



Using Rare Diseases to Teach about Scientific Inquiry

Presented by: Dr. Mark Bloom

April 25, 2011

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Section I Introduction



Biological Sciences Curriculum Study

- Established in 1958
- Nonprofit organization
- Three main areas of emphasis
 - Curriculum Development
 - Professional Development
 - Research and Evaluation

NIH Curriculum Supplement Series

Free curriculum supplements to support elementary, middle school, and high school teaching:

<http://science.education.nih.gov>



The screenshot shows the NIH Curriculum Supplement Series website. The main navigation bar includes 'Home', 'FAQ', 'Links', and 'Contact Us'. The page is divided into three columns: Title, Description, and News.

Title Column: Includes a search prompt: "Select a title to request print versions, access electronic versions, and view state education standards alignments." It lists categories: High School, Middle School, and Elementary School, each with a list of supplement titles.

Description Column: Features a featured supplement titled "Cell Biology and Cancer" for Grades 9-12. It includes a thumbnail image of the supplement cover and a brief description: "The NIH curriculum supplements are teacher's guides to two weeks of lessons on the science behind selected health topics. They combine cutting-edge biomedical discoveries with state-of-the-art instructional practices." Below this, it states: "HTML and PDF versions of each supplement are online and accessible to all. Print versions are **FREE** upon request to educators in the U.S."

News Column: Features a news item titled "Exploring Bioethics is now available!" with a thumbnail image of the supplement cover. The text reads: "Request your free copy today. Our newest curriculum supplement for high school is a collaboration between NIH and EDC. These lessons present a new model for ethical inquiry in the biology classroom. Online teacher support materials and alignments with state education standards are in the works and will be posted soon on the Exploring Bioethics summary page."

The Importance of Scientific Inquiry

- Educational research supports the idea that scientific inquiry should be viewed both as a learning goal and a teaching method.
- This approach is described in the National Science Education Standards, which lists specific understandings and abilities associated with scientific inquiry.
- Many teachers do not make scientific inquiry an integral part of their teaching.

Rare Diseases and Scientific Inquiry

Objectives

- To illustrate that the study of rare diseases is not just important to those affected individuals and their families, but also contributes knowledge that can be applied to more common diseases.
- To use the context of rare diseases to help middle school students experience scientific inquiry.
- To encourage students to think about human health as:

Knowledge + Choice = Power

Power + Behavior = Enhanced health



Let's pause for questions
from the audience





Section 2

Overview of Rare Diseases and Scientific Inquiry

Rare Diseases and Scientific Inquiry (approximately 2 weeks of instruction)

Lesson 1: What Is a Rare Disease?

Lesson 2: What Causes Rare Diseases?

Lesson 3: The Difficulty of Diagnosis

Lesson 4: The Importance of Medical Research

Lesson 5: Communicating about Rare Diseases

Engage: What Is a Rare Disease?

- Using a reality TV show scenario students consider their feelings about rare diseases and their attitudes toward people affected by a rare disease.
- Student preconceptions about disease and its causes are elicited.
- Students distinguish between diseases that be cured and those that can be managed.



Explore: What Causes a Rare Disease?

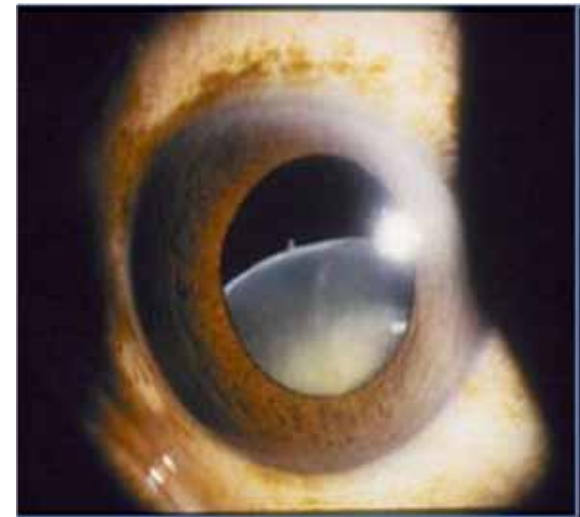
Students assume roles of medical officers at an army post. They

- analyze information about visits to the infirmary;
- consider the 3 major causes of disease;
- recognize that disease causes may interact;
- learn that a bacterium that causes a common disease may sometimes cause a rare disease.



