

Methods in behavioral genetics

Are all genes Mendelian ones?

The Mendel's rules had been discovered in experiments where strict dominance - recessivity gene relationships are clear.

After Mendel there were found out another gene states and interactions.

Multiple Alleles (ABO Blood Types) and Punnett Squares

<https://www.youtube.com/watch?v=9O5JQqIngFY>

The first example of multi-allele genes was given by Karl Landsteiner in 1900 discovered four blood groups of ABO system caused by combinations of three genes: I^A , I^B and i .

Genotypes and phenotypes in ABO system

Blood group	1	2	3	4
Phenotype	O	A	B	AB
Genotype	ii	$I^A I^A, I^A i$	$I^B I^B, I^B i$	$I^A I^B$

A test was done to determine the biological father of a child. The child's blood Type is A and the mother's is B. A man #1 has a blood type of O, and a man #2 has blood type AB. Who of them is the biological father?

Try to fill in all gaps in the table to find all probable blood groups of children in different combinations of parental ones.

Phenotype ♂	♀	O	A		B		AB
	Genotype	ii	I ^A i	I ^A I ^A	I ^B i	I ^B I ^B	I ^A I ^B
O	ii						
A	I ^A i						
	I ^A I ^A						
B	I ^B i						
	I ^B I ^B						
AB	I ^A I ^B						

Try to fill in all gaps in the table to find all probable blood groups of children in different combinations of parental ones.

Phenotype	♀	♂	A		B		AB
♂	Genotype	ii	$I^A i$	$I^A I^A$	$I^B i$	$I^B I^B$	$I^A I^B$
O	ii	ii	Ii $I^A i$	$I^A i$	Ii $I^B i$	$I^B i$	$I^A i$ $I^B i$
A	$I^A i$	Ii $I^A i$	ii $I^A i$ $I^A I^A$	$I^A i$ $I^A I^A$	ii $I^A i$ $I^B i$ $I^A I^B$	$I^B i$ $I^A I^B$	$I^A i$ $I^A I^A$ $I^B i$ $I^A I^B$
	$I^A I^A$	$I^A i$	$I^A i$ $I^A I^A$	$I^A I^A$	$I^A i$ $I^A I^B$	$I^A I^B$	$I^A I^A$ $I^A I^B$
B	$I^B i$	Ii $I^B i$	ii $I^A i$ $I^B i$ $I^A I^B$	$I^A i$ $I^A I^B$	ii $I^B i$ $I^B I^B$	$I^B i$ $I^B I^B$	$I^A i$ $I^B i$ $I^B I^B$ $I^A I^B$
	$I^B I^B$	$I^B i$	$I^B i$ $I^A I^B$	$I^A I^B$	$I^B i$ $I^B I^B$	$I^B I^B$	$I^B I^B$ $I^A I^B$
AB	$I^A I^B$	$I^A i$ $I^B i$	$I^A i$ $I^A I^A$ $I^B i$ $I^A I^B$	$I^A I^A$ $I^A I^B$	$I^A i$ $I^B i$ $I^B I^B$ $I^A I^B$	$I^B I^B$ $I^A I^B$	$I^A I^A$ $I^B I^B$ $I^A I^B$

Incomplete Dominance, Codominance, Polygenic Traits, and Epistasis!

<https://www.youtube.com/watch?v=YJHGfbW55l0>

Examples of Incomplete Dominance In Humans

A child born to a parent with straight hair and a parent with curly hair will usually have **wavy hair**, or hair that is a little curled, due to the expression of both curly and straight alleles.

Incomplete dominance can be seen in many other physical characteristics such as **skin color, height, hand size, and vocal pitch**.

Carriers of Tay-Sachs disease also show incomplete dominance.

The disease is caused by mutations are found on the **hexosaminidase enzyme** gene. Tay-Sachs causes **nerve cells to deteriorate** over time, which in turn results in the **decline of physical and mental functioning**.

