Lecture 15. Genetic Variation

EEB 2245, C. Simon, 21 March 2017

Last time…

- Uses of Evolutionary Biology (continued)
- Evolution vs Creationism

This time…

- Importance of variation, polymorphisms
- Mendel's laws
- Deviations from Mendelian ratios (genetic and non genetic causes)

This Time..

- Heritability, plasticity, selection, common garden, genetic assimilation, canalization
- Epigenetic inheritance
- H-W equation, assumptions, terminology, usefulness
- Importance of Lewontin & Hubby 1966

Genetic Variation

Variation in traits results in multiple phenotypes within a population or "polymorphism"

- Systematists work with "type specimens"
- But in nature, species are not unvarying types
- Traits vary with-in and among-populations
- Thousands of examples of polymorphic traits,
- Examples: color or color pattern
  - Albino, melanistic, green vs. yellow, brown (or pink)
  - Mimicry pattern polymorphism

Albino mutations

Green/Brown Color Polymorphism *Kikihia peninsularis* cicadas from NZ

Orange and purple morphs, *Pisaster ochraceus* starfish Pacific NW

Heliconius butterflies- color pattern polymorphisms controlled by one large region of the genome less than one megabase long containing **~ 20 genes** differing in expression among the different forms.
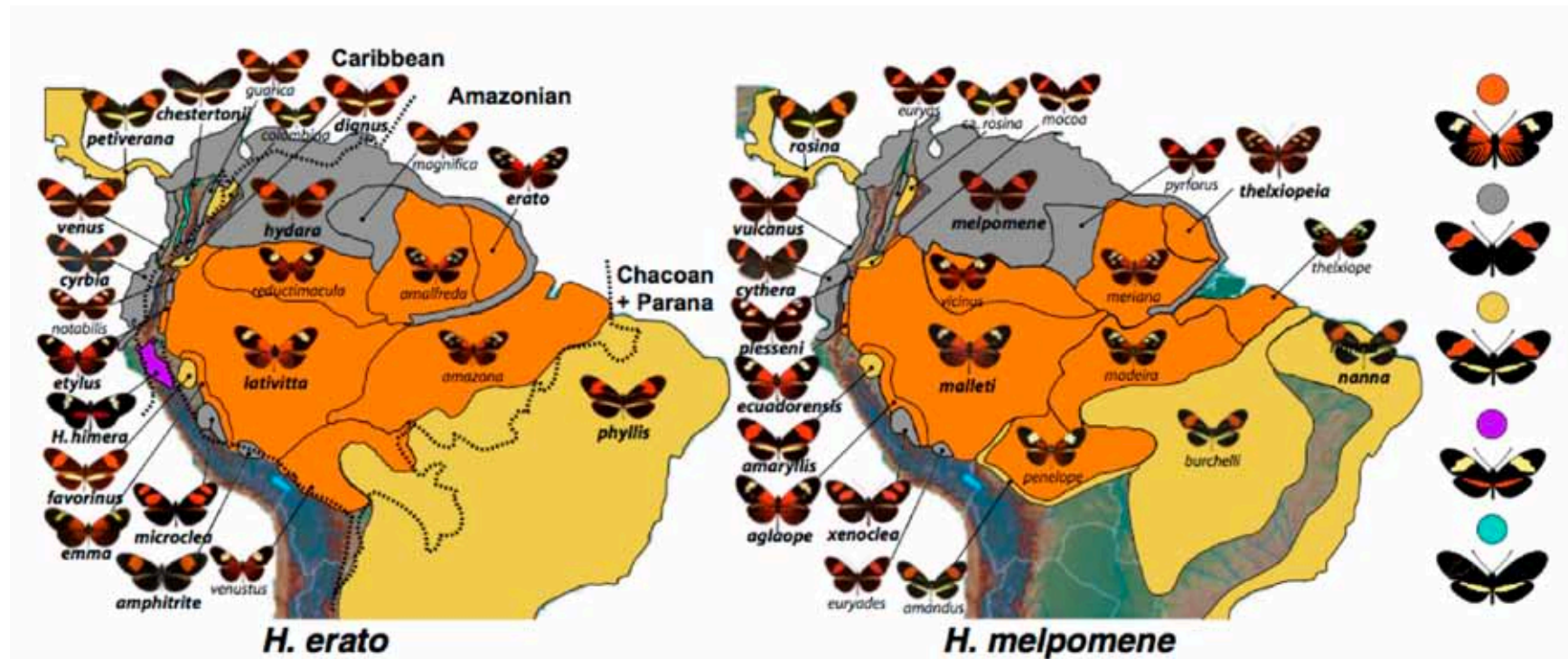
Polymorphism Examples in Futuyma:

- Blue geese vs snow geese (2 alleles, 1 locus)
- Swallowtail butterfly, *Papilio dardanus,* males non-mimetic, females mimic three very different species
- Multilocus traits w many alleles such as hair and skin color

Simple Mendelian Inheritance

Given the following mating (one trait)..   AA x aa → F_1

A - dominant, a - recessive

All F_1 genotype Aa   (phenotype A)   Aa x Aa → F_2

In F_2, recessive alleles segregate out
but minority (25%)

¼ AA, ½ Aa, ¼ aa

	A	**a**
A	AA	Aa
a	aA	aa

Review from basic biology….

Mendel's laws

Dominance- two alleles, one from each parent. recessive alleles will segregate out in the next generation (hidden variation). Dominance can also be incomplete.
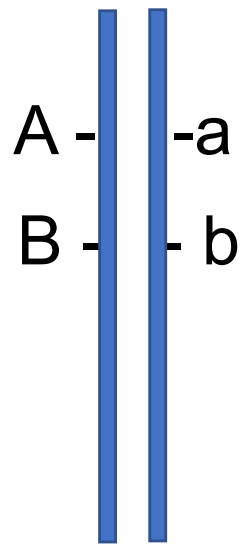
•**Segregation**- paired alleles segregate at random into gametes

•**Independent assortment-** segregation of one pair of alleles is unrelated to the segregation of any other pair of alleles

Non-Mendelian Inheritance

Many factors can cause deviations from Mendelian ratios

e.g., tight linkage- alleles close together on the same chromosome.

A - -a

B - - b

Tight linkage

A- -a

X

B- -b

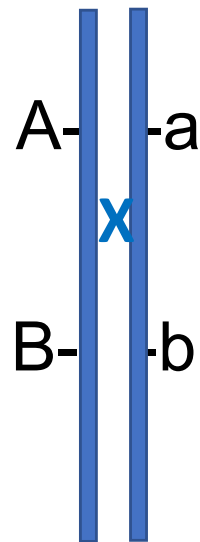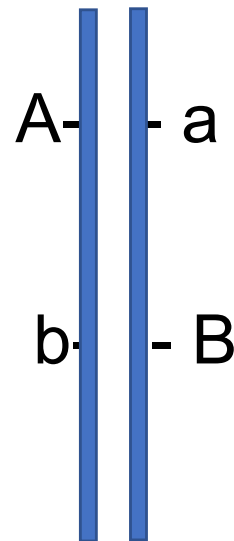Less tight

A- - a

b- - B

Two loci A & B

Non-Mendelian Inheritance

Many factors can cause deviations from Mendelian ratios

e.g., tight linkage- alleles close together on the same chromosome.

