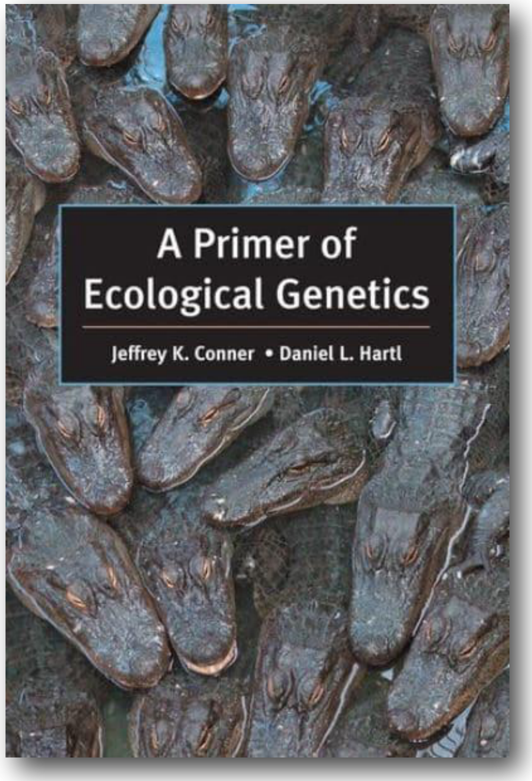
Evolutionary Genetics

LV 25600-01 | Lecture with exercises | 4KP

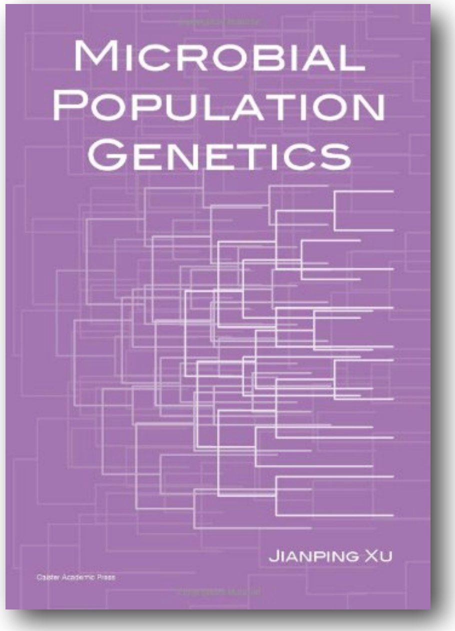
PopGen ▷ Introduction

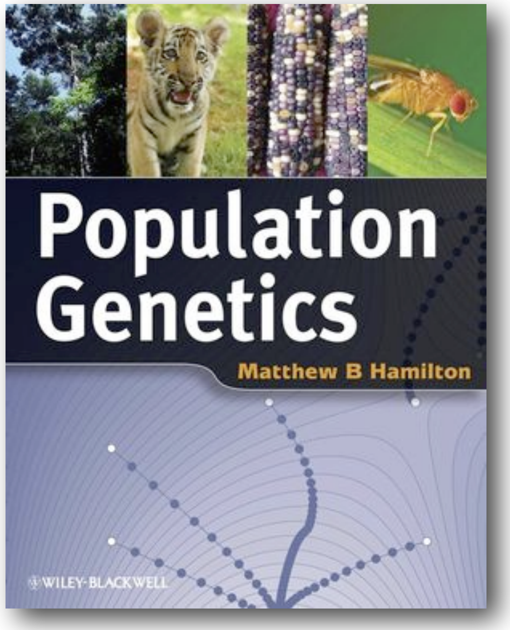

A Primer of Ecological Genetics
Jeffrey K. Conner • Daniel L. Hartl

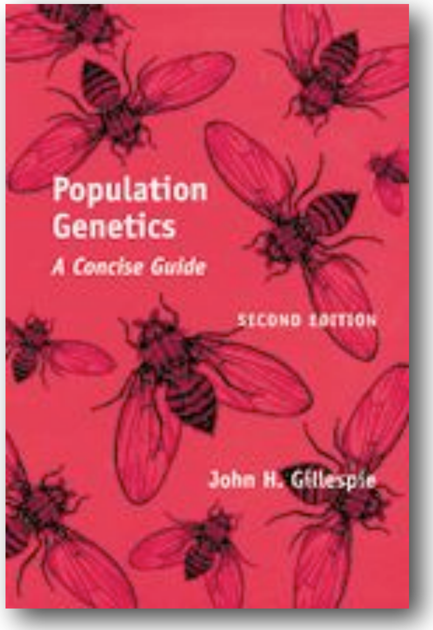
A Primer of Ecological Genetics

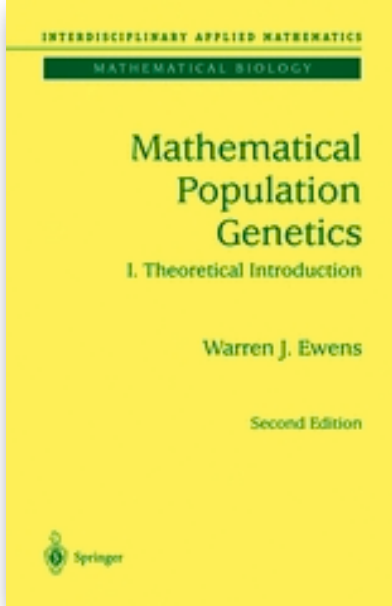
This book covers **basic concepts in population and quantitative genetics**, including measuring the selection of phenotypic traits. The emphasis is on material applicable to field studies of evolution, focusing on ecologically important traits. Topics addressed are critical for training students in ecology, evolution, conservation biology, agriculture, forestry and wildlife management.

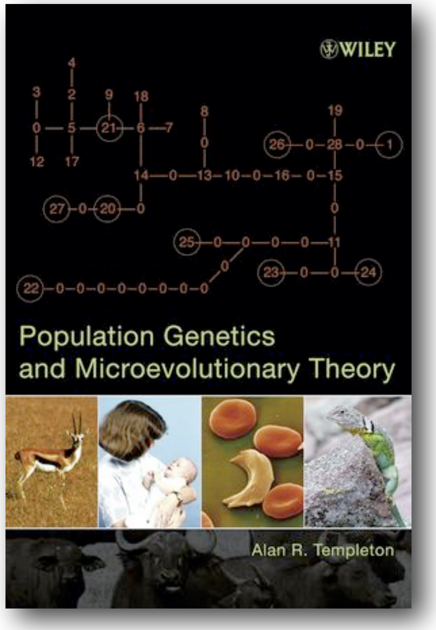
Many texts in this field are too complex and mathematical to allow the average student to grasp the key concepts. A Primer of Ecological Genetics, in contrast, employs mathematics and statistics-fully explained, but at a less advanced level-as tools to improve understanding of biological principles. Integration of the different chapters of the book shows students how diverse concepts relate to each other.


MICROBIAL POPULATION GENETICS
JIANPING XU

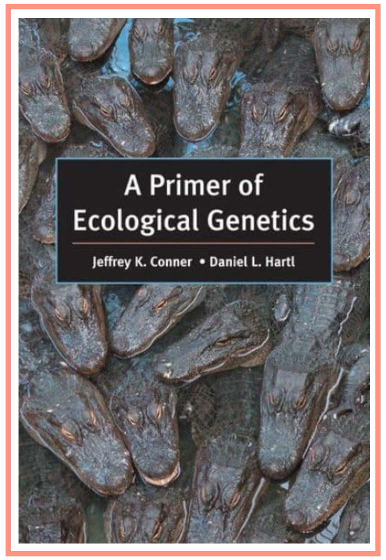

Population Genetics
Matthew B Hamilton
WILEY-BLACKWELL

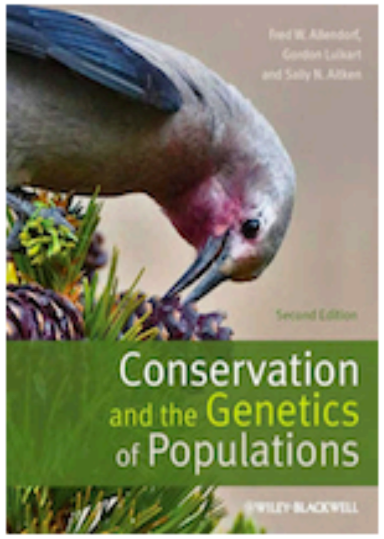

Population Genetics
A Concise Guide
SECOND EDITION
John H. Gillespie

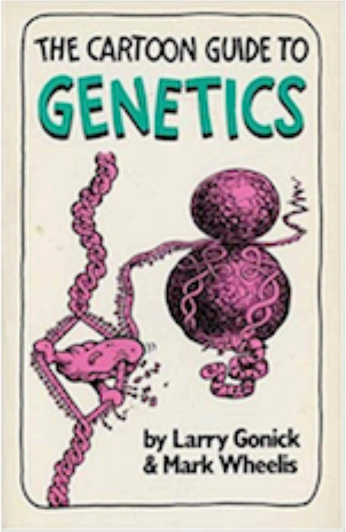

INTERDISCIPLINARY APPLIED MATHEMATICS
MATHEMATICAL BIOLOGY
Mathematical Population Genetics
I. Theoretical Introduction
Warren J. Ewens
Second Edition
Springer


WILEY
Population Genetics and Microevolutionary Theory
Alan R. Templeton

PopGen ▷ Introduction







https://www.gdc-docs.ethz.ch/UniBS/HS2022/PopGen/site/Help/

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PopGen ▷ Introduction

Population genetics is the study of the **genetic constitution (structure) of populations**, **the frequencies of alleles and genotypes** and how this constitution **changes with time** under the influence of different **evolutionary processes** (e.g. **genetic drift, natural selection, migration, mutation, assortative mating**).

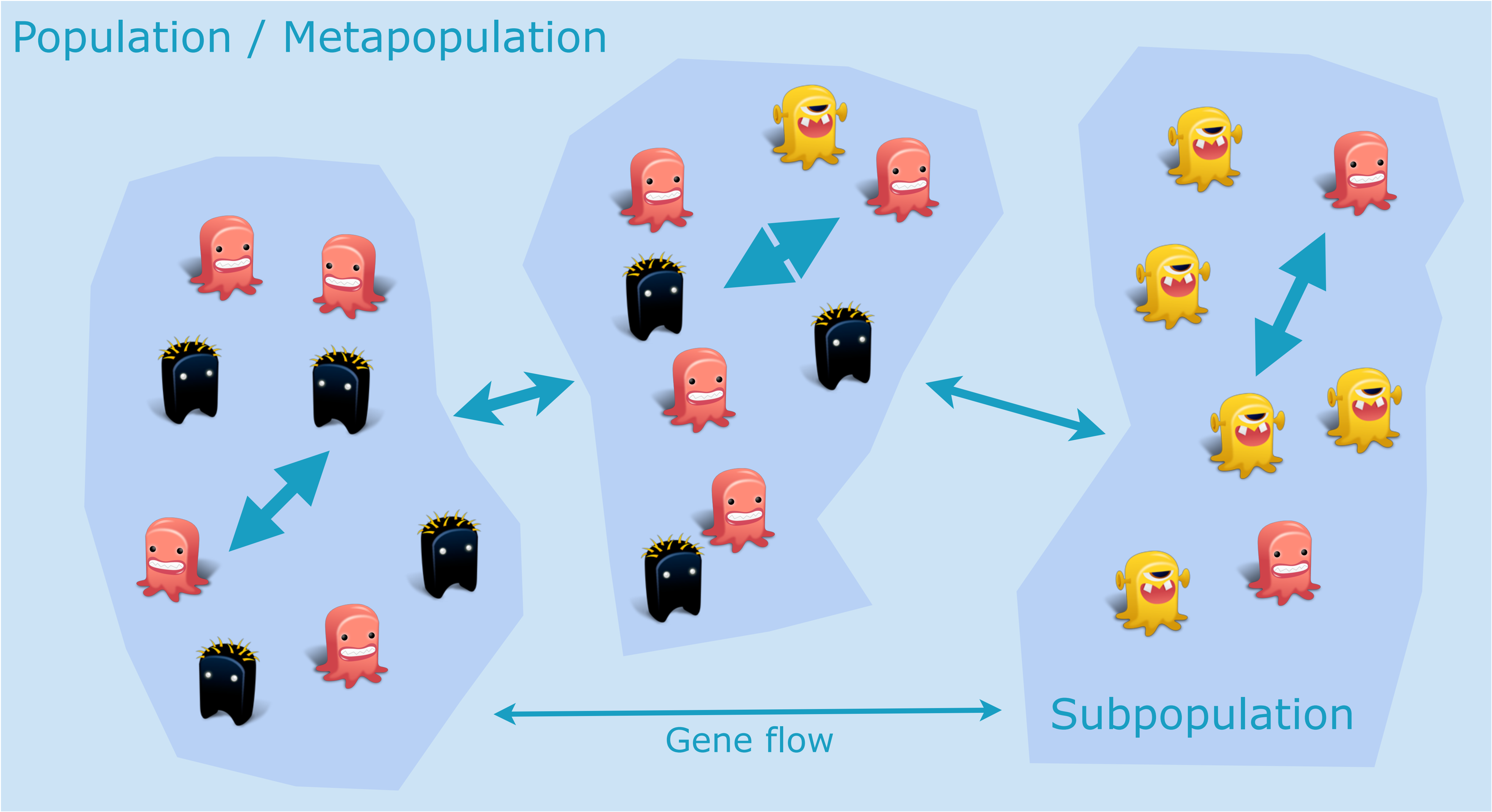
A **population** is a set of organisms in which any pair of members can breed together. This implies that all members belong to the **same species** and live near each other. Population genetics also takes into account the factors of **population subdivision and population structure**. It attempts to explain such phenomena as **adaptation** and **speciation**.

The primary goals of population genetics are to **understand the factors determining evolutionary change and stasis** and the amount and pattern of **genetic variation within and between populations**.

Hedrick, 2011

PopGen ▷ Introduction

> same species but different phenotypes



Species are divided into populations. A group of populations connected by some level of **gene flow** is often referred to metapopulation. The units within a metapopulation are called subpopulations.

PopGen ▷ Introduction

In the early part of the 20th century, **Gregor Mendel**'s experimental work on pea plants was commonly perceived to be at odds with **Darwinian natural selection**; the former, it was argued, was evidence for **discontinuous evolution** (involving large changes from parent to offspring), whereas the latter required **continuous evolution** (involving small gradual changes from parent to offspring). However, no later than 1932, the field of **theoretical population genetics** emerged as a reconciliation between Mendelism and Darwinism. Most biologists at the time accepted the fact of evolution, or Darwin's "descent with modification," but Darwin's idea of natural selection as a cause of that modification was controversial. Indeed, lacking was any generally accepted account of **genetic variation in populations** (is the variation continuous or discontinuous?), **evolutionary change** (is change gradual or saltational?), and an understanding of the appropriate use of statistical methods for studying these. The emergence of theoretical population genetics, which addressed all of these issues, is typically associated with the work of **R. A. Fisher, Sewall Wright, and J. B. S. Haldane.**

Millstein and Skipper, 2007

The foundational works that ushered in theoretical population genetics are:

Fisher (1930) The Genetical Theory of Natural Selection.

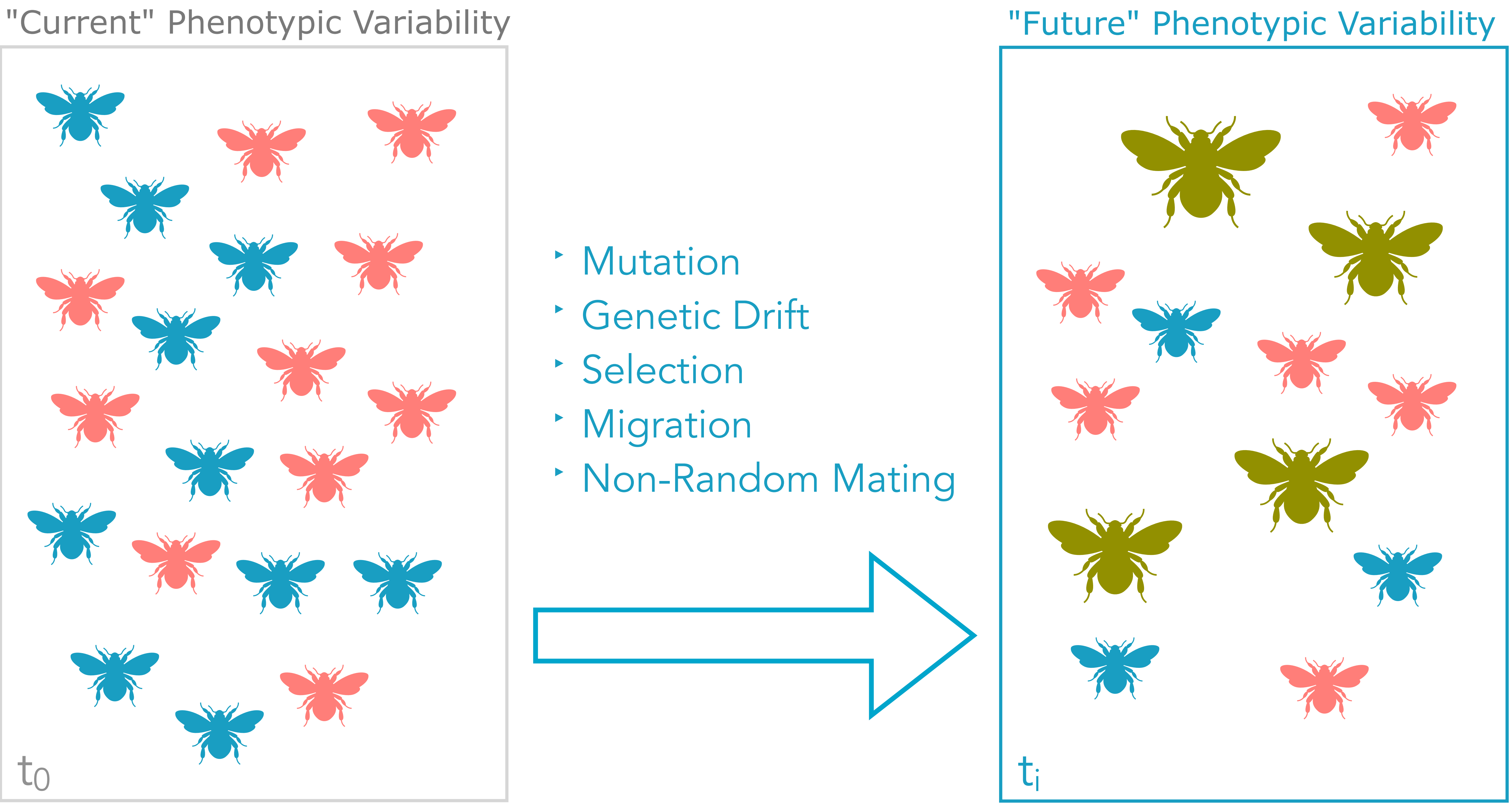
Wright (1931) Evolution in Mendelian Populations.

