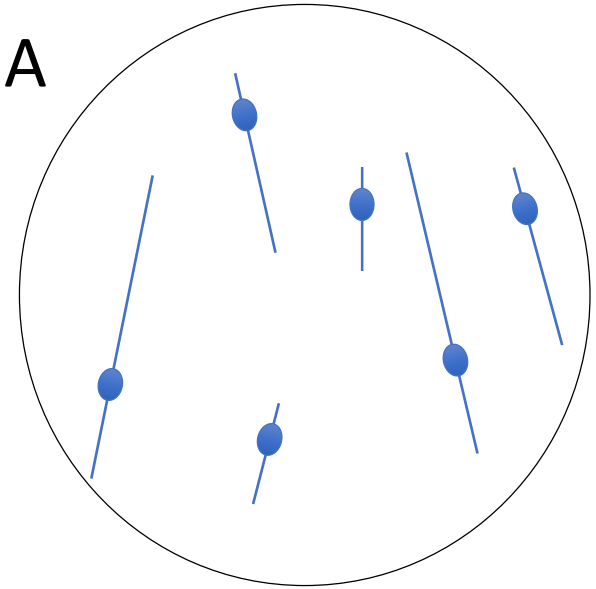
C. C

D. D

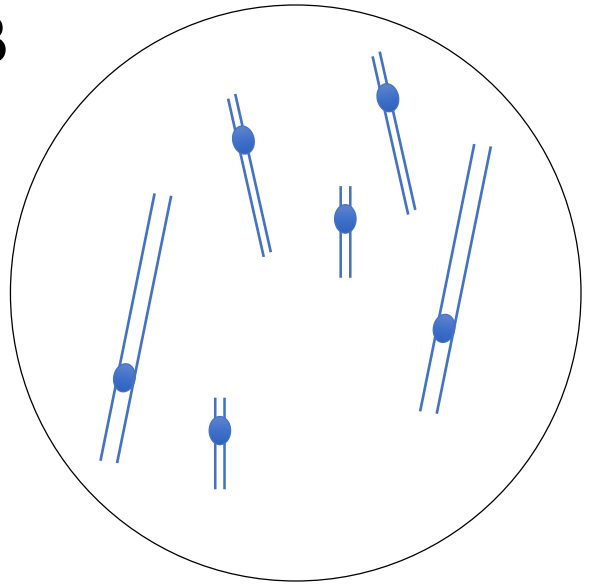
E. A & B

Answer - Which cell(s) are 2n=6?

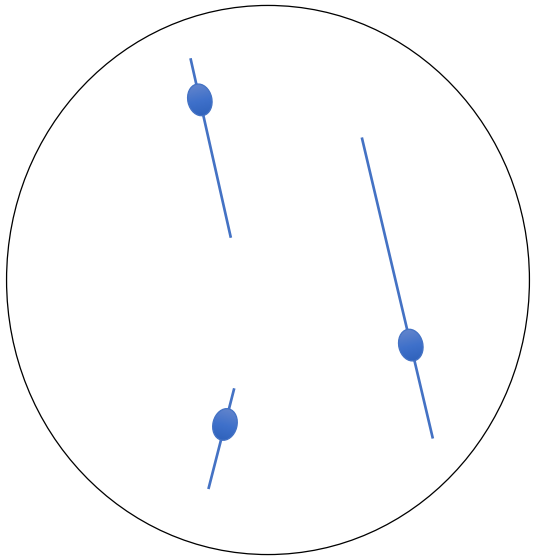
A



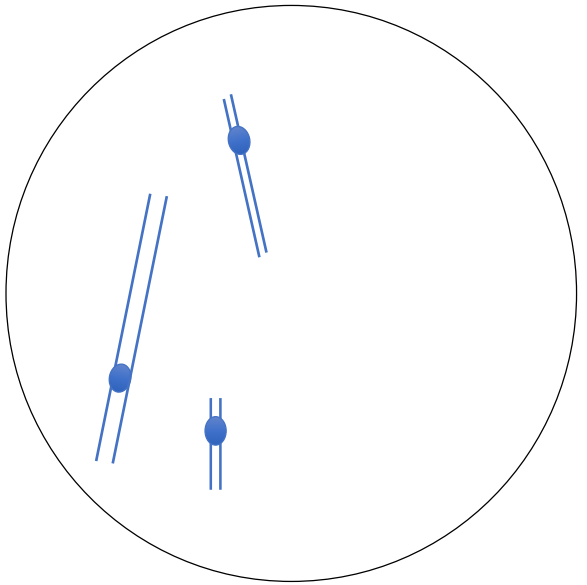
B



C



D



A. A

B. B

C. C

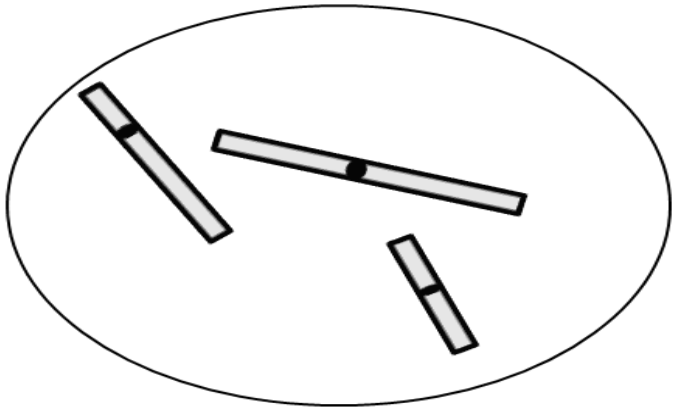
D. D

E. A & B

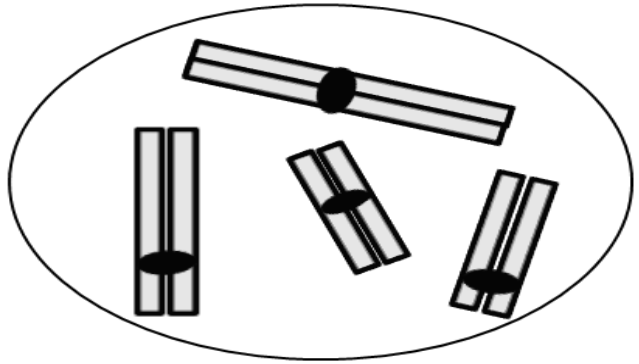
iClicker Question

Which cell is diploid?

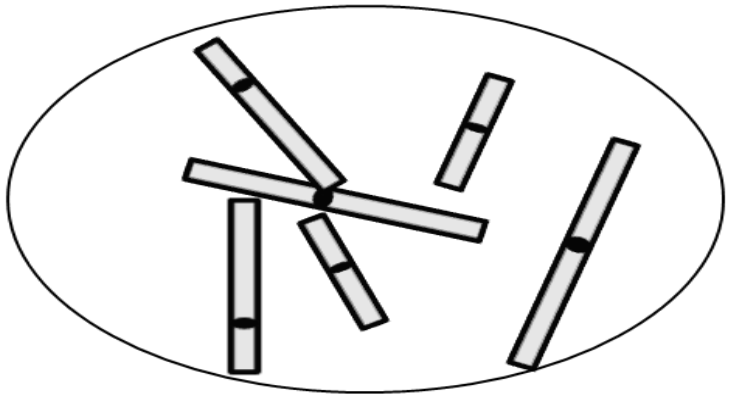
A.



B.



C.

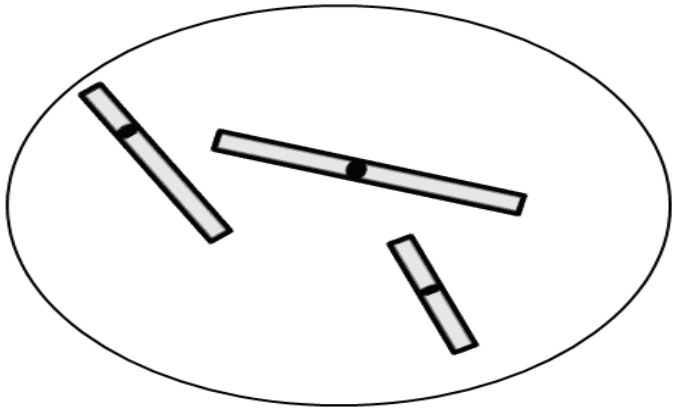


D. There is more than one diploid cell here.

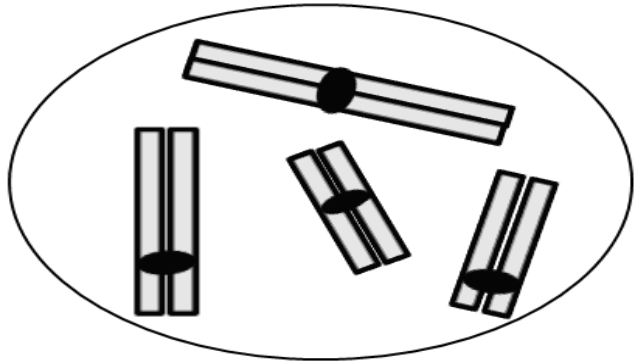
iClicker Question

Which cell is diploid?

