Fundamentals of Evolution

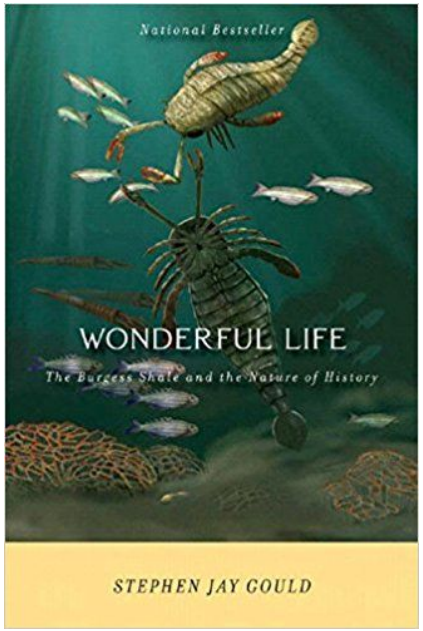
Session 22 - 11/27/2018

Contingency and Development

Contingency in evolution

- Although the influence of *chance* in evolution has been recognized since Darwin, its significance reached greater recognition from the writings by Stephen Jay Gould (1989; Wonderful Life).

- In this, Gould wrote about the Burgess Shale, a famous fossil bed from >500 Mya.



Contingency in evolution

- The *Cambrian Explosion* describes the rapid appearance of taxa which represent the ancestor of all living animal phyla approximately 540 Mya.

- The Burgess shale from British Columbia captures this period including the preservation of soft tissue.

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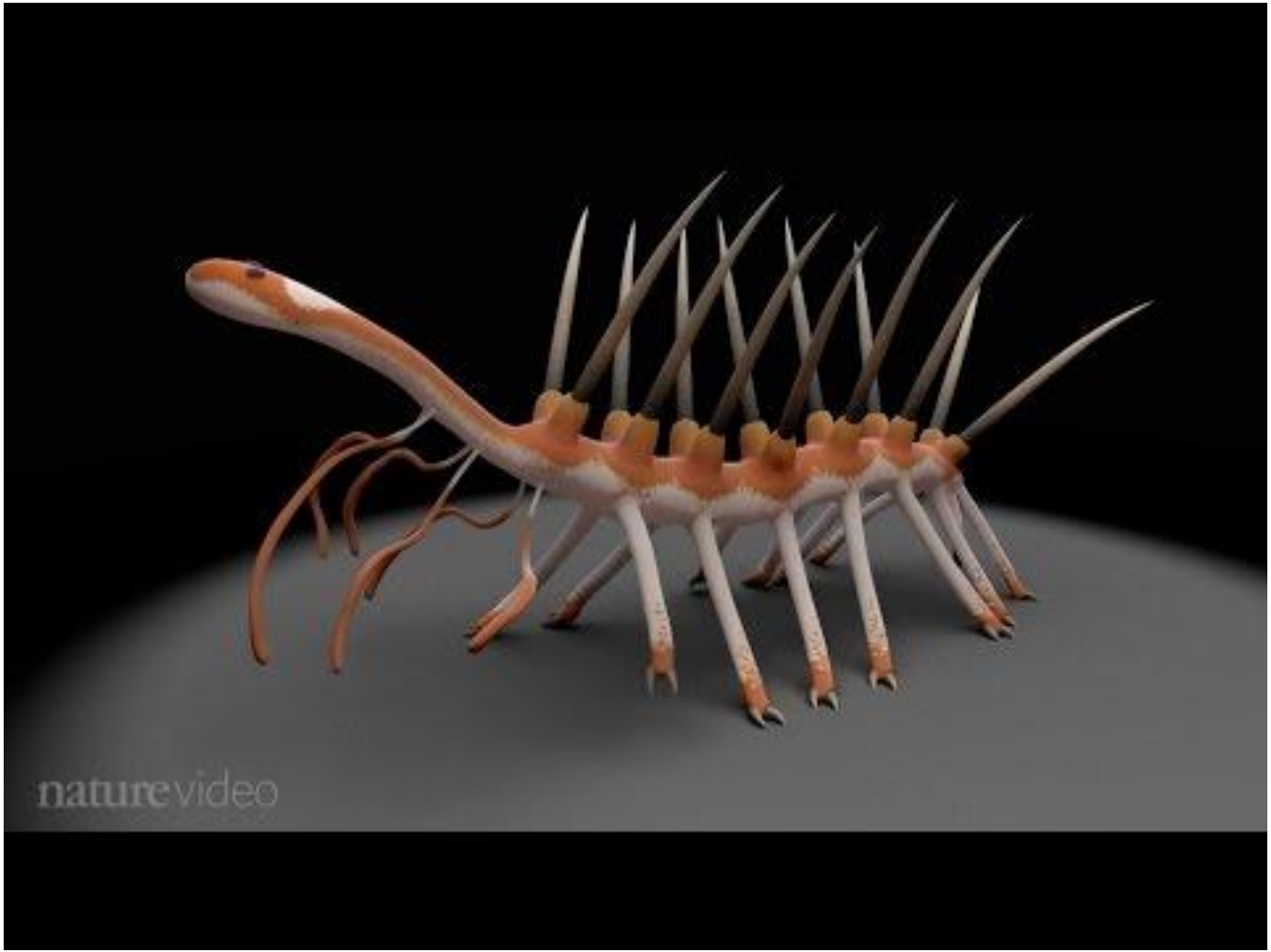
- The Burgess shale from British Columbia captures this period including the preservation of soft tissue.

