MODERN SYNTHETIC THEORY OF EVOLUTION PHYLOGENETICS & MOLECULAR EVOLUTION

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POPULATION GENETICS & EVOLUTION
LECTURE II   25TH JANUARY, 2017

What Is Consent?

When it comes to sex, silence is not consent.
Flirting is not consent.
Being in a relationship is not consent.
Consent for one thing is not consent for everything.
Dressing in sexy clothing is not consent.
Having had sex before is not consent now.
Being too drunk or high to say no is not consent.
Being passed out is not consent.
If a person is underage, it is not consent.
Being afraid to say no is not consent.
Making out is not consent.
Going into a bedroom is not consent.
"Maybe" is not consent.
"No" is not consent.
Sexual activity without consent is sexual assault.

Consent is a voluntary, conscious and mutual agreement
to engage in sexual activity.

Only "Yes" is consent.

Stop. Ask. Listen. Get consent.

Objectives of the Lecture

1. Marriage between Darwin and Mendel history and contributors and the birth of the Modern Synthetic theory of Evolution

2. Tenets of the Modern Synthetic theory of Evolution

3. To understand the relationship between phylogeny and evolution

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4. Show how molecular genetics improves understanding of phylogeny

5. The role of mutation and natural selection and evolution

6. The concept of Darwinian fitness and Neutral mutation

7. Molecular clocks, molecular evolution and molecular palaeonthology

MODERN SYNTHETIC THEORY OF EVOLUTION

- **evolutionary synthesis** (known as the **new synthesis**, the **modern synthesis**, the **evolutionary synthesis**, **millennium synthesis** or the **neo-Darwinian synthesis**) is a 20th-century synthesis of ideas from several fields of biology that provides an account of evolution which is widely accepted as the current paradigm in evolutionary biology, and reflects the consensus about how evolution works.[

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The 19th Century ideas of natural selection by Charles Darwin and Mendelian genetics by Gregor Mendel were united by Ronald Fisher, one of the three founders of population genetics, along with J. B. S. Haldane and Sewall Wright, between 1918 and 1932.

The modern synthesis solved difficulties and confusions caused by the specialisation and poor communication between biologists in the early years of the 20th century. At its heart was the question of whether Mendelian genetics could be reconciled with gradual evolution by means of natural selection.

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- A second issue was whether the broad-scale changes of [macroevolution](#) seen by palaeontologists could be explained by changes seen in the [microevolution](#) of local [populations](#) .

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- Julian Huxley invented the term in his 1942 book, *Evolution: The Modern Synthesis*. Major figures in the modern synthesis include, Theodosius Dobzhansky, E. B. Ford, Ernst Mayr, Bernhard Rensch, Sergei Chetverikov, George Gaylord Simpson, and G. Ledyard Stebbins.

- The synthesis included evidence from biologists, trained in genetics, who studied populations in the field and in the laboratory. These studies were crucial to evolutionary theory. The synthesis drew together ideas from several branches of biology which had become separated, particularly genetics, cytology, systematics, botany, morphology, ecology and paleontology.

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Tenets of the Modern Synthetic theory of Evolution

The modern synthesis bridged the gap between the work of experimental geneticists and naturalists, and paleontologists. It states that:

1. All evolutionary phenomena can be explained in a way consistent with known genetic mechanisms and the observational evidence of naturalists.

2. Evolution is gradual: small genetic changes regulated by natural selection accumulate over long periods. Discontinuities amongst species (or other taxa) are explained as originating gradually through geographical separation and extinction. This theory contrasts with the saltation theory of William Bateson (1894).

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3.   Natural selection is by far the main mechanism of change; even slight advantages are important when continued. The object of selection is the phenotype in its surrounding environment.

4.   The role of genetic drift is equivocal. Though strongly supported initially by Dobzhansky, it was downgraded later as results from ecological genetics were obtained.

- 5. Thinking in terms of populations, rather than individuals, is primary: the [genetic diversity](#) existing in natural populations is a key factor in evolution. The strength of natural selection in the wild is greater than previously expected; the effect of ecological factors such as [niche](#) occupation and the significance of barriers to gene flow are all important.

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6. In palaeontology, the ability to explain historical observations by extrapolation from microevolution to macroevolution is proposed. Historical contingency means explanations at different levels may exist. Gradualism does not mean constant rate of change.

7. The idea that speciation occurs after populations are reproductively isolated has been much debated. In plants, polyploidy must be included in any view of speciation

7. Formulations such as 'evolution consists primarily of changes in the [frequencies of alleles](#) between one generation and another' were proposed rather later.

