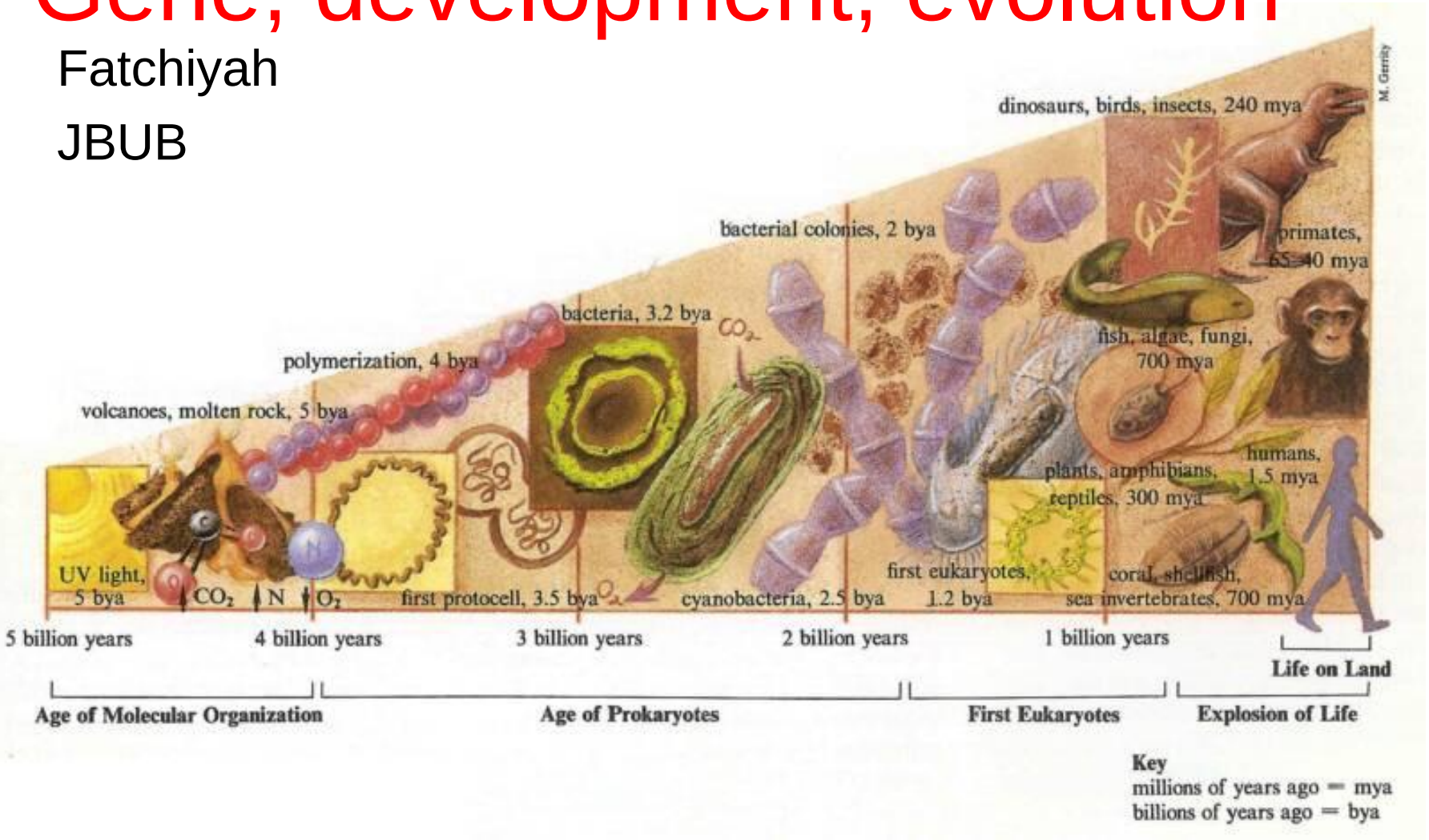
Gene, development, evolution

Fatchiyah

JBUB



Genome Evolution

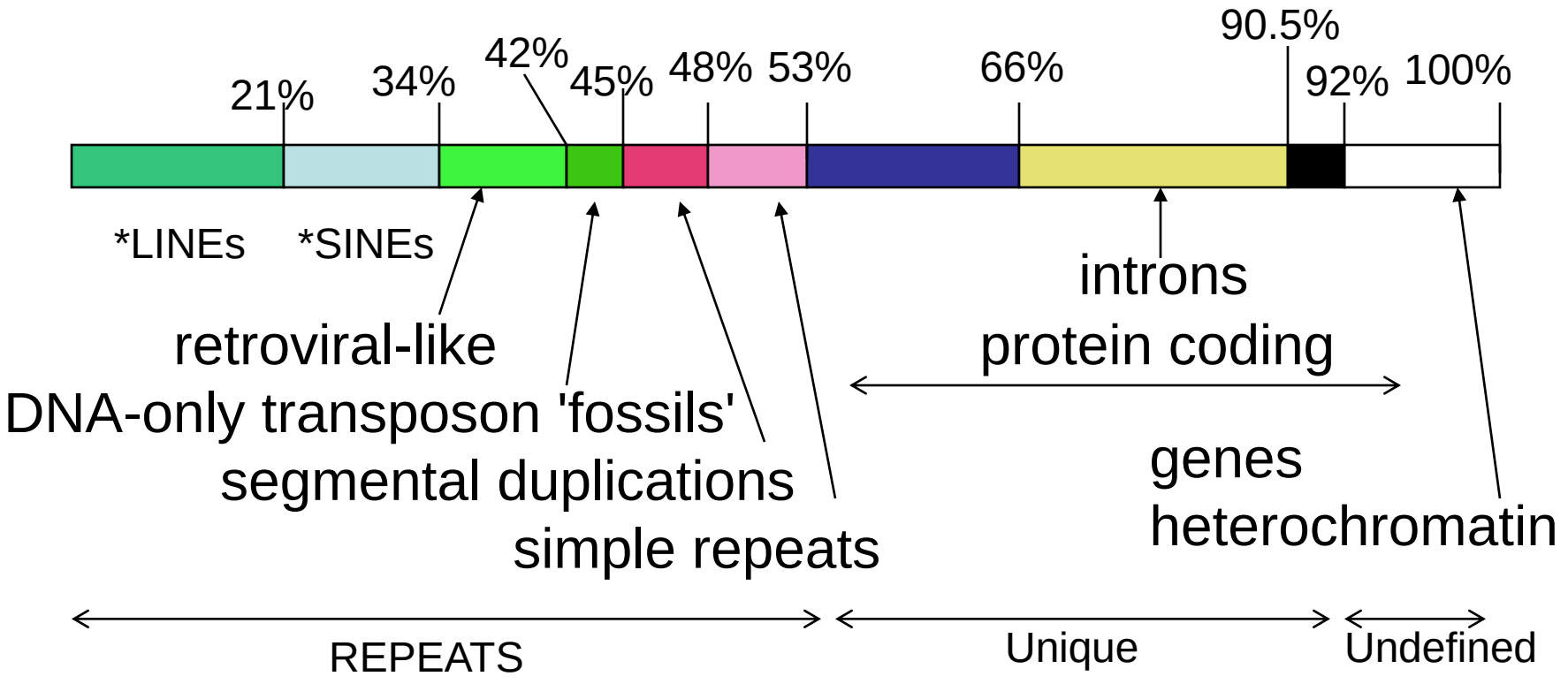
- **spontaneous mutations**
- **gene duplications, gene family expansions**
- **segmental duplications**
- **genome duplications (initially polyploidy)**
- **lateral gene transfer**
- **transposon insertions**
- **loss of not required information (extreme: obligate parasites)**

Junk DNA

potential junk:

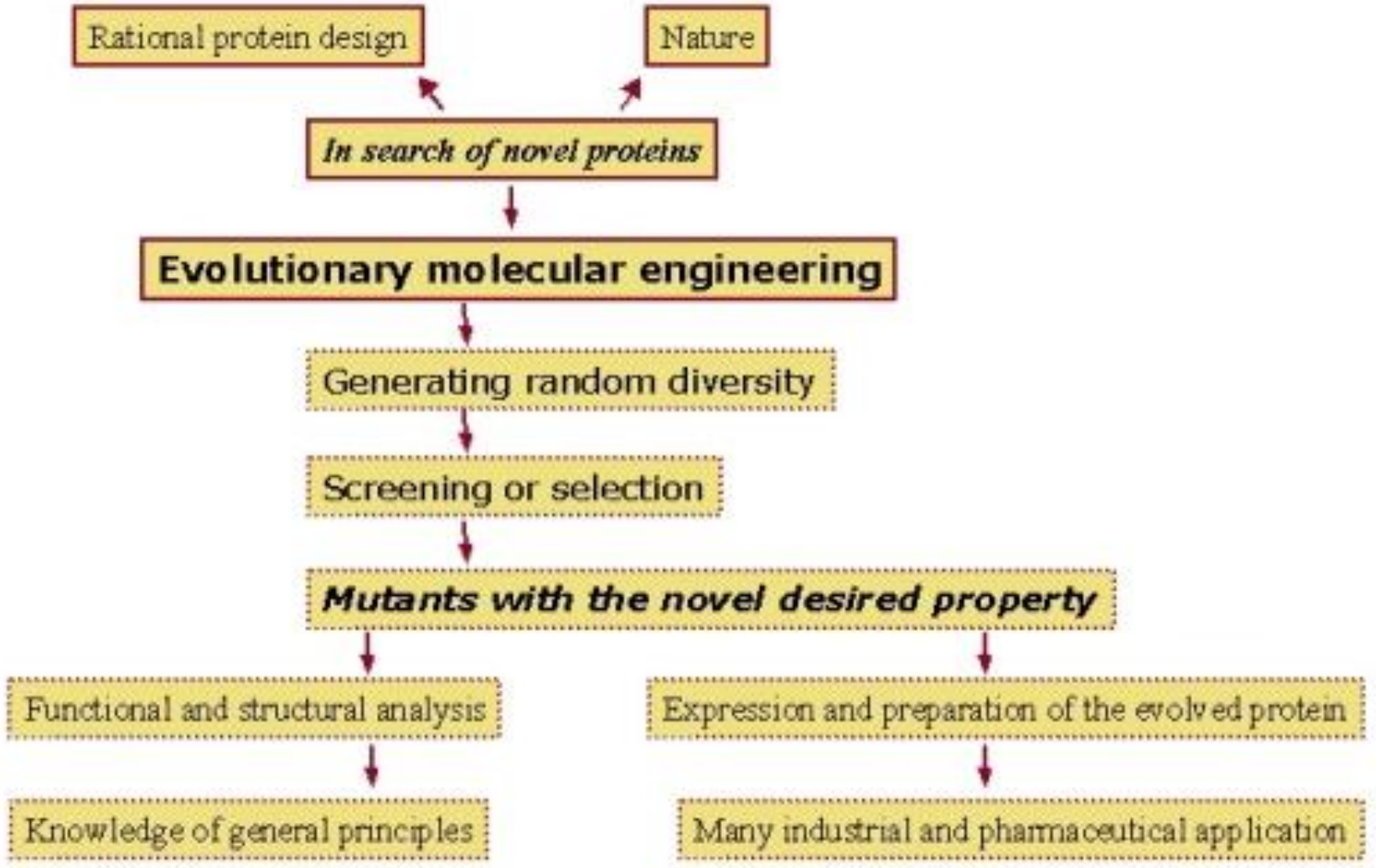
- introns
- repetitive elements
- intergenic regions
- parts of telomer and centromer structures
- ¾all parts of a genome for which a clear function is not yet defined

Percentage of 'Junk' in the Human Genome



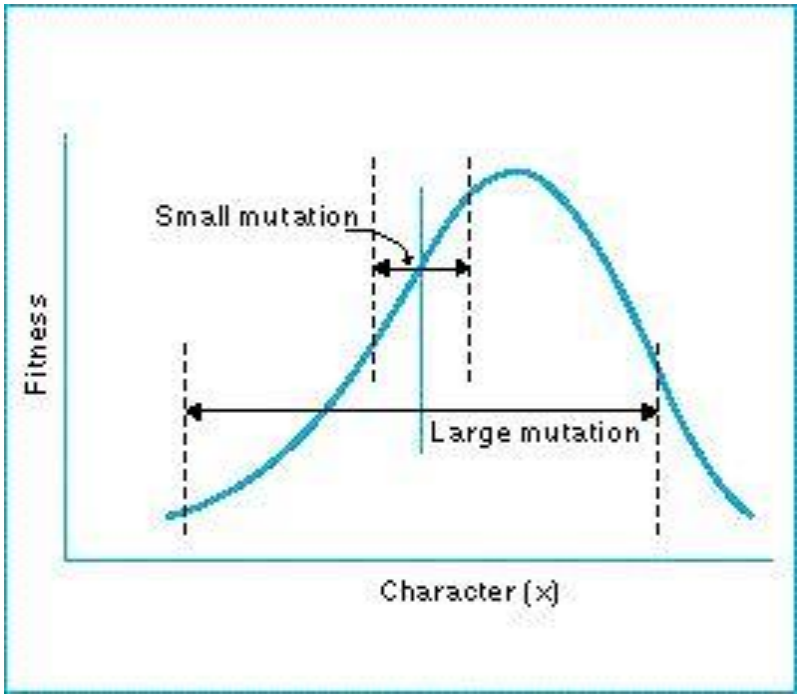
**L(S)INE: Long (Short) interspersed element, repetitive sequence Line, 6kb seq in primate genomes ; SINE, 300bp Alu-seq*

Enzymes can be tailored for optimal performance in industrial applications by evolutionary molecular engineering, also called directed evolution or *in vitro* molecular evolution.



Molecular evolution and neutral theory - Why do evolutionary rates differ?

- The fine-tuning mutants will:
- ➢ have lower selective advantage,
- ➢ and the larger proportion of selectively advantageous small mutations will be cancelled by their higher chance of random loss.



Macroevolutionary patterns

Microevolutionary processes → **?** → **Life's diversity**

How do evolutionary innovations arise?

How much genetic change is associated with phenotypic change?

<u>Evolution of development</u>

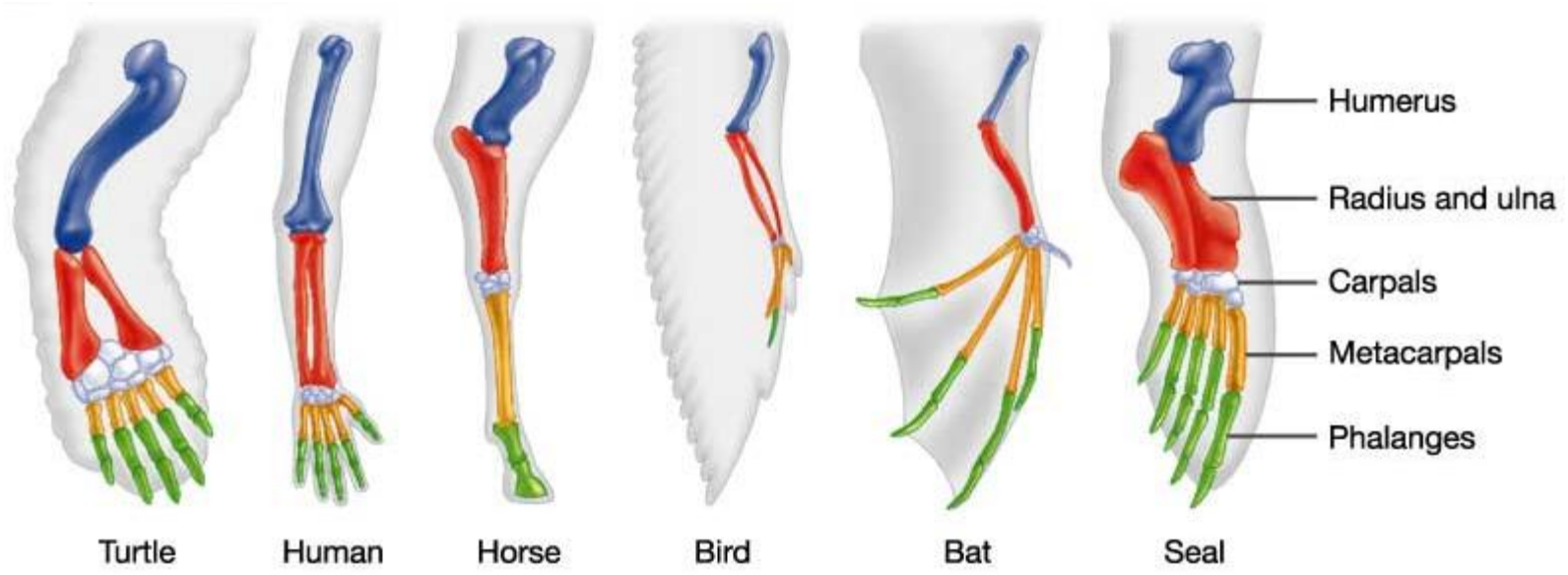
-similar development can lead to different adult forms

- small genetic change can have large phenotypic effects

- importance of cell fates

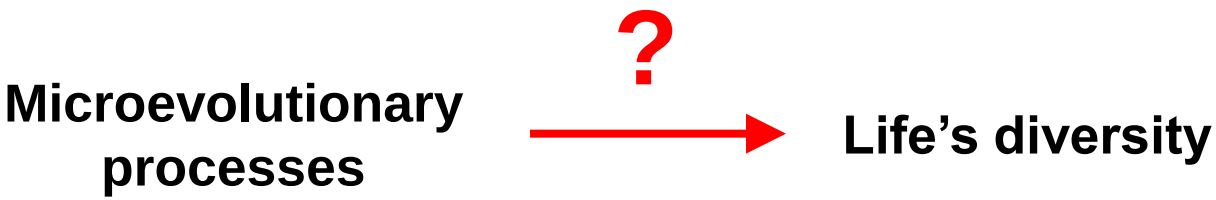
Evidence of evolution

homology – trait shared because of inheritance from common ancestor



example: similarity in limb structure among mammals

Macroevolutionary patterns



How do evolutionary innovations arise?

