

BIOL3110: Evolutionary and Conservation and genetics

Lecture 1:

Introduction to unit format/schedule/assessments & staff, foundation concepts in conservation genetics

Unit staffing

Convener:

A/Prof. Adam Stow

Email (best): adam.stow@mq.edu.au

Office: 6 Science Rd-275,

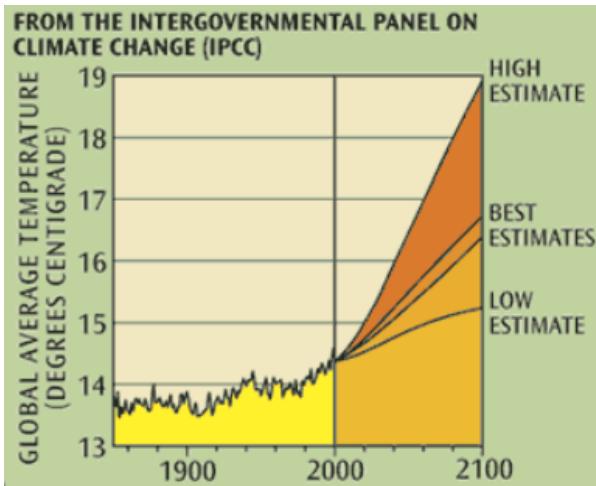
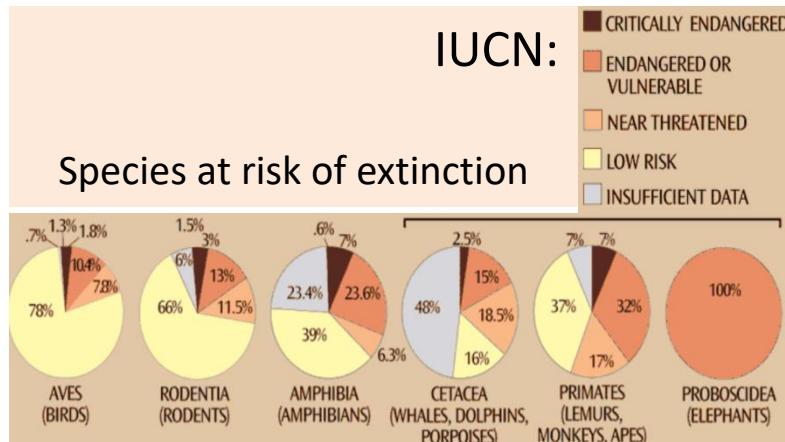
Ph: (02) 9850 8135

Tutor: Jessica O'Hare

Unit OVERVIEW

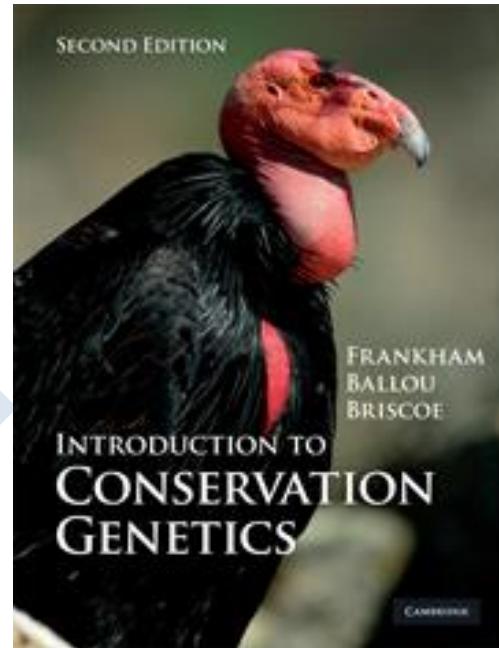
Major themes:

- **Genetic diversity (variation);**
- Determinants of genetic diversity;
- Considerations for conservation in wild & captive populations;
- Quantitative genetics, molecular genetics & genomics as tools
- Evolutionary & Ecological genetics
 - **Applied Macquarie research**

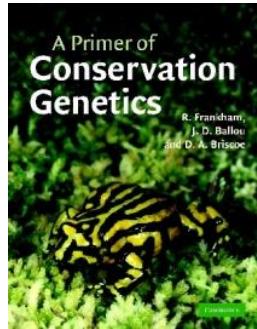


Unit TEXT(s)

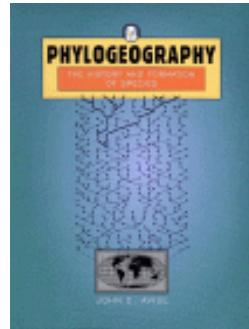
- Frankham et al. 2009
- Required & necessary
- Conceived for and strongly linked to course content
- Contains weekly problems



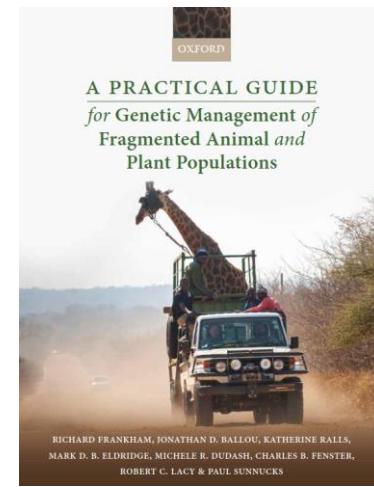
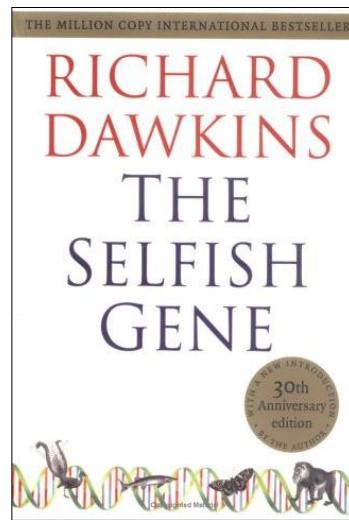
...Others:



Frankham et al. (2004) *A Primer of Conservation Genetics*, Cambridge



Avise (2000) *Phylogeography: The History and Formation of Species*, Harvard



Unit delivery

Read relevant textbook chapter & other readings (on iLearn)

- Work through problems for upcoming tutorial (weeks 2-4 & 8-10)
- Prepare assignments & seminar, study for tests & final exam

Timetable:

Lecture 1	Monday	12:00-1.00	ONLINE
Lecture 2	Tuesday	12:00-1.00	ONLINE
Tutorial [class 1]	Wednesday	10:00-12:00	14 Eastern Road 120 Science Lab
Tutorial [class 2]	Wednesday	2:00-4.00	14 Eastern Road 120 Science Lab
Tutorial [class 3]	Wednesday	4.30-6.30	ONLINE
Residential session (for infrequent attendance students)	6 - 8th of April (inclusive)		14 Eastern Rd, 160 Science Lab

Online delivery

www.iLearn.mq.edu.au

Access for:

- Announcements
- Detailed unit guide (latest **updates**)
- Assignment information & **submission**
- Grades
- Lecture materials (overheads/Echo360)
- Discussion board
- Other resources (e.g. example papers)

A screenshot of the iLearn platform interface. At the top right, it says "Week 4". Below that, there's a red box around the "Assignment 1" section. To the right of the red box, the text "Assignment 1 (5%)" is visible. A red bracket on the right side of the slide points from the word "submission" in the list above to the "Assignment 1" section in the screenshot.



General Assessment Information

Due dates, extensions, penalties and disruption to study

Overdue assignments will attract a penalty at the rate of 5 % of the total mark allocated for the assignment per day past the due date. This penalty will be capped at 75 %, which means that once your submission is more than 15-days overdue you can earn up to a maximum of 25 %.

The date and time of your submission will be taken as registered by TURNITIN.

General Assessment Information

Disruption to study

Deadlines for assessments are **not negotiable** except under circumstances when you have experienced a serious and unavoidable disruption. In such instances, you should formally lodge a **disruption to studies notification via ASK@MQ**.

1. To be eligible for special consideration, you must notify the University of a *serious and unavoidable* disruption **within five (5) working days** of the commencement of the disruption;
2. Such requests must be lodged for the **specific assessment task** for which you experienced disruption. Special consideration cannot be granted retrospectively (i.e., beyond the 5-day window of each assessment due-date);

UNIT GUIDE

See it for:

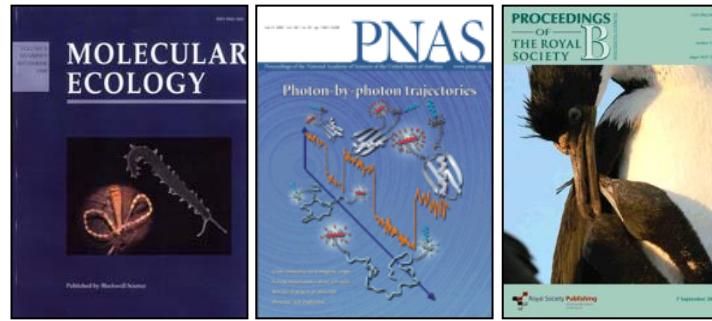
- Unit completion requirements
- Detail on upcoming assessments (including submission guidelines)
- **Due dates/penalties/special consideration**
- Plagiarism policy
- Other general policies
- Statements of:
 - Unit objectives
 - Learning outcomes
 - Graduate capabilities

Assessment at a glance

Task:	Worth	Learning Outcomes	Graduate Capabilities	Description
Assignment 1 EARLY assessment task	5%	1-3	1-9	Commentary article
Problem test 1	15%	1-4,6	1-9	Multiple choice & problems
Problem test 2	15%	1-4,6	1-9	Multiple choice & problems
Seminar	15%	1-3,5,7	1-9	Oral lab presentation
Assignment 2	20%	1-6,8	1-9	Scientific lab report
Final exam	30%	1-6	1-9	Test of all unit content (problem based) – error in unit guide

Assignment 1

Commentary article (5%)



- Commentary-style article on genetic diversity & conservation (**500-600 words**)
- Identify a paper from leading journals....examples in **iLearn** folder located in Week 3
- Read paper and its methodologies and write a commentary in the tone of those appearing in the 'Perspectives' section of the journal *Science*.
- Assessment weighted for participation, presentation and genuine effort.
- **iLearn** link for TURNITIN submission in Week 4



Assignment 1

Commentary article (5%) – see **iLearn week 3 or Unit outline**

Original article

Genetic Restoration of the Florida Panther

Warren E. Johnson,^{1*}† David P. Onorato,^{2,*†} Melody E. Roelke,^{3*} E. Darrell Land,^{2*} Mark Cunningham,² Robert C. Belden,⁴ Roy McBride,⁵ Deborah Jansen,⁶ Mark Lotz,² David Shindler,⁷ JoGayle Howard,⁸ David E. Wildt,⁹ Linda M. Penfold,⁹ Jeffrey A. Hostetler,¹⁰ Madan K. Oli,¹⁰ Stephen J. O'Brien^{1†}

The rediscovery of remnant Florida panthers (*Puma concolor coryi*) in southern Florida swamplands prompted a program to protect and stabilize the population. In 1995, conservation managers translocated eight female pumas (*P. c. stanleyana*) from Texas to increase depleted genetic diversity, improve population numbers, and reverse indications of inbreeding depression. We have assessed the demographic, population-genetic, and biomedical consequences of this restoration experiment and show that panther numbers increased threefold, genetic heterozygosity doubled, survival and fitness measures improved, and inbreeding correlates declined significantly. Although these results are encouraging, continued habitat loss, persistent inbreeding, infectious agents, and possible habitat saturation pose new dilemmas. This intensive management program illustrates the challenges of maintaining populations of large predators worldwide.

Pumas (also called cougars, mountain lions, or panthers) are currently distributed throughout western North America and much of

Central and South America (1). The endangered Florida panther (listed in 1967, table S1), the last surviving puma subspecies in eastern North Amer-

emag.org SCIENCE VOL 329 24 SEPTEMBER 2010

Science Perspectives (commentary) article

GENETICS

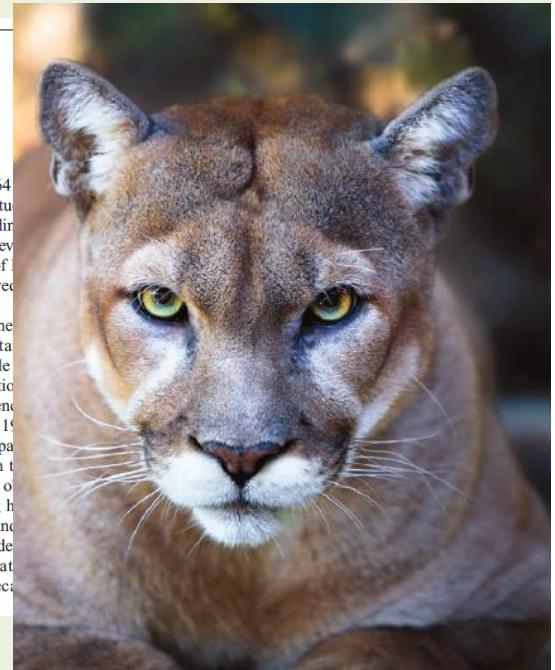
A Bit of Texas in Florida

Craig Packer

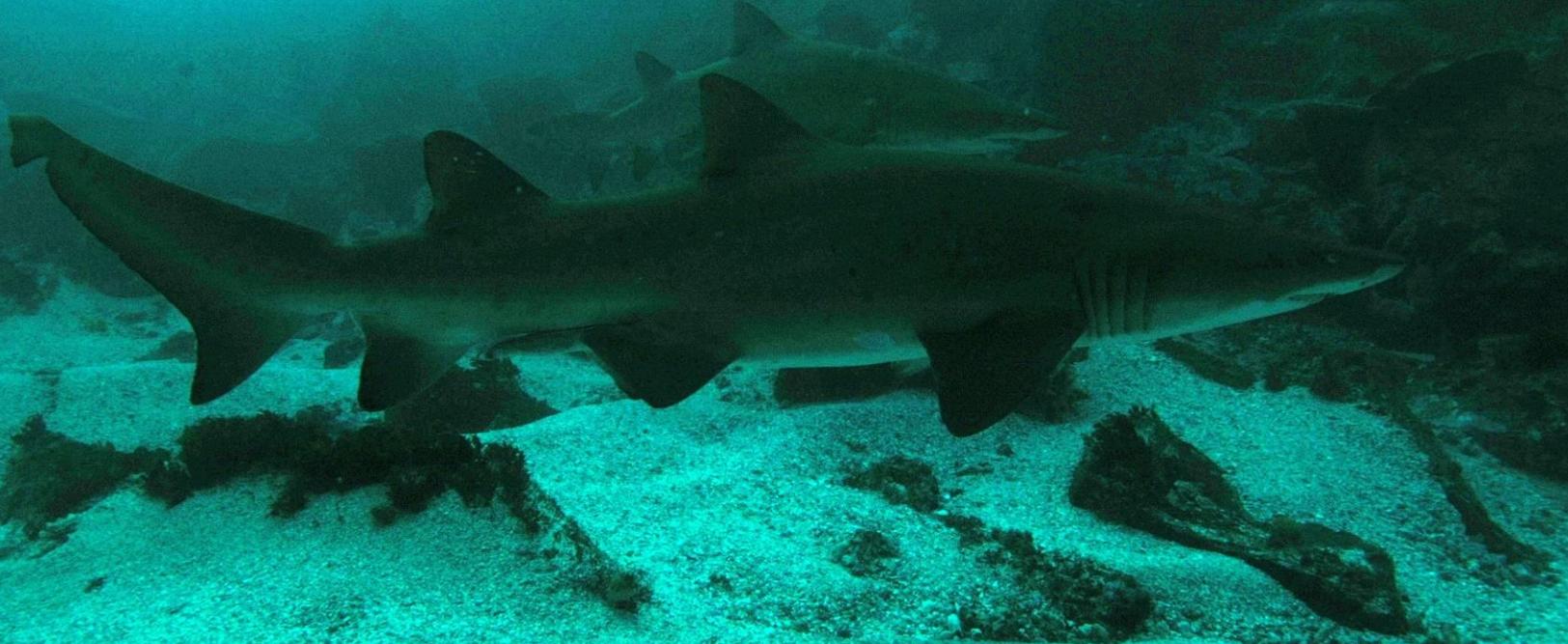
Harrassed, hunted, and restricted to ever smaller areas, most populations of large carnivores are fragmented into archipelagoes of parks and reserves. Biologists have long warned of the negative genetic consequences of inbreeding in such small populations. To restore genetic health, they have prescribed “active management,” including moving, or translocating, individuals into inbred populations. In a time of budget cuts and inadequate funding for effective conservation, however, is translocation worth the costs? Moving a lion from Namibia to South Africa is not a trivial exercise, nor is the translocation of cougars from one part of the United States to another. But it may be worth the trouble, Johnson *et al.* (1)

report on page 164 comprehensive study of the effects of inbreeding find convincing evidence of a population of panthers successfully improved theirs from Texas.

Florida panthers, pumas, or mountain lions in considerable genetic variation. By the early 1990s, of 20 to 25 adult panthers, genetic variation was low. Biologists often cite problems—including high quality, poor fecundity, and competition with one or no deer—leading to predictions that the panther could go extinct within decades.



Research Themes: A/Prof Stow

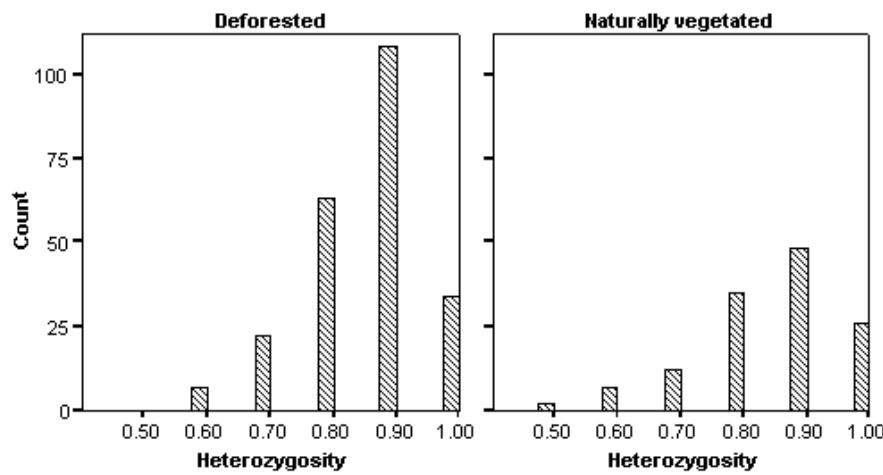


Endangered Species Management

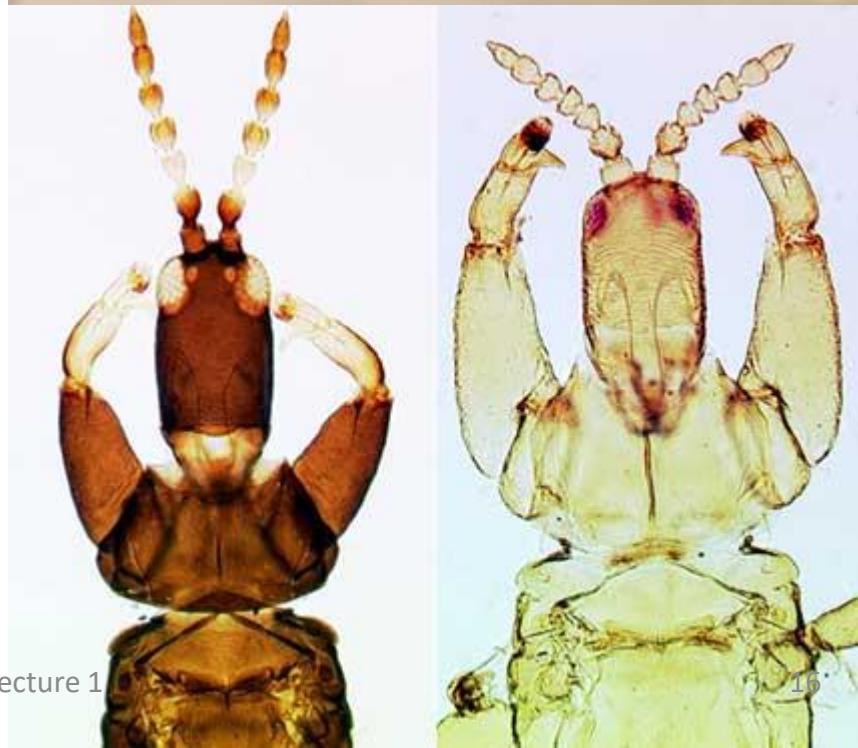
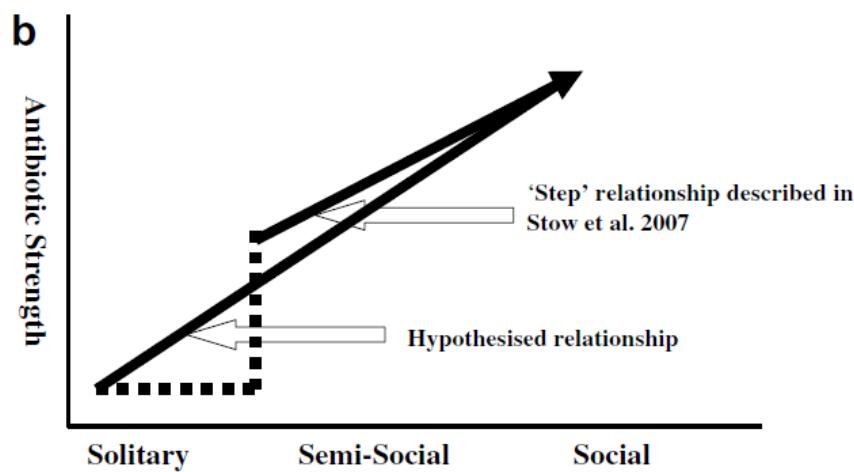
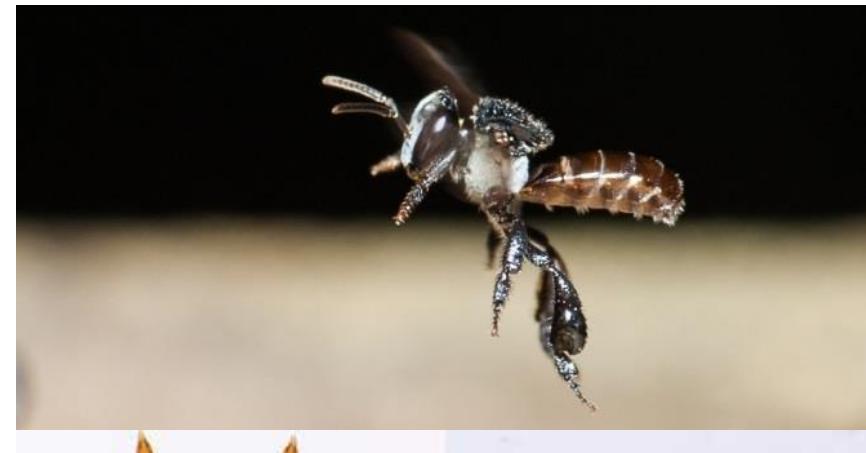
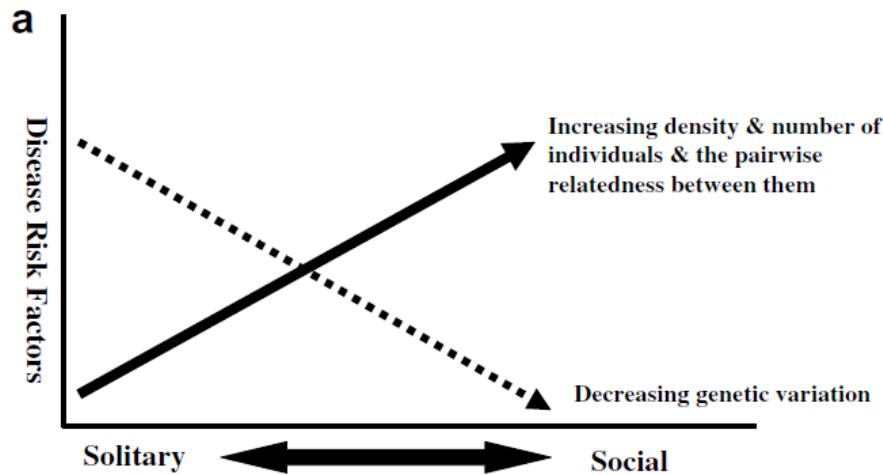
Climate change Impacts



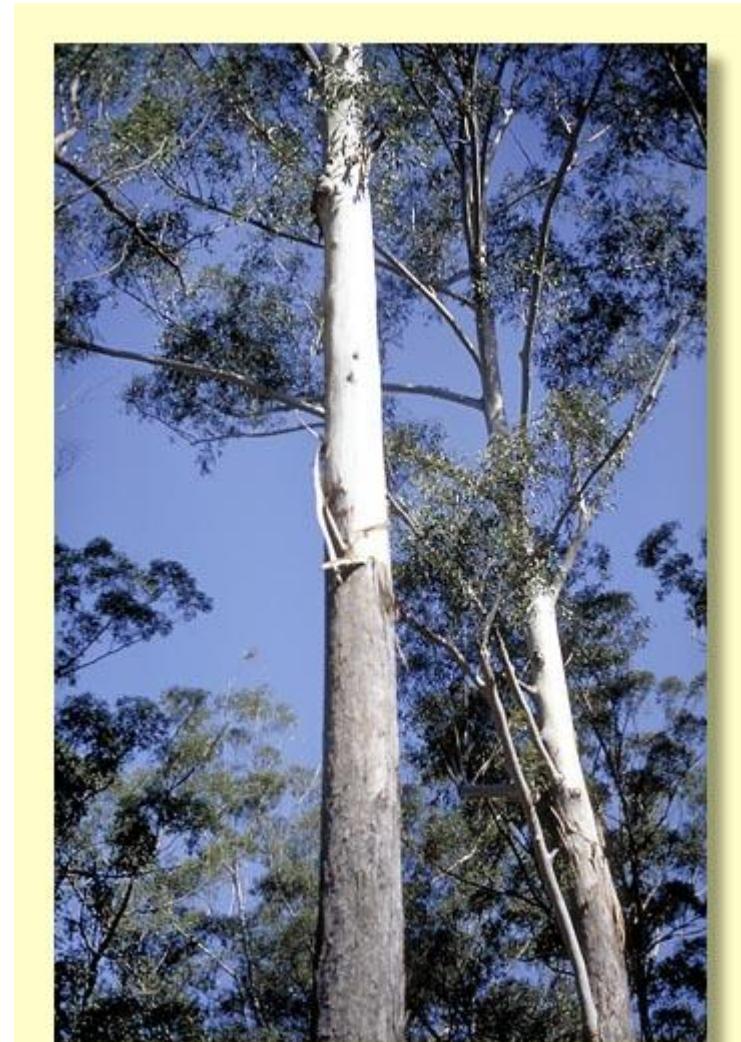
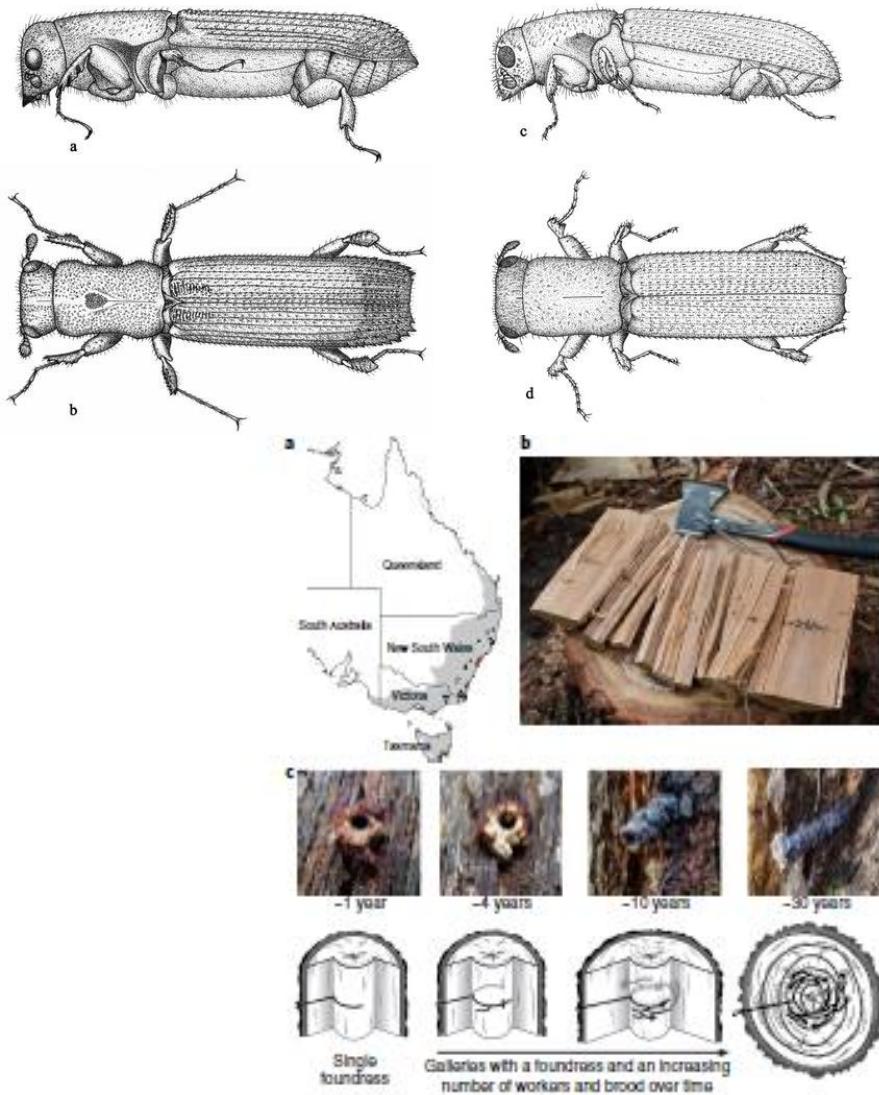
Habitat Fragmentation



Disease and evolution of sociality



Evolution of Sociality



The ‘sixth extinction’



Categories formulated by IUCN - International Union for the Conservation of Nature (www.iucn.org)



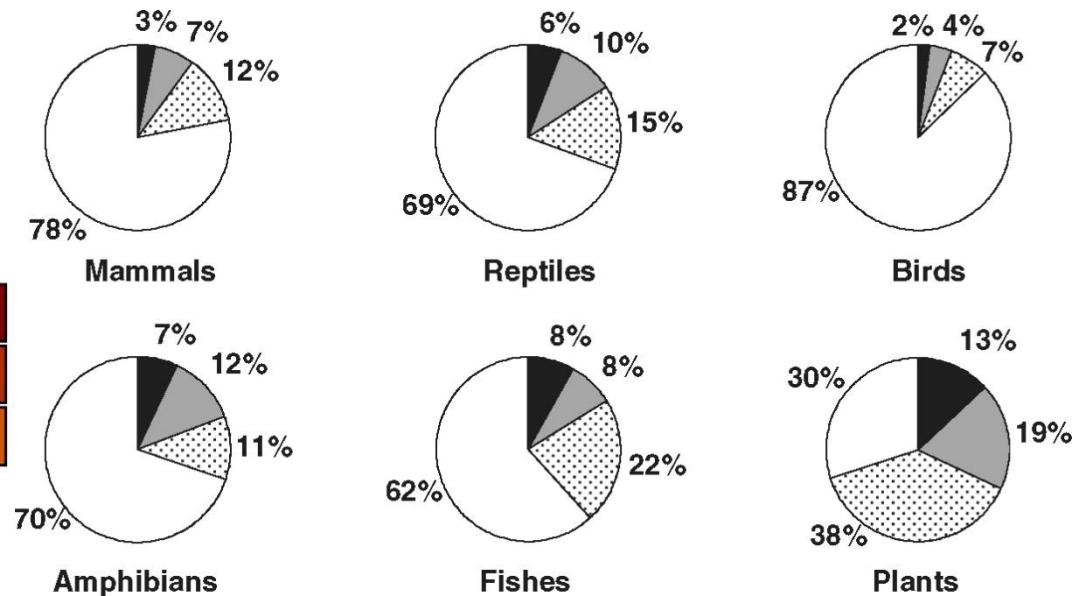
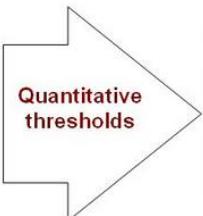
cd: conservation-dependent
nt: near threatened
lc: least concern

CRITERIA

- Population reduction
- Restricted geographic range
- Small population size & decline
- Very small or restricted population
- Quantitative analysis

THREATENED CATEGORIES

- Critically Endangered (CR)
- Endangered (EN)
- Vulnerable (VU)



The ‘sixth extinction’ – www.iucn.org



Categories formulated by IUCN - International Union for the Conservation of Nature (www.iucn.org)



The screenshot shows the IUCN Red List homepage for 2013.1. The top navigation includes links for About, Initiatives, News, Photos, Partners, Sponsors, Resources, Login, FAQ, Contact, Terms of use, and IUCN.org. A search bar allows users to enter Red List search terms and includes a 'GO' button and 'OTHER SEARCH OPTIONS'. A 'DONATE NOW!' button is also present. On the left, a circular diagram illustrates the IUCN Red List categories: LEAST CONCERN (LC), NEAR THREATENED (NT), VULNERABLE (VU), ENDANGERED (EN, highlighted in red), CRITICALLY ENDANGERED (CR), EXTINCT IN THE WILD (EW), and EXTINCT (EX). A large image of a green frog, identified as 'QUYET'S TREEFROG' (*Gracixalus quyeti*), is displayed in a circular frame. A sidebar on the left provides a user guide to the website, news articles about threatened freshwater fish and pangolin trade, and a link to the most traded wild mammal.

The IUCN Red List of Threatened Species™ 2013.1

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[About](#) [Initiatives](#) [News](#) [Photos](#) [Partners](#) [Sponsors](#) [Resources](#)

Enter Red List search term(s) [OTHER SEARCH OPTIONS](#) [Discover more](#)

[DONATE NOW!](#)

LEAST CONCERN LC NEAR THREATENED NT VULNERABLE VU ENDANGERED EN CRITICALLY ENDANGERED CR EXTINCT IN THE WILD EW EXTINCT EX

A users' guide to The IUCN Red List web site
03 April 2009 - In October 2008, the IUCN Red List web site was given a brand new look. The new site has more functionality than ever before. This also means that the site has more detailed search pages that... [more](#)

Threatened freshwater fish fall through net of mismanaged aquarium pet trade
26 July 2013 - The global trade in freshwater fish is a large and diverse industry, estimated to be worth around US \$15–30 billion a year. Supplied by captive-bred and wild-caught specimens, the... [more](#)

The most traded wild mammal - the Pangolin - is being eaten to

QUYET'S TREEFROG
Gracixalus quyeti

© Ralf Hendrix

The ‘sixth extinction’ – www.iucn.org

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The IUCN Red List of Threatened Species™ 2013.1

Enter Red List search term(s) OTHER SEARCH OPTIONS Discover more

DONATE NOW!

The IUCN Red List of Threatened Species™ 2013.1

LEAST CONCERN NEAR THREATENED VU LC NT

A users' guide to 03 April 2009 - In O new look. The new means that the site

Threatened fresh pet trade 26 July 2013 - The estimated to be w captive-bred and wil

The most traded v

Keywords

Taxonomy

ANIMALIA

FUNGI

PLANTAE

PROTISTA

Location

Systems

Habitat

Threats

Assessment

Life History

Taxa to show:

Species

Subspecies and varieties

Stocks and subpopulations

Your Search Criteria

No criteria selected

Select a tab on the left and enter text or select options.

Then, press the arrow button to build your search criteria here.

You may select options from multiple tabs.

Your selections must appear in this area to be used in your final search.

Click the "Run Search" button below to run your search using the criteria displayed above.

Run search

The ‘sixth’ extinction & genetics

Threats to extinction:

- Habitat loss
- Over-exploitation
- Alien introductions
- Pollution
- Climate change



The ‘sixth’ extinction & genetics

Threats to extinction:

- Habitat loss
- Habitat fragmentation
- Over-exploitation
- Invasive species
- Pollution
- Climate change



The ‘sixth’ extinction & genetics

Threats to extinction:

- Habitat loss
- Over-exploitation
- Alien introductions
- Pollution
- Climate change



- Small population sizes **amplify the importance of genetic factors** for extinction
- Small population sizes typify **captive populations**

The ‘sixth’ extinction & genetics

Reduced pop size

Fragmentation

Conservation Genetics

Inbreeding

Genetic drift

Inbreeding depression
Loss of fitness

Lost evolutionary
potential

Management
captive/wild pops

Reintroductions

Extinction

Eg: Florida panther

(Cougar, puma, mountain lion)



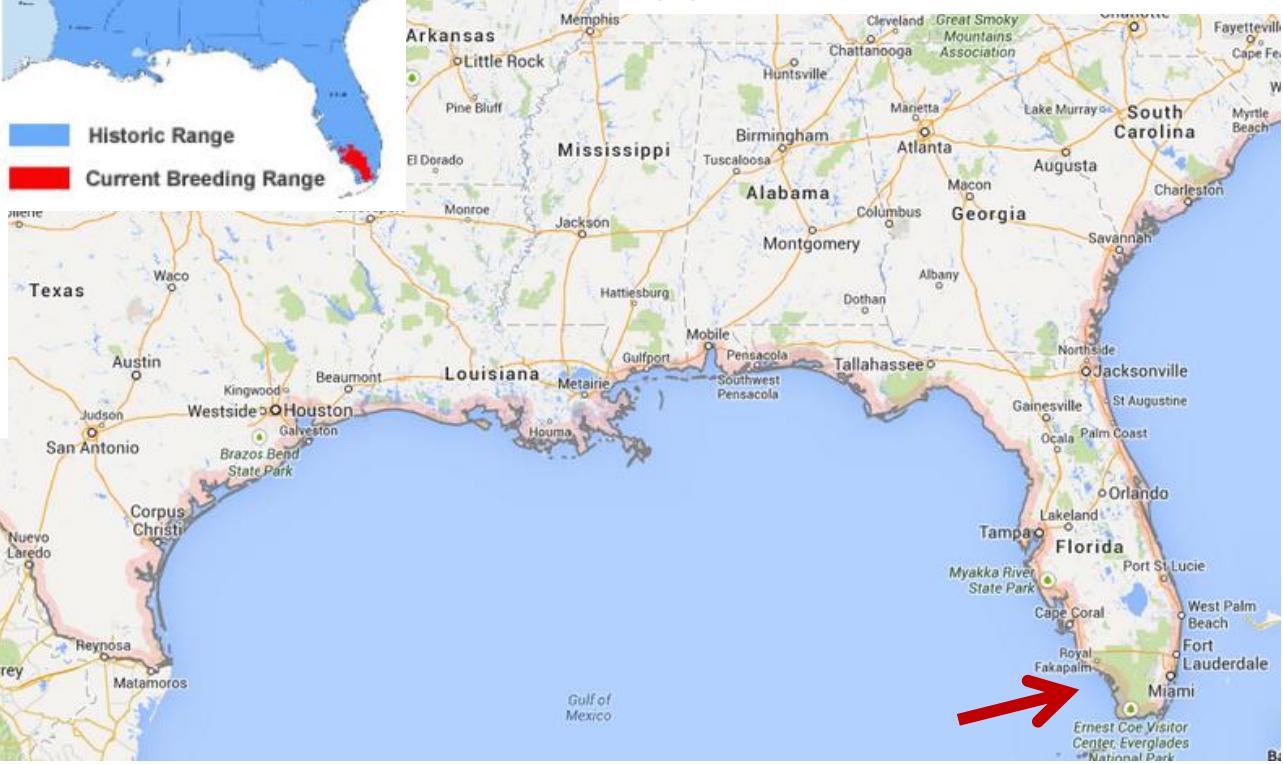
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Example: Florida panther

(Cougar, puma, mountain lion)

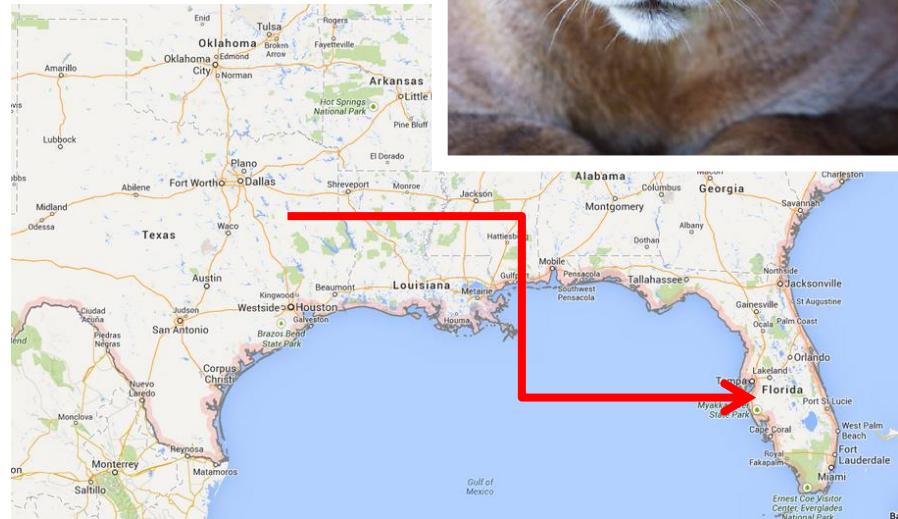
- Early 1990s, FLA pop **N = 20-25**

(including two subgroups – CFP & EVG)

- Low molecular **V_G**

- Signature of **inbreeding:**

- Poor sperm quality
- Low male testosterone levels
- Low fecundity (and recruitment)
- Spinal defects & kinked tails
- High pathogen and parasite load
- **95% Probability of extinction in next 2 decades**



Example: Florida panther

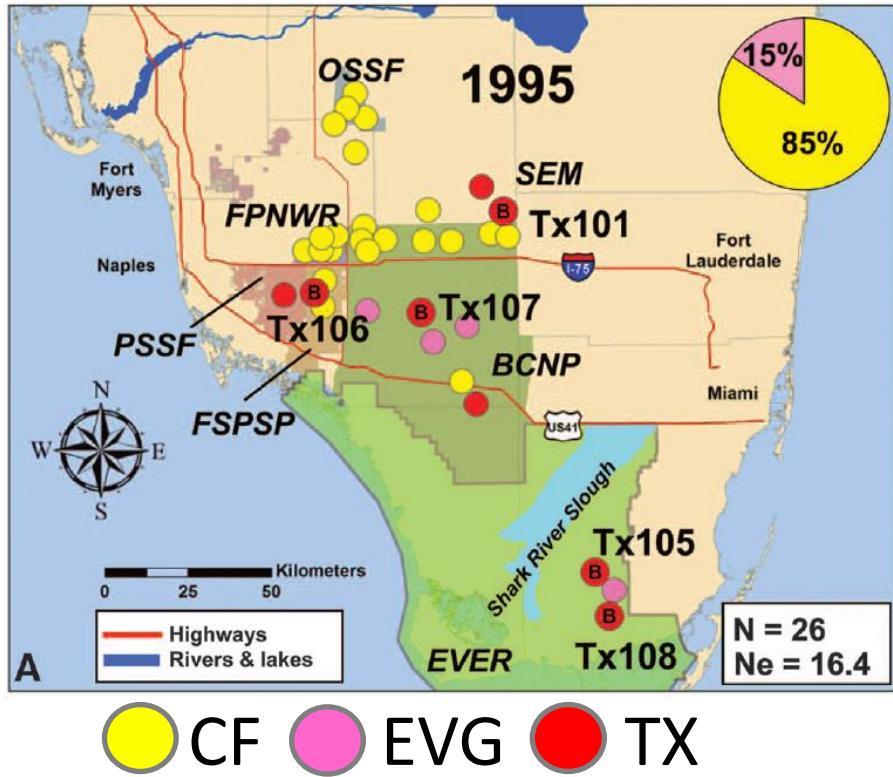
(Cougar, puma, mountain lion)

- Early 1990s, FLA pop **N = 20-25**
(including two subgroups – CFP & EVG)
- **8 TX females** introduced in 1995
- Molecular techniques
(Microsatellites [short tandem repeats]) used to track:
 - Gene flow & dispersal
 - Establishment & mating success
 - Fitness effects of outbreeding



1995

Introduction



N = 26 : Adults (breeding age)

Ne = 16.4: Effective

population size

Pie chart:

Genetic pedigree

CF = Central Florida

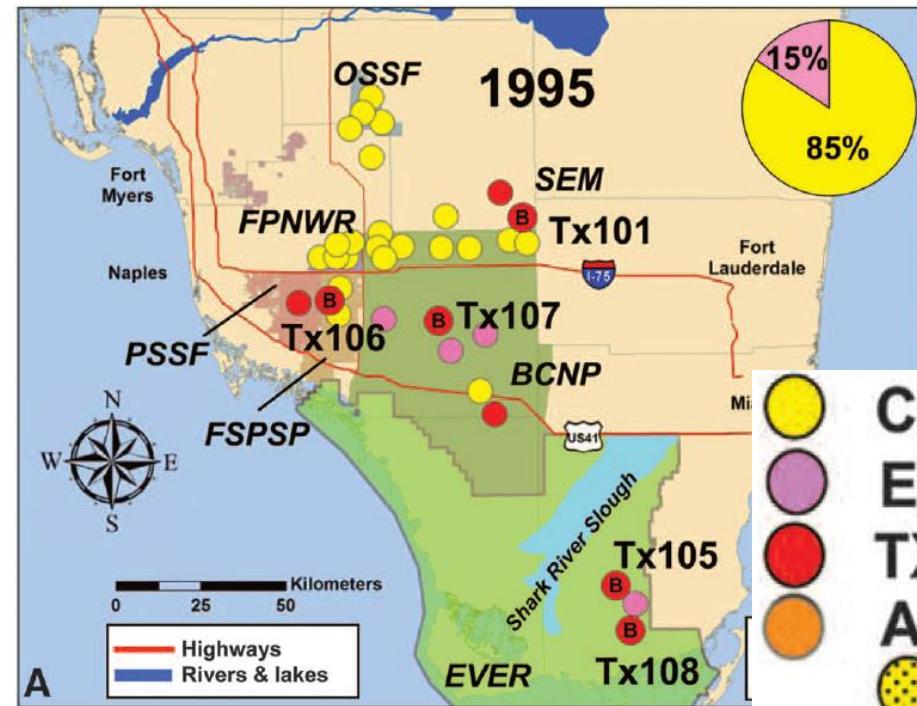
EVG = Everglades

TX = Texas

1995 versus 2007

Introduction

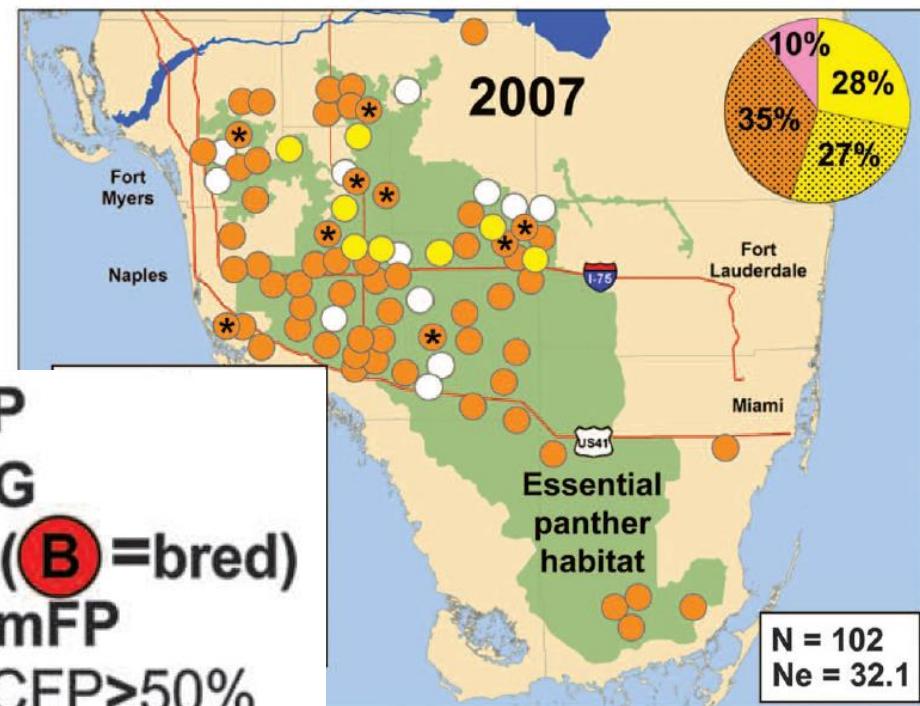
post-introduction



- CFP
- EVG
- TX (B = bred)
- AdmFP
- CFP > 50%
- CFP ≤ 50%

N = 26

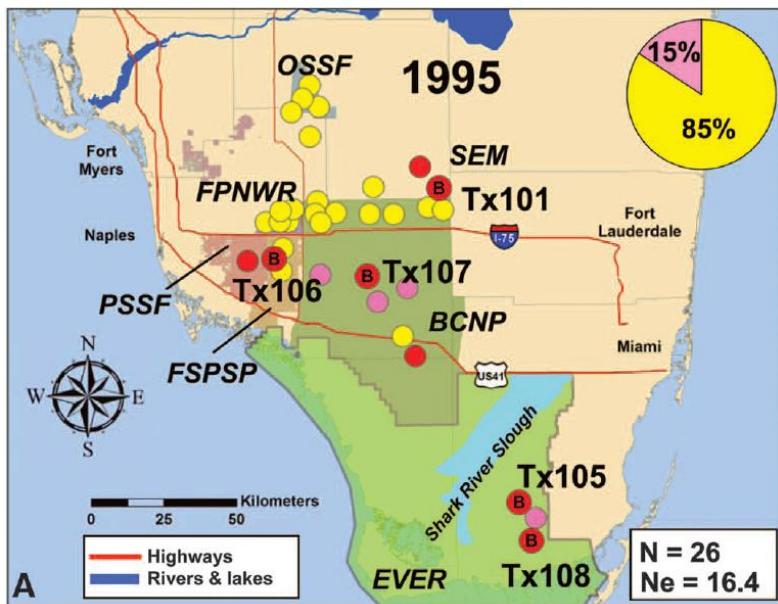
Ne = 16.4



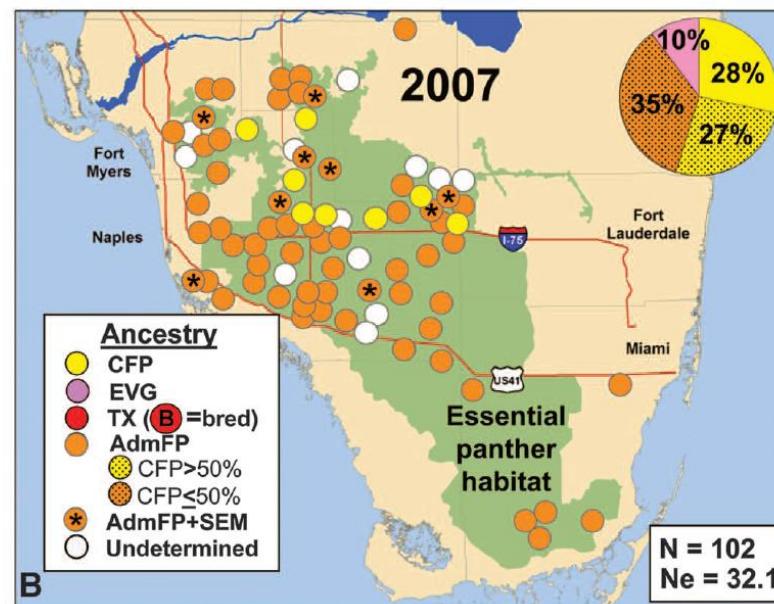
N = 102
Ne = 32.1

1995 versus 2007

Intro



post-intro



Population:

- Increased 14% per year
- Average **heterozygosity** 18 to 25%
- Average age decreased (6.6 to 4.2yrs)



Adm (hybrids) associated with:

- Dispersal and colonisation of new areas
- Increased offspring survival
- Increased competitiveness
- **Individual survival increased with heterozygosity**

Next lecture...

Genetics and extinction

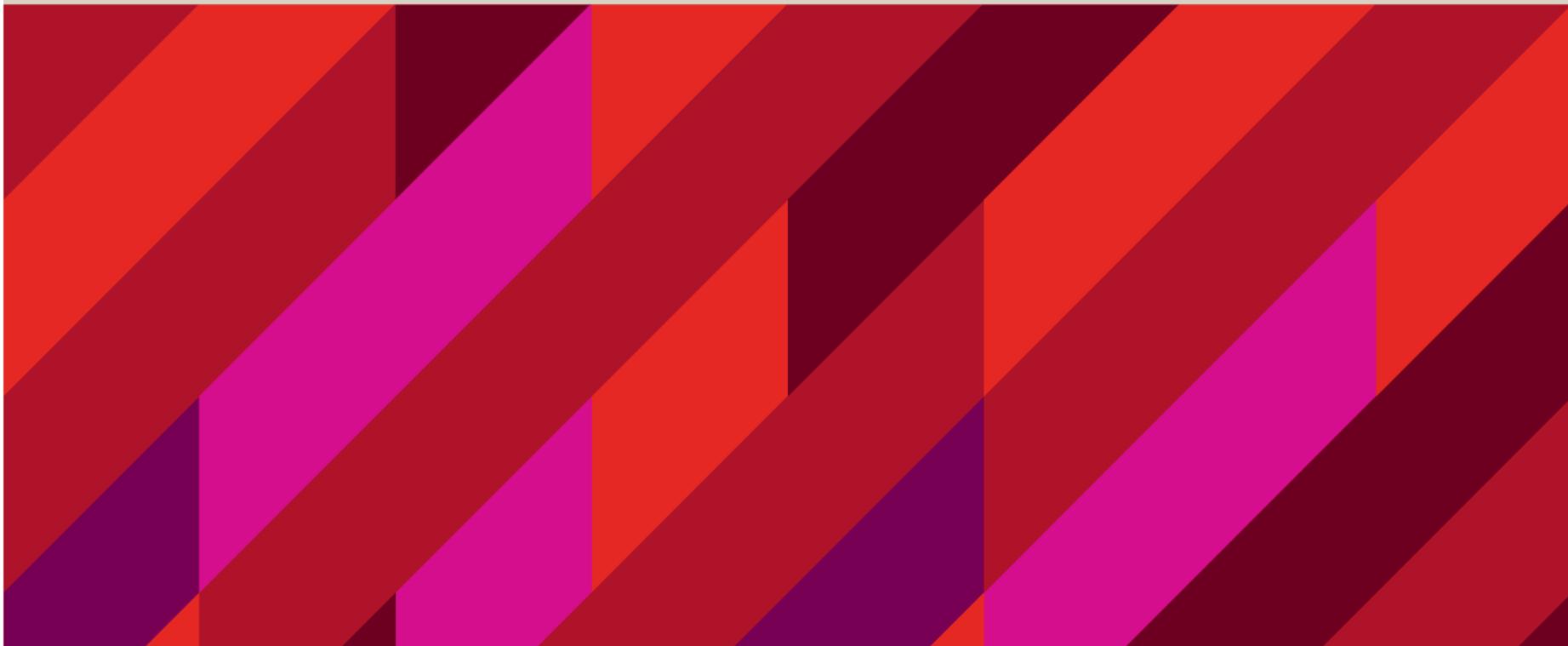




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BIOL3110 Evolutionary and Conservation Genetics

LECTURE 2: GENETICS AND EXTINCTION



Genetics & Extinction

HISTORIC VIEWPOINTS



LANDE (1988) SCIENCE:

“Demographic and environmental fluctuations and catastrophes would drive extinction before genetic factors become important”

HOWEVER:

- Small, inbred populations have low genetic variance (V_G) and are inbred
- Deliberately inbred pops go extinct (in benign lab envs)
- Inbreeding implicated in wild extinctions
- Loss of V_G >> reduced ability to track change via evolution
- Outcrossing rescues small pops (e.g. Florida panther)

Available online at www.sciencedirect.com



Biological Conservation 126 (2005) 131–140

Review

Genetics and extinction

Richard Frankham *

Bioresources, Department of Biological Sciences, Macquarie University, NSW 2109, Australia
Australian Museum, 6 College Street, Sydney, NSW 2090, Australia

Received 2 March 2005
Available online 20 June 2005

BIOLOGICAL
CONSERVATION

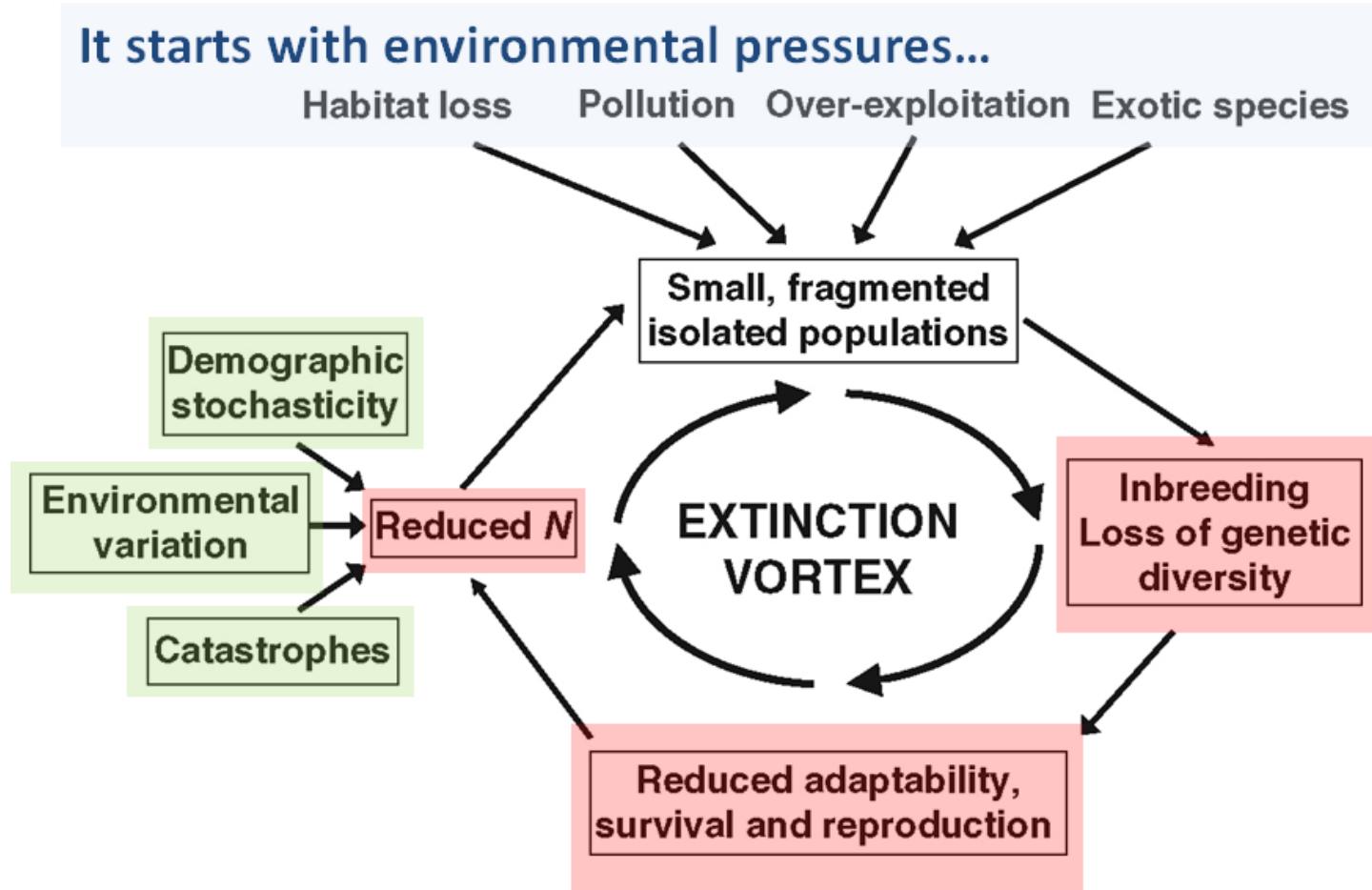
www.elsevier.com/locate/biocon

Genetics & Extinction



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EXTINCTION “VORTEX”



Genetics & Extinction

INBREEDING & INBREEDING DEPRESSION

INBREEDING

Mating between relatives (shared genomes)

INBREEDING DEPRESSION

- Increased homozygosity = greater chance of “exposing” the effects of recessive alleles
- Shown in 90% (141/157) of inbred wild pops (Crnokrak & Roff 1999; see **Box 2.4** Frankham et al)

MUTATIONAL “LOAD”

The sum of rare deleterious mutations that naturally accumulate in populations if recessive;
Effectively ~ “depression potential” 

....covered in detail in later lectures

Genetics & Extinction

INBREEDING CO-EFFICIENT (F)

FOR INDIVIDUALS:

Probability (0...1) of the same allele being inherited through shared ancestry

(or the average expected “genetic overlap” due to shared ancestry)

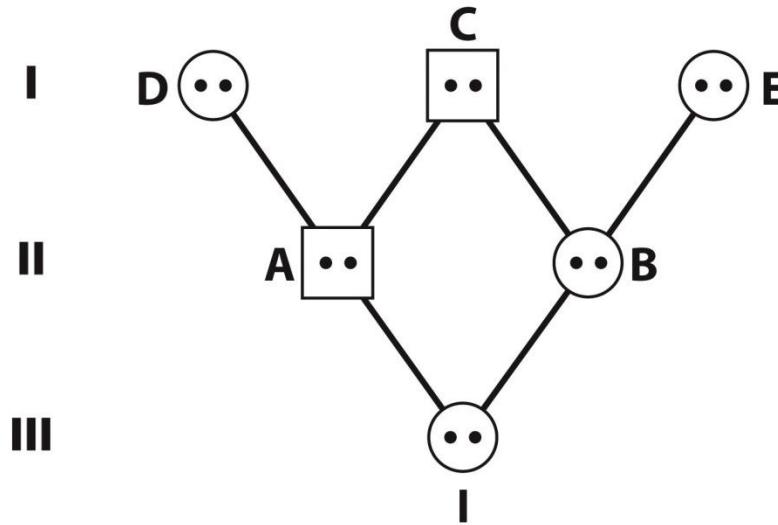
Reaches **0.986** after 20 Gen of full-sib matings (~genetic “isoline”)

Parents	F
Unrelated	0.0
Mother x Son	0.25
Father x daughter	0.25
Siblings	0.25
First cousins	0.0625
Clone (Haploid)	1.0

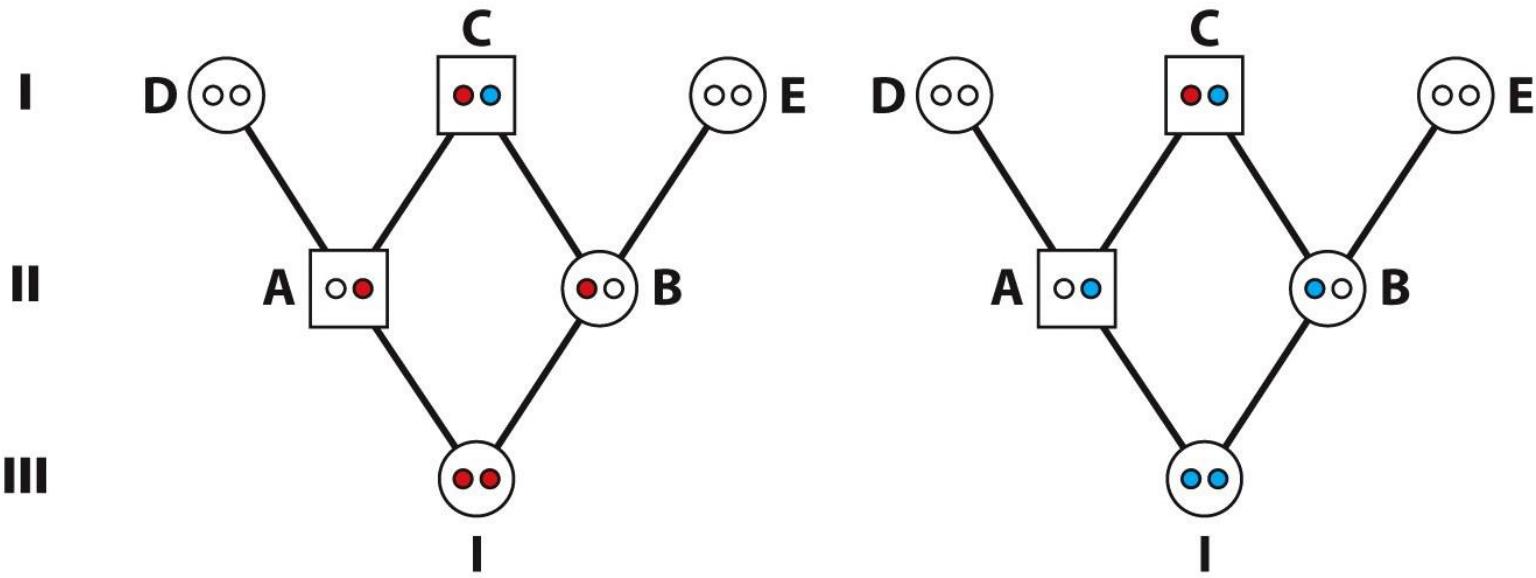
FOR POPULATIONS:

- Averaged across all individuals in a generation
- Can inform differentiation/gene-flow between sub-populations

Calculating F for a Half-Sibling Mating



- 1) Identify the common ancestor(s).
 - C is the common ancestor, so there is one inbreeding loop.
- 2) Count the number of linkages between individuals in each inbreeding loop.
 - The Loop includes C, A, and B; $n=4$
- 3) Calculate $(1/2)^n$ for each loop and sum the results.
 - There is only one loop in this case
 - $(1/2)^4 \times 2 = 1/8$, so $F = 1/8$

**Generation****Case 1****Case 2**

Genetics & Extinction



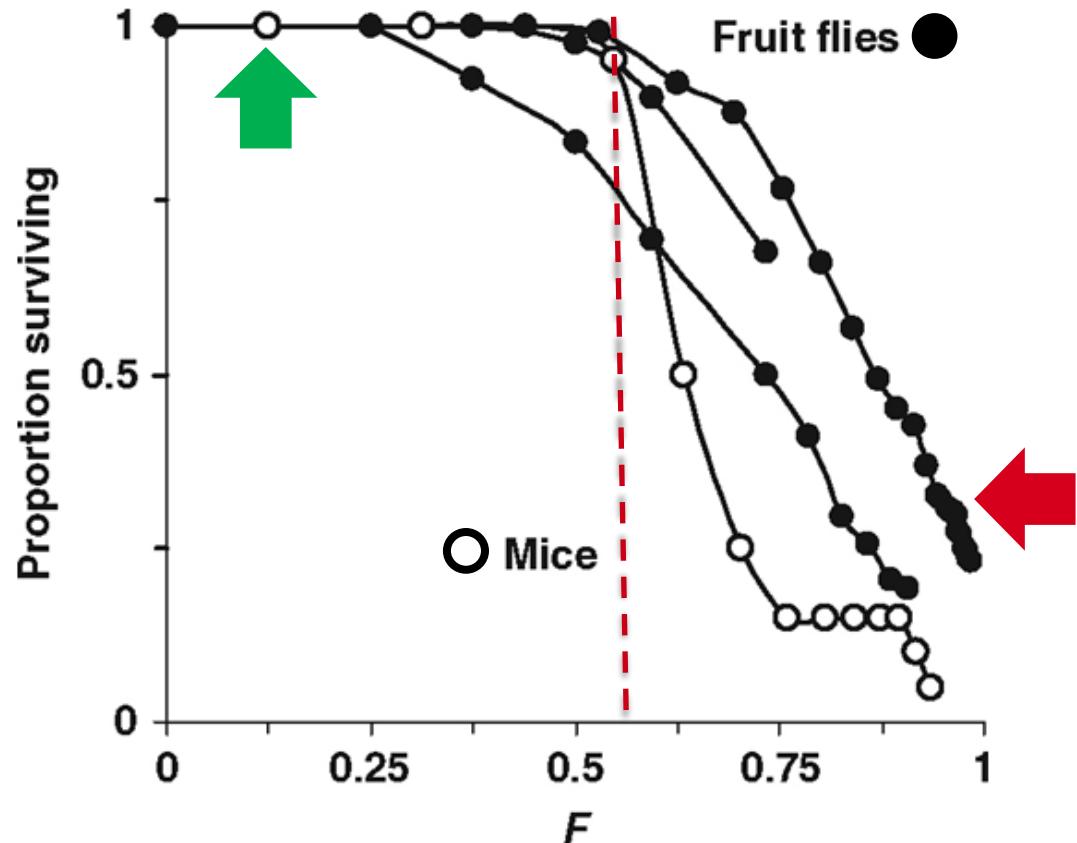
MACQUARIE
University

INBREEDING CAUSES EXTINCTION IN THE LAB

FRANKHAM (2005):

Extinction risk for replicate captive fly & mice under different levels of inbreeding

- A constant & benign environment
- Demographic fluctuations controlled



4 different experiments, multiple lines
Each line = 1 point

Genetics & Extinction

INBREEDING CAUSES EXTINCTION IN THE WILD

DIRECT EVIDENCE

Inbreeding depression, revealed in phenotypes, directly related to extinction

(e.g. observed fitness & recruitment in Fla panther & Japanese quail)

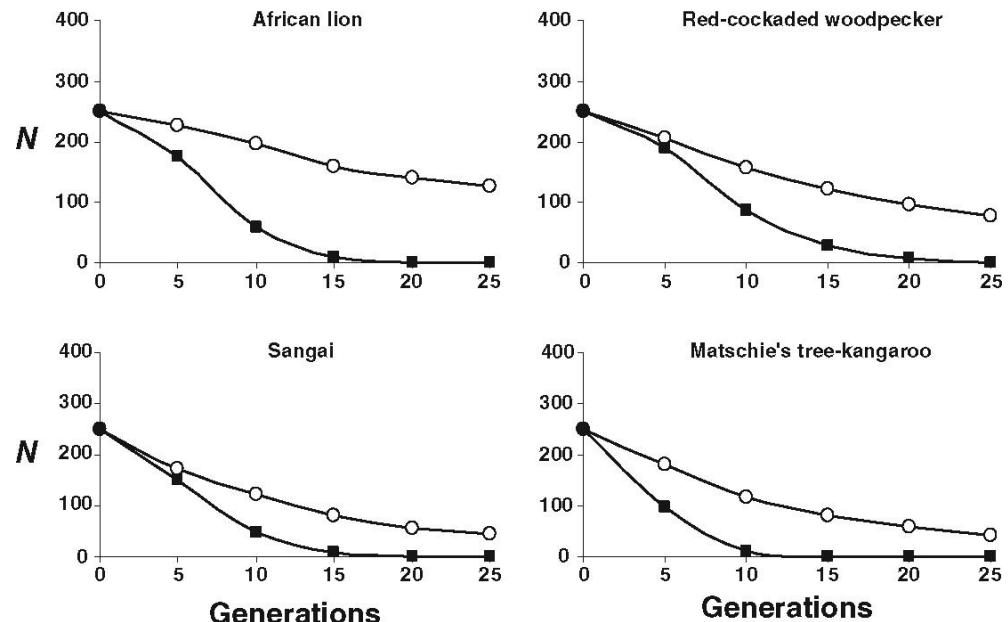


$\text{Pr}(\text{extinction}) = 0.95$ within 20-years

SIMULATION/MODELLING

Simulation based on parameters from actual wild scenarios predict extinction in small populations (due to depression)

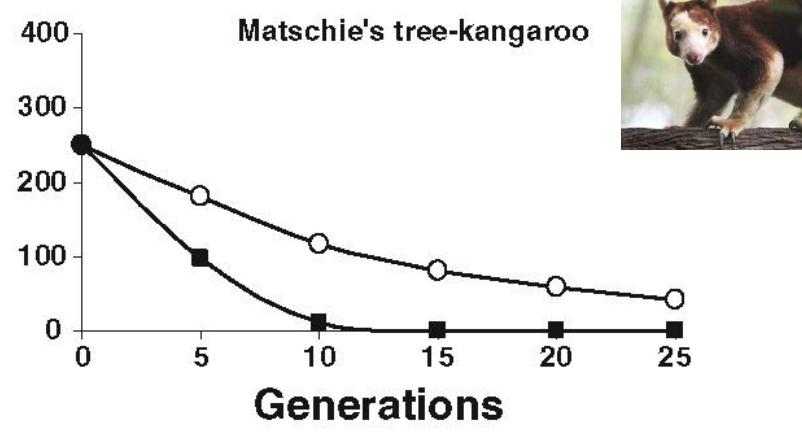
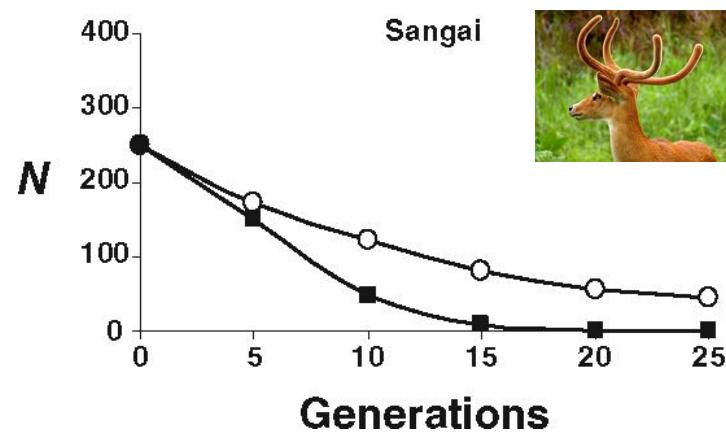
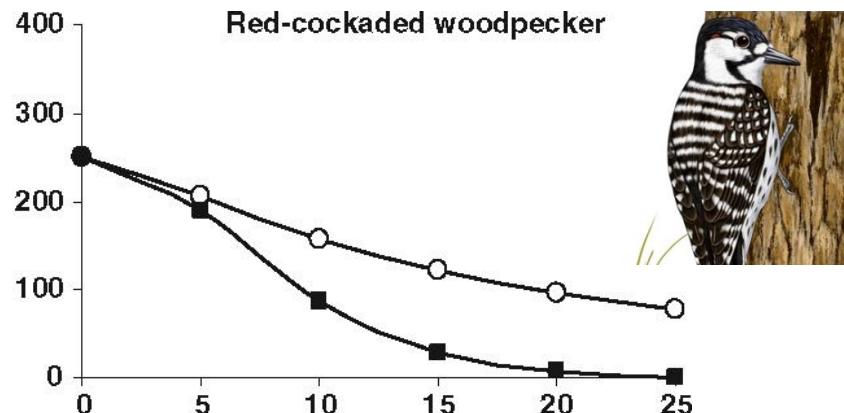
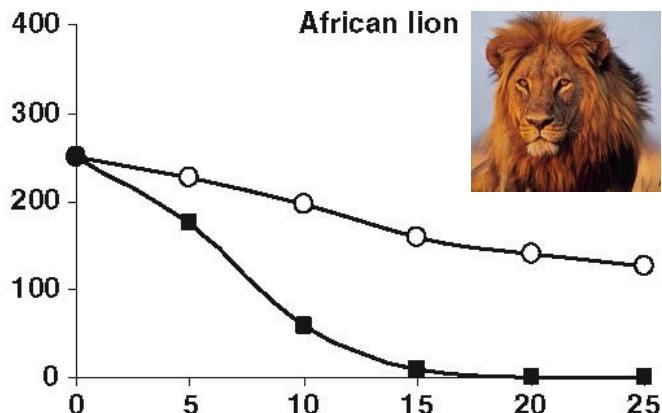
pto...



Genetics & Extinction

INBREEDING CAUSES EXTINCTION IN THE WILD

- Excluding genetic factors (population demography only)
- Accounting for genetics



Underestimate – doesn't account for accumulated inbreeding

Small, isolated populations suffer loss of Vg



Locus	Allele	Mainland	Islands					
			BI	SI	PI	MI	Wil	Wel
Pa297	102	+	—	—	—	—	—	—
	106	+	—	—	—	—	—	—
	118	+	—	—	—	—	—	—
	120	—	—	—	+	—	—	—
	124	+	—	—	—	+	—	—
	128	+	—	+	—	—	+	+
	130	+	—	—	—	—	—	—
	136	+	+	—	—	—	—	—
Pa385	157	+	—	—	—	—	—	—
	159	+	—	—	+	—	+	+
	161	+	—	+	—	—	—	—
	163	+	—	—	—	+	—	—
	165	+	—	—	—	—	—	—
	173	—	+	—	—	—	—	—
Pa593	105	+	—	—	—	—	+	+
	113	—	+	—	—	—	—	—
	123	+	—	—	—	—	—	—
	125	+	—	—	+	—	—	—
	127	+	—	—	—	—	—	—
	129	+	—	—	—	—	—	—
	131	+	—	+	—	—	—	—
	133	+	—	—	—	—	—	—
	135	+	—	—	—	—	—	—

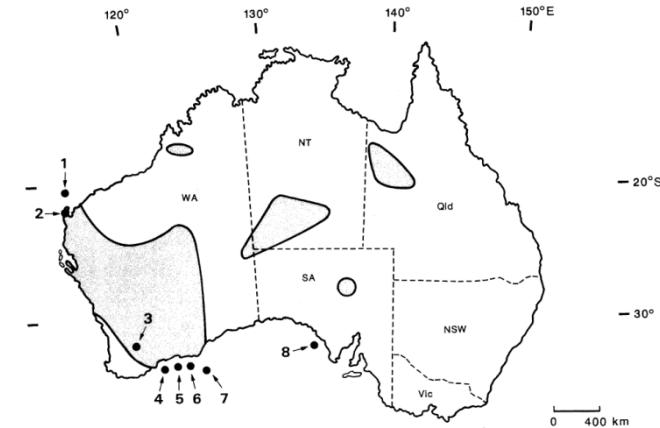
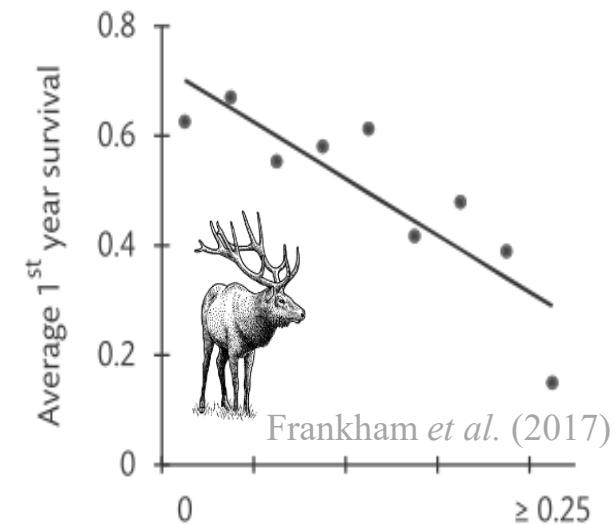
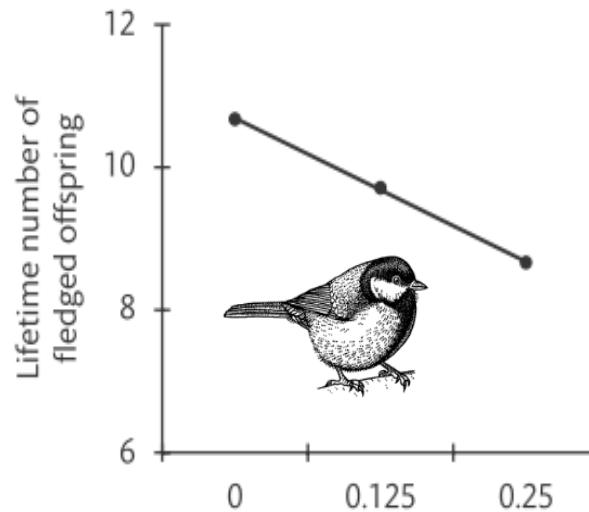
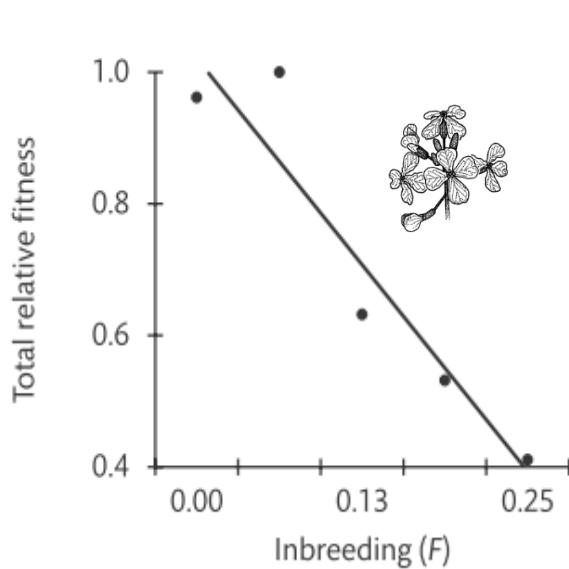


Figure 1. Distribution of the black-footed rock-wallaby (*Petrogale lateralis*) in Australia. Collection localities are Barrow Island (1), Exmouth (2), Wheatbelt (3), Wilson Island (4), Mondrain Island (5), Westall Island (6), Salisbury Island (7), Pearson Island (8).