8. The traditional view is that [evolutionary developmental biology](#) (evo-devo) played little part in the synthesis, but an account of [Gavin de Beer](#)'s work by [Stephen J. Gould](#) suggests he may be an exception

Phylogeny & molecular evolution

Fossil records and known genetic phenomena tend to support the idea that the tempo of evolution can be quite variable. In some cases, rapid evolutionary change has taken place and led to the formation of new species.

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Phylogeny & molecular evolution

- During other periods, smaller phenotypic changes may occur over a longer timescale. In conjunction with phenotypic changes, the gradual accumulation of variations in gene sequences has been revealed by molecular analyses of DNA. We will explore in this lecture the use of molecular data to analyze evolution.

Phylogeny 1

- Thus far, we have considered the various factors that play a role in the formation of new species. In this section, we will examine **PHYLOGENY**-*the sequence of events involved in the evolutionary development of a species or group of species*.

Phylogeny cont`d

- A systematic approach is followed to produce a phylogenetic tree, which is a diagram that describes a phylogeny. Such a tree is a hypothesis of the evolutionary relationships among various species, based on the information available to and gathered by biologists termed systematists.

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Phylogeny cont`d

- Historically, morphological differences have been used to construct evolutionary trees. In this approach, species that are more similar in appearance tend to be placed closer together on the tree.

Phylogeny cont`d

- In addition, species have been categorized based on differences in physiology, biochemistry, and even behaviour. While these approaches continue to be used, systematists are increasingly using molecular data to infer evolutionary relationships.

Molecular Evolution and Mutation 1

- Rates of molecular evolution can be measured by comparing DNA sequences. Rates of change vary both within and between genes. Regions with the lowest rates of change usually are those that are most functionally constrained and most subject to natural selection.

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- Mutations are changes in nucleotide sequences, whereas substitutions are mutations that have passed through the filter of selection. Synonymous substitution rates are a good indicator of the actual mutation rate operating within a genome. Selection acts at surprisingly small scales, for example, at the level of the choice of which synonymous codons are preferred.

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Molecular Evolution and Mutation 2

- Mutations are rare events, and most changes in amino acid sequence tend to be removed through natural selection.

- Relative rate tests suggest that the molecular clocks of some genes run at a steady rate over long periods of time. However, it is unreasonable to assume that all lineages in a gene tree (a tree depicting the relationship of a single gene within and between species) accumulate substitutions at the same rate.

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Molecular Evolution and Mutation 3

- Gene trees do not always correspond to species trees (a tree depicting the relationship of species based on morphological or paleontological analyses or molecular data from several genes) because some genetic polymorphisms within populations predate speciation events.

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- Increases in the number of taxa being considered dramatically increases the possible number of phylogenetic trees that can describe the relationship of those taxa. Distance matrix (statistical analysis that groups taxa on the basis of their overall similarity), parsimony (premised on the concept that the tree that invokes the fewest number of mutations is most likely to be correct because mutations are rare events), and maximum likelihood (evaluating differences in mutation frequencies) approaches are three different ways to choose which of the many possible trees are most likely to represent the true evolutionary relationship.

Molecular Evolution and Mutation cont`d

- In eukaryotic organisms, genes frequently occur in multiple copies with identical or very similar sequences. A group of such genes is called a <mark>multigene family.</mark> Duplications of genes, in whole or in part, are the principal raw material from which proteins with new functions are made.

- The functional domains of many proteins correspond to regions encoded in single exons. Many genes appear to have been derived by "mixing and matching" such functional domains of already useful proteins through exon shuffling.

- The advent of molecular approaches for analyzing DNA and gene products has revolutionized the field of evolution. Differences in nucleotide sequences are quantitative and can be analyzed using mathematical principles in conjunction with computer programs.

- Evolutionary changes at the DNA level can be objectively compared among different species to establish evolutionary relationships. Furthermore, this approach can be used to compare any two existing organisms, no matter how greatly they differ in their morphological traits. For example, we can compare DNA sequences between humans and bacteria, or between plants and fruit flies. Such comparisons would be very difficult at a morphological level.