Violates Independent assortment

segregation of one pair of alleles **will be** related to the segregation of other pairs of alleles

The farther apart two loci on a chromosome, the more likely they are to assort independently

Crossing over.

Genes on same chromosome can assort independently

Causes of deviation from Mendelian ratios of offspring allele frequencies in crosses

- Tight Linkage (genetic hitch-hiking)
- Meiotic drive (segregation distortion)
- Lethal alleles
- Epistasis (many genes affecting one trait)
- Transposable elements
- New mutations (rare)
- Non-genetic variation- Cultural inheritance
- Plastic response to the environment
- Non-genetic variation- Maternal effects
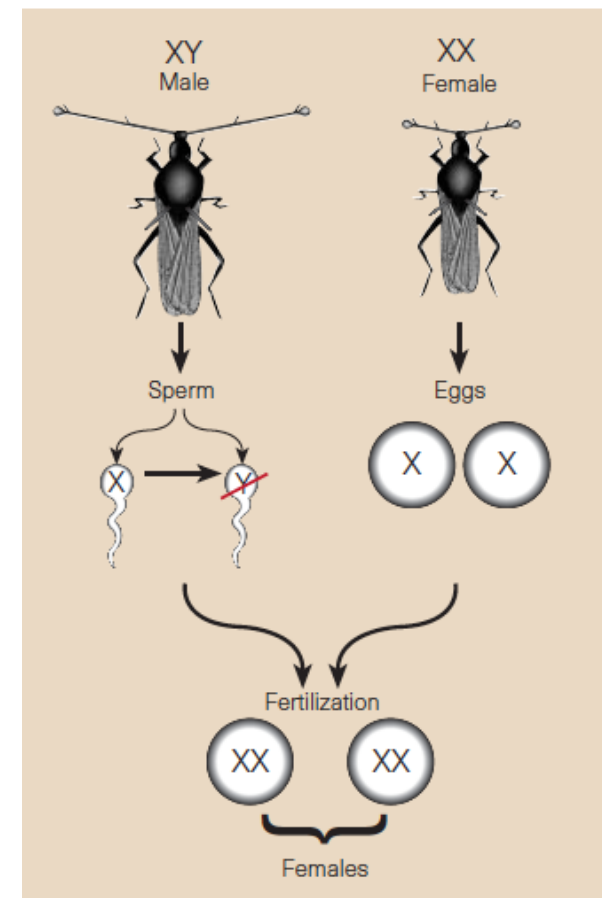- Epigenetic inheritance

Violation of Mendel's laws

- **Independent assortment-** segregation of one pair of alleles unrelated to the segregation of any other
- Violated by Meiotic Drive = Segregation distortion
- x-linked genes.
- increases the proportion of x-bearing sperm (producing females).

Normal genotypes

	X	Y
X	XX	XY
X	XX	XY

Distorted genotypes

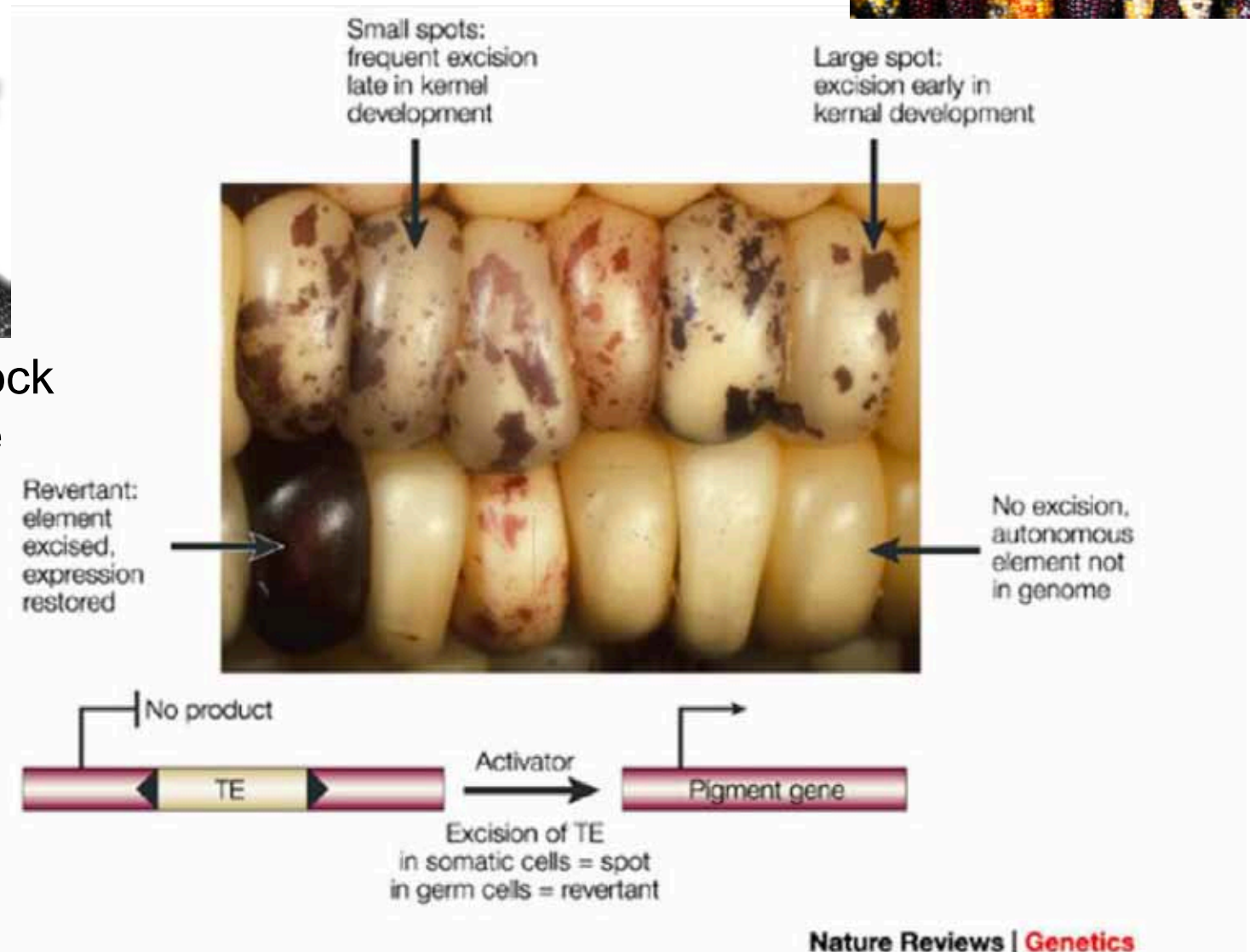# Causes of deviation from Mendelian ratios of offspring allele frequencies in crosses

