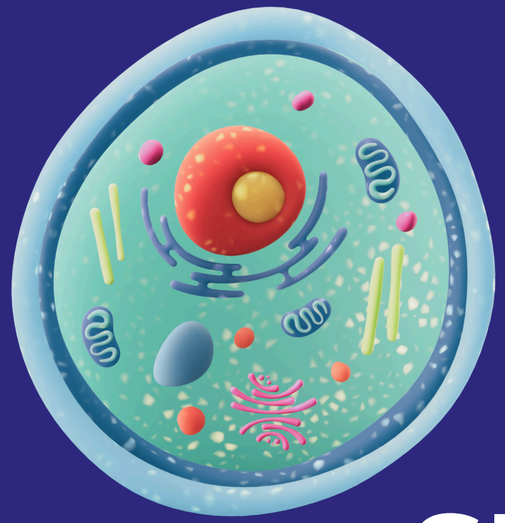


SICK CELLS: ORGANELLES & DISEASE

PROMISE

SANFORD RESEARCH

THINK, PAIR, SHARE

Think about the following questions, then **pair** up with a partner before we **share** as a class.

1) What do you already know about diseases?

- Do you know examples of diseases?
- How do diseases spread or develop?
- How are diseases diagnosed and treated?
- Do all diseases have the same symptoms?

2) What is an organelle?

- What are some functions of cells?
- Can cells have errors or mistakes?
- How do organelles help the cell work?

GUIDED NOTES

Fill in the blanks on your notes sheet with the information that is underlined on the slides.

Name: _____

SANFORD RESEARCH PROMISE

NOTES: DISEASES & ORGANELLES

Disease: an _____ condition that prevents the body from working correctly

Diseases can be:

- _____ (long-term) or _____ (short-term)
- Mild or severe
- Treatable or untreatable
- _____ or _____

What are some ways we get diseases?

- _____
- _____ - contact with something not sterile or clean
- _____ - animals/organisms that carry the disease
- _____
- _____

Cell Theory

1. _____
2. _____
3. _____

Body ← Organ Systems ← Organs ← Tissues ← Cells

WHAT IS A DISEASE?

Disease: an abnormal condition that prevents the body from working correctly

Examples of diseases:

- Influenza
- Malaria
- Cancer
- Tuberculosis
- Sickle cell disease

There are *many* types of diseases!

WHAT IS A DISEASE?

Diseases can be:

- Chronic (long-term) or acute (short-term)
- Mild or severe
- Treatable or untreatable
- Infectious or non-infectious

WHAT IS A DISEASE?

What are some ways we get diseases?

- Air
- Contamination
- Vectors
- Environment
- Genetics

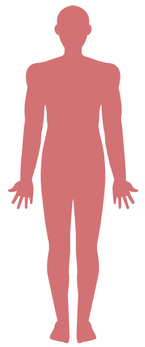
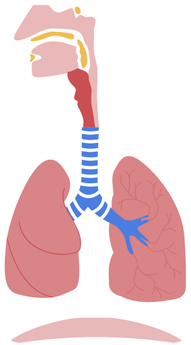
REVIEW: CELL THEORY

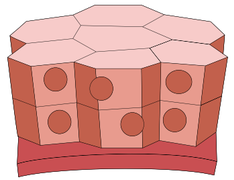
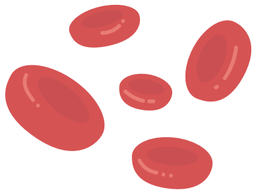
1) All living things are made of cells.

2) The cell is the smallest unit of life.

3) All cells come from other cells.

Body ← Organ Systems ← Organs ← Tissues ← Cells

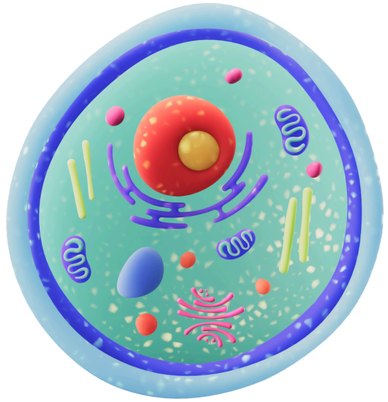



TYPES OF ORGANISMS

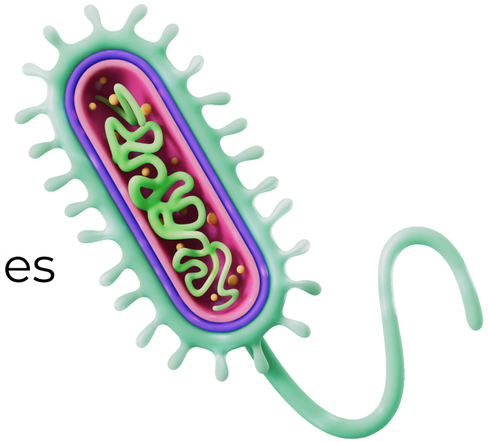
Eukaryotes

- Complex organisms
- Cells have organelles
- Includes plants, animals, fungi, and protists



Prokaryotes

- Bacteria and archaea
- No membrane-bound organelles



ORGANELLES: WHAT MAKES UP A CELL?

Organelle: small structure inside the cell that performs a job for the cell

What are some things the cell might need to do to stay healthy?

- Repair damage
- Make energy
- Move nutrients
- Get rid of waste

ORGANELLES: WHAT MAKES UP A CELL?