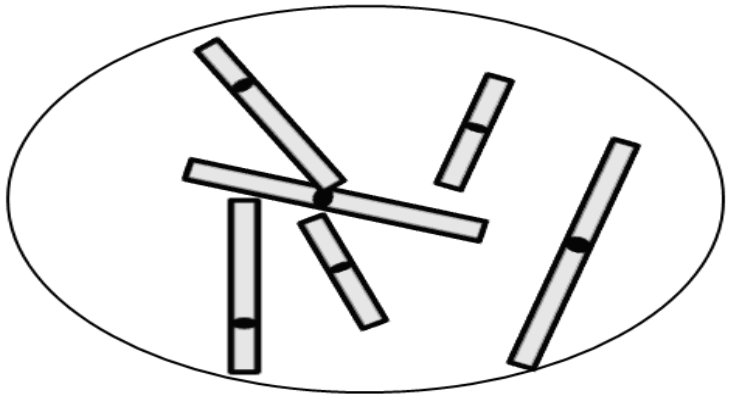
A.



B.



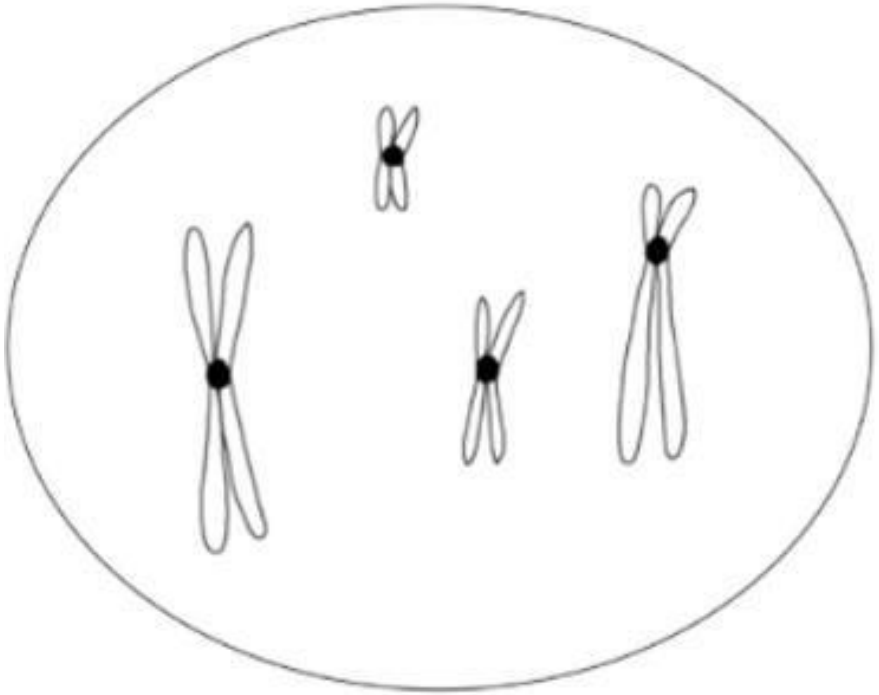
C.



D. There is more than one diploid cell here.

Explanation question

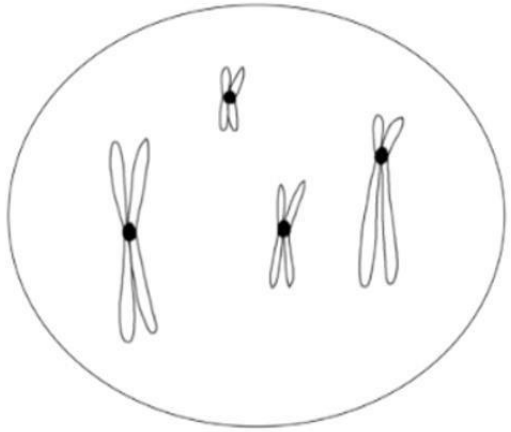
Your friend claims that the cell shown below is diploid. Is your friend correct? Explain why or why not.



2 minutes to answer question.

Example answer

Your friend this the cell shown below is diploid. Is your friend correct? Explain why or why not.



My friend is incorrect. The cell is not diploid.

Diploid means that there are two of each type of chromosome in a cell. However, there is only one of each type of chromosome in this cell, as indicated by differences in size and centromere location. Therefore, this cell is haploid (n=4), with replicated chromosomes, not diploid.

iClicker Question

What is the ploidy and haploid number of chromosomes in the cell shown below?

A. Diploid; 12
B. Diploid; 6
C. Diploid; 3
D. Haploid; 12
E. Haploid; 6

How would you write the notation for the genetic make-up of this cell?

Answer

What is the ploidy and haploid number of chromosomes in the cell shown below?

A. Diploid; 12
B. Diploid; 6
C. Diploid; 3
D. Haploid; 12
E. Haploid; 6

How would you write the notation for the genetic make-up of this cell?

Questions?

Source: www.cbc.ca

Genes & Chromosomes: learning objectives

You should be able to:

- **Identify** homologous chromosomes based on structure (size, centromere position) and genetic loci.
- **Recognize** or **describe:**
  - autosome vs. sex chromosome
  - unreplicated vs. replicated DNA
  - number of DNA molecules vs chromosome number
  - homologous chromosomes vs. sister chromatids
  - sister chromatid vs non-sister chromatid
  - cell ploidy (haploid, diploid, polyploidy)
  - ploidy versus haploid number
  - heterozygous genotype vs homozygous genotype

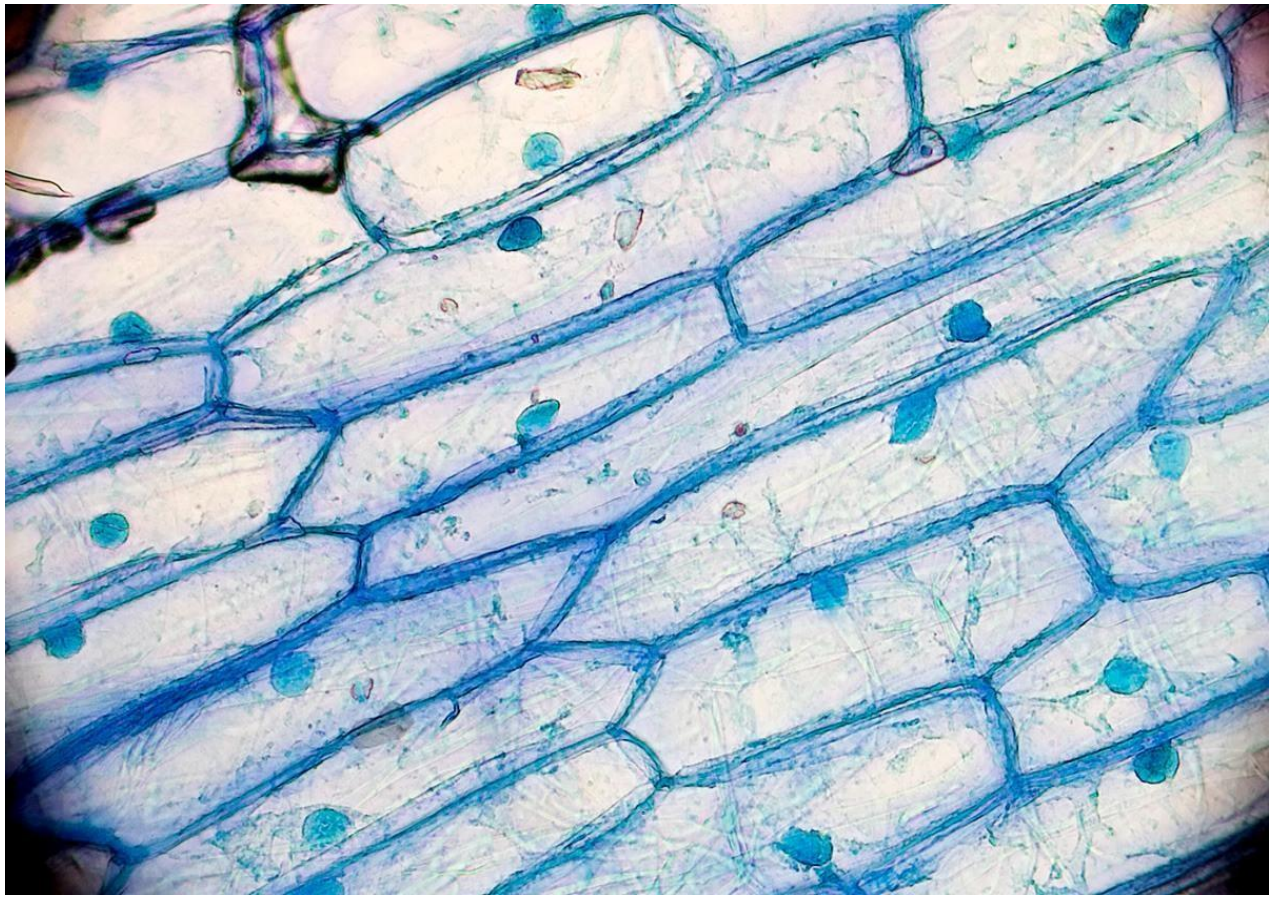
- **Diagram** or **interpret** cells with different numbers of chromosomes and cells that differ in ploidy.

