Chapter 6

Are You Only as Smart as Your Genes? Mendelian and Quantitative Genetics

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6.1 The Inheritance of Traits

- Most children are similar to their parents
- Children tend to be similar to siblings
- Each child is a combination of parental traits
- The combination of paternal traits and maternal traits is unique for each individual child

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The Inheritance of Traits

The human **life cycle:**

- _____ (a male **sperm** cell + a female **egg** cell) fuse during _____ to form a single celled _____, or embryo
- the embryo grows by cell division in mitosis
- the embryo grows into a child
- the child matures into an adult

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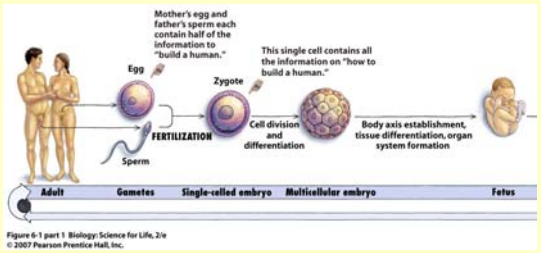

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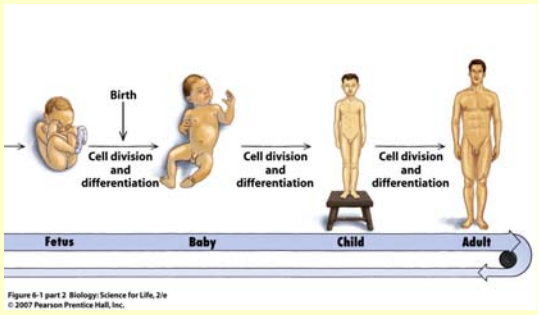

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Genes

- Most genes are segments of DNA that carry information about how to make proteins
  - _____ proteins – for things like hair
  - _____ proteins – for things like breaking down lactose

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Genes

- All cells have the same genes
- Only certain genes are active in a single cell
  - Heart cells and eye cells have genes for the protein rhodopsin, which helps to detect light
  - This is only produced in eye cells, not heart cells

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Genes and Chromosomes

- DNA is sort of like an instruction manual that shows how to build and maintain a living organism…

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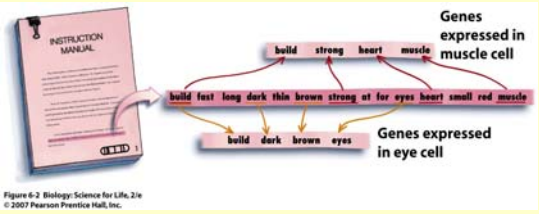

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Genes Are on Chromosomes

- The genes are located on the chromosomes
- The number of chromosomes depends on the organism
  - Bacteria – one circular chromosome
  - Humans – 23 _____ **pairs** of linear chromosomes

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Genes Are on Chromosomes

- Each of the 23 pairs of chromosomes is a homologous pair that carry the same gene
- For each homologous pair, one came from mom and the other from dad

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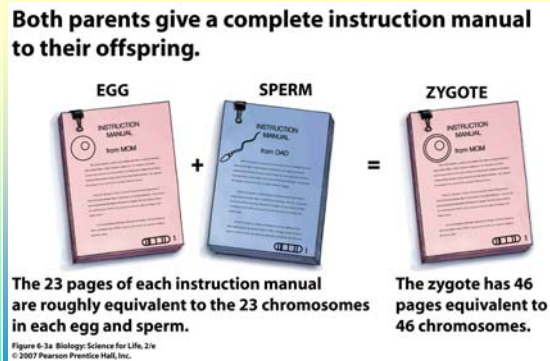

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Gene Variation Is Caused by Mutation

- Genes on a homologous pair are the same, but the exact information may not be the same
- Sometimes there are errors or _____
- Mutations in gene copies can cause somewhat different proteins to be produced
- Different gene versions are called _____