Small, isolated populations suffer loss of Vg

(1) Inbreeding depression seen in virtually all species that usually outbreed 



Loss of useful genetic variation from populations

These genetic problems cause extinction

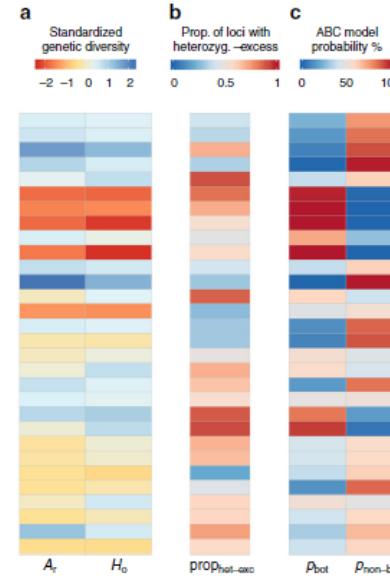
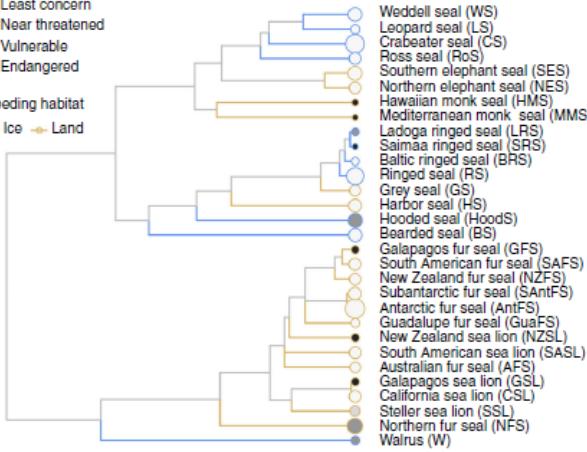
Genetic variation and endangered species



Global abundance
 $\circ 10^3$ $\circ 10^4$ $\circ 10^5$ $\circ 10^6$

IUCN rating
 \circ Least concern
 \circ Near threatened
 \bullet Vulnerable
 \bullet Endangered

Breeding habitat
 ● Ice \rightarrow Land

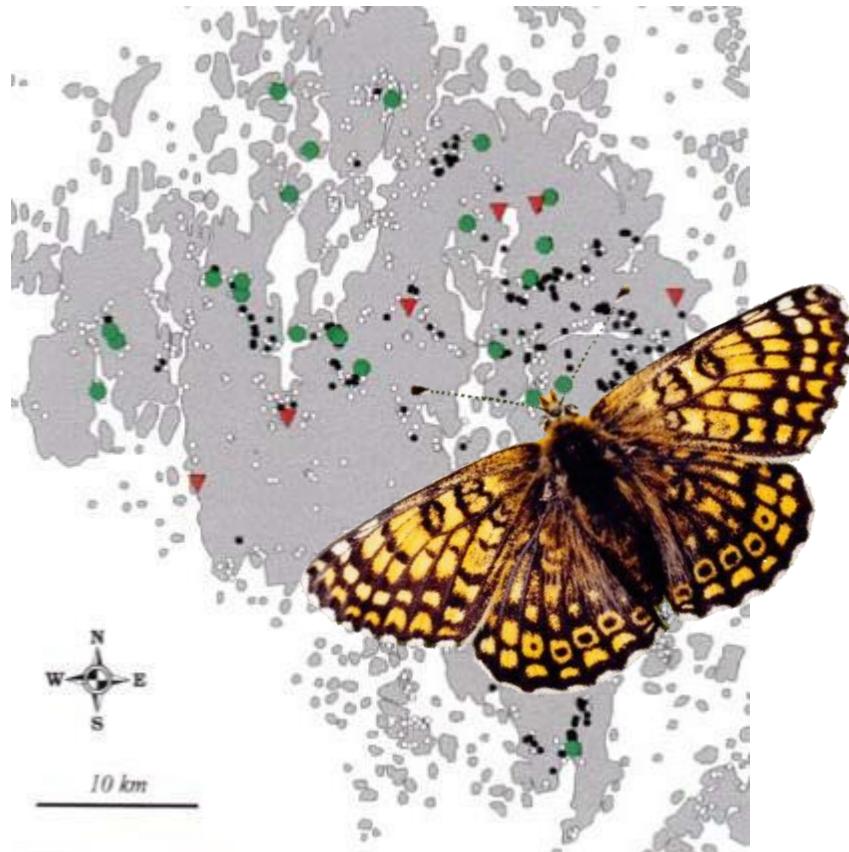


Probability of bottleneck

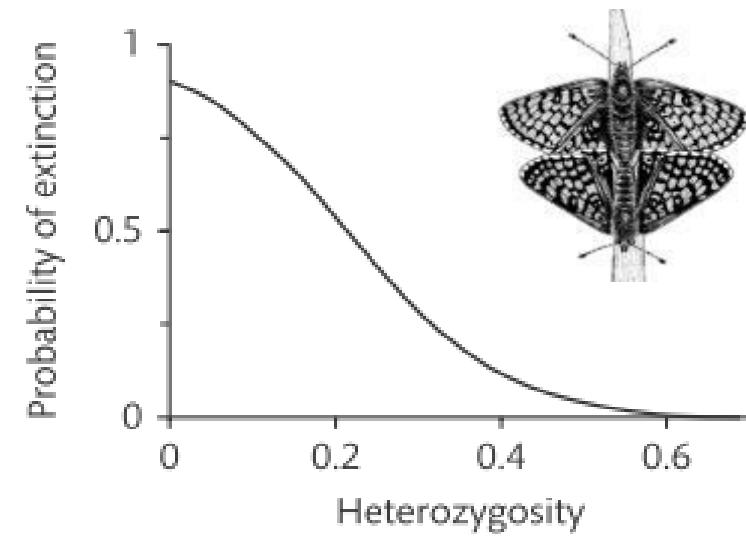
Stoffel et al 2018 Nature Communications

Glanville fritillary butterfly

Saccheri *et al.* 1998 *Nature*

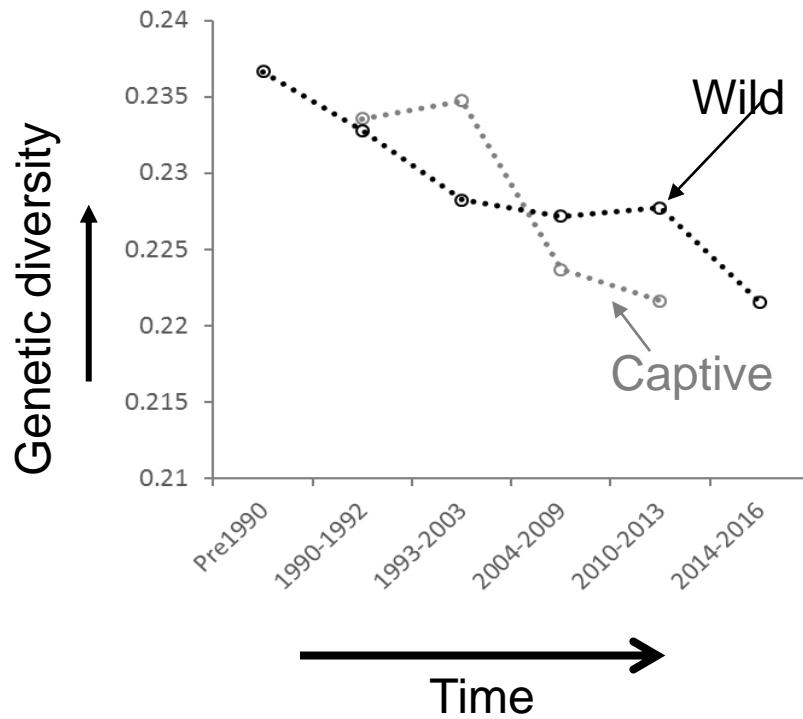


Simple genetic diversity was the best predictor of local extinction



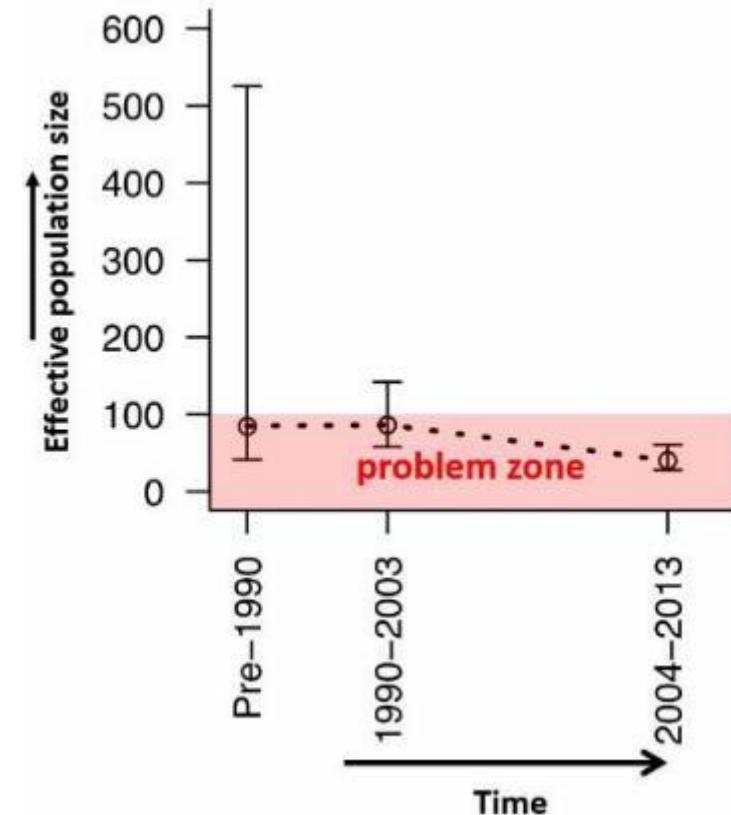
Frankham *et al.* (2017)

Genetic variation and fitness



Harrison et al. 2016
Molecular Ecology

Inbred to a degree that is usually harmful



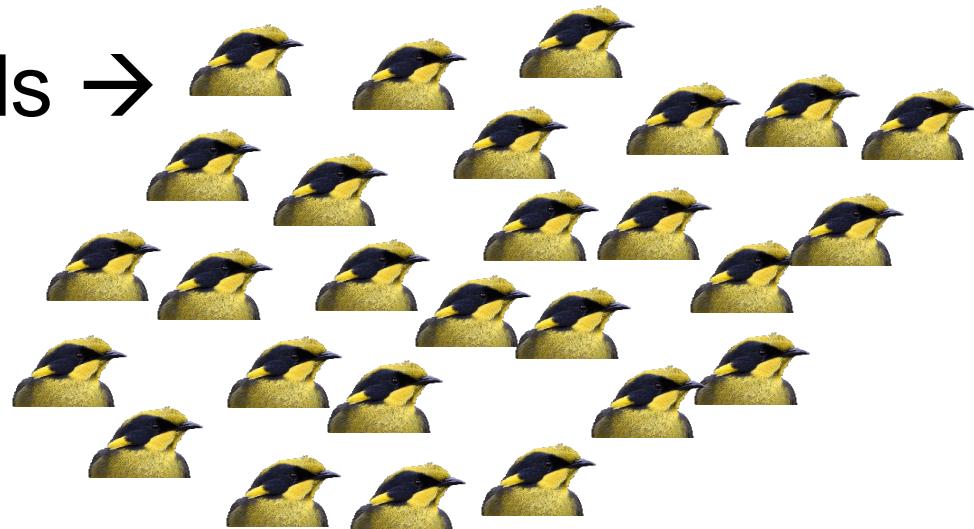
More-inbred birds have far fewer offspring in their lifetimes



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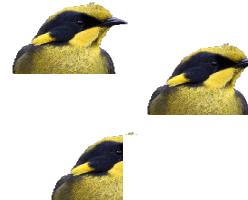
The most outbred birds →

27 fledglings



The most inbred →

2.5 fledglings



Based on 30 years of fitness data (Harrison et al Current Biology)

An example of how connectivity has been influenced the mating system of a plant

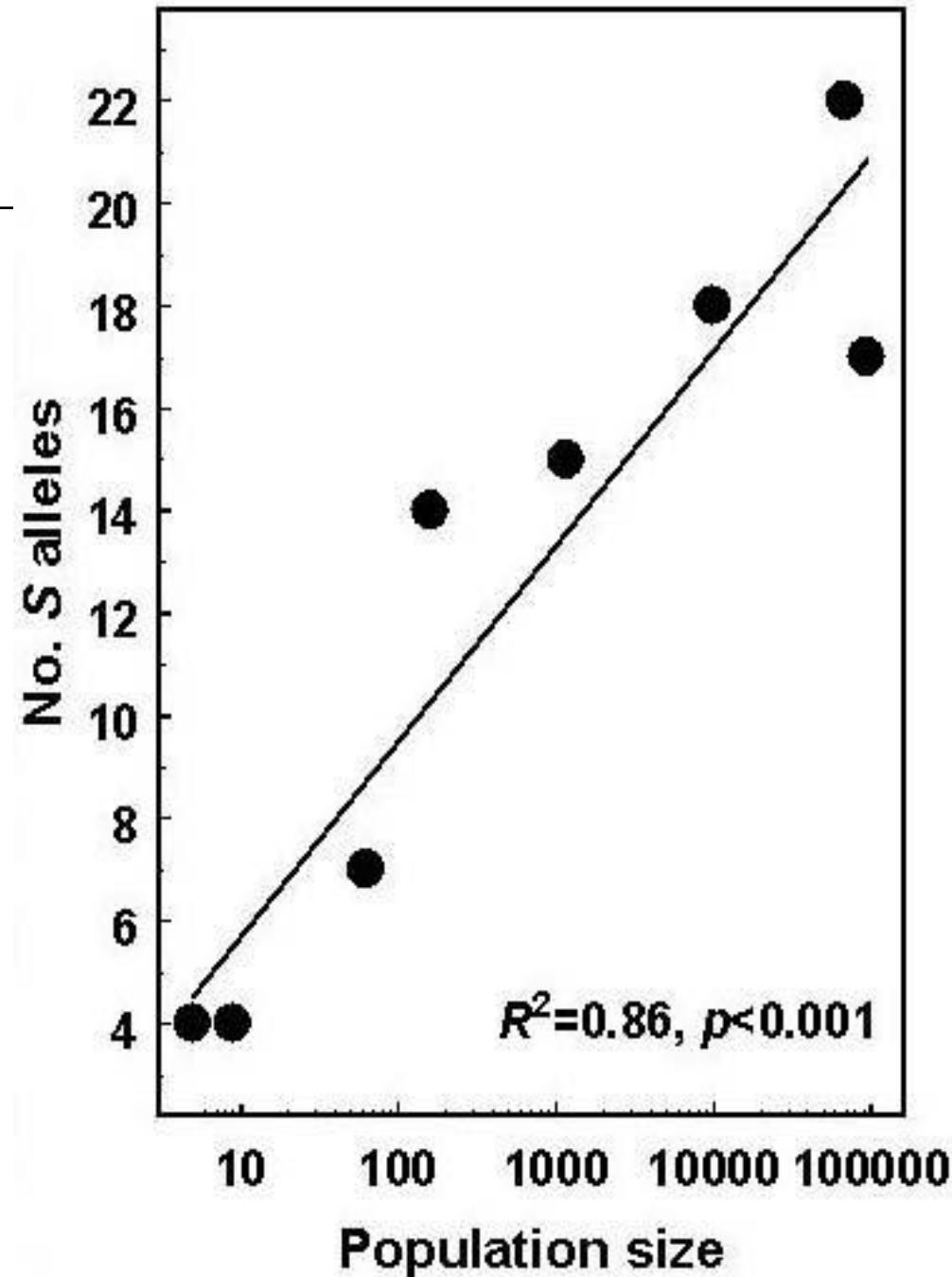
- Button wrinklewort, 24 sites in 2 groups 500Km apart
- Largely self-incompatible i.e. cannot mate with self or very similar individuals – simple genetic control (SI locus)



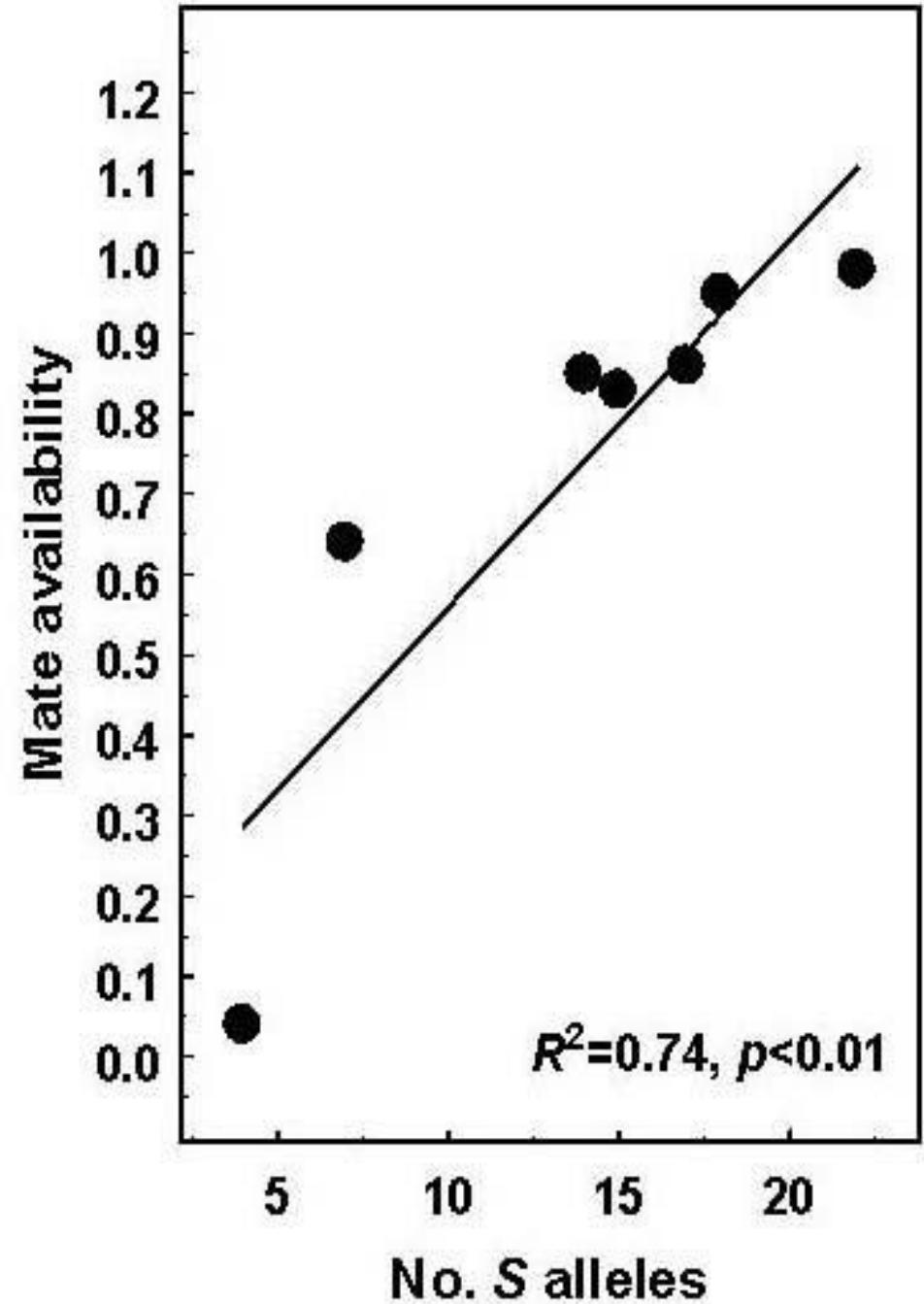
Andrew Young
Button wrinklewort *Rutidosis
leptorrhynchoides*



- genetic erosion - SI alleles lost

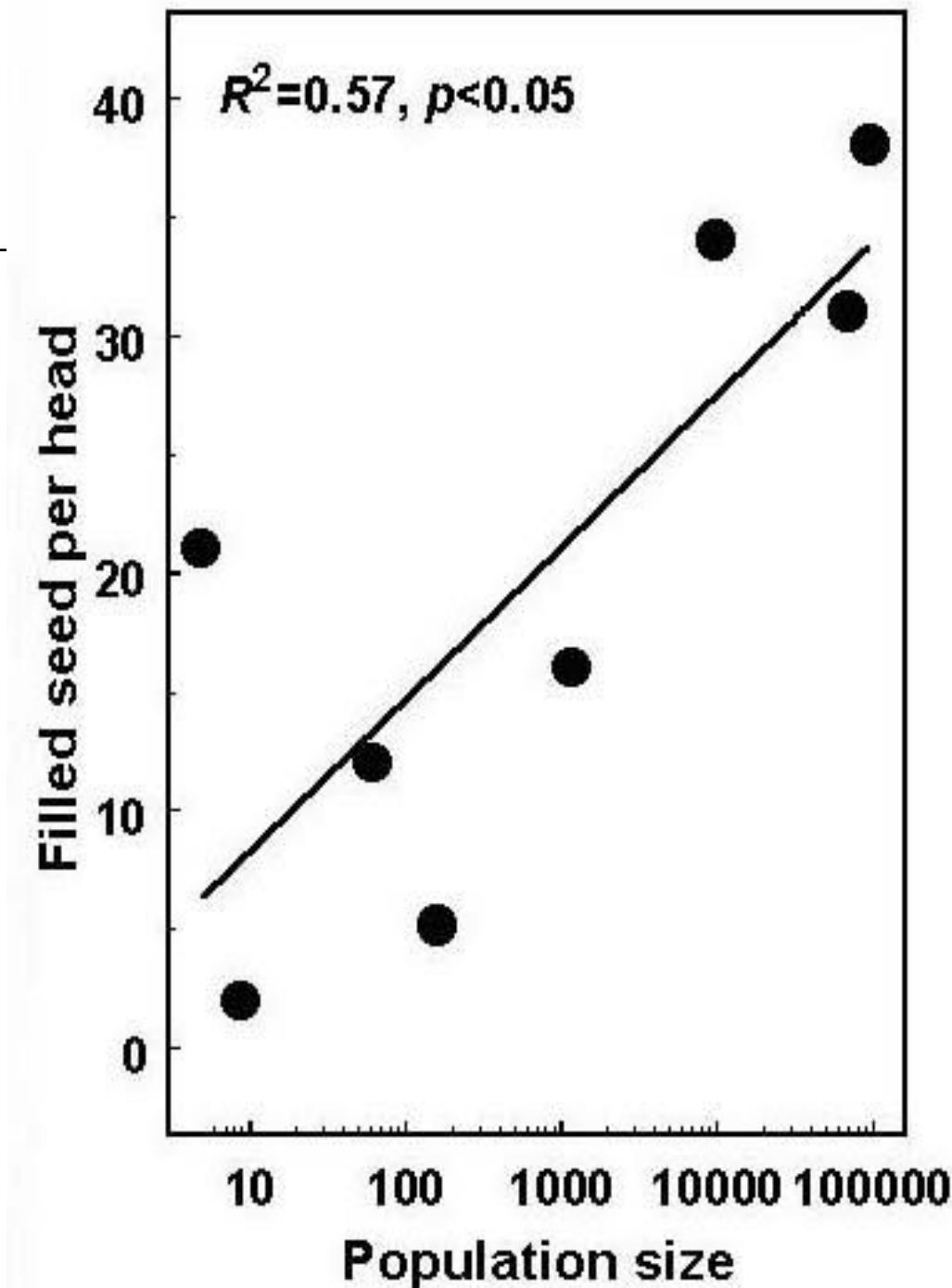


-
- lost SI alleles make many mates incompatible





- mate limitation reduces female success in small populations



Genetics & Extinction

GENETIC VARIANCE

GENETIC VARIANCE or DIVERSITY

- Denoted V_G or $\text{Var}(G)$ or V_A
- Basically the “amount” of genetic difference among individuals of a population/species
- Determines differences among phenotypes (to varying degrees)

HOW WE MEASURE IT

Molecular sequence variation

- Neutral markers via PCRs or functional loci via genomic analyses;

Pedigree-based analyses

- Statistical inference of genetic causality at the phenotypic level



Genetics & Extinction

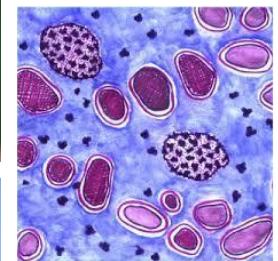
V_G AND ROBUSTNESS



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WHY IS VG SO IMPORTANT?

- (1) It informs the **potential for inbreeding depression**;
- (2) It allows **population persistence** even in extreme environments, buffering:
 - Climatic variation (droughts & flooding rains)
 - Habitat degradation & loss
 - Pollutants, pesticides, herbicides, etc
 - Novel & rapidly evolving pathogens
- (3) It determines the potential for **evolutionary adaptation**.



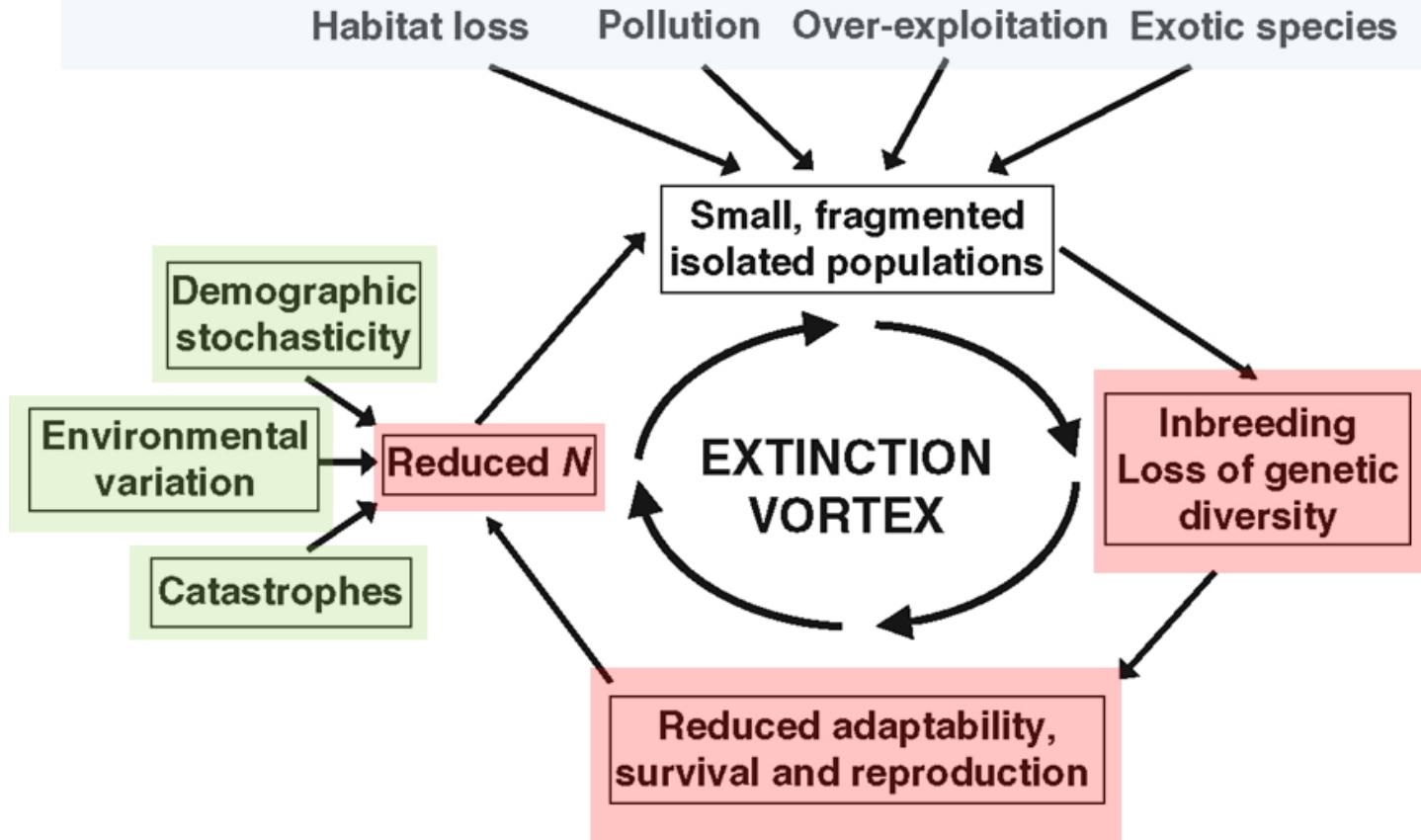
Genetics & Extinction



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EXPLICITLY IN TERMS OF V_G

It starts with environmental pressures...



Genetics & Extinction

FURTHER READINGS (ON Illearn)



Available online at www.sciencedirect.com



Biological Conservation 126 (2005) 131–140

Review

Genetics and extinction

Richard Frankham *

Bioresources, Department of Biological Sciences, Macquarie University, NSW 2109, Australia
Australian Museum, 6 College Street, Sydney, NSW 2090, Australia

BIOLOGICAL CONSERVATION 133 (2006) 42–51



available at www.sciencedirect.com

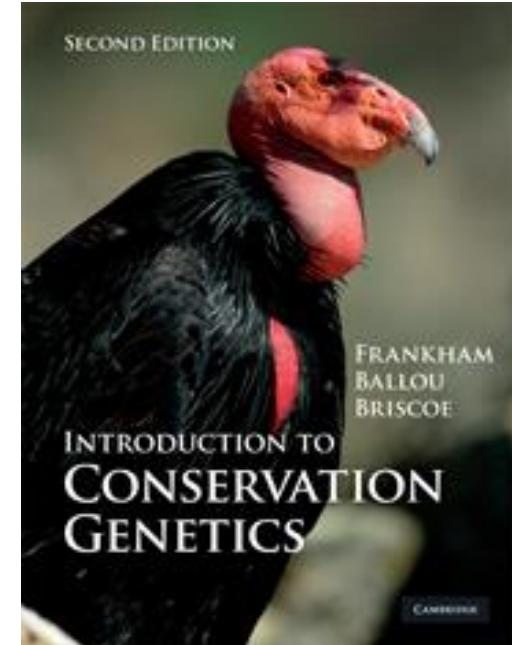


journal homepage: www.elsevier.com/locate/biocon



BIOLOGICAL
CONSERVATION

www.elsevier.com/locate/biocon



Chapter 2

Realistic levels of inbreeding depression strongly affect extinction risk in wild populations

Julian J. O'Grady^{a,*}, Barry W. Brook^b, David H. Reed^{a,1}, Jonathan D. Ballou^{a,2},
David W. Tonkyn^c, Richard Frankham^a

^aBiodiversity and Bioresources Group, Department of Biological Sciences, Macquarie University, Sydney, New South Wales 2109, Australia

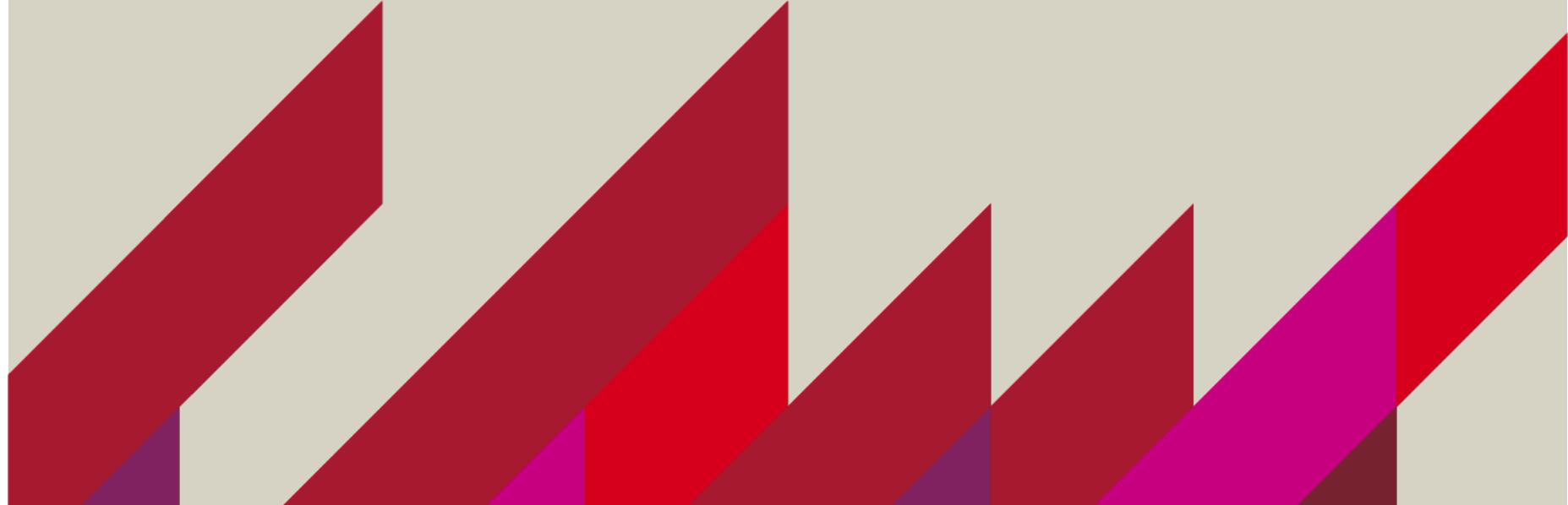
^bSchool for Environmental Research, Institute of Advanced Studies, Charles Darwin University, Darwin, Northern Territory 0909, Australia

^cDepartment of Biological Sciences, Clemson University, SC 29634, USA

Next lecture:



V_G in more detail

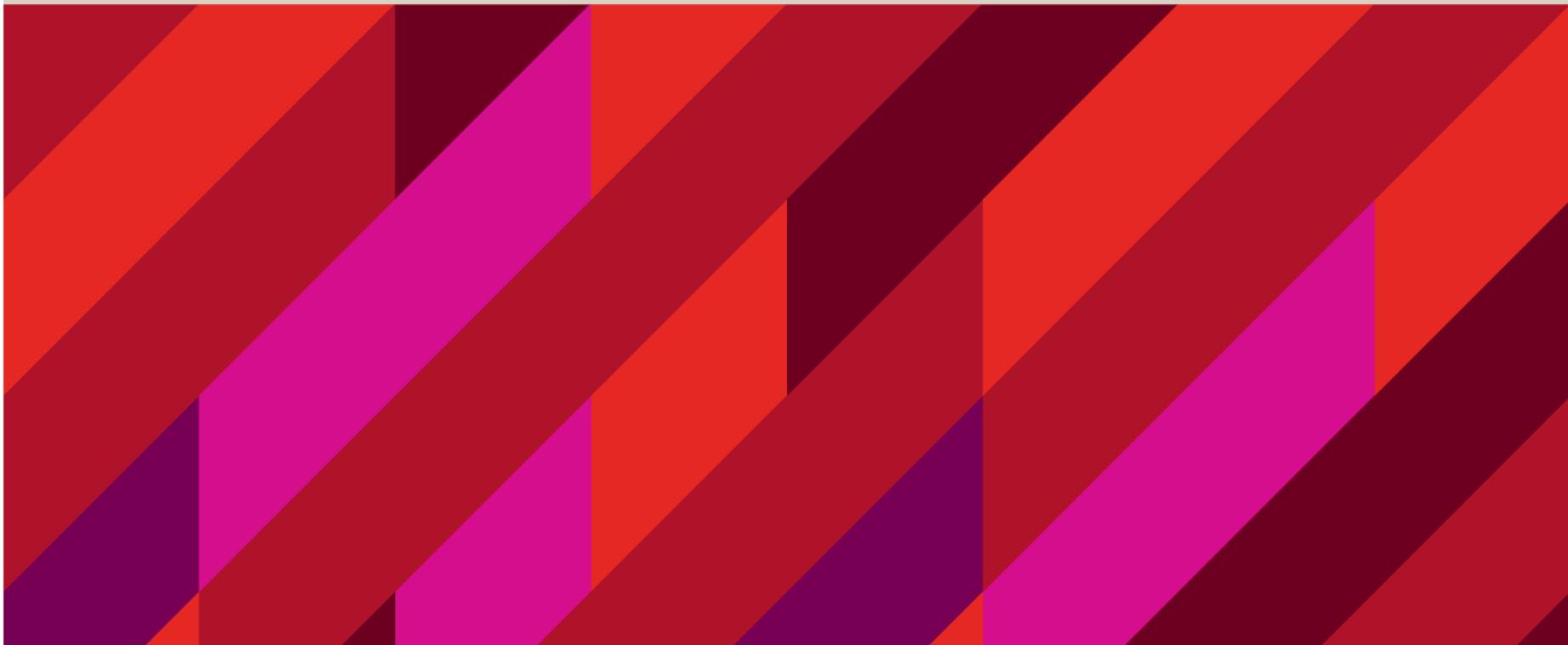




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BIOL3110 Conservation & Ecological Genetics

LECTURE 3: GENETIC VARIATION



Assignment 1: Commentary

WORTH 5 % DUE FRIDAY WEEK 3 (7/3/22) MONDAY
5PM

Criterion	Marks
Get it between 500-600 words	1
Relevance of paper selection	1
Presentation, spelling, grammar	1
Genuine effort to interpret the paper	2



TIPS:

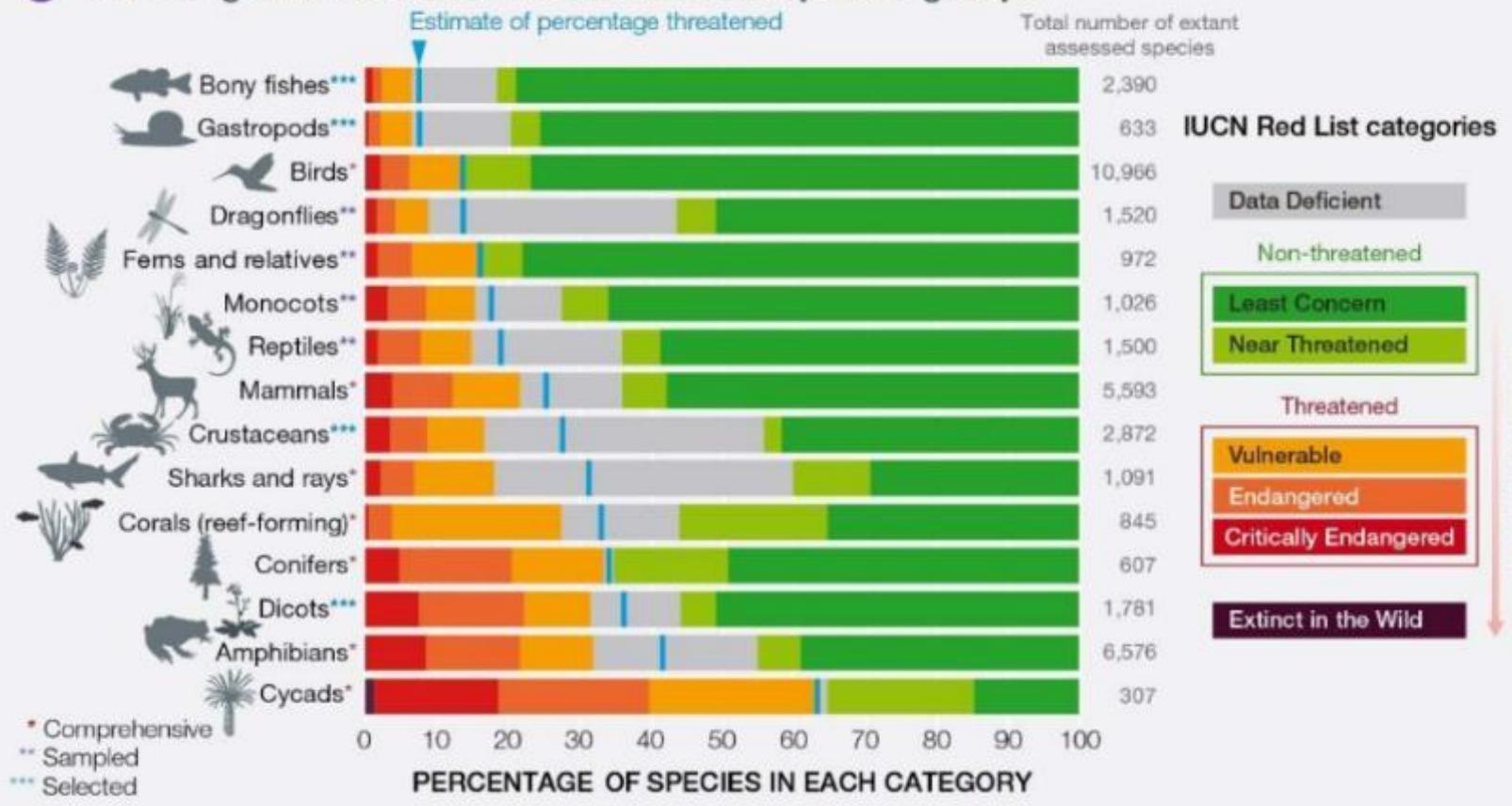
- Don't include quotations
- Cite the focal paper (at the end)
- Can include an image – plus SHORT caption
- Hone in on the relevance of VG for conservation
- Choose a paper that you find most interesting
- Consider your seminar!



Globally, > one million species are threatened



A Current global extinction risk in different species groups



IPBES



Intergovernmental Science-Policy Platform on Biodiversity and Ecosystem Services (2019)

Genetic Diversity (V_G)

AKA GENETIC VARIATION OR VARIANCE

IMPORTANCE (ACCORDING TO IUCN):

1. Key constituent of biodiversity;
2. Required for adaptation
3. Necessary for reducing inbreeding



TODAY:

- Review what it is and where it comes from
- Its role in guiding conservation
- Methods for measuring V_G >>
- Some examples of cons relevance



Genetic Diversity (V_G)

WHAT IS IT?

SIMPLY: DIFFERENCES IN DNA SEQUENCES

V_G
individual
loci

This week
(Lectures 3 & 4)

**Whole Organism
(Phenotype)**

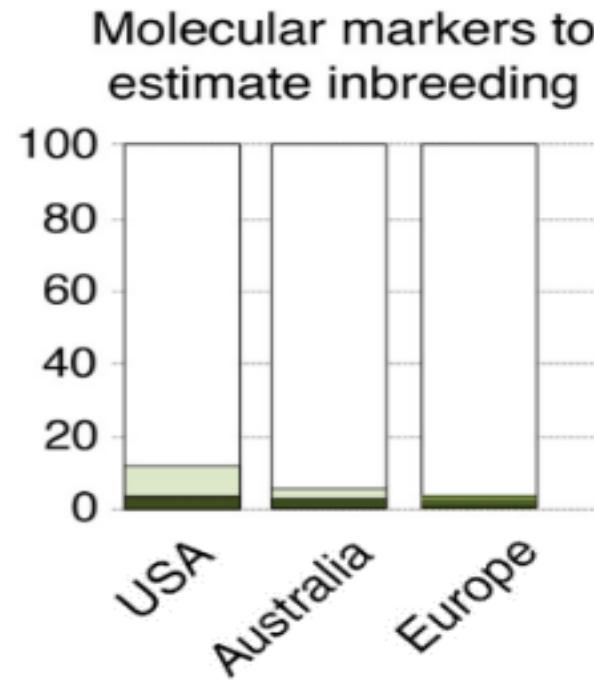
V_G
individual
traits

Next week
(Lectures 5 & 6)

170 180 190
ATCTCTGGCTCCAGCA_TCGATGAAGAACGCA
TCATTTAGAGGA_GTA_AAA_GTCGTAACAA_{GG}T
GAAC_TGTC_{AAA}ACT_{TTT}AA_CACGG_ATCT_{CTT}
TGTTGC_{TT}CGC_{GG}CG_{CC}CA_{AG}GG_TGCC_{CG}
GGC_{CT}GCC_{GT}GGC_{AG}A_TCCCCA_{AC}G_{CC}GGG_{CC}
TCT_{CTT}GG_{CT}CC_{CAG}C_AT_{CG}A_TGA_AAA_CG_{CAG}
CAGC_{AT}CG_AT_{GA}AA_GAC_GC_AG_CAA_AC_GC_GA_T
CGA_{TAC}TT_{CT}GAG_{TG}TT_{CT}AG_{CG}A_{CT}G_TCA_A
CGG_{AT}CT_{CTT}GG_{CT}CC_{CAG}C_AT_{CG}A_TGA_AAA_C
ACA_{AC}GG_{AT}CT_{CTT}GG_{CT}CC_{CAG}C_AT_{CG}A_TGA_A
CGG_{AT}CT_{CTT}GG_{CT}CC_{CAG}C_AT_{CG}A_TGA_AAA_C
GAT_{GA}AA_GAC_GC_AG_{CG}AA_{AC}G_{CG}A_TAT_{GT}TAAT

Are genetic processes appropriately considered

> 300 threatened species recovery plans assessed for consideration of evolutionary factors (genetics)



Pierson et al (2016). *Frontiers in Ecology and the Environment* 14: 433-440

Genetic Diversity (V_G)

MEASUREMENT INDICES

For individual loci:



- **Polymorphism (P)**

= proportion of polymorphic loci;



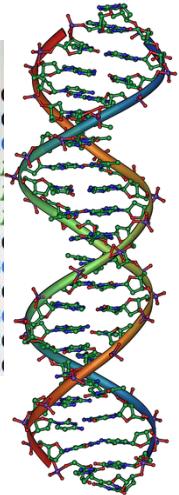
- **Average heterozygosity (H)**

= proportion of heterozygous loci per individual;

- **Allelic diversity (A)**

= average number of different alleles per locus.

170 180
ATCTCTGGCTCCAGCATCGAT
TCATTTAGAGGAAGTAAAAGTC
GAACCTGTCAAAACTTTAACAA
TGTTGCTTCGGCGGGCGCCCGCA
GGCCTGCCGTGGCAGATCCCCA
TCTCTTGGCTCCAGCATCGATG
CAGCATCGATGAAGAACGCAGC
CGATACTTCTGAGTGTTCTTAG
CGGATCTCTTGGCTCCAGCATC
ACAACGGATCTCTTGGCTCCAGC
CGGATCTCTTGGCTCCAGCATC
GATGAAGAACGCAGCGAAACGC



Genetic Diversity (V_G)

AKA GENETIC VARIATION OR VARIANCE



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POLYMORPHISM (P)

Simple ratio of the number of polymorphic loci

Eg:

Locus	Allele frequencies		
A	0.56	0.33	0.11
B	0.70	0.20	0.10
C	0.80	0.20	
D	1.0		
E	1.0		

$$\frac{\sum N_P}{N_T}$$

Where : N_P = number of polymorphic loci
 N_T = total number of loci

$$P = 3/5 = 0.6$$

Genetic Diversity (V_G)

AKA GENETIC VARIATION OR VARIANCE



HETEROZYGOSITY (H)

For a single locus

(H for monomorphic locus = 0)

Eg:

Locus	Alleles & frequencies		
	A1	A2	A3
ADA	0.56	0.33	0.11

$$H = 1 - \sum P_i^2$$

Where P_i = frequency of allele i



$$\begin{aligned} H &= 1 - (0.56^2 + 0.33^2 + 0.11^2) \\ &= 1 - (0.434) \\ &= \mathbf{0.564} \end{aligned}$$

Genetic Diversity (V_G)

AKA GENETIC VARIATION OR VARIANCE

AVERAGE HETEROZYGOSITY (H)

For multiple loci

Simple average across them Eg:

$$\frac{\sum H_i}{N_T}$$

Where : H_i = H at locus i
 N_T = total number of loci

Locus	Alleles & frequencies			H
	A1	A2	A3	
ADA	0.56	0.33	0.11	0.564
BDA	1.00	0	0	0.000

$$\begin{aligned} \text{Average } H &= (0.564 + 0.000)/2 \\ &= \mathbf{0.282} \end{aligned}$$

Genetic Diversity (V_G)

AKA GENETIC VARIATION OR VARIANCE

ALLELIC DIVERSITY (A)

Simple average number of alleles per locus. Eg:

Locus	Allele frequencies			
A	0.60	0.25	0.13	0.12
B	0.70	0.10	0.10	0.10
C	0.50	0.30	0.20	
D	0.55	0.45		
E	0.85	0.15		
F	0.90	0.10		

$$A = \frac{\sum(N_A)}{N_T}$$

Where:

N_A = number of different alleles across all loci

N_T = total number of loci examined

$$A = 17/6 = 2.83$$

Genetic Diversity (V_G)

AKA GENETIC VARIATION OR VARIANCE

AFRICAN LION POPULATION

(Example Chap 3 in textbook)

Calculate:

Polymorphism (P)

Average Heterozygosity (H)

Allelic diversity (A)



Locus	Allele frequencies			H
ADA	0.56	0.33	0.11	0.564
DIAB	0.61	0.39		0.476
ESI	0.88	0.12		0.211
GPI	0.85	0.15		0.255
GPT	0.89	0.11		0.196
MPI	0.92	0.08		0.147
20 others	1.00			0.00

Genetic Diversity (V_G)

AKA GENETIC VARIATION OR VARIANCE

6 of 26 loci polymorphic:

$$P = \frac{6}{26} = \mathbf{0.23}$$

H for the average individual:

$$\frac{\sum H}{26} = \frac{(0.564+0.476+0.211\dots)}{26} = \mathbf{0.071}$$

Allelic diversity (A):

$$\frac{[(1 \times 3)+(5 \times 2)+(20 \times 1)]}{26} = \frac{33}{26} = \mathbf{1.27}$$

Hence, an average of **1.27 alleles per locus**



Locus	Allele frequencies			H
ADA	0.56	0.33	0.11	0.564
DIAB	0.61	0.39		0.476
ESI	0.88	0.12		0.211
GPI	0.85	0.15		0.255
GPT	0.89	0.11		0.196
MPI	0.92	0.08		0.147
20 others	1.00			0.00

Individual H

- is it close to HWE expectations?
- What does this imply?

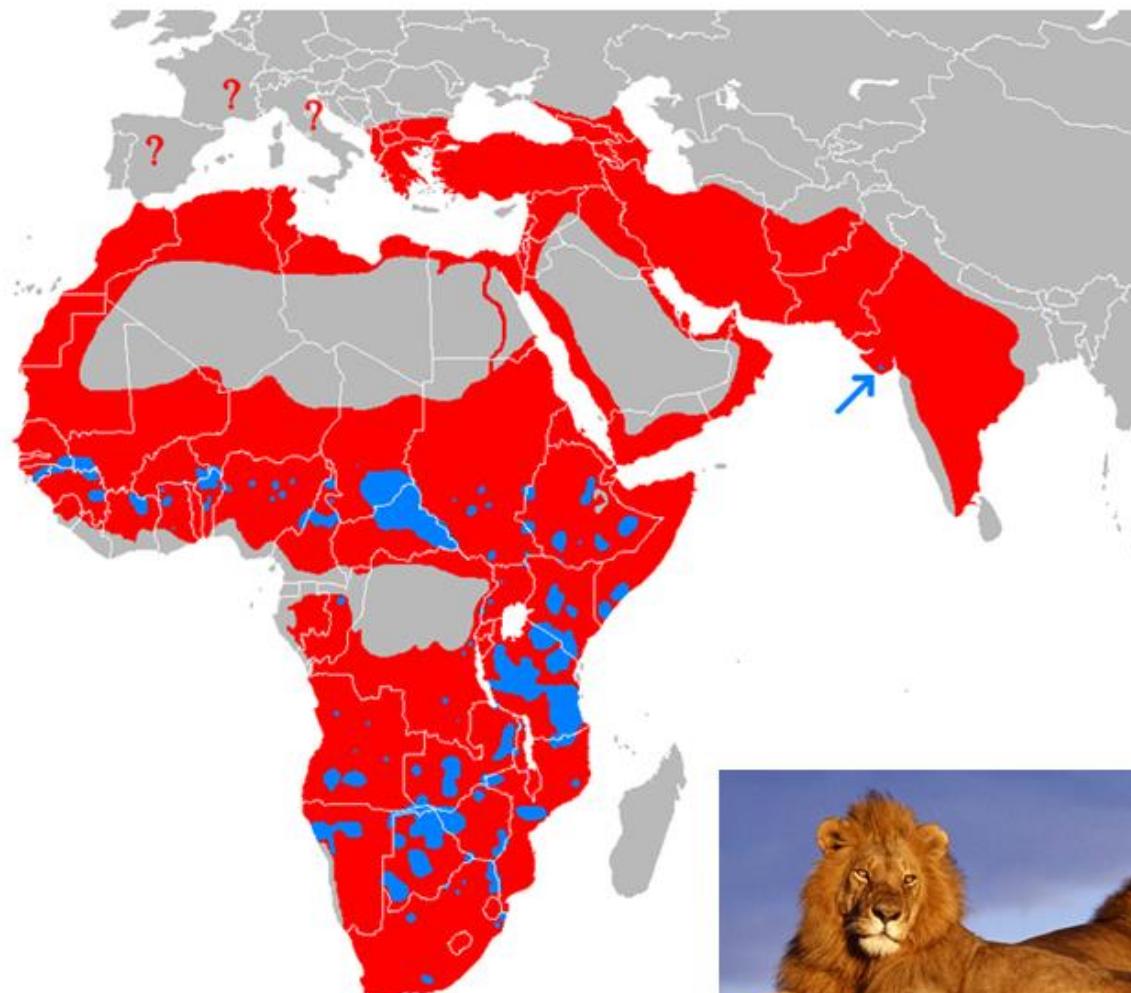
Panthera leo



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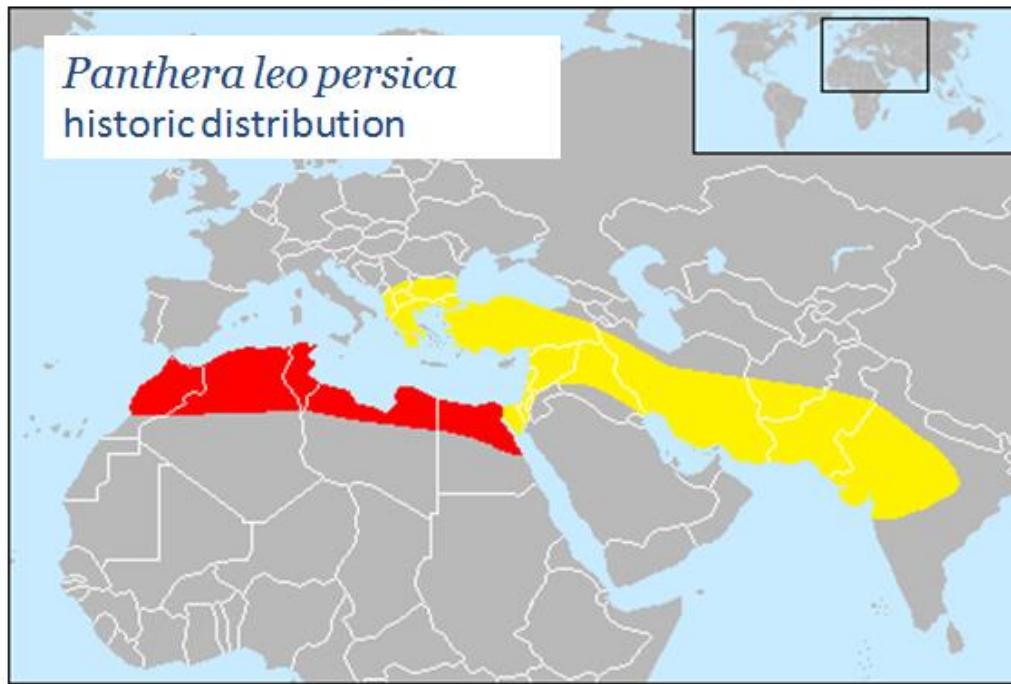
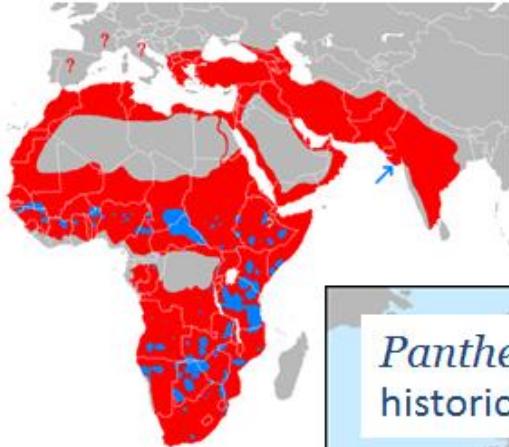
Distribution:

Historic
Current

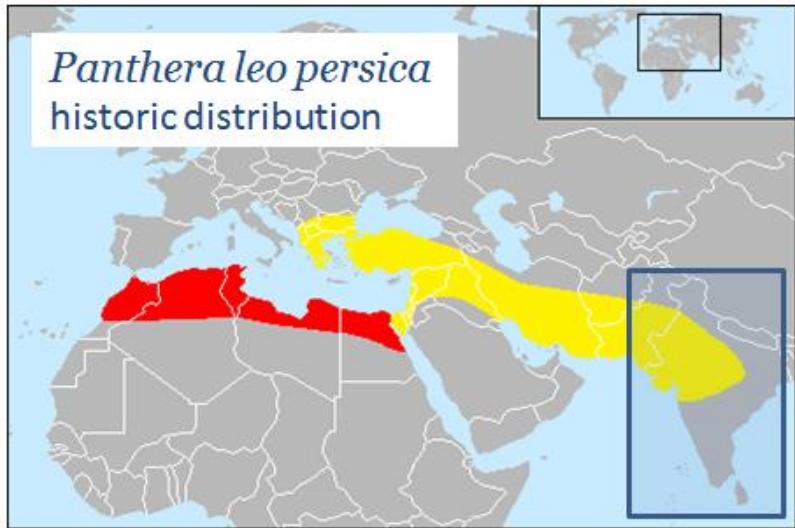


Panthera leo persica

Asiatic lion



Panthera leo persica



Gir Forest National Park:

1974: 180 individuals

2010: 411 individuals

- 97 adult males
- 162 adult females
- 75 sub-adults
- 77 cubs



Panthera leo persica

Genetic diversity

50 allozyme (protein) loci
DNA fingerprints



	Allozymes	DNA fingerprints	
	P	H	H
Asiatic lion	0.00	0.000	0.038
African lion (outbred)	0.04-0.11	0.015-0.300	0.450



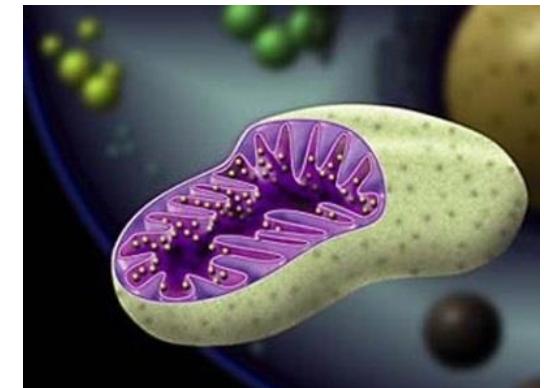
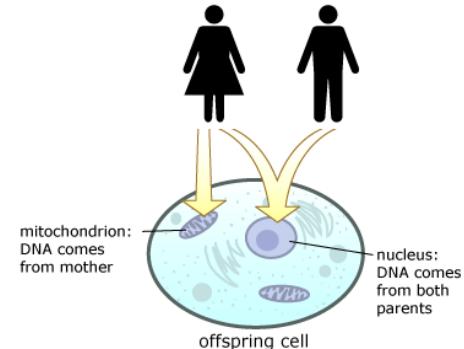
- Indicative of extreme **bottleneck** in recent past ($N \sim 20$ in early 1900's)
- Problematic for inbreeding depression, lack of population resilience and reduced adaptive potential

Genetic Diversity (V_G)

AKA GENETIC VARIATION OR VARIANCE

ASSESSMENT AT SINGLE LOCUS LEVEL

- **Proteins** (allozyme electrophoresis)
- **Nuclear DNA** amplification via PCR
 - Microsatellites (sequence repeats)
 - SNPs (single nucleotide polymorphism)
 - AFLP (amplified fragment length polymorphism)
 - RAPD (Randomly amplified polymorphic DNA)
 - Sequencing
 - Plus other techniques...
(see Box 3.3 Frankham *et al.*)
- **Mitochondrial DNA**



Genetic Diversity (V_G)

AKA GENETIC VARIATION OR VARIANCE



MICROSATELLITES

- Repeats of 1-5bp sequences
 1. Simple Sequence Repeats (STRs)
 2. Short Tandem Repeats (STRs)

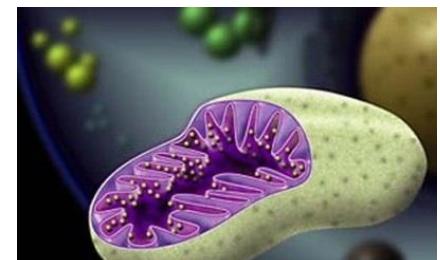
6 Repeats of 2bp segment e.g:

X C A C A C A C A C A C A Y
X G T G T G T G T G T G T Y

- **Highly variable**
- Mostly **neutral DNA** (non-coding regions)
- Potential for **non-invasive sampling**

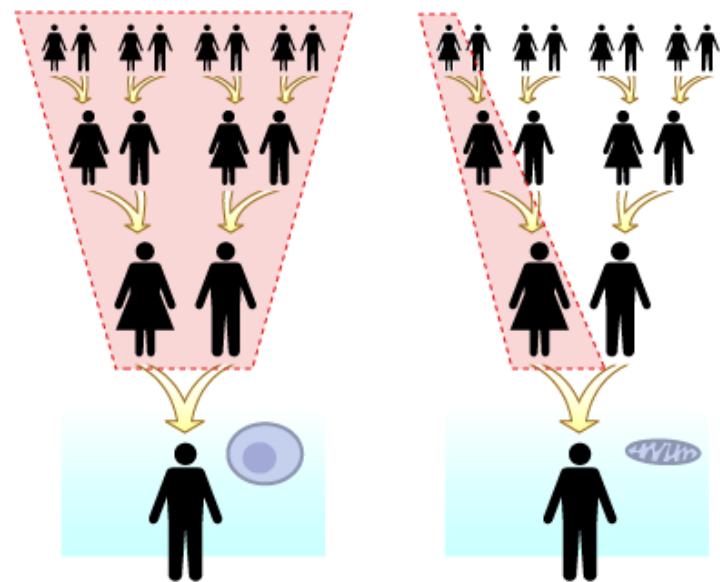
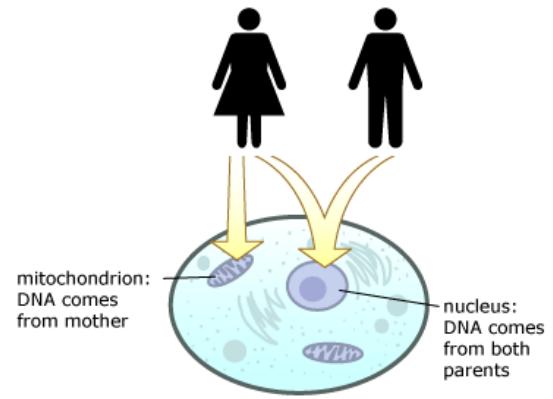
Genetic Diversity (V_G)

AKA GENETIC VARIATION OR VARIANCE



MITOCHONDRIAL DNA

- Haploid maternally-inherited DNA
- No recombination
- Useful in cons gen to:
 1. Resolve taxonomic uncertainties
 2. Define management units (haplotypes)
 3. Understand species biology



DNA in plant chloroplasts is equivalent

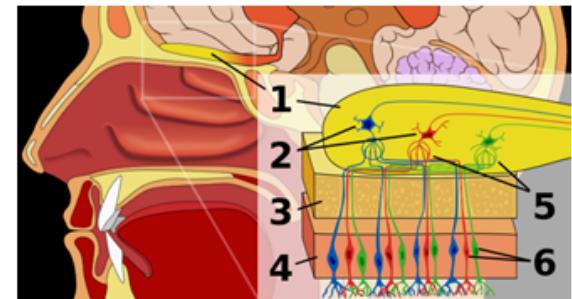
Genetic Diversity (V_G)

AKA GENETIC VARIATION OR VARIANCE

MAJOR HISTOCOMPATABILITY COMPLEX



- Code for proteins for surface antigens in 2 classes:
 - Class I Recognize intracellular pathogens and ‘tag’ the cell for immune system components (T-cells, phages etc).
 - Class II
- Highly polymorphic loci with very high allelic diversity
- Sexual selection implicated in maximising population heterozygosity: mate choice for MHC diversity (via olfactory cues in humans)

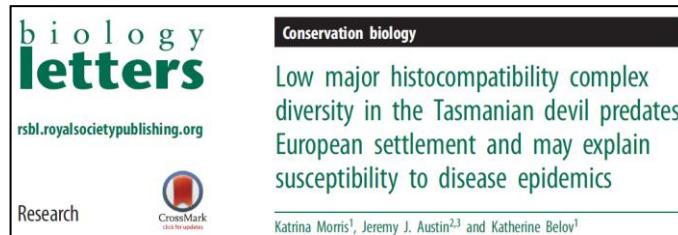


Genetic Diversity (V_G)

AKA GENETIC VARIATION OR VARIANCE

MHC DIVERSITY & DEVIL FACIAL TUMORS (DFTD)

Historically low VG in MHC:



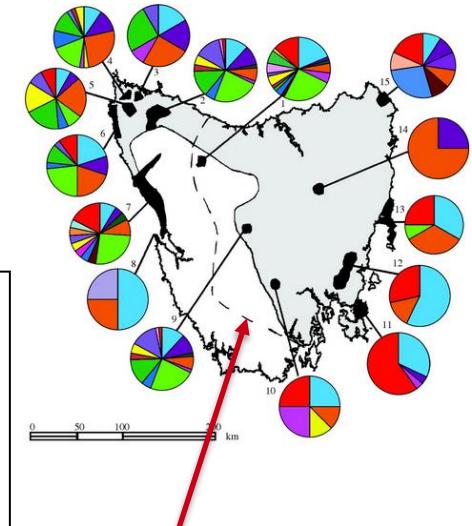
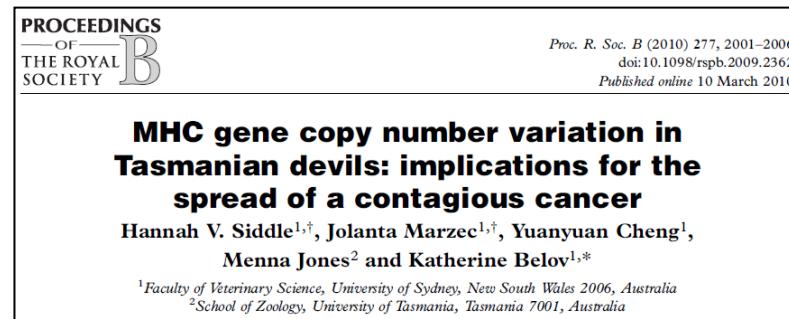
Recovery implications:

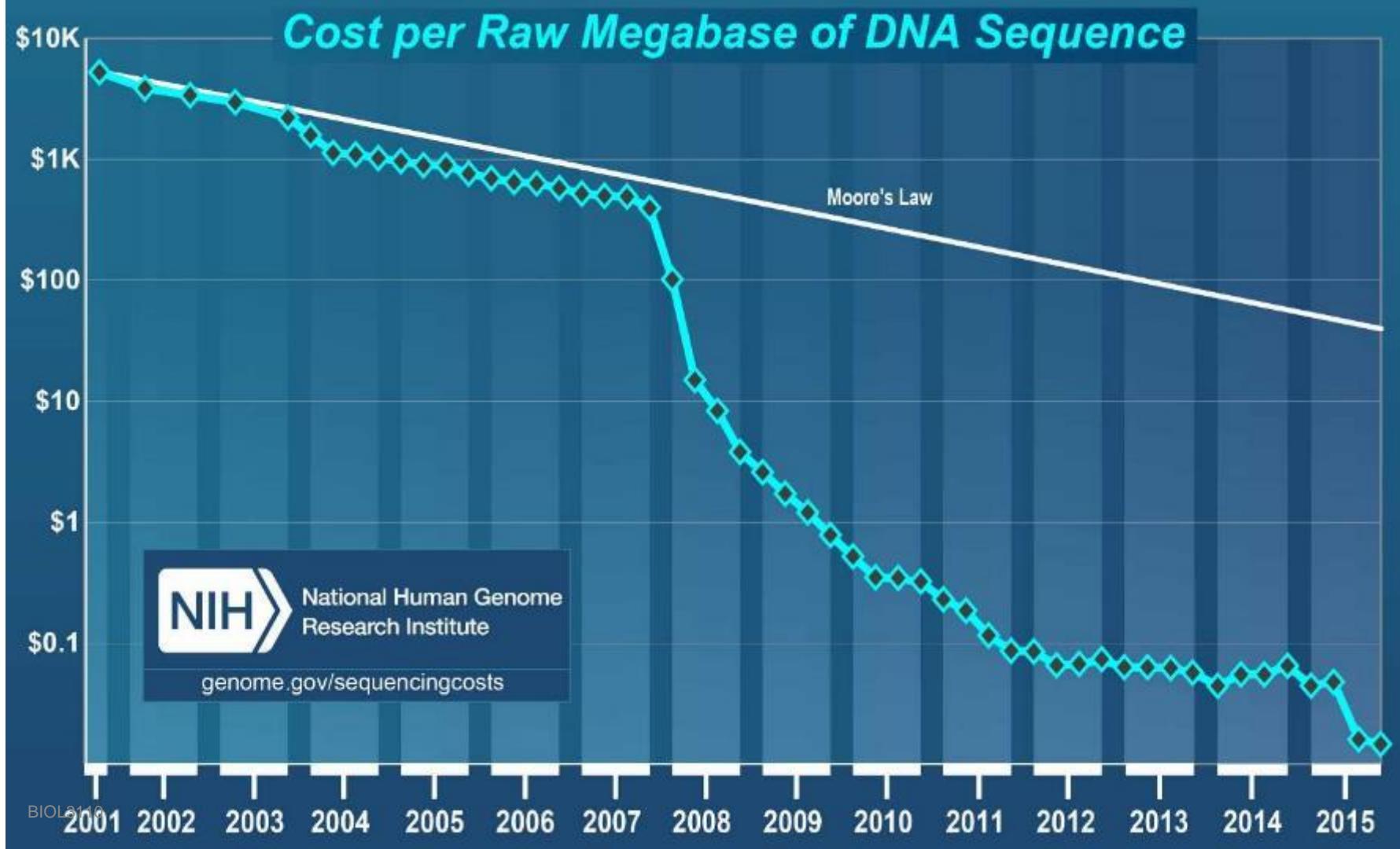
Most DFT cells express MHC classes 1 & 2

20% of devils only have class 1 or 2 genes

MHC1 devils may detect DFT cells
expressing MHC1&2

Counterintuitive example





Modern genomic workflow



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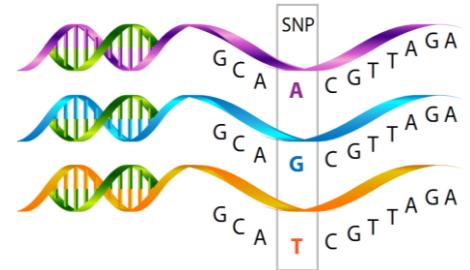
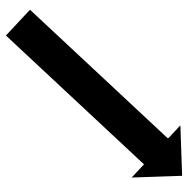
Extract
DNA



Outsource



More time to
read/think!



More time to develop bioinformatic skills

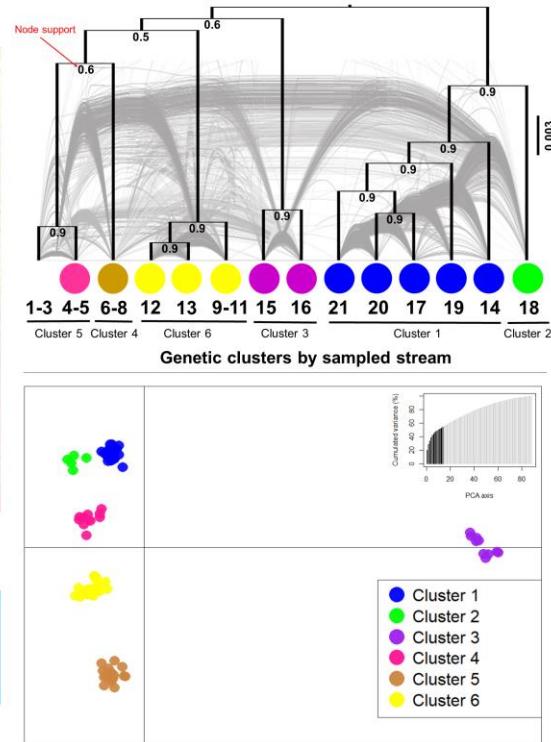
Big data:
 $10^2 - 10^6$
genome wide
markers

1) Biodiversity patterns

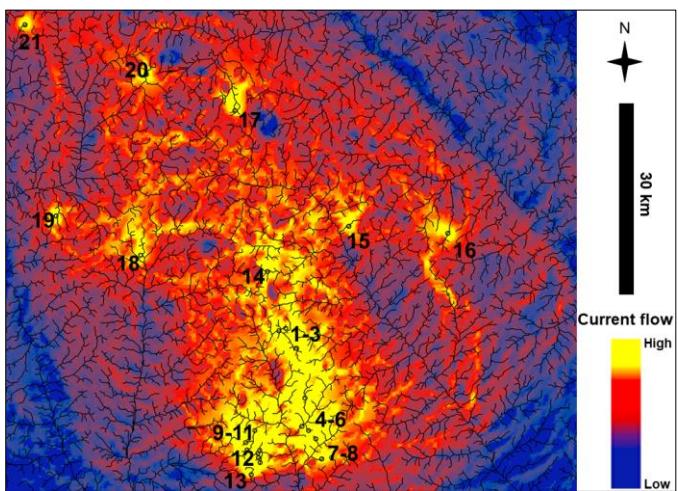
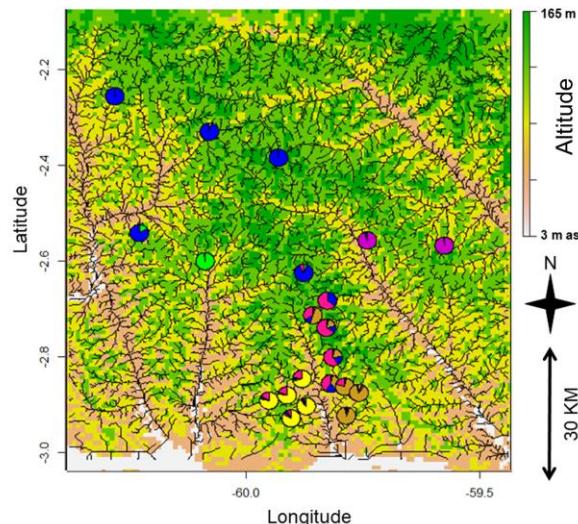
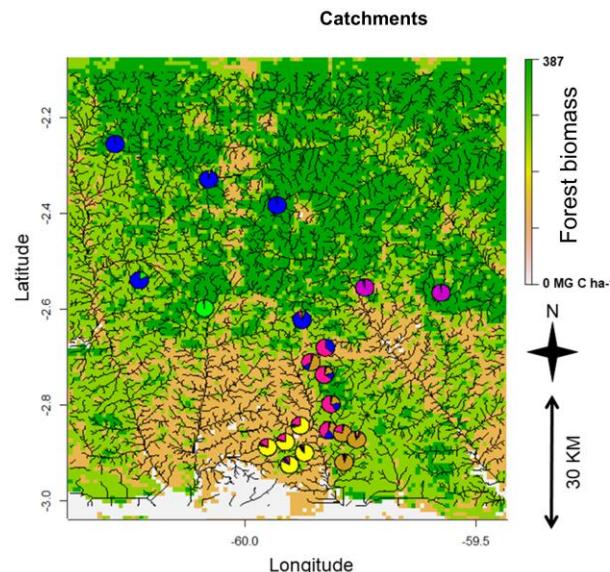
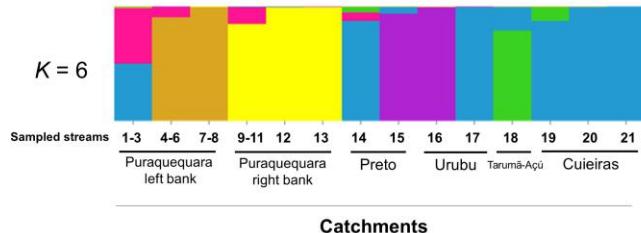
- diversity, distribution, distinctiveness



2) Estimating dispersal, genetic connectivity



3) Identifying processes

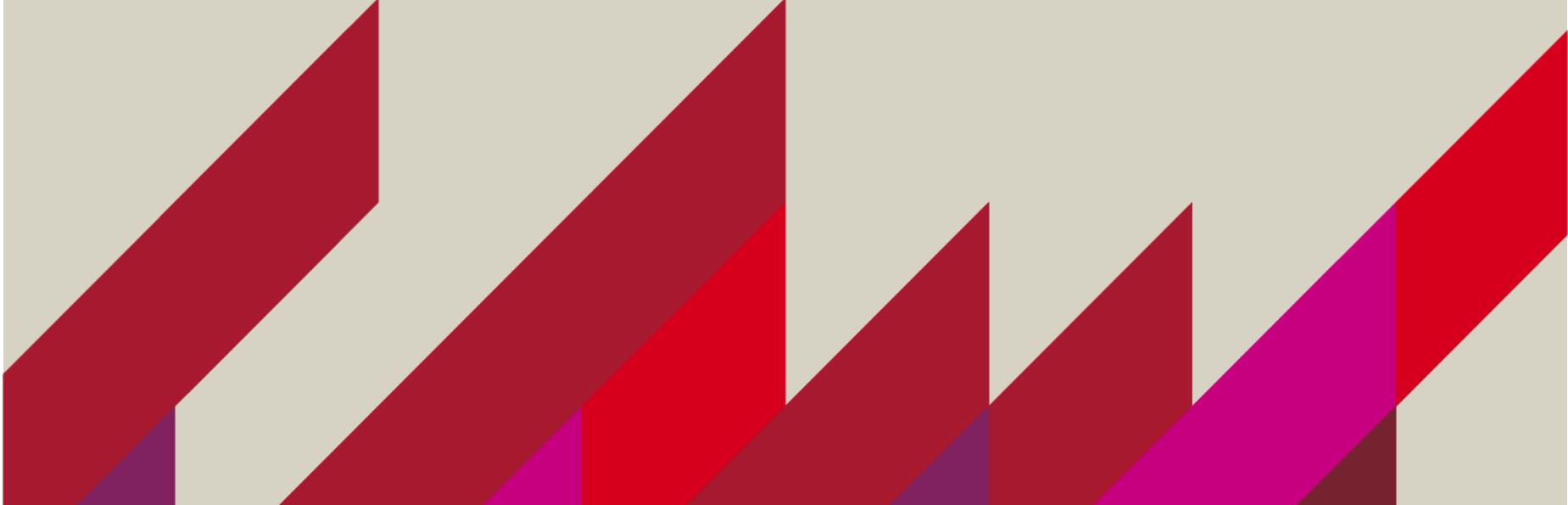


Next lecture:



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Characterising V_G for single loci
More detail & examples

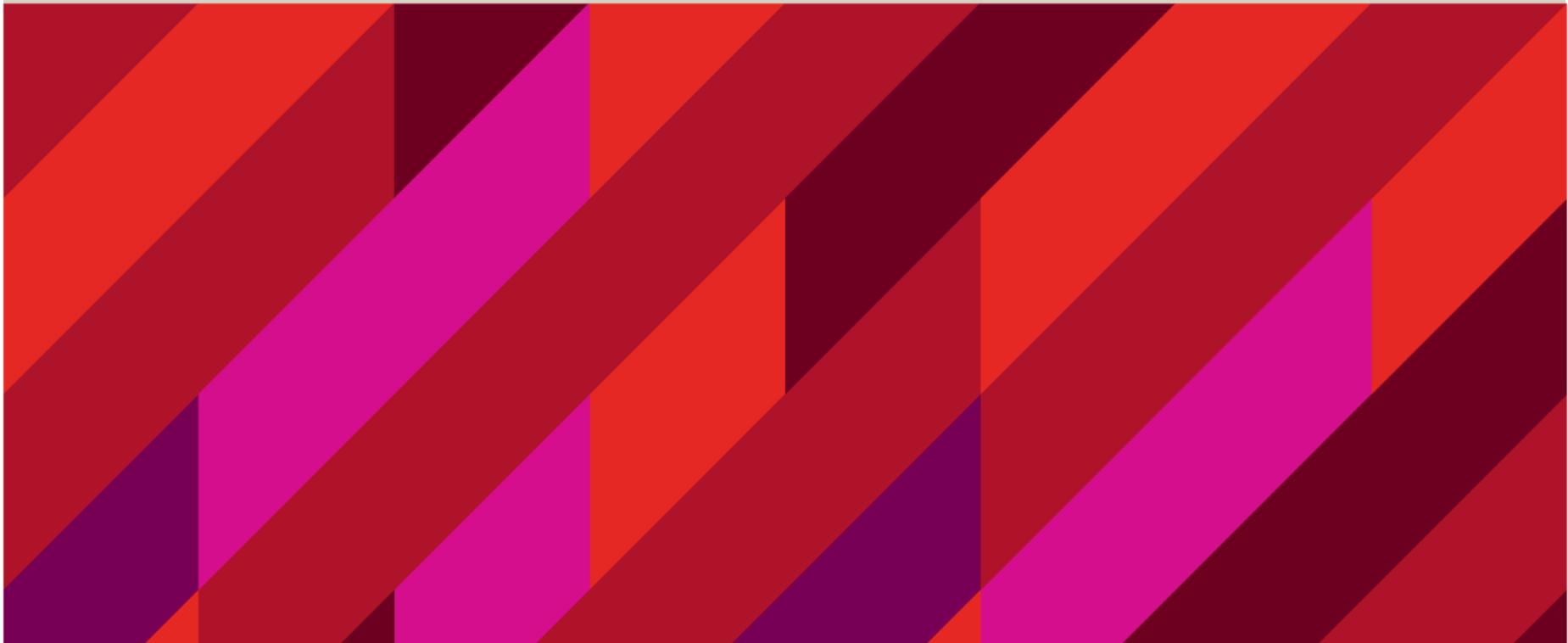




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BIOL3110 Conservation & Ecological Genetics

LECTURE 4: CHARACTERIZING GENETIC VARIATION



Genetic Diversity (V_G)

AS A “BAROMETER” OF POPULATION GENETIC HEALTH

Useful comparisons:

- (1) Among (sub)populations →
- (2) Over time (the same pop/species)
as an index of change, i.e.:



$$\frac{H_0}{H_T}$$

Where :

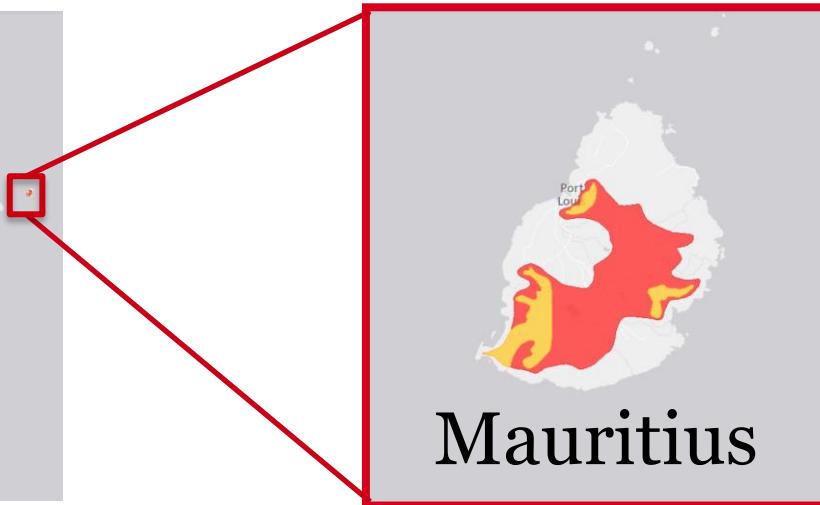
H_0 = H at the time of observation

H_T = H as known for the past or predicted
for the future...



Genetic Diversity (V_G)

MAURITIUS KESTREL (*Falco punctatus*)



- █ Extant (resident) 
- █ Possibly Extinct 

[Home](#) > *Falco punctatus* (Mauritius Kestrel)



Falco punctatus

NOT EVALUATED	DATA DEFICIENT	LEAST CONCERN	NEAR THREATENED	VULNERABLE	ENDANGERED	CRTICALLY ENDANGERED	EXTINCT IN THE WILD	EXTINCT
NE	DD	LC	NT	VU	EN	CR	EW	EX

Driven down to **4** wild birds – **1 breeding pair** in 1974!!

Genetic Diversity (V_G)

MAURITIUS KESTREL (*Falco punctatus*)

Population size:

1974: single mating pair bottleneck **N=2**

1997: N=500

Calculations using microsatellite markers
extracted from museum material:

$$\frac{H_O}{H_T} = \frac{H_{1997}}{H_{1974}} = \frac{0.10}{0.23} = 0.43$$



Loss of more than 50% of H since 1974
...even despite N increasing from 2 to ~500?

Genetic Diversity (V_G)

THE HARDY-WEINBERG (H-W) EQUILIBRIUM



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H-W equilibrium provides the “null expectation” for allele/genotype frequencies in a pop. Genotypes will accord to HWE unless:

- **Inbreeding** (>>homozygotes)
- **Selection** (not exp at neutral loci)
- **Non-random mating** (e.g. MHC)
- **Small populations** (Drift) 
- **Mutation** – takes a long time!



Genetic Diversity (V_G)

THE HARDY-WEINBERG (H-W) EQUILIBRIUM

For 2 alleles (A_1 and A_2) segregating at a locus, if we have:

- A large, closed population (no migration)
- Random mating (i.e. no selective mate choice)
- Equal fitness of genotypes
- Negligible mutation

Then we expect alleles to assort among genotypes as:

Genotypes:	A_1A_1	A_1A_2	A_2A_2
Frequencies:	p^2	$2pq$	q^2

Where $\textcolor{blue}{p}$ = frequency of A_1 and $\textcolor{red}{q}$ = frequency of A_2

Genetic Diversity (V_G)

THE HARDY-WEINBERG (H-W) EQUILIBRIUM



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Where $\textcolor{teal}{p}$ = frequency of A_1 and $\textcolor{red}{q}$ = frequency of A_2

Genotypes:	A_1A_1	A_1A_2	A_2A_2
Frequencies:	p^2	$2pq$	q^2

For a locus
with **2** alleles:

$$H_{exp} = 2\textcolor{teal}{p}\textcolor{red}{q}$$

Genetic Diversity (V_G)

ALLELLE COUNTING

Absolute frequencies	Genotypes			Total
	A_1A_1	A_1A_2	A_2A_2	
Individuals:	38	60	2	100
Alleles:	2 copies of A_1	1 copy of A_1 1 copy of A_2	2 copies of A_2	200

Genetic Diversity (V_G)

ALLELLE COUNTING

Absolute frequencies	Genotypes			Total
	A_1A_1	A_1A_2	A_2A_2	
Individuals:	38	60	2	100
Alleles:	2 copies of A_1	1 copy of A_1 1 copy of A_2	2 copies of A_2	200
$A_1:$	76	60	0	136
$A_2:$	0	60	4	64

Genetic Diversity (V_G)



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ALLELLE COUNTING

Absolute frequencies	Genotypes			Total
	A_1A_1	A_1A_2	A_2A_2	
Individuals:	38	60	2	100
Alleles:	2 copies of A_1	1 copy of A_1 1 copy of A_2	2 copies of A_2	200
$A_1:$	76	60	0	136
$A_2:$	0	60	4	64

Relative frequencies (~%) of each allele:

$$A_1 = p = 136/200 = 0.68$$

$$A_2 = q = 64/200 = 0.32 \text{ (...or simply } 1-p\text{)}$$

Genetic Diversity (V_G)

USE p & q TO CALCULATE H-W EXPECTATION

$$p (A_1) = 0.68, q (A_2) = 0.32$$

	Genotypes			Total
	A ₁ A ₁	A ₁ A ₂	A ₂ A ₂	
H-W exp frequencies:	p^2	$2pq$	q^2	

Genetic Diversity (V_G)

USE p & q TO CALCULATE H-W EXPECTATION

$$p (A_1) = 0.68, q (A_2) = 0.32$$

	Genotypes			Total
	A ₁ A ₁	A ₁ A ₂	A ₂ A ₂	
H-W exp frequencies:	p^2	$2pq$	q^2	
For p=0.68 & q =0.32:	0.68^2	$2 \times 0.68 \times 0.32$	0.32^2	1.0
=	0.462	0.435	0.102	1.0

Genetic Diversity (V_G)

USE p & q TO CALCULATE H-W EXPECTATION



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$$p (A_1) = 0.68, q (A_2) = 0.32$$

	Genotypes			Total
	A_1A_1	A_1A_2	A_2A_2	
H-W exp frequencies:	p^2	$2pq$	q^2	
For $p=0.68$ & $q = 0.32$:	0.68^2	$2 \times 0.68 \times 0.32$	0.32^2	1.0
=	0.462	0.435	0.102	1.0

Expected genotypes given a sample of $N=100$	46.2	43.5	10.2	100
---	-------------	-------------	-------------	------------

Genetic Diversity (V_G)

USE p & q TO CALCULATE H-W EXPECTATION



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$$p (A_1) = 0.68, q (A_2) = 0.32$$

	Genotypes			Total
	A_1A_1	A_1A_2	A_2A_2	
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=	0.462	0.435	0.102	1.0

Expected genotypes given a sample of $N=100$	46.2	43.5	10.2	100
---	------	------	------	-----

What we observed:	38	60	2	100
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DO THESE DISTRIBUTIONS DIFFER?

