



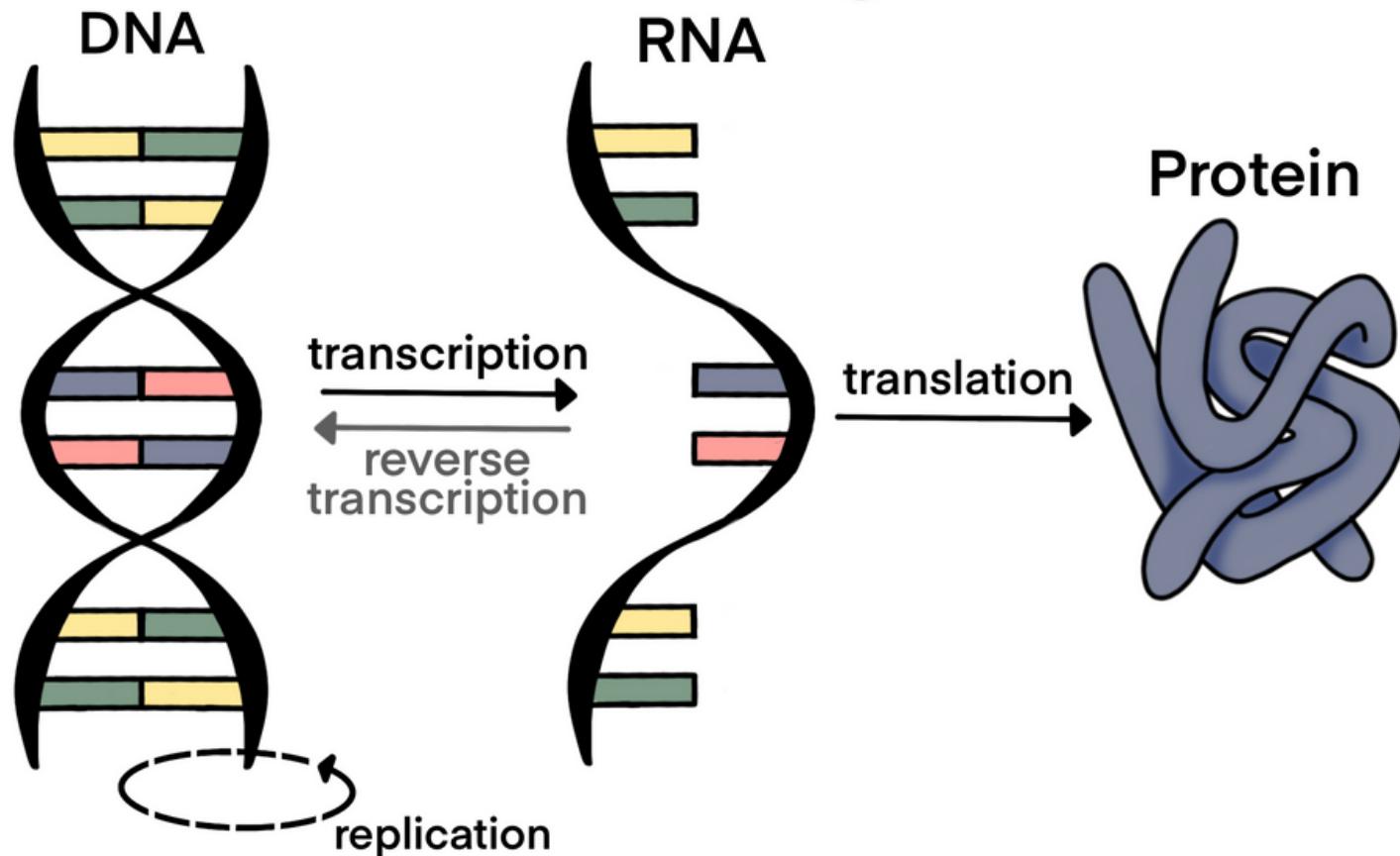
Materials of Life

Energy of Life

Information of Life



central dogma





DNA, Cell Division, & Inheritance Unit

- *Remember from chapter 1:*
 - life reproduces itself (with DNA), grows, & develops
- *Remember from chapter 3:*
 - all cells contain DNA that is duplicated & passed on
- Chapter 7 = How do we get from DNA to walking & talking us?
- Chapter 8 = How does DNA duplicate itself & how do we grow?
- Chapter 9 = How do we pass on DNA to reproduce?



Chapter 7: DNA Structure & Gene Function

- What is the structure of DNA?
- How does DNA code for traits?
- How are genes regulated?
- How do mutations affect genes?

Corresponds with OpenStax Biology 2e Chapters 15 & 16



How does DNA code for traits?

- Remember: the nucleus of a cell contains its chromosomes, which are made up of DNA (& proteins)

- Sequences of DNA that code for a specific protein/trait = **genes**

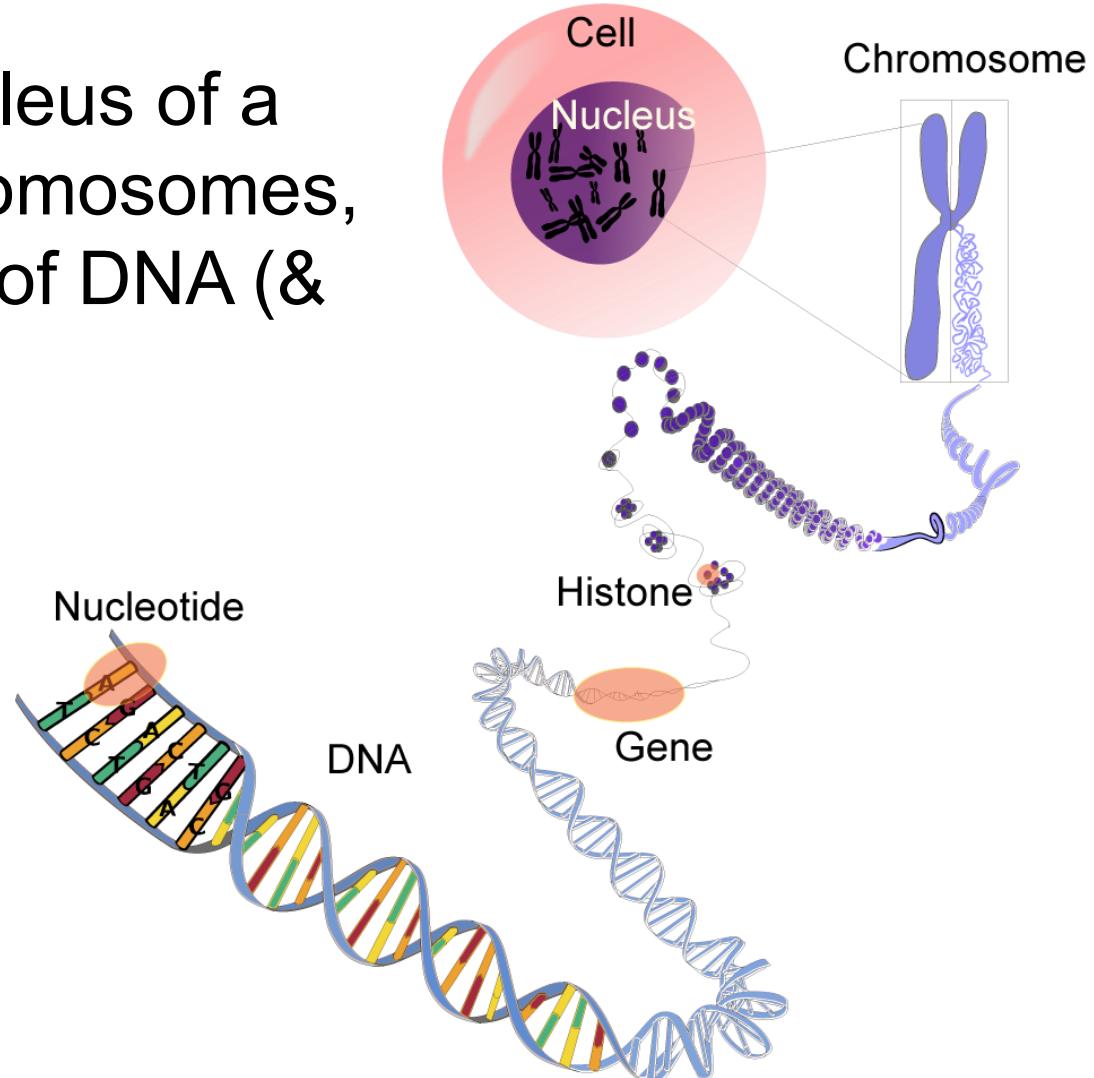
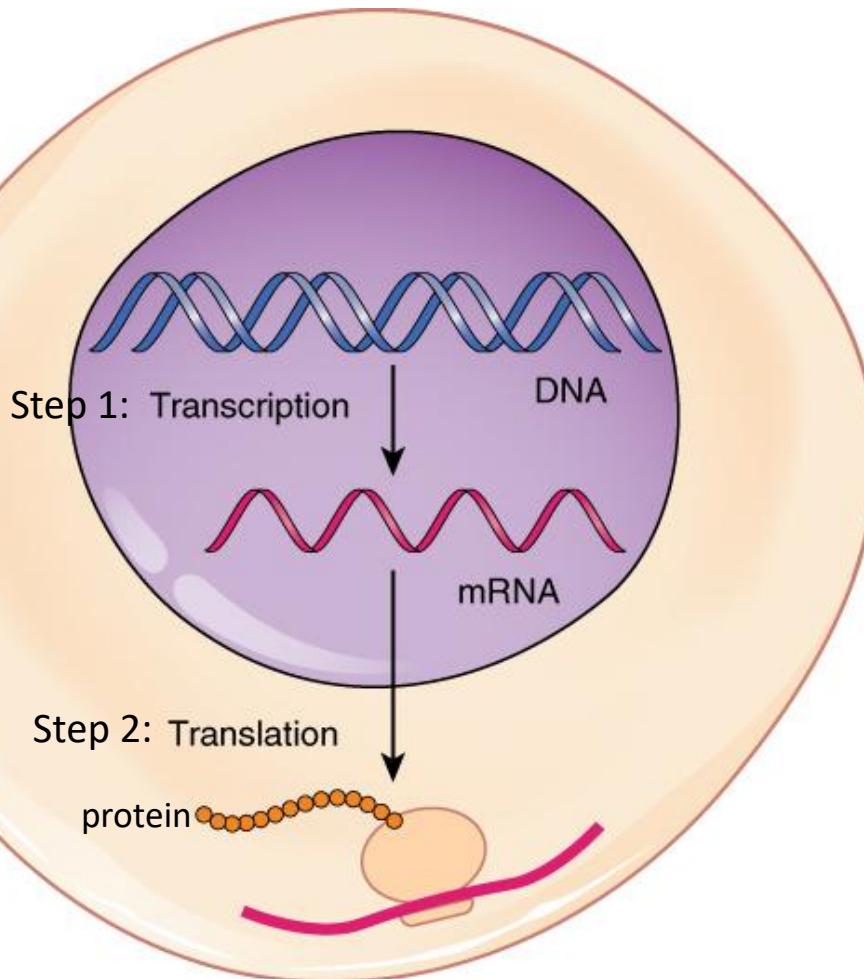


Figure: <https://commons.wikimedia.org/wiki/File:DNA-terminology.png>

- Getting from DNA to proteins is a 2 step process



Step 1: DNA → mRNA
is called **transcription**

Step 2: RNA → protein
is called **translation**

Figure: https://commons.wikimedia.org/wiki/File:0328_Transcription-translation_Summary.jpg



- DNA codes for proteins/traits: STEP 1 = transcription

- **Transcription = DNA → mRNA**

- the information in DNA is copied to RNA inside the nucleus
 - the RNA is acting as the middle man, so it is called **messenger RNA (mRNA)**
 - *there are other types of we'll bring up soon*

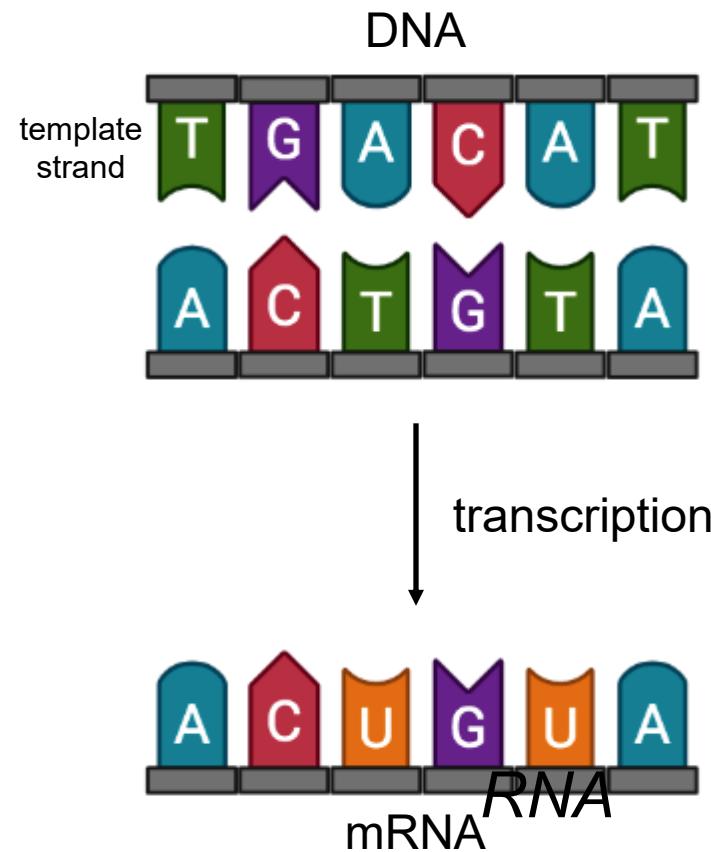


Figure made using BioRender



How do we actually transfer info from DNA to RNA?

- Transcription has 3 stages
- 1. **Initiation:** enzymes unzip the DNA double helix starting at the **promoter** (signals the start of a gene), exposing the **template strand** (the strand that will be copied)

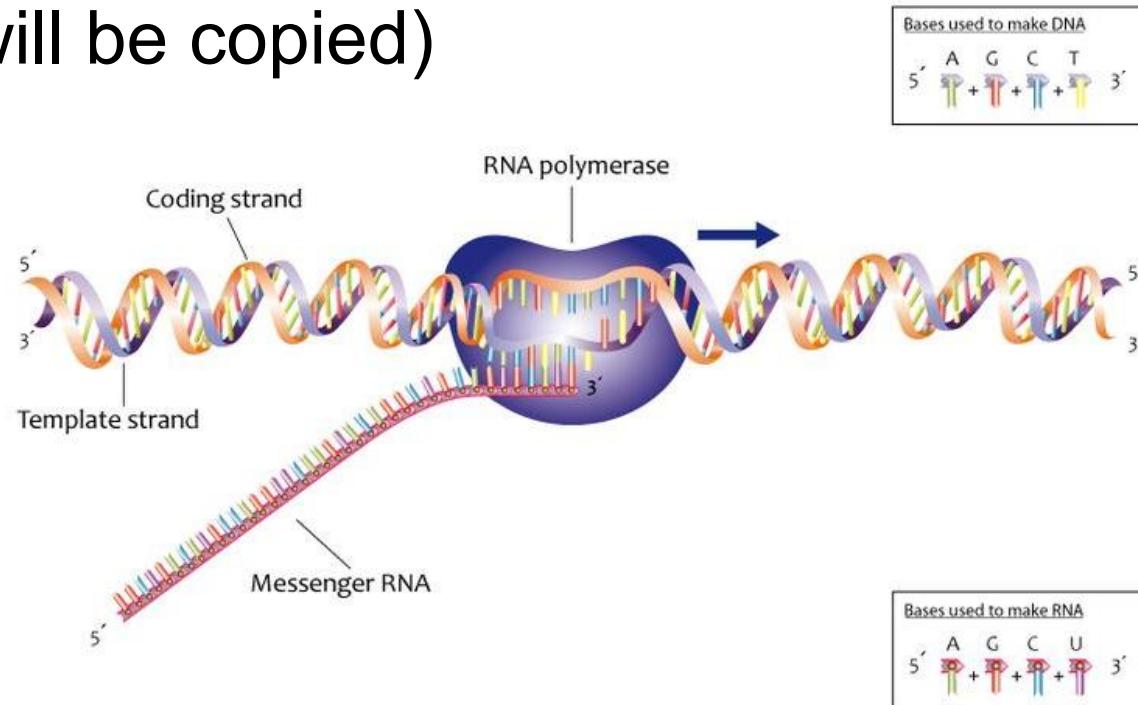


Figure: [https://commons.wikimedia.org/wiki/File:Process_of_transcription_\(13080846733\).jpg](https://commons.wikimedia.org/wiki/File:Process_of_transcription_(13080846733).jpg)



- Transcription has 3 stages

- **2. Elongation:** the enzyme **RNA polymerase** moves along the template strand pairing RNA with DNA (e.g. A with U & C with G), lengthening the mRNA strand

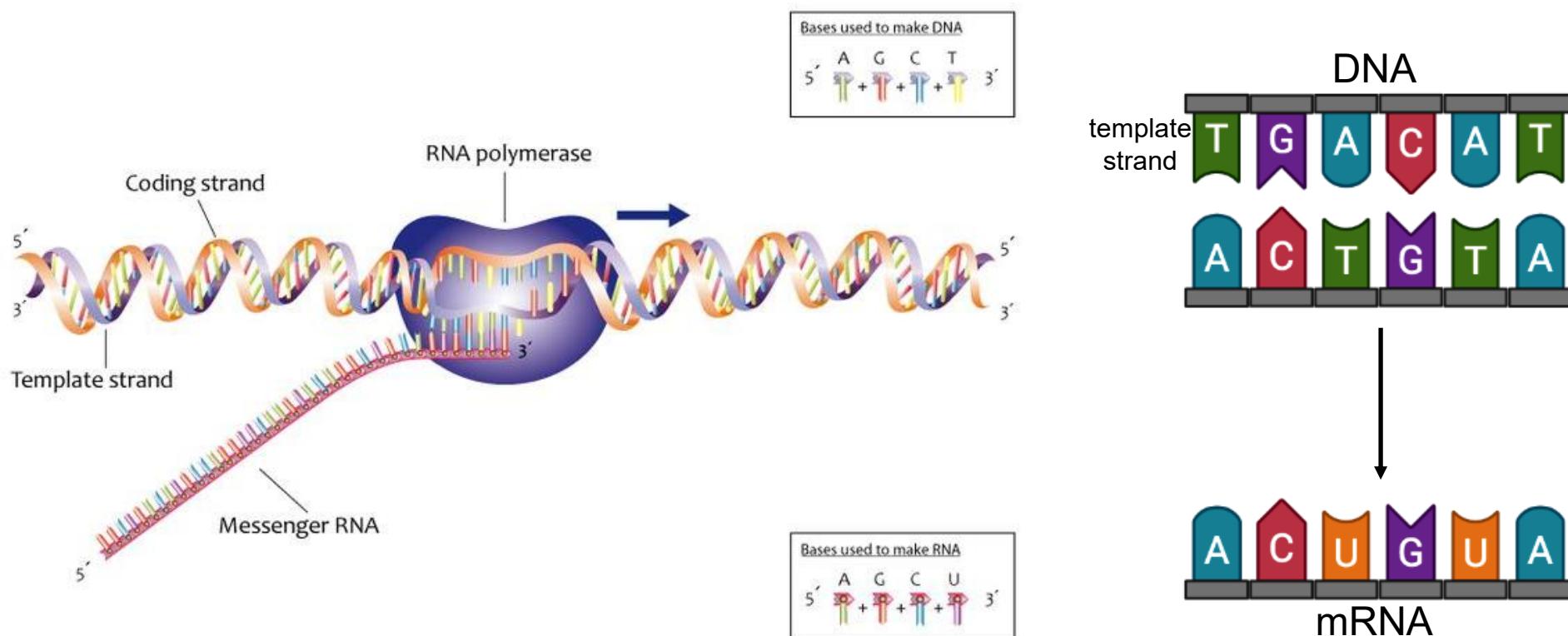
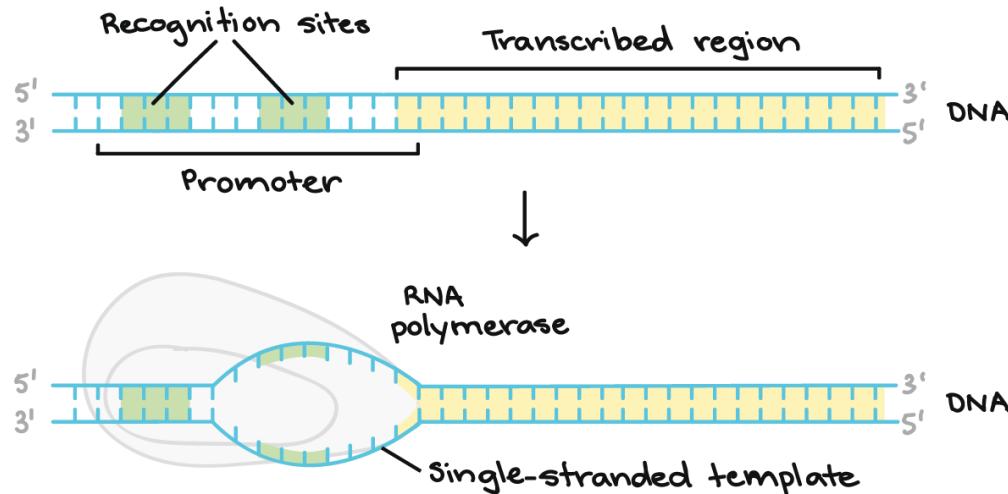


Figure: [https://commons.wikimedia.org/wiki/File:Process_of_transcription_\(13080846733\).jpg](https://commons.wikimedia.org/wiki/File:Process_of_transcription_(13080846733).jpg)



transcription initiation



transcription elongation

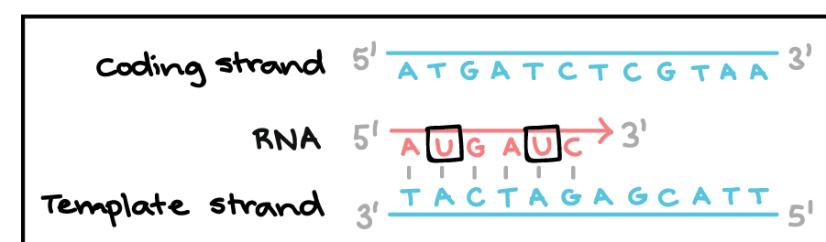
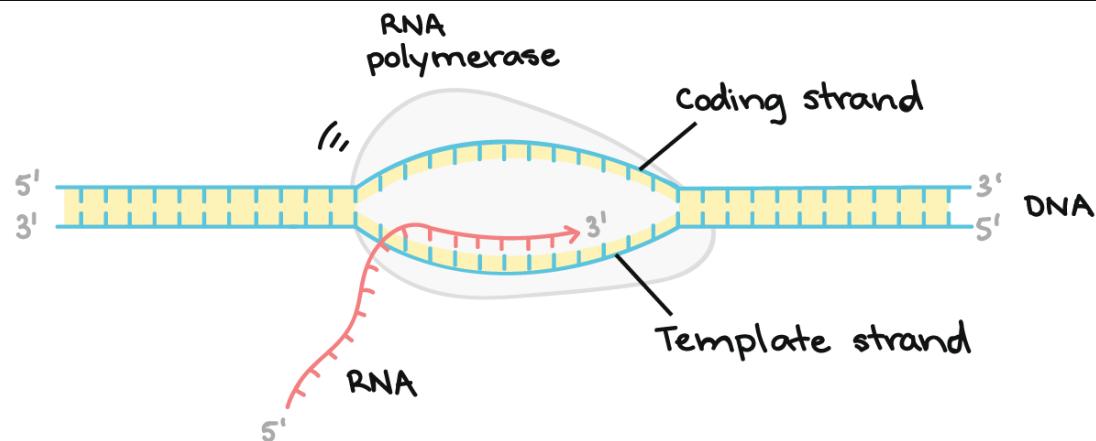


Figure: <https://www.khanacademy.org/science/biology/gene-expression-central-dogma/transcription-of-dna-into-rna/a/stages-of-transcription>



- Transcription has 3 stages
- 3. Termination: when the **terminator signal** is reached RNA, DNA, & RNA polymerase fall apart
 - DNA zips back up to a double strand
 - we now have an exact mRNA copy of a gene

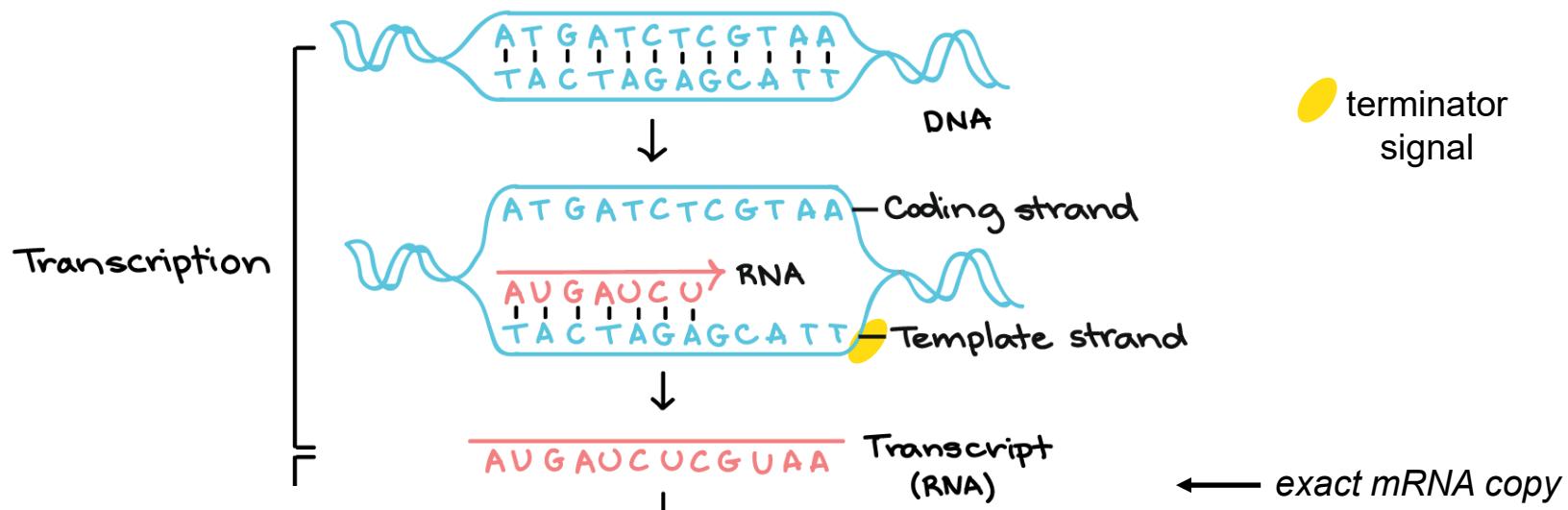
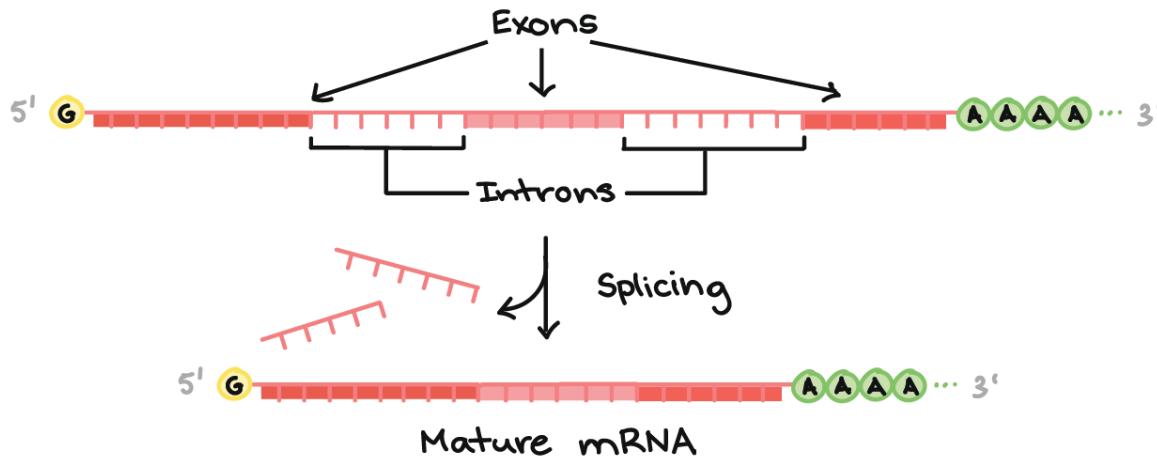


Figure: <https://www.khanacademy.org/science/biology/gene-expression-central-dogma/transcription-of-dna-into-rna/a/stages-of-transcription>



We now have an exact copy of the gene, but mRNA is not ready to leave the nucleus yet

- A gene has 2 kinds of segments: introns & exons
 - **exons** = truly coding nucleotides in DNA or RNA
 - **introns** = nucleotides that are not part of the code – must be cut out of mRNA after it's made



The process of cutting out introns = **RNA splicing**

Figure: <https://www.khanacademy.org/science/biology/gene-expression-central-dogma/transcription-of-dna-into-rna/a/eukaryotic-pre-mrna-processing>



RNA Splicing

- Immediately after transcription but before splicing, we have **pre-mRNA** – not spliced yet
- Enzymes in nucleus cut out the introns & splice together the exons to make a full strand of only coding mRNA
 - called finished or **post-mRNA**

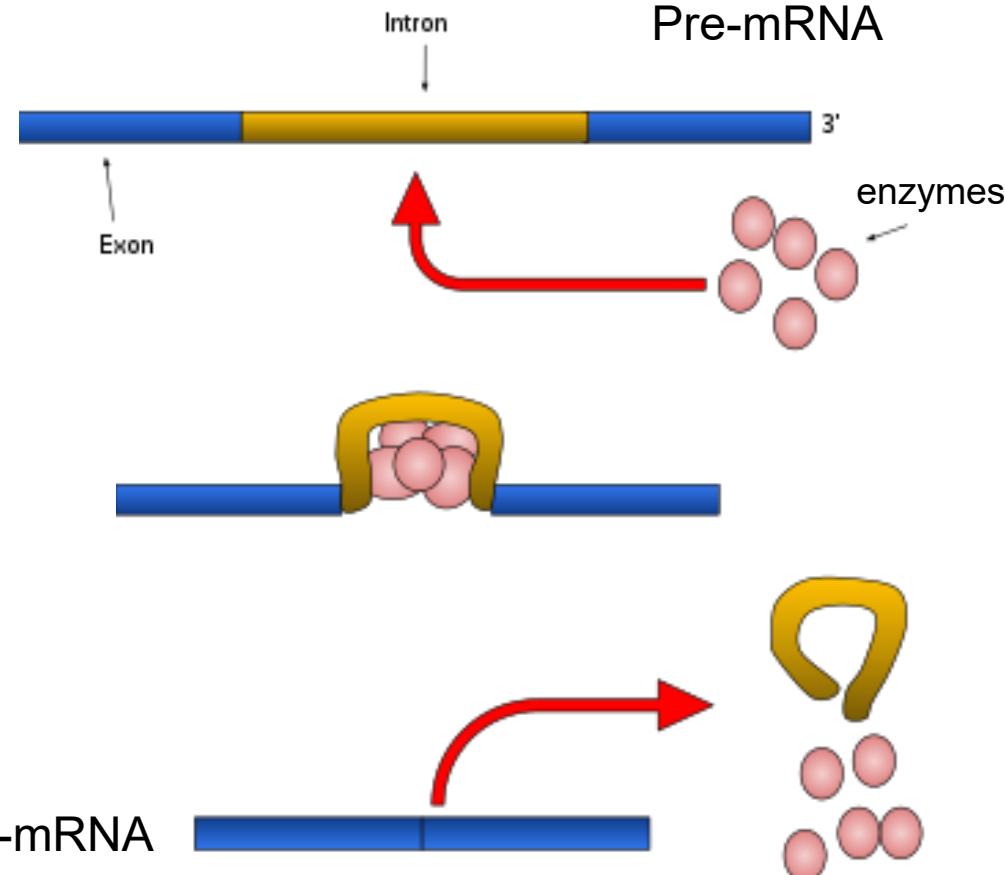


Figure: <https://en.wikipedia.org/wiki/File:Splicesome.svg>



Seems inefficient to carry around DNA we don't need – why would RNA splicing be useful?

- Exons aren't always spliced back together exactly
- This means the RNA splicing process can allow for 1 gene to make multiple different versions of a protein – very useful!