- Tight Linkage (genetic hitch-hiking)
- Meiotic drive (segregation distortion)
- Lethal alleles (in embryo)
- Epistasis (many genes affecting one trait)
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Transposable elements

Barbara McClintock
1983 Nobel Prize

Small spots: frequent excision late in kernel development

Large spot: excision early in kernal development

Revertant: element excised, expression restored

No excision, autonomous element not in genome

No product

TE

Activator

Pigment gene

Excision of TE
in somatic cells = spot
in germ cells = revertant

Nature Reviews | Genetics

Barbara McClintock (1902-1992)

- PhD, Cornell, 1927- maize genetics
- First genetic map for maize
- Role of telomere and centromere
- Cornell –Missouri (1936) -- Cold Spring Harbor (1941)
- 1940's-50's discovered transposition in maize
- National Academy of Sciences 1944
- 1953 –Due to harsh skepticism--- stopped publishing on transposition
- 1983 – awarded Nobel Prize for this work

Causes of deviation from Mendelian ratios of offspring allele frequencies in crosses

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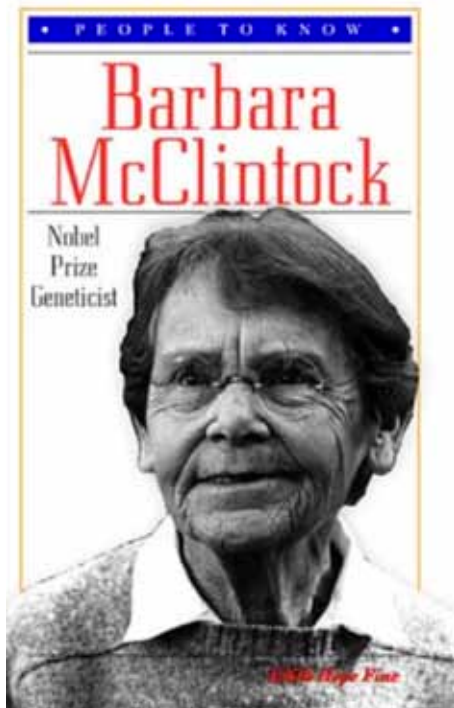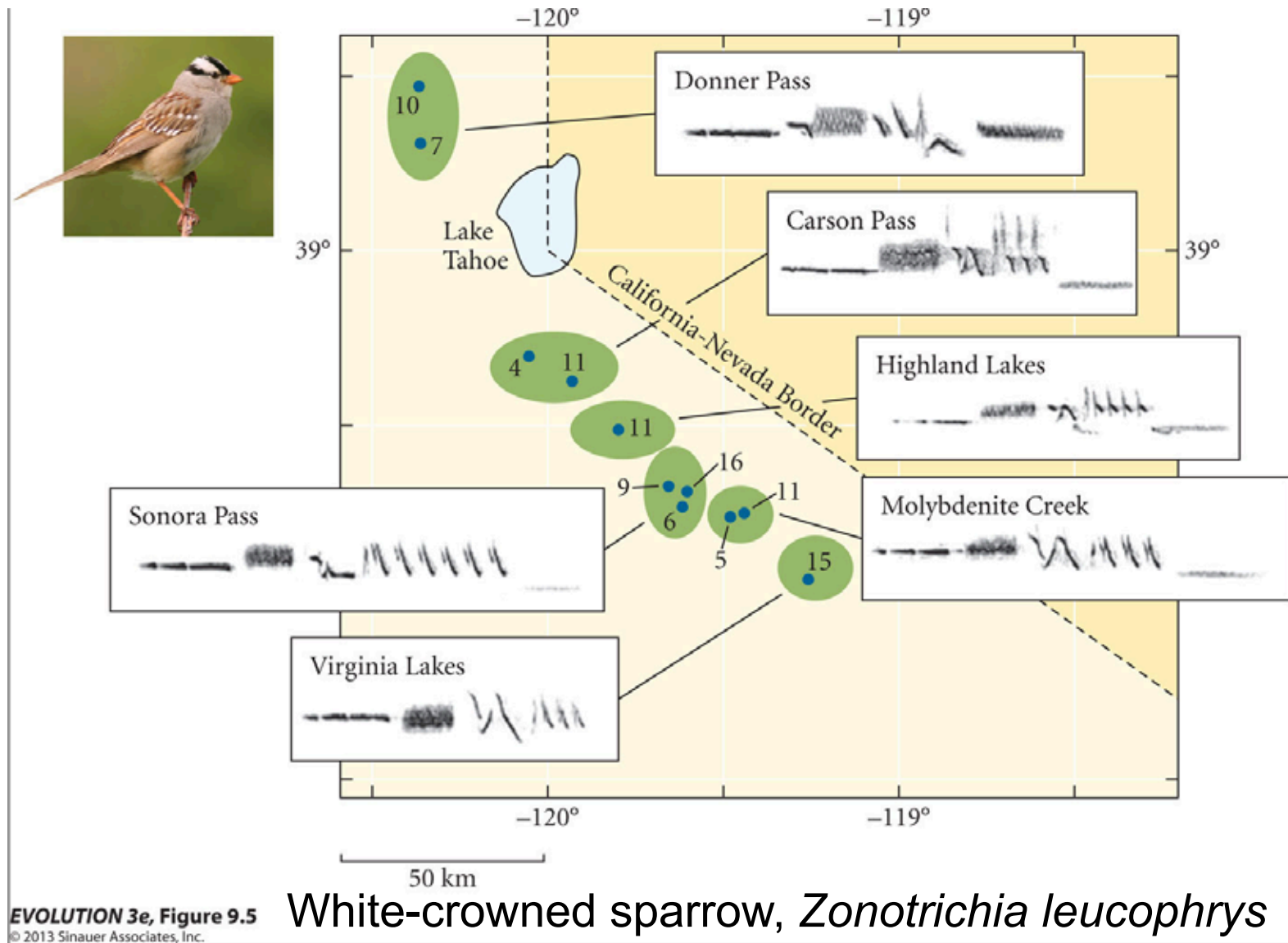- Non-genetic variation- Maternal effects
- Epigenetic inheritance

Cultural inheritance (not Mendelian)- songs are learned in birds. Dialects are common.

White-crowned sparrow, *Zonotrichia leucophrys*

EVOLUTION 3e, Figure 9.5
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In contrast songs of insects and frogs are innate – genetically programmed.