Genetic Diversity (V_G)

THE “CHI-SQUARED” TEST FOR SIGNIFICANCE

$$\chi^2 = \sum \frac{(Obs - Exp)^2}{Exp}$$

Does your observed distribution of genotypes **differ** from the H-W expected distribution?

Calculate using **absolute genotype frequencies** (the numbers that we just calculated)

Use a significance table to look up a **P-value**

Genetic Diversity (V_G)

CHI-SQUARED" TEST FOR SIGNIFICANCE

$$\chi^2 = \sum \frac{(Obs - Exp)^2}{Exp}$$

	Genotypes			Sum
	A_1A_1	A_1A_2	A_2A_2	
H-W Exp:	46.2	43.5	10.2	100
Obs:	38	60	2	100

Genetic Diversity (V_G)

CHI-SQUARED" TEST FOR SIGNIFICANCE



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	Genotypes			Sum
	A_1A_1	A_1A_2	A_2A_2	
H-W Exp:	46.2	43.5	10.2	100
Obs:	38	60	2	100
$\frac{(Obs - Exp)^2}{Exp}$	$\frac{(38 - 46.2)^2}{46.2}$	$\frac{(60 - 43.5)^2}{43.5}$	$\frac{(2 - 10.2)^2}{10.2}$	

$$\chi^2 = \sum \frac{(Obs - Exp)^2}{Exp}$$

Genetic Diversity (V_G)

CHI-SQUARED" TEST FOR SIGNIFICANCE

$$\chi^2 = \sum \frac{(Obs - Exp)^2}{Exp}$$

	Genotypes			Sum
	A_1A_1	A_1A_2	A_2A_2	
H-W Exp:	46.2	43.5	10.2	100
Obs:	38	60	2	100
$\frac{(Obs - Exp)^2}{Exp}$	$\frac{(38 - 46.2)^2}{46.2}$	$\frac{(60 - 43.5)^2}{43.5}$	$\frac{(2 - 10.2)^2}{10.2}$	
=	1.49	6.29	6.59	= 14.34

Chi Square statistic = **14.34**, $P < 0.05$ (from a table)
 Hence, conclude a significant deviation from HW...

Critical values of the Chi-square distribution with d degrees of freedom

Probability of exceeding the critical value							
d	0.05	0.01	0.001	d	0.05	0.01	0.001
1	3.841	6.635	10.828	11	19.675	24.725	31.264
2	5.991	9.210	13.816	12	21.026	26.217	32.910
3	7.815	11.345	16.266	13	22.362	27.688	34.528
4	9.488	13.277	18.467	14	23.685	29.141	36.123
5	11.070	15.086	20.515	15	24.996	30.578	37.697
6	12.592	16.812	22.458	16	26.296	32.000	39.252
7	14.067	18.475	24.322	17	27.587	33.409	40.790
8	15.507	20.090	26.125	18	28.869	34.805	42.312
9	16.919	21.666	27.877	19	30.144	36.191	43.820
10	18.307	23.209	29.588	20	31.410	37.566	45.315

Degrees of freedom =
number of genotypes -1-
number of alleles -1

Or more simply –
number of genotypes –
number of alleles

Df = 1

Genetic Diversity (V_G)

H-W EQUILIBRIUM FOR 2+ ALLELES



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2 ALLELES:

$A_1(p)$
 $A_2(q)$

Genotypes		
A_1A_1	A_1A_2	A_2A_2
p^2	$2pq$	q^2

3 ALLELES:

$A_1(p)$
 $A_2(q)$
 $A_3(r)$

Genotypes					
A_1A_1	A_1A_2	A_1A_3	A_2A_2	A_2A_3	A_3A_3
p^2	$2pq$	$2pr$	q^2	$2qr$	r^2

Genetic Diversity (V_G)

SEX-LINKED LOCI



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Loci on the Sex Chromosomes

Humans, primates,
mammals & most insects:

Females XX, males XY

(females double-dose of X-linked alleles)



Birds & butterflies:

Females ZW, males ZZ



This changes the allele counting process...

see **Frankham et al. Table 4.6 & Example 4.7**

Genetic Diversity (V_G)

AS A “BAROMETER” OF POPULATION GENETIC HEALTH



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Heterozygosity versus allelic diversity...

A: average allelic diversity

- Sensitive to sample size (N)
- Statistical control for unequal N

$$A = \frac{\sum(N_A)}{N_T}$$

Where:

N_A = number of different alleles across all loci

N_T = total number of loci examined

n_e : effective number of alleles

- Less sensitive to sample size
- Less influenced by rare alleles
- Highest when more alleles present but at equal frequencies

$$n_e = \frac{1}{\sum P_i^2}$$

Where:

P_i = frequency of each allele

Genetic Diversity (V_G)

LINKAGE DISEQUILIBRIUM

- Non-random associations between alleles at different loci is called **linkage disequilibrium (D)**



e.g.: 2 alleles at **A & B loci:**

A_1B_1	A_1B_2	A_2B_1	A_2B_2
----------	----------	----------	----------

- Can result from strong selection (e.g. mate preference & trait alleles in sexual selection)
- Can indicate bottlenecks, recent pop mixing, etc.

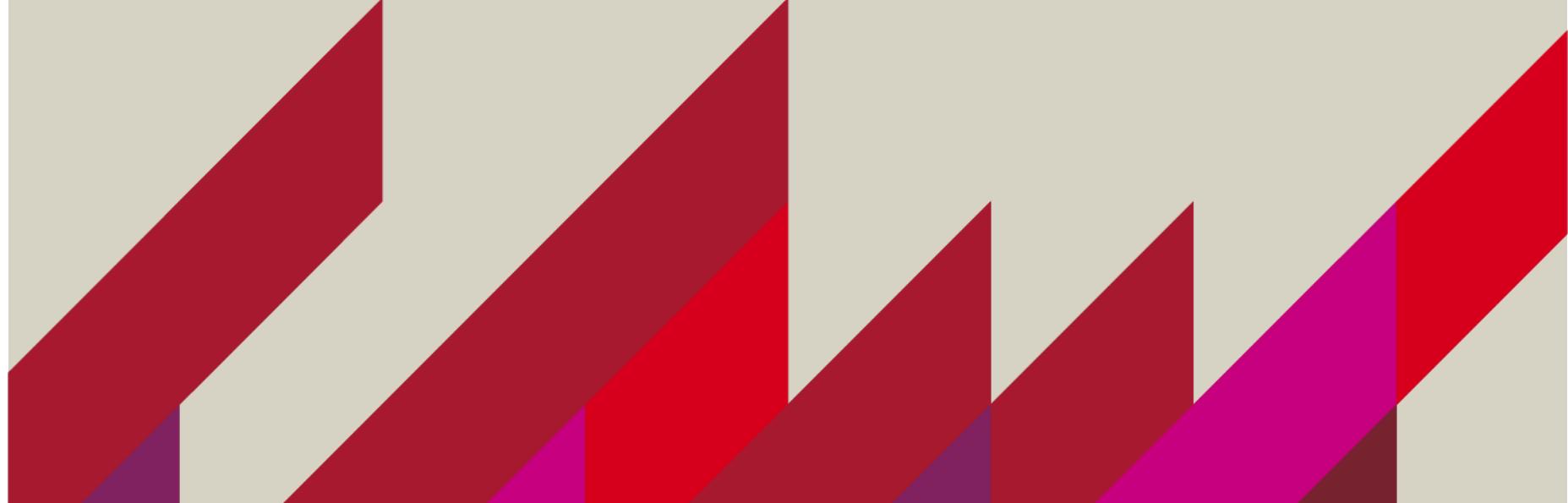
see Frankham et al. Ch. 4 (Box 4.3 & 4.4, Table 4.9)

Coming up:



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Genetics from the phenotypic level



Quantitative Genetics (1)

LECTURE 5: HERITABILITY

2022



Background

Quantifying VG

- so far limited to single locus

- H, A, ne, P,

- Looking at genetics from a phenotypic level

- Used to attribute causality to traits (nature vs. nurture)

Causal agents of phenotypes

- Alleles segregating at many loci

- Interactions among alleles (dominance and/or epistasis)

- Environmental effects

- Interactions & correlations between genetic and environmental effects

Lecture Outline

- Quantitative and Qualitative Traits
- History of Heritability
- Heritability (H^2) and Genetic Causes
- Relationship with Conservation Biology
- Worked Example for H^2 (ANOVA)
- GxE Interaction
- Phenotypic Plasticity

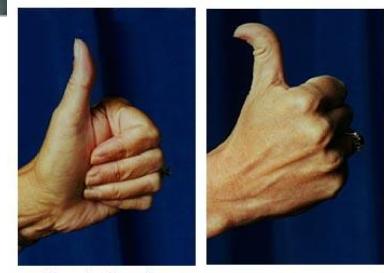
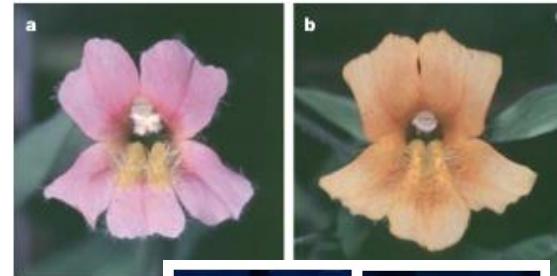
'Mendelian loci'

- Blood Type (A, B, O, AB)
- Flower colour (yellow, pink)
- Thumb shape
- Coat colour (black, orange –X linked)
- Goat beard (beard, no beard)

- Phenotypes take discrete values
- Predictable using Mendel's laws

22,480 known Mendelian traits in man
(as of 14 Aug 2014), OMIM:
<http://www.omim.org/>

3,057 known Mendelian traits in animals,
OMIA: <http://omia.angis.org.au/home/>



©Warren Photographic



Quantitative genetics

- The genetic appraisal and analysis of '**complex**', '**continuous**', or '**quantitative**' traits, where:
- A quantitative trait = a trait whose phenotypic expression is determined by many individual genes (i.e. variation at many individual loci) (polygenic)



Examples of Quantitative Traits:

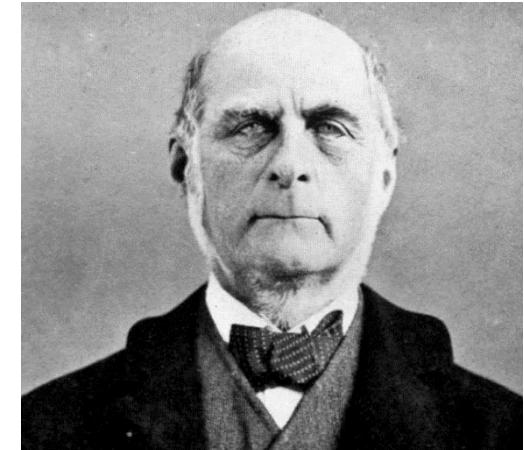
- Morphological traits:
 - Body size (height, weight)
 - Colouration (e.g. Bird plumage)
- Physiological traits
 - Enzyme activity
 - Blood pressure, cardiovascular performance
- Behavioural traits
 - Aggression, mate preferences, IQ etc.
- Life history traits
 - Developmental rates, age at maturity, etc.



History of Quantitative Genetics

Francis Galton: ‘the apostle of quantification’

- the weather (1863)
- height (1889)
- finger print patterns (1888; 1892; 1893)
- beauty (1909)
- boredom (1909)
- criminal characteristics (1885)
- the effectiveness of prayers (1872)

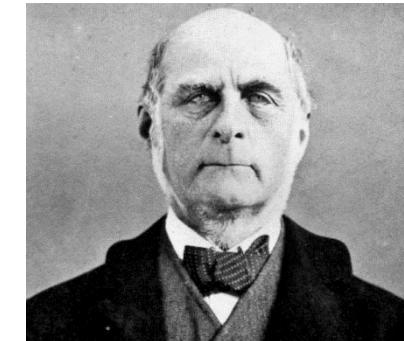


Introduced the terms ‘nature’ and ‘nurture’
(*English Men of Science*, 1874)

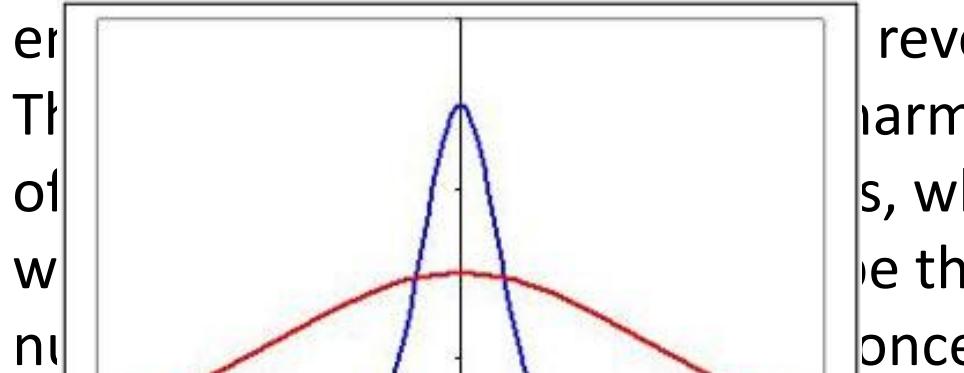
History of Quantitative Genetics

Francis Galton

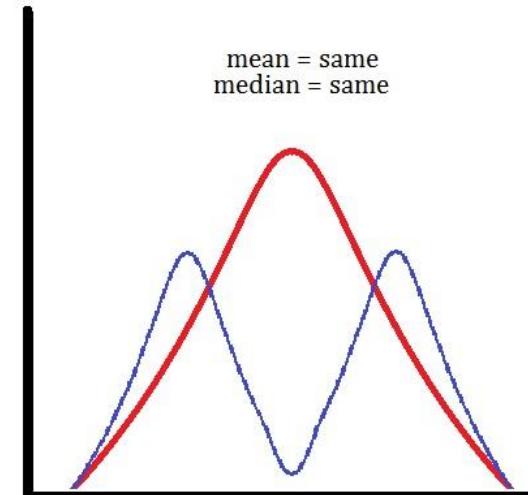
Interested in *trait variation* rather than trait means



“It is difficult to understand why statisticians commonly limit their



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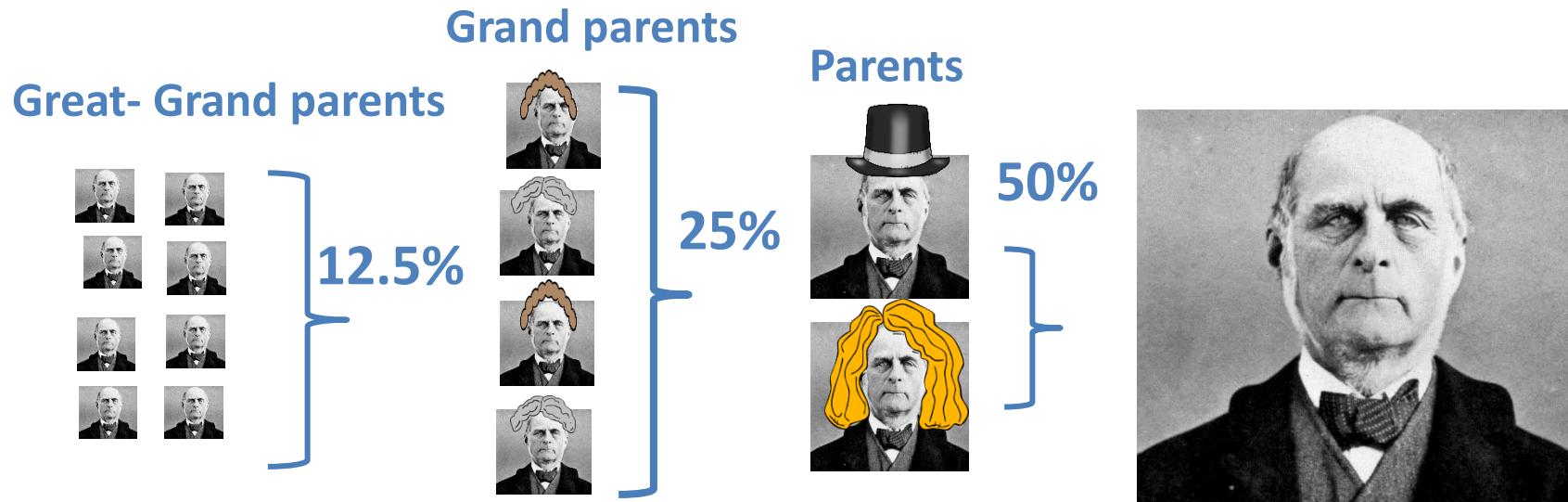
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History of Quantitative Genetics

Francis Galton

Different theory of inheritance:

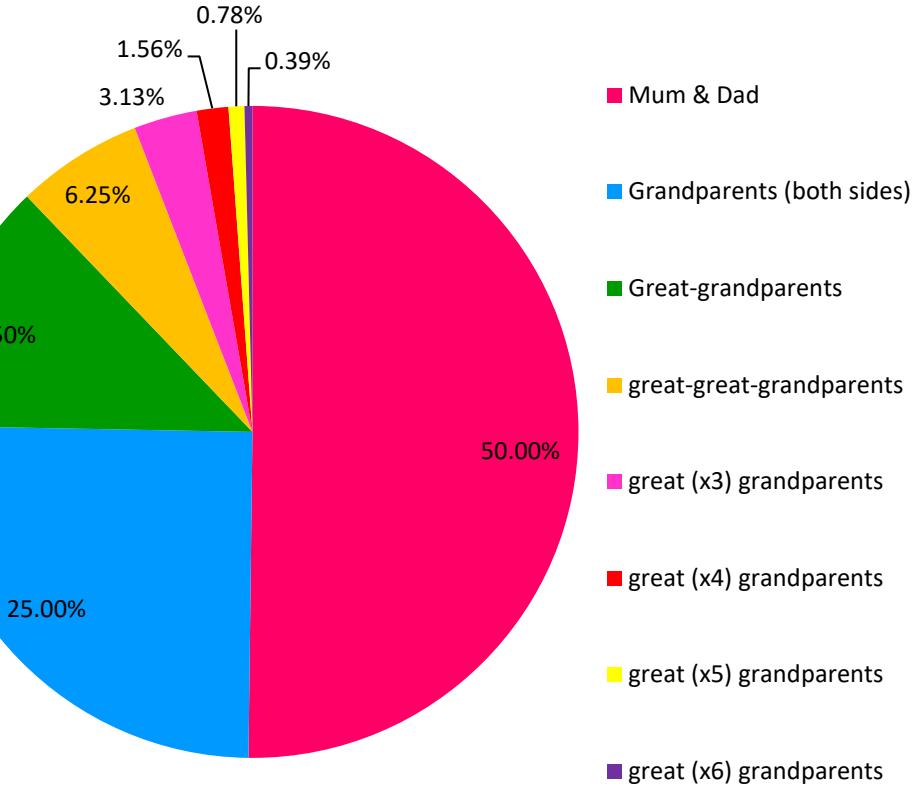
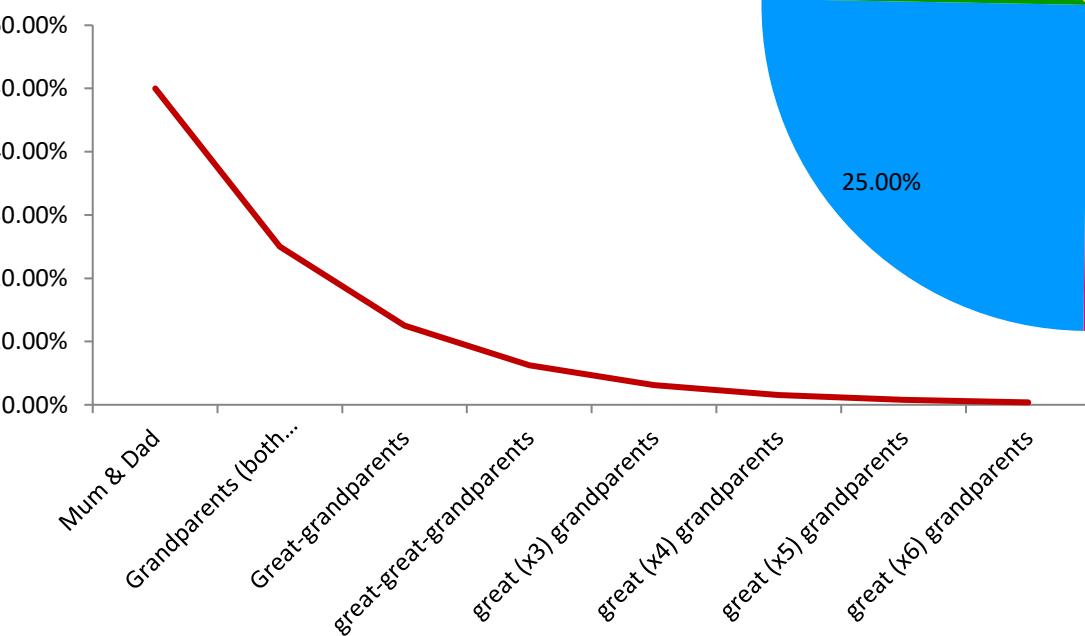
The parents of an individual together contribute on an average 50 percent of the total inherited characters, the 4 grandparents together 25 percent, the 3rd generation of ancestors together 12.5 percent, etc.



History of Quantitative Genetics

Francis Galton

Different theory of inheritance:



History: Galton vs. Mendel

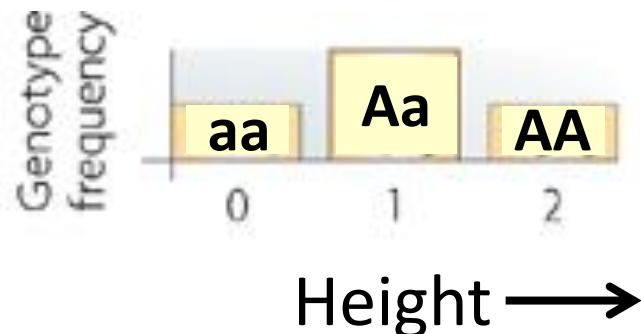


Traits inherited in a Mendelian fashion. Phenotypes discrete characteristics

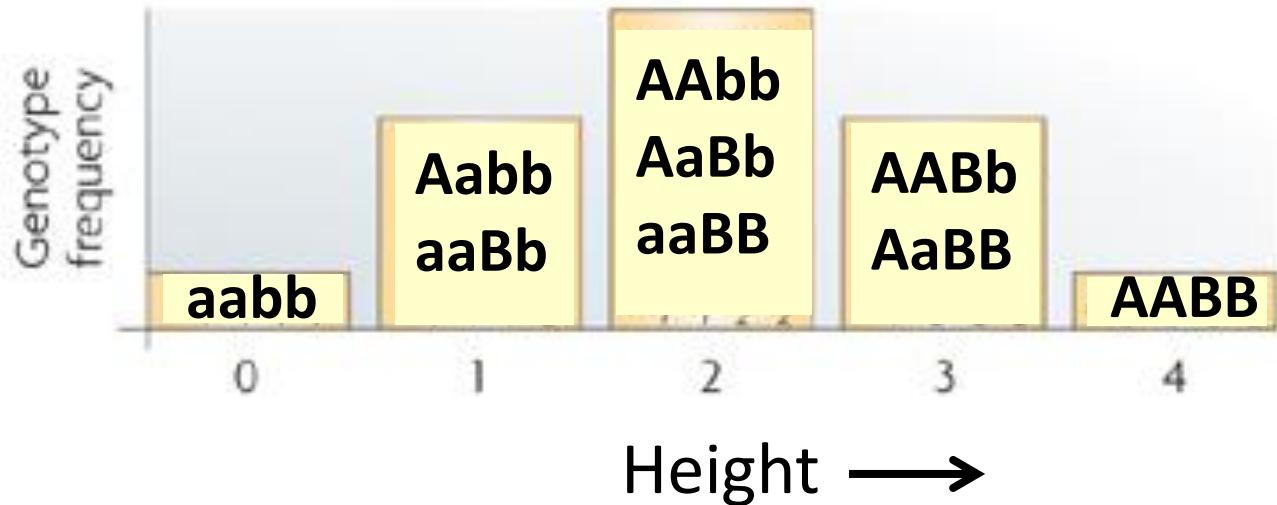
Traits inherited as complex averages of ancestral phenotypes. Continuously varying phenotypes

Quantitative traits and Mendelian laws

a One locus
(A):

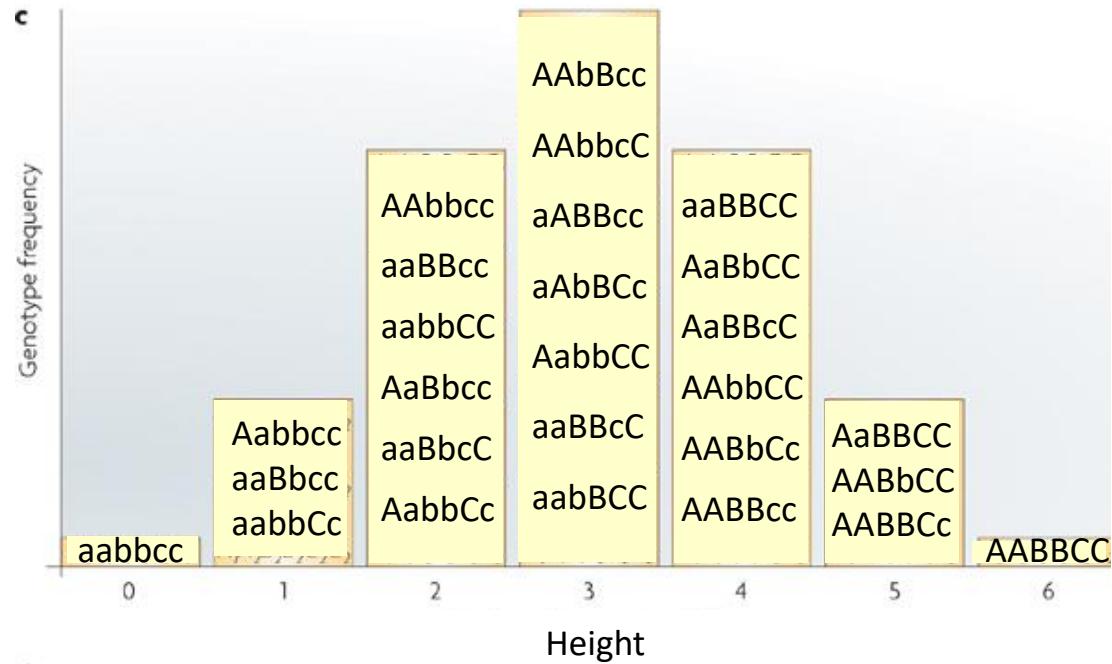


b Two Loci
(A & B):

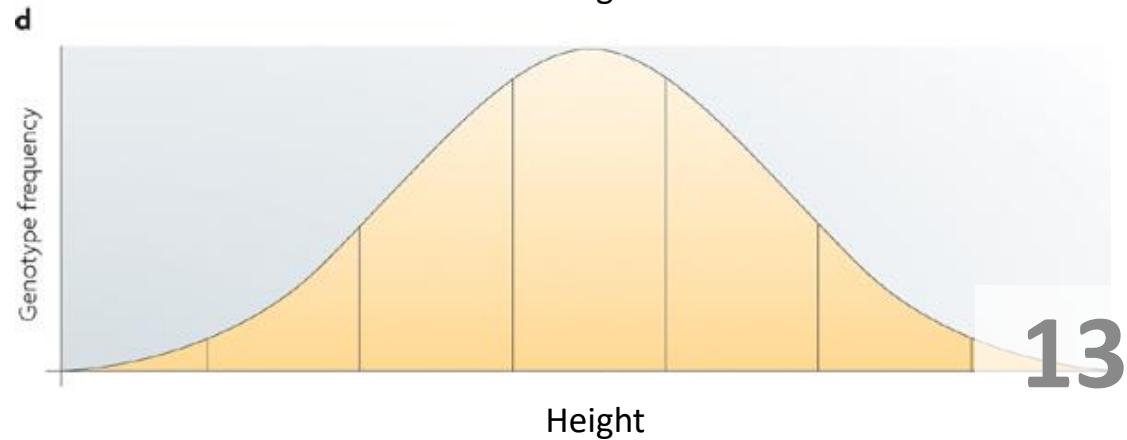


Quantitative traits and Mendelian laws

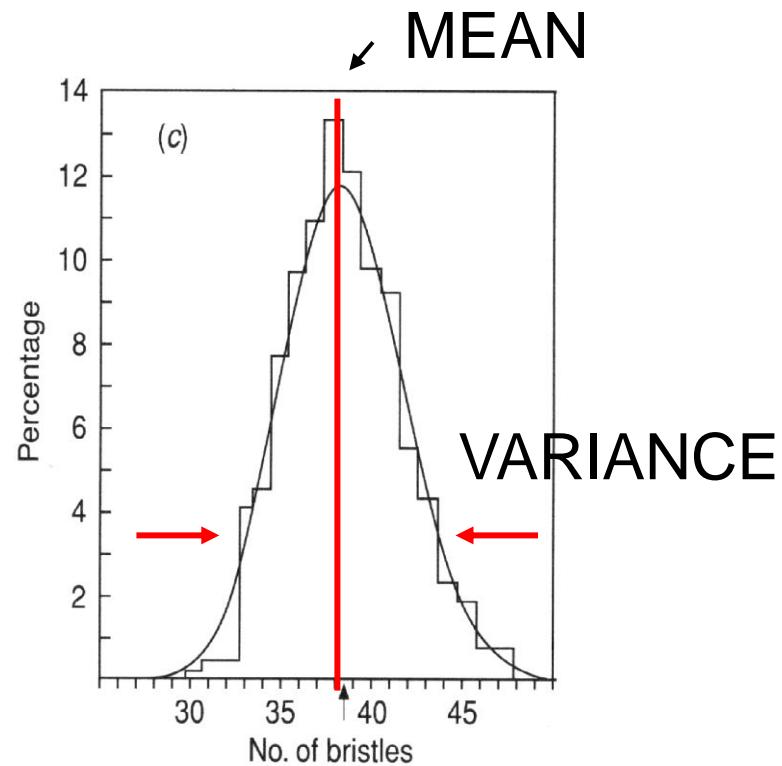
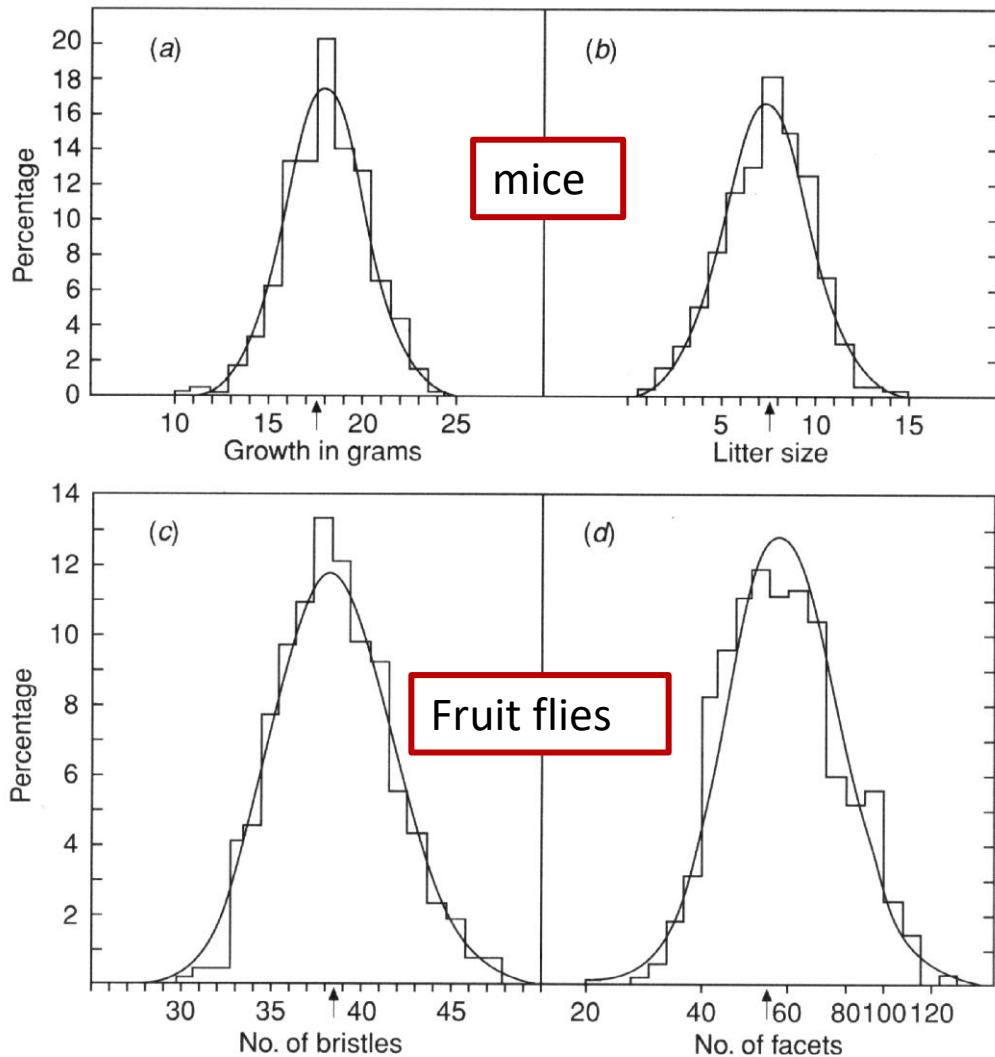
Three
Loci
(A, B & C):



Many
loci:



Normal Distribution of Phenotypes

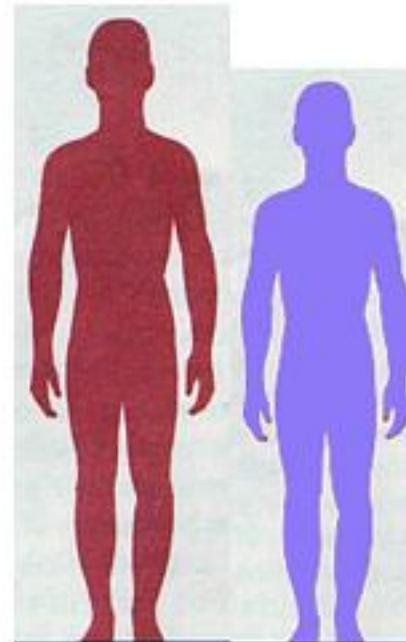


Nature versus Nurture?

- Two influences on the expression of quantitative traits:
 - (1) Genetic (effects of many genes)
 - (2) Environmental (effects of environment)

- E.g. height:

Difference due
to different
genes



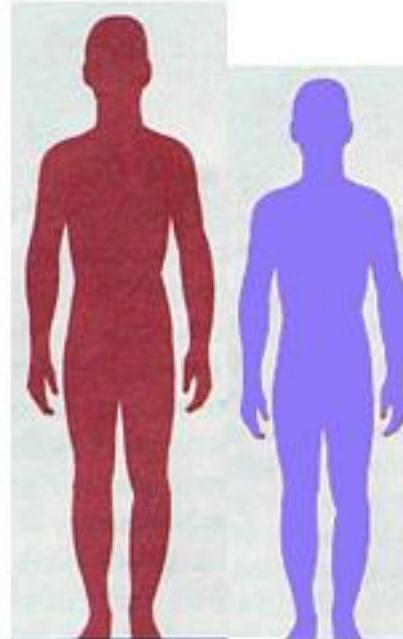
1. Normal
development

Nature versus Nurture?

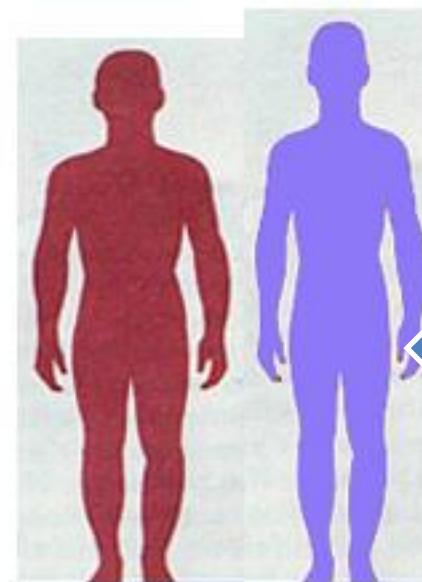
- Two influences on the expression of quantitative traits:
 - (1) Genetic (effects of many genes)
 - (2) Environmental (effects of environment)

- E.g. height:

Difference due
to different
genes



1. Normal
development

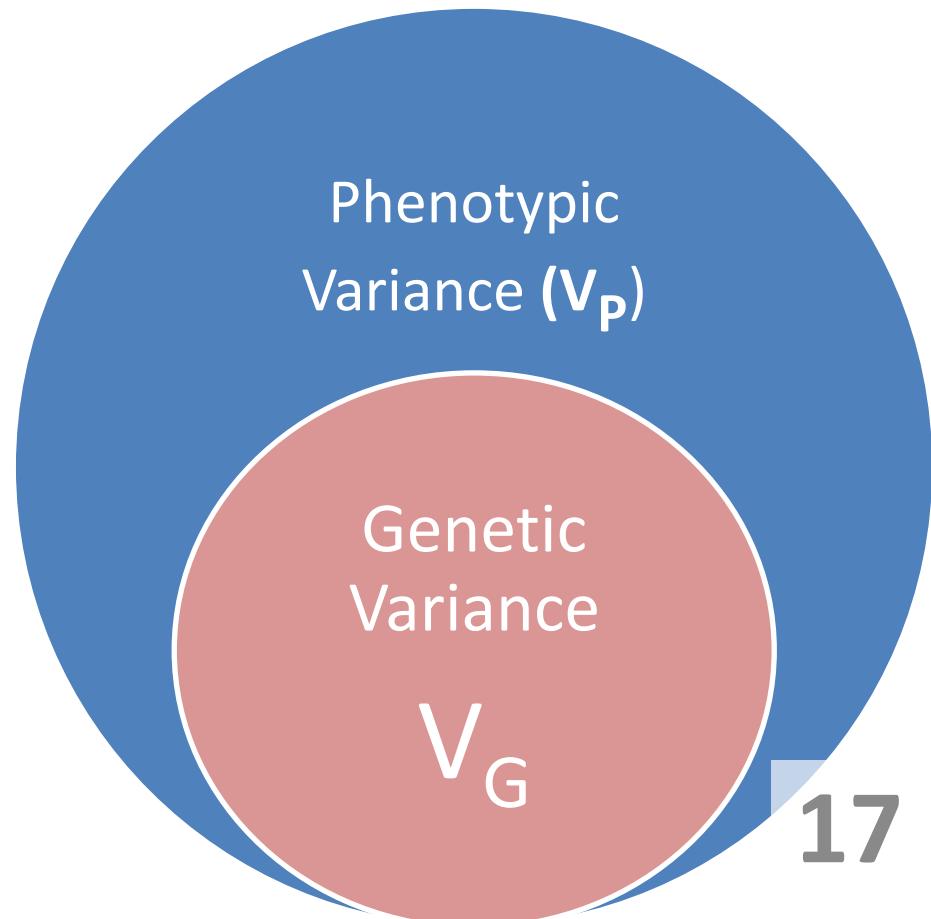


2. Red guy
smoked!

Difference
due to
genes & env.

Partitioning trait variation:

- Most traits influenced by both genes and environment (nature & nurture)
- If we represent all the **phenotypic variation** in the population as a solid circle, we can recognise the portion that is due to different genes
- This is called '**genetic variance**'



Heritability (H^2)

What Does it Measure?

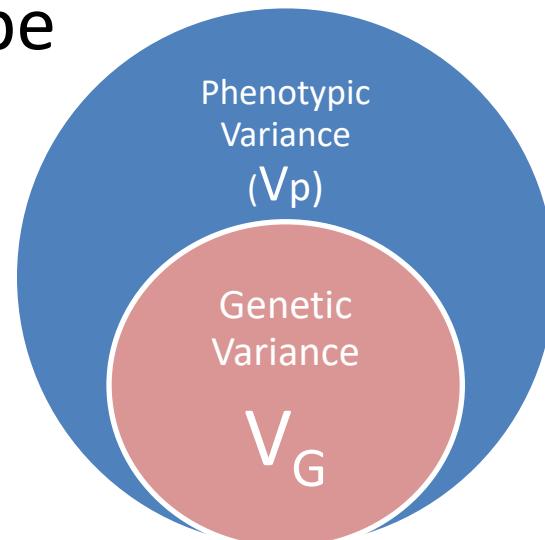
Measure of how much *variation* in a phenotype is caused by *variation* in genotype

Assuming additivity:

$$VP = VG + VE$$



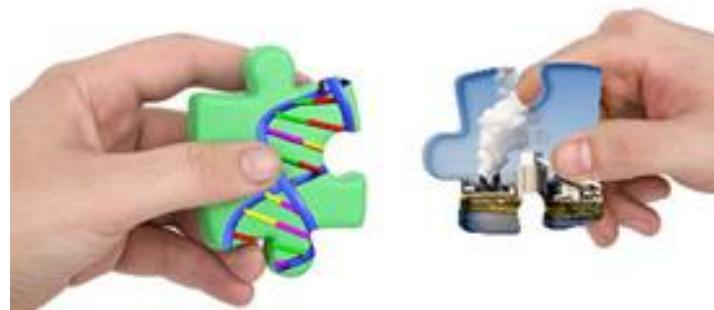
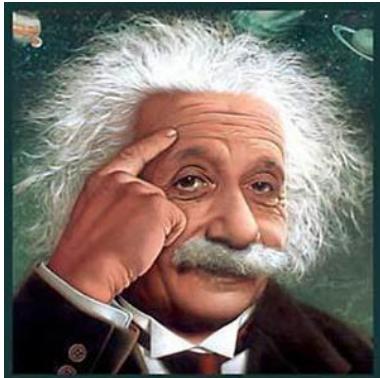
$$H^2 = VG/VP$$



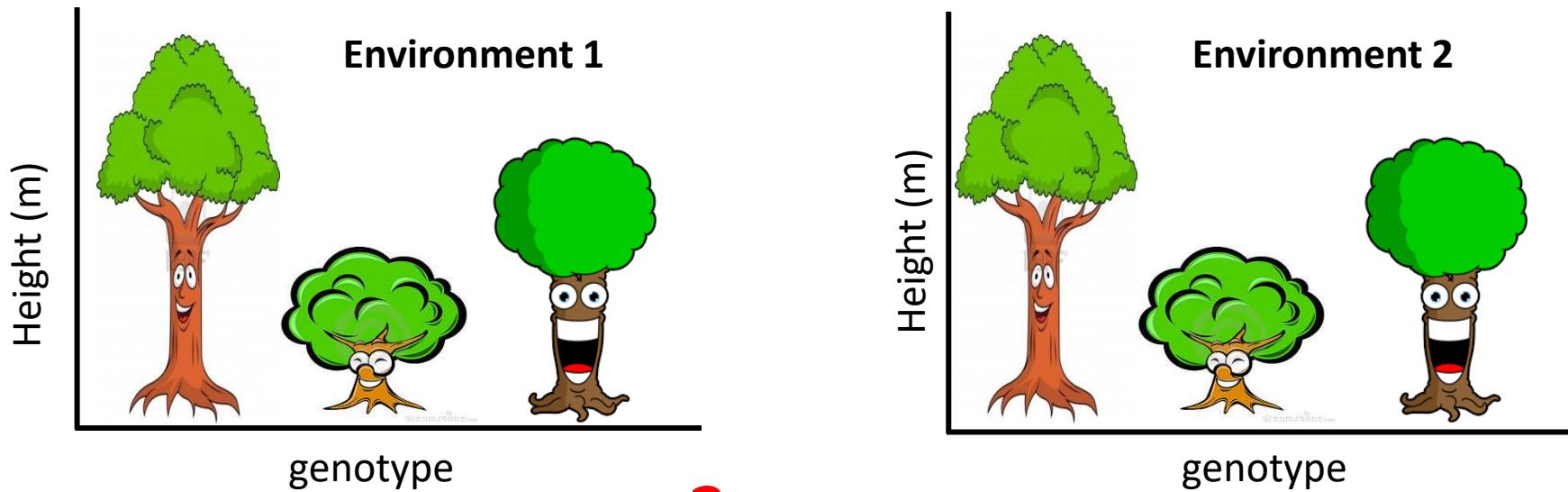
Proportion of Genetic variance to phenotypic variance

Heritability and Causation

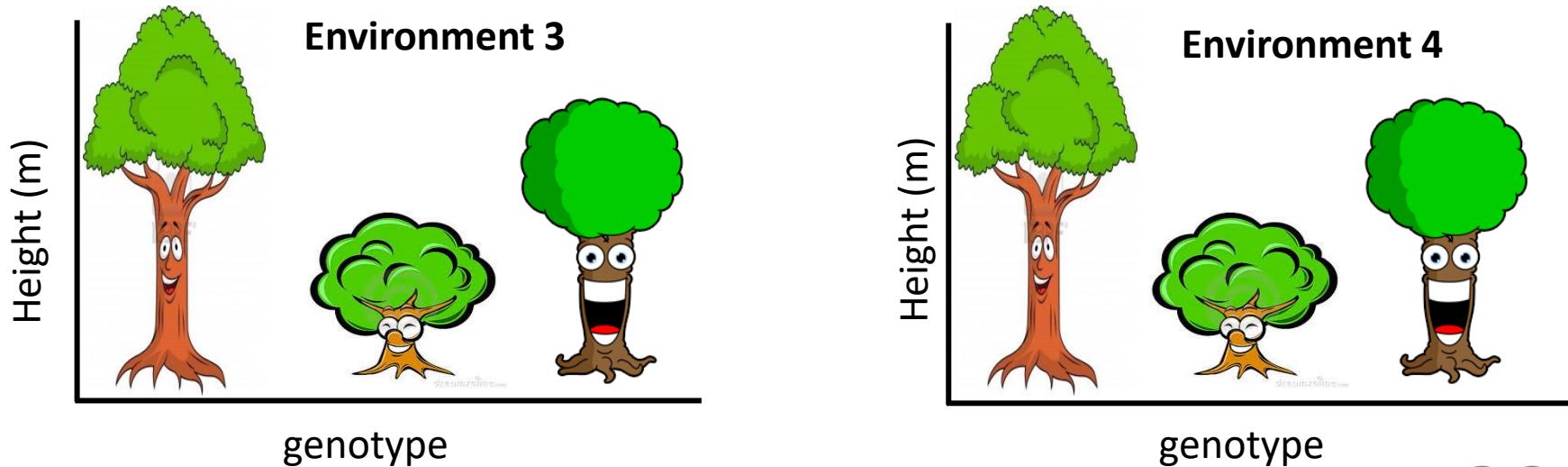
- Heritability can be used to make causal claims about the relative effects of genes (nature) and the environment (nurture)
- Which is more causally important?



- *Specific types* of causal claims

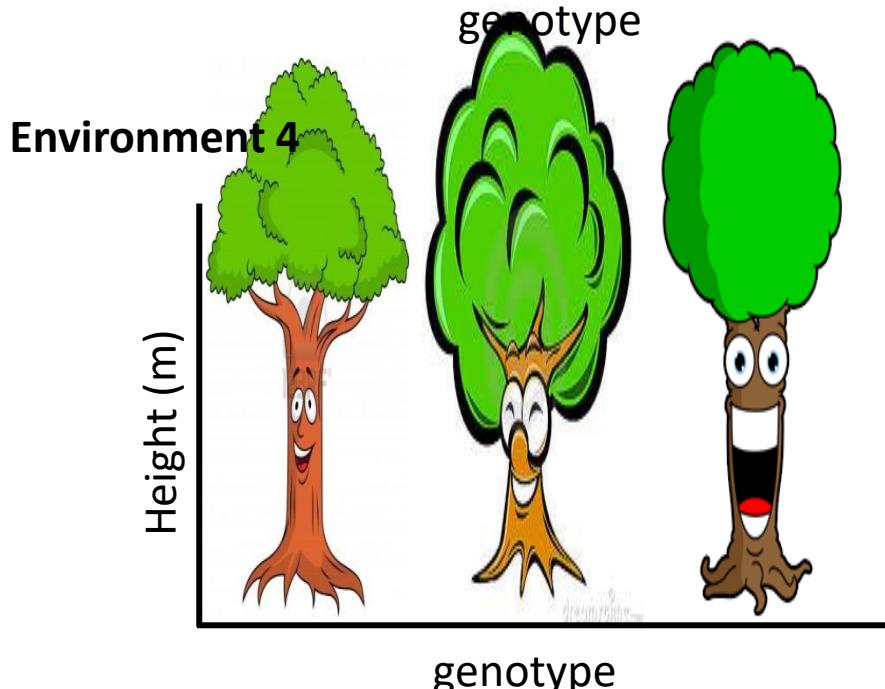
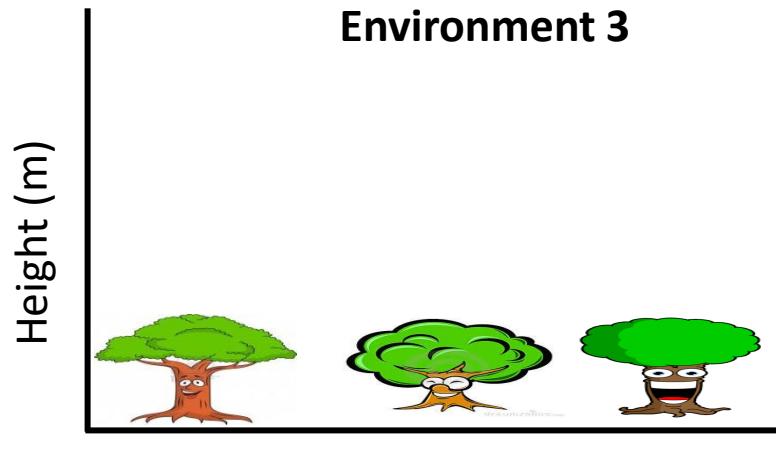
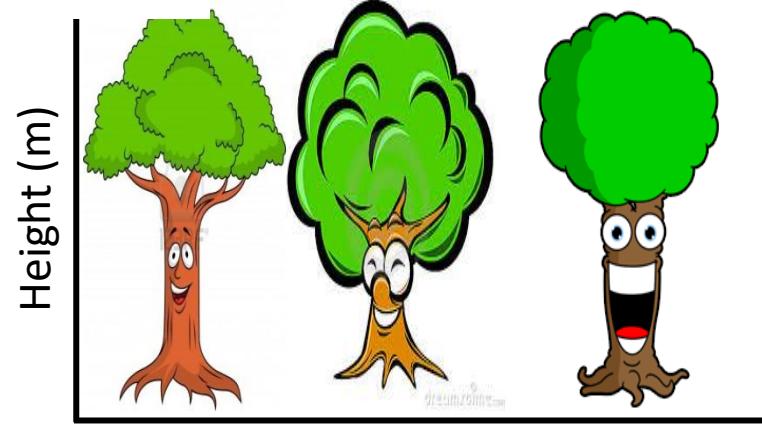


H² HIGH

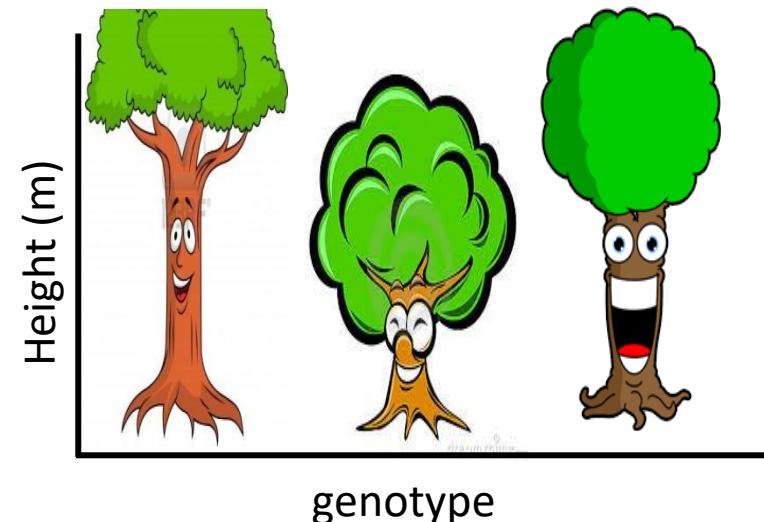
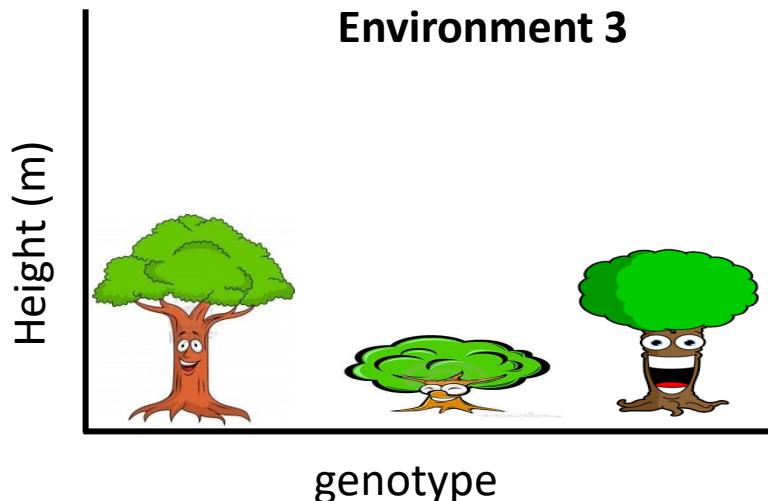
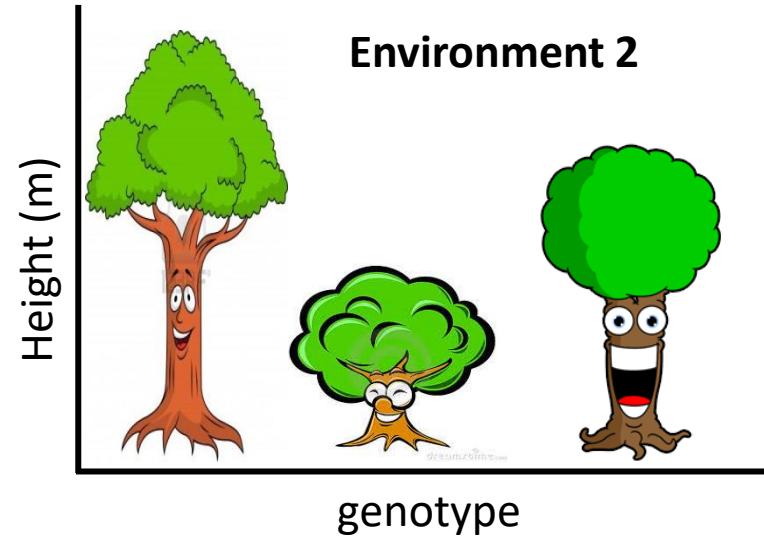
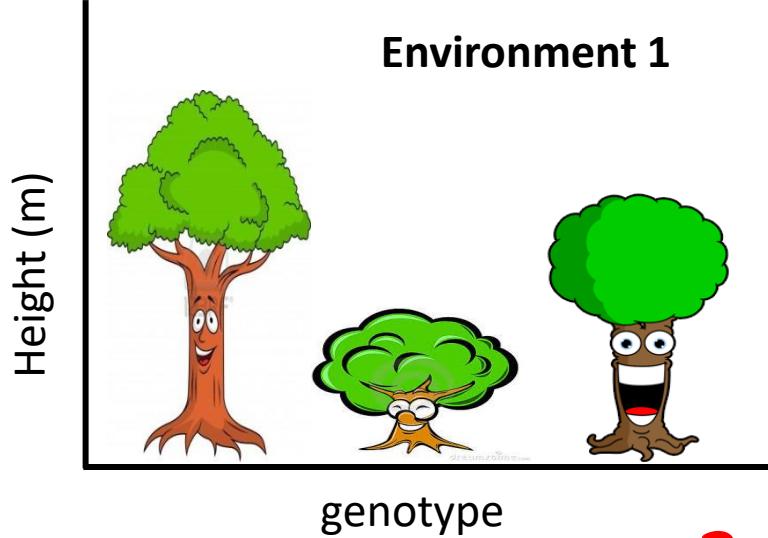


Environmental or Genetic?

H² LOW



Environmental or Genetic?



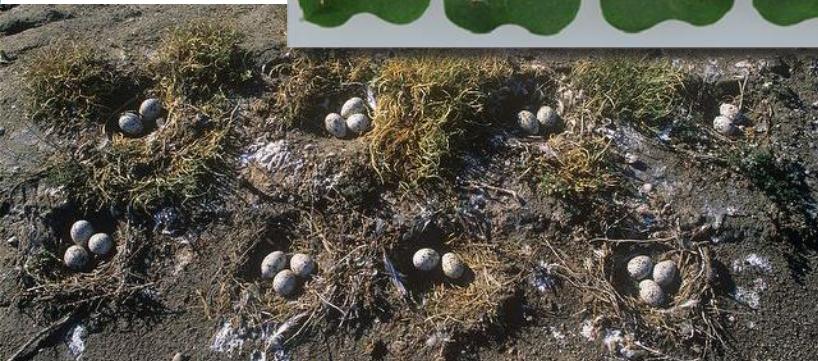
H^2 INTERMEDIATE

Environmental or Genetic?

How does this relate to Conservation Biology?

Knowledge of quantitative fitness traits

- Clutch size
- Resistance to disease / introduced organisms
- Resistance to climate change
- Lifetime fecundity
- Age of first litter
- Days to flowering



Where do the values come from?

Heritability of postchallenge survival, an indicator of disease resistance, was estimated to be 0.35 ± 0.09.

Silverstein, J. T., et al. (2009). Rainbow trout resistance to bacterial cold-water disease is moderately heritable and is not adversely correlated with growth. *Journal of animal science* 87(3), 860-867.

Broad-sense heritabilities ranged from 0.55 in the F₂ population (Fargo, N.D.) to 0.95 in the F_{2:4} progenies at

León, A. J., Lee, M., & Andrade, F. H. (2001). Quantitative trait loci for growing degree days to flowering and photoperiod response in sunflower (*Helianthus annuus* L.). *Theoretical and Applied Genetics*, 102(4), 497-503.

and the heritability estimates (95 % confidence intervals) for boldness and aggressiveness were 0.76 (0.49, 0.90) and 0.36 (0.10, 0.72) respectively. Furthermore, there were

Ariyomo, T. O., Carter, M., & Watt, P. J. (2013). Heritability of Boldness and Aggressiveness in the Zebrafish. *Behavior genetics*, 43(2), 161-167.

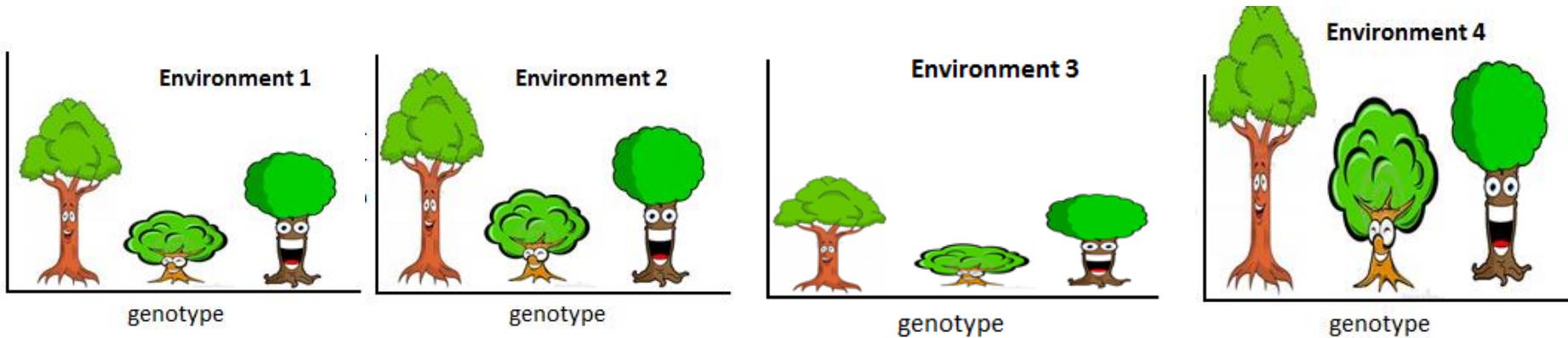
$$H^2 = VG / VP$$

$$VP = VG + VE$$

VG?

VE?

Where do the values come from?



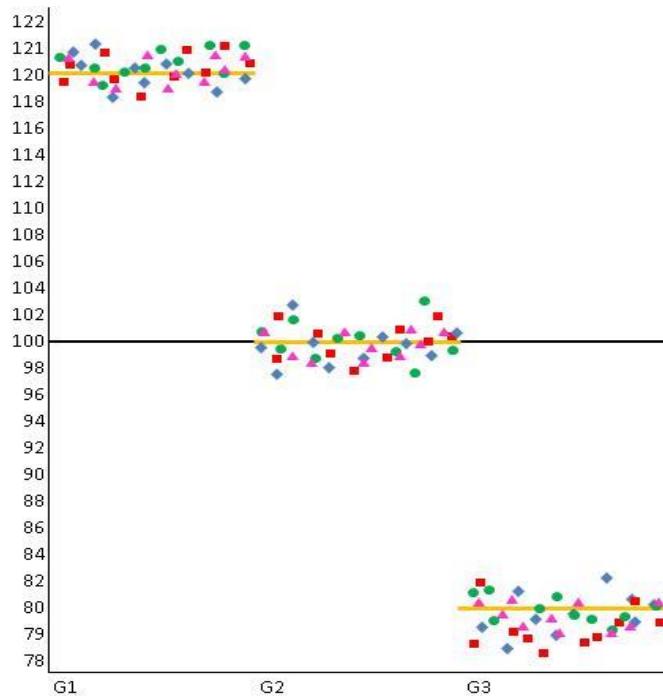
	E1 (75)	E2 (75)	E3 (75)	E4 (75)
G1 (100)				
G2 (100)				
G3 (100)				

The height of the trees are measured...

	E1 ($n_e = 75$)	E2 ($n_e = 75$)	E3 ($n_e = 75$)	E4 ($n_e = 75$)	Genotype Means
G1 ($n_g = 100$)	$\mu = 120$	$\mu = 118$	$\mu = 119$	$\mu = 123$	$\mu_{G1} = 120$
G2 ($n_g = 100$)	$\mu = 100$	$\mu = 101$	$\mu = 98$	$\mu = 101$	$\mu_{G2} = 100$
G3 ($n_g = 100$)	$\mu = 80$	$\mu = 81$	$\mu = 78$	$\mu = 81$	$\mu_{G3} = 80$
Environment Means	$\mu_{E1} = 100$	$\mu_{E2} = 100$	$\mu_{E3} = 98.33$	$\mu_{E4} = 101.67$	$\ddot{X} = 100$

Trees with G1 genotype are significantly taller than G2, who in turn are taller than G3. Differences between environments are negligible.

	E1 ($n_e = 75$)	E2 ($n_e = 75$)	E3 ($n_e = 75$)	E4 ($n_e = 75$)	$N=300$
G1 ($n_g=100$)	$\mu = 120$	$\mu = 118$	$\mu = 119$	$\mu = 123$	$\mu_{G1} = 120$
G2 ($n_g=100$)	$\mu = 100$	$\mu = 101$	$\mu = 98$	$\mu = 101$	$\mu_{G2} = 100$
G3 ($n_g=100$)	$\mu = 80$	$\mu = 81$	$\mu = 78$	$\mu = 81$	$\mu_{G3} = 80$
$N=300$	$\mu_{E1} = 100$	$\mu_{E2} = 100$	$\mu_{E3} = 98.33$	$\mu_{E4} = 101.67$	$\bar{X} = 100$



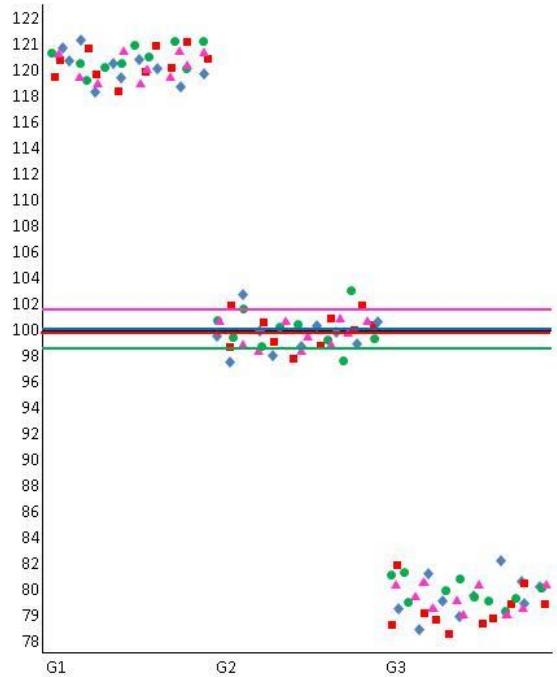
- ◆ E1
- E2
- E3
- ▲ E4

$$VG = \frac{(\mu_{G1} - \bar{X})^2 + (\mu_{G2} - \bar{X})^2 + (\mu_{G3} - \bar{X})^2}{nG}$$

$$VG = \frac{(120 - 100)^2 + (100 - 100)^2 + (80 - 100)^2}{3}$$

$$VG = 266.67$$

	E1 ($n_e = 75$)	E2 ($n_e = 75$)	E3 ($n_e = 75$)	E4 ($n_e = 75$)	$N=300$
G1 ($n_g=100$)	$\mu = 120$	$\mu = 118$	$\mu = 119$	$\mu = 123$	$\mu_{G1} = 120$
G2 ($n_g=100$)	$\mu = 100$	$\mu = 101$	$\mu = 98$	$\mu = 101$	$\mu_{G2} = 100$
G3 ($n_g=100$)	$\mu = 80$	$\mu = 81$	$\mu = 78$	$\mu = 81$	$\mu_{G2} = 80$
$N=300$	$\mu_{E1} = 100$	$\mu_{E2} = 100$	$\mu_{E3} = 98.33$	$\mu_{E4} = 101.67$	$\bar{X} = 100$



$$VE = \frac{(\mu_{E1} - \bar{X})^2 + (\mu_{E2} - \bar{X})^2 + (\mu_{E3} - \bar{X})^2 + (\mu_{E4} - \bar{X})^2}{n_E}$$

$$VE = \frac{(100 - 100)^2 + (100 - 100)^2 + (98.33 - 100)^2 + (101.67 - 100)^2}{4}$$

$$VE = 1.38$$

Getting the heritability estimate

The phenotypic variance is assumed to be additive

$$VP = VG + VE$$

Heritability (H^2) is the proportion of the genotypic variance from the total phenotypic variance

$$H^2 = VG / VP \quad \text{or} \quad VG / (VE + VG)$$

Usually a high heritability estimate accurately reflects an example in which differences in genotype are the major cause of differences in phenotype

$$VG = 266.67$$

$$VE = 1.38$$

$$VP = 268.05$$

Genetic var (H^2)

$$= VG / VP$$

$$= 266.67 / 268.05$$

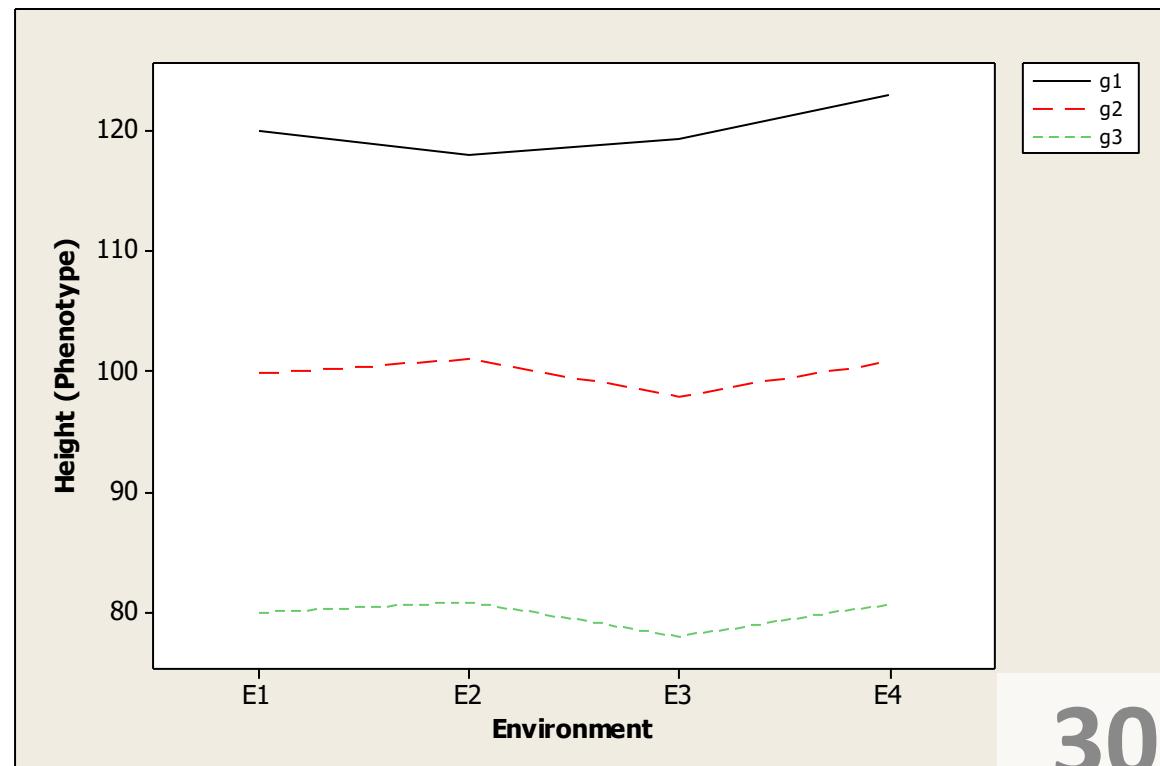
$$= 0.9948$$

Environmental var

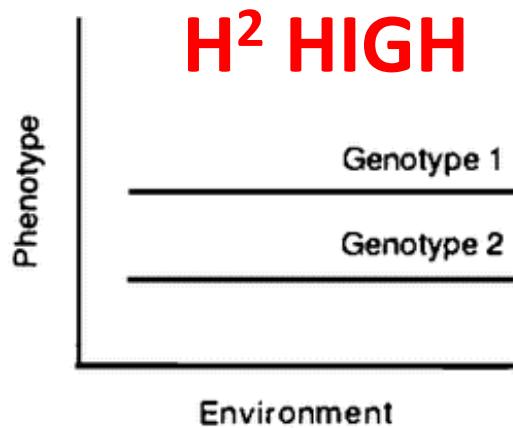
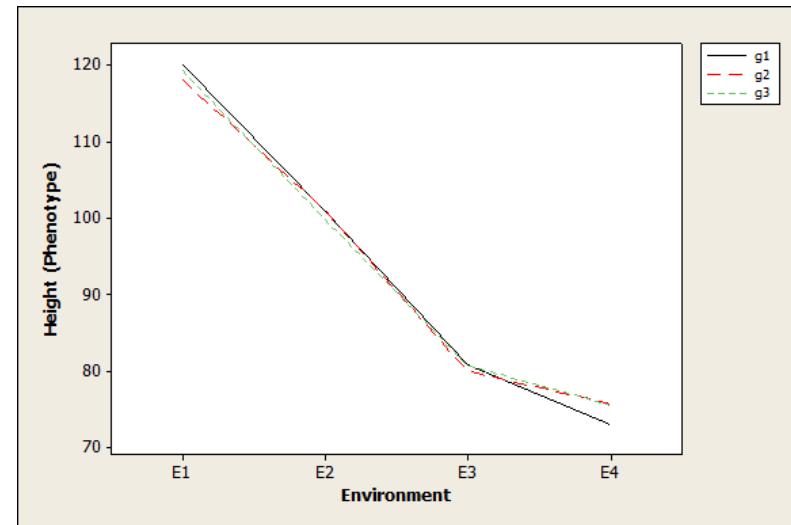
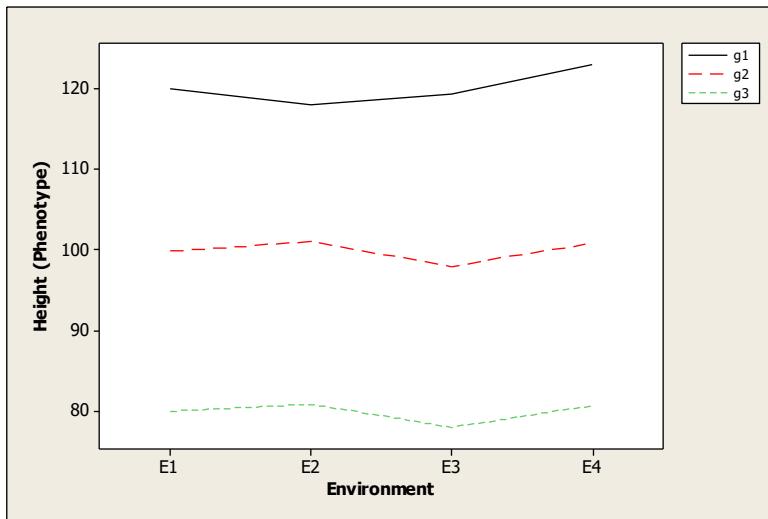
$$= VE / VP$$

$$= 1.38 / 268.05$$

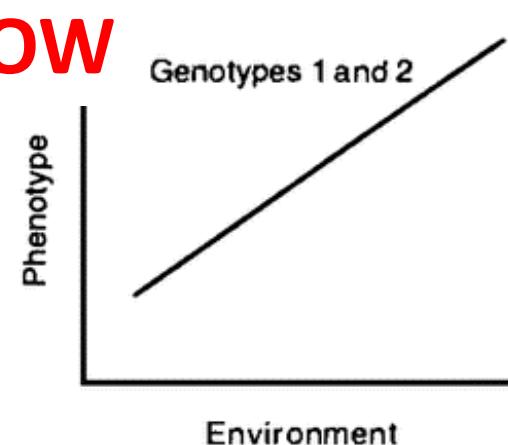
$$= 0.0052$$



Norm of Reaction

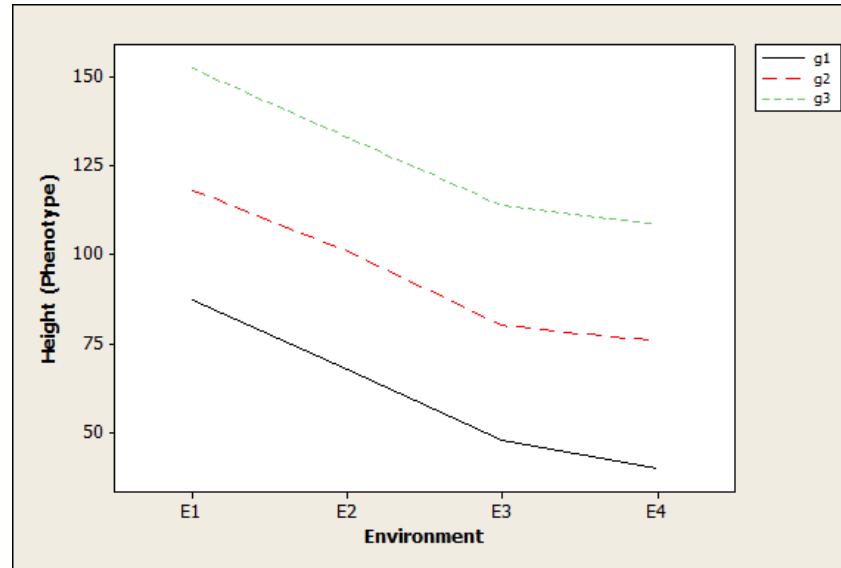


a) Biological determinism

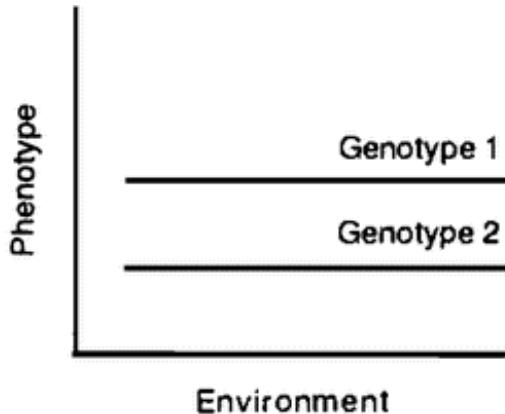


b) Social determinism

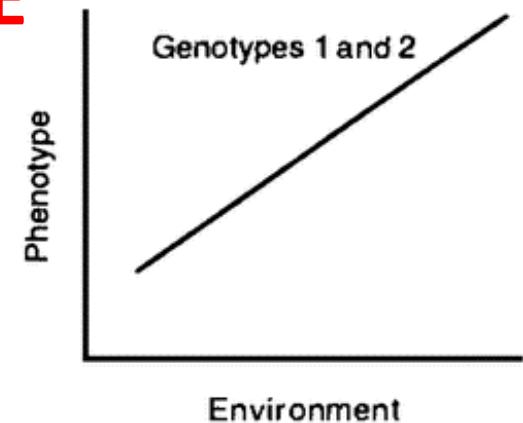
Norm of Reaction



H² INTERMEDIATE

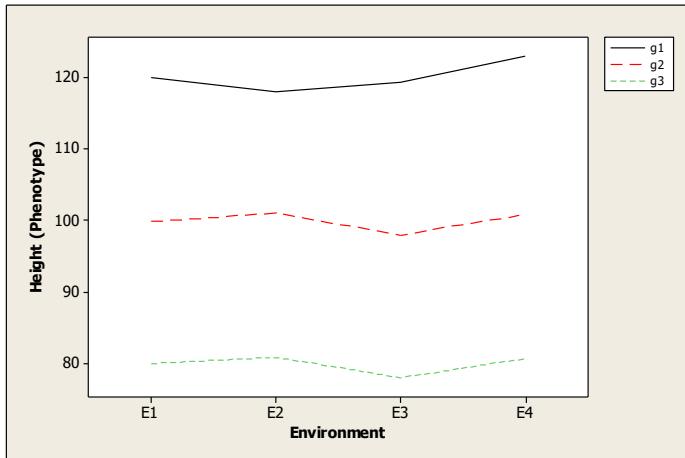


a) Biological determinism

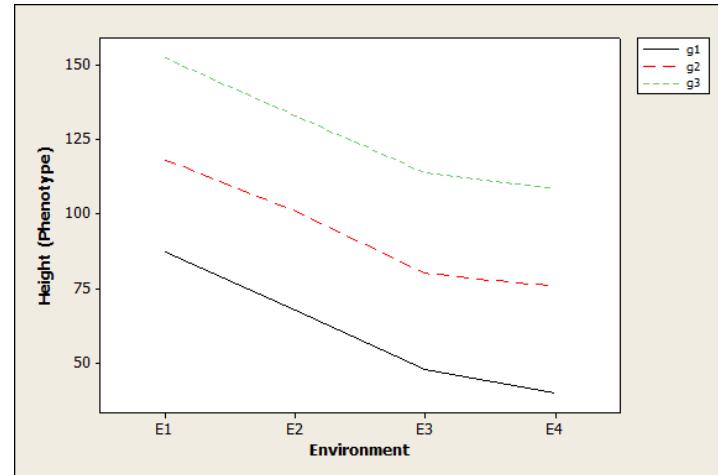


b) Social determinism

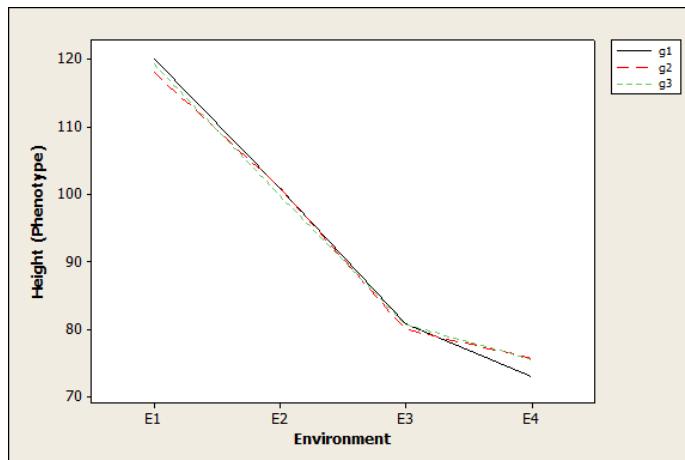
Norm of Reaction - GxE (Interaction)



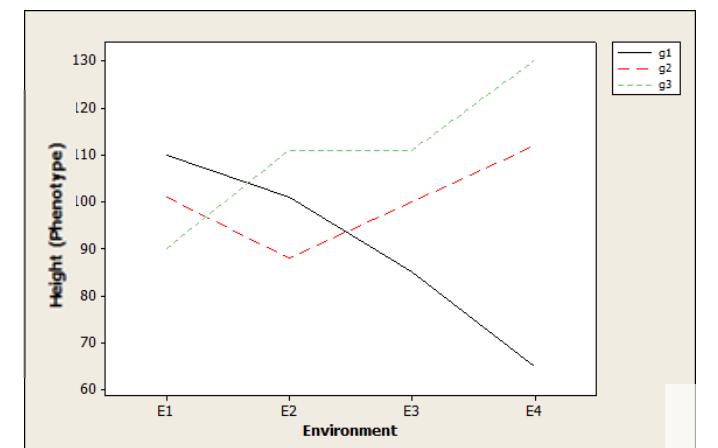
H² HIGH



H² INTERMEDIATE

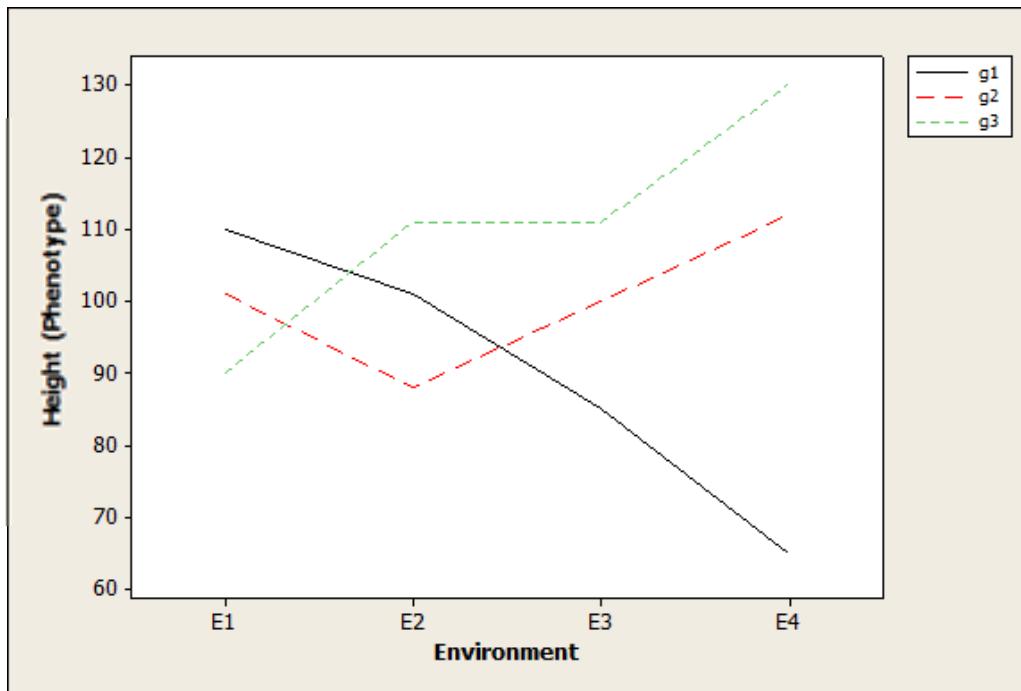


H² LOW



H² ??

$$H^2 = 0.968 \text{ (VERY HIGH!)}$$



$$VG = 266.67$$

$$VE = 8.8089$$

$$VP = 275.4789$$

$$266.67 / 275.4789$$

$$H^2 = 0.968$$

	E1	E2	E3	E4	Means
G1	110	100	85	65	90
G2	100	88	100	112	100
G3	90	110	110	130	110
Means	100	99.33333	98.33333	102.3333	$\ddot{X} = 100$

$$VG = \frac{(\mu_{G1} - \ddot{X})^2 + (\mu_{G2} - \ddot{X})^2 + (\mu_{G3} - \ddot{X})^2}{n_G}$$

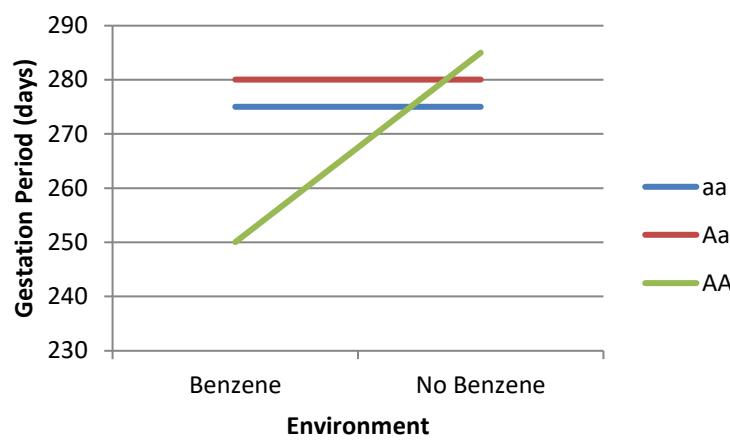
$$VG = 266.67$$

$$VE = \frac{(\mu_{E1} - \ddot{X})^2 + (\mu_{E2} - \ddot{X})^2 + (\mu_{E3} - \ddot{X})^2 + (\mu_{E4} - \ddot{X})^2}{n_E}$$

$$VE = 8.8089$$

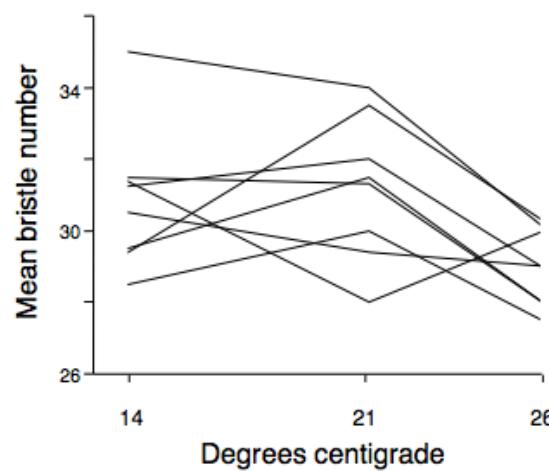
GxE (Gene-Environment Interaction)

- Trait expression varies over different environments – may not be directional
- Genome produces different phenotypes depending on developmental environment
- Example 1: benzene exposure is significantly associated with shorter gestation periods in pregnant women possessing the AA allele of *CYP1A1*, while there is no association for those with Aa or aa (Wang et al. 2000)



GxE (Gene-Environment Interaction)

- Trait expression varies over different environments – may not be directional
- Genome produces different phenotypes depending on developmental environment
- Example 2: Bristle number in drosophila is determined by genotypic differences, and temperature. How temperature affects bristle number depends on the flies genetic background (Gupta and Lewontin 1981)



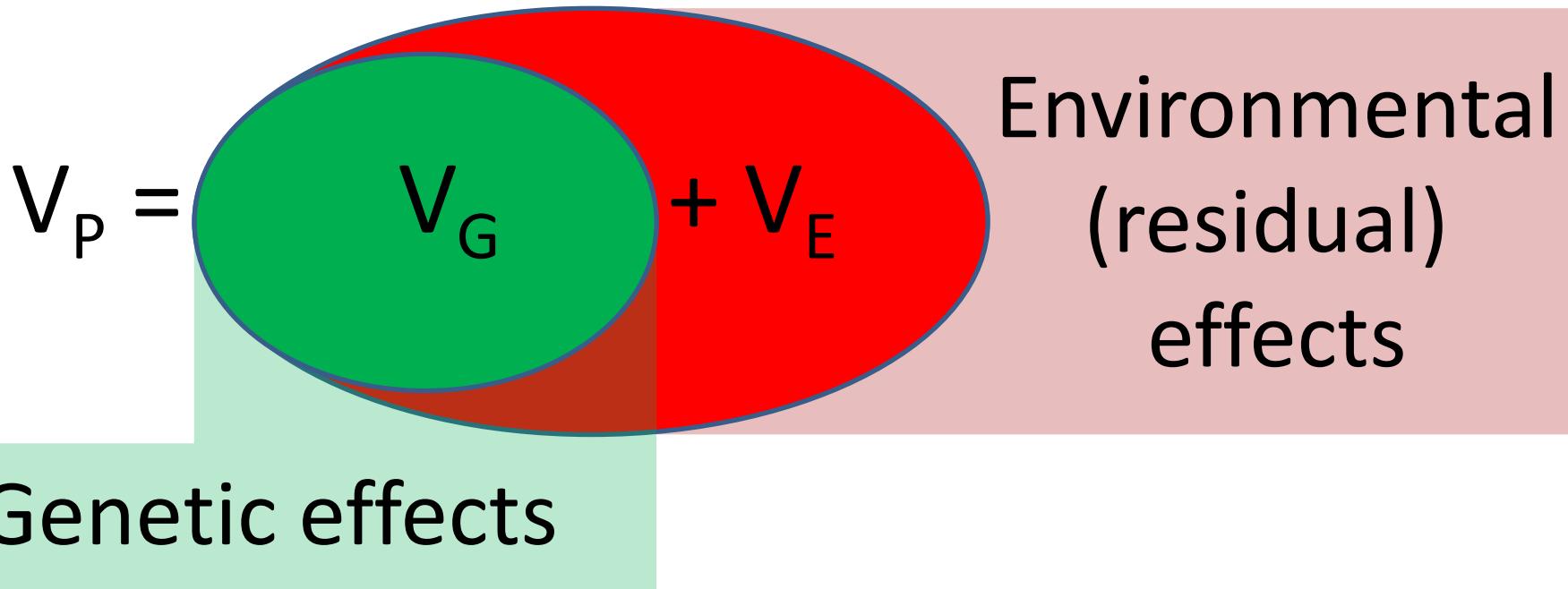
GxE (Gene-Environment Interaction)

- GxE doesn't always mean an high heritability
- Could also result in low H^2 if not accounted for 
- Demonstrates when interaction occurs and is unaccounted for – one cannot get a good understanding without looking at the NOR.
- GxE can lead to a heritability estimate that does not accurately reflect the contribution of environmental or genetic variation
- Can add GxE term into heritability model so it no longer skews results

Non-Additive

$$VP = VG + VE + \textcolor{red}{V_{GxE}} + 2\text{CovGE} + \text{Error}$$

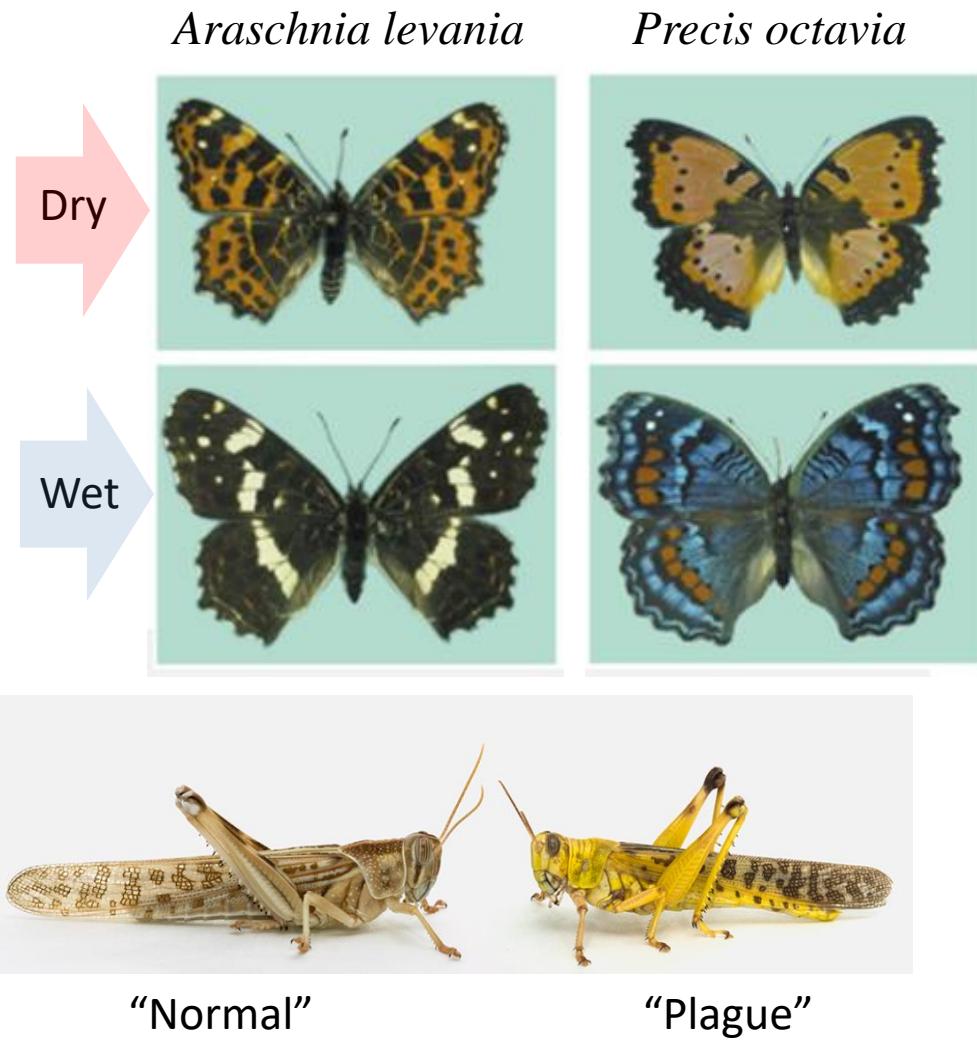
NATURE... versus ...NURTURE



....assuming that genetic and environmental effects are **additive**

Phenotypic plasticity

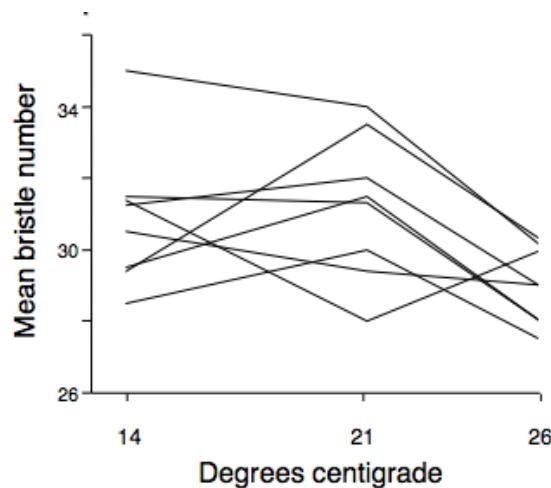
The ability of a single genome to produce different phenotypes depending upon the environment experienced during development



GxE (Gene-Environment Interaction)

GxE = A form of phenotypic plasticity

Single genotype produces multiple phenotypes, depending on environmental interaction.