Explain: The Difficulty of Diagnosis

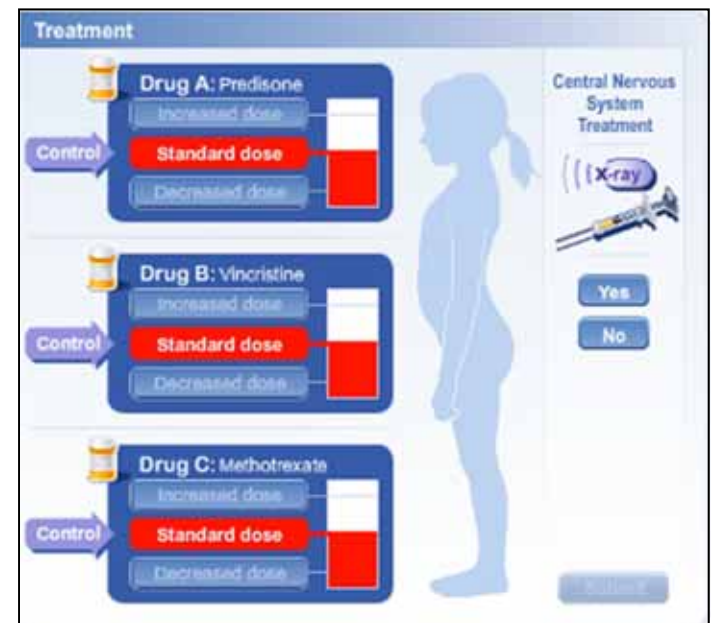
- Students learn about a teenager that wants to try out for the basketball team but has health problems.
- Students assume the roles of medical specialists. They combine their findings to look for a factor that connects the patient's different health problems.



Elaborate: The Importance of Medical Research

Students are introduced to parents whose daughter has been diagnosed with leukemia. They

- perform a simulated Web search to learn about the disease and identify reliable sources of information.
- perform simple karyotype analysis to see the relationship between leukemia and genetics.
- design a clinical trial to guide treatment of the disease.



Evaluate: Communicating about Rare Diseases

- Students use what they learned to create an informational poster about one of the diseases covered in this supplement.
- Students evaluate a poster made by their classmates.
- Students view a video made by a person with a rare disease, revisit their initial ideas about rare diseases, and discuss how their thinking has changed.



Let's pause for questions
from the audience



Section 3

Lesson 3: The Difficulty of D

- Introducing the scenario



Patrick is a 13-year-old middle school student who loves to play basketball. Excited, he came home from school and explained to his parents that the school basketball team will be holding tryouts next month and he wants to participate.

Patrick's parents are both happy and concerned for him. They are happy because they know Patrick loves sports and feel that the exercise will be good for him. They also know that Patrick has been occasionally teased because he is tall and thin. Maybe by joining the basketball team he will make new friends and feel more accepted by his classmates.

Patrick's parents are concerned because he has some health problems. When Patrick was a toddler, the family doctor diagnosed him with a heart murmur. The doctor explained that a heart murmur refers to a sound that the blood makes as it flows through the heart. She further explained that heart murmurs are usually harmless and that Patrick could lead a normal life.

When he was nine, Patrick developed a problem with his eyesight and it was discovered that one of his eye lenses was detached and had to be repaired.

When Patrick was 10, he was diagnosed with asthma. The doctor explained that asthma causes the tubes carrying air in and out of the lungs to become sore and swollen. This can cause coughing and wheezing and make it difficult to breathe. She also explained that with proper management of his asthma, Patrick could play sports and that the exercise might even improve his condition.

Finally just last year Patrick was diagnosed with scoliosis or curvature of the spine. The doctor explained that Patrick's scoliosis was moderate and as with most children the cause was unknown. He further explained that in 90% of cases no future treatment is needed.

Explain: The Difficulty of Diagnosis

- Patrick's parents wonder if his medical problems have a common cause.
- They take him to see a medical geneticist.
- Students act as specialists helping a medical geneticist make a diagnosis of a rare genetic disease.
- The question:

“Do Patrick's health problems result from an underlying genetic mutation?”



Let's pause for questions
from the audience



Section 4

Lesson 3: The Difficulty of Diagnosis

- Analyzing the evidence

Explain: The Difficulty of Diagnosis

Let's see what the medical specialists have to say.