- Animal body plan diversity was greater at that time than it is in today's oceans (in terms of of *body plan diversity* only).

Hallucigenia sparsa from the Burgess Shale. Credit: Jean-Bernard Caron


nature video

Contingency in evolution

- Gould argues that few of the disparate clades of organisms from the Burgess shale left descendants that exist today.
- He argues that all of these taxa were *adapted to their environment,* but that does not guarantee long-term survival -- many catastrophic extinction events can occur that wipe out entire clades.
- *"traits that enhance survival during mass extinction do so in ways that are incidental and unrelated to the causes of their evolution in the first place."*

Contingency in evolution

- Therefore, much of the major trends in life -- which groups expand to become diverse or dwindle to extinction -- ***is random.***

- If we rewind the tape of life and replay it from some point in the past we should expect a very different outcome.

- Contingency has also been proposed as an explanation for parallel evolution, and constraints. *Chance historical events, like the fixation of neutral mutations, may affect later responses to selection.*

Testing Contingency

- Experimental evolution can tell us a lot about contingency, at least on the *micro-evolutionary timescale*

- The Lenski lab has investigated contingency using long-term experiments with bacteria and phage viruses.

Historical contingency and the evolution of a key innovation in an experimental population of *Escherichia coli*

Zachary D. Blount, Christina Z. Borland, and Richard E. Lenski*

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- Twelve initially identical populations of *Escherichia coli* were founded in 1988. They have since evolved in a *glucose-limited medium that also contains citrate*, which *E. coli* cannot use as a carbon source under oxic conditions.
- No population evolved to use citrate for the first 30K generations. A cit+ mutant evolved in one population in generation 31,500.
- Was this caused by a very rare variant, or did it require multiple mutations such that some contingent changes need to take place before cit+ can evolve?

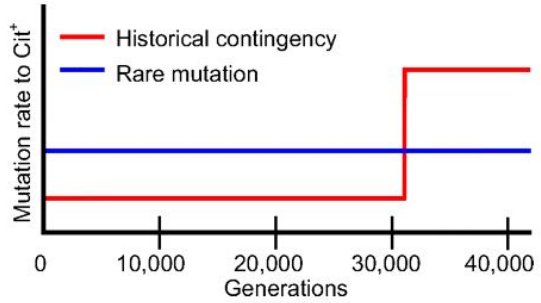

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Fig. 3.

Alternative hypotheses for the origin of the Cit^+ function. According to the rare-mutation hypothesis, the probability of mutation from Cit^- to Cit^+ was low but constant over time. Under the historical-contingency hypothesis, the probability of this transition increased when a mutation arose that produced a genetic background with a higher mutation rate to Cit^+.

Historical contingency and the evolution of a key innovation in an experimental population of *Escherichia coli*

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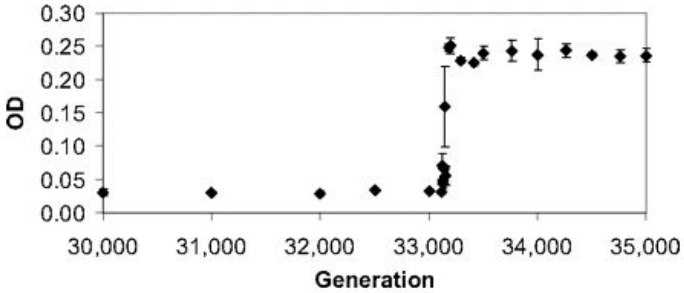
- Contingency was tested by “replaying evolution” from frozen records that occurred before generation 31,500.
- Restarting from generation 15,000 did not lead to any cit+ mutants over many independent replicate tests.
- However, restarting after 20,000 generations led to cit+ mutants many times, suggesting that potentiating mutations were present at this time.



Evolutionary Development

History of developmental biology

- *Developmental biology* -- the study of how an individual organism's morphology changes over time -- was broadly studied before Darwin (1859), most famously by many German scientists including Von Baer (1828).

