

Changing Perception of Human Differences

- I. **Examples of traits that vary among human populations and between individuals**
 1. Skin color
 2. Hair form, eye color
 3. Body size and proportions
 4. Biochemical differences
- II. **Interpretations of the origin and nature of variation**
 - A. **Early hunter-gatherer beliefs**
 1. Human lineage are descended from a variety of animal ancestors
 2. Some humans, such as shaman can transform themselves into animals
 3. **Pseudo-speciation**: Outsiders are symbolically transformed into "non-human" humans for purposes of exploitation
 - B. **Theories of later agricultural societies**
 1. **Variation is Cyclical**: The world is composed of cycles with varying periodicities. Golden ages alternate with periods of decay and rebirth
 2. **Pythagoras** (582-500 BC) theorized that the earth was a perfect sphere that could be understood through mathematics
 3. **Plato** (ca 427-347): the world is the product of a divine artisan
 4. **Epicurus** (342-270 BC): Epicureans believed the world is composed of random swiftly swirling atoms
 1. **Aristotle** (384-322 BC): Argues the world is static and of unlimited duration. He espoused the scientific approach of collecting data and speculating upon it. Argued that the highest and most satisfying form of human existence is that in which man exercises his rational faculties to the fullest extent.
 2. **ÊStoics**: the world is designed for the benefit of man
 3. The Created World of Judeo-Christian tradition: the world is static, and has only existed for a short time
 - C. **Darwin's world view: the world is gradually evolving through the process of natural selection**
- III. **Laying the framework for a changing in universe**
 - A. **Natural historians of the 17th and 18th centuries**
 1. Believed that the world is static and that the variations that do exist are a reflection of divine wisdom
 2. Classification of plants and animals was considered a religious activity designed to reveal God's wisdom: For example, **John Ray's** book (1627-1705) "The Wisdom of God as Manifested in the Works of Creation"
 3. Interpretations of variation within a static world
 - a) *Cyclical variation- rotation of planets, etc.*
 - b) *Random variation such as the differences between individuals is God's way keeping the world interesting and thus perfect*
 - c) *Differences between human populations reflect a decline form original perfection*

d) *The "Great Chain of Being": Organisms can be ranked according to their closeness to God*

4. Problems of intermediate forms: continuous variation makes some animals difficult to pigeonhole
5. **Carolus Linnaeus** (Carl von Linné), 1707-78 .The Swedish botanist who developed the binomial system of nomenclature in which animals are classified within a nested hierarchy
6. **Georges-Louis De Buffon** (1707-1788) French naturalist remembered for his comprehensive work on natural history, *Histoire naturelle, générale et particulière* Believed that species were the highest level at which animals could be organized. Realized the environment influenced lifeforms. Looked at how variation was patterned and how those patterns were created
7. **Gorges Cuvier** (1767-1832) "magician of the charnel house" identified animals in the fossil record that no longer exhibit. An anti-evolutionist, the religious dilemma posed by the assumptions of Linnaeus's hierarchical system

B. The inconstant heavens

1. **Nicholas Copernicus** (1473-1543) discovered that the earth rotated around the sun. Developed the heliocentric theory of astronomy
2. **Tycho Brahe** (1564-1601) discovered new heavenly bodies
3. **Galileo Galilei** (1564-1642) found irregularities in the surface of the moon. Emphasized the development of general laws based on observation and experiment rather than the word of authority

C. Expanding the spatial dimension

1. The voyages of the early explorers such as Marco Polo and Columbus revealed an unexpected diversity plants and animals
2. The discovery of the microscope made visible a previously unknown fauna of microscopic organisms

D. Expanding the temporal dimension

1. **Neptunian concepts** of geology: geological deposits were the result of catastrophic floods of the Bible
2. **James Ussher** (1581-1656): Calculated that creation occurred at midday on October 23, 4004 BC
3. **Issac Newton** (1642-1727): Using physical principals calculated that the earth must 75,000 or more years old
4. **Charles Lyell** (1797-1875): developed the theory of **uniformitarianism** which postulates that geological deposits are a result of process like those seen today operating over immense periods of time.

