Pleiotropy

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Pleiotropy

The term "pleiotropie" was coined in a 1910 by Festschrift .
The term pleiotropy comes from the Greek pleion, meaning "more", and tropos, meaning “character“

Pleiotropy

Defination-

- Pleiotropism is defined as when one gene influences multiple, unrelated phenotypic traits .
- Pleiotropy describes the genetic effect of a single gene on multiple phenotypic traits .
- Mutation in a pleiotropic gene may have an effect on some or all traits
- Mechanism of pleiotropy in most cases is the effect of a gene on metabolic pathways that contribute to different phenotypes.

Pleiotropy

- Mechanism of pleiotropy in most cases is the effect of a gene on metabolic pathways that contribute to different phenotypes.
- Genes showing pleiotropy produce a single polypeptide just like other non pleiotropic genes .
- But their polypeptide governs such a biochemical reaction, which is basic to many developmental events.
- As a result impairment of the function.

Pleiotropy

Example Of pleiotrpy:

Phenylketonuria:

- A classic example of pleiotropy is the human disease phenylketonuria (PKU). .
- This disease can cause mental retardation and reduced hair and skin pigmentation.
- phenylketonuria is due to mutation in a single gene that codes for the enzyme phenylalanine hydroxylase .
- Phenylalanine hydroxylase converts the amino acid phenylalanine to tyrosine
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Pleiotropy

Phenylketonuria(cont....)

- Due to mutation, conversion of phenylalanine to tyrosine is reduced or ceases entirely.
- Phenylalanine in the bloodstream is toxic to the developing nervous system of newborn and infant children and it causes mental retardation.
- Where as tyrosine is used by the body to make melanin (an important component of the pigment found in hair and skin) .
- The failure to convert normal levels of phenylalanine to tyrosine results in less pigmentation of hair and skin.

Pleiotropy

Phenylketonuria(contd....)

- Where as tyrosine is used by the body to make melanin an important component of the pigment found in hair and skin .
- The failure to convert normal levels of phenylalanine to tyrosine results in less pigmentation of hair and skin.
- Thus Without the enzyme phenylalanine hydroxylase, levels of the amino acid phenylalanine increase in the blood and damage the nervous system in infants.
- And also result in several conditions in infants including intellectual disabilities, seizures, heart problems, and developmental delays etc.

Pleiotropy

- **sickle cell disease:**
- Another example of pleiotropy is sickle cell disease.
- Sickle cell disorder 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.

Pleiotropy

- Hemoglobin helps red blood cells to bind and transport oxygen to cells and tissues of the body.
- Sickle cell is a result of a mutation in the beta-globin gene.
- This mutation results in red blood cells that are 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.

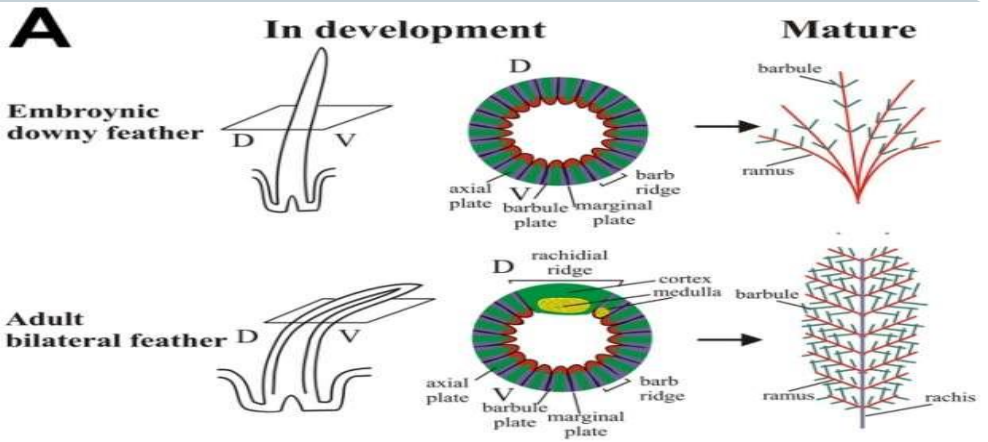
Pleiotropy

Frizzled feather trait:

