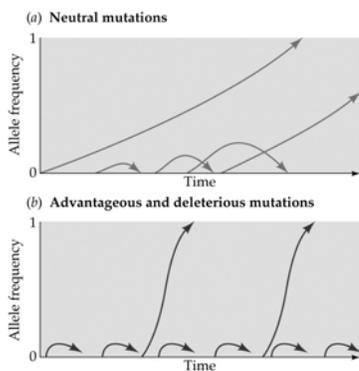


Molecular Evolution & the Origin of Variation



What Is Molecular Evolution?

- **Molecular evolution** differs from phenotypic evolution in that mutations and genetic drift are much more important determinants of molecular evolution.
- The goals of **molecular evolution** studies are to determine patterns of evolutionary change in organisms' molecules, determine the processes that caused the changes, and use those insights to solve other biological problems.
- **Neutral alleles** are fixed slowly, whereas **advantageous and disadvantageous alleles** are fixed rapidly.

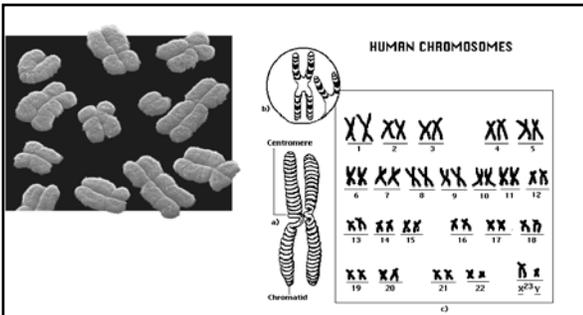


Mechanisms that Act on the Diversity of Genes and Alleles

- Mutation
- Drift (Dominant in Neutral theory)
- Selection (Dominant in Selectionist Theory)

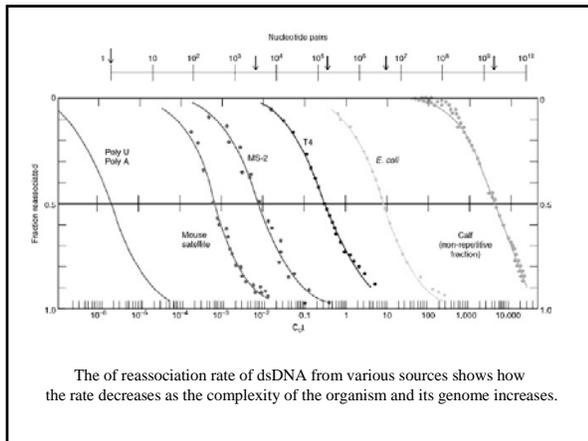
Genome Organization

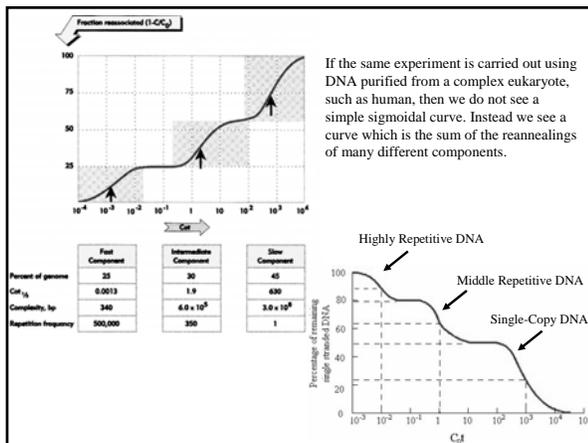
- C₀t curves – Three levels of structure in Eukaryotes.
- Size does not affect complexity of a Genome: “C-value Paradox.”
- Zuckerkandl & Pauling – Clock-like thru time supporting Neutral Theory.