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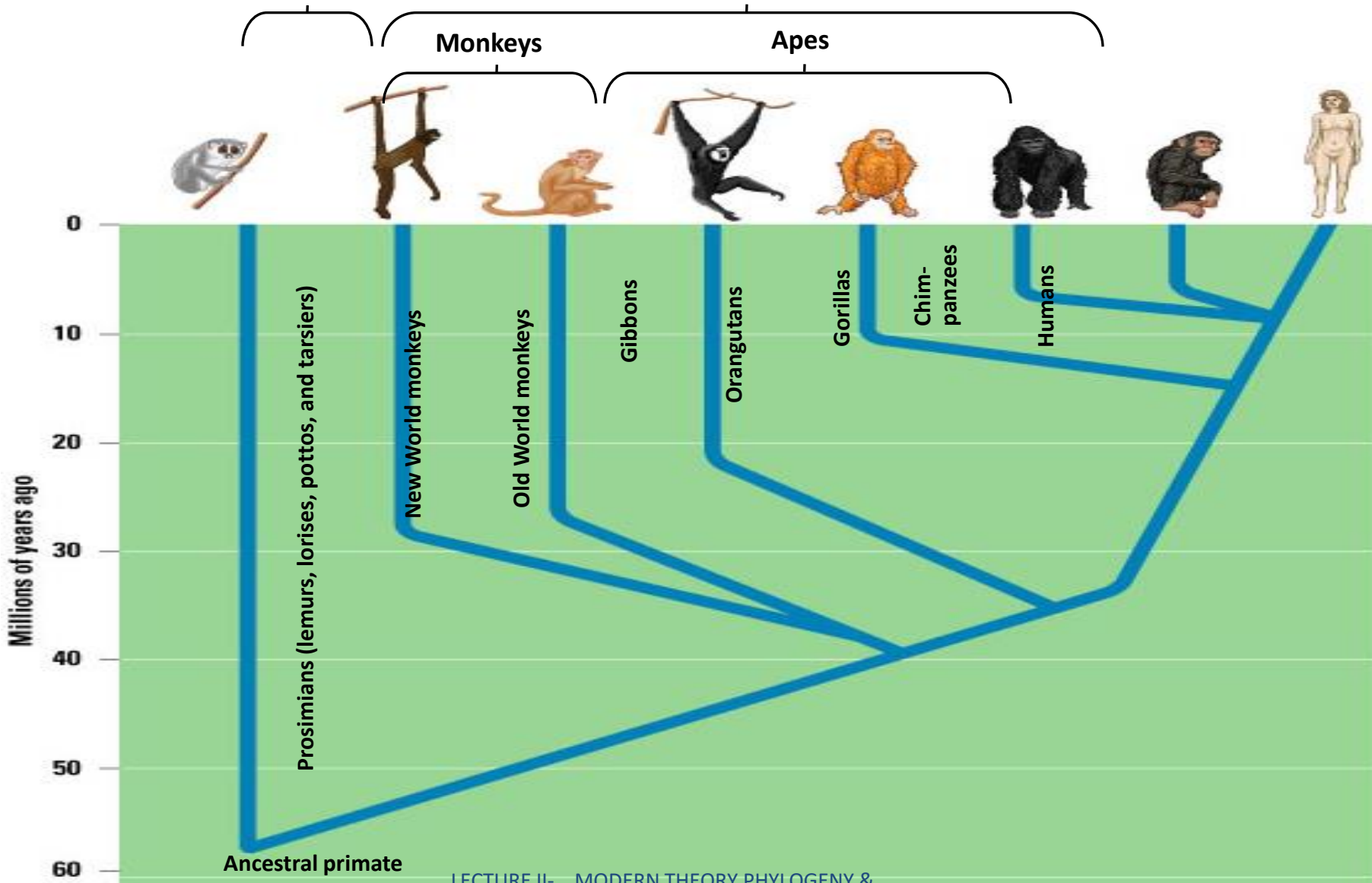PHYLOGENY & MOLECULAR EVOLUTION

DEFINITIONS

Anagenesis, in which a single species evolves into a different species

Cladogenesis, in which a species diverges into two or more species. In cladogenesis, the original species may remain in existence when a second new species is formed

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A monophyletic group, also known as a **clade,** is a group of species consisting of all descendents of the group's most common ancestor. The present-day descendents of a common ancestor can also be called a clade. smaller and more recent clades are subsets of larger and olderclades.

Prosimians

Anthropoids

Monkeys

Apes

Millions of years ago

Prosimians (lemurs, lorises, pottos, and tarsiers)

New World monkeys

Old World monkeys

Gibbons

Orangutans

Gorillas

Chim-panzees

Humans

0

10

20

30

40

50

60

Ancestral primate

Homologous Genes Are Derived from a Common Ancestral Gene

- Researchers typically study homology at the level of morphological traits or at the level of genes. We will focus on genetic homology. Two genes are said to be homologous if they are derived from the same ancestral gene. During evolution, a single species may become divided into two or more different species. When two homologous genes are found in different specie these genes are termed orthologs.