Summary

- Quantitative traits are characterised by heritability estimates (H^2)
- Estimates partition the relative effects of environmental variation (VE) and genetic variation (VG) on differences in a phenotype (VP) *within a population.*

$$VP = VG + VE$$

$$H^2 = VG / VP$$

- Can estimate the influence of genes just by studying the phenotype (no molecular techniques needed)
- Conservation biologists are interested in the genetic basis of fitness traits
- Limitations to estimates: non-additivity (GxE interaction)

$$VP = VG + VE + V_{GxE}$$

$$H^2 = VG / VP$$

Quantitative Genetics (2)

LECTURE 6: BROAD AND NARROW HERITABILITY & ADAPTIVE POTENTIAL

2022



Previous Lecture

- Quantitative traits are characterised by H^2
- Estimates partition the relative effects of environmental variation (VE) and genetic variation (VG) on differences in a phenotype (VP) *within a population.*

$$VP = VG + VE$$

$$H^2 = VG / VP$$

- Can estimate the influence of genes just by studying the phenotype
- Non-additivity (GxE interaction)

$$VP = VG + VE + V_{GxE}$$

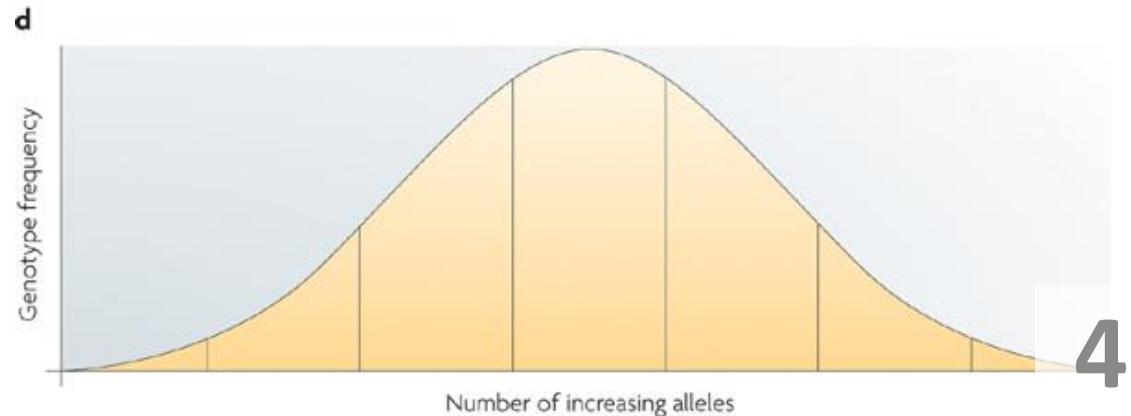
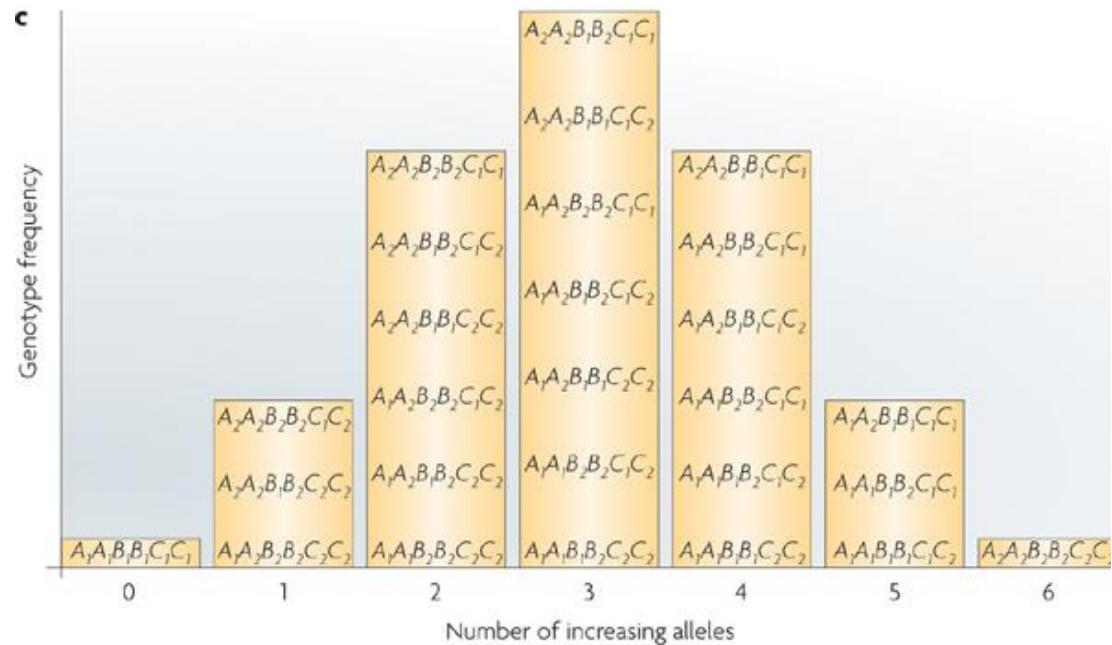
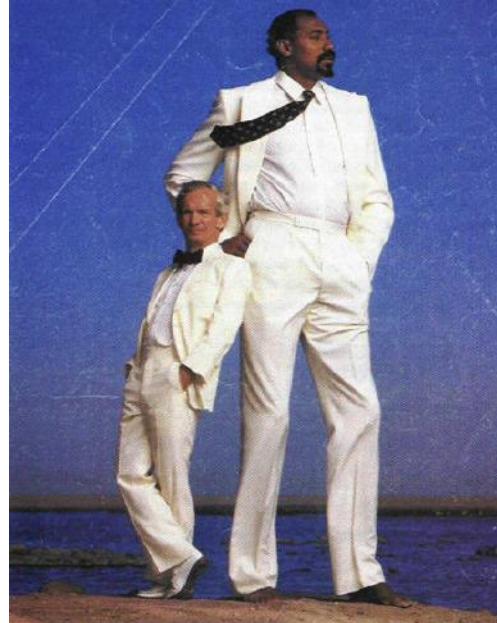
$$H^2 = VG / VP$$

Lecture Outline

- Types of Genetic Variance
- Broad and Narrow Heritability
- Estimation of H^2 and h^2 via Regression
- Heritability and Adaptive Potential
- Estimation of Adaptive Potential (breeders equation)

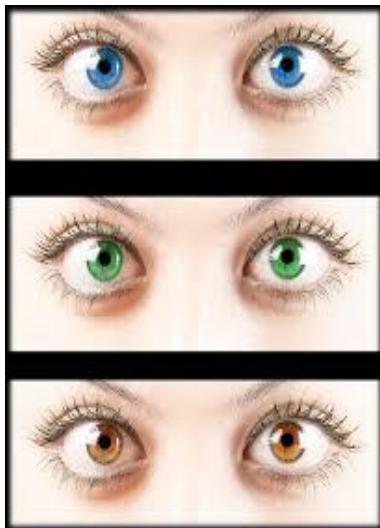
Types of Genetic Variance

Remember that VG is a cumulative effect of many Mendelian interactions...

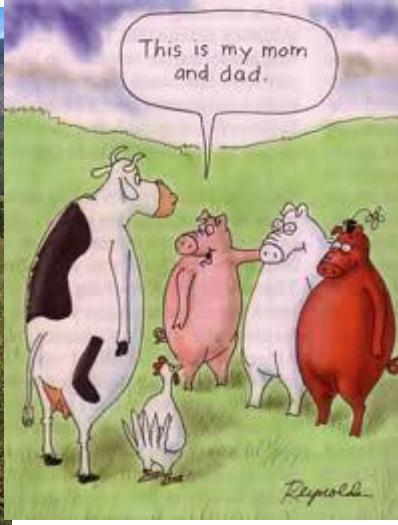
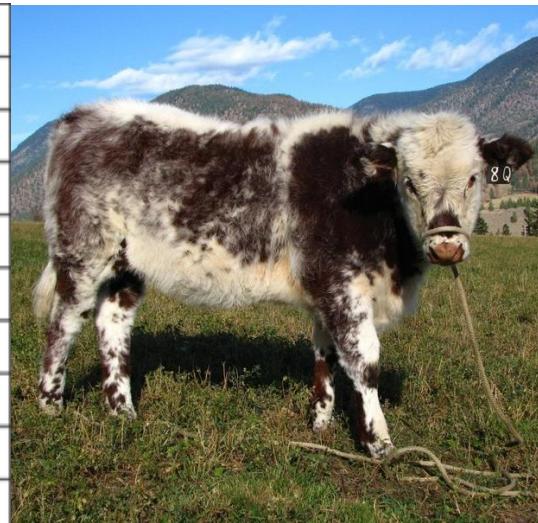


Varieties of Mendelian Genetics

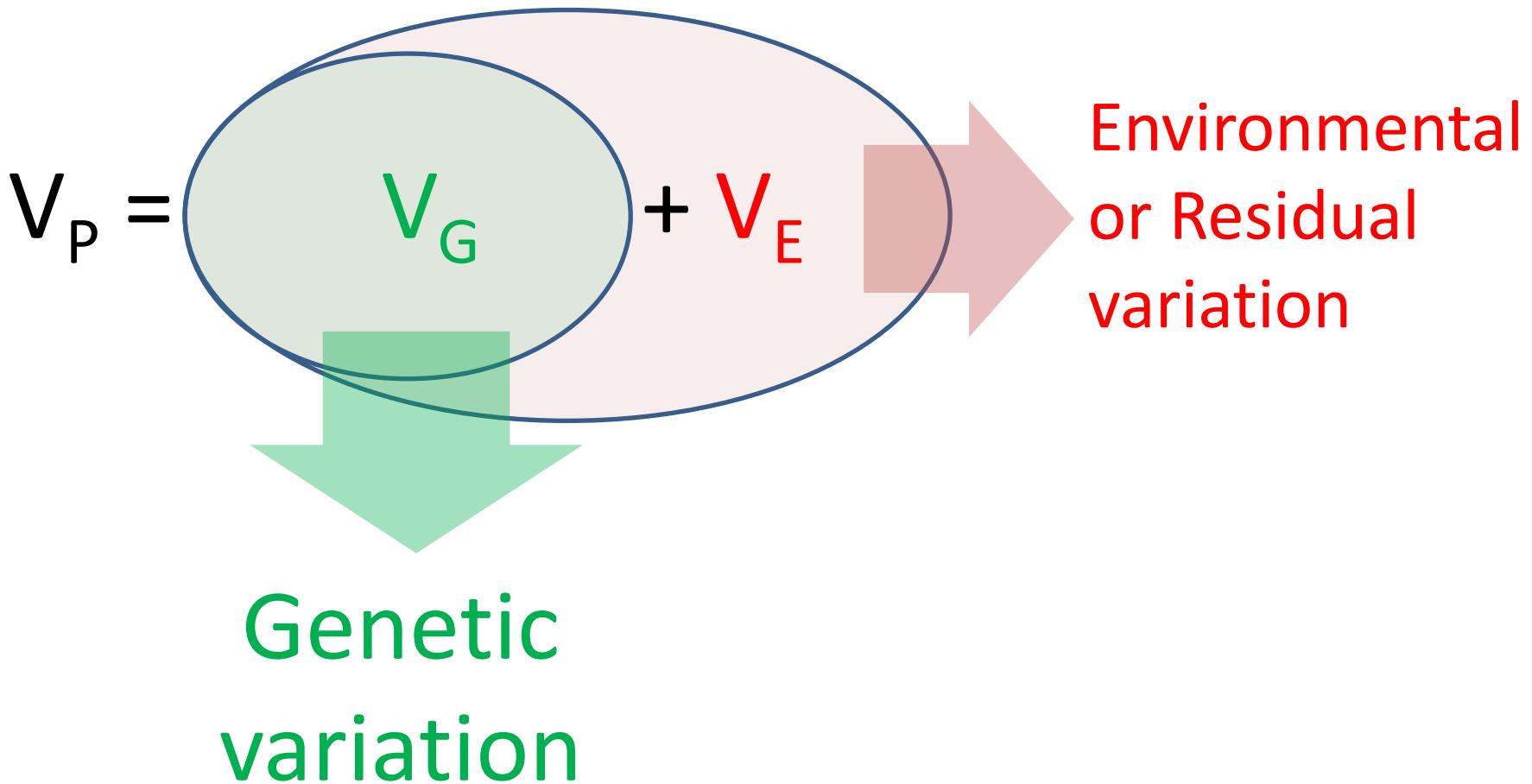
1. Dominance: interaction of alleles at a single loci
2. Epistasis: interaction of alleles at different loci
3. no interaction of alleles or loci



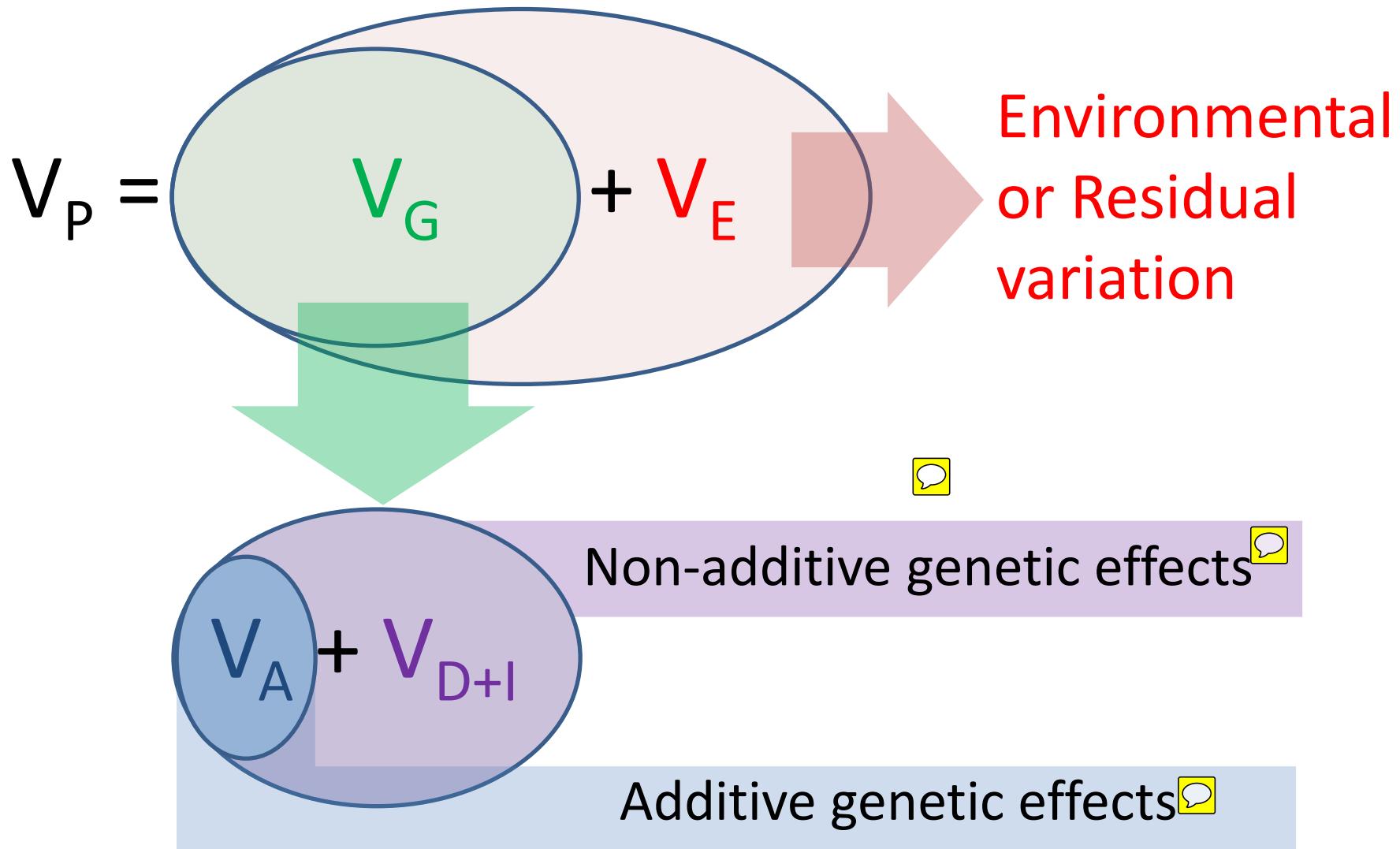
<i>HERC2</i>	<i>Gey</i>	Eye Color
BB	GG	Brown
BB	Gb	Brown
BB	bb	Brown
Bb	GG	Brown
Bb	Gb	Brown
Bb	bb	Brown
bb	GG	Green
bb	Gb	Green
bb	bb	Blue



Variance partitioning



Variance partitioning



Variance partitioning

Phenotypic variance: $V_P = V_G + V_E$

Genetic variance (V_G) = $V_A + V_{D+I}$

Overall: $V_P = [V_A + V_{D+I}] + V_E$

Types of Genetic Variance

VP = Phenotypic Variance

VE = Environmental Variance

VD = Dominance Variance

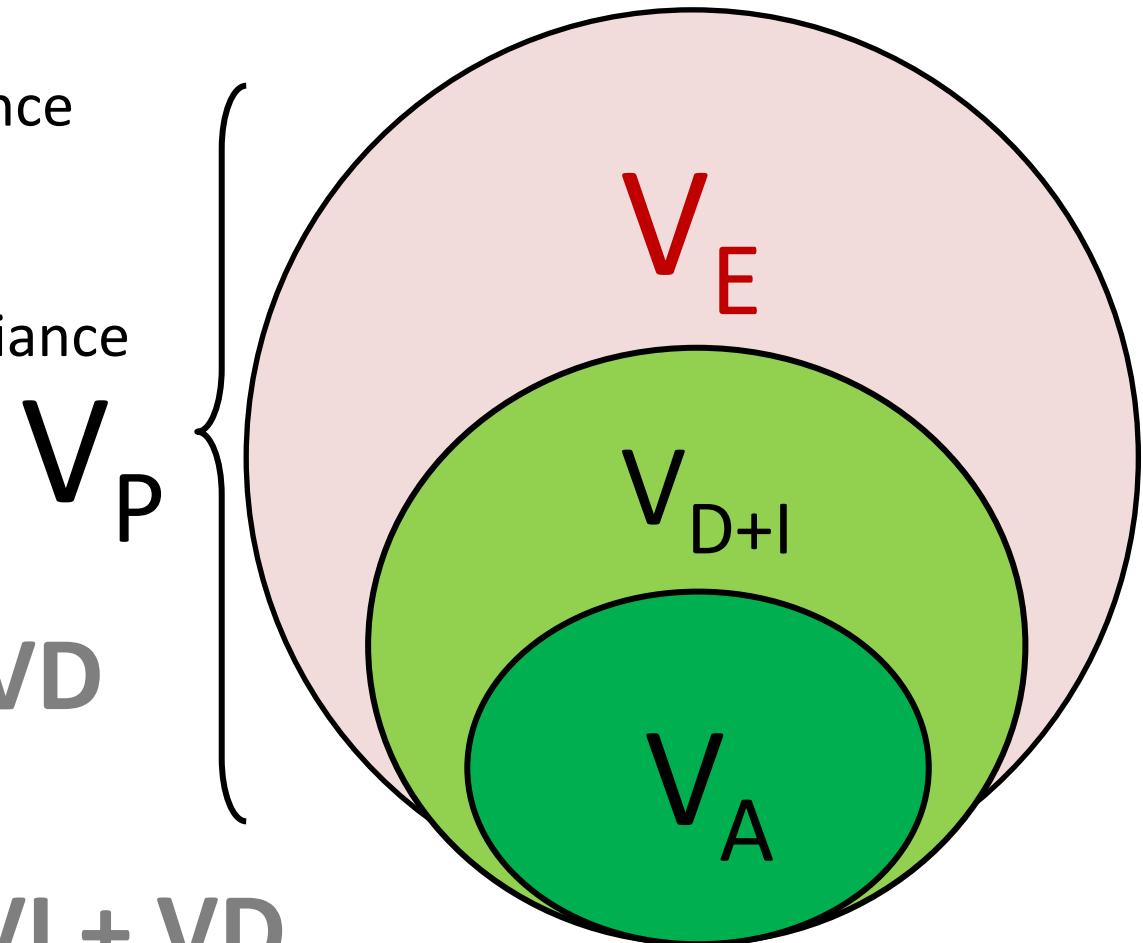
VI = Epistatic Variance

VA = Additive Genetic Variance

$$VG = VA + VI + VD$$

$$VP = VE + VG$$

$$VP = VE + VA + VI + VD$$



Why partition different types of genetic variance?

Different types of genetic variance can be used to estimate different types of heritability

VD + VI:

- phenotypic consequences are all visible at the next generation
- “refresh” each generation
- Not consistently inherited



Used often for psychological traits: we just want to know which is doing more causal work: genes or environment

Broad Heritability (H^2) = VG / VP

$VG = VD + VI + VA$

Why partition different types of genetic variance?

Different types of genetic variance can be used to estimate different types of heritability



VA:

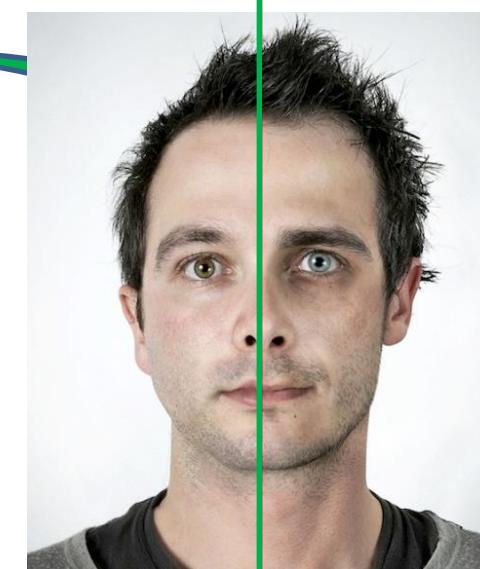
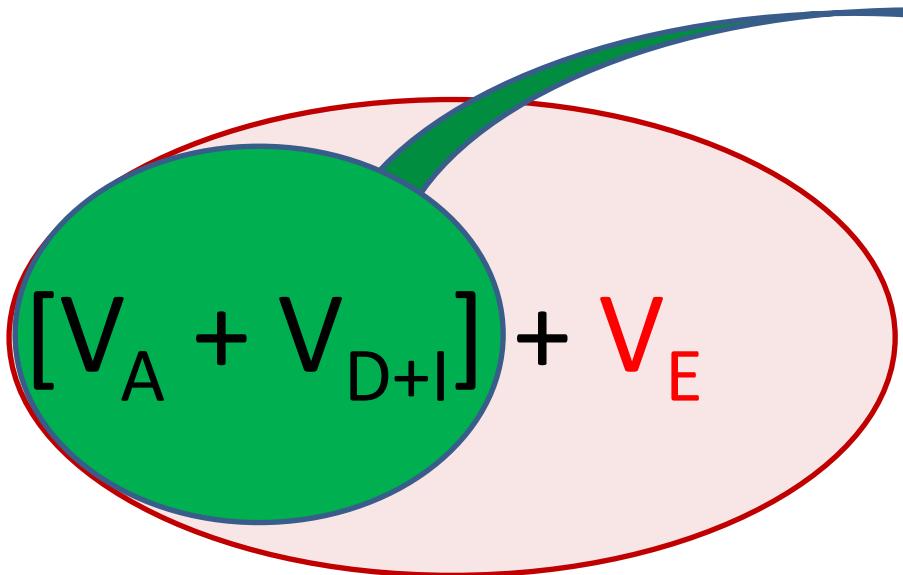
- phenotypic consequences are all visible at the next generation *and at subsequent generations*
- Phenotypic Effects always visible to selection pressures
- Consistently inherited

Used to forecast the evolution of traits over longer time scales: Multiple generations

Can inform us about the ‘evolvability’ of trait

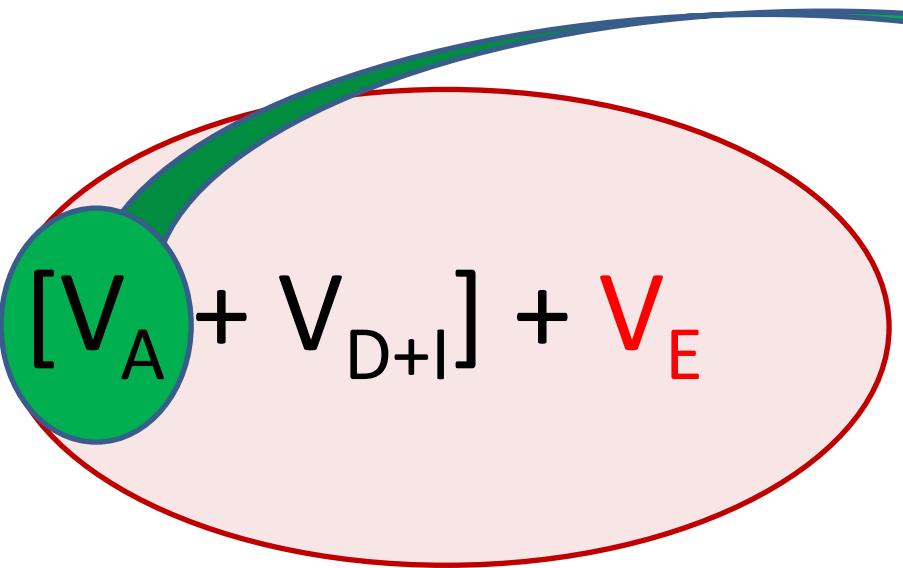
Narrow Heritability (h^2) VA / VP

$$V_P = [V_A + V_{D+I}] + V_E$$

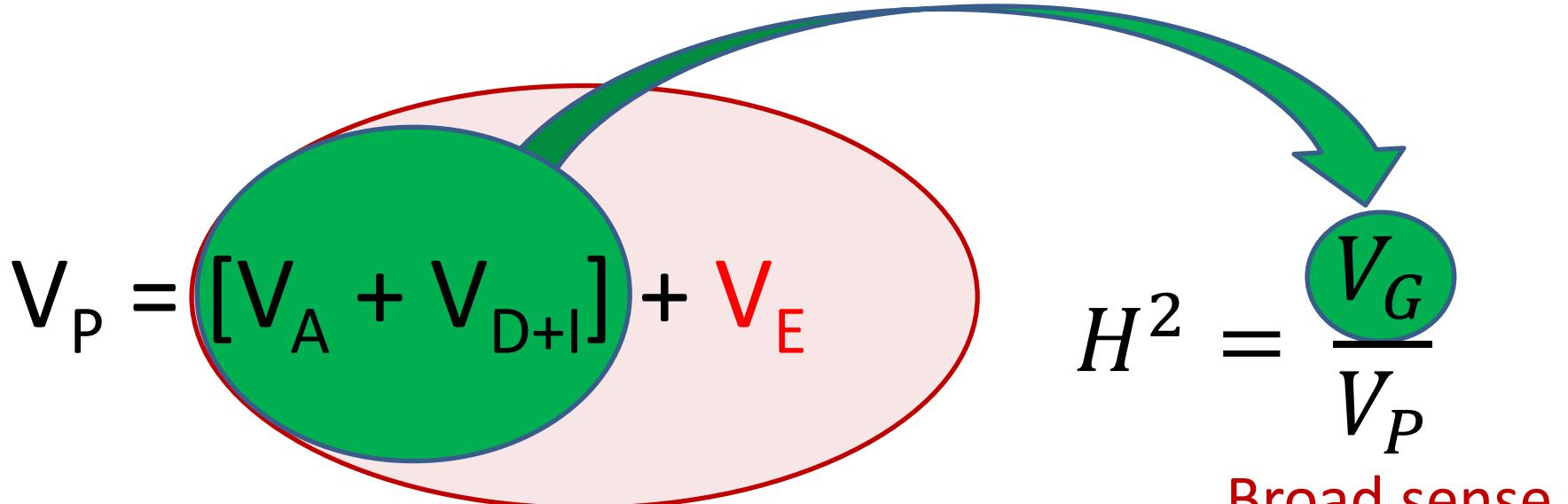


Sibling A Sibling B

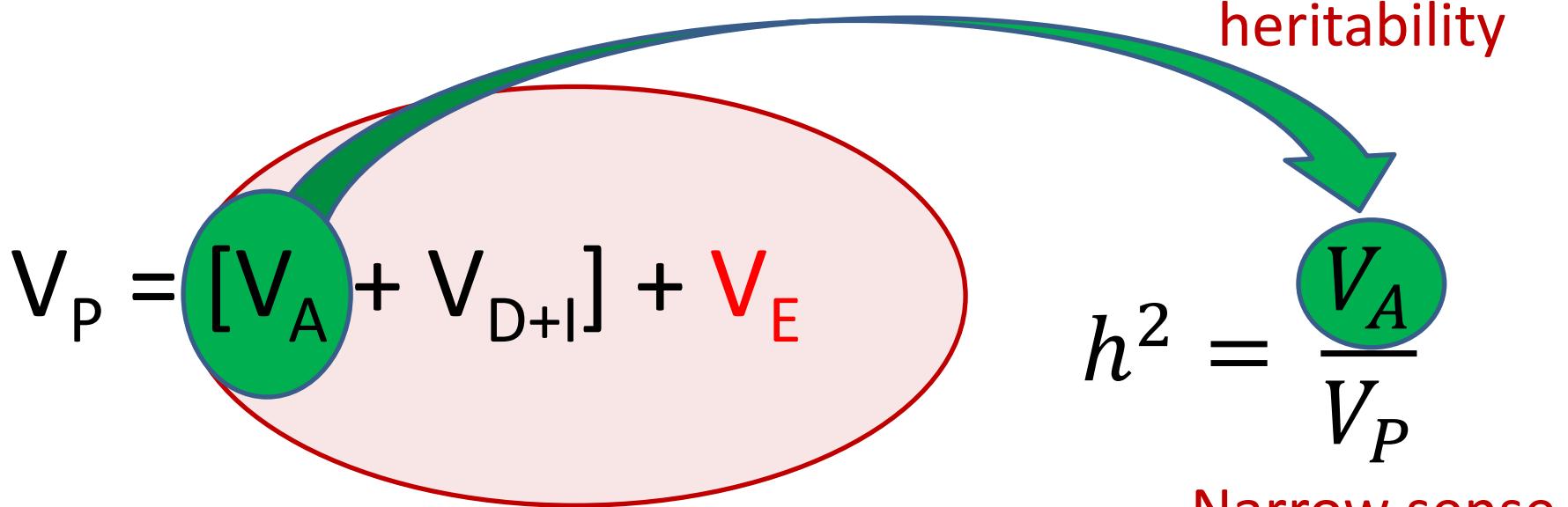
$$V_P = [V_A + V_{D+I}] + V_E$$



Parent Offspring



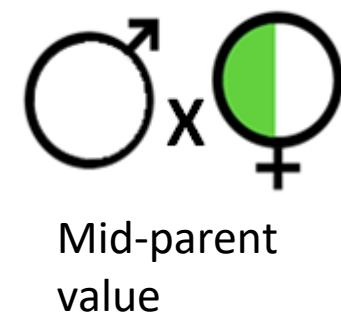
Broad sense
heritability



Narrow sense
heritability

Additive effects model

Quantitative genetic basis of phenotypic “greeness”...



Mean Parental Phenotype
0.25

Sire	Dam
--	++
--	++
--	--
--	++
--	+-
--	-+
--	+-
--	--
--	+-
--	+-

Sire	Dam
20 -	10 +
10 -	

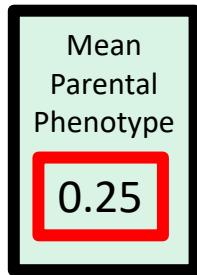
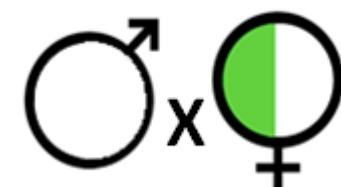
Greeness hypothetically measured from 0 - 1.

Decided by genes at 10 diploid loci
= 10 “on/off” switches

Regardless of genomic position,
+ = more green
- = less green

Additive effects model

Quantitative genetic basis of phenotypic “greeness”...



Sire	Dam
--	++
--	++
--	--
--	++
--	+-
--	-+
--	+-
--	--
--	+-
--	+-

Sire	Dam
20 -	10 +
10 -	

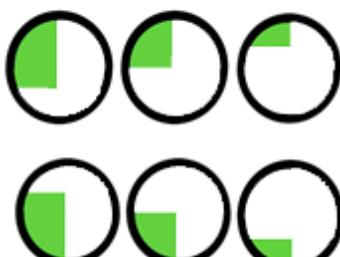
For each offspring:

Half (10) genes from father
Other half from mother

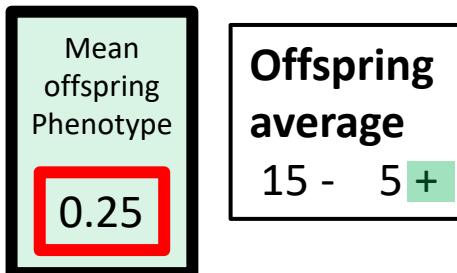
Here,

All paternal genes “off” for greeness
50 % of maternal genes “off”

Average of 5 (25%) “on” genes per
offspring = 0.25 greeness

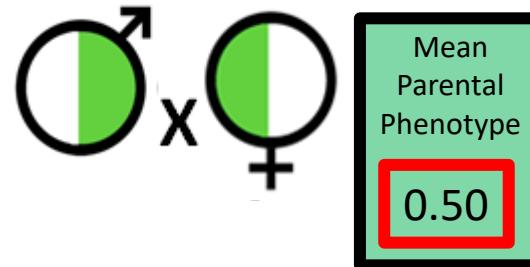
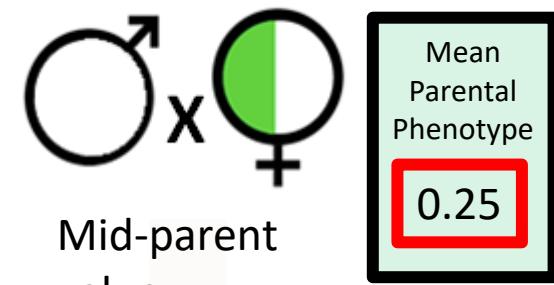


Family 1



Additive effects model

Quantitative genetic basis of phenotypic “greeness”...

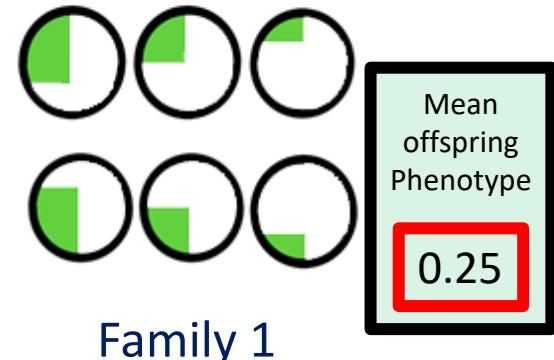


Sire	Dam
-+	-+
--	++
++	--
-+	++
+-	+-
-+	-+
--	+-
++	--
--	-+
++	-+

Sire Dam

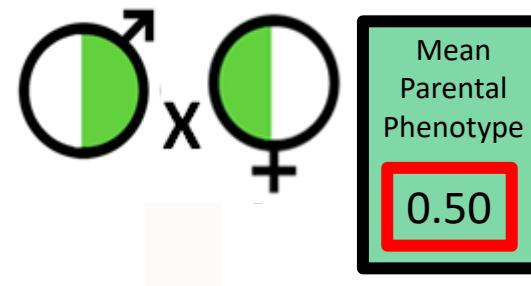
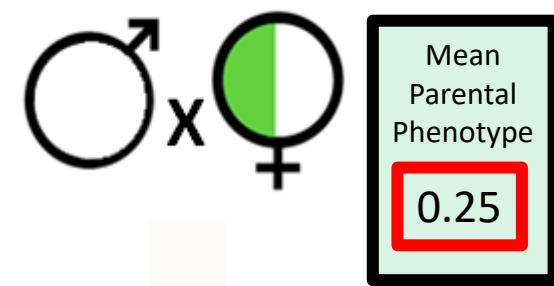
10+ 10+

10- 10-



Additive effects model

Quantitative genetic basis of phenotypic “greeness”...

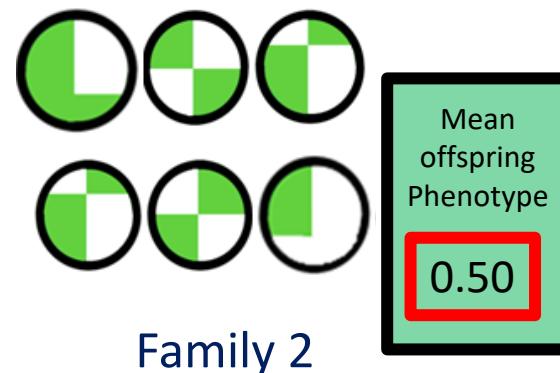
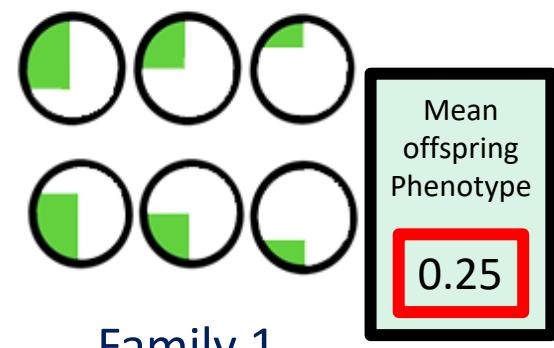


Sire	Dam
-+	-+
--	++
++	--
-+	++
+-	+-
-+	-+
--	+-
++	--
--	-+
++	-

Sire Dam

10+ 10+

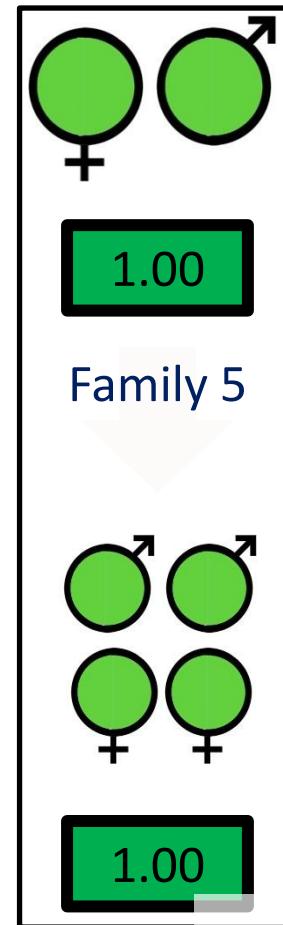
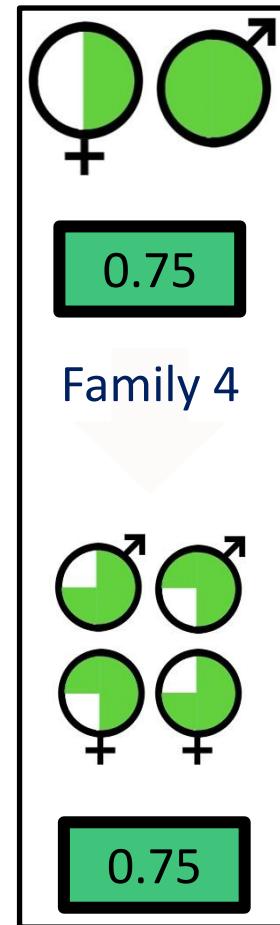
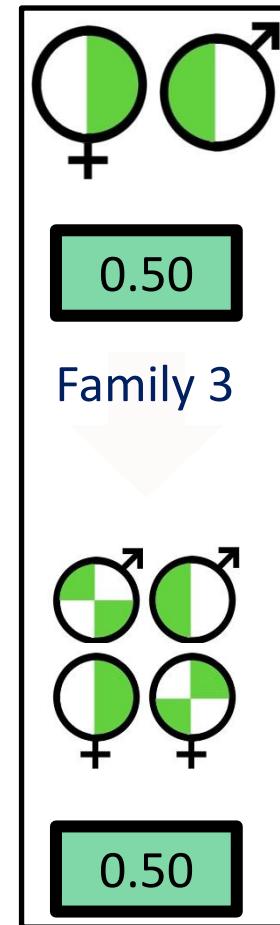
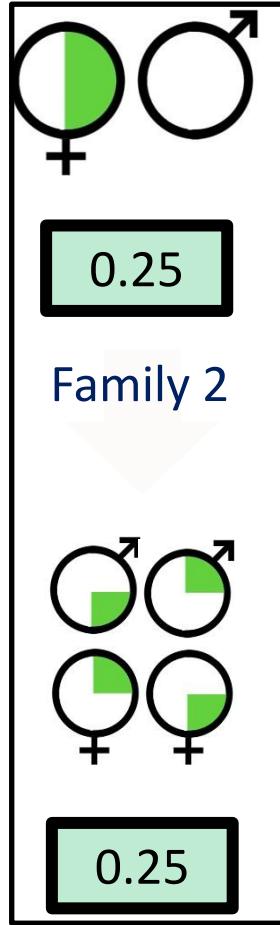
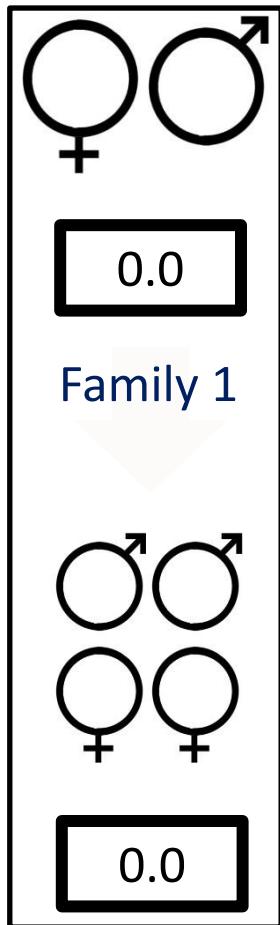
10- 10-



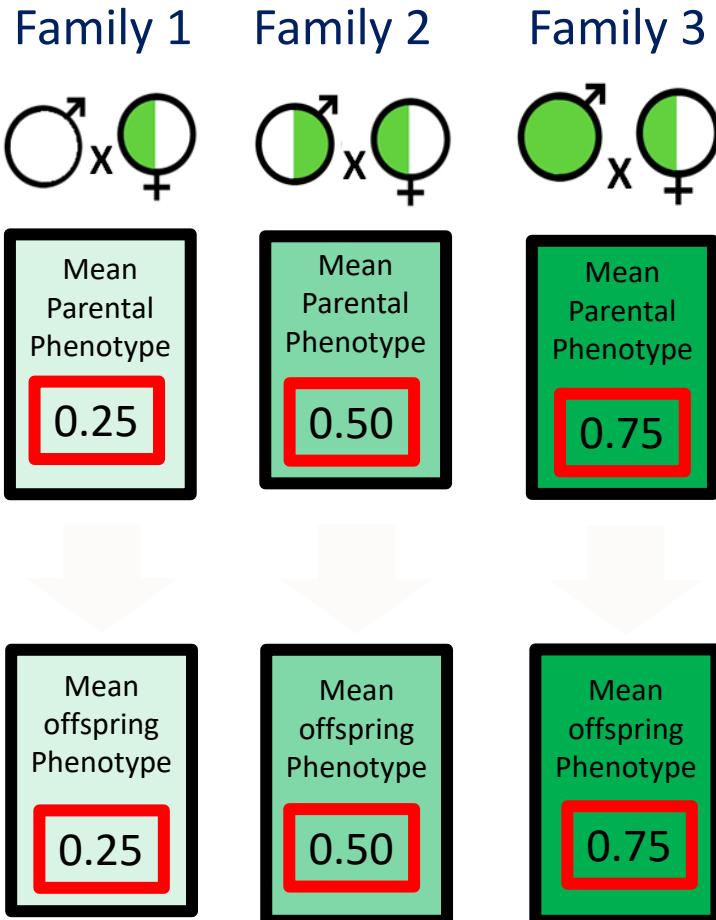
Offspring average
10 - 10 +

h^2 from a parent-offspring regression:

$$h^2 = \frac{V_A}{V_P}$$

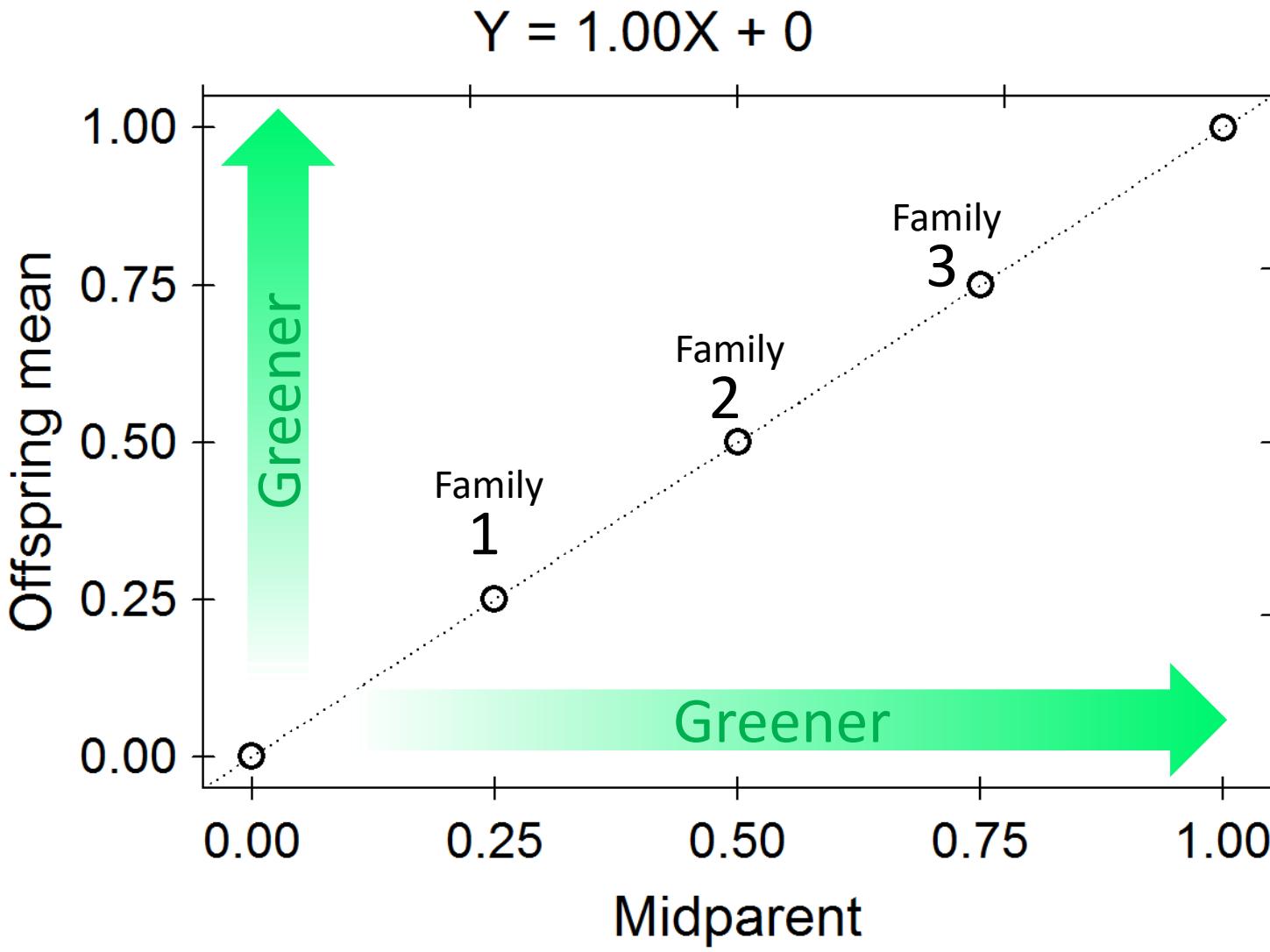


Regression: parents vs. offspring

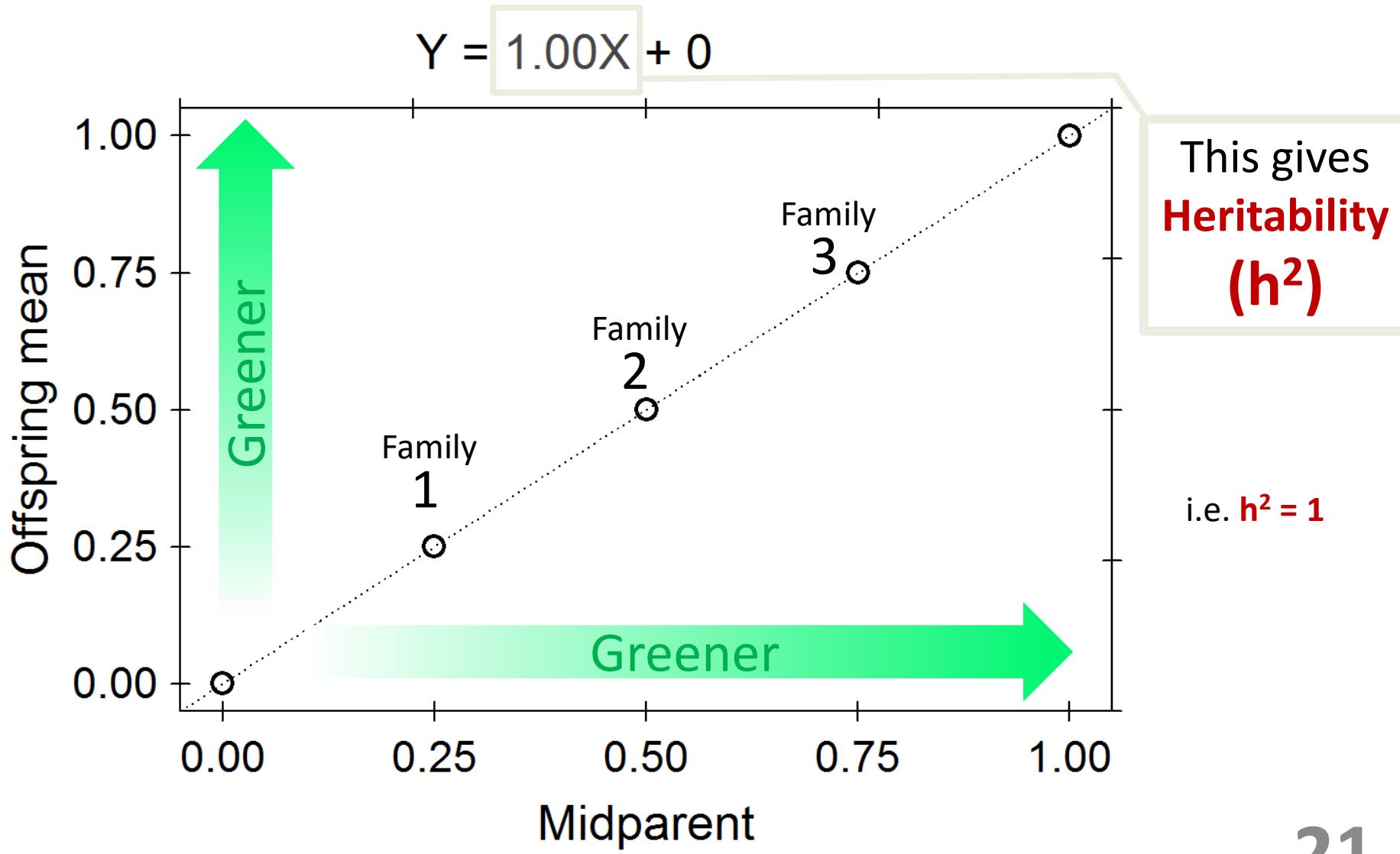


Family	Mid-parent (X-variable)	Offspring mean (Y)
1	0.25	0.25
2	0.50	0.50
3	0.75	0.75

Parent-offspring regression:



Parent-offspring regression:

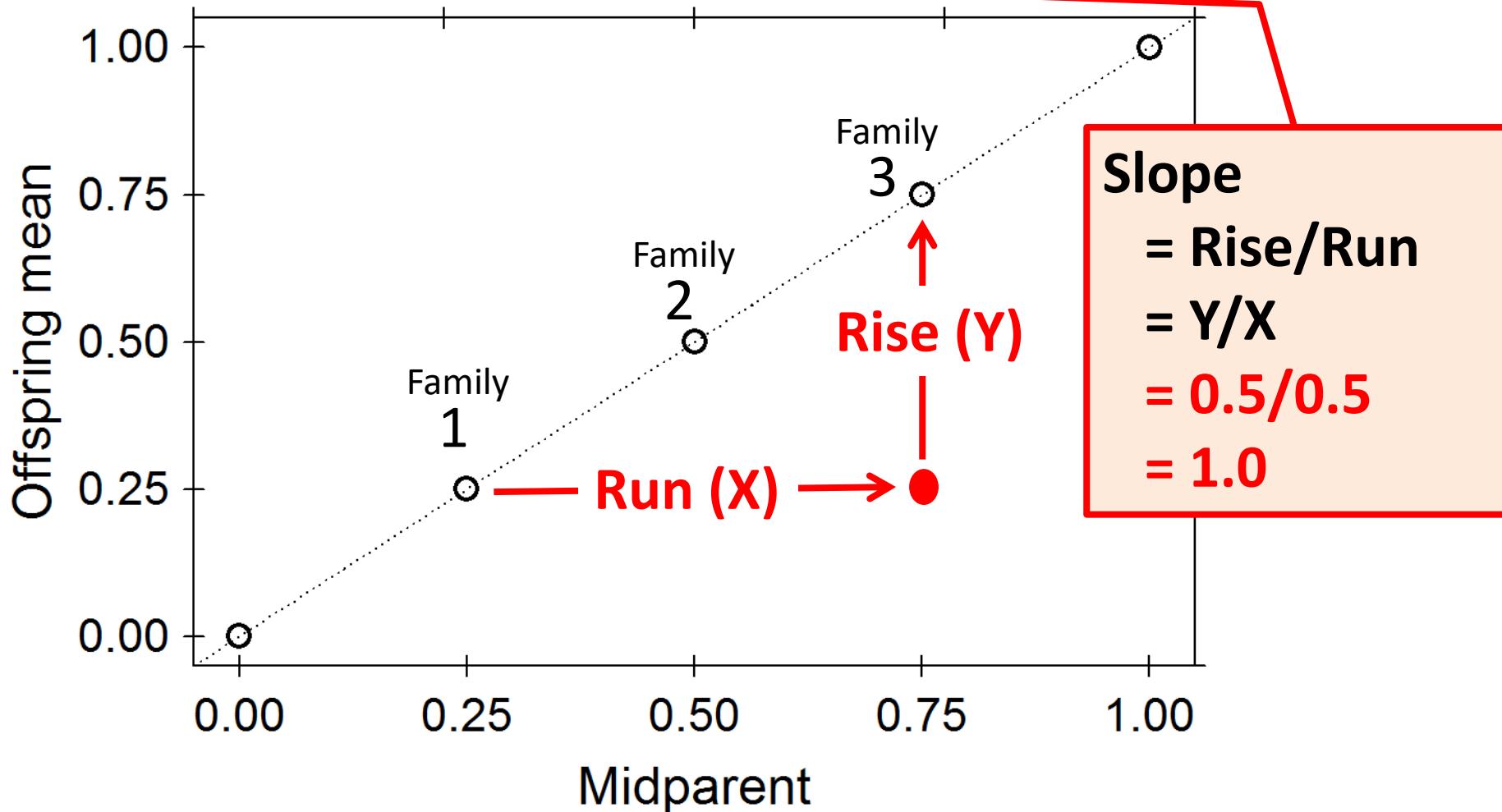


$$h^2 = \frac{V_A}{V_P}$$

Example 1:

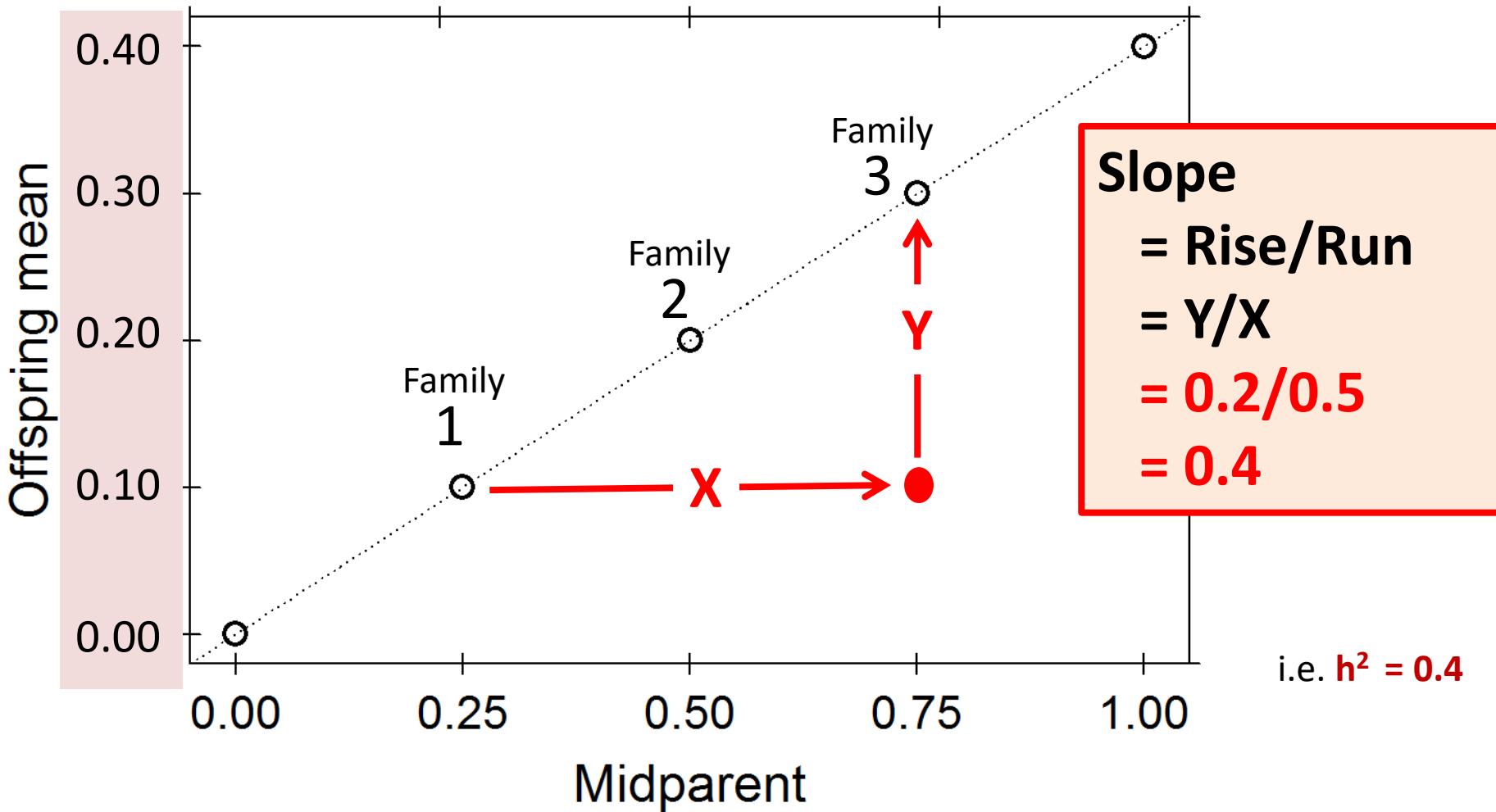
h^2 from a Parent-offspring regression

$$Y = 1.00X + 0$$



$$h^2 = \frac{V_A}{V_P}$$

Example 2: h^2 from a Parent-offspring regression



Heritability and Evolution

- The heritability of a trait determines its ‘ability’ to respond to selection, according to the breeder’s equation (a prediction of evolutionary change):

$$R = h^2 \times S \quad h^2=0, \text{ then } R=0$$

- R = response to selection
- S = selection differential (difference between selected mean and population mean)
- h^2 = heritability

Needs to be heritable for selection to act

Breeders Equation – Example 1

Example:

- F0 Bunnies can jump, on average, to **20cm** high
- We know that jumping in bunnies is heritable (has some genetic basis), $h^2=0.3$
- We want to breed the best (highest jumping) bunnies for the next generation (F1) in order to win the kanninhopning championships
- What do we do?
- Select the highest jumping bunnies!



Breeders Equation – Example 1

Step 1: Select the highest jumping bunnies!
Breed those with mean jumping height of
30cm

Step 2: Estimate selection differential:

$$(S) = 30 - 20 = 10$$

Step 3: Using the breeders equation, and the heritability estimate for jumping ($h^2 = 0.3$), calculate the response to selection

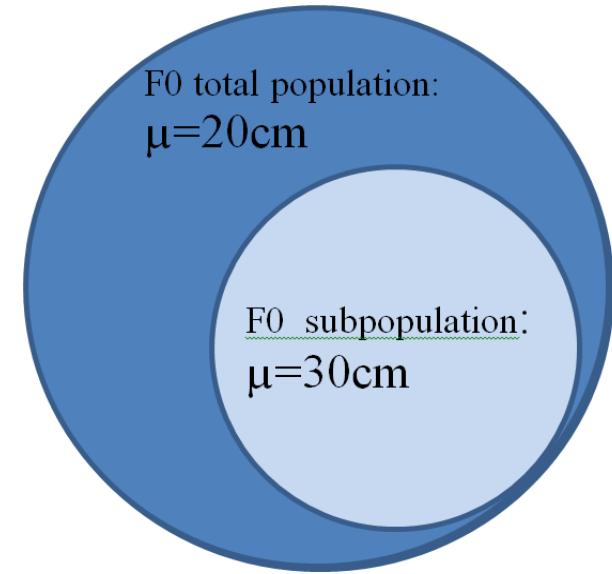
$$R = h^2 S$$

$$R = 0.3 \times 10 = 3$$

Step 4: estimate the next generation by adding R to the original (F0) mean (20cm)

$$F0 + R = 20 + 3 = 23$$

Expected F1 mean = **23**. We have increased the mean of the trait by 3 units.



Breeders Equation – Example 2

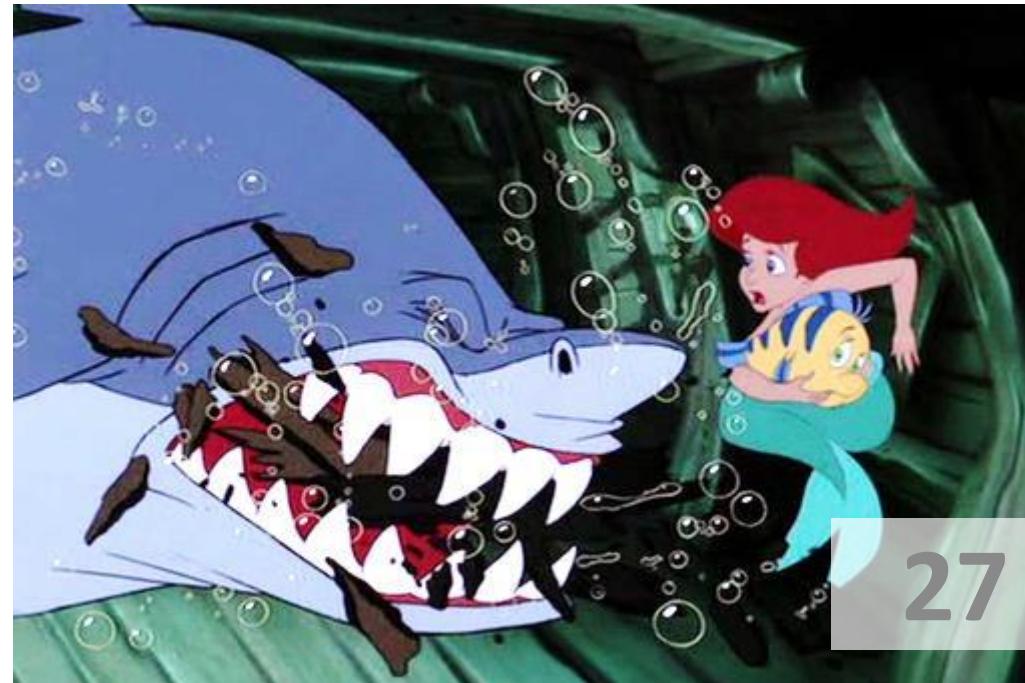
F₀ mermaids can swim, on average, 10km before exhausting

We know that swimming distance is heritable, $h^2=0.65$

In a high predation environment, only mermaids that can swim for 11km or more survive being eaten by sharks.

Of the remaining mermaids,
their average swimming
distance was 12.5km

What is the expected mean
swimming distance for the
offspring (F₁) of these
surviving mermaid's?



Breeders Equation – Example 2

F0 mermaids $\mu=10\text{km}$

$h^2=0.65$

Breeding F0 $\mu=12.5\text{km}$

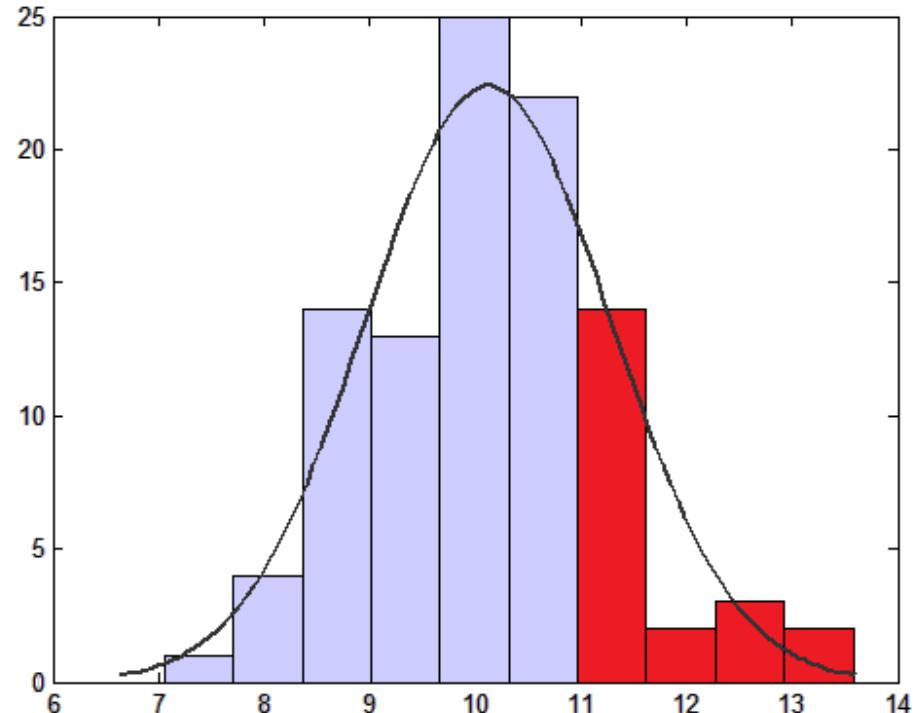
Selection Differential:

$$(S) = 12.5 - 10 = 2.5$$

Response to Selection:

$$R = h^2 S$$

$$R = 0.65 \times 2.5 = 1.625$$



Estimate the next generation (F1) mean (μ) by adding R to the original (F0) μ (10km)

$$F0 + R = 10 + 1.625 = 11.625$$

Expected F1 mean=11.625km. We have increased the mean of the trait by 1.625km.

Quantitative genetics, heritability, natural selection & adaptive potential

Genetic basis



Heritability (h^2)



Potential to respond to selection

Described by:

Breeder's equation $R = h^2 S$

h^2 , adaptive potential & climate change....

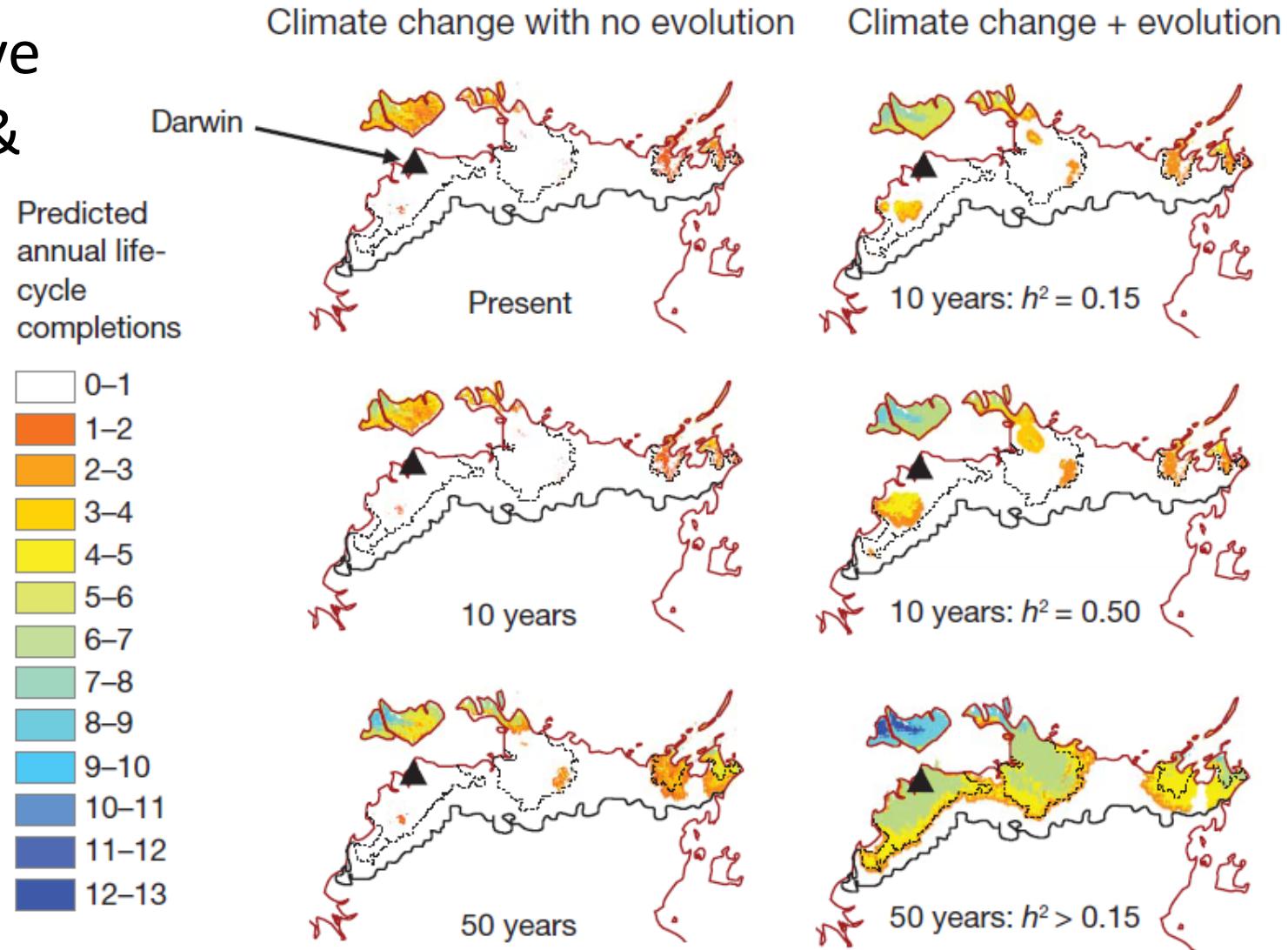


Figure 1 | Potential effect of evolution in egg desiccation resistance on life-cycle completions of the mosquito *Aedes aegypti* in the region around Darwin, Australia. Predictions are based on a mechanistic model of mosquito

Some positive news in the face of Climate Change?

