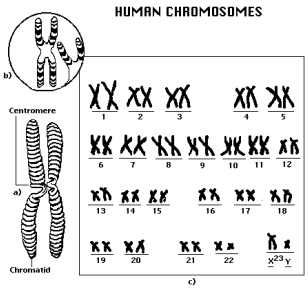
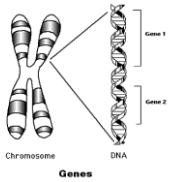


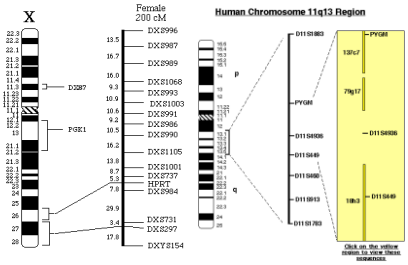
The Human Genome

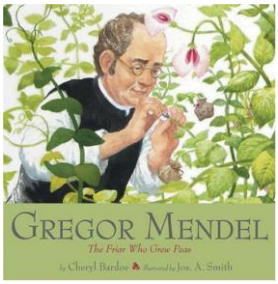
Humans have 23 pairs of chromosomes with 1 of each pair coming from Mom, the other from Dad.

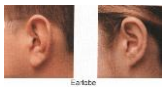


- Chromosomes consist of long strands of DNA, whose structure is often described as a **double helix or twisted ladder**.
- "Genes" or genetic instructions are portions of this "twisted ladder". A particular chromosome may contain over 1000 different genes down its length.

Each Chromosome Pair Shows Distinctive Banding When Stained



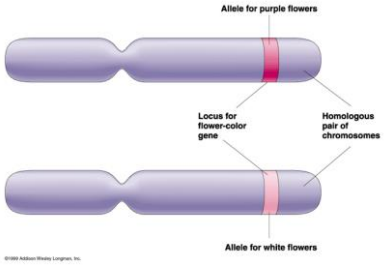




Simple Mendelian Genetics

- Studied traits and how they are passed on to offspring
- 2 possible "alleles" or variants of a gene for simple traits
- The combination you have (1 from mom, 1 from dad) = your "genotype" for that trait
- 2 matching alleles = "homozygous"
- 2 different alleles = "heterozygous"

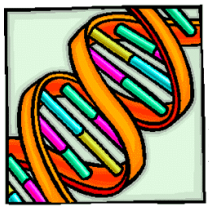
Heterozygous Example



Mendelian Genetics

- If you are heterozygous for a trait, the stronger or "dominant" allele is "expressed" (seen in individual's appearance)
- The traits that are expressed or seen are your "phenotype"- e.g. brown eyed
- The weaker allele not expressed is the "recessive" allele. Though not expressed, it is part of your "genotype" & can be passed on to your kids. Recessive trait is usually only expressed when you inherit the recessive allele from both parents.
- Dominant alleles indicated by upper-case letters
- More than 2 varieties of alleles for many traits

"Nucleotide Base Pairs" Form the "Rungs" of the Double Strand DNA "Ladder"



- Human genome contains about 3 billion of these nucleotide base pair "rungs". Sequences of these base pairs makeup our ~ 25,000 genes.

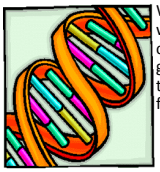
Pairs:

Adenine - Thymine

Guanine – Cytosine

These 4 are the "letters" of our genetic alphabet

Nucleotide "Rungs" of DNA



We share 99.8% genes in common with other humans, 98% genes in common with chimps, and lots of genes in common with all living things (e.g. ½ the banana genome found in the human genome)

Where Do Your Genes Comes From?

- http://www.youtube.com/watch?v=lJzZ7p-47P8&feature=related

Punnett's Square

- A tool for visualizing the possible genotypes of offspring (or, conversely, to help you figure out the genotype of a parent)
- Letters used are arbitrary but Uppercase=dominant & lowercase = recessive
- (If a parent shows the recessive trait , all kids will have a recessive allele in their genotype)

Father's genotype \ Mother's genotype	n	n
N	Nn	Nn
n	nn	nn

Another Example

- If both parents express the dominant trait, but they have a child showing the recessive trait, then both parents must be heterozygous.