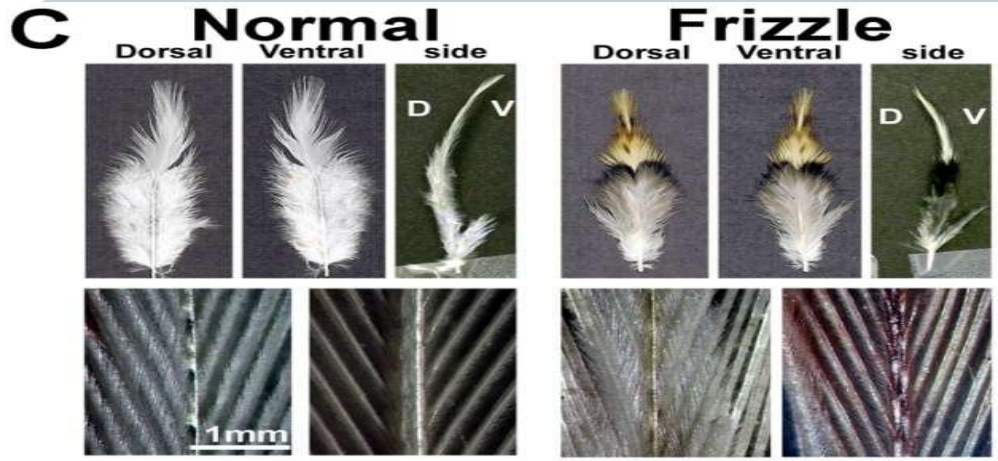
The frizzled feather trait is an example of pleiotropy seen in chickens.

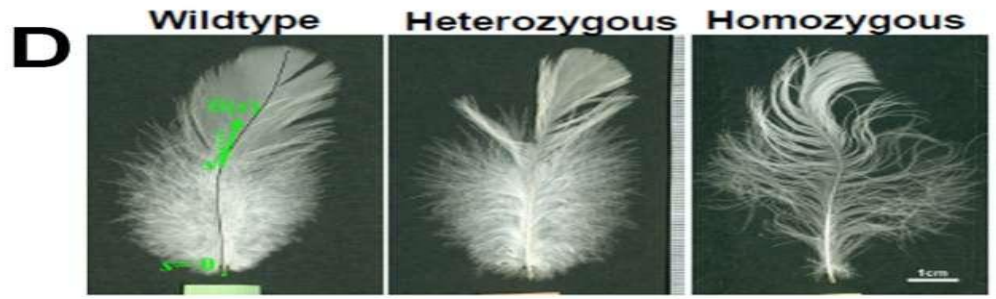
- Chickens with this particular mutated feather gene display feathers that curl outward as opposed to lying flat.
- In addition to curled feathers, other pleiotropic effects include a faster metabolism and enlarged organs.
- The curling of the feathers leads to a loss of body heat requiring a faster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

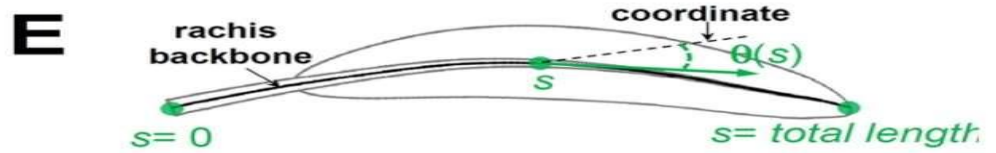
Pleiotropy


A
In development
Mature
Embroynic
downy feather
D
V
barbule
ramus
axial
plate
barbule
plate
marginal
plate
barb
ridge
Adult
bilateral feather
rachidial
ridge
cortex
medulla
rachis