Haldane (1932) The Causes of Evolution.

PopGen ▷ Introduction

Population genetics is the study of **genetic variation within populations** and involves the **examination and modelling of changes in the frequencies of genes and alleles in populations over space and time.** Many of the genes found within a population will be polymorphic, they will occur in several different forms (or alleles). **Mathematical models are used to investigate and predict the occurrence of specific alleles or combinations of alleles in populations**, based on developments in the molecular understanding of genetics, Mendel’s laws of inheritance and modern evolutionary theory. **The focus is the population or the species - not the individual.**

PopGen ▷ Natural Variation



"Current" Phenotypic Variability

- Mutation
- Genetic Drift
- Selection
- Migration
- Non-Random Mating

"Future" Phenotypic Variability

PopGen ▷ **Natural Variation**

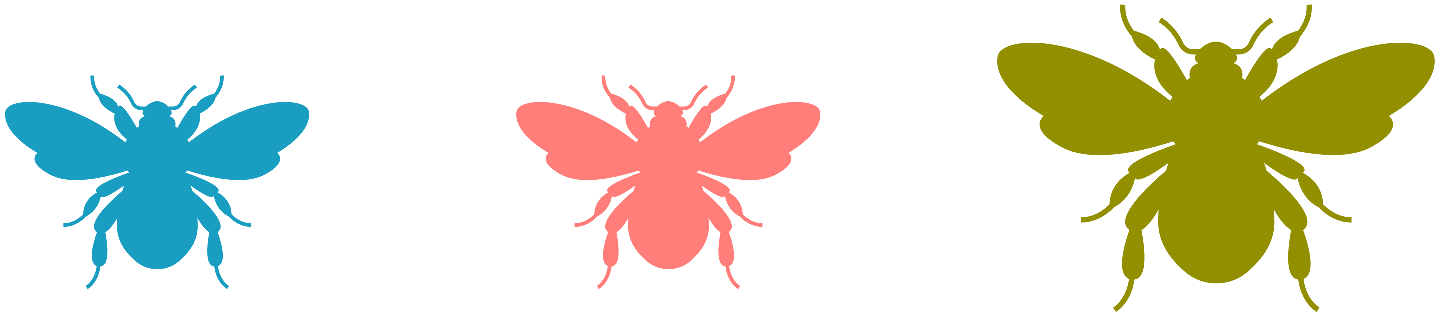
Number of Alleles

Genetic Diversity

- ⇅ Mutation
- ⇅ Genetic Drift
- ⇅ Selection
- ⇅ Migration
- ⇅ Non-Random Mating

PopGen ▷ Natural Variation

Phenotypic variability



PopGen ▷ Natural Variation

Phenotypic variability for a trait within a populations:

$$V_P = V_G + V_E$$

$$V_P = V_G + V_E + (V_G \times V_E)$$

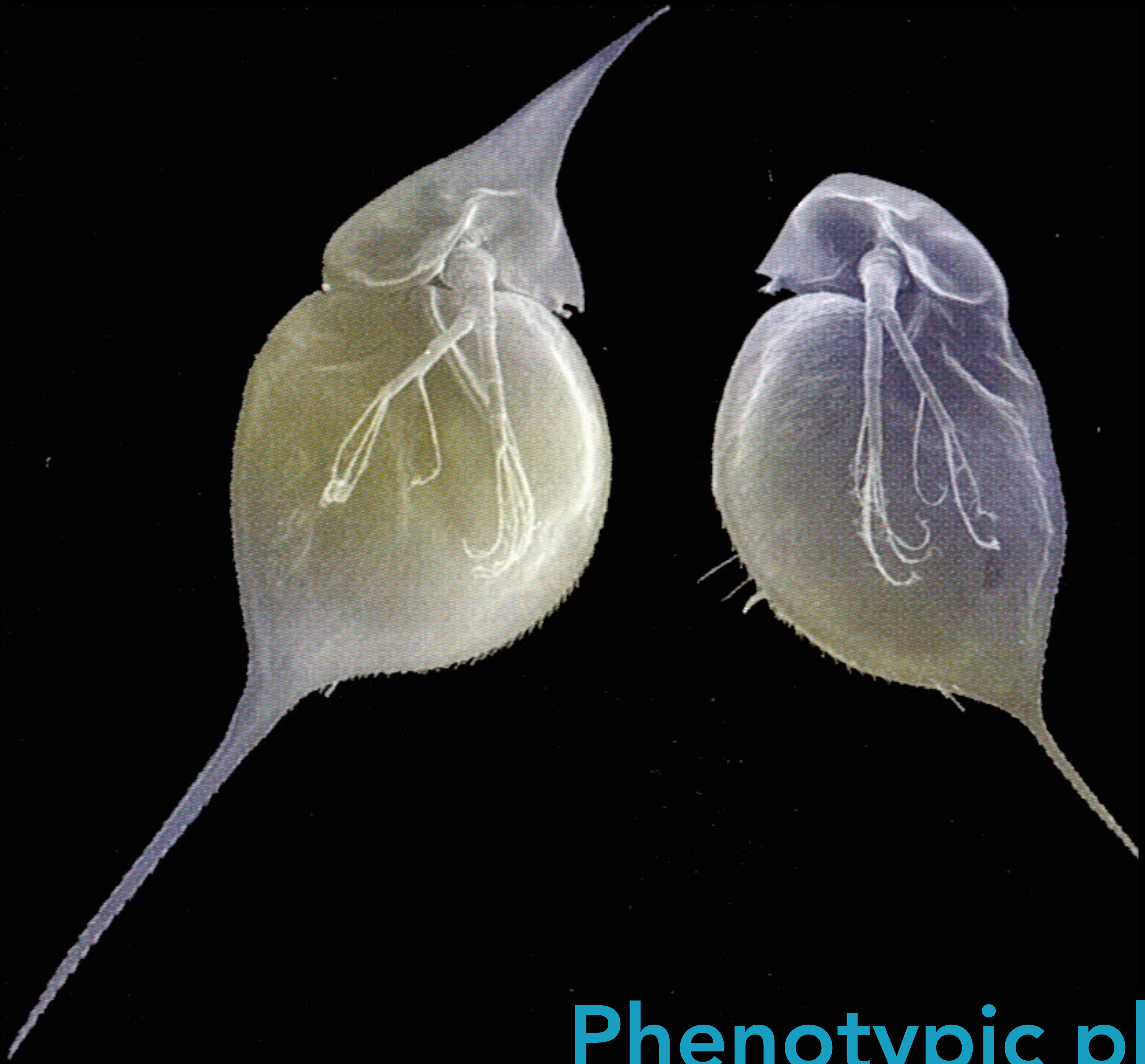
The heritability of a trait: $\dfrac{V_G}{V_E}$

V_G: genotypic variation

V_E: environmental variation

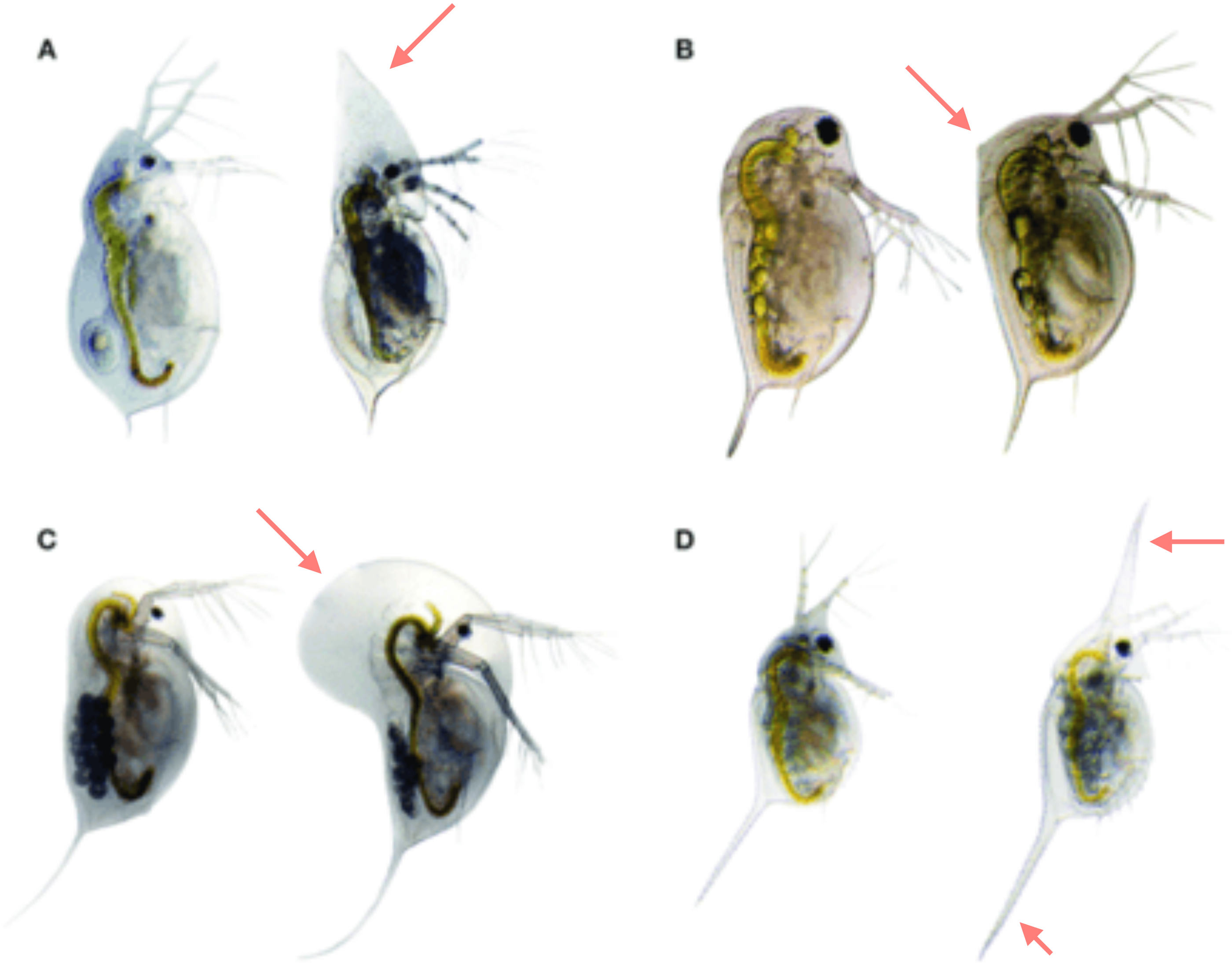
PopGen ▷ Natural Variation

Phenotypic differences between populations observed in the wild are **not a reliable indicator of genetic differences** between populations without additional information! In some cases, all of the phenotypic differences between populations may result from e.g. environmental conditions. → **Non-Genetic Inheritance**!



Phenotypic plasticity

PopGen ▷ Introduction

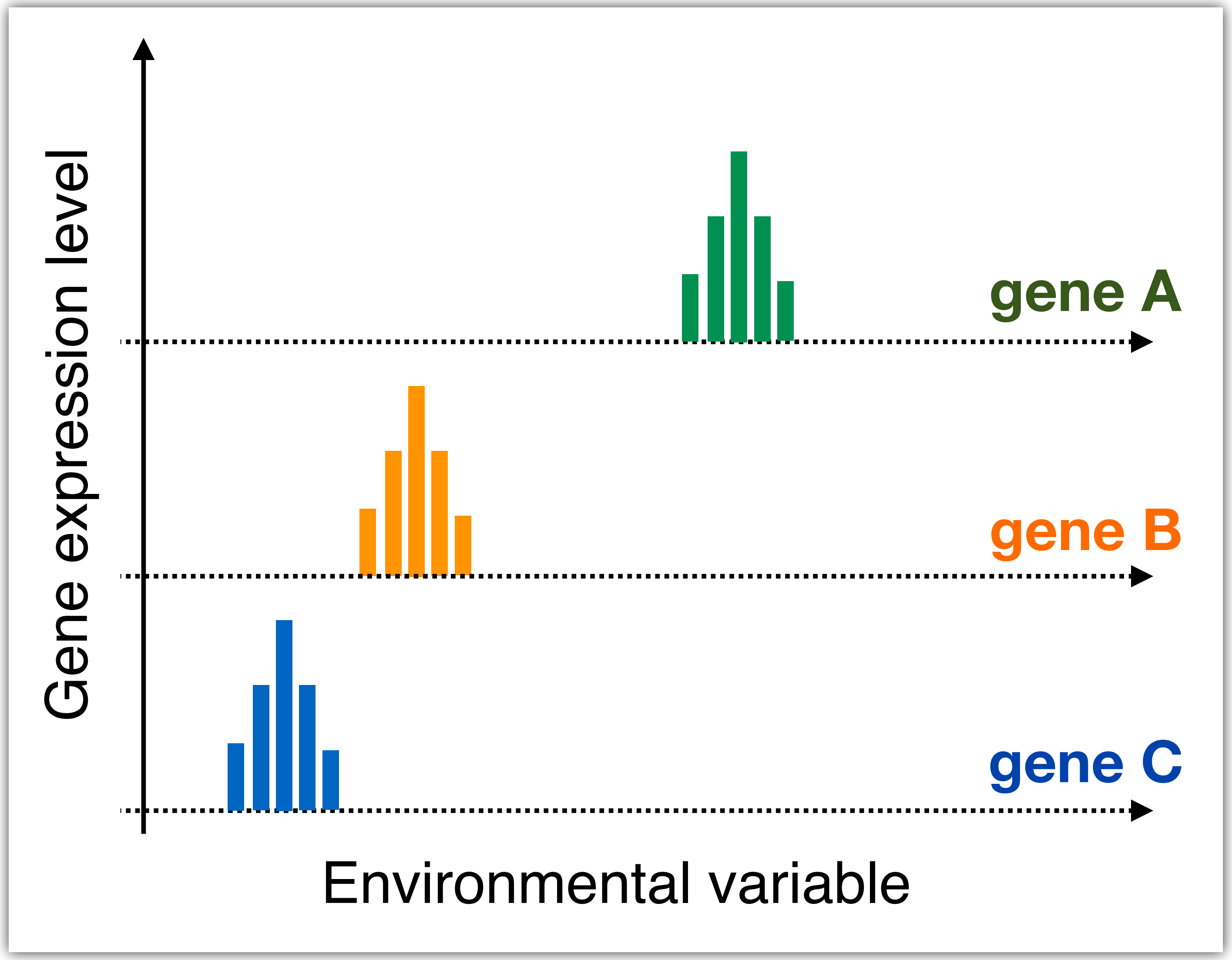


Inducible morphological defences are manifold in the genus Daphnia. The listed examples show helmet expression in *D. cucullata* (A); neckteeth expression in *D. pulex* (B); crest expression in *D. longicephala* (C); head- and tail-spine formation in *D. lumholtzi* (D). Undefended morphotypes are displayed on the left side, and the defended morphotype on the right side.

