Mendel's Laws Not Perfect

- Shortly people began to notice that not all traits are "Mendelian"
  - –This means, they do NOT follow **Mendel's laws**

Mendel's Laws Not Perfect

–Was he just plain wrong?

- Truth is, his laws are correct and did explain how genetics works
  - – Real life is just more complicated than peas!

Altering Mendel's Ratios

Two different types of complications:

1. Genotypic ratios follow Mendel's laws, but phenotypes do not
    - Somehow the underlying genotypic ratios are hidden

2. Mendel's laws do not apply
    - Both genotypes and phenotypes are not following Mendel's laws

Incomplete dominance

- Neither allele is dominant and heterozygous individuals have an intermediate phenotype.

- For example, A cross between red and white flowered plants produced plants with intermediate flower colour i.e. pink colour in F1 and a modified ratio of 1 red: 2 pink: 1 White in F2.

Incomplete Dominance

C^RC^R

F_1 generation

All C^RC^W

C^WC^W

Gametes

C^R C^W

C^R

Gametes

C^W

	C^R	C^W
C^R	 C^RC^R	 C^RC^W
C^W	 C^RC^W	 C^WC^W

F_2 generation

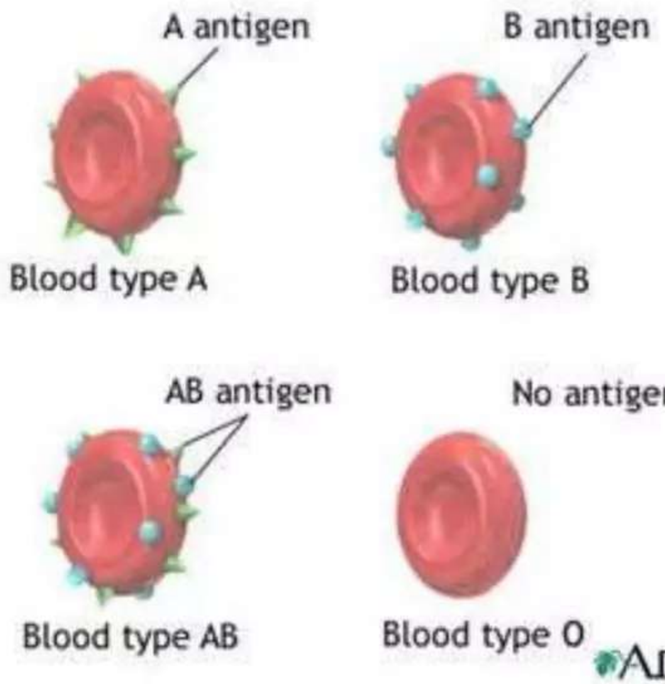
1 : 2 : 1

Hair Type

- **Straight Hair**: Homozygous: Incomplete Dominant
- **Curly Hair**: Homozygous: Incomplete Dominant
- **Wavy Hair**: Heterozygous: A mix (Straight and Curly)

CoDominance

- Neither allele is dominant
- Heterozygote expresses both alleles equally
- In flowers a red and white flower
- ABO blood groups



Co-dominance

Example : human blood type

3 different alleles (= versions of genes coding for one trait) include:

- I^A
- I^B
- -and i

that code for A, B, and “O” blood types.

Genotype → Phenotype

- I^Ai or I^AI^A → Type A blood

- I^Bi or I^BI^B → Type B blood

- ii → Type O Blood

- I^AI^B → Type AB blood (Co-dominant)