Map from www.geographx.com

2152 bp mtDNA COI+COII
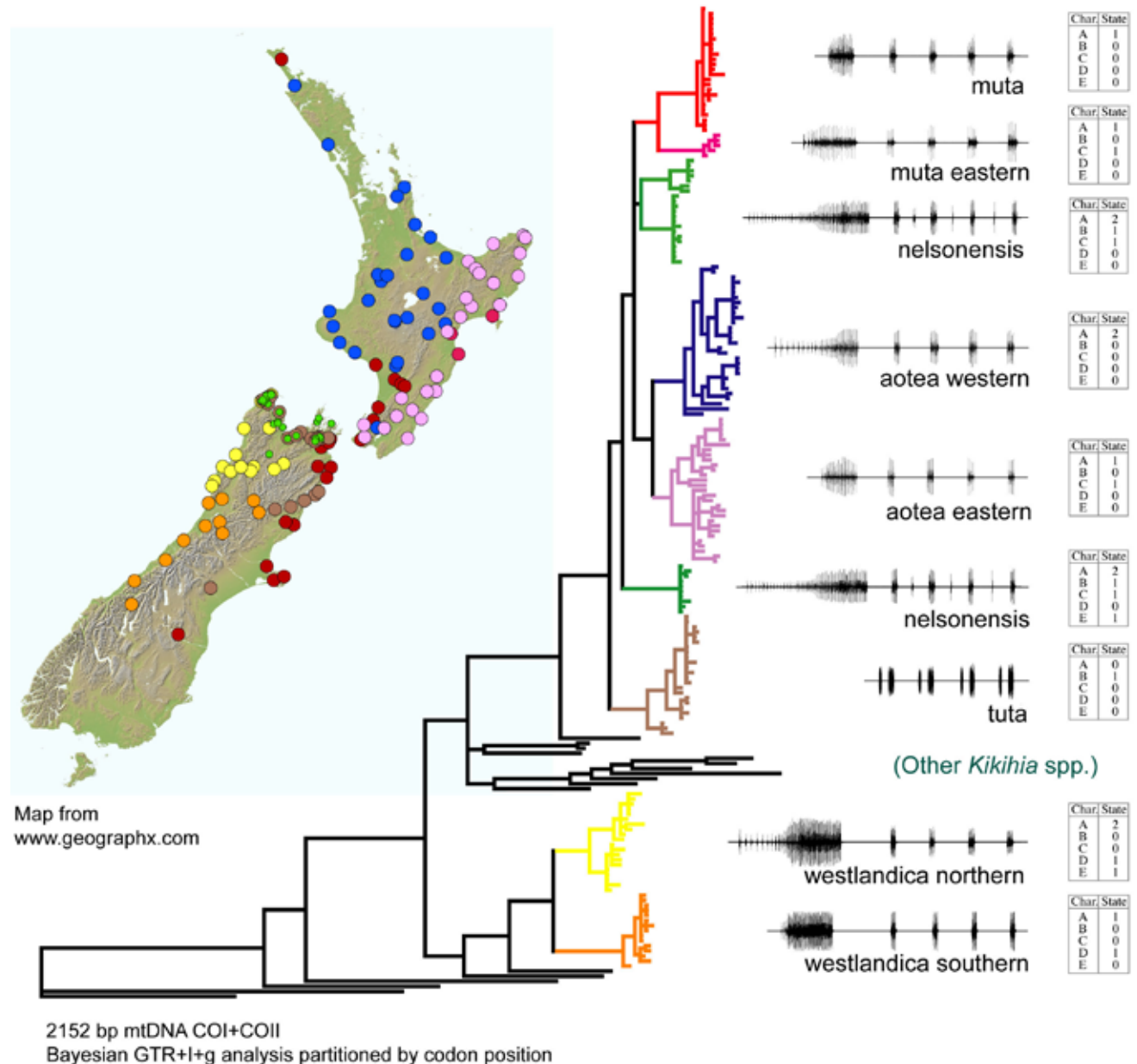Bayesian GTR+I+g analysis partitioned by codon position

Causes of deviation from Mendelian ratios of offspring allele frequencies in crosses

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Phenotypic plasticity

- The capacity of an organism, of a given genotype, to express different phenotypes under different environmental conditions.

Not all variation is heritable.

Some variation comes solely from a plastic response to environmental stimuli.

$$V_{total} = V_{genetic} + V_{environment}$$

$$V_{total} = V_{genetic} + V_{environment}$$

Drosophila

Body weight

Larval temperature

female
male
embryo
pupa
1st instar larva
prepupa
2nd instar larva
3rd instar larva

Aphids

Wing state

Winged

Wingless

Population density

c

apple aphid
(Aphis pomi)

winged adult

wingless adult

© 2013 Encyclopædia Britannica, Inc.