Please see Canvas > Course Information > Learning Objectives

- The remaining slides include content on the cell cycle (interphase and the cell cycle).
- I am including these slides for people who want to read ahead.
- I will likely update slides before Tuesday’s class.

According to cell theory

- All cells arise from pre-existing cells



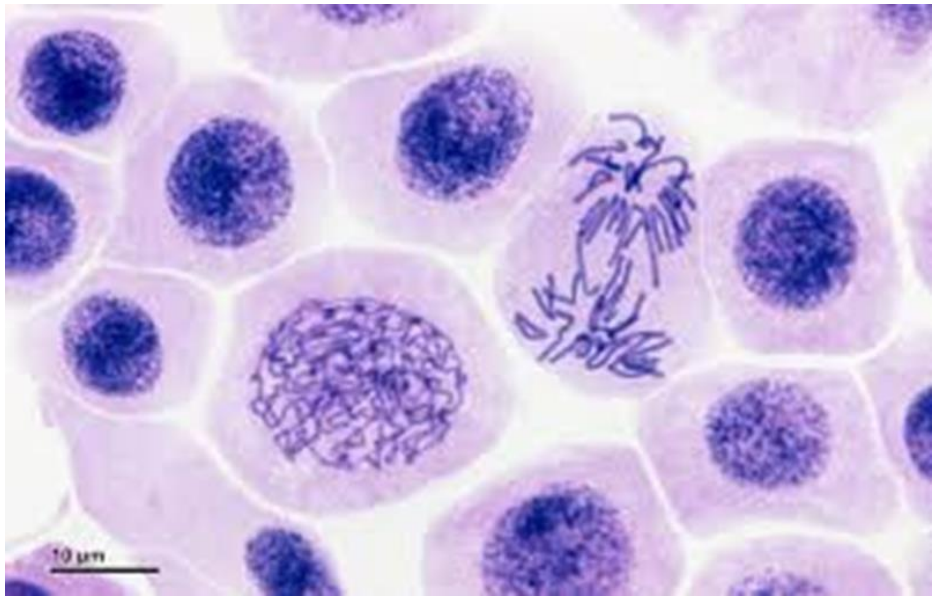
https://commons.wikimedia.org/wiki/File:Onion_Cells.jpg

New cells arise through cell division

- Two types of cell division:

Mitosis + cytokinesis

Meiosis + cytokinesis



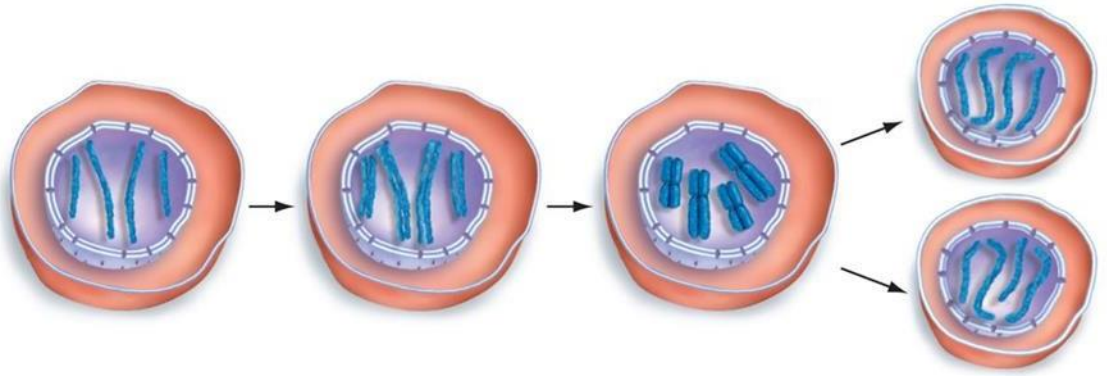
Mitosis & Meiosis = nuclear divisions

Cytokinesis = division of cytoplasm

Mitosis & Cytokinesis

- Goal – to produce 2 genetically identical daughter cells

Parent cell



2 genetically identical daughter cells

One round of nuclear division

Why would you want your somatic cells to be able to undergo mitosis?

Without mitosis (and cytokinesis)

- All organisms (including you) would be single celled;
- You could not grow
- Dead and damaged cells and worn out cells could not be replaced;

https://en.wikipedia.org/wiki/Zygote

Table 4.1.1: Life Span of Cells in Selected Organs of the Human Body

Data after KLIMA 1967, RUCKER 1967

Organ	Average life span (days)
Stomach (pylorus)	1.8– 1.9
Stomach (cardia)	9.1
Small intestine	1.3– 1.6
Liver	10.0– 20.0
Large intestine	10.0
Rectum	6.2
Anus	4.3
Trachea	47.5
Lungs (alveoli)	8.1
White blood cells	1.0– 3.0
Red blood cells	120.0
Bladder	64.0
Epidermis	
Lips	14.7
Soles	19.1
Abdominal skin	19.4
Ear	34.5
Nervous system	No regeneration

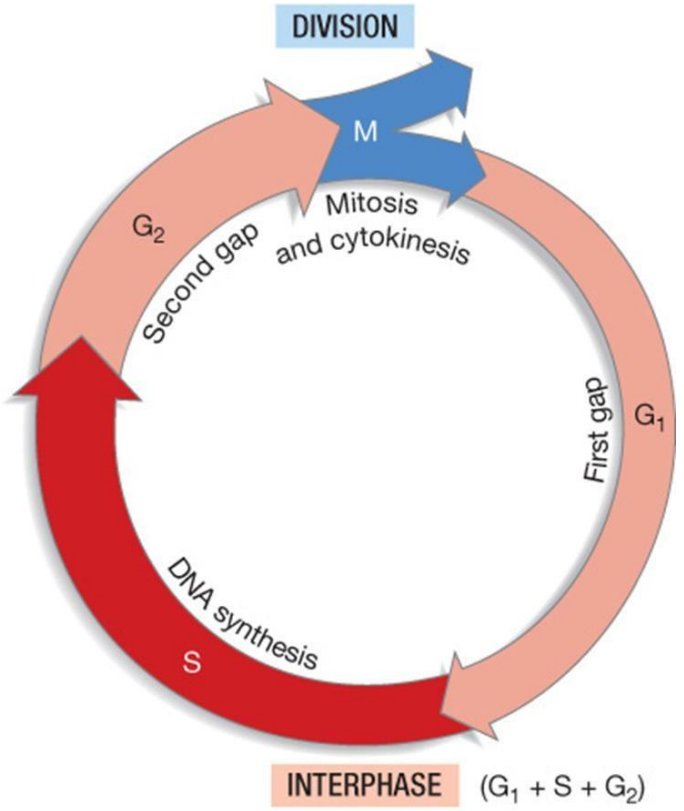
https://bionumbers.hms.harvard.edu/bionumber.aspx?s=n&v=2&id=101940

Note – red blood cells and (most) white blood cells cannot divide – not testable

Some eukaryotic organisms also <u>reproduce</u> via mitosis
e.g. mosses produce spores

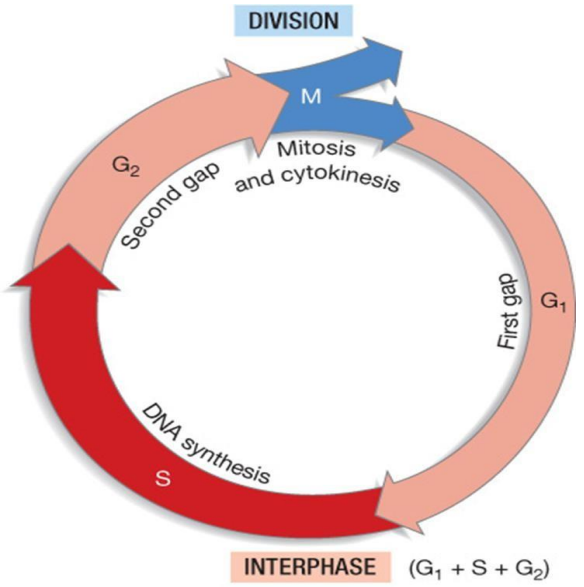
The Cell Cycle

- Mitosis is part of the cell cycle
- Cell cycle is defined as an ordered sequence of events that a cell goes through between one cell division and the next. (not testable)
- Cell cycle has two major phases: Interphase and Mitosis plus cytokinesis



Interphase

- Interphase = longest part of a cell's life.
- In Interphase, the cell is performing its normal functions (e.g., being a skin cell, lung cell, liver cell, etc.)
- This is also when the cell will start to prepare to divide if given the signal to divide.
- 3 stages to Interphase: G1 phase
  S phase
  G2 phase