- He compared embryos (embryology) to show that morphological similarities in embryos often match taxonomic groupings (phyla) more clearly than adult morphologies do.

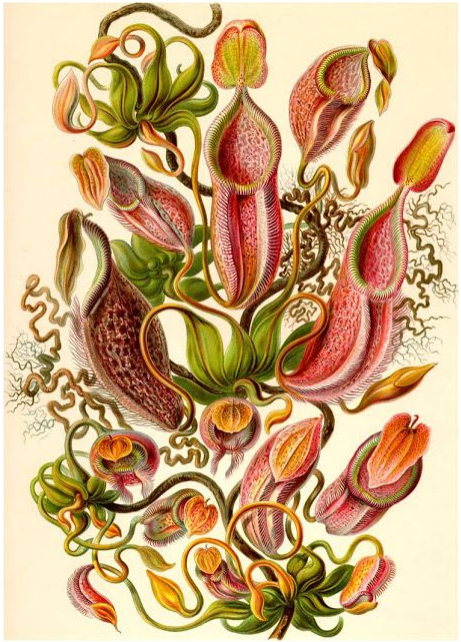
History of developmental biology

- *Ernst Haeckel* took this a step further with the claim that *“ontology recapitulates phylogeny”*, and used embryology to infer phylogenetic relationships (in a pre-cladistic analysis).

- However, many shared embryological characters represent plesiomorphies (derived characters are not yet expressed in the embryos) and they in fact provide somewhat poor phylogenetic characters.

History of developmental biology

- *That's ok though, because Haeckel was a great artist.*



Evolutionary developmental biology

- *With advances in molecular genetics of the 80s evo-devo was revived and has led to many advances in our understanding of how genetics -> phenotypes.*

- Key Question: *How does genetic variation lead to the morphological diversity that we observe?*

- Key Question: *Given that all cells in a body have the same DNA, how do tissues and organs differentiate to become so different?*

Evolutionary developmental biology

- *Proximate causes: mechanisms that operate within an individual organism to regulate development based on genetic and environmental signaling. e.g., programmed cell death causes the skin between digits to be lost in humans but not ducks.*

- *Ultimate causes: mechanisms that operate on populations over generations. e.g., natural selection. Explains how proximate causes evolve, by changes in allele frequencies.*

Evolutionary developmental biology

- Terms in evo-devo. There's a lot of them.
  - Ontogeny: *development of an individual*
  - Allometry: *differential growth of different parts*
  - Heterochrony: *change in timing of development.*
  - Heterotopy: *change in position of development.*
  - Paedomorphosis: *retain expression of juvenile phenotype.*
  - Neoteny: *slowed process of development (more juv. state)*

  - E.g., Human's are *neotenic* compared to their closest relatives, showing a prolonged juvenile stage of development.

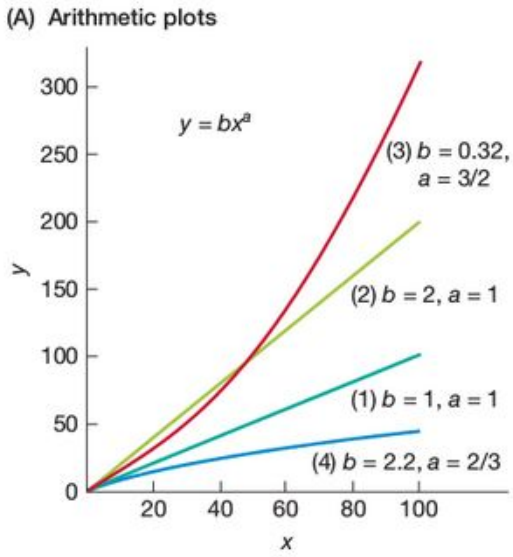
Allometry and comparative biology

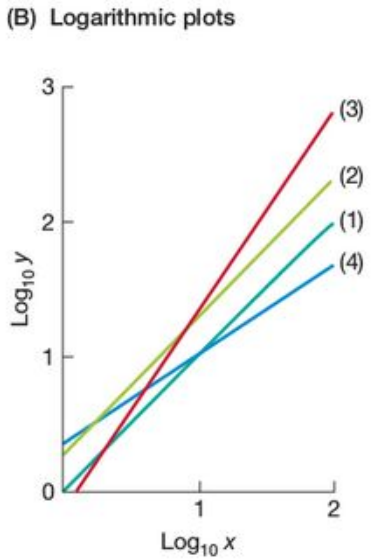
- It is important to take allometry into account when doing comparative biology, because in general, we are interested in quantifying *relative change, and not just scale.*

- In animals, typically, measurements are made relative to body size, because just about everything correlates with body size. e.g., we might study the residuals of toe length versus body size rather than toe length itself.

