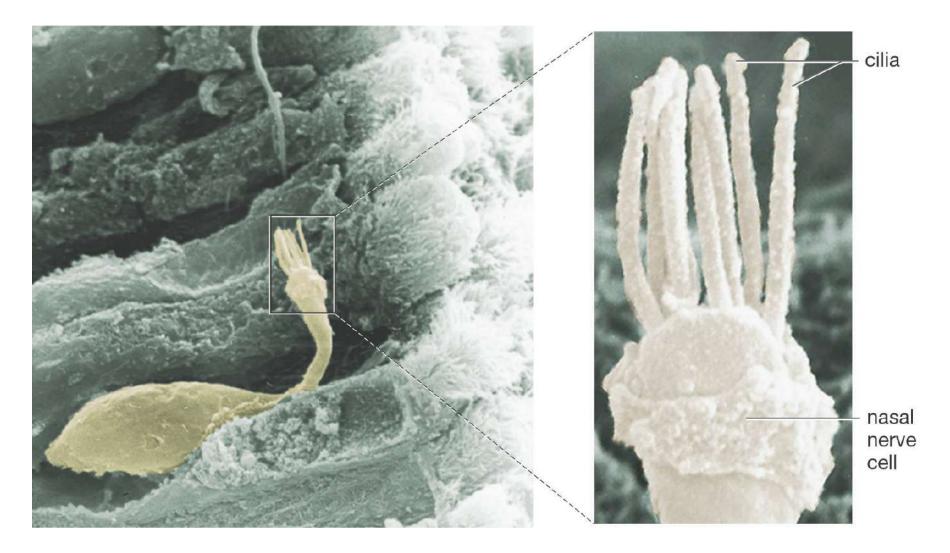
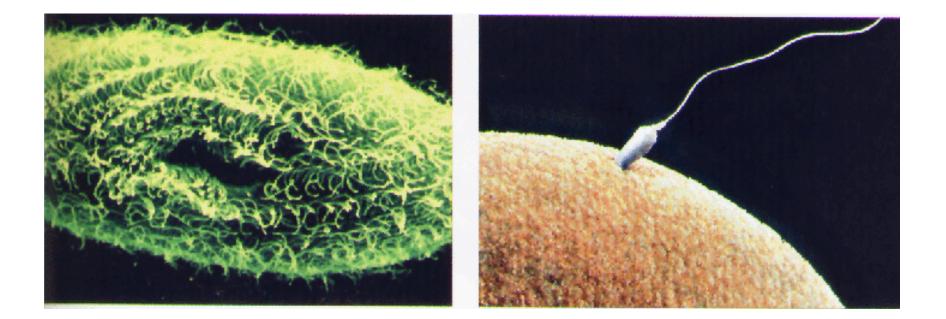
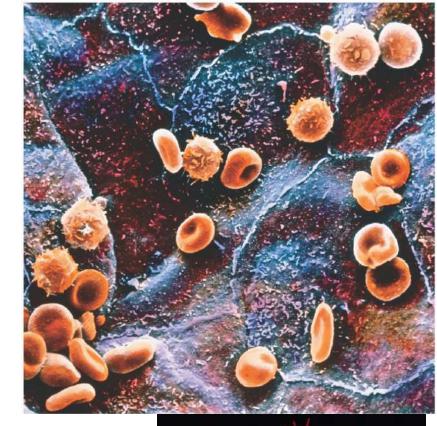
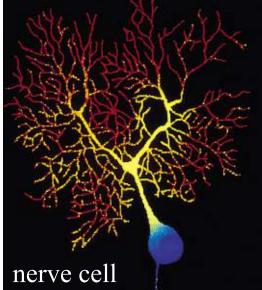


Figure 9–14. Molecular Biology of the Cell, 4th Edition.