- Cardiologist (heart)
- Ophthalmologist (eyes)
- Pulmonologist (lungs)
- Orthopedist (bones)

Cardiologist

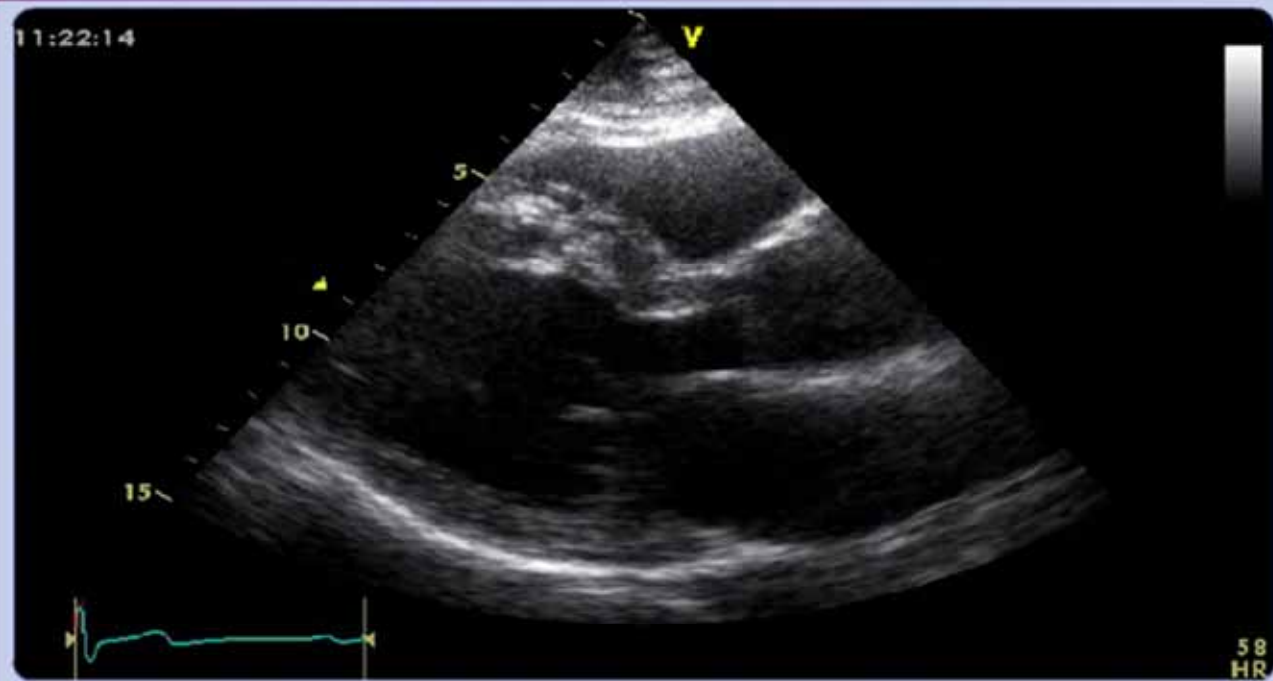
Medical history

Patient was previously diagnosed with a heart murmur. An echocardiogram reveals mitral valve prolapse and an enlarged aorta.

Physical exam

The presence of a heart murmur was confirmed. An echocardiogram revealed the presence of mitral valve prolapse.