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Evolution of development

-similar development can lead to different adult forms

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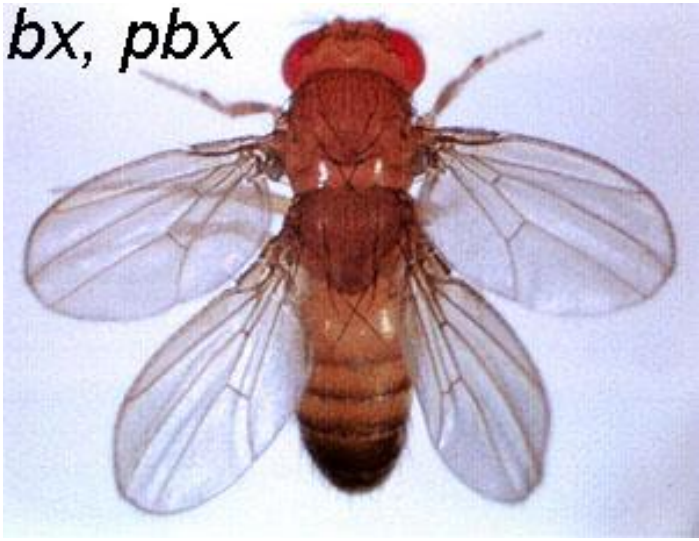
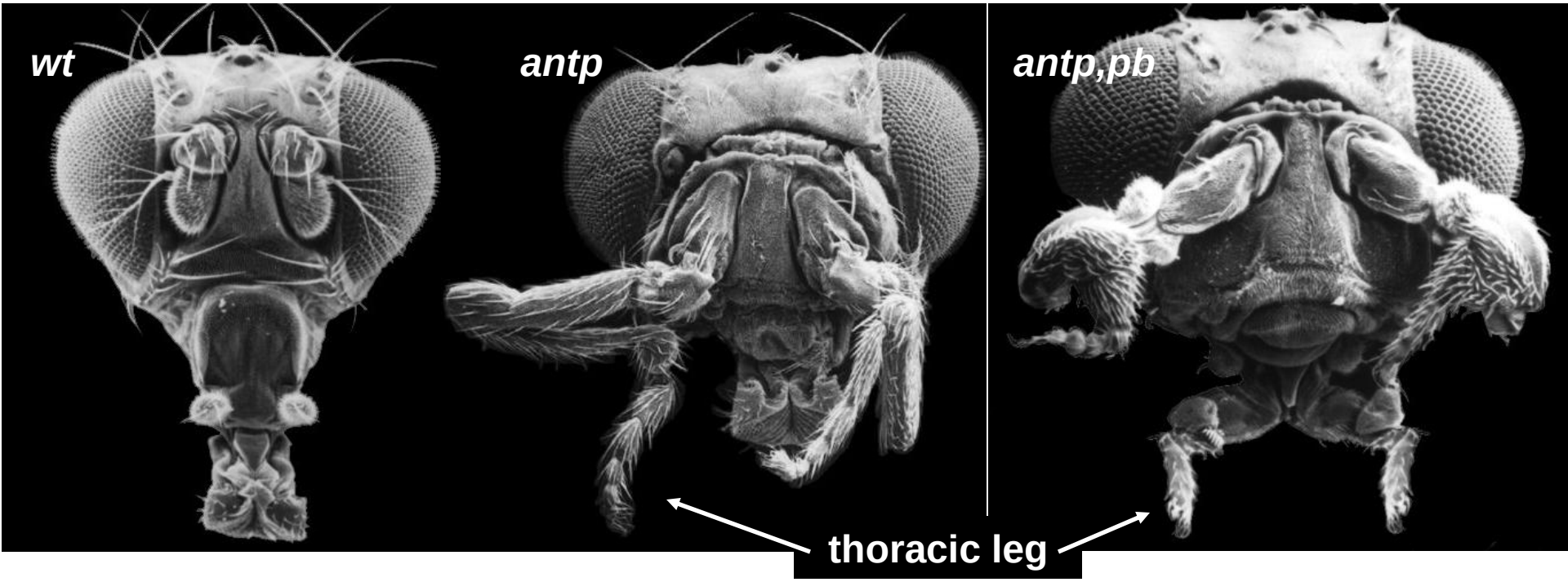
- importance of cell fates

Diversity of body plans reflects changes in number and interactions of a few genes associated with pattern formation in embryos.

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

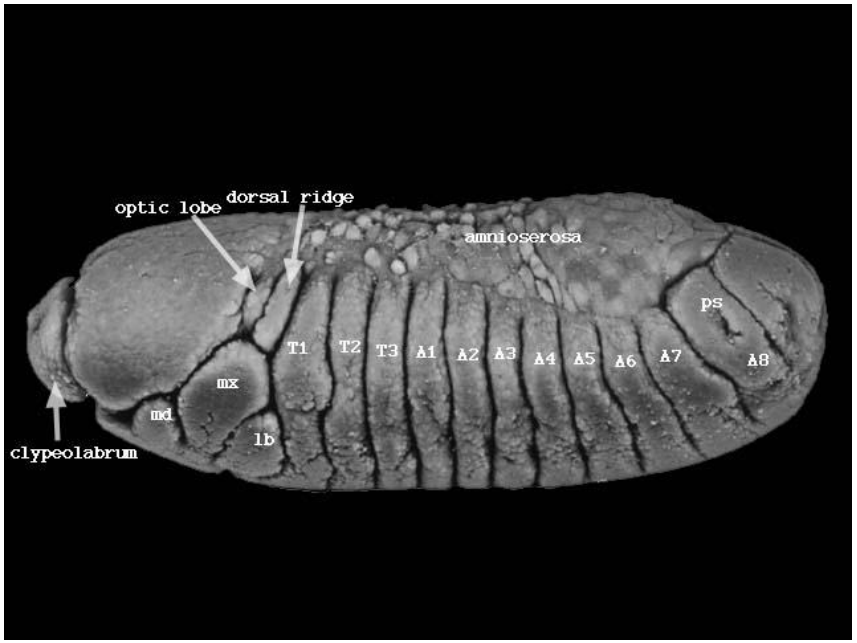


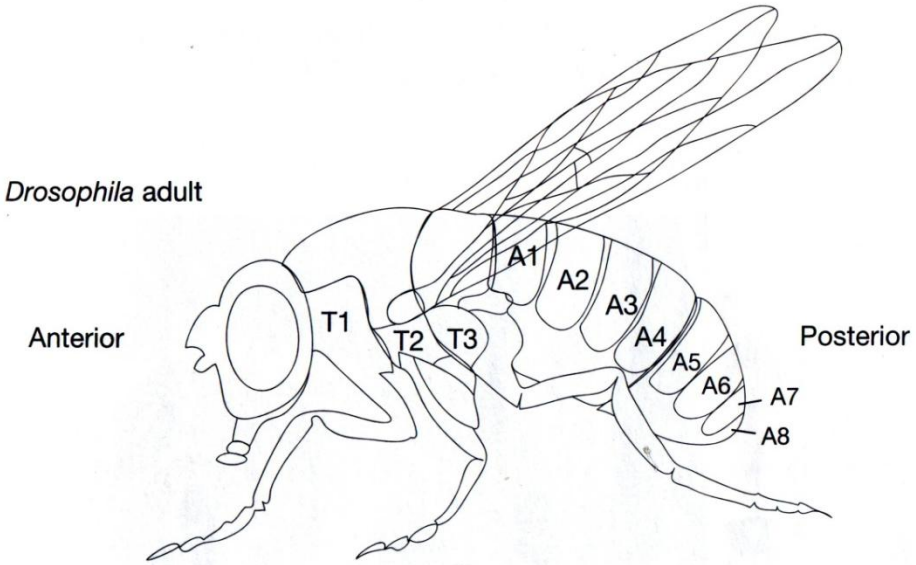
Homeotic mutations in *Drosophila*

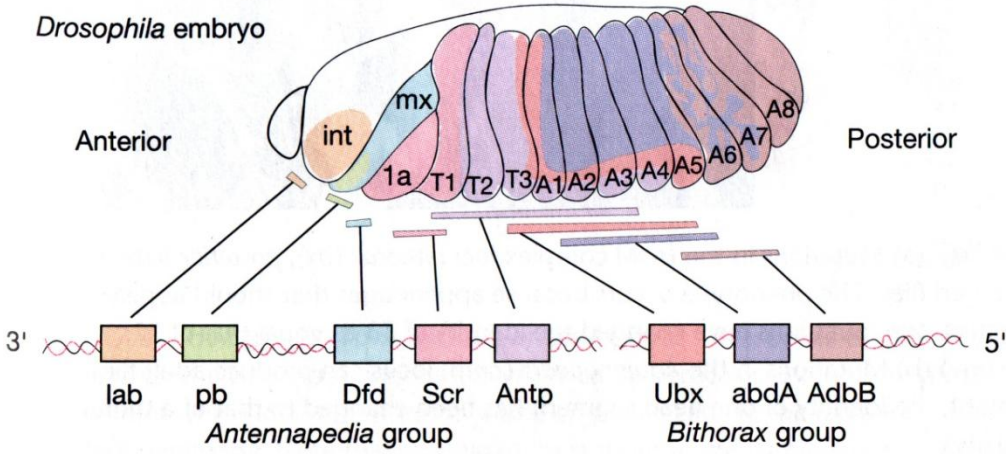


<u>Homeotic gene complexes</u> →
code for transcription factors that regulate expression of other genes

