

1 Traits of Simple Inheritance

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- Traits of **monogenic** control (as opposed to **polygenic** control)

3 Polymorphisms

- Discrete genetic traits in which there are at least two alleles at a locus having frequencies greater than 0.01 (too large to result from mutation alone)

4 Polymorphisms in Human Blood

5 Blood

- 7-8% of human body weight
- Performs critical functions of transporting oxygen and nutrients to our cells and getting rid of carbon dioxide and other wastes
- Also plays a vital role in our immune system and in maintaining a relatively constant body temperature

6 The Four Components of Blood

- **Red Blood Cells (Erythrocytes)**
- **White Blood Cells (Leukocytes)**
- **Platelets (Thrombocytes)**
- **Plasma**

7 Red Blood Cells (Erythrocytes)

- 40-50% of the total blood volume
- Main function is to transport oxygen to the tissues located throughout the body and to transport carbon dioxide from tissues
- 95% of each red blood cell is made up of the gas transporting protein called **hemoglobin**

8 White Blood Cells (Leukocytes)

- Only make up about 1% of the total blood volume
- Three main types of white blood cells:
 - **Lymphocytes:** white blood cells that form a major part of the immune system
 - **Granulocytes and Macrophages:** white blood cells that protect our bodies from infection by surrounding and destroying microorganisms in the body

9 **Platelets (Thrombocytes)**

- Cells that clot blood at the site of wounds
- Also release proteins that fight bacteria
- Also stimulate the immune system

10 **Plasma**

- A relatively clear liquid protein and salt solution that makes up about 55% of the total blood volume
- Contains about 95% water
- Carries nourishment and removes waste products throughout the body
- About 500 different proteins identified in human blood plasma
 - **Albumin:** a group of large protein molecules that combine with and transport a number of substances
 - **Fibrinogen:** blood clotting agents
 - **Globulin:** small granular bodies – alpha, beta and gamma globulins (antibodies)
 - **Several clotting factors**

11 **Antibodies/Antigens**

- **Antibodies:** are protein molecules that are produced by the body and have the ability to identify and attach to foreign antigens on the surface of microorganisms that cause the organisms to cling together (**agglutinate**)

- **Antigens:** are substances that stimulate the synthesis of antibodies in the host's body

12 **ABO Blood Group**

Simple Genetic Systems

13 **Karl Landsteiner and the Discovery of the ABO Blood Group**

- Systematically analyzed the pattern of agglutination between blood donors and recipients
- Mixed the blood serum from one individual with the blood cells of another individual
- Found two antibodies: anti-A and anti-B
- Found that the ABO blood group passed on by Mendelian Inheritance
- Antigens are under control by three alleles (A, B, O) at a locus on Chromosome 9

14 **Relationship between Antigen, Antibody, and Blood Type**

Genotype	Phenotype	Antigen	Antibody
AA	A	A	anti-B
AO	A	A	anti-B
BB	B	B	anti-A
BO	B	B	anti-A
AB	AB	A,B	none
OO	O	none, H	anti-A, anti-B (smaller, weaker)

15 **Blood Donating**

Genotype	Phenotype	Donate To	Receive From
AA, AO	A	A or AB	A or O
BB, BO	B	B or AB	B or O

AB	AB	AB	A, B, AB, and O
OO	O	A, B, AB, or OO	

Universal Donor – O
 Universal Recipient - AB

16 **Biological Function of the ABO Blood Group**

- Distribution of Allelic Frequencies in All Populations:
 - Allele O: averages at 62.5%; ranges from 46 to 100%
 - Allele A: averages at 21.5%; ranges from 0 to 50%, highest frequencies occur in small unrelated populations
 - Allele B: is the rarest blood ABO allele with averages at 16%; ranges from 0 to 34%
 - Considerable interpopulation variation

17 **Some Insight Through An Analysis of Its Distribution**

- In general, A and B are greater than zero but usually less than 50% (Australian Aboriginals, some Native Americans, and the Lapps are the exceptions)
- Type A is more frequent in Europe
- Type B is more frequent in Asia
- Type O is more frequent in the New World

18 **World Distribution of the A Allele**

19 **World Distribution of the B Allele**

20 **World Distribution of the O Allele**

21 **Balanced Polymorphism**

- Selective forces that maintain a balance between the frequency of two or more alleles

22 **Selective Mechanisms in the ABO Blood System**

No clear consensus on the kind of selection – **balanced polymorphism** (selective forces that maintain a balance between the frequency of 2 or more alleles)

- **Differential Fertility**
 - **Prezygotic Selection**
 - **Infant Mortality**
- **Differential Mortality**

23 **Differential Fertility in the ABO Blood System**

- **Type O** mothers produce fewer children than expected when fathers were Type A or B blood
- **Type O** mothers produced larger numbers of offspring than expected when fathers were of the genotype AO (Type A) or BO (Type O)
- **Type B** mothers produced more children for all male parent blood types than Type O mothers

24 **Differential Fertility Acts in Two Ways**

- Selection at the prezygotic stage (prior to fertilization)
- Infant Mortality

25 **Selection at the Prezygotic Stage**

- Type O mothers produced more Type O offspring when mated with heterozygous males (AO and BO) possibly because of the presence of antibodies (anti-A and anti-B) in vaginal secretions

26 **Selection: Infant Mortality**

- Fetal loss due to anti-A and anti-B in the Type O mother (some mothers produce antibodies that can cross the placental membranes into the fetus – not all mothers) – ABO Incompatibility
- ABO Incompatibility leads to spontaneous abortion of the fetus (miscarriage)

27 **ABO Incompatibility**

Mother's Genotype	Incompatible Fetal Genotype (all heterozygotes)
AA	AB
AO	AB, BO
BB	AB
BO	AB, BO
AB	none
OO	AO, BO

28 **ABO Incompatibility and Fixation of the O Allele**

29 **Differential Mortality in the ABO Blood Group**

- An association between ABO blood types and certain diseases
 - Non-infectious diseases (particularly digestive system related diseases)
 - Infectious diseases
 - Insect vectors
 - Population size

30 **ABO Blood Group and Mortality Due to Non-Infectious Diseases**

31 **ABO Blood Group and Mortality Due to Infectious Diseases**

- Low incidence of Type O in plague areas (Europe, Asia, India, and Mesopotamia; the bacteria carries a H like antigen)
- Low incidence of A in smallpox areas (carries an A like antigen)
- Type B highest in India with long history of smallpox and plague

32 **ABO Blood Group and Mortality Associated with Insect Vectors**

- Blood sucking insects that carry diseases and parasites (such as Malaria) prefer Type O blood

33 **ABO Blood Groups and Mortality Related to Population Size**

- Infectious diseases most common in large densely populated areas
- Result in the lowest type A frequencies

34 **Secretors vs. Non-Secretors**

Secretors secrete the soluble ABH antigens into bodily fluids including saliva, tears, semen, milk, gastric juice, vaginal fluids, and other watery secretions (75-80% of the population are secretors)

Genotype	Phenotype
SE SE	secretor
SE se	secretor
se se	non-secretor