HOX genes orthologs, paralogs & gene families

Hox genes refers to the situation where several homologous genes were identified in the fruit fly and the mouse. In addition, two or more homologous genes can be found within a single species. These are termed paralogous genes, or paralogs. This can occur because abnormal gene duplication events can produce multiple copies of a gene and ultimately lead to the formation of a gene family. A gene family consists of two or more paralogs within the genome of a particular species.
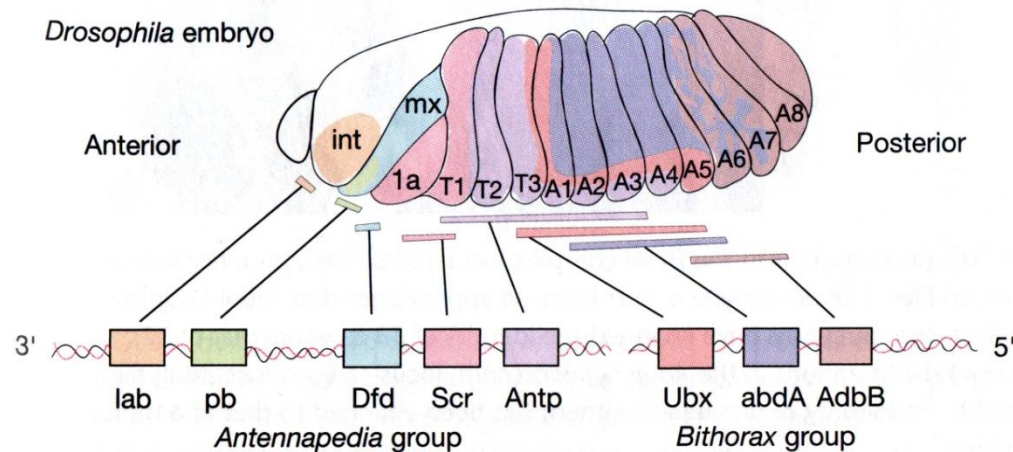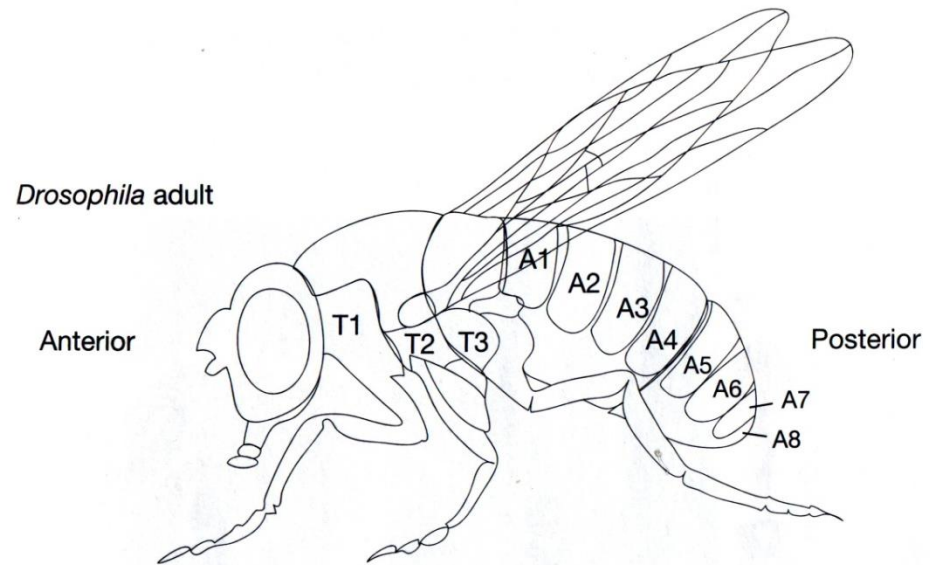
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Homeotic gene complexes →
code for transcription factors that
regulate expression of other genes