	A	a
A	AA	Aa
a	Aa	aa

Genetics Assignment

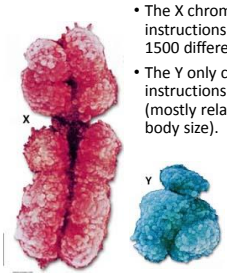
- Explore family member phenotypes and predict the genotypes for 6 traits, such as
- Genetics assignment due 9/7 BUT the practice genetics quiz linked to syllabus is due by this Wed 8/29

Family Member	Toes
Linda (mom)	2nd toe shorter (phenotype) tt (genotype)
Jim (dad)	2nd toe longer T__
Jen (daughter)	2nd toe longer Tt
Sara (daughter)	2nd toe longer Tt
Annie (daughter)	2nd toe longer Tt

Fill out your assignment this way.

Family Member	Tongue Rolling
Linda	Can't roll (phenotype) rr (genotype)
Jim	Can roll R__
Jen	Can roll Rr
Sara	Can roll Rr
Annie	Can roll Rr

X and Y Chromosomes



- The X chromsome contains instructions for building ~ 1500 different proteins.
- The Y only contains instructions for 27 proteins (mostly related to testes & body size).

Sex-Linked Genes (X-linked genes)

- In the case of genes located on the X , females (who have 2 X's) would have the usual 2 alleles for each gene.
- Males, however, only have 1 X chromosome and thus 1 allele at many loci If a male inherits a recessive gene on his X, it is expressed
- Example – X-linked recessive red/green color blindness gene; X-linked hemophilia

Single Nucleotide Differences in a Genetic Instruction: A Genetic Typo

- "Single Nucleotide Polymorphisms" or SNPs
- http://www.youtube.com/watch?v=5raJePXu0OQ&feature=related

Recent data suggests that small genetic errors ("microdeletions" and "microduplications" in brain related genes may explain schizophrenia>

Genetic Does Not Necessarily Mean Unmodifiable
Example: Phenylketonuria (PKU)

- Metabolic disorder transmitted by recessive gene on Chromosome 12, causing a lack of the enzyme *phenylalanine hydroxylase* and a toxic buildup of *phenylalanine*.
- This impairs brain development & causes mental retardation, hyperactivity, hyperirritability, possible seizures. Also low levels of dopamine.

Characteristics of PKU

- How common the recessive gene is varies in different populations
- About 1 in a 100 Caucasians of European ancestry (especially Scots and Irish), fewer Asians, and almost no Blacks carry the recessive gene.
- ~1 in 10,000 babies born with PKU (those who get the recessive gene from each of their parents).

But remember:

- Even in genetic disorders "Nature" may interact with "nurture"
- PKU's effects depend on the environment (diet consumed)
- With a special diet (avoiding protein & aspartame), most of PKU's effects are prevented
- All states require screening of newborns before they leave the hospital

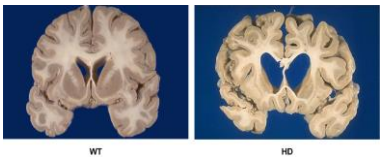
http://www.youtube.com/watch?v=KUJVujhHxPQ

Huntington's Disease

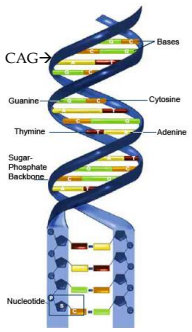
(p. 361,364-365 & syllabus links)

- Hereditary disease transmitted by a dominant gene on chromosome 4 (about 30,000 US cases with 150,000 at risk kids)
- Causes progressive deterioration of the basal ganglia caudate & putamen & cortex, with some damage to additional regions as well. Symptoms usually begin at age 30-50.
- Characterized by involuntary movements (chorea) that interfere with normal movement and speech, depression, progressive dementia, psychosis, circadian rhythm problems
- Death due to health complications in 15-20 yrs

Normal vs Huntington's Disease

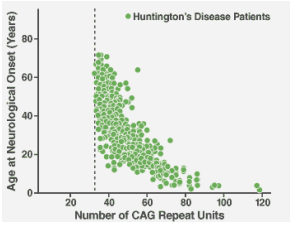


Abnormal protein leads to death of basal ganglia neurons



- On chromosome 4 there is an abnormal # of CAG repeats, >35 instead of usual 24 or fewer, resulting in an **abnormal form** of protein known as huntingtin
- If there are >60 CAGs, symptoms appear earlier
- If just 36-38, symps may not occur until old age
- CAG repeats in genes also occur in several other neurodegenerative conditions.