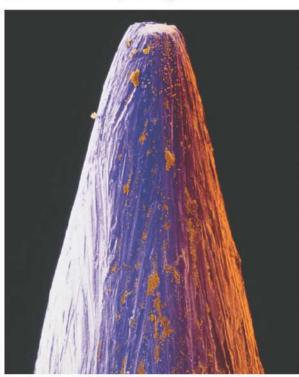




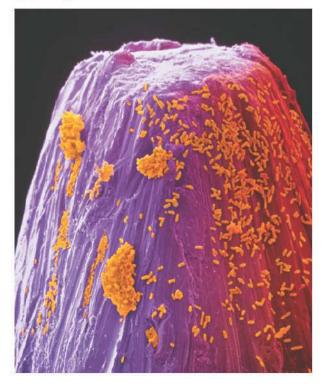




### (a) Bacteria on a pin, magnified x 85

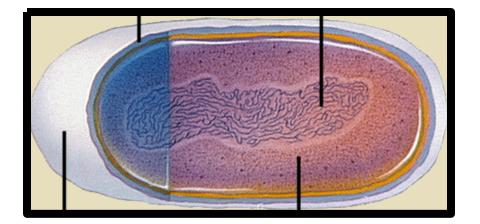


## (b) Magnified x 425

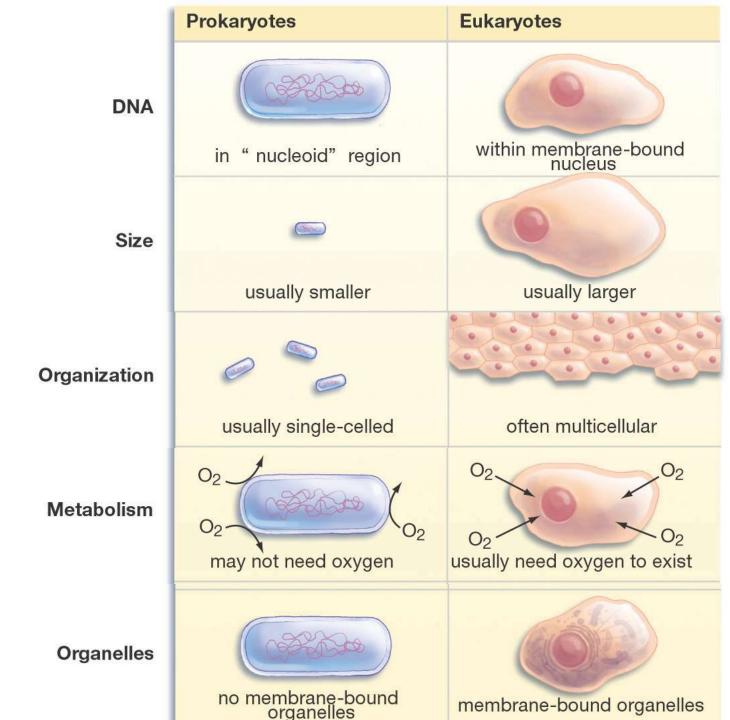


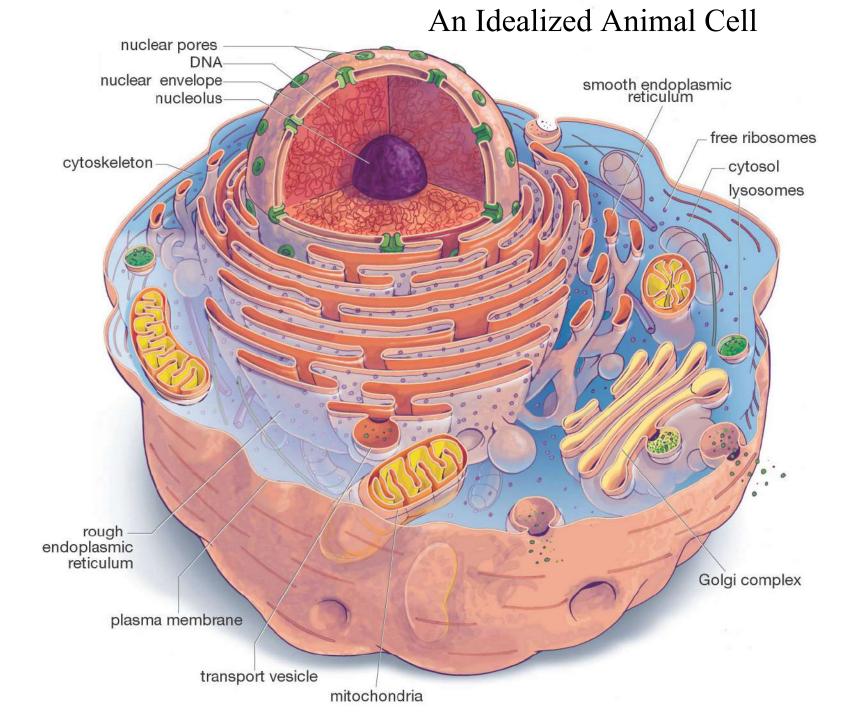
## (c) Magnified x 2100





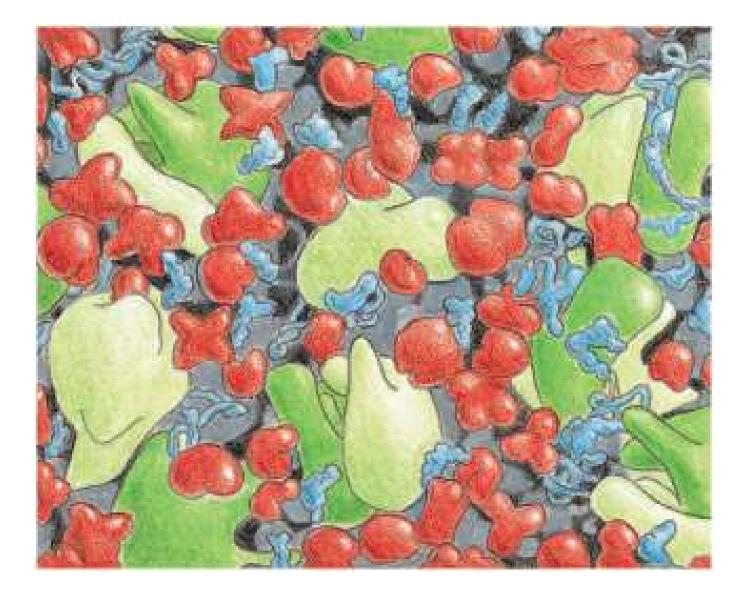


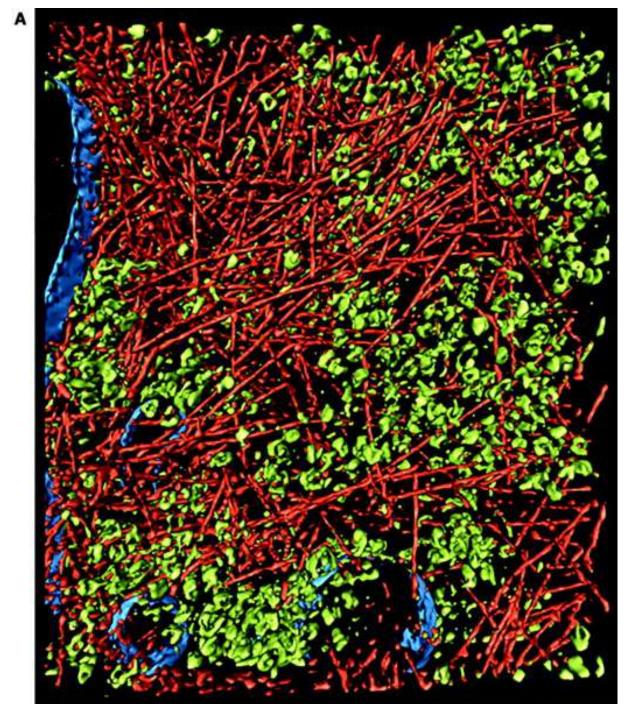


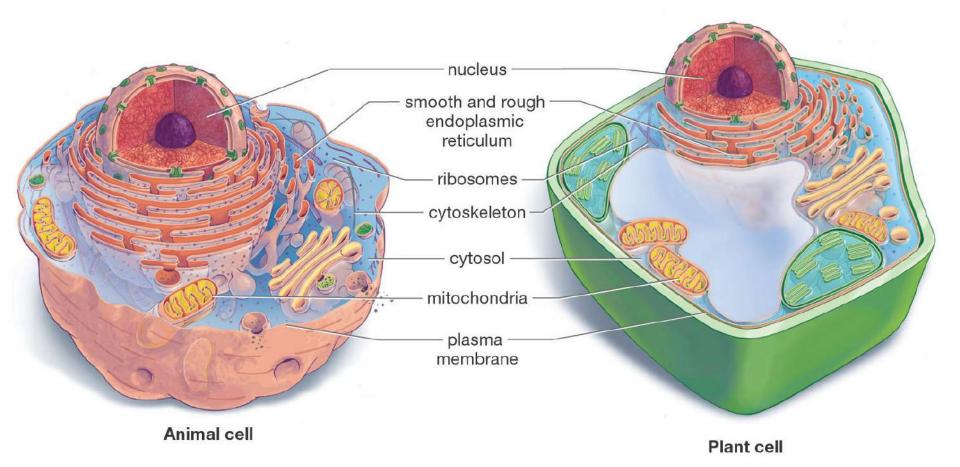


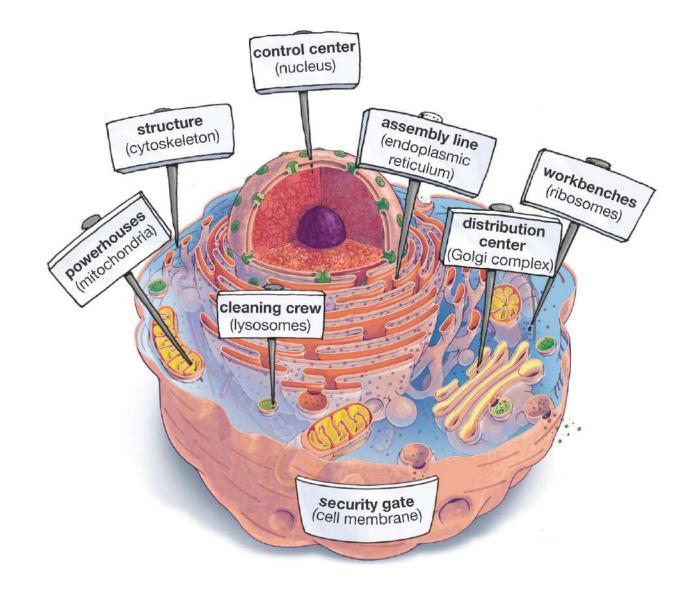


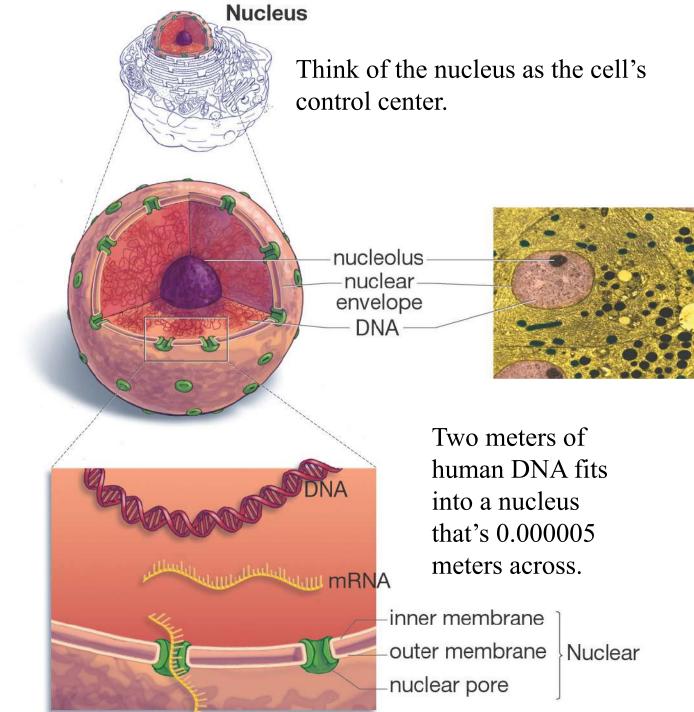


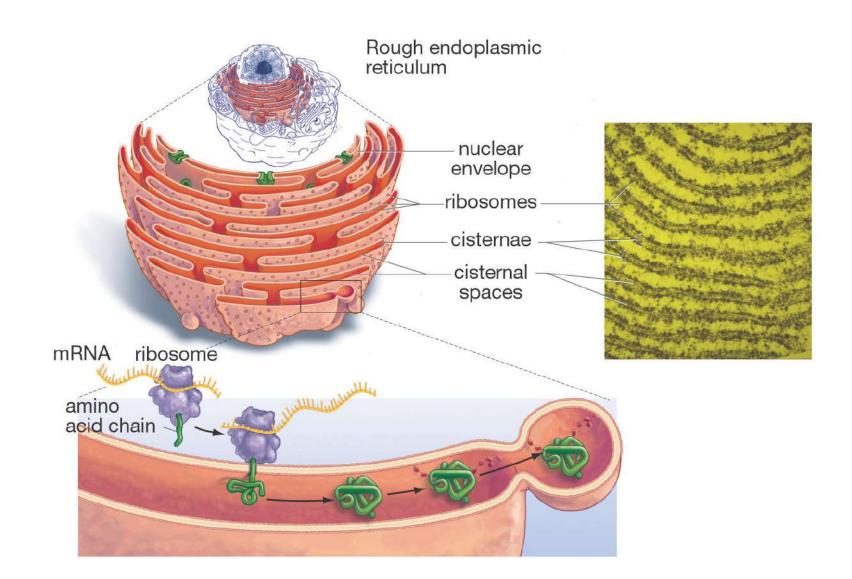


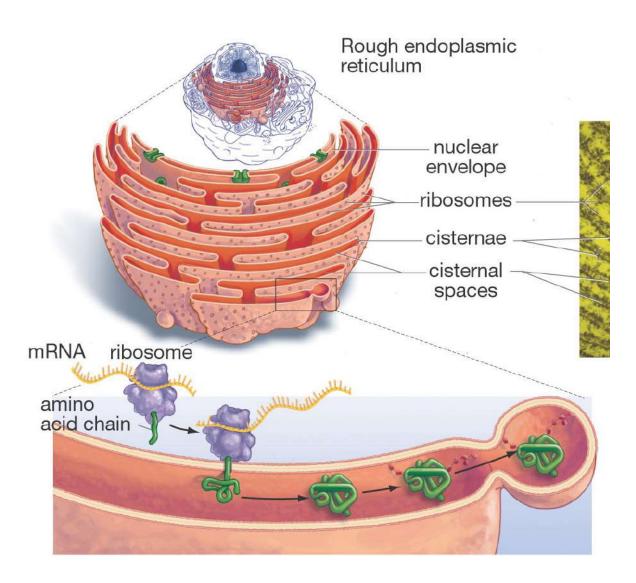












HHAMI HOWARD HUGHES MEDICAL INSTITUTE



GLOSSARY

#### Stalking a Lethal Gene

A Gifted Young Patient Seeks His Own Genetic Flaw

Finding the Faulty Gene's Fellow Travelers