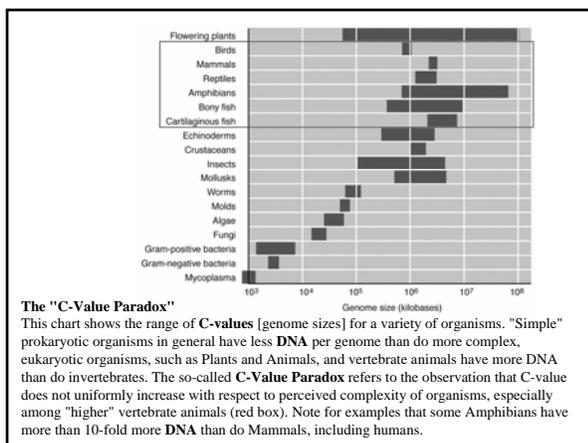


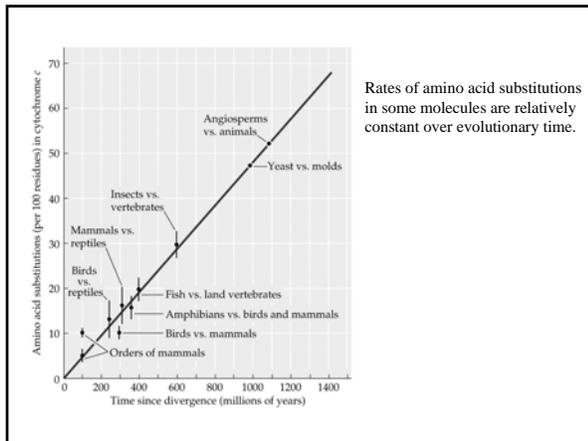
The image contains a micrograph of human chromosomes on the left, showing various sizes and shapes. On the right is a karyotype diagram titled "HUMAN CHROMOSOMES" showing 22 pairs of autosomes and one pair of sex chromosomes (X and Y). A detailed diagram of a single chromosome is shown in the center, with labels for "Centromere" and "Chromatid".

The sum of all the chromosome information is known as a **karyotype** with 22 pairs of **autosomes** and 1 pair of **sex chromosomes**.



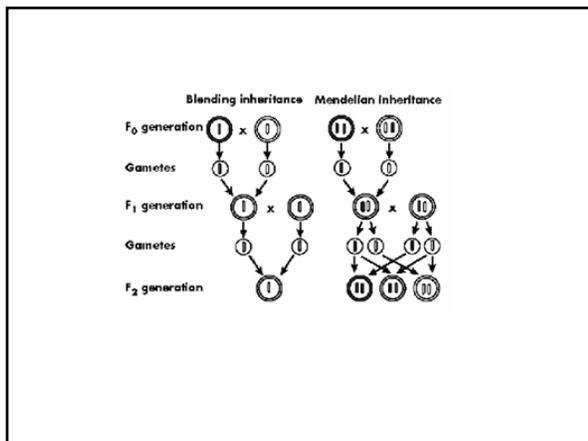


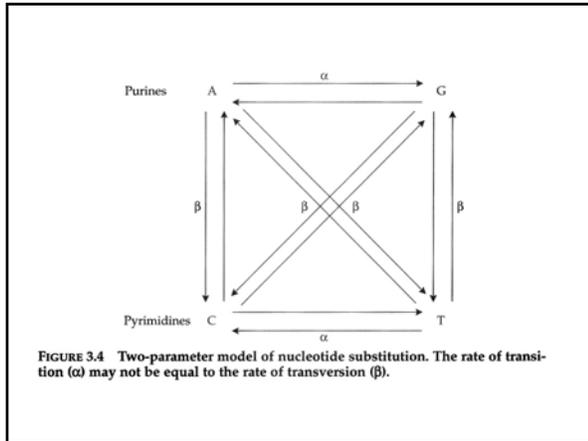




Source of New Genes and Alleles

- Old view: Inheritance of acquired characters.
- New view: Mutation is ultimate source of all variation.
- Rem: Mutations in somatic vs. germ line cells.





Examples of **point mutations** and consequences for mRNA & amino acid sequences

Direction of transcription →

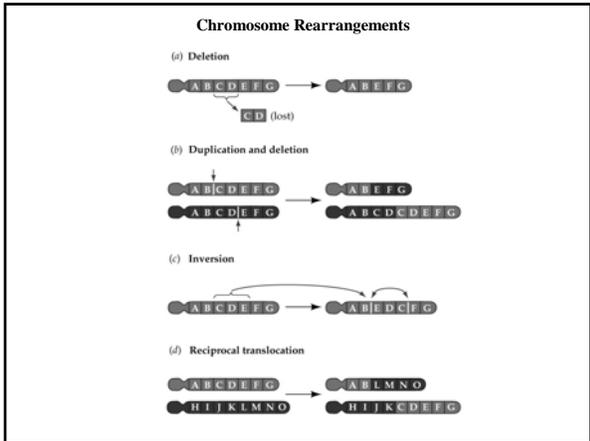
Original sequence: DNA: AGA TGA CCG TTT GCA
 RNA: UCU ACU GCC AAA CGU
 Protein: Ser Thr Ala Lys Arg