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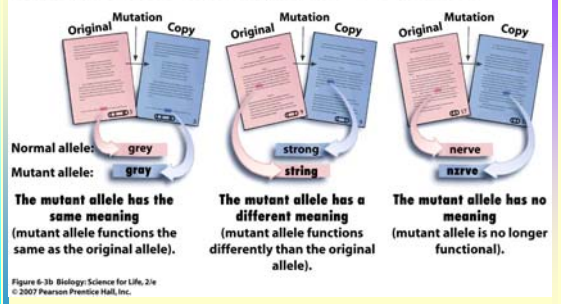
Mutations are errors in copying the instructions.



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Diversity in Offspring

- It is the combination of alleles from both parents that helps determine what traits an individual has
- Environment also plays a role
  - One reason non-twin siblings are dissimilar is that each developed in dissimilar conditions
    - ✓ Mother's nutrition during pregnancy
    - ✓ Presence of toxic compounds in her environment
    - ✓ Number of siblings in the family at the time of birth
- Although differences in the environment of development can lead to differences between siblings, the primary reason non-twin siblings are not identical is that their parents did not give all of their offspring exactly the same set of alleles

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Diversity in Offspring

Non-twin siblings:

- The combination of alleles each individual receives dependes on the gametes that were part of the fertilization event
- Remember that each gamete has 1 copy of each homologous pair

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Segregation

- When a gamete is formed, the homologous pairs are separated and segregated into separate gametes (this is called the **law of segregation**)
- This results in gametes with only 23 chromosomes
  - 1 of each homologous pair

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Independent Assortment

- Due to **independent assortment**, parents contribute a unique subset of alleles to each of their non-identical twin offspring

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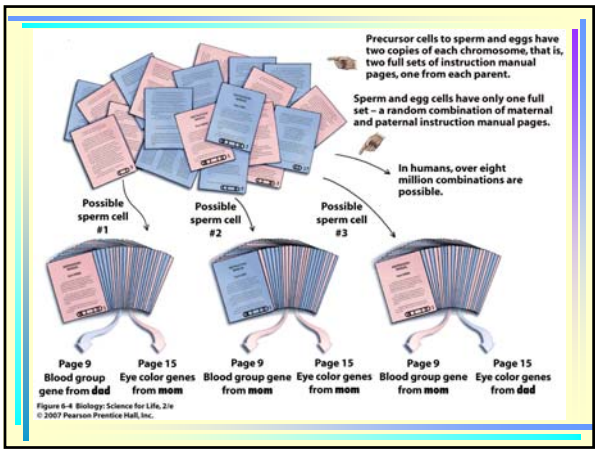

Diversity in Offspring

- See also Figure 6.5 (p. 144)
- Since each gamete is produced independently, the combination of genes is unique

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Diversity in Offspring

- That means a unique egg will be fertilized by a unique sperm to produce a unique child
- For each gene, there is a 50% chance of having the same allele as a sibling
- There are 2^{23} combinations for the way the homologous chromosomes could line up and separate
- This is more than 8 million combinations

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Crossing Over

- In addition, crossing over in meiosis can increase diversity
- The chromosomes trade information, creating new combinations of information
- Each chromosome that results from crossing over consists of a combination of alleles that have never been found together before
- Independent assortment and crossing over create almost limitless variations in eggs or sperm from a single parent

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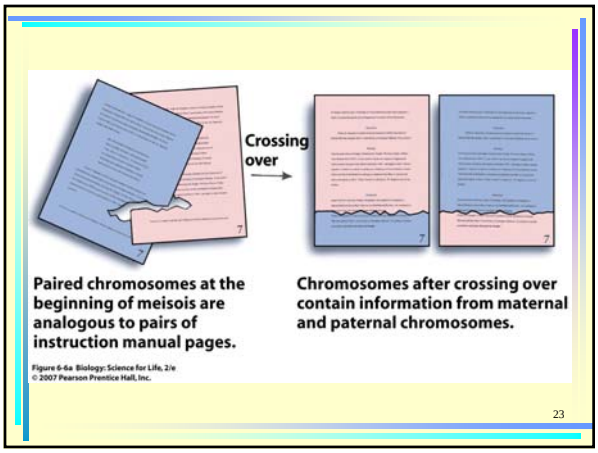