Plastic response, e.g., to presence of predators

- Water containing chemical cues from predators (kairomones) …

- induces changes in exoskeleton over life of *Daphnia cucullata,* water fleas

- Helmet length,

- body length,

- tail spine length
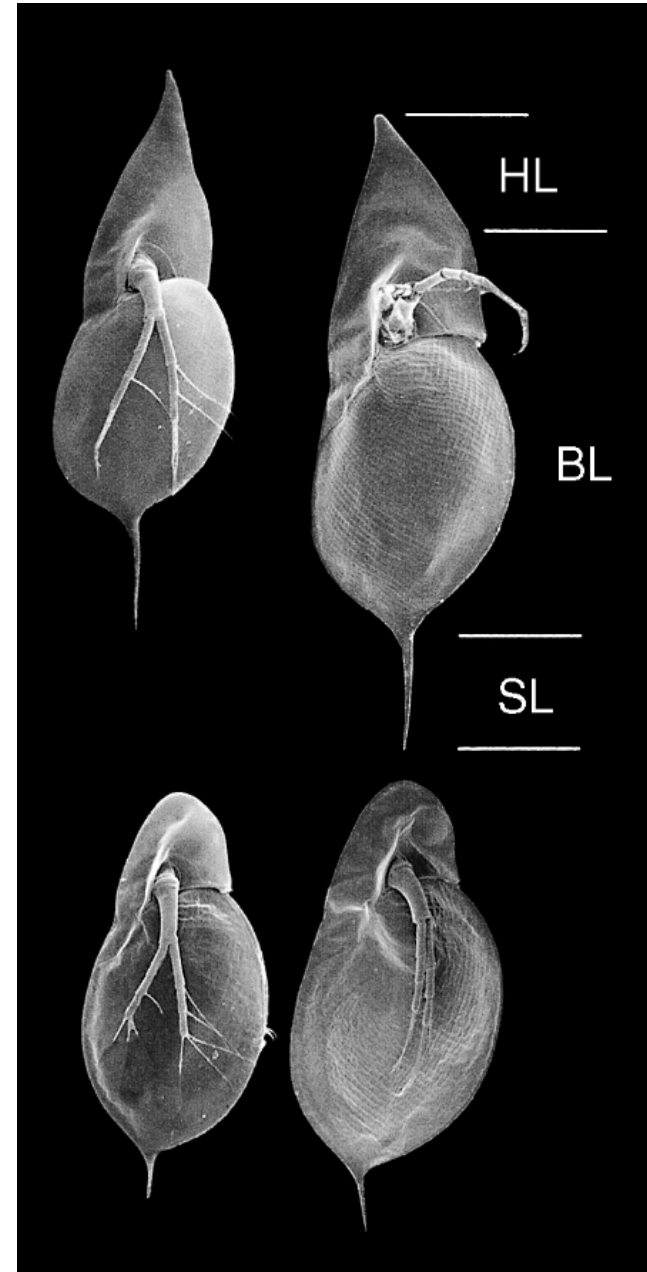
Daphnia cucullata

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- **Non-genetic variation- Maternal effects**
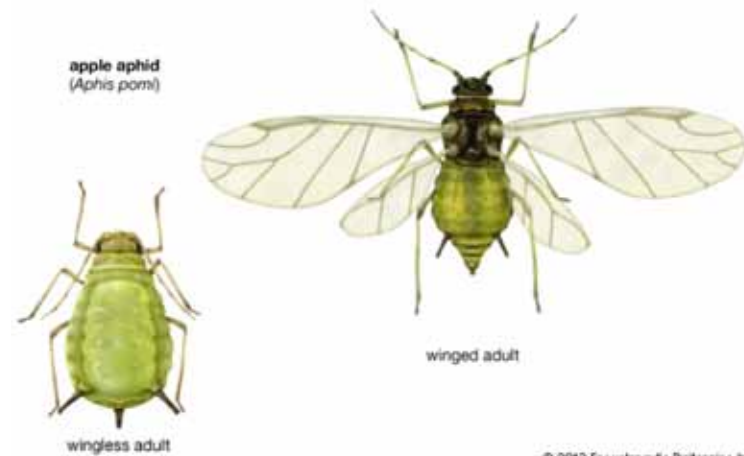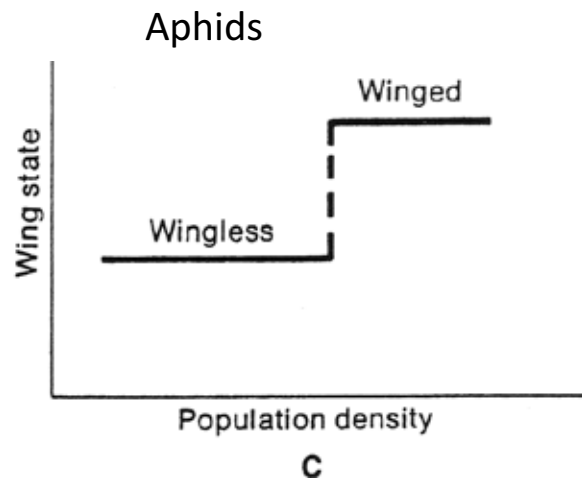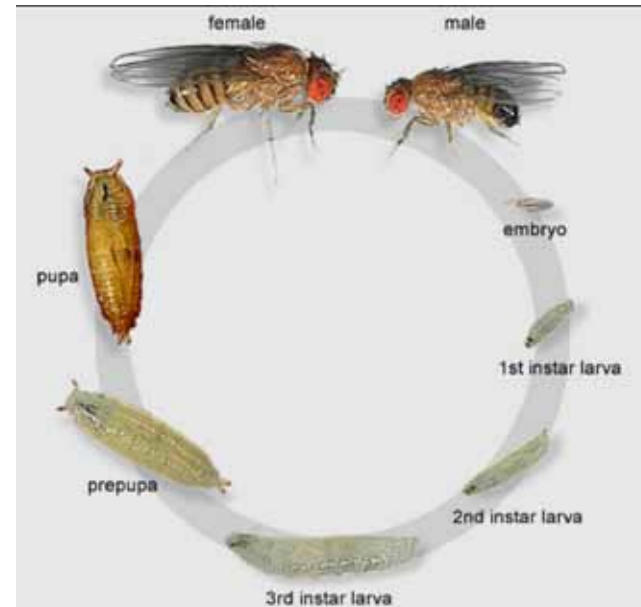- Epigenetic inheritance

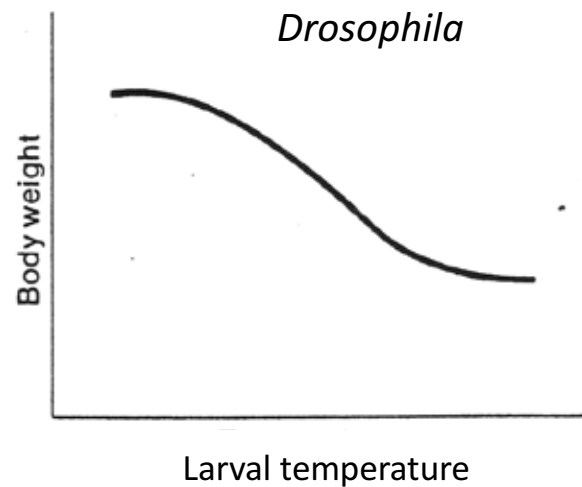# Plasticity can be amplified by Maternal Effects

EVOLUTION 3e, Figure 9.6
© 2013 Sinauer Associates, Inc.
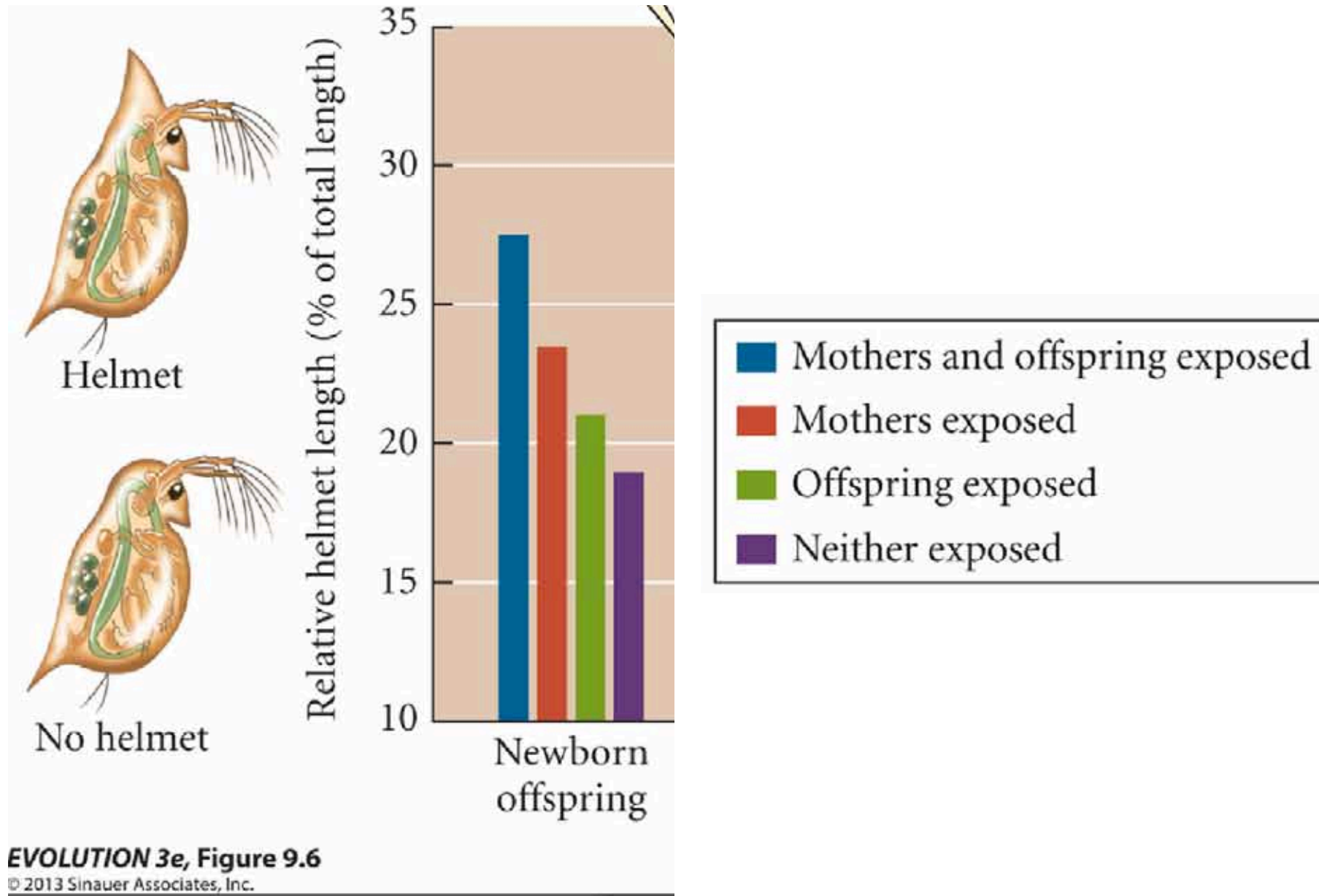
Plastic response to the environment