Echocardiogram



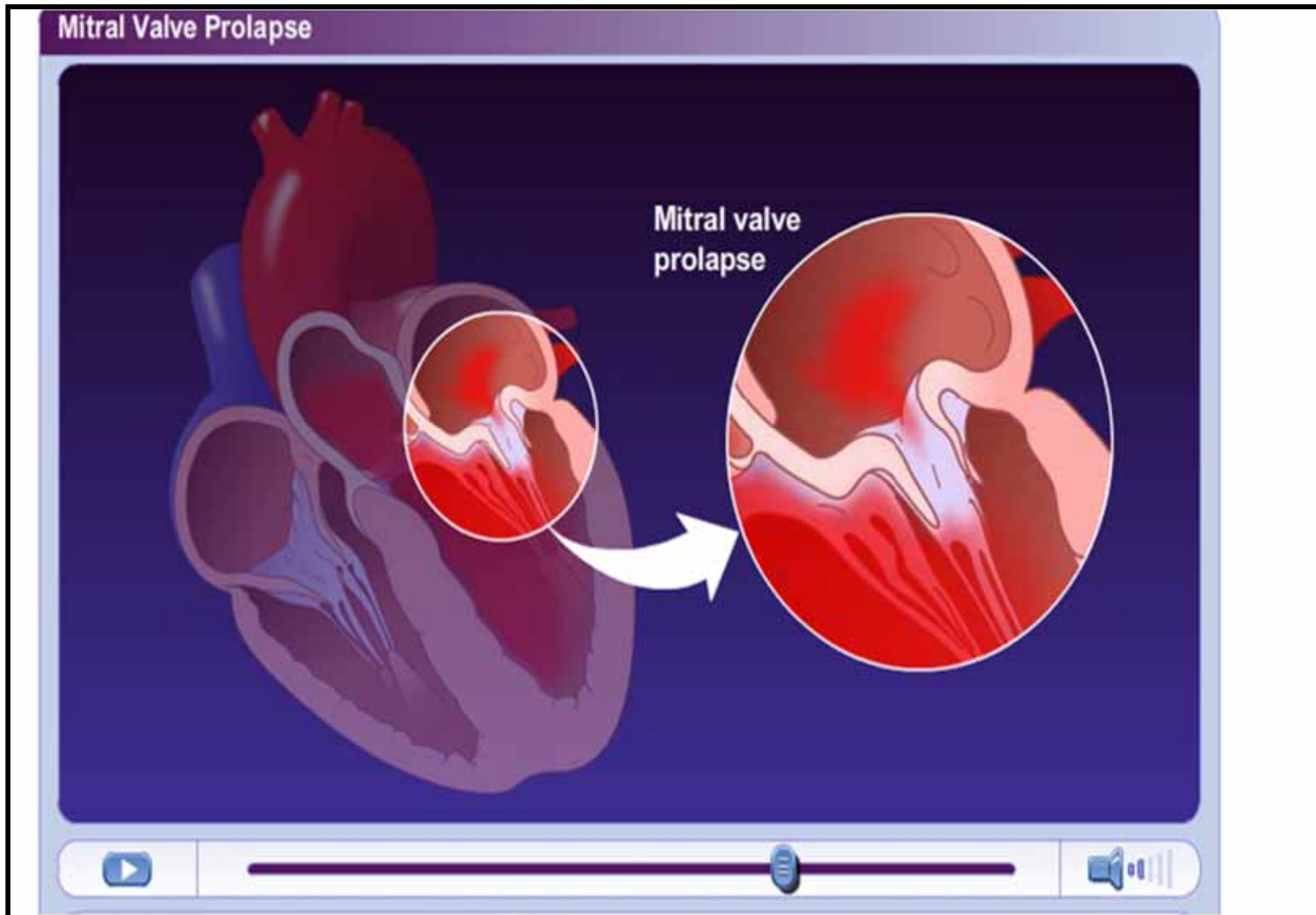
Heart Murmur

When doctors use a stethoscope to listen to the heart, they hear a lub-DUB sound made by heart valves opening and closing as blood flows through the heart. The term heart murmur refers to an unusual whooshing sound heard by doctors listening to the heart.

Mitral Valve Prolapse

This is a condition where one of the heart's valves doesn't work properly. The valve flaps are "floppy" and don't close as they should. This sometimes causes blood to flow backward from its normal direction. This backward flow of blood may be associated with shortness of breath or chest pain.

The cause of mitral valve prolapse is not known. Most people with the condition are born with it. It tends to run in families and is associated with connective tissue disorders such as Marfan syndrome.





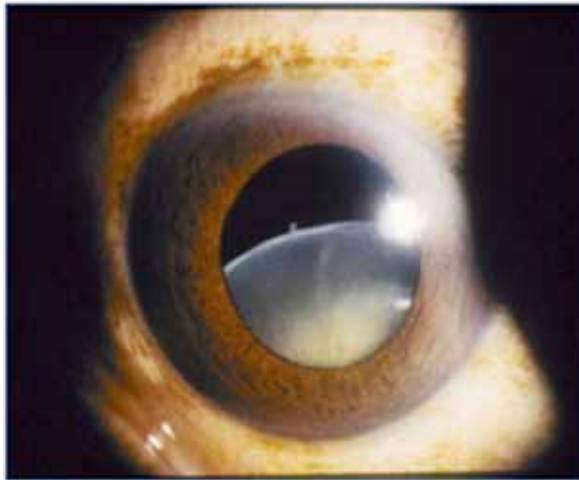
Questions about Patrick and his heart?



Ophthalmologist

Medical history

The patient is myopic (nearsighted). When he was 9 years old he was being fitted for eye glasses when an exam revealed that his left lens was dislocated.



Slit lamp exam showing detached lens

Physical exam

An eye exam confirmed myopia and a repaired detached left lens.