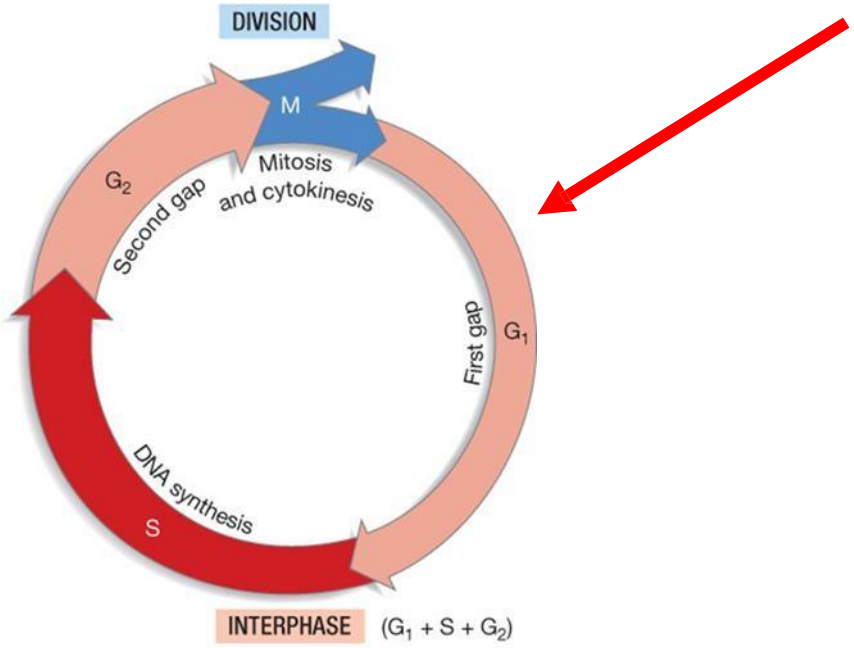


G1 stage

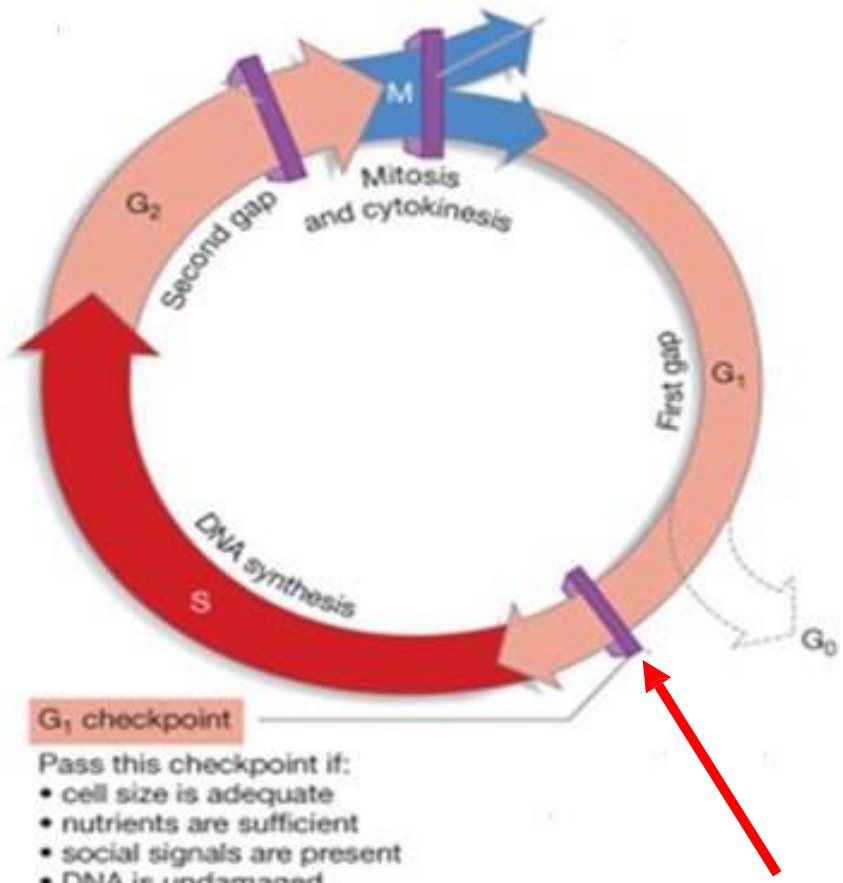
G1 – if cell is signaled to start dividing, cell begins:

- growing
- duplicating organelles
- accumulating nucleotides
- obtains energy reserves

i.e., gathering all of the resources needed for the DNA duplicate and for the cell to divide



G1 Checkpoint (not testable)



Before the cell can enter the next phase (S phase), the cell must pass checkpoint 1.

- Is the cell size adequate?
- Nutrients/cytoplasm sufficient?
- DNA undamaged?
- Social signals present (go ahead)?

The G1 checkpoint is when a cell irreversibly commits to the cell division process.

Many types of cancers involve defects in the G1 checkpoint, i.e. cells begin dividing in the absence of the “go ahead” signal.

11.14

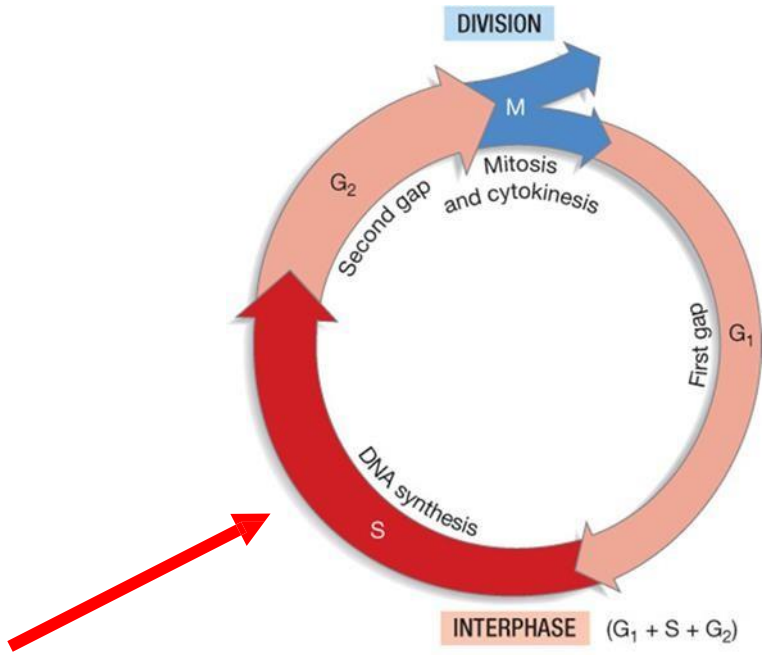
Fun fact: Not testable

- In humans, the DNA itself signals that it has been damaged by increasing the activity of certain genes, including the tumor suppressor gene p-53
- p-53 gene sometimes called “the guardian of the genome” – determines whether the cell can continue into S-phase to protect the genome from a proliferation of damaged DNA.

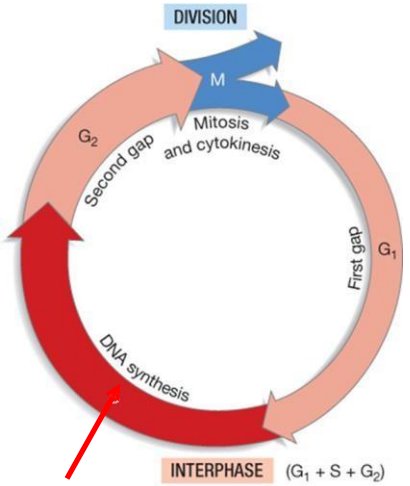
S phase (or stage)

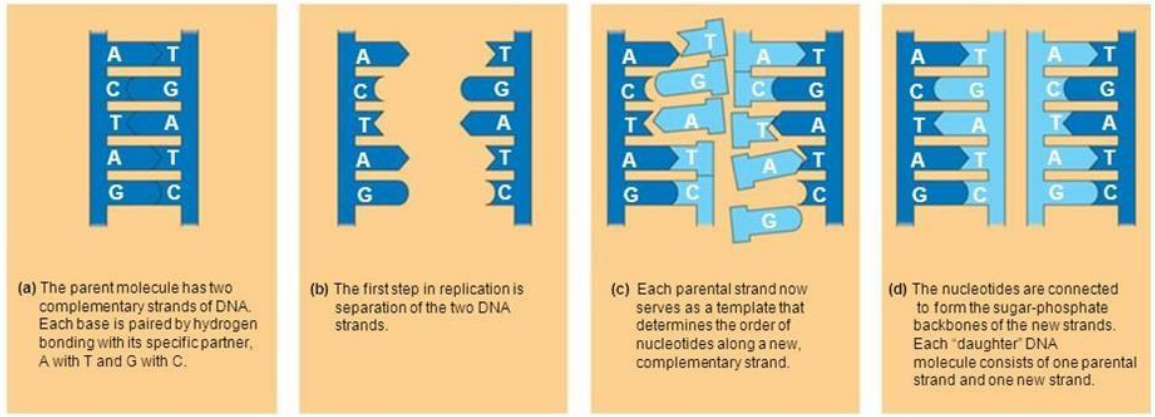
S= Synthesis

- This is when the DNA replicates (sister chromatids are formed)



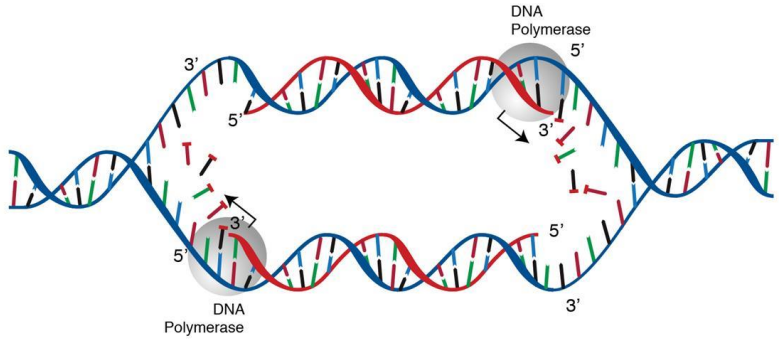
S-stage





Replicated chromosome

Consists of two copies of the same chromosome.



https://www.genome.gov/genetics-glossary/DNA-Replication

This is called semi-conservative DNA replication because each daughter DNA molecule consists of a **one old strand and one new** strand.

iClicker Question

Students in Dr. Jennifer Klenz's genetics class (Biol 234) were asked to draw two replicated homologous chromosomes <u>just after DNA replication</u>. One homolog carries the B2 gene and one homolog carries the B5 gene. Which diagram is correct?

