

How is genotype determined?

- How does DNA code for the making of proteins?
- How do the two copies of DNA you carry work together to create your phenotype?
- How do you get your two copies of any chromosome or locus through meiosis?

Meiosis

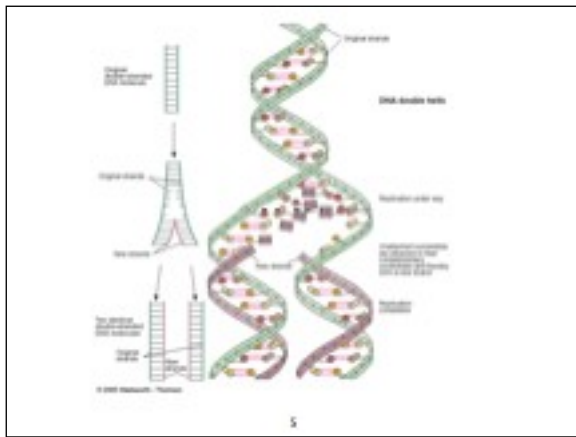
- How does meiosis divide cells?
- What are haploid and diploid cells?
- Describe the process of meiosis?
- When and how during meiosis is variation introduced?
- How do you get new genotypes?

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

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Do all mutations change the protein created?

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Mutations?

- "Bad" - reduces the protein's ability to function causing reduction in fitness
- Neutral - no change in protein form or function
- "Good" - increases protein's ability to function, enhances fitness

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Figure 3.15

How common is mutation?

- happens all the time
- assume a rate of one in 100 million bases
- repair mechanisms fix 99% for effective mutation rate of 10^{-10}
- gives a rate of 130 mutations per individual per generation
- (<http://sandwalk.blogspot.com/2010/11/human-mutation-rates.html>)₁₀

How is phenotype created?

- phenotype = genotype + environment
- genotype = combination of the two particles of inheritance we carry for each locus
- plus other involved loci

Most human traits...

- Polygenic
 - controlled by 2 or more loci
- Affected by the environment
- Many genes PLEIOTROPIC
 - a single gene has multiple effects

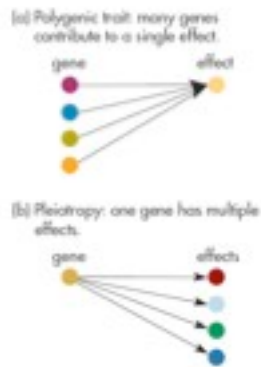
Example: Height

- phenotype = genotype + environment
- environment = diet, altitude, sleep, health
- genotype = pairs of alleles at at least 5 loci (4q35, 9p24, 13q12, 18q21, 22q13)



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Polygeny vs. Pleiotropy



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Pleiotropy in Marfan Syndrome

Mistake making fibrillin-1 → builds elastic tissue

- thinness
- joint hypermobility
- limb elongation
- lens dislocation
- increased risk of heart disease



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Modern Synthesis

- production and redistribution of variation
- Natural selection is one force than can act on this variation to cause change

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Evolution =
changes in gene frequencies
over time

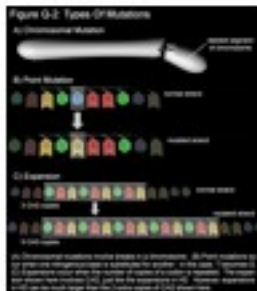
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Four Forces of Evolution

- Mutation
- Gene Flow
- Genetic Drift
- Natural Selection

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Mutation



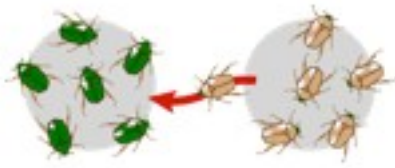
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Mutation

- The only way to introduce new genetic variation
- Very common
 - many neutral
 - many deleterious enough to get removed quickly
 - some are incorporated

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Gene Flow



Movement of alleles within and between populations

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Genetic Drift

- The random factor
- Greatest effect in small populations
- Founder effect

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Genetic Drift - Founder Effect



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Clinodactyly



Retinitis pigmentosa

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