IV. Theories of organic changes

A. Lamarck (1744-1829): Adaptation through inheritance of acquired traits

1. Concluded based on his geological studies that the earth is very old and that conditions on it are constantly changing

2. Since organisms are adapted to the environment, he concluded that organisms must changed to adapt to their changing environment
3. Proposed that this adaptation occurred through the inheritance of acquired traits
4. **Lysenko (1898-1976):** Russian Lamarckian geneticist responsible for the suppression of Mendelian genetics for political purposes

B. Charles Darwin (1809-1882): Adaptation through natural selection

1. Variations exist among individuals of the same species
2. Variations are hereditary
3. Species tend to over-reproduce
4. There is a struggle for existence
5. Survival of the fittest
6. Through accumulation of favorable hereditary characters, new species eventually develop

C. The Galapagos Finches: natural selection and adaptive radiation as an explanation for diversity

1. Colonization of a single island by an ancestral finch from the South American mainland
2. Population expansion owing to lack of competitors
3. Colonization of new islands owing to population pressure
4. Adaptation to different environmental conditions on each island through natural selection
5. Eventual development of reproductive isolation

Important terms in Marks Chapter 1 and Pages Where They Occur			ÊÊÊ
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		Great Chain of Being	9
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Mitosis	29
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Phenotypes	32

Mendelian Inheritance

V. Pre-Mendelian theories of inheritance

- A. ÊDarwin-pangenesis and gemmules
- B. ÊBlending of bloods

VI. Gregor Mendel (1822-1887) and his pea experiments

- A. ÊHeredity is governed by a large number of independent units (genes)
- B. ÊWhen each parent contributes the same gene a constant character is produced in the offspring
- C. ÊWhen each parent contributes a different form of the same gene, hybrid offspring are produced that possess both alleles
- D. ÊThese alleles maintain their identity since they reappear in succeeding generations

VII. Dominant, Recessive, and Semidominant alleles

- A. Dominant traits are expressed when homozygous or heterozygous
- B. Recessive traits are only expressed when homozygous
- C. Semidominant both alleles produce the same product but in lesser quantity compared with dominant alleles (example:red,white and pink four o'clocks)

VIII. Mendelian inheritance in humans

A. Dominant Traits

- 1. Phenylthiocarbamide (PCT): taste sensitivity to this synthetic compound is dominant
- 2. Ear Wax: "wet sticky" is dominant, "dry, flaky" is recessive
- 3. Incisor teeth missing
- 4. Sickle cell anemia
- 5. White forelock
- 6. Polydactyly (having extra digits)
- 7. Blood groups: ABO, MNS, Rh
- 8. Achondroplastic dwarfism
- 9. Von Recklinghausen disease- autosomal dominant, this is the disease that afflicts Man"

B. Recessive Traits

- 1. Albinism- failure to synthesize melanin in the skin owing to a lack of the enzyme necessary to convert tyrosine to melanin- recessive
- 2. Tay Sachs syndrome- the enzyme hexosaminidase is not produced and a complex lipid, ganglioside GM2 builds up in nerve cells. About 1 out of 27 Ashkenazic Jews are carriers
- 3. Deaf-mutism

C. ÊTraits whose mode of inheritance is not well established.

- 1. ÊDominant inheritance?
 - a) *Tongue rolling, and folding (see reader article)*
 - b) Ê*Darwin's Point*
 - c) Ê*Attached earlobes- free earlobes may be dominant (see reader article).*
 - d) Ê*Handedness*

- e) *Widow's peak (a point formed by the hair at the middle of the forehead).*
- f) *Relative finger lengths*
- g) *Hand clasping*
- 2. *Recessive inheritance?*
 - a) *Whorl pattern of the hair- counter clockwise is thought to be recessive.*
 - b) *Eye color*
 - c) *Hair color (blond)*
 - d) *Hair color (red)*
 - e) *Hitch-hiker's thumb*