Cell membrane: protective layer around the cell

Nucleus: holds DNA (genetic information)

Mitochondria: produces ATP (energy) for the cell

Ribosome: makes proteins

Endoplasmic reticulum (ER)

- Smooth ER: makes lipids (fats)
- Rough ER: makes proteins

Golgi: checks proteins for errors and packages them up to be moved

ORGANELLES: WHAT MAKES UP A CELL?

Cytoplasm: fluid inside the cell where the organelles are suspended

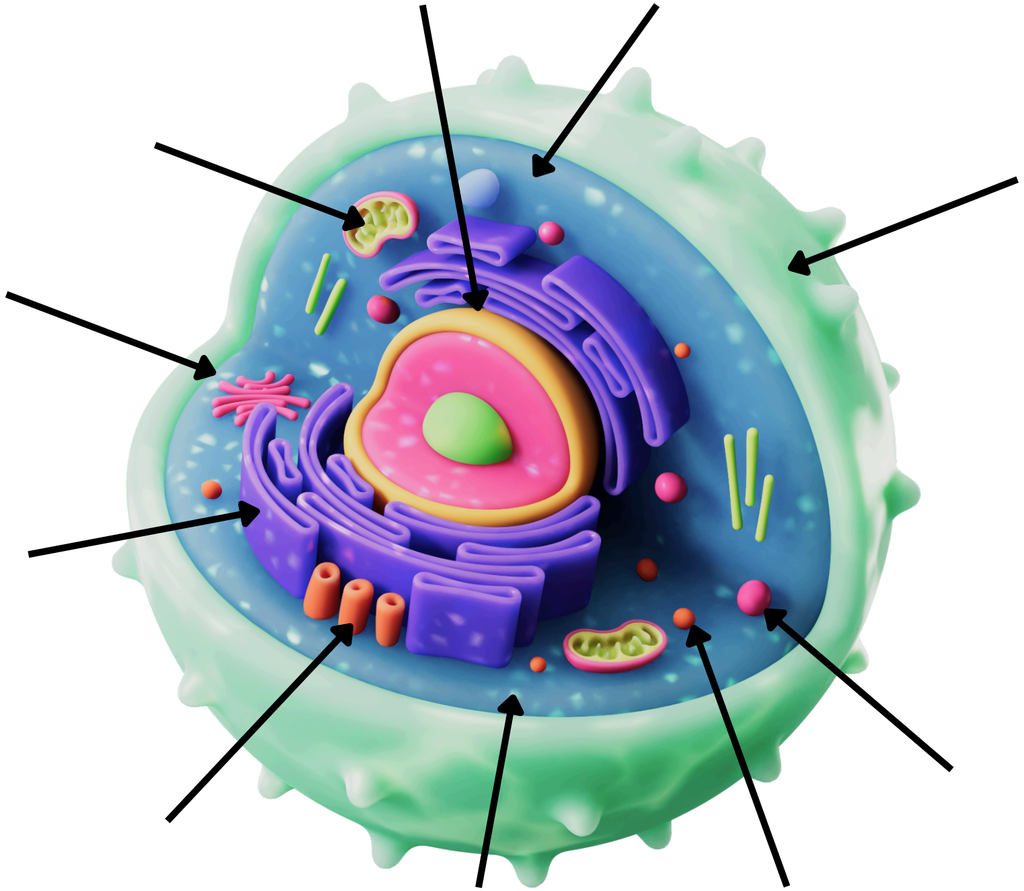
Vesicle: “bubble-like” structures that transport materials around the cell

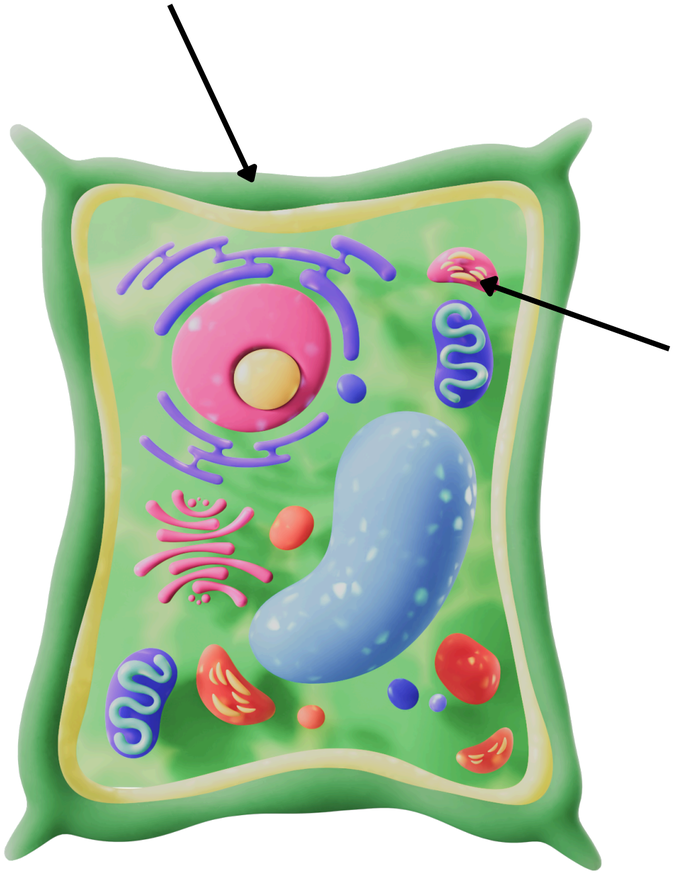
Lysosome: degrade cell waste and harmful material

