The Human Heritage:

Genes and Environment

1/25/01

Outline

- **Sexual Reproduction and Genetic Transmission**
- **Genotype and Phenotype**
- **Mutations & Genetic Abnormalities**
- **Biology and Culture**

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We are all Unique - Why?

- Unique Genetics
  - Most of us
    - Exception: Identical Twins (MZ)
- Unique Environment
  - Varying degrees of similarity
- Combination of Genetics and Environment
  - All of us!

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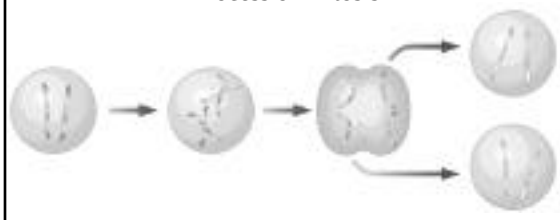
Sexual Reproduction and Genetic Transmission

- **Mitosis: A Process of Cell Replication**
- **Meiosis: A Source of Variability**
  - **Crossing-over**
- **Sexual Determinism: A Case of Variability**

Process of Mitosis



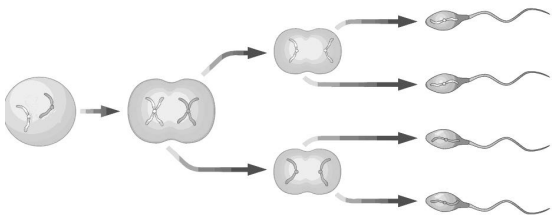
Cell nucleus with a pair of chromosomes

Chromosomes split and replicate to produce two identical pairs

The pairs separate, and the cell divides

Each daughter cell now has a pair of chromosomes that is identical to the original pair

Meiosis

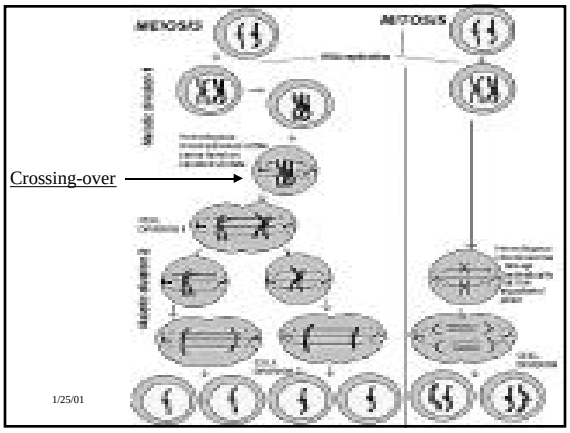


One pair of homologous chromosomes

First meiotic cell division begins, but does not proceed as in mitosis. Instead of the replicated chromosome splitting apart, one member of each homologous pair becomes a part of the first-generation daughter cell.

The second meiotic division proceeds after the first is completed; now the replicated chromosome acquired in the first-generation daughter cell splits apart.

Each of the four gametes produced by the two-step process now has acquired one member of the pair of homologous chromosomes.


MEIOSIS
MITOSIS
Crossing-over
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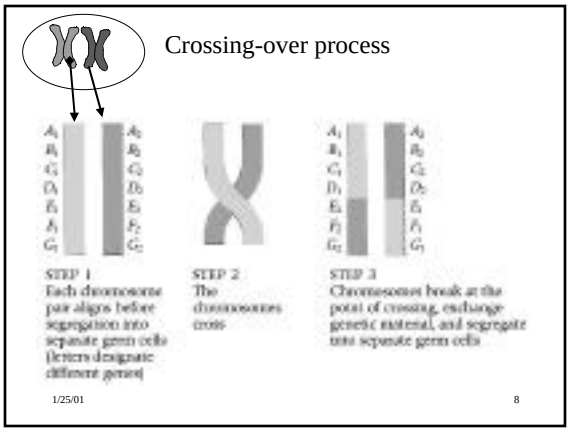
Crossing-over process
STEP 1
Each chromosome pair aligns before segregation into separate germ cells (letters designate different genes)
STEP 2
The chromosomes cross
STEP 3
Chromosomes break at the point of crossing, exchange genetic material, and segregate into separate germ cells
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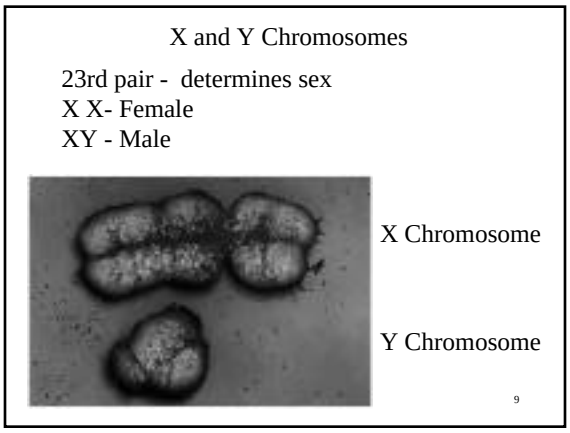
X and Y Chromosomes
23rd pair - determines sex
X X- Female
XY - Male
X Chromosome
Y Chromosome
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Genotype and Phenotype