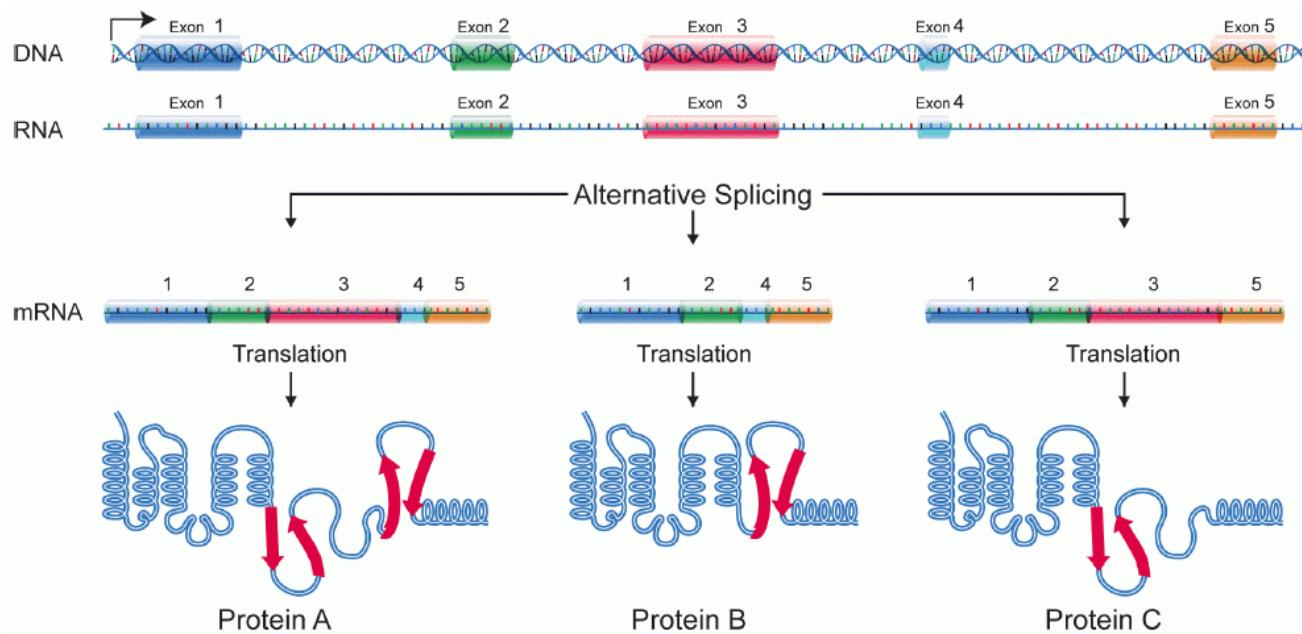


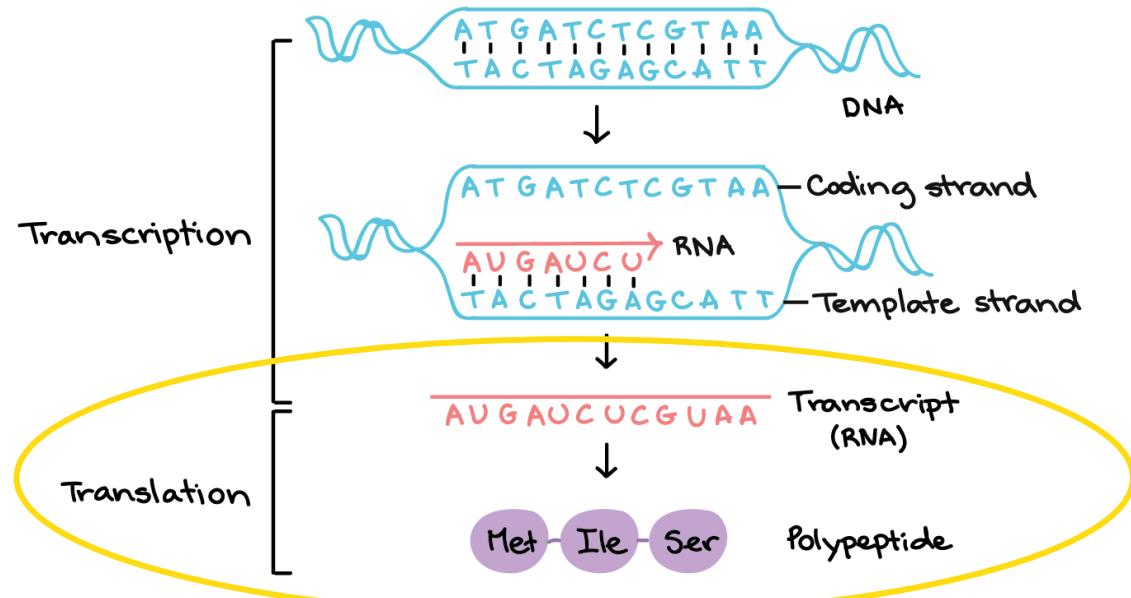
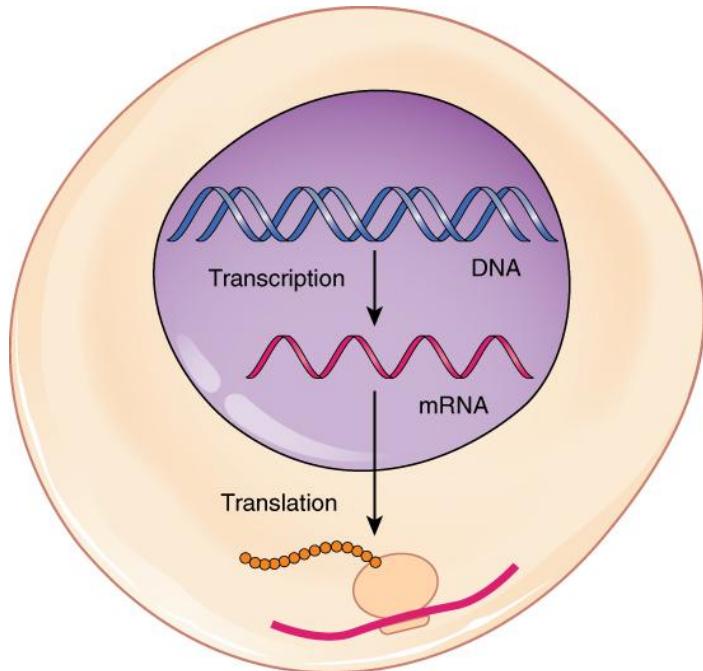
Figure: <https://www.khanacademy.org/science/biology/gene-expression-central-dogma/transcription-of-dna-into-rna/a/eukaryotic-pre-mrna-processing>



DNA codes for proteins/traits: STEP 2 = translation

– Translation = mRNA → protein

- mRNA moves out of the nucleus into the cytoplasm to direct protein synthesis (make a protein/polypeptide)





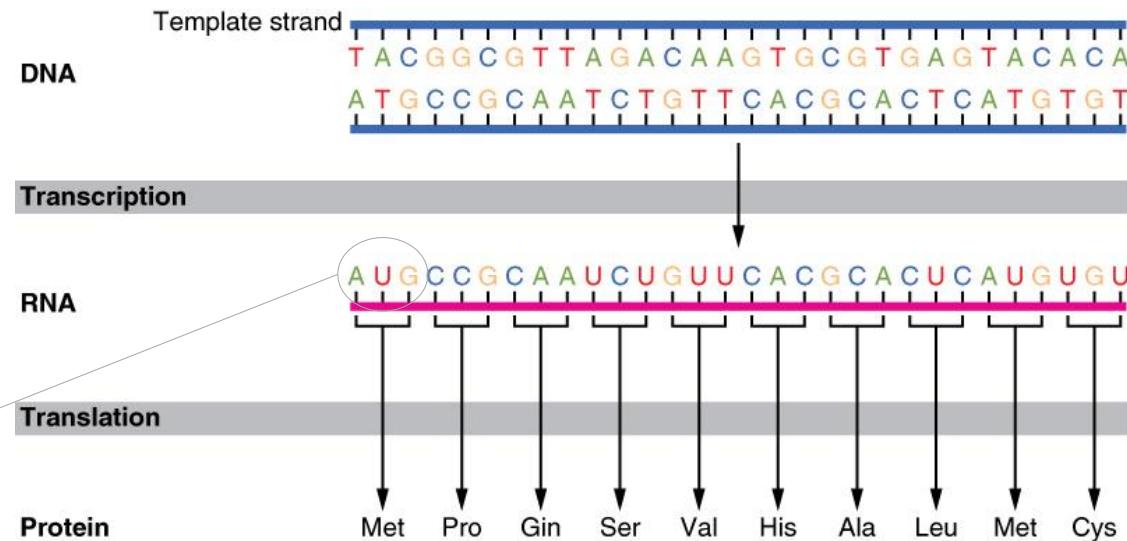
We can try to match up every 2 bases of RNA to every 1 amino acid (e.g. AG = 1 amino acid)

- there are only 16 possible 2-base combinations
 - we'd only use 16 amino acids & have 4 extra

- Must use 3 bases to code for 1 amino acid

- e.g. AUG will code for 1 amino acid

- each 3-base code is called a **codon**



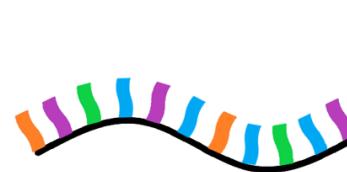
Translation requires use of the genetic code

- Each **codon** specifies that a unique amino acid should be added to build the protein

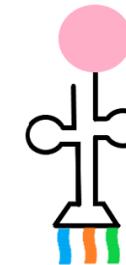
		Second Base							
		U	C	A	G				
First Base	U	UUU UUC UUA UUG	UCU UCC UCA UCG	UAU UAC UAA UAG	UGU UGC UGA — STOP UGG — Trp	U C A G			
	C	CUU CUC CUA CUG	CCU CCC CCA CCG	CAU CAC CAA CAG	CGU CGC CGA CGG	U C A G			
	A	AUU AUC AUA AUG — Met or Start	ACU ACC ACA ACG	AAU AAC AAA AAG	AGU AGC AGA AGG	U C A G			
	G	GUU GUC GUA GUG	GCU GCC GCA GCG	GAU GAC GAA GAG	GGU GGC GGA GGG	U C A G			
Third Base									



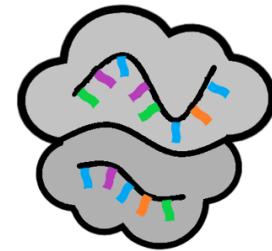
How do we physically match RNA to amino acids?



Messenger RNA



Transfer RNA



Ribosomal RNA

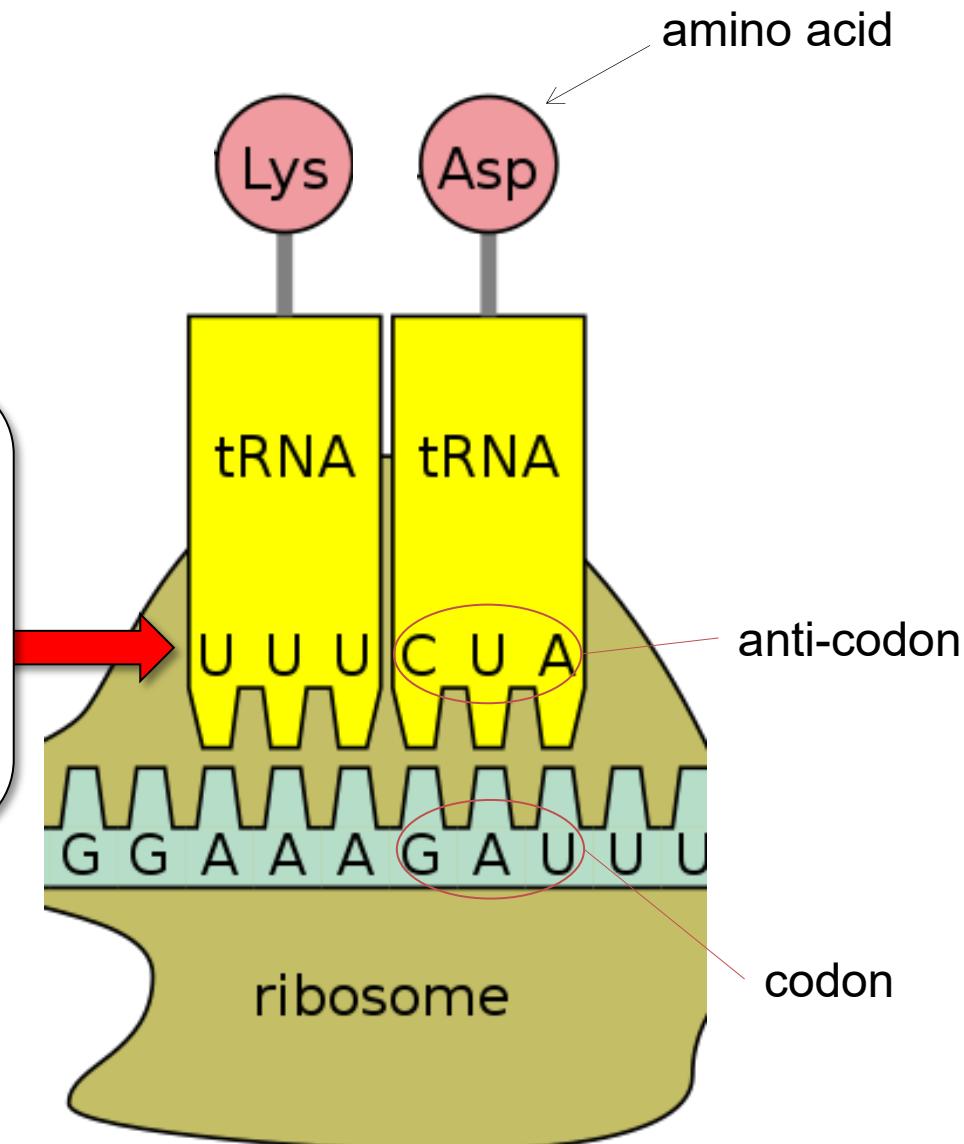
■ Key players in translation

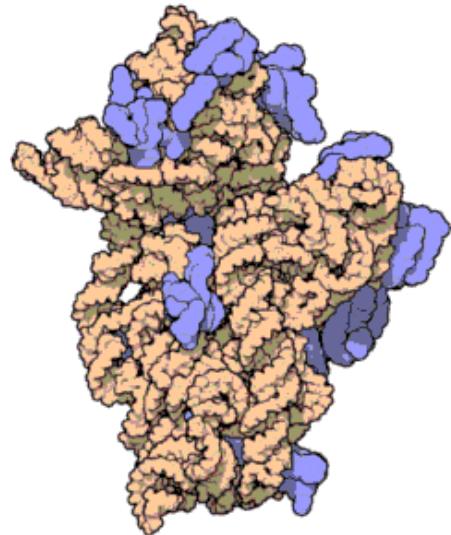
- **mRNA** (messenger RNA): brings instructions (in codons) for the amino acid sequence
- **rRNA** (ribosomal RNA): works with proteins to make **ribosomes** (on rough ER) where proteins are made
- **tRNA** (transfer RNA): helps match up each codon to the amino acid it codes for
 - can do this because it has an **anti-codon** (to bind to a codon) & each is attached to a specific amino acid



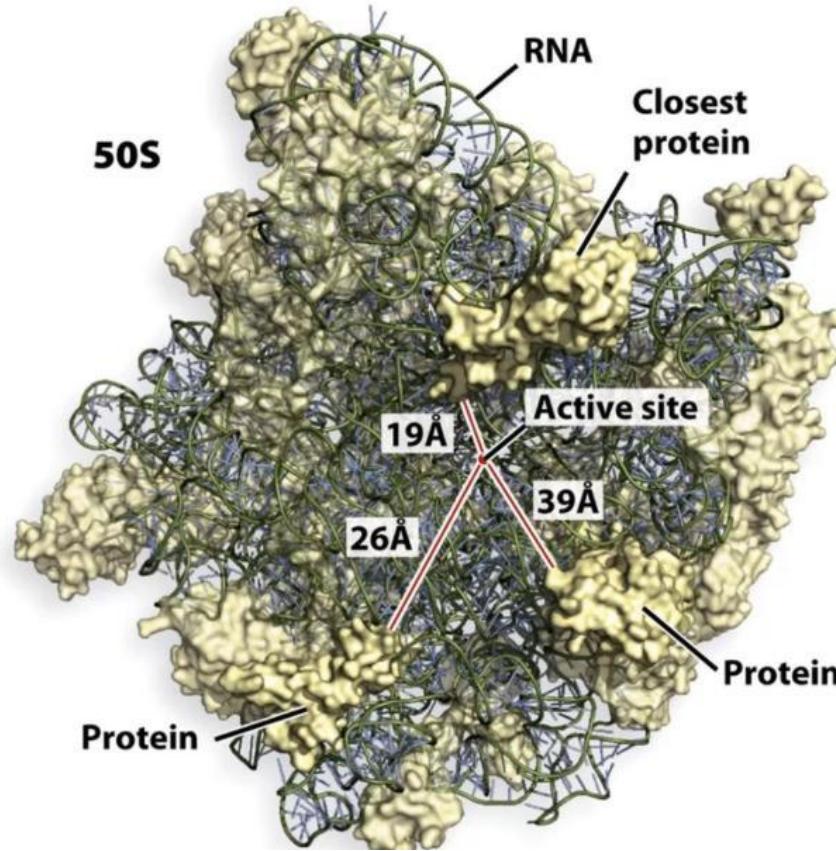
Transfer RNA (tRNA) molecules translate the mRNA code by linking specific bases on the mRNA with specific amino acids that will be used to build a protein.

The anti-codon is a 3-base sequence that matches up with the 3-base sequence on the mRNA strand. Each 3-base sequence in mRNA—called a codon—matches with a tRNA that carries one particular amino acid.





THE RIBOSOME'S CATALYTIC SITE IS FAR FROM RIBOSOMAL PROTEINS



Ribosomal Catalytic Site

The active site—where the peptidyl transferase forms peptide bonds—is 18 Å away from the closest ribosomal protein (very far!). RNA is the only molecule in the active site, evidence that the ribosome is a ribozyme.

[Source: PDB ID 1Q7Y.]

What lines of evidence I've shown you suggest that RNA is the critical molecule for ribosomal function?

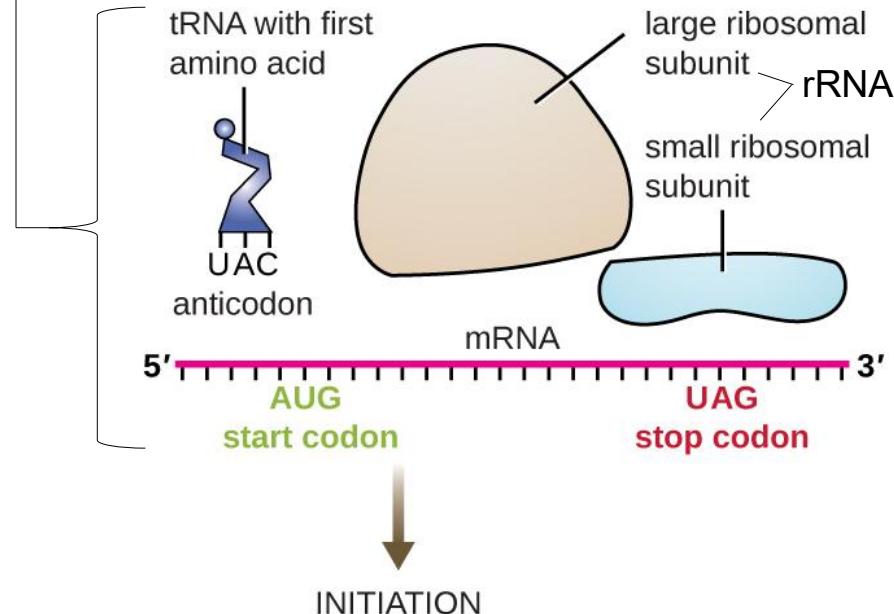


Translation has 3 stages

- 1. **Initiation:**
mRNA, tRNA, &
rRNA/ribosomes all
come together starting
at a **start codon**

- *the tRNA with the anticodon to the start codon is the first to bind inside the ribosome*

Starting components



Transitional complex forms, and tRNA brings first amino acid in polypeptide chain to bind to start codon on mRNA.

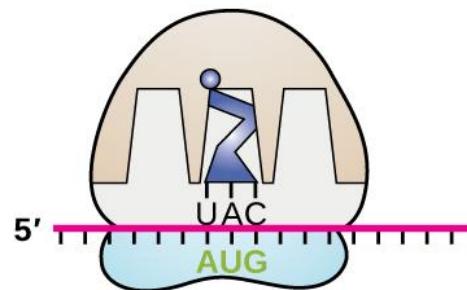
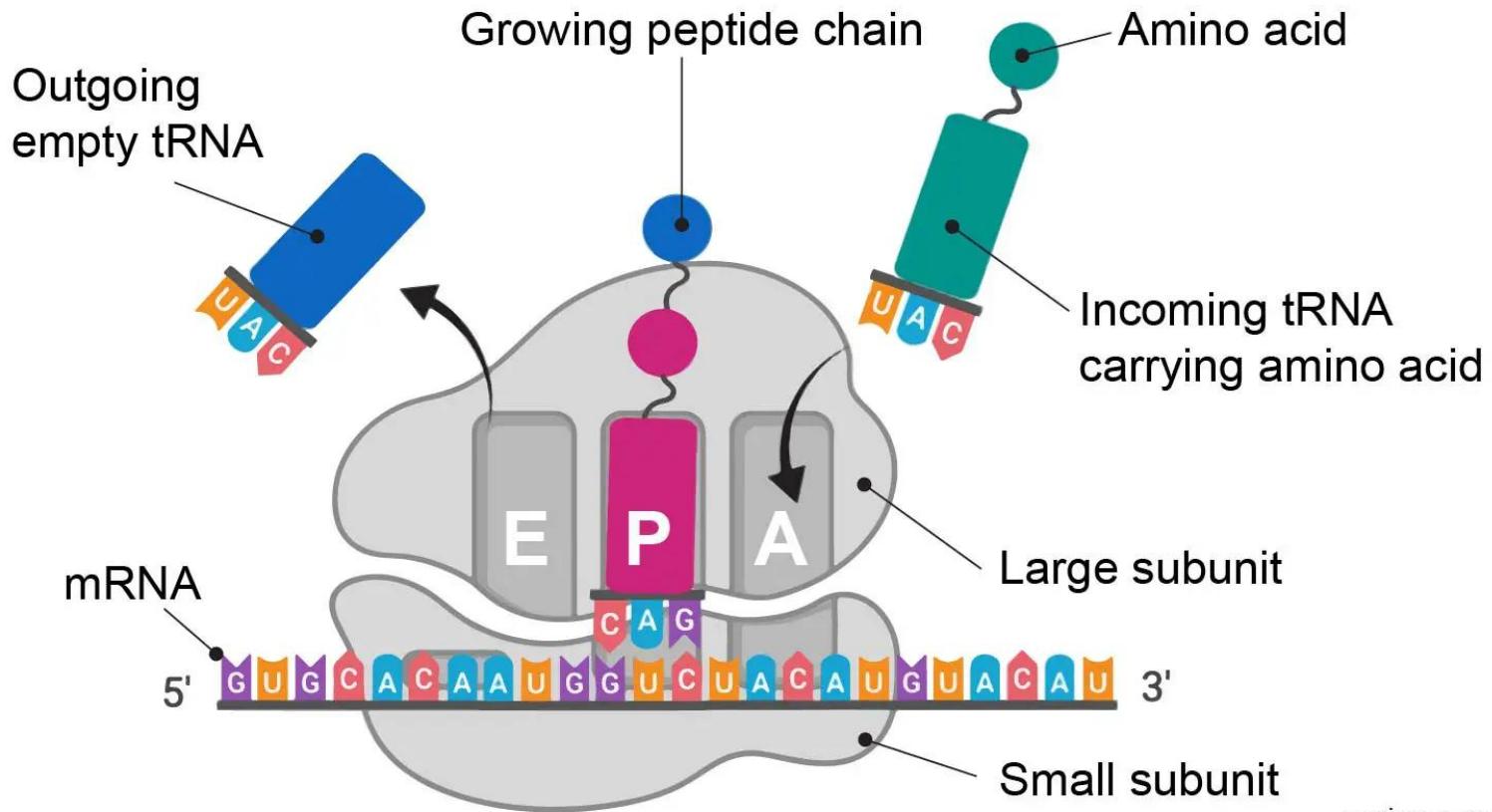


Figure: https://commons.wikimedia.org/wiki/File:OSC_Microbio_11_04_TInInit.jpg



rsscience.com

A: aminoacyl site

P: peptidyl site

E: exit site



Translation has 3 stages

- **2. Elongation:** tRNAs move in & out of the ribosome, leaving their amino acid behind attached to the last amino acid
 - *each time a tRNA leaves, the mRNA shifts over, making space for a new tRNA to bind*

INITIATION

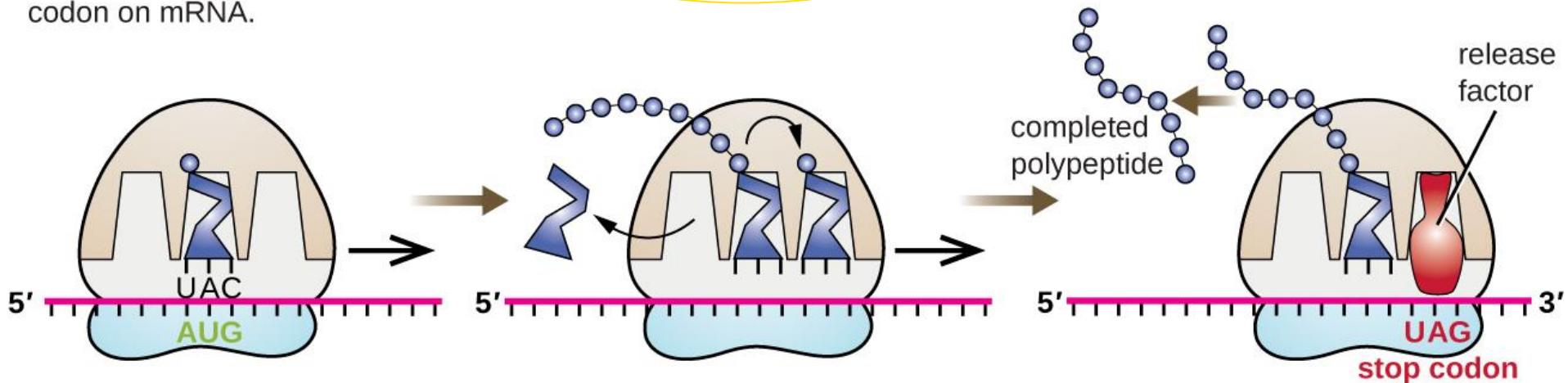
Transitional complex forms, and tRNA brings first amino acid in polypeptide chain to bind to start codon on mRNA.

ELONGATION

tRNAs bring amino acids one by one to add to polypeptide chain.

TERMINATION

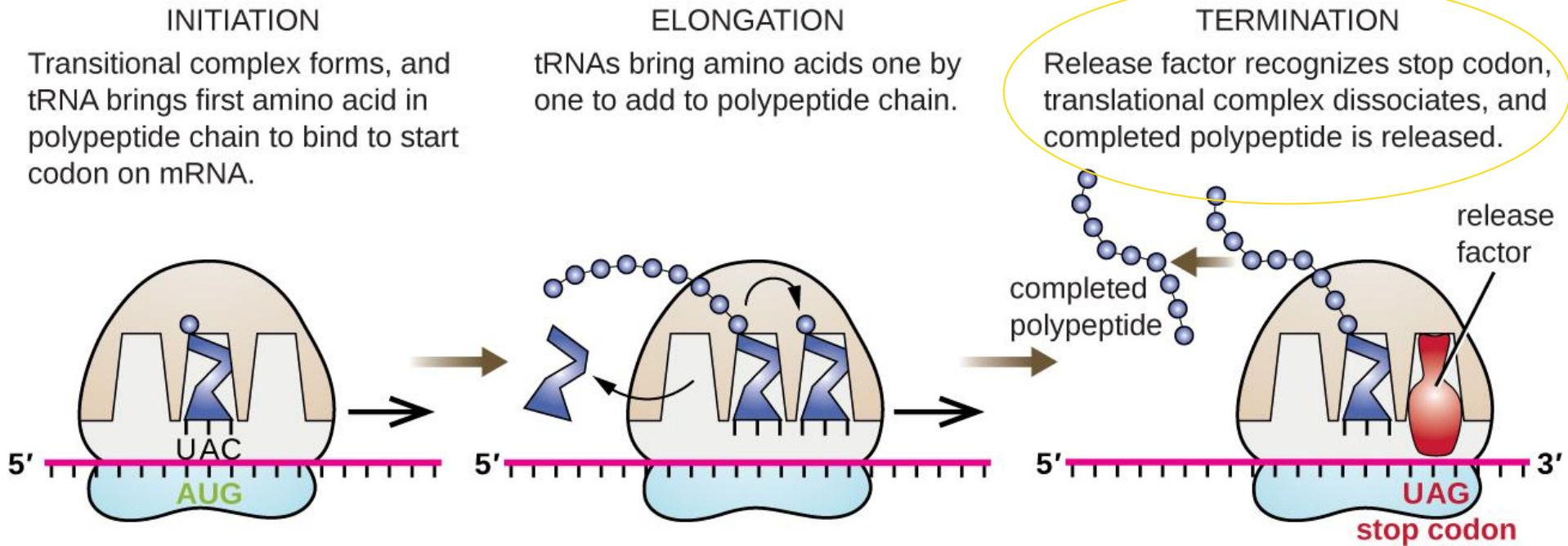
Release factor recognizes stop codon, translational complex dissociates, and completed polypeptide is released.





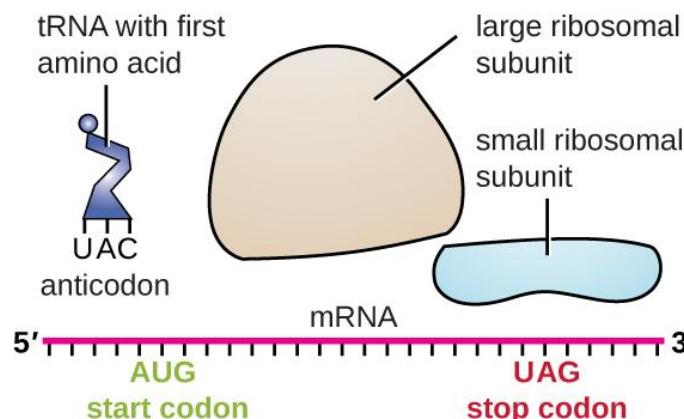
Translation has 3 stages

- **3. Termination:** when a **stop codon** is reached, all components separate & the amino acid chain will move through the rough ER & golgi apparatus to be folded & functional





The mRNA strand can be translated over & over again before it is degraded – can produce dozens or even hundreds more proteins



INITIATION

Transitional complex forms, and tRNA brings first amino acid in polypeptide chain to bind to start codon on mRNA.

ELONGATION

tRNAs bring amino acids one by one to add to polypeptide chain.

TERMINATION

Release factor recognizes stop codon, translational complex dissociates, and completed polypeptide is released.

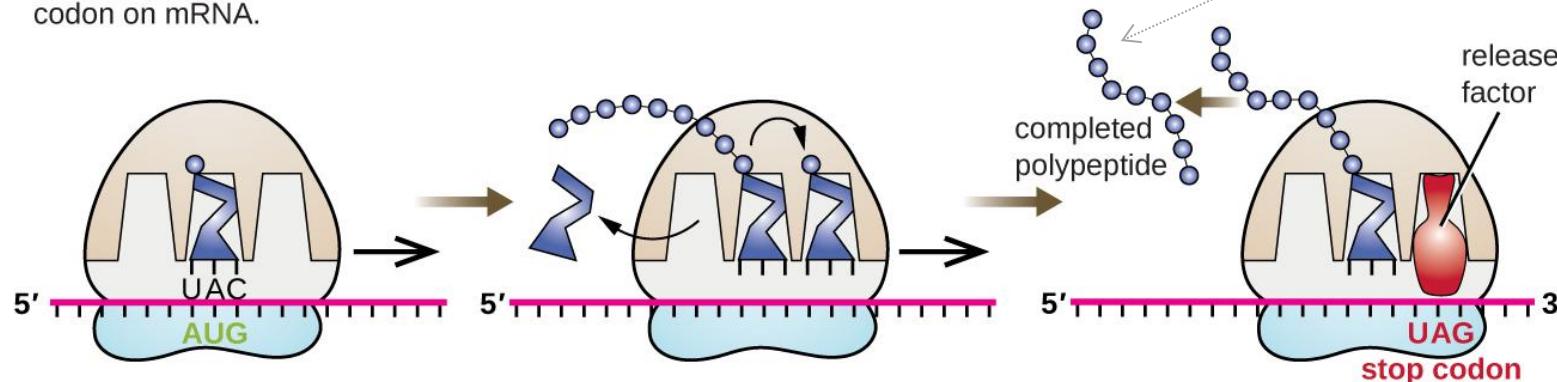
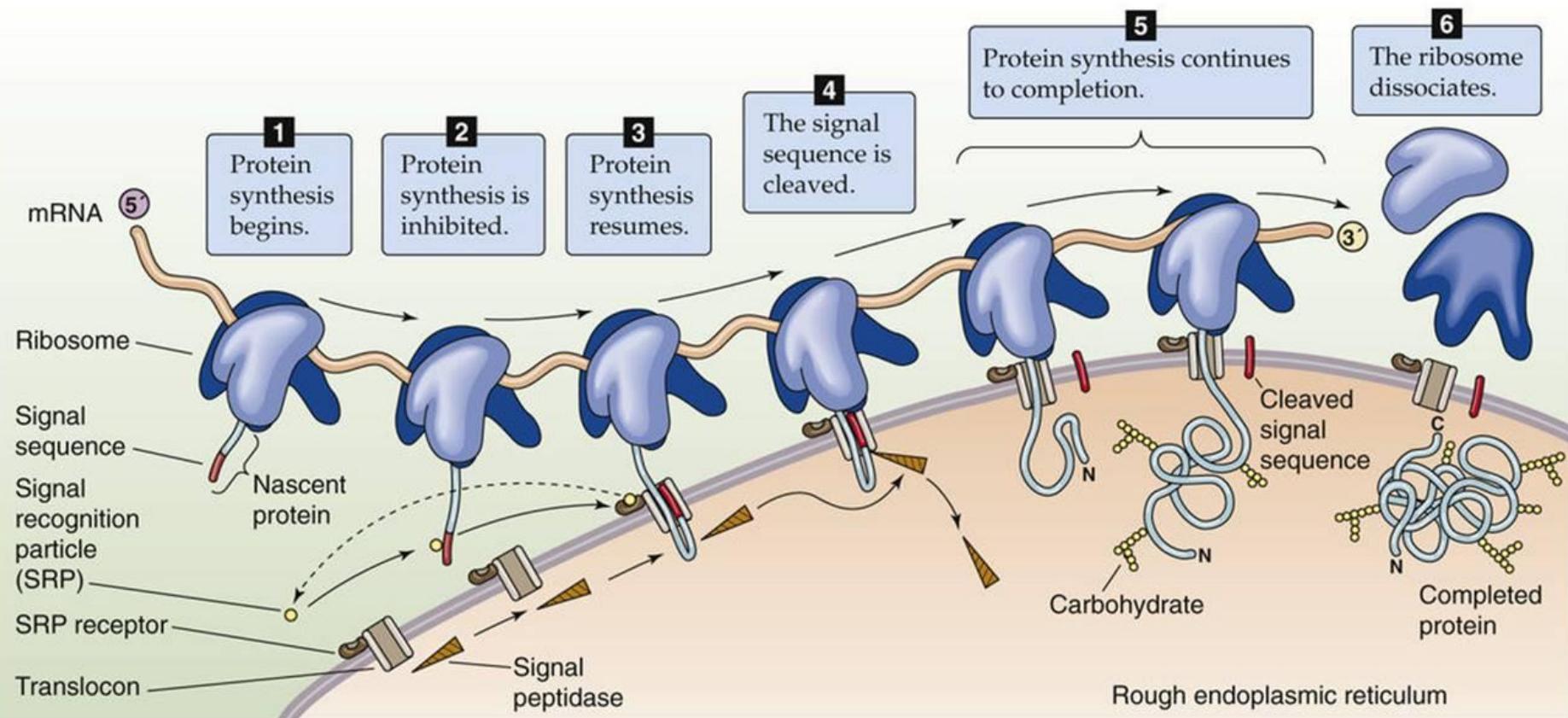


Figure: https://commons.wikimedia.org/wiki/File:OSC_Microbio_11_04_TlInit.jpg

The newly formed amino acid sequence will become folded into a functional protein



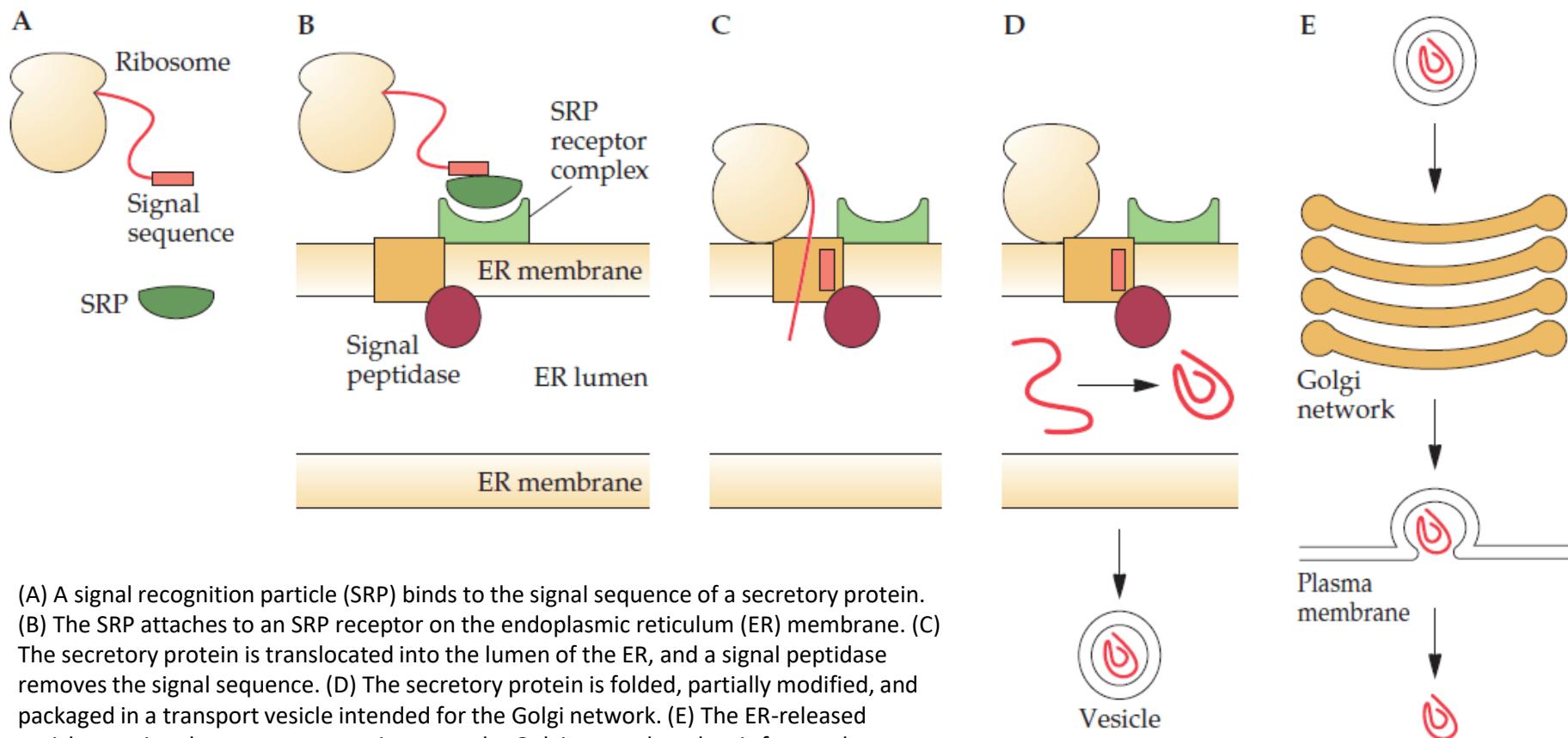
Protein Secretion Pathways



<https://doctorlib.info/physiology/medical/medical.files/image045.jpg>



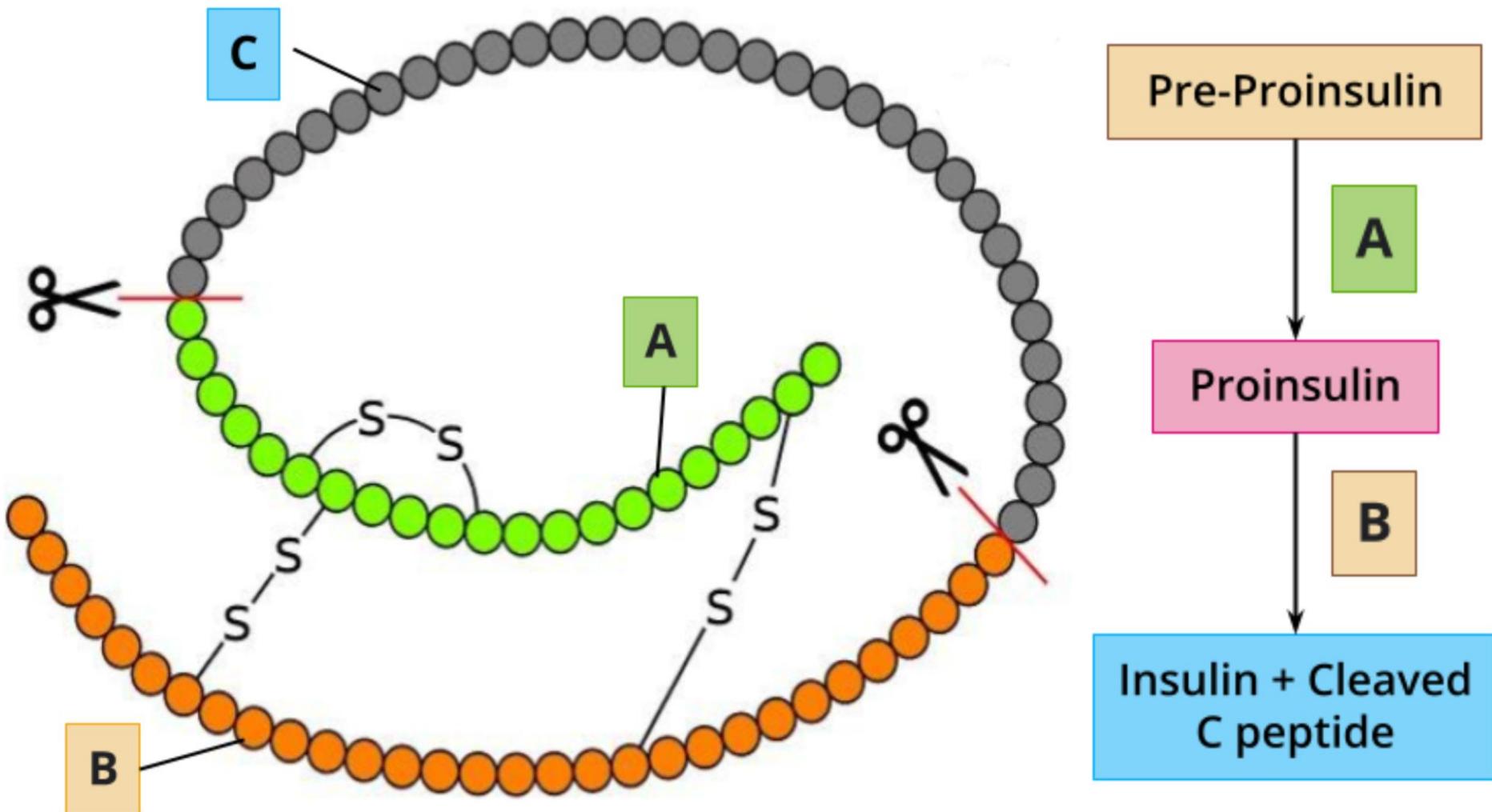
Protein Secretion Pathways



(A) A signal recognition particle (SRP) binds to the signal sequence of a secretory protein. (B) The SRP attaches to an SRP receptor on the endoplasmic reticulum (ER) membrane. (C) The secretory protein is translocated into the lumen of the ER, and a signal peptidase removes the signal sequence. (D) The secretory protein is folded, partially modified, and packaged in a transport vesicle intended for the Golgi network. (E) The ER-released vesicle carrying the secretory protein enters the Golgi network at the cis face and passes through the Golgi stack, where it is further modified; after it is sorted, a plasma membrane-specific vesicle is formed at the trans face of the Golgi network. The secretory transport vesicle fuses with the plasma membrane and releases the secretory protein to the extracellular environment.



