



GENETICS

CHAPTER 21

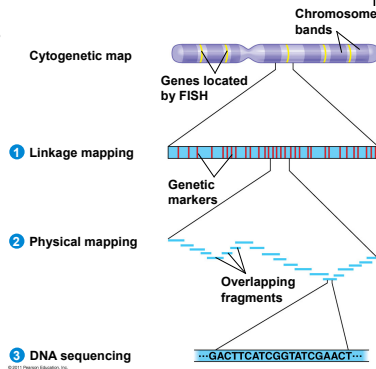
GENOMES AND THEIR EVOLUTION

DATE 2013 COURSE AP BIOLOGY

1

- * Comparisons of genomes provide information about the evolutionary history of genes and taxonomic groups
- * Genomics - study of whole sets of genes and their interactions
- * Bioinformatics - application of computational methods to storage and analysis of biological data
- * Human Genome Project - officially began in 1990 and was completed in 2003
 - * Linkage map - maps location of genes on each chromosome using recombination frequencies
 - * Physical map - expresses distance between genetic markers by cutting up DNA and arranging them by overlapping fragments

Tree of Life



THREE-STAGE PROCESS FIG. 21.2

2

Whole Genome/Shotgun Approach

- * Skips genetic and physical mapping and sequences random DNA fragments directly
- * Powerful computer programs are used to order fragments into a continuous sequence

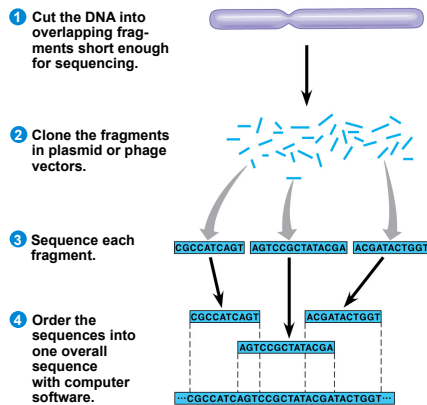


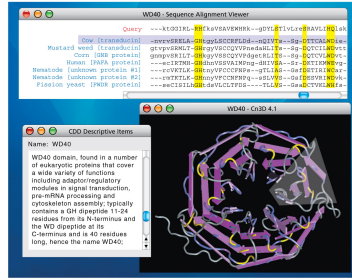
FIG. 21.3

3

Bioinformatics

FIG. 21.4

- * Many new resources since completion of Human Genome Project
- * Many nations have data resources (National Library of Medicine, NIH (National Center for Biotechnology Information-NCBI), European Molecular Biology Laboratory, DNA Data Bank of Japan)
- * NCBI doubles its database every 18 months and provides 3-D views of all protein structures that have been determined



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Proteomics

- * Study of all proteins encoded by a genome

FIG. 21.5

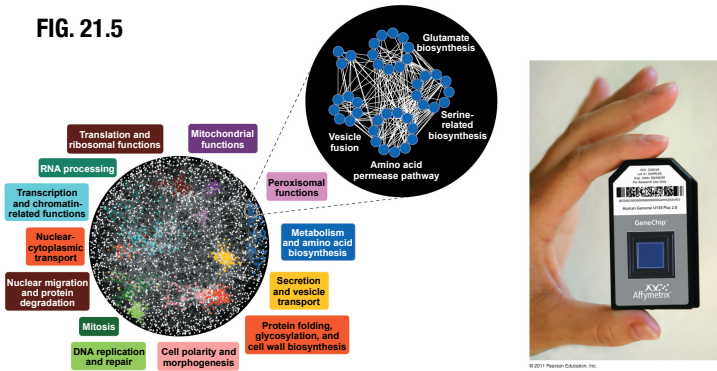


FIG. 21.6

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Genomes

- * Genomes vary in size, number of genes and gene density
- * By early 2010, over 1,200 genomes were completely sequenced (mostly bacteria)
- * Genomes of most bacteria are between 1-6 million base pairs (Mb), plants are more than 100 Mb, humans have 3,000 Mb
- * No relationship between genome size number of genes (alternative splicing)
- * Mammals (including humans) have the lowest gene density