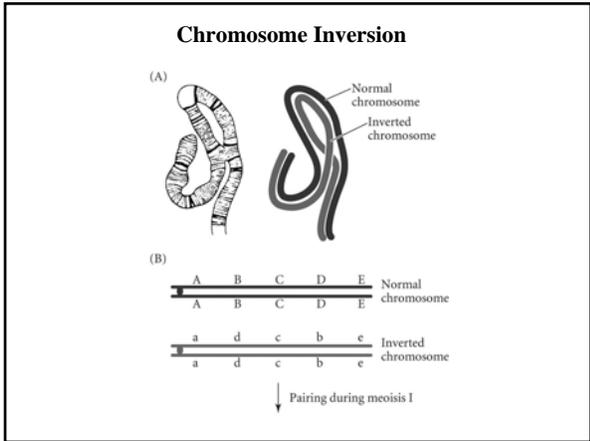
<p>Base pair substitutions</p> <p>Transition (A → G)</p> <table border="1" style="font-size: small;"> <tr><td>GGA</td><td>TGA</td><td>CGG</td><td>TTT</td><td>GCA</td></tr> <tr><td>CCU</td><td>ACU</td><td>GCC</td><td>AAA</td><td>CGU</td></tr> <tr><td>Pro</td><td>Thr</td><td>Ala</td><td>Lys</td><td>Arg</td></tr> </table> <p>Transversion (A → T)</p> <table border="1" style="font-size: small;"> <tr><td>TGA</td><td>TGA</td><td>CGG</td><td>TTT</td><td>GCA</td></tr> <tr><td>ACU</td><td>ACU</td><td>GCC</td><td>AAA</td><td>CGU</td></tr> <tr><td>Thr</td><td>Thr</td><td>Ala</td><td>Lys</td><td>Arg</td></tr> </table>	GGA	TGA	CGG	TTT	GCA	CCU	ACU	GCC	AAA	CGU	Pro	Thr	Ala	Lys	Arg	TGA	TGA	CGG	TTT	GCA	ACU	ACU	GCC	AAA	CGU	Thr	Thr	Ala	Lys	Arg	<p>Frameshifts</p> <p>Insertion (T)...</p> <table border="1" style="font-size: small;"> <tr><td>AGT</td><td>ATG</td><td>AGG</td><td>GTT</td><td>TGG</td><td>A_</td></tr> <tr><td>UCA</td><td>UAC</td><td>UGC</td><td>CAA</td><td>ACG</td><td></td></tr> <tr><td>Ser</td><td>Tyr</td><td>Cys</td><td>Glu</td><td>Thr</td><td></td></tr> </table> <p>...followed by deletion (T)</p> <table border="1" style="font-size: small;"> <tr><td>AGT</td><td>ATGA</td><td>CGG</td><td>TTT</td><td>GCA</td></tr> <tr><td>UCA</td><td>UCU</td><td>GCC</td><td>AAA</td><td>CGU</td></tr> <tr><td>Ser</td><td>Ser</td><td>Ala</td><td>Lys</td><td>Arg</td></tr> </table>	AGT	ATG	AGG	GTT	TGG	A_	UCA	UAC	UGC	CAA	ACG		Ser	Tyr	Cys	Glu	Thr		AGT	ATGA	CGG	TTT	GCA	UCA	UCU	GCC	AAA	CGU	Ser	Ser	Ala	Lys	Arg
GGA	TGA	CGG	TTT	GCA																																																												
CCU	ACU	GCC	AAA	CGU																																																												
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TGA	TGA	CGG	TTT	GCA																																																												
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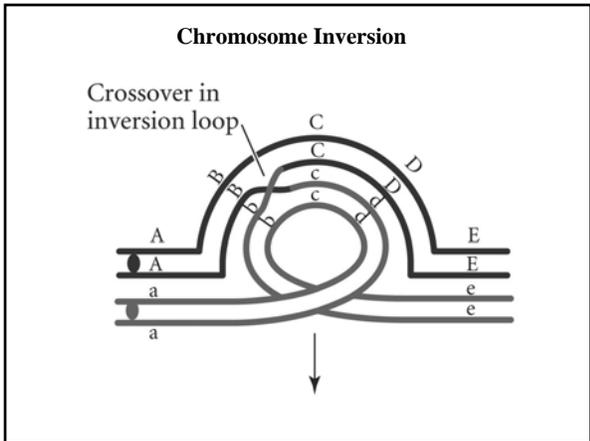
Mutation Rates (rare for most part)