Homeotic mutations in *Drosophila*
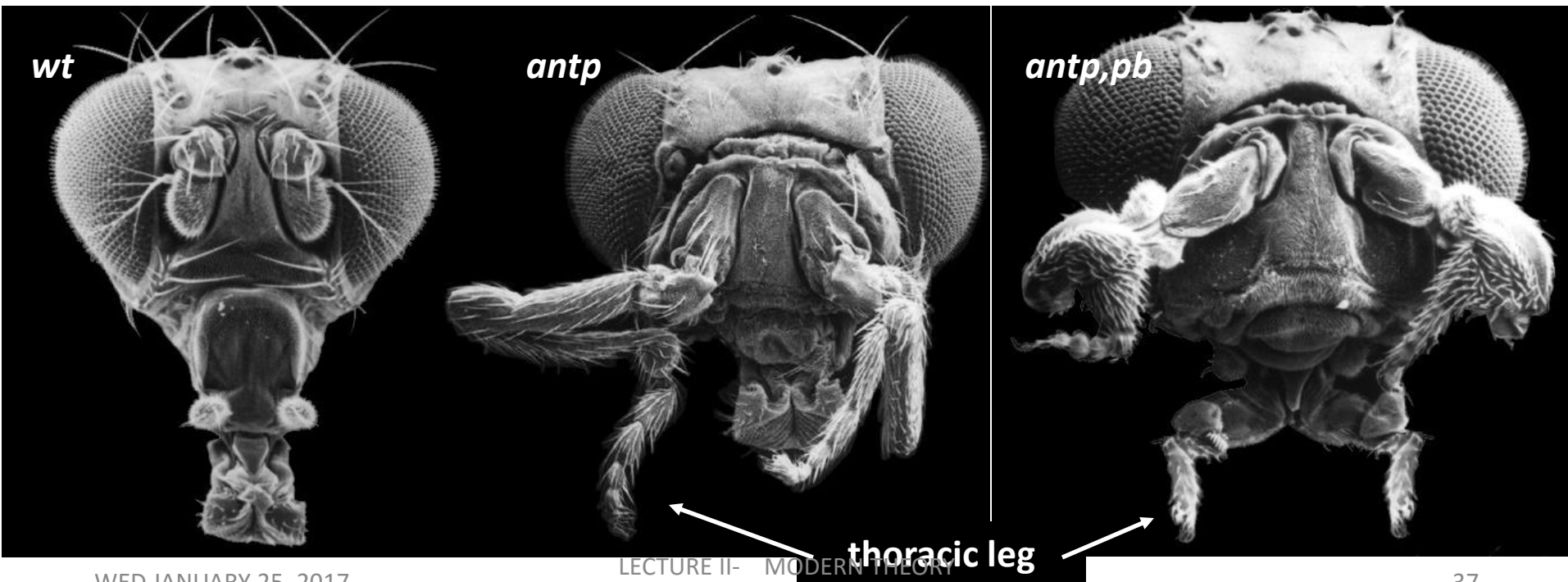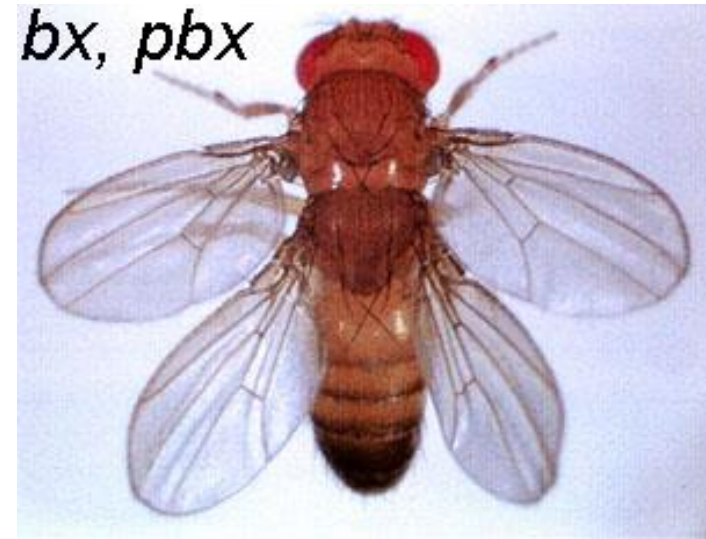
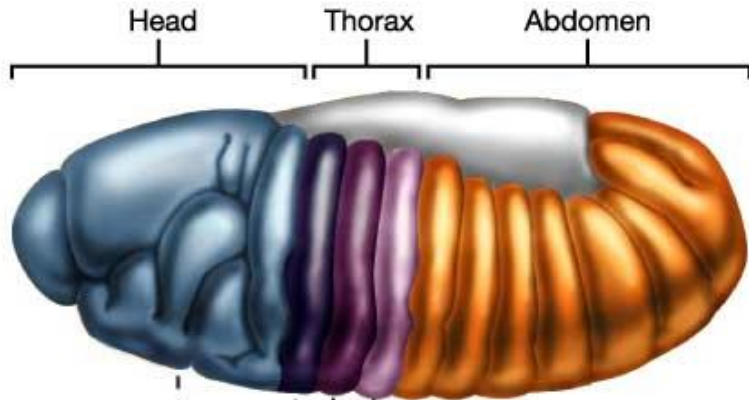
bx, pbx

wt   *antp*   *antp,pb*

thoracic leg

Homeotic gene complexes →
code for transcription factors that
regulate expression of other genes