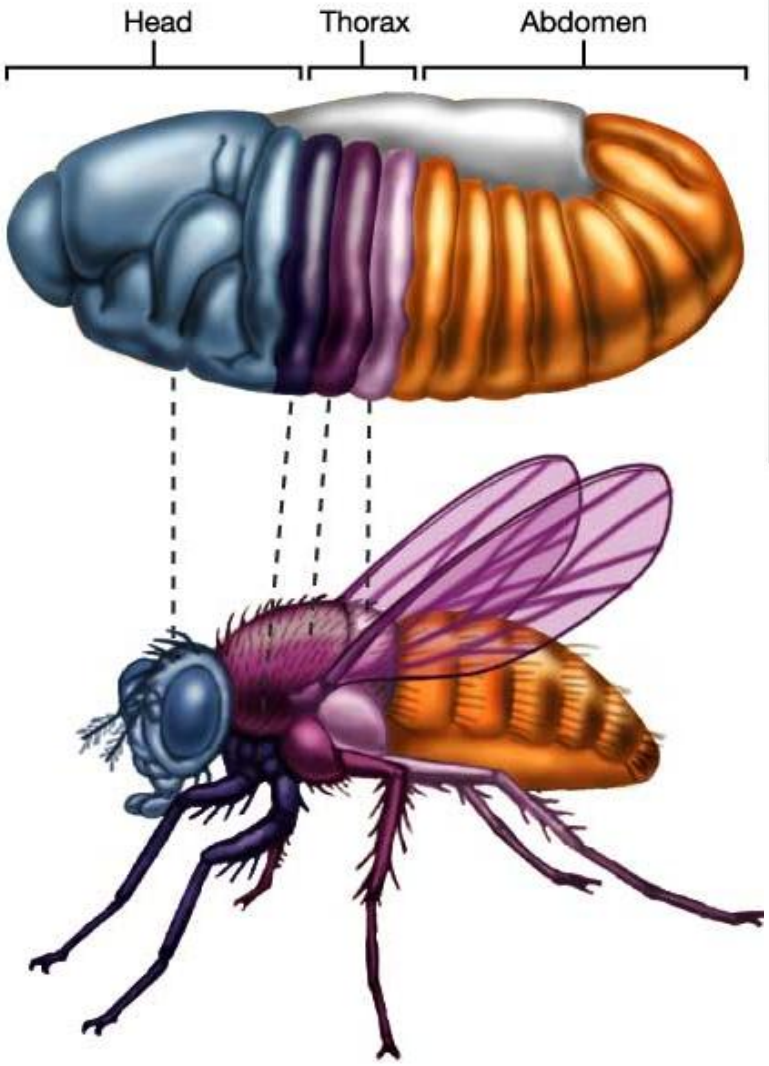
Hox gene expression in *Drosophila*



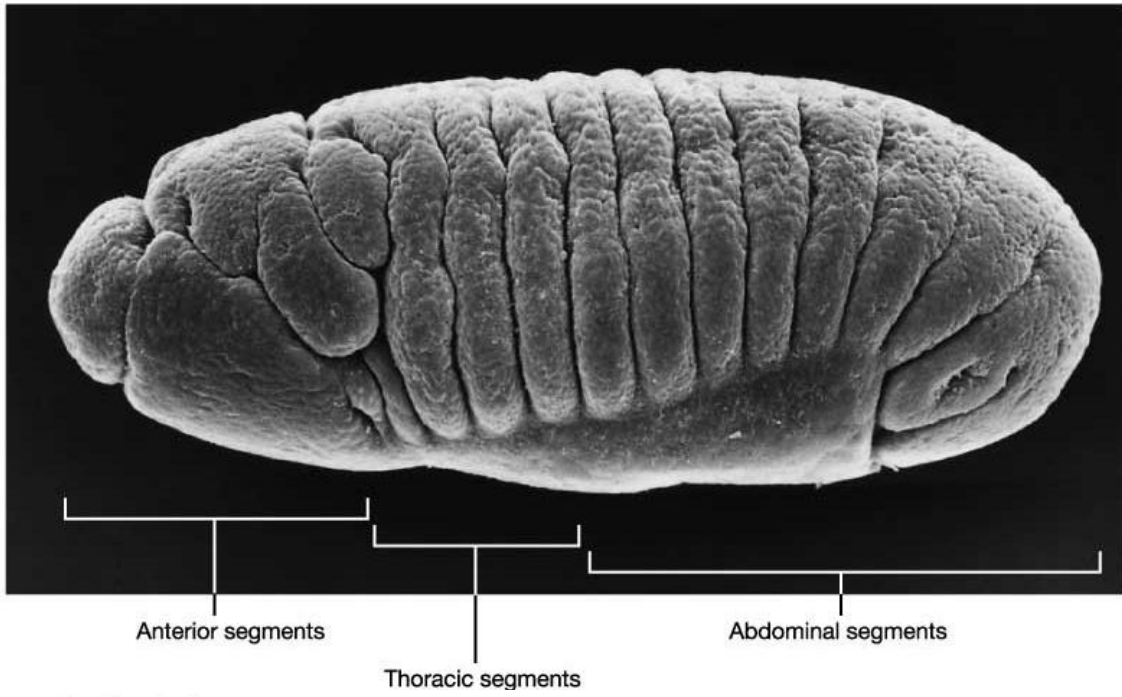




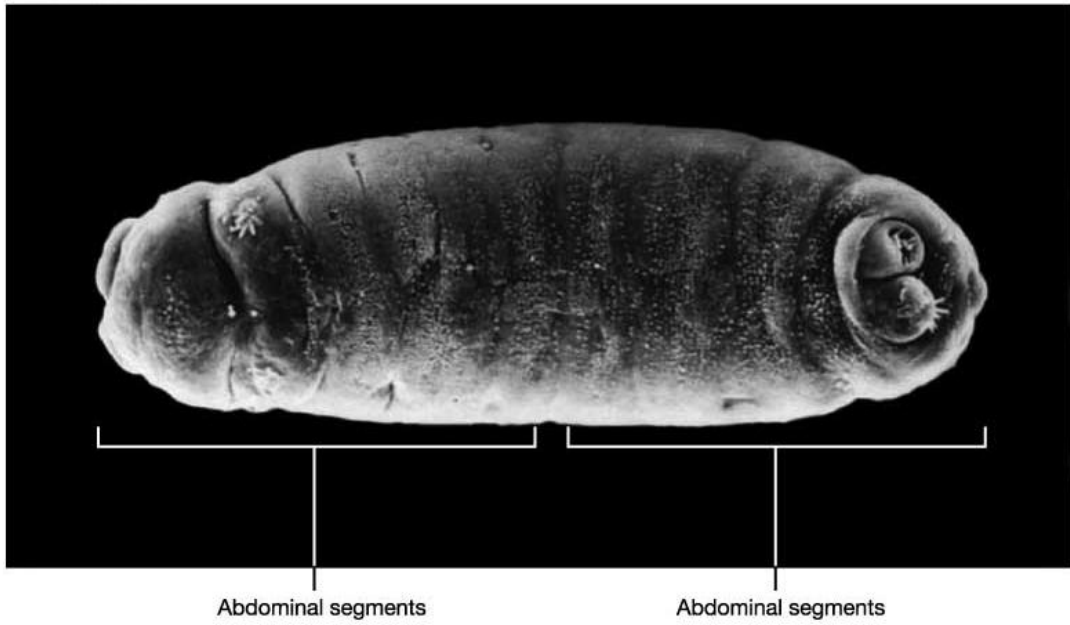
regulate genes to develop structures In the appropriate segments