Allometry and comparative biology

- Morphometrics relies on allometric comparisons using body size, snout-vent-length, leaf surface area, etc.





Identity and homology

- Heterotopy -- *evolutionary change in the position of a feature*
- Experimental manipulations that alter the placement or type of organismal features have shown that the “identity” of a feature is sometimes controlled by few genes. (more on this later).
- Heterotopic differences among species are common, especially in plants: e.g., stems, leaves, or flowers in different positions in different species.

Modules and homology

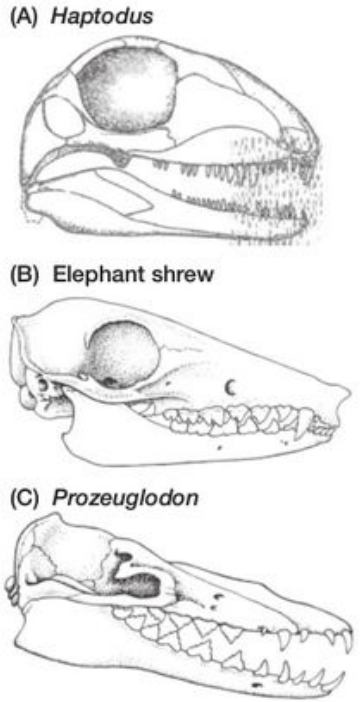
- The bodies of most organisms consist of ***modules*** -- distinct units that have genetic specifications, developmental patterns, locations, and interactions with other modules.

- Developmental modules have historically been defined on the basis of being *similar* across species (Huneman 2013).

- Evo-devo is typically more concerned with *differences* among species, and identifying the genetic basis of modules.

Modules and homology

- Teeth in vertebrates are *serially homologous*: “repetitive relation of segments in the same organism”
- In mammals, teeth have become differentiated into incisors, canines, premolars, etc, by *individualization*.
- Distinct genes are active in developing primordia of different teeth.



Signalling

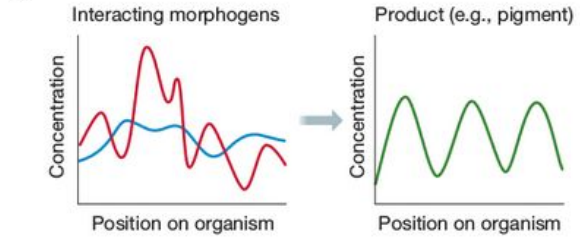
- All cells have the *same set of genes*, but do different things with them based on signals.
- Many aspects of development are controlled by signals that bind to cells and initiate signal transduction to affect gene expression.
- Signals can be extrinsic: environmentally induced (GxE), or intrinsic: hormones (chemicals) exchanged between cells.

Signalling

- Experiments in animals show that development for many cell types depends on preceding events, e.g., differentiation of neighboring cells, which therefore changes the signals which they transmit.

- Mathematical models about the diffusion of signaling molecules -- simply the interaction of chemicals along gradients -- can create complex patterns of development (e.g., Turing models).

Example of diffusion gradient from the textbook.