Both child and adult-onset forms of the disease occur, and **children** with the disease **usually die before the age of four**. About 1 in 320,000 newborns in the **United States** develop Tay-Sachs. It occurs in higher frequencies in **Ashkenazi Jews, Cajuns, and French Canadians** (about 1 in 3500 in these populations), although the mutations associated with the disease are different in each population.

There is currently **no treatment or cure**.

Sickle-cell anemia is a recessive disorder caused by a single substitution in the gene that creates hemoglobin, which carries oxygen in the blood.

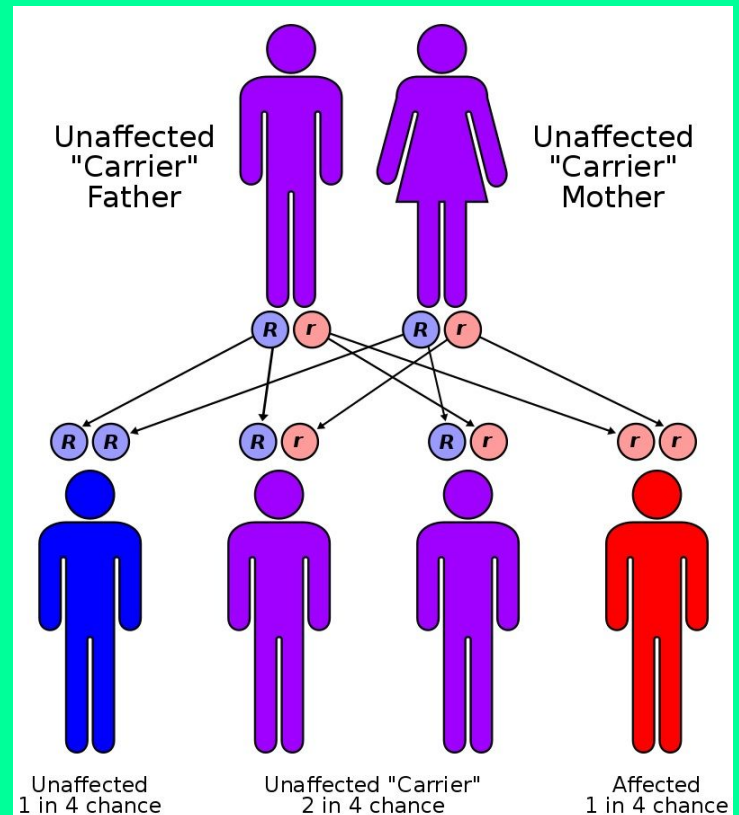
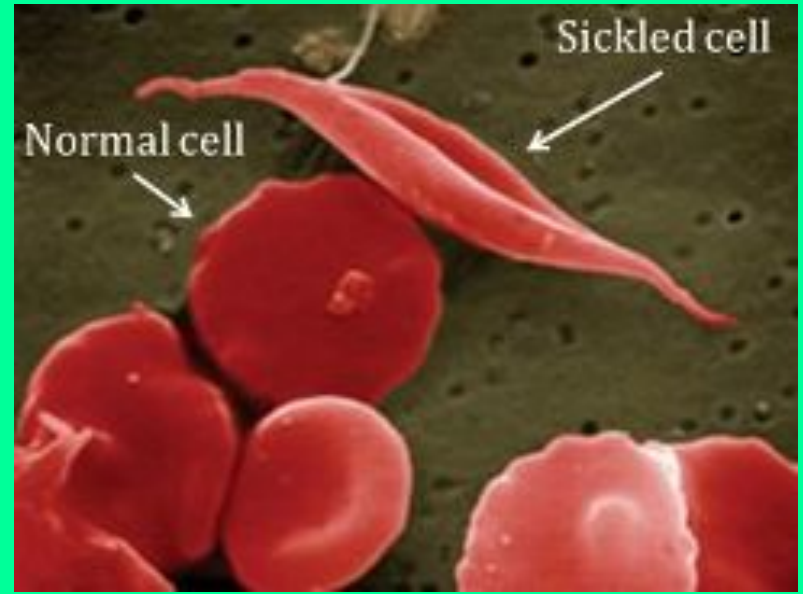
The reason to this disease is replacing of glutamic acid by valine in hemoglobin alpha-chain.