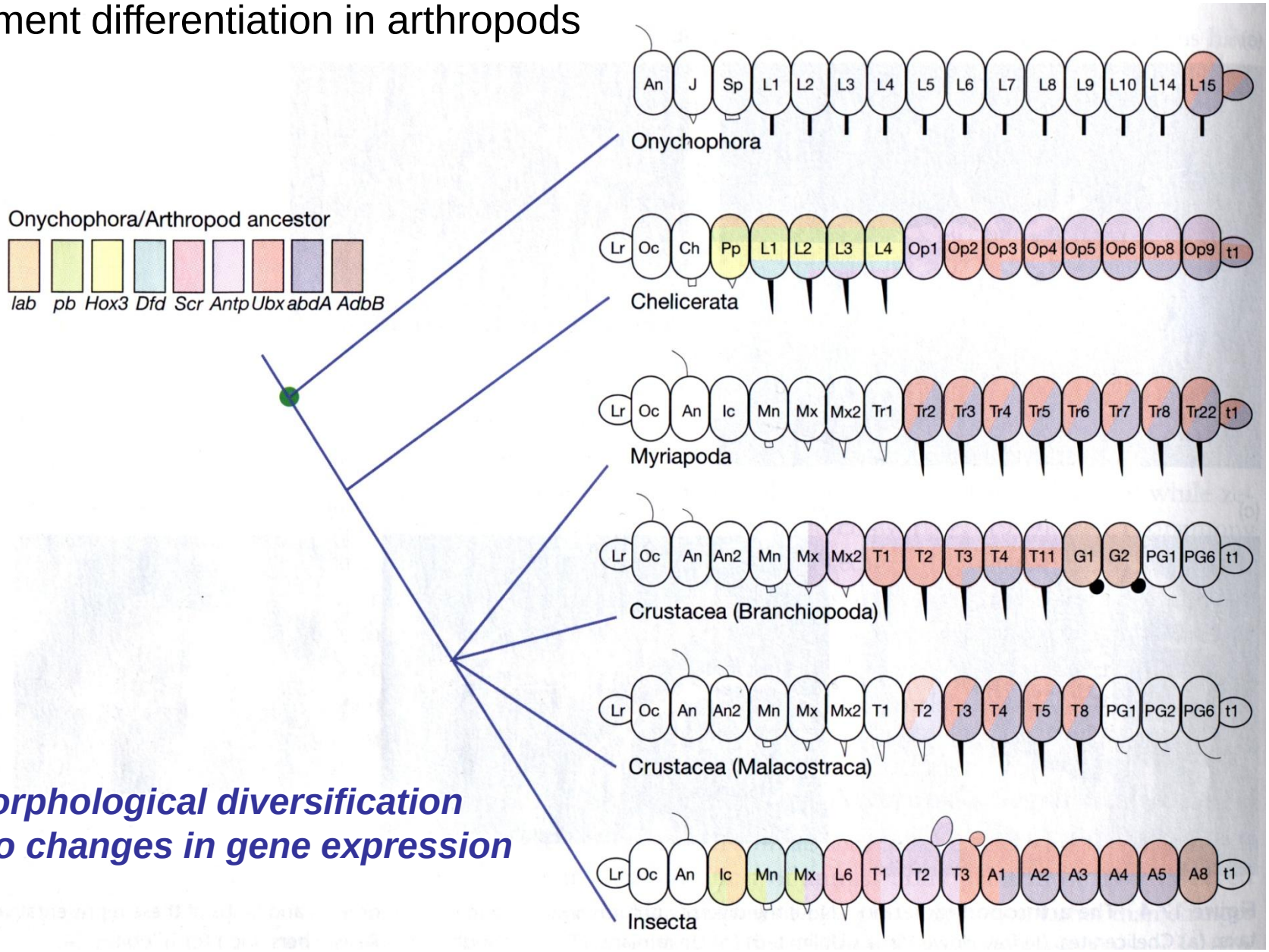
Normal fruit fly embryo



bicoid mutant



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



morphological diversification due to changes in gene expression

What we can learn from multiple sequence alignments

- An alignment is a hypothesis about the relatedness of a set of genes
- This information can be used to reconstruct the evolutionary history of those genes
- The history of the genes can provide us with information about the structure and function, and significance of a gene or family of genes

Can we reconstruct evolution

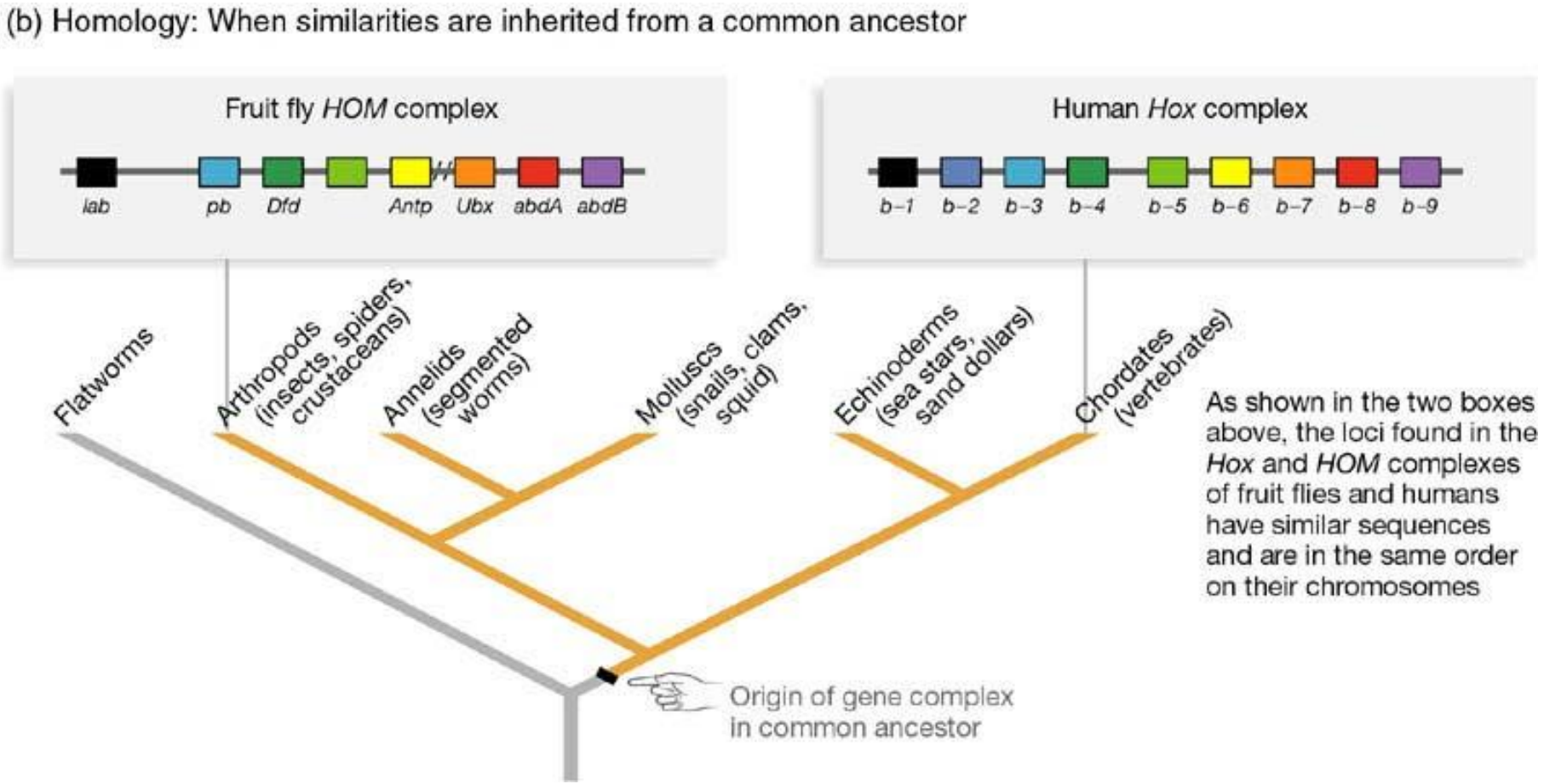
- We can also use the reconstructed history to test hypotheses about evolution itself:
  - Rates of change
  - The degree of change
  - Implications of change, etc
- We can then pose and test hypotheses about the evolution of phenomena unrelated to the genes
  - Evolution of flight in insects
  - Evolution of humans
  - Evolution of disease

Assumptions made by phylogenetic methods:

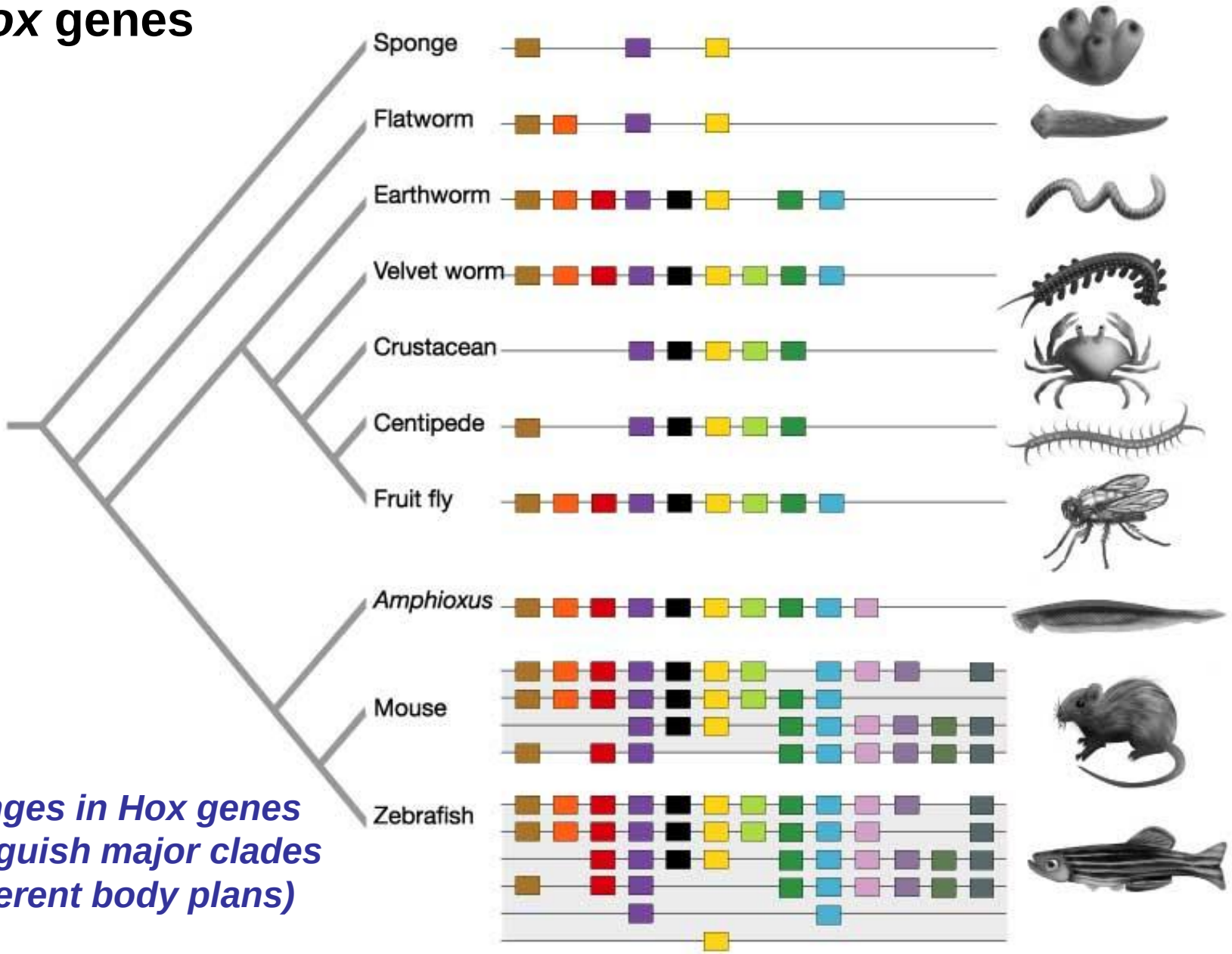
- The sequences are correct
- The sequence are homologous
- Each position is homologous
- The sampling of taxa or genes is sufficient to resolve the problem of interest
- Sequence variation is representative of the broader group of interest
- Sequence variation contains sufficient phylogenetic signal (as opposed to noise) to resolve the problem of intereest
- Each position in the sequence evolved independently