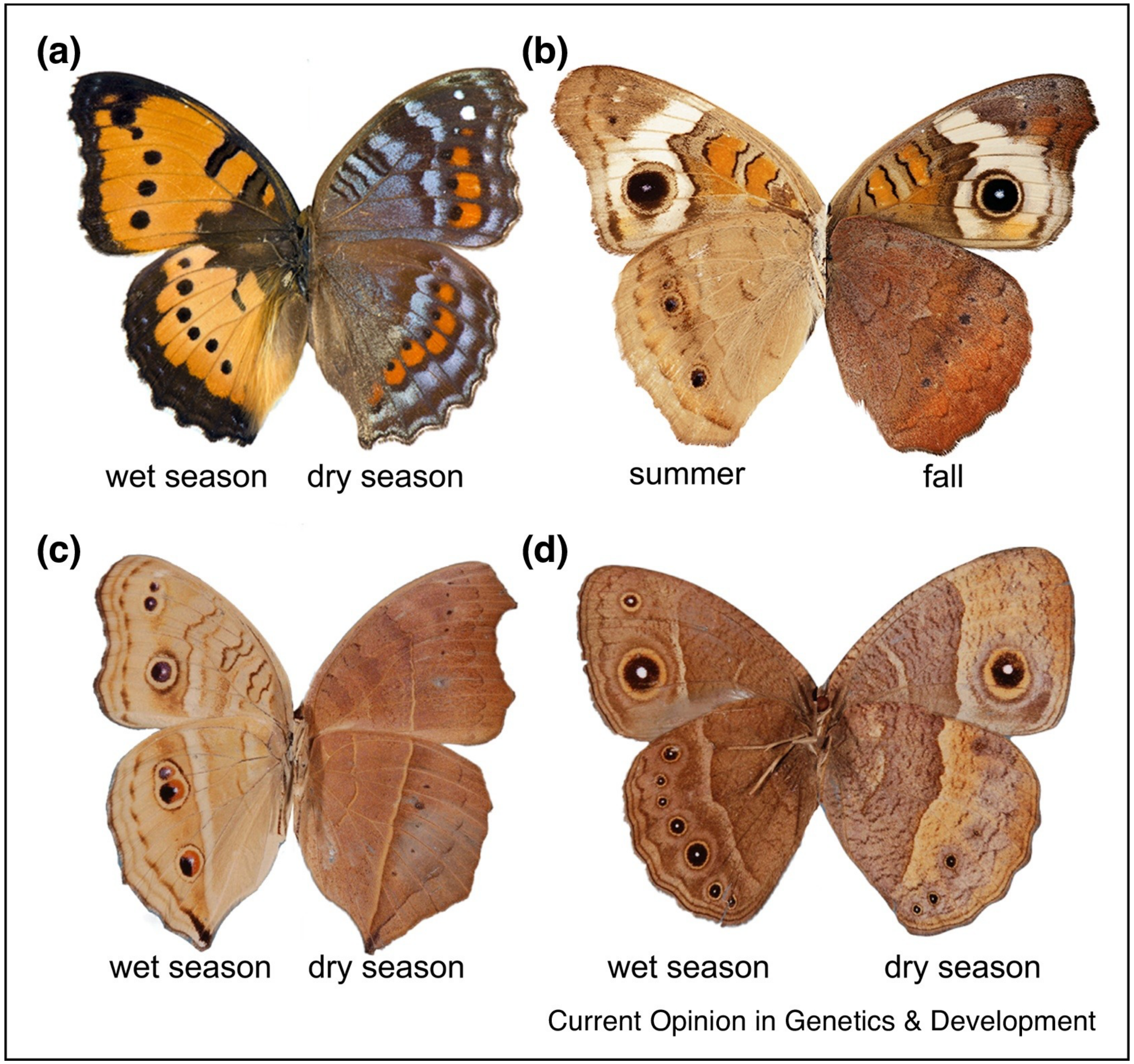
Source: Weiss (2019). Sensory Ecology of Predator-Induced Phenotypic Plasticity. Frontiers in Behavioral Neuroscience.

Introduction

Phenotypic plasticity is the ability of an organism to change its phenotype in response to changes in the environment. A condition in which the **same genotype** produces **different phenotypes** in **different environments**.



Seasonal plasticity



Source: van der Burg & Reed (2021) Seasonal plasticity: how do butterfly wing pattern traits evolve environmental responsiveness? Current Opinion in Genetics & Development. 69: 82-87.

Butterfly wing phenotypic plasticity comes in many forms; wing pattern, wing color, wing shape and eyespot size can all change. (a) *Precis octavia*, (b) *Junonia coenia*, (c) *Junonia almana*, (d) *Junonia coenia.*

Highlights:

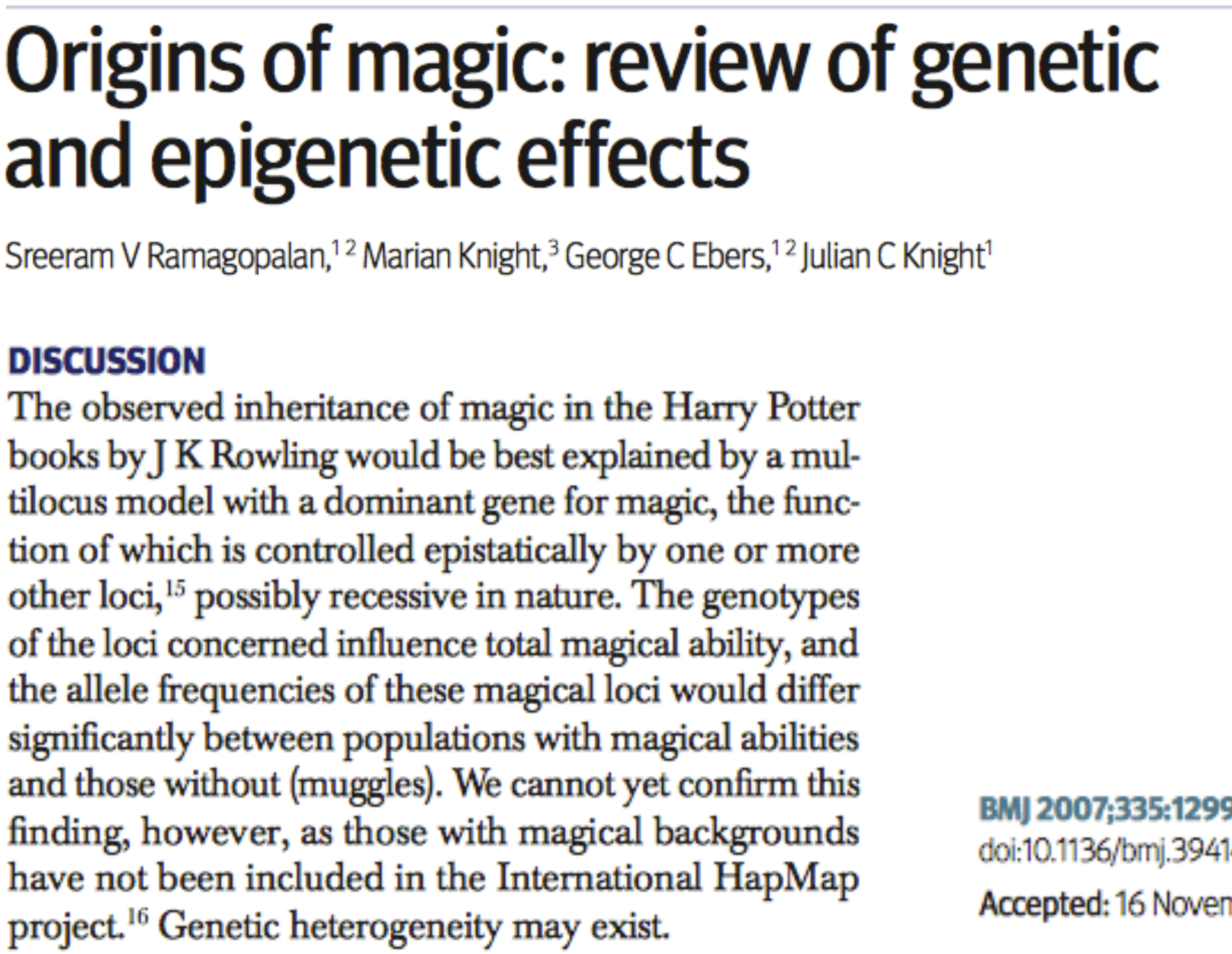
Phenotypic plasticity in butterfly wings is widespread, and different wing pattern traits evolve plastic responses independently.

Seasonal plasticity in butterflies is mediated by the hormone ecdysone.

Ecdysone signalling has been shown to affect the chromatin landscape in other insect species.

PopGen ▷ Introduction

Epigenetics is the study of phenotypic effects of changes in gene expression caused by mechanisms other than changes in the underlying DNA sequence, such as inherited changes to **DNA structure** and **packaging including DNA methylation and histone modifications**.



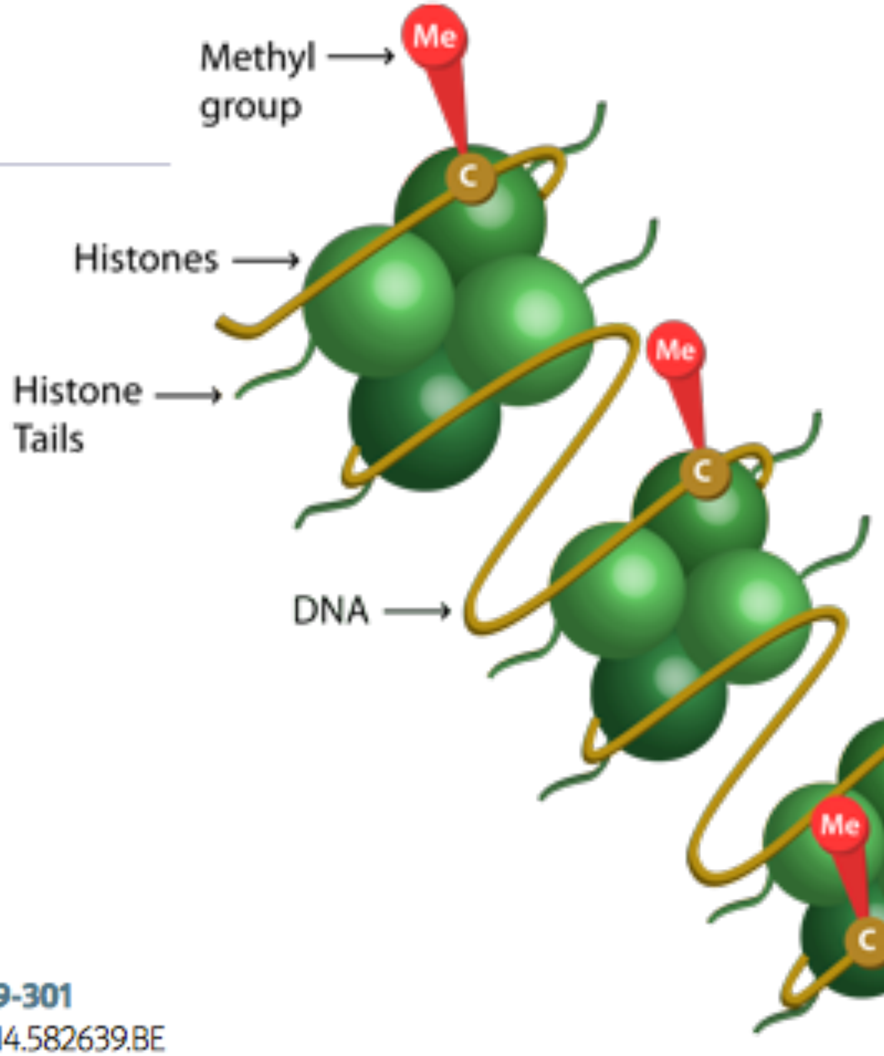
Origins of magic: review of genetic and epigenetic effects

Sreeram V Ramagopalan,[1 2] Marian Knight,[3] George C Ebers,[1 2] Julian C Knight[1]

DISCUSSION

The observed inheritance of magic in the Harry Potter books by J K Rowling would be best explained by a multilocus model with a dominant gene for magic, the function of which is controlled epistatically by one or more other loci,[15] possibly recessive in nature. The genotypes of the loci concerned influence total magical ability, and the allele frequencies of these magical loci would differ significantly between populations with magical abilities and those without (muggles). We cannot yet confirm this finding, however, as those with magical backgrounds have not been included in the International HapMap project.[16] Genetic heterogeneity may exist.

BMJ 2007;335:1299-301
doi:10.1136/bmj.39414.582639.BE
Accepted: 16 November 2007



PopGen ▷ Introduction

It has been suggested that understanding genetic change in populations requires the incorporation of **cultural inheritance**: the part of the phenotypic variation that is inherited socially or learned from others (Danchin et al. 2011).

Evidence for Cultural Differences between Neighboring Chimpanzee Communities

Humans aren’t the only primate species with cultural differences: even in the same environment, different groups of chimpanzees use different tools (Luncz et al. 2012).

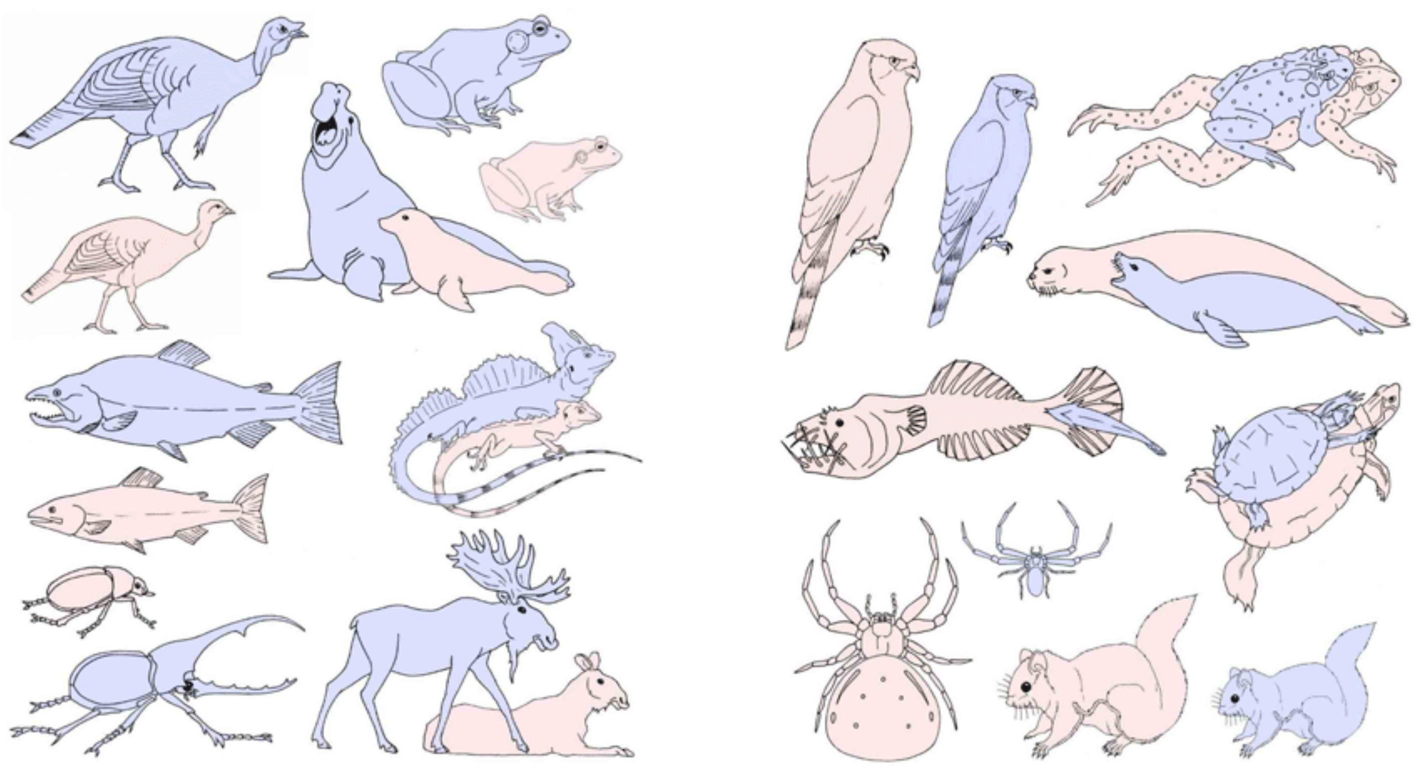
Chimps living in a national park in Cote d’Ivoire like eating Coula nuts. They hammer them open with stone or wood. At the beginning of the season, the nutshells are harder. So you might expect all the chimps in the forest to initially use stone hammers and then switch to easy-to-find wooden tools when the nut-cracking requires less force.

But researchers examined the tool use of three different chimpanzee communities and found that despite sharing genes and a habitat, each group chose their hammers differently.

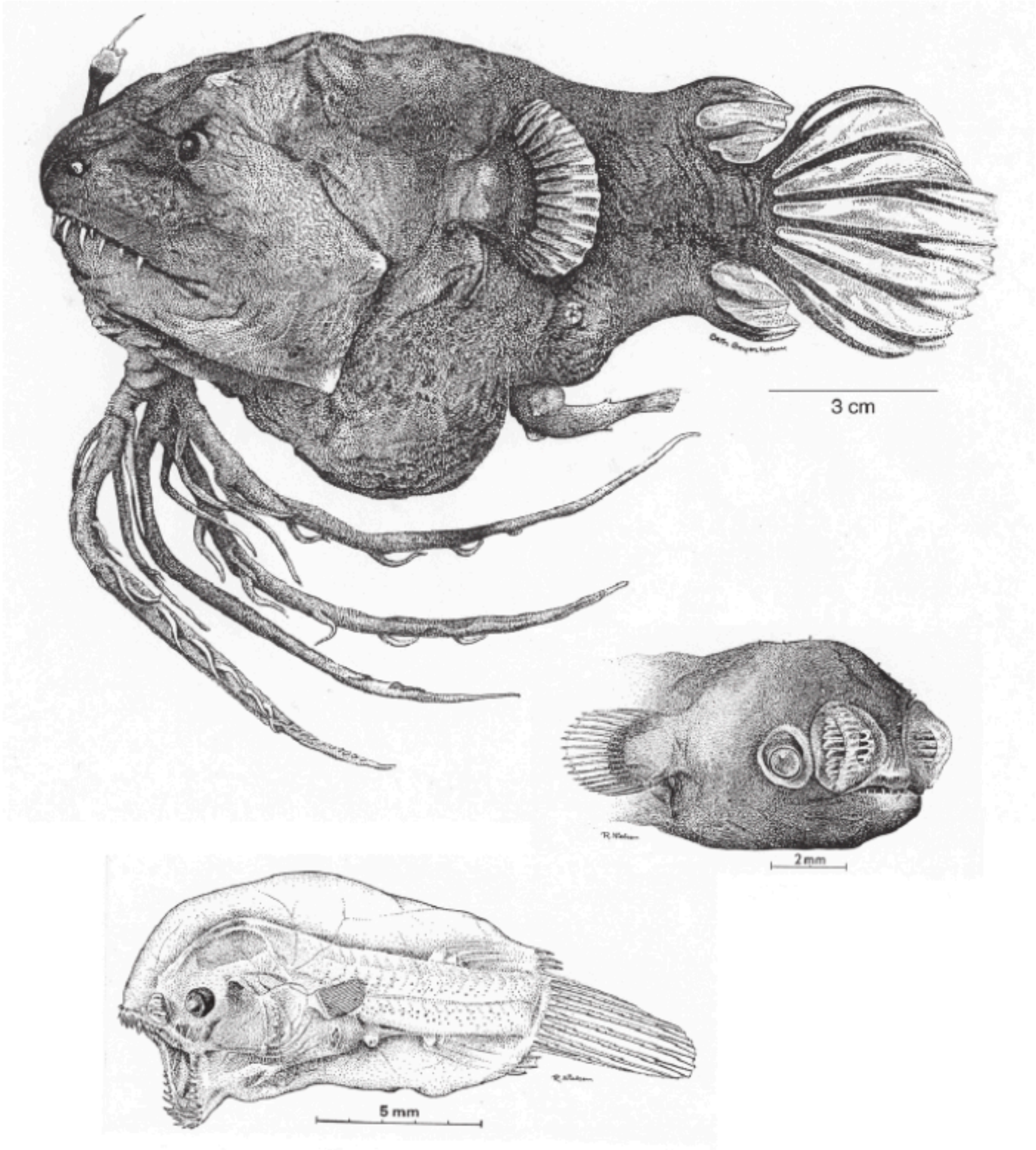
Current Biology 22, 922–926, May 22, 2012 ©2012 Elsevier Ltd All rights reserved DOI 10.1016/j.cub.2012.03.031

PopGen ▷ Introduction

Sexual (size) dimorphism



PopGen ▷ Introduction



Deep-sea anglerfishes of the suborder Ceratioidei. Top: Female Linophryne brevibarbata with parasitic male attached to her venter (after Bertelsen 1980); middle: dwarf male of the Linophryne arborifera -group and bottom: Haplophryne mollis (both after Munk & Bertelsen 1983).