- Genotype: Set of genetic traits a person inherits; a person's inborn capacity or potential
- Phenotype: Set of genetic traits a person inherits; a person's inborn capacity or potential

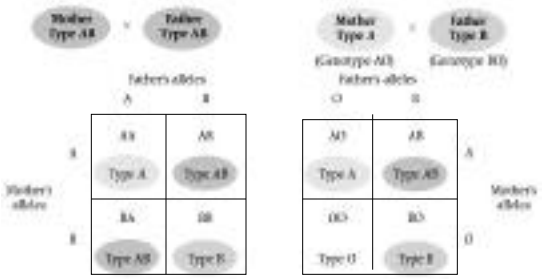
The Laws of Genetic Inheritance

- Simple Form
  - Single pair of genes contributes to inherited characteristic (allele)
  - Alleles can be the same (homozygous) or different (heterozygous)
  - When the pair is the same - trait will be displayed

The Laws of Genetic Inheritance

- When alleles are different (heterozygous):
  - 1. Child will express characteristics of one of the alleles (Dominant allele)
  - 2. Child will express both (averaging, intermediate)
  - 3. Child is affected by both but the characteristic displayed is different from both - co-dominance

Inheritance of alleles for blood type



Type AB = Codominance

AO or BO = dominance

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Inheritance of a Dominant Gene Disorder

Affected Parent (Has the Disorder)

Normal Father	D	r
r	**Dr Affected (25%)**	**rr normal (25%)**
r	**Dr Affected (25%)**	**rr normal (25%)**
	(50%)	**(50%)**

From Seifert/Hoffnung, *Child and Adolescent Development, 5/e*, Figure 3.10, p. 73. Used by permission of Houghton Mifflin Company.

Dominant Gene

Disorder	Incidence
Polydactyly	1/300 - 1/100
Achondroplasia	1/2,300
Huntington disease	1/15,000 - 1/5,000

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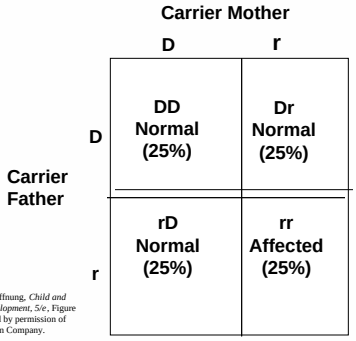
Inheritance of a Recessive Gene Disorder



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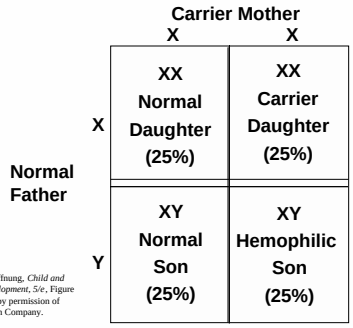
Recessive Gene

Disorder	Incidence
Cystic fibrosis	1/2,500 white persons (carrier risk: 1/25)
Sickle-cell disease	1/625 African Americans (carrier risk: is 1/10)
Tay-Sachs disease	1/3,600 Eastern European Jews (carrier risk: 1/30 - 1/300)

Sex-linked Genetic Effects

- Genes found only on the X or Y chromosome
- Most carried on the X chromosome (Why?)
- Males more susceptible to sex-linked genetic defects

Inheritance of Hemophilia, a Sex-Linked Disorder



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X-Linked

Disorder	Incidence
Hemophilia	1/2,500 male babies
Duchenne's MD	1/3,500 male babies

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Film: Tackling a Killer Disease

Things to look for:

1. Genetic and Chromosomal abnormalities
   - X-linked gene disorders
2. Nature of experimentation
   - A. Cause and effect relationships
   - B. Independent and Dependent variables
   - C. Experimental and Control conditions
   - D. Double-blind Procedure
   - E. Placebo

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Multifactorial Conditions

- Normal variant of normal gene
- Indirect Genetic effects
  - Operate on risk factors not disease itself
  - Risk features continuously distributed attributes
  - Several genes involved
  - Genetic effects are probabilistic
- Example: Smoking

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Multifactorial

Disorder	Incidence
Congenital heart disease	1/125
Neural tube defect	1 - 2/1,000
Cleft lip/cleft palate	1/1,000 - 1/5,000

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Mutations & Genetic Abnormalities

- Additional Source of Variability
  - Can be inherited and passed on
    - Mutation affects the sperm or ovum
  - Can be limited to the individual
    - Mutation limited to specific types of cells
      - E.g., Exposure to environmental elements.

