Mendel

- What was Mendel's contribution to our understanding of heredity?
- What is the Law of Segregation?
- What is the Law of Independent Assortment?
- What is a Punnett square and how is it used to illustrate the principles of inheritance?

Human Mendelian Traits

- OMIM estimates that about 20,000 genes are inherited in a simple Mendelian way.
- Many blood characteristics and diseases and disorders - some simple physical traits
- Examples: Sickle cell, tented eyebrows, hitchhiker's thumb
- Most human "traits" are more complicated, but particles of inheritance - genes - are passed on in a simple Mendelian way.

Terminology

- Genotype
- Phenotype
- Gene
- Allele
- Locus
- DNA
- Chromosomes
- Bases

Locus = the location of a gene on a chromosome
Allele = alternative form of a locus
Homozygous = having the same allele at the locus on both chromosomes
Heterozygous = having different alleles at the locus on both chromosomes
Heritability - a 3 Part question

• How come we resemble our parents? That is, how is our heritable information passed from generation to generation?
• How does the genetic code create a characteristic?
• Where does variation in the code come from?

Human Karyotype

• MITOSIS - somatic cell division
• MEIOSIS - gametic cell division

Mitosis
Heritability - a 3 Part question

- How come we resemble our parents? That is, how is our heritable information passed from generation to generation?
- How does the genetic code create a characteristic?
- Where does variation in the code come from?
Variation comes from

- Recombination
- Crossing Over
- Mutation

Recombination
23 chromosomes
2 possibilities for each
= 2 to the 23rd power
= 8,388,608
Variation comes from
- Recombination
- Crossing Over
- Mutation

Crossing over during meiosis

Chromosomal mutations
- Down's syndrome -21
- Klinefelter's syndrome -Sex
- Turner's syndrome - sex
- William's Syndrome - 7

Klinefelter's Karyotype
Down's Karyotype

Trisomy 13
Karyotype from a female with Trisomy 13 (47,XX,+13)
- small head
- small eyes
- cleft lip
- ear shape
- palm differences
- extra fingers/toes
- heart defects
- kidney defects
- etc.

Variation comes from
- Recombination
- Crossing Over
- Mutation

Mutation
- Change in base sequence of DNA
- Occurs during replication stage of meiosis (or mitosis)
- MAY change the amino acid change and therefore the protein
Kinds of Mutations

- Substitution - replace one base with another
- Frame Shift -
  - Insertion - an extra base gets pulled in
  - Deletion - a base gets omitted

How common is mutation?

- Happens all the time
- Assume a rate of one in a million per locus per gamete
- Assume approximately 50,000 loci
- \((1 \times 10^{-6}) \times (5 \times 10^{-4}) = 0.05\)
  5% of gametes have a mutation
- An individual is a combination of two gametes
  \(2 \times 0.05 = 0.1 - 10\%\)