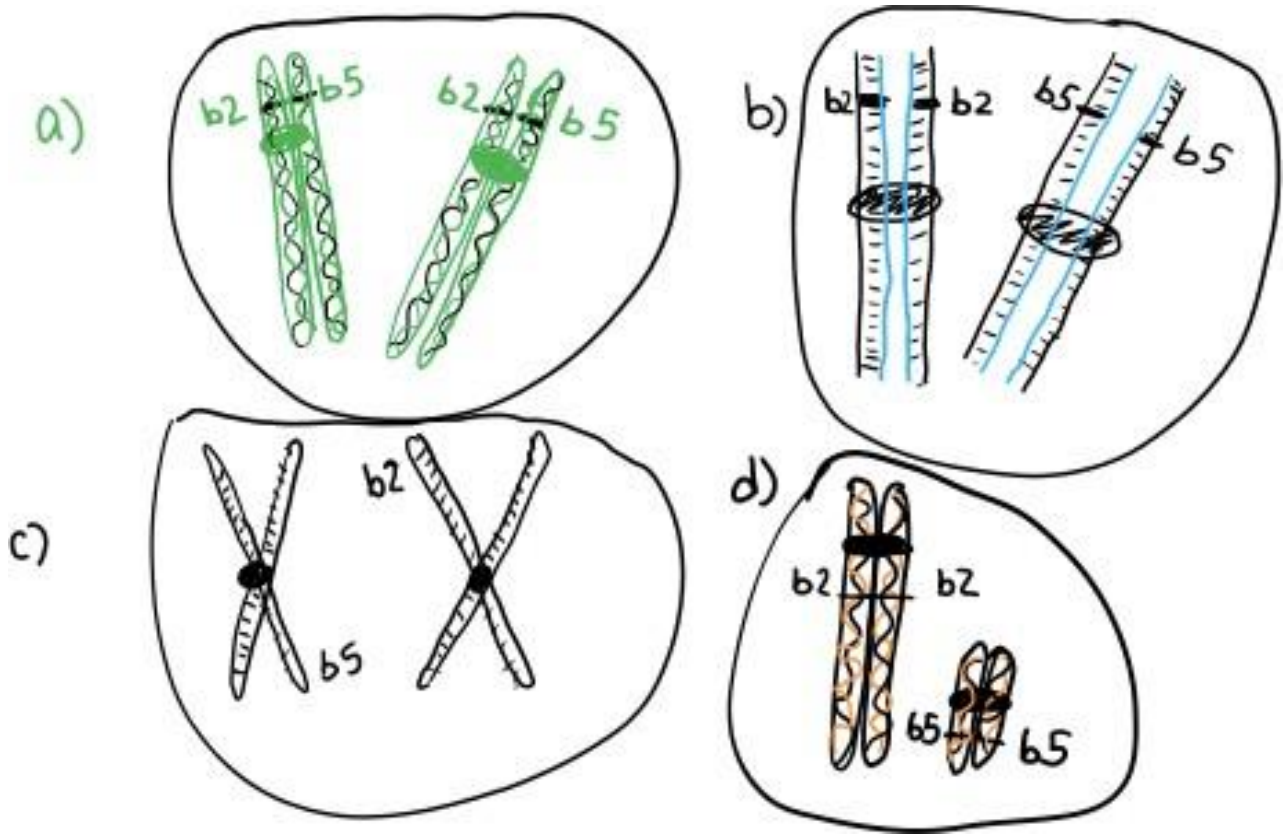
A. = a

B. = b

C. = c

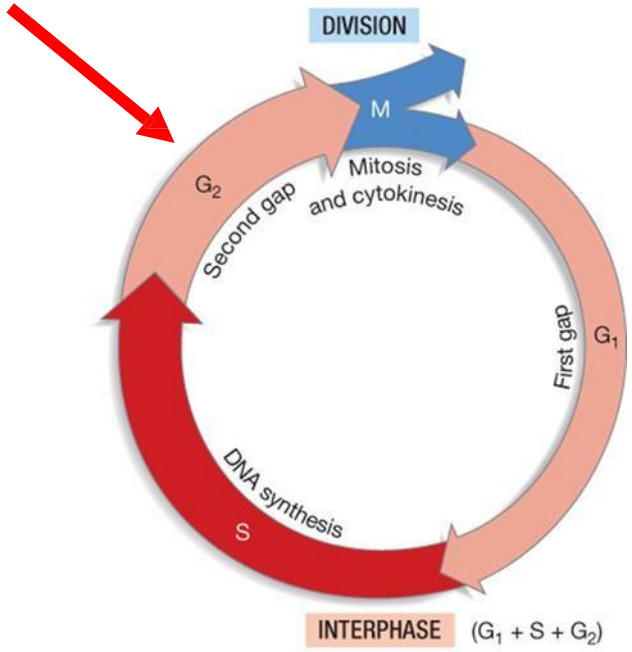
D. = d

E. = not sure

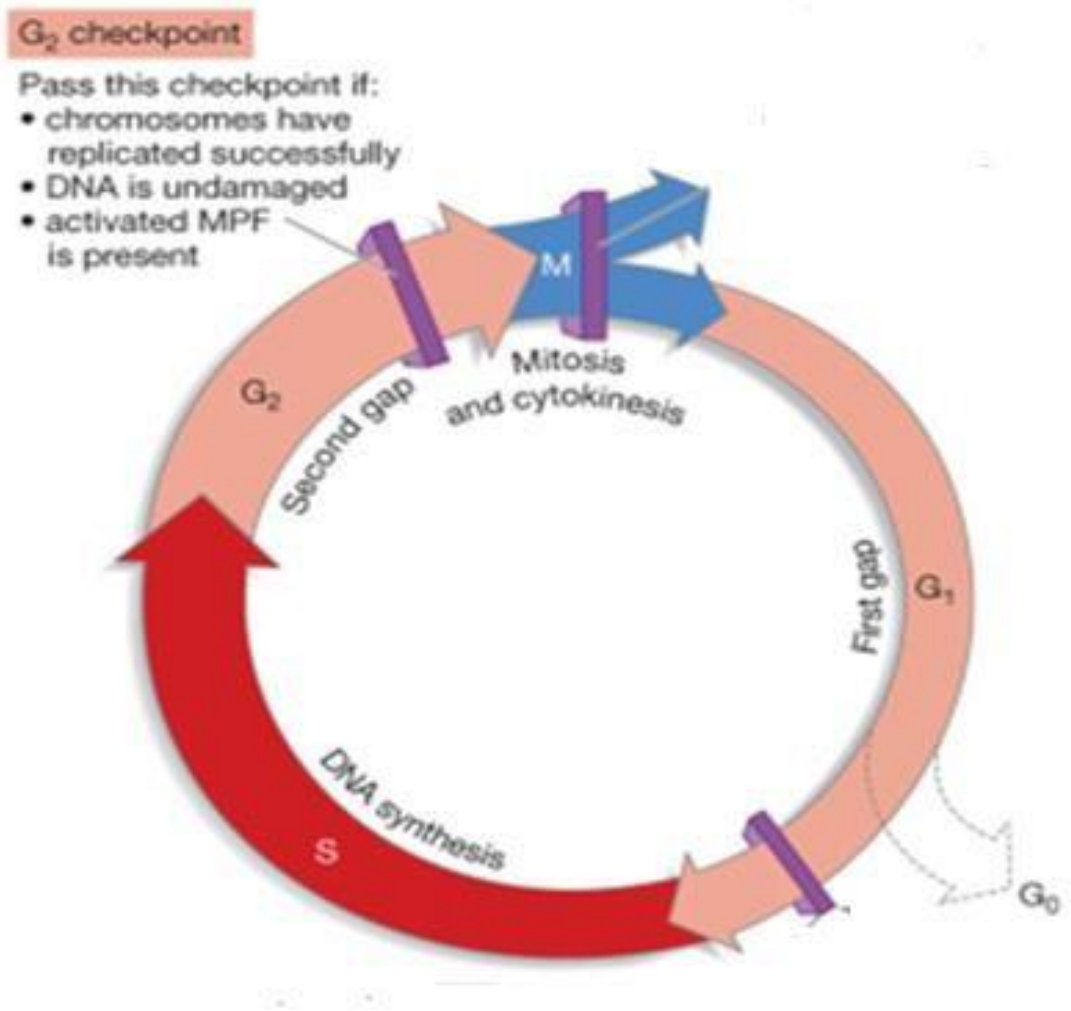


G2 stage

- final preparations are made before cell divides.
- may be additional growth
- more organelles may be duplicated



G2 checkpoint (not testable)



Before entering mitosis the cell must pass another checkpoint.