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Random Fertilization

- Gametes combine randomly—without regard to the alleles they carry in a process known as **random fertilization** (any sperm produced by your father had an equal chance of fertilizing any egg produced by your mother
- You are one out of 64 trillion genetically different children that your parents could produce

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Diversity in Offspring

- Mutation, independent assortment, crossing over, and random fertilization result in unique combinations of alleles
- These processes produce the diversity of individuals found in humans and all other sexually reproducing biological populations

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Twins

- Fraternal (non-identical)
  - _____ – two separate fertilized eggs (2 separate eggs fuse with different sperm)
  - Not genetically the same

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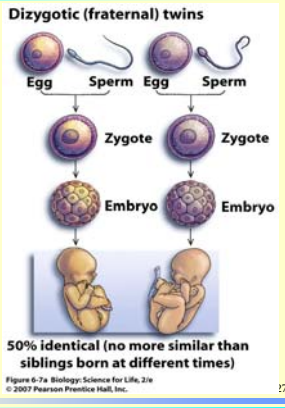
In humans, about 1 in every 80 pregnancies produce dizygotic twins



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Twins

- Identical
  - _____: one single fertilized egg that separates
  - Genetically the same

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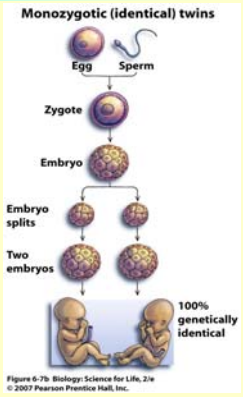
In humans, ~ 1 of every 285 pregnancies results in identical twins



6.2 Mendelian Genetics: When the Role of Genes Is Clear

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Gregor Mendel

- Determined how traits were inherited
- Used pea plants and analyzed traits of parents and offspring

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Mendelian Genetics

- Mendelian genetics – the pattern of inheritance described by Mendel – for single genes with a few distinct alleles
- Sometimes inheritance is not so straightforward
- Many of the Mendelian traits identified in humans are the result of genes with mutant alleles that result in some type of disease or dysfunction

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Genotype

- **Genotype** – combination of alleles
  - _____: two of the same allele
  - _____: two different alleles

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Phenotype

- **Phenotype**
  - _____
  - Depends on nature of alleles

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Mendelian Genetics

- _____ – can mask a recessive allele
- _____ – can be masked by a dominant allele
- _____ – alleles produce an intermediate phenotype
- _____ – both alleles are fully expressed

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Mendelian Genetics

Dominant alleles – capital letter

- For example: ***T*** for tall

Recessive alleles – lower case letter

- For example: ***t*** for short

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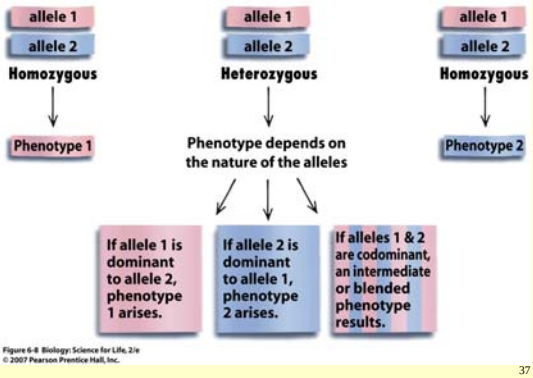