PopGen ▷ Introduction

Polygenic traits, also known as **multifactorial traits**, are traits that are typically controlled by the combined effects of several **non-allelic genes**, each contributing to some aspect of the trait. The effect of a single gene is difficult to distinguish, especially since a polygenic trait involves multiple genes. Many polygenic traits are also influenced by the environment and are called multifactorial. Because multiple genes are involved, polygenic traits do not follow the patterns of Mendelian inheritance, but result in a range of phenotypes. The study of polygenic traits is challenging because of the interaction of multiple genes and environmental factors.

Non-allelic genes, also known as non-allelic loci or non-allelic genetic markers, are genes located on different chromosomes or at significant distances from each other on the same chromosome. These genes are not closely linked and their **inheritance is not influenced by each other.** The term "non-allelic" indicates that these genes are not alternative forms (alleles) of the same gene but rather different, unrelated genes that can independently assort and undergo recombination during sexual reproduction. This independent assortment allows for greater genetic diversity in offspring since the combination of alleles from different genes is not constrained by linkage.

Introduction

Examples of polygenic traits in humans include:

- Several genes influence an individual's **height**, and the combined effects of these genes lead to a wide range of heights within a population.
- **Skin colour** is determined by several genes that control the production of melanin, with variations in these genes contributing to different skin tones.
- **Intelligence** is thought to be influenced by a complex interplay of multiple genetic and environmental factors.
- Traits such as the risk of developing heart **disease**, diabetes and some cancers are polygenic, with multiple genetic and environmental factors contributing to an individual's susceptibility.

PopGen ▷ Introduction

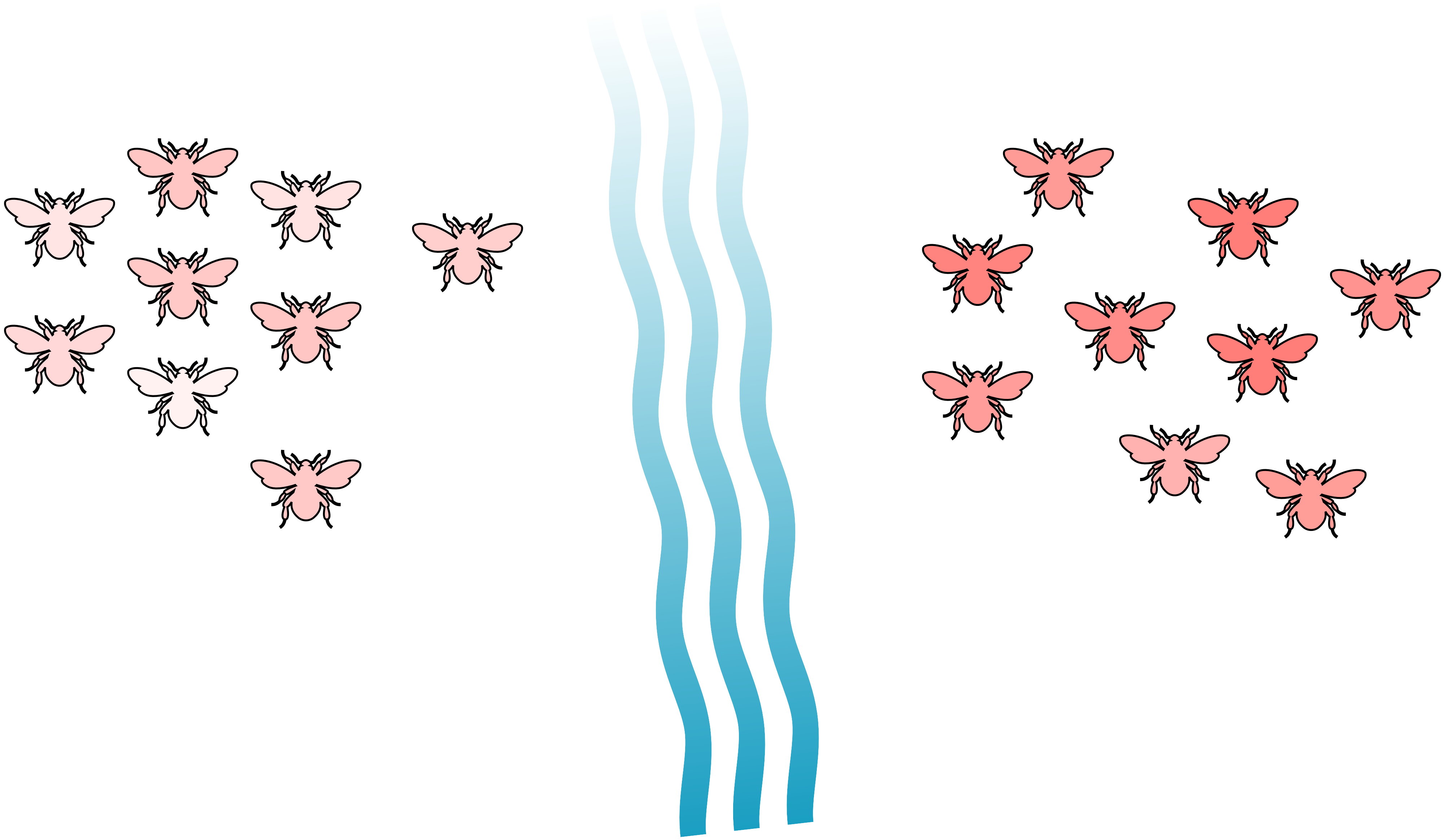
In many genetic diseases, including those that are autosomal dominant, specific mutations associated with the same disease present in different families may be uniform, such that every affected individual carries exactly the same molecular defect (**allelic homogeneity**), or they may be heterogeneous, such that tens or even hundreds of different mutations, all affecting the same gene, may be seen in the affected population (**allelic heterogeneity**). In some cases even mutations in different genes can lead to the same clinical disorder (**genetic heterogeneity**).

Encyclopædia Britannica

It's important to note that while there may be allelic homogeneity at one locus, there may still be genetic diversity throughout the genome and at other loci. Different populations may have different levels of allelic homogeneity at different genetic loci, and genetic diversity is a crucial factor in evolution and adaptation.

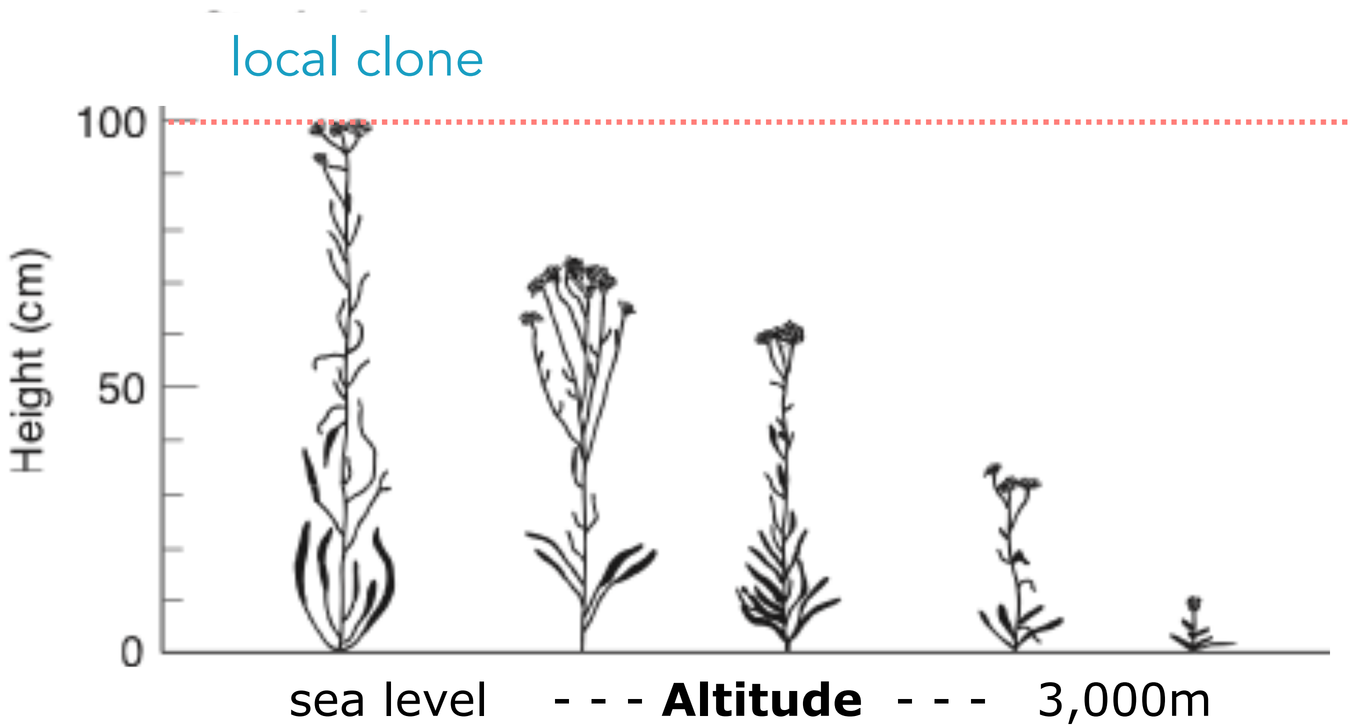
Differentiation

V_G or V_E or V_G+V_E



PopGen ▷ Introduction

Populations from different geographic areas are detectably different for many phenotypic attributes in almost all species. **Gradual changes across geographic or environment gradients are found in many species.** However, there is no simple way to determine if such a cline for a particular phenotype results from genetic or environmental differences between populations. One way to **test for genetic differences between populations is to eliminate environmental differences by raising individuals under identical environmental differences in a so-called common-garden experiment**.



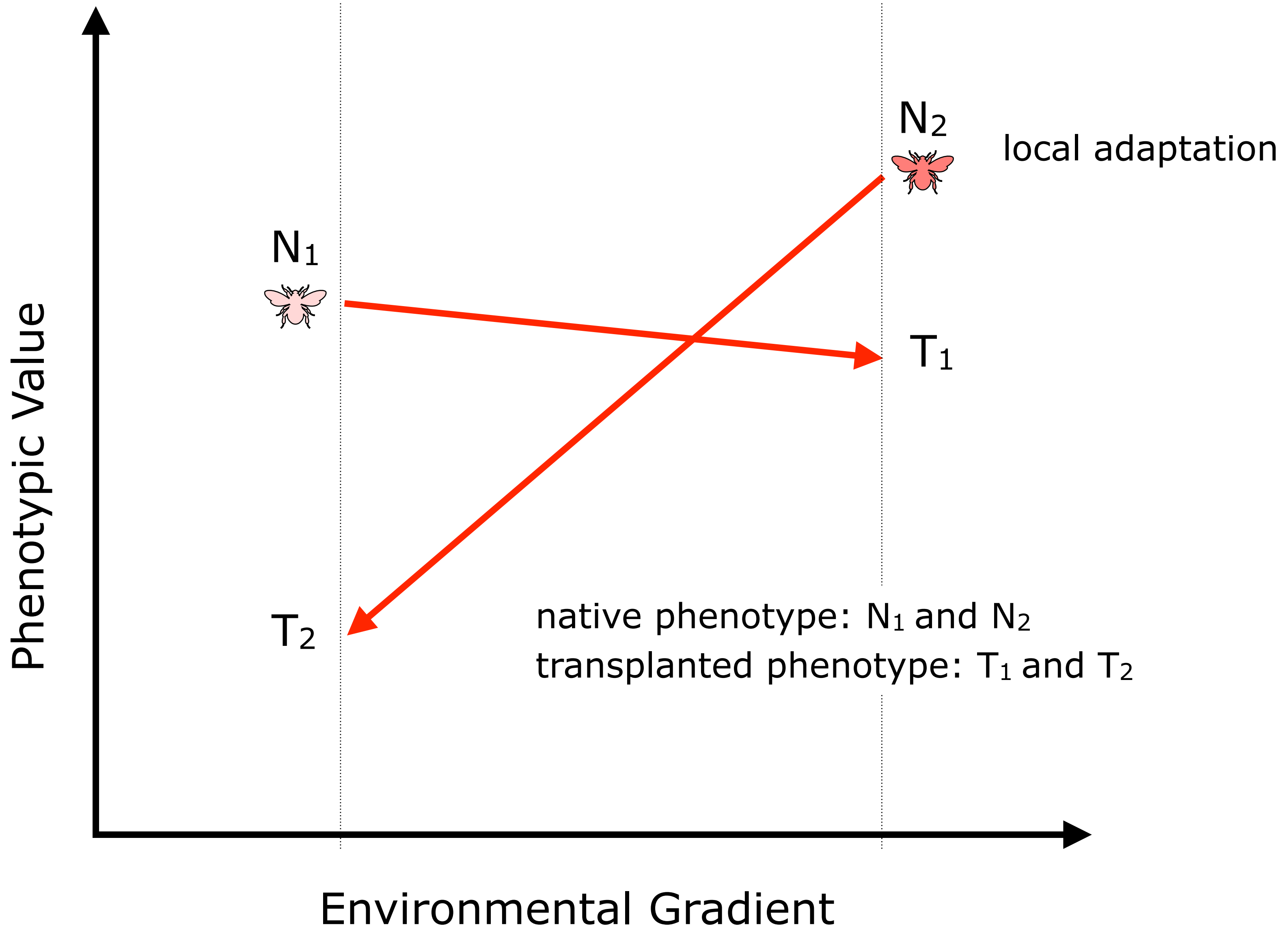
Representative clones of yarrow plants originating from five different altitudinal locations grown at 30 m above sea level at Stanford.

Clausen (1951) Stages in the Evolution of Plant Species.

$$V_P = V_G + V_E$$

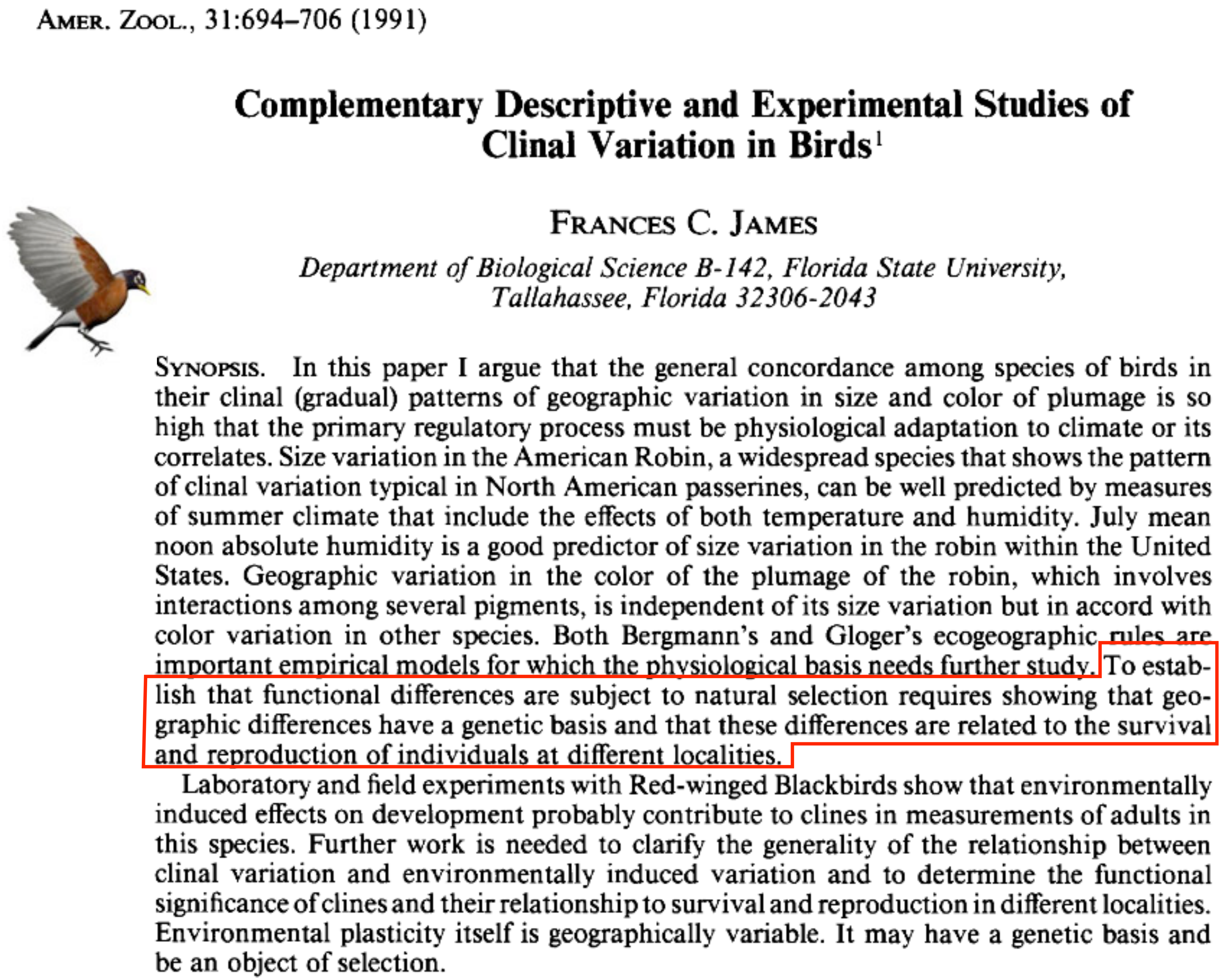
$$V_E \approx 0$$

Gradient variation



native phenotype: N_1 and N_2
transplanted phenotype: T_1 and T_2

PopGen ▷ Introduction



AMER. ZOOL., 31:694–706 (1991)

Complementary Descriptive and Experimental Studies of Clinal Variation in Birds[1]

FRANCES C. JAMES

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Tallahassee, Florida 32306-2043

SYNOPSIS. In this paper I argue that the general concordance among species of birds in their clinal (gradual) patterns of geographic variation in size and color of plumage is so high that the primary regulatory process must be physiological adaptation to climate or its correlates. Size variation in the American Robin, a widespread species that shows the pattern of clinal variation typical in North American passerines, can be well predicted by measures of summer climate that include the effects of both temperature and humidity. July mean noon absolute humidity is a good predictor of size variation in the robin within the United States. Geographic variation in the color of the plumage of the robin, which involves interactions among several pigments, is independent of its size variation but in accord with color variation in other species. Both Bergmann's and Gloger's ecogeographic rules are important empirical models for which the physiological basis needs further study. To establish that functional differences are subject to natural selection requires showing that geographic differences have a genetic basis and that these differences are related to the survival and reproduction of individuals at different localities.