Blood type A

AO

Blood type B

BO

A allele

B allele

Codominance

O allele — Recessive

AO

AB

BO

OO

Blood type A

Blood type AB

Blood type B

Blood type O

Polygenic Traits

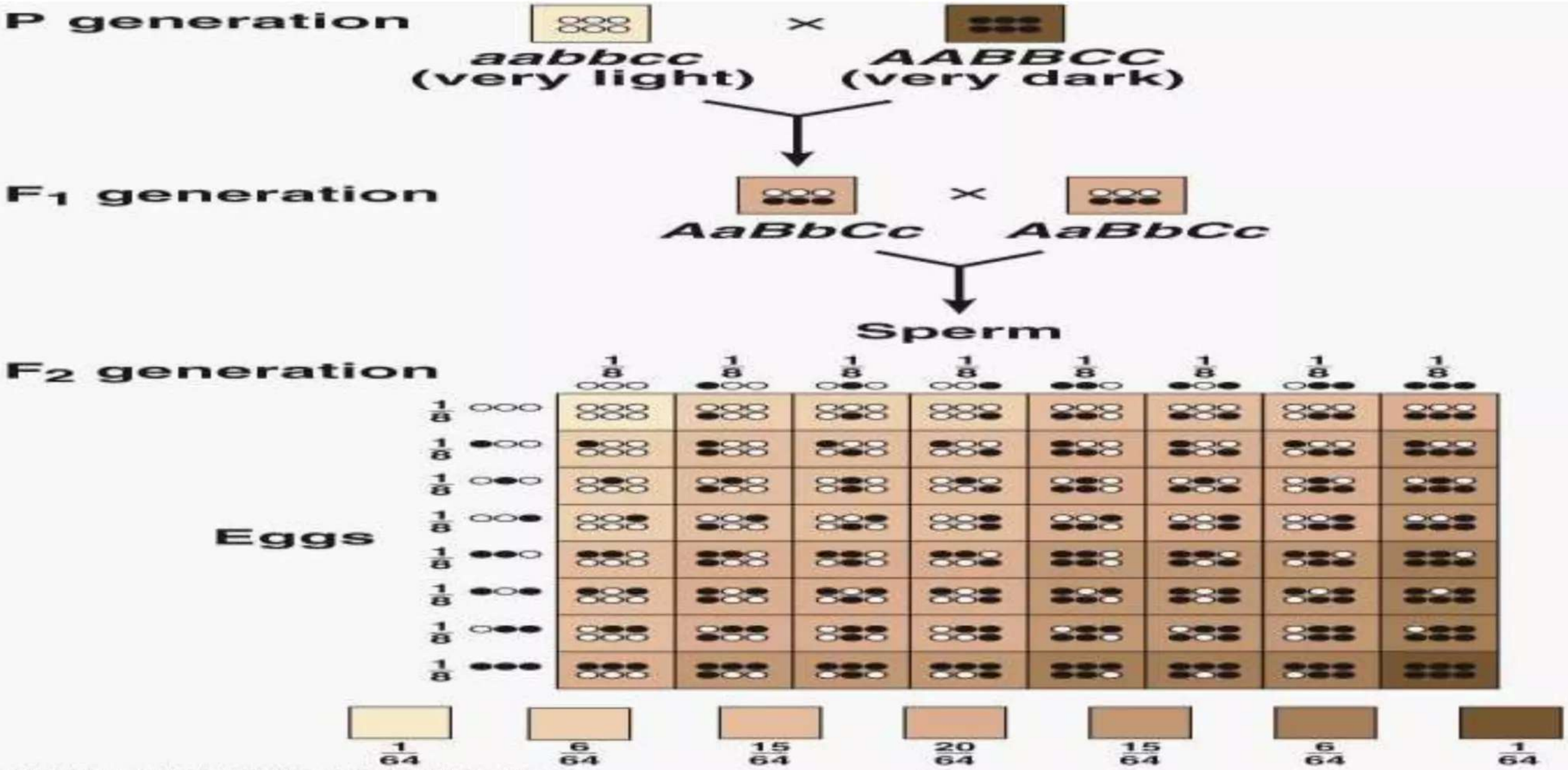
- traits are not controlled by a single gene locus, but by the combined interaction of many gene loci.

- characters show a range of continuous phenotypes instead of discrete, defined phenotypes

- In this case many genes have an additive effect. The characteristic or trait is the result of the combined effect of several genes.

Ex: human skin color, eye color, hair color and height.