"Jumping" Toward the Gene

Discovering the Gene for Cystic Fibrosis

Seeking New Treatments

Who Should Be Tested?

In Search of Large Families

Reading the Human Blueprint

Why So Many Errors in Our DNA?

How Genetic Disorders Are Inherited

How to Conquer a Genetic Disease

Of Mice and Men

Progress Continues

HHMI Home

# A Gifted Young Patient Seeks His Own Genetic Flaw

In the summer of 1990, 20-year-old Jeff Pinard set out to find the flaw in his genes that causes him to have cystic fibrosis.

He already knew quite a lot about <u>genetic diseases</u>, especially his own. Cystic fibrosis (CF) is a fatal disorder that clogs the lungs and other organs with a viscous, sticky mucus that interferes with breathing and digestion. It is the most common lethal inherited disease among white children and young adults, attacking about 30,000 Americans. Until recently, most patients died before reaching the age of 30. But Pinard, a microbiology major at the University of Michigan, was full of hope and could hardly contain his excitement at the thought of working with top scientists at the cutting edge of research on CF.

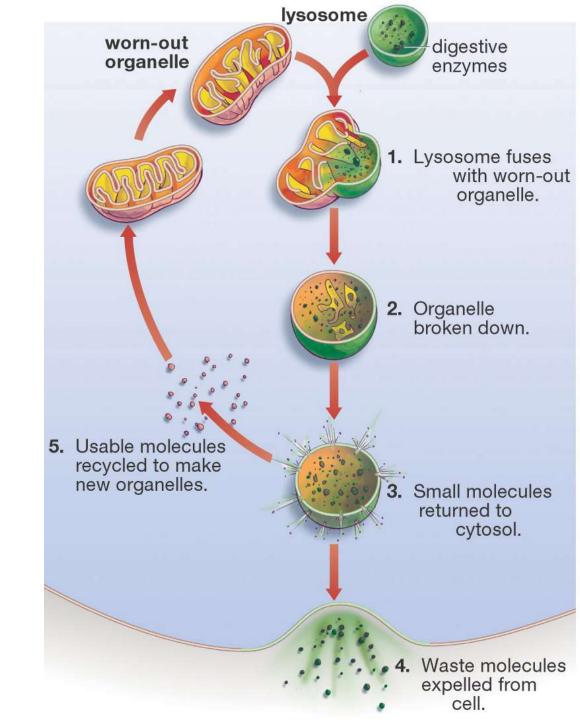
A year earlier—in a triumph of molecular genetics—a research team headed by Francis Collins, who was then an HHMI investigator at the University of Michigan, and Lap-Chee Tsui and John Riordan of Toronto's Hospital for Sick Children had discovered an errant gene that is responsible for CF. The researchers also identified the specific mutation, a missing snippet of genetic material, involved in most cases of CF.