Protein processing



Biotechnology in medicine — lesson. Science State Board, Class 10.



How are genes regulated?

- We now see how our DNA codes for our traits, making us who we are
- BUT: not all of our genes are expressed at all times
 - Humans have around 25,000 pairs of genes (1 from each parent) spread across 23 pairs of chromosomes

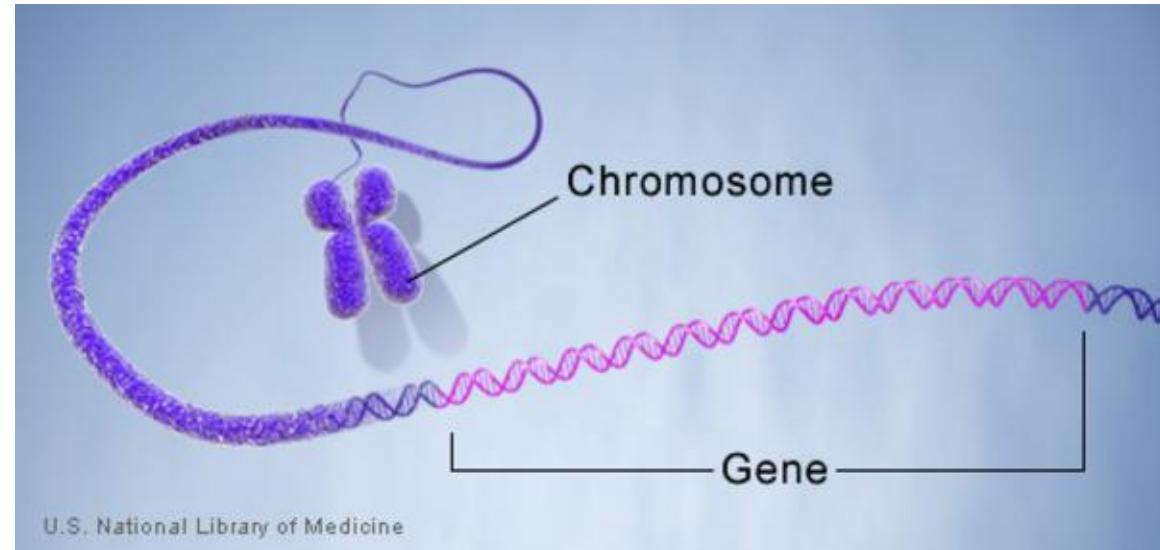


Figure: <https://medlineplus.gov/genetics/understanding/basics/gene/>



Gene expression:

- Some genes are only expressed at certain times
 - e.g. every human has 2 genes that code for milk proteins during lactation, but only some people produce these proteins at only specific times
- Some are never used by a cell
 - e.g. skin cells never need hemoglobin (for blood)
- Some are always expressed in every cell
 - e.g. cellular respiration / mitochondria enzymes
 - (all cells need ATP)

- Cells can regulate gene expression at 3 levels

- **Level 1: Transcription**

- cells control which genes are actually transcribed from DNA → RNA

- no transcription = no expression at all (e.g. milk proteins in cells that aren't lactating)

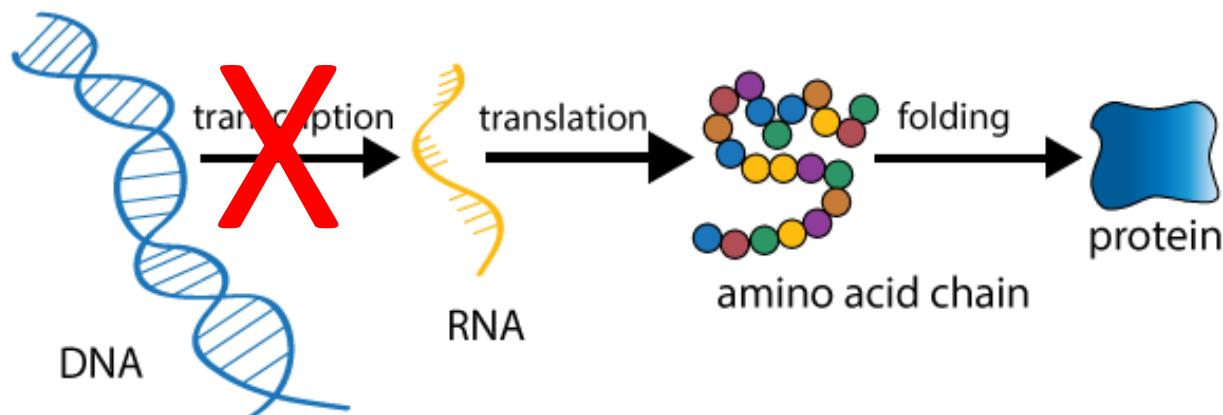


Figure: <https://biosocialmethods.isr.umich.edu/research-support/videos-tutorials/epigenetics-tutorial/>



■ RNA splicing

- Exons aren't always spliced back together exactly
- This means the RNA splicing process can allow for 1 gene to make multiple different versions of a protein – very useful!

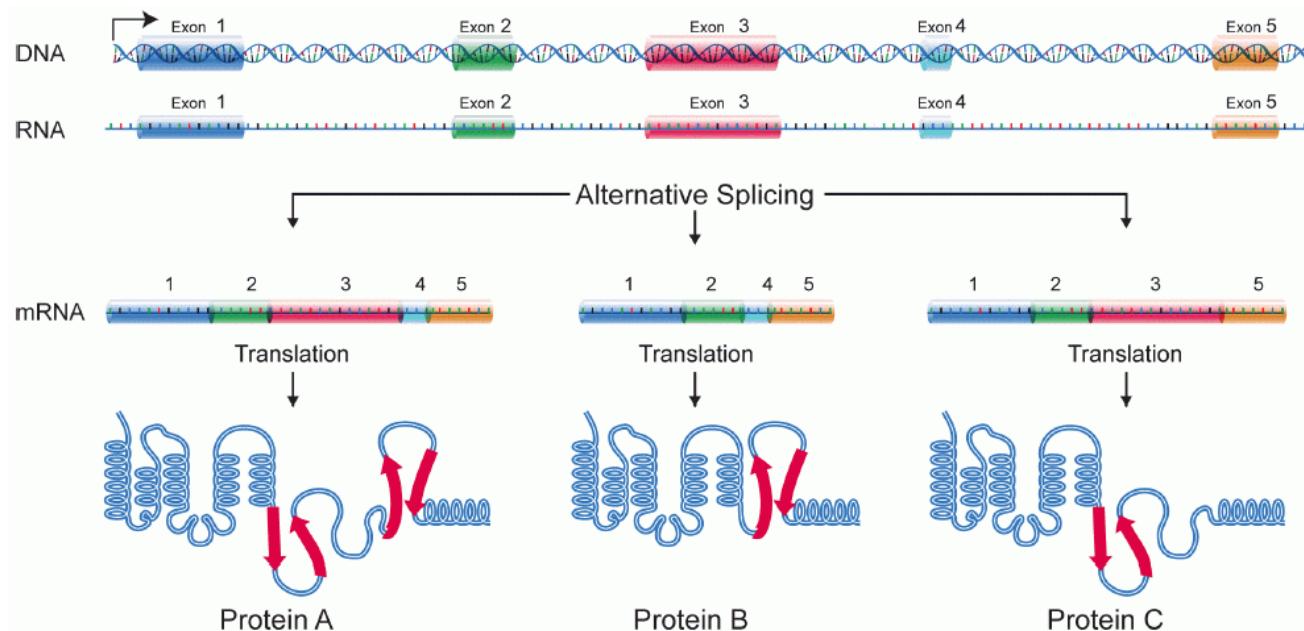


Figure: <https://www.khanacademy.org/science/biology/gene-expression-central-dogma/transcription-of-dna-into-rna/a/eukaryotic-pre-mrna-processing>

- Cells can regulate gene expression at 3 levels

- **Level 2: Translation**

- cells control how many proteins are made from mRNA after transcription

- does 1 mRNA strand make 1 protein
& stop, or should it make 100 proteins?

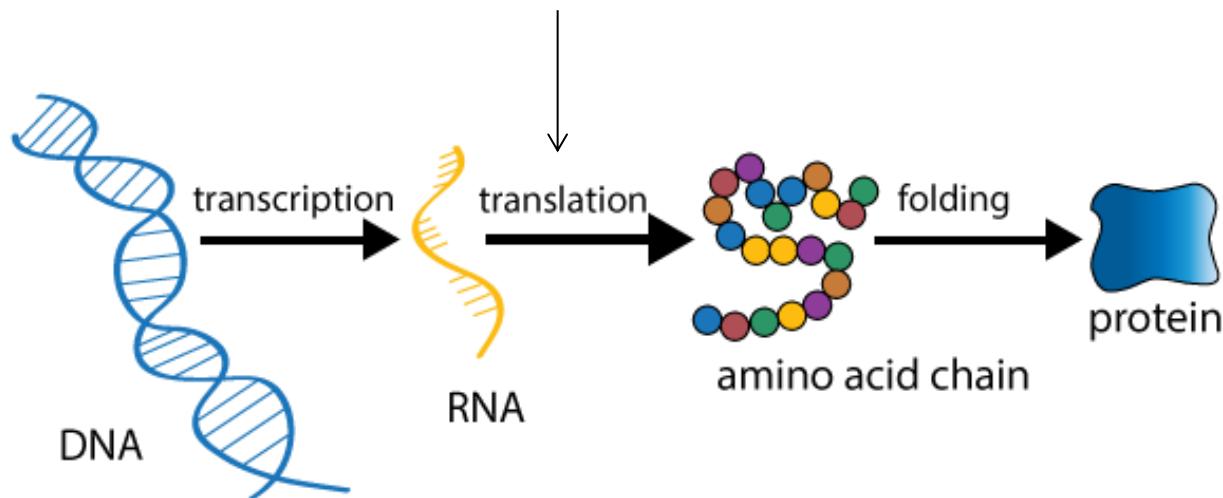


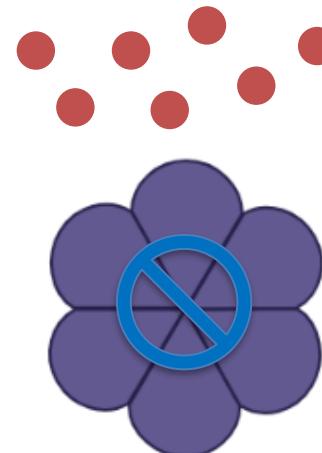
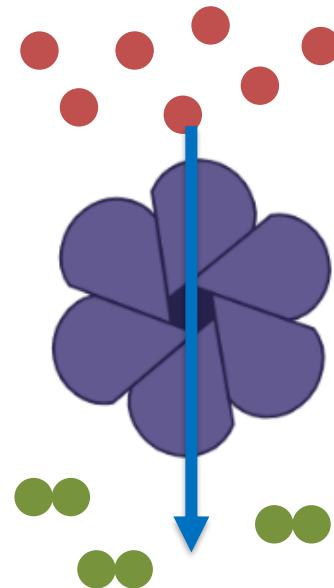
Figure: <https://biosocialmethods.isr.umich.edu/research-support/videos-tutorials/epigenetics-tutorial/>

- Cells can regulate gene expression at 3 levels

- **Level 3: Protein activity**

- *Remember:* cells can control metabolism by activating or inactivating proteins that have already been made

A regularly functioning protein



A protein that has been inactivated



How do mutations affect genes?

- **Mutations** = changes in the sequence of DNA bases
 - Can occur naturally, but are usually caused by carcinogens & mutagens
 - **Mutagen** = a substance that causes mutations
 - **Carcinogen** = a substance capable of causing cancer, usually by mutating DNA (*more on cancer in next chapter*)

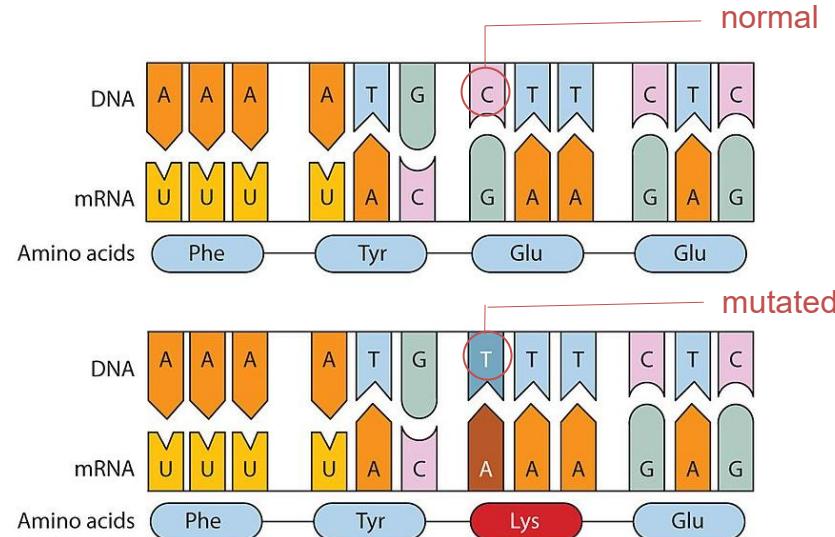


Figure: <https://commons.wikimedia.org/wiki/File:Block-mutations.jpg>



- We come into contact with many carcinogens & mutagens in our environment all the time
- Many types of mutations can occur
 - Sometimes only one base changes
 - e.g. substitution
 - e.g. insertion
 - e.g. deletion
 - Sometimes large pieces of DNA are moved
 - e.g. inversion
 - e.g. translocation

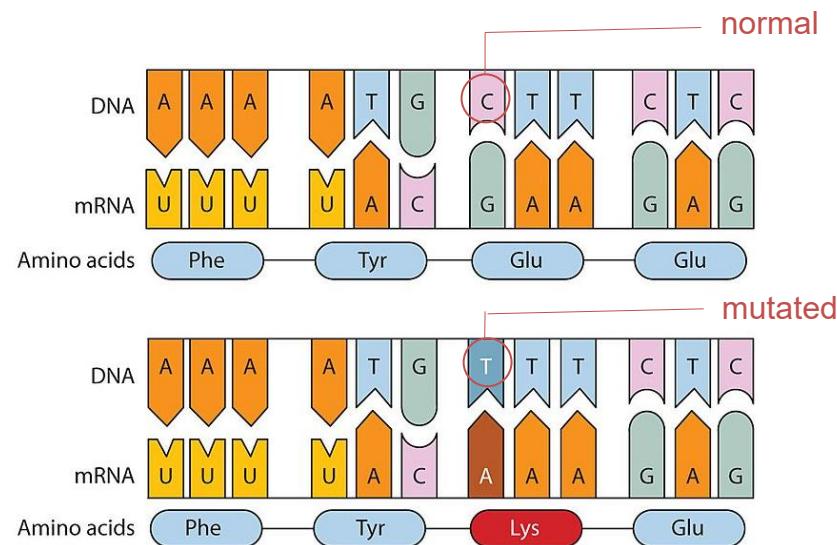


Figure: <https://commons.wikimedia.org/wiki/File:Block-mutations.jpg>



■ Substitution mutations:

- one base is swapped out for a different base
 - e.g. an A is swapped (substituted) for a T

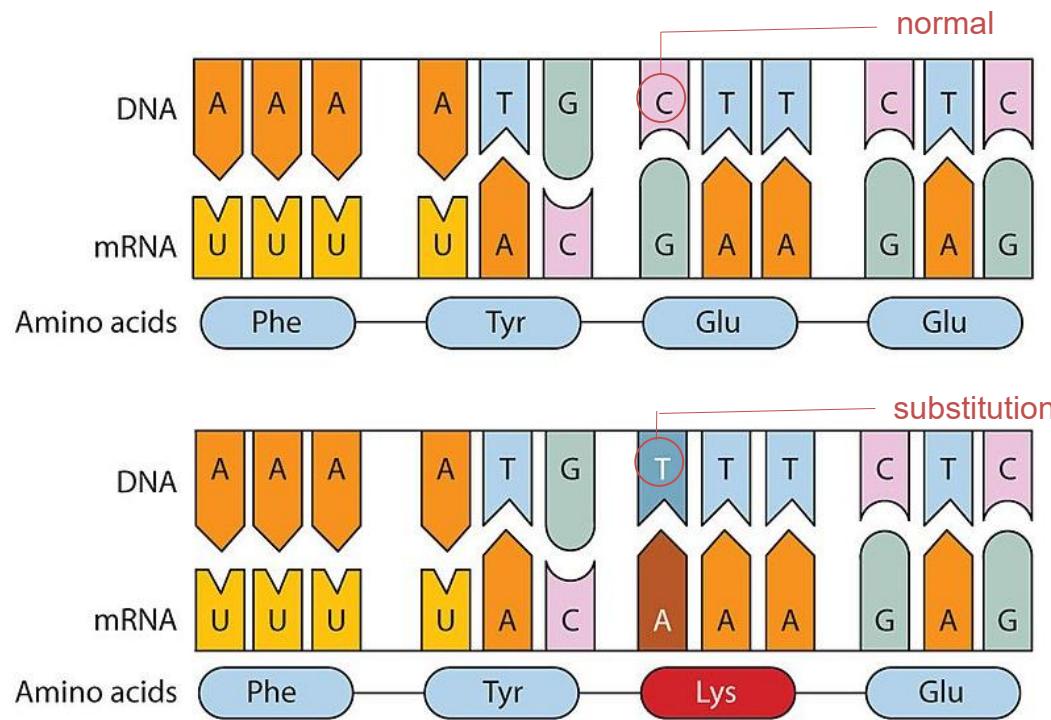


Figure: <https://commons.wikimedia.org/wiki/File:Block-mutations.jpg>



■ Insertion & deletion mutations:

– 1 or more nucleotides are inserted or deleted into the DNA sequence

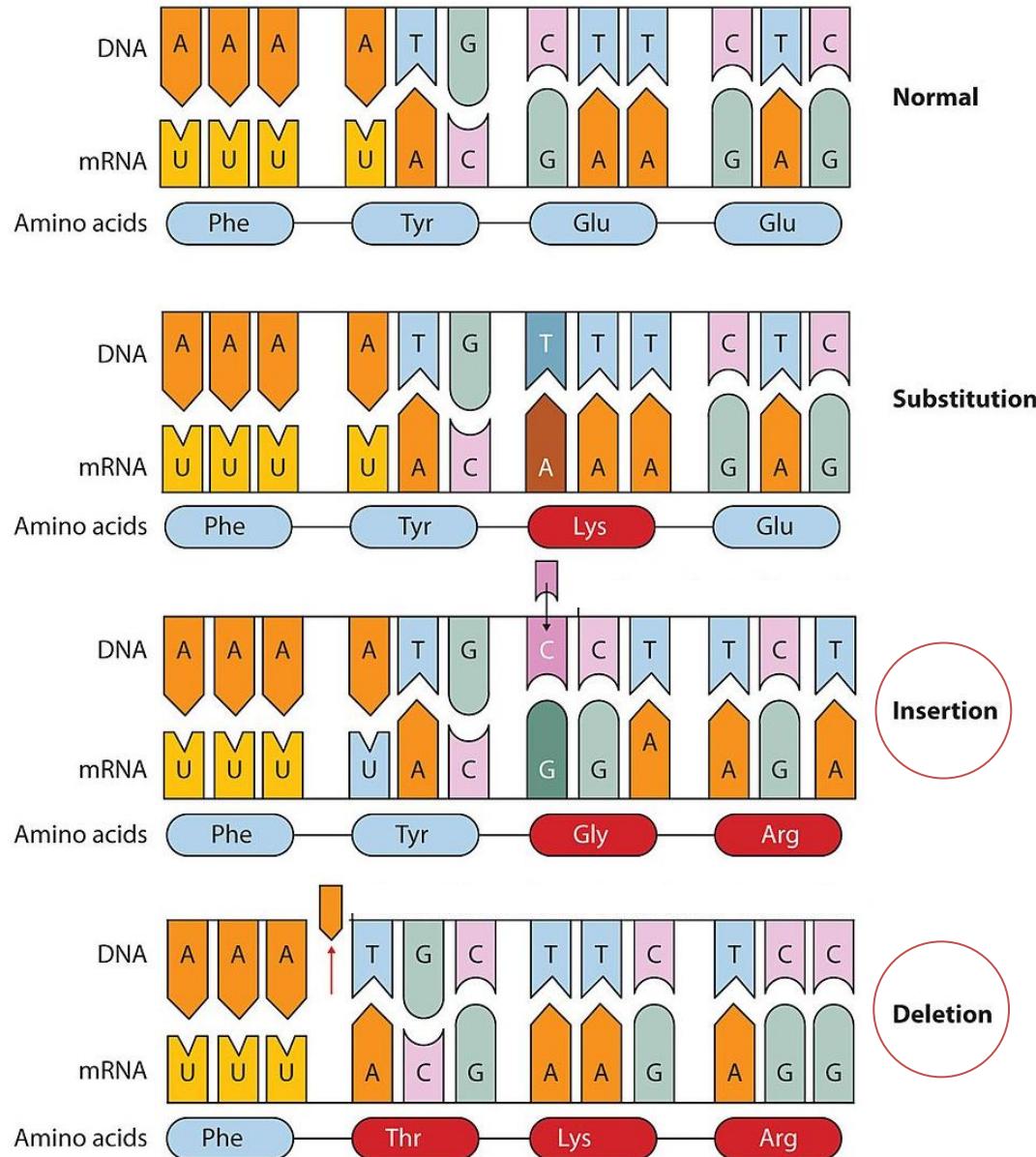


Figure: <https://commons.wikimedia.org/wiki/File:Block-mutations.jpg>



■ Inversion mutations:

- a piece of DNA is accidentally cut out of a chromosome, gets turned around, & is re-inserted into the gap upside down

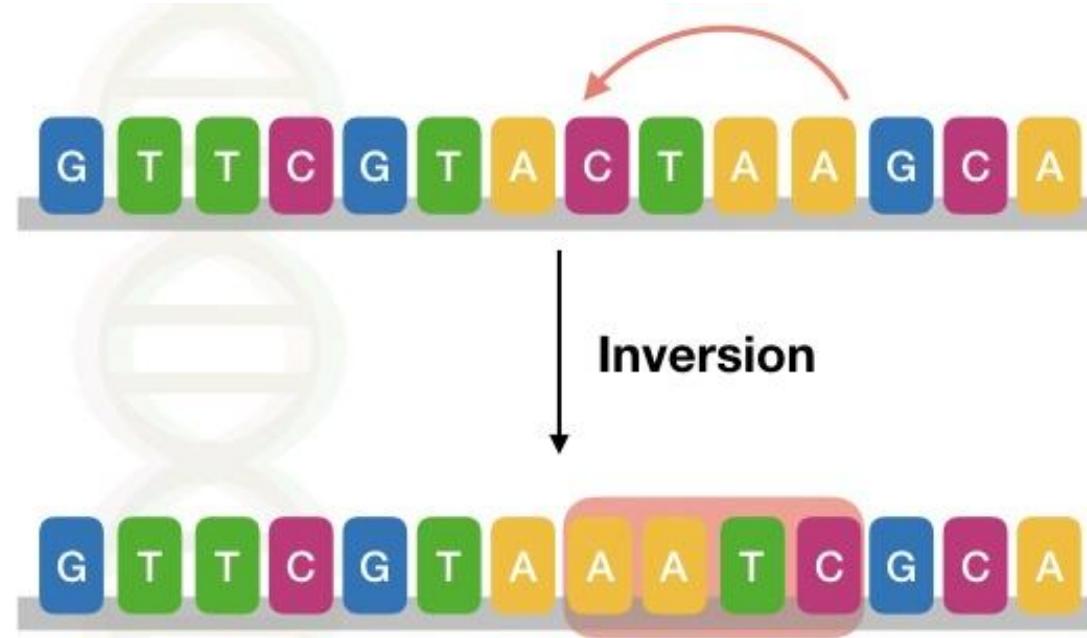


Figure: <https://geneticeducation.co.in/genetic-mutations-definition-types-causes-and-examples/>

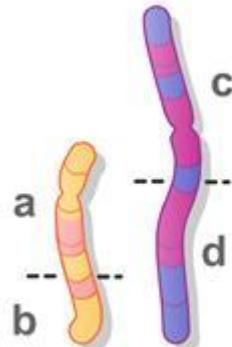


■ Translocation mutations:

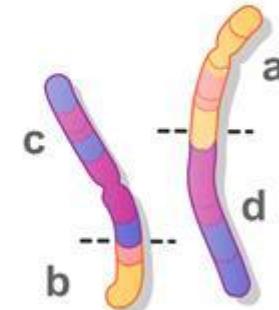
– a piece of DNA (often very large) is accidentally removed from 1 chromosome & attached to another chromosome

- only time more than one chromosome is involved

Normal chromosomes

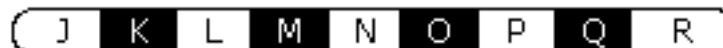


Chromosomes with translocation



© AboutKidsHealth.ca

2 different healthy chromosomes



2 different chromosomes after translocation



Figure: <https://www.aboutkidshealth.ca/Article?contentid=2828&language=English>



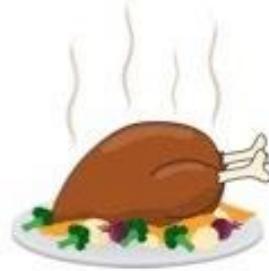
A silly summary of the different types of mutations:

Normal



BEAST

Substitution



FEAST

Insertion



BREAST

Deletion



BEST

A

Inversion



BEATS



■ What are the effects of mutations?

– **Inversion & translocation** mutations involve large pieces of DNA

- These may be neutral if the entire gene is moved from one place to another – very rare!
- Most common: a gene is split in 2
 - when transcription occurs, the message is cut short & no functional protein can be made: harmful

Normal vs. Mutation	Hypothetical visualization of effects
Normal DNA codons	THE ONE BIG FLY HAD ONE RED EYE
Translocation	THE ONE BIG F



■ What are the effects of mutations?

- One **insertion** or **deletion** mutation causes all following nucleotides to shift over – this changes every codon after the mutation = **frameshift**
 - changing the codons means changing the amino acids coded for during translation, & almost always = no functional protein: harmful

Normal vs. Mutation	Hypothetical visualization of effects
Normal DNA codons	THE ONE BIG FLY HAD ONE RED EYE
Insertion (of Q)	THE ONE B QI GFL YHA DON ERE DEY E..
Deletion (of G)	THE ONE BI F LYH ADO NER EDE YE.



■ What are the effects of mutations?

– Substitution

- *Remember:* the genetic code is **redundant**
– e.g. UUA & UUG both code for leucine
- the codon is changed but may still code for the same amino acid: the mutation can be neutral

Normal vs. Mutation	Hypothetical visualization of effects
Normal DNA codons	THE ONE BIG FLY HAD ONE RED EYE
Substitution (S for G)	THE ONE BIQ FLY HAD ONE RED EYE

If BIG & BIQ both code for the same amino acid, the final protein is unchanged & perfectly functional



■ What are the effects of mutations?

– Substitution

- If the mutation causes a different amino acid to be coded for, the mutation can lead to a nonfunctional protein: harmful
- It may even code for a premature stop codon, which leads to an incomplete protein: harmful

Normal vs. Mutation	Hypothetical visualization of effects
Normal DNA codons	THE ONE BIG FLY HAD ONE RED EYE
Substitution (Q for G)	THE ONE BIQ FLY HAD ONE RED EYE

If BIG & BIQ code for different amino acids, the final protein is changed & may not be functional



Some mutations can be beneficial: e.g. 1 substitution on the hemoglobin gene codes for proteins that cause **sickle cell anemia (SCA)**

- Can be harmful
- However: sickle cell anemia provides resistance to malaria
 - for people that live in malaria-prone areas of the world, this mutation is beneficial

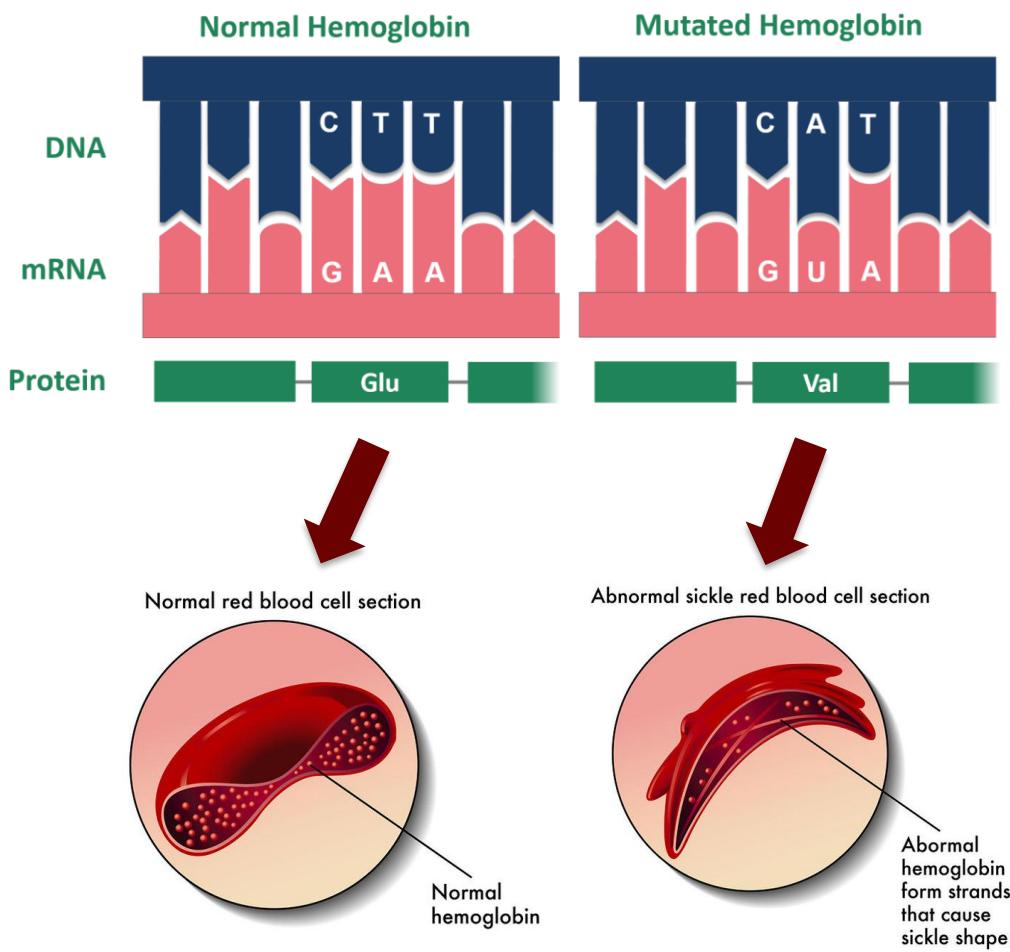


Figure: https://commons.wikimedia.org/wiki/File:Point-Mutation-Sickle-Cell-Normal_and_Mutated-Hemoglobin.png
Figure: [https://commons.wikimedia.org/wiki/File:Risk-Factors-for-Sickle-Cell-Anemia_\(1\)2.jpg](https://commons.wikimedia.org/wiki/File:Risk-Factors-for-Sickle-Cell-Anemia_(1)2.jpg)



Chapter 8: DNA Replication & Mitosis

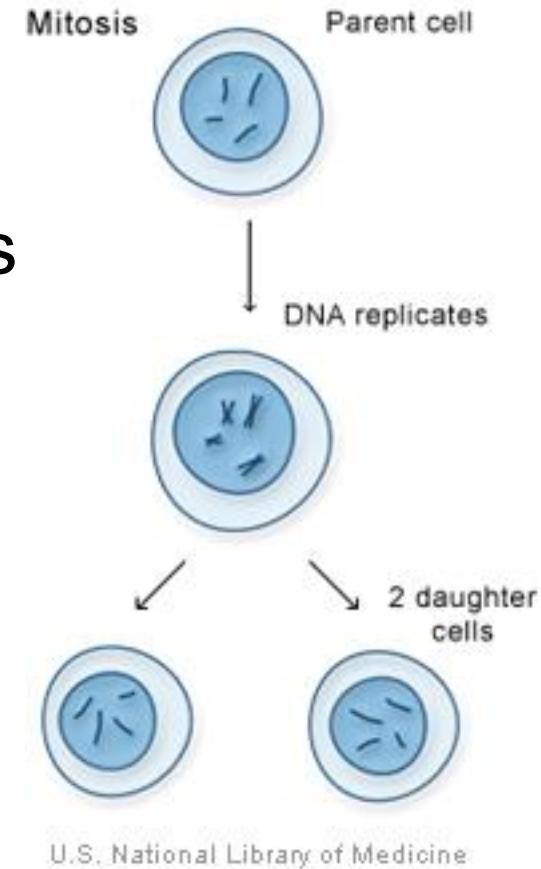
- Why do cells divide?
- Before cells divide, they must replicate their DNA
- Mitosis = cell division
- How is mitosis regulated?
- What happens when regulation goes wrong?