When people have **two copies** of changed gene, it results in **thin sickle-shaped** blood cells that sometimes cannot carry oxygen properly.

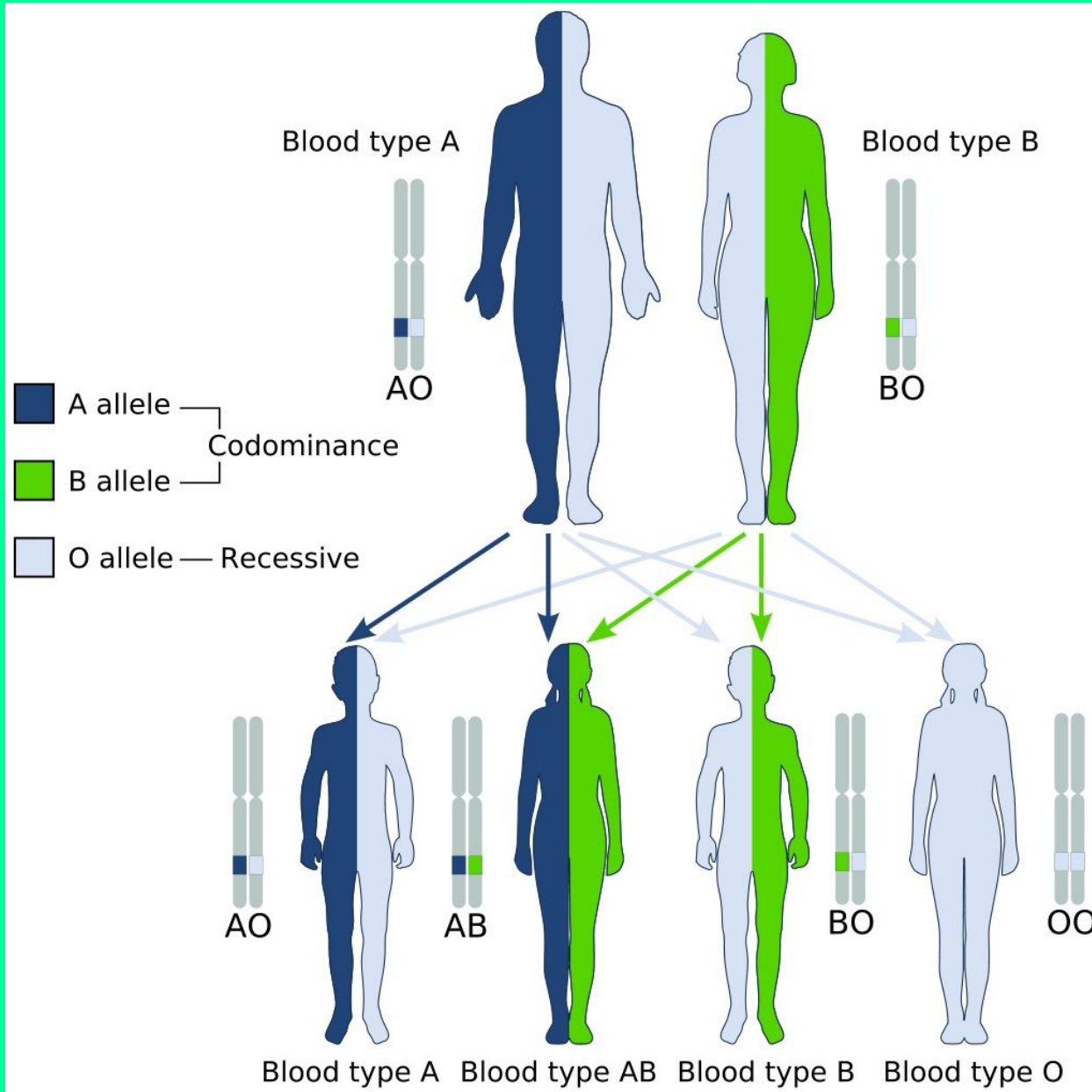
About **80%** of people with sickle-cell disease are in **sub-Saharan Africa**, where being a **carrier for sickle-cell anemia** (having only one copy of the gene, not two) actually helps **protect against malaria**.