IX. The ABO blood group

- A. Carl Landsteiner (1864-1943) in 1901 showed the nature of transfusion incompatibilities
- B. Blood cells possess specific surface proteins called antigens
- C. Plasma, cell free portion of the blood contains complimentary antibodies:

Blood Type	SurfaceAntigens	Antibodies in Plasma
EA	A antigens	Anti -B antibodies
EB	B antigens	Anti -A antibodies
EAB	A and B antigens	No anti -A or B antibodies
EO	No A or B antigens	Anti-A and B antibodies

X. Rh Maternal fetal incompatibility

- A. Rh- mothers become sensitized by Rh+ babies
- B. The second pregnancy with a Rh+ results in Erythroblastosis

XI. Sex Linked Inheritance

- A. Males have 22 autosomes + xy, females 22+ xx
- B. Females carry traits on their x chromosomes that appear recessive in their female children and dominant in male children. This is because males only have one x chromosome
- C. Examples: Red green color blindness, hairy ears, hemophilia

Sources of Genetic Variation

XII. Mutations: copying errors in the reproduction of genetic material. Most are harmful (analogy with a random change in a watch)

- A. Germinal mutations: occur during meiosis, heritable.
- B. Somatic Mutations: occur during mitosis, not heritable. McCune-Albright's syndrome is a non-heritable genetic disease caused by a somatic mutation.
- C. The Value of Mutations: Without the variation they generate, adaptation through natural selection is impossible

XIII. Gross chromosome level mutations: Chromosomal aberration ("cytogenetic aberration"): mutation in which the wrong number of copies of some or all of a chromosome,

- A. About 1% of newborns, and maybe 50% of spontaneous abortions, possess an abnormal karyotype.
- B. Down's syndrome: trisomy-21, 1 in 700 births. It affects around 1 child in 700. Advanced maternal age is a risk factor. Maybe 1 in 25 live births to mothers over 45 have Down's.
- C. Cri du Chat syndrome: deletion of part of a chromosome 4 or 5, 1 in 10000 births, Children are profoundly retarded, but some survive into adulthood.
- D. Klinefelter's syndrome: xxy, xxxy, etc., One man in about 850 is affected.
- E. Turner's syndrome: ox, 1 in 2500 births
- F. Females with extra x's: xxx, xxxx, etc., 1 in 1000 births
- G. Males with extra y's: xyy, xyxy, etc., 1 in 1000
- H. Crossing over: exchange of genetic material between homologous chromatids. An important source of genetic variation. It is common and geneticists do not consider it a mutation.

XIV. Point Mutations: Changes at the molecular level, often involving a single base substitution

- A. The DNA molecule: Phosphate, sugar, base pairs (adenine, thymine, cytosine, guanine)
- B. Examples:
 - 1. Sickle cell anemia: Caused by the substitution of amino acid valine for Glutamic acid
 - 2. Neurofibromatosis (Von Recklinghausen disease): Autosomal dominant, appears to be caused by a gene on chromosome 17
 - 3. Huntington's disease: caused by a dominant gene located on chromosome 4
- C. The role of somatic mutations in cancer: several are often necessary to deregulate cell growth- Example: colon cancer
- D. Causes of Mutations
 - 1. Genetic regulation of mutation rates

2. ÊChemical mutagens: Mustard gas, benzene, pesticides, formaldehyde, meat preservatives (nitrites) produce nitrosamines in the stomach
- E. ÊRadiation:Ê X-rays: In 1926 Hermann Muller discovered that X-rays produced them in fruit flies.
- F. Other Sources: cosmic rays, air travel, high voltage cables, microwaves, TV and computer screens, atomic bombs

Mutation Rates (per million gametes per generation)

ÊDominant Traits

ÊAnirida (no iris)	5
ÊMarfan's syndrome	5
ÊChondrodystrophic dwarfism	42
ÊNeurofibromatosis	7.3-10.5