Models use CURRENT behaviour and physiological limits

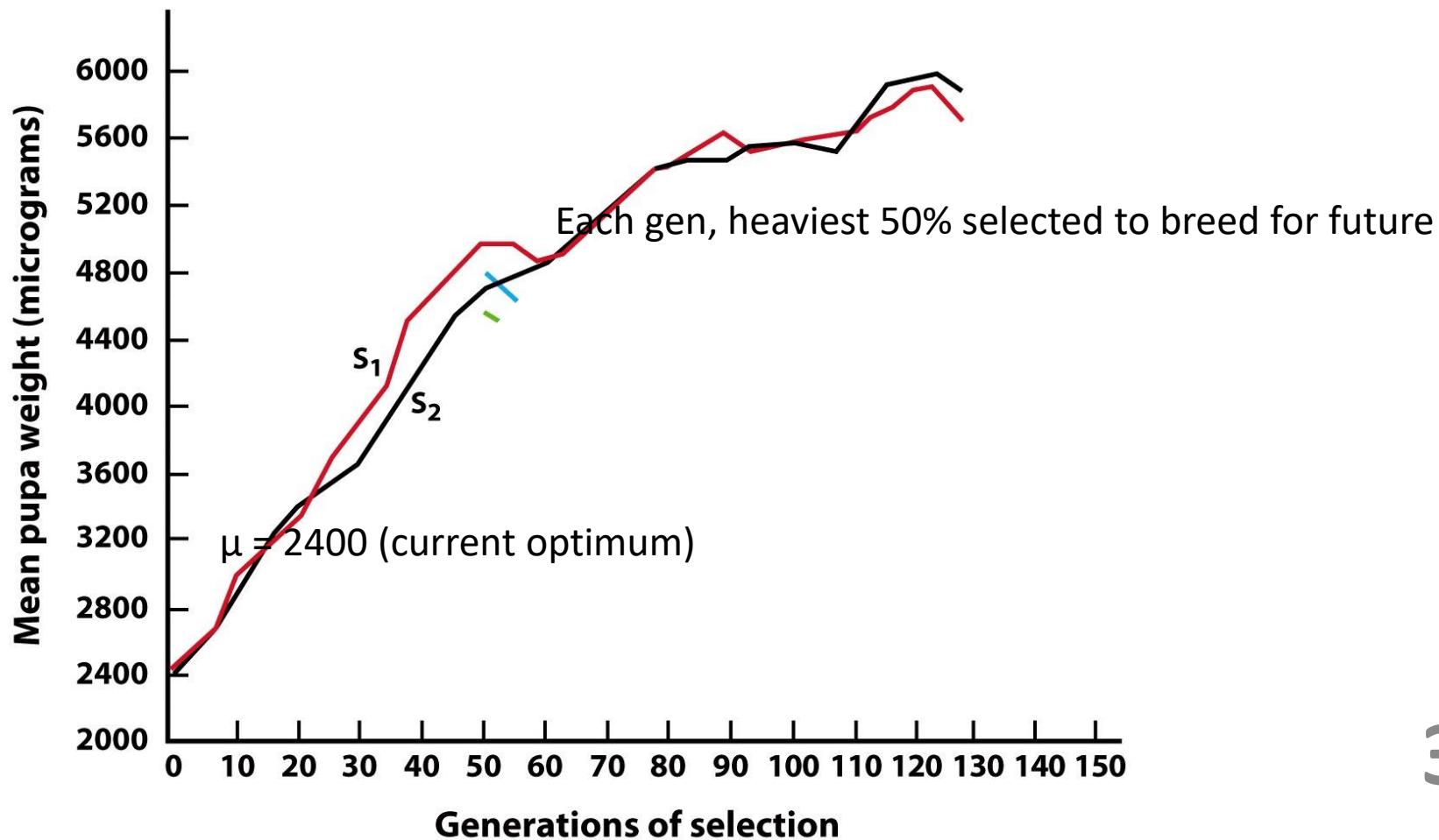
Adaptive potential -> fitter individuals are selected and represented in the next generation

Dependent on: degree of heritability, speed of environmental change, amount of existing genetic variation, generation time



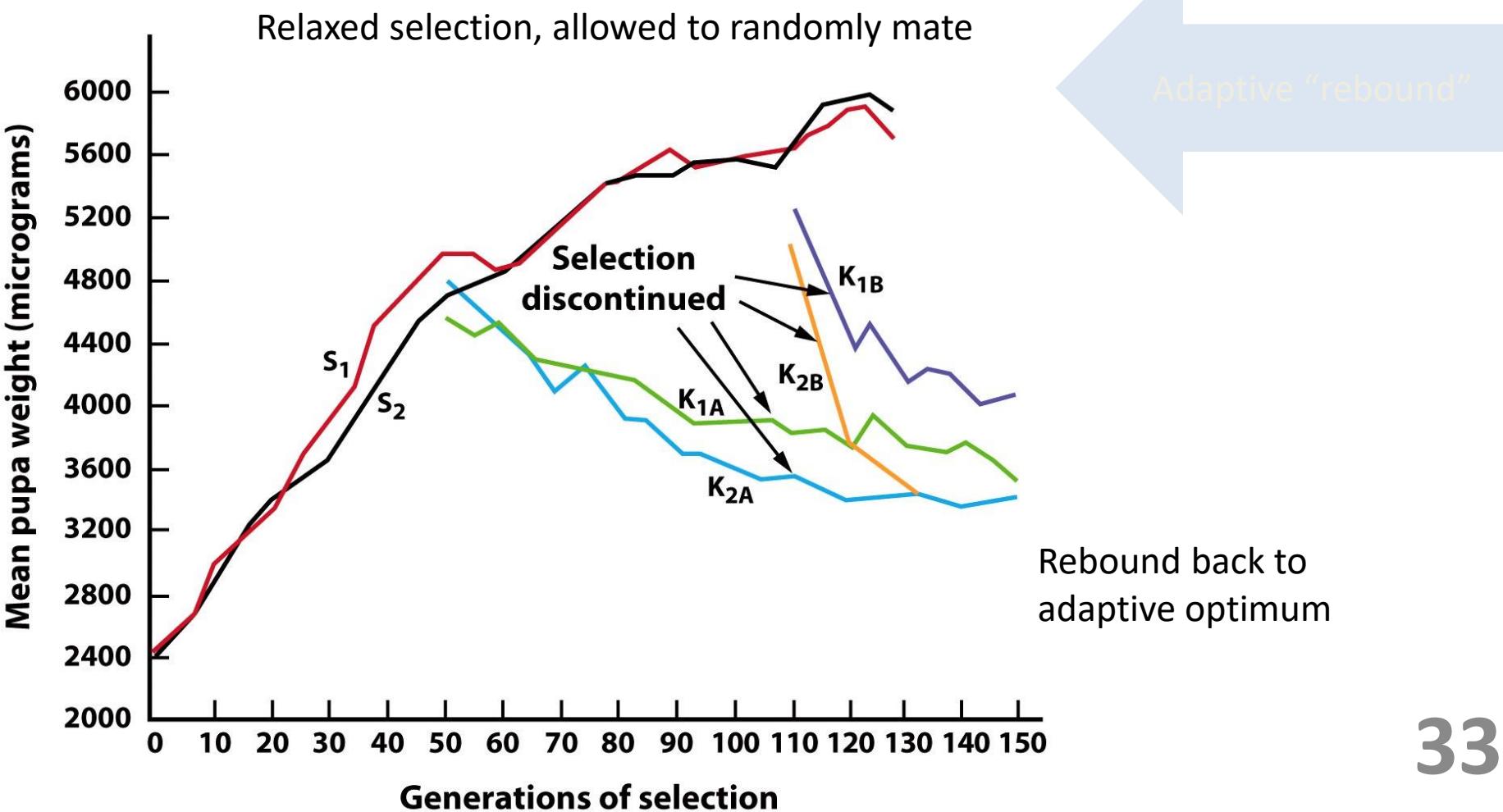


Artificial selection on pupal weight in flour beetles

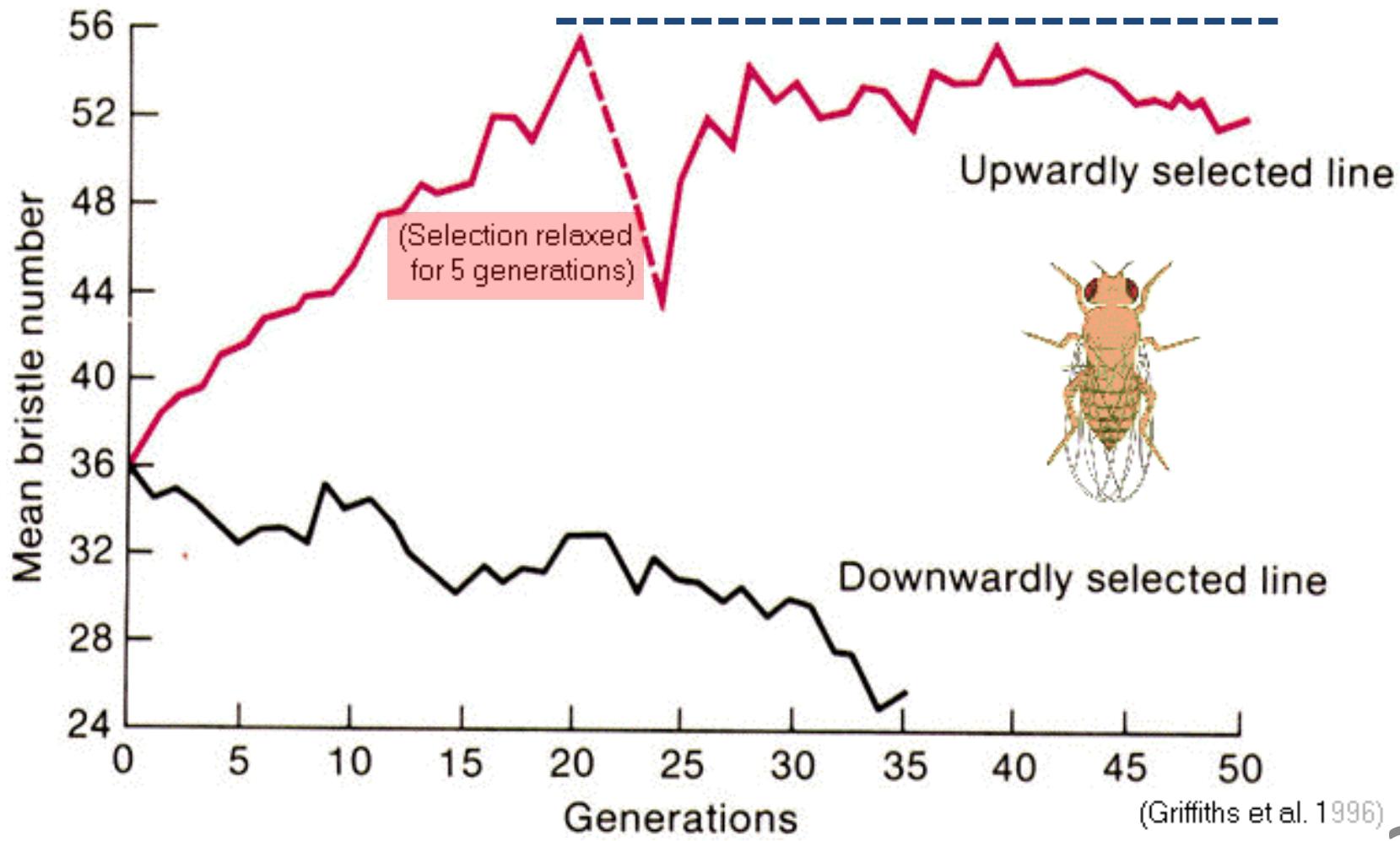




Artificial selection on pupal weight in flour beetles



Selection eventually hits a limit (asymptote)...



Need to Know Formulae

Broad Sense

$$VP = VG + VE$$

$$H^2 = VG/VP$$

VP = phenotypic variance

VG = genotypic variance

VE = environmental variance

VA = additive genetic variance

VI = epistatic genetic variance

VD = dominance genetic variance

Narrow Sense

$$VP = VG + VE$$

$$VG = VA + VD + VI$$

$$h^2 = VA/VP$$

H^2 = broad heritability

h^2 = narrow heritability

Adaptive Potential

$$R = h^2 S$$

S = response to selection

Summary

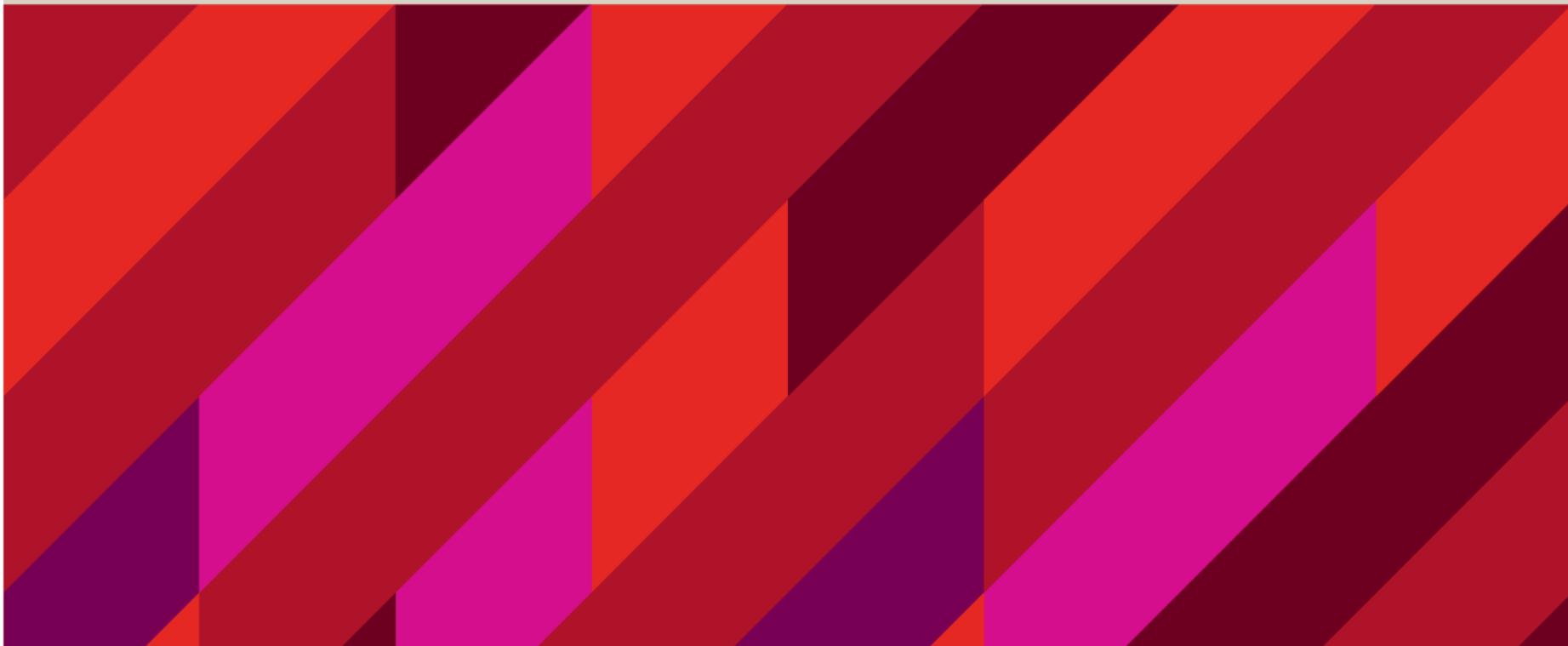
- $VG = VD + VI + VA$
- Narrow (h^2) and Broad (H^2) Heritability
- Narrow: (mid)parent offspring regression
- Broad: sibling regression
- Traits must be heritable to be selected upon (breeders equation – h^2)
- Heritability can tell us about the genetic ‘robustness’ of an organism in changing environments



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BIOL3110 Conservation & Ecological Genetics

LECTURE 7: GENETIC VARIATION & EVOLUTION



V_G and Evolution

IMPORTANCE OF QUANTITATIVE GENETICS



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1. Quick review of key Quant Gen concepts

2. Importance for conservation

3. Case studies of evolution

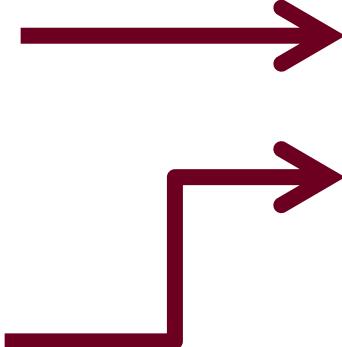


V_G and Evolution

QUANTITATIVE GENETICS

From studying phenotypes, we can partition:

- **Additive** genetic variation
- Non-additive genetic variation
- Environmental variation
 - (+ interactions/covariance – later)
- Total **Phenotypic** variation



$$\frac{V_A}{V_P} = h^2$$

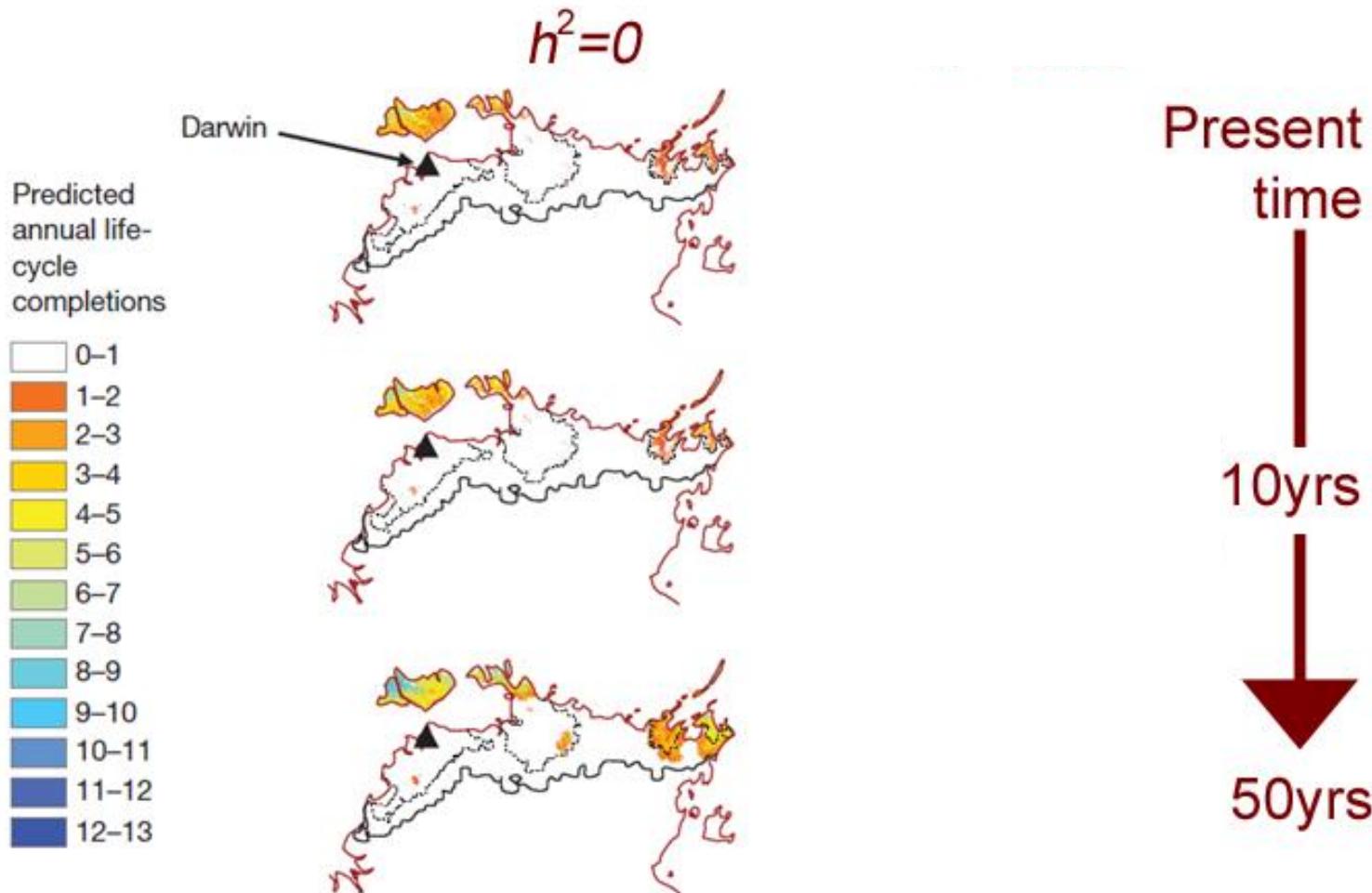
Narrow-sense heritability

- Then, predict **Response to Selection**:
(via the “breeder’s equation”)

$$R = h^2 S$$

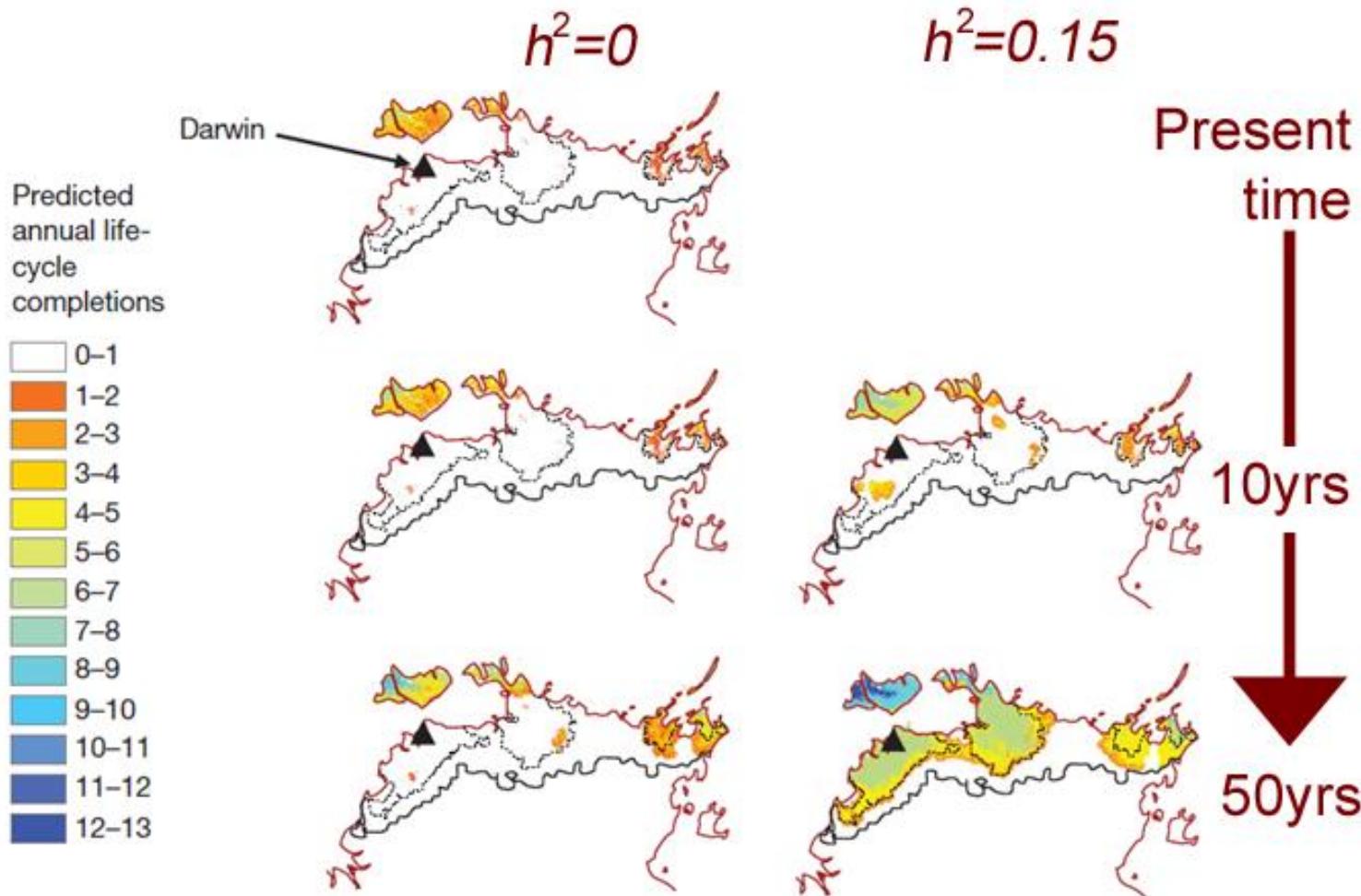
V_G and Evolution

IMPORTANCE OF QUANTITATIVE GENETICS



V_G and Evolution

IMPORTANCE OF QUANTITATIVE GENETICS



V_G and Evolution

Bufo marinus INVASION



<http://www.cairnsunlimited.com>

Cane toad (*Bufo marinus*)



Cane beetle (*Dermolepida albohirtum*)

V_G and Evolution

Bufo marinus INVASION



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<http://www.frogwatch.org.au>

Biology

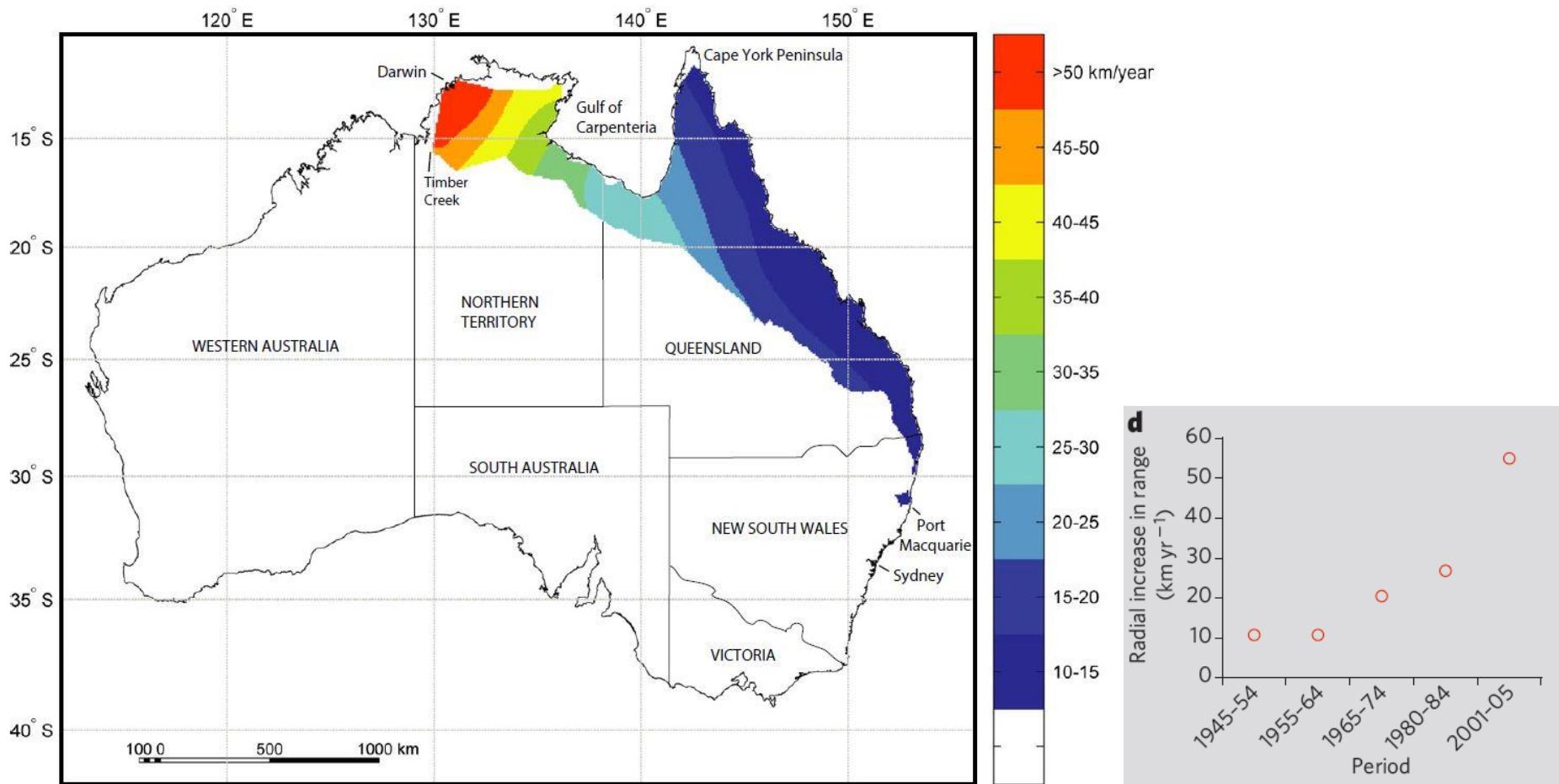
- Classic r-selected species
- >30,000 eggs per clutch
- Metamorphosis ~12-days
- Lifespan 10-15 years
- Adults chemically protected by bufotoxin (incl. bufotenin: a class 1 hallucinogen)

V_G and Evolution

Bufo marinus INVASION



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Rollins *et al.* 2015

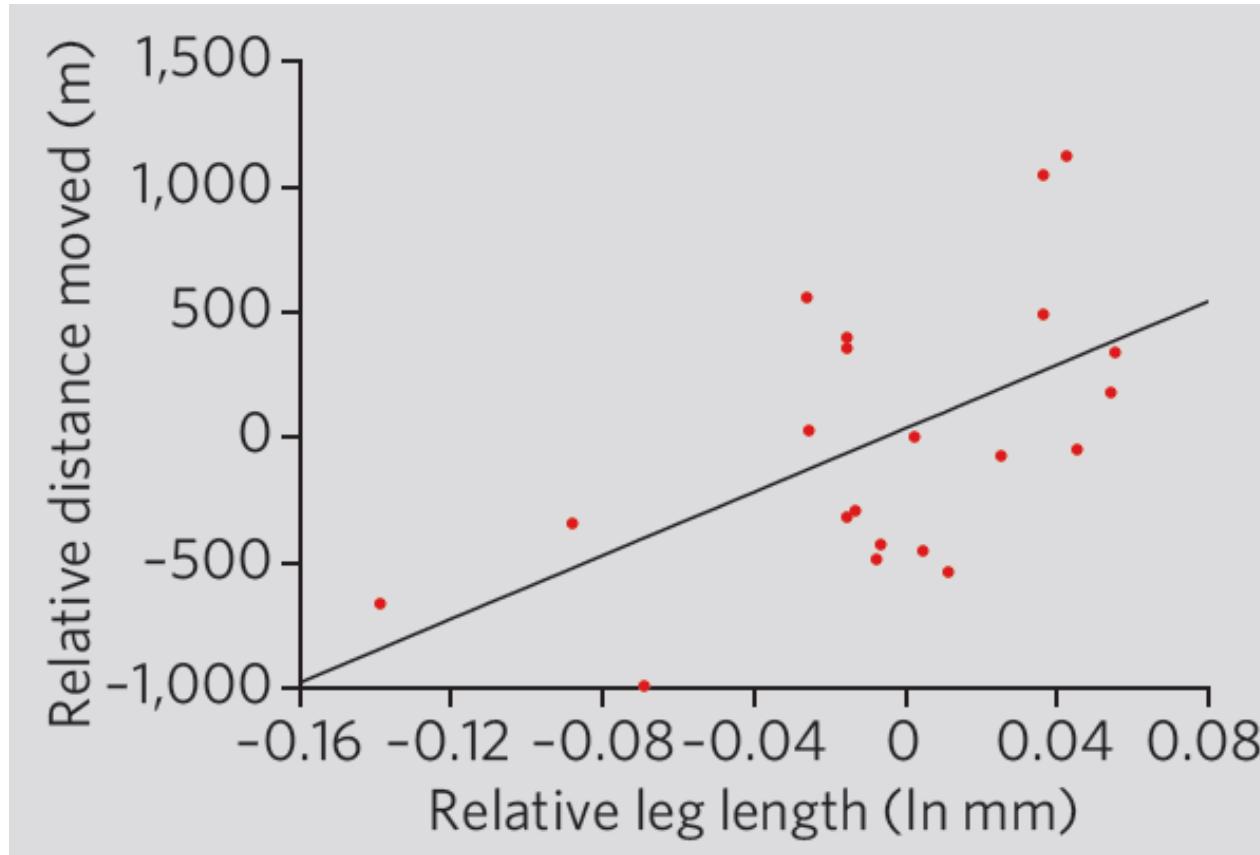
Phillips *et al.* 2006

V_G and Evolution

Bufo marinus INVASION



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Phillips *et al.* 2006

Longer-legged toads disperse more quickly

V_G and Evolution

Bufo marinus INVASION

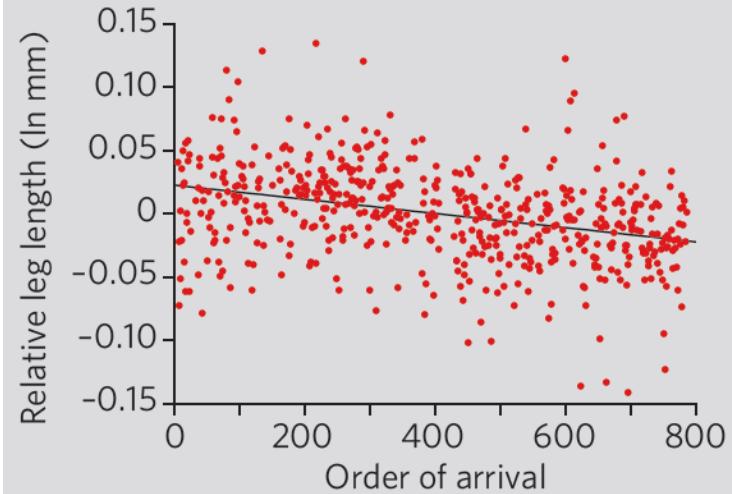


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At the invasion FRONT:

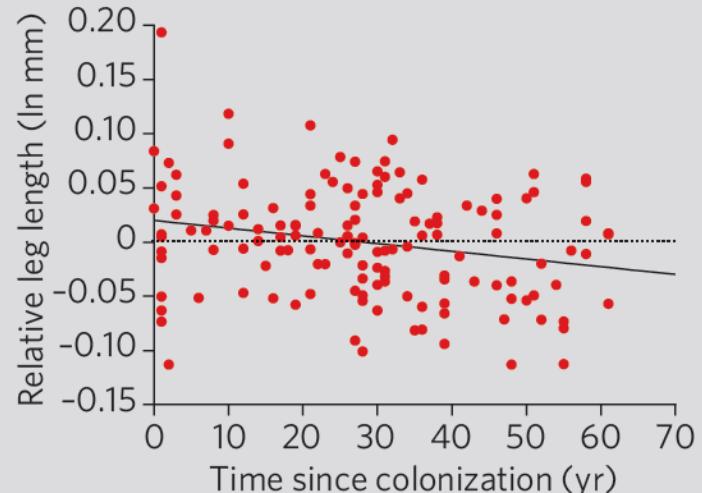
- Long-legged toads arrive first
- Higher fitness
- Dispersal genes favoured at the front



After long establishment:

- Leg length declines
- Fitness 'target' shifts
- Dispersal genes disfavoured back from the front

Phillips *et al.* 2006



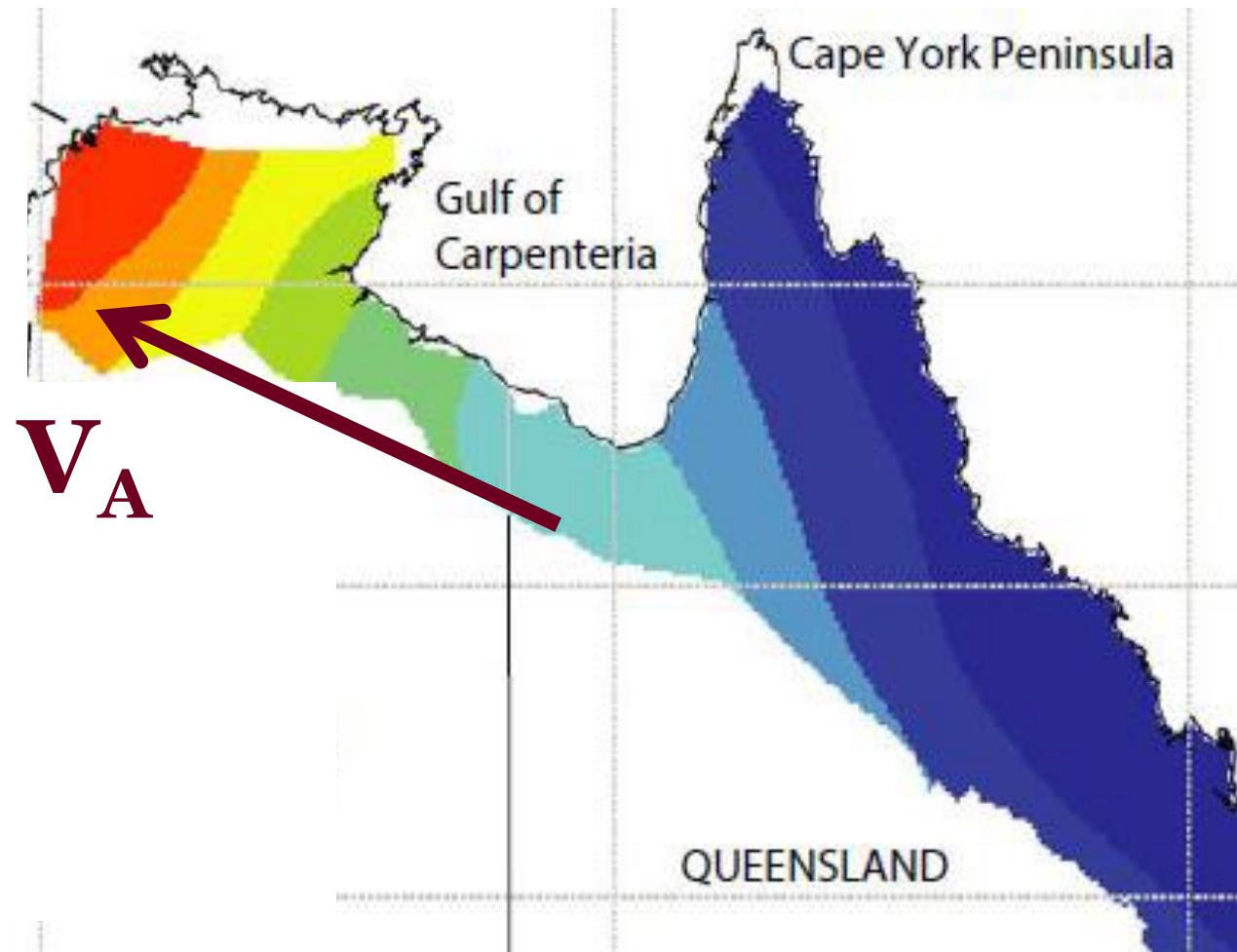
V_G and Evolution

Bufo marinus INVASION



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“colocation”
of dispersal-
related
genes



V_G and Evolution

Bufo marinus INVASION



Red-bellied black snake (*Pseudechis porphyriacus*)



Freshwater crocodile
(*Crocodylus johnsoni*)



Red-cheeked dunnart
(*Sminthopsis virginiae*)



Corvis spp.)

V_G and Evolution

Bufo marinus INVASION



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Under strong directional selection:

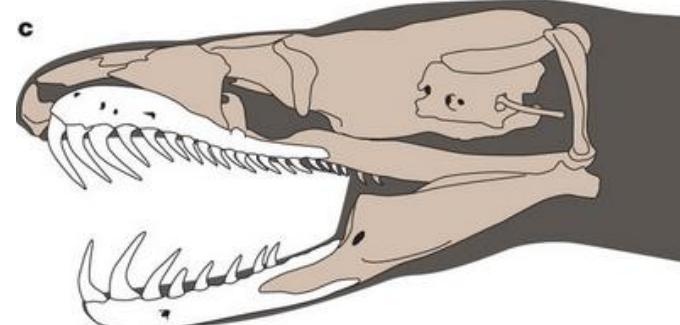
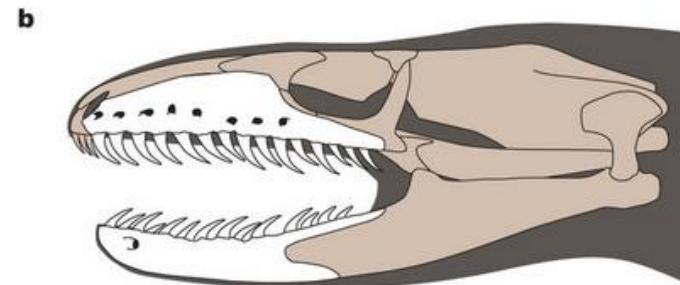
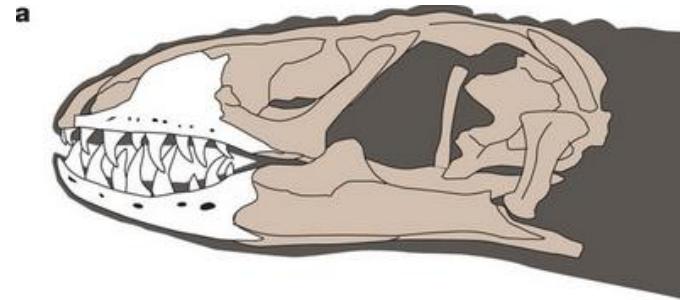
- Snakes avoid toads as prey
(a **non-learned** behaviour)
- Morphological evolution.....?



Pseudechis porphyriacus



Dendrelaphis punctulatus



V_G and Evolution

Bufo marinus INVASION

Under strong directional selection:

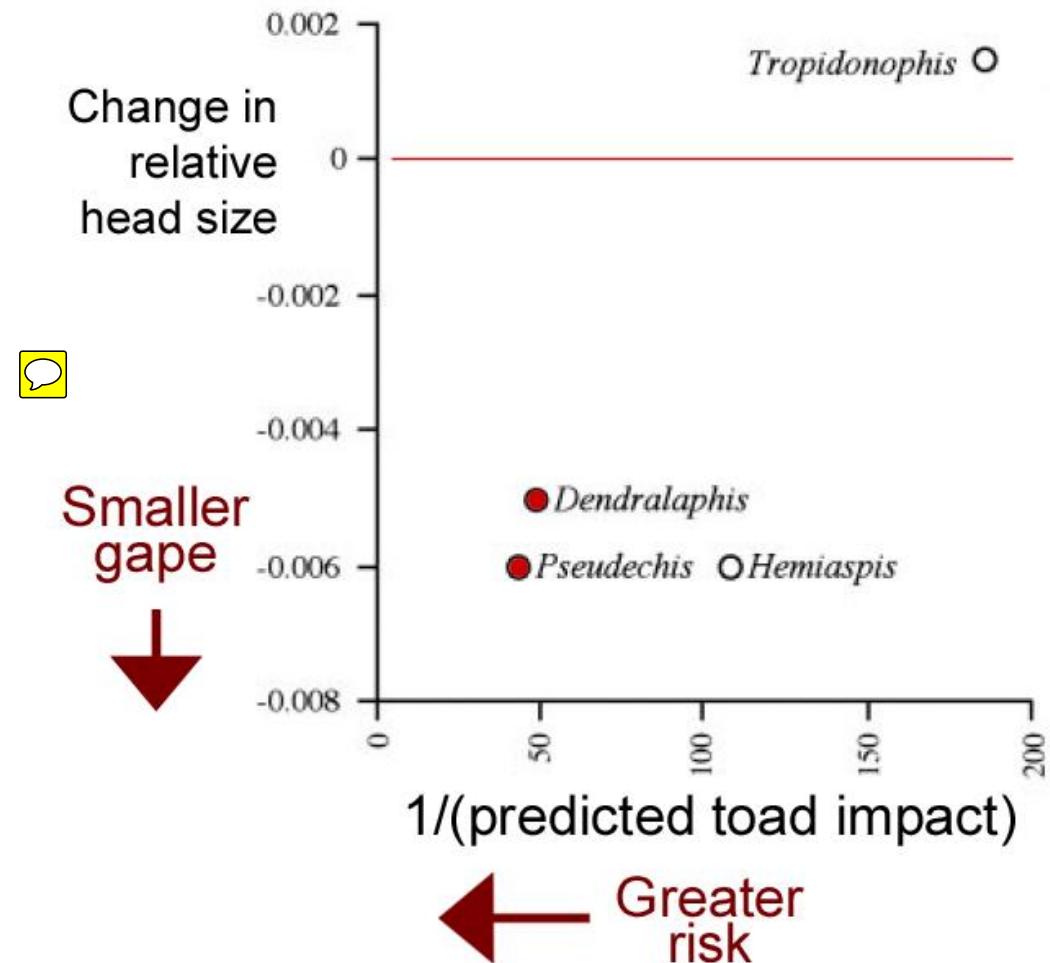
- Snakes avoid toads as prey (a **non-learned** behaviour)
- Snake morphology has evolved in **~23 generations**



Pseudechis porphyriacus



Dendrelaphis punctulatus



V_G and Evolution

Bufo marinus INVASION



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MOLECULAR ECOLOGY

Molecular Ecology (2015) 24, 2264–2276

doi: 10.1111/mec.13184

INVASION GENETICS: THE BAKER AND STEBBINS LEGACY

A genetic perspective on rapid evolution in cane toads (*Rhinella marina*)

LEE A. ROLLINS,* MARK F. RICHARDSON* and RICHARD SHINE†

*Centre for Integrative Ecology, School of Life & Environmental Sciences, Deakin University, P.O. Box 22023, Geelong, Victoria 3216, Australia, †School of Biological Sciences A08, University of Sydney, Sydney, NSW 2006, Australia

Vol 439|16 February 2006

nature

BRIEF COMMUNICATIONS

Invasion and the evolution of speed in toads

Cane toads seem to have honed their dispersal ability to devastating effect over the generations.

Adapting to an invasive species: Toxic cane toads induce morphological change in Australian snakes

Ben L. Phillips* and Richard Shine

School of Biological Sciences A08, University of Sydney, New South Wales 2006, Australia

Edited by David B. Wake, University of California, Berkeley, CA, and approved October 27, 2004 (received for review August 31, 2004)

The arrival of invasive species can devastate natural ecosystems, but the long-term effects of invasion are less clear. If native organisms can adapt to the presence of the invader, the severity of impact will decline with time. In Australia, invasive cane toads

much less research has been conducted on counteradaptation by native species (17, 18).

Many species of Australian snake have been severely impacted by the invasion of highly toxic cane toads (*Bufo marinus*).

PNAS

Evolving resistance and prey discrimination



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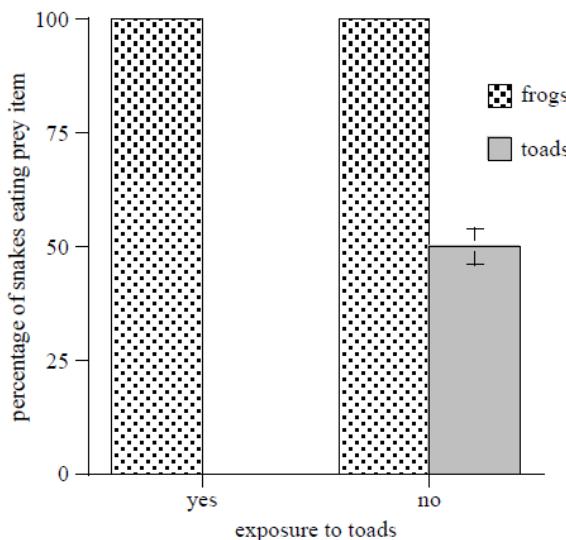


Figure 1. The percentage of black snakes from toad-exposed and toad-naive populations willing to eat a toad or a frog. No snake from a toad-exposed locality would consume a toad. Error bars represent a standard error.

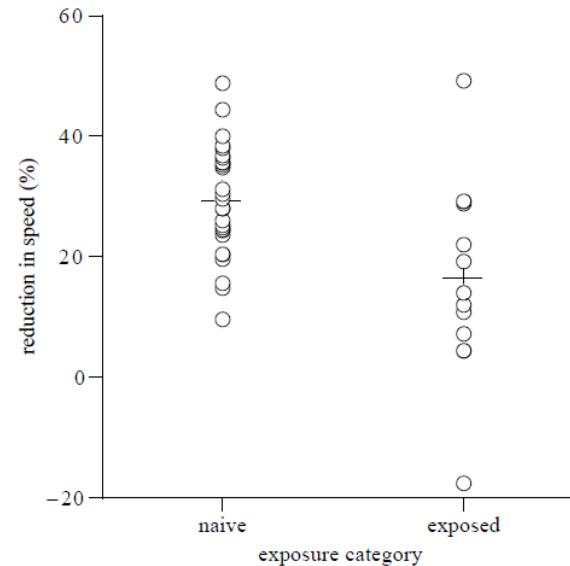


Figure 2. Resistance to toad toxin in toad-exposed and toad-naive populations. A large percentage reduction in speed indicates low resistance to toxin. Hence, snakes from toad-exposed populations exhibited higher resistance to toad toxin.

The standard equation used to estimate adaptive potential

The Breeder's equation, $R = h^2 S$

- applies only to conditions under which heritability was measured
- does not assess several major components of evolutionary genetic variation: dominance, epistasis, plasticity and epigenetics

Can we use genomics to estimate adaptive potential?



Using genomics to characterize evolutionary potential for conservation of wild populations

Katherine A. Harrisson, Alexandra Pavlova, Marina Telonis-Scott and Paul Sunnucks

Evolutionary Applications

doi:10.1111/eva.12149

Given our current understanding of genomes - screening genome wide diversity will usually give a better estimate of evolutionary potential rather than trying to get estimates from particular components of the genome, eg focussing on genes of known function

Two distinct components of evolutionary potential

- **Genetic** (DNA-sequence-based)
- **Epigenetic** (non-DNA-sequence-based)

Molecular basis of evolutionary potential

Epigenetic

*Histone modification,
DNA methylation,
small non-coding RNAs
(microRNA, small
interfering RNA)*

Genetic (sequence-based)

*Distal/long-range transcription factor
binding sites (enhancers), splice
junctions, splicing enhancers,
synonymous mutations, silencers,
tandem repeats, non-coding variation of
unknown function*

*Non-synonymous
coding changes, known
transcription factor
binding sites
(enhancers/promoters)*

Unknown

small

LARGE

effect size on phenotype

effect size on phenotype



Integrative and Comparative Biology

Integrative and Comparative Biology, volume 53, number 2, pp. 351–358
doi:10.1093/icb/ict007

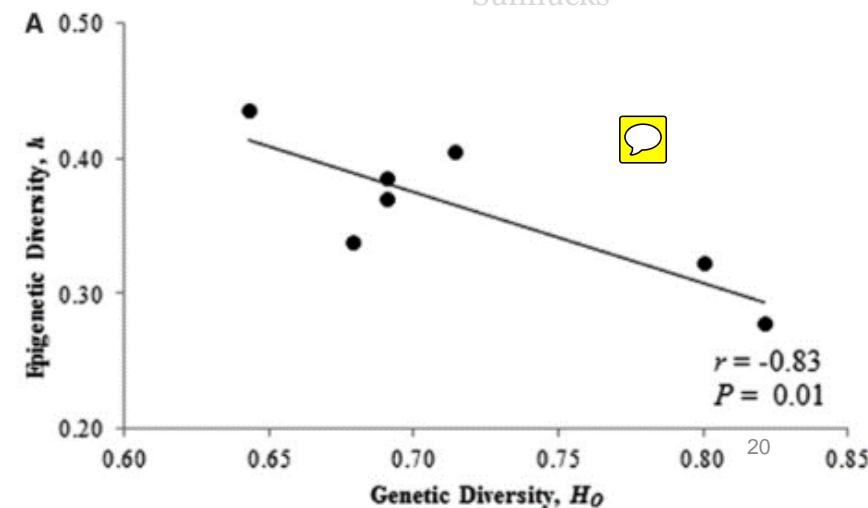
Society for Integrative and Comparative Biology

Patterns of DNA Methylation Throughout a Range Expansion of an Introduced Songbird

Andrea L. Liebl,^{1,*} Aaron W. Schrey,^{*†} Christina L. Richards^{*} and Lynn B. Martin^{*}

- 1950s, house sparrow introduced into Kenya
- High levels of variation in DNA methylation (epigenetic marker)
- Epigenetics could be important part of phenotypic variability and adaptation

Epigenetic variation was greatest in the populations with the lowest genetic variation – may compensate for low genetic variation



Methods to infer selection are complex, numerous, emerging

Epigenetic

Genetic (sequence-based)

Biochemical

Function hard to infer

Known function

MS-AFLP 

Expression profiling

Genomic selection analysis

Animal model

Quantitative trait nucleotides/
Loci analysis

Genome-wide selection scans

Gene-environment associations

Genome-wide association studies

Environmental correlation methods

Three main insights into evolutionary potential
summarized by Harrisson et al. 2014:

- (1) Rapid adaptive evolution is mainly from changes in gene expression
- (2) Most traits are polygenic (built by lots of genes of small effect)
- (3) Most recent adaptation is due to subtle shifts of allele frequencies

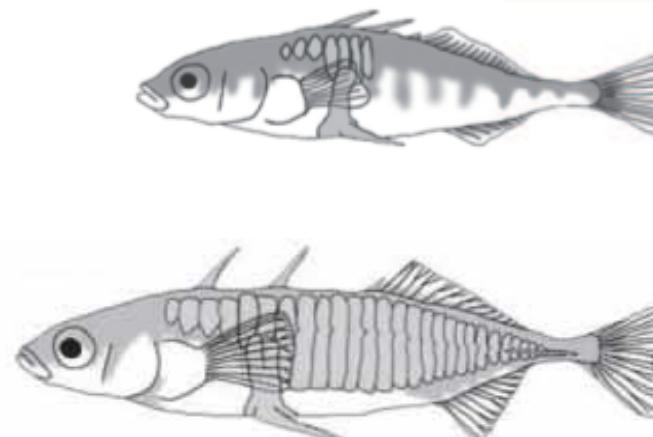
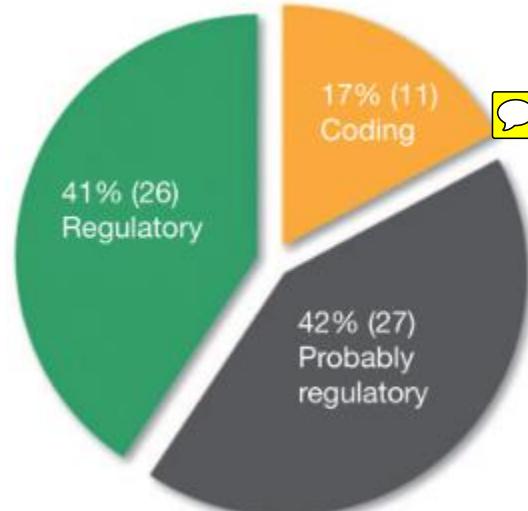
(1) Rapid adaptive evolution is driven predominantly by changes in gene expression

The genomic basis of adaptive evolution in threespine sticklebacks

5 APRIL 2012 | VOL 484 | NATURE | 55

Felicity C. Jones^{1*}, Manfred G. Grabherr^{2,3*}, Yingguang Frank Chan^{1†*}, Pamela Russell^{2*}, Evan Mauceli^{2†}, Jeremy Johnson², Ross Swofford², Mono Pirun^{2†}, Michael C. Zody², Simon White⁴, Ewan Birney⁵, Stephen Searle⁴, Jeremy Schmutz⁶, Jane Grimwood⁶, Mark C. Dickson⁶, Richard M. Myers⁶, Craig T. Miller^{1†}, Brian R. Summers¹, Anne K. Knecht¹, Shannon D. Brady¹, Haili Zhang¹, Alex A. Pollen¹, Timothy Howes¹, Chris Amemiya⁷, Broad Institute Genome Sequencing Platform & Whole Genome Assembly Team[†], Eric S. Lander², Federica Di Palma², Kerstin Lindblad-Toh^{2,3} & David M. Kingsley^{1,8}

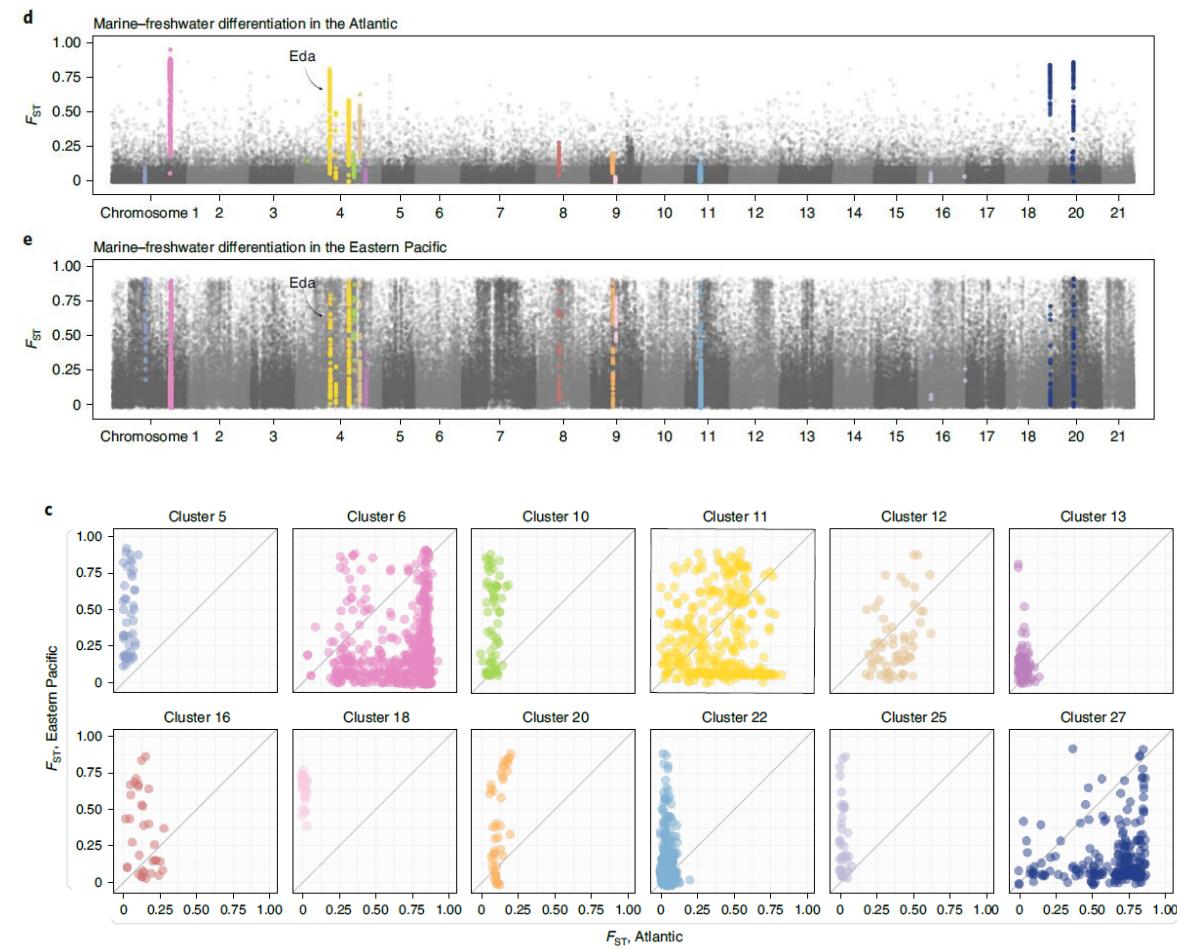
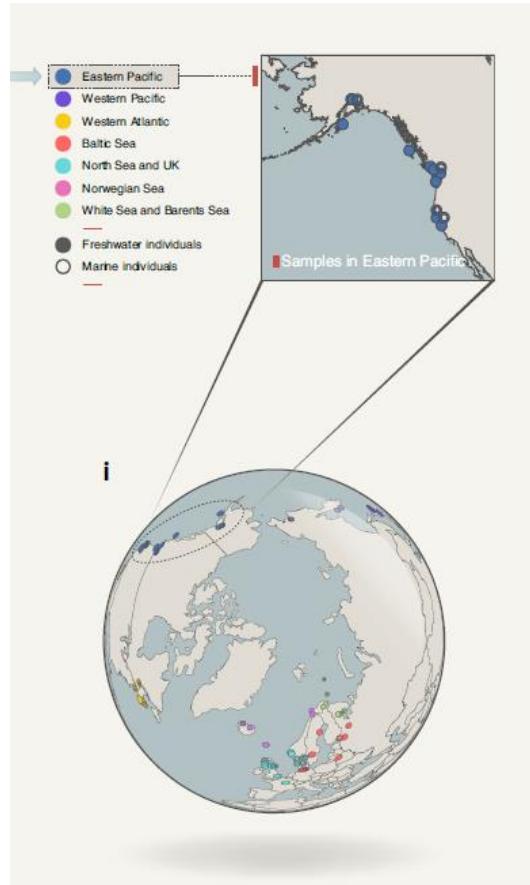
Found 64 strong candidate genes for evolutionary transitions between sea & freshwater
Mostly regulatory



Freshwater forms

Marine forms

Ectodysplasin A (EDA) gene is known to be responsible for marine–freshwater differences in lateral armour plate development worldwide



(2) Most traits are polygenic



Genomic dissection of variation in clutch size and egg-mass in a wild great tit (*Parus major*) population

ANNA W. SANTURE,*¹ ISABELLE DE CAUWER,*†¹ MATTHEW R. ROBINSON,*
JOCELYN POISSANT,* BEN C. SHELDON‡ and JON SLATE*

MOLECULAR ECOLOGY

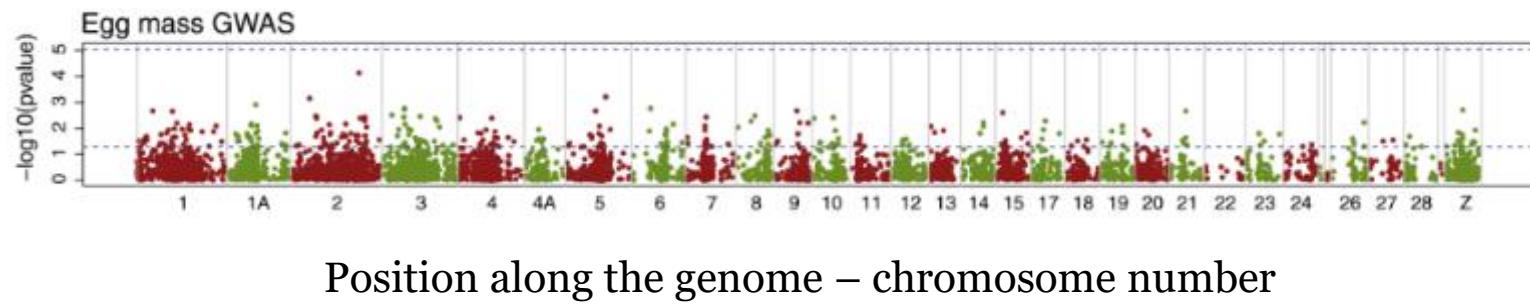
Molecular Ecology (2013) 22, 3949–3962



e.g. egg characteristics (number, mass) are predicted by variation at many genes – egg characteristics are polygenic

e.g. as done by genome-wide association studies (GWAS)

Probability
that each
marker is
associated
with egg mass



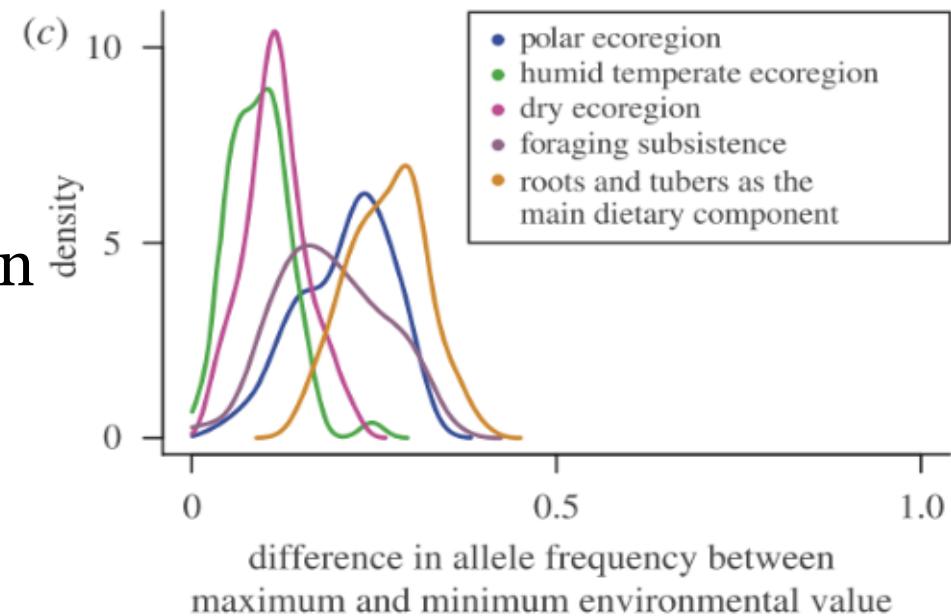
Can find specific candidate genes - the ones above the lower dotted blue line

(3) Most recent adaptation is due to subtle shifts of allele frequencies

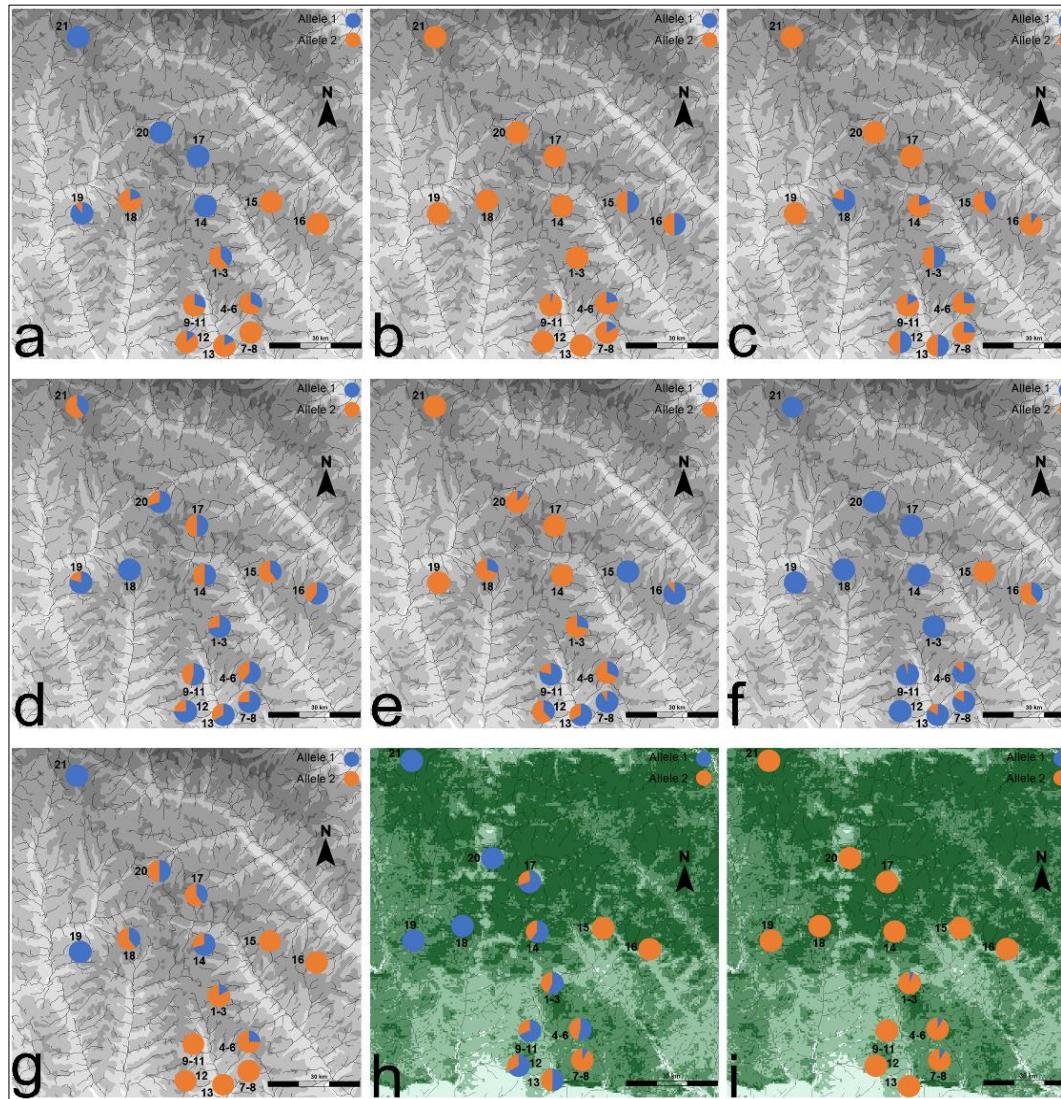
Subtle shifts in frequencies – need huge data sets, so many earlier studies are related to human health

E.g.

Adaptations to a diet rich in roots and tubers show association with changes in starch and sucrose metabolism and an ability to synthesize folic acid (tubers are low in this)



Hancock et al. (2010) Phil Trans Roy Soc B 365 2459-2468



Gray - altitude

functions for genes associated with forest biomass were annotated to blood, cell and chromatin functions, whereas genes with allele frequencies associated with altitude were annotated to the immune system, catabolic regulation, reproduction and functioning of sensorial organs (i.e., vision)

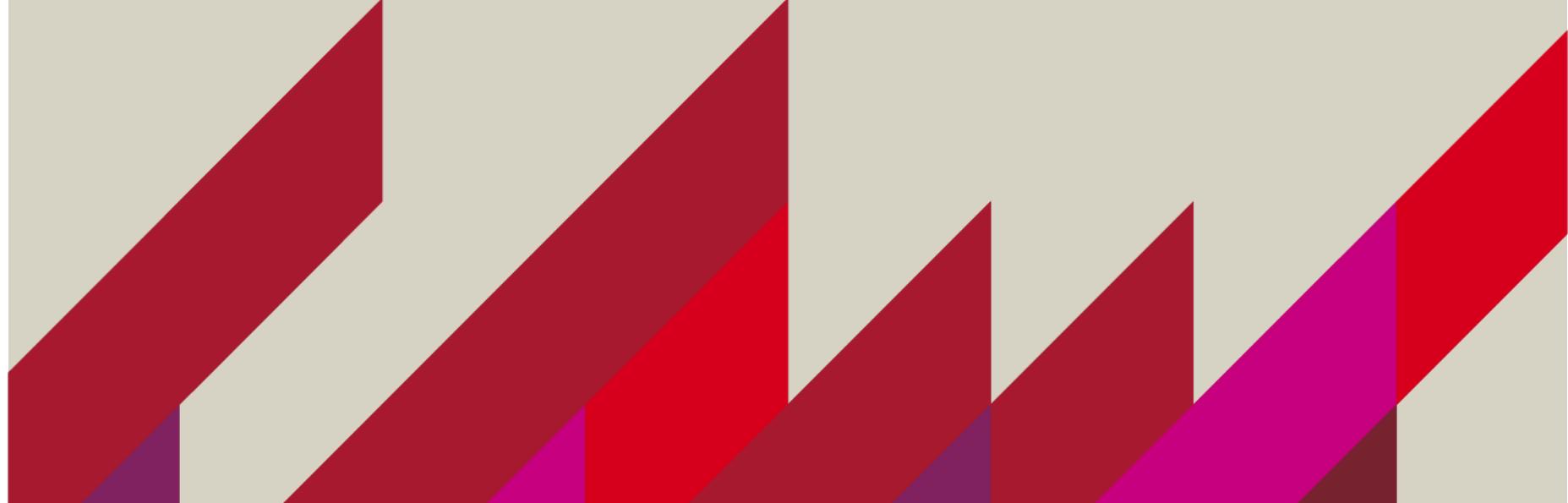
Green – forest density

Next:



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Evolution part 2

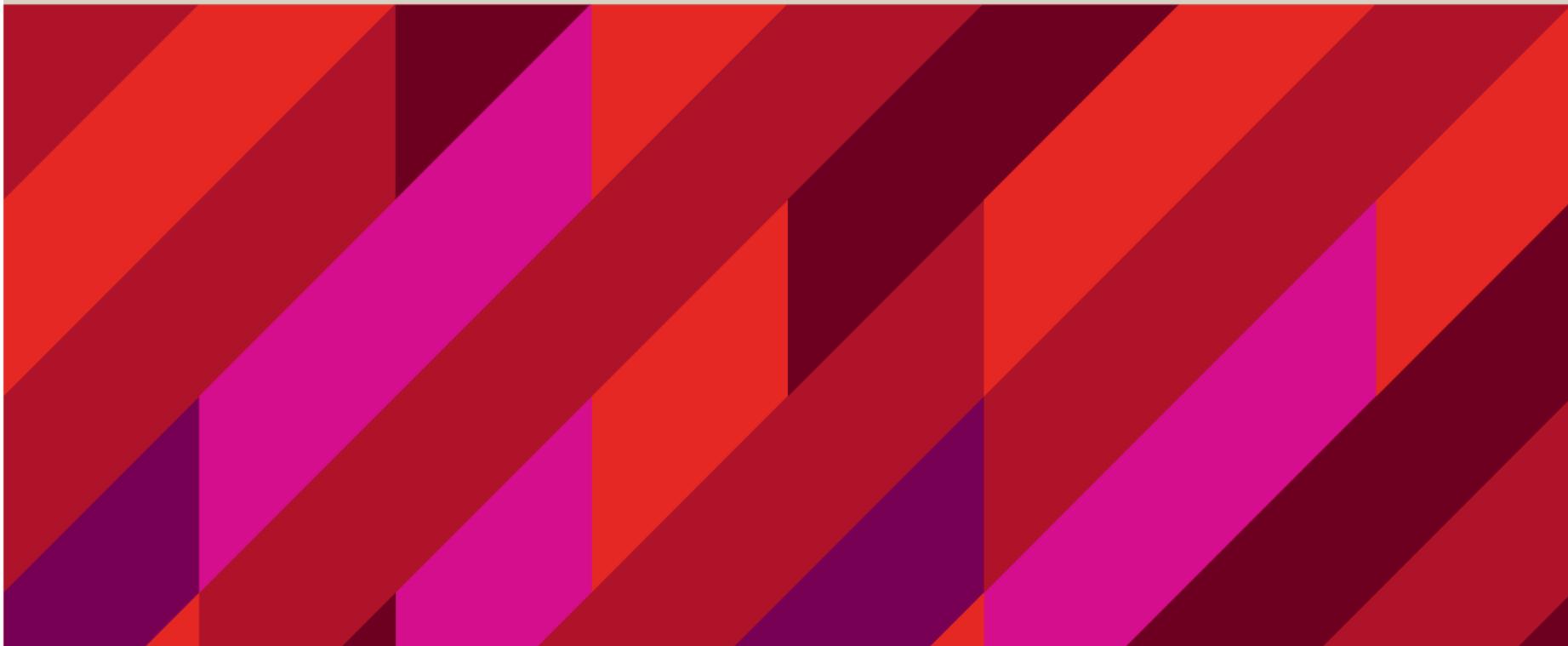




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BIOL3110 Conservation & Ecological Genetics

LECTURE 8: MUTATION, MIGRATION & SELECTION



Mutation, Migration & Selection



DEFINITIONS wrt GENETICS

Mutation: Copying errors – rare (beneficial mutations even rarer). Nevertheless, mutation is the core generator of V_G for Darwinian evolution.

Migration: among populations – brings new genes in or takes genes out. 

Selection: Different forms of selection. Can either reduce or increase V_G under different circumstances.

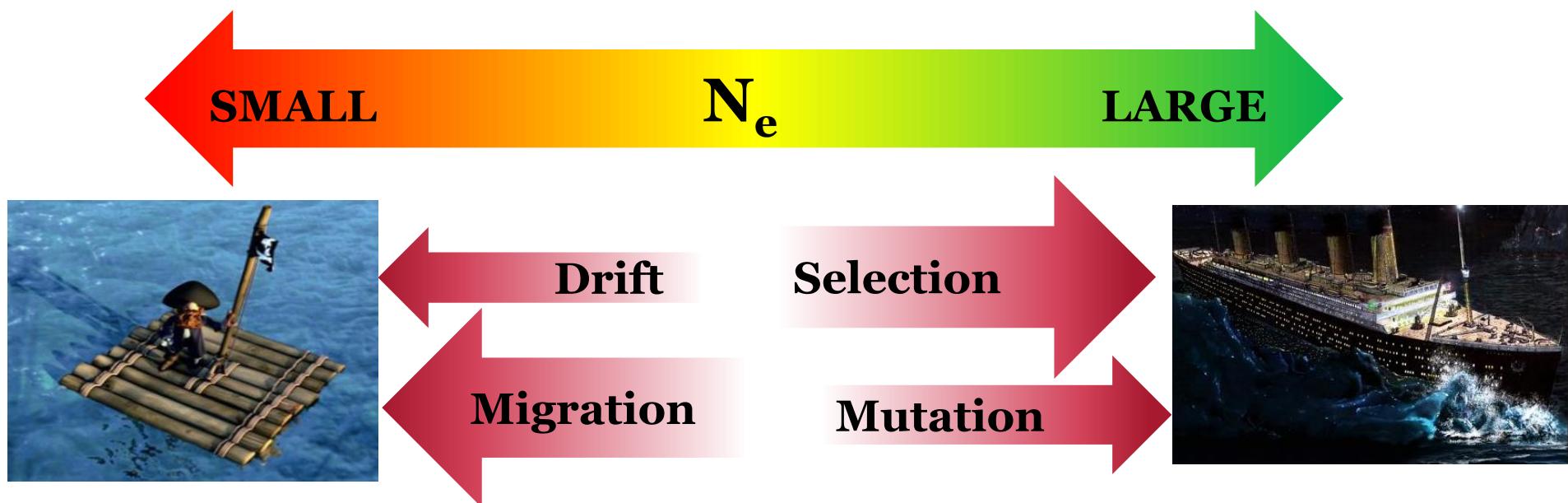
Mutation, Migration & Selection



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MORE DETAIL NEXT WEEK:

The critical importance of (effective) population size for these parameters.



Mutation, Migration & Selection

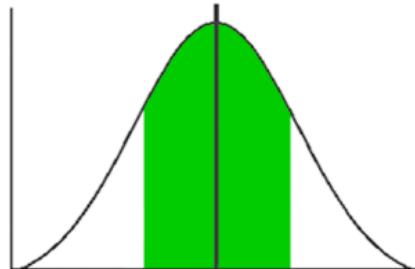


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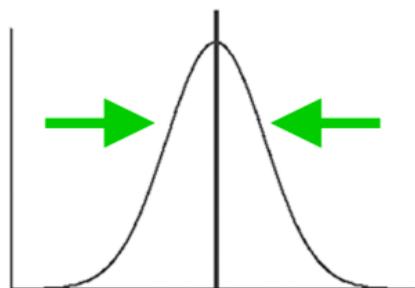
TYPES OF SELECTION (I)

Favoured

Stabilizing
Before selection:



After selection:

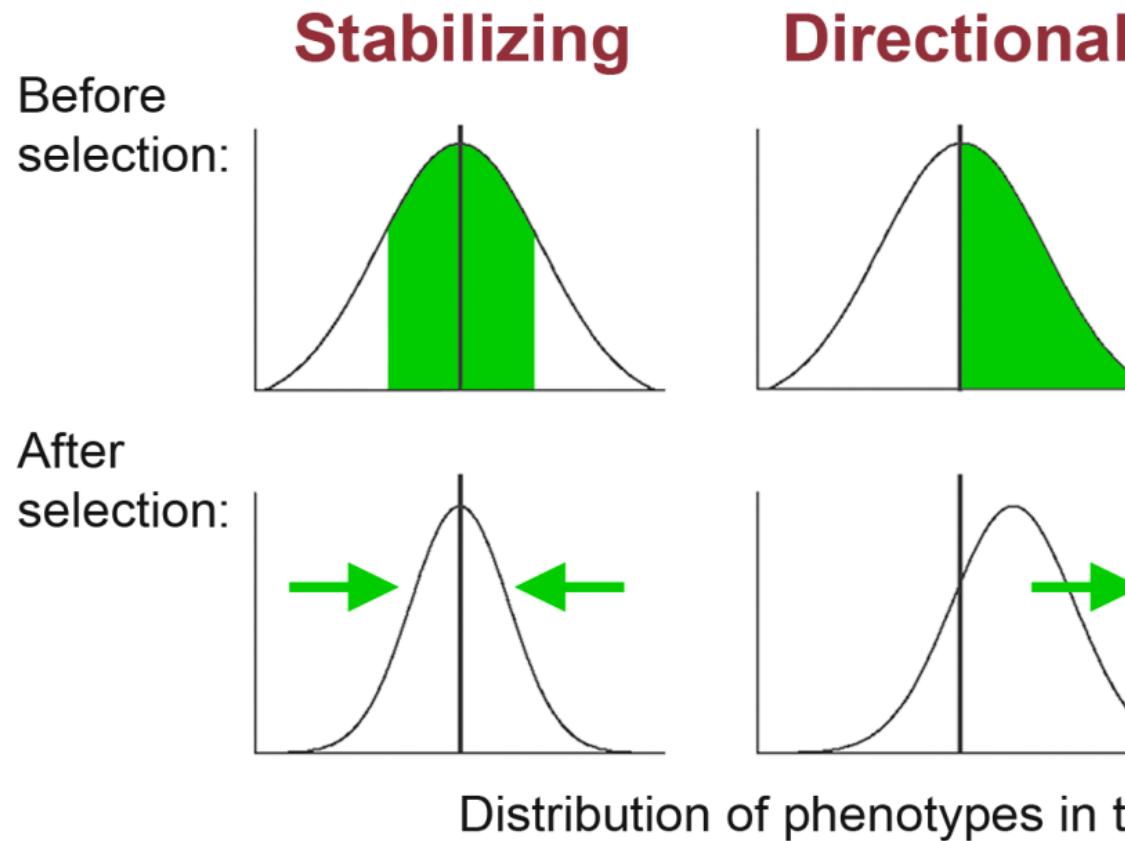


Distribution of phenotypes in the population

Mutation, Migration & Selection

TYPES OF SELECTION (II)

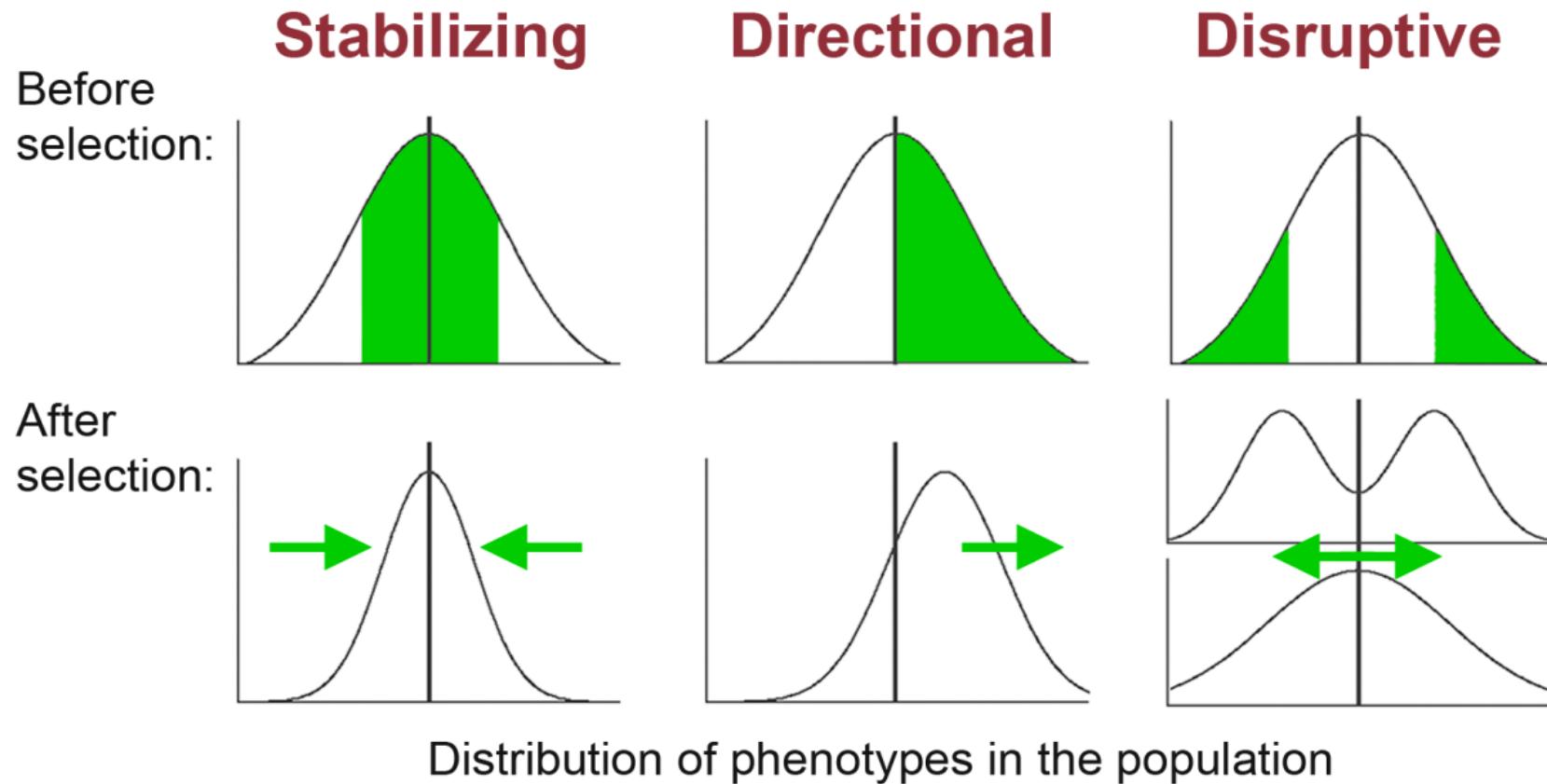
■ Favoured



Mutation, Migration & Selection

TYPES OF SELECTION (III)

■ Favoured



Mutation, Migration & Selection

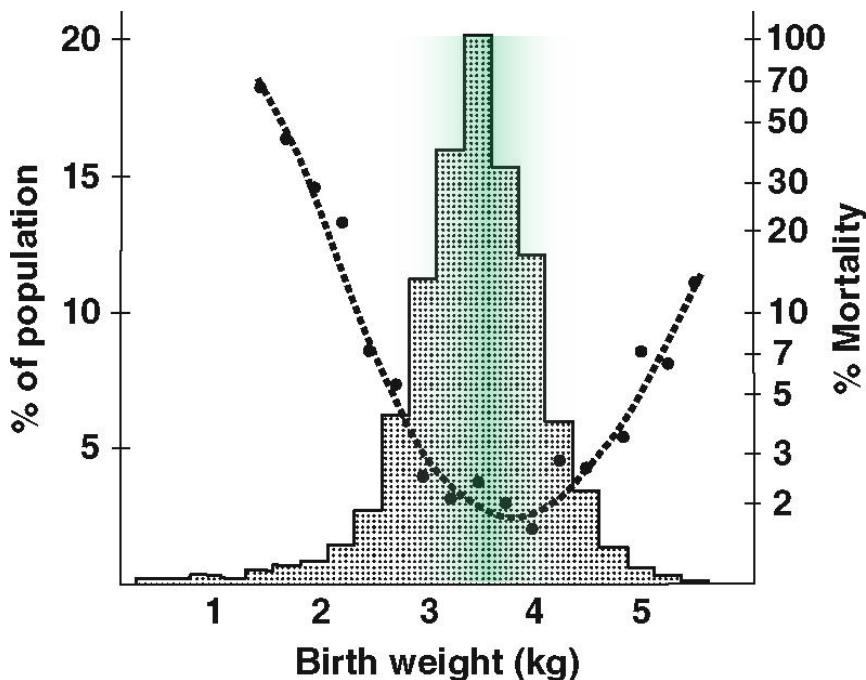


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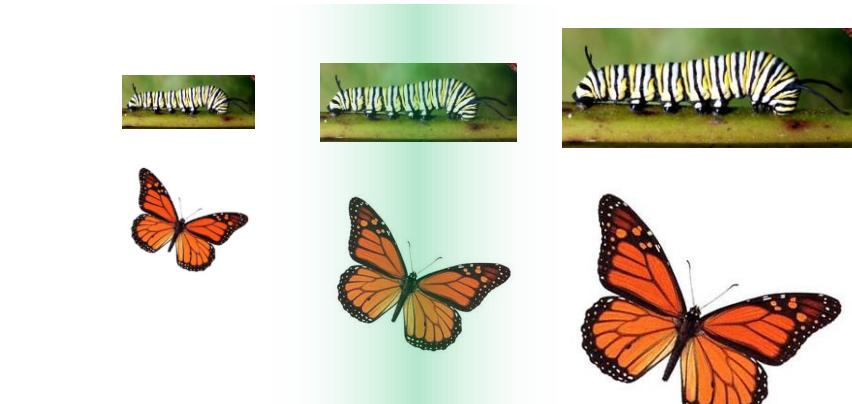
STABILIZING SELECTION

Fitness is under directional selection, BUT:

Most individual traits in wild populations are subject to **NET stabilizing selection**.