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Genetic Diseases in Humans

- Most alleles do not cause diseases in humans
- There are some diseases that are genetic:
  - Recessive, such as cystic fibrosis
  - Dominant, such as Huntington's Disease
  - Codominant, such as sickle-cell anemia

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Cystic Fibrosis

- Affects 1 in 2500 individuals in European populations
- Recessive condition: individuals have 2 copies of cystic fibrosis allele
- Carriers – have one cystic fibrosis allele but do not have cystic fibrosis
  - Can pass the allele to children

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Cystic Fibrosis

- Produces nonfunctioning protein
- Normal protein transports chloride ion in and out of cells in lungs, intestines and other organs
- Result – balance between sodium and chloride in the cell is disrupted, and thus a thick mucus layer is produced instead of the think slick mucus produced by cells with normal allele
  - This is difficult to clear out of lungs and interferes with absorption of nutrients in intestines

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Huntington's Disease

- Dominant condition
- Fatal condition
- Only one Huntington's allele needed
- Produces abnormal protein that clumps up inside nuclei of cells – especially nerve cells in the brain
- Symptoms: restlessness, irritability, and difficulty in walking, thinking, and remembering
- Disease is progressive and incurable – the nervous, mental, and muscular symptoms gradually become worse and eventually result in death

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Sickle-Cell Anemia

- Codominant – both alleles are expressed
- One allele codes for normal hemoglobin and the other codes for sickle-cell hemoglobin

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Sickle-Cell Anemia

- If you have two normal hemoglobin alleles, you do not have the disease
- If you have two sickle-cell hemoglobin alleles, you have sickle-cell disease
- If you have one of each, you are a carrier
  - These people are resistant to malaria – the parasite is unable to reproduce within cells containing the abnormal hemoglobin; thus infection does not cause severe malarial disease in heterozygotes

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Punnett Squares

- **Punnette square** – a table that lists the different kinds of sperm or eggs parents can produce relative to the gene or genes in question and then predicts the possible outcomes of a **cross**, or mating, between these parents
- Punnett squares are used to predict offspring phenotypes
- Uses possible gametes from parents to predict possible offspring

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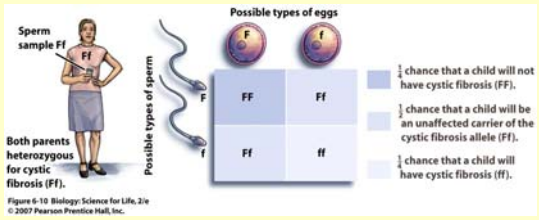

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Punnett Squares: Single Gene

- A parent who is heterozygous for a trait
  - Aa can produce two possible gametes
    A or a
- A parent who is homozygous for a trait
  - AA can only produce gametes with A

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Punnett Squares

- The possible gametes are listed along the top and side of the square
- The predicted offspring genotypes are filled in the center boxes of the square

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Punnett Squares

- The offspring can be homozygous or heterozygous
- It all depends on the parents and the possible gametes
- Punnet squares can be used to predict possibilities of inheriting genetic diseases

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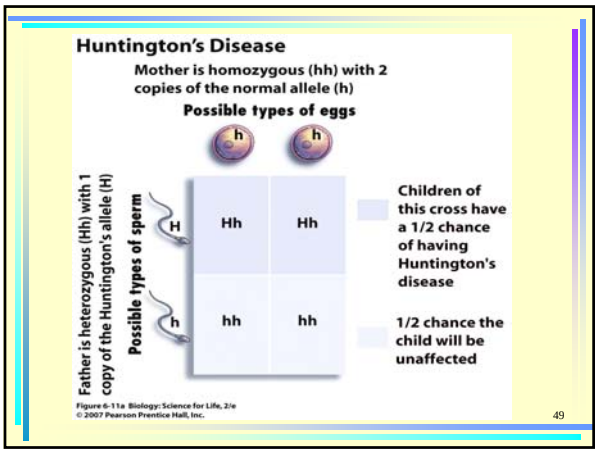