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Mutations & Genetic Abnormalities

- Gene-Environmental Interaction
  - Sickle-Cell Anemia
- Chromosomal Error
  - Down Syndrome
- Sex-Linked Chromosomal Abnormalities
  - Phenylketonuria (PKU): A Treatable Genetic Disease

Down Syndrome: Chromosomal Error

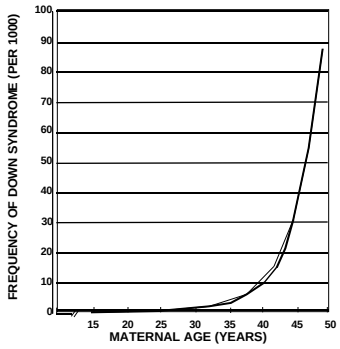
A. Caused by:
1) Trisomy 21
- accounts for 95%

2) Translocation
- accounts for 3-4%

3) Mosaicism -
Accounts for 1%

B. Incidence
- 1/800 live births
- maternal age factor

Relationship Between Maternal Age and the Incidence of Down Syndrome



PKU - Single gene disorder

- Recessive pattern of inheritance (Chr. 12)
  - 1/8000 - 10,000 births
- Affects body's metabolization of proteins
  - Lack enzyme that converts amino acids
  - Leads to toxic levels of phenylalanine
- Damage to nervous system
  - Untreated - effects evident by 3 - 5 months, retarded by 1 yr.
- Treatment - restricted diet

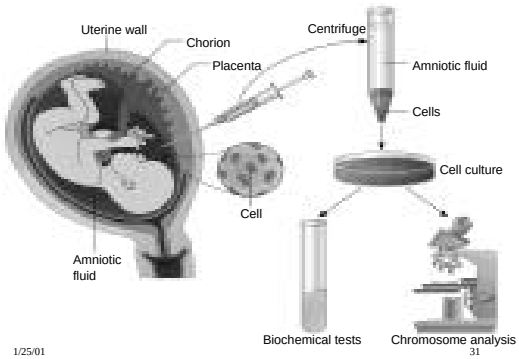
Examples of Sex Chromosome Abnormalities

Turner syndrome (XO) 1/5,000 women	Short stature, limited secondary sex characteristics, infertile, near-average IQ, deficiencies in spatial abilities
Triple-X syndrome (XXX) 1/1,200	Can exhibit delays in speech and language development, coordination problems, academic and behavioral difficulties
Klinefelter syndrome (XXY) 1/600 men	Tall, female body contour, usually sterile, some evidence for short-term memory and reading problems
"Supermale" (XYY) 1/1000	Above-average height, near-average IQ, some have learning disabilities

Prenatal Screening Tests

Prenatal	When Usually Administered (gestational age)	Typical Waiting Period for Results	Other Comments
Fetal blood sampling	18 weeks or later	24–48 hours	Possibly somewhat greater risk than associated with amniocentesis.
Amniocentesis	15–18 weeks	About 2 weeks	Can be administered in weeks 11–14 but normally is not because the available supply of amniotic fluid is more limited.
Chorionic villus sampling	10–12 weeks	24–48 hours	Possibly a slightly greater risk than associated with amniocentesis, including limb deformities.
Ultrasonography	About 6 weeks and later	None	Provides picture of growing fetus. Not definitive for identifying many disorders. Little evidence of any risk. Often used to accompany other test procedures.

The Process of Amniocentesis



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Who Should Seek Prenatal Counseling?

1. Couples who already have a child with some serious defect such as Down syndrome, spina bifida, congenital heart disease, limb malformation, or mental retardation
2. Couples with a family history of a genetic disease or mental retardation
3. Couples who are blood relatives (first or second cousins)
4. African Americans, Ashkenzzi Jews, Italians, Greeks, and other high-risk ethnic groups
5. Women who have had a serious infection early in pregnancy (rubella or toxoplasmosis) or who have been infected with HIV
6. Women who have taken potentially harmful medications early in pregnancy or habitually use drugs or alcohol
7. Women who have had X rays taken early in pregnancy
8. Women who have experienced two or more of the following: stillbirth, death of a newborn baby, miscarriage
9. Any woman thirty-five years or older

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Source: Adapted from Fienbloom & Forman (1987) p. 129