Table 21.1 Genome Sizes and Estimated Numbers of Genes*

Organism	Haploid Genome Size (Mb)	Number of Genes	Genes per Mb
Bacteria			
<i>Haemophilus influenzae</i>	1.8	1,700	940
<i>Escherichia coli</i>	4.6	4,400	950
Archaea			
<i>Archaeoglobus fulgidus</i>	2.2	2,500	1,130
<i>Methanosarcina barkeri</i>	4.8	3,600	750
Eukaryotes			
<i>Saccharomyces cerevisiae</i> (yeast, a fungus)	12	6,300	525
<i>Caenorhabditis elegans</i> (nematode)	100	20,100	200
<i>Arabidopsis thaliana</i> (mustard family plant)	120	27,000	225
<i>Drosophila melanogaster</i> (fruit fly)	165	13,700	83
<i>Oryza sativa</i> (rice)	430	42,000	98
<i>Zea mays</i> (corn)	2,300	32,000	14
<i>Mus musculus</i> (house mouse)	2,600	22,000	11
<i>Ailuropoda melanoleuca</i> (giant panda)	2,400	21,000	9
<i>Homo sapiens</i> (human)	3,000	<21,000	7
<i>Fritillaria assyriaca</i> (lily family plant)	124,000	ND	ND

*Some values given here are likely to be revised as genome analysis continues. Mb = million base pairs. ND = not determined.

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Multicellular Eukaryotes

- * Most of the eukaryotic genome does not code for proteins or functional RNAs (98.5% of human genome is noncoding)
- * Noncoding DNA plays an important role in cells
- * Pseudogenes - former genes that have accumulated mutations and are nonfunctional
- * Repetitive DNA - present in multiple copies in the genome

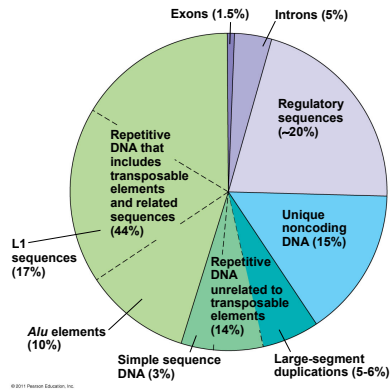
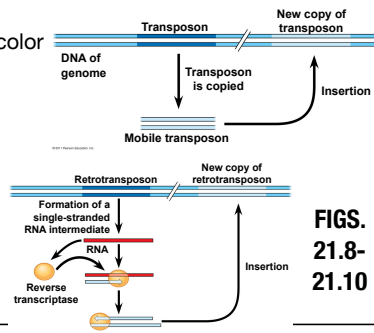


FIG. 21.7

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Transposable Elements

- * First evidence of mobile DNA segments came from research of Barbara McClintock breeding experiments with Indian corn
- * She identified changes in color of corn kernels that only made sense if genetic elements were moving from other locations into the genes for kernel color
- * These transposable elements are present in both prokaryotes and eukaryotes
- * Transposons - move via a DNA intermediate
- * Retrotransposons - move via a RNA intermediate



FIGS. 21.8-21.10

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Genome Evolution

- * Mutation is the basis for genomic evolution
- * Accidents in meiosis can lead to one or more extra sets of chromosomes (polyploidy) which can allow for genes in one set to diverge and accumulate mutations which may persist if organism reproduces
- * Combining of chromosomes also shows evolutionary relationships
- * Gene duplication can be caused by unequal crossing over (Fig. 21.13)

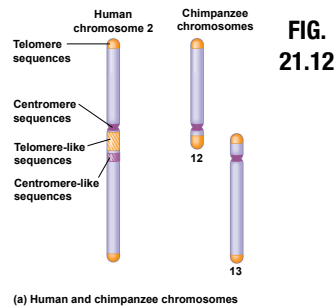
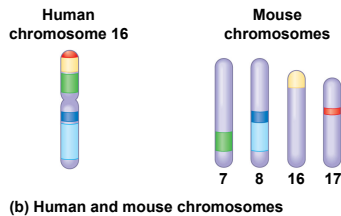