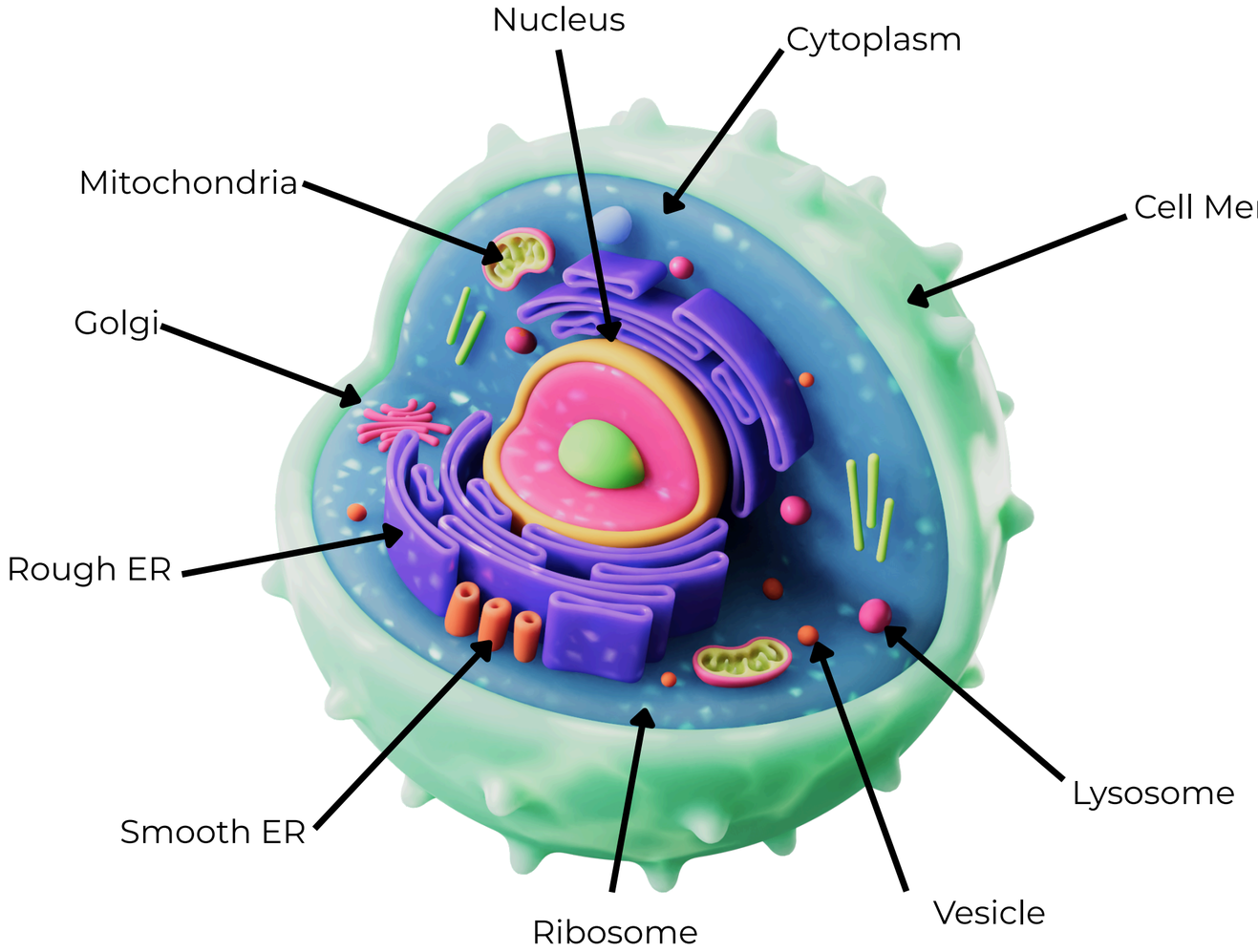
Cilia: hair-like structure that controls movement of cells and things around cells, like in the respiratory tract

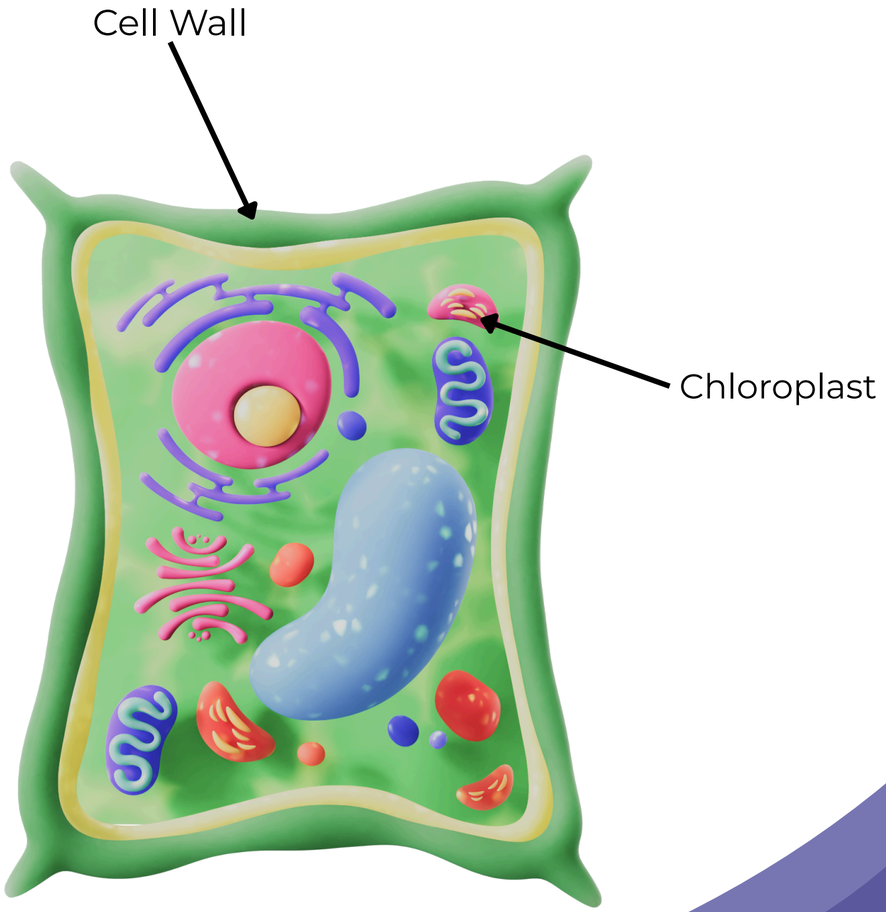
Chloroplast: converts sunlight into ATP for plants