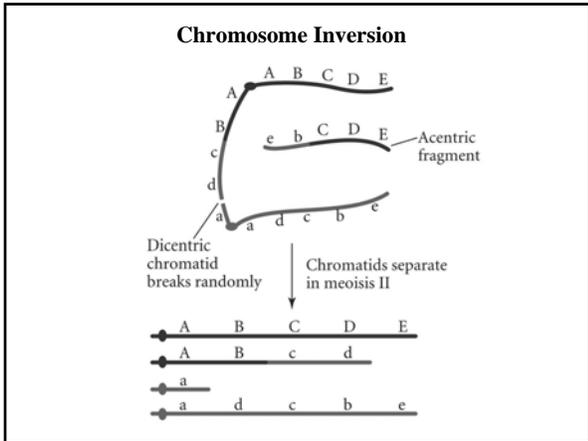
Organism	Base pairs		Mutation rate			
	in haploid genome	in effective genome ^a	per base pair per replication	per replication per haploid genome	per replication per effective genome ^a	per sexual generation per effective genome ^b
T2, T4 phage	1.7 × 10 ⁵	—	2.4 × 10 ⁻⁴	0.0040	—	—
Escherichia coli	4.6 × 10 ⁹	—	5.4 × 10 ⁻¹⁰	0.0025	—	—
Saccharomyces cerevisiae (yeast)	1.2 × 10 ⁷	—	2.2 × 10 ⁻¹⁰	0.0027	—	—
Neurospora crassa (bread mold)	4.2 × 10 ⁷	—	7.2 × 10 ⁻¹⁰	0.0030	—	—
Caenorhabditis elegans	8.0 × 10 ⁷	1.8 × 10 ⁷	2.3 × 10 ⁻¹⁰	0.018	0.004	0.006
Drosophila melanogaster	1.7 × 10 ⁸	1.6 × 10 ⁷	3.4 × 10 ⁻¹⁰	0.058	0.005	0.14
Mouse	2.7 × 10 ⁸	8.0 × 10 ⁷	1.6 × 10 ⁻¹⁰	0.49	0.014	0.9
Human	3.2 × 10 ⁹	8.0 × 10 ⁷	5.0 × 10 ⁻¹⁰	0.16	0.004	1.6

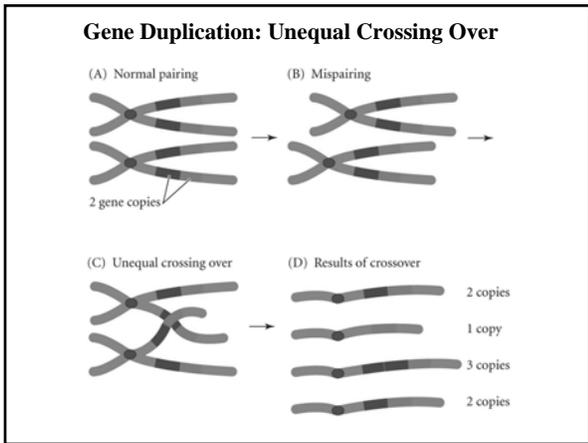
Source: Albert Drake et al., 1988.
^a The effective genome is the number of base pairs in functional sequences that could potentially undergo mutations that reduce fitness.
^b Calculated for multicellular organisms in which multiple DNA replication events occur in development between zygote and gametogenesis.