Laboratory and field experiments with Red-winged Blackbirds show that environmentally induced effects on development probably contribute to clines in measurements of adults in this species. Further work is needed to clarify the generality of the relationship between clinal variation and environmentally induced variation and to determine the functional significance of clines and their relationship to survival and reproduction in different localities. Environmental plasticity itself is geographically variable. It may have a genetic basis and be an object of selection.

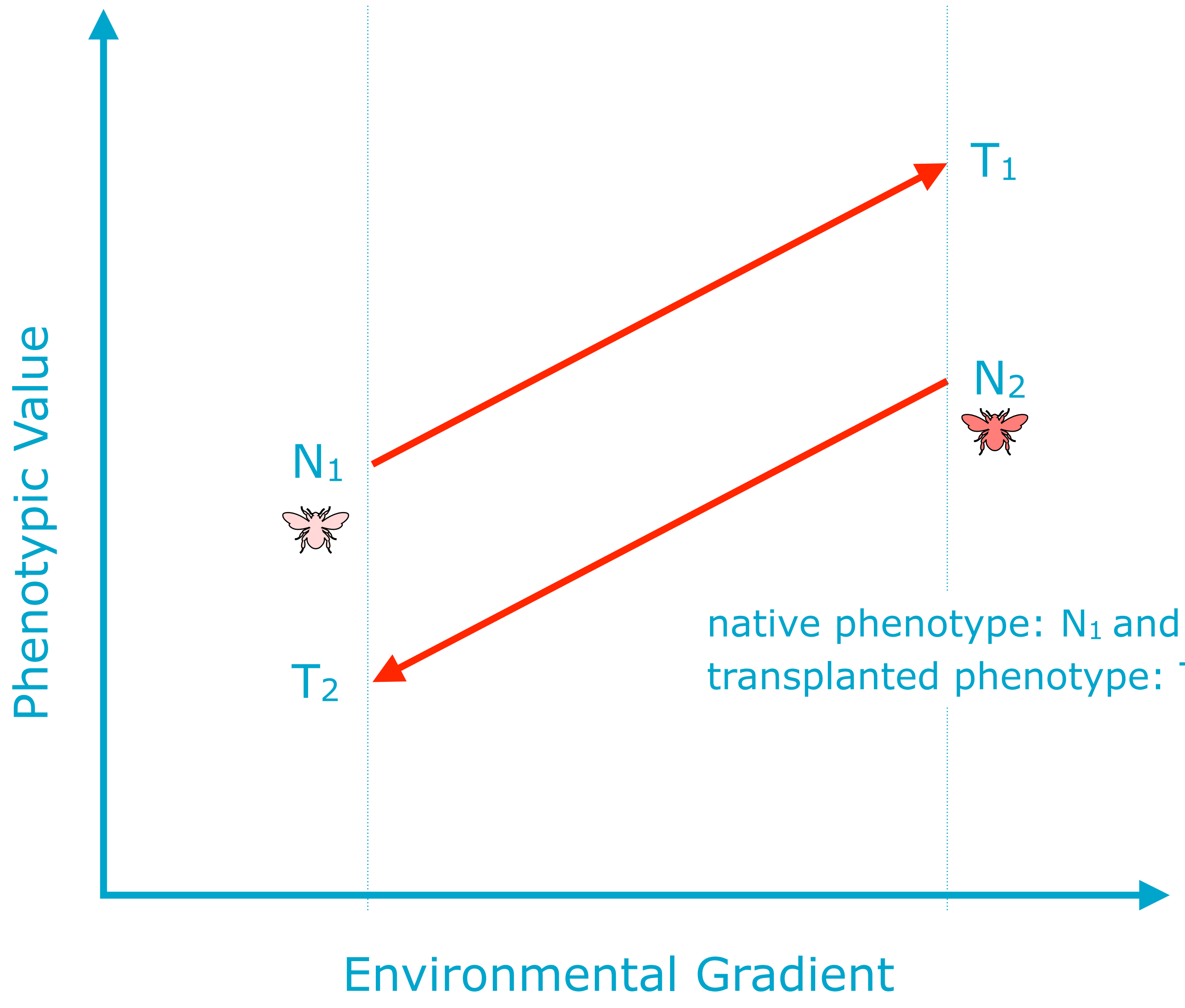
← Size Variation

← Local Climate

← Local Adaptation

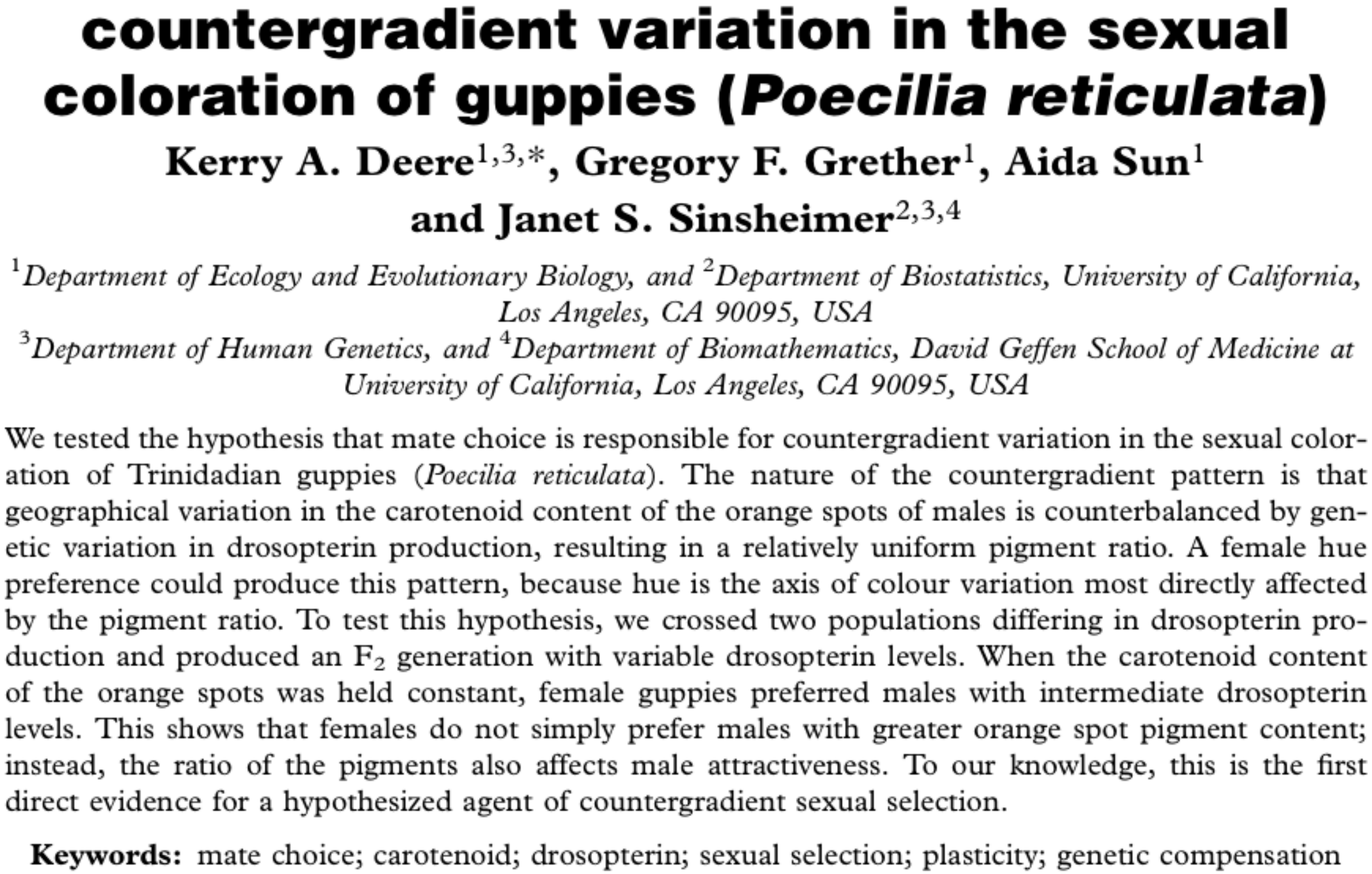
In biology and ecology, an **ecocline** or simply cline describes an ecotone in which a series of biocommunities display a continuous gradient. The term was coined by the English evolutionary biologist Julian Huxley in 1938. (source: WIKIPEDIA)

Counter-gradient variation



native phenotype: N_1 and N_2
transplanted phenotype: T_1 and T_2

Counter-gradient variation is an interesting concept in population genetics because it highlights how genetic adaptation can offset the effects of different environmental challenges, ultimately allowing a species to occupy a wide range of habitats. This phenomenon demonstrates the complex interplay between genetics and the environment in shaping the characteristics of different populations within a species.



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Female mate preference explains countergradient variation in the sexual coloration of guppies (*Poecilia reticulata*)

Kerry A. Deere[1,3,*], Gregory F. Grether[1], Aida Sun[1]
and Janet S. Sinsheimer[2,3,4]

[1] *Department of Ecology and Evolutionary Biology, and* [2] *Department of Biostatistics, University of California, Los Angeles, CA 90095, USA*
[3] *Department of Human Genetics, and* [4] *Department of Biomathematics, David Geffen School of Medicine at University of California, Los Angeles, CA 90095, USA*

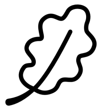
We tested the hypothesis that mate choice is responsible for countergradient variation in the sexual coloration of Trinidadian guppies (*Poecilia reticulata*). The nature of the countergradient pattern is that geographical variation in the carotenoid content of the orange spots of males is counterbalanced by genetic variation in drosopterin production, resulting in a relatively uniform pigment ratio. A female hue preference could produce this pattern, because hue is the axis of colour variation most directly affected by the pigment ratio. To test this hypothesis, we crossed two populations differing in drosopterin production and produced an F_2 generation with variable drosopterin levels. When the carotenoid content of the orange spots was held constant, female guppies preferred males with intermediate drosopterin levels. This shows that females do not simply prefer males with greater orange spot pigment content; instead, the ratio of the pigments also affects male attractiveness. To our knowledge, this is the first direct evidence for a hypothesized agent of countergradient sexual selection.

Keywords: mate choice; carotenoid; drosopterin; sexual selection; plasticity; genetic compensation

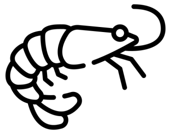
← Sexual coloration
← Mate choice
← spot size **and** ratio

PopGen ▷ Introduction

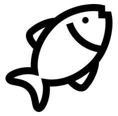
Berven et al. (1979) Countergradient Selection in the Green Frog, Rana clamitans. Evolution 33 (2): 609-623.



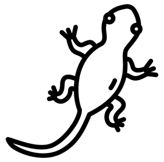
Caignard et al. (2021) Counter-gradient variation of reproductive effort in a widely distributed temperate oak. Functional Ecology 35 (8): 1745-1755.



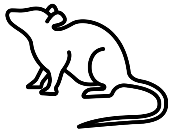
Campos et al. (2009) Latitudinal variation in growth of Crangon crangon (L.): Does counter-gradient growth compensation occur? Journal of Sea Research 62 (4): 229-237.



Gardiner et al. (2010) Counter-Gradient Variation in Respiratory Performance of Coral Reef Fishes at Elevated Temperatures. PLoS ONE 5(10): e1329.



Hodgson & Schwanz (2019) Drop it like it's hot: Interpopulation variation in thermal phenotypes shows counter-gradient pattern. Journal of Thermal Biology 83: 179-187.



Hodgson & Schwanz (2023) Counter-gradient variation and the expensive tissue hypothesis explain parallel brain size reductions at high elevation in cricetid and murid rodents. Scientific Reports 13-5617.

What are the limits of a common-garden experiment?

PopGen ▷ Introduction

Common-garden experiments are a valuable tool in ecology, evolution, and plant science to study how environmental factors, such as soil quality or climate, affect the growth, development, and traits of organisms. While they provide valuable insights, common-garden experiments have certain limitations:

- The Simplicity and Reductionism may not capture the full complexity of natural ecosystems, potentially leading to oversimplified conclusions.
- These experiments often run for a limited duration, which may not allow researchers to assess long-term effects on organisms and ecosystems.
- Organisms in common gardens are often grown in controlled, artificial environments that do not fully mimic their natural habitats.
- When organisms from different environments are transplanted into a common garden, they may experience stress due to the change in conditions.
- Common gardens are typically small-scale experiments, limiting their ability to capture large-scale ecological processes or the movement of species across landscapes.
- Organisms may not immediately adapt to the common-garden conditions, and their responses may lag behind changes in environmental conditions.
- What works in one location may not hold in another due to differences in context. Common gardens may not account for all potential interactions between species and environmental factors, leading to unforeseen outcomes in natural ecosystems.
- Conducting common-garden experiments can be resource-intensive, limiting the number of conditions or species that can be studied simultaneously.
- Genetic variation can have a significant influence on the responses of organisms to environmental changes.

Despite these limitations, common-garden experiments remain a valuable tool for understanding ecological and evolutionary processes, especially when they are designed and interpreted with these constraints in mind. They provide controlled conditions that allow researchers to investigate cause-and-effect relationships and gain insights into how environmental factors influence the traits and performance of organisms.