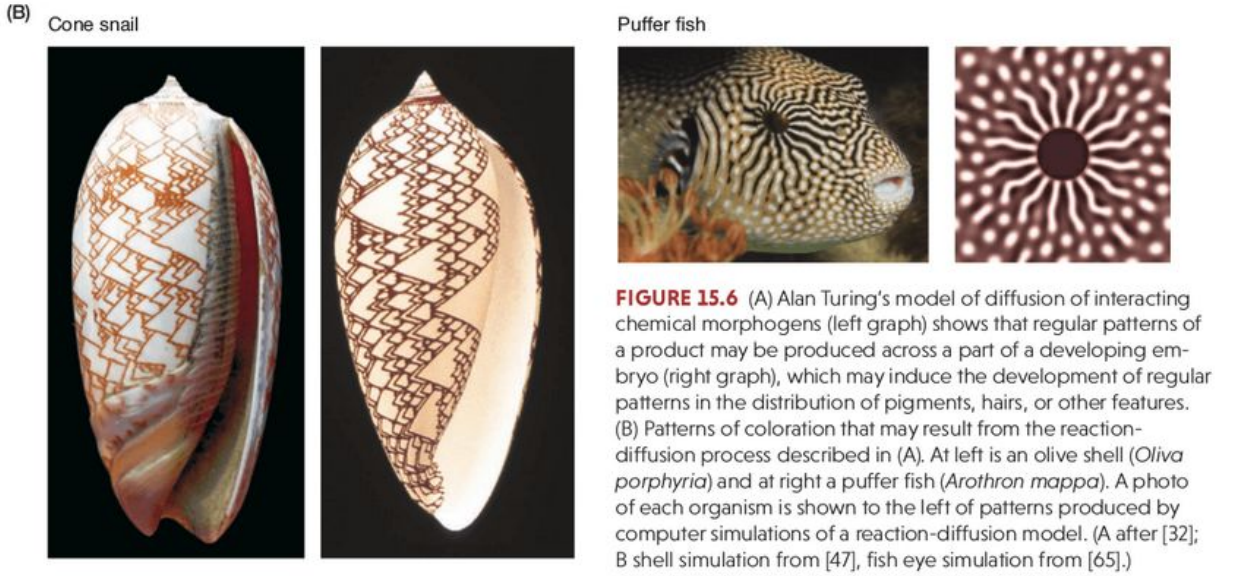
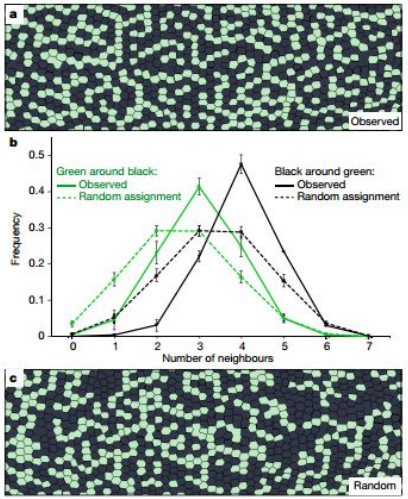
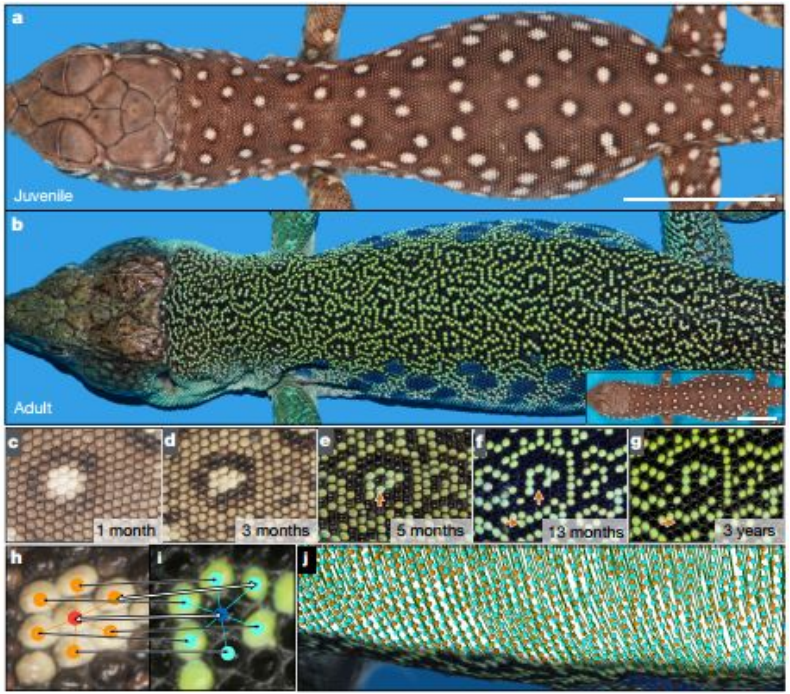


FIGURE 15.6 (A) Alan Turing's model of diffusion of interacting chemical morphogens (left graph) shows that regular patterns of a product may be produced across a part of a developing embryo (right graph), which may induce the development of regular patterns in the distribution of pigments, hairs, or other features. (B) Patterns of coloration that may result from the reaction-diffusion process described in (A). At left is an olive shell (*Oliva porphyria*) and at right a puffer fish (*Arothron mappa*). A photo of each organism is shown to the left of patterns produced by computer simulations of a reaction-diffusion model. (A after [32]; B shell simulation from [47], fish eye simulation from [65].)

Signalling

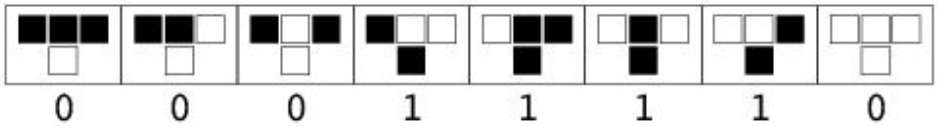
- Another cool diffusion gradient
- Cellular automaton models (Manukyan et al. 2017)