E.g. Birth weight in humans



E.g. Size at maturity in insects

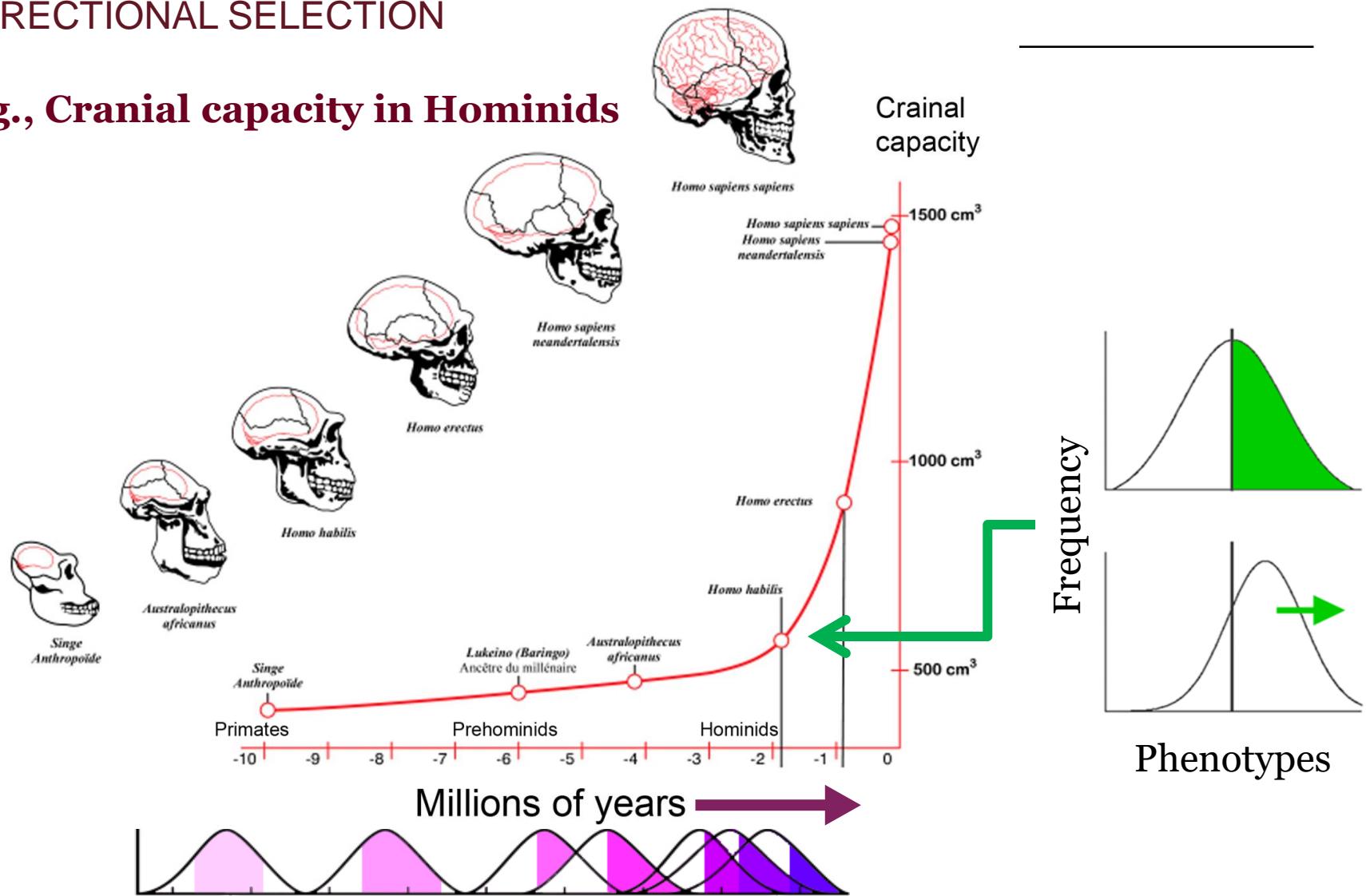
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DIRECTIONAL SELECTION

E.g., Cranial capacity in Hominids



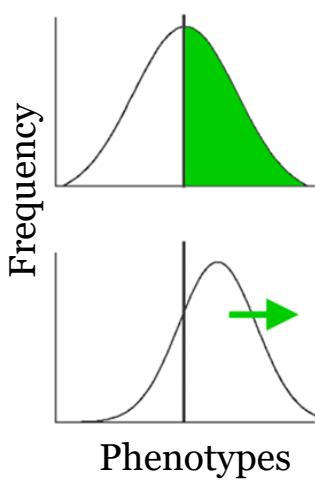
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DIRECTIONAL SELECTION

E.g. Sexual selection



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DISRUPTIVE SELECTION

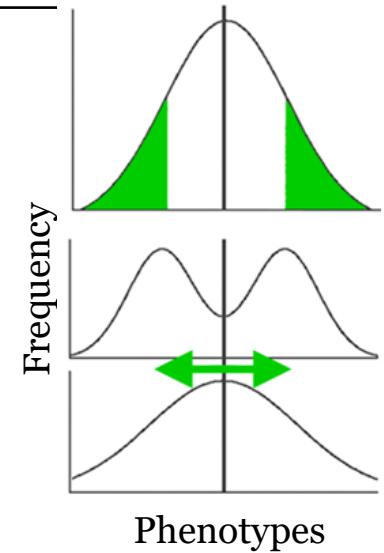
Often maintains V_G . May also favour di-morphism or poly-morphism or phenotypic plasticity.



Sexual dimorphism (e.g. *Eclectus*)



E.g. Dimorphism in
Rhino beetles



E.g. Plastic wing
color in
butterflies



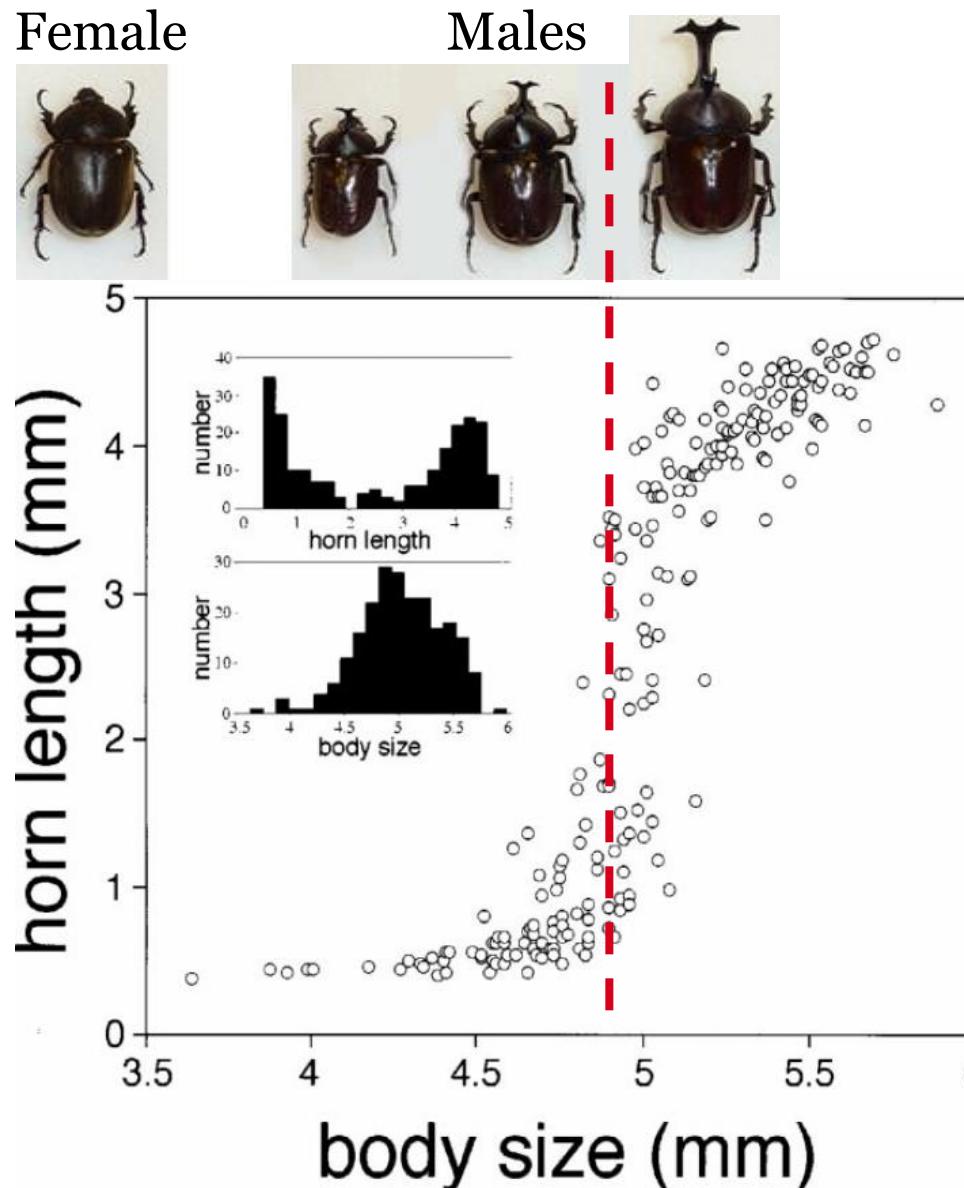
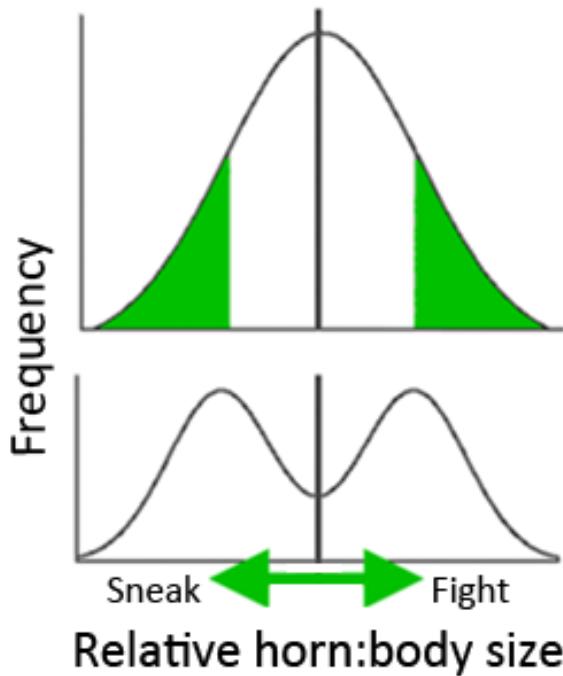
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DISRUPTIVE SELECTION

Example:
Horn polyphenism in male
Onthophagus taurus



Mutation, Migration & Selection



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MIGRATION

In whole populations:



Among-populations:



Inter-pop migration = **gene flow**

20 individuals contain 95% of V_G

Little gene flow needed to maintain substantial V_G

Higher levels required to maintain rare alleles

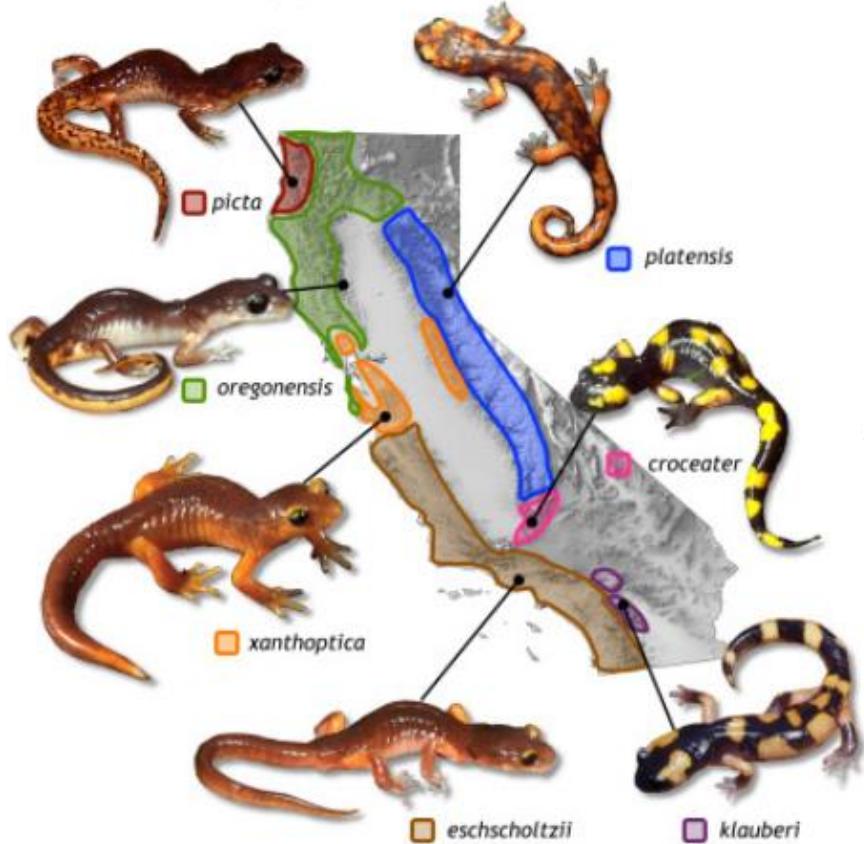
Mutation, Migration & Selection



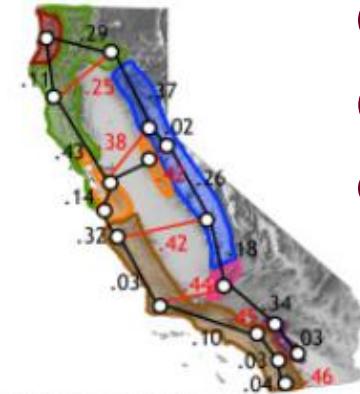
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MIGRATION versus SELECTION

a) *Ensatina* ring species



b) Genetic divergence



c) Ecological divergence



Cline:

Geographic gradient
of phenotypes

Arises as the balance
between selection for
localized adaptation
and/or drift versus
gene flow

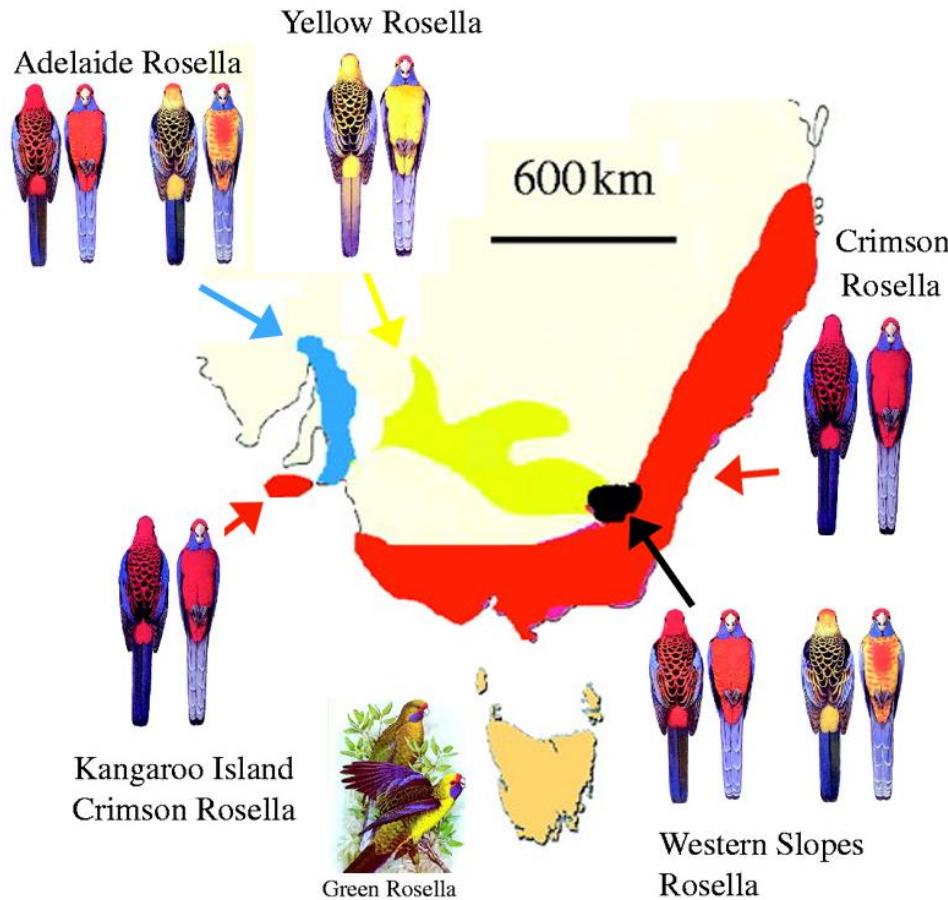
e.g. *Ensatina* salamander “ring species” in California

Mutation, Migration & Selection



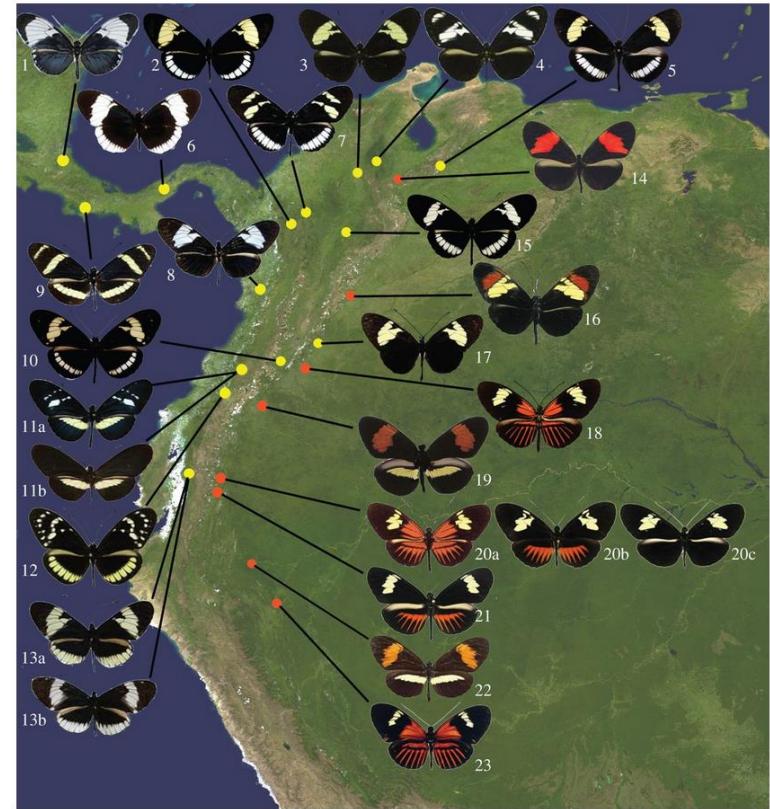
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MIGRATION versus SELECTION



Platycercus elegans

Joseph *et al.* (2008) Proc. R. Soc. Lond. (B)



Heliconius cydno

Brower (2012) Proc. R. Soc. Lond. (B)

Mutation, Migration & Selection

MUTATION

Copying errors in germ-line replication



At the genomic level:

- Single-Nucleotide Polymorphisms (**SNPs**)
- Additions, deletions & duplications (e.g., microsats)
- Insertion of transposable or “mobile” elements



Rates of point mutation:

1 mutation per locus per 100,000 gametes per generation

= **10 mutations/individual** (typical eukaryote ~1M loci)

Mutation among microsatellites **~10x higher rate**

Mutation, Migration & Selection



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MUTATION

Rates of point mutation (1 mutation/locus/100,000 gametes/generation)

Corroboree frog:

250 individuals

16-40 eggs per clutch



$250 \times 1M \text{ loci} = 250,000,000 \text{ loci}$

$125 \text{ pairs} \times 40 \text{ gametes} = 5,000 \text{ gametes}$

1 generation per year

= 50,000 (**5 x 10⁴**) mutations per year

Bufo marinus:

200,000,000 individuals

30,000 eggs per clutch



$2 \times 10^{14} \text{ loci}$

$3 \times 10^{12} \text{ gametes}$

~4 clutches per year

= **1 x 10¹⁴** mutations per year

A spatial analogy:

3m²

versus:



Mutation, Migration & Selection



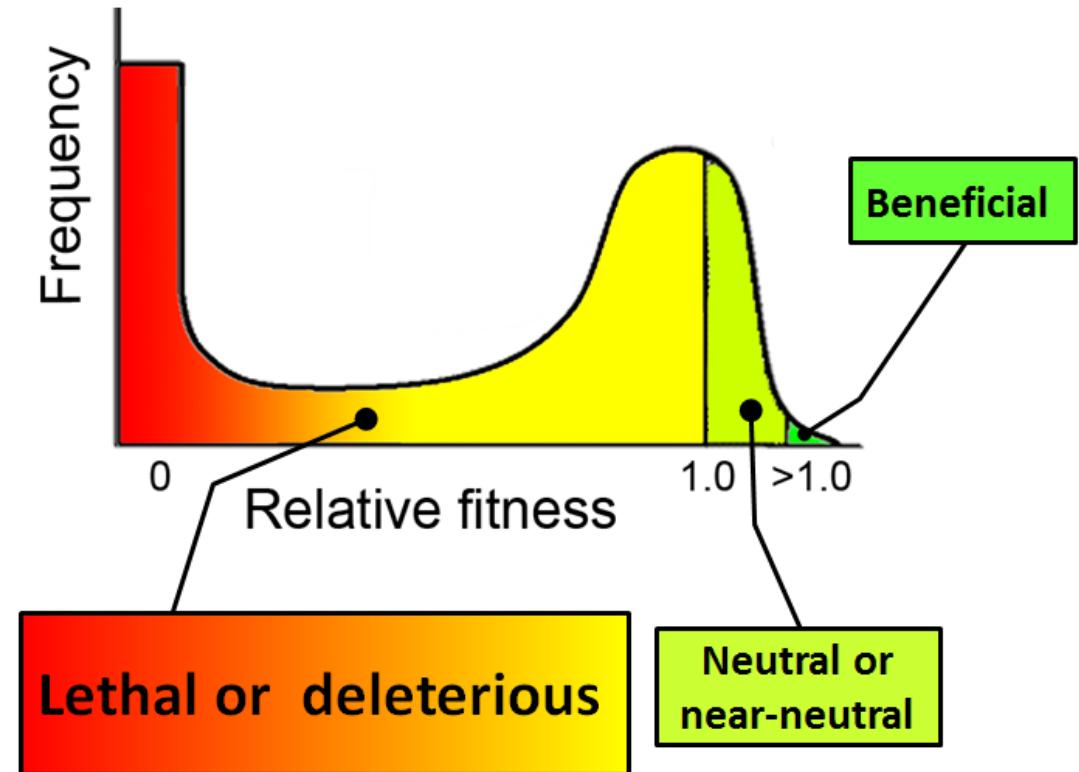
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MUTATION

Generates V_G in the long term

Most mutations deleterious

Eliminated quickly by selection
unless recessive...



Mutation, Migration & Selection



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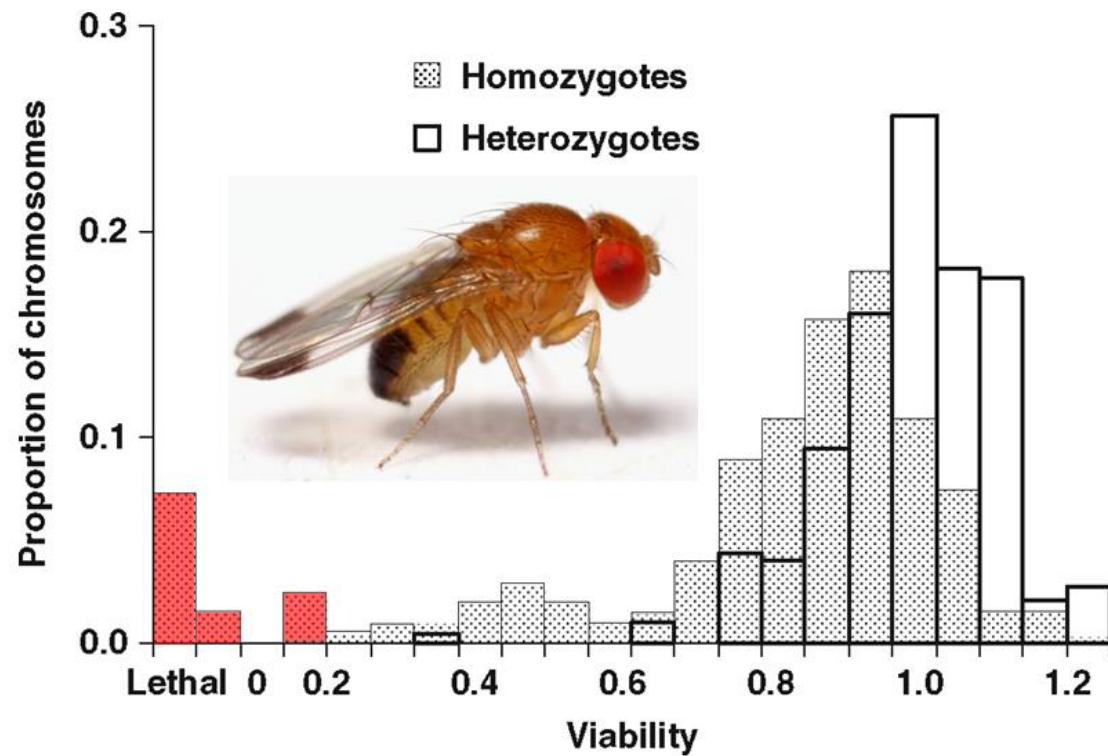
MUTATIONAL LOAD

1. Low frequency (<1% per locus) of deleterious mutations
2. Exist as recessives (rarely exposed)
3. Thought to occur in most species & loci

**Sum for population
called
“Mutational load”**



Fitness effects experimentally revealed using inbred lines (=increased homozygotes):



Mutation, Migration & Selection



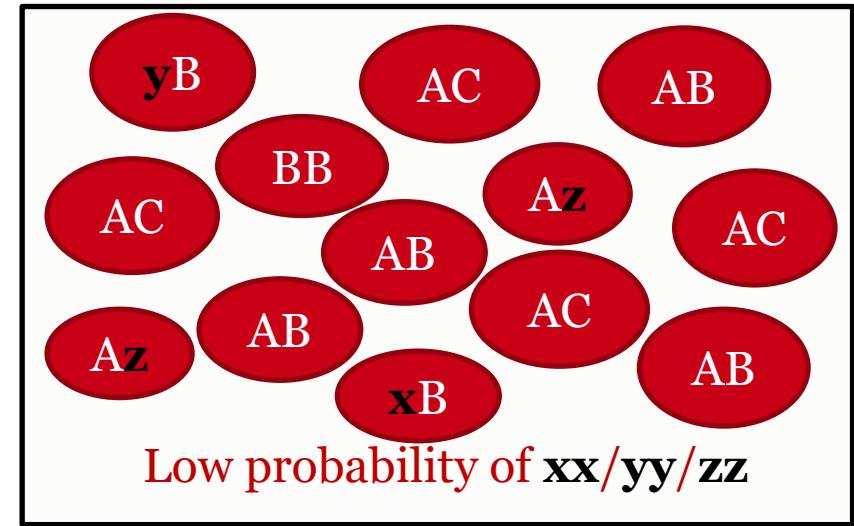
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MUTATIONAL LOAD

Mutational load is increasingly revealed as populations become smaller

Via the increased chance of deleterious recessives pairing together, and being expressed

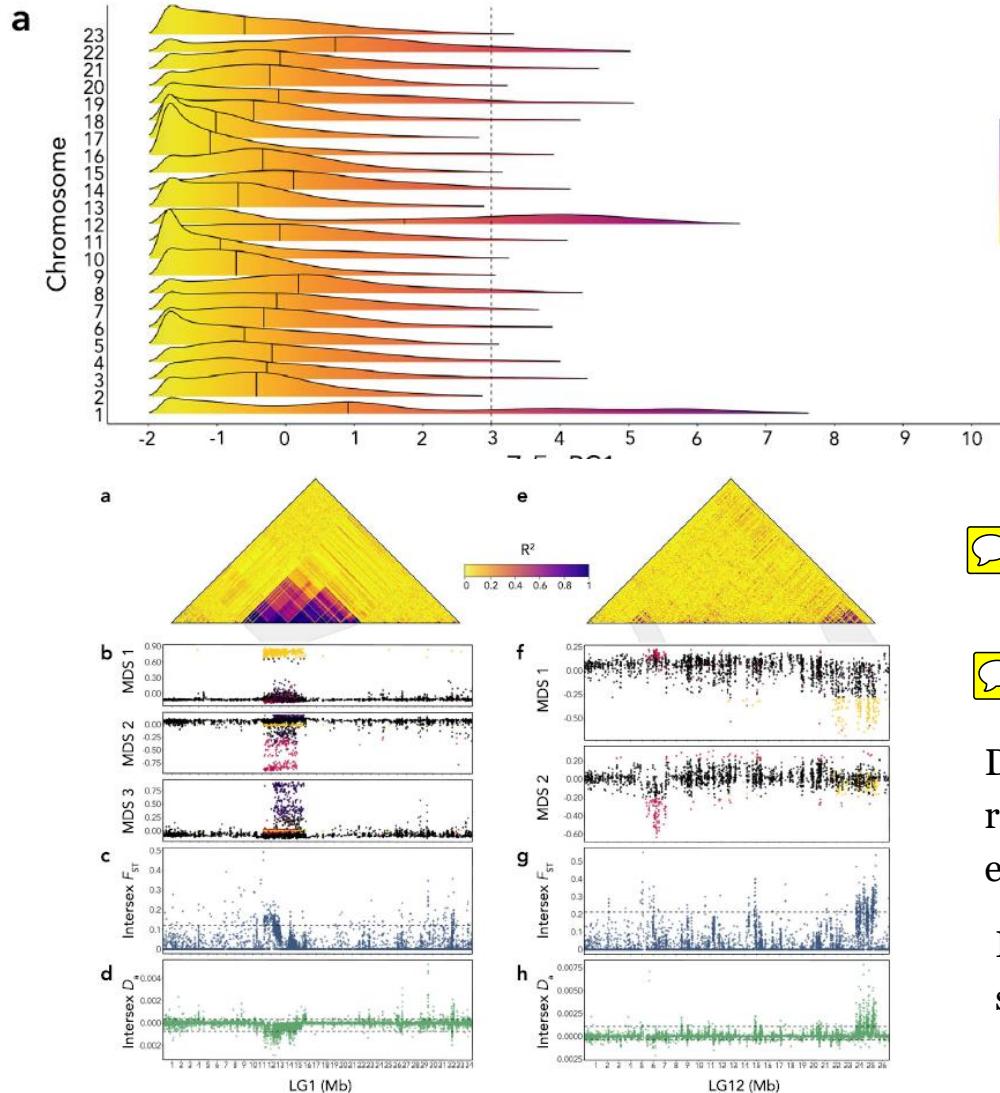
This is the mechanism of inbreeding depression



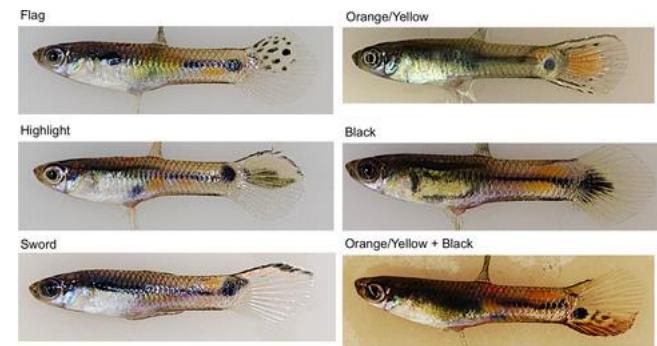
Size reduction



Chromosome 1 (autosome) and 12 (Y chromosome) sig differentiated – cr 1 more so



Trinidadian guppy –
males colorful – 4 ‘Iso Y lines’
True breeding 40 gen



Differentiation (cr 1 & 12) associated with regions of low recombination – physical linkage – except for intersex comparisons

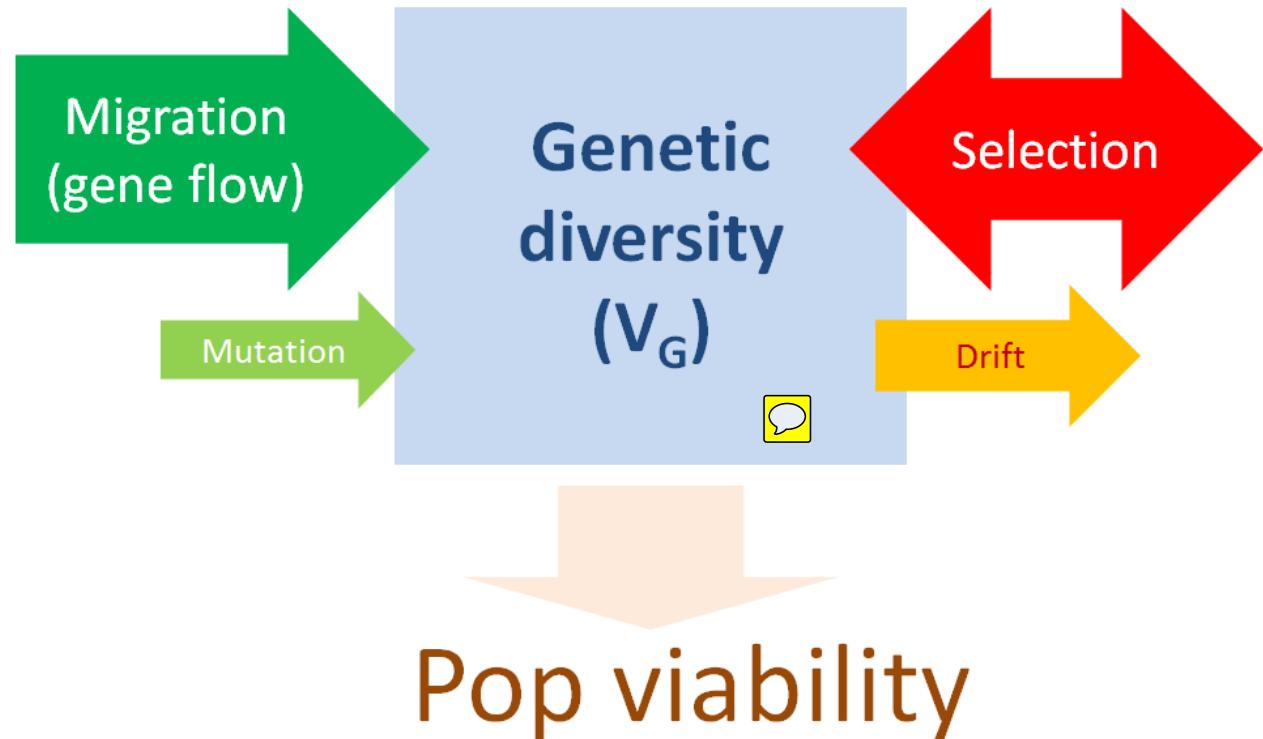
Epistasis between 1 & 12 – increases diversity & shields Y region diversity from drift

Mutation, Migration & Selection



MORE ON THIS NEXT WEEK...

V_G is a **balance** between selection, drift, migration & mutation;



Increasingly **stable** with increasing N.

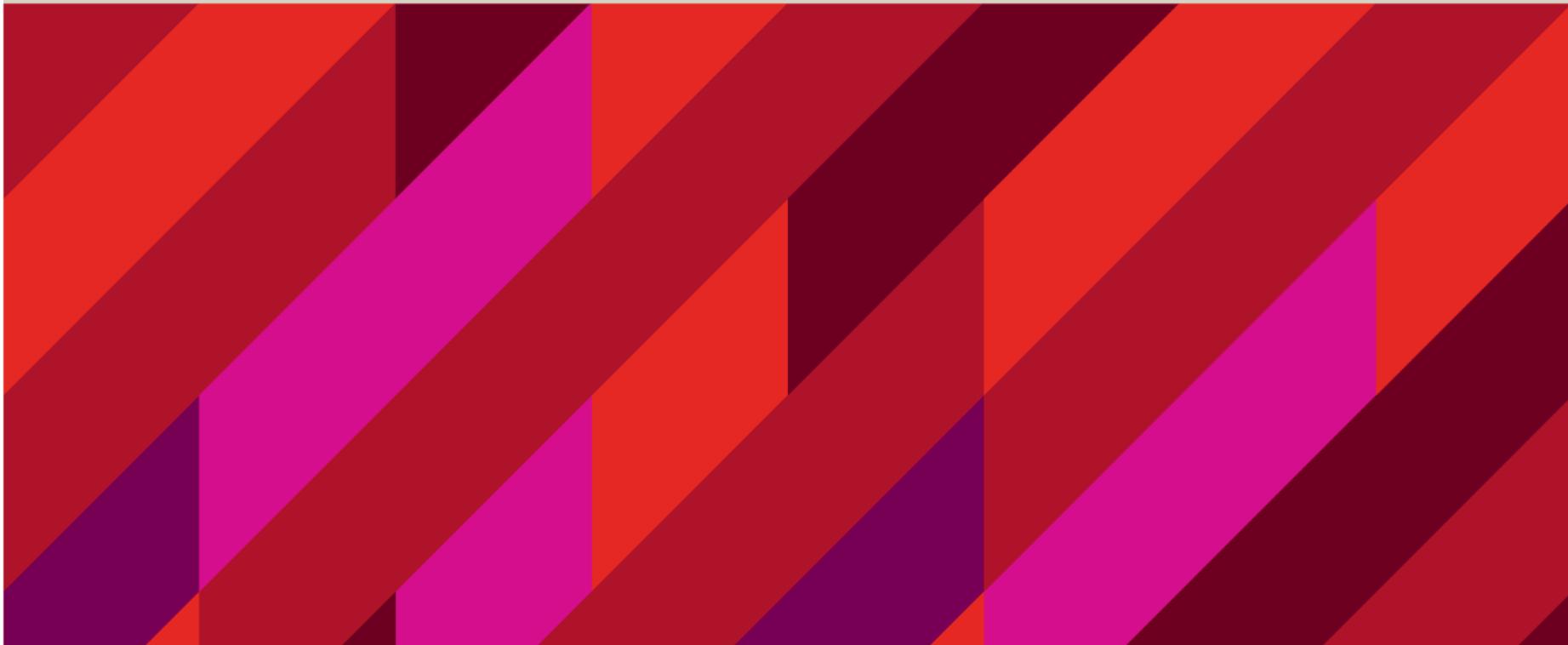
Long-term accumulated negative mutations revealed by small N.



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BIOL334 Conservation & Ecological Genetics

LECTURE 9: MUTATION, MIGRATION & SELECTION II



Mutation, Migration & Selection

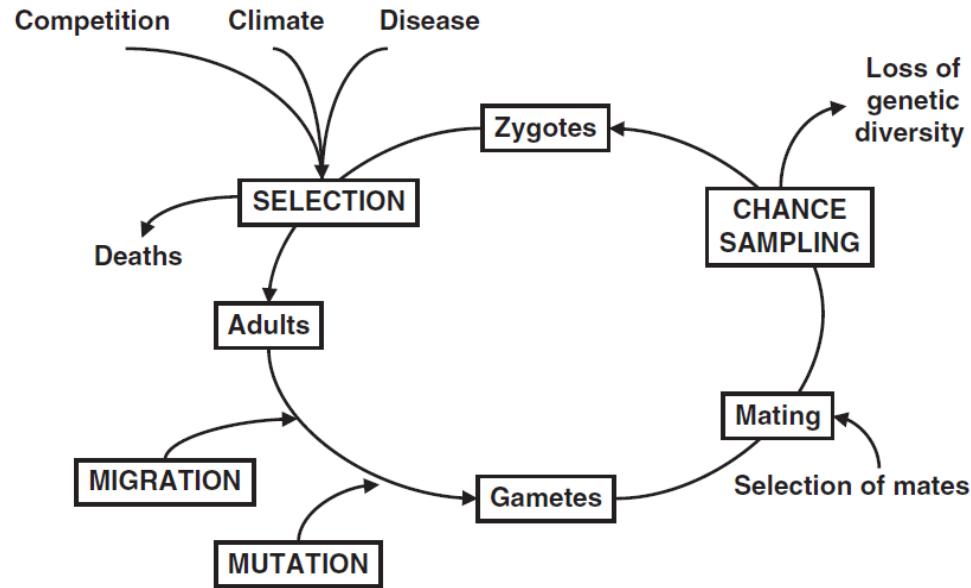


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Assignment #1 grades on iLearn

Continuing from Lecture 8...

Fig. 6.1 An evolving population as a complex system.



Genetics & Population size

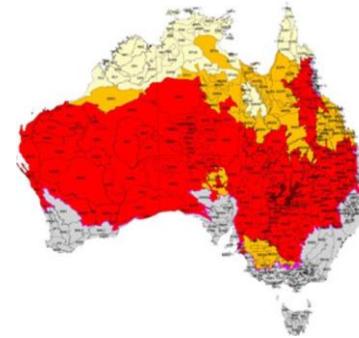
LARGE vs SMALL POPs

Large N

High levels of V_G
= “variant” alleles
= **resilience** to change

High mutation potential
= **NEW** V_G

Power for selection (all forms)
= **adaptive potential**



Rabbit
N~10 Billion (1920)



Water buffalo
N~100,000

Genetics & Population size

LARGE vs SMALL POPS

Large N

High levels of V_G
= “variant” alleles
= **resilience** to change

High mutation potential
= **NEW** V_G (over time)

Power for selection (all forms)
= **adaptive potential**

Small N

1. Low V_G
= **vulnerable** to change
2. few new mutations, **BUT**
existing mutations exposed
3. **Selection** overpowered
by **genetic drift...**

Genetics & Population size

GENETIC DRIFT

Random **luck of the draw...**

Chance decides allele frequencies from one generation to the next;



These effects accumulate – once a rare gene is lost, it is gone forever!

Causes small populations to evolve in ‘random’ directions – i.e. to **drift...**

Loss of rare alleles via drift



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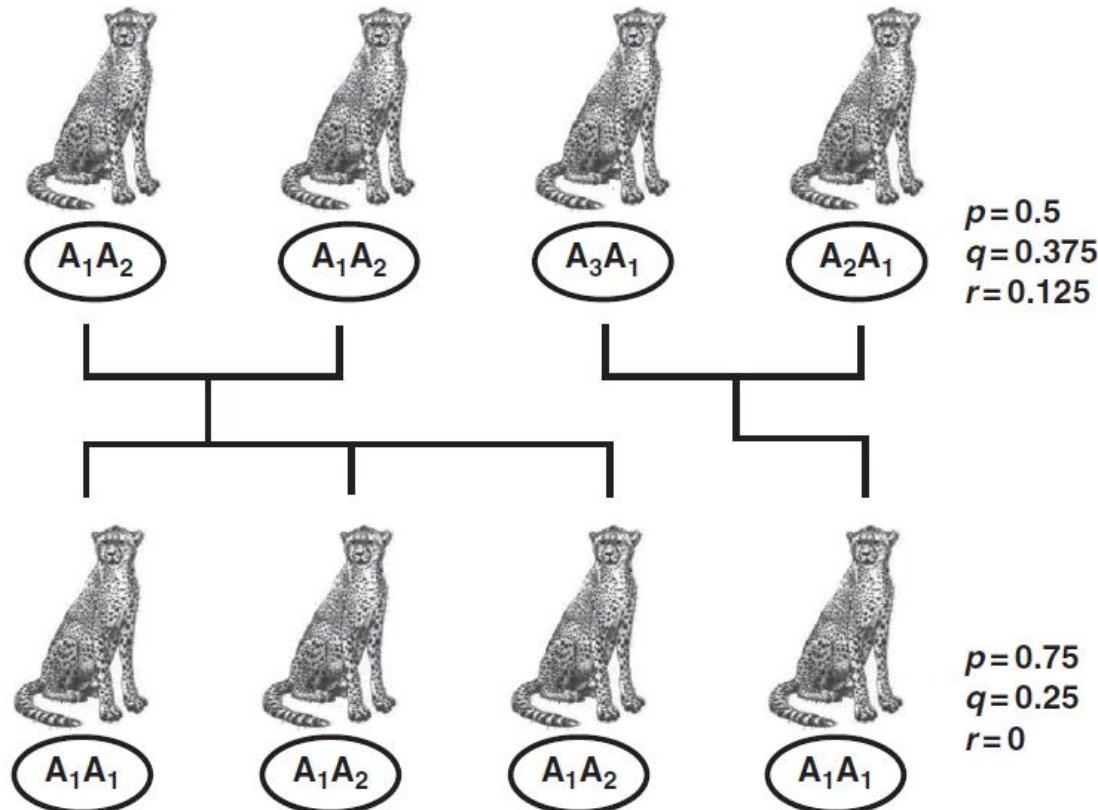


Fig. 8.1 Genetic drift in allele frequencies in a small population of cheetahs. p , q and r are the frequencies of alleles A_1 , A_2 and A_3 , respectively. Allele A_3 is lost by chance. Further, the frequencies of A_1 and A_2 change from one generation to the next, with A_1 rising and A_2 falling.

Genetics & Population size



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GENETIC DRIFT

Population N=36

AZ	x	AB	AB	x	BB
AB	x	AA	ZA	x	AB
AA	x	BA	AB	x	AA
AB	x	AZ	AA	x	BZ
AB	x	AB	AB	x	BA
ZB	x	AA	BB	x	AA
AA	x	BZ	AA	x	BZ
ZA	x	BA	AB	x	BA
BB	x	BA	BB	x	AZ

Allele frequencies

A: 36 (50.0%)
B: 27 (37.5%)
Z: 9 (12.5%)

Gen
+1

Nine 50:50
chances of
retaining
the Z allele

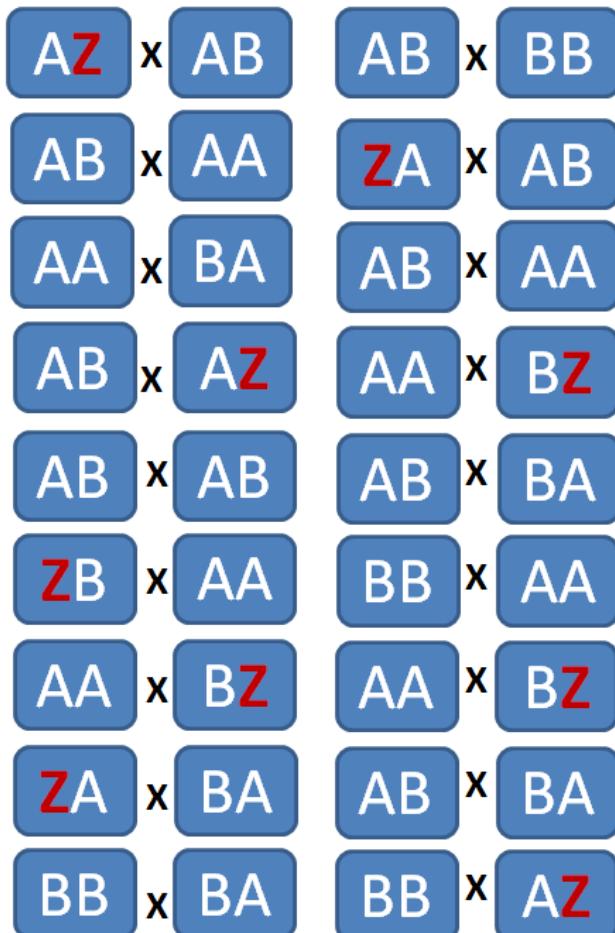
Genetics & Population size



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GENETIC DRIFT

Population N=36

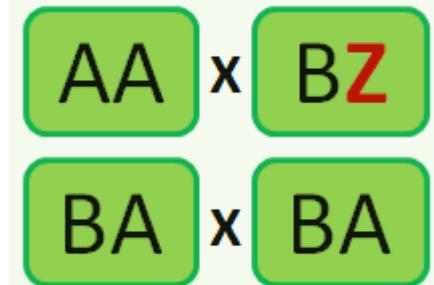


Allele frequencies

A: 36 (50.0%)
B: 27 (37.5%)
Z: 9 (12.5%)

Gen
+1

Nine 50:50
chances of
retaining
the Z allele



N = 4 (8 alleles)
A: 8 (50.0%)
B: 7 (37.5%)
Z: 1 (12.5%)

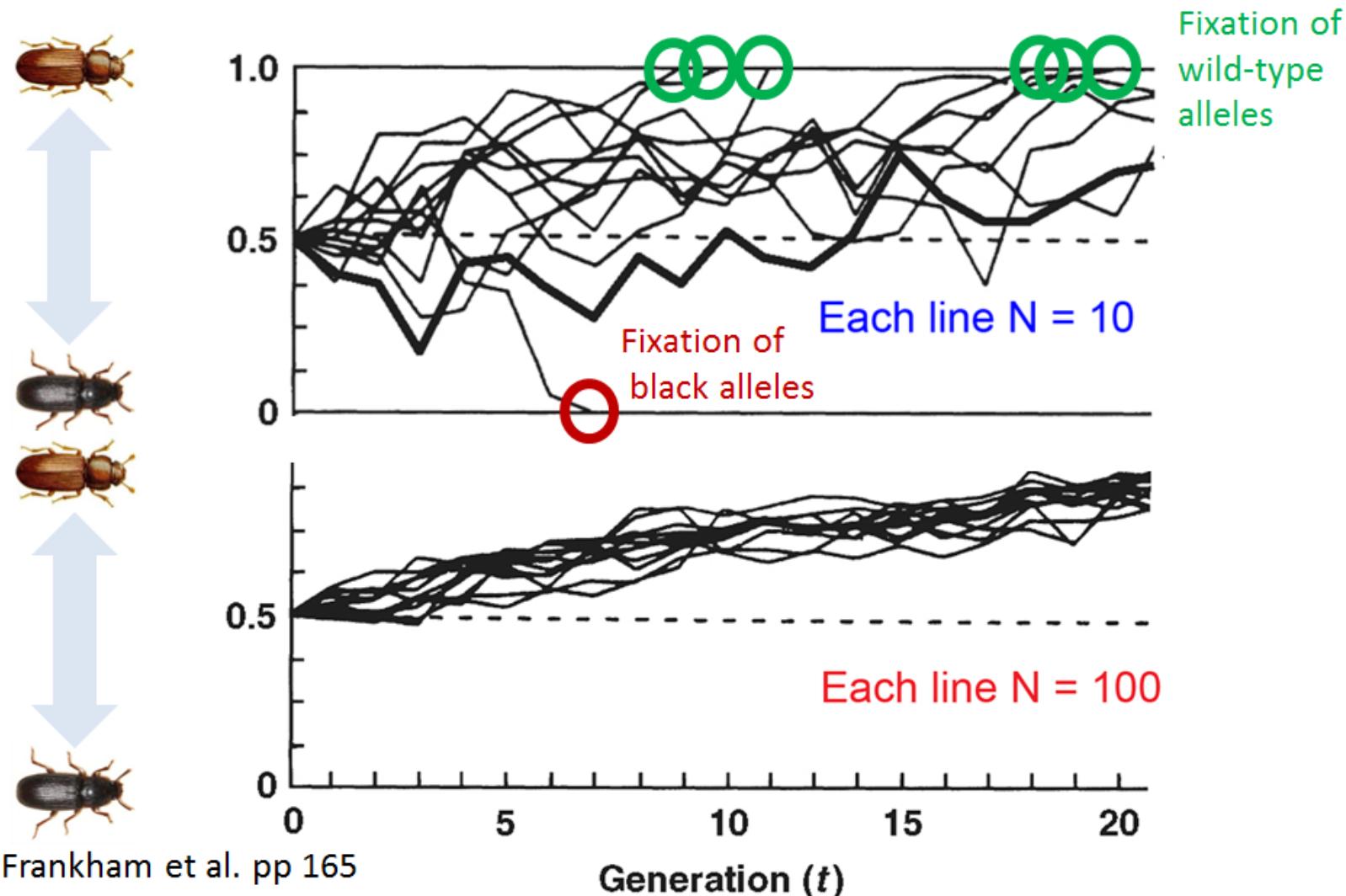
ONE 50:50
chance of
retaining
the Z allele

Genetics & Population size



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GENETIC DRIFT vs SELECTIVE POWER

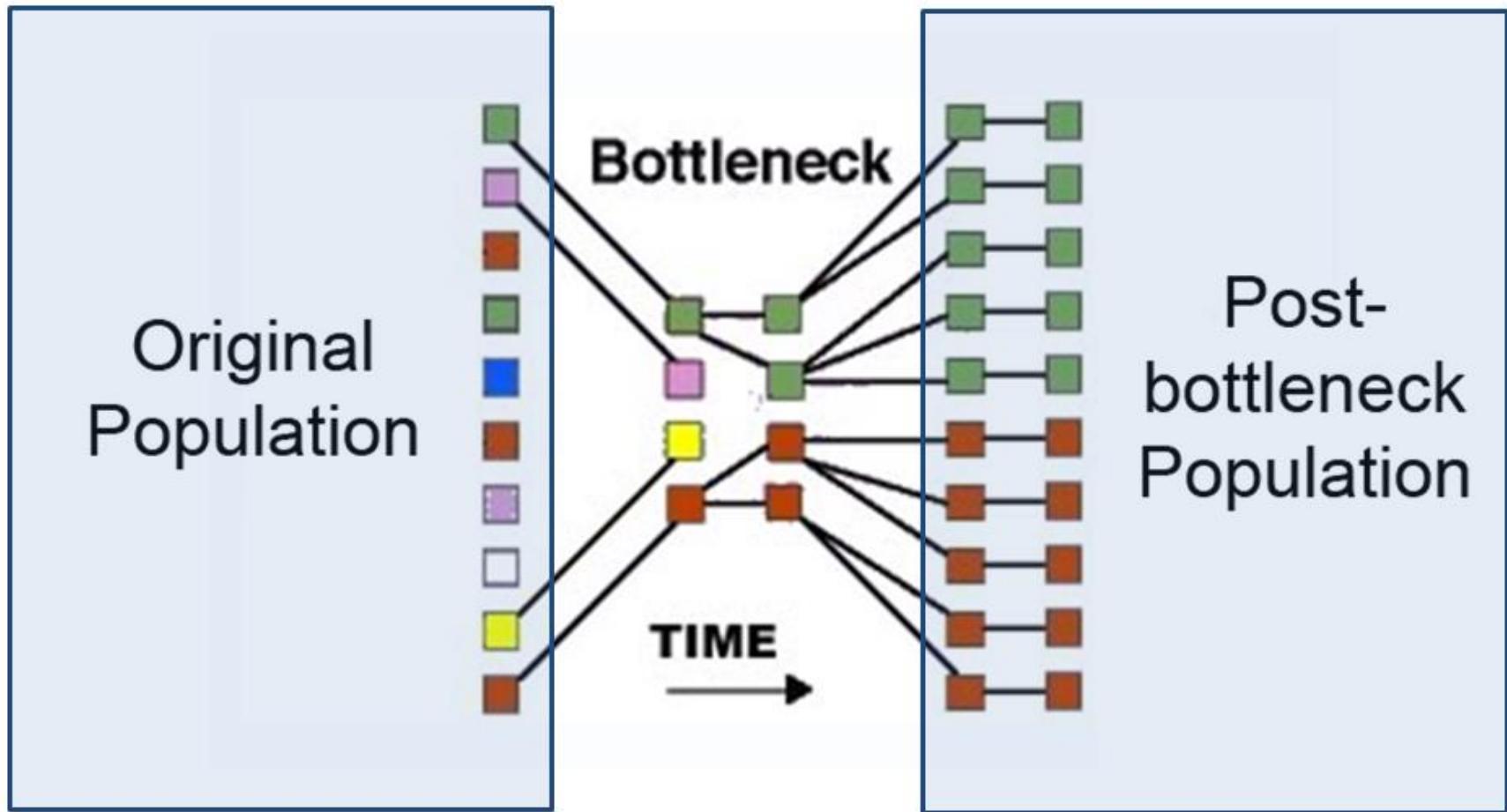


Genetics & Population size



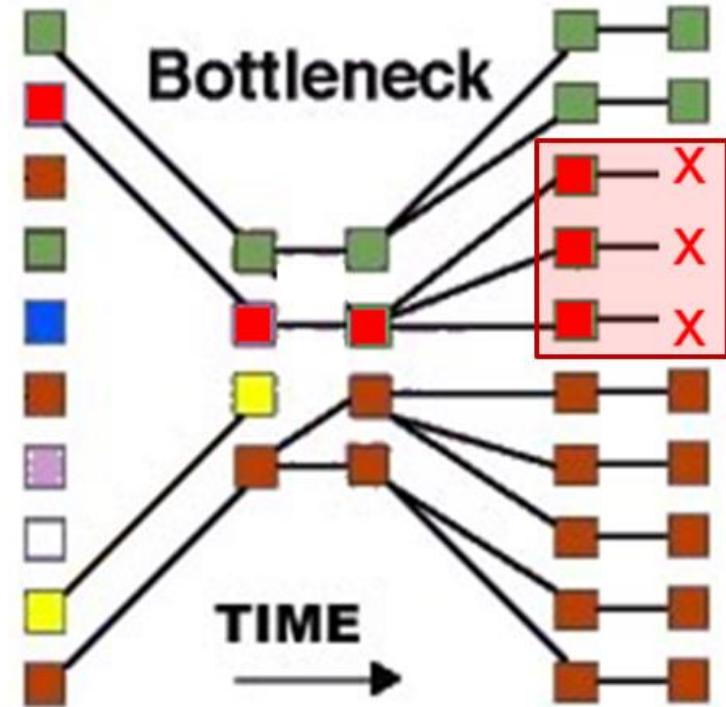
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BOTTLENECKS IN THE PAST...



Genetics & Population size

SUBHEADING

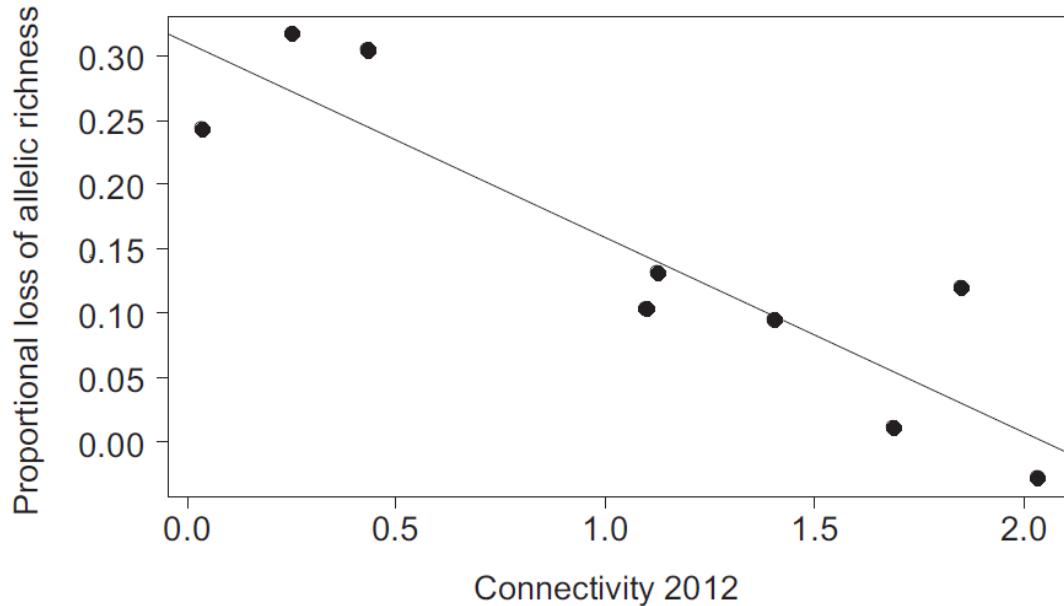


Northern Elephant Seal:
20-30 individuals in late 1800s
>175,000 now

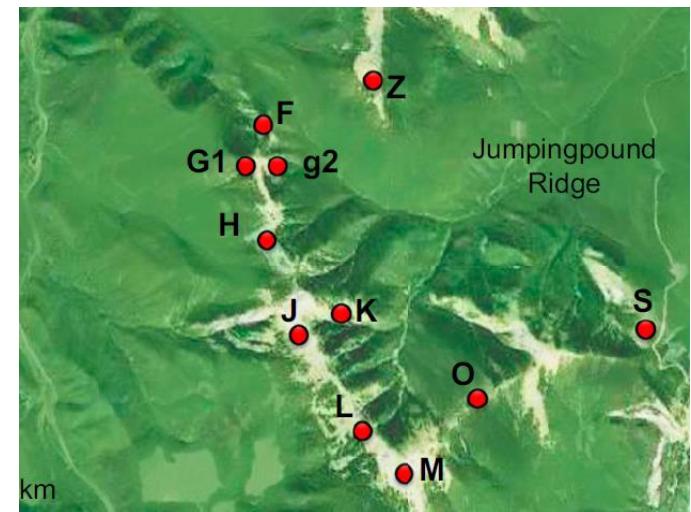
Importance of connectivity



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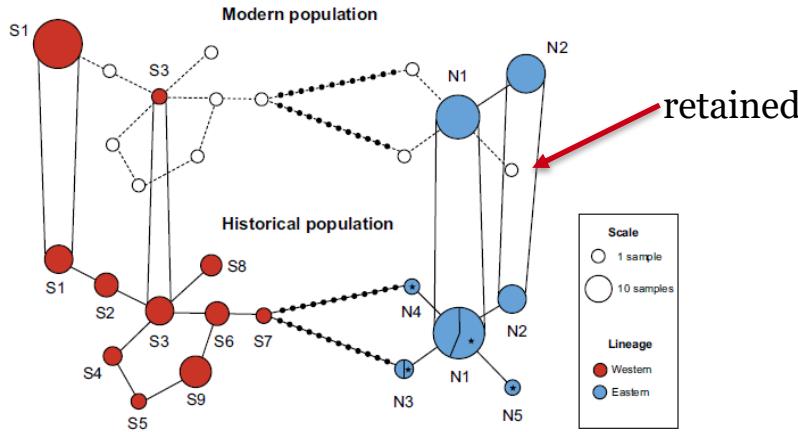
Jangjoo et al. 2016 PNAS



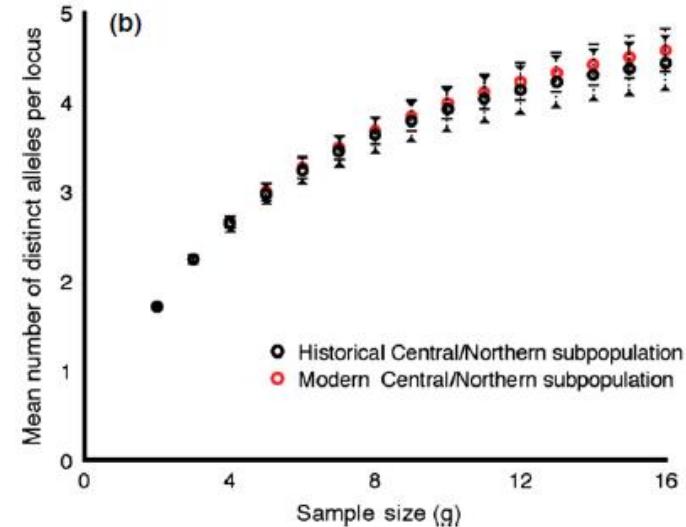
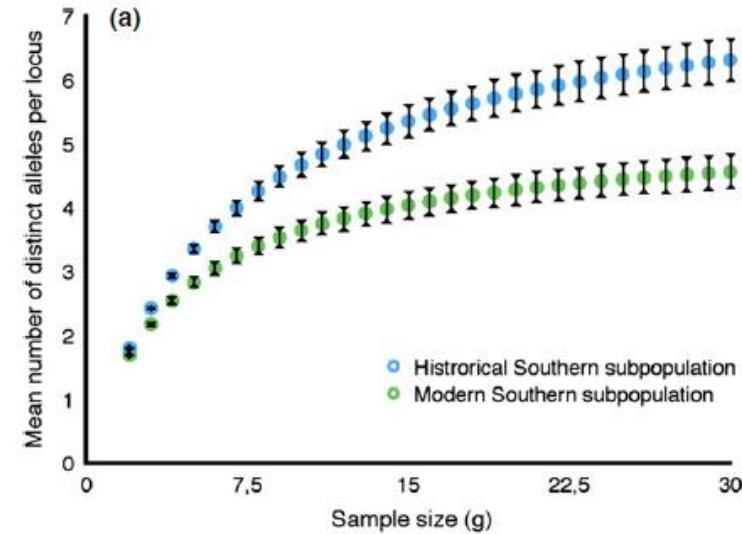
Magnitude of bottleneck effects vary across a distribution



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Xenidoudakis et al.
2015 Mol Ecol



Genetics & Population size

LARGE vs SMALL POPS

Original release by Thomas Austin in 1859 (Vic).

Austin hunted rabbits on his weekends in England. Upon arriving in Australia, Austin requested **12 grey rabbits, 5 hares, 72 partridges and some sparrows** to fuel his hobby.

His nephew sent **grey & domestic rabbits** to meet this order.

Many other releases of rabbits following Austin.



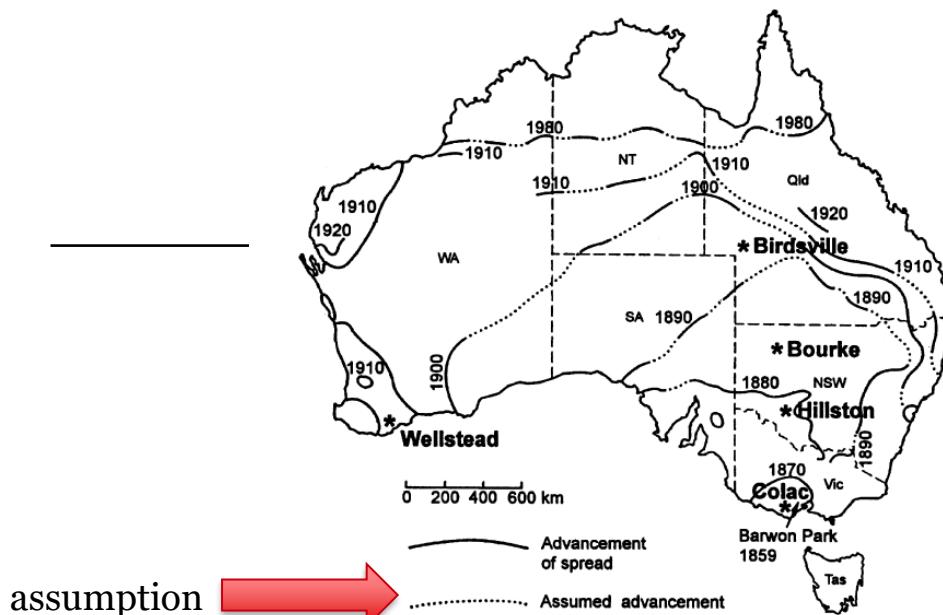


Fig. 1 The summary spread of the rabbit over mainland Australia (after Stodart & Parer 1988). Map also shows geographical location of sample sites.



Table 1 Estimates of genetic diversity (Mean \pm SE) for rare alleles (rA ; = 0.05 frequency), number of alleles (A), observed and expected heterozygosity (H_O and H_E) and unique alleles (uA) across all loci for all countries (**bold**) and across individual Australian populations. Results of heterozygosity excess test (HET), mode shift and M ratio analysis where ‘–’ indicates no significant reduction in N_e

Sample location	n	rA	A	H_O	H_E	uA	HET TPM	Mode shift	M ratio§ (M)
Spain†	110	4.10 ± 0.32	9.62 ± 0.39	0.66 ± 0.03	0.79 ± 0.02	4.86 ± 0.70	—	—	—
France†	257	1.23 ± 0.20	5.03 ± 0.26	0.63 ± 0.03	0.64 ± 0.03	0.86 ± 0.26	—	—	$0.477\text{--}0.554^*$
England‡	340	1.68 ± 0.22	5.05 ± 0.29	0.44 ± 0.06	0.64 ± 0.04	1.00 ± 0.60			
Australia	252	0.97 ± 0.17	5.03 ± 0.21	0.66 ± 0.02	0.67 ± 0.01	0.29 ± 0.18			
Wellstead	49	1.84 ± 0.55	5.57 ± 0.61	0.64 ± 0.06	0.65 ± 0.03	0.71 ± 0.42	—	—	0.519^*
Birdsville	49	1.14 ± 0.26	5.00 ± 0.53	0.64 ± 0.04	0.66 ± 0.03	0.43 ± 0.20	—	—	0.514^*
Bourke	47	1.00 ± 0.22	4.86 ± 0.26	0.69 ± 0.04	0.68 ± 0.02	0.14 ± 0.14	—	—	0.491^*
Hillston	57	0.43 ± 0.30	5.14 ± 0.46	0.72 ± 0.04	0.72 ± 0.03	0.14 ± 0.14	*	—	0.525^*
Colac	50	0.43 ± 0.20	4.43 ± 0.30	0.63 ± 0.05	0.65 ± 0.01	0	*	*	0.437^*

†Data obtained from Queney *et al.* (2001).

‡Data based on sat5, sat7 and sat8 from Surridge *et al.* (1999).

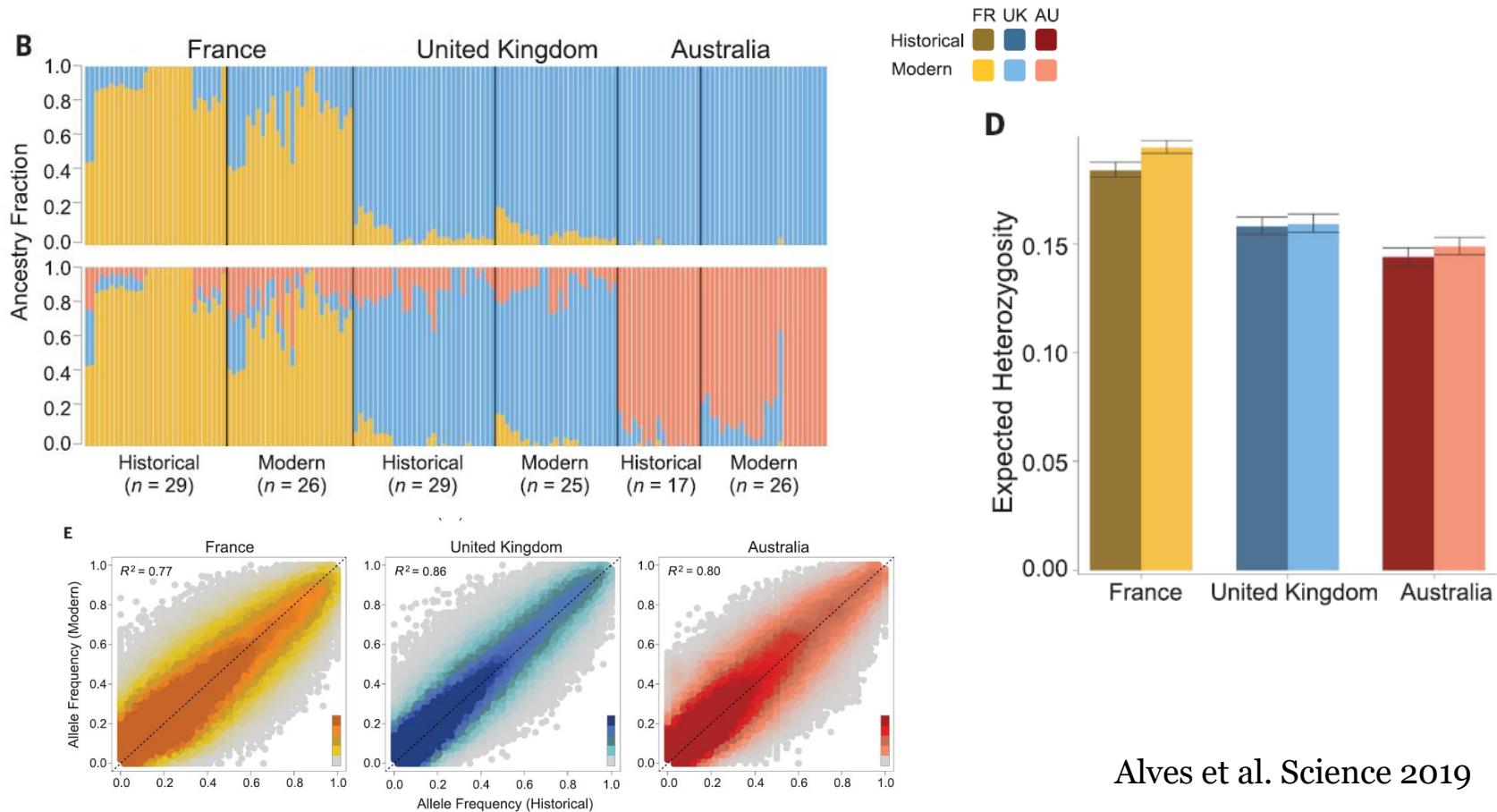
§Significance based on $M < M_C$ where $M_C = 0.712\text{--}0.828$; * $P < 0.05$.

No sequential loss in variation?

Lower Australian Vg supported by large scale SNP analysis

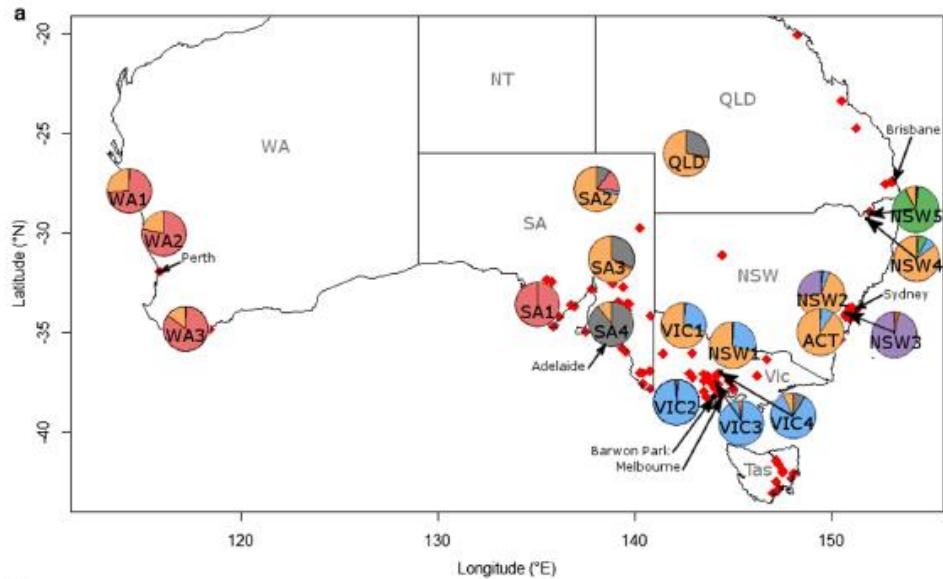


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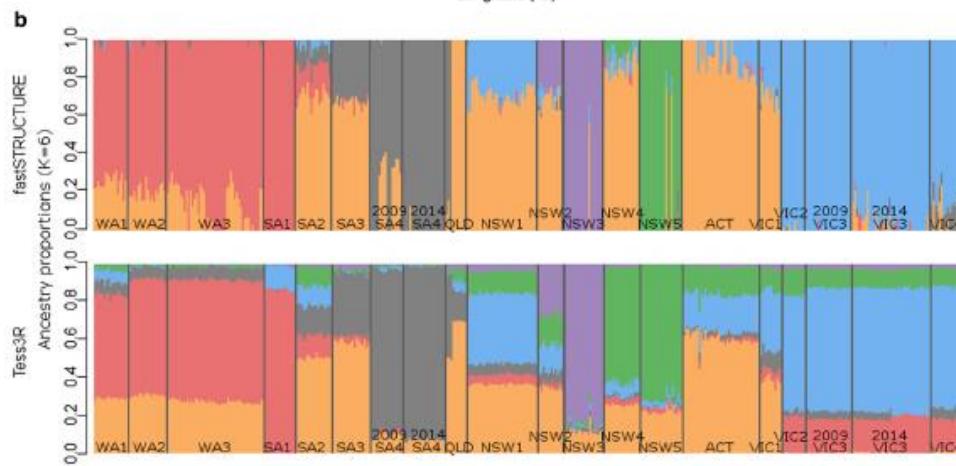


Alves et al. Science 2019

Multiple Introductions

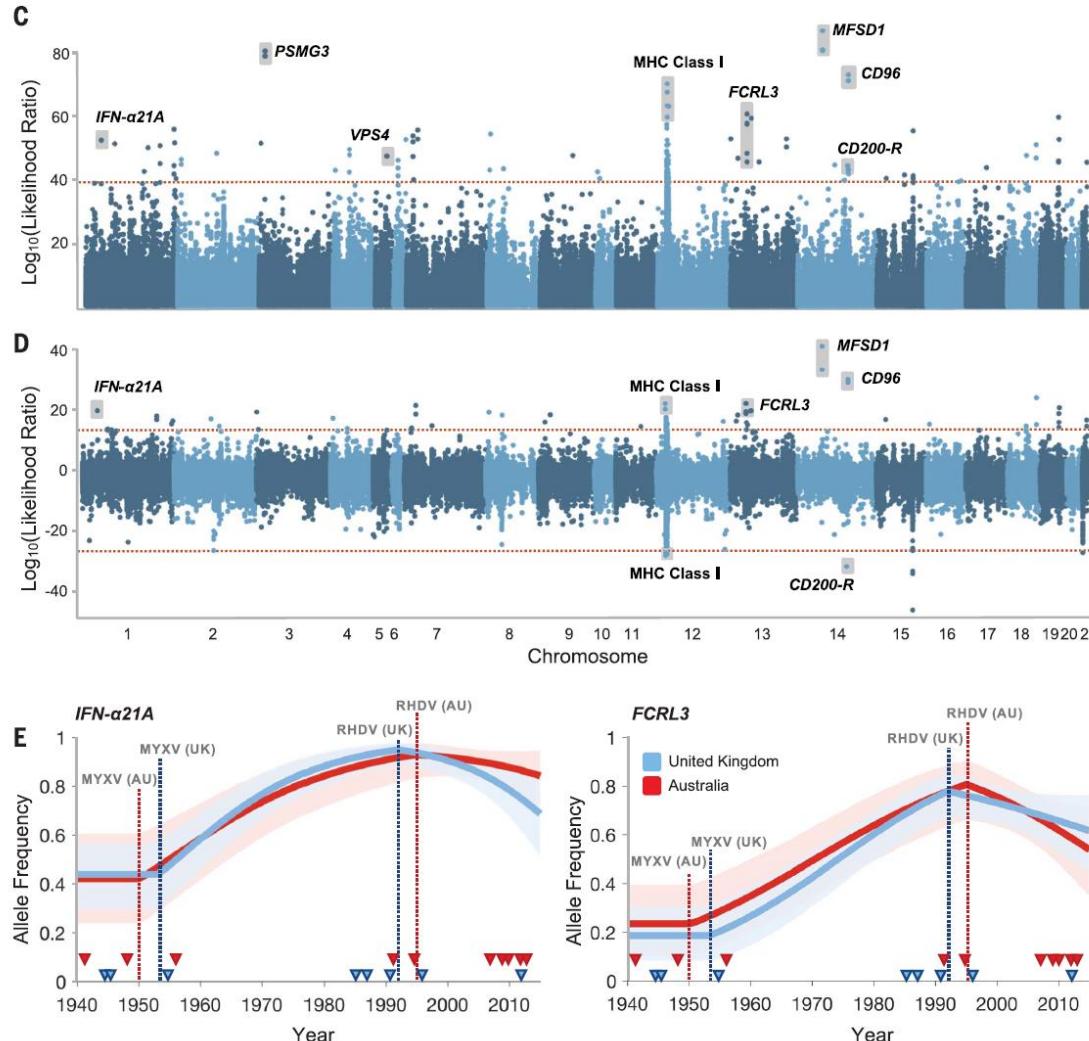


~ 40000 SNPs



Iannella et al.
Biological Invasions 2019

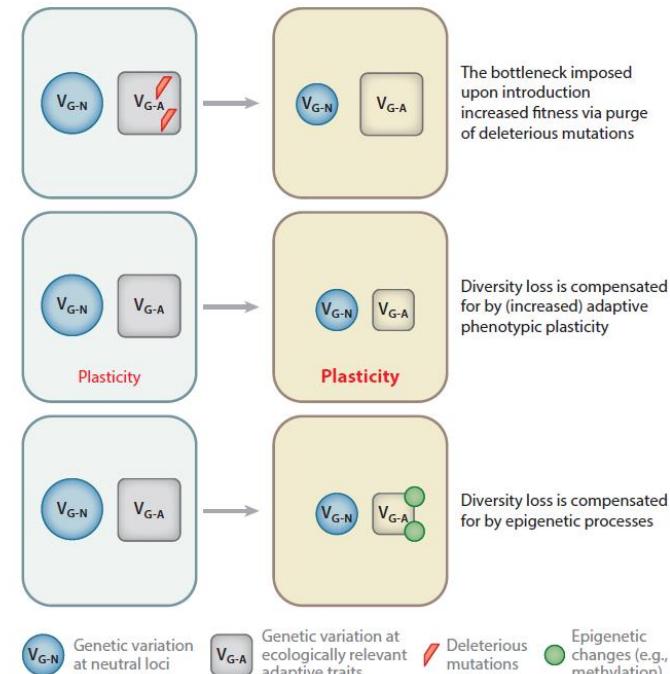
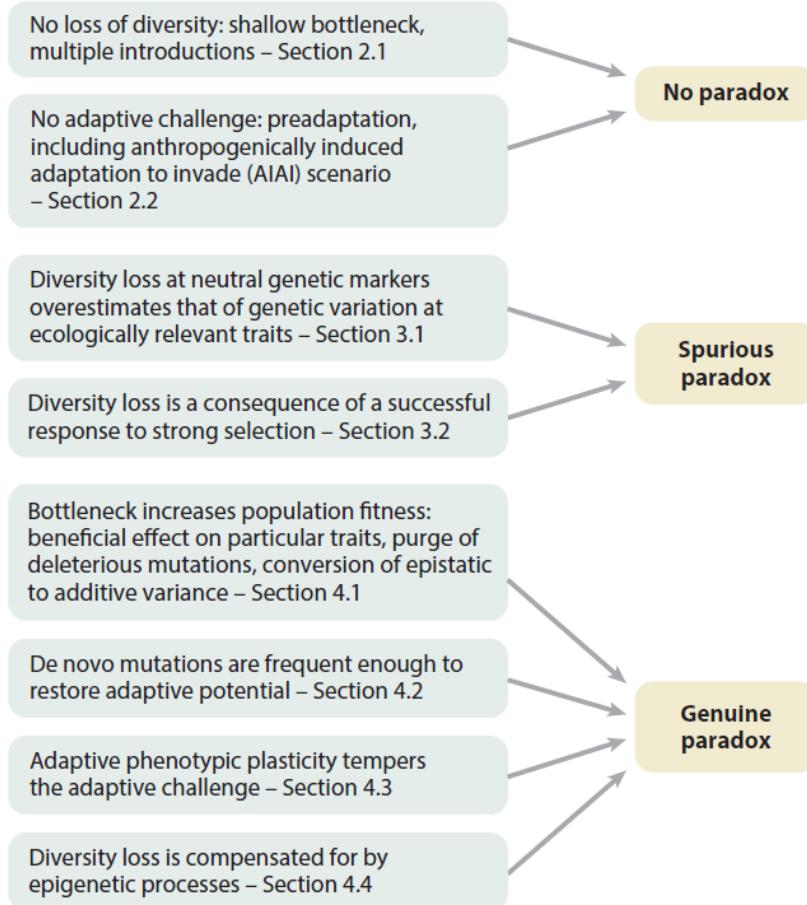
Enough standing genetic variation for evolution



Alleles selected for
by myxoma virus

Decrease in
Myxoma virulence

Invasive species success- a genetic paradox?