PopGen ▷ Natural Variation

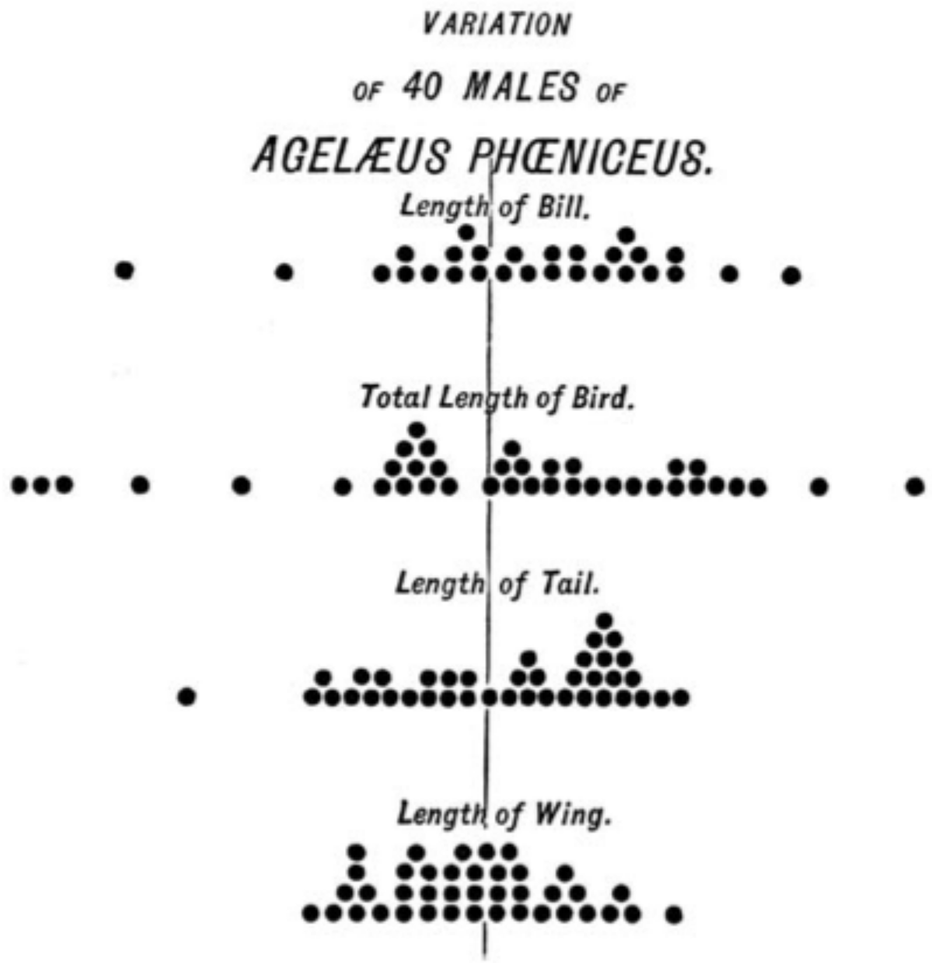
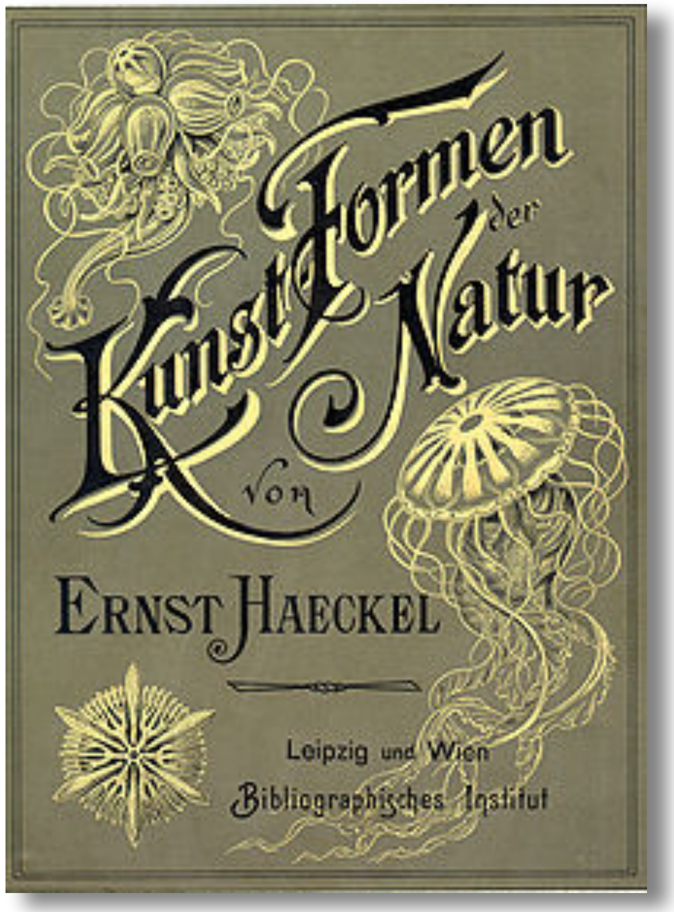
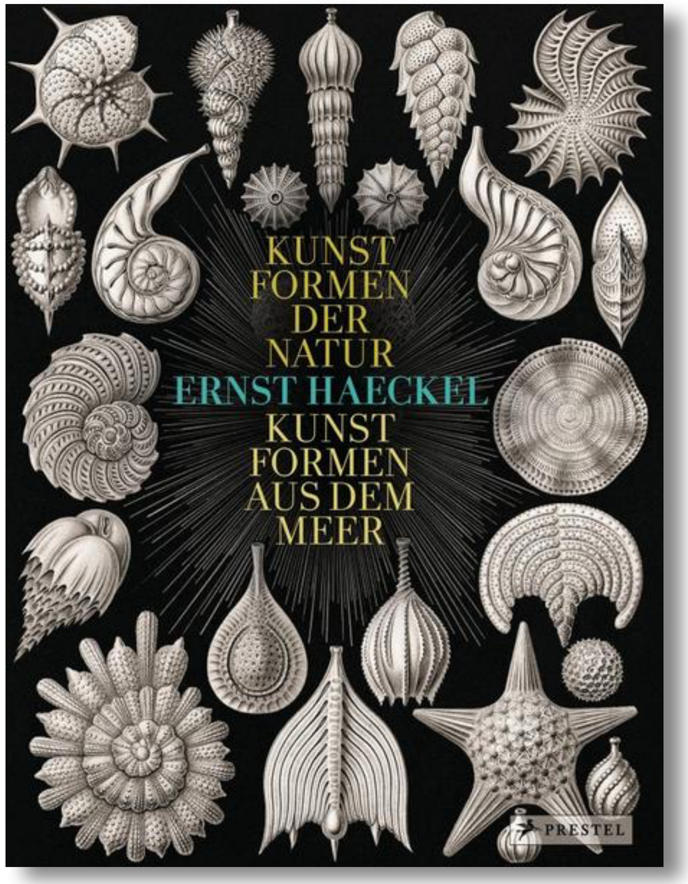


Diagram from Alfred Russel Wallace of variation in body dimensions of 40 red-winged blackbirds in the United States. From Wallace (1923, p. 64).

PopGen ▷ **Natural Variation**

Variation **between** Species

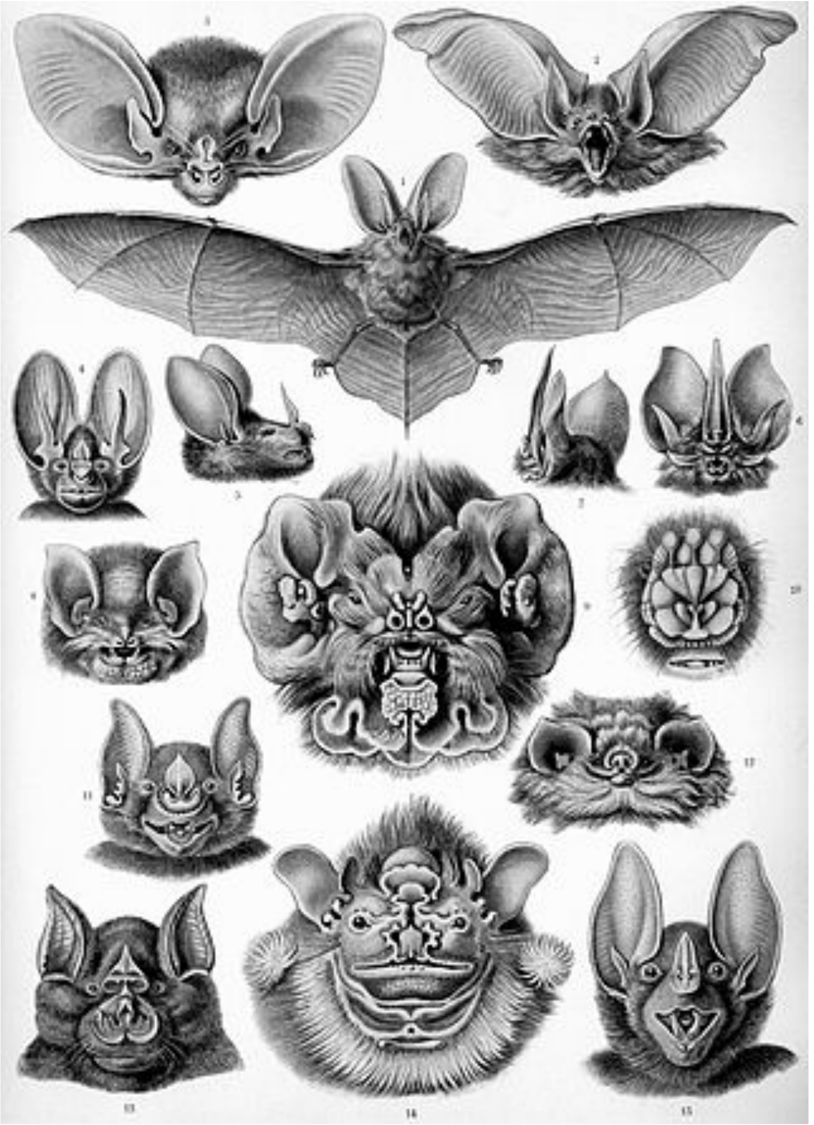




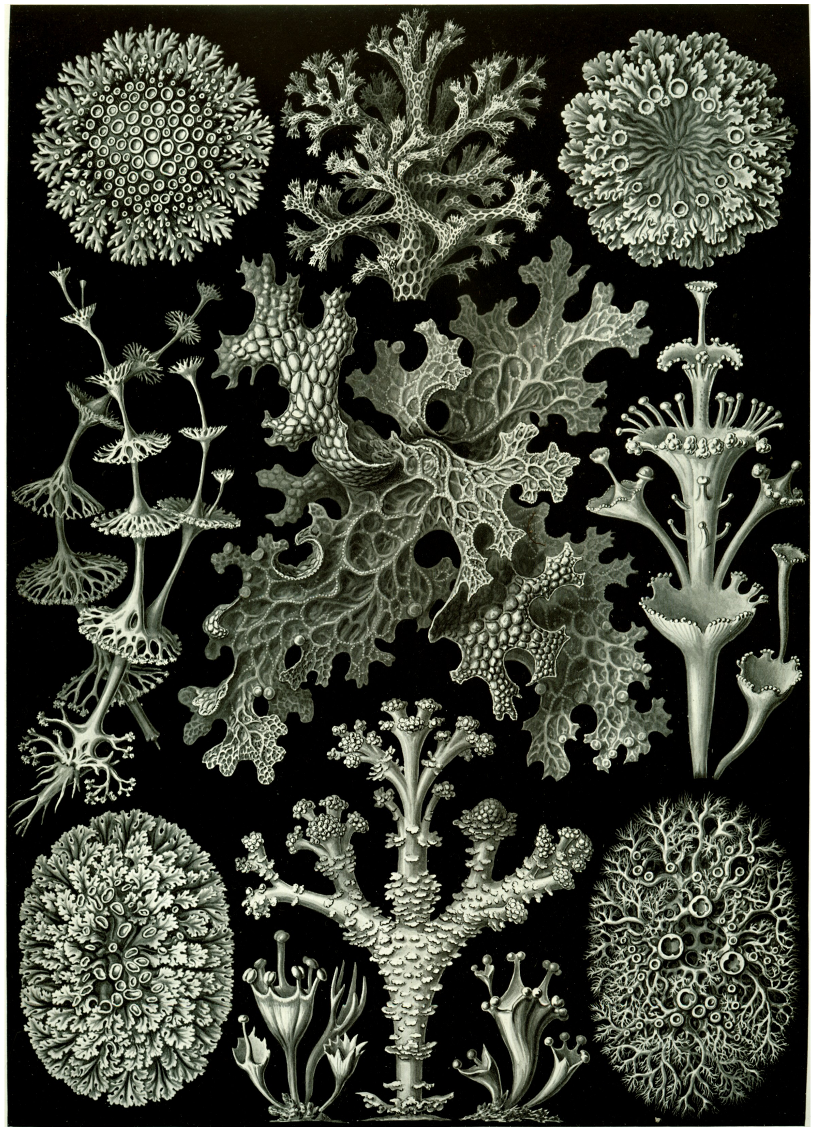
Ernst Heinrich Philipp August **Haeckel** 1834-1919) German zoologist and evolutionist who was a strong proponent of Darwinism and who proposed new notions of the evolutionary descent of human beings.

ENCYCLOPÆDIA BRITANNICA

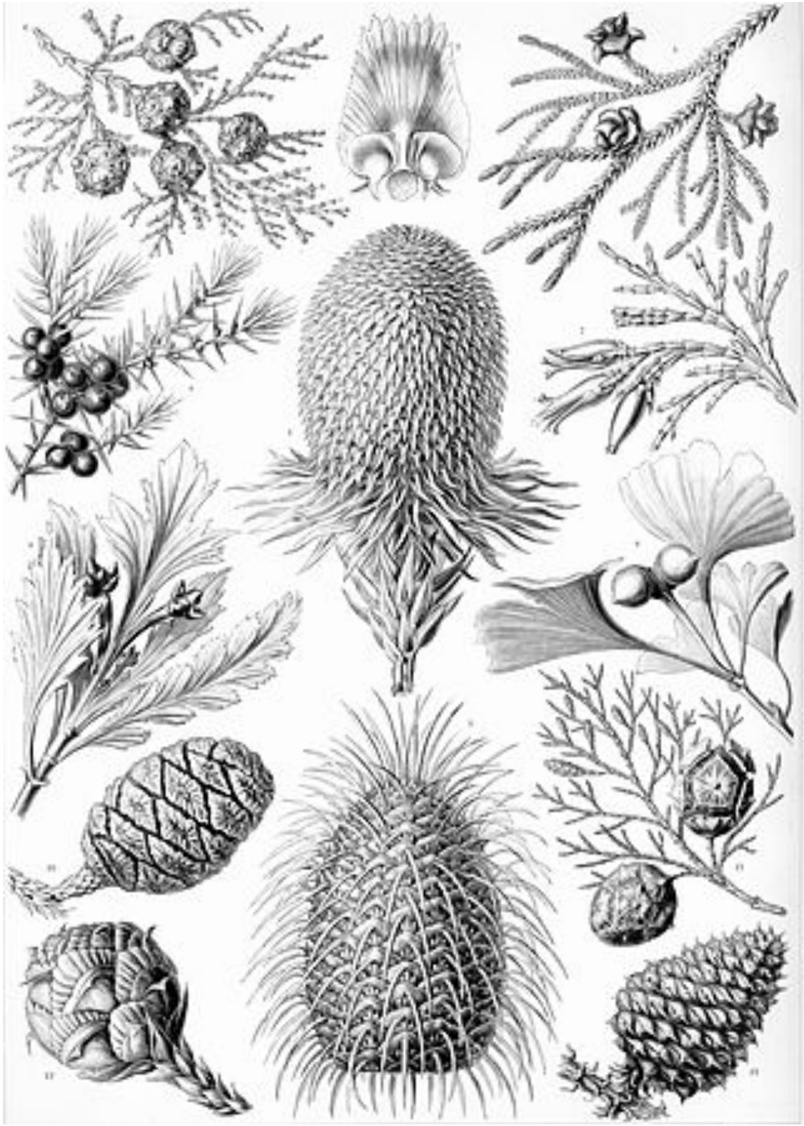
PopGen ▷ Natural Variation



Chiroptera



Lichenes



Coniferae

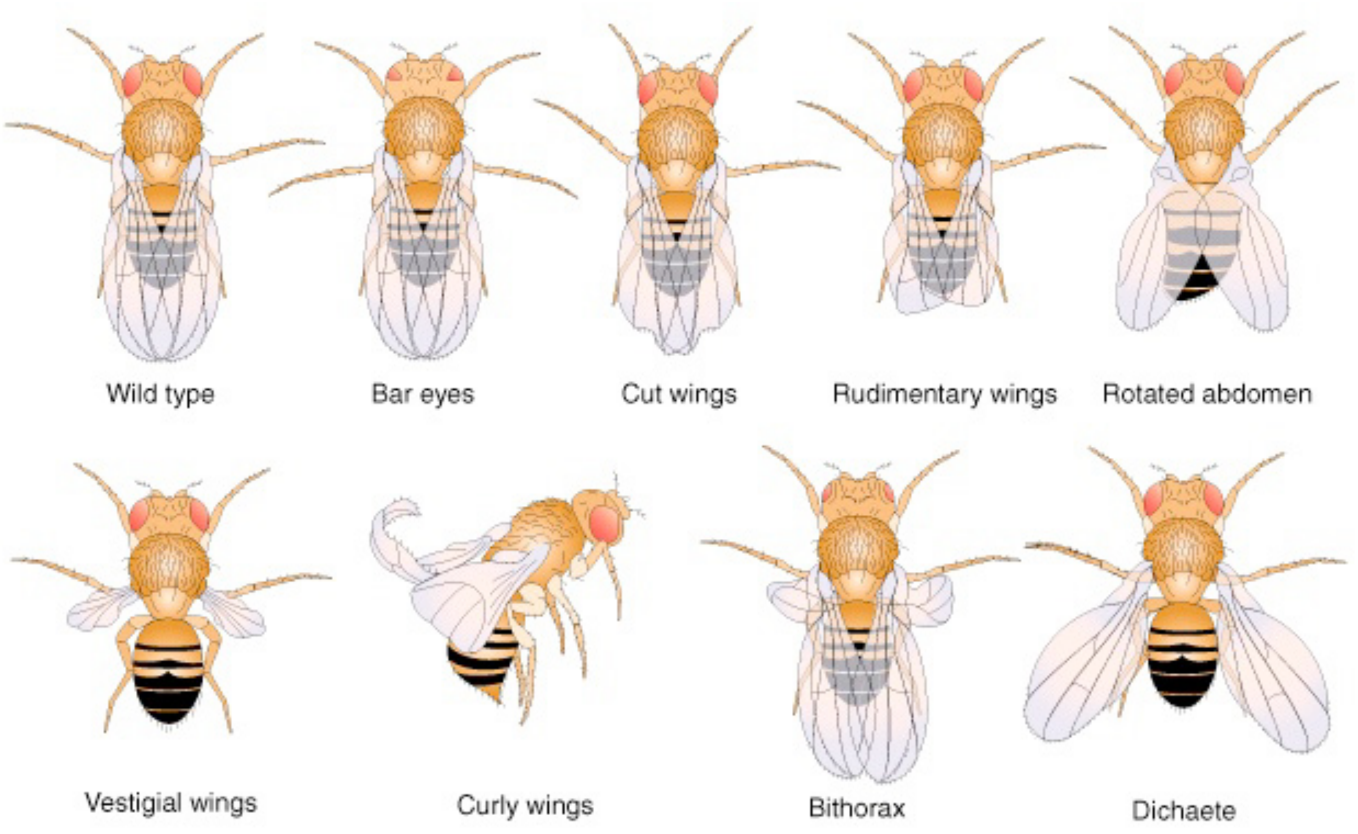
PopGen ▷ Natural Variation

Alfred Russel Wallace (co-founder of the principle of natural selection) was perhaps the first biologist to emphasise the extent and importance of **variability within natural populations**. His idea was in opposition to the commonly held view of naturalists in the 19th century that individual variation was comparatively rare in nature.