Spring vs Fall geometrid caterpillars, *Nemoria arizonaria*

Water-crowfoot leaves, *Ranunculus aquitilis*

(A) Catkins   Catkin-like   Caterpillars   Stick-like

Caterpillars

(B)   Submerged   Air-water interface   Aerial
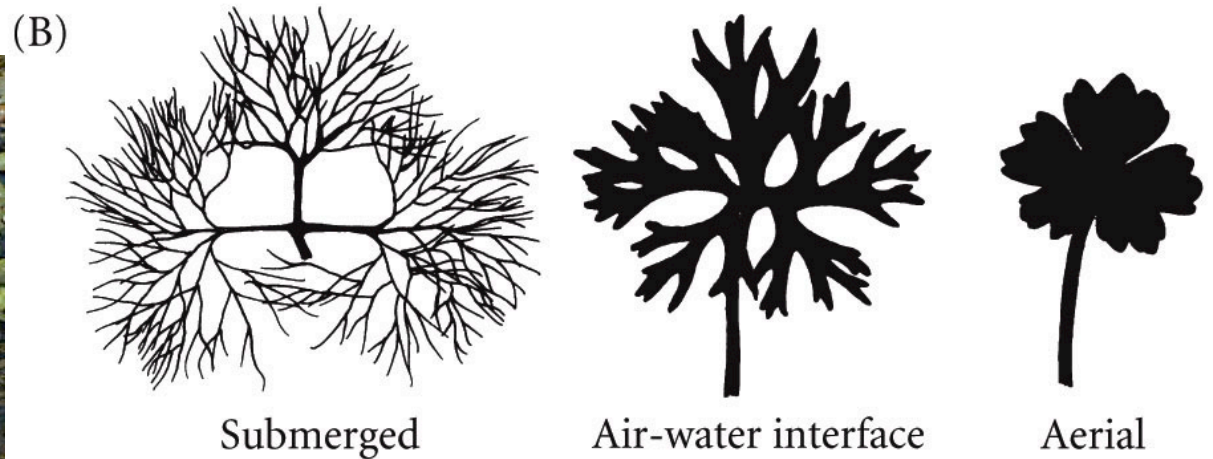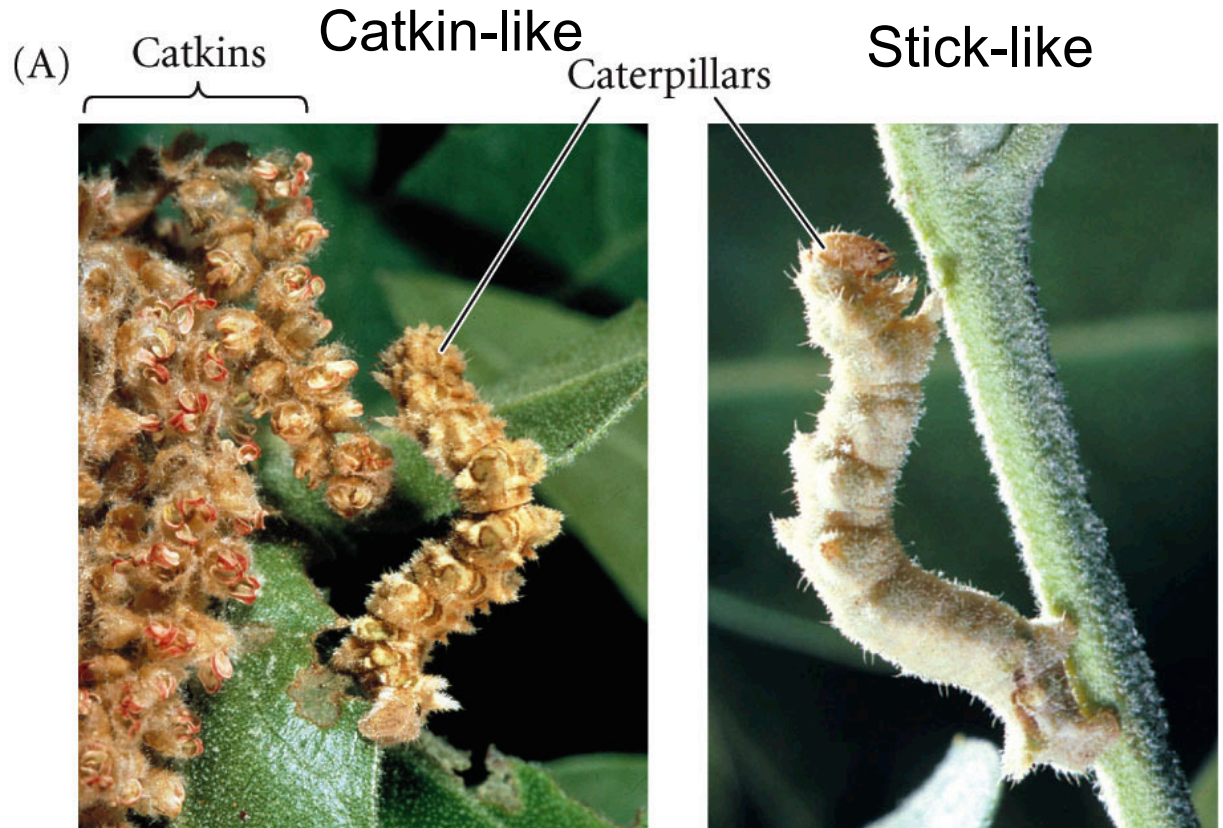
Evolution 2e Futuyma,   ©Sinauer Press.