- Have chromosomes replicated successfully?
- Is the DNA undamaged?

- Cell size and reserves are also checked

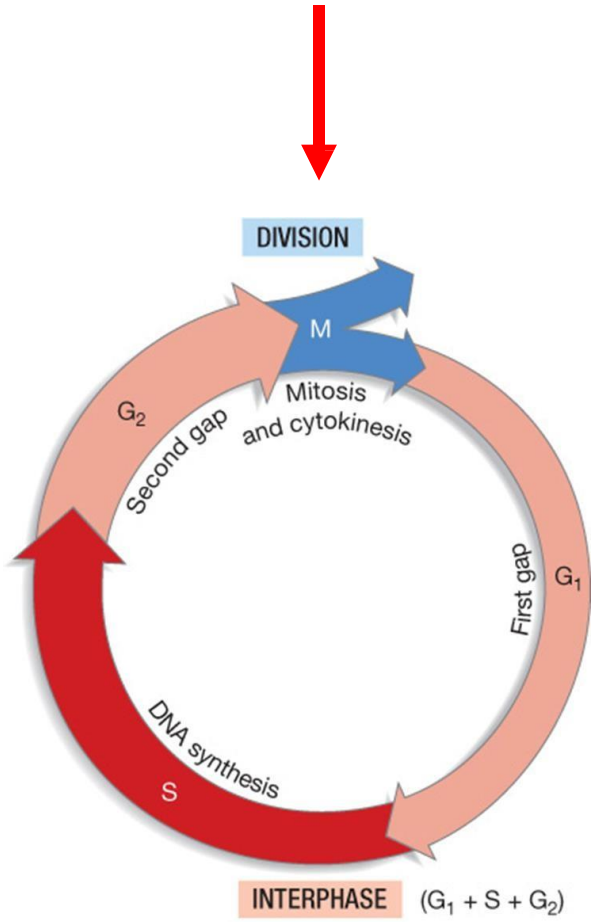
M stage (Mitosis)

Goal: To produce 2 genetically identical daughter cells

This requires that the content of the nucleus and the cytoplasm must be divided equally.

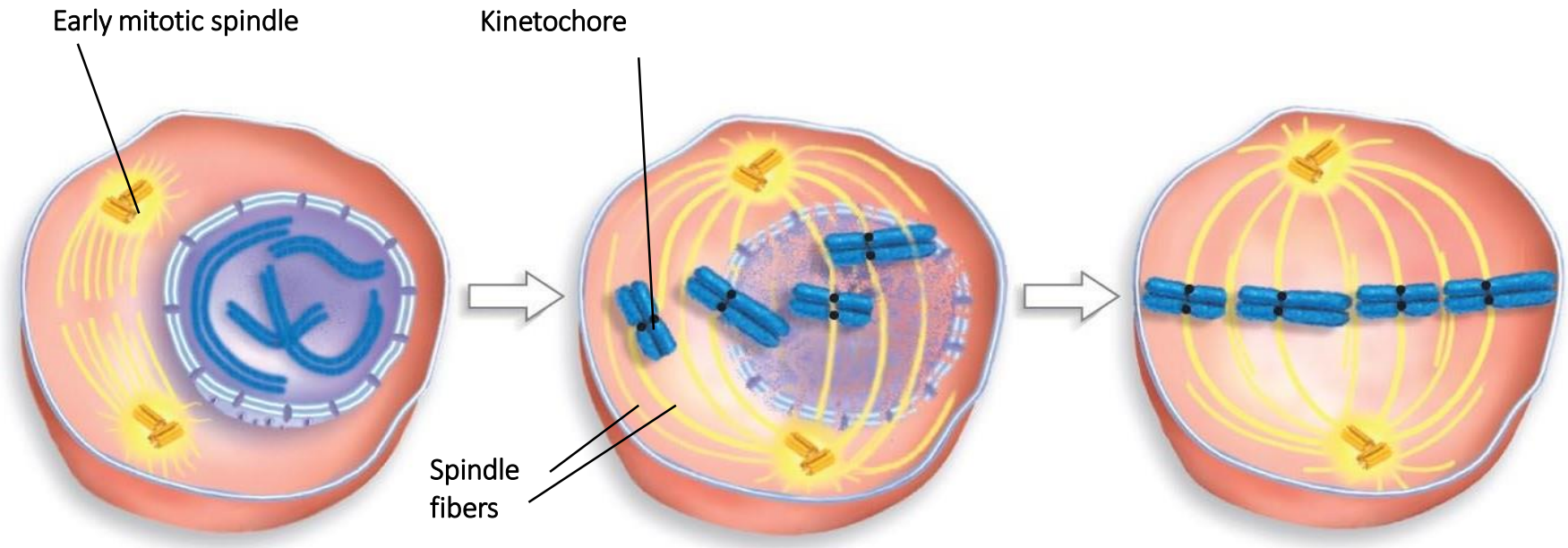
Mistakes can result in:

- impaired cell function, cell and organ disease or cell death
- worst case scenario – uncontrolled cell division = cancer
    - as many as 50% of cancers may be caused by errors in the cell cycle/mitosis (British Society for Cell Biology)



Mitosis – one round of nuclear division (4 or 5 phases)

Prophase > Prometaphase > Metaphase > Anaphase > Telophase



2. Prophase: Chromosomes condense, and mitotic spindle begins to form.

3. Prometaphase: Nuclear envelope breaks down. Spindle fibers contact chromosomes at kinetochore.

4. Metaphase (M=middle): Chromosomes complete migration to middle of cell.

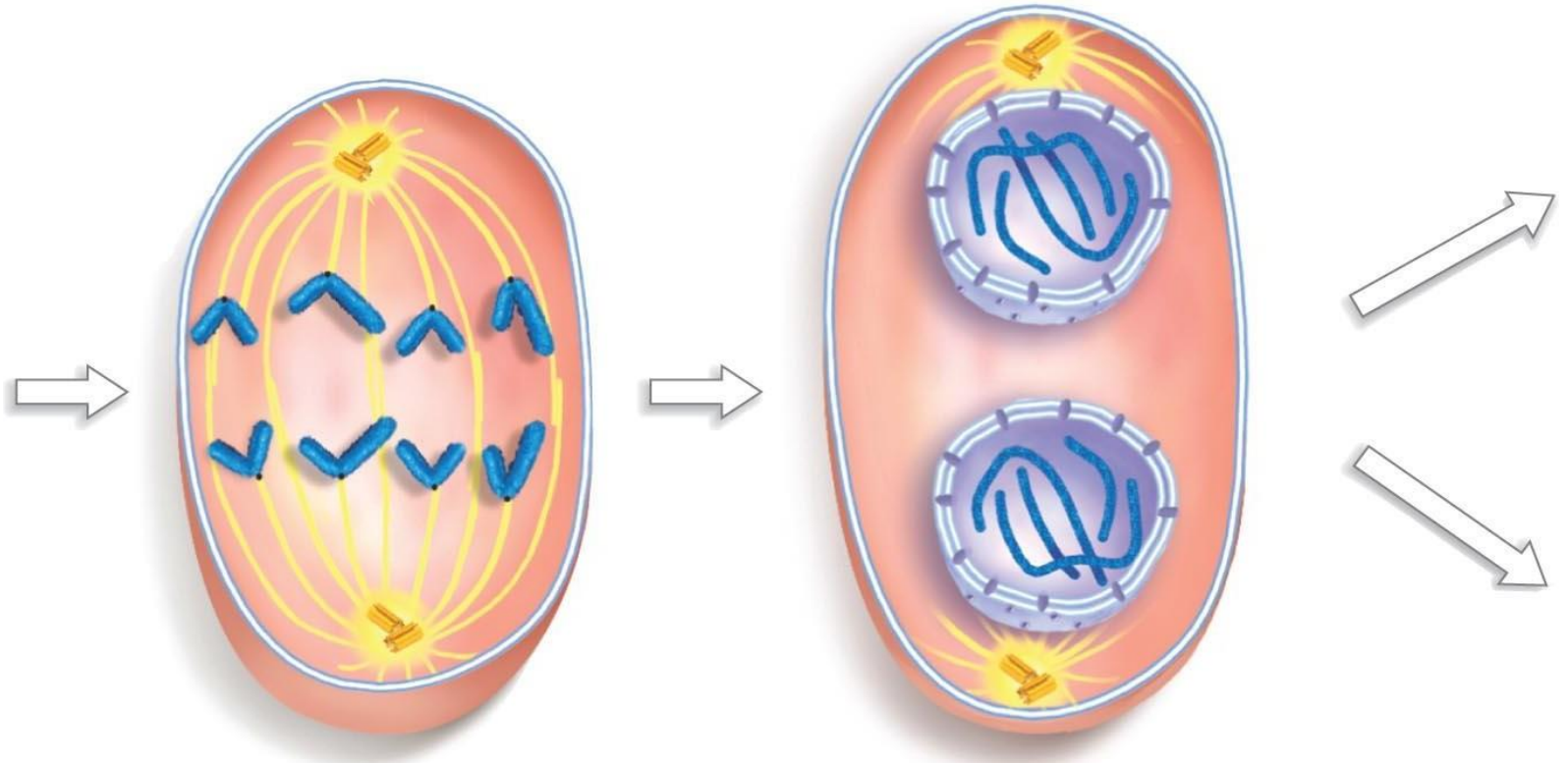
M checkpoint (not testable)

- Near the end of metaphase the M checkpoint occurs (also called the spindle fiber checkpoint).

