

Anthropology 215

GENETIC POLYMORPHISMS

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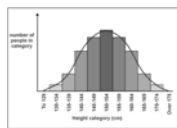
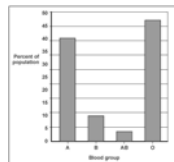
Variability in Human Populations

Discontinuous (or qualitative) variation

- traits which are simply inherited (a single gene locus is responsible)

Continuous (quantitative or multifactorial traits) variation

- controlled by many genes
- mode of inheritance is obscured because the environment and other developmental processes strongly influence the phenotypes

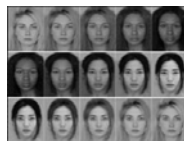


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Genetic polymorphism

"The occurrence together in the same locality of two or more discontinuous forms of a species in such proportions that the rarest of them cannot be maintained by recurrent mutation." (E.B. Ford)

Genetic traits which occur fairly regularly (at least 1-5% or 10%) in the population and whose frequency is too common to be due to repeated generation to generation mutations.



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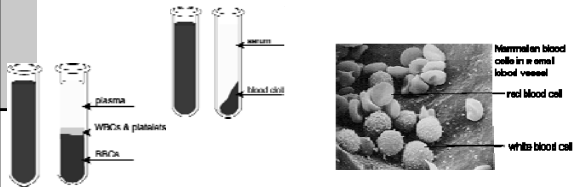
Genetic polymorphisms

- 13-14 human blood cell systems: e.g., ABO, Rh, MN, Kell, Duffy, Lewis, Diego, I, Xg, Lutheran, P, and Auberger etc.
- 160+ red blood cell antigens
- 30+ serum proteins (e.g., hatoglobins, transferrins, albumins)
- Hemoglobin molecule

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Blood

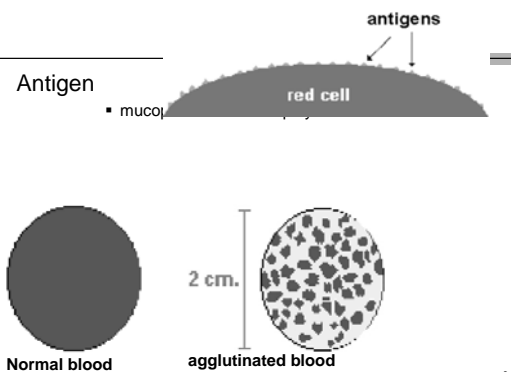
- elements (r.b.c., w.b.c., Hb, antigens etc.)
- serum (plasma)



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Antigen

- mucopolysaccharides



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Immunology

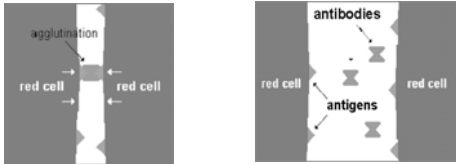
Study of antigen-antibody reactions

Antibody

- substances produced to counteract antigens

Antigen

protein that causes the production of an antibody or reacts with an already existing antibody



ABO Blood Group

- discovered by observing the results of earlier transfusions
- Landsteiner (1900)



anti-B	anti-A	Blood Type
-	-	O
+	-	B
-	+	A
+	+	AB

Genetics of ABO Blood Group

- Bernstein (1924)
 - described genetic basis for antigen-antibody reactions

Alleles	Genotype	Phenotype	Antigen on r.b.c.	Antibody in serum
A,B,O	AA, AO	A	A	anti-B
	BB, BO	B	B	anti-A
	AB	AB	A, B	--
	OO	O	--	anti-A, anti-B

ABO

Universal donors (O)

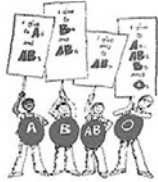
- no antigens on their r.b.c. and therefore no clumping in the major cross

Universal recipients (AB)

- have both A and B antigens but no antibodies in their serum

major cross: donor's cells (antigens) mixed w/ recipient's serum

minor cross: donor's serum mixed w/ recipient's cells



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Other Blood Groups

MN Blood type (co-dominant):