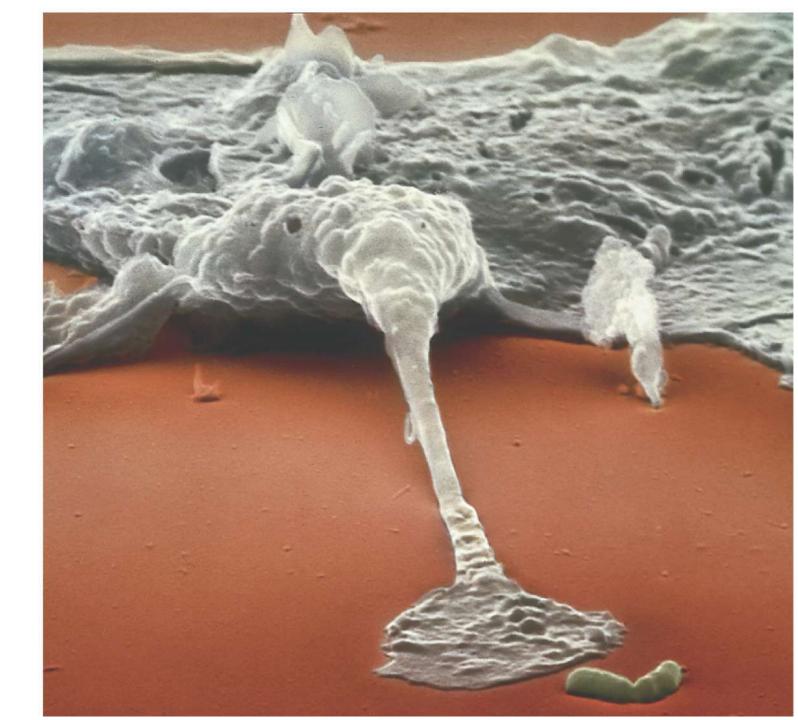
As many as 1 in 25 Americans of northern European descent—some 10 million people—carry a gene with a CF-causing defect. Babies who inherit a defective gene from both parents develop the disease.



Pinard adds fragments of a CF patient's DNA to a dye that will help track these fragments as the move through a porous gel.

## Click here to see the article.





# Lysosomal Storage Disease Center

Programs and Services

Lysosomal Storage Disease Center

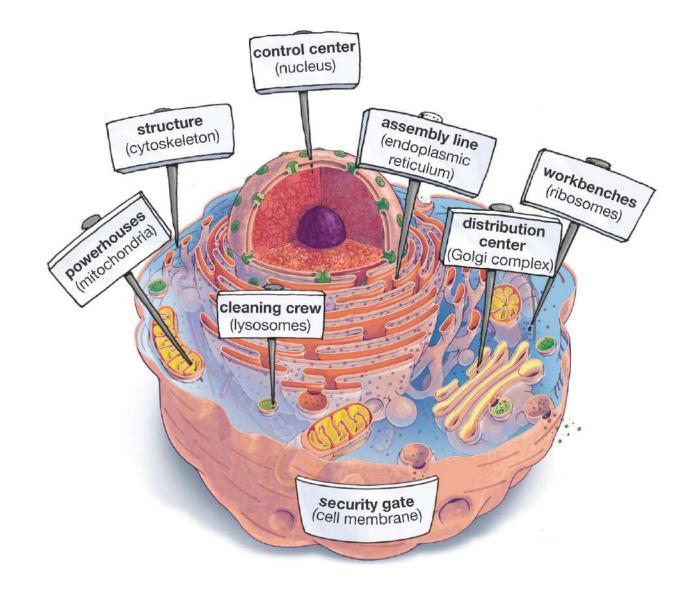
## Diagnosing, Managing and Treating Complex Conditions