- Are the spindle fibers correctly attached to the kinetochore of each sister chromatids or not? If not, cell division does not proceed.

- Why this checkpoint is important - the next step is Anaphase, when the sister chromatids separate from each other; so, very important that the spindle fibers are correctly attached.

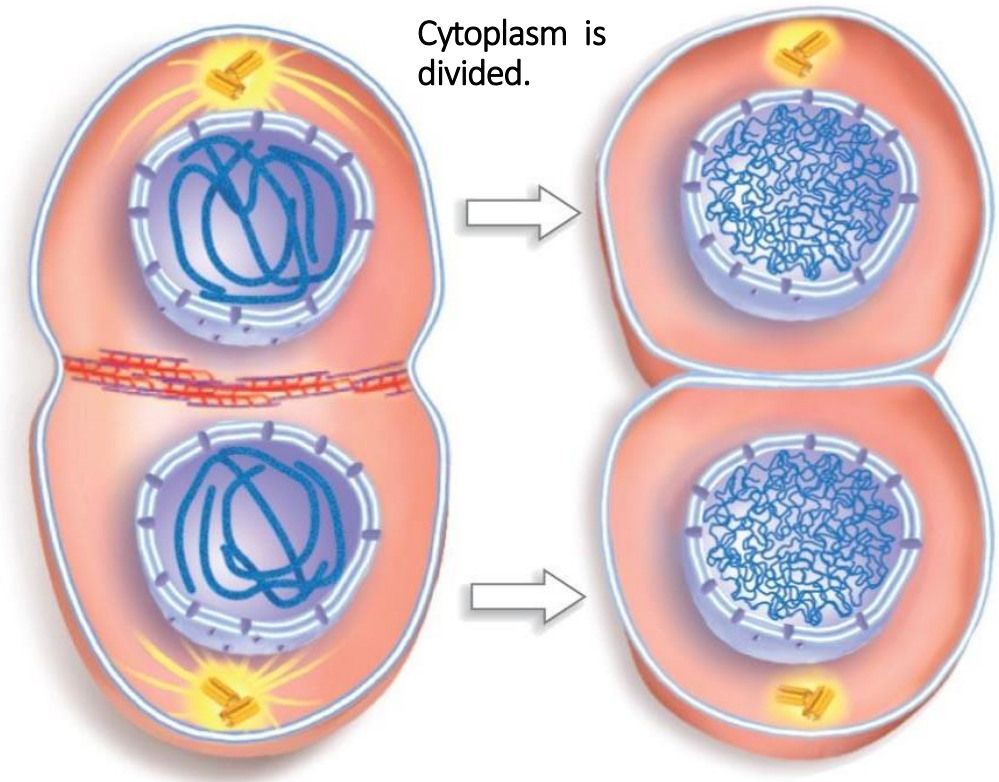
Sister chromatids separate in mitosis



5. Anaphase (A=Apart):
Sister chromatids separate. Upgraded to chromosomes. Chromosomes are pulled to opposite poles of the cell.

6. Telophase: The nuclear envelope re-forms, and the spindle apparatus disintegrates. DNA starts to decondense.

Cytokinesis



7. Cytoplasm is divided: Actin-myosin ring causes the plasma membrane to begin pinching in. (not testable)

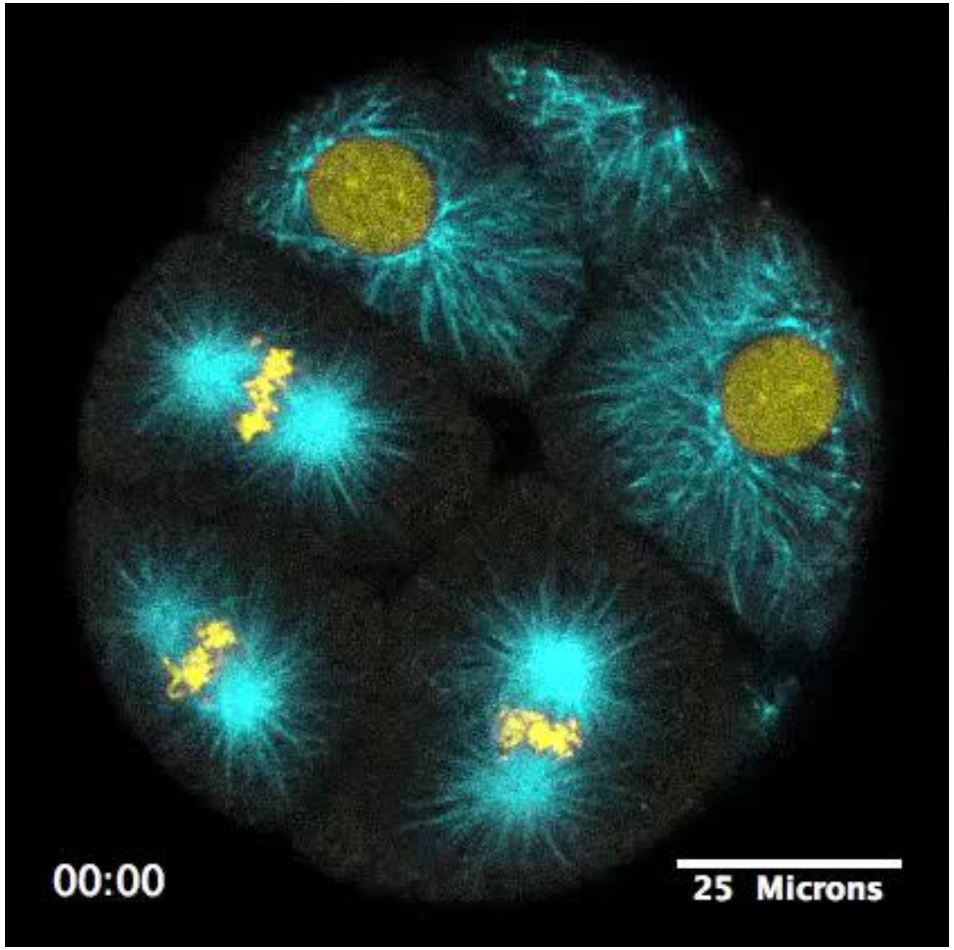
8. Cell division is complete: Two daughter cells form.

Mitosis fingers – sister chromatids

Mitosis happens very quickly (sea urchin cells)

- Watch the two cells near the top first.

- What structures are gold in colour?

