

Name: _____ Date: _____

Life Science Notes 15
Human Genetics: Recessive Genetic Disorders

The Rise of Recessive Disorders

- Genes can mutate from time to time to cause an entirely new trait.

Sometimes they are helpful to an organism, but most often, they are

fatal or harmful

- Over hundreds of thousands of years, many recessive genetic disorders have come into the human gene pool due to mutation of normal genes.
- They are called recessive traits, because to have the disorder, a person must be pure recessive for that particular trait. A person who is hybrid ~~heterozygous~~ is often called a carrier, because even though they do not have the disorder, they can still pass on the recessive allele to their offspring.

Examples of recessive Disorders

1. Sickle-cell Anemia
 - a. Red blood cells pick up and deliver oxygen to every cell in the body through the blood. Their hat-like shape works well for picking up rather large oxygen molecules and holding onto them tight until delivery.



Red Blood Cells



- b. A person who is pure recessive for red-blood cell shape will have sickle-shaped cells. These cells cannot pick up and hold onto Oxygen as well and a person suffering from this disease will likely not live as long as a person with normal alleles.
- c. This disorder is more common among people of African decent than others.

2. Cystic fibrosis

- a. In most people, the body creates a thin mucus to coat the lungs and digestive tract.
- b. People who are ^{pure} homozygous recessive for cystic fibrosis have a thick mucus instead. This thick mucus makes it harder to breathe and harder for the body to absorb Oxygen. People with cystic fibrosis often die in or before their twenties.
- c. This disorder is most common in whites.

Other examples to research on your own

- Phenyl ketonuria (PKU)
 - The inability to metabolize phenylalanine, which is found in many artificial sweeteners.
- Albinism
 - A total lack of pigment.
- Tay Sachs Disease
 - Does not allow proper brain development and patients of the disease are not expected to live beyond the age of 5.