Corresponds with OpenStax Biology 2e Chapter 10



Why do cells divide?

- There are 2 types of cell division
- 1. **Mitosis** makes identical cell copies
 - process by which:
 - organisms grow
 - *Remember: to grow we don't get bigger cells, we get more cells*
 - old cells can be replaced
 - some organisms can asexually reproduce
 - some organisms can regenerate body parts



U.S. National Library of Medicine

Figure: <https://medlineplus.gov/genetics/understanding/howgeneswork/cellsdivide/>



2. Meiosis makes gametes (egg & sperm) which are not identical

- only for sexual reproduction
 - *will come back to meiosis next chapter*

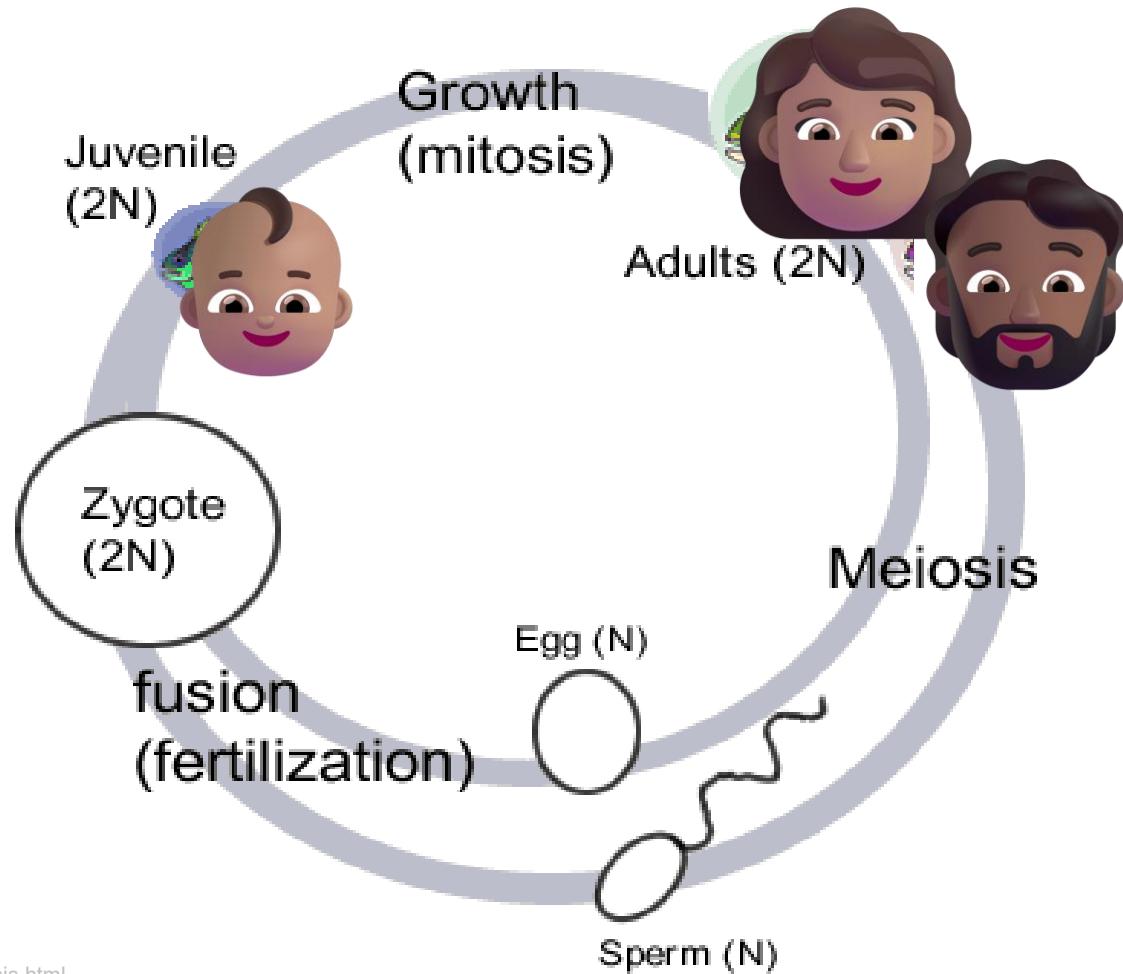


Figure: https://www.biologycorner.com/APbiology/inheritance/10-1_meiosis.html



Before cells divide, they must replicate their DNA

- *Remember:* all cells have DNA
 - In order to make a new cell, we must first make a copy of our DNA to put inside it
 - So before any type of cell division (mitosis or meiosis) cells must duplicate all of their DNA
- = DNA replication**

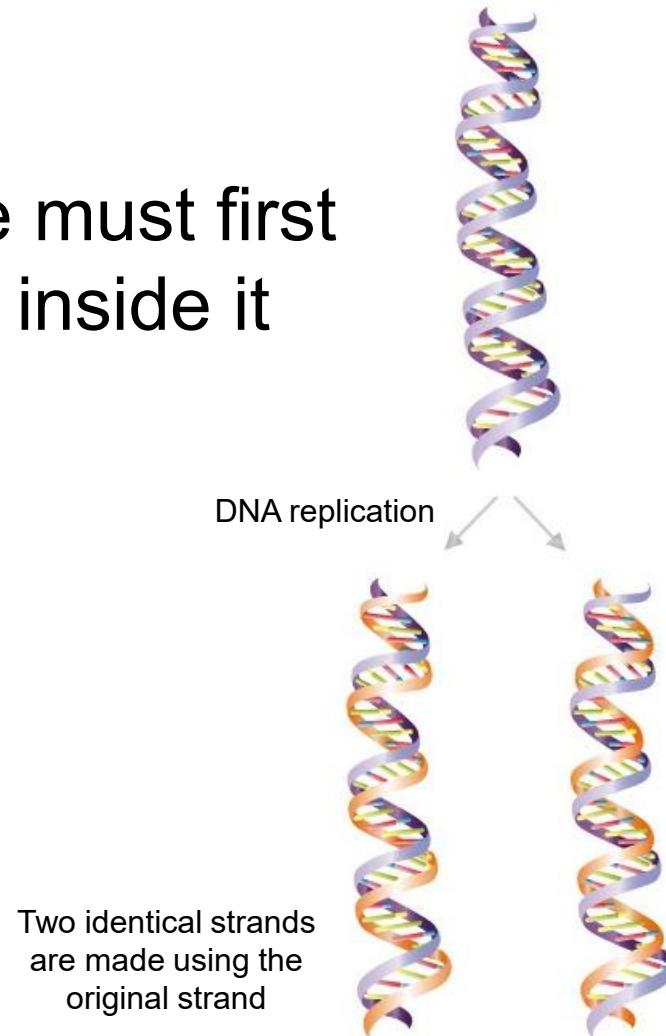


Figure: [https://commons.wikimedia.org/wiki/File:Semi_conservative_reproduction_of_DNA_\(13081032424\).jpg](https://commons.wikimedia.org/wiki/File:Semi_conservative_reproduction_of_DNA_(13081032424).jpg)



■ DNA replication requires 3 ingredients

- the original DNA double helix
- free nucleotides (A, T, G, C)
- several enzymes
 - e.g. DNA helicase & DNA polymerase

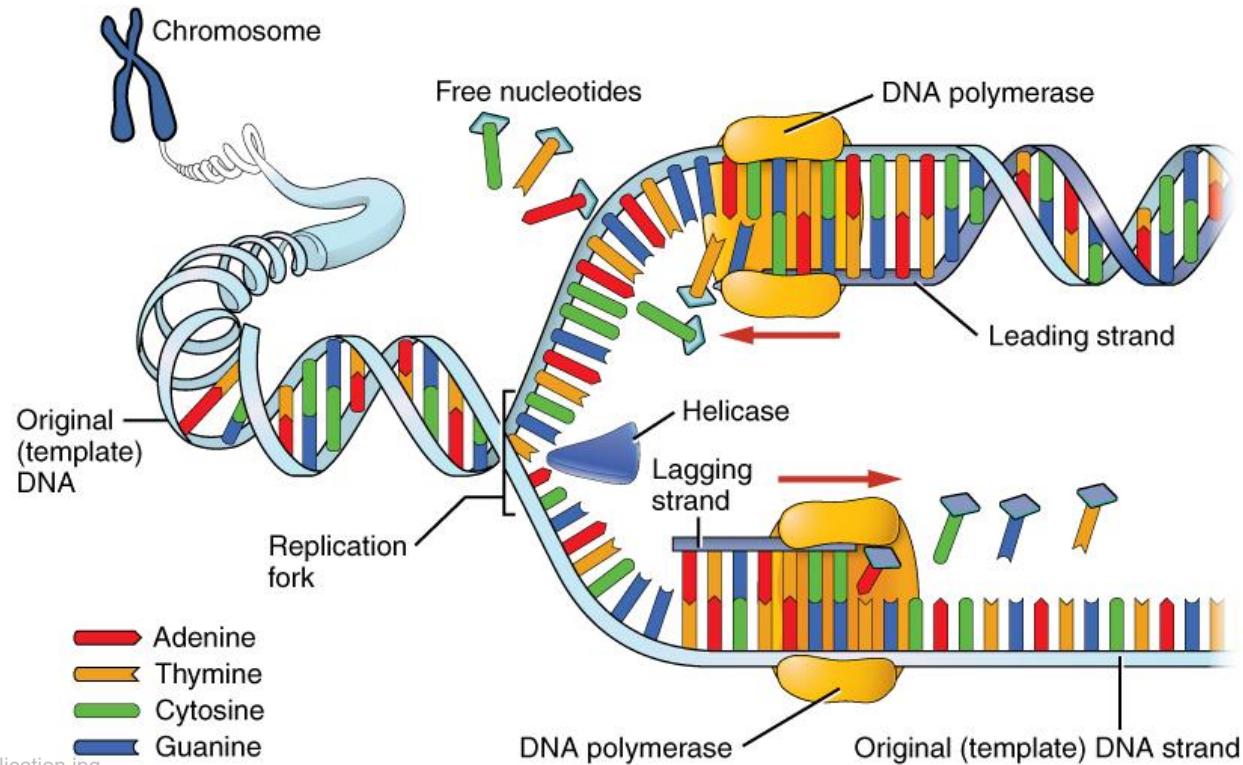


Figure: https://commons.wikimedia.org/wiki/File:0323_DNA_Replication.jpg



▪ DNA replication happens in 2 main steps

1. the original double helix is pulled apart (unzipped) by the enzyme **DNA helicase**

2. the enzyme **DNA polymerase** pairs free nucleotides (A, T, G, C) with each of the now single-stranded sides, building new double-stranded DNA

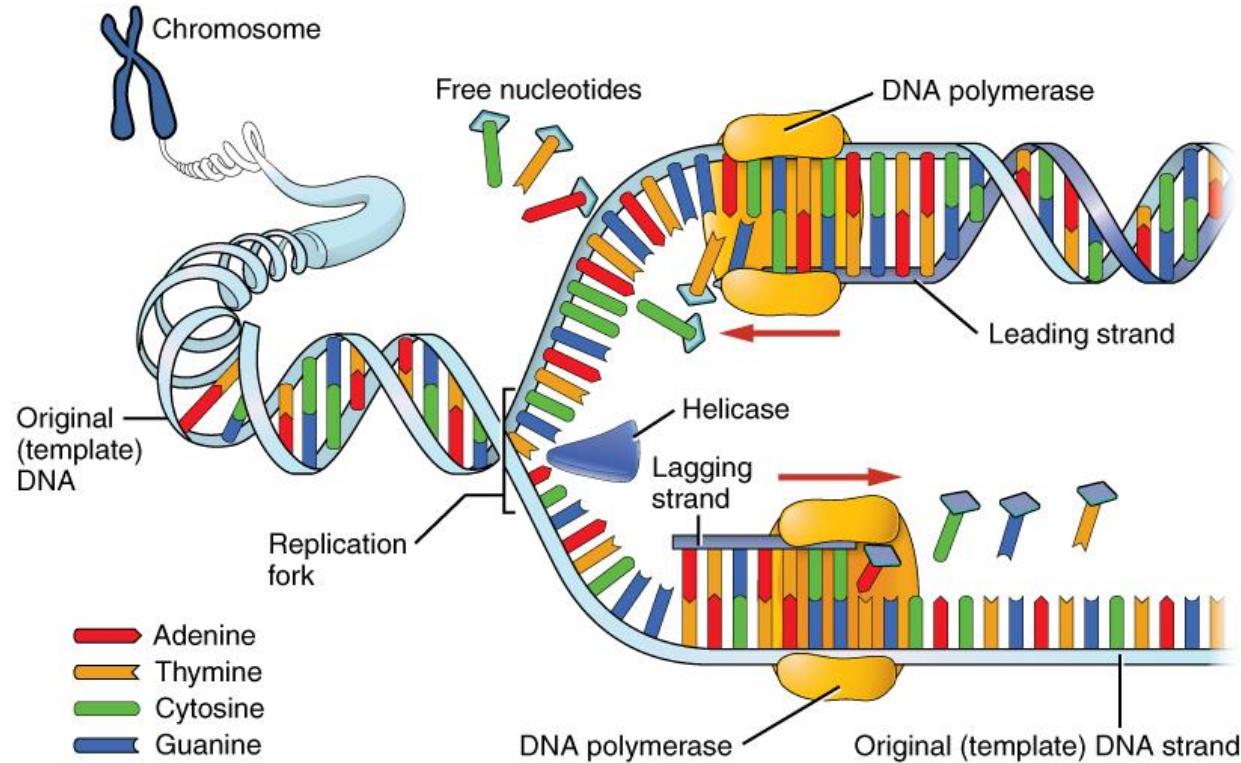
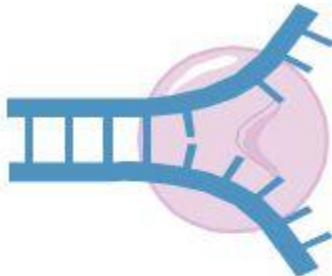
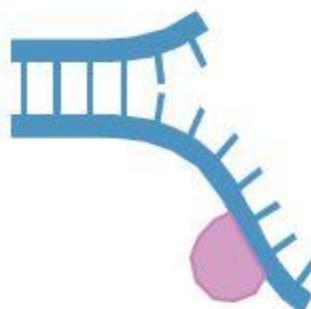


Figure: https://commons.wikimedia.org/wiki/File:0323_DNA_Replication.jpg

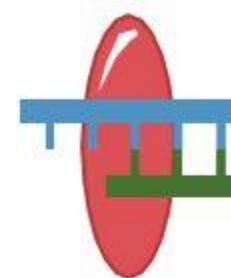
Enzymes in DNA replication



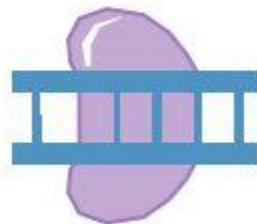
Helicase unwinds
parental double helix



Binding proteins
stabilise separate
strands



Primase adds
short primer
to template strand



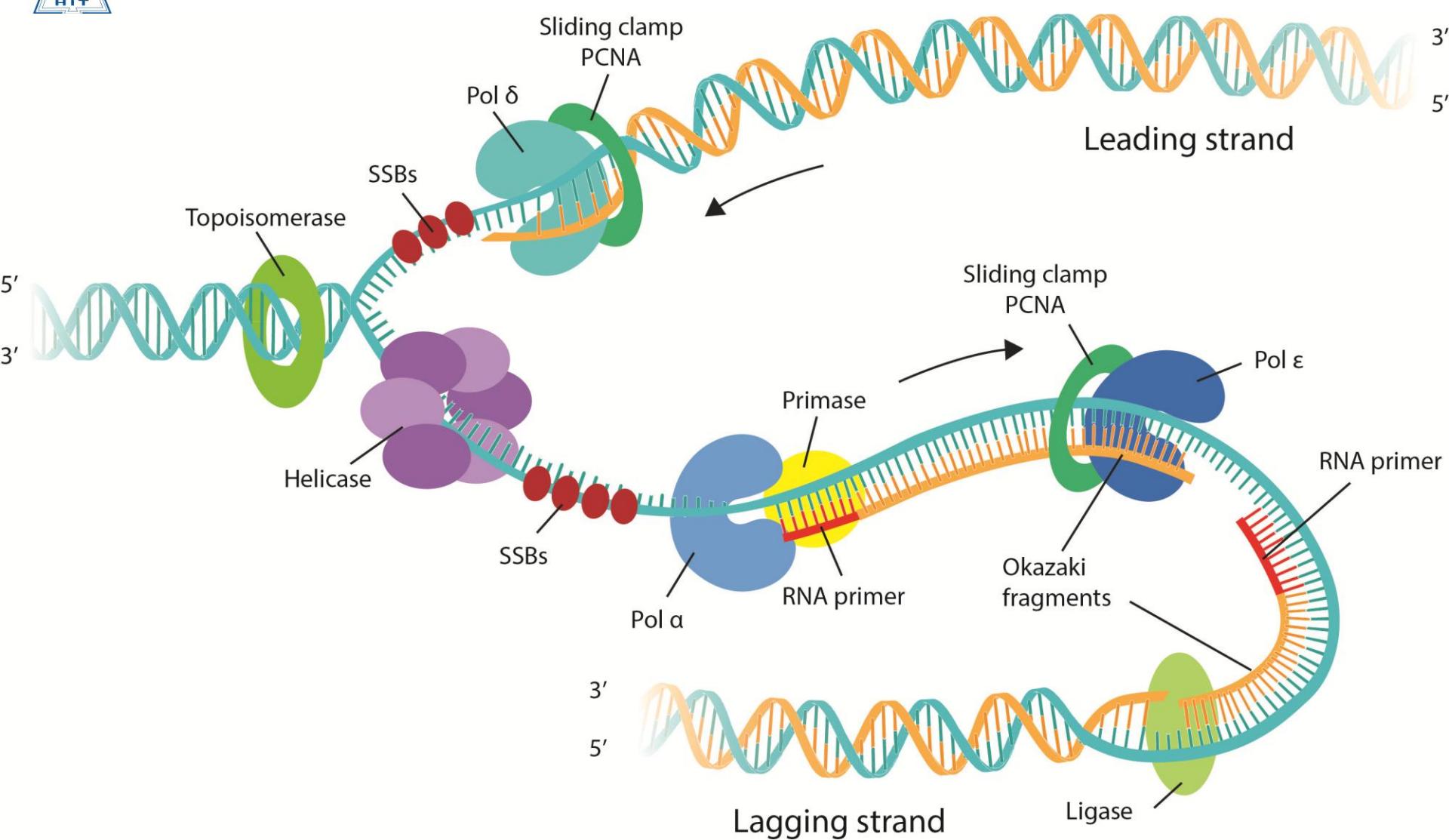
DNA polymerase
binds nucleotides
to form new strands



DNA polymerase I
(Exonuclease) removes
RNA primer and inserts
the correct bases



Ligase joins Okazaki
fragments and seals
other nicks in sugar-
phosphate backbone





One DNA double helix

■ DNA replication

- When replication is complete, each of the final DNA copies have one old original strand & one newly made strand
= **semiconservative replication**

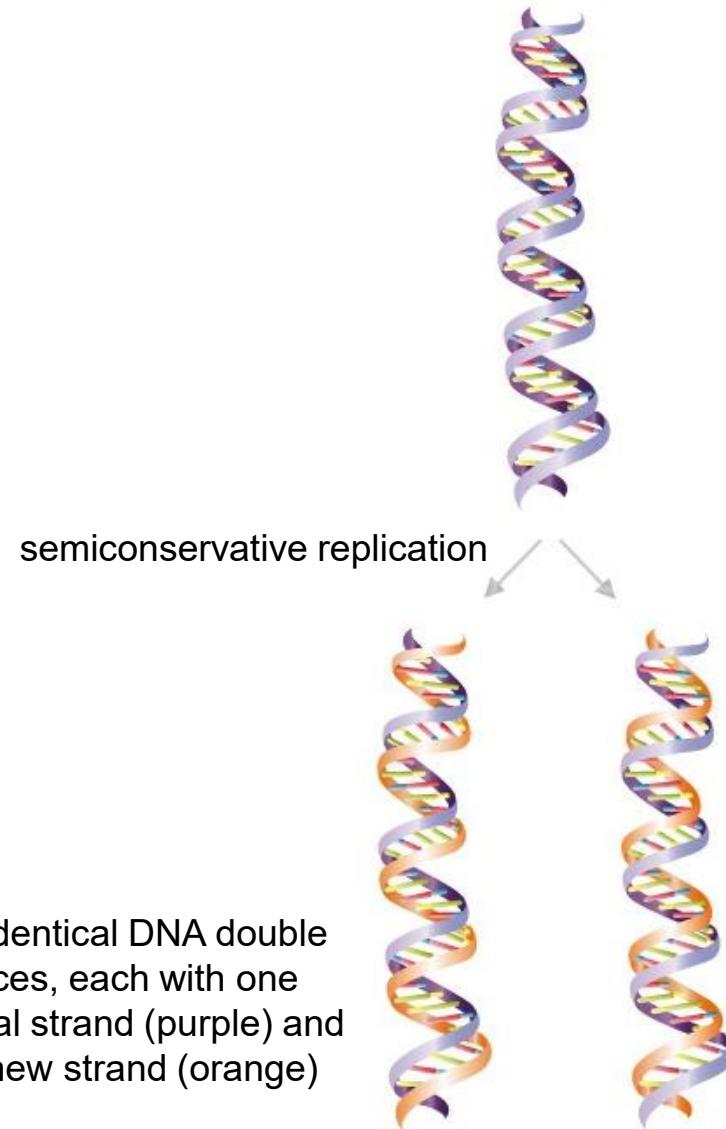


Figure: [https://commons.wikimedia.org/wiki/File:Semi_conservative_replication_of_DNA_\(13081032424\).jpg](https://commons.wikimedia.org/wiki/File:Semi_conservative_replication_of_DNA_(13081032424).jpg)

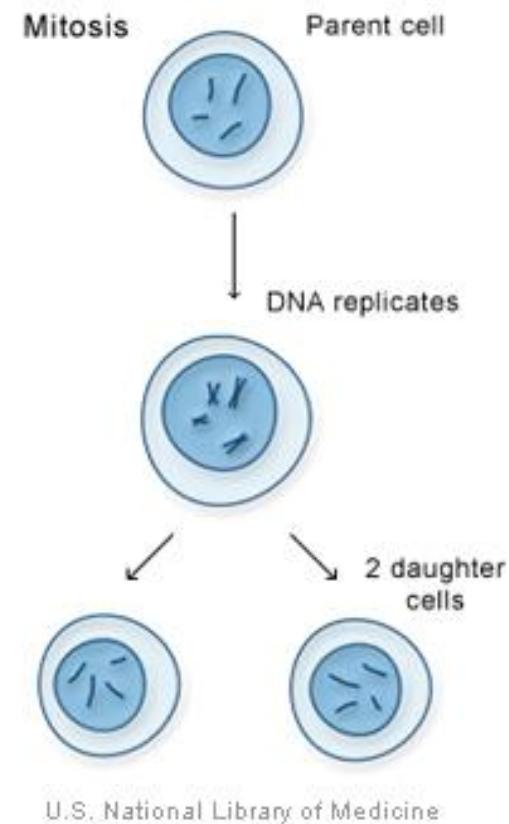


Mitosis = cell division

- Now that there are 2 copies of DNA, the cell can divide in half, placing 1 copy in each cell

– **Mitosis** (also called mitotic cell division) = process by which 1 parent cell gives rise to 2 identical daughter cells

- *Cells that undergo mitosis follow a cell cycle*

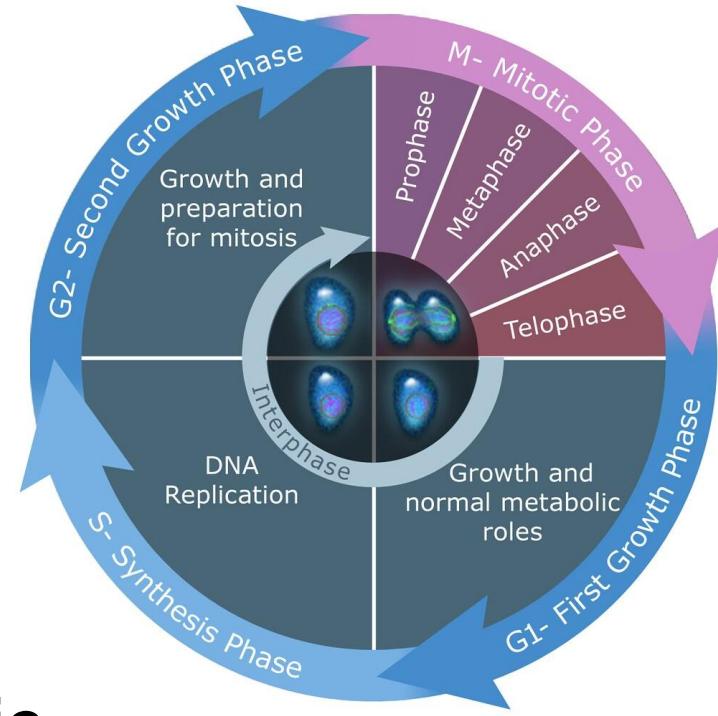


U.S. National Library of Medicine



Cells that undergo mitosis go through a repeating pattern: **cell cycle**

- Divide
- Grow
- (in some cases) Differentiate
- Divide again, etc...
 - some cells **differentiate** (only once): they become specialized cells for specific functions
 - e.g. skin cells, or heart cells, or liver cells



- *BUT: not all cells divide*



Organisms have cells that can & can NOT divide

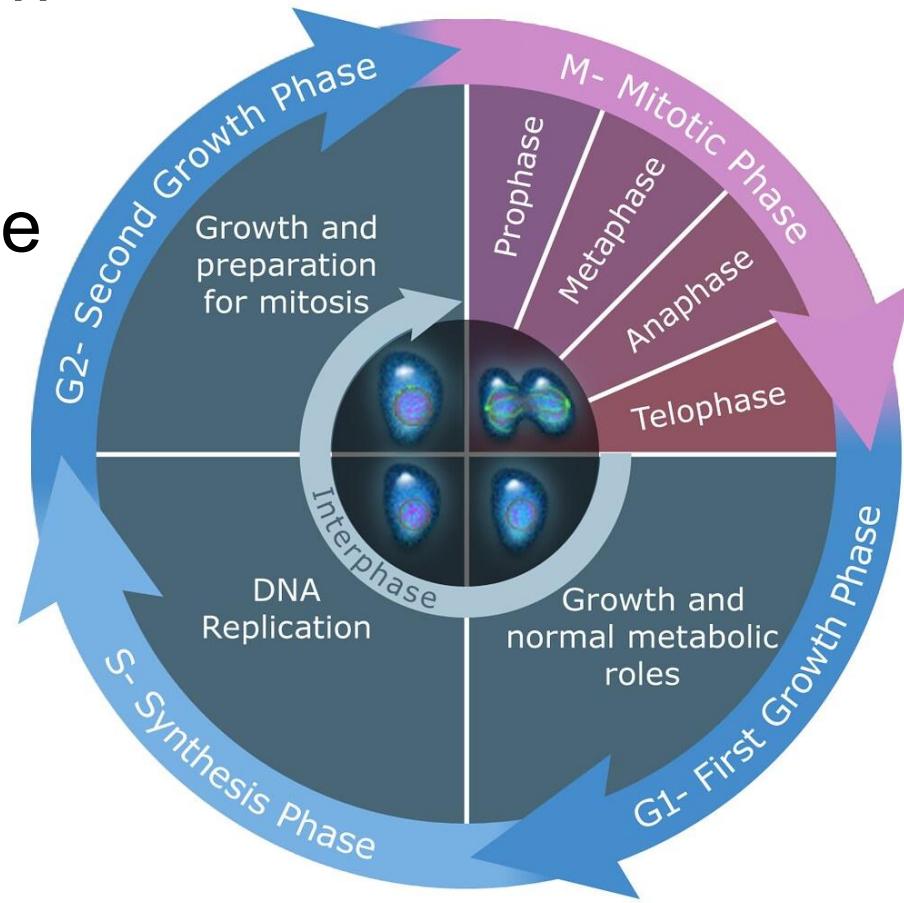
- Stem cells & most other cells in animals can divide: they follow the cell cycle
 - Stem cells have not differentiated yet – can eventually differentiate into a variety of cell types
 - Regular cells have differentiated but keep dividing: liver cells produce more liver cells, etc.
- Certain animal cells have differentiated & will never divide again: leave the cell cycle at the earliest growth phase
 - e.g. most heart & brain cells can NOT divide



Cells that divide through mitosis follow the cell cycle

- **Interphase** = a time for acquisition of nutrients, growth, & DNA replication

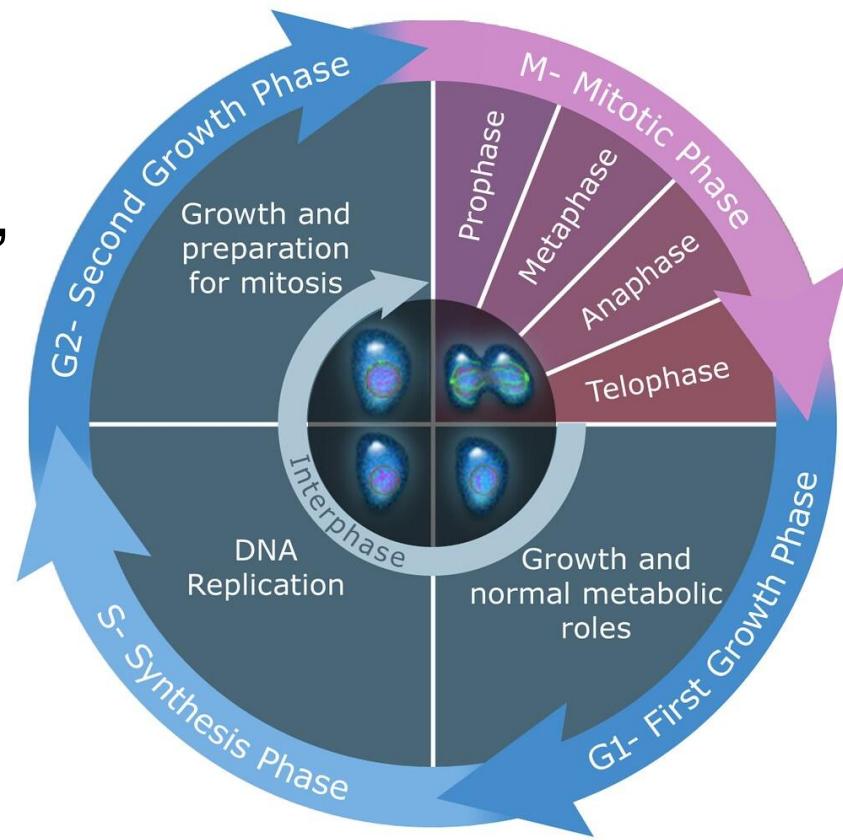
- Cells spend most of their time in interphase
 - (*blue in figure →*)
- Includes G1, S, & G2 phases





Interphase is divided into 3 phases

- **G₁** (growth phase 1) = acquisition of nutrients & growth to proper size
- **S** (synthesis phase) = DNA replication: all DNA is copied, or “synthesized”
- **G₂** (growth phase 2) = completion of cell growth & final preparation for mitosis



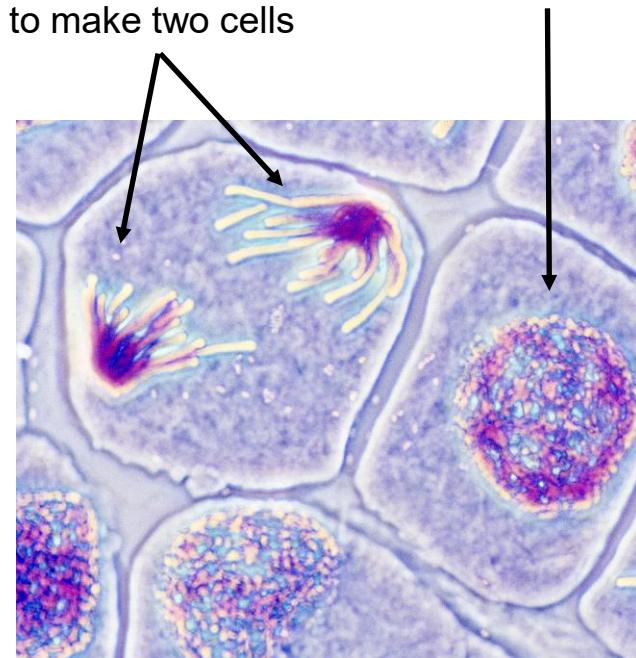


Mitosis = actual cell division: 1 copy of DNA & half of cytoplasm goes into each of 2 daughter cells

- Duplicated DNA/chromosomes are separated in half & pulled to opposite ends of the cell
- Then the cell divides along the middle
 - *has several phases (in pink on figure in the prior slide): we will not cover these phases in detail*

Two copies of DNA being pulled to either side in a cell about to divide down the middle to make two cells

DNA inside nucleus of a cell not dividing





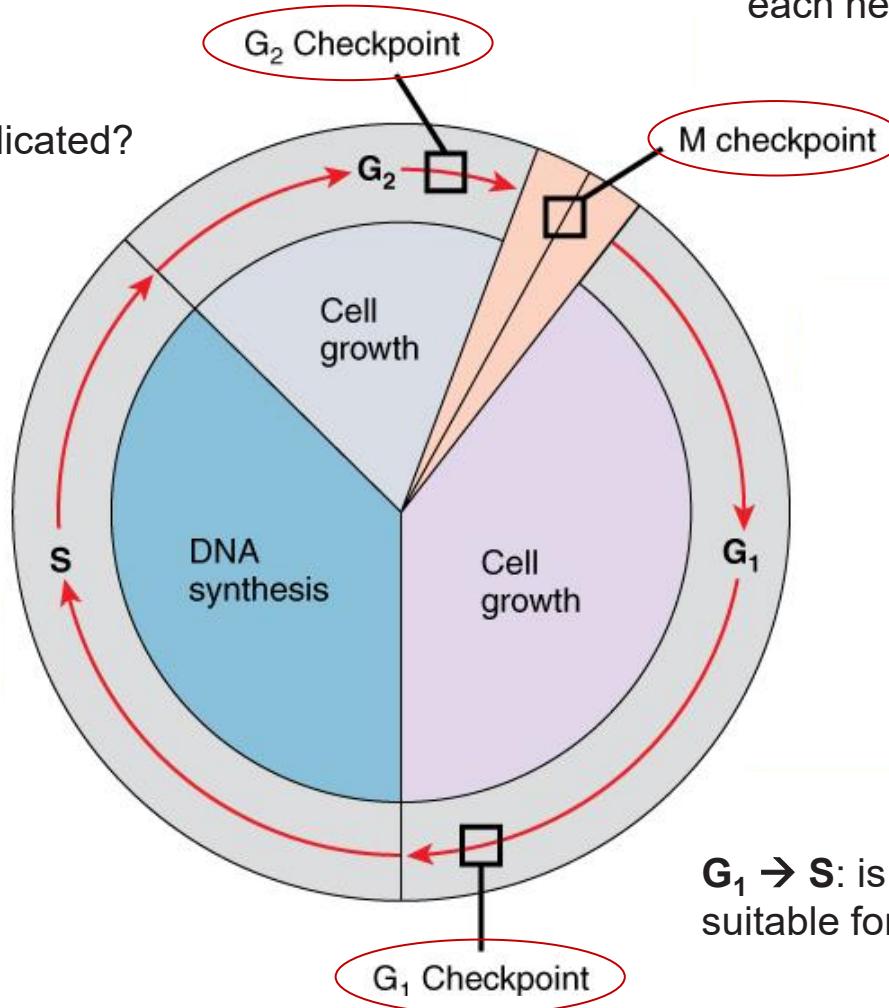
How is mitosis regulated?