Medical Reference Manual Ophthalmology

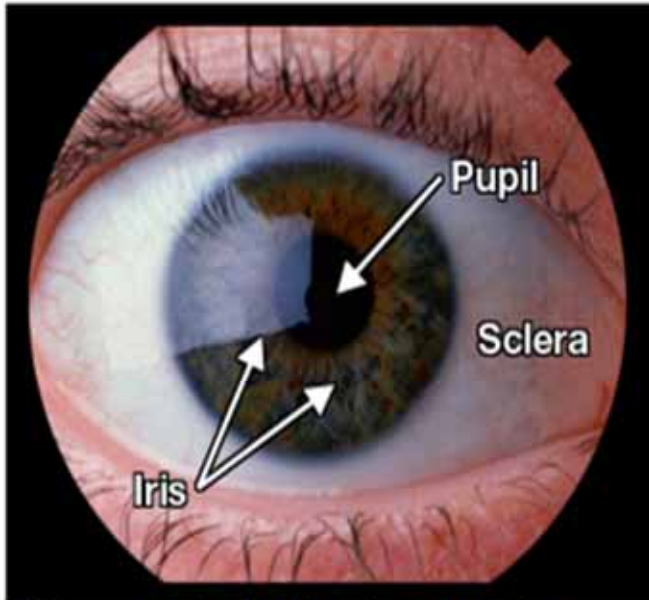
Ophthalmologist

Nearsightedness is caused by a change in the shape of the eyeball so that it is egg-shaped instead of round. This egg-shaped eyeball focuses light a little in front of the retina instead of directly on it resulting in blurry vision. Myopia is a common condition affecting between 30-40% of the American population.

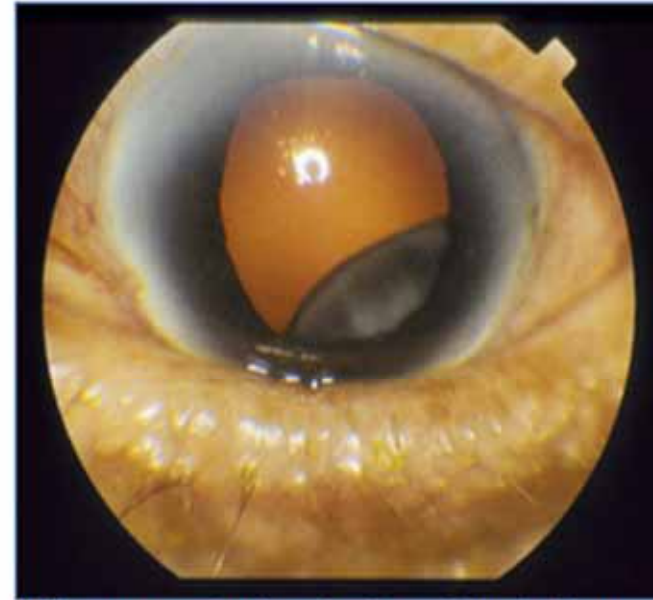
Detached Lens

A detached lens means that the lens has shifted from its normal position centered behind the pupil. If the detachment is moderate, the problem may be corrected with glasses. A severe lens detachment may require surgery to correct.

Detached lenses are rare in the general population. They are often caused by a blow to the eye. The condition is much more common among people with certain diseases involving connective tissue such as Ehlers-Danlos Syndrome and Marfan Syndrome.



Slit lamp examination showing a normal eye



Slit lamp examination showing a detached lens



Questions about Patrick and his eyes?



Pulmonologist

Medical history

Patient has been diagnosed with asthma. When 7 years old, he experienced a collapsed lung.

