

# Pleiotropy

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# Introduction

- ❑ Pleiotropy refers to the expression of multiple traits by a single gene which may or may not be related. Pleiotropy was first noticed by geneticist Gregor Mendel, who noticed that plant flower color (white or purple) was always related to the color of the leaf axil (area on a plant stem consisting of the angle between the leaf and upper part of the stem) and seed coat.
- ❑ The study of pleiotropic genes is important to genetics as it helps us to understand how certain traits are linked in genetic diseases.
- ❑ Phenotypes are traits that are physically expressed such as color, body shape, and height. It is often difficult to detect which traits may be the result of pleiotropy unless a mutation occurs in a gene. Because pleiotropic genes control multiple traits, a mutation in a pleiotropic gene will impact more than one trait.
- ❑ Typically, traits are determined by two alleles (variant form of a gene). Specific allele combinations determine the production of proteins which drive the processes for the development of phenotypic traits. A mutation occurring in a gene alters the DNA sequence of the gene. Changing gene segment sequences most often results in non-functioning proteins. In a pleiotropic gene, all of the traits associated with the gene will be altered by the mutation.

# Forms of Pleiotropy

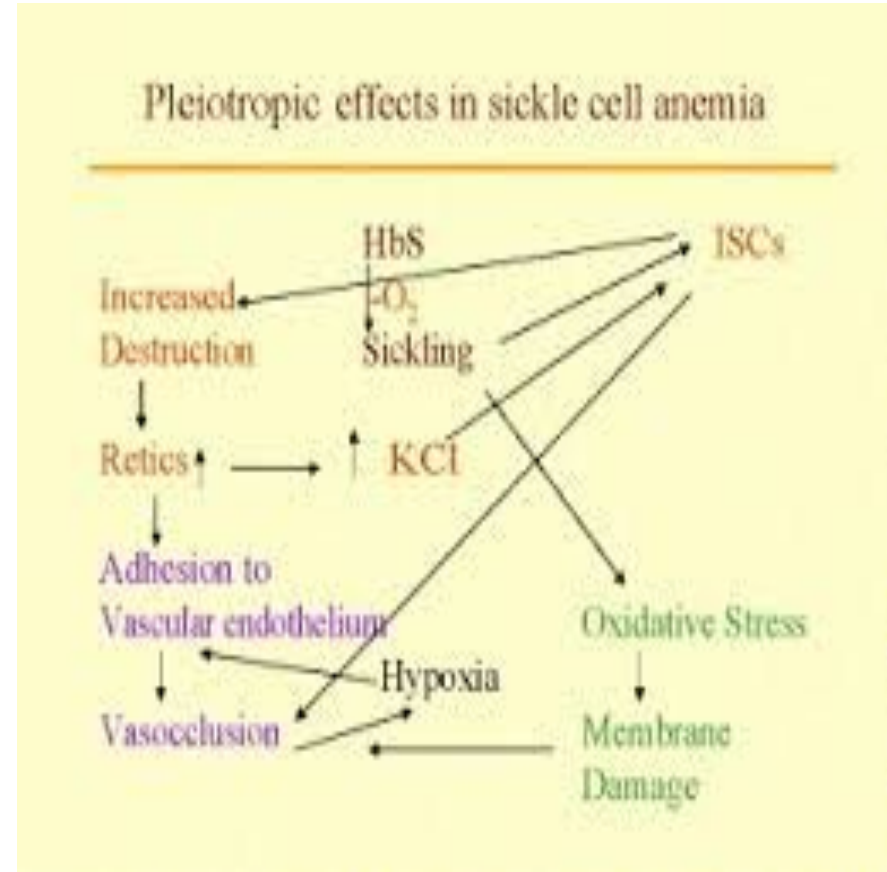
- ★ **Gene pleiotropy**, also referred to as molecular-gene pleiotropy, focuses on the number of functions of a particular gene. The functions are determined by the number of traits and biochemical factors impacted by a gene. Biochemical factors include the number of enzyme reactions catalyzed by the protein products of the gene.
- ★ **Developmental pleiotropy** focuses on mutations and their influence on multiple traits. The mutation of a single gene manifests in the alteration of several different traits. Diseases involving mutational pleiotropy are characterized by deficiencies in multiple organs that impact several body systems.
- ★ **Selectional pleiotropy** focuses on the number of separate fitness components affected by a gene mutation. The term fitness relates to how successful a particular organism is at transferring its genes to the next generation through sexual reproduction. This type of pleiotropy is concerned only with the impact of natural selection on traits.
- ★ **Antagonistic pleiotropy** is focused on the prevalence of gene mutations that have advantages early in life and disadvantages later in life.

# Antagonistic Pleiotropy

- Explains how senescence, or biological aging, can be attributed to natural selection of certain pleiotropic alleles.
- In antagonistic pleiotropy, an allele that has a negative impact on an organism can be favored by natural selection if the allele also produces advantageous effects.
- Antagonistically pleiotropic alleles increase reproductive fitness early in life but promote biological aging later in life and tend to be selected for by natural selection.
- The positive phenotypes of the pleiotropic gene are expressed early when reproductive success is high, while the negative phenotypes are expressed later in life when reproductive success is low.
- Sickle cell trait is an example of antagonistic pleiotropy in that the Hb-S allele mutation of the hemoglobin gene provides advantages and disadvantages for survival. Those who are homozygous for the Hb-S allele, meaning that they have two Hb-S alleles of the hemoglobin gene, have a short life span due to the negative impact (damage to multiple body systems) of the sickle cell trait. Those who are heterozygous for the trait, meaning that they have one Hb-S allele and one normal allele of the hemoglobin gene, do not experience the same degree of negative symptoms and show resistance to malaria.