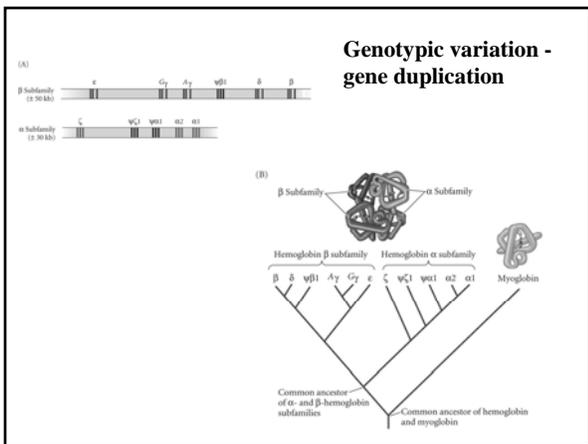


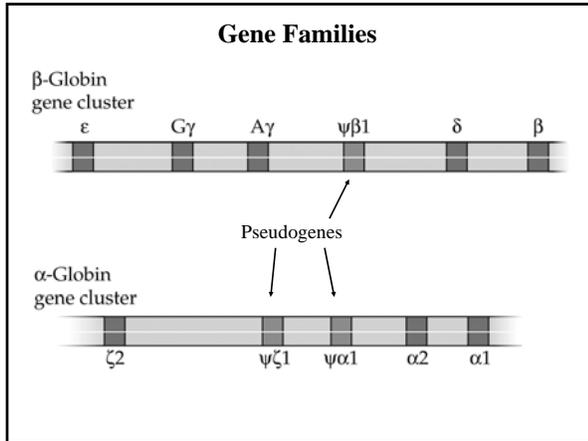


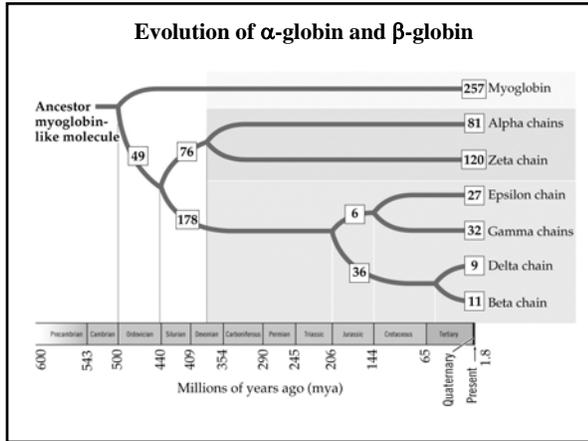


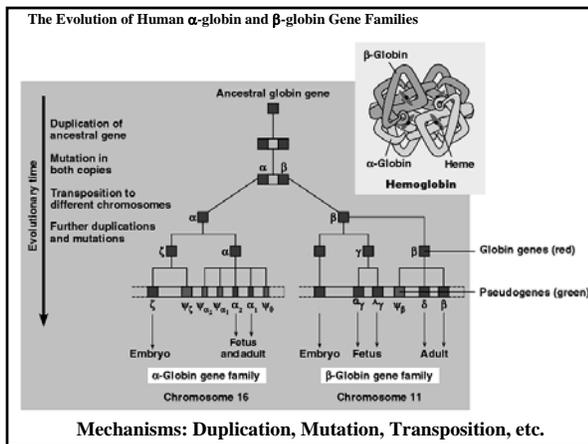


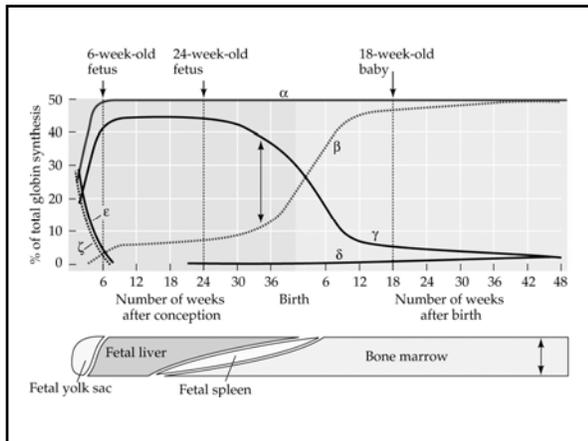


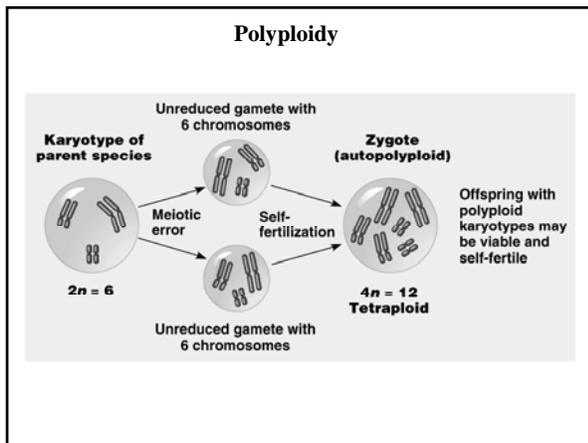


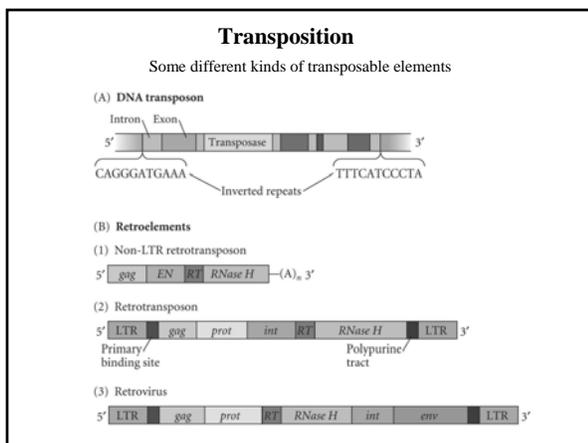






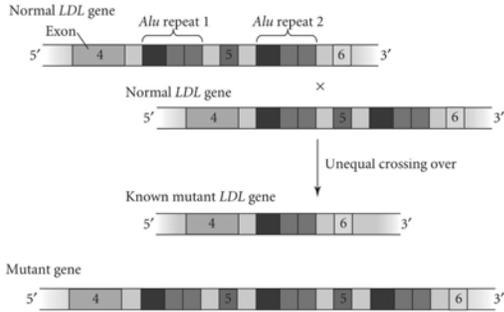