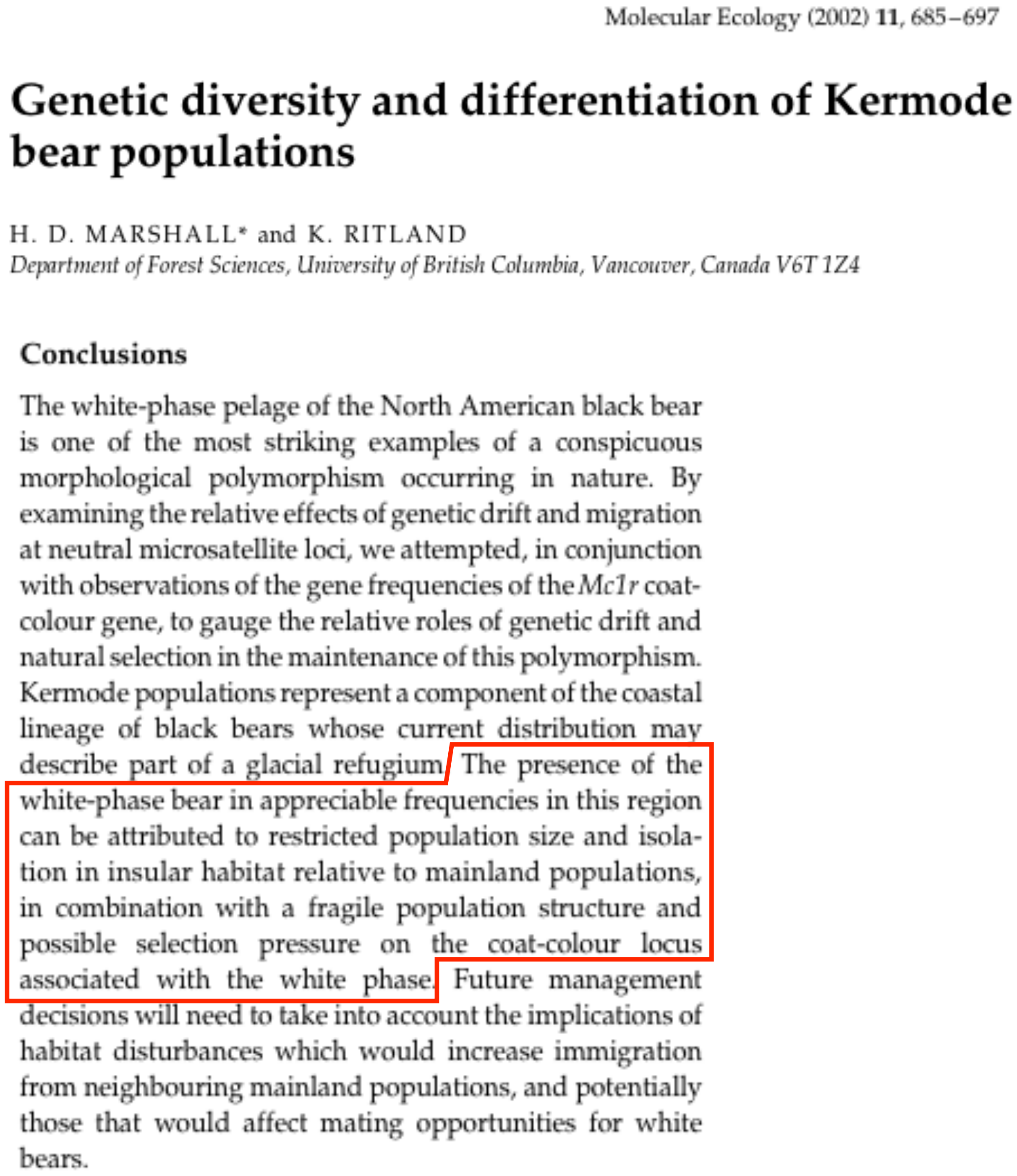
Ahlgren et al. 2013, Biol. Lett. Vol. 9

Variability among individuals is essential for adaptive evolutionary change. In other words, natural selection cannot operate unless there are phenotypic differences between individuals.

Morphological Phenotype



Color-Pattern Phenotype



Molecular Ecology (2002) **11**, 685–697

Genetic diversity and differentiation of Kermode bear populations

H. D. MARSHALL* and K. RITLAND
Department of Forest Sciences, University of British Columbia, Vancouver, Canada V6T 1Z4

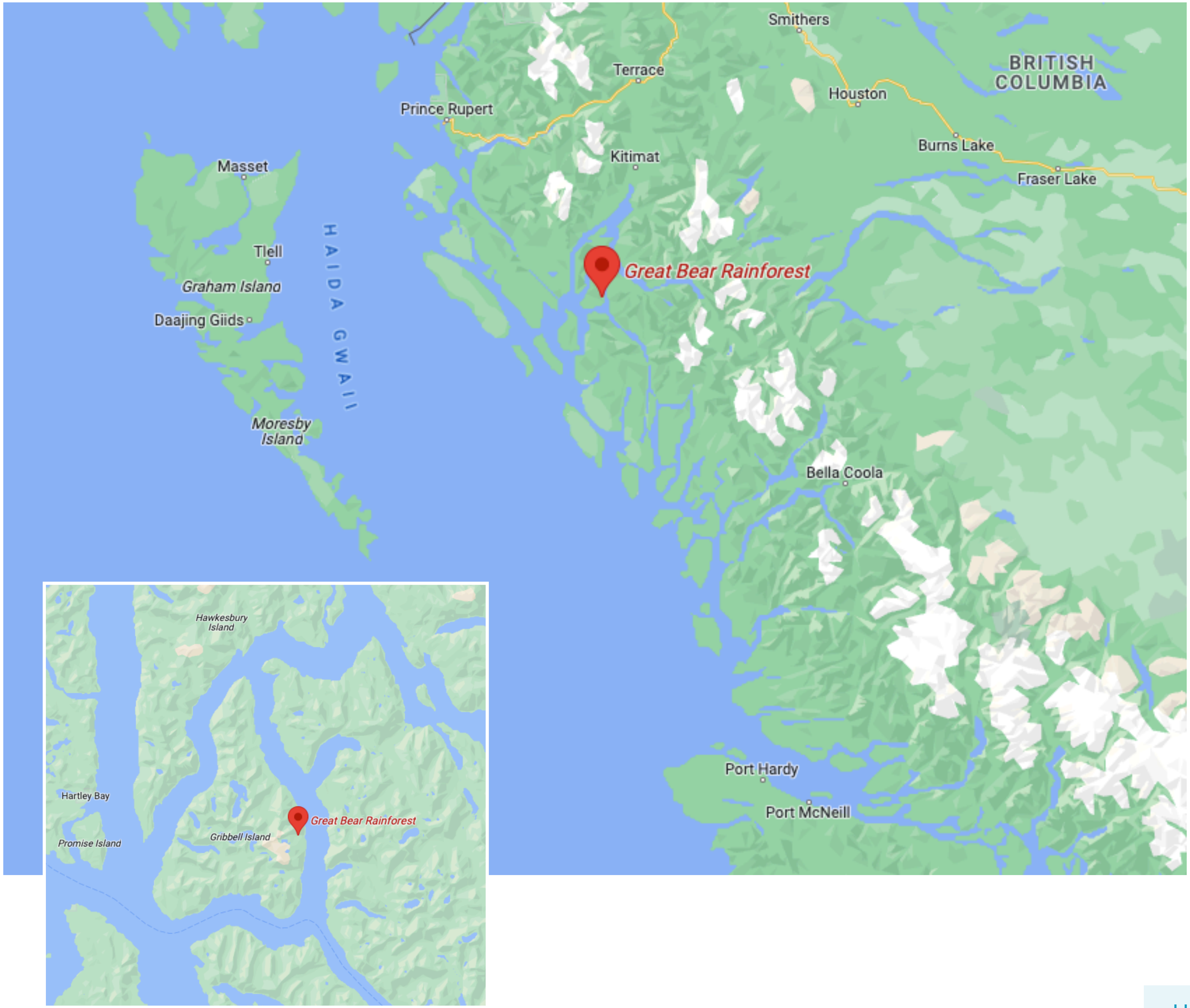
Conclusions

The white-phase pelage of the North American black bear is one of the most striking examples of a conspicuous morphological polymorphism occurring in nature. By examining the relative effects of genetic drift and migration at neutral microsatellite loci, we attempted, in conjunction with observations of the gene frequencies of the *Mc1r* coat-colour gene, to gauge the relative roles of genetic drift and natural selection in the maintenance of this polymorphism. Kermode populations represent a component of the coastal lineage of black bears whose current distribution may describe part of a glacial refugium. The presence of the white-phase bear in appreciable frequencies in this region can be attributed to restricted population size and isolation in insular habitat relative to mainland populations, in combination with a fragile population structure and possible selection pressure on the coat-colour locus associated with the white phase. Future management decisions will need to take into account the implications of habitat disturbances which would increase immigration from neighbouring mainland populations, and potentially those that would affect mating opportunities for white bears.

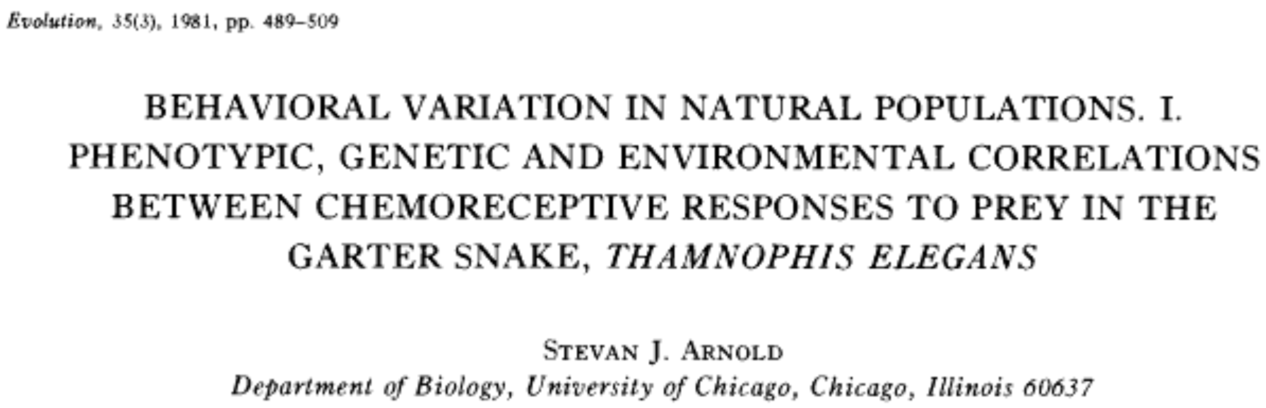
PopGen ▷ Natural Variation

- Spirit bears, also known as Kermode bears (*Ursus americanus kermodei*), are a subspecies of North American black bear (not albino bears!) with a rare **recessive** gene that makes their fur white or creamy.
- Spirit bears are found only in the Great Bear Rainforest, a 6.4 million hectare ecosystem on the north and central coast of British Columbia. It is the largest intact temperate rainforest in the world.
- Pacific salmon are the lifeblood of the Great Bear Rainforest. When the salmon return from the ocean to spawn in the autumn, the spirit bears emerge from the forest to feast on pink, chum and coho salmon before hibernating. When they're not hibernating and there are no salmon around, spirit bears eat a variety of foods including fruit, berries, nuts, grasses, roots, other plants, insects, fawns and carrion. Spirit bears are classic omnivores, capable of eating a wide variety of foods.
- White bears are more successful at catching salmon in daylight than black bears. Pale bears are better camouflaged during the day than dark ones because they contrast less with the bright sky, so salmon don't notice them as much.

PopGen ▷ Natural Variation



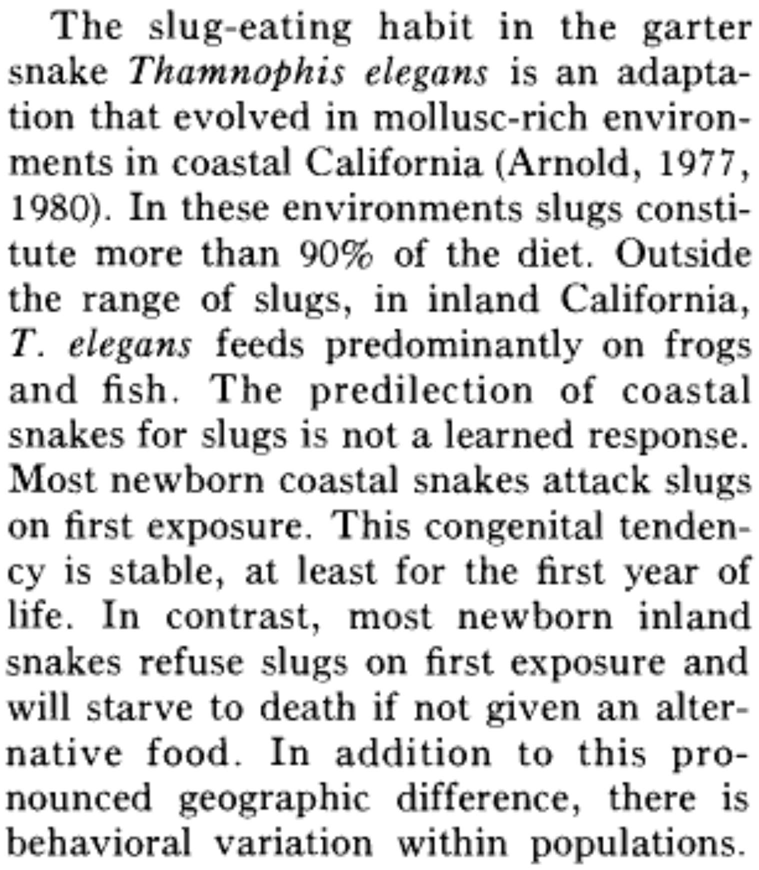
Behavioural Phenotype



Evolution, 35(3), 1981, pp. 489–509

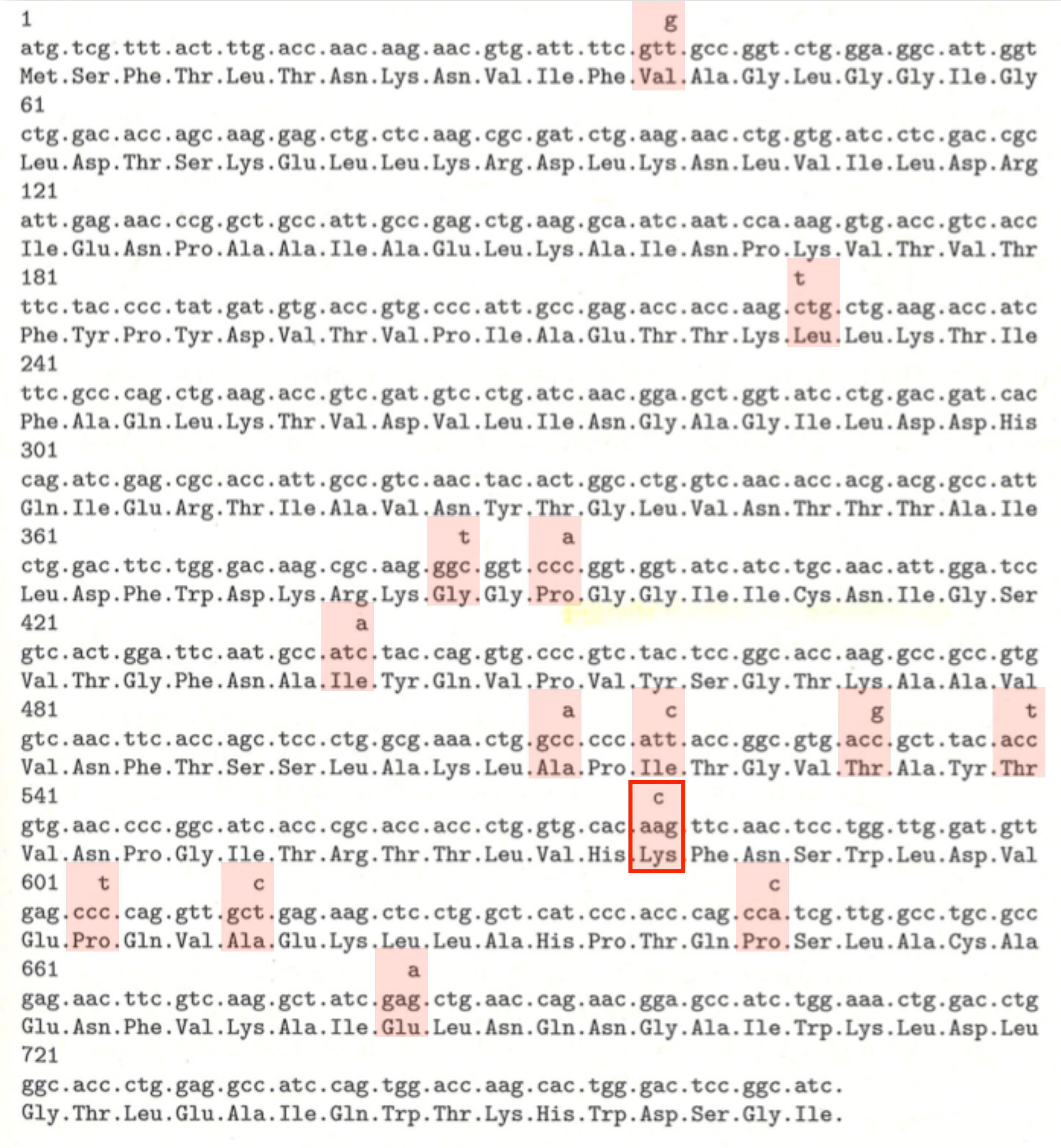
BEHAVIORAL VARIATION IN NATURAL POPULATIONS. I.
PHENOTYPIC, GENETIC AND ENVIRONMENTAL CORRELATIONS
BETWEEN CHEMORECEPTIVE RESPONSES TO PREY IN THE
GARTER SNAKE, *THAMNOPHIS ELEGANS*

STEVAN J. ARNOLD
Department of Biology, University of Chicago, Chicago, Illinois 60637



The slug-eating habit in the garter snake *Thamnophis elegans* is an adaptation that evolved in mollusc-rich environments in coastal California (Arnold, 1977, 1980). In these environments slugs constitute more than 90% of the diet. Outside the range of slugs, in inland California, *T. elegans* feeds predominantly on frogs and fish. The predilection of coastal snakes for slugs is not a learned response. Most newborn coastal snakes attack slugs on first exposure. This congenital tendency is stable, at least for the first year of life. In contrast, most newborn inland snakes refuse slugs on first exposure and will starve to death if not given an alternative food. In addition to this pronounced geographic difference, there is behavioral variation within populations.