Hox gene expression in

regulate genes to develop structures in the appropriate segments

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Normal fruit fly embryo

Head Thorax Abdomen

Anterior segments

Thoracic segments
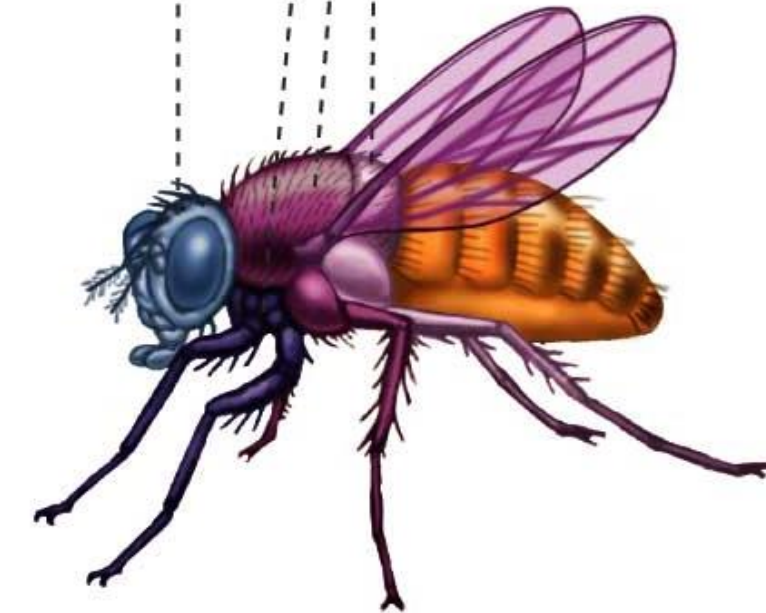
Abdominal segments

bicoid mutant

Abdominal segments

Abdominal segments

Changes in *Hox* gene expression → segment differentiation in arthropods

***morphological diversification
due to changes in gene expression***
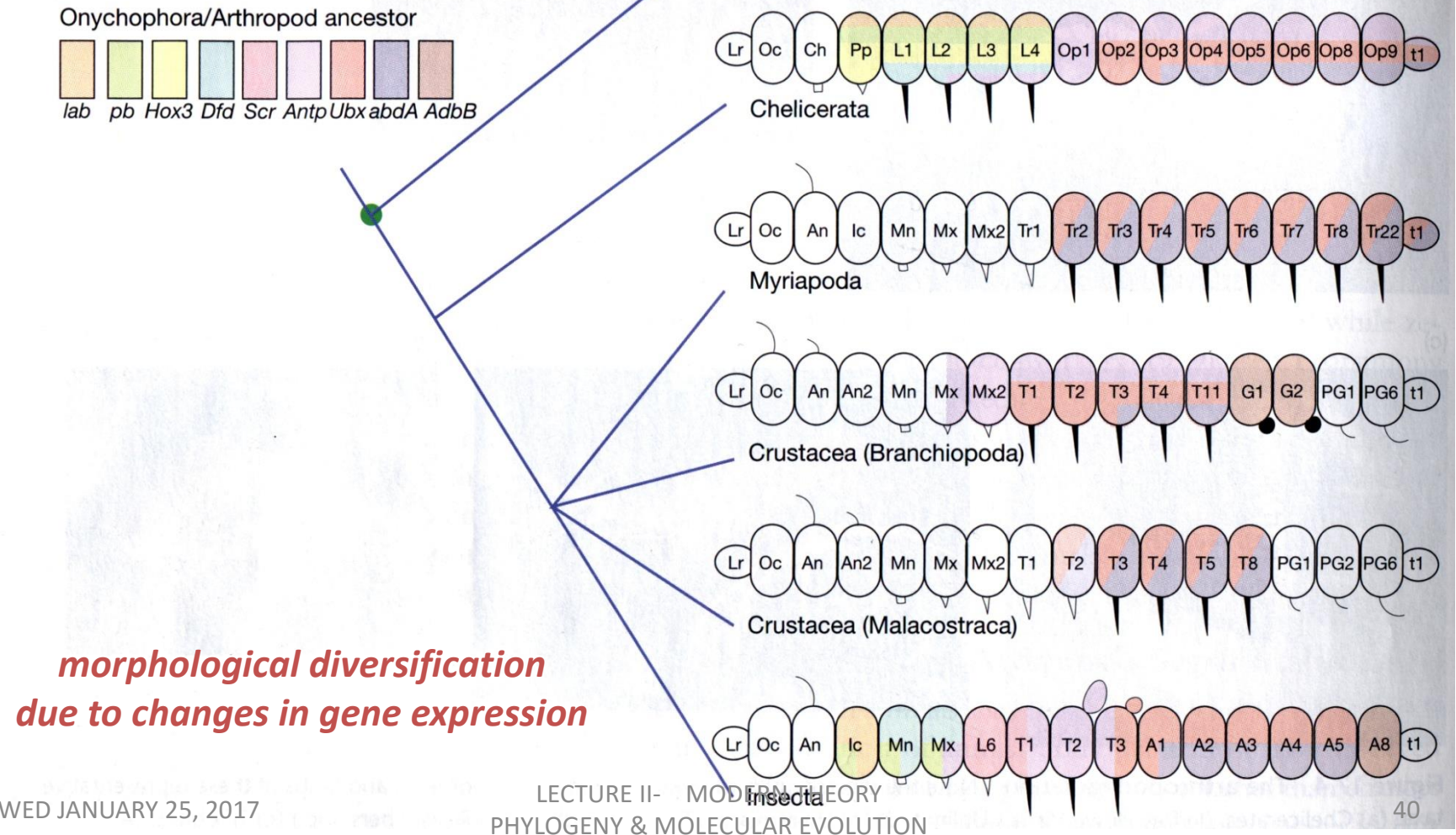
Shared traits