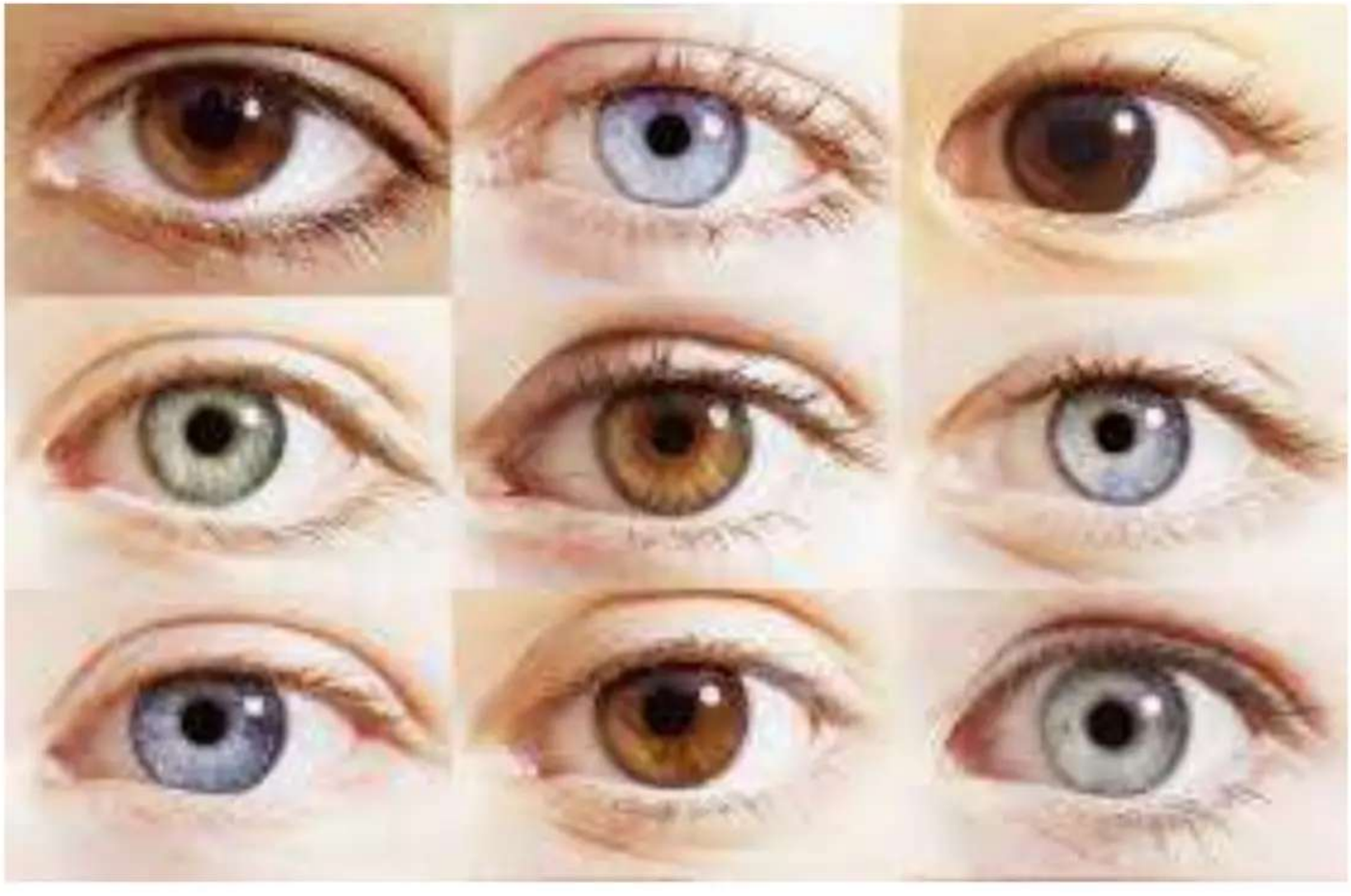
Pigmentation in humans is controlled by at least three (3) separately inherited genes.

