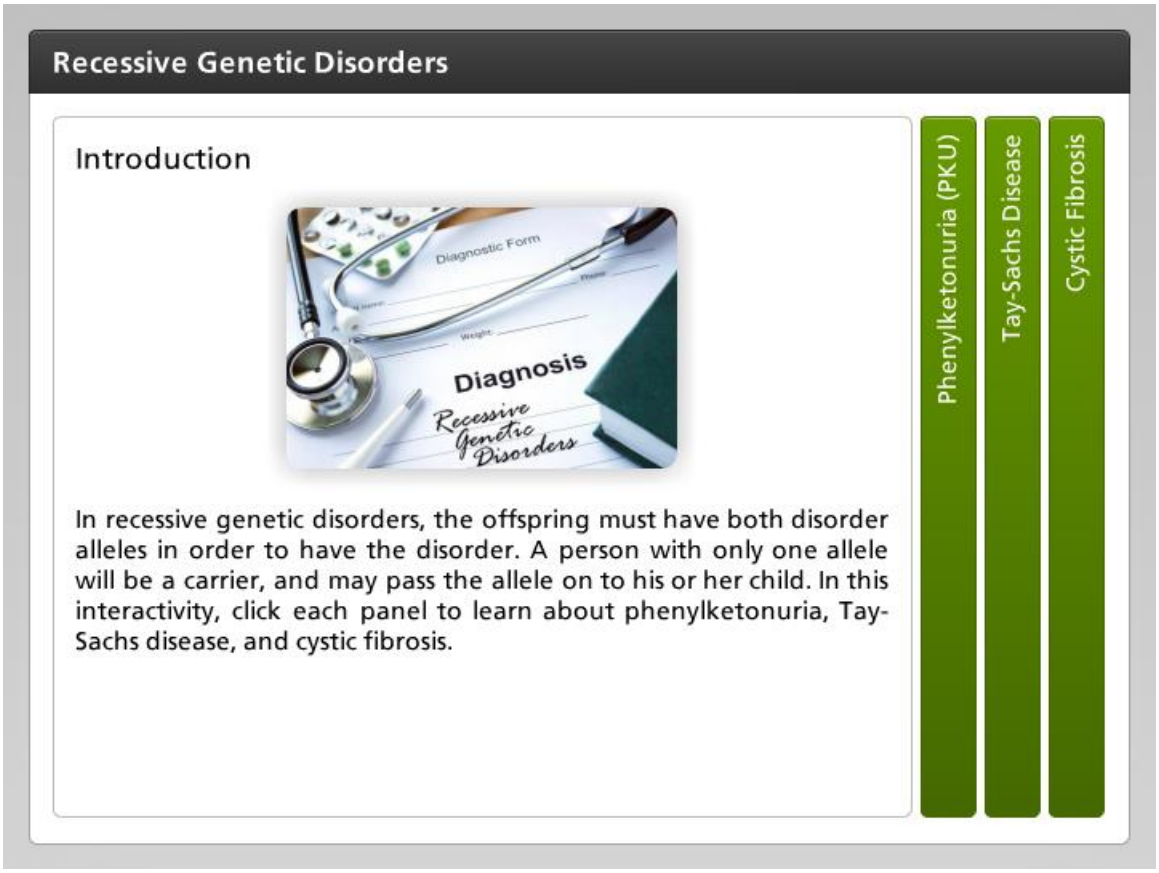
Module 5: Mendelian Genetics and Genetic Disorders

Topic 3 Content: Recessive Genetic Disorders Notes

Introduction



In recessive genetic disorders, the offspring must have both disorder alleles in order to have the disorder. A person with only one allele will be a carrier, and may pass the allele on to his or her child. In this interactivity, click each panel to learn about phenylketonuria, Tay-Sachs disease, and cystic fibrosis.