Transposition

A mutated low-density lipoprotein (*LDL*) gene in humans lacks exon 5



Translocation

Muntiacus reevesii (2N = 46)



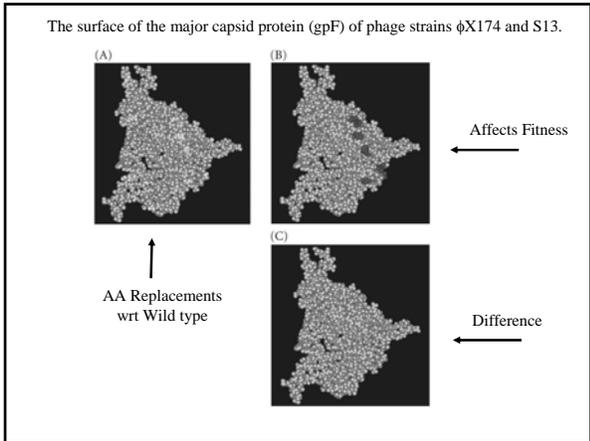
Muntiacus muntiacus (2N = 8)

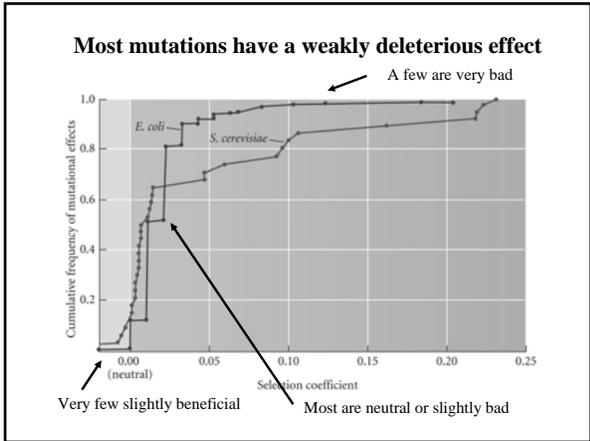


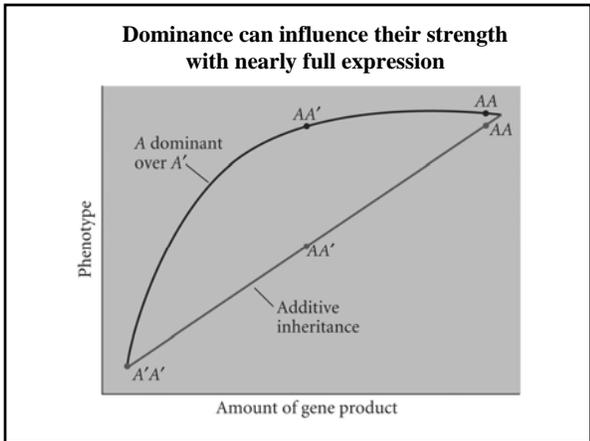
Barking Deer: Similar phenotype, dissimilar karyotype.

