

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



<http://www.popartuk.com/q/1/g3734+brothers-albino-tiger-and-bengal-tiger-poster.jpg>; <http://www.hedgehogs.org/albino-hedgehog.jpg>
<http://media.ebaumsworld.com/picture/DamianRules/albino1.jpg>; <http://rebel5ive.lbbhost.com/AlbinoFawn/AlbinoSquirrel.jpg>

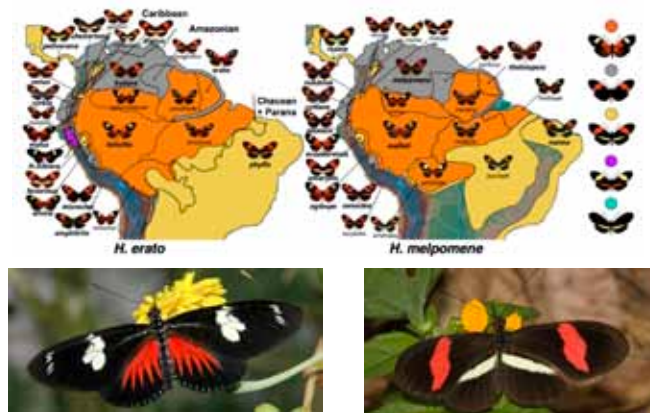
Green/Brown Color Polymorphism *Kikihia peninsularis* cicadas from NZ



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

http://resweb.llu.edu/sdunbar/students/Perumal_clip_image002_0002.jpg

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).. $AA \times aa \rightarrow F_1$
A - dominant, a - recessive

All F_1 genotype Aa (phenotype A) $Aa \times Aa \rightarrow F_2$

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

$\frac{1}{4} AA, \frac{1}{2} Aa, \frac{1}{4} aa$

	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.

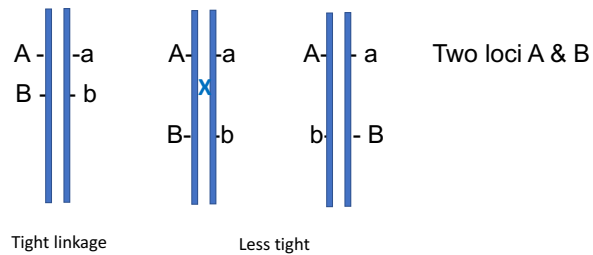
•**Segregation**- paired alleles segregate at random into gametes

•**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.



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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

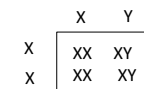
Causes of deviation from Mendelian ratios of offspring allele frequencies in crosses

- 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

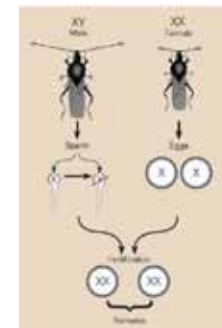
Violation of Mendel's laws

- **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



Distorted genotypes



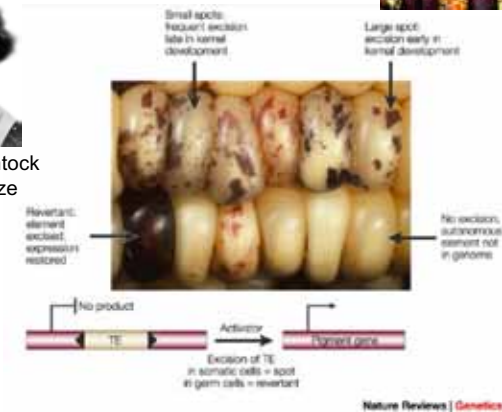
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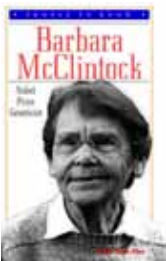
Transposable elements



Barbara McClintock
1983 Nobel Prize



Barbara McClintock (1902-1992)