FIG. 21.12



(b) Human and mouse chromosomes

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Human Globin Genes

- * Caused by gene duplication and subsequent mutation

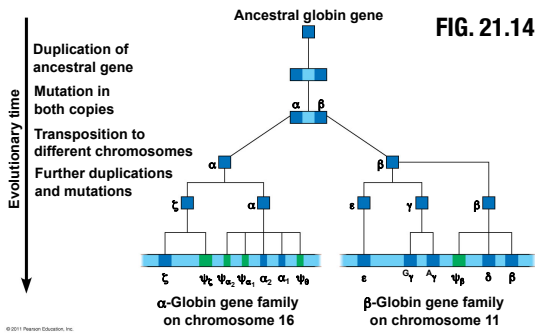
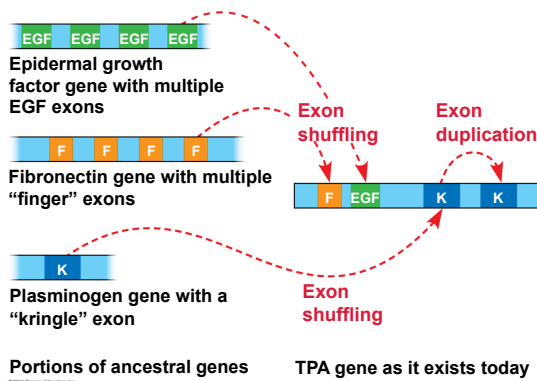


Table 21.2 Percentage of Similarity in Amino Acid Sequence Between Human Globin Proteins

	α-Globins		β-Globins		
	α	ζ	β	γ	ε
α-Globins	α	—	42	39	37
	ζ	58	34	38	37
β-Globins	β	42	—	73	75
	γ	39	73	—	80
	ε	37	75	80	—

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Exon Duplication and Shuffling



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Transposable Elements' Contributions to Evolution

- * Multiple copies of similar elements may cause recombination or crossing over between chromosomes
- * Insertion of a transposable element within a protein-coding sequence may block, increase, or decrease protein production
- * May carry a gene or groups of genes to a new position
- * May create new sites for alternative splicing
- * Changes are usually detrimental but may prove advantageous

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Comparing Genomes

- * Provides clues of evolutionary relationships
- * Comparing closely related species helps understanding of recent evolutionary events
- * Allows for correlation with phenotypic differences
- * Comparing distantly related species helps understanding of ancient evolutionary events
- * Highly conserved genes change little over time

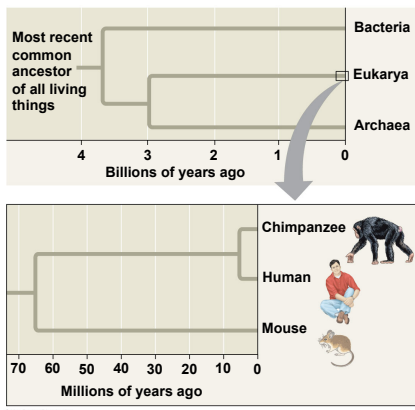


FIG. 21.16

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Ex. *FOXP2*

- * Involved in vocalization

FIG. 21.17

EXPERIMENT

Wild type: two normal copies of *FOXP2*

Heterozygote: one copy of *FOXP2* disrupted

Homozygote: both copies of *FOXP2* disrupted

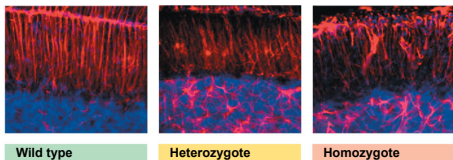


Experiment 1: Researchers cut thin sections of brain and stained them with reagents that allow visualization of brain anatomy in a UV fluorescence microscope.

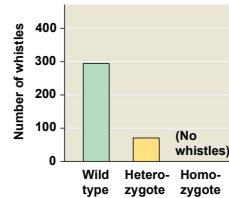
Experiment 2: Researchers separated each newborn pup from its mother and recorded the number of ultrasonic whistles produced by the pup.

RESULTS

Experiment 1



Experiment 2



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Evo-devo

- * Evolutionary developmental biology studies the evolution of developmental processes in multicellular organisms
- * Analysis of homeotic genes in *Drosophila* show that all include a sequence called a homeobox which is identical or similar in all vertebrates and invertebrates
- * Homeobox genes code for a domain that allows proteins to bind to DNA as transcription regulators
- * In animals these homeotic genes are called *Hox* genes

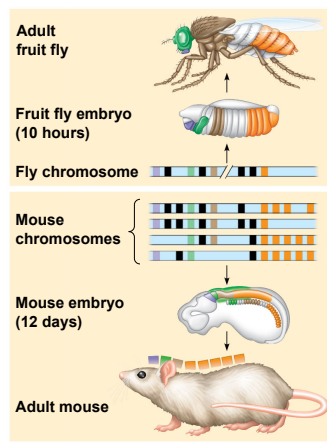


FIG. 21.18

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