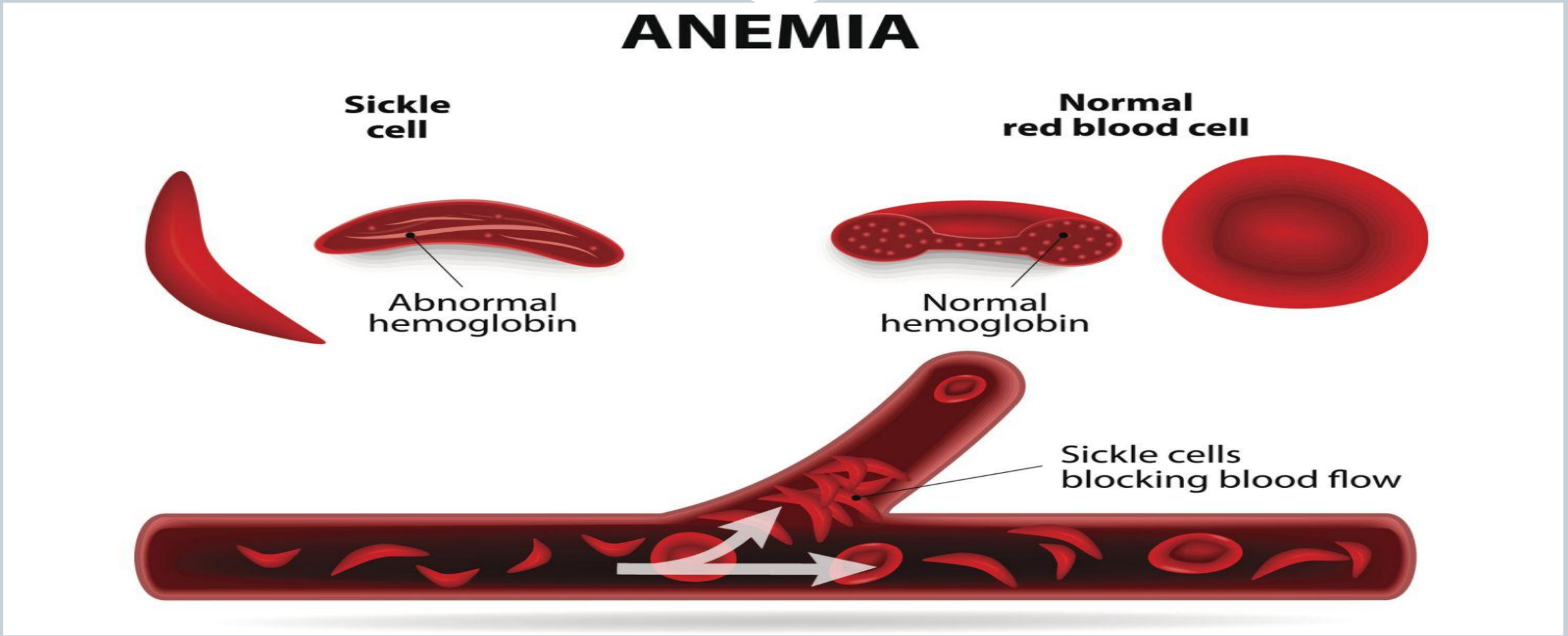

B
Adult Frizzle
Frizzle newborn
Frizzle 1 month old
wing
body
Frizzle 1 month old


C
Normal
Frizzle
Dorsal
Ventral
side
1mm


D
Wildtype
Heterozygous
Homozygous


E
rachis
backbone
coordinate
θ(s)
s
s= 0
s= total length

Pleiotropy


ANEMIA
Sickle cell
Normal red blood cell
Abnormal hemoglobin
Normal hemoglobin
Sickle cells blocking blood flow

Thank You