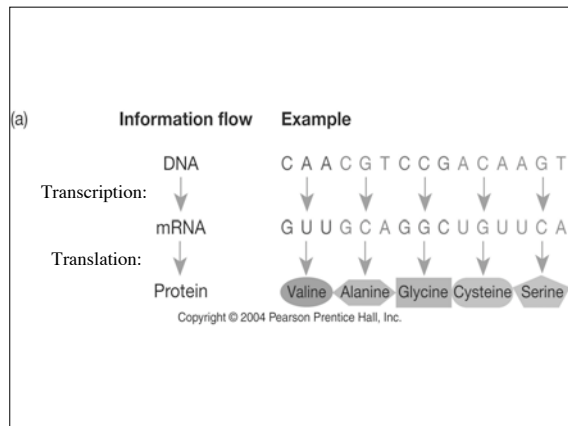


b) The 62 3-letter condons which specify the 20 amino acids. Note The redundancy built in.

| First base | Second base | | | | Third base |
|------------|------------------------|---------------|-------------------|----------------|------------|
| | U | C | A | G | |
| U | UUU Phenylalanine | UCU Serine | UAU Tyrosine | UGU Cysteine | U |
| | UUC Phenylalanine | UCC Serine | UAC Tyrosine | UGC Cysteine | C |
| | UUA Leucine | UCA Serine | UAA Stop | UGA Stop | A |
| | UUG Leucine | UCG Serine | UAG Stop | UGG Tryptophan | G |
| C | CUU Leucine | CCU Proline | CAU Histidine | CGU Arginine | U |
| | CUC Leucine | CCG Proline | CAC Histidine | CGC Arginine | C |
| | CUA Leucine | CCA Proline | CAA Glutamine | CGA Arginine | A |
| | CUG Leucine | CCG Proline | CAG Glutamine | CGG Arginine | G |
| A | AUU Isoleucine | ACU Threonine | AAU Asparagine | AGU Serine | U |
| | AUC Isoleucine | ACC Threonine | AAC Asparagine | AGC Serine | C |
| | AUA Isoleucine | ACA Threonine | AAA Lysine | AGA Arginine | A |
| | AUG Start (Methionine) | ACG Threonine | AAG Lysine | AGG Arginine | G |
| G | GUU Valine | GCU Alanine | GAU Aspartic Acid | GUU Glycine | U |
| | GUC Valine | GCC Alanine | GAC Aspartic Acid | GUC Glycine | C |
| | GUA Valine | GCA Alanine | GAA Glutamic Acid | GGA Glycine | A |
| | GUG Valine | GCG Alanine | GAG Glutamic Acid | GGG Glycine | G |

Codon Amino acid

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Definitions

- Gene-particular stretch of DNA that codes for a distinctive type or RNA or protein
- Allele-versions of the same gene that differ in their base sequence
- Mutations-any type of change in the base sequence of DNA

Point Mutation

- Substitution of one base by another
 - Caused by:
 1. Random error in DNA synthesis
 2. Random errors in the repair of sites damaged by chemical mutagens or high energy radiation

Types of Point Mutations

1. Transition: a purine (A/G) is substituted for another, or a pyrimidine (T/C) is substituted for another
2. Transversion: a purine is substituted for a pyrimidine or *vice versa*
(transitions outnumber transversions 2:1--less disruptive and so less likely to be recognized as an error and fixed)

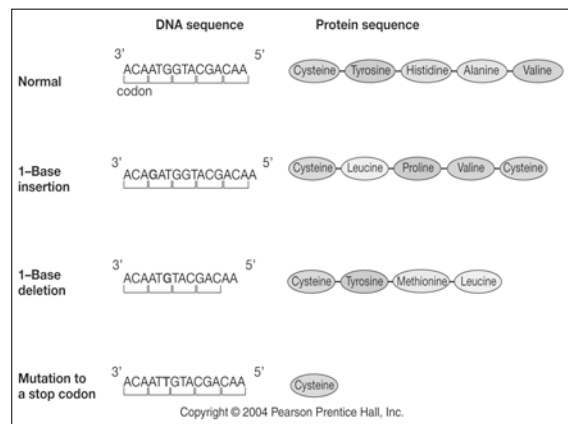
Point Substitutions

1. Replacement substitution--point mutation that results in an amino acid change
2. Silent site substitution--point mutation that does not change an amino acid

Both these result in new alleles.