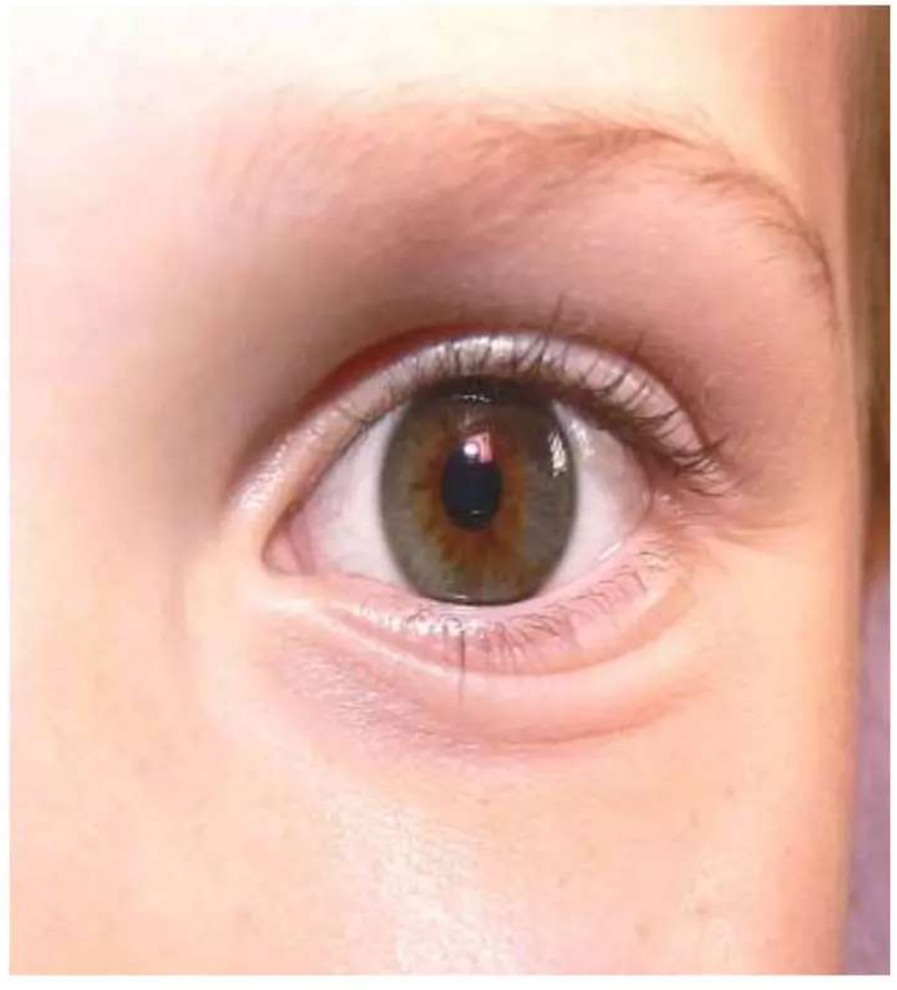


A human rainbow of skin colors
Photograph by Sarah Leen

NATIONAL GEOGRAPHIC

Eye color is influenced by many genes coding for different kinds of pigment as well as where in the iris those pigments are found .





Human Eye Color

Mother

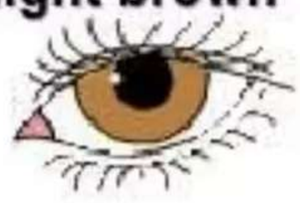
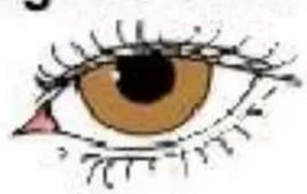
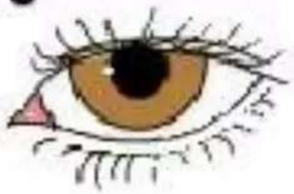
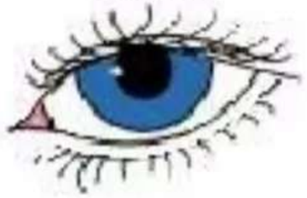
AaBb