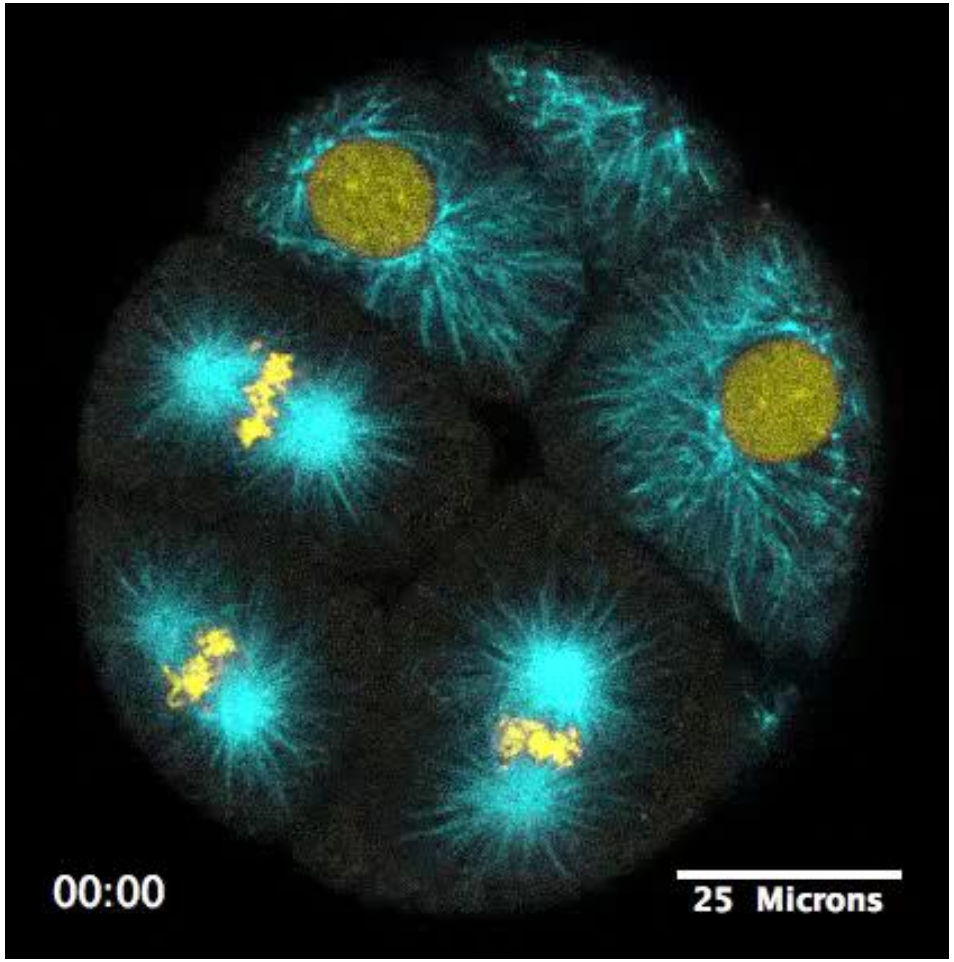


http://cellimagelibrary.org/images/15792?fbclid=IwAR07DWz9LDLIT5MnVzkeEo5_Rzom5U67LFVMrRkKmyxQxLiV0aoGCeEy508

Mitosis

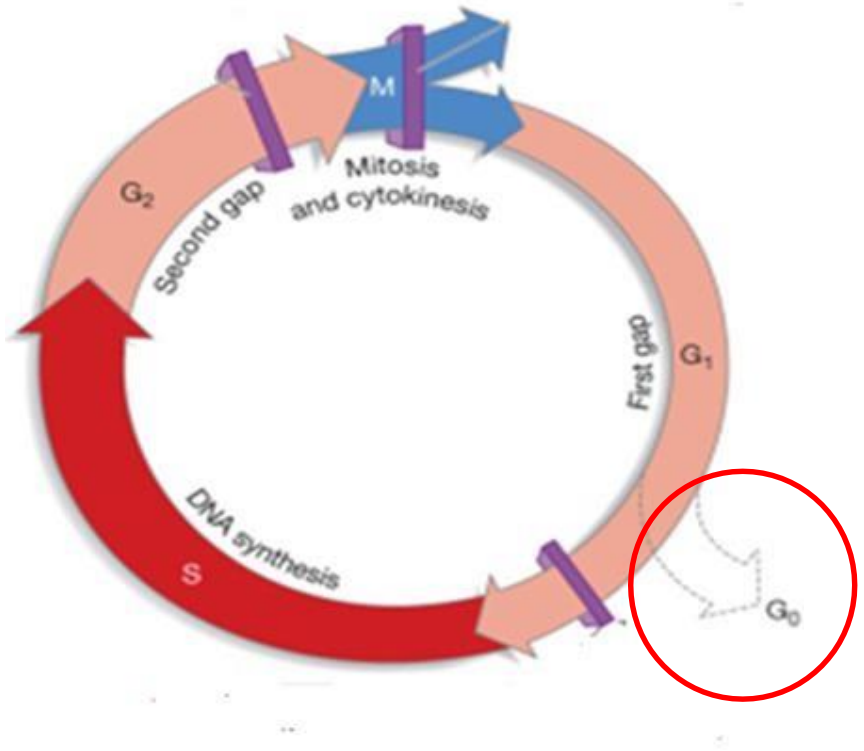
- Watch the three cells near the bottom of the video

- What stage of mitosis are those cells in at the start of the video?



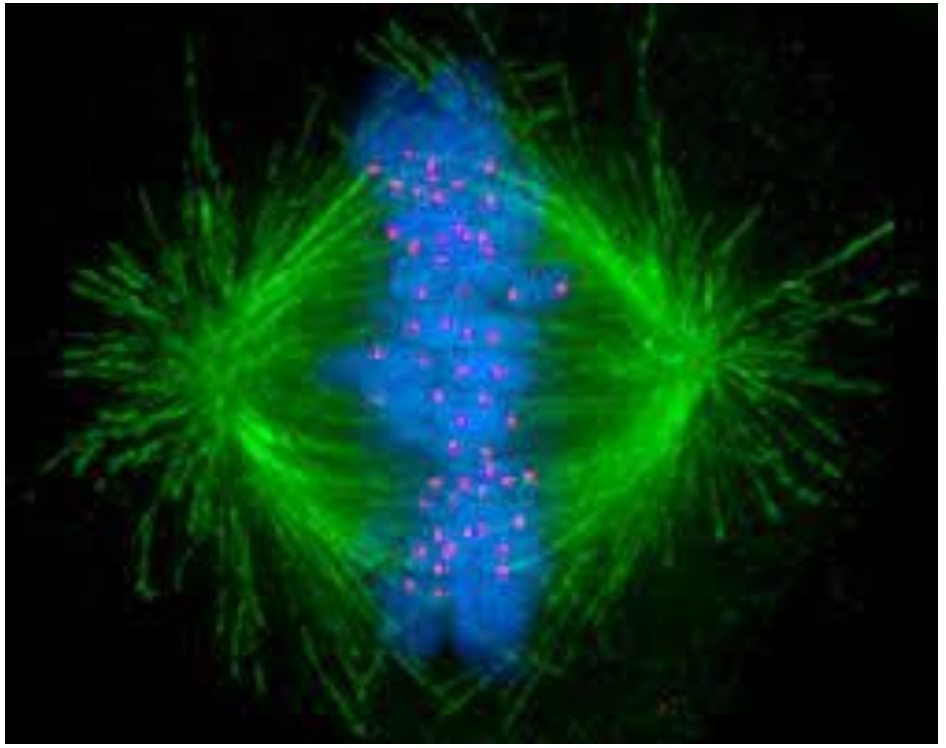
http://cellimagelibrary.org/images/15792?fbclid=IwAR07DWz9LDLIT5MnVzkeEo5_Rzom5U67LFVMrRkKmyxQxLiV0aoGCeEy508

One phase not discussed - G_o (resting phase)



- A cell in G1 phase can enter Go
- A cell in Go is neither dividing nor preparing to divide
- Performs main function indefinitely
- e.g., neurons, muscles cells in your heart are in G_o phase.
- Cells may return to G_1

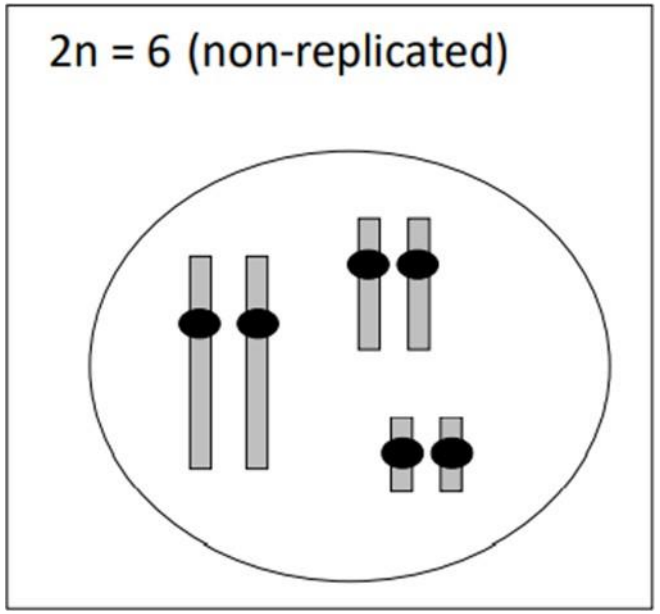
Questions?



https://en.wikipedia.org/wiki/Spindle_apparatus

iClicker Question

The cell is in which stage of the cell cycle?



A. G1

B. S

C. G2

D. Mitosis

E. Not sure

iClicker Question

- From a population of dividing human cells, biologists isolate cells at various phases of the cell cycle. They find some normal cells that have 1.5 times the DNA compared to normal non-dividing human cells. These cells are in which cell-cycle phase?

A. G1 phase
B. S phase
C. G2 phase
D. M phase
E. E phase

iClicker Question

A diploid species of animal has three pairs of chromosomes in the nucleus of their adult cells. How many total molecules of DNA do the nuclei of these cells have during G2 phase?

A. 3
B. 6
C. 12
D. 24
E. a bazillion

Cell Cycle & Mitosis – Learning Objectives

Understand/know:

- The stages of the cell cycle (interphase and mitosis)
  - How ploidy, number of chromatids and number of DNA molecules changes through mitosis

Be able to:

- Interpret and/or draw diagrams of chromosomes at different stages of the cell cycle and stages of cell division (mitosis).
- Identify and diagram the stages of the cell cycle and mitosis.

You should now be able to complete:

- Quiz #1
- Worksheet #1

Due date will be changed to Sunday, January 22nd @ 11:59 pm.

Next Class – Mitosis & Meiosis