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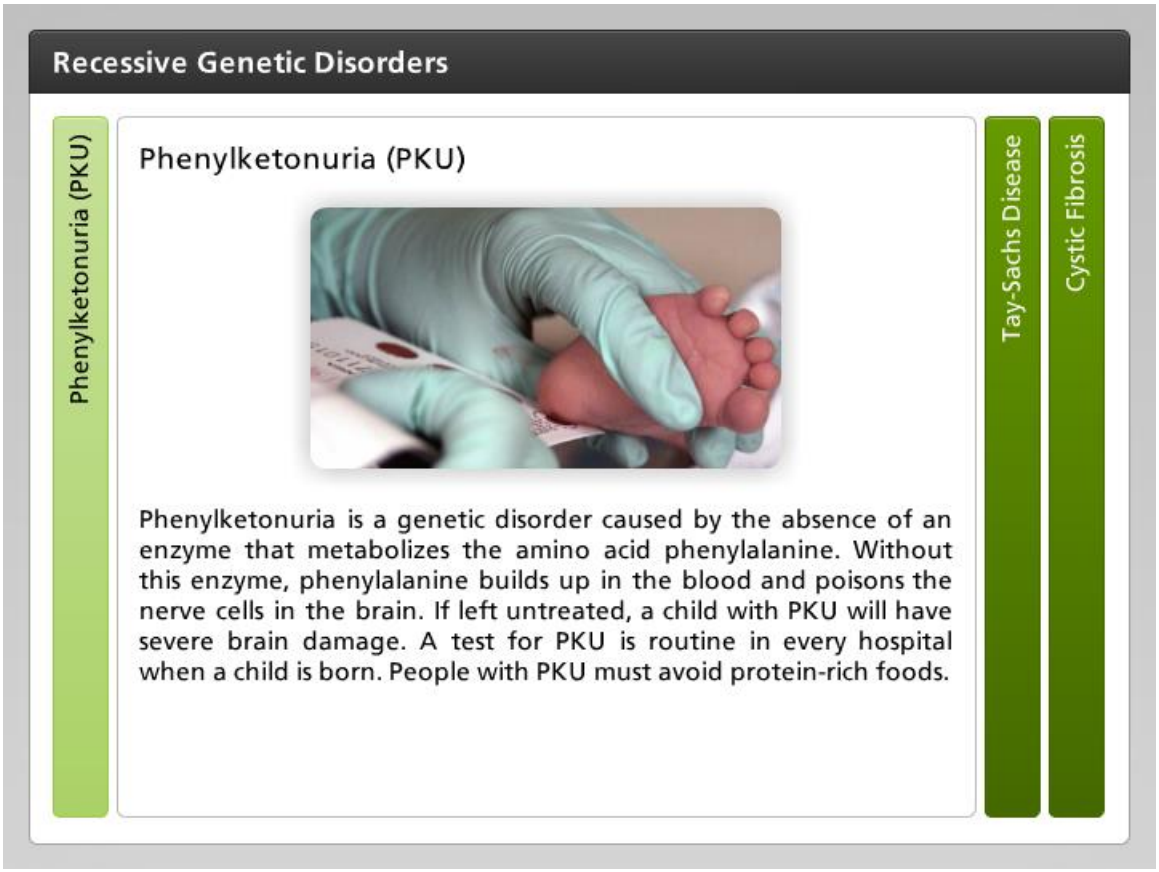
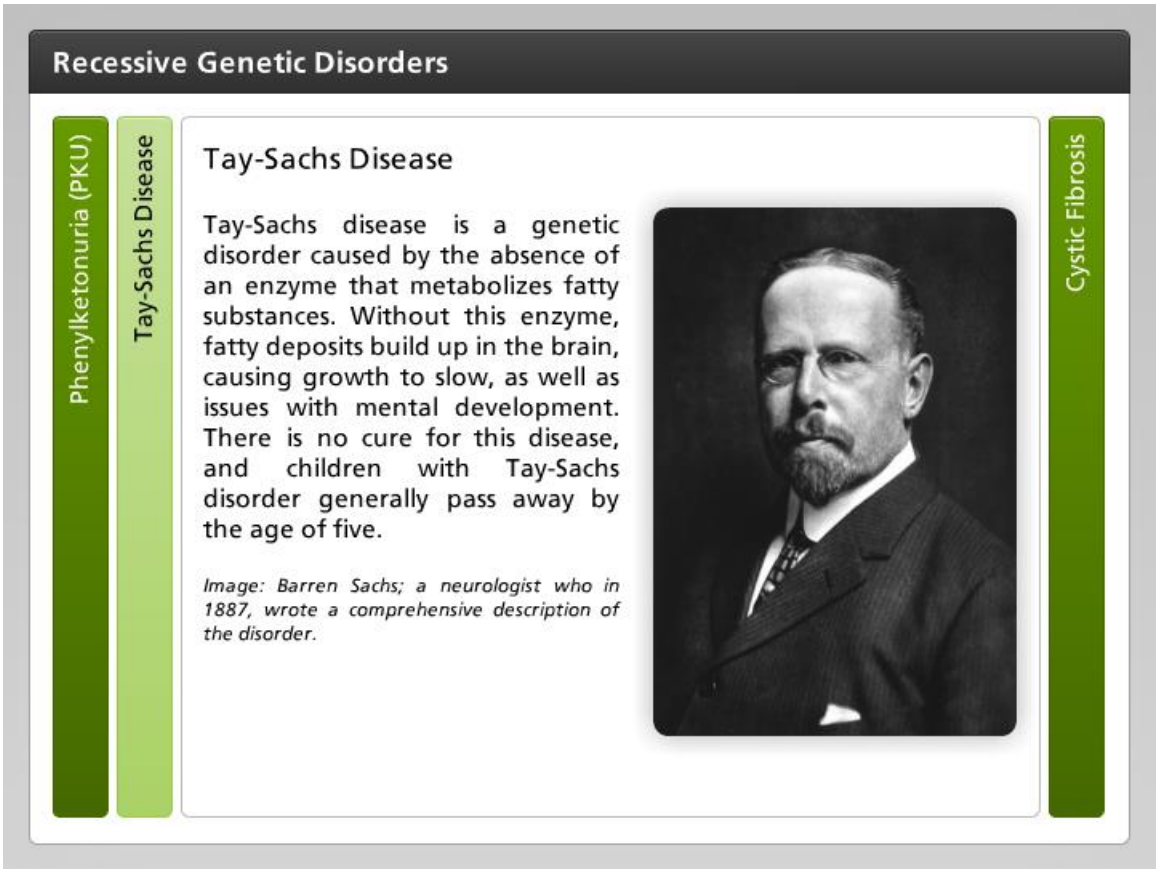
Phenylketonuria (PKU)