- The cell cycle is regulated by several checkpoints
 - **Checkpoints** = enzymes (proteins) that move through the cell to make sure it is healthy before it divides
 - 3 big checkpoints check to be sure:
 - the DNA is healthy & suitable for replication
 - DNA has been fully replicated correctly
 - DNA/chromosomes have split in half perfectly so that each cell will have a full copy



Cell cycle checkpoints:

G₂ → M: has the DNA been completely & accurately replicated?



mid-mitosis: are all of the chromosomes split in half so that each new cell gets a full copy?

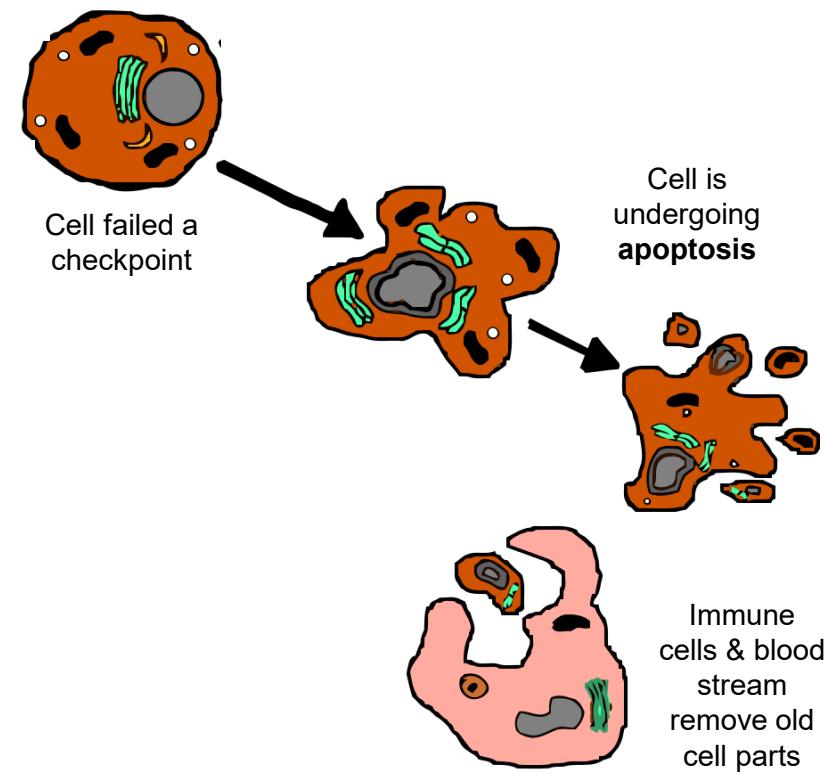
G₁ → S: is the DNA healthy & suitable for replication?



What happens if a cell fails a checkpoint?

- **Apoptosis** = programmed cell death (cell suicide): it seems extreme that a cell would kill itself, but it is very useful for 3 big reasons

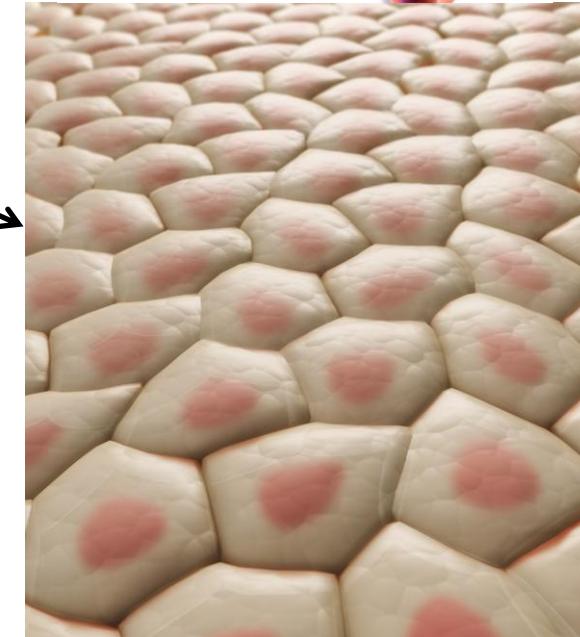
- 1. destroys cells that are not in good condition (infected with a virus, has mutations, etc.)





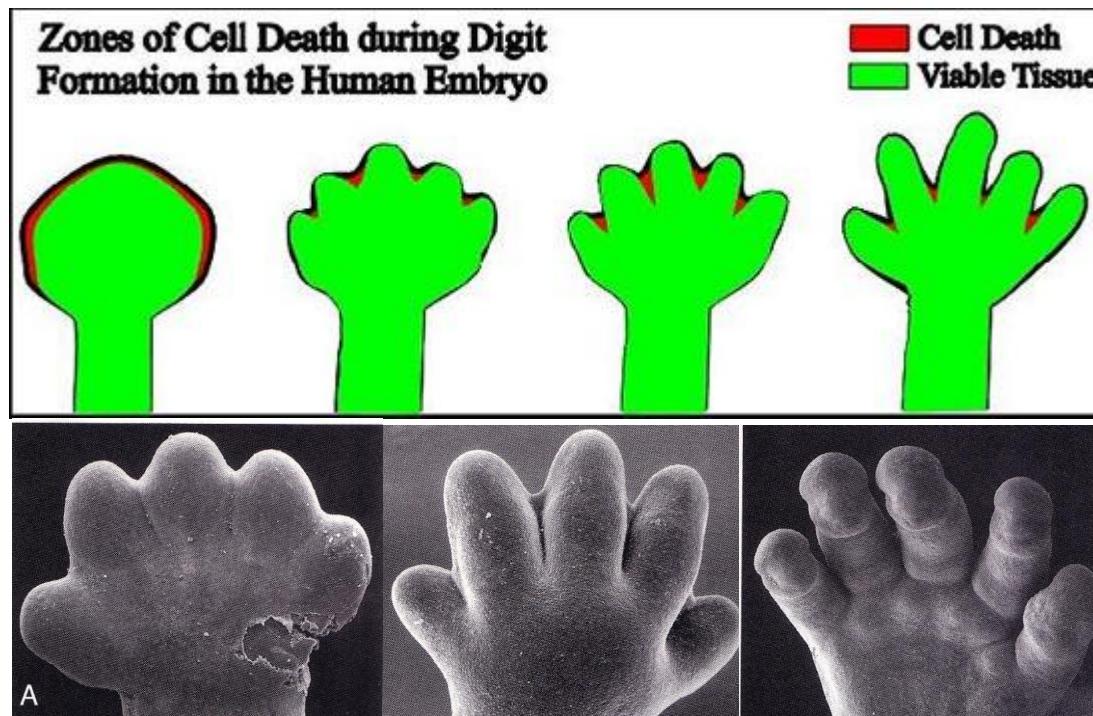
2. removes old cells so that new ones can take their place

- red blood cells only live →
~120 days before undergoing apoptosis & being replaced
- skin cells only live ~20 days
- colon cells only live for ~4 days
before being replaced!



3. removes unneeded cells during development

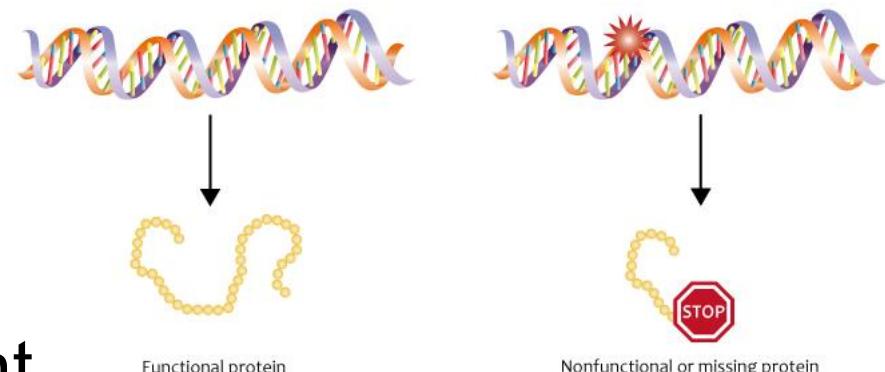
- e.g. hands & feet start out paddle-shaped in early embryos – apoptosis removes unneeded cells, allowing for individual fingers & toes





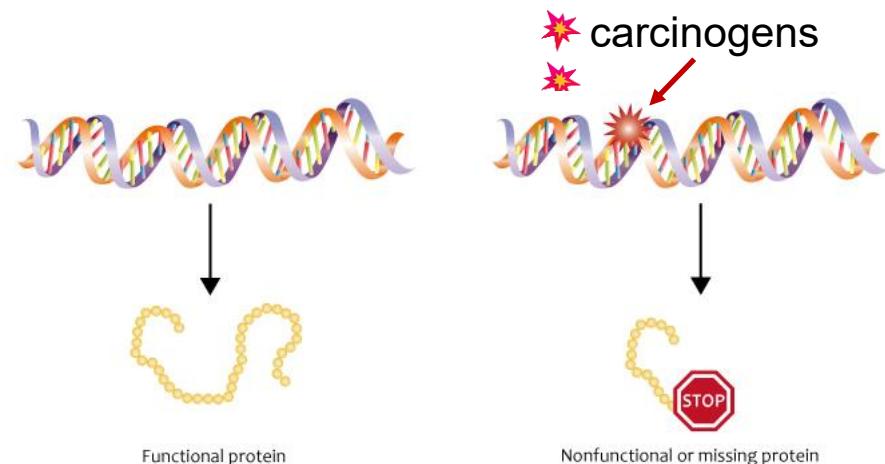
What happens when regulation goes wrong?

- *Remember:* genes (made of DNA) code for proteins
- *Remember:* mutations in genes can = nonfunctional proteins
- *Remember:* the checkpoints regulating the cell cycle are proteins
 - So a mutation to the gene that codes for a checkpoint protein can = a nonfunctional checkpoint



If 1 checkpoint is not working, there are still 2 others

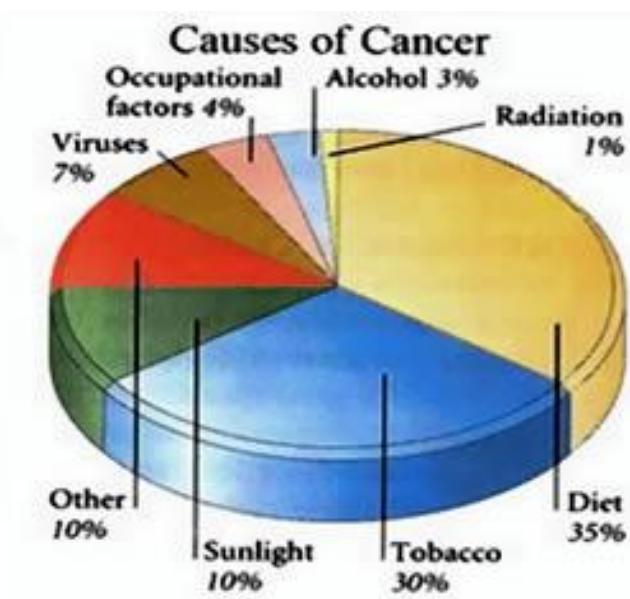
- But what if all 3 fail?
 - Seems unlikely that 1 cell would have multiple mutations at all 3 genes coding for the checkpoints: odds of winning the lottery
 - But: BILLIONS of cells are dividing in our bodies every day – that's a lot of lottery tickets!
 - plus, more & more **carcinogens** are in our environment, increasing the risk of mutations





Carcinogens are common in our environment:

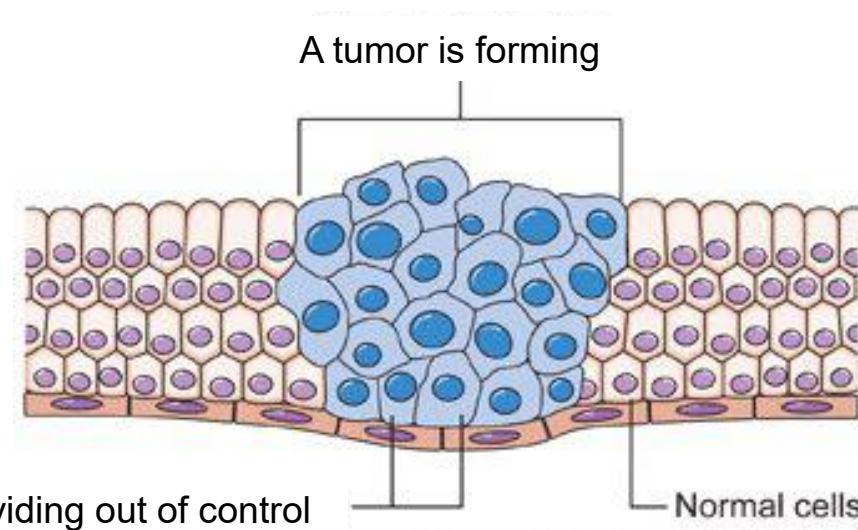
- Some **viruses** (HPV mutates DNA in cervical cells)
- Some **chemicals** (asbestos, pesticides, etc.)
- **Tobacco** (2nd leading cause of death in US)
- **Radiation & sunlight** (x-rays, UV light)
- **Animal proteins & fats**
 - e.g. hormones fed to animals linked to mutations in breast cells
 - e.g. normal red meat linked to mutations in colon cells





So what happens if all of the checkpoints fail to show up & the cell cycle becomes unregulated?

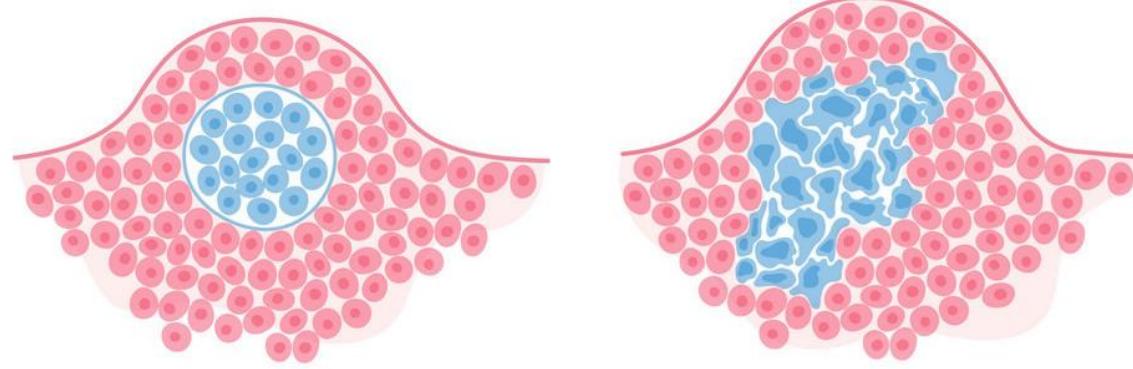
- Cells can grow out of control & form a **tumor**
 - There are 2 types of tumors:
 - **Benign tumor** = large mass of cells that is stationary
 - only dangerous if it damages the surrounding tissue





Malignant tumor = cells dividing out of control that are able to break off & move around the body, invading & damaging other organs & starting new tumors – this movement is called **metastasis**

– **Cancer** = diseases involving malignant tumor cells



Benign tumor

- Non-cancerous
- Capsulated
- Non-invasive
- Slow growing
- Do not metastasize (spread) to other parts of the body
- Cells are normal

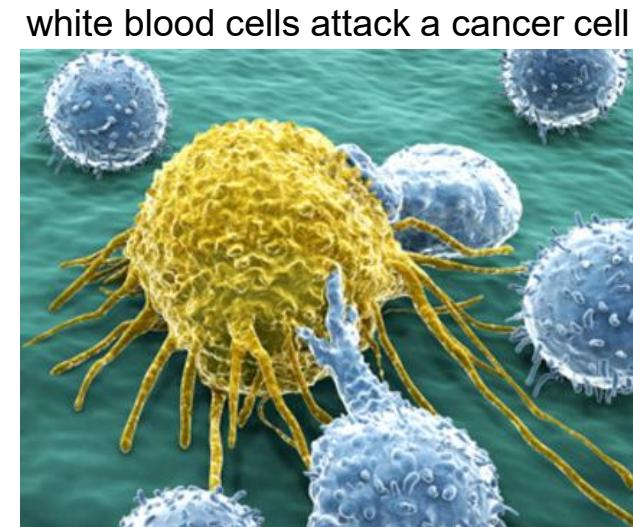
Malignant tumor

- Cancerous
- Non-capsulated
- Fast growing
- Metastasize (spread) to other parts of the body
- Cells have large, dark nuclei; may have abnormal shape



Cells with mutations form in our bodies all the time, especially since carcinogens are so common – so why haven't we all be diagnosed with cancer?

- The immune system recognizes & destroys mutated & cancerous cells before they can spread
- Then why does anyone get cancer?
 - there are some cancerous cells that evade the immune system
 - the immune system may not be functioning at optimal levels
 - too many carcinogens may overwhelm the immune system





The majority of cancers are caused by some factors that are under our control

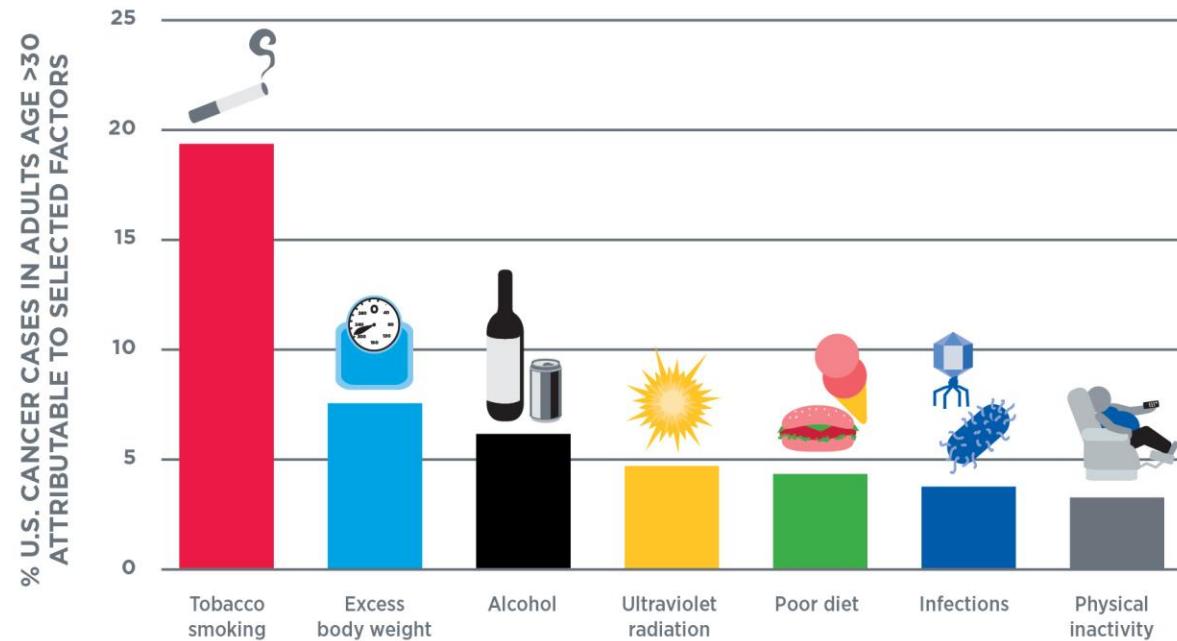
- e.g. exposing ourselves to carcinogens & not keeping our bodies in good health
 - *tobacco use, diet (fried foods, animal protein), sun exposure, environmental pollutants, infections, stress, & physical inactivity*
- In the media there tends to be more focus on *treatments* rather than *prevention*, but cancer is considered a “largely preventable disease”





FIGURE 2

INCREASING CANCER RISK



Research has identified numerous factors that increase an individual's risk for developing cancer. By modifying behavior, individuals can eliminate or reduce many of these risks and thereby reduce

their risk of cancer. Developing and implementing additional public education and policy initiatives could help further reduce the burden of cancers related to preventable cancer risk factors.

Data from (46). Figure adapted from (15)

American Association for Cancer Research (AACR) Cancer Progress Report 2019



How do we prevent (or reduce our risk of) cancer?

- 1. Healthy diet & physical activity
(keeps immune system at peak)
- 2. Avoid carcinogens as much as possible
(UV rays, tobacco, pesticides & other pollutants, etc.)
- 3. Vaccinations
(HPV & Hepatitis B)

REDUCE YOUR RISK FOR CANCER BY MAINTAINING A HEALTHY WEIGHT, BEING PHYSICALLY ACTIVE, AND CONSUMING A BALANCED DIET

Research shows that about one-fifth of all cancers diagnosed in the United States can be attributed to being overweight or obese, being physically inactive, eating poorly, and drinking excessively. Based on current evidence experts from the World Cancer Research Fund International recommend people:

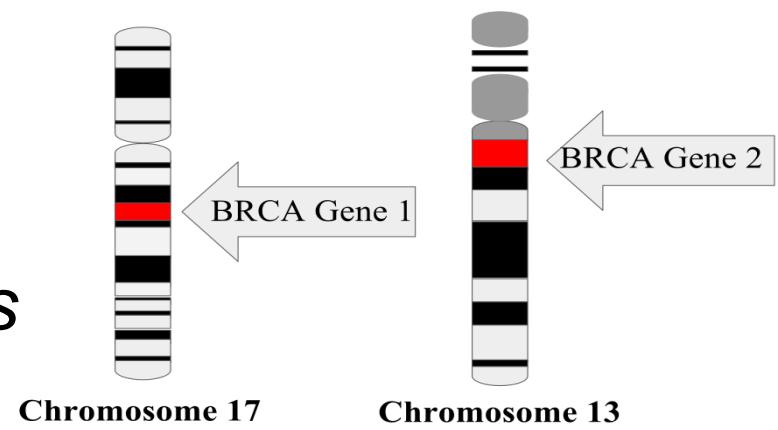
Maintain a healthy weight (body mass index [BMI] between 18.5 and 24.9) because 15 types of cancer have been causally linked to being obese or overweight (see Figure 4 , p. 30). 	Limit intake of red and processed meats (e.g., hot dogs, bacon, and salami) because these foods can increase risk for colorectal cancer.
Be physically active as part of everyday life; regular physical activity can decrease risk for eight types of cancer (see Figure 4 , p. 30, and sidebar on Physical Activity Guidelines , p. 32). 	Limit intake of sugar-sweetened drinks since these lead to weight gain; drink mostly water.
Eat a diet rich in vegetables, fruits, whole grains, and beans, because these foods have a low energy density and, therefore, promote healthy weight. 	If consumed at all, limit alcoholic drinks, because alcohol consumption can increase risk for six types of cancer (see Figure 5 , p. 33).
Limit consumption of "fast foods" and other processed foods high in fat, starches, or sugars because these contribute to weight gain. 	

Source: <https://www.wcrf.org/dietandcancer/resources-and-toolkit>



~5% of all cancer has a genetic link

- There are genes a person can inherit which alter the cell cycle, making it more likely that malignant cells will occur
- e.g. BRCA1 & 2 are genes that produce proteins that help repair damaged DNA – a damaged gene in either location leads to increased risk of breast & ovarian cancer
- *only a small percent of breast & ovarian cancer are due to these mutations – they are not common*





What are the treatments for cancer?

- Treating cancer is challenging: hard to target & kill some of our cells without killing the healthy ones
 - **Surgery:** cutting out the harmful cells is always the 1st line of defense
 - **Radiation:** if a malignant tumor hasn't spread (metastasized), radiation directed at the tumor kills the cells
 - cannot be used on whole body (would kill all cells, even healthy ones)

A cancer patient receives a beam of radiation energy from this large machine





What are the treatments for cancer?

- **Chemotherapy**: drugs that stop cells from dividing (mitosis) – affects whole body so possible after metastasis
 - But these stop ALL cells in the body from dividing, which causes unpleasant side effects
 - e.g. no new hair follicle cells = loss of hair
 - e.g. no new intestinal cells = nausea
 - e.g. no new red blood cells = fatigue & shortness of breath

This woman is being treated with chemotherapy for breast cancer. Cooling mitts are placed on her hands and feet to ease deleterious effects on the nails. Similar strategies can be used to slow down hair loss.





What are the treatments for cancer?

- **Changes to diet & lifestyle:** can improve the bodies own immune response – can better fight cancer cells
- Researchers are always searching for new options:
 - e.g. **Immunotherapy** = stimulating the immune system to recognize the cancer cells
 - e.g. **Targeted therapy** = attempts to develop drugs that ONLY target & kill cancer cells
 - e.g. **Angiogenesis inhibitors** = stop blood vessel growth – cuts off blood supply to tumor



Chapter 9: Sexual Reproduction & Meiosis

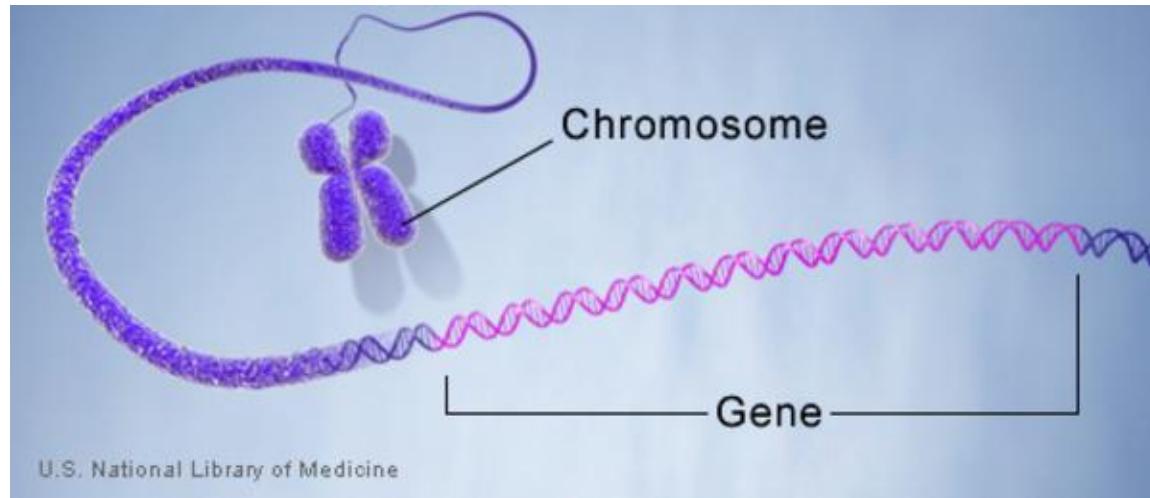
- What are characteristics of the average cell?
- Meiosis = a unique type of cell division
- How is sex determined in offspring?
- What happens when meiosis goes wrong?

Corresponds with OpenStax Biology 2e Chapter 11



What are characteristics of the average cell?

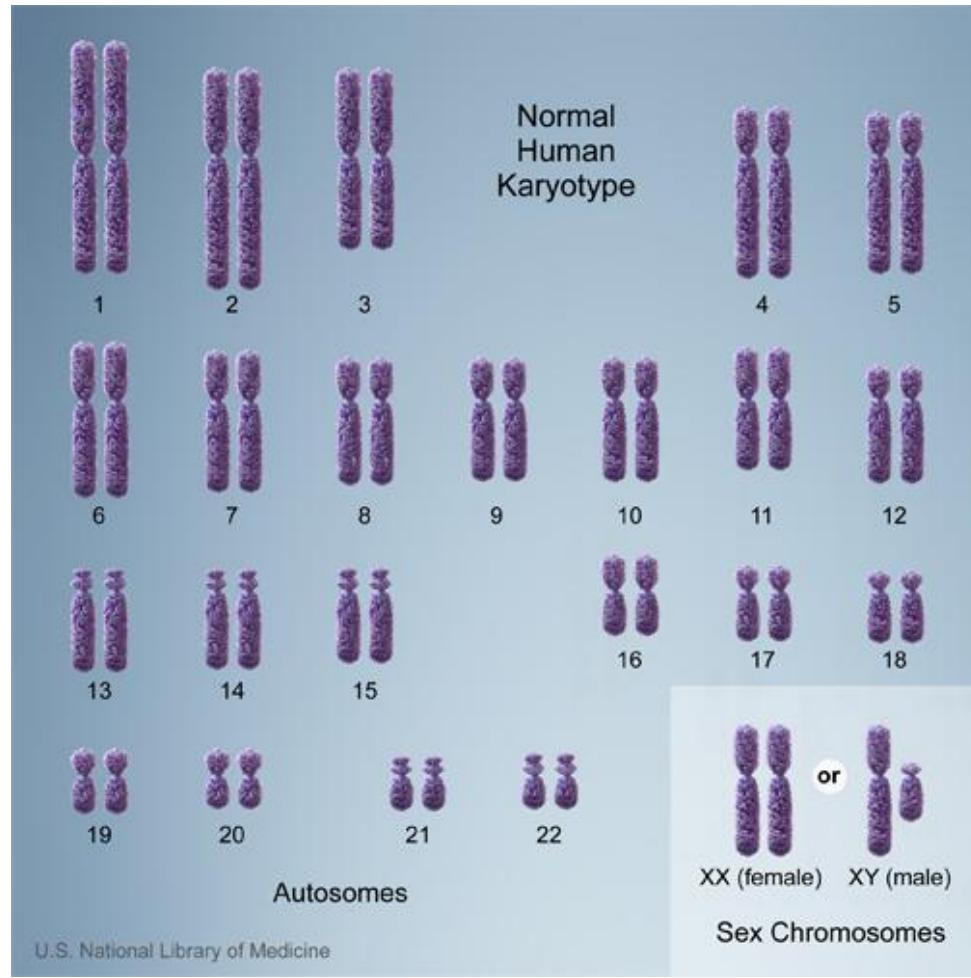
- The average human cell has **23 pairs of chromosomes**
 - 1 from each parent, for a total of 46 per cell
- *Remember:* we have about 25,000 pairs of genes spread across our 23 pairs of chromosomes





We can visualize our 23 pairs of chromosomes in a **karyotype**: a size-ordered chart of all the chromosomes in one cell

- Cells with 2 sets of chromosomes (1 set from each parent) = **diploid**
- 22 of our 23 chromosomes are **autosomes**
 - chromosomes that are the same for all people

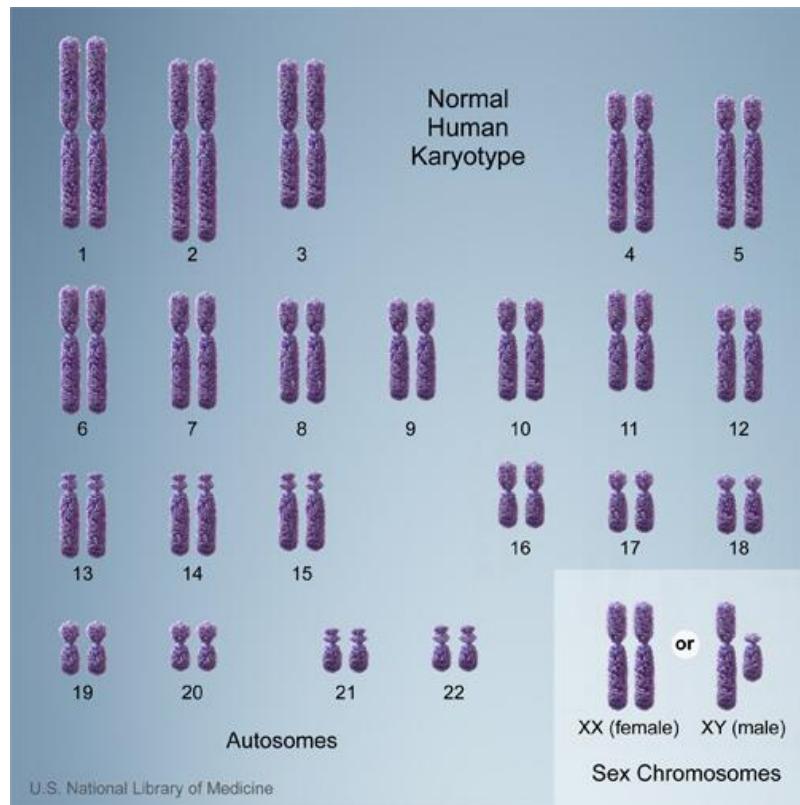




For autosomes, each chromosome from 1 parent has a matching chromosome from the other parent
= **homologous chromosomes**

– homologous = “having the same basic structure”

- e.g. chromosome 21 includes 367 genes in a very specific order
 - this is exactly the same for both copies, 1 from each parent

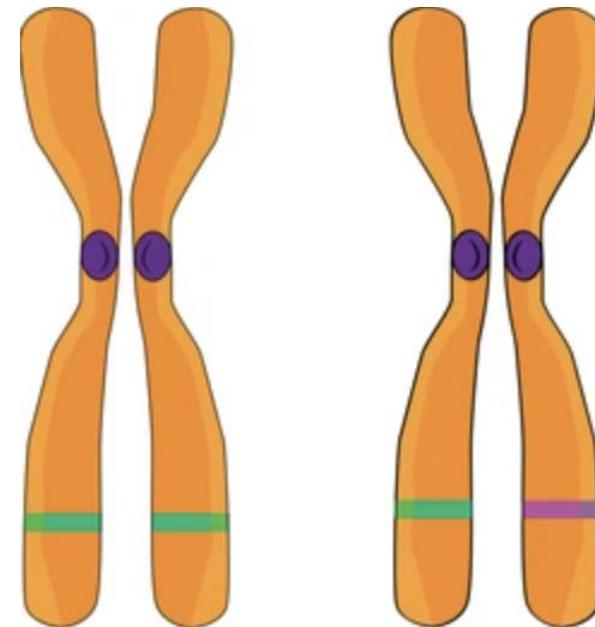




Homologous chromosomes match even though they are from different parents

– HOWEVER: homologous chromosomes are not necessarily identical

- Have the same genes in the same order
- Do NOT necessarily have the same alleles
 - **Alleles** = different versions of a gene



2 genes,
same alleles

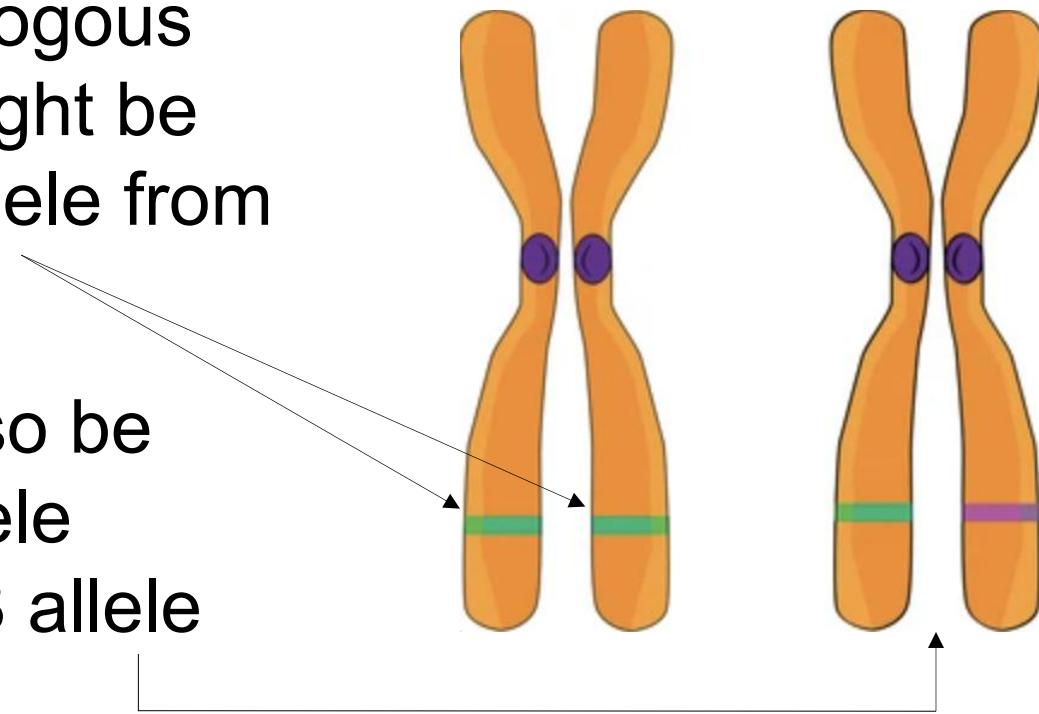
2 genes,
2 different alleles



Alleles = different versions of a gene

– e.g. all of us have 2 genes for blood type, but we have some combination of the different possible alleles that code for blood type: A, B, or O

- a person's homologous chromosomes might be identical: 1 "O" allele from each parent
- but they might also be different: an A allele from 1 parent & B allele from the other