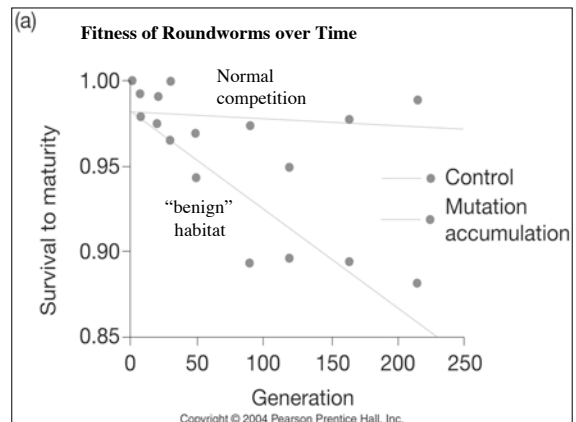
How often do mutations occur?

- Data from **loss of function mutations**--any type of mutation that deactivates a gene
 - Frameshift mutation
 - Insertion
 - Deletion
 - Either of these might create a stop codon and all three would change all subsequent codons creating a dysfunctional gene



DNA sequencing

- With this technique we can look directly for mutations
- Roundworm study looking at mitochondrial DNA in offspring compared to parents and found about 15 new mutations in every individual in every generation
 - Might be higher than in nuclear DNA

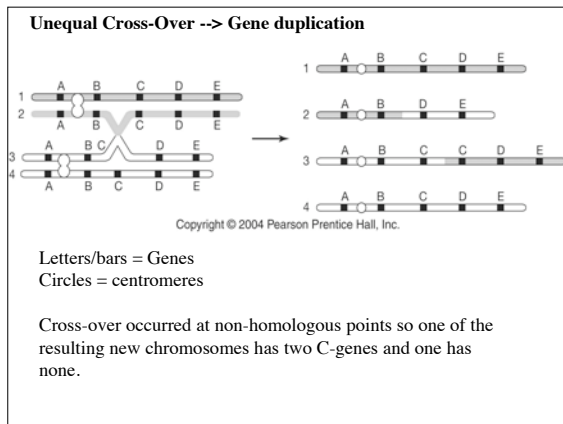


Mutation rates may be selected for:

- Different species have different rates
 - *C. elegans* translation enzymes insert an incorrect base about once in every 100 million nucleotides!
 - Suggests the enzymes themselves have been subject to natural selection
- The rate of repair also varies and so is perhaps also selected for
- Higher mutation rates may be selected for in organisms subject to changing environments

Where do new genes come from?

- Gene duplication resulting from unequal crossover which occurs during meiosis.....



What happens to this new DNA?

1. Retains original function, providing an extra copy of the parent allele
2. Gains a new function through mutation and selection
3. Becomes a functionless pseudogene

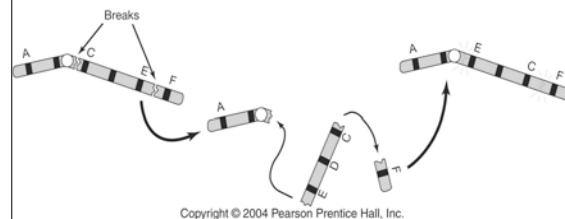
- Human hemoglobin "gene family" of 5 essentially duplicate genes: changes in gene expression producing hemoglobin in embryonic vs. infant humans
- Can determine relatedness based on who shares which duplicate genes: mice share 2 of these duplicates with humans

Chromosome Alterations

- Changes in gross morphology of chromosomes might do the following:
 - Affect just gene order and organization
 - Produce duplications or deletions
 - Involve the entire DNA molecule or just segments

Chromosome alterations important in evolution:

1. Inversions---affect genetic linkage



An example of a **cline**: a regular change in the frequency of an allele or inversion over a geographic area.