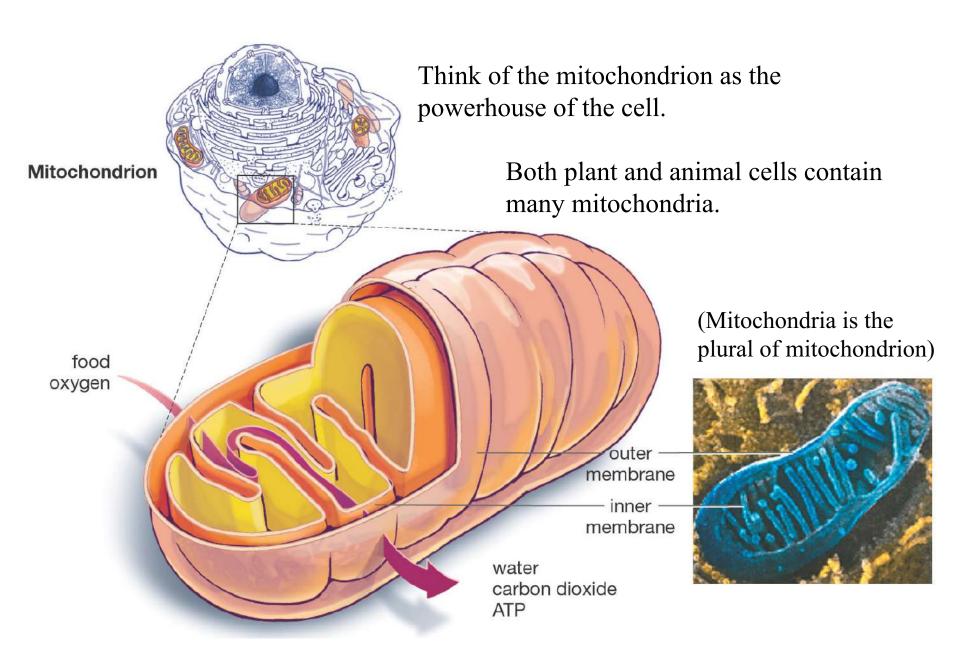
#### Lysosomal Storage Diseases

Lysosomal storage diseases are caused by a lack of enzymes that normally eliminate unwanted substances in the cells of the body. The enzymes are found in sac-like structures in cells called lysosomes. Lysosomes act as the "recycling center" of each cell, breaking down unwanted material into simple products for the cell to use to build new material. The lack of certain enzymes causes a buildup of the substance that the enzyme would normally eliminate, and deposits accumulate in many cells of the body. Abnormal storage causes inefficient functioning and damage of the body's cells, which can lead to serious health problems.

There are more than 40 known lysosomal storage diseases, including:

- Fabry disease causes kidney and heart problems, pain and a skin rash
- Gaucher disease causes the spleen to enlarge, anemia and bone lesions if untreated
- Hurler syndrome causes deformities of the skeleton and facial features, enlargement of the spleen and liver, joint stiffness,





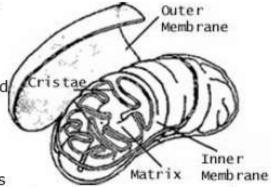




"To promote research and education for the diagnosis, treatment and cure of mitochondrial disorders and to provide support to affected individuals and families."

#### **Basis of the Disease**

Mitochondrial diseases result from failures of the mitochondria, specialized compartments present in every cell of the body except red blood cells. Mitochondria are responsible for creating more than 90% of the energy needed by the body to sustain life and support growth. When they fail, less and less energy is generated within the cell. Cell injury and even cell death follow. If this process is repeated throughout the body, whole systems



begin to fail, and the life of the person in whom this is happening is severely compromised. The disease primarily affects children, but adult onset is becoming more and more common.

Diseases of the mitochondria appear to cause the most damage to cells of the brain, heart, liver, skeletal muscles, kidney and the endocrine and respiratory systems.

Depending on which cells are affected, symptoms may include loss of motor control, muscle weakness and pain, gastro-intestinal disorders and swallowing difficulties, poor growth, cardiac disease, liver disease, diabetes, respiratory complications, seizures, visual/hearing problems, lactic acidosis, developmental delays and susceptibility to infection.

#### NEWS OF THE WEEK

#### MEDICINE

## Low-Power Mitochondria May Raise Risk of Cardiovascular Problems

Try as we might, only an elite few will ever win the Tour de France or even the local 10-K foot race. People simply vary widely in their ability to perform aerobic exercise. New work

with rats now suggests that individuals with a low tolerance for aerobic exercise may have a lot more to worry about than just their inability to run fast and long. The same underlying defect that reduces aerobic capacity may also predispose a person to a witch's brew of medical problems that could increase the possibility of heart attacks and strokes.

On page 418, a research team including Ulrik Wisløff of the Norwegian University of Science and Technology in Trondheim. Sonia Naijar of first time researchers have linked it to all of them at once. "This is an incredibly provocative study," says Vamsi Mootha of Massachusetts General Hospital in Boston, either high or cise. They ide ity to run on with one ano animals with oxygen meta biology, def pathology," e The anim

report, the p



Running for their lives. These rats, bred to have high aerobic capacity, appear to have fewer cardiovascular risk factors than their couch-potato cousins.