Shared traits

1) Homology – inherited from common ancestor



Homology of *Hox* genes



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

Evolution and society

Science – addresses questions about natural world (empirical, material)
– does not address questions beyond the natural world
-these questions are left to religion and ethics

Hypothesis – an idea to be tested
– can be supported but cannot be "proven"
b/c always the possibility of falsifying the hypotheses

Testable (falsifiable) hypothesis – material evidence could disagree

Theory – hypothesis that's been supported over and over again
(e.g., germ theory, cell theory, theory of gravity, theory of evolution)

Evolution and society

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"natural" ≠ good, right, just, moral

Does an evolutionary explanation for infidelity justify this behavior?
murder ?
discrimination ?

Evolution and Religion

What is the evolution versus creationism dispute about?

1) **What is the origin of humans? Are we special?**

   If we're "just" animals does this justify immorality?

   → Science does not deal with whether behaviors are moral or immoral.

2) **Is God excluded?**

   If evolution can explain the origins of life, is there a role for God?

   → Science does not test hypotheses about the spiritual world.

SCIENCE & RELIGION CAN BE COMPATIBLE.

Evolution and Religion

What about "creation science" and "intelligent design"?

- Recent movements in the U.S.
- Response to immorality of modern society

- Not scientific: don't provide falsifiable hypotheses

- Goal: to influence laypeople (not to convince scientists)

SCIENCE & RELIGION CAN BE COMPATIBLE.

physical world ⟵ ⟶ ***morality, spirituality***

Evolution and society

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Does evolutionary biology justify social policies?

"Social Darwinism" – 19th c. extension of natural selection to society

- "fittest" (most competitive) should gain the most power and wealth
- justification of inequity as "survival of fittest"

- **flaw:** equating a "natural" process with good

Evolution and society

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Does evolutionary biology justify social policies?

Eugenics – early 20th c. application of artificial selection to humans

- successful, wealthy families:
  - interpreted as evidence that intelligence and talent are heritable
  - denies any influence of education, money, privilege
- in US, sterilized institutionalized people with "undesirable" traits
  - "feeblemindedness", immorality, alcoholism
  - no evidence of these traits heritability
- in Nazi Germany, extended eugenic practices to genocide
  - used science to justify hatred of another group

- **scientific flaw:** lack of information about heritability of traits
- **fundamental flaw:** denies people's right to self-determination

Evolution and society

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Does evolutionary biology justify social policies?

Biological determinism:

- belief that individual differences are biologically determined and fixed.

  biological influences >> social influences

- modern biology replaces "nature versus nurture" debate with understanding that genes, environment, and G x E affect traits

- **flaw:** assuming that genetically influenced traits are immutable

- **biology does not rule out environmental influences like culture**

Evolution and Gender

Are sex differences biological?

genetic

environmental (cultural)

uterus

aggression?

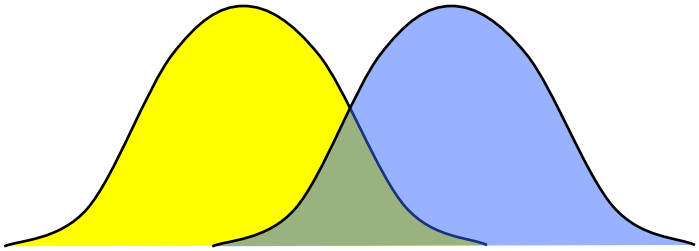
promiscuity?

intelligence?

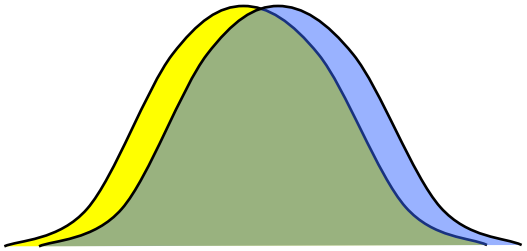
high heels

Do measurable differences justify different treatment?

Men tend to be stronger than women.
Should women be allowed to be firefighters?



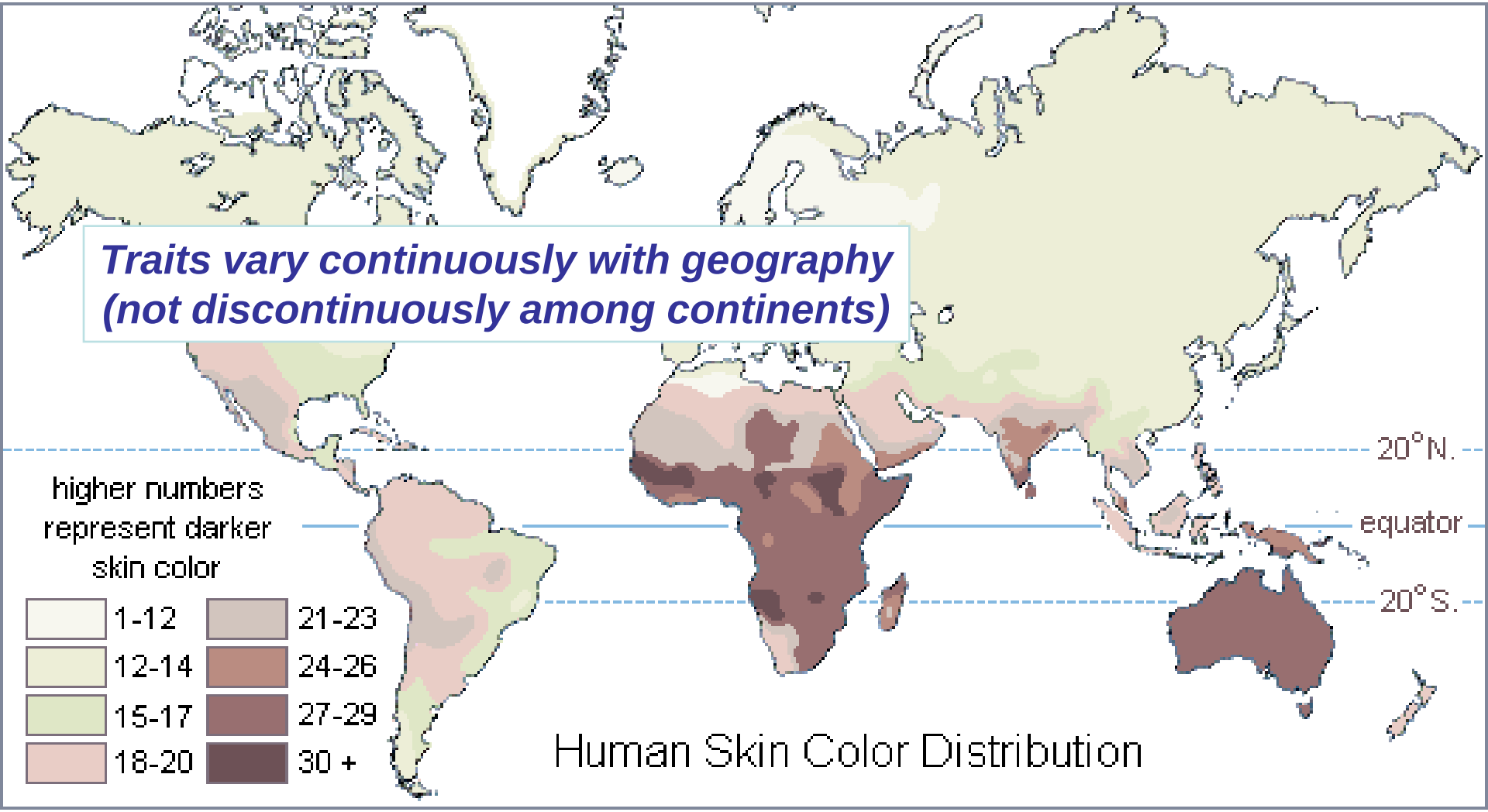
some predictability



lack of predictability

Evolution and Race

Is race a biological reality?



Evolution and Race

Is race a biological reality?