ÊAutosomal Recessive

ÊAlbinism	28
ÊEpidermolysis bulosa (blisters on feet)	50

ÊX-linked Recessive

ÊHemophilia A	32
ÊMuscular dystrophy, Duchenne type	43

Important terms in Marks chapter 4

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Blumenbach , Johann Friedrich	57
Coon, Carleton S.	49
Gobineau, Arthur de	64
Homo erectus	58
Platonism	55
Thrifty genotype hypothesis	62

Natural Selection and Human Variation

XV. Forms of Natural Selection

A. Stabilizing selection

1. Reduces genetic variability by selecting out extremes
2. Examples: The effects of the winter of 1898 on sparrows, Clutch size in birds? The fact that humans usually give birth to one child. The distribution of birth weights in human babies?

B. Directional selection

1. Selection that favors one extreme
2. Responsible for evolutionary change
3. Examples:
 - a) *Artificial selection is a special form of directional selection in which humans provide the selective pressure*
 - b) *Penicillin resistant Micrococcus*
 - c) *Industrial melanism*
 - d) *Adaptation of humans and horses to environmental change*

C. Diversifying selection

1. Selection favoring more than one adaptive norm
2. The fishing net analogy
3. The role does diversifying selection in species formation

D. Balancing Selection

1. **Balanced Polymorphism** a condition of equilibrium among different alleles at a given chromosomal locus as in cases where heterozygotes have a selective advantage over homozygotes
2. Sickle cell anemia and malaria
 - a) *People homozygous for the trait (ss) are severely anemic*
 - b) *anemic*
 - c) *Normal people (SS) are susceptible to malaria*
 - d) *Heterozygous people are slightly anemic but have the advantage of malaria resistance*
 - e) *have the advantage of malaria resistance*
 - f) *Human modification of the environment may be reducing the advantage of having the sickle cell trait*

XVI. **Sexual Selection According to Darwin (1871)** *"sexual selection depends not on a struggle for existence in relation to other organic beings or to external conditions, but on a struggle between the individuals of one sex, generally the males for possession of the other sex."*

- A. **Intrasexual selection:** competition for mates among members of the same sex: this may favor horns, large canines etc.
- B. **Intersexual selection ("sex appeal"):** This kind of sexual selection may explain traits such as the peacock's tail.
- C. Sexual selection and natural selection can reinforce each other as well as act at cross-purposes

- XVII. **Darwinian Fitness:** The adaptive value of a trait measured in terms of its effects on reproductive capacity
- A. Fitness is measured relative to the reproductive success of a reference population
 - B. The fitness value of a trait depends on the environment in which it occurs. The reproductive value of a trait may be high in one environment and low in another
 - C. Fitness varies with the time frame used to measure reproductive success. Short-term success may mean long-term failure
 - A. Adaptedness versus adaptability: generalists have a great deal of adaptability, specialists are highly adapted.

Important terms in Marks chapter 5

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Davenport , Charles B.	81
Dobzhansky	91
Feeble-mindedness	82
Galton, Francis	77
Hereditarianism	89

Important terms in Marks chapter 6

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Nuremberg laws	112
Polymorphism	101
Polytypism	101

Population Genetics

XVIII. Processes that alter the composition of a gene pool

- A. Immigration: immigrants may differ genetically from members of the gene pool
- B. Emigration: alleles can be lost through emigration
- C. Natality: births change gene pools through the introduction of mutations and differential reproductive success
- D. Mortality: alleles are constantly being removed from gene pools through deaths

XIX. **The Hardy-Weinberg law:** The allele frequencies in a population will remain the same over time (in equilibrium) if the following conditions are met:

- A. Random mating
- B. No Genetic Drift
- C. No migration
- D. No natural selection
- E. No mutations

XX. **The Hardy-Weinberg Equation:** a means of calculating gene frequencies expected if the population is in genetic equilibrium. These frequencies can then be compared with gene frequencies actually observed to identify the effects of selection, drift, migration, and so on.

$$P + Q = 1$$

P= frequency of dominant alleles in population

Q= frequency of recessive alleles in population

P and Q account for 100% of the alleles i.e: $P+Q=1$

$$P^2 + 2PQ + Q^2 = 1$$

P^2 = frequency of homozygous dominant individuals

Q^2 = frequency of homozygous recessive individuals

$2PQ$ = frequency of heterozygous individuals

XXI. **Genetic Drift:** Variation in gene frequencies of small populations due to accidents in sampling. The larger a population the lower the probability of genetic drift.