Structure and Function Considerations

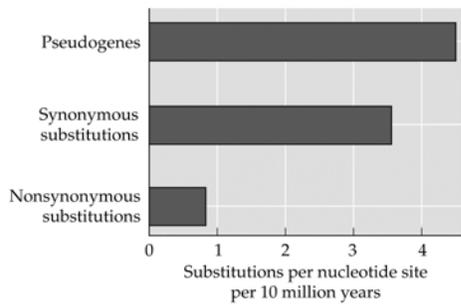
- Magnitude of genetic and phenotypic changes are not necessarily correlated, most have little effect on fitness.
- Repair mechanisms are not random, directed to specific exons.
- Point mutations at first and second position, usually replacement.
- Point mutations at third position, usually silent.
- Most populations harbor considerable allele diversity.

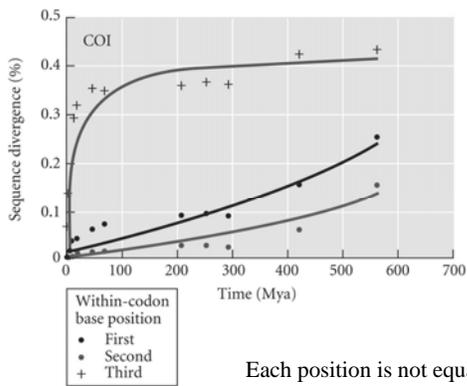




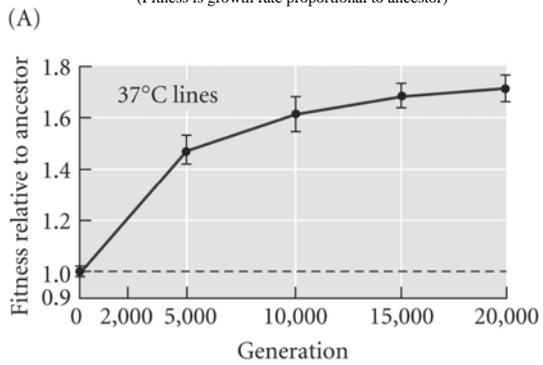


Changes evolve slowly in regions of functionally significant molecules, but more rapidly in regions where base substitutions do not affect molecule functioning.

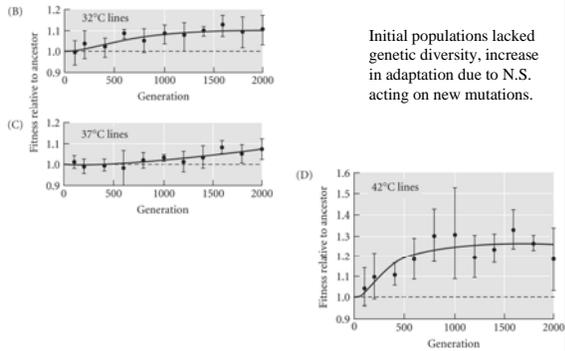




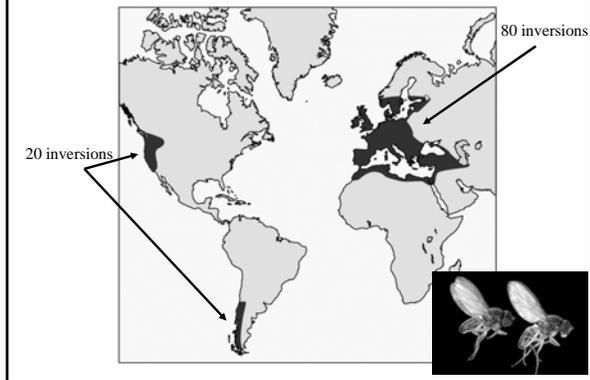
Adaptation in experimental populations of *E. coli*
(Fitness is growth rate proportional to ancestor)



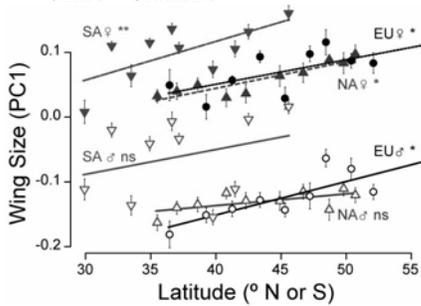
Adaptation in experimental populations of *E. coli*



Founder Effect in *Drosophila subobscura*



Clines = Gradients



NS: Larger sizes in colder wetter climates, greater number of inversions.