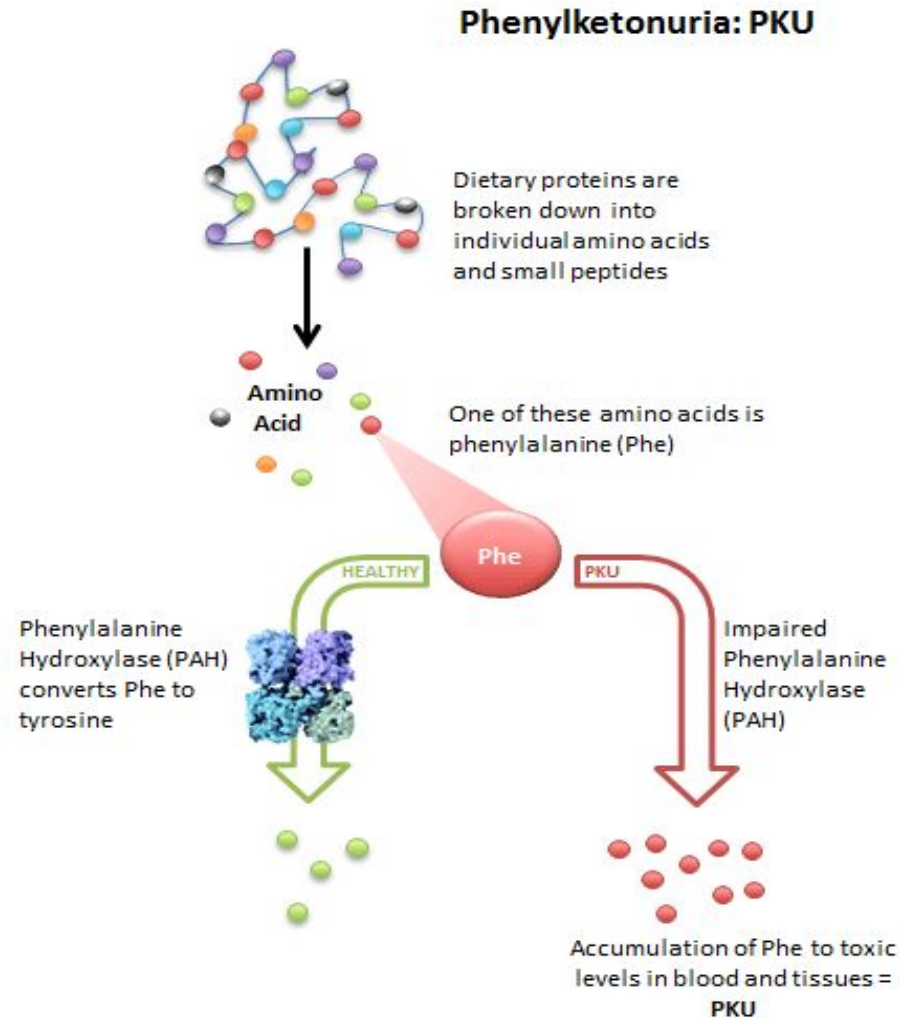
# Pleiotropy- Sickle Cell Disease

- Occurs in humans
- Results from the development of abnormally shaped red blood cells. Normal red blood cells have a biconcave, disc-like shape and contain enormous amounts of a protein called hemoglobin. Hemoglobin helps red blood cells bind to and transport oxygen to cells and tissues of the body.
- Sickle cell is a result of a mutation in the beta-globin gene
- Red blood cells become sickle-shaped, which causes them to clump together and become stuck in blood vessels, blocking normal blood flow.
- The single mutation of the beta-globin gene results in various health complications and causes damage to multiple organs including the heart, brain, and lungs.



# Pleiotropy- PKU

- ❖ **Phenylketonuria (PKU)**, is another disease resulting from pleiotropy.
- ❖ Caused by a mutation of the gene responsible for the production of an enzyme called phenylalanine hydroxylase.
- ❖ This enzyme breaks down the amino acid phenylalanine that we get from protein digestion.
- ❖ Without this enzyme, levels of the amino acid phenylalanine increase in the blood and damage the nervous system in infants.
- ❖ PKU disorder may result in several conditions in infants including intellectual disabilities, seizures, heart problems, and developmental delays.



# Pleiotropy- Frizzled Feather Trait

- ★ This an example of pleiotropy seen in chickens.
- ★ Chickens with this particular mutated feather gene display feathers that curl outward as opposed to lying flat.
- ★ In addition to curled feathers, other pleiotropic effects include a faster metabolism and enlarged organs.
- ★ The curling of the feathers leads to a loss of body heat requiring a faster basal metabolism to maintain homeostasis.
- ★ Other biological changes include higher food consumption, infertility, and sexual maturation delays.



Thank you