What are Enzymes Made Of?

Extension Activity

The discovery that enzymes were made of protein was a significant step toward determining where they come from.

We now know that genes determine the enzymes that we possess in our bodies. When an enzyme is not working properly it infers that the gene has been altered or mutated. A mutation in a gene coding for an enzyme almost always impairs or destroys enzyme function (*Audesirk 1999, page 216).*

Here are some scenarios of conditions that result from genes that code for abnormal enzymes or proteins. Select ***one*** of the scenarios and write an abstract that contains the

- name of the condition,
- number of the chromosome that the gene causing the condition is found on, and
- name of the enzyme or protein that causes the condition.

Note: *Some of the scenarios are more difficult to research than others; the teacher may wish to limit the scenarios from which the students can choose.*

Scenario #1
The red blood cells of a person may appear normal until the person exercises or travels to high altitudes. The red blood cells may change shape and block capillaries, causing pain, stroke, or a heart attack. Some believe that this condition provides resistance to malaria. *[The condition is sickle cell anemia. Valine is substituted for glutamic acid as the seventh amino acid of hemoglobin.]*

Scenario #2
This condition prevents a person from breaking down a certain amino acid. The amino acid, phenylalanine, is converted to other chemicals that accumulate in the bloodstream. These abnormal substances are harmful to infants because they interfere with the development of brain cells. Infants with this disorder suffer severe mental retardation and rarely live more than thirty years. When detected early enough, individuals with this condition can be given special diets and develop and mature normally. The condition is not usually harmful to adults (*Raven & Johnson 1992, page 270).*
[The disorder is Phenylketonuria. A defective form of the enzyme phenylalanine hydroxylase is produced.]

Scenario #3
This incurable human disease causes a slow, progressive deterioration of the brain. It results in the loss of motor coordination, flailing movements, personality disturbances, and eventual death. The symptoms typically do not appear until 30 to 50 years of age *(Audesirk 1999). [The condition is called Huntington's disease. It is on chromosome 4; the gene's product is a protein dubbed "huntingtin".]*

Scenario #4
Some people cannot manufacture all of the molecules needed for blood to clot properly. This means that they bleed excessively from the slightest wound. There are "clotting

factors" that can be given to a person with this condition. *[The condition is hemophilia. It is found on the X chromosome.]*

Scenario #5
This condition is not considered to be life threatening but it may prevent you from being a boat captain. It is one of the reasons that stoplights are standardized – that is, the color of the lights always appear in the same order. *[The condition is red-green color blindness. It is carried on the X chromosome.]*

Scenario #6
This life-threatening disease causes individuals to secrete thick mucus that clogs the airways of their lungs and the passages of their pancreas and liver *(Raven & Johnson, 1992). [The disorder is called cystic fibrosis.]*

Scenario #7
There is deterioration of the central nervous system in infancy. There is also a "late onset" form of this condition that occurs between adolescence and the mid-30's. In either case, fat (lipid) accumulates in nerve cells and affects the functioning of the nervous system. *[The disorder is Tay-Sachs disease. Individuals produce a defective form of the enzyme hexosaminidase A. The gene is located on chromosome 15.]*

Scenario #8
There is a wasting away of muscles. *[The condition is Duchenne's muscular dystrophy.]*

Scenario #9
There are excessive cholesterol levels in the blood, leading to heart disease. *[This is hypercholesterolemia and causes an abnormal form of cholesterol cell-surface receptor.]*