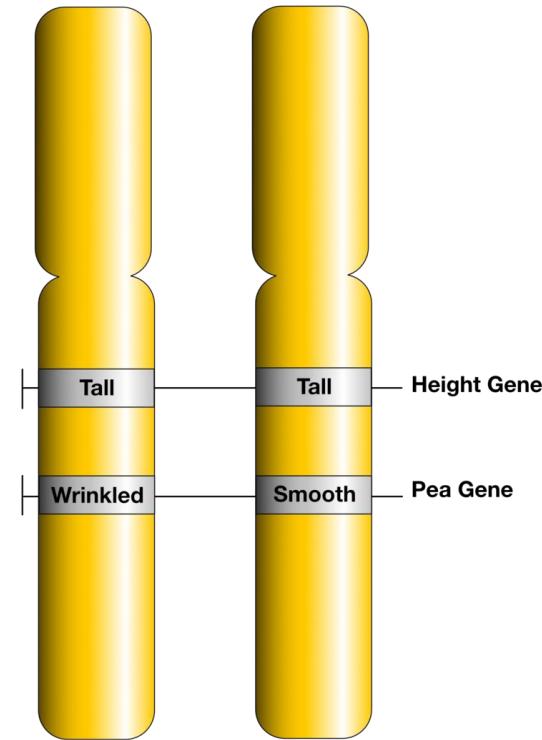




Other allele examples:

- cats have 2 genes for fur length, which has the possible alleles coding for short & long fur
- e.g. plants have 2 genes for flower color, but there can be many different possible alleles, coding for red, yellow, or purple flowers

Chromosome from Parent 1 Chromosome from Parent 2



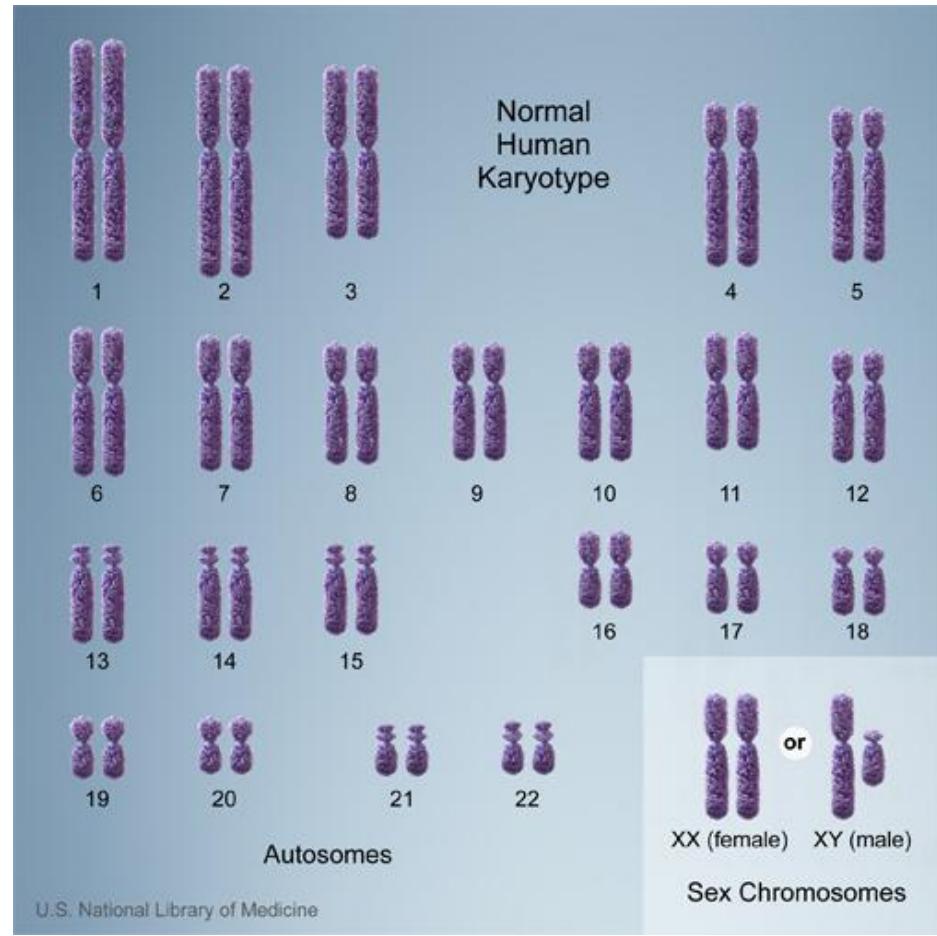
- e.g. pea plants have 2 genes for pea shape, which has the possible alleles coding for wrinkled or smooth peas



The last pair of chromosomes = **sex chromosomes**

– these usually determine whether a person will develop:

- ovaries – X & X
or
- testes – X & Y



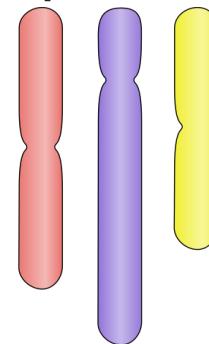


Not all cells are diploid, with 2 sets of chromosomes

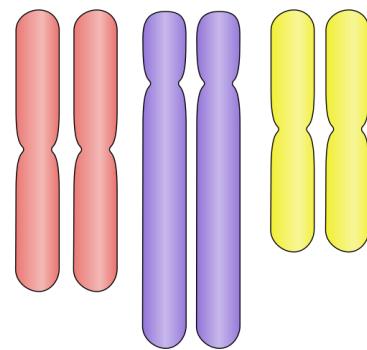
- Some cells are **haploid**: they only have 1 set of chromosomes (half)
 - these cells are **gametes**: sperm or egg cells
- gametes are made in ovaries (eggs) or testes (sperm) through a type cell division called **meiosis**

In egg or sperm cells (gametes), we have only one of each chromosome

Haploid (N)



Diploid (2N)

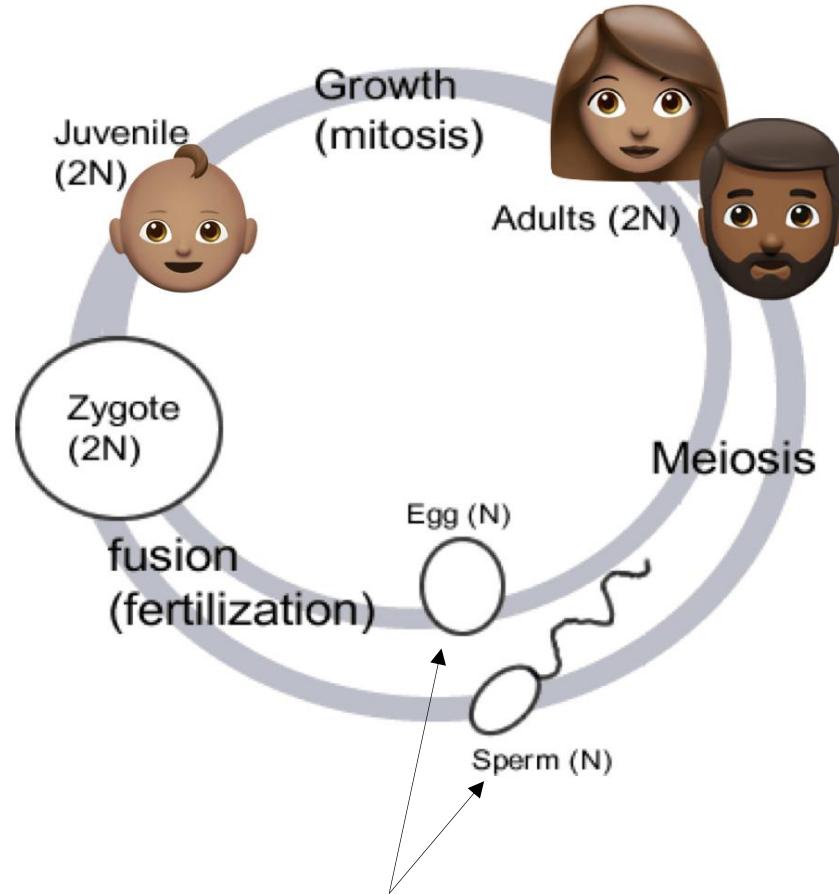


In regular cells, we have one chromosome from each parent (two of each = diploid)



Why would we make haploid cells?

- Sexual reproduction means that 2 cells (sperm & egg) must meet to make 1 new organism
 - when a haploid sperm fertilizes a haploid egg, the offspring will have 2 sets of chromosomes: now diploid



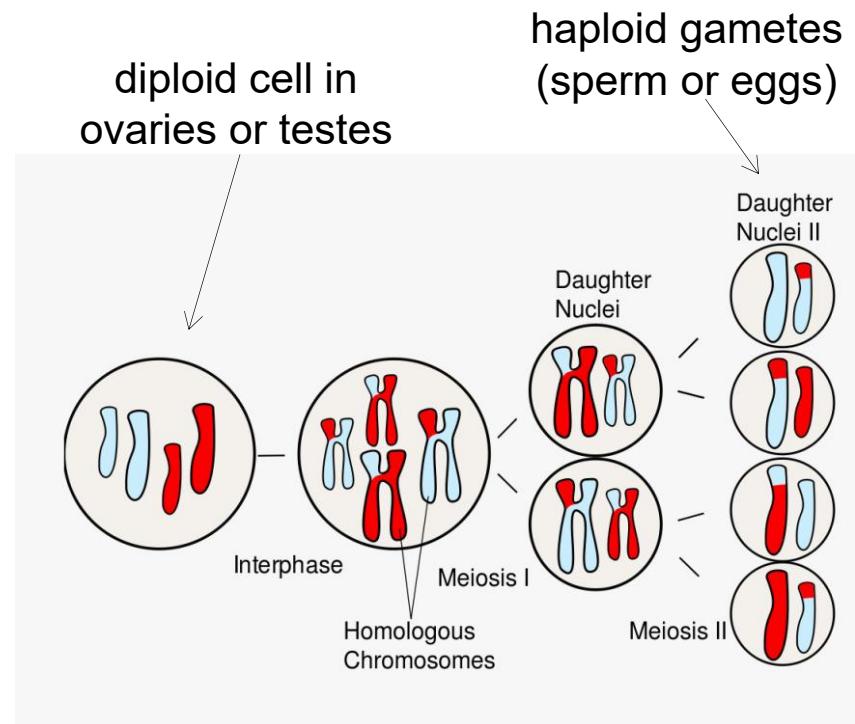
Haploid gametes fuse:
 $1N + 1N = 2N$

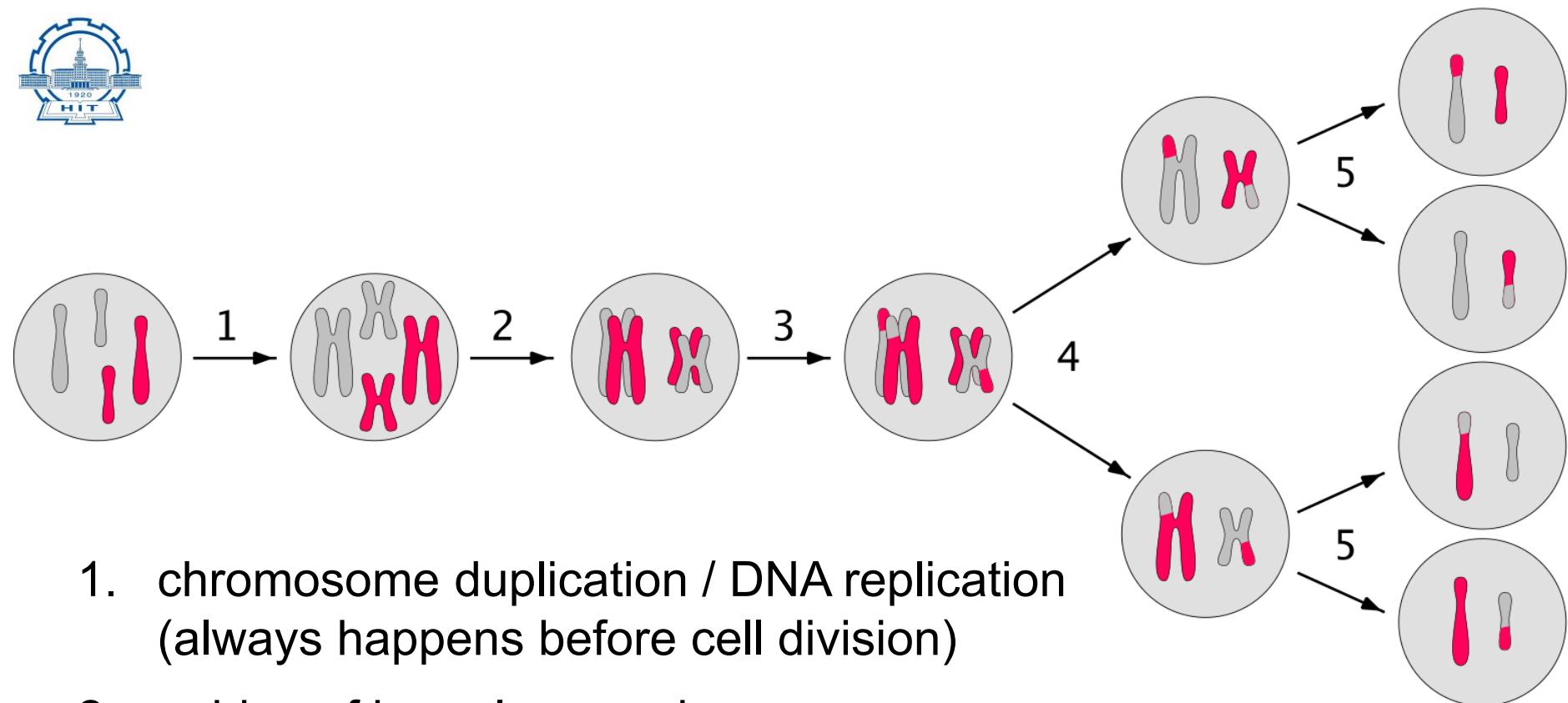
So offspring is now 2N
(diploid) like their parents



Meiosis = a unique type of cell division

- Haploid gametes for sexual reproduction are made through **meiosis**
 - Through 2 divisions, 1 parent cell produces 4 daughter cells / gametes
 - Each gamete has half the genetic material of the parent cell (haploid)
 - Gametes are NOT genetically identical to the parent cell



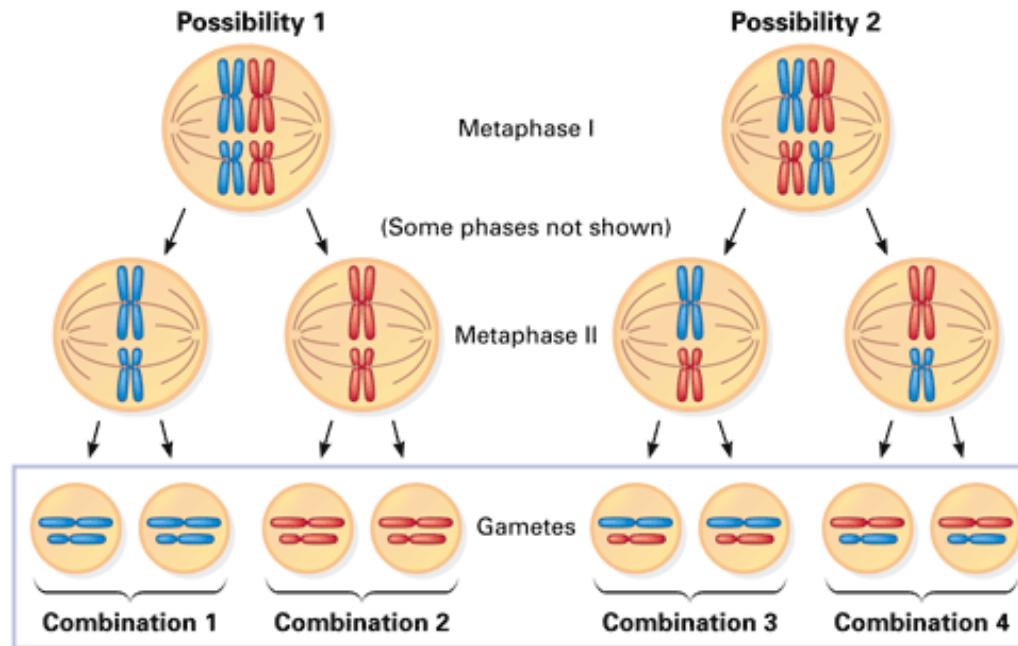


1. chromosome duplication / DNA replication
(always happens before cell division)
2. pairing of homologous chromosomes
3. crossing-over (*more on this soon*)
4. meiosis I (first division) – one of each duplicated chromosome per daughter cell
5. meiosis II (second division) – one of each chromosome per daughter cell (gamete)



Gametes are NOT genetically identical to each other

- Chromosomes are randomly lined up & split during meiosis
 - this means there are several possible combinations

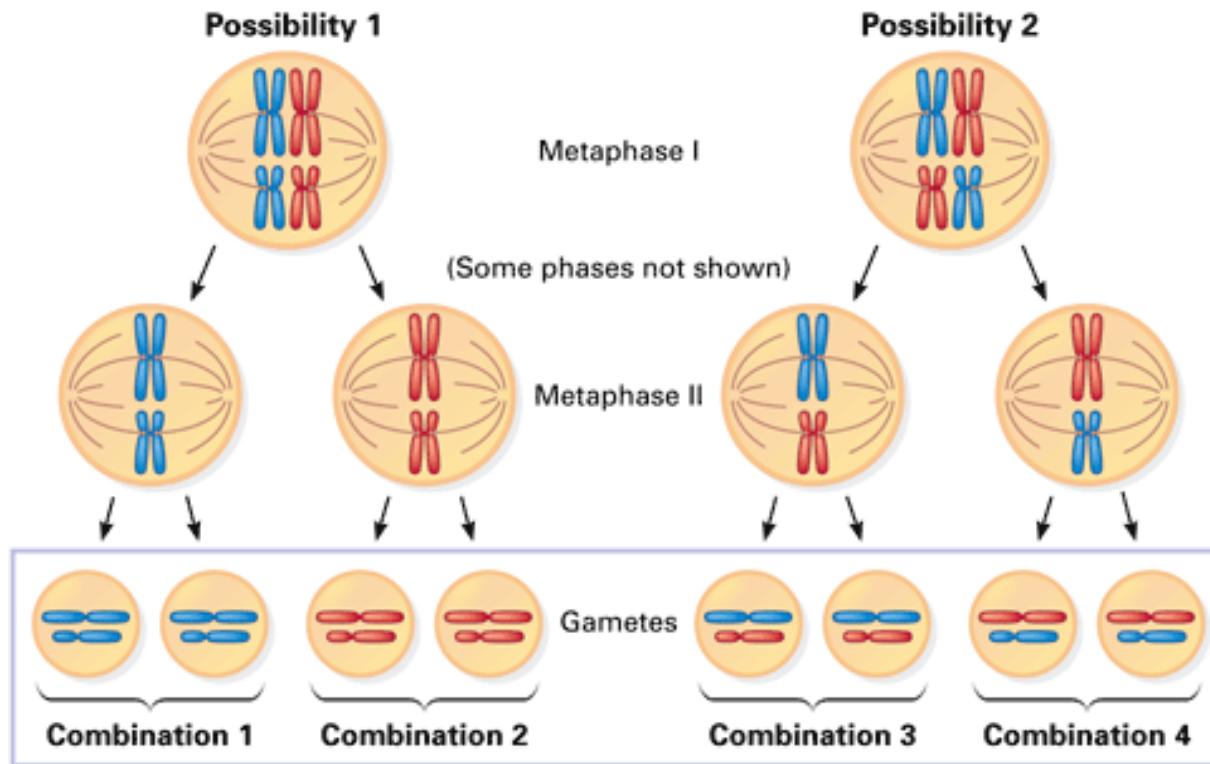


This figure shows that only one change in how two chromosomes are lined up create four possible gametes. But we have 23 chromosomes, not just two, leading to huge variation in gametes.



Gametes are NOT genetically identical to each other

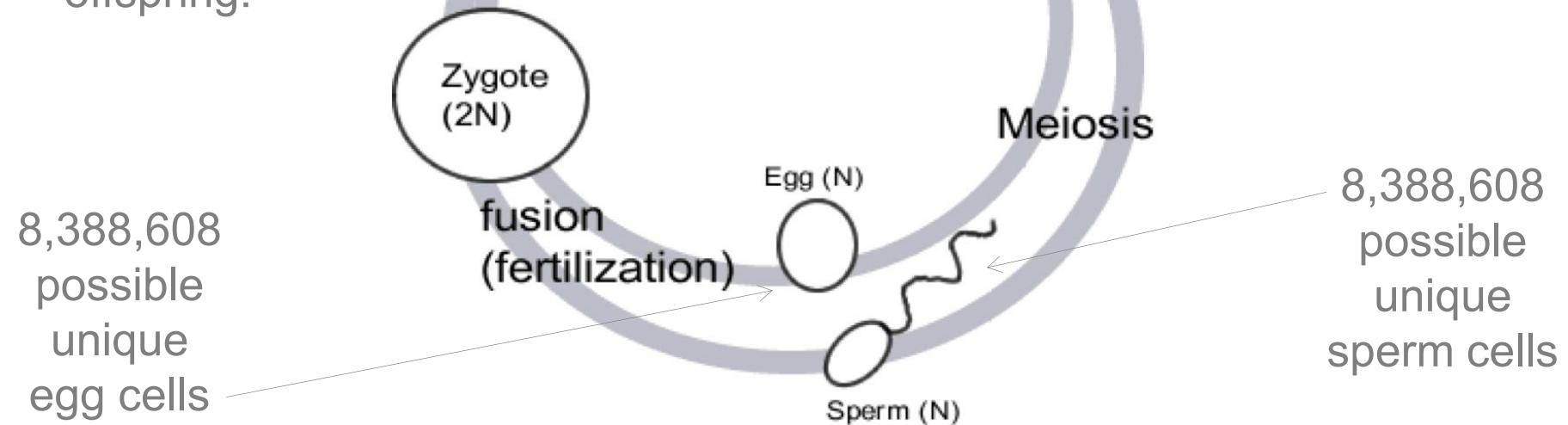
- Humans have 23 pairs of chromosomes – when those can all line up randomly, we can make 8,388,608 possible unique gametes, all equally likely





With 23 chromosomes, incredible human diversity is possible

>70 trillion unique possible offspring!

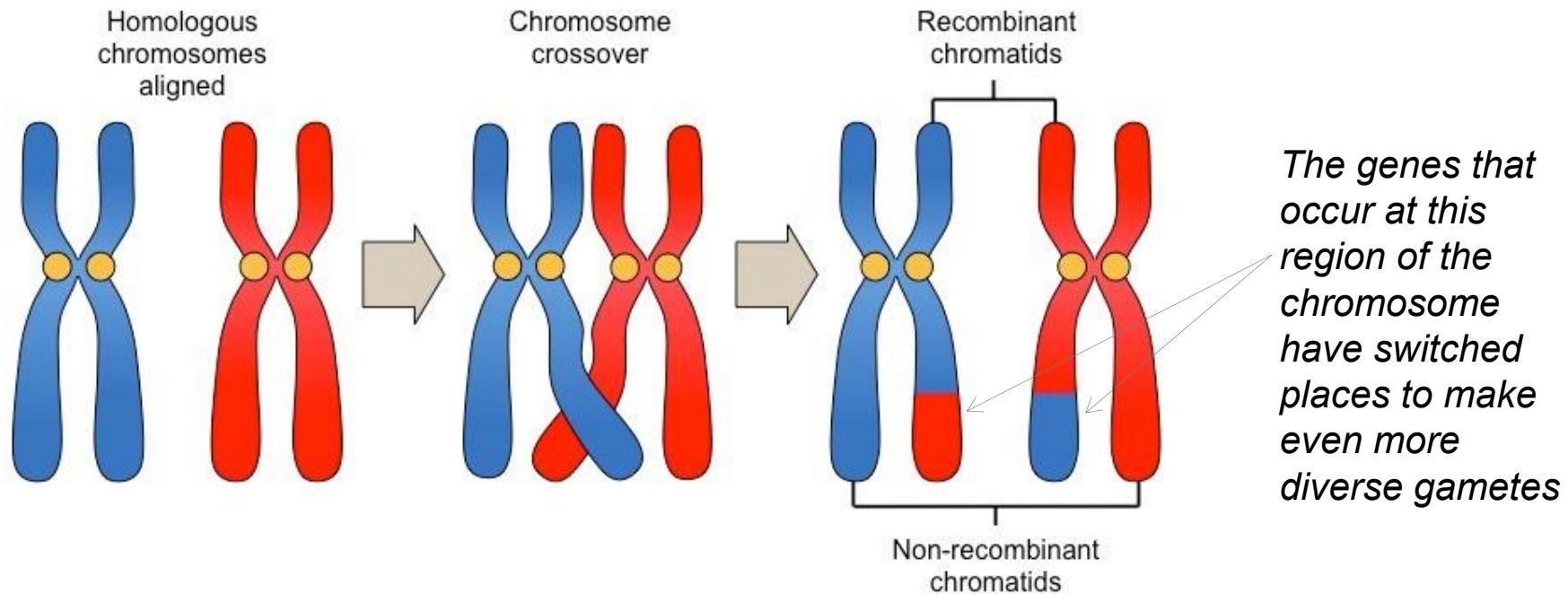


$$8,388,608 \times 2 \text{ (egg} \times \text{sperm)} = \text{more than } 70 \text{ trillion unique individuals!}$$



Diversity is increased even more by crossing over

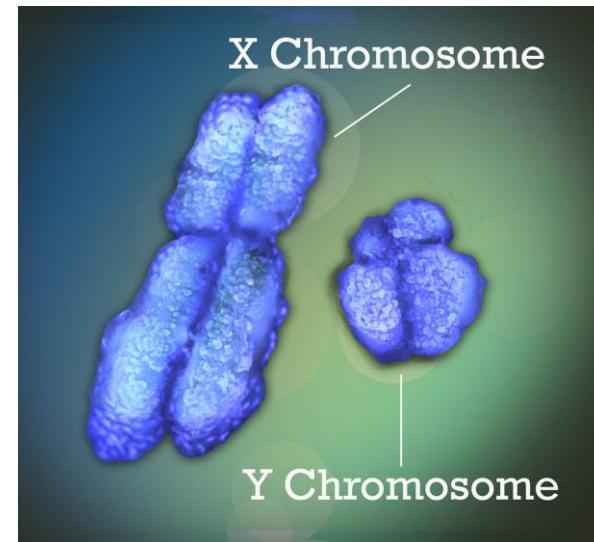
- **Crossing over** = the exchange of DNA during meiosis – genes sometimes change places



Because of crossing over, 70 trillion unique individuals is a vast understatement!

How is sex determined in offspring?

- Mammals have a set of **sex chromosomes** that determine sex: the biological makeup of an individual's chromosomes & reproductive anatomy
 - We usually describe 2 sexes
 - Females: two X chromosomes, development of ovaries
 - Males: an X & Y chromosome, development of testes
 - *Not everyone fits into these two categories*
 - *Sex is different than gender*





Remember: after meiosis each gamete only has 1 of each chromosome

- All female eggs will carry an X chromosome
- In male sperm, half carry an X & half carry a Y

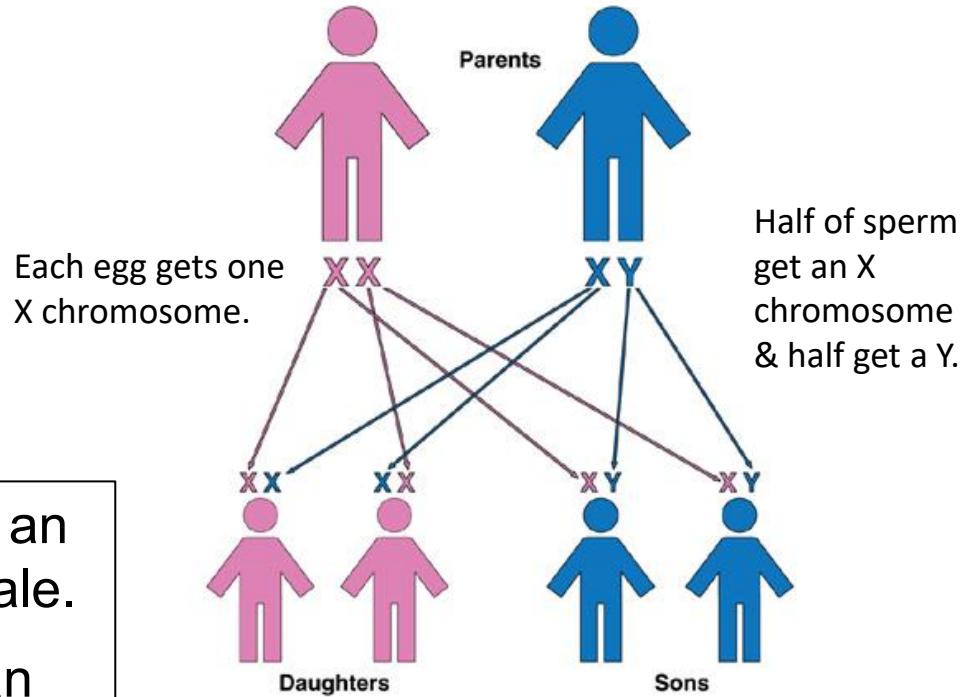
If an X-carrying sperm meets an egg, the offspring will be female.

If a Y-carrying sperm meets an egg, the offspring will be male.

Individuals have two copies of the sex chromosomes in every cell.

Females have two copies of the X chromosome.

Males have one X chromosome and one Y chromosome.

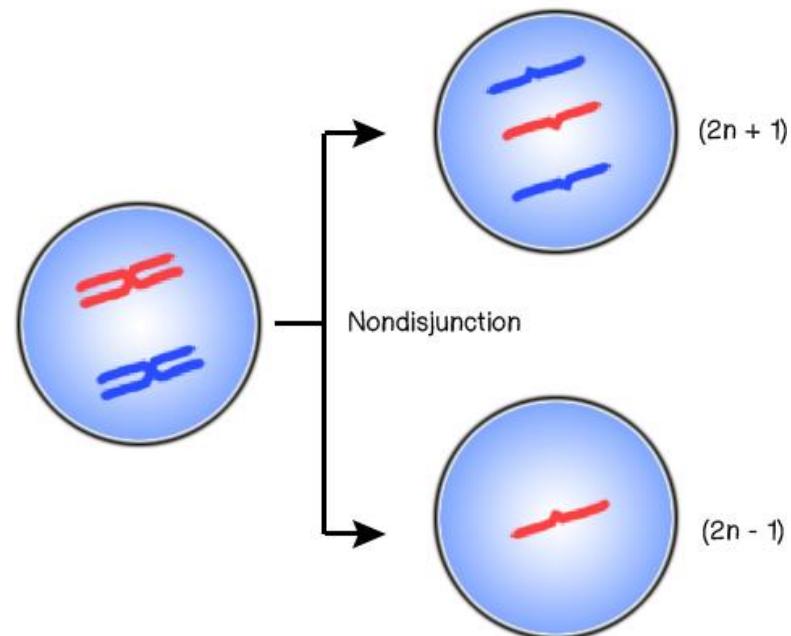




What happens when meiosis goes wrong?

- **Nondisjunction** = the incorrect separation of chromosomes at some point during meiosis
 - The result is an inappropriate number of chromosomes in the gametes (0 or 2 instead of 1)
 - Can happen in sperm or egg cells

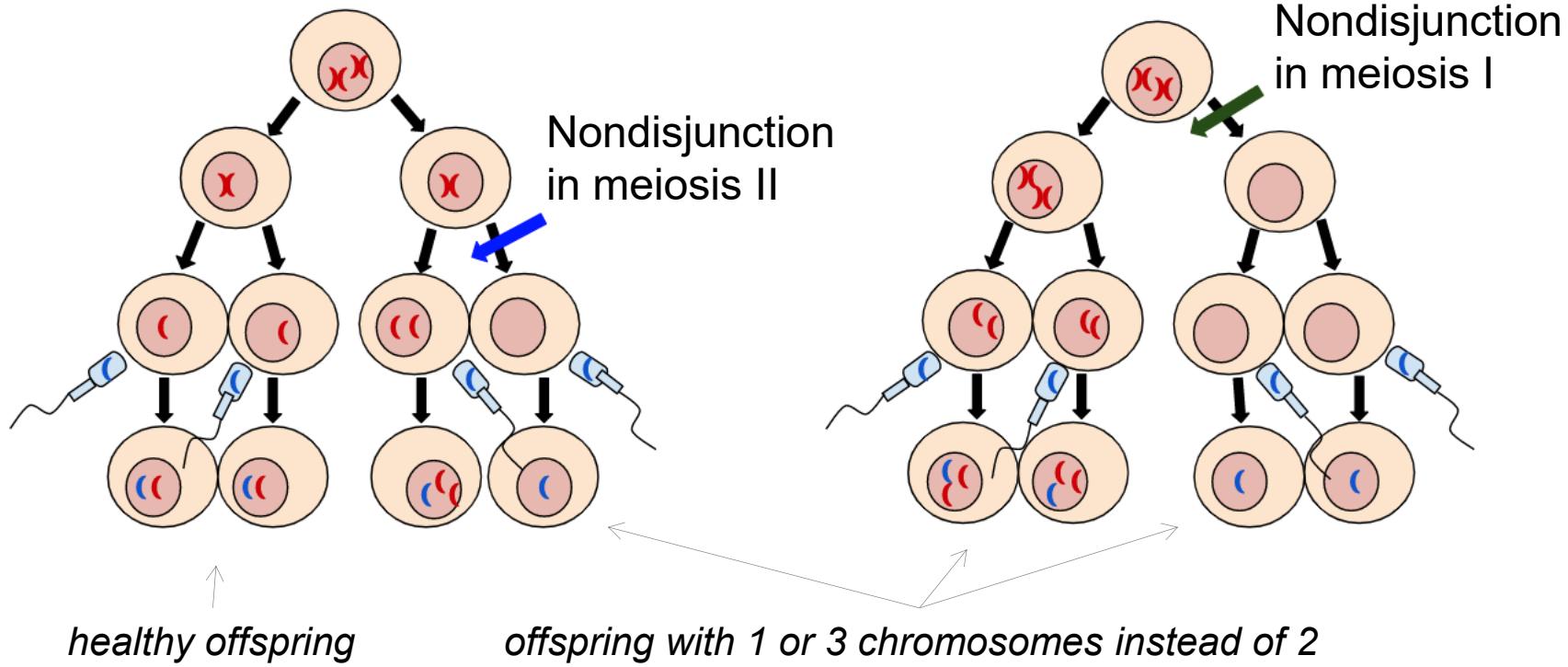
Nondisjunction in Mitosis





Nondisjunction can occur at either division in meiosis

– *in either case, when fertilization happens the offspring will have 1 or 3 chromosomes instead of 2 (diploid)*





What are the consequences of nondisjunction?