Father

AaBb

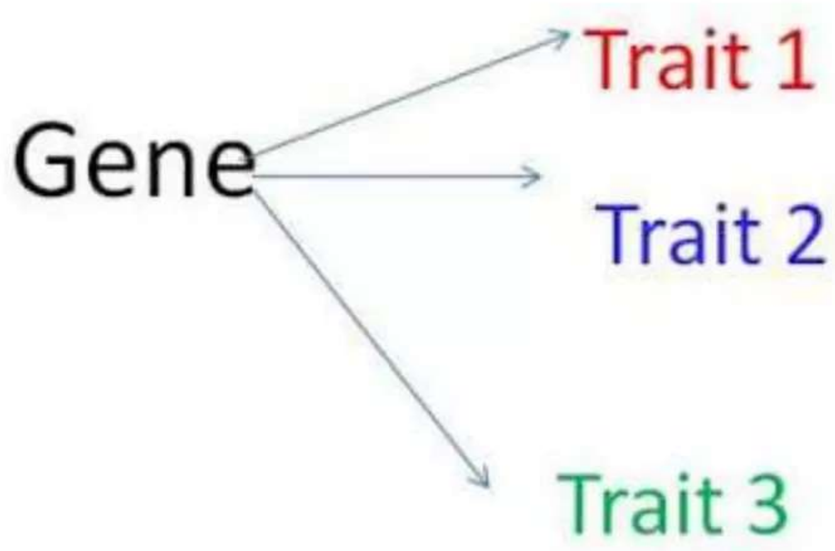
SPERM

EGGS

	AB	Ab	aB	ab
AB	black  AABB	dark brown  AABb	dark brown  AaBB	light brown  AaBb
Ab	dark brown  AAbB	light brown  AAbb	light brown  AabB	blue  Aabb
aB	dark brown  aABB	light brown  aABb	light brown  aaBB	blue  aaBb
ab	light brown  aABb	blue  aABb	blue  aaBb	light blue  aabb

Pleiotropy

- Alleles at a single locus may have effects on two or more traits.
- When a single gene affects the expression of more than one phenotypic character, the phenomenon is known as **pleiotropy** (from the Greek *pleio* meaning "more" and "*trop*, meaning "change").



Examples of pleiotropsm

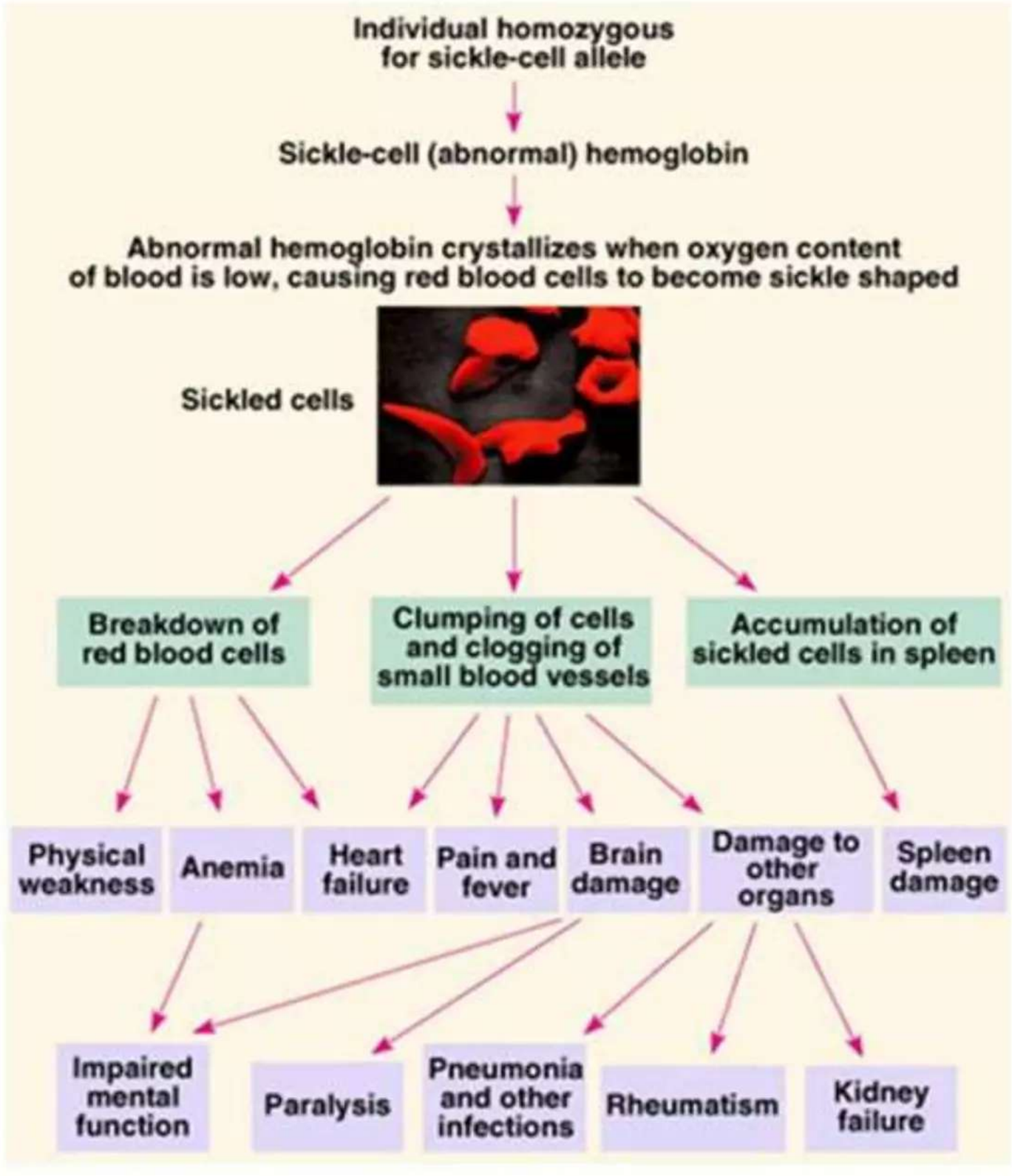
- **<u>Sickle cell disease</u>**
- Homozygotes have two copies of abnormal gene leading to formation of crescent shaped or sickle shaped hemoglobin (Hbs).
- These cells have very low oxygen carrying capacity. Thus impair circulation. This often led to renal failure, cardiac failure and thrombosis.