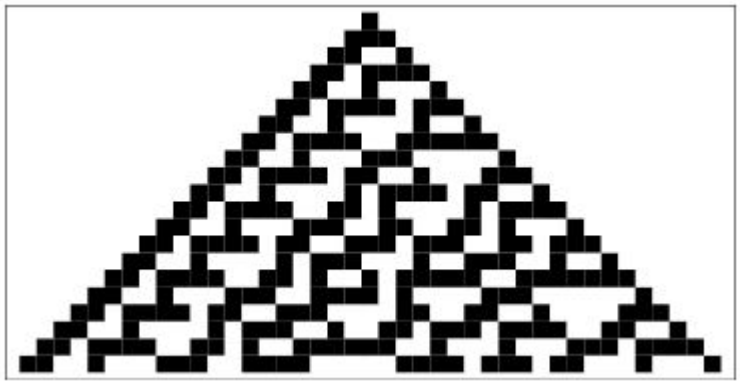




Cellular automata

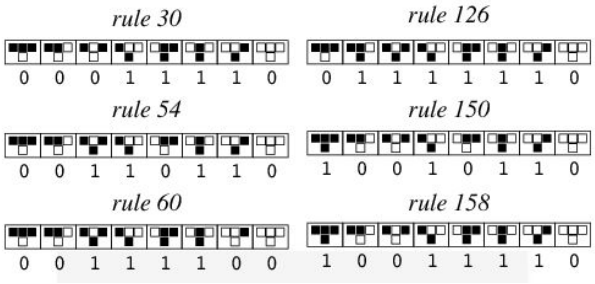
- 1-dimensional example: The state of cell in the next step depends on the state of its neighbors this step.
- Depending on the set of rules a different deterministic pattern can result.

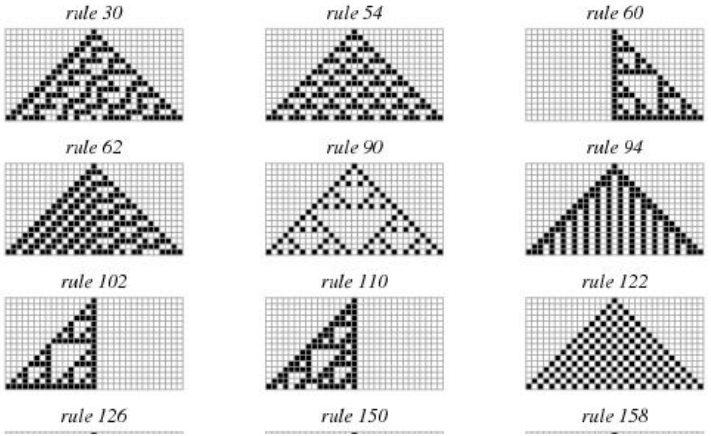




Cellular automata

- 1-dimensional example: The state of cell in the next step depends on the state of its neighbors this step.
- Depending on the set of rules a different deterministic pattern can result.
- This can be a *proximate cause* of differences between taxa, based on (rules of) how signals are interpreted.





Determinate versus indeterminate growth

- Many plants have indeterminate growth, meaning that they have a flexible *bauplan* (body plan) composed of modules that can be easily interchanged and replicated, with no determined end point (for the most part).

- Most animals have determinate growth, meaning there is a set *bauplan* that if not developed in the correct order and timing will typically result in major abnormalities. But, some organisms, like fish, have a set plan but less determinate end points -- they seem to continue growing indefinitely.

Gene regulation

- In eukaryotes, transcription of a protein coding gene is initiated when a protein (RNA-polymerase II) binds to an upstream region, the promoter.

- This is *regulated* by certain regulatory proteins -- transcription factors (TFs) -- which bind to an enhancer region upstream of the promoter.

- Enhancers are *cis regulatory elements*
- Transcription factors are *trans regulatory elements*

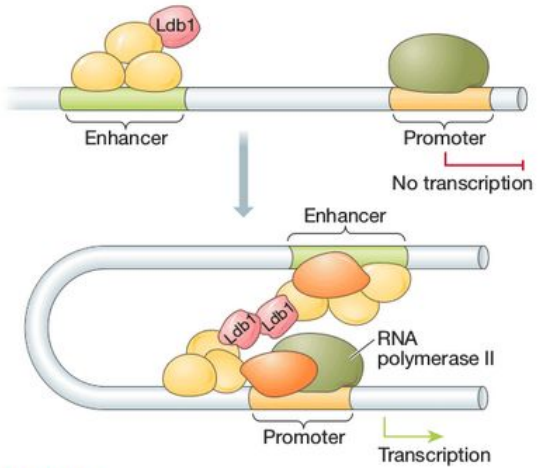


FIGURE 15.7 RNA polymerase II binds to a gene's promoter and initiates transcription of the gene into mRNA, but only after transcription factor proteins bind to both the promoter and an enhancer sequence. The promoter and enhancer may be linked to each other by other transcription factors (labeled Ldb1 here). (From [19], after [13].)