Estoup et al. 2016 Annual Reviews

Decrease in methylation expected in new invasives



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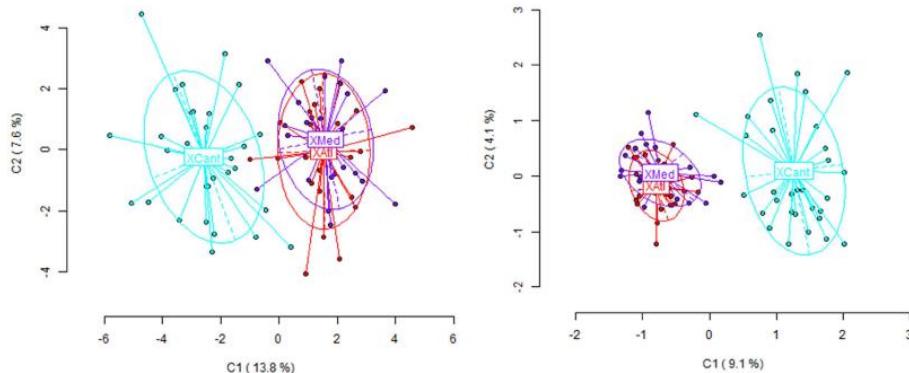
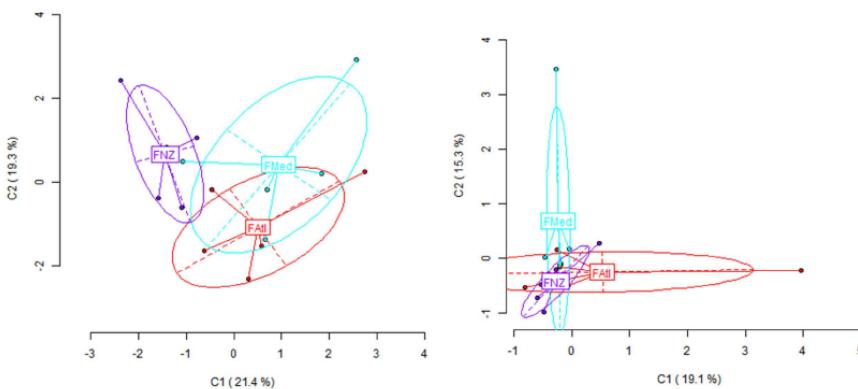


Figure 2. Two-dimensional visualization of the Principal Component Analysis (PCoA) of the detected methylation patterns in *Xenostrobus securis*, with the epigenetic variation (methylation-sensitive loci) on the left and the genetic variation (no methylated loci, NML) on the right. The individuals of each population are represented by the acronyms XAtl, XCant and XMed for the Atlantic international port, Cantabric international port and Mediterranean lagoon populations respectively.

New



old



Figure 4. Two-dimensional visualization of the Principal Component Analysis (PCoA) of the detected methylation patterns in *Ficopomatus enigmaticus*, with the epigenetic variation (methylation-sensitive loci) on the left and the genetic variation (no methylated loci, NML) on the right. Each population is represented by the acronyms FNZ, FCant and FMed for samples, respectively, from the international Napier port in New Zealand, Cantabric fishing port and Mediterranean lagoon locations.

B

Next Lecture

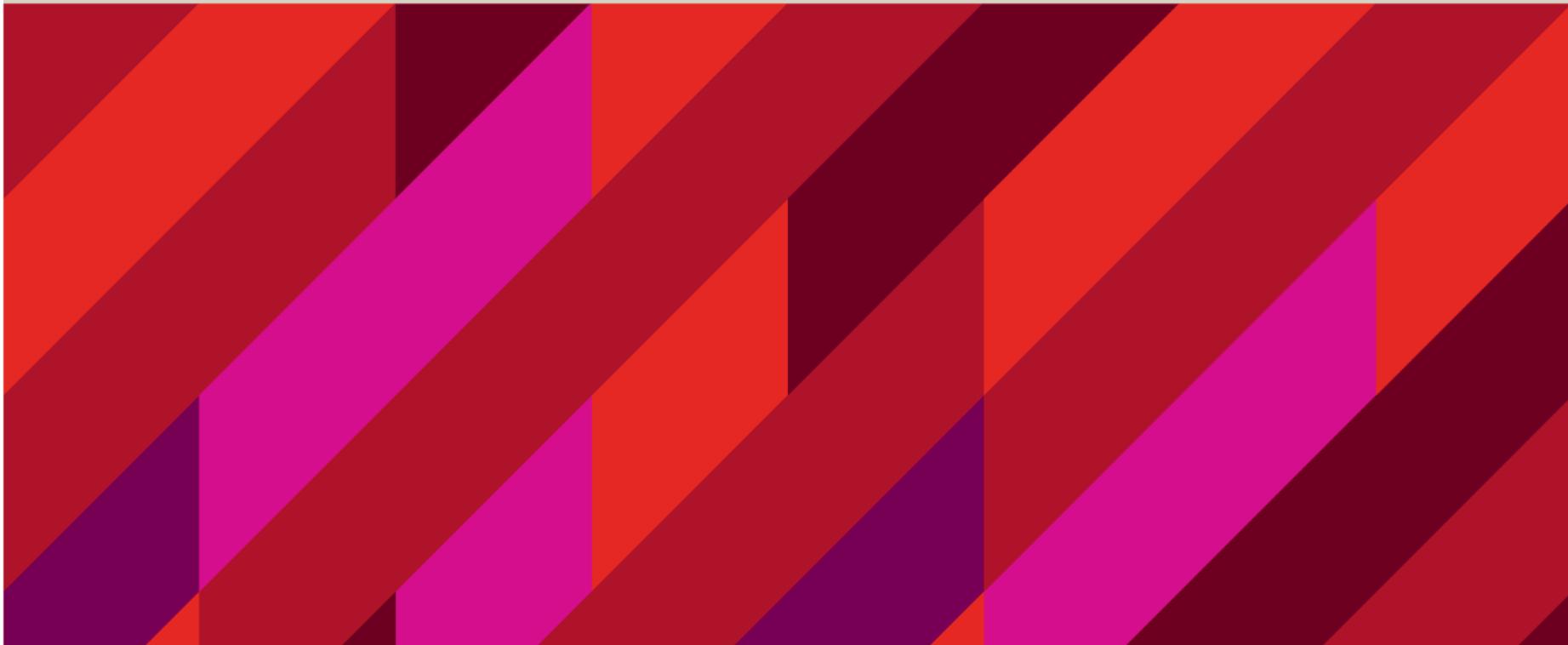
Inbreeding



MACQUARIE
University

Inbreeding

BIOL3110 Conservation and Evolutionary Genetics



Invasion & Population Genetics



MACQUARIE
University

ISLAND ANOLIS LIZARDS



Invasion & Population Genetics

ISLAND ANOLIS LIZARDS



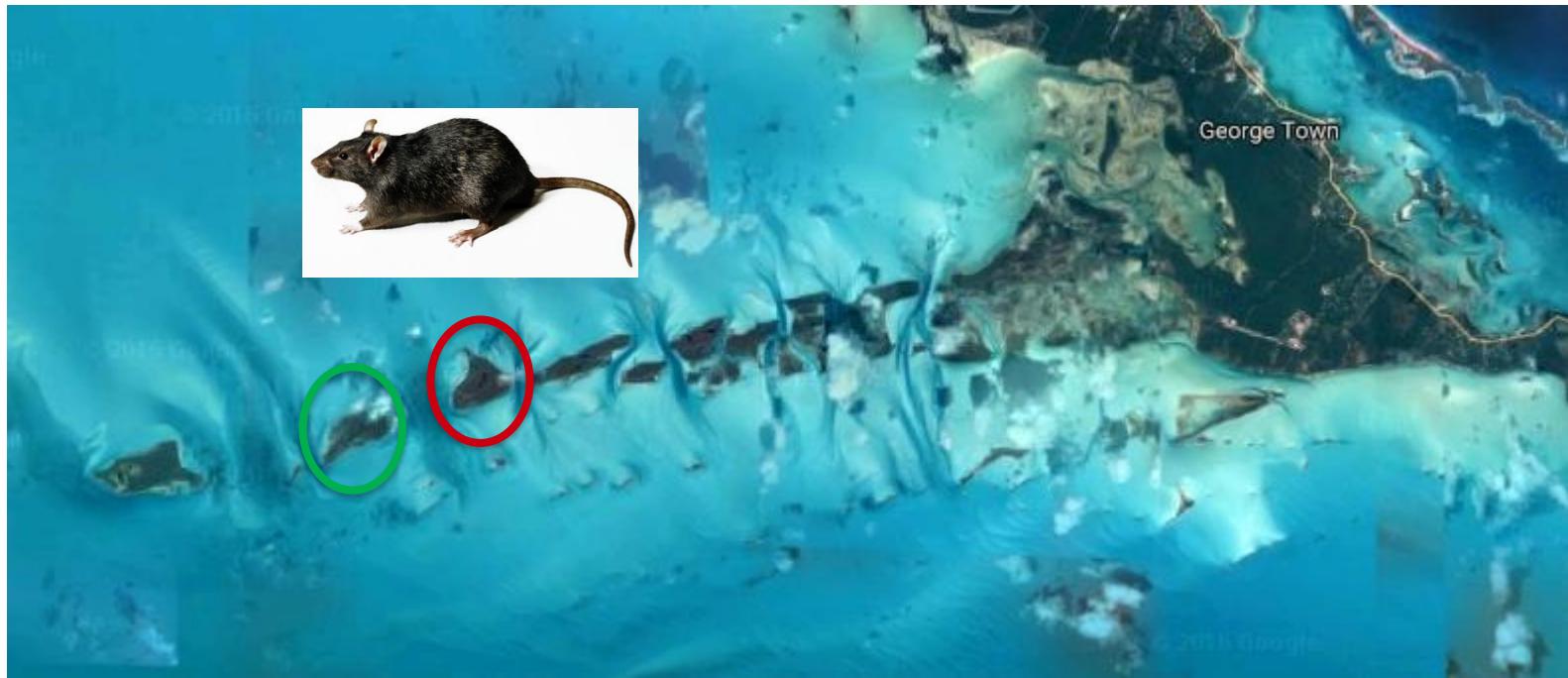
- Bahamas: Great Exuma Island + archipelago

Invasion & Population Genetics



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ISLAND ANOLIS LIZARDS



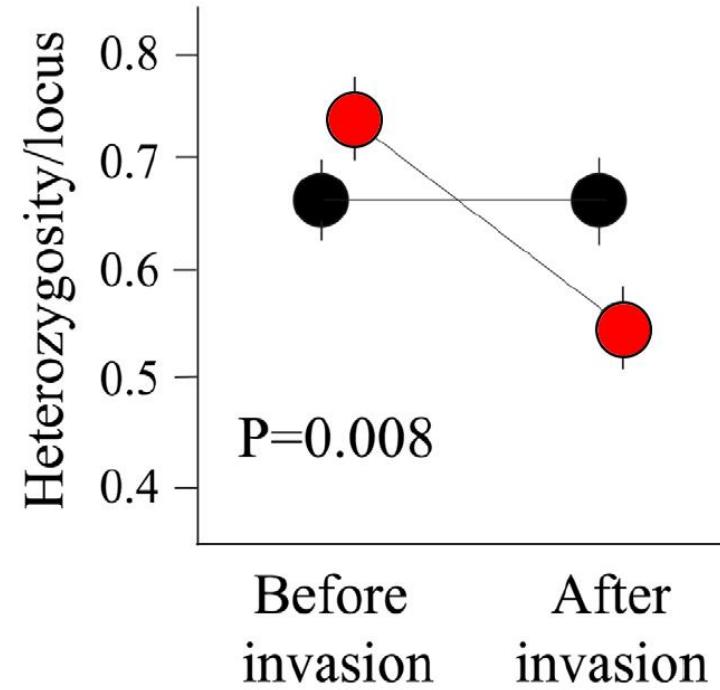
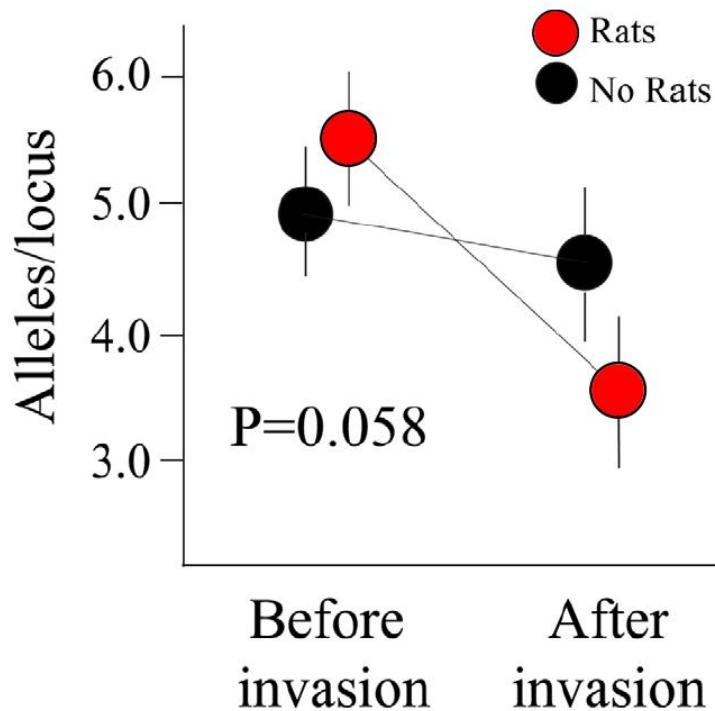
- Rats **infested** 1 island for 12-mo, then exterminated, leaving 13 individuals
- Nearby **non-infested** island served as a control

Invasion & Population Genetics



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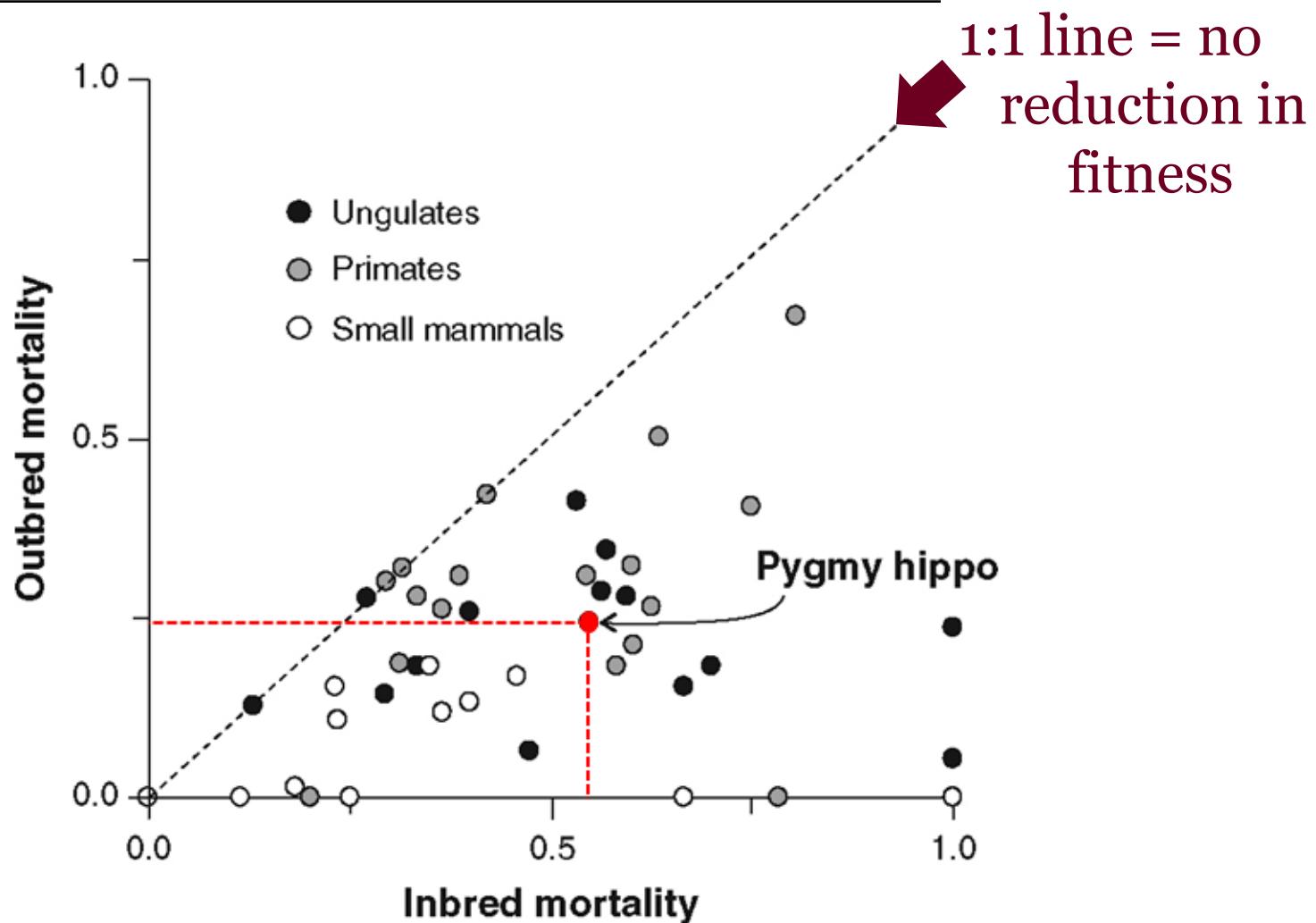
ISLAND ANOLIS LIZARDS



- Significant reductions in V_G post-population “recovery”

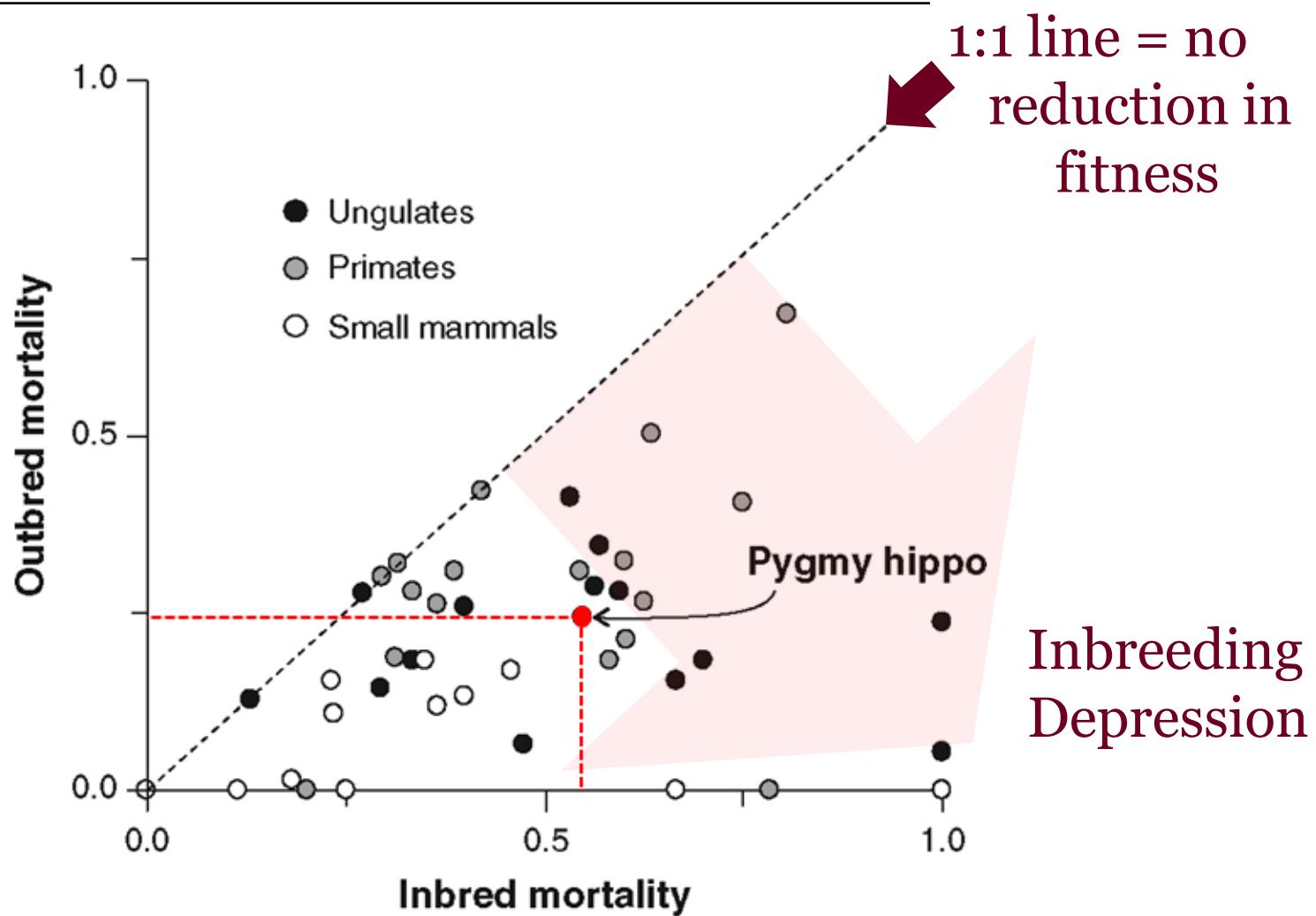
Inbreeding

SPECIES VARIATION IN INBREEDING DEPRESSION



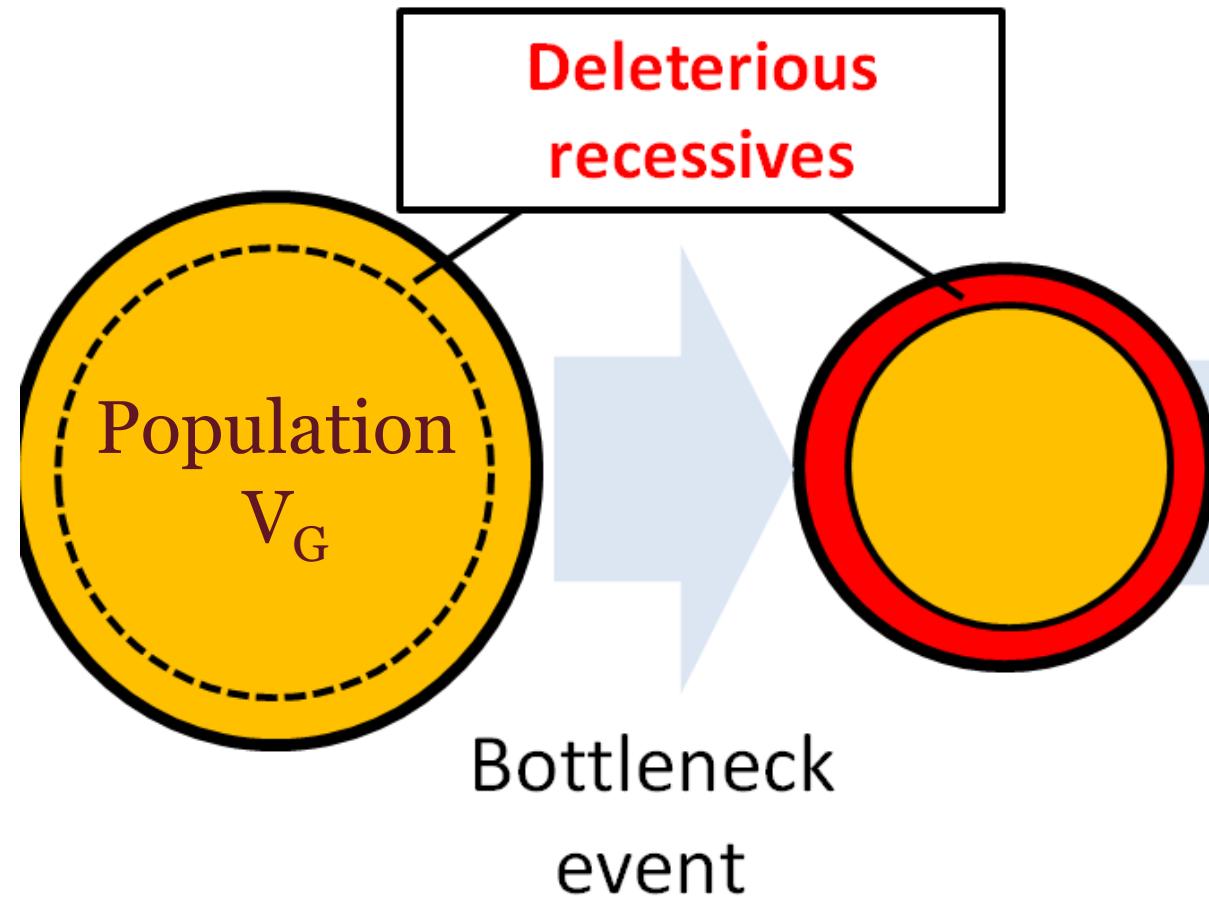
Inbreeding

SPECIES VARIATION IN INBREEDING DEPRESSION



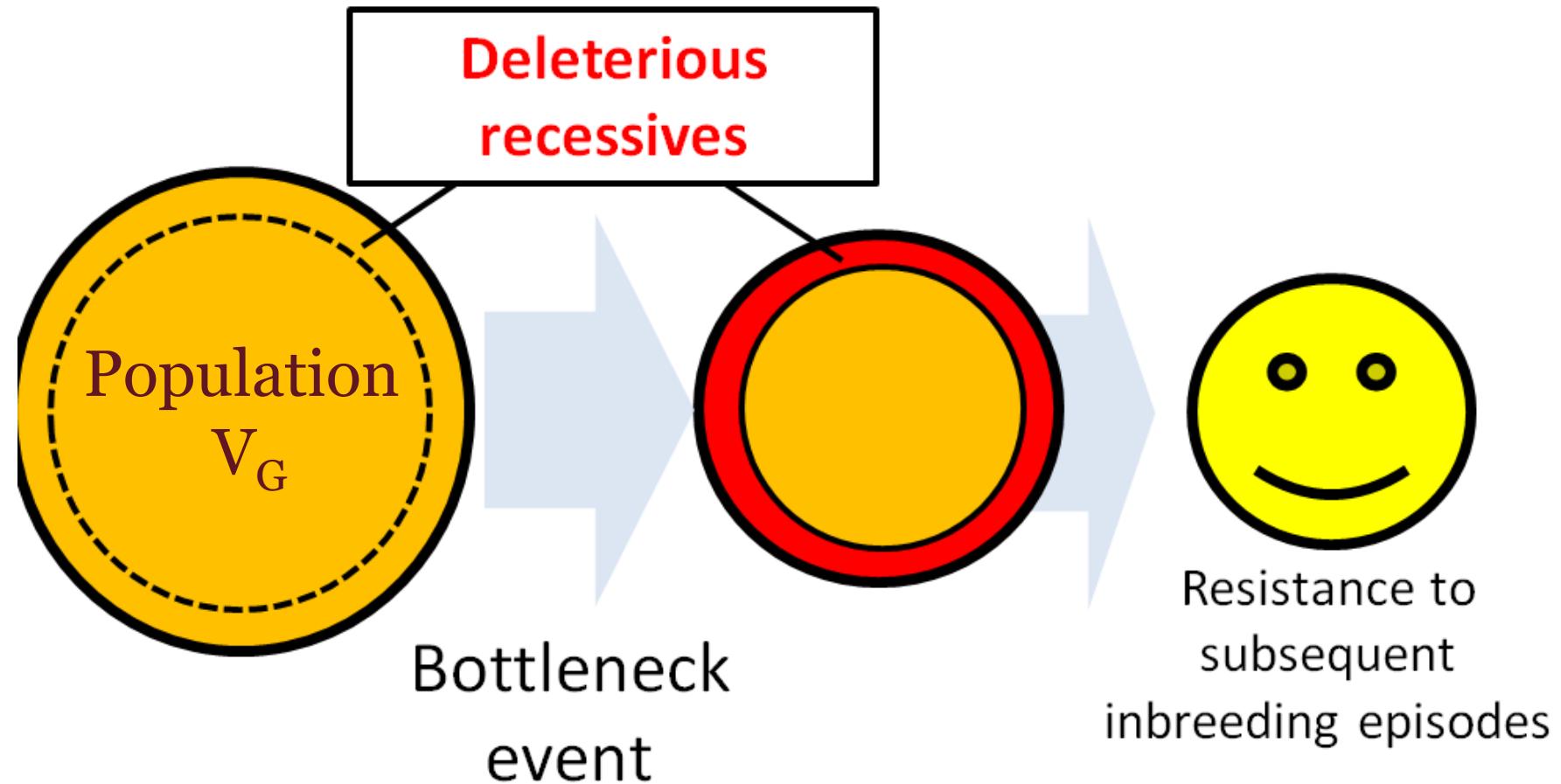
Inbreeding:

Mutational load exposed & purged



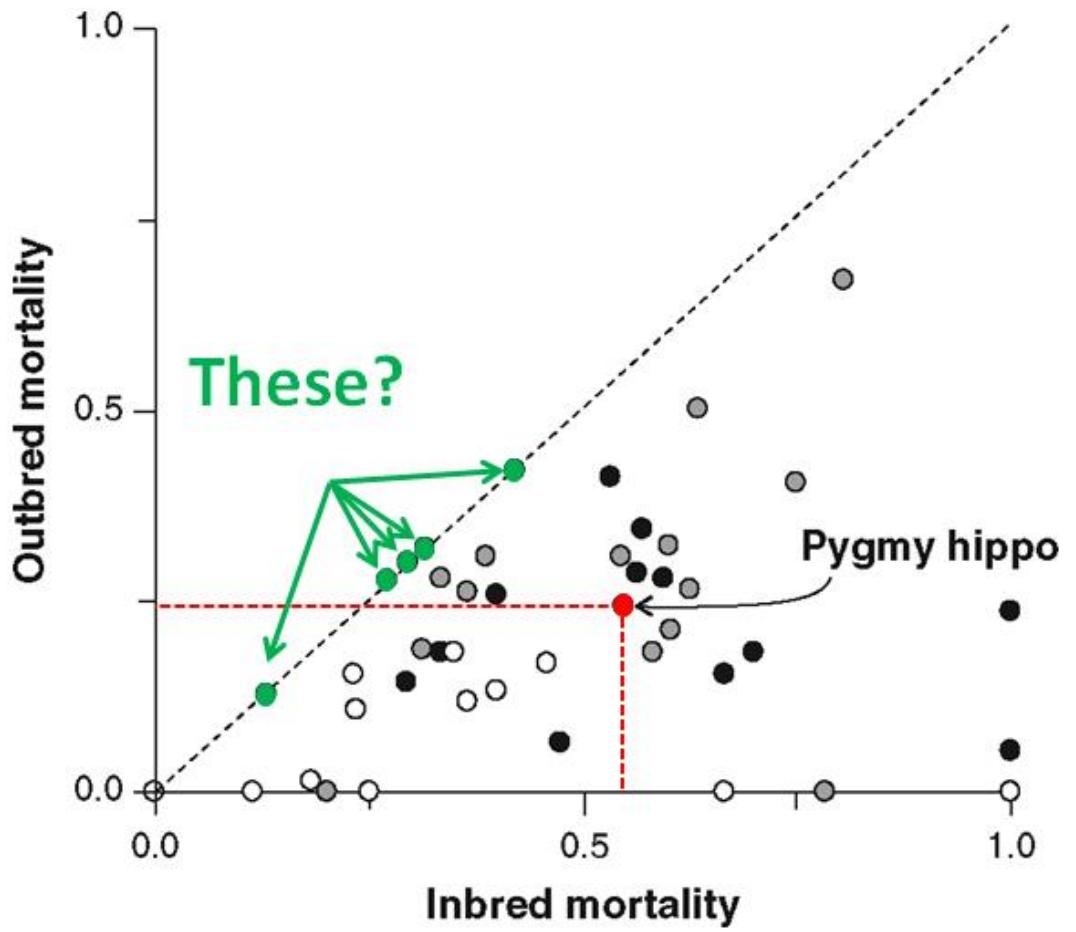
Inbreeding:

Mutational load exposed & purged



Inbreeding:

Mutational load exposed & purged



Inbreeding



Inbreeding & Evolutionary Potential

e.g. *Evolution* 66:2384–2398

ORIGINAL ARTICLE

doi:10.1111/j.1462-2469.2012.03721.x



RESPONSE TO SELECTION ON COLD TOLERANCE IS CONSTRAINED BY INBREEDING

Anneke Dierks,^{1,2} Birgit Baumann,¹ and Klaus Fischer¹

¹Zoological Institute and Museum, University of Greifswald, J.-S.-Bachstraße 11/12, D-17489 Greifswald, Germany

²E-mail: anneke.dierks@uni-greifswald.de

Inbreeding & Evolutionary Potential

ORIGINAL ARTICLE

doi:10.1111/j.1558-5646.2012.01604.x

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No inbreeding
($F = 0.00$)

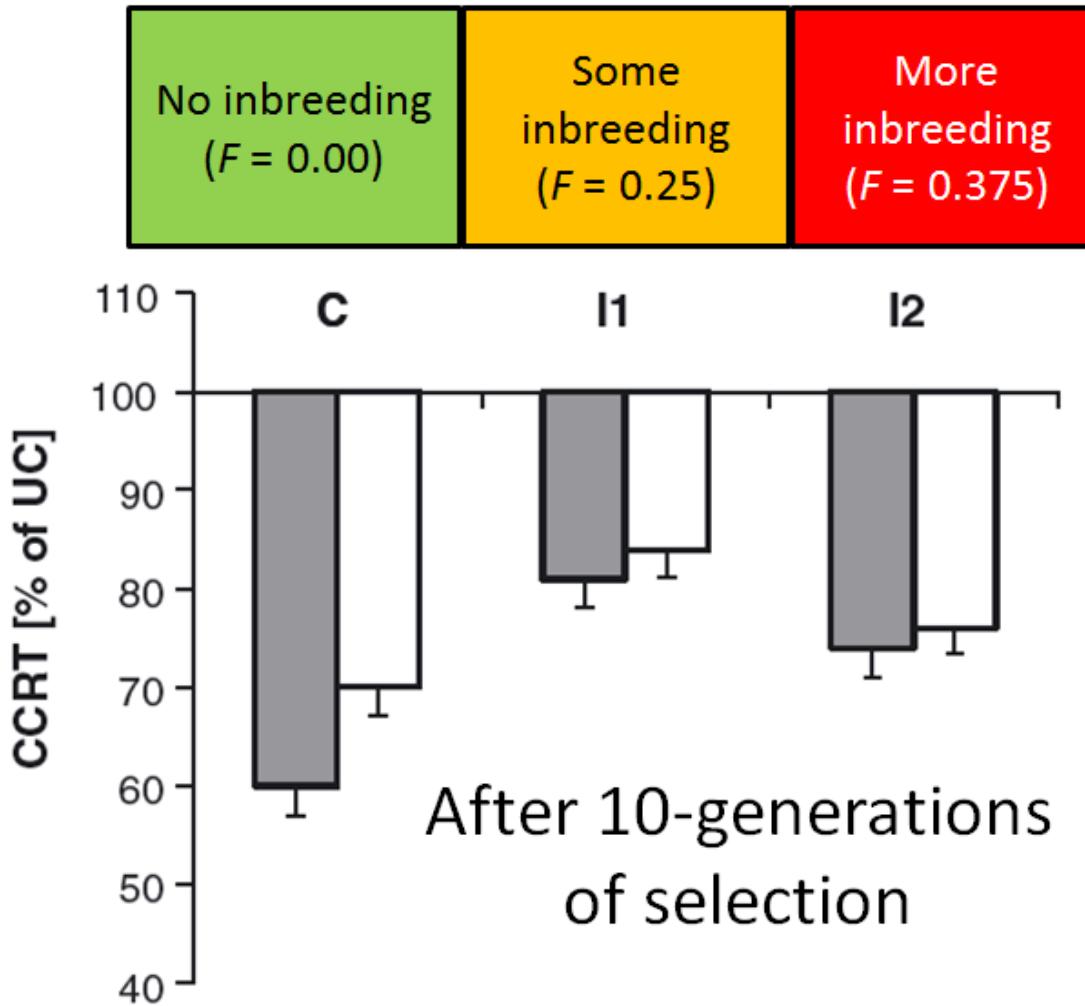
Some inbreeding
($F = 0.25$)

More inbreeding
($F = 0.375$)

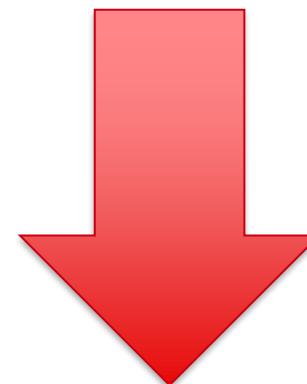
10 Generations of
directional selection for
cold tolerance

Assessed:
- Cold tolerance
Other stuff, such as:
- Heat tolerance
- Fecundity
- Egg hatching
- Develop time
- Adult size
- Growth rate

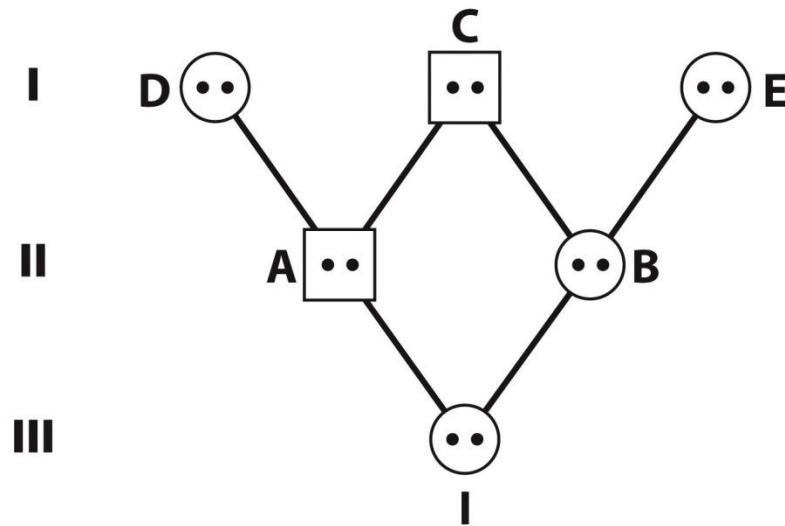
Inbreeding & Evolutionary Potential



Selective response



Genetic Analysis of Inbreeding



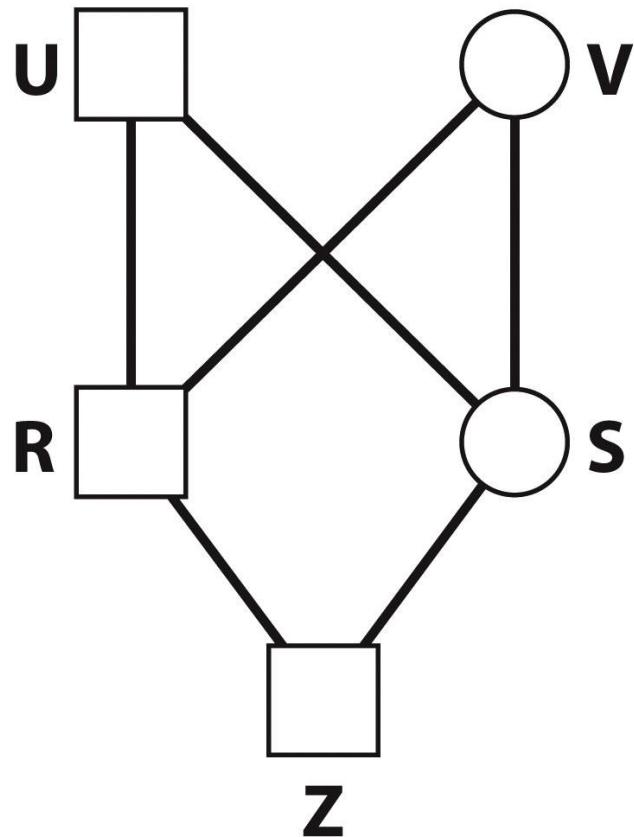
- Individuals A and B are half-siblings.
- Their offspring, I, is inbred, and inherited one copy of her genes from A and one copy from B.
- These copies may be **identical by descent** if they are identical copies inherited from individual C.
- C is the common ancestor of I.

The Inbreeding Coefficient



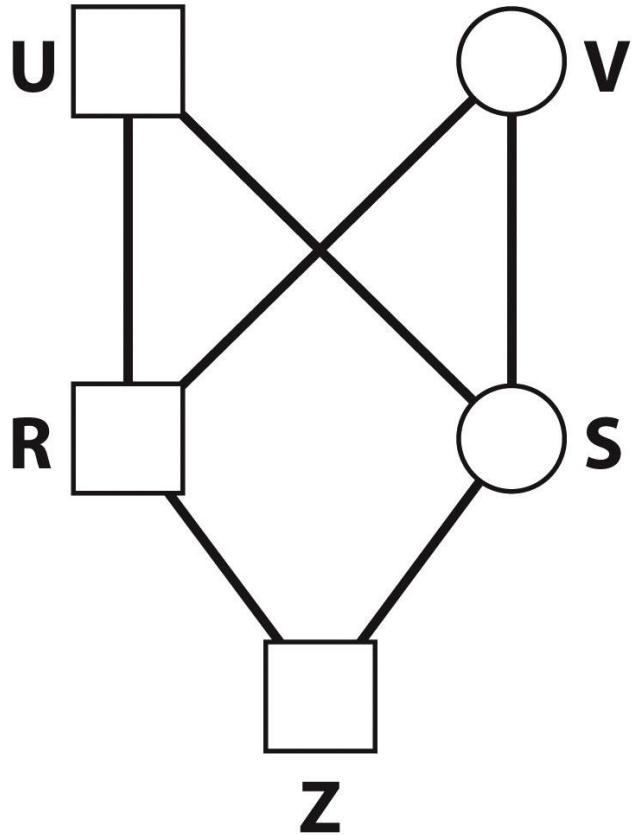
-
- The Inbreeding Coefficient, F , is the probability that the two gene copies in an individual are identical by descent from a common ancestor.
 - Calculation of the Inbreeding Coefficient
 - 1) Identify the common ancestor(s) of an inbred individual.
 - 2) Count the number of linkages between individuals (n) in each inbreeding loop.
 - 3) Calculate the quantity $2 \times (1/2)^n$ for each inbreeding loop and sum the results.

Calculating F for a Full-Sibling Mating



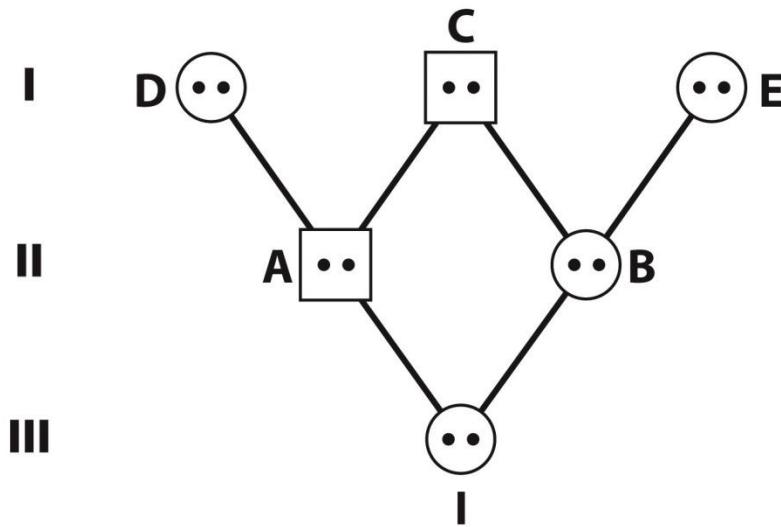
- 1) Identify the common ancestor(s).
 - U and V are both common ancestors, so there are two inbreeding loops.
- 2) Count the number of links between individuals in each inbreeding loop.
 - Loop 1: U, R, S, Z; $n=4$
 - Loop 2: V, R, S, Z $n=4$)

Calculating F for a Full-Sibling Mating



- 3) Each individual is diploid...therefore the probability of **Z** receiving two copies of either allele from a common ancestor is $(1/2)^n \times 2$ for each loop and sum the results.
- Loop 1: $(1/2)^4 \times 2 = 1/8$
 - Loop 2: $(1/2)^4 \times 2 = 1/8$
 - $F = 1/8 + 1/8 = 1/4$

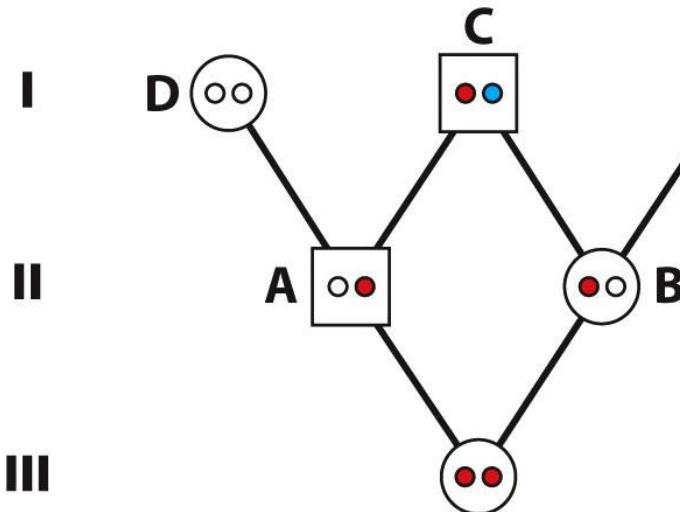
Calculating F for a Half-Sibling Mating



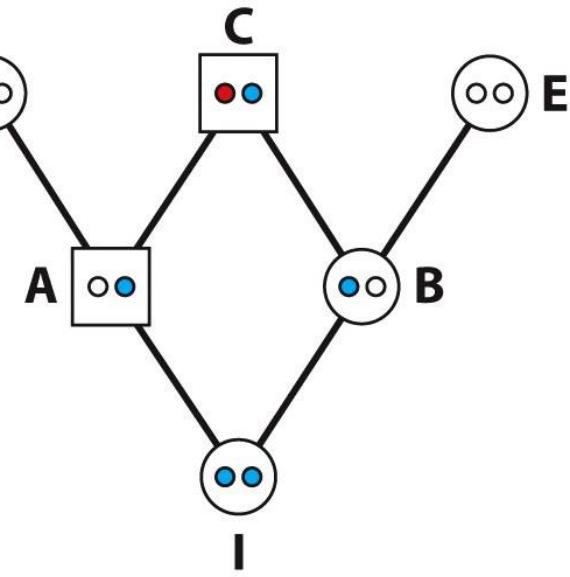
- 1) Identify the common ancestor(s).
 - C is the common ancestor, so there is one inbreeding loop.
- 2) Count the number of linkages between individuals in each inbreeding loop.
 - The Loop includes C, A, and B; $n=4$
- 3) Calculate $(1/2)^n$ for each loop and sum the results.
 - There is only one loop in this case
 - $(1/2)^4 \times 2 = 1/8$, so $F = 1/8$

Generation

Case 1



Case 2



Probability

1/16

+

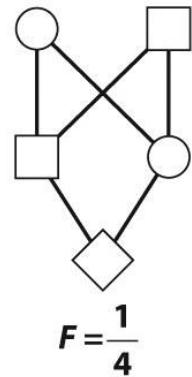
1/16

=

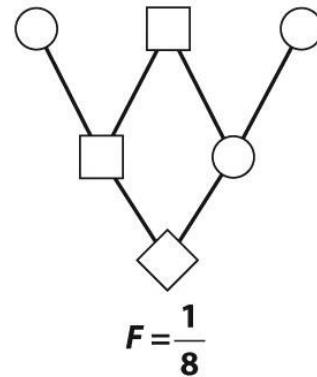
1/8



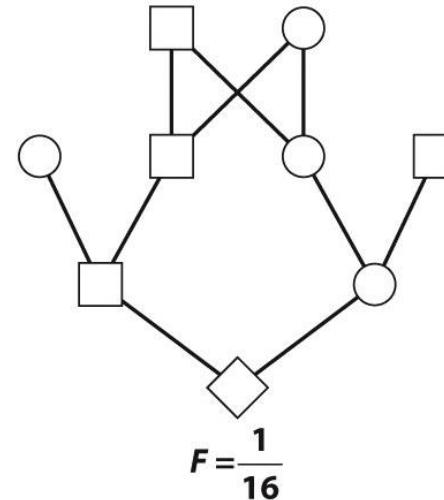
Full siblings



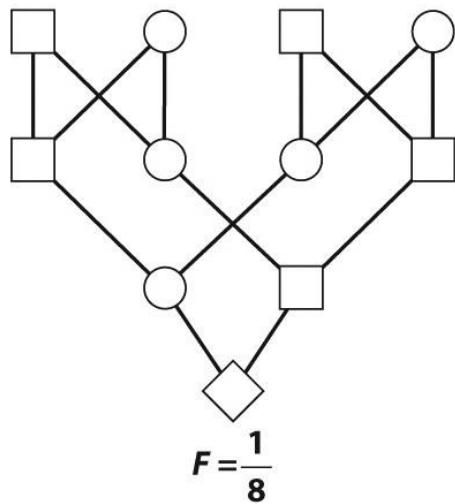
Half siblings



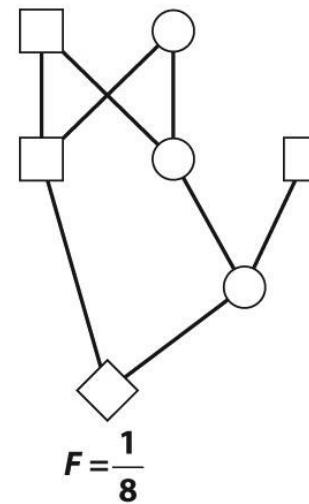
First cousins



Double first cousins

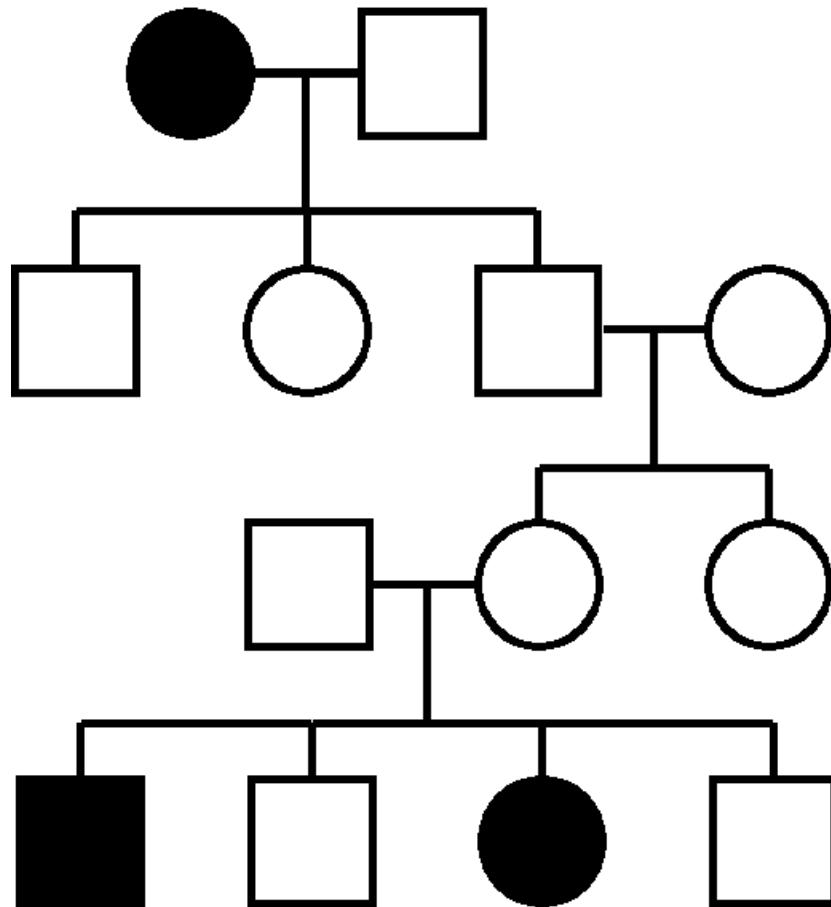


Uncle-niece



Inbreeding

TRACED VIA PEDIGREE DIAGRAMS

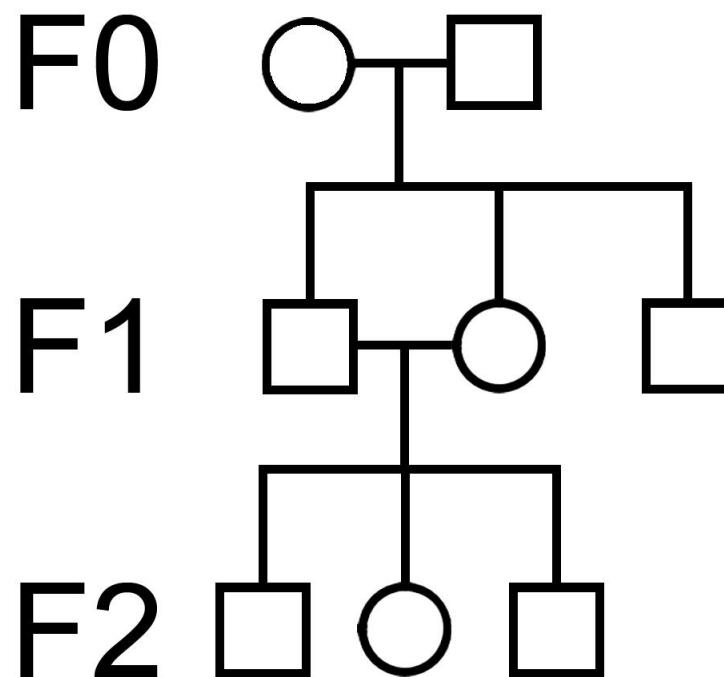


Inbreeding

SIMPLY FROM PEDIGREE DIAGRAMS:

Count the nodes:

$$F = 0.5^n$$

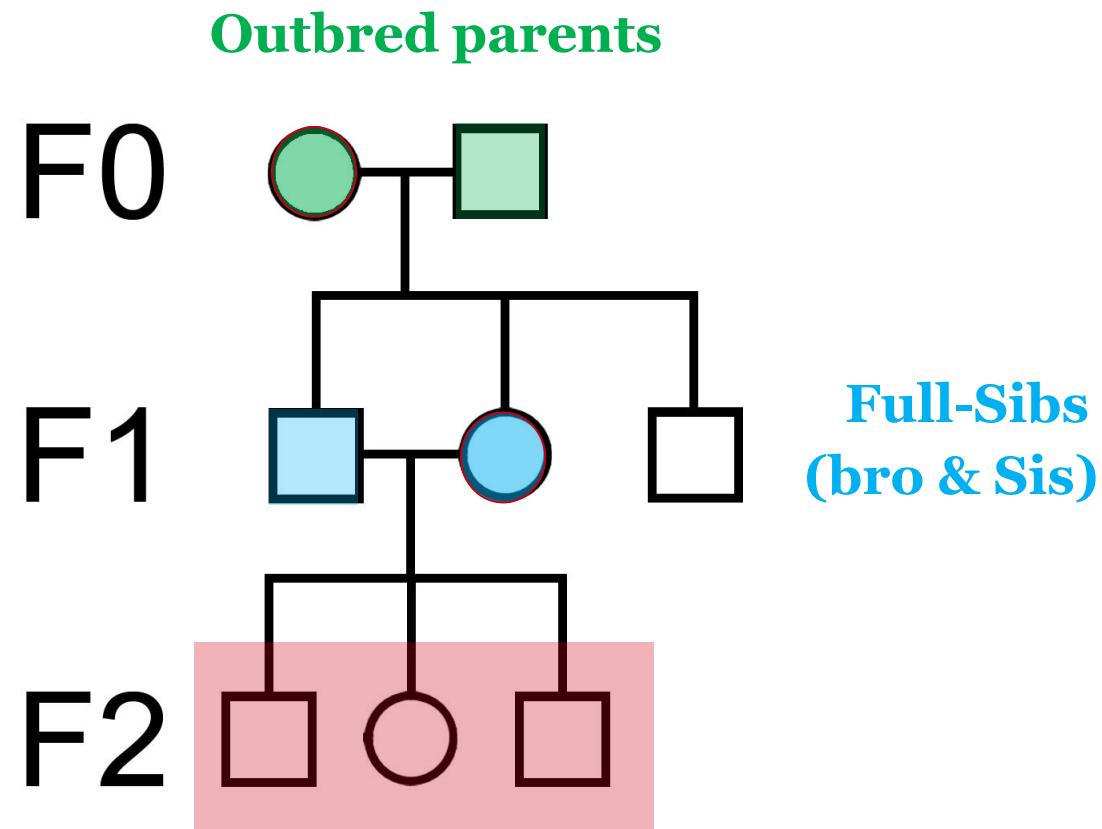


Inbreeding

OFFSPRING OF FULL-SIBLINGS:

Count the nodes:

$$F = 0.5^n$$



Inbreeding

OFFSPRING OF FULL-SIBLINGS:

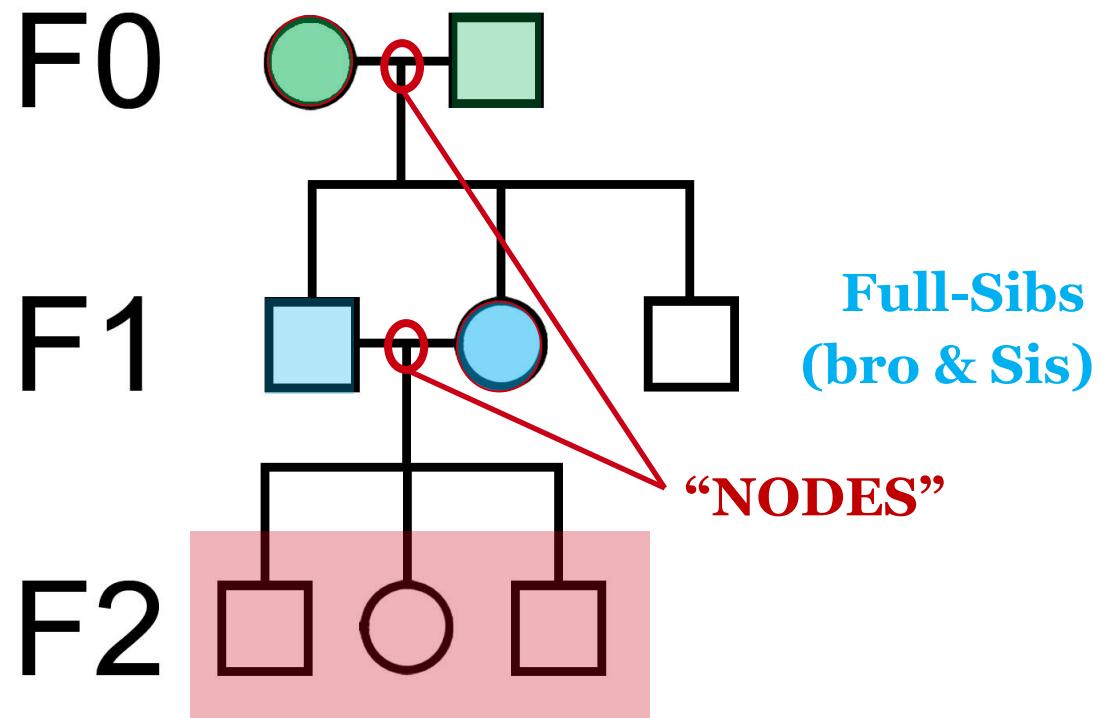
Count the nodes:

$$F = 0.5^n$$

$$F = 0.5^2$$

$$F = 0.25$$

Outbred parents



Inbreeding

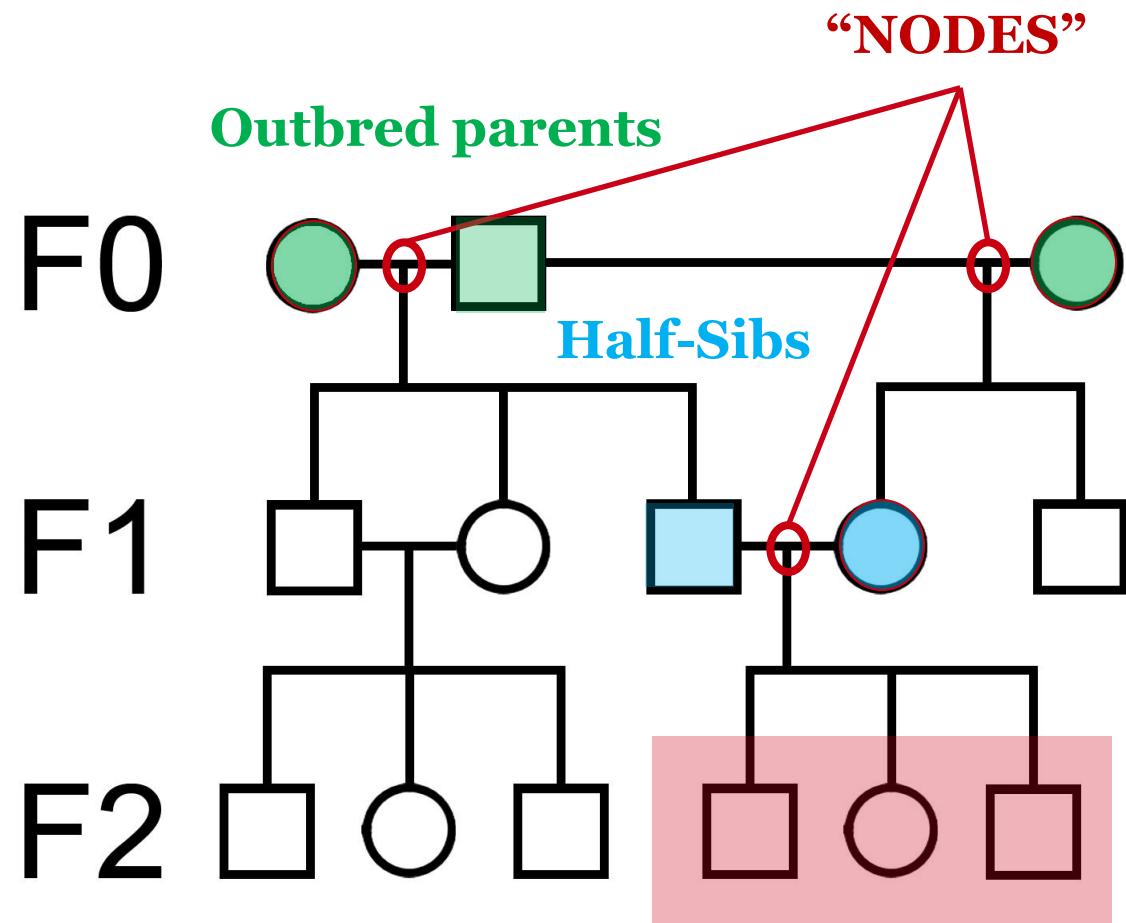
OFFSPRING OF HALF-SIBLINGS:

Count the nodes:

$$F = 0.5^n$$

$$F = 0.5^3$$

$$F = 0.125$$



Inbreeding

RELATIONSHIP TO GENETIC VARIANCE

$$F = 1 - \frac{H_{obs}}{H_{exp}}$$

H-W expected V_G

As H_{obs} nears H_{exp} F nears zero.

As $H_{obs} << H_{exp}$ F nears one.

Inbreeding



RELATIONSHIP TO GENETIC VARIANCE

$$F = 1 - \frac{H_{obs}}{H_{exp}}$$

H-W expected V_G

$$F = 1 - \frac{H_{t=1}}{H_{t=0}}$$

Change in V_G
over time

Inbreeding

RELATIONSHIP TO (DELTA) GENETIC VARIANCE

$$F = 1 - \frac{H_{t=1}}{H_{t=0}}$$

Inbreeding

RELATIONSHIP TO (DELTA) GENETIC VARIANCE

$$F = 1 - \frac{H_{t=1}}{H_{t=0}}$$

$$\frac{H_{1997}}{H_{1974}} = \frac{0.10}{0.23} = 0.43$$



Inbreeding

RELATIONSHIP TO (DELTA) GENETIC VARIANCE

$$F = 1 - \frac{H_{t=1}}{H_{t=0}}$$

$$F = 1 - 0.43$$

$$F = 0.57$$

$$\frac{H_{1997}}{H_{1974}} = \frac{0.10}{0.23} = 0.43$$



Genetic problems are usually eased by restoring genetic variation

‘genetic rescue’ / ‘gene pool mixing’ works

Nearly every one of 156 cases (Frankham 2015)



Applications of Genetic Tools and Approaches in Conservation Biology and Management

CASE STUDY
Augmenting
genetic
variation
reverses
a
population
decline

Madsen *et al.* (1999)
Nature



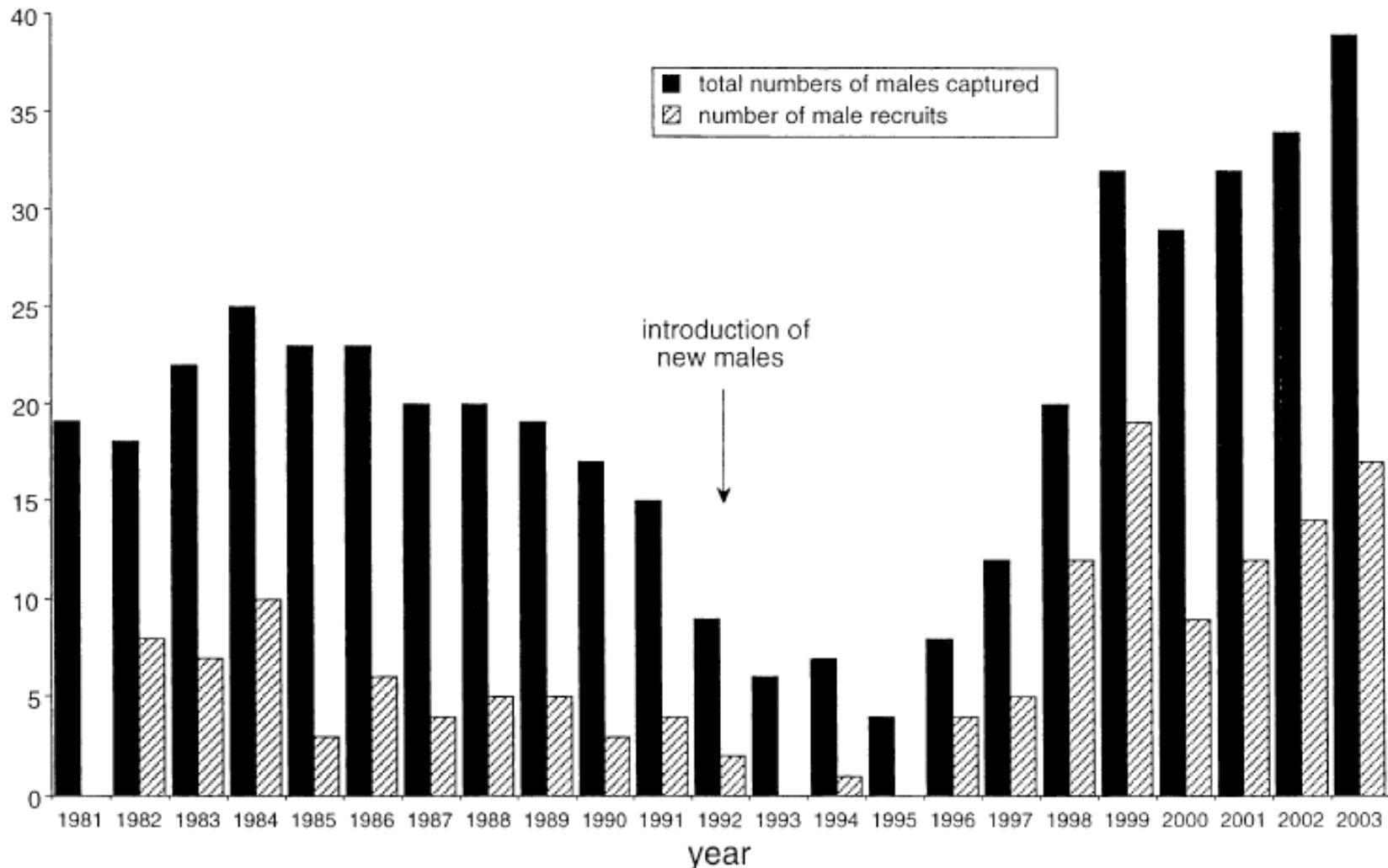
- population of adders ($N_e < 15$)
- isolated for 100 years
- pop. decline over 35 years
- high proportion of stillborn & deformed young
- low genetic variation

- added 20 males from diverse nearby population
- watched population change & re-examined genetic variation

- migrant males settled in & mated, removed after 4 breeding seasons
- from first breeding season, stillbirths declined, population began dramatic growth...

NB: numbers exclude ones added

T. Madsen et al. / Biological Conservation 120 (2004) 145–147



Many other high profile successes



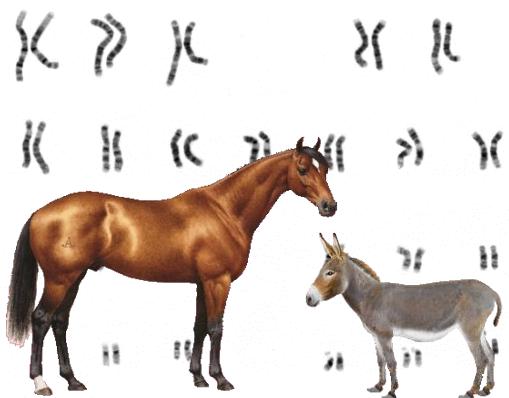
But still, genetic rescue has rarely been used

Fear of outbreeding depression, but it's

- Rare
- Usually mild / transient
- Predictable and avoidable

Three ‘red flags’ for outbreeding depression

Major
chromosomal
differences



Long isolation



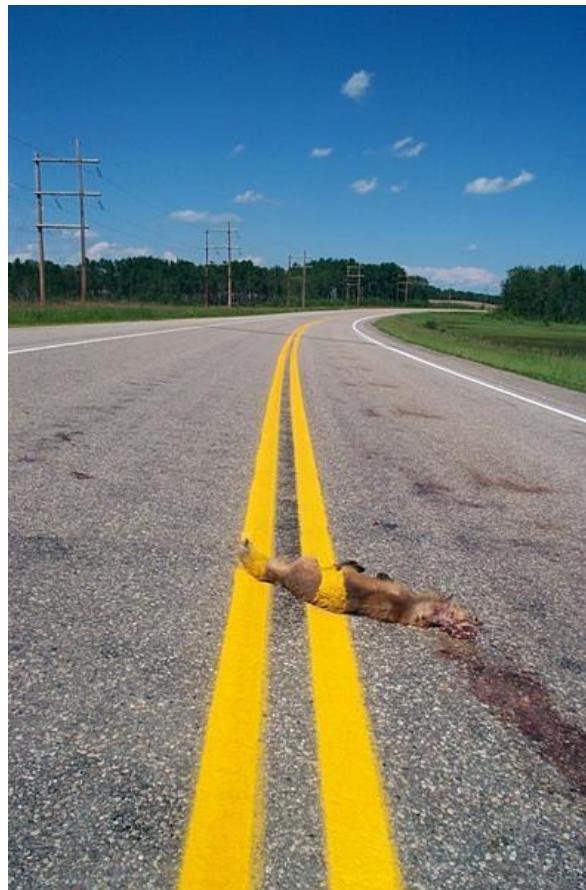
Adaptive
differentiation



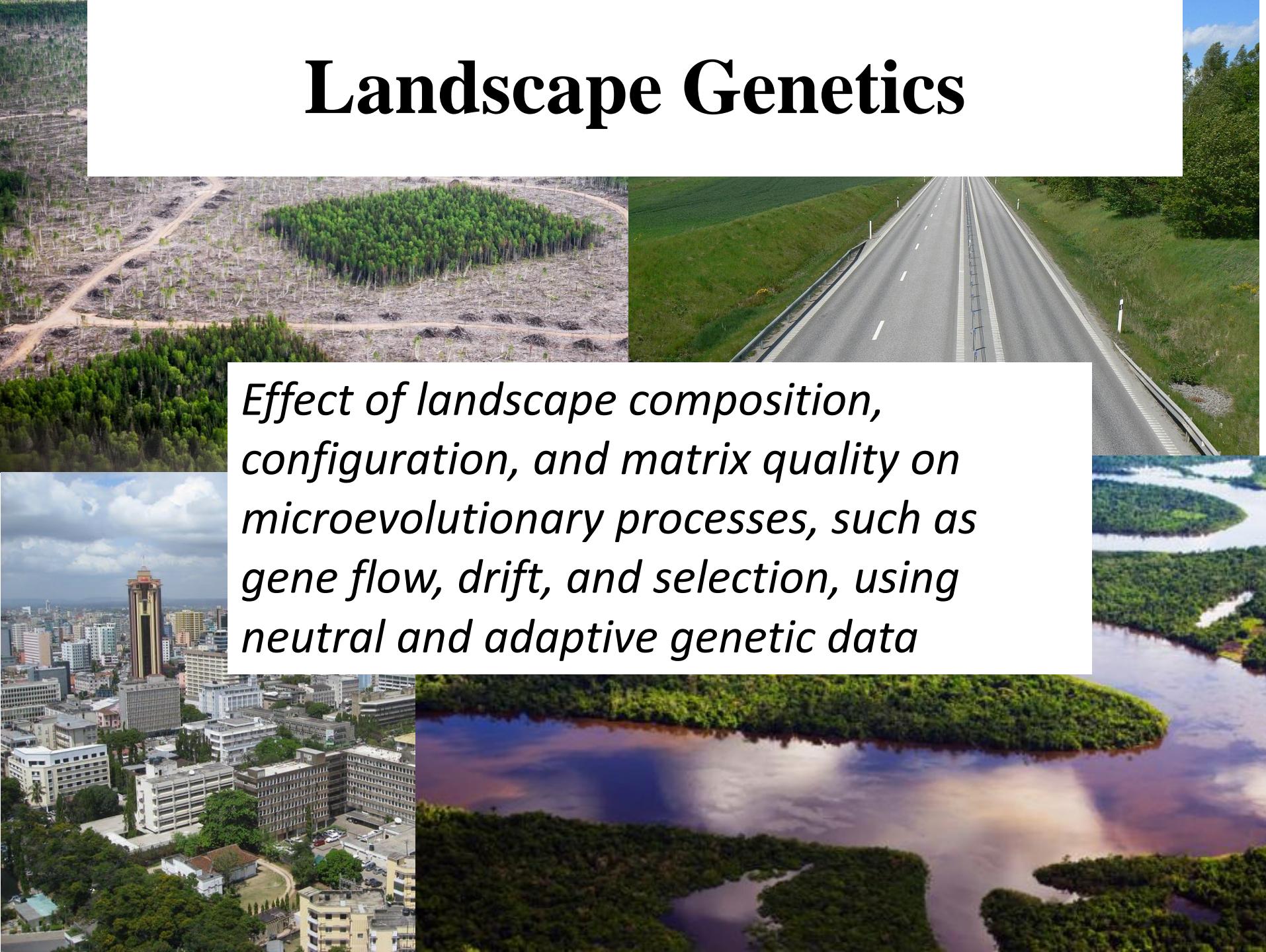
Next Lecture: Genetic consequences of Habitat Fragmentation



Habitat Fragmentation



Landscape Genetics



*Effect of landscape composition,
configuration, and matrix quality on
microevolutionary processes, such as
gene flow, drift, and selection, using
neutral and adaptive genetic data*

Connectivity/ Landscape Genetics

- Genetic connectivity
- Demographic connectivity

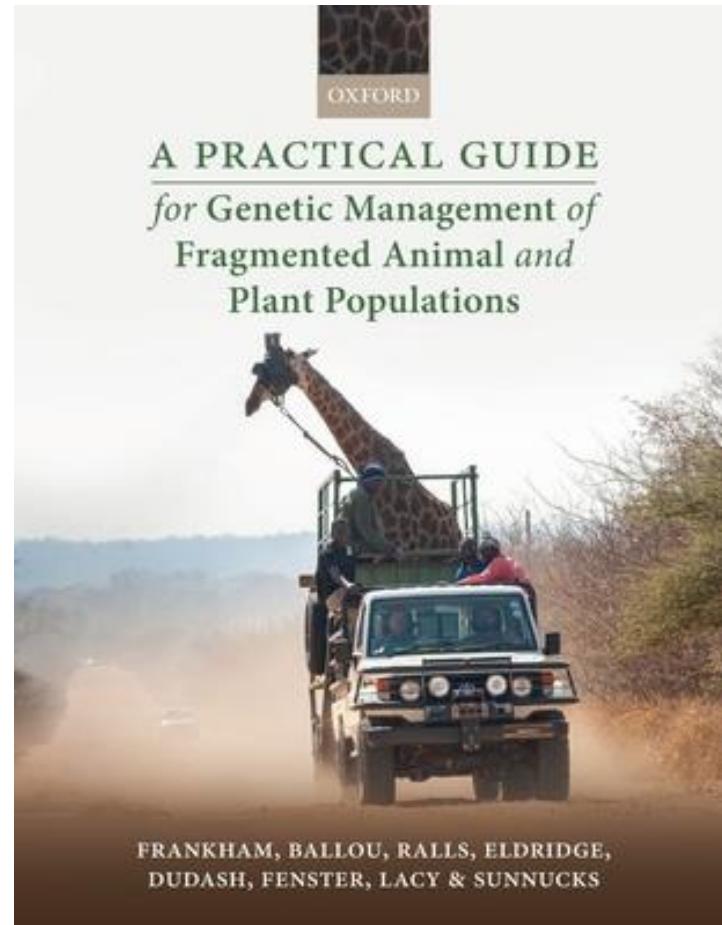
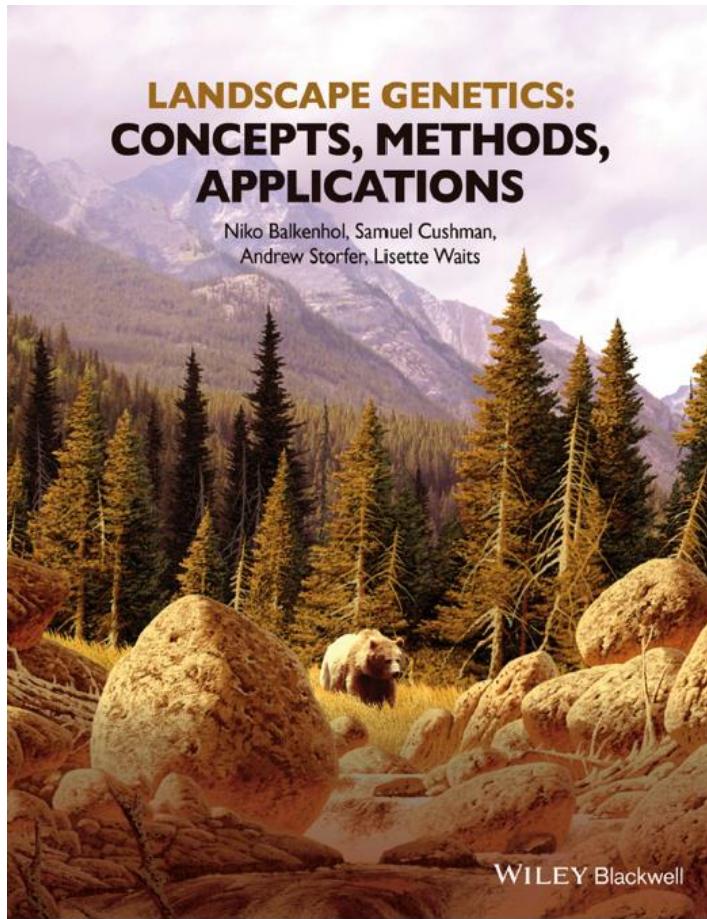
Key themes:

1. Infer underlying ecological processes, such as dispersal or disease transmission.
2. Understanding how landscape characteristics affect evolutionary processes

Connectivity - definitions

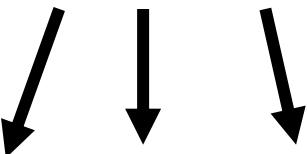
1. Genetic connectivity – Degree to which gene flow affects evolutionary processes
2. Demographic connectivity – Degree to which individual movements influence demography.