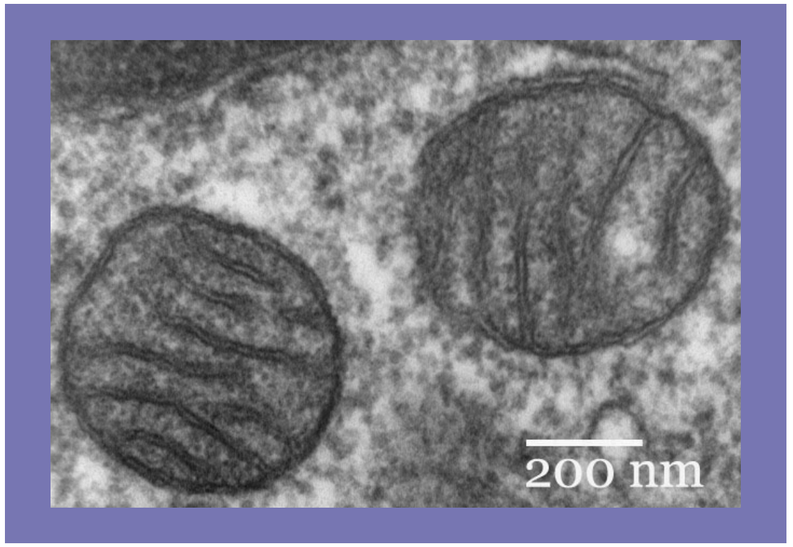
Cell wall: rigid outside layer that gives plants support and shape



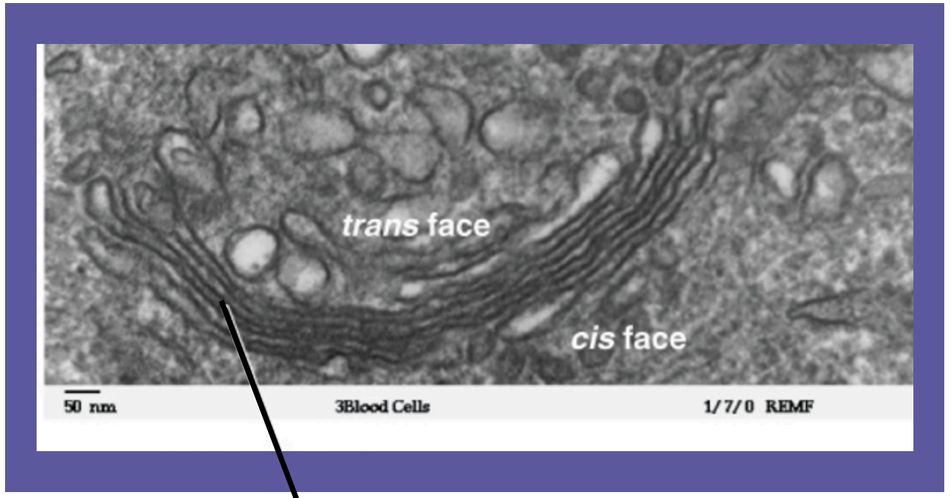



Nucleus
Cytoplasm
Mitochondria
Cell Membrane
Golgi
Rough ER
Smooth ER
Ribosome
Vesicle
Lysosome