21 JANUARY 2005 VOL 307 SCIENCE www.sciencemag.org

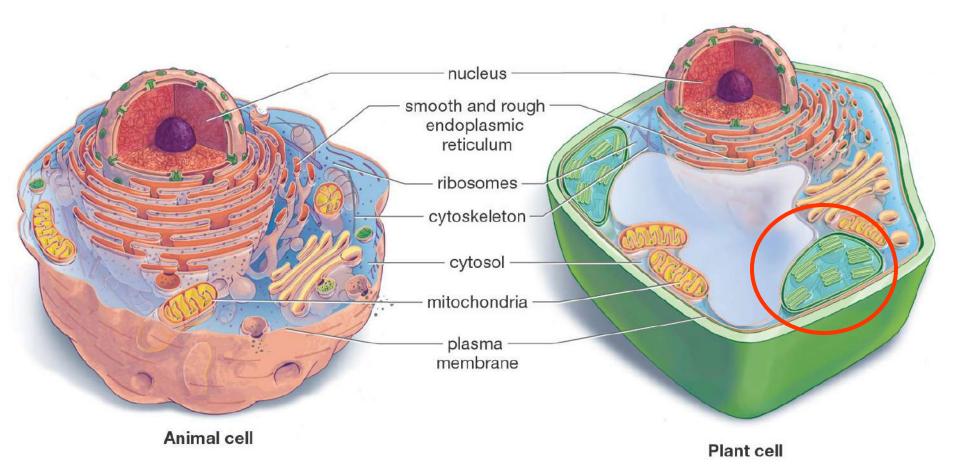
NATURE VOL 429 27 MAY 2004 357

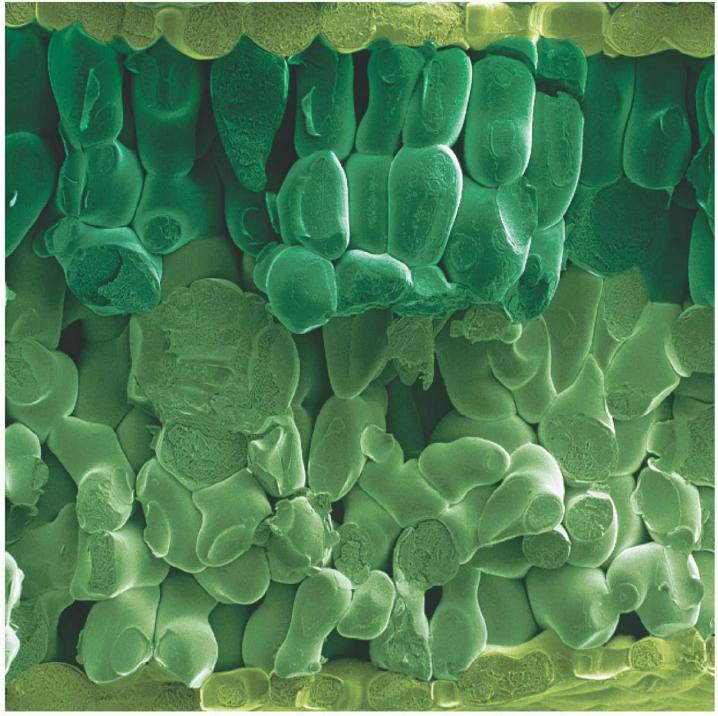
#### Ageing

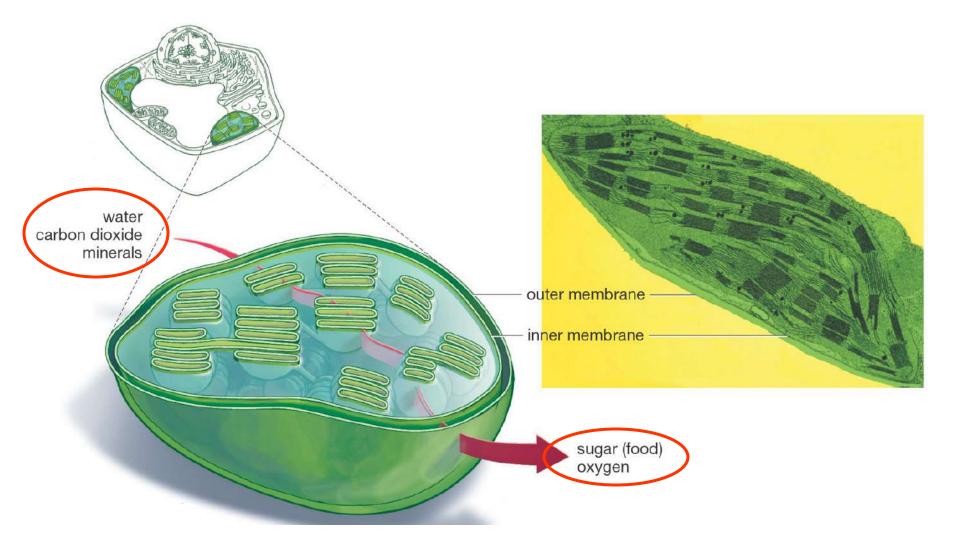
# Mice and mitochondria

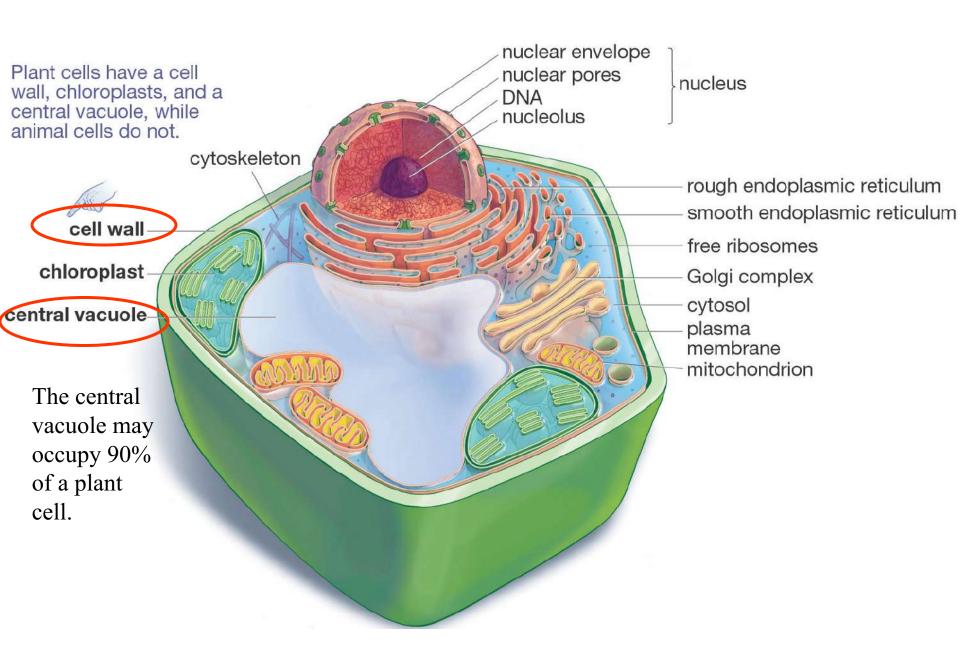
George M. Martin and Lawrence A. Loeb

It can be hard to work out whether particular events are a cause or a correlate of ageing — do mutations in mitochondrial DNA, for instance, speed up the process of growing old? Some clever studies suggest so.