Hox genes and the genetic toolkit

- Homeotic mutations were first discovered in Drosophila, where mutants were discovered for which major structural differences existed.
- Homeotic mutations change the *identity* of segments in Drosophila (see figure).
- These genes affect anterior-posterior axis development, and were found to encode *transcription factors.*
- The part of the sequence that encodes the protein that *binds* to DNA is called the homeobox, so these are homeobox selector genes or **Hox genes.**

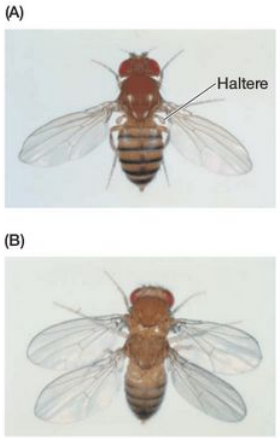
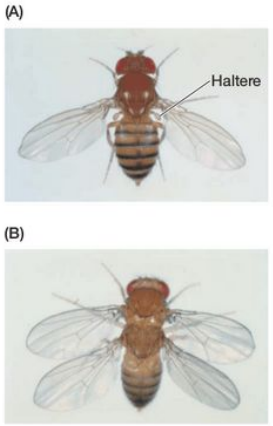


FIGURE 15.8 A homeotic mutation. (A) A wild-type *Drosophila melanogaster* has a single pair of wings, borne on the second thoracic segment, and a pair of small winglike structures called halteres, borne on the third thoracic segment. (B) In a fly carrying mutations in the *Ultrabithorax* (*Ubx*) gene, the third thoracic segment has been transformed into another second thoracic segment, bearing wings instead of halteres. (Photos courtesy of E. B. Lewis.)

Hox genes and the genetic toolkit

- Hox genes are highly conserved across bilaterial animals, suggesting that they existed in their common ancestor.
- Individualization has happened many times through differential expression of Hox genes in different space or time, which can differentiate tissues, colors, shape, and size.



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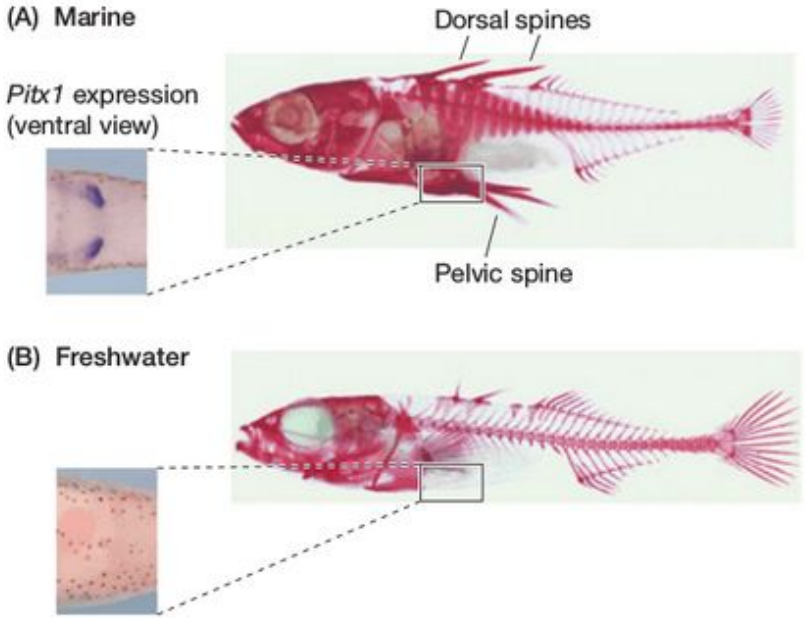
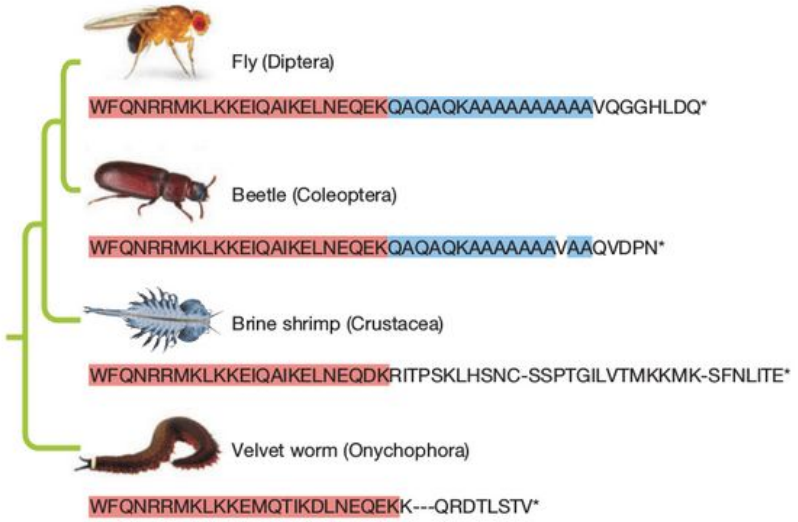
Evolution by cis-regulatory elements

- Mutations in the regulatory enhancer element can affect the expression of a gene.
- Example: three-spine sticklebacks. Mutation in *Pitx1* causes loss-of-function mutation that leads to loss of armored plating.