Genes, The Organism, and the Environment

- Studying Gene-environment Interactions
- Range of Reaction
- Canalization
- The Study of Genetic Influences on Human Behavior
- Estimating Genetic Influence Through Kinship Studies

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Gene-environment Interactions

- Two-way interaction
  - Genes predispose you to display characteristic
    - Environment can increase/decrease likelihood
  - Genetic predisposition can change your reaction to the environment

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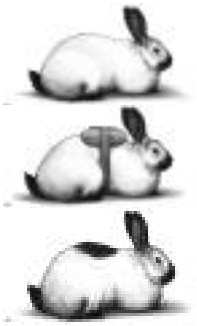
The Effect of the Environment on Fur Color

1. Normally only feet, tail, ears and nose are black.

2. Remove fur on back & place icepack

3. New fur is black.



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Range of Reaction

- Combination of genes and environments can lead to many possible outcomes
  - Example: If you have the genes that code for obesity but are in a time of famine, you will not show signs of obesity but if you are in a bountiful time you will.
- Researchers try to find the range of reaction for a given genotype by manipulating the environment (done with plants, animals)
  - For ethical reasons can't be done with humans

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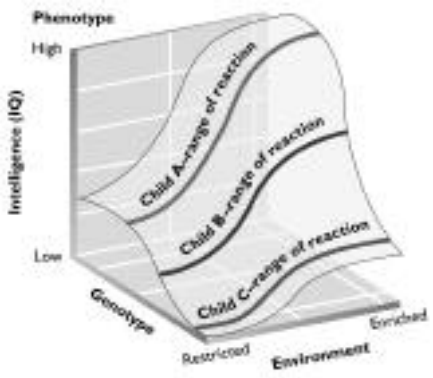
The Concept of Range of Reaction for Intellectual Performance



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Adapted from Turkheimer, Goldsmith, & Gottesman, "Commentary", *Human Development*, 38 pp. 142-153. Copyright © 1995. Reprinted by permission of S. Karger AG, Basel, Switzerland.

Canalization

- Certain characteristics resistant to environmental input
- Narrow bandwidth of change regardless of a wide range of environmental contexts
  - Example: Language Acquisition

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Gene-Environment Relationships

- **Passive links**: parents transmit traits through genes, the environments they provide, or both.
- **Evocative links**: people react to the characteristics of the child's genotype.
- **Active links (niche-selection)** : children seek out environments compatible with their genotypes.

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Studying Genetic Influence

- Difficult
  - can't control environments thus no cause-effect analysis
  - Don't know specific gene-behavior relationship
  - Can't detail the environment of gene expression
  - Many behaviors controlled by multiple genes in interaction with the environment

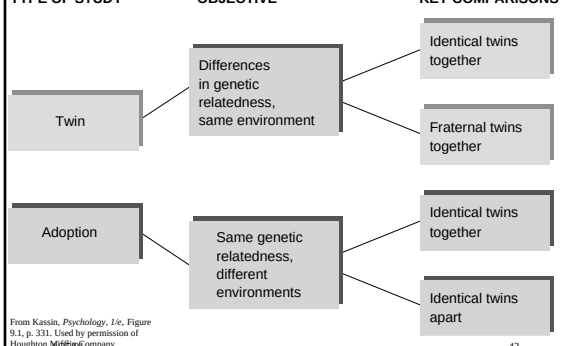
Methods to estimate Genetic Influence

- Kinship Studies
  - Determine degree of genetic closeness
    - Parents-children - 50%
    - Siblings - 50%
    - MZ twins - 100%
    - Half siblings - 25%
  - Relate genetic closeness with trait similarity
    - If trait similarity increases with genetic similarity then evidence for heritability

Kinship Study Designs

- Family Studies
  - Relatives within a household are compared
  - Problem: Shared environment also
- Twin Studies
  - Comparison of MZ and DZ twins
- Adoption Studies
  - Comparison of child to biological and adopted parents, siblings
  - Comparison of twins reared apart v.s. together

Measuring the Effects of Nature and Nurture: Twin and Adoption Studies



From Kassin, *Psychology, 1/e*, Figure 9.1, p. 331. Used by permission of Houghton Mifflin Company.

Concordance rates for some Behavioral & Personality Disorders

Twin Concordances	Identical Twins	Fraternal Twins
Conduct disorder	.85	.70
Manic depression	.65	.20
Autism	.65	.10
Unipolar depression	.45	.20
Alcoholism—males	.40	.20
Schizophrenia	.40	.10
Alcoholism—females	.30	.25

Source: Data from Plomin, 1994.

* MZ twins raised apart are close to but below MZ twins raised together

What does this mean?

- There does seem to be a genetic influence on many traits (Evidence?)
- The environment still plays a major role. (Evidence?)