There are genetic differences in superficial traits that vary geographically.

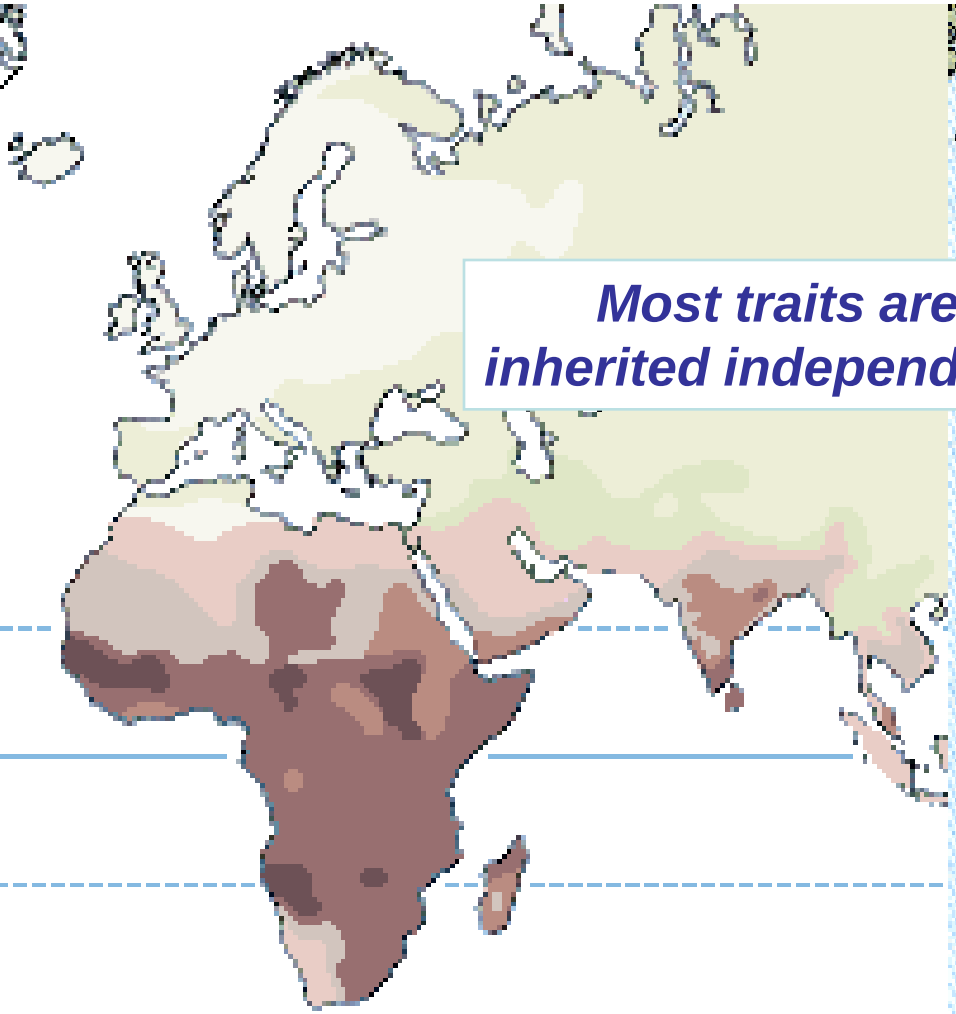
e.g., skin color

BUT, the traits used to infer “race”:

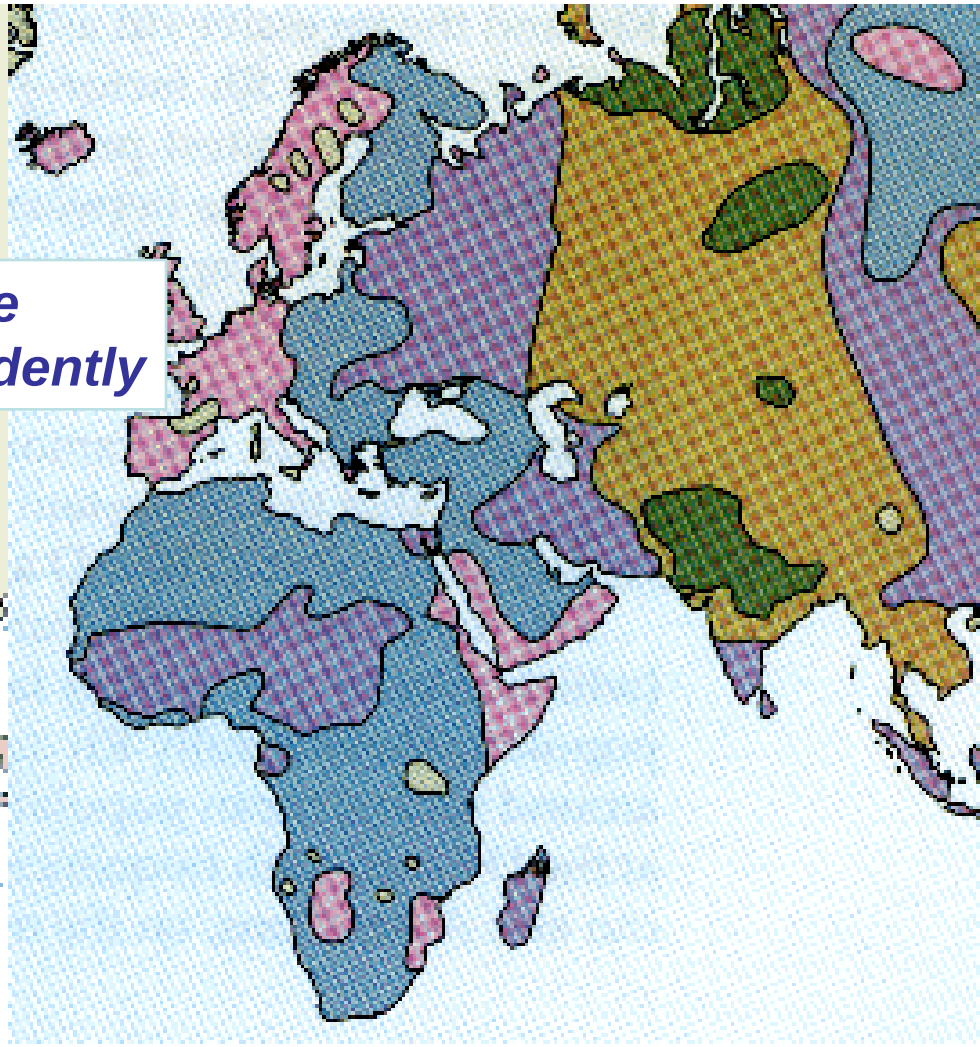
- show continuous variation across geographic range
- *do not* differentiate the different continents

Do these visible differences reveal any other more substantial differences?

Evolution and Race



Most traits are inherited independently



Skin color

Type B blood allele frequency

Evolution and Race

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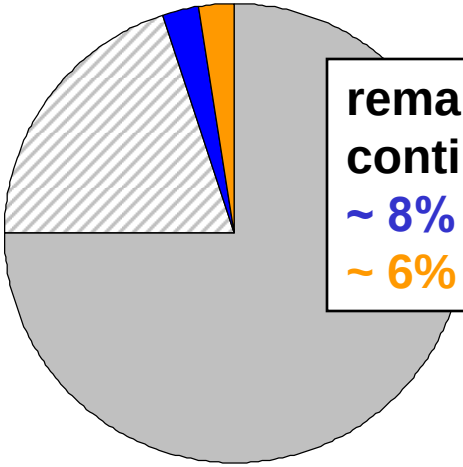
Evolution and Race