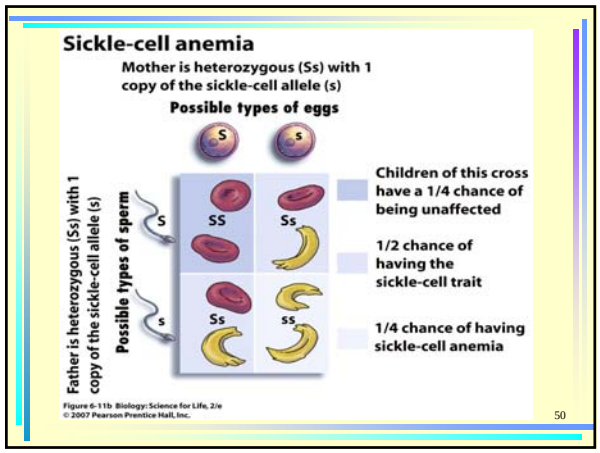

Punnett Squares

- This is a probability for each individual offspring
- If there is a 25% chance an offspring will have cystic fibrosis – this means that – for every fertilization event, there is a 25% chance of cystic fibrosis

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Punnett Squares: Multiple Genes

- You can also use Punnett squares to predict the offspring with multiple genes
- It is significantly more difficult as the number of genes being studied increases

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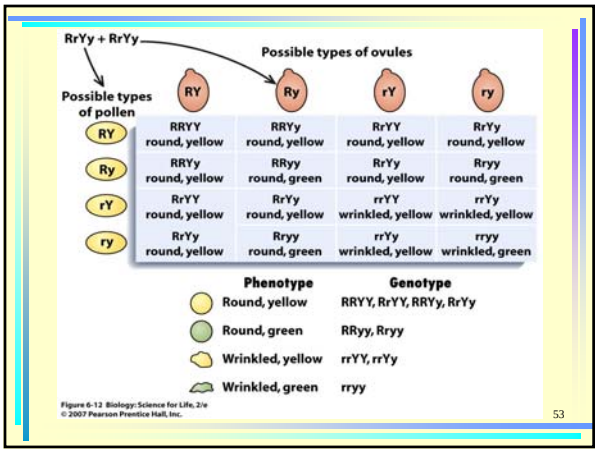

6.3 Quantitative Genetics: When Genes and Environment Interact

- Single-gene traits have a distinct "off or on" character; individuals have either one phenotype or the other
  - These traits are known as qualitative traits
- Many traits do not have this off-or-on character – these traits are called quantitative traits
- Quantitative traits include weight, musical ability, susceptibility to cancer, intelligence, and height
- **Quantitative traits** show _____; we can see a large range of phenotypes in the population
- The amount of variation in a population is called _____

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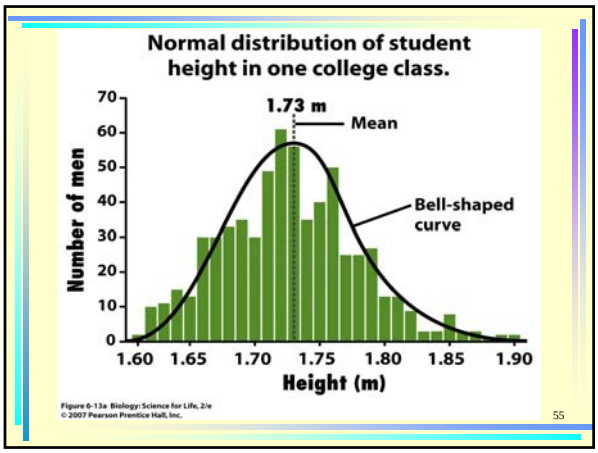