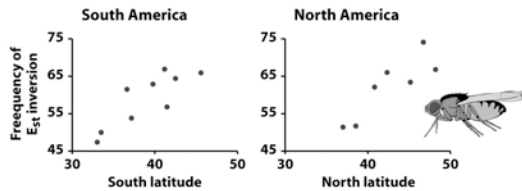


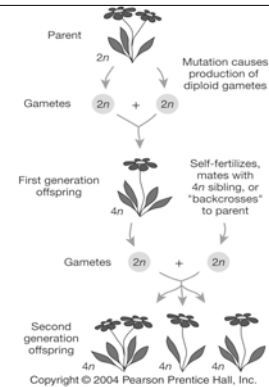
Figure 5-10 Evolutionary Analysis, 4/e
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Different alleles or combinations are better suited to different climate conditions.

Polyploidy--

- instead of being haploid or diploid, an individual can have 4 or 8 sets of chromosomes, or more (tetraploid, octaploid)

- Common in plants
 - Nearly half of all angiosperms and nearly all ferns are polyploidy



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Table 5.2 Types of mutation with significant evolutionary impact: A summary

| Name | Description | Mechanism | Significance |
|----------------------|--|--|---|
| Point mutation | Base pair substitutions in DNA sequences | Chance errors during DNA synthesis or during repair of damaged DNA | Creates new alleles |
| Chromosome inversion | Flipping of a chromosome segment, so order of genes along the chromosome changes | Breaks in DNA caused by radiation or other insults | Alleles inside the inversion are likely to be transmitted together, as a unit |
| Gene duplication | Duplication of a short stretch of DNA, creating an extra copy of the sequence | Unequal crossing-over during meiosis or retrotransposition | Redundant new genes may acquire new functions, by mutation |
| Genome duplication | Addition of a complete set of chromosomes | Errors in meiosis or (in plants) mitosis | May create new species; massive gene duplication |

Table 5.2 Evolutionary Analysis, 4/e
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How much genetic diversity exists in a typical population

1. Percentage of polymorphic loci (a population is polymorphic at a particular locus if more than one allele exists at that particular locus)
2. Heterozygosity (H) is either:
 1. Average frequency of heterozygotes or
 2. Fraction of genes that are heterozygous in the genotype of an average individual

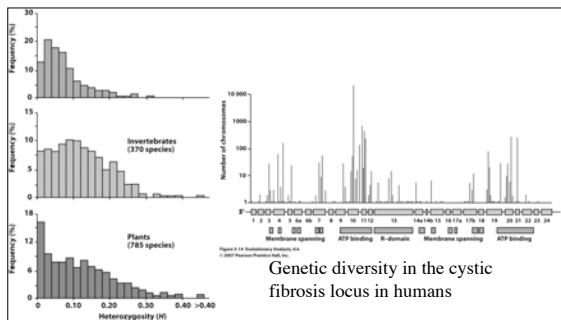


Figure 5-13 Evolutionary Analysis, 4/e
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Genetic diversity in the cystic fibrosis locus in humans

-plus the CCR5+/CCR5-Δ32 HIV-AIDS example

To summarize on diversity:

- In a typical natural population between 30-50% of the loci are polymorphic
- The average individual is heterozygous at 4-15% of its loci

So how is this diversity maintained?

Two Theories of Why There is so Much Genetic Diversity

1. Balance or Selectionist Theory-genetic diversity is maintained by natural selection favoring heterozygotes or rare individuals
2. Neutral Theory-most alleles at polymorphic loci are functionally and therefore selectively equivalent so there is no selection against them