Evolution by trans-regulatory elements

- Mutations in the transcription factors that bind to enhancers can affect the expression of a gene.
- Example: the Hox gene *Ubx* has evolved differences among arthropod lineages which changes its function -- in insects it suppresses leg development in the abdomen.
- Experimental insertion of crustacean or velvet worm *Ubx* into Drosophila did not stop leg development.

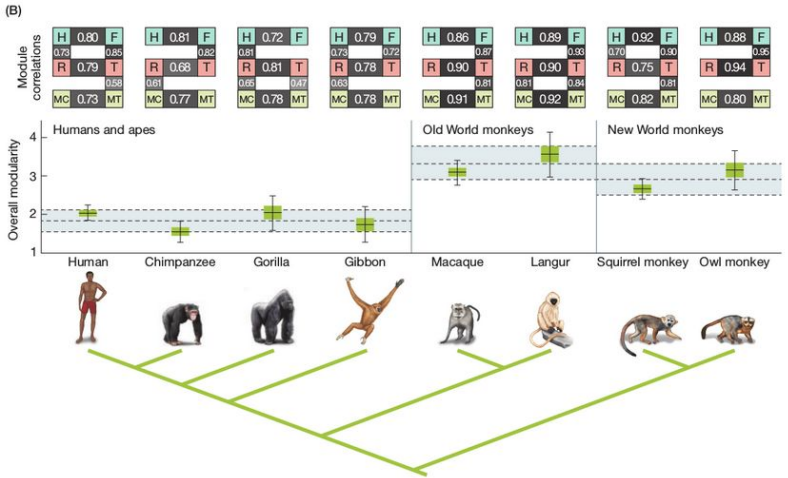


The basis of phenotypic evolution?

- Discuss and debate.
- What is Hoekstra & Coyne's argument? List several examples.
- What is Carrol and colleagues' argument? List several examples.
- Who is more convincing?
- Do we have enough evidence yet to decide? It's been almost 10 years since this debate started, do you think we know more now?

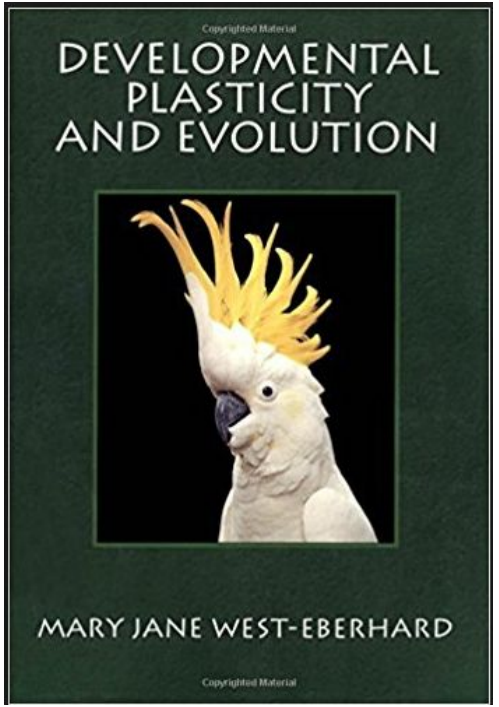
Pleiotropy and development

- Pleiotropy -- when one gene affects multiple things -- such as in transcription factors, can *constrain* evolution, since any change will affect many processes.
- This is likely the reason why TFs such as Hox genes are so highly conserved.
- However, characters that are genetically correlated may also be more *evolvable* if a change in one necessarily means a change in its corresponding parts. This is termed *phenotypic integration.*
- The evolution of integration of distinct modules through individualization.



Plasticity and Canalization

- Phenotypic variation ***before*** genetic variation?
- Mary Jane West-Eberhard made the argument that plasticity can allow organisms to colonize a habitat in which a certain extreme of their phenotype can survive.
- *Later,* genetic mutations can arise that allow the individuals to express that phenotype as the norm, as opposed to an extreme (perhaps less costly or variable). It becomes canalized.



Plasticity and Canalization

- Not all integration is a result of specific carefully evolved mechanisms, but rather, integration may result of developmental plasticity.
- In this way, genetic correlations between characters is a spandrel that appears to be necessary but is actually a result of a specific environment.
- Slijper's two-legged goat example and thought experiment:(http://www.pnas.org/content/102/suppl_1/6543)