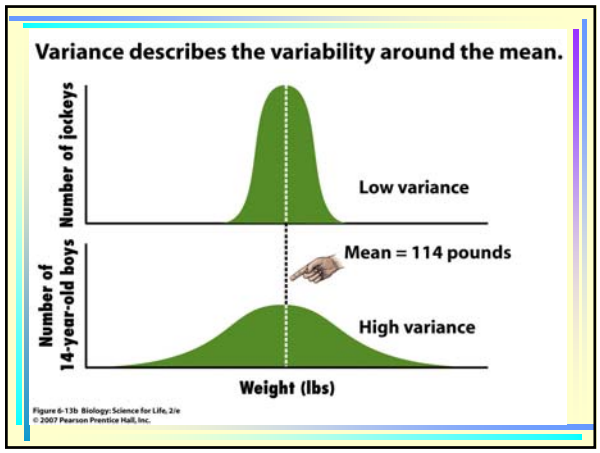

Why Traits Are Quantitative

- One reason we see a range of phenotypes in the human population is because numerous genotypes exist among individuals in the population
  - This occurs when a trait is influenced by more than one gene
- _____ – those traits influence by more than one gene
  - Ex: eye color – influenced by at least 3 genes, each with more than one allele; these genes help produce and distribute the pigment melanin to the iris
  - This produces a range of eye colors

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Why Traits Are Quantitative

- Continuous variation also may occur in a quantitative trait due to the influence of environmental factors
  - In this case, each genotype is capable of producing a range of phenotypes depending on outside influences
  - Thus, even if all individuals have the same genotype, many different phenotypes can result if they are raised in a variety of environments
  - Ex: identical twins with the same genotypes may not have exactly the same appearance…

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Why Traits Are Quantitative

- Most traits that show continuous variation are influenced by both genes and the effect of differing environmental factors
  - Skin color is affected by both genes and environment
  - A number of genes have an effect on skin-color phenotype – both those that influence melanin production and those that affect the distribution of melanin in the skin
  - The environment, particularly the amount of sun exposure to the sun during a season or lifetime, also influences the skin color of individuals

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Genes

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Environment

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Using Heritability to Analyze Inheritance

- Inheritance patterns for quantitative traits are difficult to understand (unlike qualitative traits where the relationship between genes and traits is very clear)
- Researchers use plants and domestic animals to study _____ – the amount of variation for a trait in a population that can be explained by differences in genes among individuals

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Using Heritability to Analyze Inheritance

- **Artificial selection**:
  - technique of controlling the reproduction of individual organisms to influence the phenotype of the next generation (to achieve the desired offspring)

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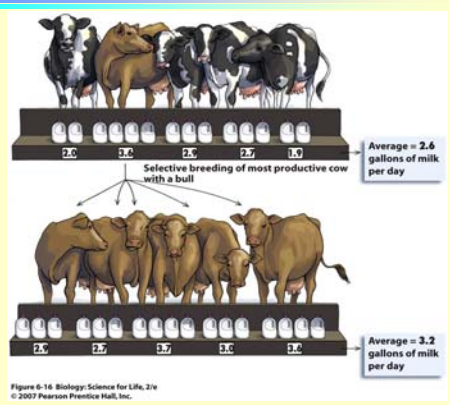

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Calculating Heritability in Human Populations

- Studies of the relative influence of genes and environment that use response to artificial selection cannot be performed in human population
  - It is ethically and socially unacceptable to design breeding programs to produce people with various traits, or select men and women who will produce the next generation
- So we look at **correlations**

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Correlations between Parents and Children

- For human IQ, the correlation between parents and offspring is 0.42 – in other words, 42% of IQ variation among people is the result of differences among them in their genes
- There is also an effect of society and environment on IQ
- Nature versus nurture debate – "born that way" or because they were "raised that way"

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Correlations between Parents and Children

- The impossibility of using traditional selection studies, and the difficulty of separating genetic and environmental influences in most families compels researches interested in the heritability of traits in humans to use **natural experiments**
  - Situations where unique circumstances allow a hypothesis test without prior intervention by researchers
  - Human twins are one source of a natural experiment to test hypotheses about the heritability of quantitative traits