Both genetics and plasticity can affect a phenotype simultaneously

Long vs short wing planthoppers- 3 lines

(A)

Percent long winged

Nymphal density

Selected Long

Control

Selected short

m4
c4
b4
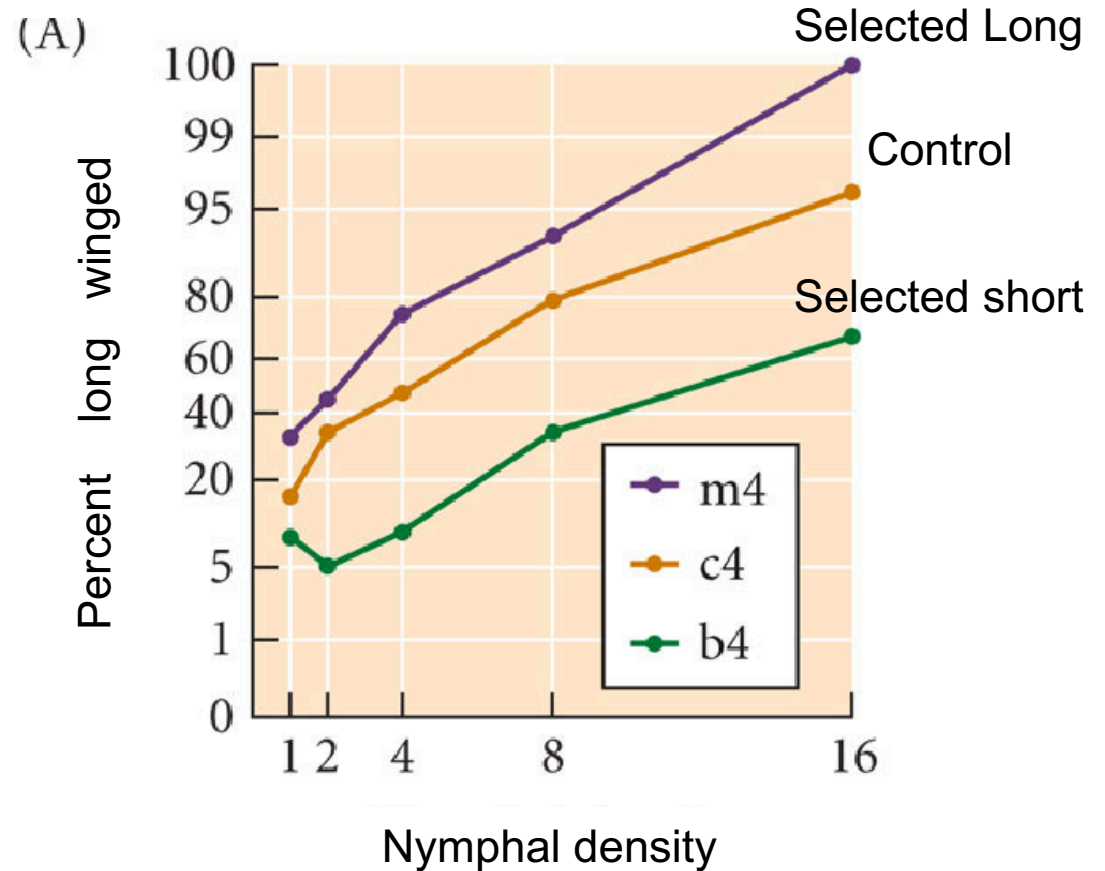
The trait "wing length".....
- can be selected in the lab
- has a genetic basis
- is variable
- responds to density environment (plastic)

c = control lines
m = lines selected for long wings
B = lines selected for short wings

Clausen, Keck, & Hiesey. 1940. Common garden experiments. Are these plastic varieties or different genetically (ecotypes)?

(A) *Potentilla glandulosa*

P.g. *typica*
Low elevation

P.g. *hanseni*
mid elevation

P.g. *nevadensis*
high elevation

Different Elevations

(B)

hanseni

nevadensis

typica

No plants survived

Height of plants (cm)

Stanford
El. 30 m

Mather
El. 1400 m

Timberline
El. 3050 m
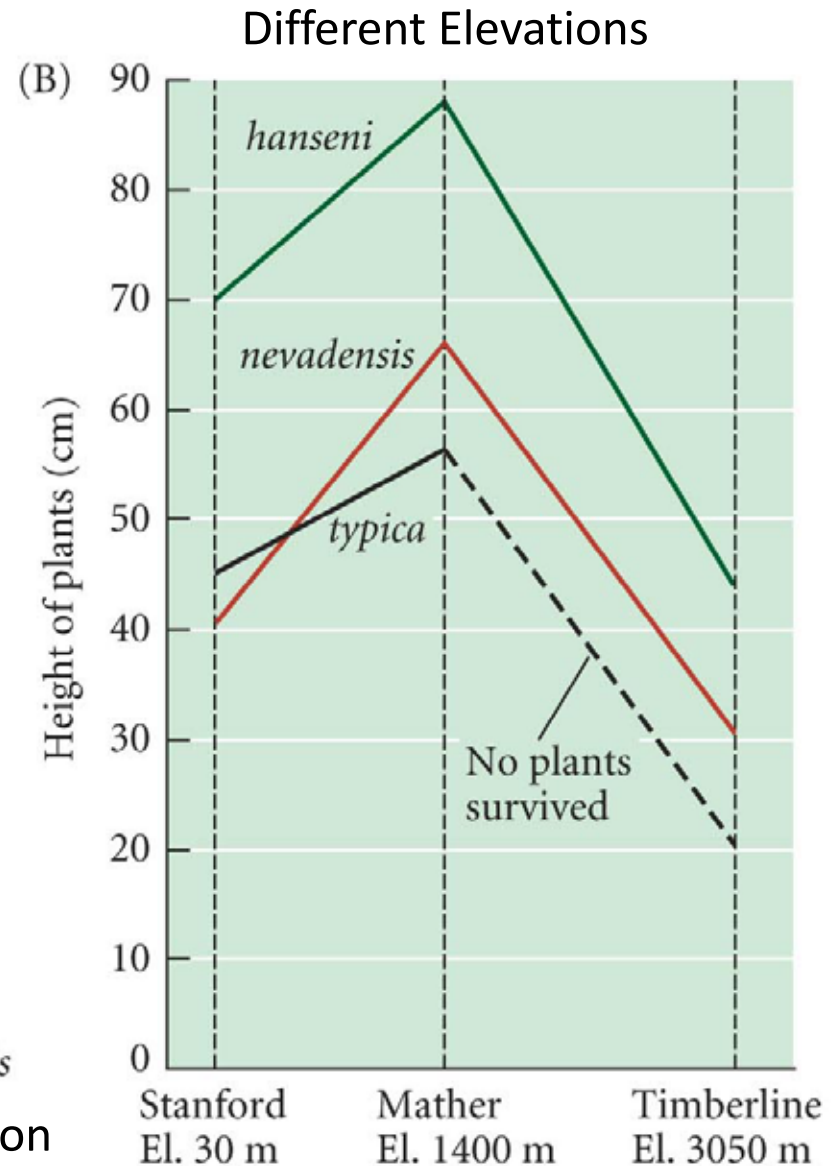
Two theories for the effect of phenotypic plasticity on evolution…

- Plasticity will allow optimal phenotypic response to a wide variety of environments and therefore shield the genotype from selection.