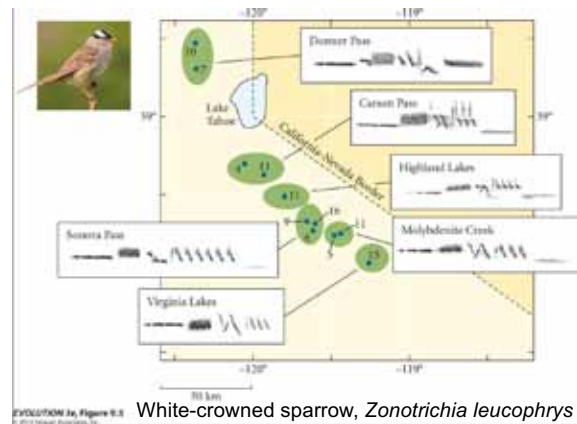


- PhD, Cornell, 1927- maize genetics
- First genetic map for maize
- Role of telomere and centromere
- Cornell –Missouri (1936) -- Cold Spring Harbor (1941)
- 1940's-50's discovered transposition in maize
- National Academy of Sciences 1944
- 1953 –Due to harsh skepticism--- stopped publishing on transposition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

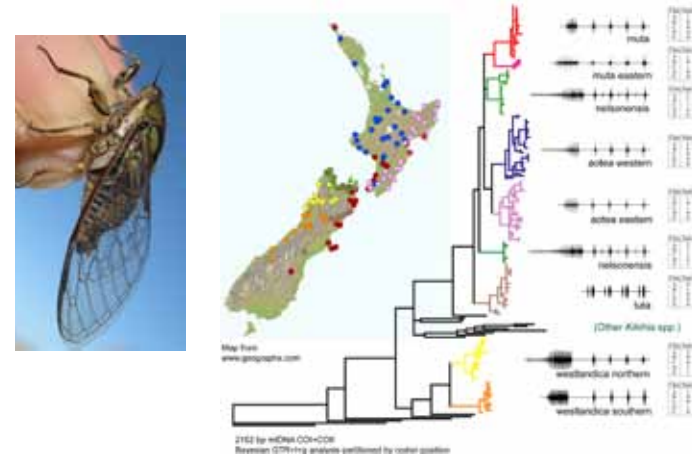
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Cultural inheritance (not Mendelian)- songs are learned in birds. Dialects are common.



In contrast songs of insects and frogs are innate – genetically programmed.



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Phenotypic plasticity

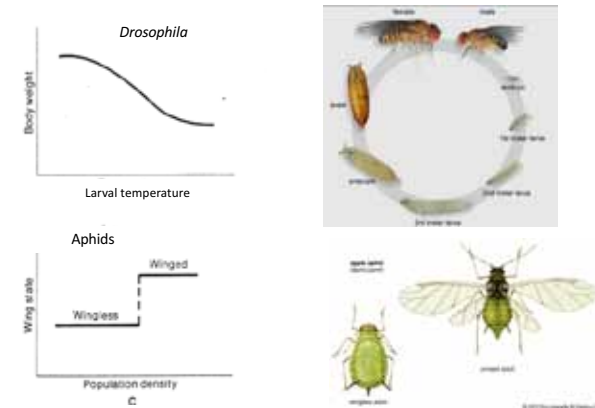
- The capacity of an organism, of a given genotype, to express different phenotypes under different environmental conditions.

Not all variation is heritable.

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

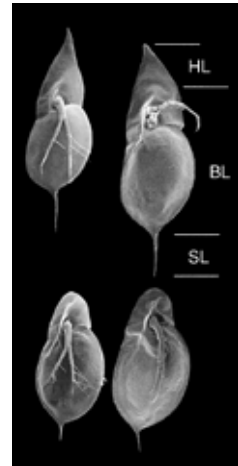
$$V_{\text{total}} = V_{\text{genetic}} + V_{\text{environment}}$$