- anti-M
- anti-N

Other blood groups: Diego, P, Lutheran, Kell, Lewis, Duffy, Kidd, Auberger, X_g

Distribution of Blood Groups

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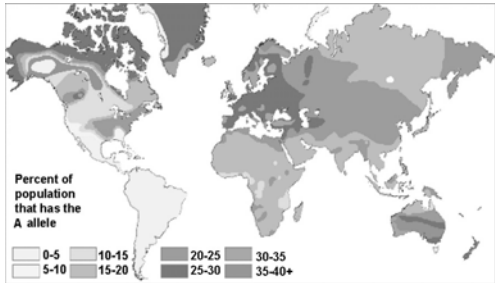
Distribution of the B type blood allele in native populations of the world



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Distribution of the A type blood allele in native populations of the world



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Distribution of the O type blood in native populations of the world



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ABO & Disease

Syphilis & A antigen

- smallpox & A, AB
- typhoid fever, influenza, bubonic plague & O
- infant diarrhea & B

ABO & non-infectious diseases

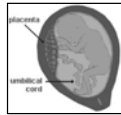
- O - duodenal and gastric ulcers
- O - birth pill
- A - cancer of stomach, cervix, ovarian tumors, pernicious anemia

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Erythroblastosis Fetalis

- Rh Incompatibility
- maternal-fetal incompatibility
- Mother (dd) Rh- X Father (DD or Dd) Rh+
- fetus (Dd) Rh-

	father		father	
	D	D	D	d
mother	d	Dd	Dd	100% Rh+ children
	d	Dd	Dd	50% Rh+ children



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ABO Incompatibility

Mother-fetus ABO incompatibilities

- O Mother & A or B fetus

ABO incompatibility

- of mother and father can also lead to fetal wastage

Mother	Father
AA	AB
AO	AB, BO
BB	AB
BO	AB, AO
AB	None
OO	AO, BO

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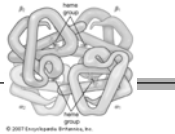
Rhesus (Rh) Blood Type

- Rh Blood Type
 - more complicated mode of inheritance
 - D, C, and E
 - DD, Dd = Rh+
 - dd = Rh-



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Hemoglobin



found in the blood

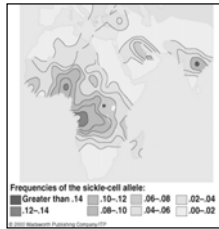
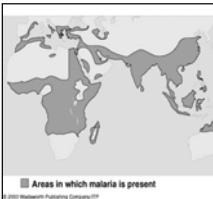
- carry oxygen that picks up in the lungs and transports it to all parts of the body by way of the circulatory system

Alleles	Genotypes	Phenotypes
A,S	AA	normal hemoglobin
	AS	sickle-cell trait (sicklers)
	SS	sickle-cell anemia (deadly)

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Malaria

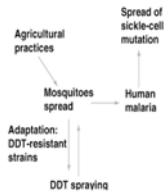
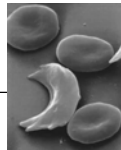
A.C. Allison (1954)



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Malaria and HbS

- sickle-cell trait (HbAS)
 - statistics
 - biochemical and physiological basis
 - mosquito



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Other Hemoglobin Variants

- Hb^C - lysine is substituted for glutamic acid at the 6th position of the beta chain
- Hb^E - lysine is substituted for glutamic acid at the 26th position on the beta chain

G-6PD Deficiency (X-linked)

- Glucose-6-Phosphate deficiency
- anti-malaria drugs (primaquine) sulfonamides, anti-bacterial agents
- favism (fava bean)
- American Black Servicemen, given primaquine, developed severe hemolytic anemia

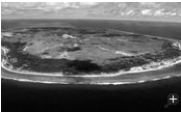

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Tay-Sachs disease

(NIDD) Noninsulin-dependent diabetes:

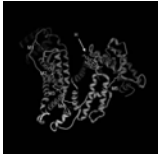
- Nauru Island - 30% of the people over age 15 have disease

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Serum Proteins

- Albumins - maintain proper osmotic pressure in blood and also protein reserves
- Haptoglobin - bind free Hb in plasma and prevent Hb from passing to the kidneys
- Transferrins - binds iron for transport to bone marrow and other tissues
- Group-specific proteins (Gc) - function not clear



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