Did Environmental Exposure Cause a Disease Cluster?

In this video segment from Greater Boston, Liz Lombard describes her struggle with a painful, progressive autoimmune disease called scleroderma. Scleroderma is rare, so Lombard was surprised to learn that other people in her South Boston neighborhood also had the disease. Health officials explain that there is a genetic predisposition to scleroderma, and there may be environmental factors involved as well. However, officials don't know whether this particular cluster is a result of environmental hazards or simply a coincidence.

BACKGROUND ESSAY

When an unexpected number of people from the same area get a rare disease like scleroderma, it’s called a disease cluster. The scientists who investigate disease clusters are called epidemiologists. Epidemiology is the study of human health in populations, rather than in individuals. It studies the causes of illness, how diseases spread, and how to control them. Using their knowledge of diseases, environmental science, and statistics, epidemiologists try to determine whether a disease cluster is caused by the environment, by genetics, or simply by chance.

Epidemiology has limits. It can show a correlation between two phenomena, but it rarely proves one phenomenon caused the other. For example, the exact causes of scleroderma are still unknown. But there is some correlation between the disease and exposure to silica dust, industrial solvents, and certain chemotherapy drugs. Does this mean that everyone exposed to industrial solvents will get the disease? No. To prove that solvents cause the disease, scientists would have to expose test subjects to solvents to see if they developed scleroderma. This kind of study would be unethical.

Epidemiological studies are important. Even if they cannot prove that something causes a disease, they might point the way for further research. Take the case of scleroderma in Choctaw Native Americans. In 1996, epidemiologists found that a population of Choctaws living in Oklahoma were 20 times more likely than the general population to develop scleroderma. However, Choctaws living in Mississippi were not. This seems to indicate that something in the environment was causing scleroderma in Oklahoma. But when researchers looked around Oklahoma for clues, they found that non-Choctaws in the studied area did not appear to have a high incidence of scleroderma. So scientists turned to genetics.

By studying the Choctaw genome, researchers found more than 10 gene regions associated with scleroderma. While cases of scleroderma in Oklahoma Choctaws trace back to just five families that relocated from Mississippi to Oklahoma in the 1800s, exposure to something in the environment, such as a virus or bacteria, may trigger the disease among people with these genetic markers. If that’s the case, then epidemiology will have had a strong hand in piecing together the puzzle of this disease.

DISCUSSION QUESTIONS
Other than the environment, what might explain why people in the same South Boston neighborhood were experiencing the same health condition? What else do they have in common?

Do you think the cluster of scleroderma cases in South Boston could be coincidence? What additional information would you need to determine that?

What are the challenges in linking a disease such as scleroderma to an environmental factor? How could you test the hypothesis that an industrial agent causes scleroderma?

How could you eliminate genetics as an explanation for the cluster of scleroderma patients in a particular neighborhood?


SEE VIDEO FILE TITLED SCLERODERMA