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

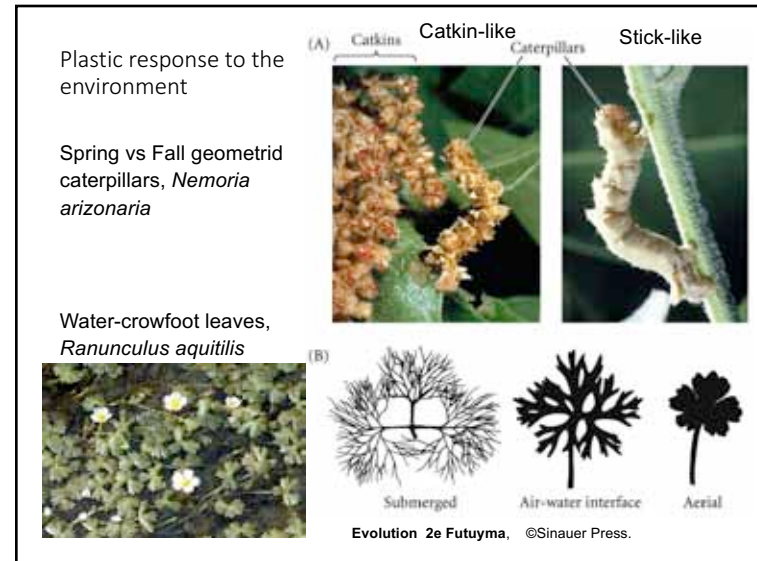
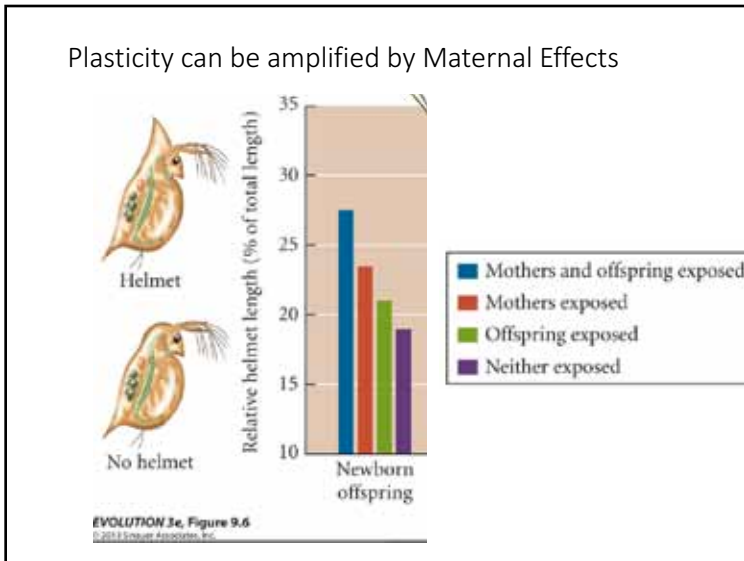
- Water containing chemical cues from predators (kairomones) ...
- induces changes in exoskeleton over life of *Daphnia cucullata*, water fleas
- Helmet length,
- body length,
- tail spine length



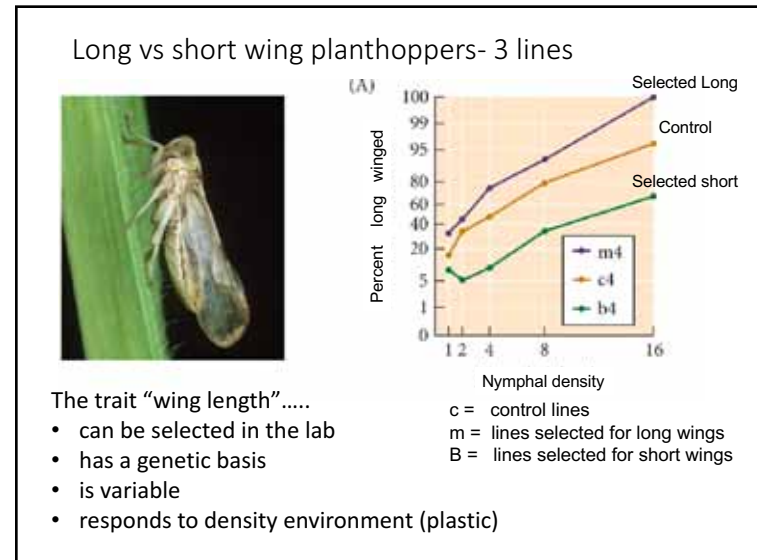
Daphnia cucullata

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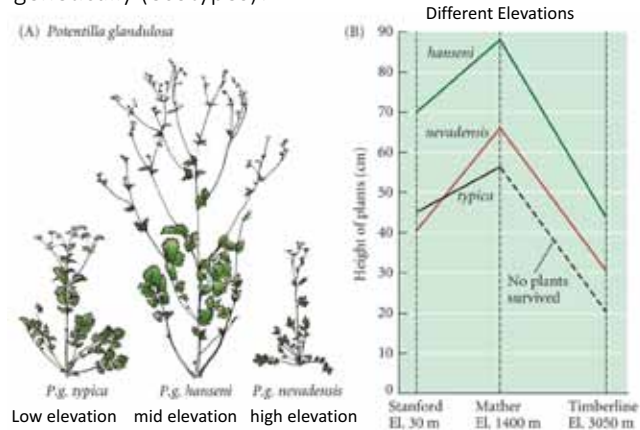
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Both genetics and plasticity can affect a phenotype simultaneously