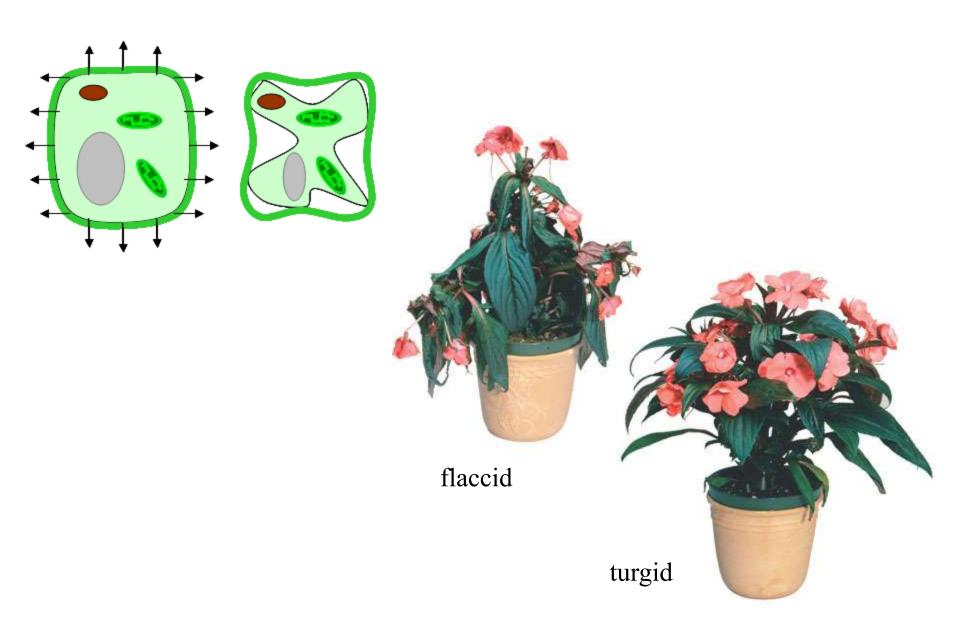


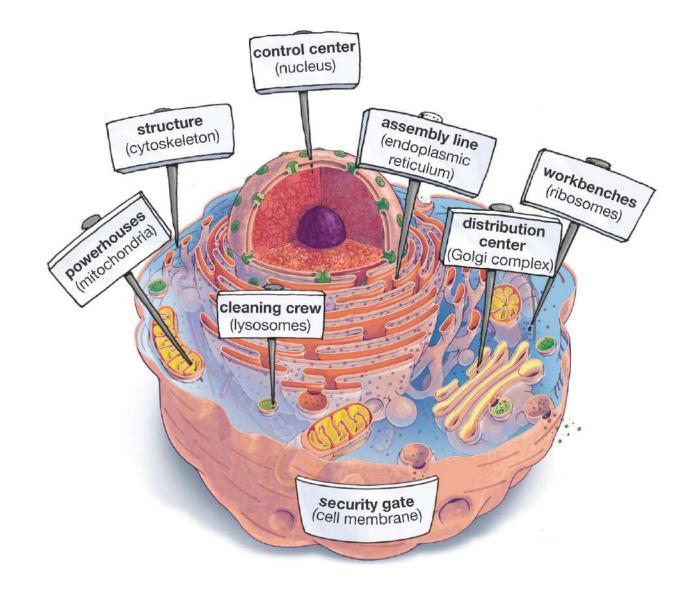
(a) Wood is mostly cell walls



(b) A magnified view of bark







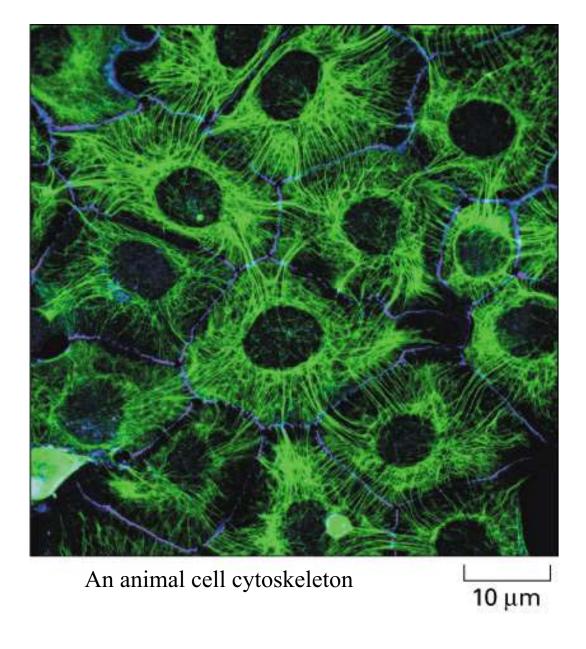


Figure 16–18. Molecular Biology of the Cell, 4th Edition.