1) Homology – inherited from common ancestor

(b) Homology: When similarities are inherited from a common ancestor

Fruit fly *HOM* complex

lab   pb   Dfd   Antp   Ubx   abdA   abdB

Human *Hox* complex

b–1   b–2   b–3   b–4   b–5   b–6   b–7   b–8   b–9

Flatworms

Arthropods (insects, spiders, crustaceans)

Annelids (segmented worms)

Molluscs (snails, clams, squid)

Echinoderms (sea stars, sand dollars)

Chordates (vertebrates)

As shown in the two boxes above, the loci found in the *Hox* and *HOM* complexes of fruit flies and humans have similar sequences and are in the same order on their chromosomes

Origin of gene complex in common ancestor

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Homology of *Hox* genes

changes in Hox genes distinguish major clades (different body plans)

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Alpha and beta Haemoglobin and evolution

Based on the analysis of genetic sequences, evolutionary biologists have estimated that the gene duplication that created the a-globin and b-globin gene lineages occurred approximately 400 million years ago, while the speciation events that created different species of mammals occurred less than 200 million years ago. Therefore, the a-globin and b-globin genes have had much more time to accumulate changes relative to each other.

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Darwinian fitness and Beneficial mutation

According to Darwin, natural selection is the agent that leads to evolutionary change in populations. It selects for individuals with the highest Darwinian fitness and often promotes the establishment of beneficial alleles and the elimination of deleterious ones. Therefore, many geneticists have assumed that natural selection is the dominant factor in changing the genetic composition of natural populations, thereby leading to variation.

Neutral mutation and Evolution

Motoo Kimura proposed the neutral theory of evolution. According to this theory, most genetic variation observed in natural populations is due to the accumulation of neutral mutations that do not affect the phenotype of the organism and are not acted on by natural selection. For example, a mutation within a structural gene that changes a glycine codon from GGG to GGC would not affect the amino acid sequence of the encoded protein.

Because neutral mutations do not affect phenotype, they spread throughout a population according to their frequency of appearance and to random genetic drift.

This theory has been called the "survival of the luckiest" and also non-Darwinian evolution to contrast it with Darwin's theory, which focuses on fitness. Kimura agreed with Darwin that natural selection is responsible for adaptive changes in a species during evolution. His main argument is that most modern variation in gene sequences is neutral with respect to natural selection.

Kimuras five point proposition for molecular evol.

1. For each protein, the rate of evolution, in terms of amino acid substitutions, is approximately constant with regard to neutral substitutions that do not affect protein structure or function.

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Evidence: As an example, the amount of genetic variation between the coding sequence of the human a- globin and b-globin genes is approximately the same as the difference between the a-globin and b-globin genes in the horse . This type of comparison holds true in many different genes compared among many different species.

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2. Proteins that are functionally less important for the survival of an organism, or parts of a protein that are less important for its function, tend to evolve faster than more important proteins or regions of a protein. In other words, during evolution, less important proteins accumulate amino acid substitutions more rapidly than important proteins.