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



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



Manduca sexta: Tobacco Hornworm
Green at all temperatures.



High temperature morph

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.

Yan et al. 2015. ann. Rev. Entomol.

Epigenetic inheritance

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



Futuyma Example- mutant toadflax, caused by extensive methylation of floral symmetry gene.

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

Methylation- location in gene varies across lineages

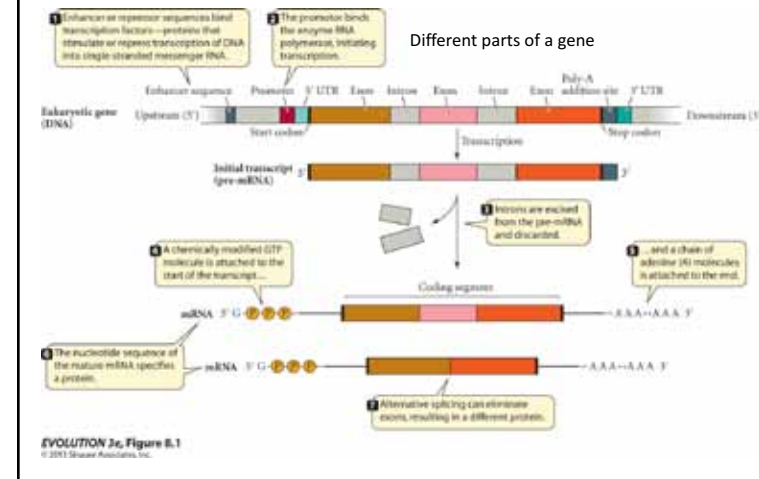
Table 1 Evolutionary overview of DNA methylation

Species	Common name	CG methylation (%) ^a	Methylation status			Reference(s)
			Gene body	Promoter	Transposons	
<i>Homo sapiens</i>	Human	70-80	Yes	Yes	Yes	10
<i>Mus musculus</i>	House mouse	74	Yes	Yes	Yes	38
<i>Apis mellifera</i>	Honey bee	<1	Yes	No	No	88, 88
<i>Harpegnathus saltator</i>	Jardón's jumping ant	<0.2	Yes	No	No	17
<i>Camponotus floridanus</i>	Florida carpenter ant	<0.2	Yes	No	No	17
<i>Nasutia cornutus</i>	Jewel wasp	1-2	Yes	No	No	132
<i>Arabidopsis thaliana</i>	Thale cress	22-24	Yes	Yes	Yes	25, 33

^aCG methylation levels listed here in insects (*A. mellifera*, *H. saltator*, *C. floridanus*, *N. cornutus*) 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 (37, 88, 132).

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

Futuyma 3e chapter 8. Reviews basic genetics.



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.

What does the H-W equation state?

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

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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).. $AA \times aa \rightarrow F_1$

All the F_1 will be Aa (phenotype A) $Aa \times Aa \rightarrow F_2$

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

$\frac{1}{4} AA, \frac{1}{2} Aa, \frac{1}{4} aa$

	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 aa		50	.25 = R
Total number of indiv. = 100	Total number of alleles = 200		

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Two ways to calculate allele frequencies....

$$p = \text{freq of "A" allele} = \#A \text{ alleles} / \text{total \# alleles} = 100/200 = 0.50$$

$$p = (D + \frac{1}{2} H) = 0.25 + \frac{1}{2} (0.50) = 0.50$$

$$q = \text{freq of "a" allele} = \# a \text{ alleles} / \text{total \# alleles} = 100/200 = 0.50$$

$$q = (R + \frac{1}{2} H) = 0.25 + \frac{1}{2} (0.50) = 0.50$$