- Most embryos formed from gametes with abnormal chromosome numbers result in miscarriage or die very shortly after birth
- Some can survive: specifically if the nondisjunction happened with chromosomes 13, 18, 21, or the sex chromosomes

- these individuals may have impairments

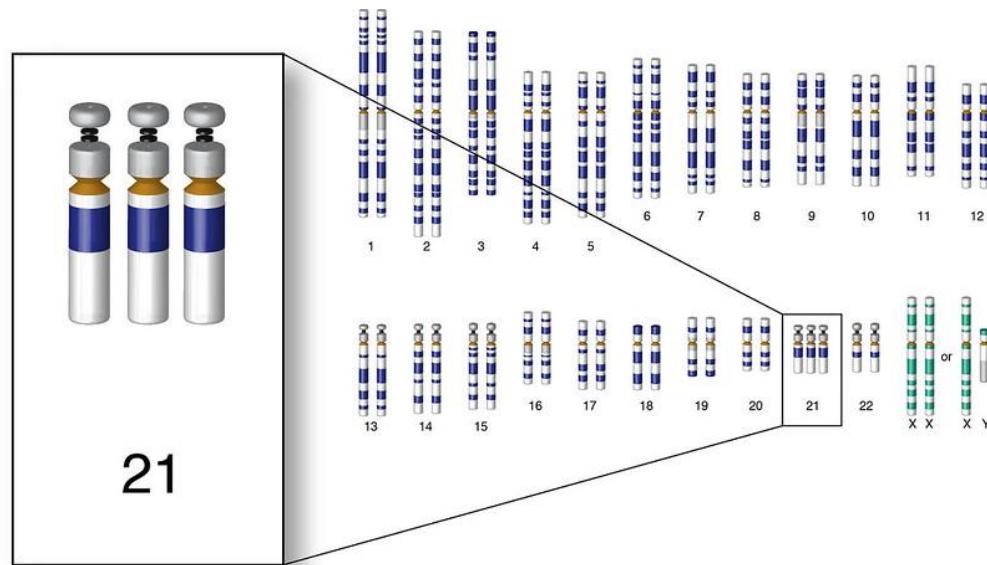


This person has nondisjunction in the sex chromosomes



A common example of nondisjunction = down syndrome

- Also called trisomy 21 because affected individuals have 3 copies of chromosome 21
 - includes several characteristics, including distinctive facial features & often varying degrees of learning difficulties





Other common examples of nondisjunction occur with sex chromosomes

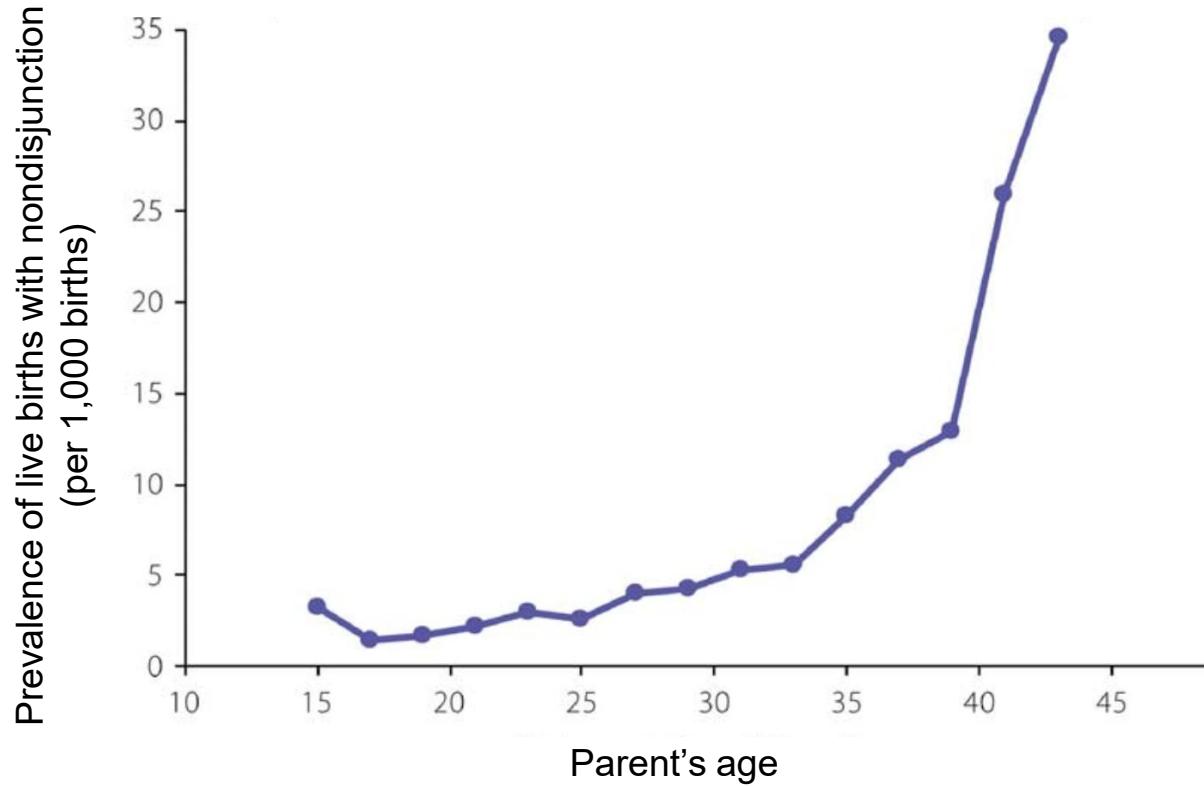
– these can cause problems with reproduction & development but can also have no symptoms at all

Genotype	Syndrome characteristics
XO	Turner Syndrome (female, one X) <ul style="list-style-type: none">• short height• underdeveloped ovaries, often infertile• hormone deficiencies can lead to prevention of puberty & menstruation
XXY	Klinefelter Syndrome (male) <ul style="list-style-type: none">• underdeveloped testes• lower testosterone levels, usually infertile• possible development of some female features• however, many males show no symptoms
XYY	XYY (male) <ul style="list-style-type: none">• most have normal development & offspring• more prone to tallness & acne but most males show no symptoms
XXX	XXX / Trisomy X (female) <ul style="list-style-type: none">• most have normal development & offspring
YO	One Y - not viable (offspring would not survive)



The frequency of nondisjunction in both males & females increases with age

- Doctors will often run more tests on pregnant females older than 35 to find out early about nondisjunction





Chapter 10: Patterns of Inheritance

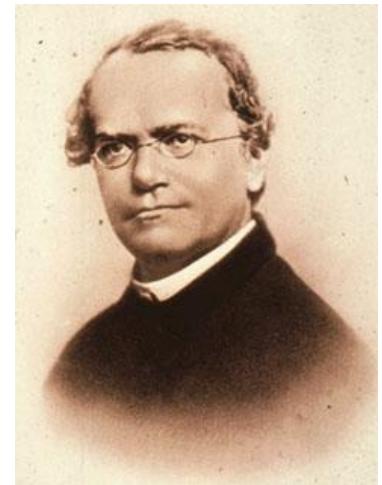
- The genetic secrets that Mendel discovered
- Can we predict the chances of passing on certain genes to offspring?
- Sex-linked genes
- 5 realms of non-Mendelian genetics
- Genetic anomalies
- The environment affects traits too

Corresponds with OpenStax Biology 2e Chapter 12 & 13



The genetic secrets that Mendel discovered

- In the 1800s, no one knew anything about inheritance – only that offspring resembled their parents
 - Gregor Mendel, an Austrian monk, discovered some of the most basic principles of genetics
 - used pea plants in all of his experiments



Gregor Mendel (1822–1884)





Why use pea plants?

- They are easy to maintain & breed
- They reproduce quickly, so multiple generations can be observed
- They have many easy to see traits with only two variants each

Easy to see traits, only 2 variants each

Characteristics of pea plants Gregor Mendel used in his inheritance experiments							
Seeds		Flower colour		Pod		Stem	
form	cotyledons	colour		form	colour	position of inflorescences	size
round roundish			white		yellow		long
			violett-red		green		short



What did Mendel discover?

- *Remember:* we knew that offspring resemble parents → “heredity”
 - So something must be passed on from parents to offspring, but what?
 - Mendel’s work uncovered the secrets of “genes”
 - definition for gene then was simple: a “unit of information” passed on to offspring
(we didn’t know DNA even existed, so we definitely didn’t know it made up genes or was involved with inheritance)





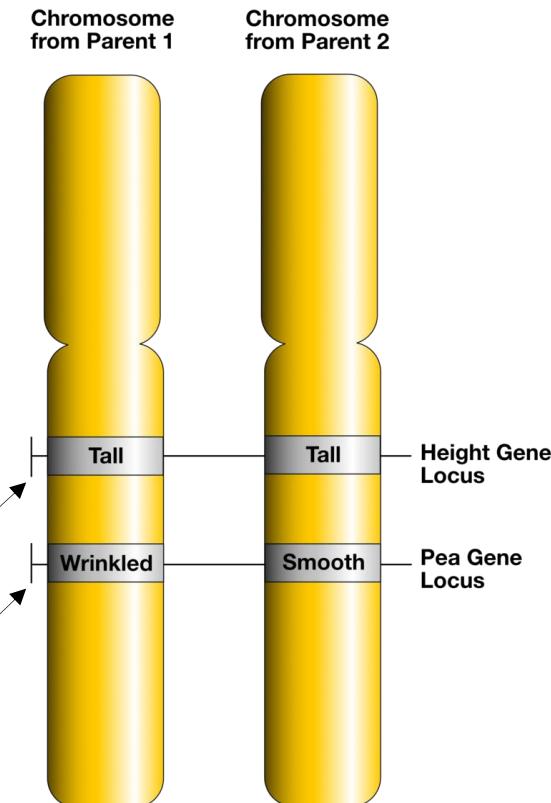
1. All organisms have 2 of every gene

- Through very careful notes on thousands of pea plant crosses, Mendel discovered that all organisms have **2 of every gene**

- 1 from each parent

*– we now know they are
on homologous
chromosomes*

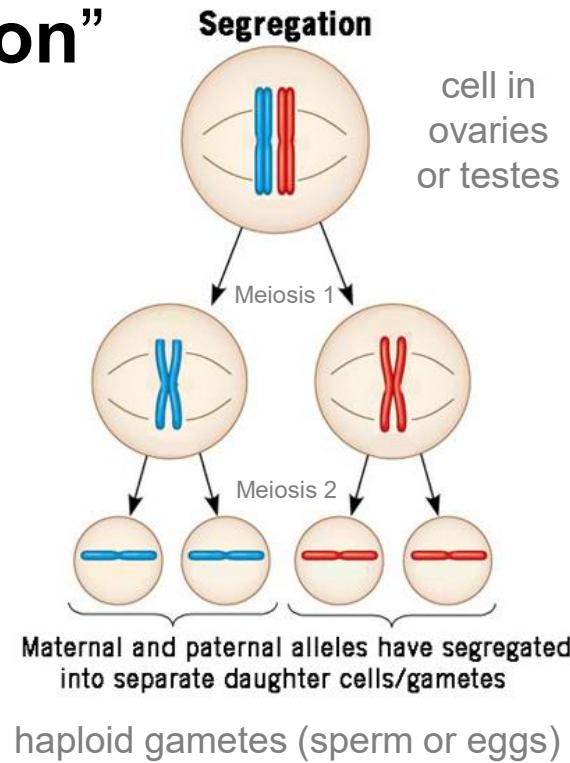
2 genes in a plant with the same allele to make it tall
2 genes in a plant with different alleles, one for wrinkled & one for smooth peas





2. Only 1 of an organism's 2 genes is passed on to offspring

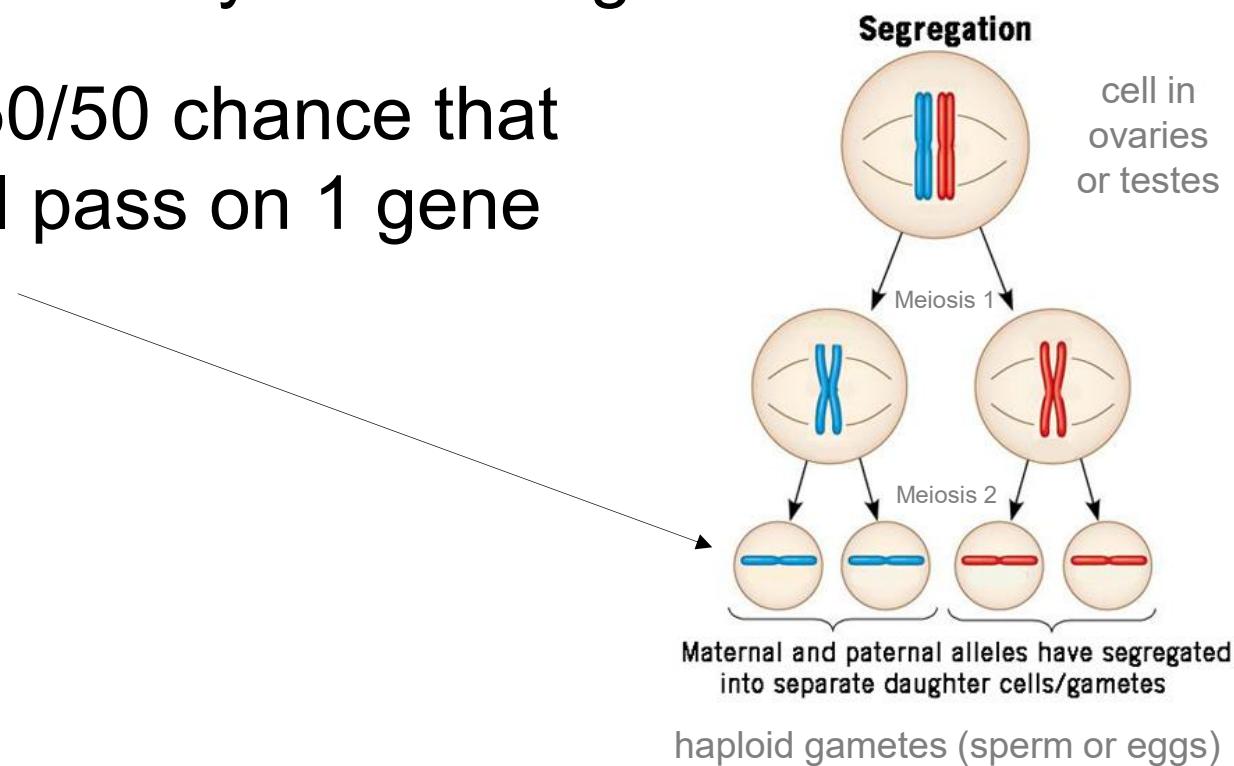
- Even though a parent has 2 of every gene, they only pass 1 on to their offspring
- Also called the “**Law of segregation**”
 - = each gene is separated during gamete formation, so that gametes only have 1 gene or the other
- *We now know this occurs because of meiosis*





3. The 1 gene that is passed on is random – 50/50 chance

- 50% of gametes carry 1 of the genes
- 50% of gametes carry the other gene
 - There is a 50/50 chance that a parent will pass on 1 gene or the other

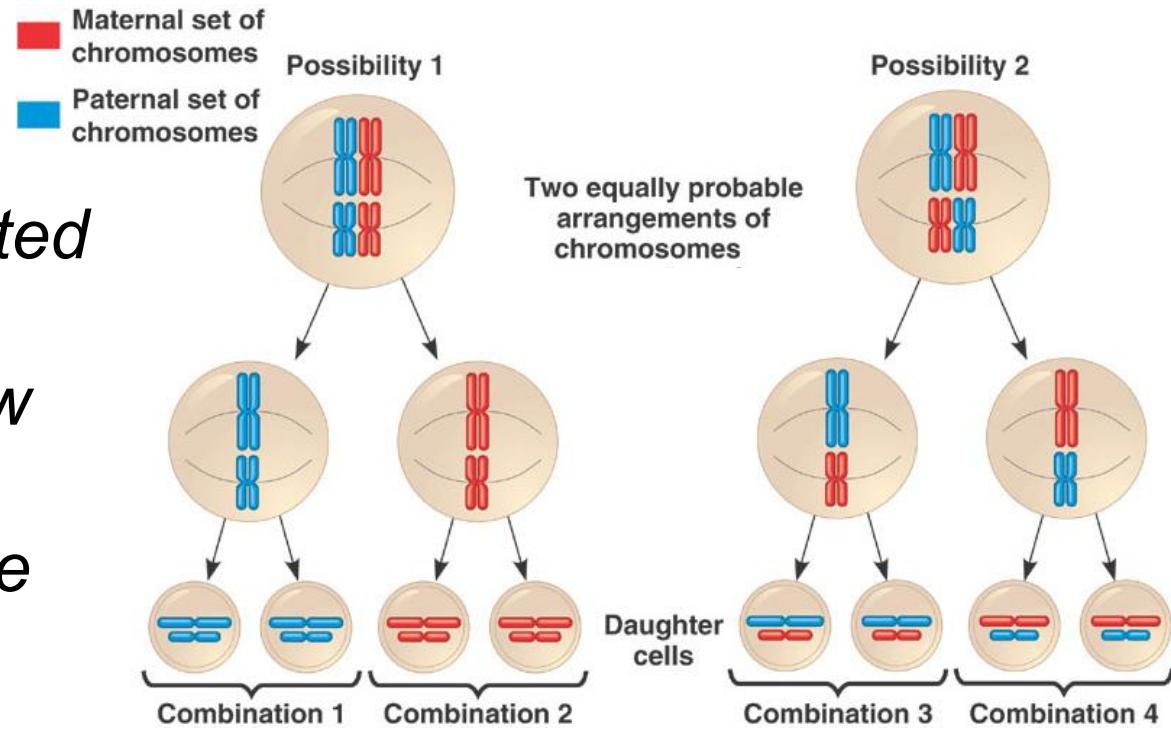




3. The 1 gene that is passed on is random – 50/50 chance (cont.)

- Also part of the “**law of independent assortment**”
 - Because it is independent of the other genes passed on

– *Genes are separated from each other & don't influence how other genes are separated – they're all 50/50*

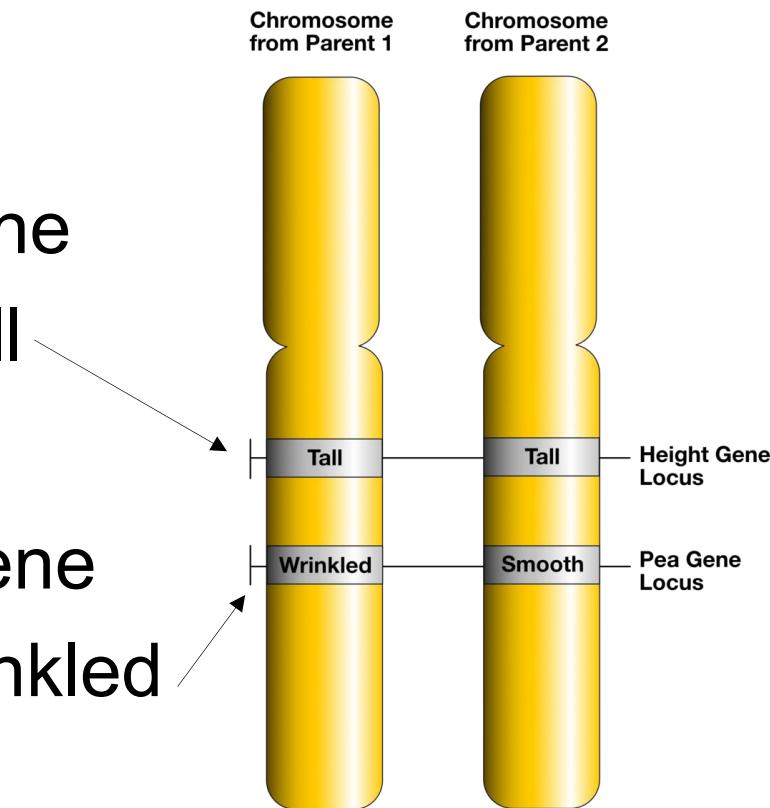




4. An organisms genes can be homozygous or heterozygous

– Since organisms have 2 copies of every gene, it is possible to have:

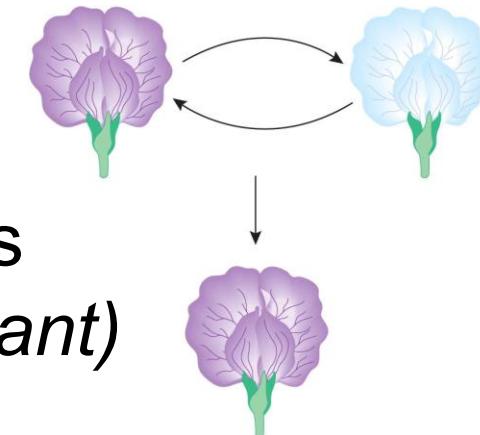
- 2 matching alleles =
homozygous for that gene
 - e.g. plant height for tall
- 2 different alleles =
heterozygous for that gene
 - e.g. pea shape for wrinkled AND smooth





5. Some alleles are dominant or recessive

- If an organism is *homozygous* for a gene, they will only show that 1 possibility
 - e.g. purple flower allele + purple flower allele = having purple flowers
- BUT: in a *heterozygous* case, **the dominant allele will mask the recessive allele**
 - e.g. purple flower allele + white flower allele = having purple flowers
(because the purple allele is dominant)





5. Some alleles are dominant or recessive (cont.)

– We usually denote dominant alleles with capital letters & recessive alleles with lowercase letters

- Purple allele = dominant = “P”
- White allele = recessive = “p”

– *Homozygous dominant = PP*



Genotype

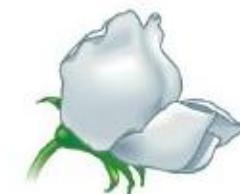
PP
(homozygous)

– *Heterozygous = Pp*



Pp
(heterozygous)

– *Homozygous recessive = pp*

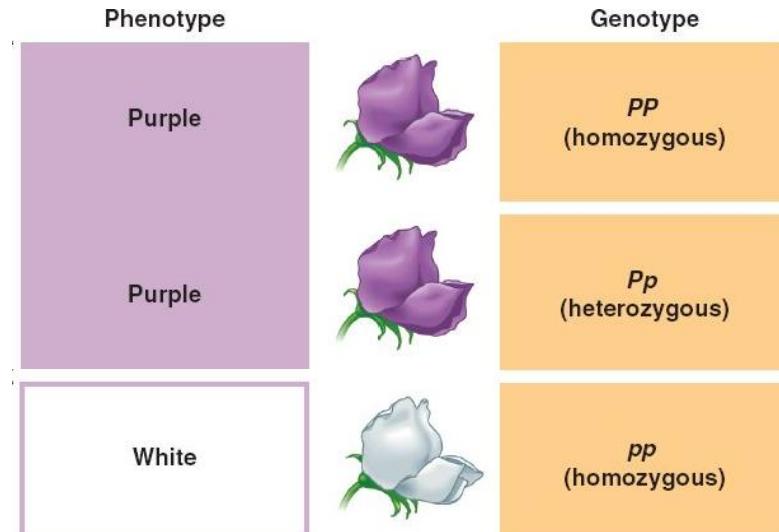


pp
(homozygous)



Since a dominant allele can mask a recessive allele, we now see that the genes we have inside our cells don't always match the traits we see

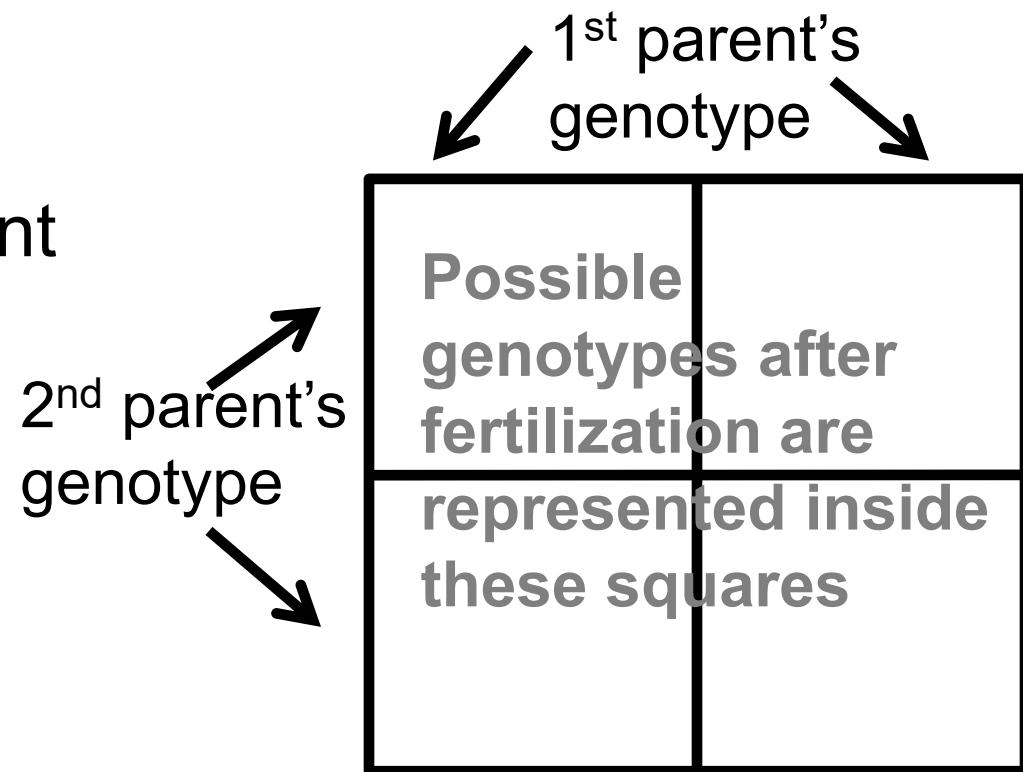
- The 2 alleles carried by an organism = the **genotype** (*what you have*)
 - e.g. *purple & white alleles, or “Pp”*
- The physical expression of the genotype = the **phenotype** (*what you show*)
 - e.g. *purple flowers*





Can we predict the chances of passing on certain genes to offspring?

- The **Punnett square** method predicts the probability of offspring genotypes & phenotypes using the genotype of each parent





Let's try it for the flower color trait:

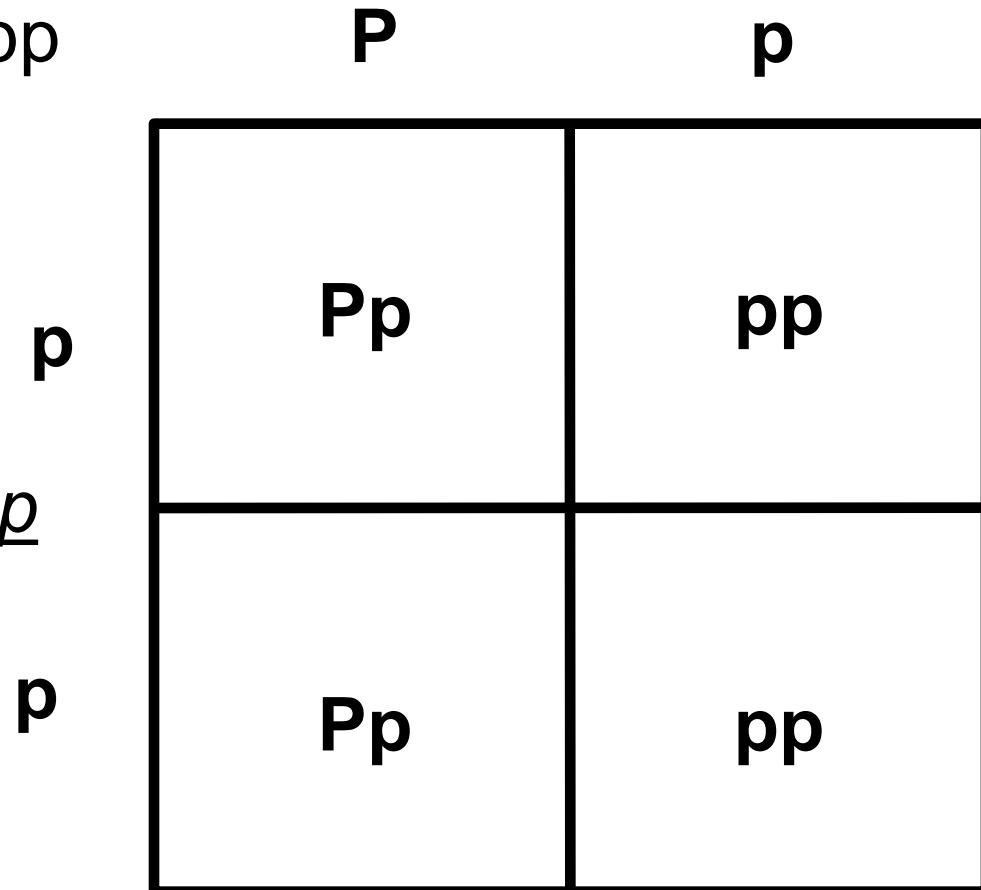
- 1 parent has genotype Pp
(has purple flowers)
- Other has genotype pp
(has white flowers)



One parent is pp



One parent is Pp



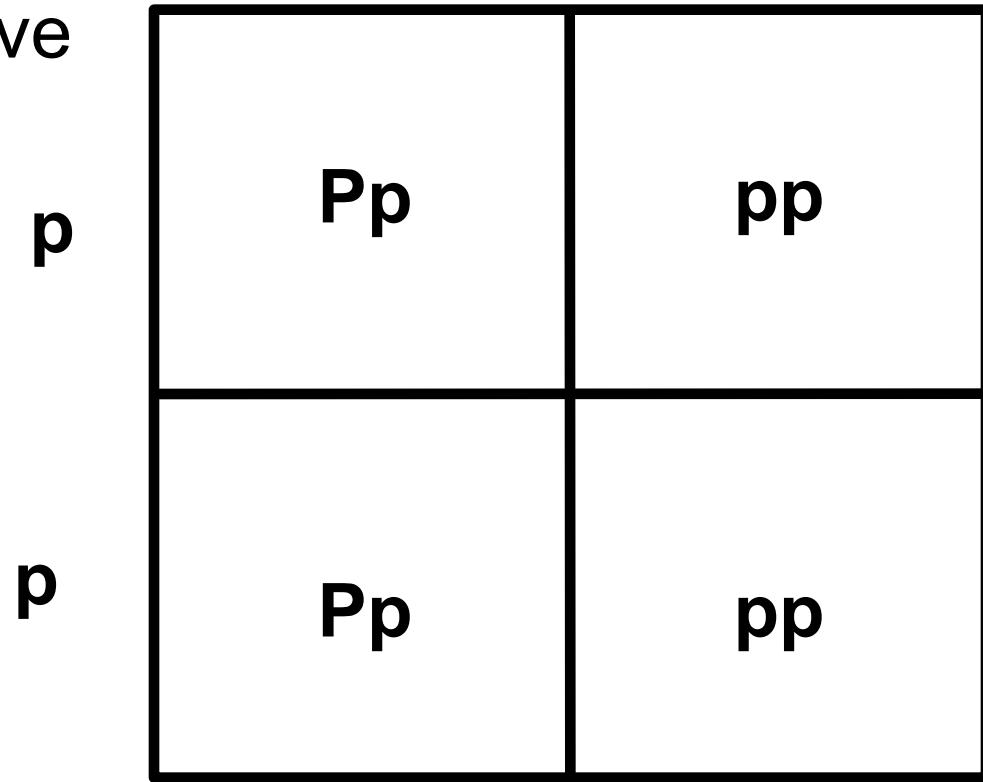


What is the % chance of each genotype?

- Homozygous dominant (PP) = 0%
- Heterozygous (Pp) = 50%
- Homozygous recessive (pp) = 50%



P p





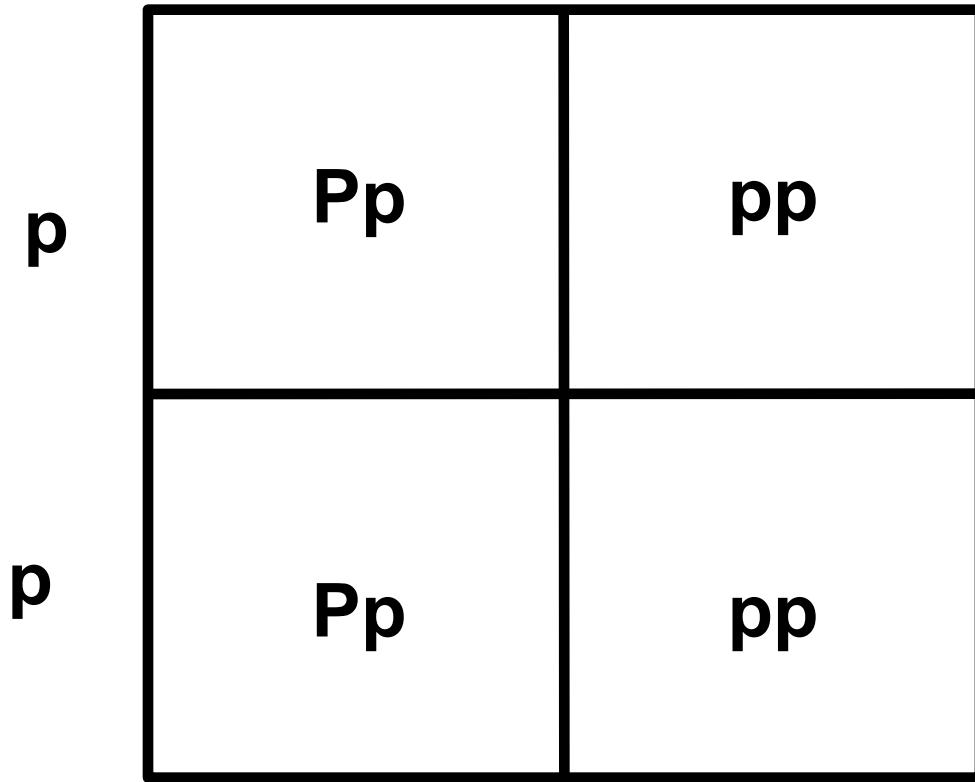
What is the % chance of each phenotype?

- Purple-flowered offspring = 50%
- White-flowered = 50%



P

p





Let's try it with another trait:

- The gene for fur length in cats has 2 alleles:
 - Short fur = dominant (F)
 - Long fur = recessive (f)
- 1 parent is homozygous dominant with short fur (FF) → 
- 1 parent is homozygous recessive with long fur (ff) → 
- What would the genotypes & phenotypes of their offspring be regarding fur?



What is the % chance of each genotype?

- Homozygous dominant =
- Heterozygous =
- Homozygous recessive =



One parent is FF



One parent is ff



What is the % chance of each phenotype?

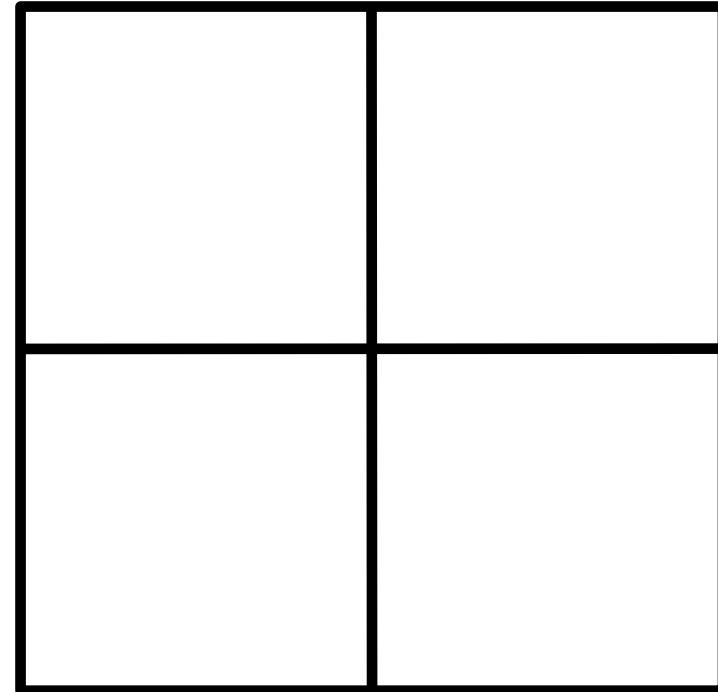
- Short fur =
- Long fur =



One parent is FF



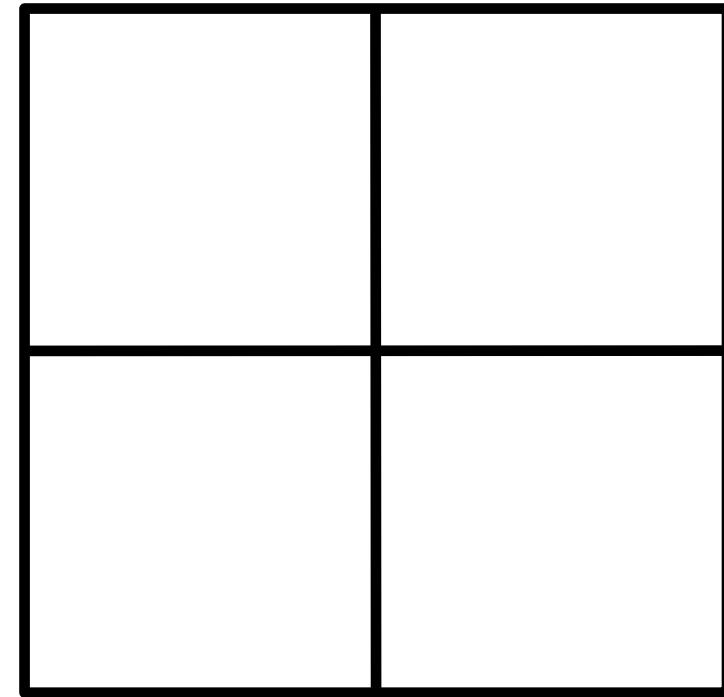
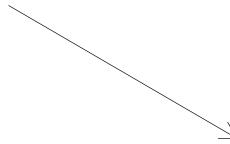
One parent is ff





NEW PRACTICE

- 1 parent is heterozygous with short fur
- The other parent is also heterozygous with short fur





What is the % chance of each genotype?