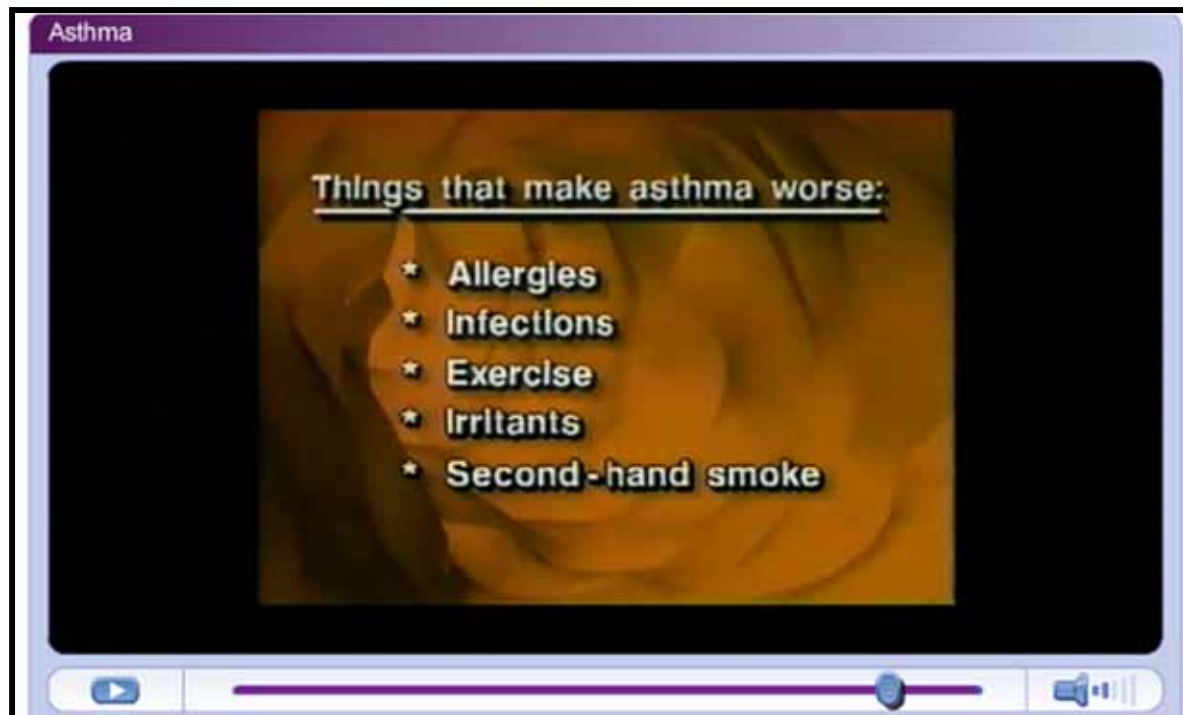
Physical exam

Exam confirmed the diagnosis of asthma. Lung volume was normal. Chest X-ray was normal.



Asthma

Asthma is a medical condition that causes the tubes bringing air to the lungs to become sore and swollen. This causes the affected person to wheeze and cough and experience shortness of breath. Asthma often starts in childhood but affects all ages. Over 22 million people in the United States have asthma.



Orthopedist

Medical history

Patient was previously diagnosed with mild scoliosis (curvature of the spine).

Physical exam

A physical exam and X-rays confirmed the presence of mild scoliosis. The curvature was measured to 8.3 degrees. It was noted that the patient has unusually long, slender arms, fingers, and feet.

Medical Reference Manual Orthopedics



X-ray of spine, age 5.

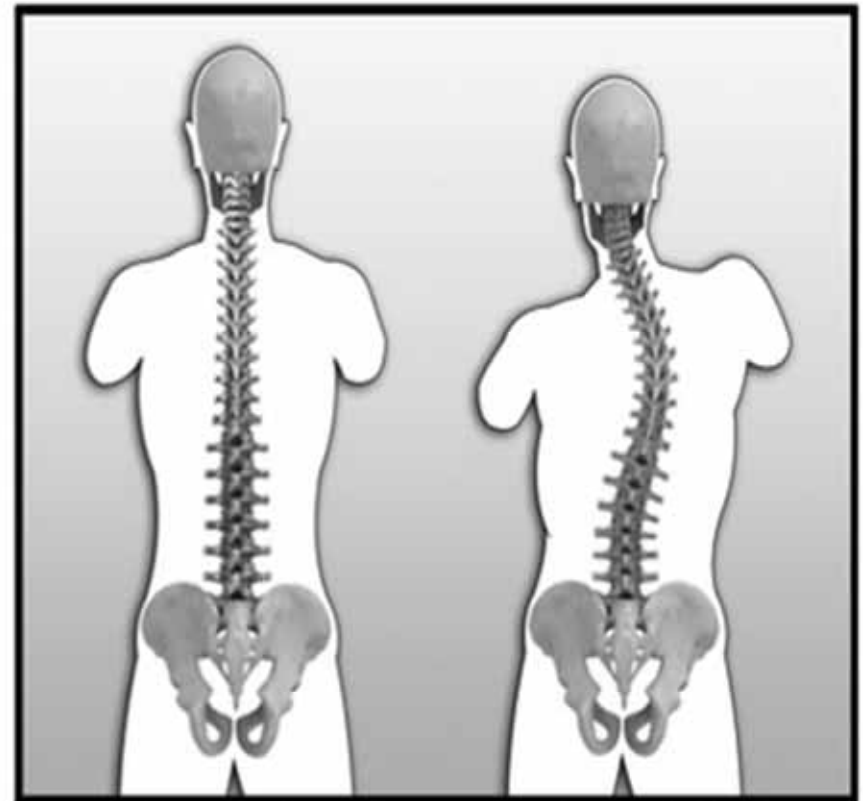
Scoliosis (Curvature of the spine)