Genetic Variation



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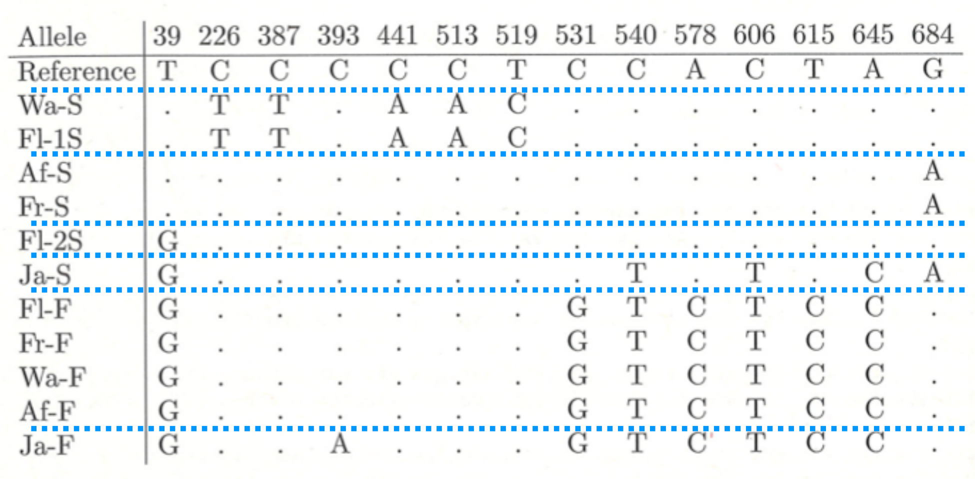
```

The 1983 paper "Nucleotide polymorphism at the alcohol dehydrogenase (*Adh*) locus of *Drosophila melanogaster,*" by Marty Kreitman, was a milestone in evolutionary genetics because it was the first to describe **sequence variation** in a sample of alleles obtained from nature.



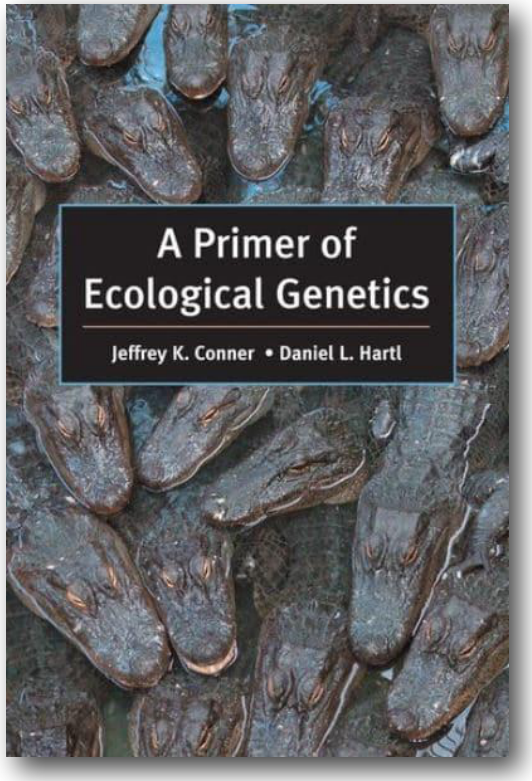
Allele	39	226	387	393	441	513	519	531	540	578	606	615	645	684
Reference	T	C	C	C	C	C	T	C	C	A	C	T	A	G
Wa-S	.	T	T	.	A	A	C	.	.	.	.	.	.	.
Fl-1S	.	T	T	.	A	A	C	.	.	.	.	.	.	.
Af-S	.	.	.	.	.	.	.	.	.	.	.	.	.	A
Fr-S	.	.	.	.	.	.	.	.	.	.	.	.	.	A
Fl-2S	G	.	.	.	.	.	.	.	.	.	.	.	.	.
Ja-S	G	.	.	.	.	.	.	.	T	.	T	.	C	A
Fl-F	G	.	.	.	.	.	.	G	T	C	T	C	C	.
Fr-F	G	.	.	.	.	.	.	G	T	C	T	C	C	.
Wa-F	G	.	.	.	.	.	.	G	T	C	T	C	C	.
Af-F	G	.	.	.	.	.	.	G	T	C	T	C	C	.
Ja-F	G	.	.	A	.	.	.	G	T	C	T	C	C	.

> Organism : *Drosphila melanogaster*
> Samples: 11
> Gene : *Adh*
> Size : 768nt (256aa)
> Number of segregating sites : 14
> Number of alleles : 6 (7)

Source: Kreitman (1983) Nature. doi: 10.1038/304412a0.

Note: There are 194 known classical and insertional alleles for the gene that codes for ADH in *Drosophila melanogaster.*

AAG - Lysine (K) / ACG - Threonine (T)

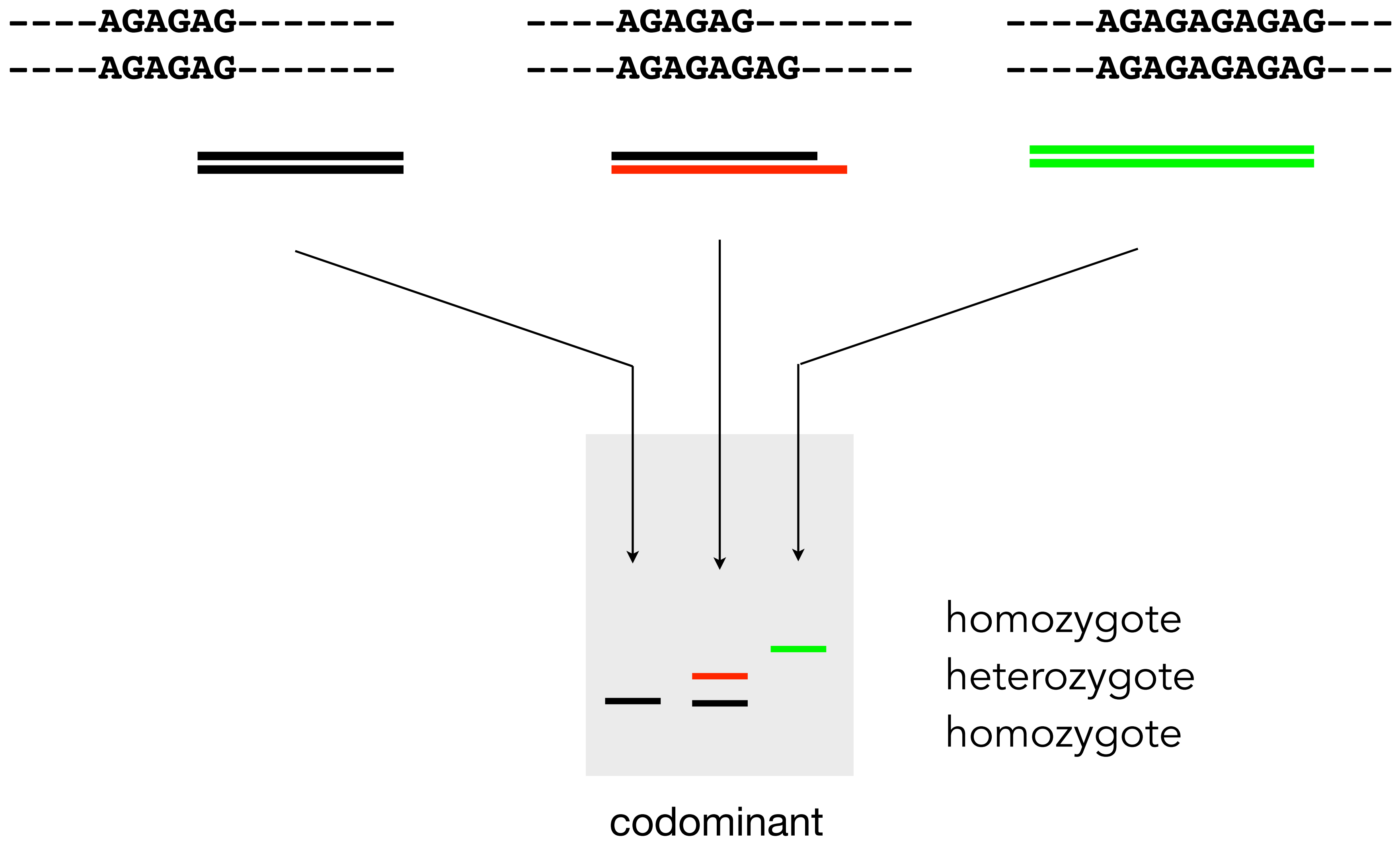

A Primer of Ecological Genetics
Jeffrey K. Conner • Daniel L. Hartl

A Primer of Ecological Genetics

Chapter 2 - Genetic variation - Page 9-22

Molecular markers : Microsatellites

(simple sequence repeats, usually 1-6bp long)



```
----AGAGAG-------        ----AGAGAG-------        ----AGAGAGAGAG---
----AGAGAG-------        ----AGAGAGAG-----        ----AGAGAGAGAG---
```

homozygote
heterozygote
homozygote

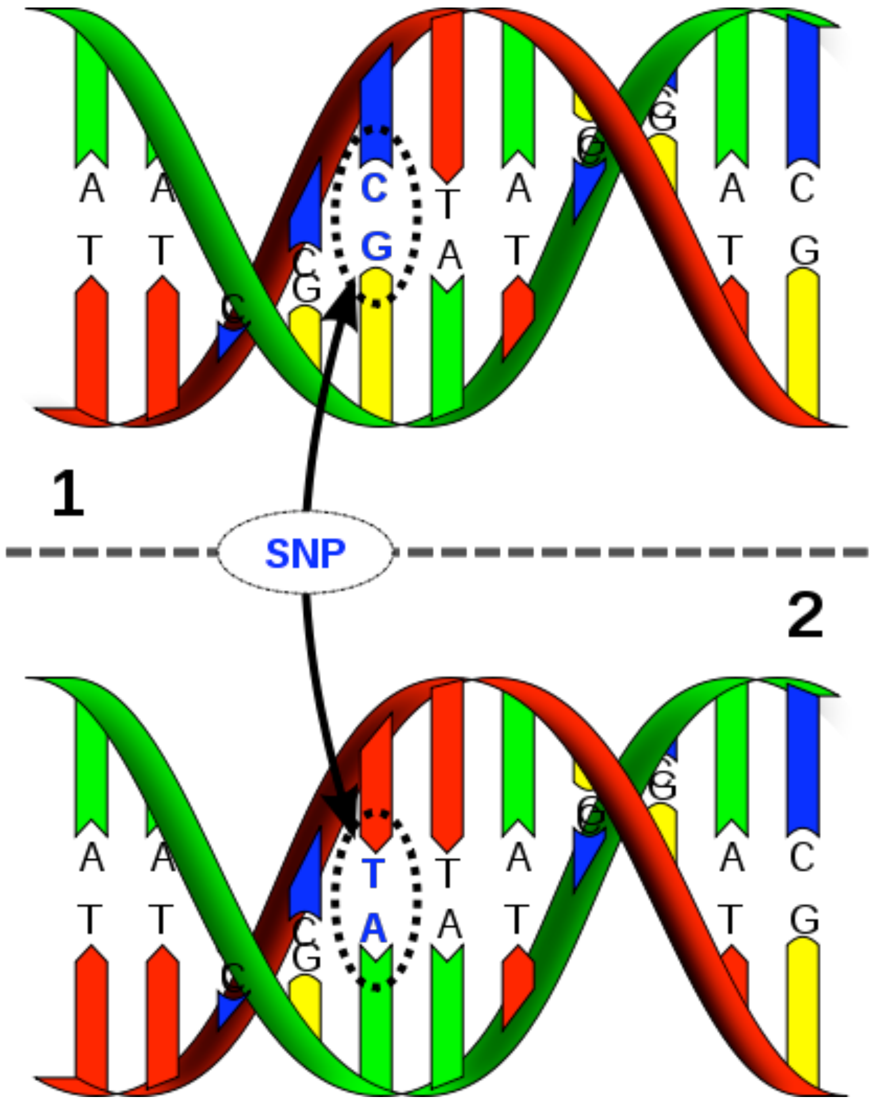
codominant

SNPs (Single-Nucleotide Polymorphism)

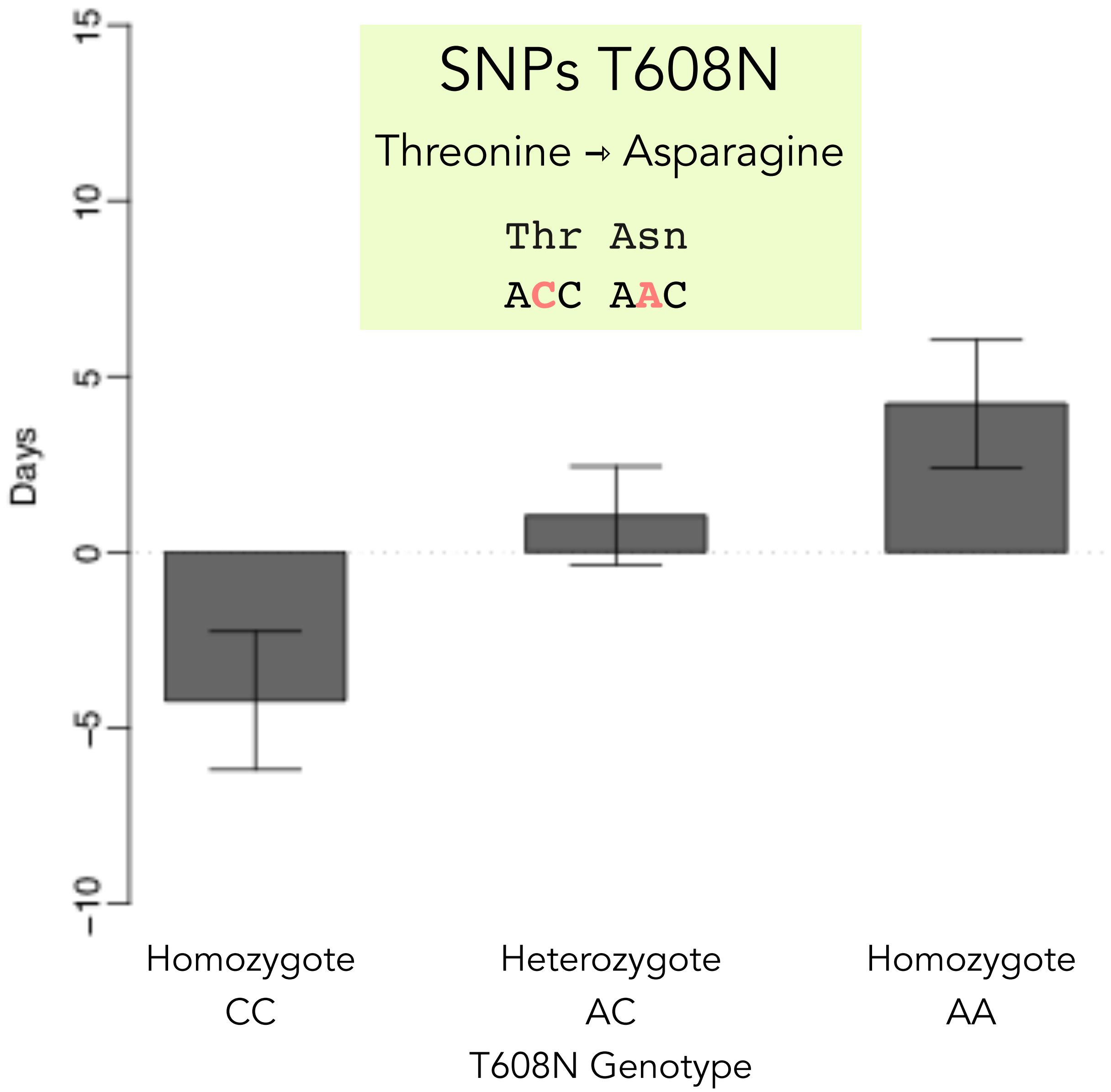
SNPs refer to DNA sequence variation occurring when a single nucleotide in the genome differs between members of the same species (or between paired chromosomes in an individual) - **a SNP is a single base-pair substitution (mutation) at a specific locus.**

The genetic sequences of even non related people are remarkably similar. When the chromosomes of two humans are compared, their DNA sequences can be identical for hundreds of bases. But at about one in every 1000 bases the sequences will differ.

Association studies (e.g. QTL) can determine whether a genetic variant (SNP) is associated with a disease or trait.



Genotypic effects of **phytochrome B**



SNPs T608N

Threonine → Asparagine

`Thr Asn`
`ACC AAC`

Association between genotypes at a SNP at phytochrome B and the date of **bud set** in the Eurasian aspen (*Populus tremula*).

Ingvarsson et al.(2008) Genetics.