Phenylketonuria is a genetic disorder caused by the absence of an enzyme that metabolizes the amino acid phenylalanine. Without this enzyme, phenylalanine builds up in the blood and poisons the nerve cells in the brain. If left untreated, a child with PKU will have severe brain damage. A test for PKU is routine in every hospital when a child is born. People with PKU must avoid protein-rich foods.

Tay-Sachs Disease



Tay-Sachs disease is a genetic disorder caused by the absence of an enzyme that metabolizes fatty substances. Without this enzyme, fatty deposits build up in the brain, causing growth to slow, as well as issues with mental development. There is no cure for this disease, and children with Tay-Sachs disorder generally pass away by the age of five.

Image: Barren Sachs; a neurologist who in 1887, wrote a comprehensive description of the disorder.

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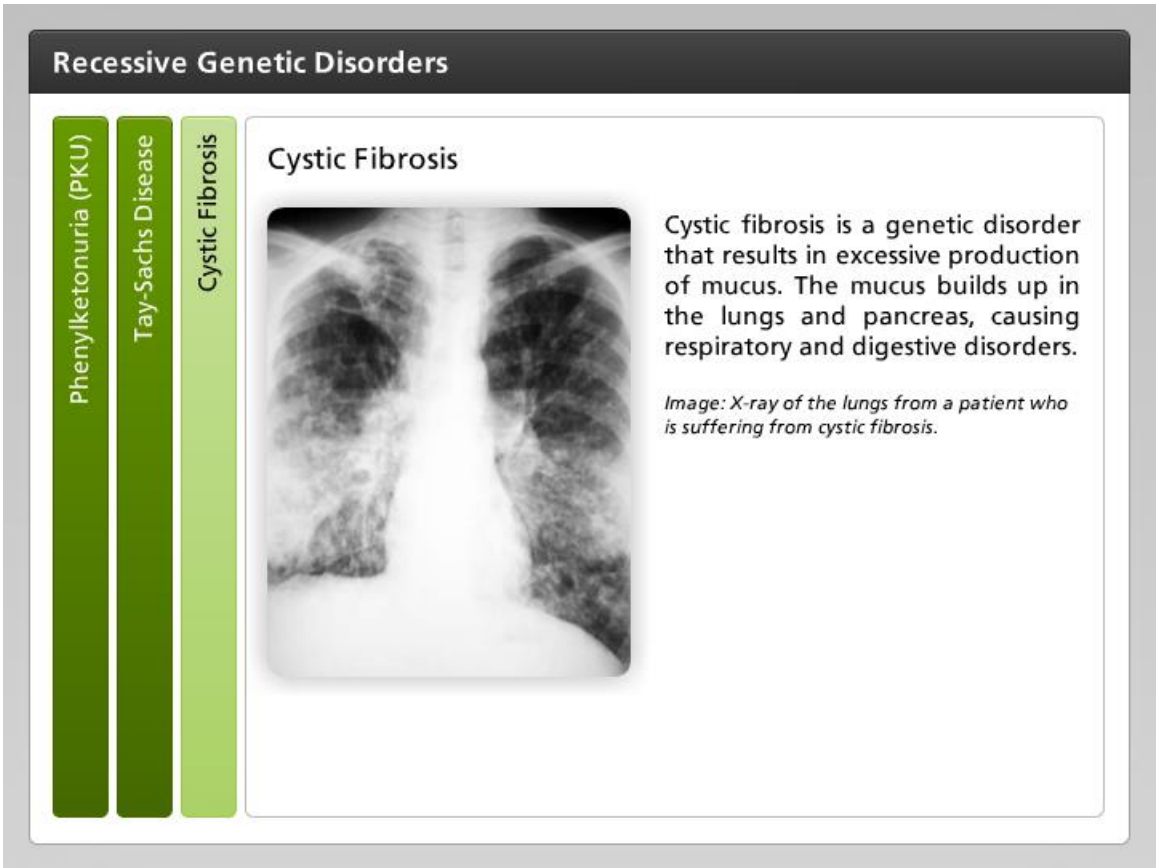
Cystic Fibrosis



Cystic fibrosis is a genetic disorder that results in excessive production of mucus. The mucus builds up in the lungs and pancreas, causing respiratory and digestive disorders.

Image: X-ray of the lungs from a patient who is suffering from cystic fibrosis.