Scoliosis refers to an abnormal curvature of the spine. About 10% of adolescents show some degree of scoliosis but less than 1% need treatment for the condition. The severity of scoliosis is described by the extent of the curvature.

Curvature less than 20 degrees	Mild
Curvature between 20-70 degrees	Moderate
Curvature greater than 70 degrees	Severe

Patients with mild scoliosis usually don't require treatment beyond examination to see if the condition worsens. Patients with moderate and severe scoliosis are treated by using back braces or surgery.

In most cases the cause of scoliosis is not known, however it does seem to run in families. In some cases the condition is caused by an injury. In other cases, the condition is a result of a muscle, nerve, or connective tissue disease.



Normal spinal column (left) and scoliosis (right)



Questions about Patrick and
his lungs and/or spine?



Section 5

Lesson 3: The Difficulty of Diagnosis

- Exploring connective tissue



Is there evidence that Patrick's medical problems may have a common cause?



Connective Tissue Disorders

Connective tissues are made of proteins and fats. They support your body's organs and give them their shapes. Cartilage is an important connective tissue. It is stiff but more flexible than bone. Cartilage helps your bones move and glide over each other. It also gives shapes to body parts such as your nose and ears.

Connective tissue may be damaged by injury or through infection. It can also be damaged by a large number of rare genetic diseases.



What is connective tissue?

- Students use rubber bands as a model system.
- Each student team compares the behavior of two different rubber bands.

Ehler-Danlos Syndrome

Ehlers-Danlos syndrome refers to a collection of related disorders that weaken connective tissues. Symptoms may be mild or life threatening. They include:

- Heart valves that leak
- Weakened blood vessels
- Loose joints
- Soft, stretchy skin that bruises easily
- Muscle weakness

Ehlers-Danlos syndrome is an inherited disorder. Treatment involves managing symptoms and learning how to protect the joints and prevent injuries.

Marfan Syndrome

Marfan syndrome is a disorder of connective tissue that is due to mutations in a gene that codes for a protein called fibrillin. Symptoms may be mild to severe. Often people with Marfan syndrome are tall, thin, and have loose joints. Their fingers and feet may be unusually long. Other symptoms may include:

- Heart valves that leak
- Heart murmur
- Weakened blood vessels
- Curvature of the spine
- Sudden lung collapse
- Nearsightedness and problems with the eye lens
- Flat feet
- Teeth that are crowded together

Treatment for Marfan syndrome involves managing symptoms and adopting physical activity guidelines that are specific to each person.

Scleroderma

Scleroderma is a group of related disorders involving abnormal growth of connective tissue. One type of scleroderma affects only the skin. Another type can also affect other body systems. The cause of scleroderma is not known. It is more common in females than males. Other symptoms may include:

- Calcium deposits in connective tissues
- Narrowing of blood vessels in the hands and feet
- Swelling of the esophagus (tube between the throat and stomach)
- Thick, tight skin on fingers
- Red spots on hands and face

Treatment involves managing the symptoms.

Osteogenesis Imperfecta

Osteogenesis imperfecta is an inherited disorder that causes bone weakness. The disorder is caused by mutations to a gene involved with making the protein collagen. Sometimes bones break for no apparent reason. Other symptoms may include:

- Muscle weakness
- Curvature of the spine
- Loose joints
- Hearing loss
- Skin that bruises easily
- Brittle teeth

Treatment involves exercise, physical therapy, braces, and surgery.



Which disorder best fits Patrick's symptoms?

- A. Ehlers-Danlos syndrome
- B. Marfan syndrome
- C. Scleroderma
- D. Osteogenesis Imperfecta



Questions about disorders of connective tissue?