The more abnormal the genetic instruction, the earlier the onset of symptoms. Less than 35 repeats – don't develop HD



Heritability

- An estimate of the amount of the variance in the behavior/characteristics **of a specific group/population** is due to genetic variation.
- Heritability ranges from 0 (none of variance due to heredity) to 1 (all of the variance due to heredity)
- Heritability depends on the population or group studied
- Heritability may not be constant over time

Variability in Populations Differs

Some populations have less genetic variability, so genetics cannot play as much of a role is explaining variability of traits.

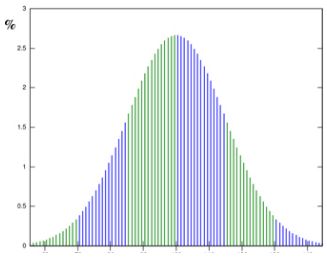
Similarly, some populations have less variability in environment/experiences. Others contain individuals with dramatically different environments or experiences so "nurture" accounts for more variability of traits.

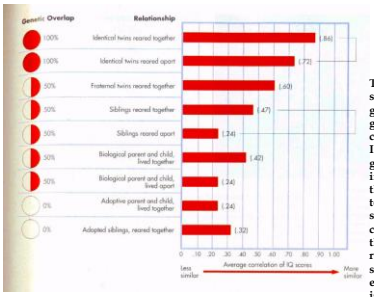
Some populations have more genetic variability and this variability contributes more to the variability in traits.

Methods for Studying Heritability in Humans

- Comparing the similarity of pairs of identical twins vs. the similarity of pairs fraternal twins
- Comparing the similarity of child/ biological parent vs child/adopted parents
- Comparing the similarity of family members of different degrees of relatedness

Variability of IQ Scores





That those sharing more genes show greater correlation in IQ shows genetics is important; that those reared together show stronger correlation than those reared apart shows environment is important.

- It is not just that genetic differences AND environmental differences shape the characteristics of the individual.
- Environmental differences can directly alter gene expression!
- *Epigenetics* is the study of sometimes heritable changes in gene *expression* triggered by *environmental factors.* There is no change in the underlying DNA sequence — a change in **phenotype** occurs (without a change in **genotype**) due genes being switched on or off.

Epigenetics & the Epi-genome

http://www.youtube.com/watch?v=LcaRTDsLmiA&feature=related

- OR this url
- http://video.pbs.org/video/1525107473/
- (can go to 2:30 to start)
- It is not just a matter of what genes you inherit, but also which of those genes are switched on or off.
- (The mice are discussed on p. 181)
- Notice over time environment is having a greater & greater influence.

Identical Twins Can Differ in Gene Expression

- 80 pairs of identical twins of diff. ages
- Examined DNA from several areas
- Few epigenetic diffs in toddler twins
- The older the twin pair, the more diffs in gene *expression*
- The more diff their environments, the more differences in expression.

Sex-Limited Genes

- Some genes, present in both sexes, are only turned on if we are exposed to sufficient levels of a particular sex hormone.
- Sex hormones can act as epigenetic factors influencing which & when genes are expressed
- Example: Any of us may carry the gene for male pattern baldness, but it is only expressed in the presence of significant androgens.
- Other examples: genes for chest hair, breast development, crowing in roosters

What About the Nature-Nurture Issue?

Understanding the influence of genetics vs environment on behavior has gotten complicated!

We now know:

Environment influences genes (certain stimuli can turn on or off genetic instructions)

Genes, in turn, can make one seek out particular environments, or can affect the response to an environmental influence.

The environment, in turn, can magnify or multiply the impact of genes

- Genetic technologies have allowed the mapping of species' genomes.
- Now can alter individual genes (e.g. the knockout technique on p. 114) to study the influence of each gene in animal research.
- Applying genetic manipulation to medical treatment is just beginning to yield "gene therapies" for certain disorders.