XXII. **The Founder Effect:** When emigrants from a population do not possess a random sample of the genes of that population, they will differ genetically from the ancestral population.

- A. The Dunkers: A religious isolate founded by 50 families. They have deviant frequencies of attached earlobes and hitchhiker's thumb and so on that that may be explained by the founder effect

Dunker's ABO allele frequencies	O	A	B
Dunkers	60%	38%	2%
West Germans	64%	29%	7%
Americans	70%	26%	4%

B. Alternative explanations of the high frequency of type O blood in American Indians:

1. The founder effect: Was the O allele exceptionally common among the first Americans.
2. Natural Selection: Malaria resistance of A and B vs. losses of OA and OB fetuses

XXIII. The Bottleneck Effect: This is a sampling error that results when a catastrophic event reduces the size of a population to a few individuals, the survivors may differ genetically from the ancestral population in traits unrelated to their survival and give rise to a genetically distinctive population.

A. Example: The population of Pingelap island in Micronesia has an unusually high frequency of achromatopsia in the population. The survivors of a hurricane appear to have had an unusually high frequency of the recessive allele for this form of color blindness.

XXIV. Non-random mating and its causes

A. Geographical Isolation

1. Spatial proximity makes the heart grow fonder: Islands, mountains, and glaciers isolate local populations
2. The development of transportation technology has resulted in the breakup of local genetic isolates

B. Behavioral isolation

1. Territoriality
2. Cultural factors: Language, Religion, Social class regulate gene flow between groups.

C. Marriage patterns

1. Polygyny: more than one wife
2. Polyandry: more than one husband
3. Monogamy

D. Assortative mating

1. Positive assortative mating: "like marries like"
2. Negative assortative mating: like repels like

E. Inbreeding

1. (1)Genetic consequences
 - a) *Reduction in genetic variability*
 - b) *Increase in frequency of homozygous recessives*
 - c) *Recessive alleles exposed to natural selection*
2. (2)Social consequences
 - a) *Concentration of wealth and power*
 - b) *Reduction of social ties with other groups*

Human Biodiversity Study Questions

Chapter 1

1. Explain how the Great Chain of Being paradigm was used as a tool to understand nature. How does it differ from the "nested hierarchy" idea of biological variation.
2. What were Carolus Linnaeus's contributions to science?
3. How did the ideas of Linnaeus, Buffon and Lamarck differ? How were they the same.
4. Explain what Darwin meant by "descent with modification."
5. What is the difference between phenetic and cladistic (phylogenetic) forms of analysis? What are some of the weaknesses of cladistic analyses?
6. What is the importance of cultural relativism?

Chapter. 2

1. What are the biological processes that produce genetic diversity?
2. Define adaptation and adaptability.
3. Explain genetic drift. Under what conditions can it violate the Hardy-Weinberg equilibrium assumptions.
4. What is the difference between microevolutionary and macroevolutionary processes?
5. How does culture affect human biodiversity? Give some examples.

Chapter 3

1. How did the idea of race originate and how has evolved during the past few centuries?

Chapter 4

1. What are the advantages of separating the issues biological and cultural origins? Can you see any disadvantages?
2. What was Darwin's impact on the notion of historical and biological progress.
3. According to F. Boas, why could biological evolution not be the cause of cultural evolution?

Chapter 5

1. What distinguishes science from pseudoscience? How is eugenics a pseudoscience?
2. After enjoying such popularity, why did the eugenics movement fail?

Chapter 6

1. Explain the importance of the distinction between polymorphism and polytypism in the issues surrounding race and eugenics
2. How do racial studies differ from racist studies?
3. What are some of the problems in IQ tests to measure performance and ability?

Important terms in Marks**Chapter 5**

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