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- *Evidence:* Certain proteins are critical for survival, and their structure is precisely suited to their function. Examples are the histone proteins necessary for nucleosome formation in eukaryotes. Histone genes tolerate very few mutations and have evolved extremely slowly. By comparison, fibrinopeptides, which bind to fibrinogen to form a blood clot, evolve very rapidly. Presumably, the sequence of amino acids in this polypeptide is not very important for allowing it to aggregate and form a clot.

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Kimuras five point proposition for molecular evol.

3. Amino acid substitutions that do not significantly alter the existing structure and function of a protein are found more commonly than disruptive amino acid changes.

- *Evidence:* When comparing the coding sequences within homologous genes of modern species, nucleotide differences are more likely to be observed in the wobble base than in the first or second base within a codon. Mutations in the wobble base are often silent because they do not change the amino acid sequence of the protein. In addition, conservative substitutions (i.e., a substitution with a similar amino acid, such as a nonpolar amino acid for another nonpolar amino acid) are fairly common.

- Nonsense and frameshift mutations are very rare within the coding sequences of genes. Also, intron sequences evolve more rapidly than exon sequences.

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4. Gene duplication often precedes the emergence of a gene having a new function.

Evidence: When a single copy of a gene exists in a species, it usually plays a functional role similar to that of the homologous gene found in another species. Gene duplications have created gene families in which each family member can evolve somewhat different functional roles. Examples include the globin family and the *Hox* genes.

- 5.Selective elimination of definitely deleterious mutations and the random fixation of selectively neutral or veryslightly deleterious alleles occur far more frequently in evolution than selection of advantageous mutants.

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Evidence: As mentioned in principle 3, silent and conservative mutations are much more common than nonconservative substitutions. Presumably these nonconservative mutations usually have a negative effect on the phenotype of the organism, so they are effectively eliminated from the population by natural selection.

On rare occasions, however, an amino acid substitution due to a mutation may have a beneficial effect on the phenotype. For example, a nonconservative mutation in the b-globin gene produces the *HbA/HbS* allele, which gives an individual resistance to malaria in the heterozygous condition.

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Molecular clocks

In 1963, Linus Pauling and Emile Zuckerkandl were the first to suggest the use of molecular data to establish evolutionary relationships. When comparing homologous genes in different species, the DNA sequences from closely related species are more similar to each other than are the sequences from distantly related species. A molecular clock.

Molecular clocks

According to the concept of a molecular clock, neutral mutations accumulate over evolutionary time. When comparing homologous genes between species, those species that diverged more recently tend to have fewer differences compared to those whose common ancestor occurred in the very distant past.

rRNA and Molecular evolution

For molecular evolutionary studies, the DNA sequences of many genes have been obtained from a wide range of sources. Several different types of gene sequences have been used to construct phylogenetic trees. One very commonly analyzed gene is that encoding 16S rRNA, an rRNA found in the small ribosomal subunit. This gene has been sequenced from thousands of different species.

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- It is as reliable a molecular measure of phylogenetic relationships among organisms as is now available. Because rRNA is universal in all living organisms, its function was established at an early stage in the evolution of life on this planet, and its sequence has changed fairly slowly.

- Presumably, most mutations in this gene are deleterious, so few neutral or beneficial alleles can occur. This limitation causes this gene sequence to change very slowly during evolution. Furthermore, 16S rRNA is a rather large molecule, and therefore it contains a large amount of sequence information.

Horizontal gene transfer & Evolution

- The types of phylogenetic trees considered thus far are examples of vertical evolution, in which species evolve from preexisting species by the accumulation of gene mutations and by changes in chromosome structure and number.

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- Vertical evolution involves genetic changes in a series of ancestors that form a lineage. In addition to vertical evolution, however, species accumulate genetic changes by another process called ==horizontal gene transfer. This involves the exchange of genetic material among different species.==

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- An analysis of many genomes suggests that horizontal gene transfer was prevalent during the early stages of evolution, when all organisms were unicellular, but continued even after the divergence of the three major domains of life. With regard to modern organisms, horizontal gene transfer remains prevalent among prokaryotic species. It is believed the evolution of sexual reproduction in eukaryotes is a barrier to such horizontal transfer

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Molecular Palaenthology

Since the mid-1980s, many researchers have become excited about the information that might be derived from sequencing DNA obtained from older specimens. Currently there is debate concerning how long DNA can remain significantly intact after an organism has died.

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Molecular Palaenthology

Over time, the structure of DNA is degraded by hydrolysis and the loss of purines. Nevertheless, under certain conditions (e.g., cold temperature, low oxygen), DNA samples may remain stable for as long as 50,000 to 100,000 years and perhaps longer.

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