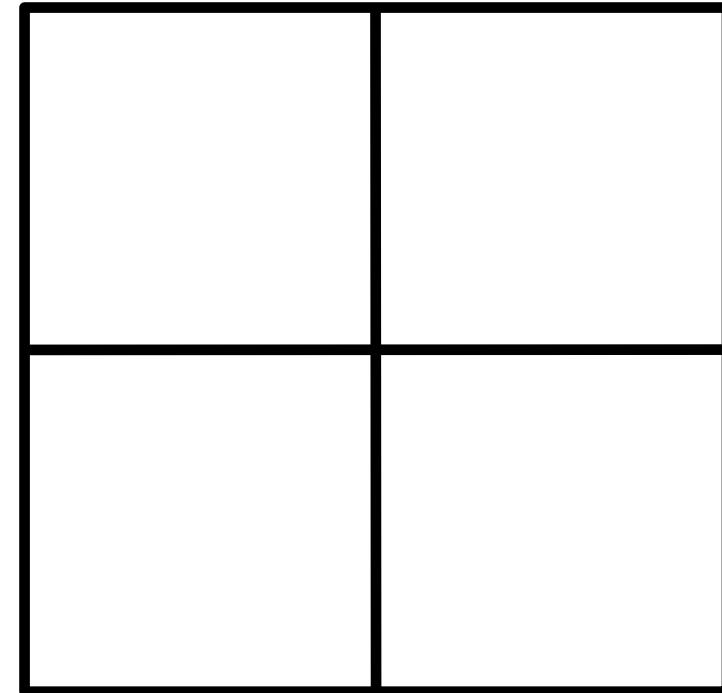
- Homozygous dominant =
- Heterozygous =
- Homozygous recessive =





What is the % chance of each phenotype?

- Short fur =
- Long fur =



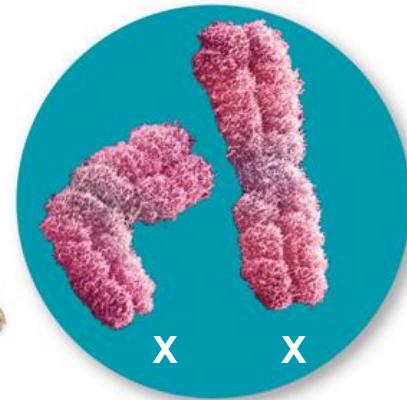
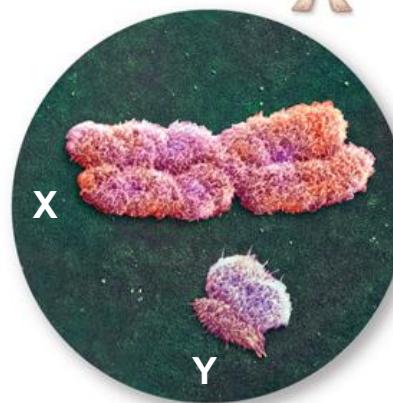


Sex-linked genes

- Remember from chapter 9:
sex is determined by sex chromosomes

– Also remember: not everyone fits into these 2 categories

– Also remember:
sex is not gender



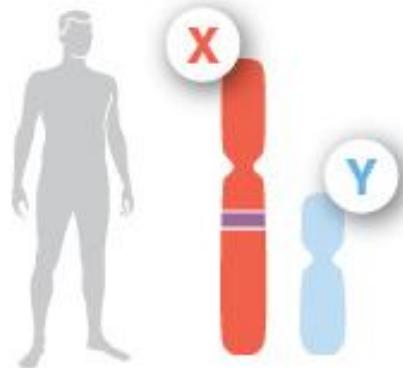
Female gametes		Male gametes	
X	X	X	XX female
		Y	XY male

Female (XX) : 50% chance
Male (XY) : 50% chance

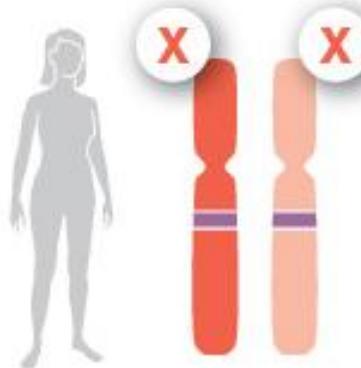


Sex-linked genes are found only on the X or on the Y sex chromosomes (not both)

- Some genes found only on the X chromosome are important to both sexes
 - e.g. genes for color vision, blood clotting, & certain proteins in muscles



Males will have only 1 of these important genes, inherited from his female parent

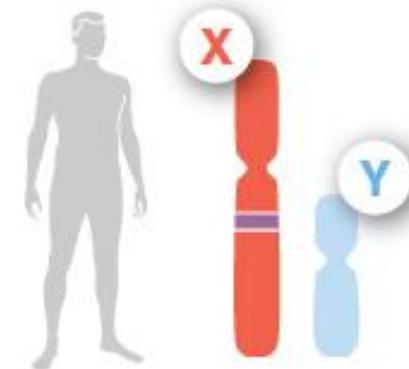
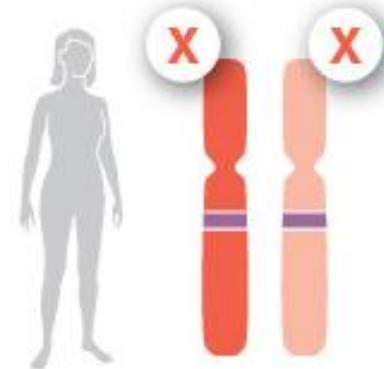


Females will have 2 of these important genes, 1 from each parent



Males only have one copy of the X chromosome

- Females (XX) can be homozygous or heterozygous for a characteristic
 - If heterozygous: the dominant allele shows up
- Males (XY) have only 1 copy of each gene - there is no 2nd copy to mask recessive genes
 - Fully express all the X-linked genes inherited from their female parent, whether the alleles are dominant or recessive

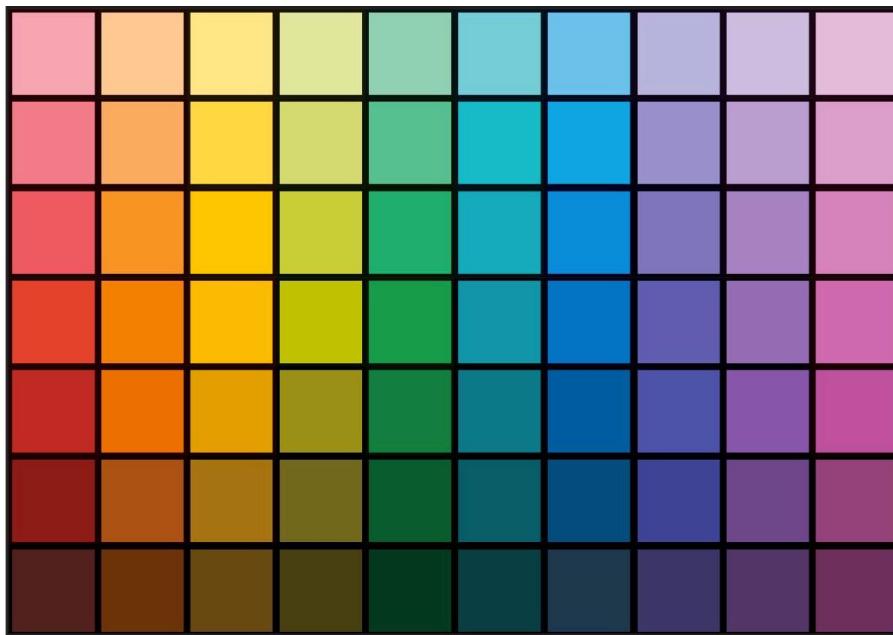




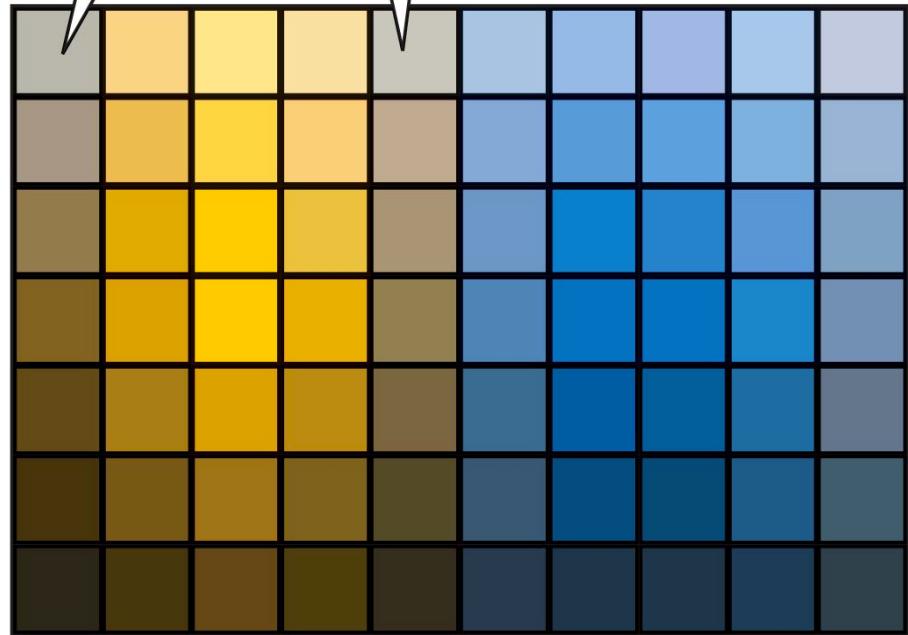
A gene on the X-chromosome codes for the protein in the eye that detects red & green = *sex-linked gene*

- Red-green color blindness in humans is caused by a recessive allele on the X chromosome that does not properly code for the protein

The individual cannot distinguish red from green



(a) Normal color vision



(b) Red-green color blindness



Inheriting sex-linked genes

- X_R = dominant allele = color vision
- X_r = recessive allele = R/G color blind
- **Females** can have [X_R - X_R], or [X_R - X_r], or [X_r - X_r]
 - Only the [X_r - X_r] combination = R/G color blind
 - *A female with R/G colorblindness inherited it from BOTH parents*
The male parent must also be R/G color-blind





But **males** have only 1 of every gene on the X chromosome

- If they inherit [X_R-Y], then they have normal vision
- If they inherit [X_r-Y], then they are R/G color blind
 - There is no option for a dominant allele (X_R) to mask a recessive one
 - This means males = 16 times more likely to be color blind than females
 - *A male with R/G colorblindness inherited it from his female parent ONLY – the male parent can have regular color vision*

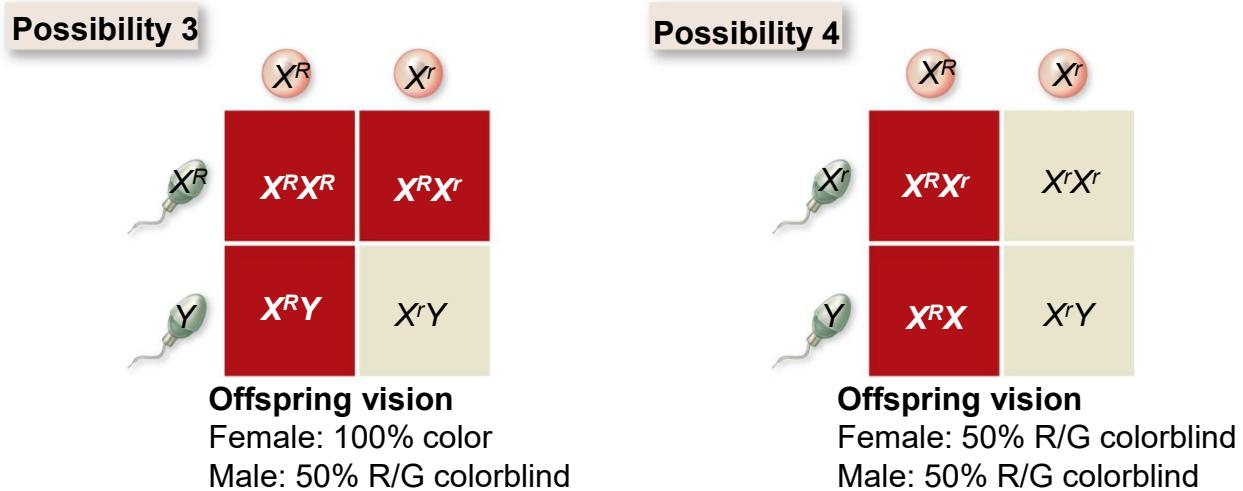
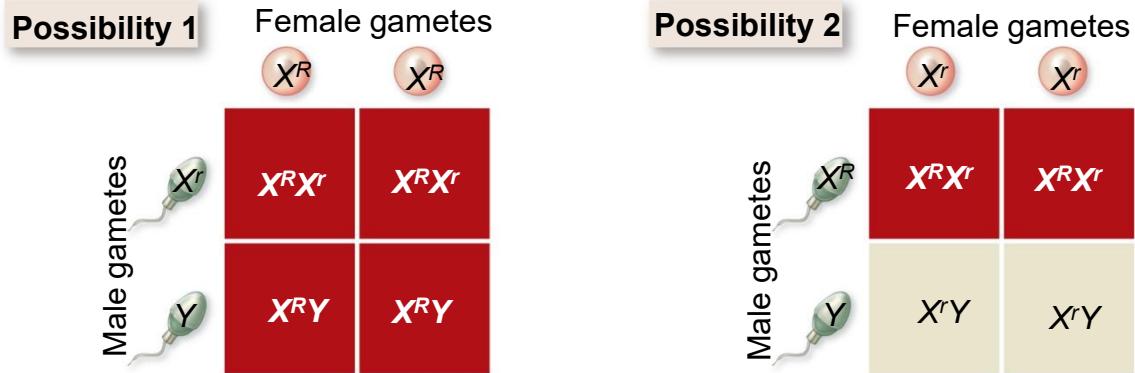




Possible parental combinations & resulting offspring

– Notice how much less likely it is to produce a female that is R/G colorblind

*There are
more possible
combinations -
here are only 4*





Other sex-linked genes (on X chromosome), which affect males 16x more, include:

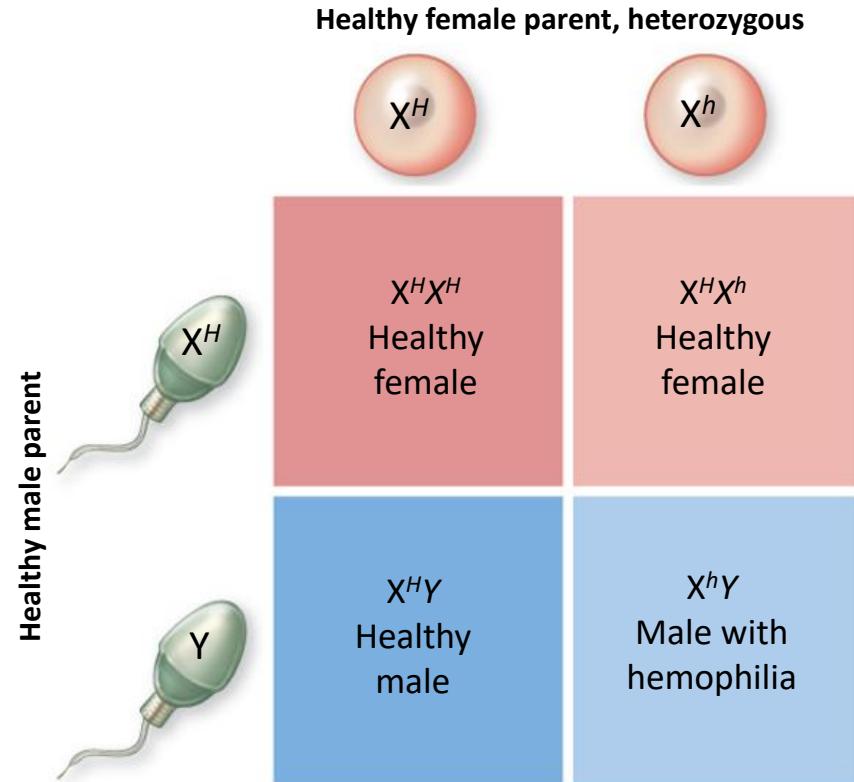
- **Muscular dystrophy** (progressive weakening of the muscles)
- **Pattern Baldness** (most common type of baldness)
 - “pattern” because it recedes from the hairline 1st, then becomes thinner, then is only left on back & sides of head





Other sex-linked genes (on X chromosome), which affect males 16x more, include:

- **Hemophilia** (in which the ability of the blood to clot is severely reduced)





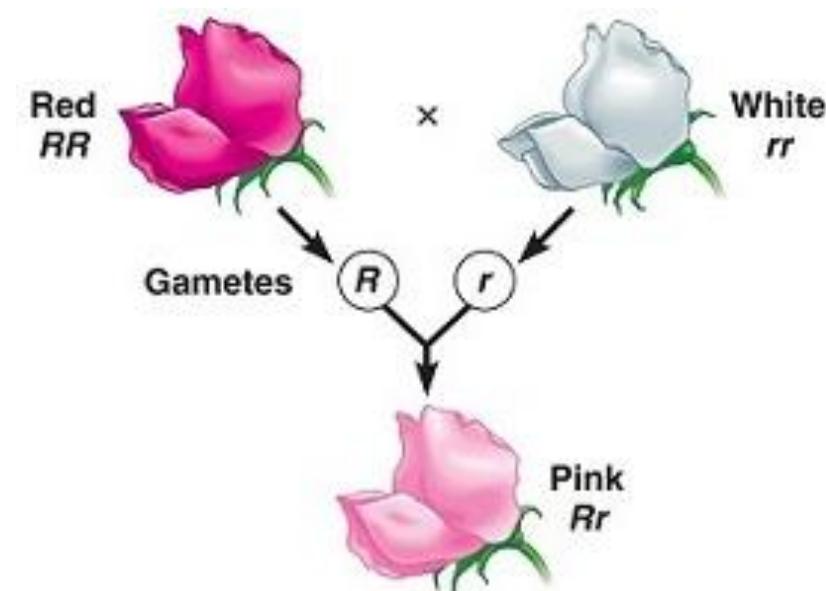
5 realms of non-Mendelian genetics

- Many traits do not follow the simple Mendelian rules of inheritance
 - There are 5 realms where genetics are a bit more complex: **non-Mendelian genetics**
 - 1. Incomplete dominance
 - 2. Multiple alleles
 - 3. Codominance
 - 4. Polygenic inheritance
 - 5. Pleiotropy



1. Some alleles are not dominant & recessive, but are *incompletely* dominant over others
 - When the heterozygous phenotype is an intermediate between the 2 homozygous phenotypes, the pattern of inheritance is called **incomplete dominance**

e.g. RR = red rose color
 WW = white rose color
 RW = pink rose color



*Incomplete dominance results in an *in-between* phenotype*



Another example of incomplete dominance:

e.g. CC = curly hair

SS = straight hair

CS = wavy hair

*Notice that we
don't use the same
letter capitalized &
lowercase,
because neither is
dominant over the
other*



*Incomplete dominance results
in an *in-between* phenotype*



2. A single gene may have **multiple alleles**

- The human blood types are an example of multiple alleles of a single gene
 - Human blood type genes have 3 possible alleles: A, B, and O
- Consequences of multiple alleles:
 - instead of having only 3 possible genotypes (e.g. RR, Rr, rr), there are many possible genotypes:

<ul style="list-style-type: none">• AA• AB• AO	<ul style="list-style-type: none">• BB• BO• OO
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3. Some alleles are **codominant**: both are expressed in entirety

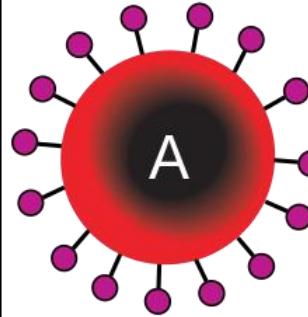
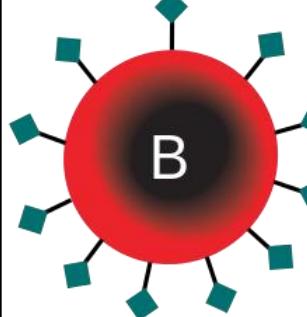
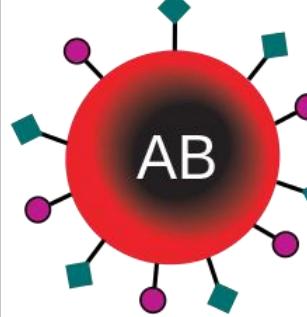
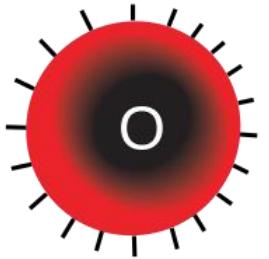
- Human blood types are another good example of this: A & B are codominant over O, which is recessive
 - leads to an extra phenotype: blood type AB

Genotype (2 alleles)	Phenotype (blood type)	Description
AA	A	homozygous
BB	B	homozygous
OO	O	homozygous
AO	A	heterozygous – A is dominant
BO	B	heterozygous – B is dominant
AB	AB	heterozygous – codominance



What does blood type mean?

- Blood type refers to the antigens a cell makes
 - Antigens = recognition proteins that sit on the outside of a cell & prevent the immune system from attacking them

Red blood cell type				
Antigens in Red Blood Cell	A antigen	B antigen	A and B antigens	None



We make antibodies against antigens we don't have

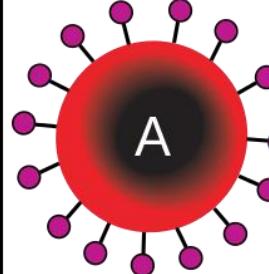
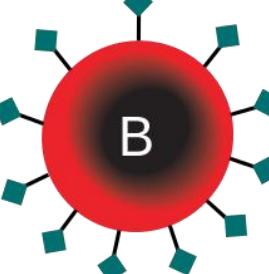
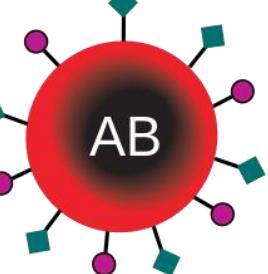
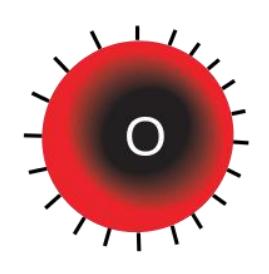
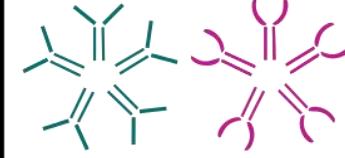
- AB has both antigens, so makes no antibodies
- O has no antigens, so makes both antibodies

Red blood cell type	A	B	AB	O
Antibodies in Plasma	Anti-B	Anti-A	None	Anti-A and Anti-B
Antigens in Red Blood Cell	A antigen	B antigen	A and B antigens	None



What does this mean for blood donation?

- Type O blood is not attacked by anyone: **universal donor**
- Type AB blood has no antibodies, so will not attack any kind of blood: **universal recipient**

Red blood cell type				
Antibodies in Plasma			None	 Anti-A and Anti-B



Phenotypes & genotypes in US population

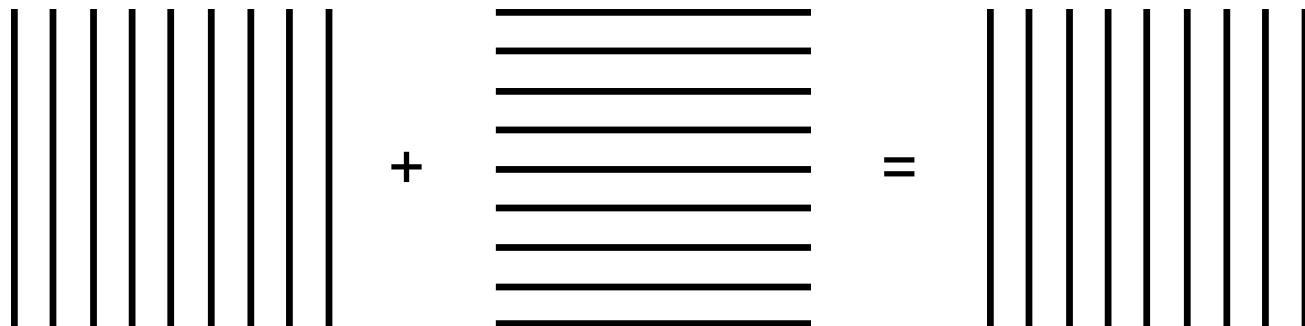
- Type O blood makes antibodies for both A & B, so they can ONLY get blood transfusions with other type O blood
 - Fortunately, almost half the US population has type O blood

“Blood type” Phenotype	Genotypes	Antibodies in the blood	% of US pop. w/blood type
Type A	AA and AO	Anti-B only	40%
Type B	BB and BO	Anti-A only	10%
Type AB	AB	None	4%
Type O	OO	Anti-A and Anti-B	46%

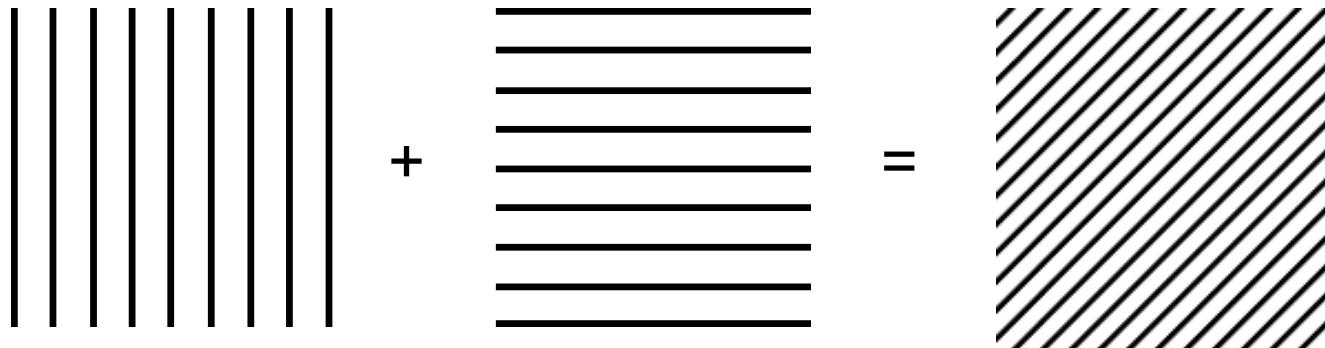


Understanding the 3 possibilities of dominance

Dominant & recessive:



Incomplete dominance:



Codominance:





4. Polygenic inheritance “polygenic” = many genes

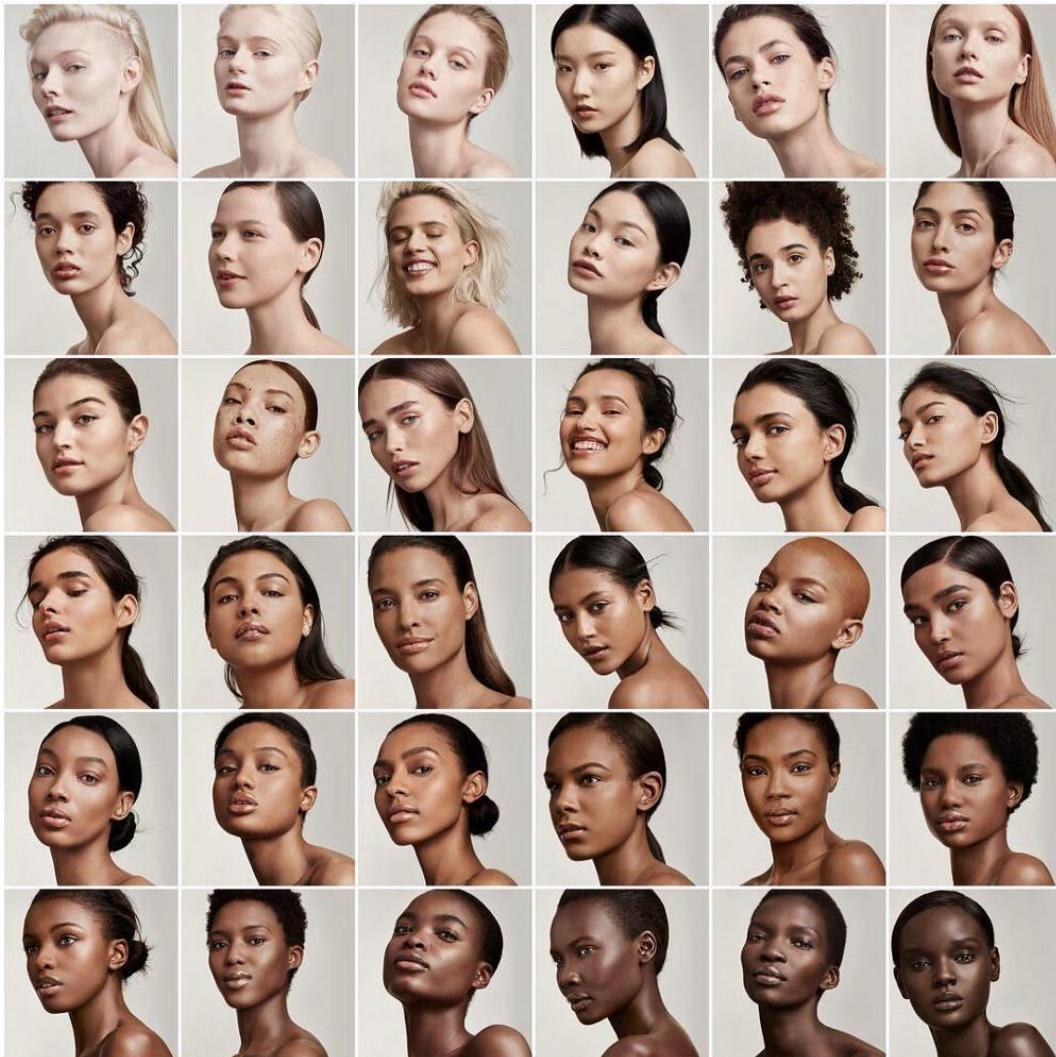
- Traits based on 1 gene have specific phenotypes
 - e.g. short fur or long fur
 - e.g. blood type A, B, AB, or O
- But some traits don’t have specific phenotypes – they have a range
 - e.g. height doesn’t come only in 5’5”, 5’10”, & 6’2”
 - it comes in a range from 4’ all the way up to 7’ or more





Phenotypes that have a range (instead of specific phenotypes) are caused by multiple genes: **Polygenic inheritance**

- Human examples
 - = height, skin color, eye color, & body build
 - Skin color is due to at least 3 different genes
 - many genes, one trait*



5. Sometimes 1 gene doesn't only have 1 trait – sometimes 1 gene affects many traits = **pleiotropy**

– e.g. in chickens, a gene called “frizzle” affects:

- Feather pattern
- Metabolic rate
- Body temperature
- Digestive ability

– *one gene, many traits*
• *opposite of polygenic*





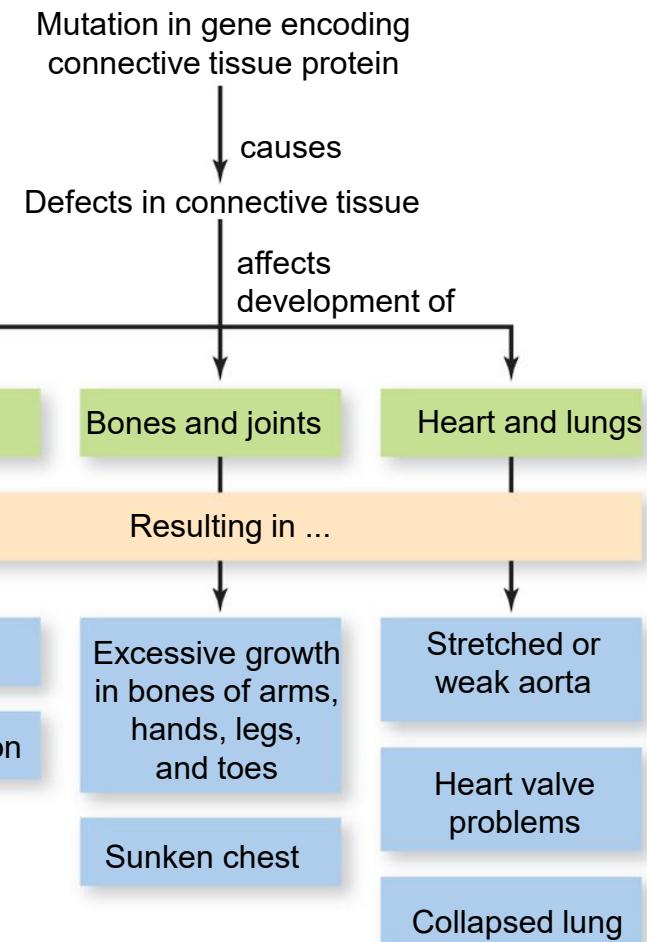
A human example of pleiotropy = Marfan syndrome

- A mutated gene on chromosome 15 causes an error in coding for a protein important for connective tissue

Bradford Cox of the band Deerhunter has Marfan syndrome

Abraham Lincoln is suspected to have had it too

One gene, affecting many traits





Genetic anomalies

- **Genetic anomalies** are often referred to as “genetic disorders” even though in some cases they are neutral or even advantageous
 - Anomalies that are inherited can be recessive or dominant – changes the risk for offspring
 - e.g. Huntington’s disease & an allele increasing the risk of breast cancer (*BRCA1*) are **dominant genetic anomalies**
 - people that are heterozygous or homozygous dominant will be affected
 - e.g. albinism is a **recessive genetic anomaly**
 - only homozygous recessive will be affected



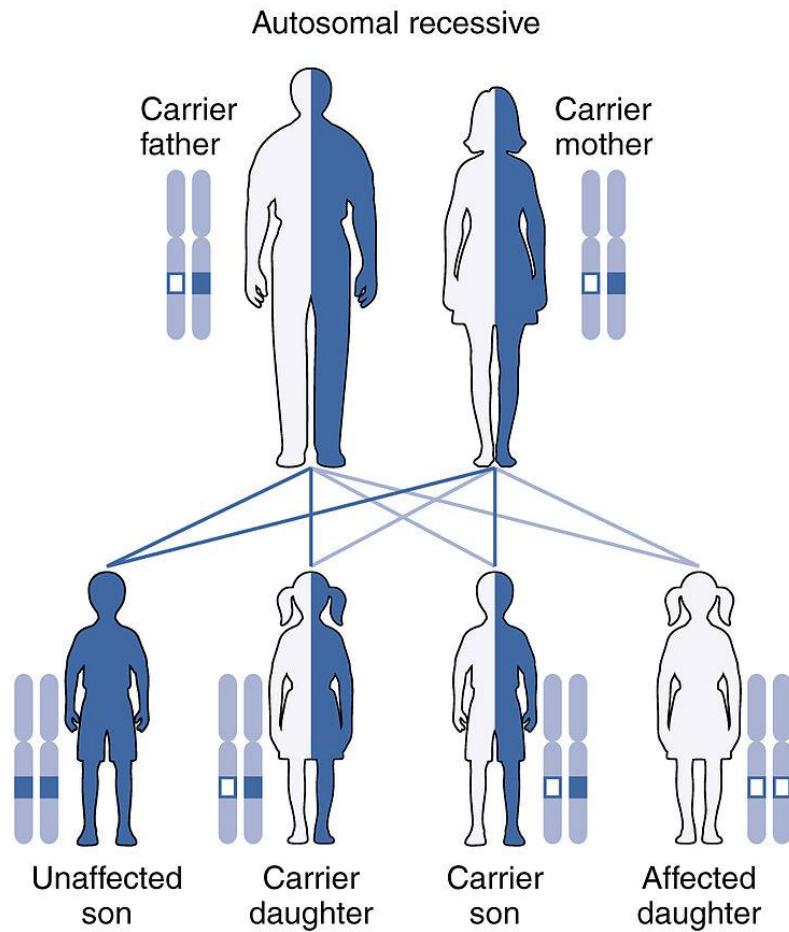
Recessive genetic anomalies only show up in the phenotype if the person is homozygous recessive

- e.g. albinism = mutation that causes the pigment protein melanin to be nonfunctional – leading to little or no pigment in the eyes, hair, & skin
- Both parents can be regularly pigmented but be *carriers*
 - **Carrier** = heterozygous for a regular & a recessive allele, but unaffected/not shown in their phenotype – can pass it to offspring though





Another example of a recessive genetic anomaly = **cystic fibrosis**: a life-threatening disorder that damages the lungs & digestive system



2 healthy parents
that are **carriers**
can have offspring
with cystic fibrosis

		Mother	
		C	c
Father	C	CC	Cc
	c	Cc	cc

Probabilities:
75% cystic fibrosis
not expressed
25% cystic fibrosis



The environment affects traits too

- You may have noticed before that identical twins aren't quite identical
 - This is because some traits are affected by the environment
 - Obvious traits affected by environment code for things like personality, intelligence, or body shape & weight





Other traits are affected by environment too:

- e.g. people who are pregnant sometimes notice changes in their **hair texture** – this is because the environment (in this case, circulating hormones) affects gene expression of hair proteins
- e.g. some animals have “heat-sensitive **hair color**”
 - the dark-pigment proteins are made from the genes but only activated under cool temperatures, like those found in the extremities (ears, tail, & paws)