Pleiotropic Effects Seen if Hb^S present

Hb^AHb^A = normal

Hb^AHb^S = sickle cell trait

Hb^SHb^S = sickle cell disease/anemia



- **Phenylketonuria**
- One of the most widely examples in pleiotropy in humans

- Inborn metabolic disorder in which the homozygous recessive individual lacks the enzyme **Phenylalaine hydroxylase** needed to change phenylalanine (amino acid) to tyrosine (amino acid).
- Lack of the enzyme phenylalanine hydroxylase is due to the abnormal gene on chromosome 12.

- In children, symptoms are mental retardation, decrease pigmentation of hair and skin and eczema.
- The heterozygous individuals are normal but carriers.

Clinical manifestations of PKU

- Mental retardation
- seizure, irritability
- abnormal EEG
- fair skin
- unpleasant odor of urine

Epistasis

- One gene effecting or masking another gene
- or
- •Two genes controlling same phenotype

- •Mendel's Laws are still working for each individual gene, but phenotype is not determined by that single gene's genotype alone

Red hair

- Let's look at the example of red hair color in human beings.
- **Blond, brown**, and **black** hair in humans is determined by the amount of a pigment called **eumelanin**.
- Without much eumelanin, you get blond hair; an intermediate amount will be brown; a lot will get you black hair.

- A second pigment called **pheomelanin** makes red hair. But usually people don't build up a lot of that because of a totally different gene called ***MCR1*.**

- *MCR1* allows the conversion of pheomelanin into eumelanin, making redheadedness rare. The genes for eumelanin and MCR1 interact to produce one single phenotype.

- So why do we ever get redheads?

- Well, a certain variation of the *MCR1* gene will **stop the conversion of pheomelanin into eumelanin**, allowing the build up of pheomelanin in the hair, which leads to red hair.

- **<u>Albinism</u>**
- Albinism, a lack of pigment resulting in pale white individuals, is another example of epistasis.

- The albino condition occurs due to an entirely different gene than the genes that encode skin color and tone.

- If the albinism gene is present, the organism will not have any pigment - no matter what skin color is encoded by other genes.

- an important protein named **tyrosinase** is necessary for the production of the **pigment melanin**.

- A gene named ***TYR*** codes for **tyrosinase**.

- A variation of the ***TYR*** gene has a mutation that code for a non-functional tyrosinase that **dosent work**.

- If a person only has nonfunctional **tyrosinase**, then no melanin will be made in the body. Even though that person's genetic code might have genes for dark skin, if there is no pigment to make it with, the person will be an albino.