- Phenotypic change induced by a new environment may prove adaptive and become genetically assimilated (Mary Jane West-Eberhard).

Genetic Assimilation- Waddington 1953

- Drosophila cross-vein sometimes fails to appear if pupa given a heat shock.

- Waddington artificially selected individuals that responded to the heat shock over many generations.

- Eventually, a large proportion of the population was cross-veinless even without heat shock.

- The trait was genetically assimilated (no longer plastic).

Does plasticity become genetically assimilated?

Low temperature morph

Manduca quinquemaculata
Tomato hornworm

High temperature morph

Manduca sexta: Tobacco Hornworm
Green at all temperatures.

Definition: canalization

- Environmentally canalized- insensitive to alteration by environmental changes.

- Genetically canalized- a phenotype with a low sensitivity to the effects of mutation

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Epigenetic signals

- DNA methylation,
- Histone modifications (acetylation)
- Noncoding RNAs
- Transcription factors (TFs)

Control heritable cell memory and maintain cell identity.   Effects behavior and lifespan.

Epigenetic inheritance

- E.g., DNA methylation
  - Methyl group joined to C-G couplet during replication
  - Can prevent or lower gene expression
  - Can be influenced ay environment, aging, maternal & paternal condition.

Standard *Linaria vulgaris*

peloria form

EVOLUTION 3e, Figure 9.7
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Futuyma Example- mutant toadflax, caused ay extensive methylation of floral symmetry gene.

Methylation is lost in some lineages, e.g., Drosophila and Nematodes.

Methylation- location in gene varies across lineages

Table 1   Evolutionary overview of DNA methylation

Species	Common name	CG methylation (%)[a]	Gene body	Promoter	Transposons	Reference(s)
Homo sapiens	Human	70–80	Yes	Yes	Yes	10
Mus musculus	House mouse	74	Yes	Yes	Yes	38
Apis mellifera	Honey bee	<1	Yes	No	No	38, 88
Harpegnathos saltator	Jerdon's jumping ant	<0.2	Yes	No	No	17
Camponotus floridanus	Florida carpenter ant	<0.2	Yes	No	No	17
Nasonia vitripennis	Jewel wasp	1–2	Yes	No	No	132
Arabidopsis thaliana	Thale cress	22–24	Yes	Yes	Yes	25, 38

[a]CG methylation levels listed here in insects (*A. mellifera, H. saltator, C. floridanus, N. vitripennis*) have been measured by bisulfite conversion followed by genome-wide sequencing (BS-seq). Another approach, amplified fragment length polymorphism (AFLP), is likely to overestimate DNA methylation (85, 88, 123).

Yan et al. 2015. Ann. Rev. Entomol.

Futuyma 3e chapter 8. Reviews basic genetics.

1 Enhancer or repressor sequences bind transcription factors—proteins that stimulate or repress transcription of DNA into single-stranded messenger RNA.

2 The promotor binds the enzyme RNA polymerase, initiating transcription.

Different parts of a gene

Enhancer sequence   Promoter   5′ UTR   Exon   Intron   Exon   Intron   Exon   Poly-A addition site   3′ UTR

Eukaryotic gene (DNA)   Upstream (5′)   Downstream (3′)

Start codon   Stop codon

Transcription

Initial transcript (pre-mRNA)   5′   3′

3 Introns are excised from the pre-mRNA and discarded.

4 A chemically modified GTP molecule is attached to the start of the transcript…

5 …and a chain of adenine (A) molecules is attached to the end.

Coding segment

mRNA   5′ G-P-P-P-   — A A A··A A A 3′

6 The nucleotide sequence of the mature mRNA specifies a protein.

mRNA   5′ G-P-P-P-   — A A A··A A A 3′

7 Alternative splicing can eliminate exons, resulting in a different protein.

Epigenetic inheritance

- All ants in a colony share the same genes regardless of caste

- DNA methylation, juvenile hormone, nutrition → castes of social insects

Queens, males, workers (minors, majors, super majors)

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A knowledge of Mendelian genetics led population geneticists Hardy and Weinberg to independently come up with the same equation to predict genotype frequencies in a population.

These processes equivalent to the assumptions necessary for the HW equation to work

- Diploid population
- No Natural selection (or tight linkage w another gene under selection)
- No Migration of alleles
- No New Mutations
- Random Mating
- Only one population has been sampled.

Replace previous slide with this one.

Assumptions of Hardy Weinberg

- Diploid population
- No New Mutations
- No Migration of alleles
- Random Mating (no inbreeding)
- "**infinite" population size (no drift)**
- No Natural selection (or tight linkage w another gene under selection)
- Only one population has been sampled.

What does the H-W equation state?

Given that the assumptions of H-W are not violated then…

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- Allele frequencies will not change from one generation to the next, and

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- Genotype frequencies of the offspring can be predicted by the equation: $p^2 + 2pq + q^2 = 1$

What does the H-W equation state?

Given that the assumptions of H-W are not violated then…

- Allele frequencies will not change from one generation to the next, and

- Genotype frequencies of the offspring can be predicted by the equation: $p^2 + 2pq + q^2 = 1$

- Where p = freq of allele A

    q = freq of allele a, and

    p + q = 1

A Naïve geneticist asked...

Given the following mating (one trait)..   AA x aa → F_1

All the F_1 will be   Aa   (phenotype A)   Aa x Aa → F_2

In the F_2 recessive alleles will segregate out but will be in the minority (25%)

¼ AA, ½ Aa, ¼ aa

	A	a
A	AA	Aa
a	aA	aa

In future generations, will "a" disappear?

No, Allele frequencies will not change from one generation to the next.

Calculations (but first some terminology...)

Terminology

Number indiv. each genotype	Number of each allele		Frequency of each genotype
	A	a	
25 AA	50		.25 = D
50 Aa	50	50	.50 = H
			.25 = R
25 aa		50	

Total number of indiv. = 100

Total number of alleles = 200

Terminology

Number indiv. each genotype	Number of each allele		Frequency of each genotype
	A	a	
25 AA	50		.25 = D
50 Aa	50	50	.50 = H
25 aa		50	.25 = R

Total number of indiv. = 100	Total number of alleles = 200

Two ways to calculate allele frequencies….

p = freq of "A" allele = #A alleles / total # alleles = 100/200 = 0.50

p = (D + ½ H) = 0.25 + ½ (0.50) = 0.50

q = freq of "a" allele = # a alleles / total # alleles = 100/200 = 0.50

q = (R + ½ H) = 0.25 + ½ (0.50) = 0.50

End of Lecture 15