It is also found in other parts of the world such as **India** and the **Middle East**, and affects about **1 in 500 African Americans**.

Symptoms include **anemia**, **obstruction of blood vessels**, and **chest pain**, and it is treated with **folic acid**, **blood transfusions**, **bone marrow transplants**, and certain **prescription drugs**.



Examples of Codominance In Humans



Examples of Multiplied Alleles In Humans

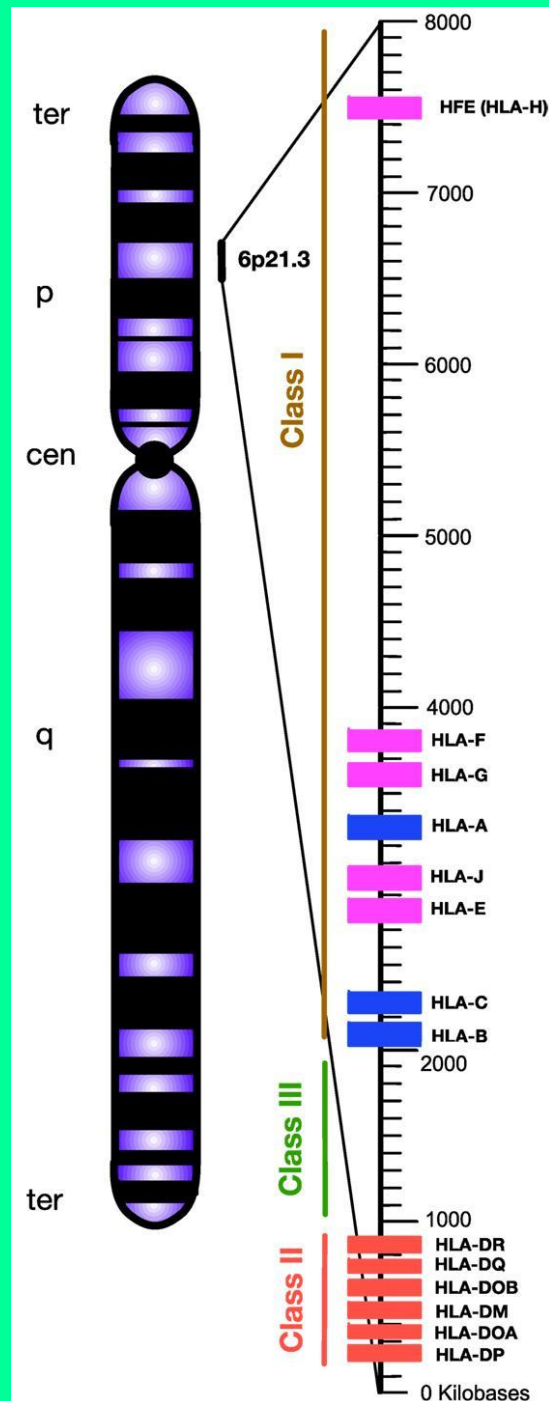
Except ABO blood system there are another example that is the human-leucocyte-associated antigen system (HLA).

HLA genes code for protein antigens that are expressed in most human cell types and play an important role in immune responses.

Six loci of it have over 100 alleles that have been detected in the human population and each allele differs from all other ones in at least one base.

These antigens are also the main class of molecule responsible for organ rejections following transplantations — thus their alternative name: major histocompatibility complex (MHC) genes.