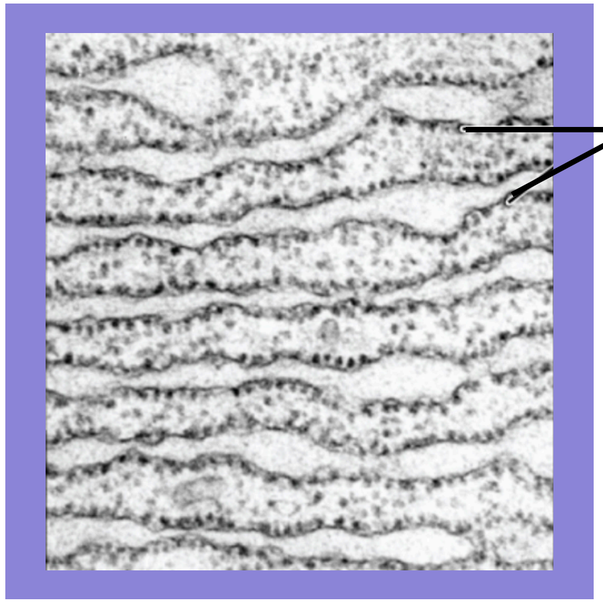

Cell Wall
Chloroplast



Mitochondria

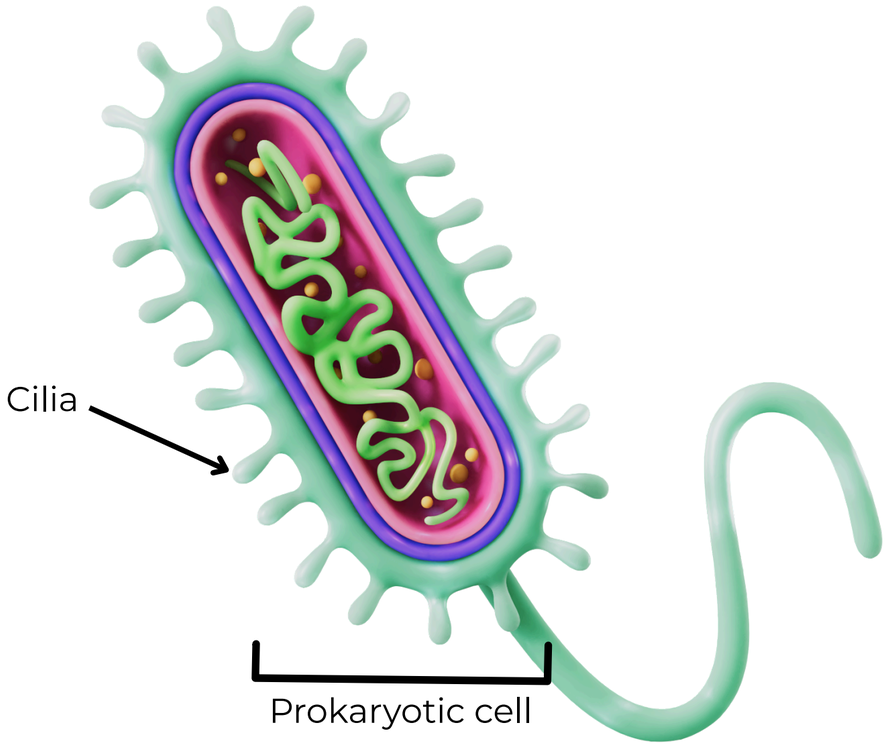


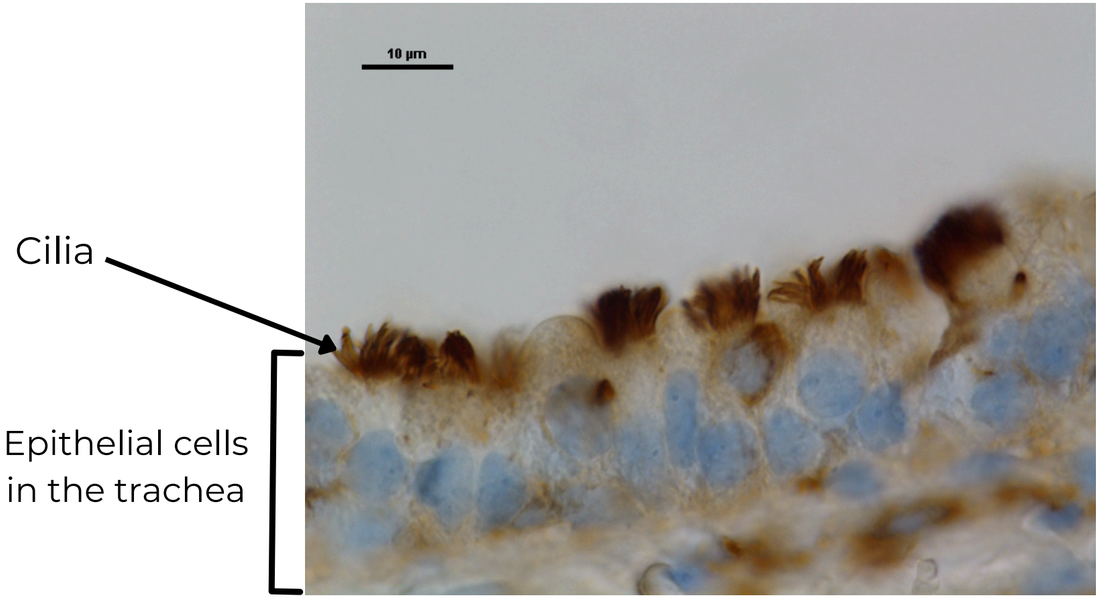
Golgi



Ribosomes

Rough ER



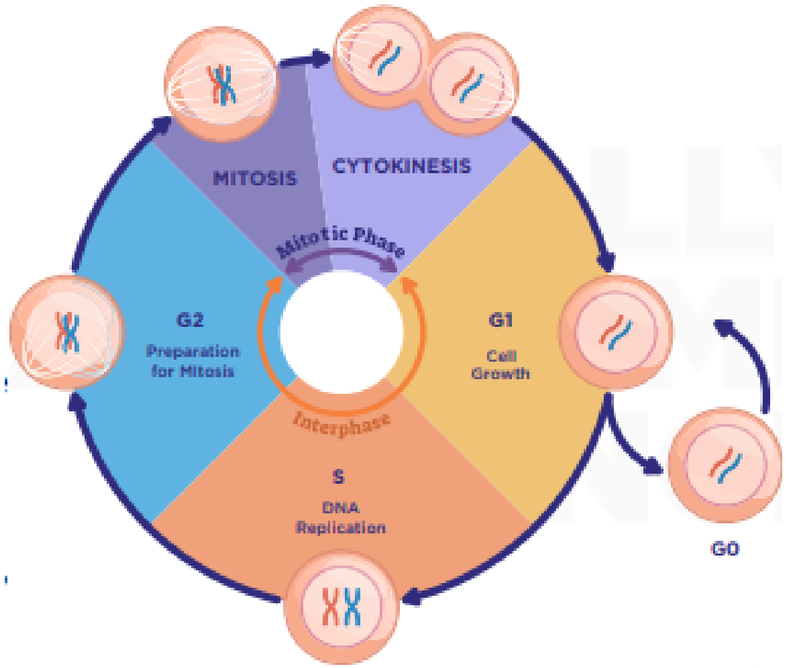


From Dr. Lance Lee's Lab, Sanford Research

WHEN CELLS MALFUNCTION

If the an organelle or process inside the cell is disrupted, this could lead to...

- Cell damage
- Uncontrolled division - cell cycle error
- Cell death



These outcomes can cause many types of diseases!

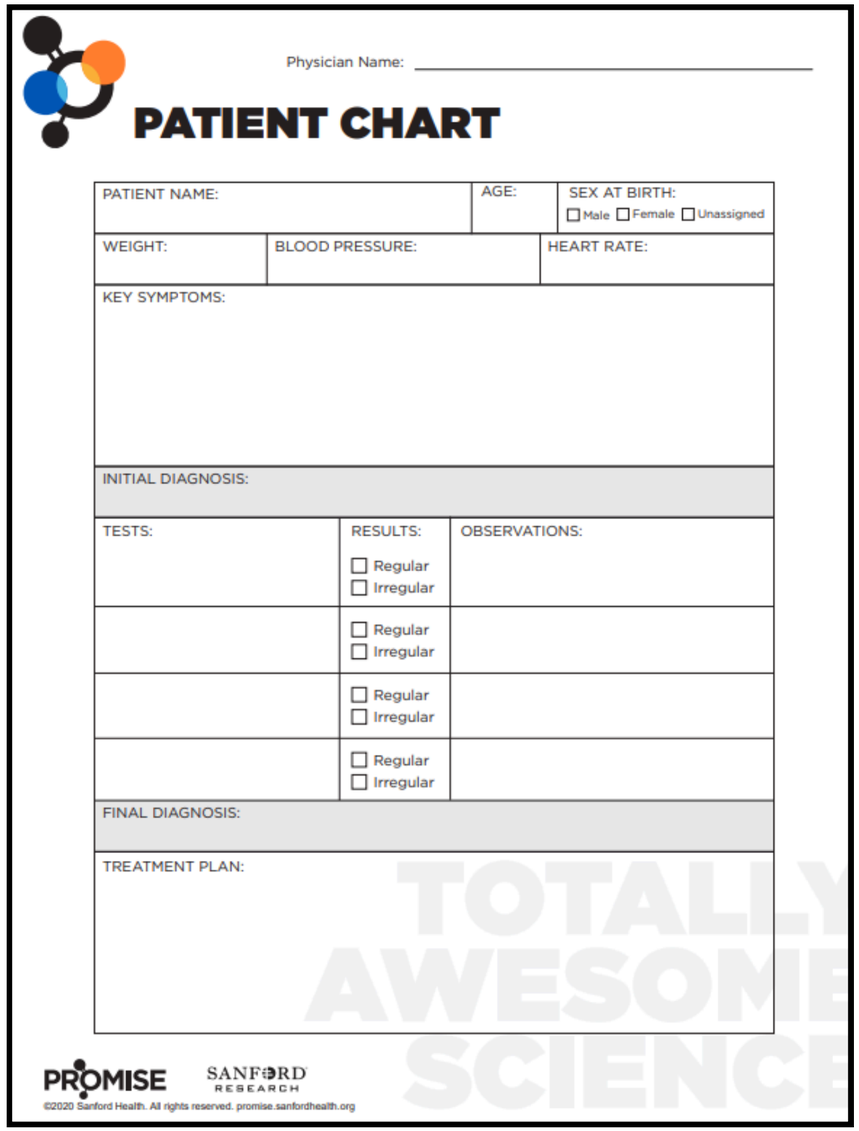
CASE STUDY: SICK CELLS

- A patient comes to the hospital with a problem, and you are the doctor that is going to help diagnose them.

- After hearing why they were admitted, you will figure out their medical history, which tests they may need, and come to a conclusion about what the patient's problem is.

- Lastly, you will identify the malfunctioning organelle that is causing the problem to help the patient and their family better understand why they are sick.

YOUR PATIENT CHART



Physician Name: _____

PATIENT CHART

PATIENT NAME:		AGE:	SEX AT BIRTH: ☐ Male ☐ Female ☐ Unassigned
WEIGHT:	BLOOD PRESSURE:		HEART RATE:
KEY SYMPTOMS:			
INITIAL DIAGNOSIS:			
TESTS:	RESULTS: ☐ Regular ☐ Irregular		OBSERVATIONS:
	☐ Regular ☐ Irregular		
	☐ Regular ☐ Irregular		
	☐ Regular ☐ Irregular		
FINAL DIAGNOSIS:			
TREATMENT PLAN:			

PROMISE SANFORD RESEARCH

CASE #1

PATIENT INFORMATION & REASON FOR VISIT

- Fred and Jill Smith have a five year old son named John
- John was brought into the hospital because he keeps falling a lot.
- John was also having problems forming sentences and responses when he talks to people, but his parents thought this was related to his ADHD diagnosis.
- Vital signs were taken when John came to the hospital
  - His heart rate was 80 beats per minute
  - His temperature was 98.7 ℉
  - His blood pressure was 108/70
  - His weight is 35 pounds

Record this patient information and his vitals in your Patient Chart.