Effects of Environment on Phenotype

- Genes play an important role in determining an organism's characteristics.

- However, for many characteristics, the individual's phenotype is influenced by other factors as well.

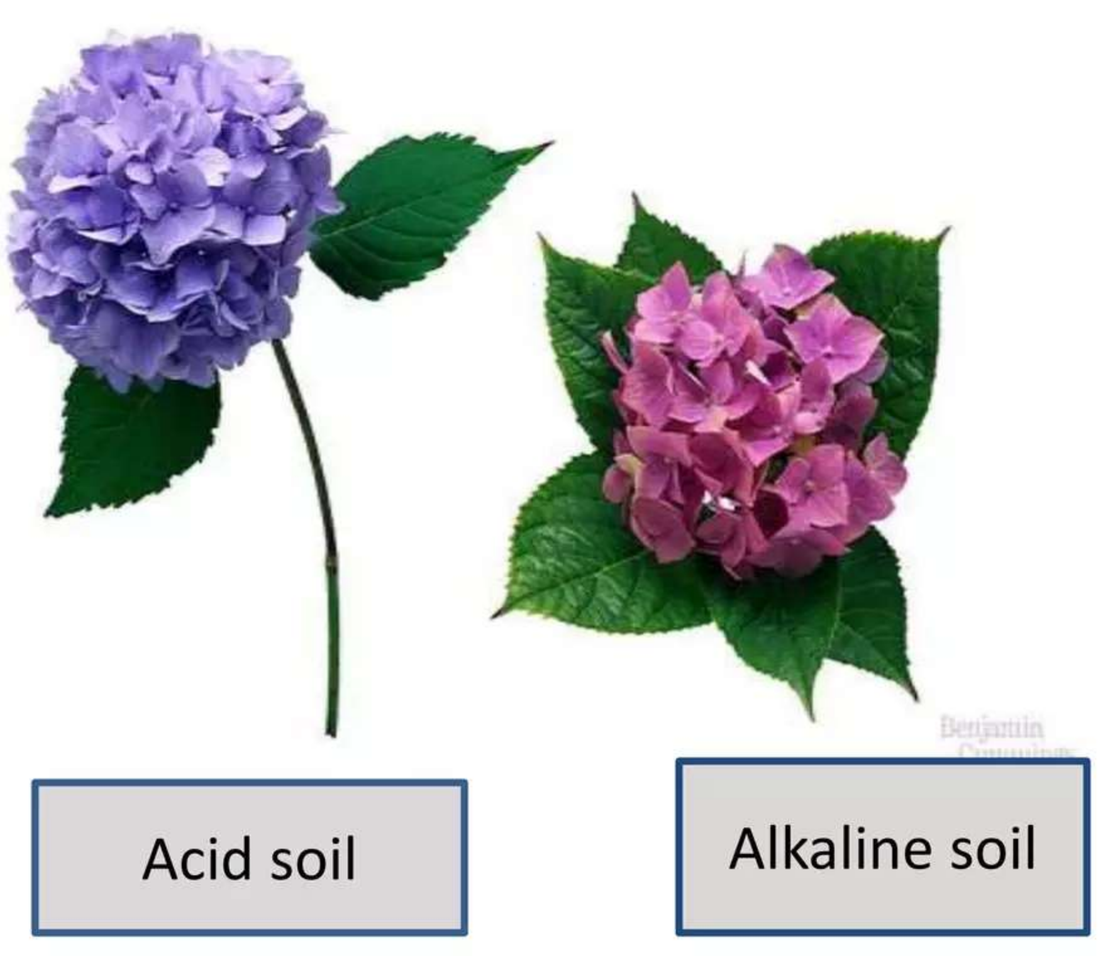
- Environmental factors, such as sunlight and food availability, can affect how genes are expressed in the phenotype of individuals.

Environmental Effects on Plant Phenotype

- *Hydrangea macrophylla*

- Action of gene responsible for floral color is influenced by soil acidity

- Flower color ranges from pink to blue



Acid soil

Alkaline soil

in human:

- Genes are a major determinant of human skin color.

- However, exposure to ultraviolet radiation can increase the amount of pigment in the skin and make it appear darker.