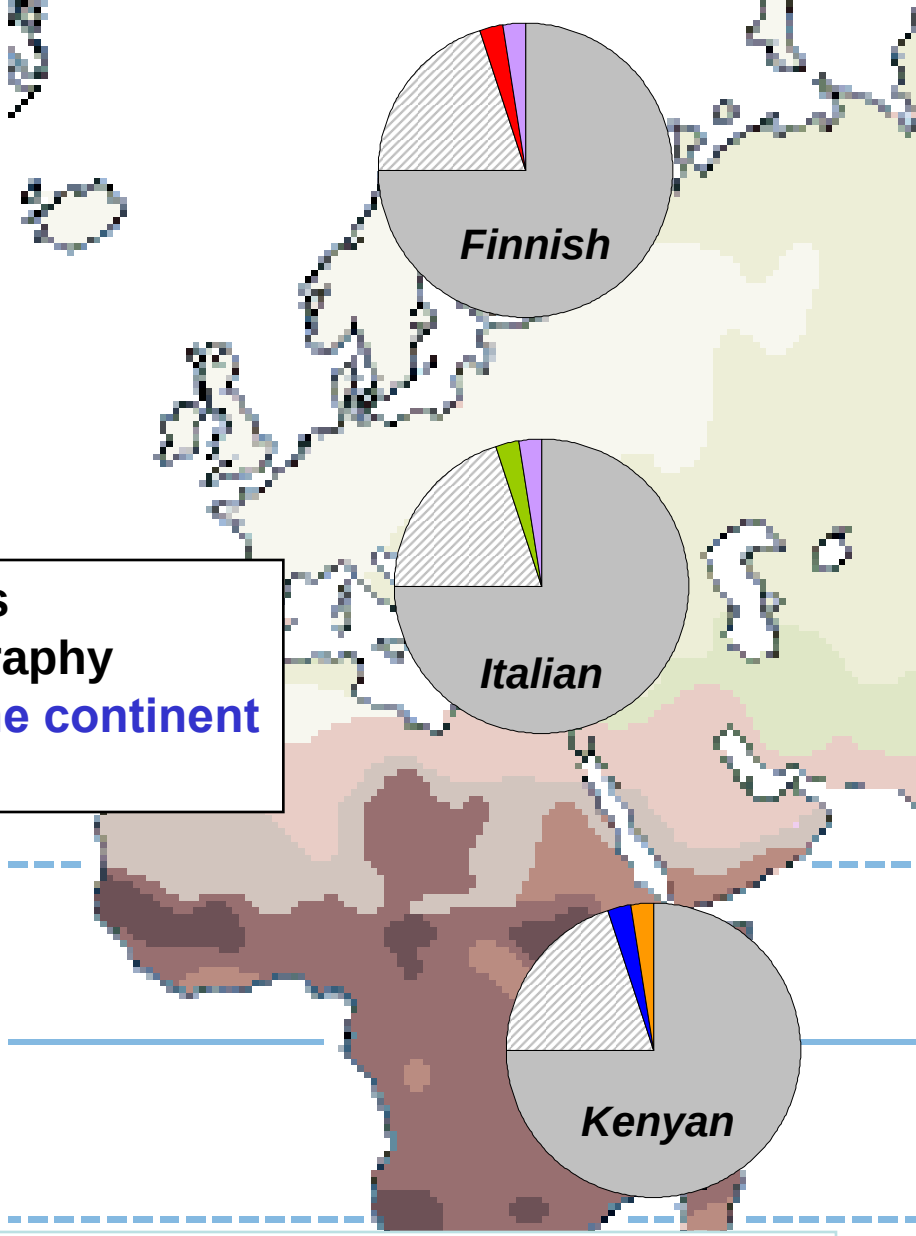
Is race a biological reality?

most (~ 85%) human genetic variation is shared among populations
ex: A,B,O blood alleles



remaining (~ 15%) varies continuously with geography
~ 8% btwn pop's on same continent
~ 6% btwn continents

~ 75% of human genes are fixed (identical among all humans)



~ 94% of all human variation can be found on each continent
~ 84% can be found in a single population

Evolution and Race

Is race a biological reality?

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e.g., skin color

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Human variation is real, but "race" isn't an effective way to organize it.

Why not?

Evolution and Race

Human populations are very similar genetically.

→ "Races" are not subspecies.

Humans are a very young species –

- modern humans evolved: 150,000 – 200,000 years ago
- Out of Africa migration: 50,000 – 100,000 years ago

→ very little time for geographic divergence

Humans have always had gene flow –

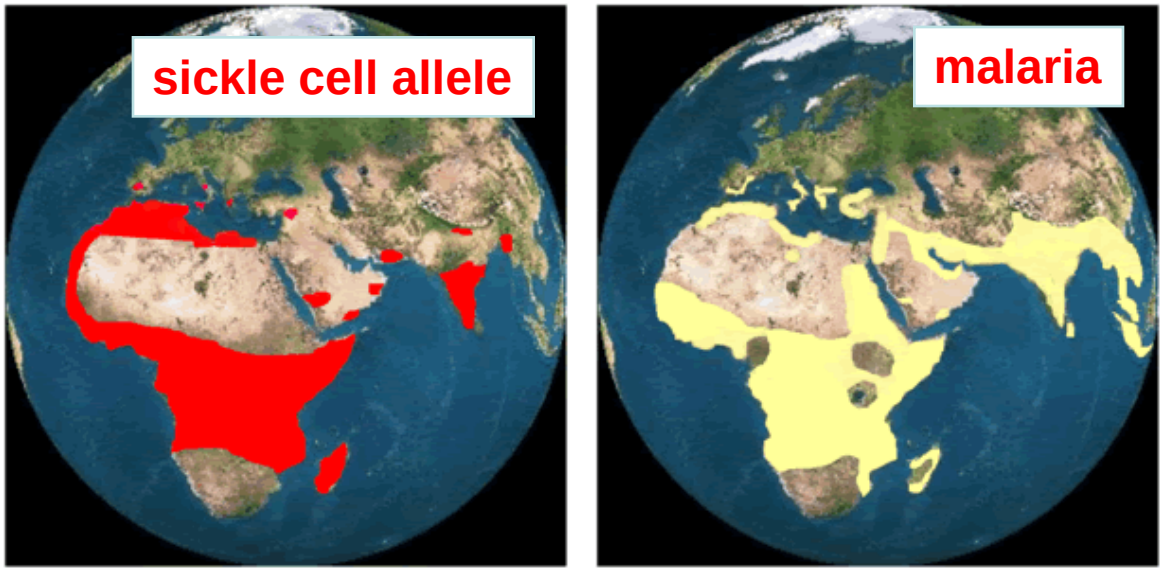
- small scale migrations among nearby villages
- large scale migrations due to exploration, trade, wars, etc.

Without isolation and without a lot of time, very little divergence.

Evolution and Race

But doesn't disease risk vary among people of different races?

Sickle cell:
people with ancestors from areas with malaria



Are all disease risks genetic?

Does focusing on genetics lead scientist to ignore other (societal) sources of variation in disease risk?

Is it ethical to focus screening on a more susceptible group, even when members of other groups may still get the disease?

How can evolutionary biology help fight disease?

How do pathogens evolve to be harmful?

Can we stop pathogens from evolving harmful traits?

- **Evolution of <u>drug resistance</u>**

  mutation – rare mutations for resistance genes

  natural selection – resistant individuals have higher fitness in environments with the drug

  → *changing the **<u>selective environment</u>** can slow the evolution of resistance*
  (presence of drug)

- **Evolution of <u>virulence</u>**

  virulence – how harmful a pathogen is to its host

  depends on **natural selection** and **migration**

  → *decreasing opportunities for **<u>migration</u>** can make virulence less adaptive*
  (spread to new host)

When should pathogens evolve high virulence?

Pathogen populations that grow quickly are more harmful (virulent)

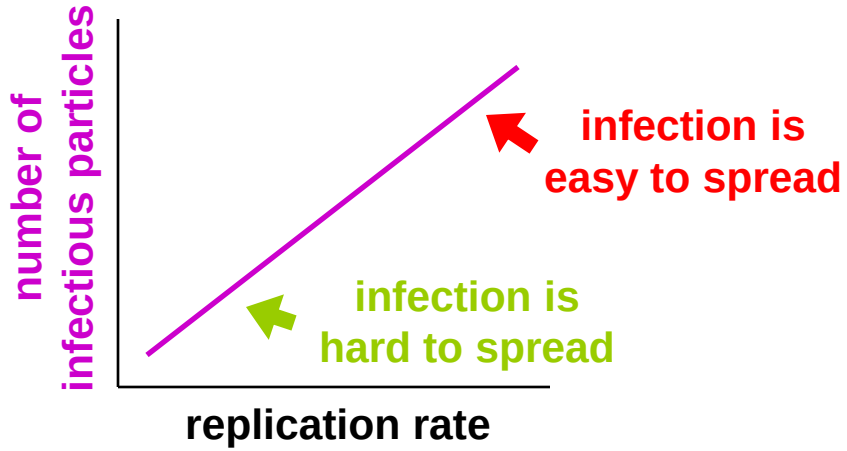
Should natural selection favor alleles that promote high replication rates?

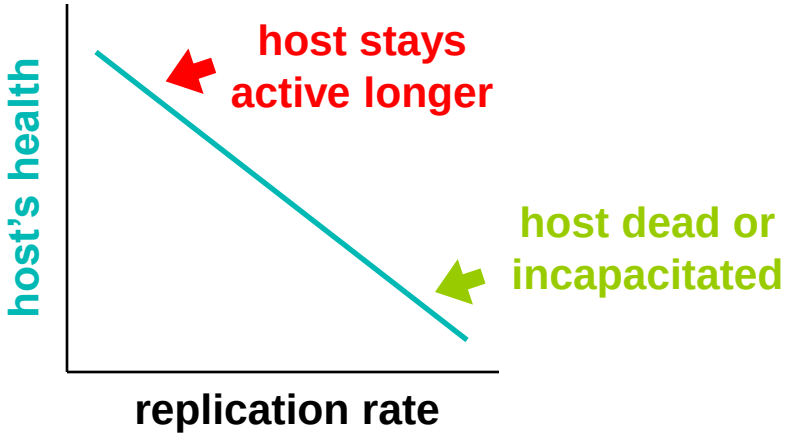
Pathogen fitness depends on spread to new hosts *(migration)*

high replication → more infectious particles produced

→ more likely to kill the host

trade-off





When should pathogens evolve high virulence?

Virulent pathogens → many infectious particles in host → **easy to spread** → **host very sick**

Avirulent pathogens → few infectious particles in host → **hard to spread** → **host not very sick**

trade-off

transmission rate (trade-off) hypothesis: **transmission requires opportunities for pathogen to spread to new hosts**

many transmission opportunities

→ contact with many potential hosts
→ if infectious, can transmit to many new hosts in short time
→ **favors high virulence**

few transmission opportunities

→ contact with few potential hosts
→ must live a long time to have transmission opportunities
→ **favors low virulence**