KNOWN MEDICAL HISTORY

- John was recently diagnosed with ADHD after his teacher noticed he had difficulty paying attention when reading and writing.
- Medication did not improve John’s attention, so he no longer takes it.
- There is no family history of seizures or other serious health complications in his close family that would explain his symptoms.

Make an initial diagnosis for John’s illness in your Patient Chart.

TESTING

- An EEG of John’s brain activity does not show signs of a seizure
- An MRI shows that John’s brain is smaller than the normal size for his age
- No notable masses are visible in the brain
- An initial blood test returned with normal results
- The doctors took a biopsy of John’s skin cells to look at under the microscope. They found abnormal clumped structures of lipids that built up in the cell.

Fill out the Tests section in the Patient Chart. Make any adjustments needed to your initial diagnosis.

DIAGNOSIS

- John was diagnosed with Batten Disease, a neurodegenerative disorder
- Early signs of this disease often include clumsiness, problems with attention, and language production issues.
- The clumped lipid masses that the doctors saw in his cells are also a sign of this disease.
- Genetic analysis could determine which form of the disease John has.
- It is predicted that John’s condition will progress.

Fill in the Final Diagnosis section. Then, list treatments that help some of John’s symptoms. Which predictions were closest to the actual disease?

TREATMENT PLAN

- John has been referred to a neurologist for future care and planning with disease management

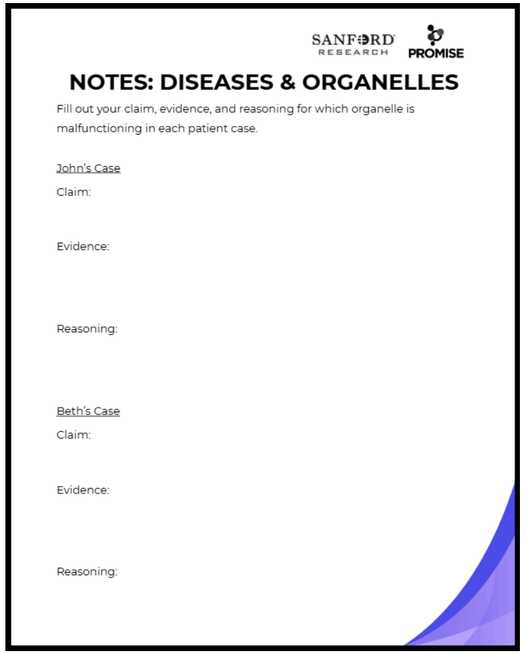
- He was also referred to physical therapy to work on balance and motor control

Update your Treatment section with this information.

WHICH ORGANELLE IS AFFECTED?

On the third page of your Guided Notes sheet:

1. Make a **claim** about which organelle is malfunctioning.
2. List the **evidence** from your Patient Chart that supports your claim.
3. Explain the **reason** why your evidence supports your claim.



SANFORD RESEARCH PROMISE

NOTES: DISEASES & ORGANELLES

Fill out your claim, evidence, and reasoning for which organelle is malfunctioning in each patient case.

John's Case

Claim:

Evidence:

Reasoning:

Beth's Case

Claim:

Evidence:

Reasoning:

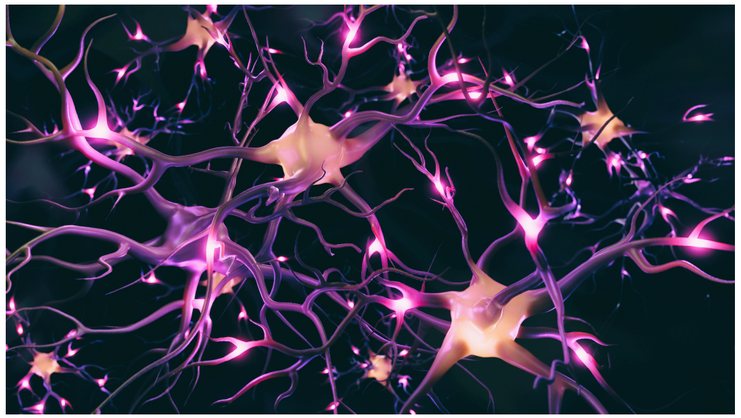
GENETIC RESULTS

- John has a mutation in the ***CLN6*** gene, which is responsible for making lysosome enzymes needed to destroy cell waste.

- With the mutation, these enzymes cannot be made, so waste like lipids will build up in cells.

- The waste could interfere with cell function, so the cells begin to die if they can’t get rid of it

- Cells like neurons in the brain cannot divide and replicate, so cell death causes neurological and motor issues.



GENETIC RESULTS

- Batten disease is a recessive disorder, meaning John has two mutated copies of the gene. He inherited one copy from each of his parents.

- Since his parents are not affected by the disease, they must be carriers of the mutated copy.