Section 6

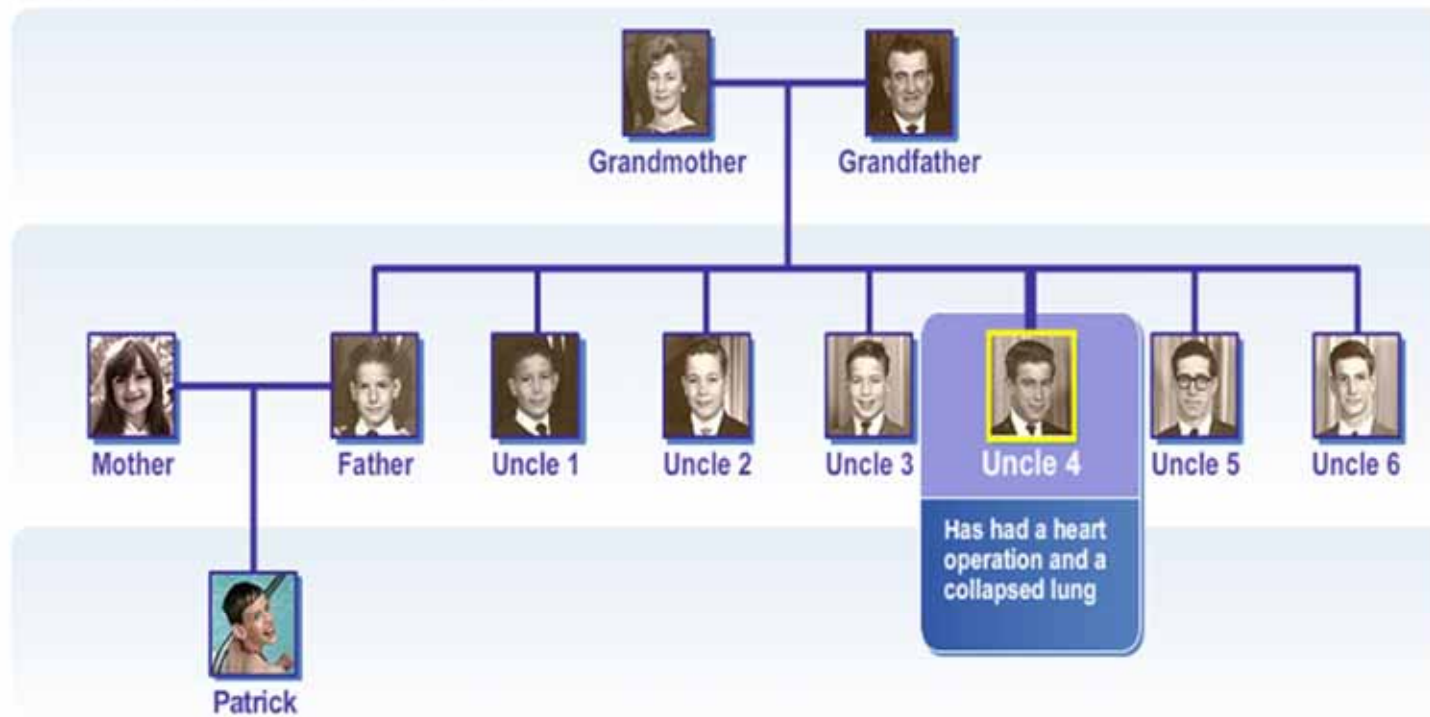
Lesson 3: The Difficulty of Diagnosis

- Marfan syndrome and heredity

Patrick may have Marfan syndrome. Is there evidence that it runs in his family?

Patrick's Family Tree

Roll over each photo to read about that person's medical history.



Evidence of Marfan syndrome in Patrick's family:

1. Grandfather had several heart operations
2. Father had two heart operations and has mild scoliosis
3. One uncle has had a heart operation and a collapsed lung
4. Another uncle had several heart operations and a detached eye lens



Questions about Marfan syndrome and heredity?



Section 7

Lesson 3: The Difficulty of Diagnosis

- Living with Marfan syndrome

People with Marfan Syndrome



Quotes from students with Marfan syndrome

- “When they measured my heart with the echocardiogram they told my mom they don’t think I should do marching band. I was wondering if I don’t exert myself too much, if I take it at my own pace, do you think I will still will be able to do it?”
- “The most frustrating thing for me is ...I can’t drive. I tried to get my permit and I couldn’t pass the vision test because I did have my retina detached.”
- “When it comes to how you’re socially accepted, high school is really lame. In a couple of years it’s not going to matter what sport you played or anything. It is going to matter what you know and what you do with the knowledge that you know.”

Revisiting ideas and attitudes about rare diseases

Me and Antoine B



I can't work my heart too hard, like I can't like run a bunch of miles... That's a horrible answer. "I can't run a bunch of miles?"

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