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Correlation between Twins

Twin studies:

- Allow scientists to test the effects of environment
- The DNA is identical in identical twins but the environment may be different
- Compare monozygotic (identical) twins to dizygotic (fraternal) twins
- Study twins raised together
- Study identical twins raised apart and compare to fraternal twins raised apart

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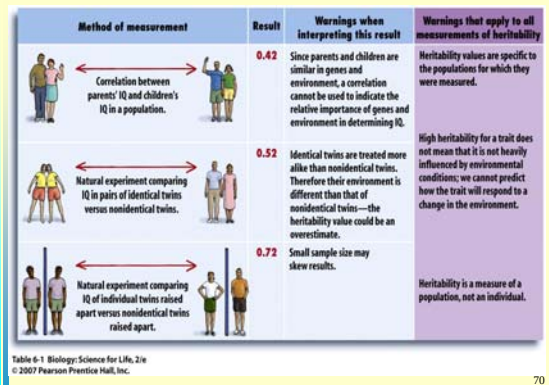

Method of measurement	Result	Warnings when interpreting this result	Warnings that apply to all measurements of heritability
Correlation between parents' IQ and children's IQ in a population.	0.42	Since parents and children are similar in genes and environment, a correlation cannot be used to indicate the relative importance of genes and environment in determining IQ.	Heritability values are specific to the populations for which they were measured.
Natural experiment comparing IQ in pairs of identical twins versus nonidentical twins.	0.52	Identical twins are treated more alike than nonidentical twins. Therefore their environment is different than that of nonidentical twins—the heritability value could be an overestimate.	High heritability for a trait does not mean that it is not heavily influenced by environmental conditions; we cannot predict how the trait will respond to a change in the environment.
Natural experiment comparing IQ of individual twins raised apart versus nonidentical twins raised apart.	0.72	Small sample size may skew results.	Heritability is a measure of a population, not an individual.

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6.4 Genes, Environment, and the Individual

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The Use and Misuse of Heritability

- Calculated heritability values are unique to a particular environment
- Therefore, we must be cautious when using heritability to measure the general importance of genes to the development of a trait
- The environment may cause large differences among individuals, even if a trait has high heritability
- A "thought experiment" involving mice can show this…

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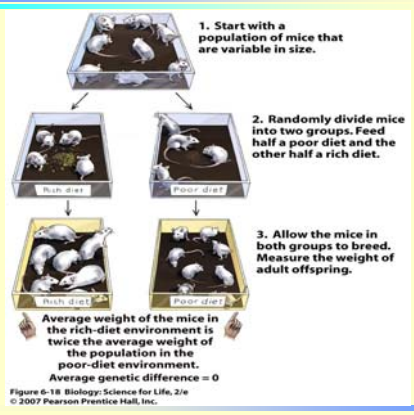

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The Use and Misuse of Heritability

- Highly heritable traits can respond to environmental change
- Traits can be both highly heritable and strongly influenced by the environment
- Knowing the heritability of a trait does not tell us why two individuals differ for that trait
- Our current understanding of the relationship between genes and complex traits does not allow us to predict the phenotype of a particular offspring from the phenotype of its parents

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How Do Genes Matter?

- We know that genes can have a strong influence on eye color, the risk of genetic diseases, susceptibility to heart attack, and even on the structure of the brain
- But what really determines who we are – nature or nurture?
- Both our genes and environment have a profound influence on our physical and mental characteristics

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How Do Genes Matter?

- Possessing functional genes is imperative to the proper development of a human being
- Our cells carry instructions for all of the essential characteristics of humanity, but the process of developing from embryo to adult takes place in a physical and social environment that influences how these genes are expressed
- Scientists are still a long way from understanding how all of these complex, interacting circumstances result in who we are

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