Positions and organization of human leukocyte antigen (HLA) genes on human chromosome 6

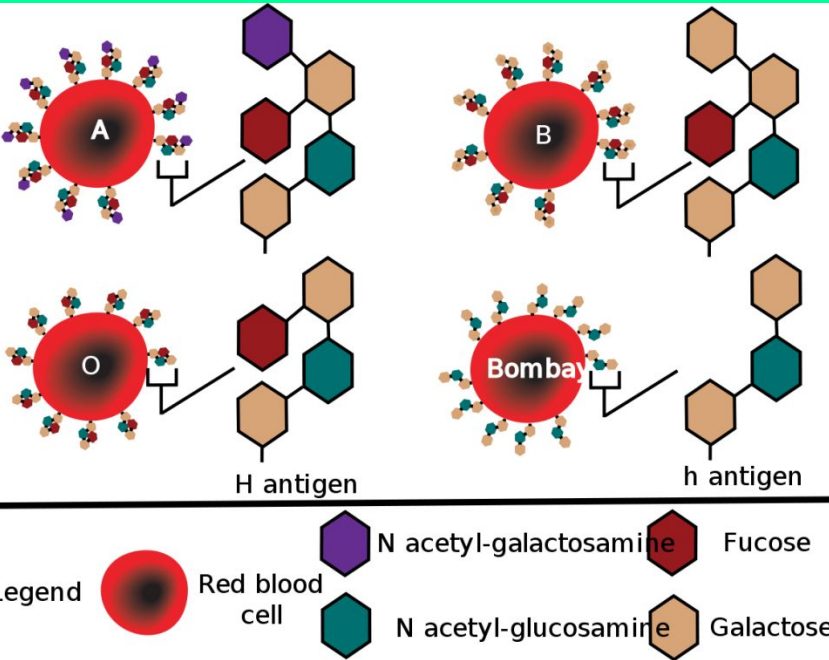
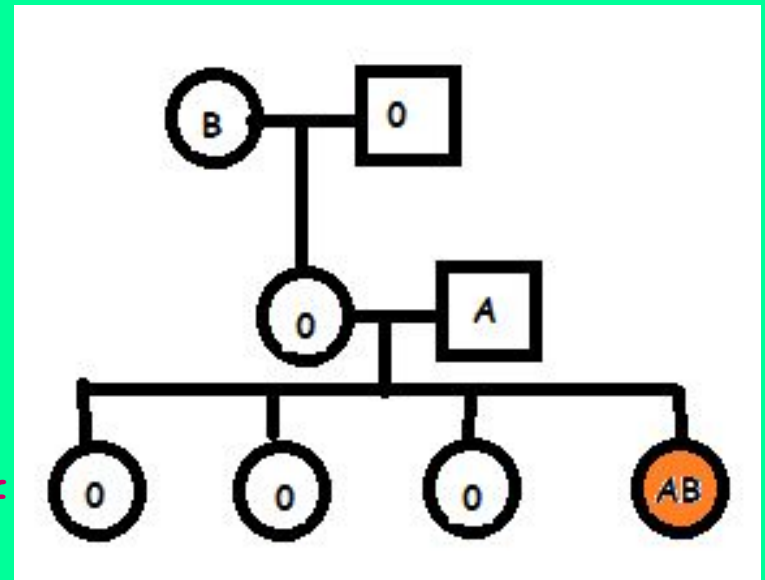


Example of Epistasis In Humans

In India in 1952 a doctor, conducting the study, noticed that the parents have some blood groups (the father had the first and the mother had the second), and the born child had a third.

The doctor was able to determine that the father had the first group.

The modification occurred due to the lack of an enzyme is necessary to synthesize the desired protein, which allows to determine the antigen.



This is due to the formation in humans pair of genes h. If a person is a heterozygote for a given gene, then the symptom does not appear. Due to an incorrect recessive combination of parental genes **the Bombay phenomenon** takes place.



Thank you