CASE #2

PATIENT INFORMATION & REASON FOR VISIT

- Sally and Paul have a five year old daughter named Beth
- Beth was brought to the hospital for respiratory distress
- Beth usually experiences chronic coughing and congestion, which has been diagnosed as symptoms of asthma
- Her parents note that her inhaler and medication has not helped her respiratory distress
- Vital signs were taken when she was admitted to the hospital:
  - Her heart rate was 85 beats per minute
  - Her temperature was 98.6 ℉
  - Her blood pressure was 100/65 mmHg
  - Her weight was 40 pounds

Record this patient information and her vitals in your Patient Chart.

KNOWN MEDICAL HISTORY

- When she was three, Beth was diagnosed with asthma due to her persistent wet cough and congestion
- She has a history of recurring ear and respiratory tract infections
- Her close family does not have a history of respiratory issues, and no one in her home smokes

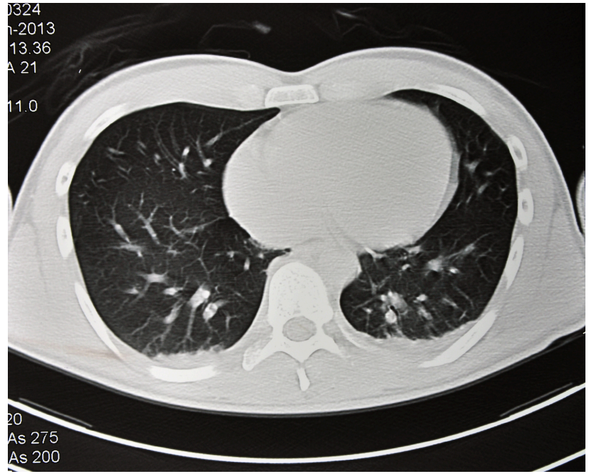
Make an initial diagnosis for Beth’s illness in your Patient Chart.

TESTING

- From listening to her lungs, fluid is built up, which is causing her wet coughing

- A CT scan of her lungs confirms the presence of fluid

- A nasal nitric oxide test was performed, and the level breathed out was low
  - Low levels indicate poor function of the lungs



- The doctors considered taking a lung biopsy, but instead opted for genetic testing first, since this is less invasive

Fill out the Tests section in the Patient Chart. Make any adjustments needed to your initial diagnosis.

DIAGNOSIS

- Beth was diagnosed with PCD, which is a respiratory illness that is often misdiagnosed as asthma or Cystic Fibrosis

- Early signs of this disease are breathing difficulty at birth, along with frequent ear and respiratory infections

- Unlike asthma, which can be triggered by things like exercise, seasonal allergies, or environmental pollution, PCD symptoms occur year-round and are not brought on by environmental conditions

- With proper treatment, PCD patients can live more comfortably

Fill in the Final Diagnosis section. Then, list treatments that could help some of Beth's symptoms. Which predictions were closest to the actual disease?

TREATMENT PLAN

- Beth was referred to begin chest physical therapy to help manually clear the fluid from her lungs

- Additionally, unlike asthma, moderate exercise is recommended because this can help loosen mucus

- She was prescribed anti-inflammatory medication and bronchodilators, which will help her breathe easier

Update your Treatment section with this information.

WHICH ORGANELLE IS AFFECTED?

On the third page of your Guided Notes sheet:

1. Make a **claim** about which organelle is malfunctioning.
2. List the **evidence** from your Patient Chart that supports your claim.
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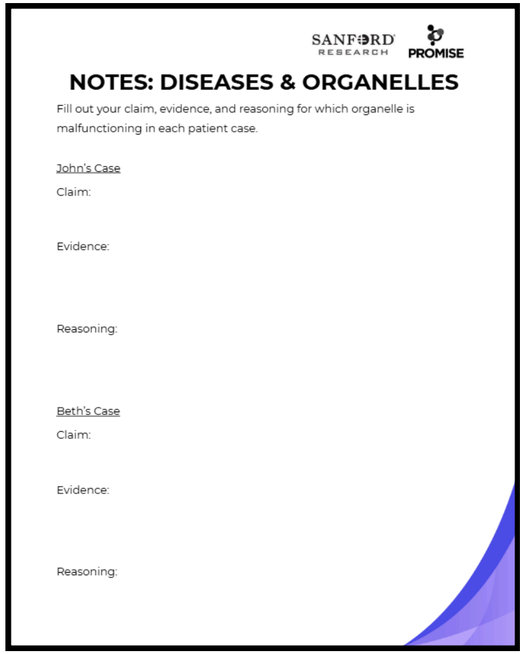

SANFORD RESEARCH PROMISE

NOTES: DISEASES & ORGANELLES

Fill out your claim, evidence, and reasoning for which organelle is malfunctioning in each patient case.

John's Case

Claim:

Evidence:

Reasoning:

Beth's Case

Claim:

Evidence:

Reasoning:

GENETIC RESULTS

- PCD stands for Primary Ciliary Dyskinesia, which means that the cilia in the lungs are not able to move correctly to push mucus and particles out of the lungs

- Beth has a mutation in the gene ***DNAH5***, which stops the lungs from being able to clear out mucus and debris
  - This gene makes a protein that normally tells the cilia to move

- Because this debris accumulates, it often leads to respiratory infections and distress if untreated

- This mutation is inherited in an autosomal recessive pattern, which means she got one copy of the mutated gene from each of her parents

REAL-LIFE RESEARCH

- Researchers are working to understand how rare diseases develop and how to treat them
- At Sanford Research, some diseases that our scientists focus on are Batten Disease and Primary Ciliary Dyskinesia
- Our scientists must study the diseases on a cell-level and in the whole body to understand what is going wrong
- This research helps us find the best ways to treat rare diseases in the clinic