Useful References



Genetic variation and a hierarchy of biodiversity

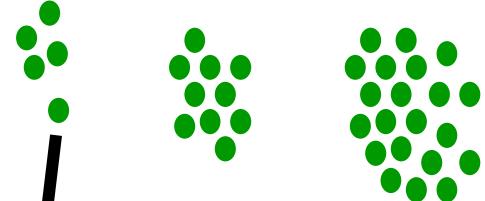
Species:



'Populations'
(habitat fragments)



Individuals:



Genes:



Research themes

Biogeography / speciation
(species and above)



genetic structure
/phylogeography



Paternity / relatedness
(individual / social groups)

Different analyses address different time scales

- 1/ *Genotypic analyses* are based on DNA ‘signatures’ of individual organisms that are reshuffled in every generation of a sexual species
- 2/ *Genic analyses*, where the focus is on frequencies of genetic variants in grouped samples, not individuals. Gene frequencies change more slowly than the generational shuffling of genotypes – measures ongoing restriction of mobility and gene flow
- 3/ *Genealogical analyses*, A new variant arises at a point in time and space, and will come to be distributed over landscapes according to gene flow, drift and extinction – slow process and therefore can detect processes far back in time

Analyzing groups of individuals

- Within Patches
 - Levels of genetic variation
 - Level of inbreeding
 - Effective population size (N_e)
- Between Patches
 - Degree of genetic differentiation
 - Measures of gene flow and/or dispersal

F-statistics

Inbreeding and drift both decrease heterozygosity and increase homozygosity. But:

- inbreeding impacts at the individual level
- drift impacts at the group level (population)

Total population variation can be partitioned into differentiation among individuals within populations and differences among populations

F-statistics measure level of heterozygosity relative to HWE expectations at different hierarchical levels:

F_{IS} : within patches

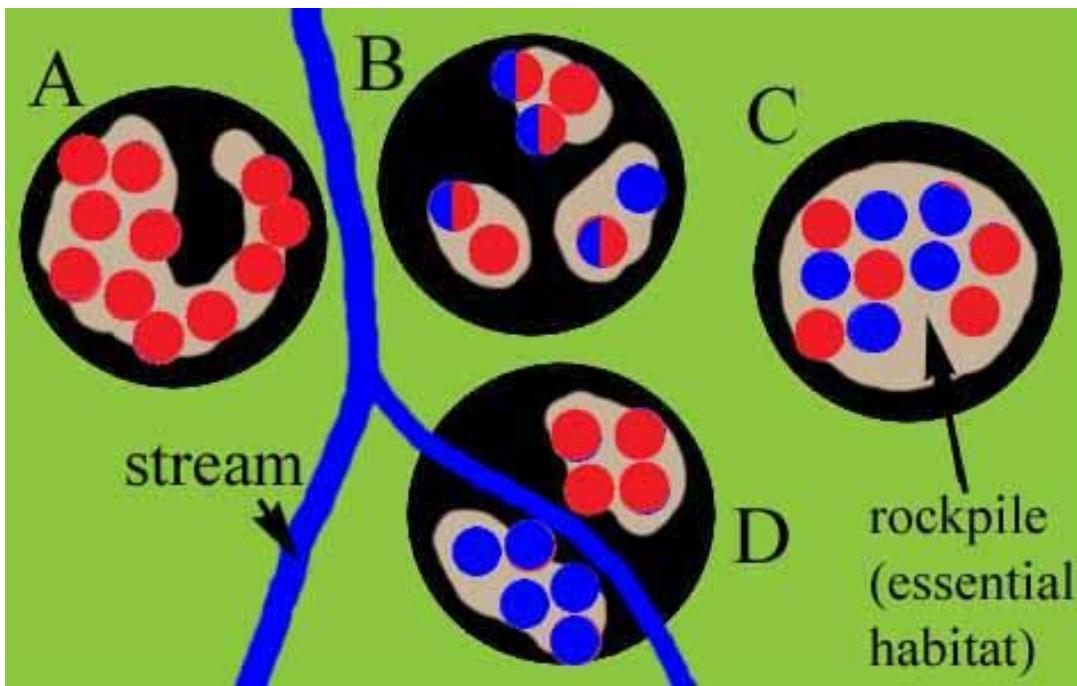
F_{ST} : among patches

F_{IS} is a measure of inbreeding ‘within a patch’

- $1 - H_o$ (within pop)/ H_e (all samples)
- ranges from -1 to +1
- positive: excess of homozygotes
- negative: excess of heterozygotes
- At HWE F_{IS} is 0

F_{ST} is a measure of genetic subdivision

- 1- $\text{He (of fragment)}/\text{He (Total)}$
- ranges from -1 to +1 
- measures the proportional reduction in heterozygosity of the entire network of subpopulations relative to HWE (due to differentiation among subpopulations)
- large: at least some subpopulations have different allele frequencies
- small or 0: subpopulations have similar allele frequencies

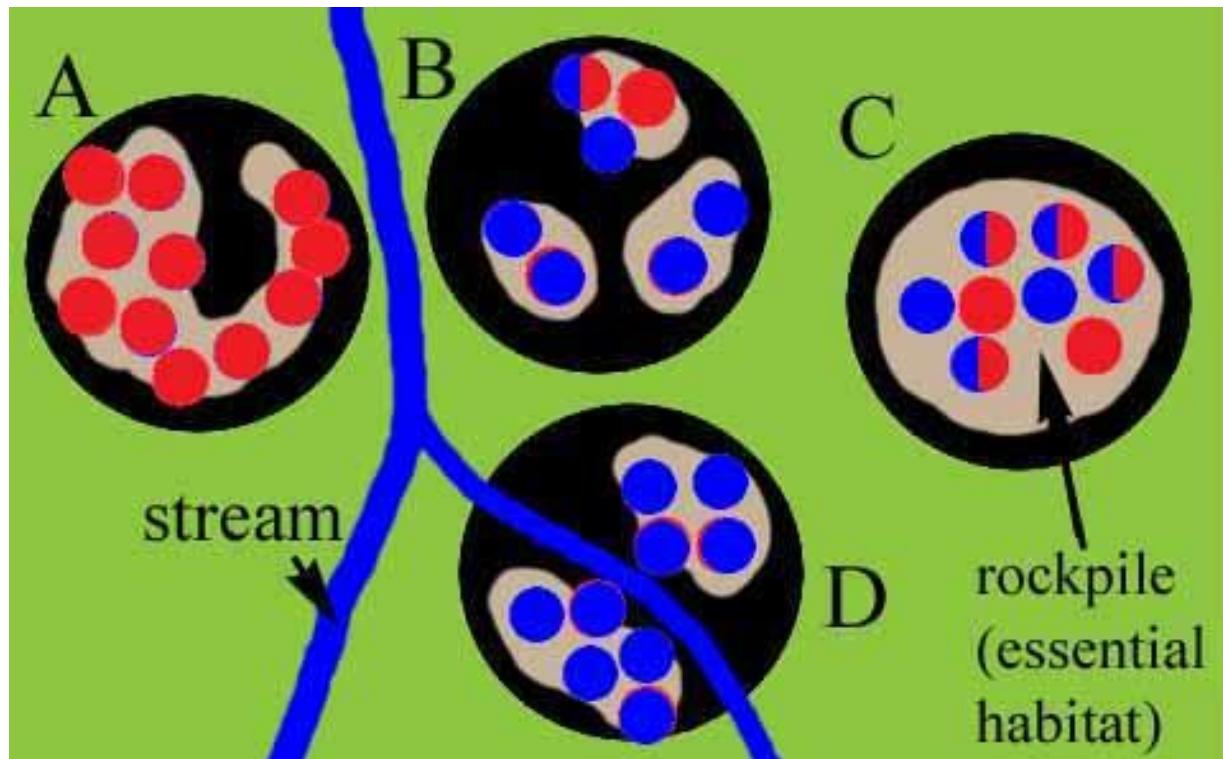


F_{ST} contributions of the subpops A, B, C, D to this would be...

- A. Large - subpop A has very different allele freqs. compared to the others
- B. Little - subpop B has similar allele freqs. to C & D
- C. Little - same reasons as B despite inbreeding in C
- D. Little - same reasons as B despite two inbred groups in D

F_{ST} and F_{IS} can operate separately...

Scenario 1



F_{IS}

low, because each subpop
is not far from HWE.

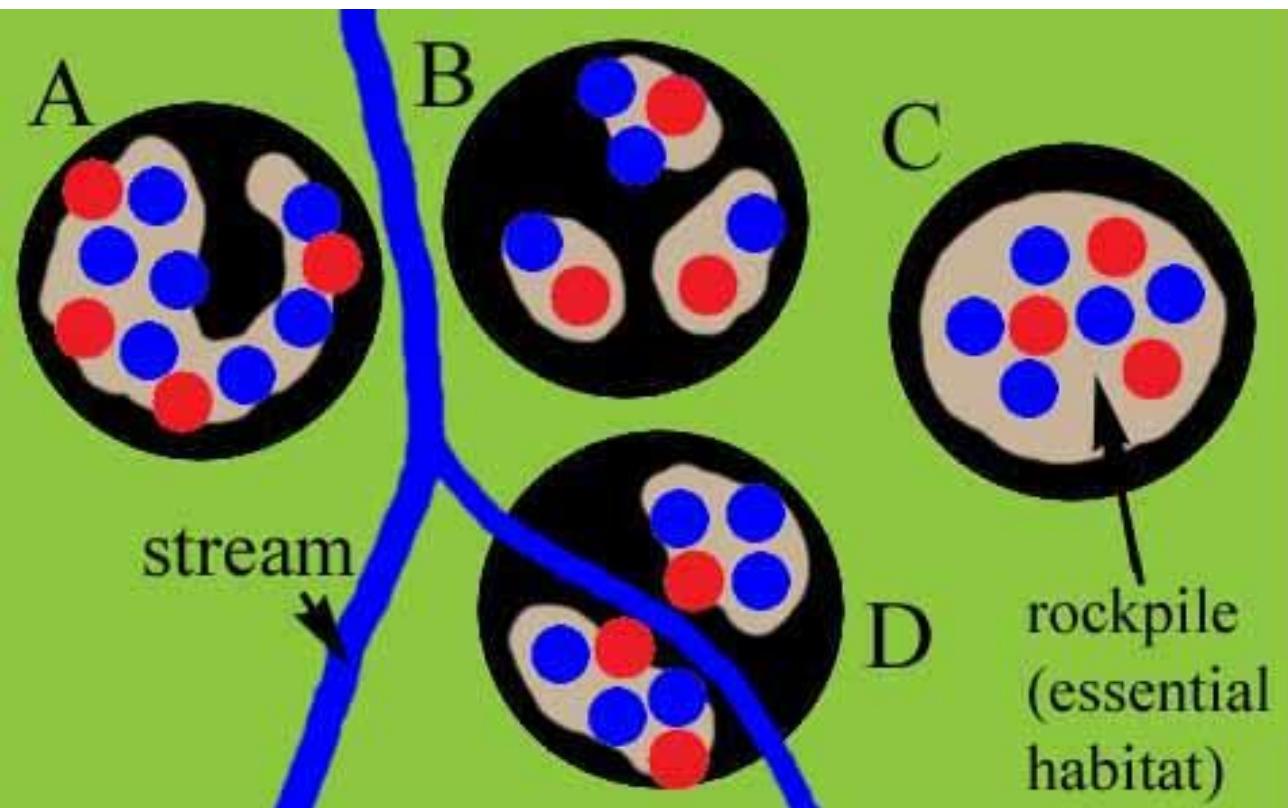
F_{ST}

is high, because the
allele freqs. in each
subop are very different:

- A is fixed for red
- B has high freq blue
- C has equal freqs
- D is fixed for blue

F_{ST} and F_{IS} can operate separately...

Scenario 2



F_{IS}

very high, because the reds and blues appear not to interbreed - there are no heterozygotes even though reds and blues live together

F_{ST}

very low, because there are few differences among the subpops A, B, C, D. All have reds and blues

The importance of the spatial scale of sampling ...



“*Wahlund Effect*”
reduction of heterozygosity in a
population caused by genetic
structure

F_{IS}

Contribution to F_{ST}

Sampling scale

D as one unit

large +(ve)

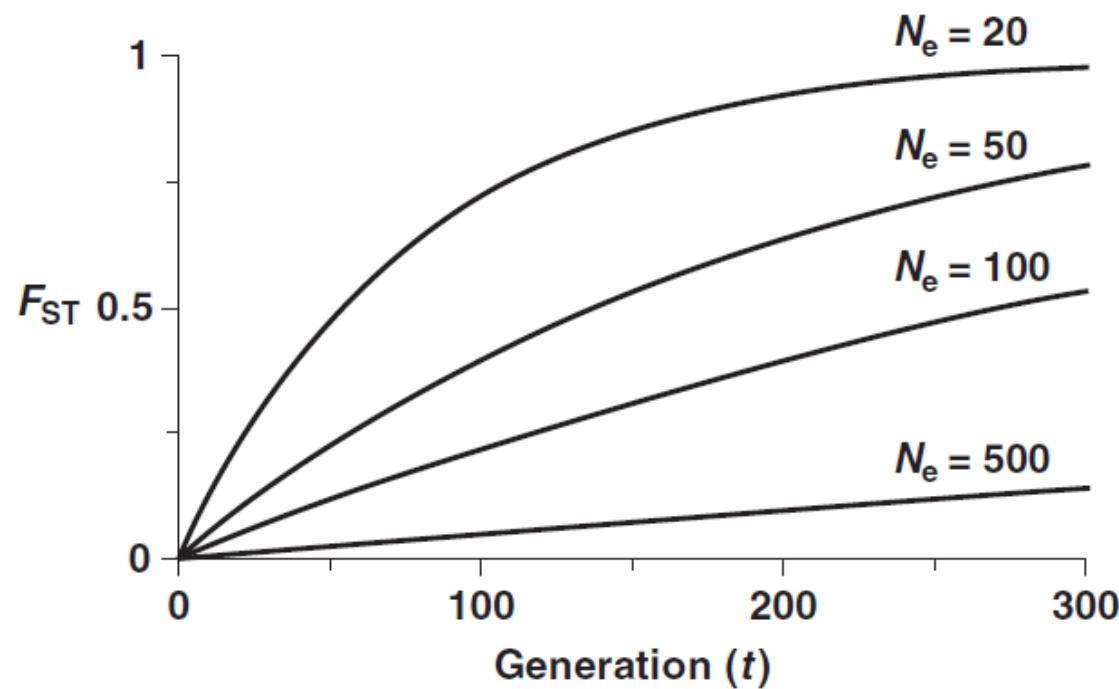
very little

D split into 2

~ 0

very large

Relationship between F_{ST} and N_e



Interpreting F-statistics

Generalizations about the biological meaning of F_{ST}

Table 2 Definitions for three types of genetic connectivity. The values for m , Nm , and F_{ST} are the approximate values associated with each type of connectivity with the island model of migration. The actual values for each parameter will differ with other models of population structure

	Adaptive connectivity	Inbreeding connectivity	Drift connectivity
Criterion	Sufficient gene flow to spread advantageous alleles	Sufficient gene flow to avoid harmful effects of local inbreeding	Sufficient gene flow to maintain similar allele frequencies
m	?	?	?
Nm	>0.1	>1.0	>10
F_{ST}	<0.35	<0.20	<0.02

Lowe and Allendorf 2010

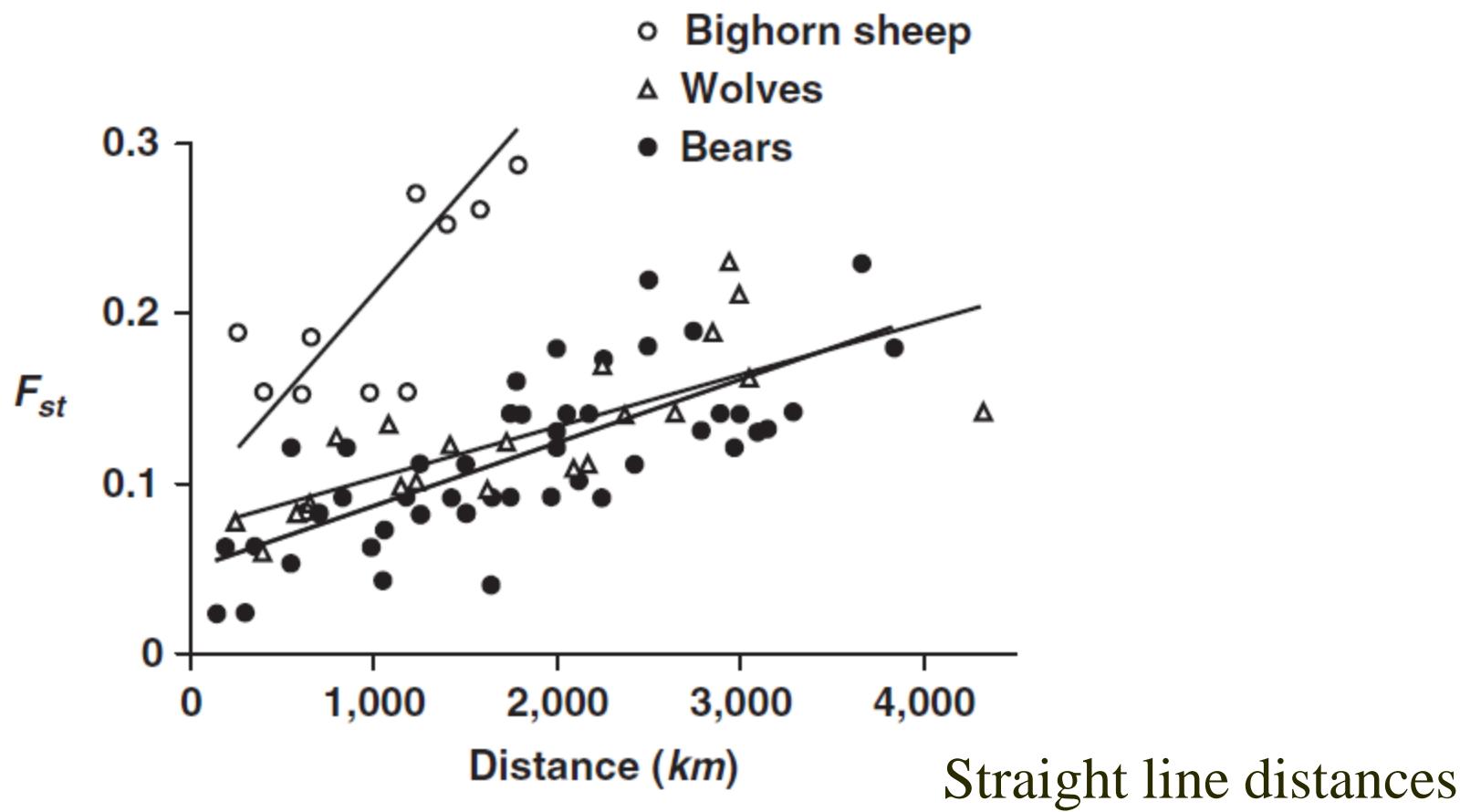


Fig. 14.12 Isolation by distance. Relationship between degree of genetic differentiation (F_{ST}) at microsatellite loci and geographic distance among bighorn sheep, brown bear and gray wolf populations in North America (after Forbes & Hogg 1999). F_{ST} increases with distance in all three species.

Measuring landscape effects on gene flow

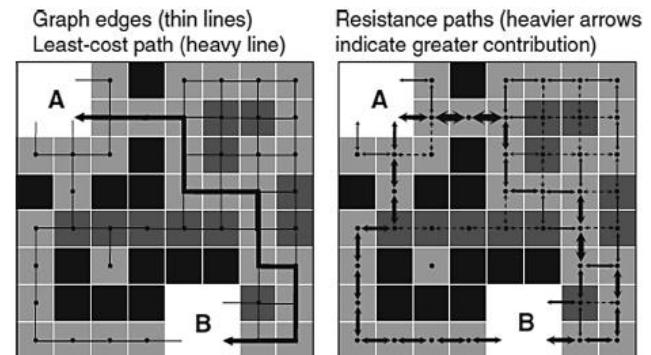
Can be characterised as:

Least Cost Path: A distance measure of the path of ‘least resistance’ i.e. the easiest

Resistance: degrees to which the environment impedes movement – accumulates with geographic distance

Balkenhol et al. landscape Genetics 2016

Continued week 10



Demographic Connectivity

$$N_{t+1} = N_t + \text{Births} - \text{Deaths} + \text{Immigrants} - \text{Emigrants}$$

Local recruitment Net immigration
Total recruitment

Measuring dispersal at the scales required can be problematic



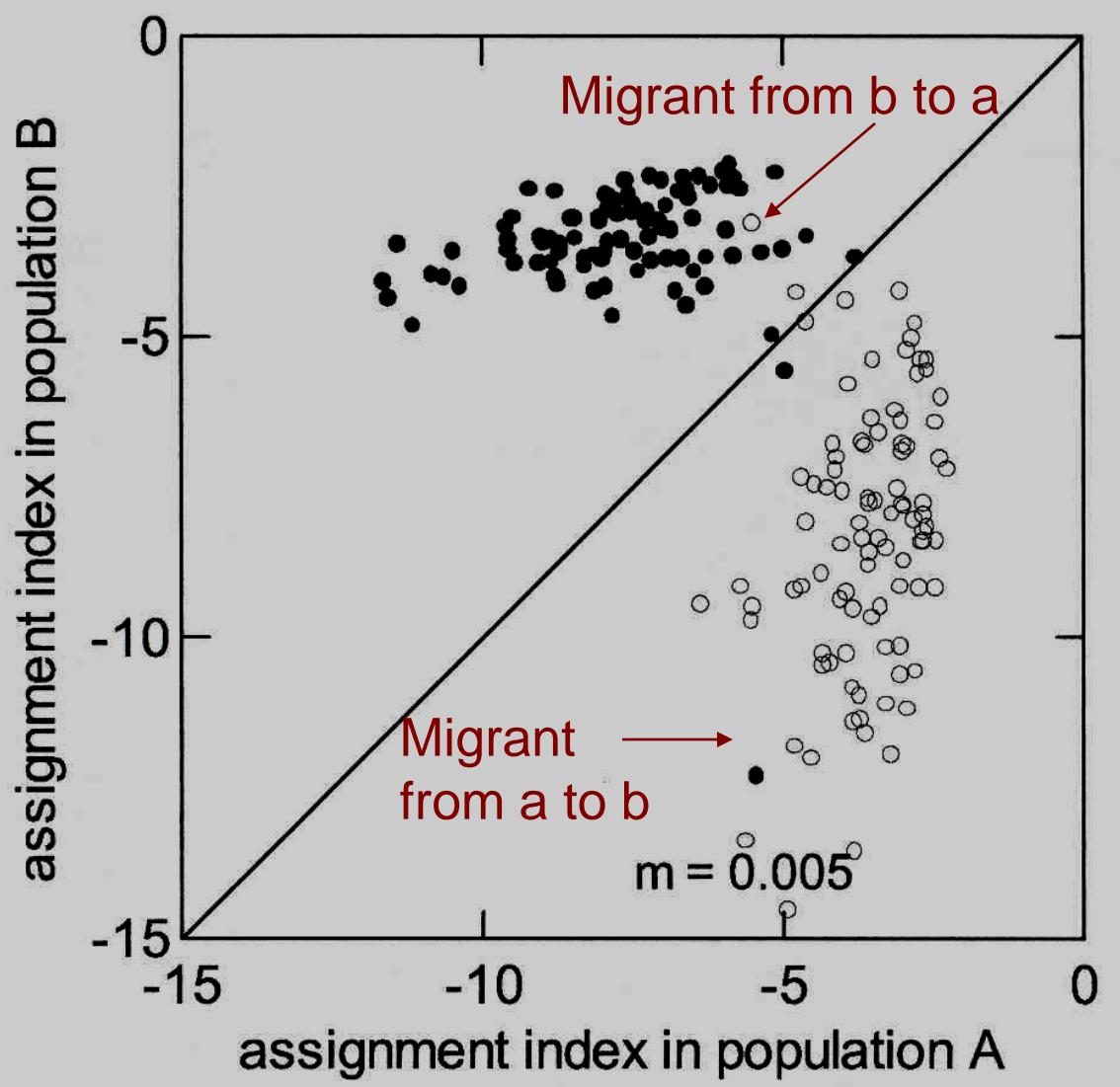
Genotypic Approaches

Direct Measures of Migration Rate Between Fragments

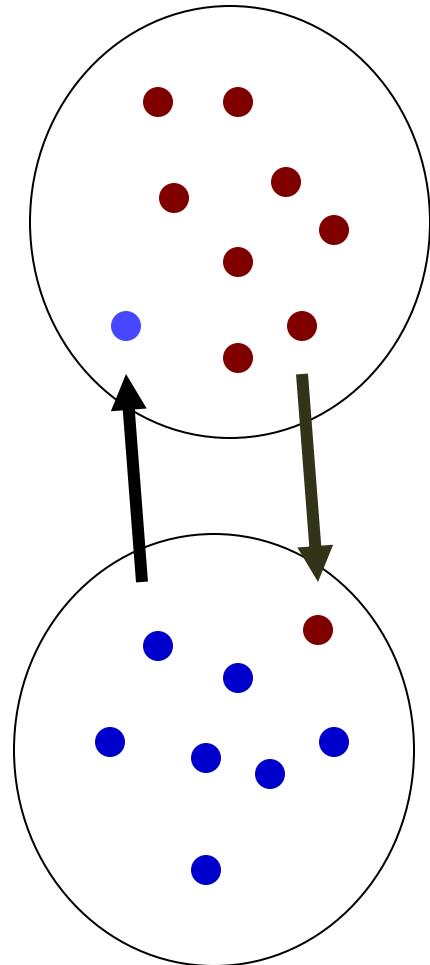
Assignment Tests

likelihood-based technique

1. assume HWE
2. calculate population allele frequencies
3. compute the likelihood of an individual multi-locus genotype belonging to each population in a candidate set of populations
4. assigns that individual to the population where the likelihood of its genotype is the highest



Pop A



Pop B

		Same Location	Movement > 11m	Median distance (Range)
Parentage*	Sib	184	1	48m
	Father	17	3	70m (22-170)
	Mother	33	0	-
Recapture	Imm	85	2	20m (18-22)
	♂	16	7	43m (15-200)
	♀	17	1	40m

If sampling is sufficient then dispersal can be characterized by the distribution of parent-offspring pairs



Next – maintaining genetic variation

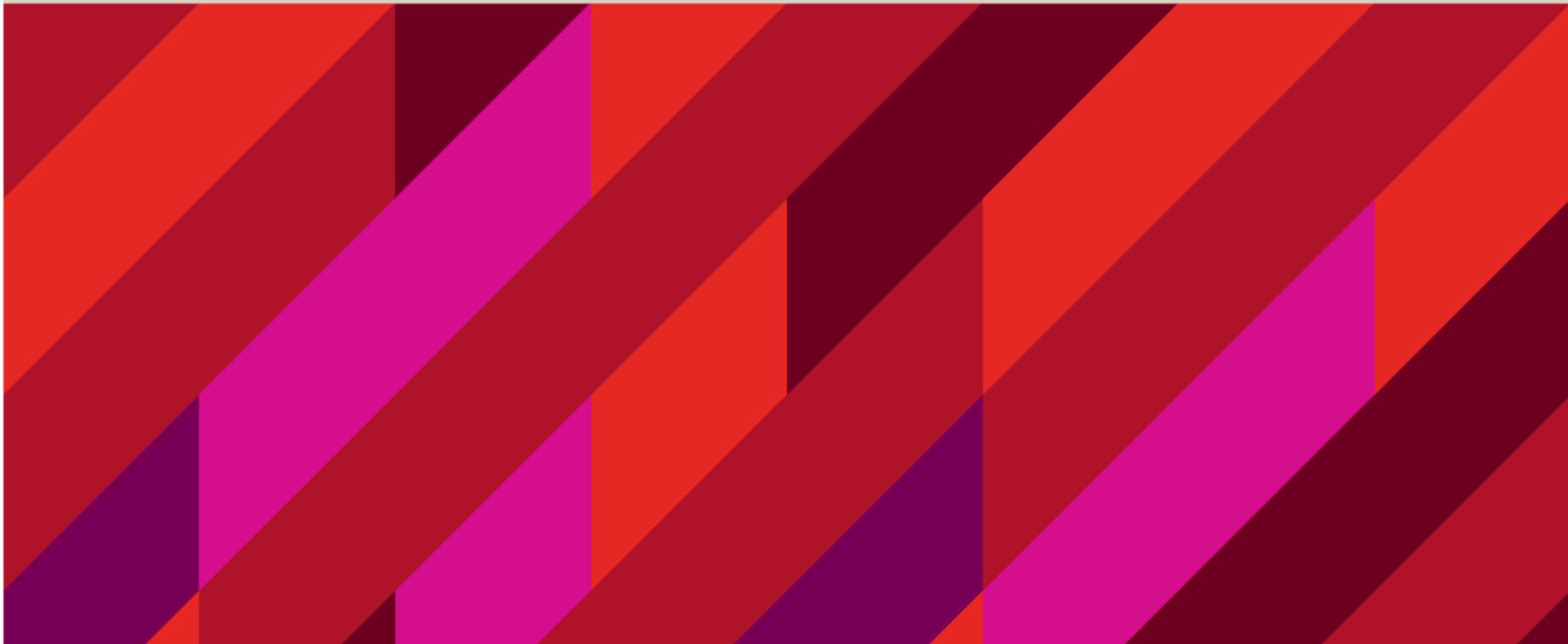




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BIOL3110 Conservation & Ecological Genetics

LECTURE 12: MAINTENANCE OF GENETIC VARIATION

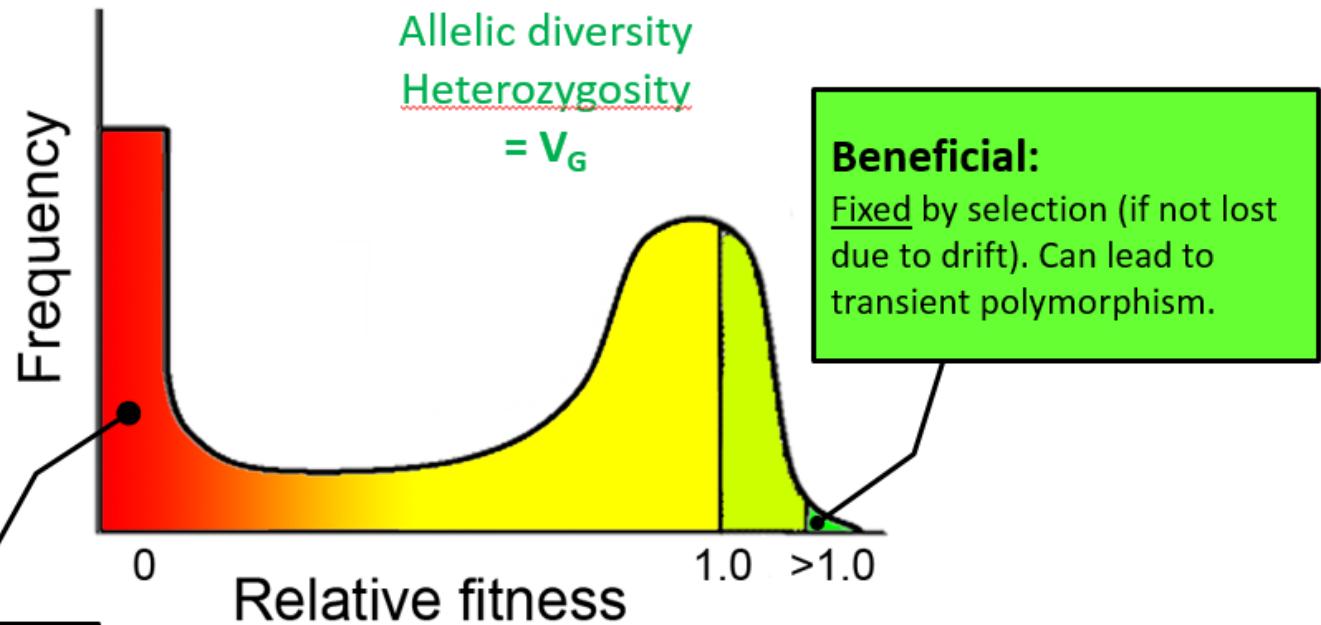


Fate of Mutations



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KEY GENERATOR OF V_G



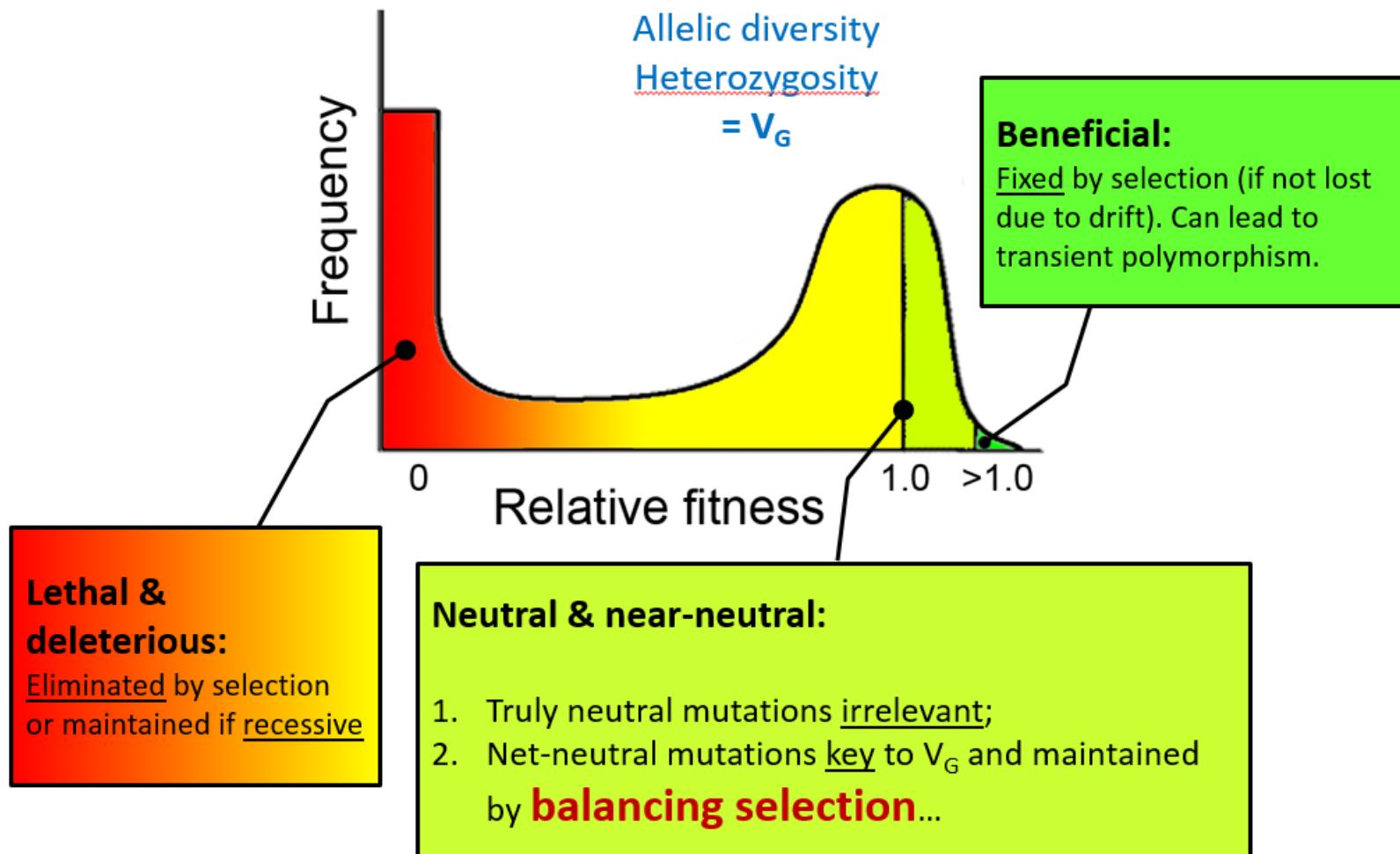
Lethal & deleterious:
Eliminated by selection or maintained if recessive

Fate of Mutations



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KEY GENERATOR OF V_G

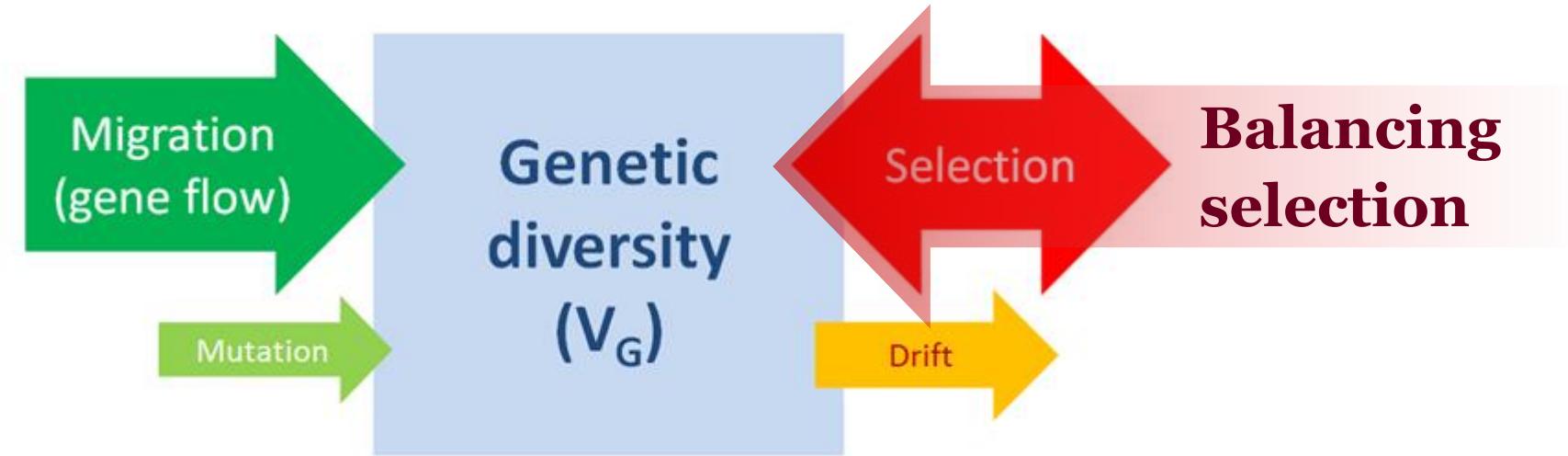


Balancing Selection



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KEY IMPORTANCE TO MAINTAINING V_G



Three mechanisms:

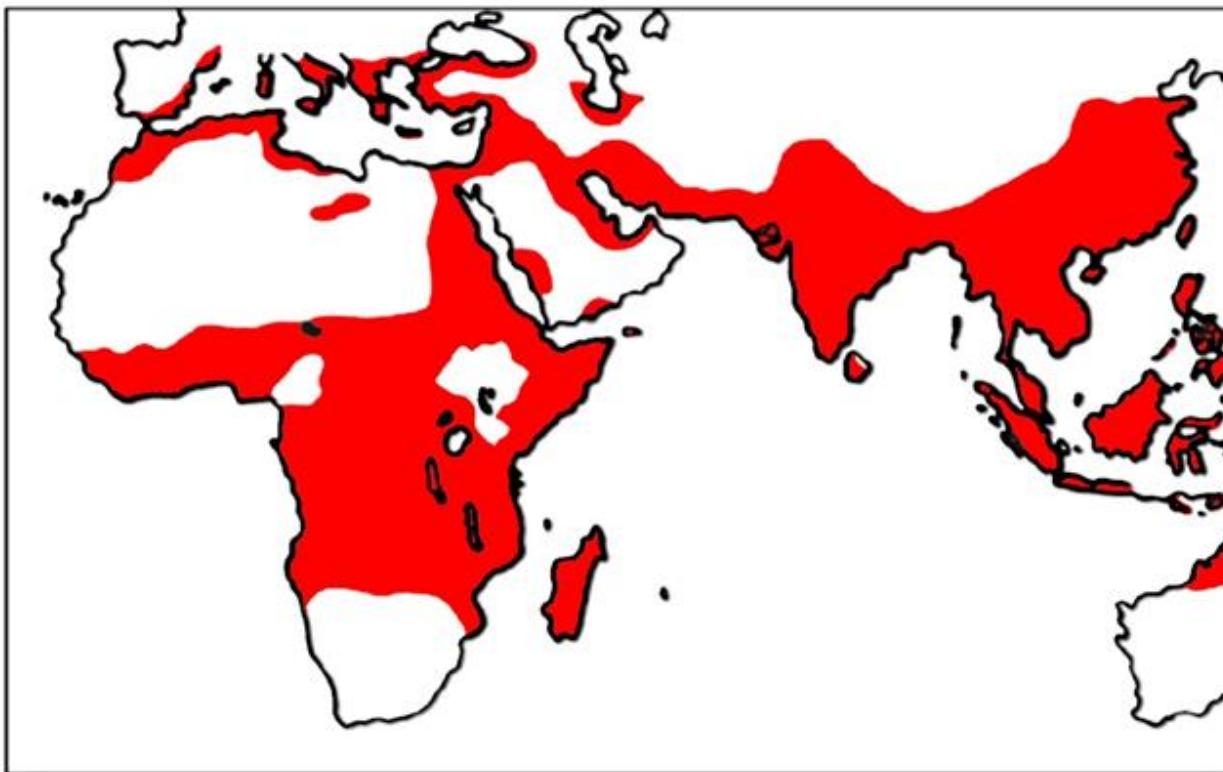
1. Heterozygote advantage (overdominance)
2. Rare allele advantage
3. Gene-environment interaction

Balancing Selection

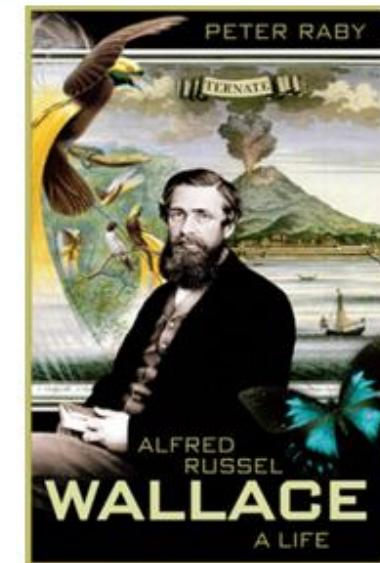


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1. HETEROZYGOTE ADVANTAGE – eg MALARIA RESISTANCE



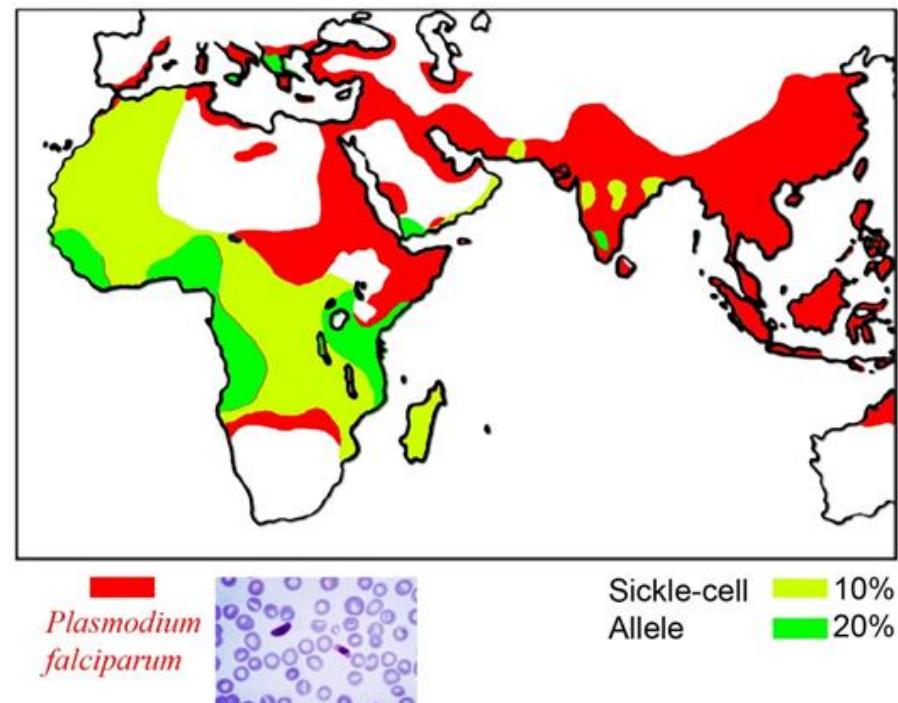
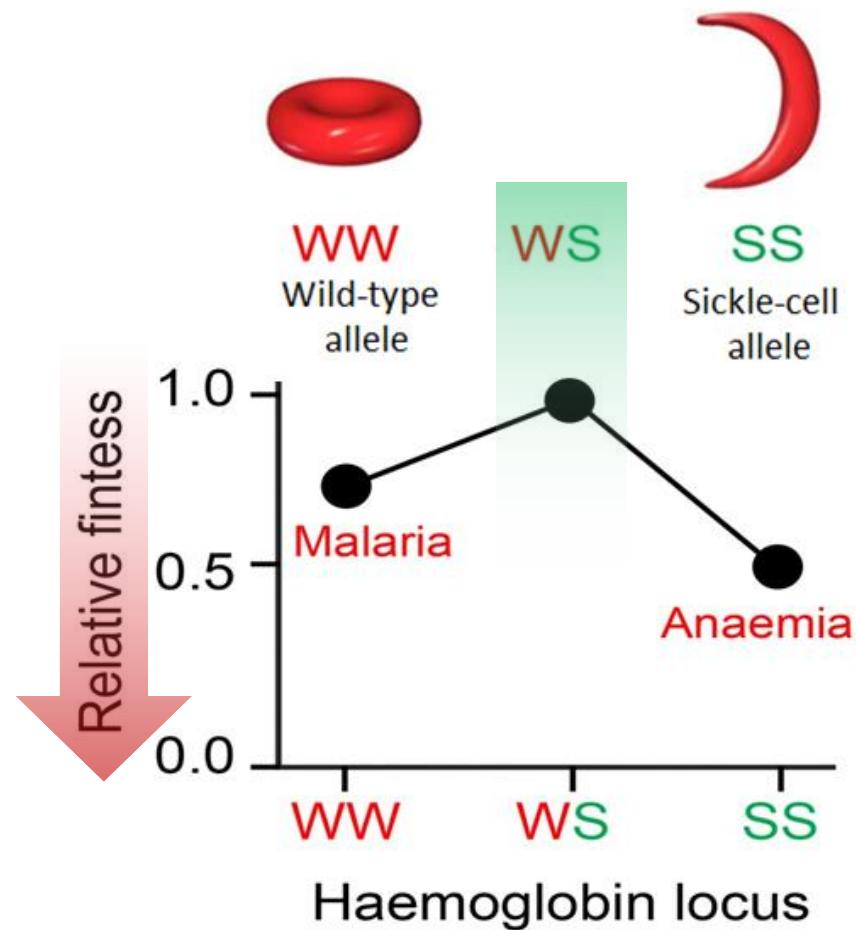
—
*Plasmodium
falciparum*



Balancing Selection



1. HETEROZYGOTE ADVANTAGE – eg MALARIA RESISTANCE

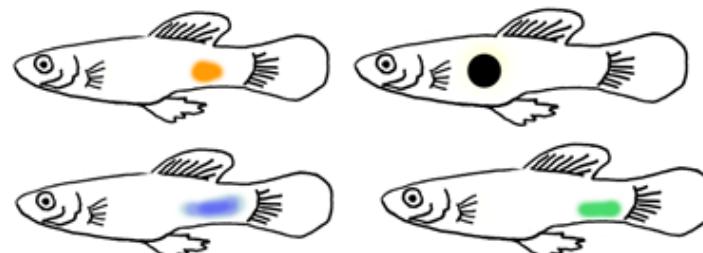
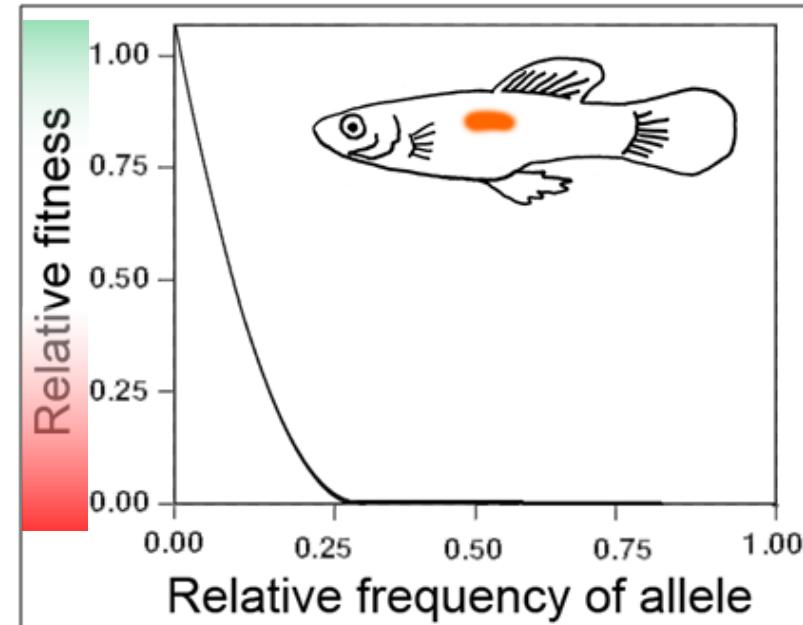
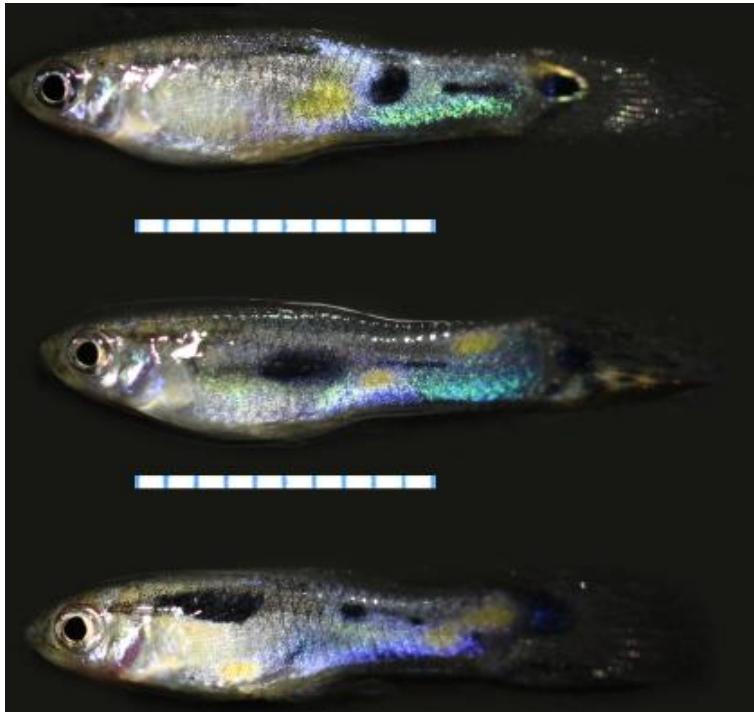
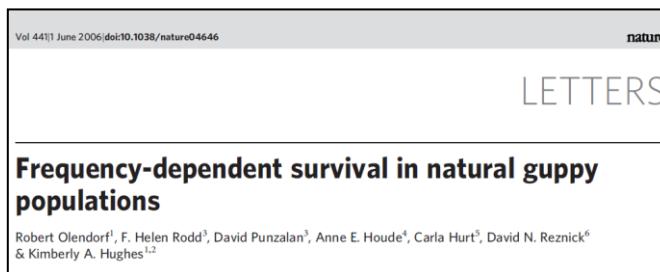


Balancing Selection



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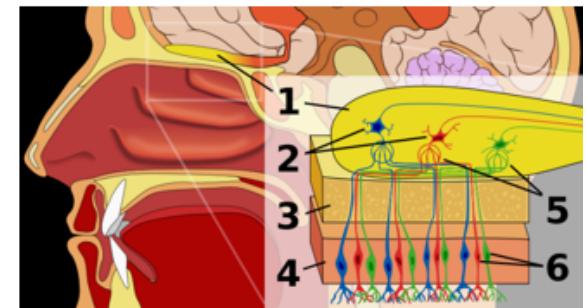
2. RARE ALLELE ADVANTAGE



Balancing Selection

2. RARE ALLELE ADVANTAGE

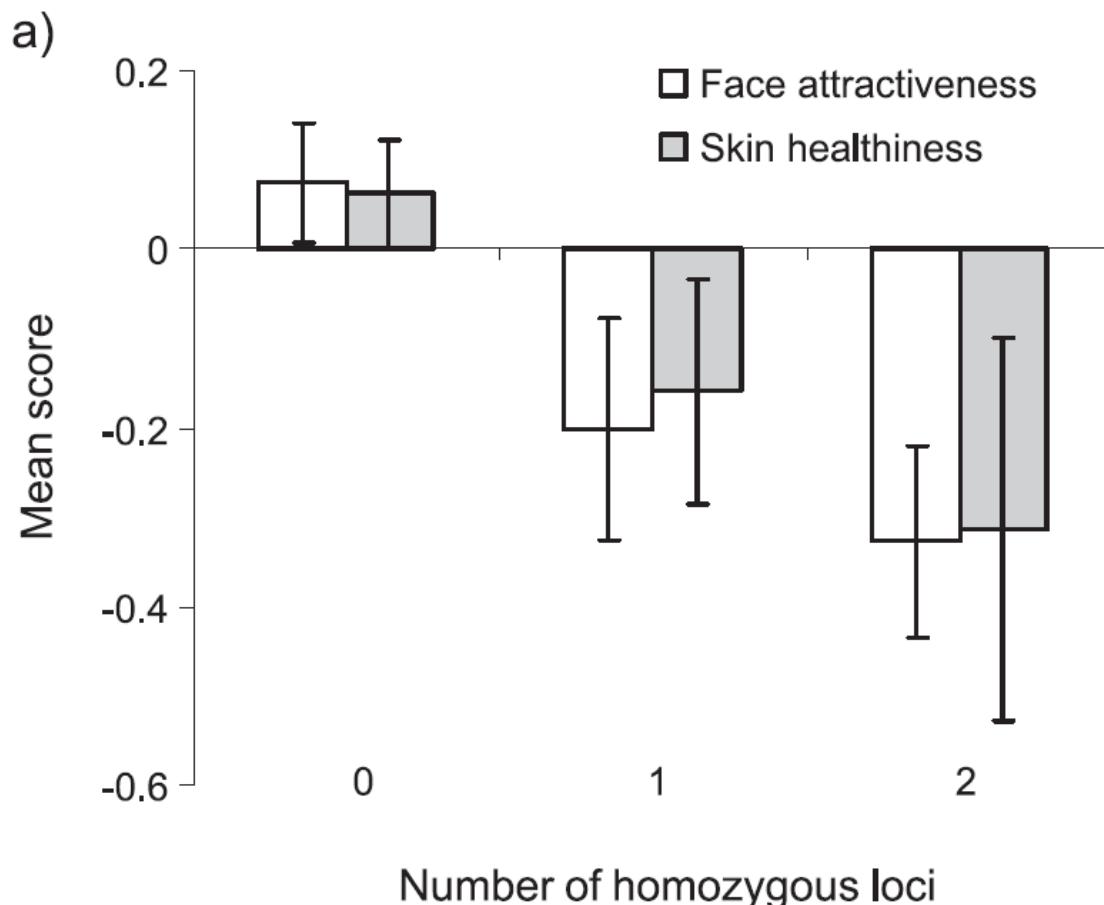
- Code for proteins for surface antigens in 2 classes:
 - Class I Recognize intracellular pathogens and ‘tag’ the cell for immune system components (T-cells, phages etc).
 - Class II
- Highly polymorphic loci with very high allelic diversity
- Sexual selection implicated in maximising population heterozygosity: mate choice for MHC diversity (via olfactory cues in humans)



MHC heterozygosity associated with increased attractiveness in both males and females



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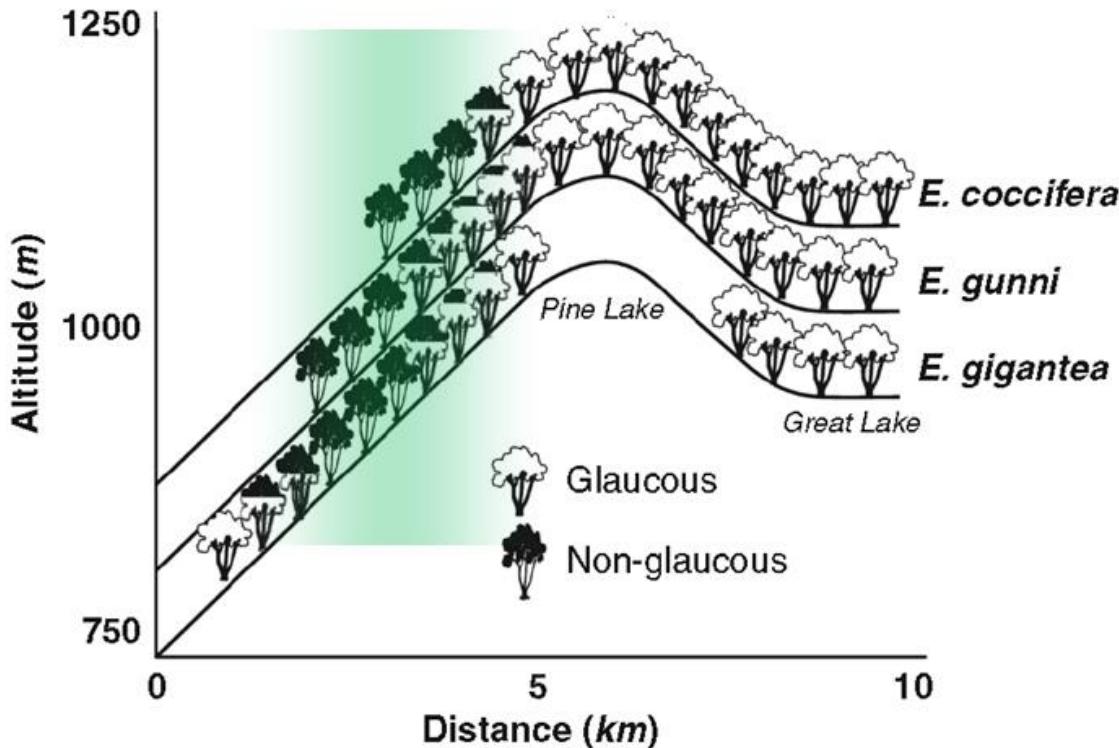


Balancing Selection

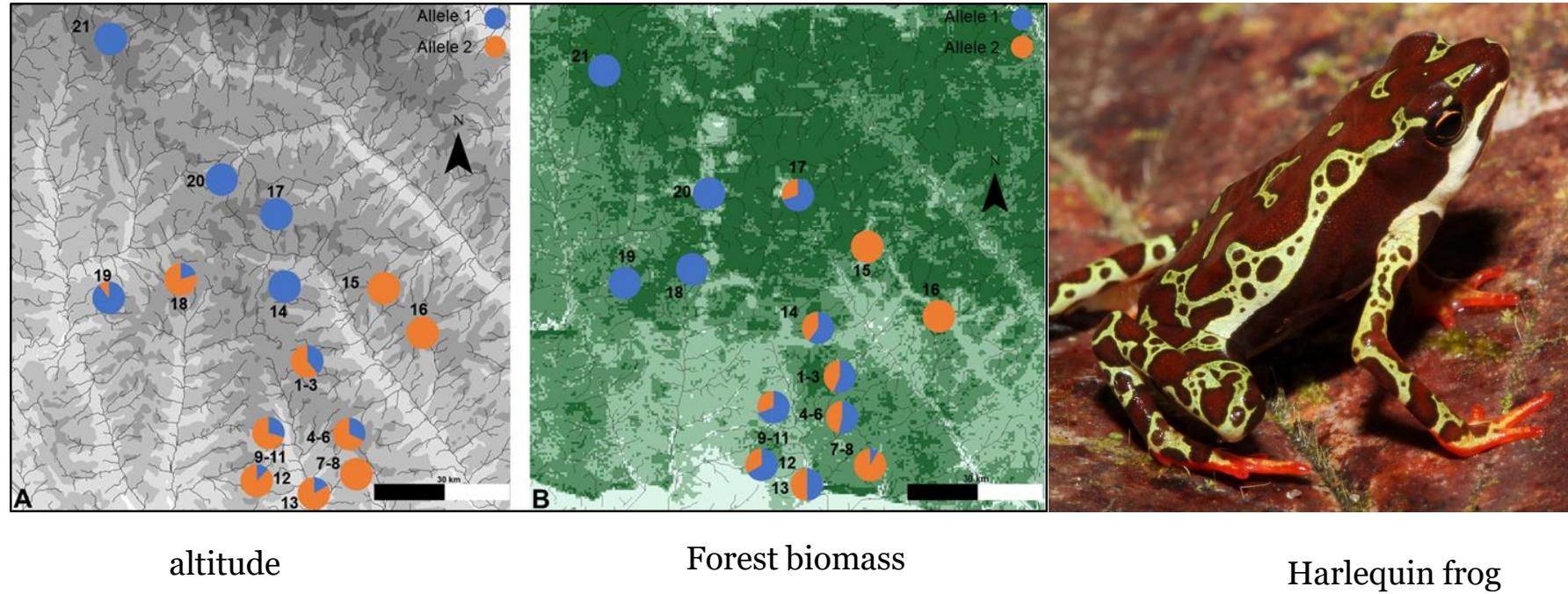


3. GENE ENVIRONMENT INTERACTION

Fitness consequences of variants depend upon the environment

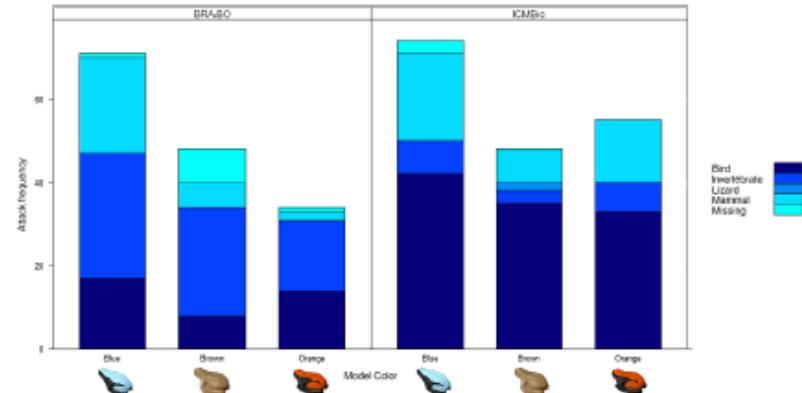
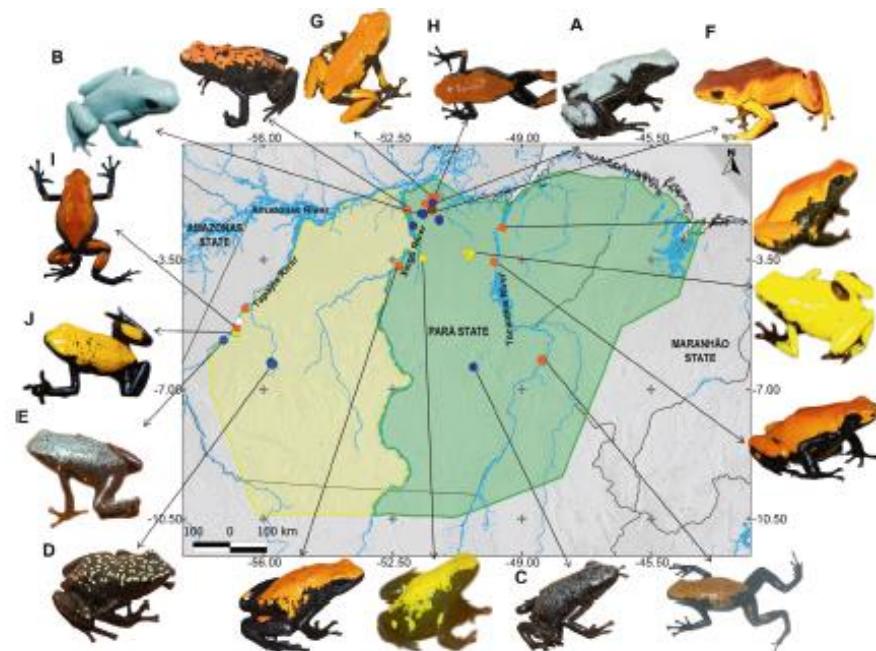


Gene environment interaction



Rafael Jorge et al 2022

Color Polymorphisms *Adelphobates galactonotus* Rojas et al 2020

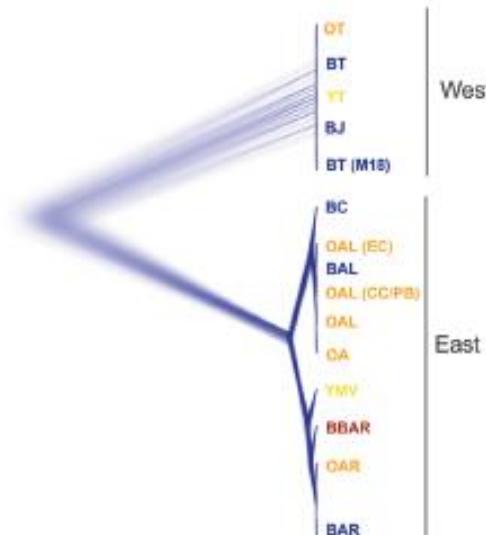
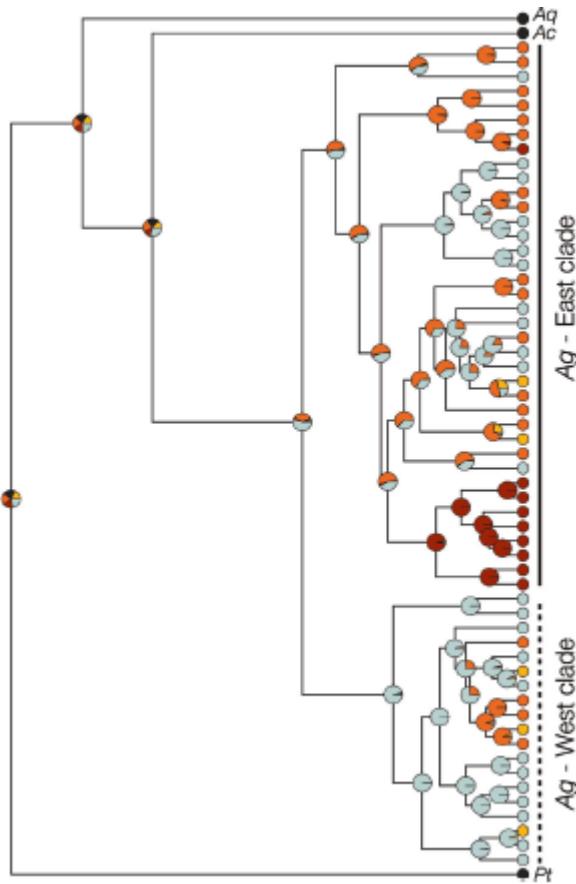


‘Translocation experiments (wax model)
– no difference in predation

GENE ENVIRONMENT INTERACTION



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Same color type arose independently several times

Hypothesis: lower hybrid fitness selected for mate choice based on same coloration

Next Lecture

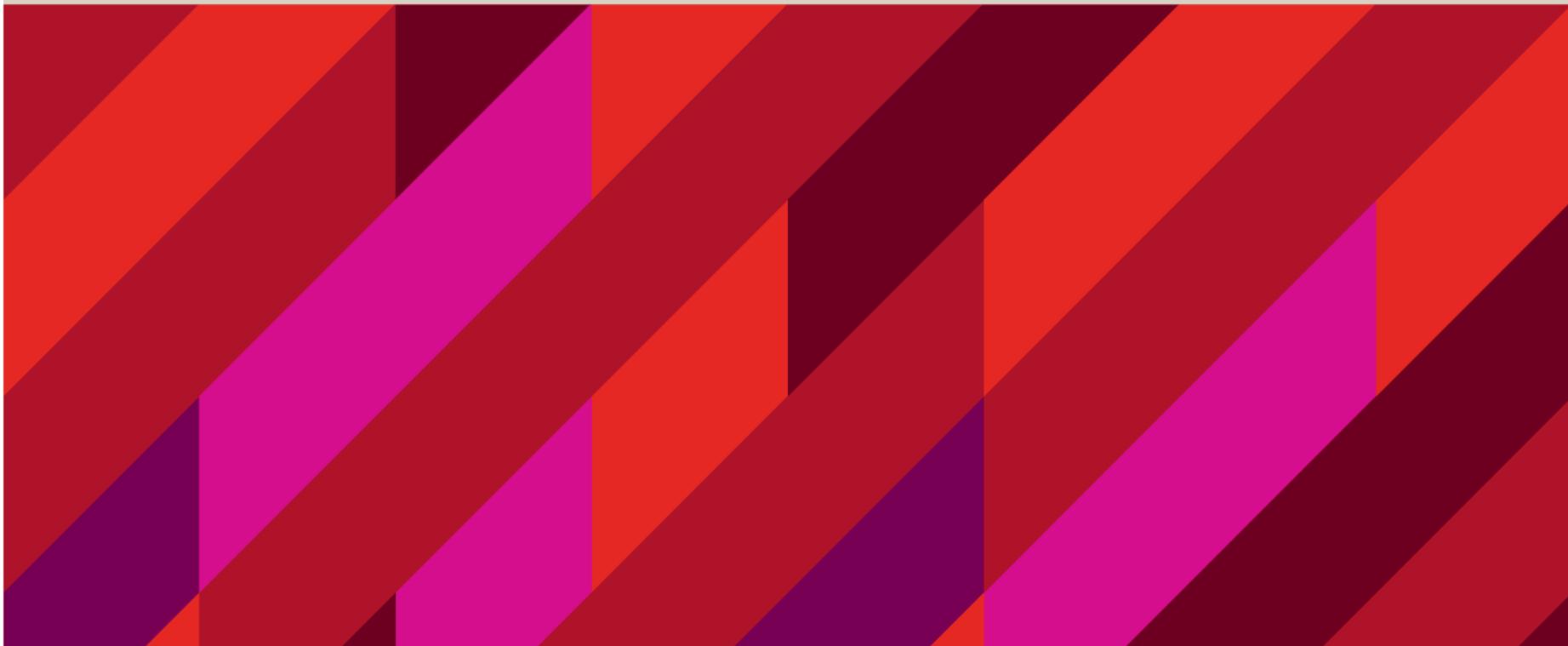
Effective population size



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BIOL3110 Conservation & Ecological Genetics

LECTURE 13: EFFECTIVE POPULATION SIZE



Effective Population Size (N_e)



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SPECIAL EDITION OF “*Heredity*”



EDITORIAL

Effective population size in ecology and evolution

Heredity advance online publication, 24 August 2016; doi:10.1038/hdy.2016.75

Effective population size (N_e) is one of the most important parameters in population genetics and conservation biology. It translates census sizes of a real population into the size of an idealized population showing the same rate of loss of genetic diversity as the real population under study. Several conceptually different types of N_e can be distinguished, but the most commonly used ones are those based on the loss of genetic diversity through inbreeding (inbreeding N_e) and through genetic drift (variance N_e). Often, these two differ considerably, as do census and effective sizes, although, contrary to what is taught in many introductory courses and textbooks, N_e is not always lower than census size (Braude and Templeton, 2009, see also Nunney in this issue). Inbreeding and variance N_e particularly differ when demography changes. However, these relationships are not all that well understood yet, and require further theoretical and empirical attention.

Many recent developments in theory, modeling and statistics as well as in molecular biology have sparked new interest in prediction and estimation of N_e and have led to a number of papers documenting

to show that linkage does not overall increase N_e . Nevertheless, some loci inevitably show authors present a method to reduce this bias by number and length into account. Yet, the precision on SNPs is lower than expected. Jones *et al.* test the reliability of confidence intervals (CIs). The methods to generate CIs and compare them with jackknife techniques. The new methods are when using large numbers of genomic SNPs valuable alternatives for genome-wide data sets. use the LD method, but employ it to test for using SNP markers. The authors test their empirical data set of marine fish in which a recombination has occurred. They show that even small and rare detected and thus provide a valuable new tool for A more specific problem with estimating N_e is a whose paper clarifies the distinction of N_e and population's neighborhood (N_h) and also investi-

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REVIEW

Prediction and estimation of effective population size

J Wang¹, E Santiago² and A Caballero³

Effective population size (N_e) is a key parameter in population genetics. It has important applications in evolutionary biology, conservation genetics and plant and animal breeding, because it measures the rates of genetic drift and inbreeding and the efficacy of systematic evolutionary forces, such as mutation, selection and migration. We review the developments in predictive equations and estimation methodologies of effective size. In the prediction part, we focus on the equations for populations with different modes of reproduction, for populations under selection for unlinked or linked loci and for their applications to conservation genetics. In the estimation part, we focus on methods developed for estimating the current effective size from molecular marker or sequence data. We discuss some underdeveloped areas in predicting and estimating N_e for future research.

Heredity advance online publication, 29 June 2016; doi:10.1038/hdy.2016.43

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NEWS

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ORIGINAL ARTICLE

Long-term effective population size dynamics of an intensively monitored vertebrate population

A-K Mueller, N Chakarov², O Krüger and JI Hoffman

Long-term genetic data from intensively monitored natural populations are important for understanding how effective sizes (N_e) can vary over time. We therefore genotyped 1622 common buzzard (*Buteo buteo*) chicks sampled over years (2002–2013 inclusive) at 15 microsatellite loci. This data set allowed us to both compare single-sample whole-population approaches and explore temporal patterns in the effective number of parents that produced each cohort in relation to observed population dynamics. We found reasonable consistency between linkage disequilibrium-based single-sample temporal estimators, particularly during the latter half of the study, but no clear relationship between annual N_e and census sizes. We also documented a 14-fold increase in \hat{N}_e between 2008 and 2011, a period during which the population size doubled, probably reflecting a combination of higher adult survival and immigration from further afield. Our analysis reveals appreciable temporal heterogeneity in the effective population size of a natural vertebrate population, corroborating the need for long-term studies and cautions against drawing conclusions from a single sample.

Heredity advance online publication, 24 August 2016; doi:10.1038/hdy.2016.67

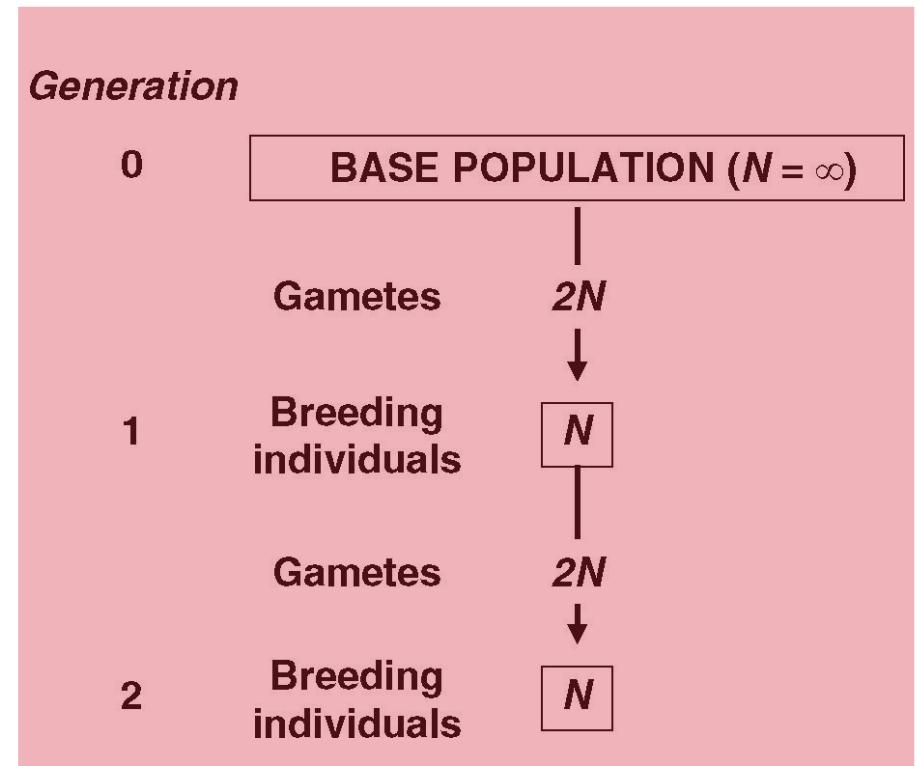
Effective Population Size (N_e)

THE IDEALIZED POPULATION

1. Constant size over time
2. No migration/emigration
3. Random mating
4. Non-overlapping generations
5. Selection ~ 0
6. Mutation ~ 0

A given level of F

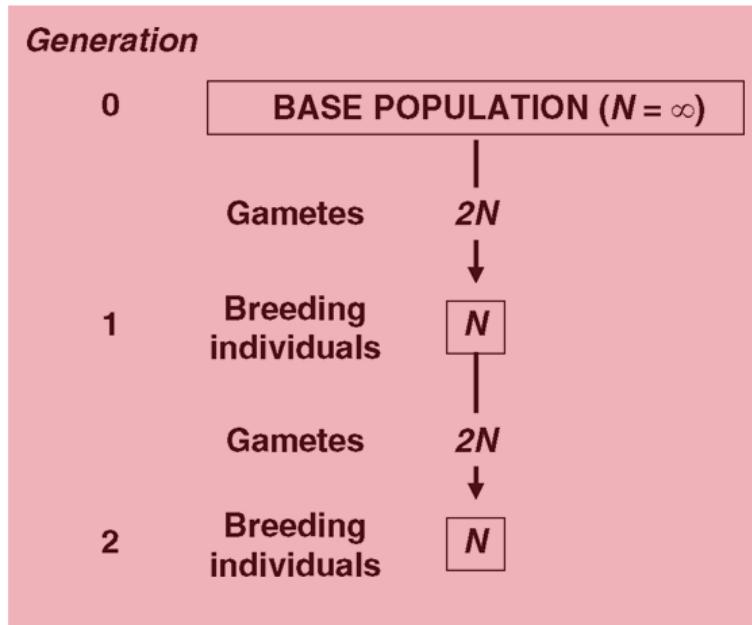
A given dynamic of V_G



Effective Population Size (N_e)

THE IDEALIZED POPULATION

As N departs from ∞ in the idealized population:



1. Loss of V_G due to drift
2. Increased chance of inbreeding ($F > 0$)
3. $H_{\text{obs}} < H_{\text{exp}}$
(ie. H-W expected H)

N_e defined as the size of an idealized population that would **lose V_G at the rate observed** for the study population

Effective Population Size (N_e)



DEFINITION

N_e defined as the size of an idealized population that would **lose V_G at the rate observed** for the study population

Example:

- For a study population of **$N = 500$**
- We measure change in V_G over time and find it reduces at the same rate as an **idealized population** of **$N = 50$**
- Then we define **$N_e = 50$**

Effectively (for purposes of V_G) dealing with N of 50 rather than 500

Effective Population Size (N_e)

DEFINITION

Meta-analysis suggests that N_e much lower than census size

Terrestrial $\sim 11\%$ (0.11) of N for many real-world populations.

Spawning run in Chinook Salmon:

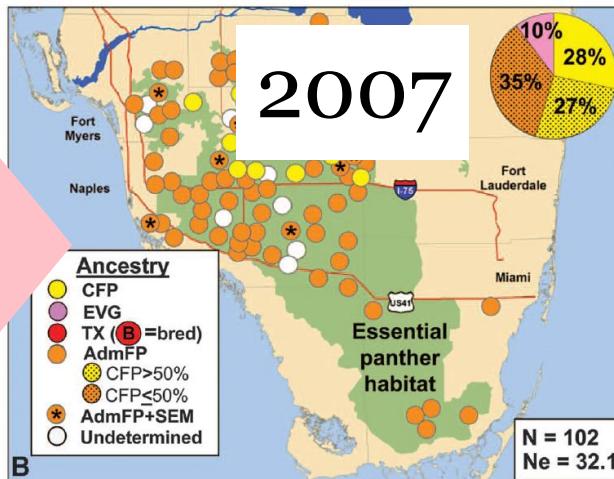
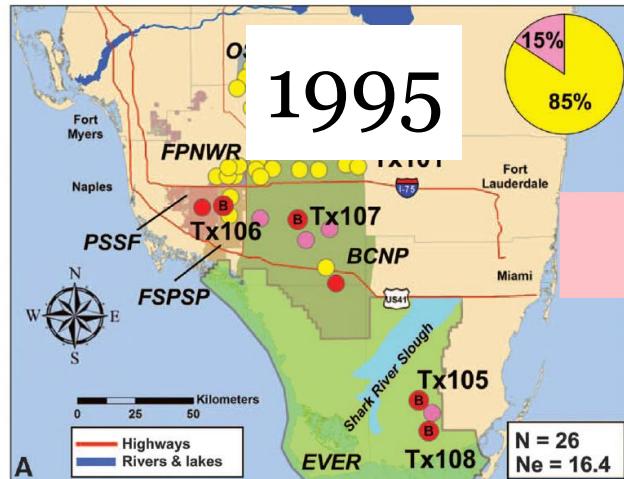
$N = 2000$ but $N_e = 85$

$N_e/N \sim 4\%$



Effective Population Size (N_e)

OFTEN OF INTEREST IS N_e/N



$$N = 26$$

$$N_e = 16.4$$

$$N_e/N = \mathbf{0.63}$$

$$N = 102$$

$$N_e = 32.1$$

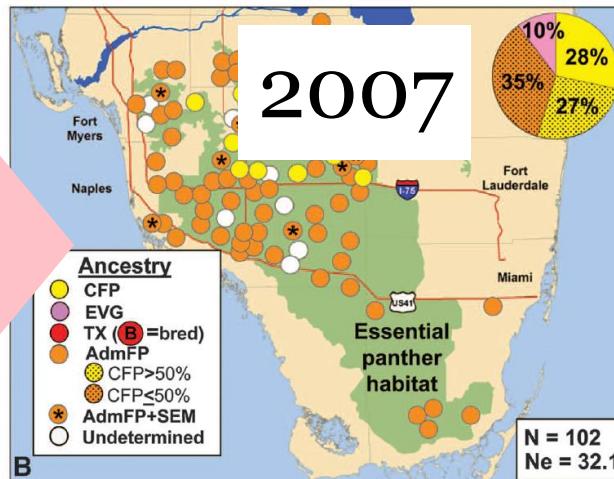
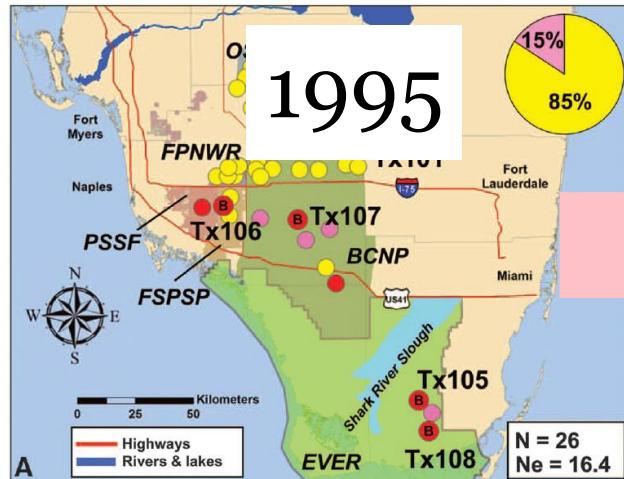
$$N_e/N = \mathbf{0.31}$$

Effective Population Size (N_e)



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OFTEN OF INTEREST IS N_e/N



$$N = 26$$

$$N_e = 16.4$$

$$N_e/N = \mathbf{0.63}$$

$$N = 102$$

$$N_e = 32.1$$

$$N_e/N = \mathbf{0.31}$$

BUT if: $N = 102$

$$N_e = 16.4$$

$$N_e/N = \mathbf{0.16}$$

Effective Population Size (N_e)



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ESTIMATION

Two bases of estimation:

(1) Knowledge of **population genetics** over time;

Changes in:

- Heterozygosity (**H**) or allelic diversity (**A**)
- Inbreeding coefficient (**F**) or Linkage Disequilibrium (**LD**)

And others including changes in neutral allele frequencies due to drift (see Frankham Ch. 11 pp.252)

(1) Knowledge of **population demography** over time;

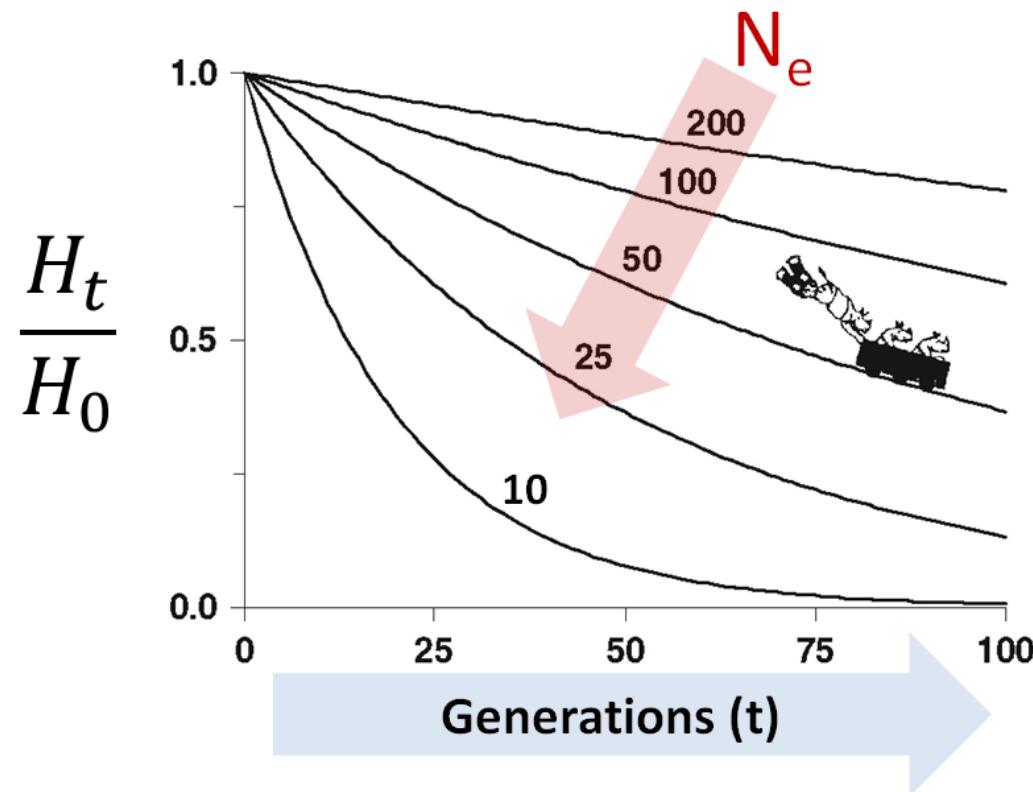
Effective Population Size (N_e)



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ESTIMATION – GENETIC FACTORS

1. Larger (effective) pops retain more V_G ...
2. Ie. H decays as the **inverse of N_e**



Effective Population Size (N_e)



ESTIMATION – GENETIC FACTORS

1. Larger (effective) populations retain more genetic diversity...
2. H decays as the **inverse** of N_e

Described by the equation:

$$\frac{H_t}{H_0} = \left(1 - \frac{1}{2N_e}\right)^t$$

Effective Population Size (N_e)



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ESTIMATION – GENETIC FACTORS

1. Larger (effective) populations retain more genetic diversity...
2. H decays as the **inverse** of N_e

Described by the equation:

Hence, if we know this
we can calculate N_e

$$\frac{H_t}{H_0} = \left(1 - \frac{1}{2N_e}\right)^t$$

Effective Population Size (N_e)



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ESTIMATION – GENETIC FACTORS

We can make predictions about H over time...

Example: If we know $N_e = 500$, then can predict the loss of H in **one generation** (i.e. at $t = 1$):

$$\frac{H_t}{H_0} = \left(1 - \frac{1}{2N_e}\right)^t = \left(1 - \frac{1}{1000}\right)^1 = 0.999$$

Where: t = number of generations

~0.1% of
 H lost

Effective Population Size (N_e)



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ESTIMATION – GENETIC FACTORS

We can make predictions about H over time...

Example: If we know $N_e = 500$, then can predict the loss of H in **one generation** (i.e. at $t = 1$):

$$\frac{H_t}{H_0} = \left(1 - \frac{1}{2N_e}\right)^t = \left(1 - \frac{1}{1000}\right)^1 = 0.999$$

Where: t = number of generations

~0.1% of
 H lost

Or after, say **50 generations** (i.e. at $t = 50$):

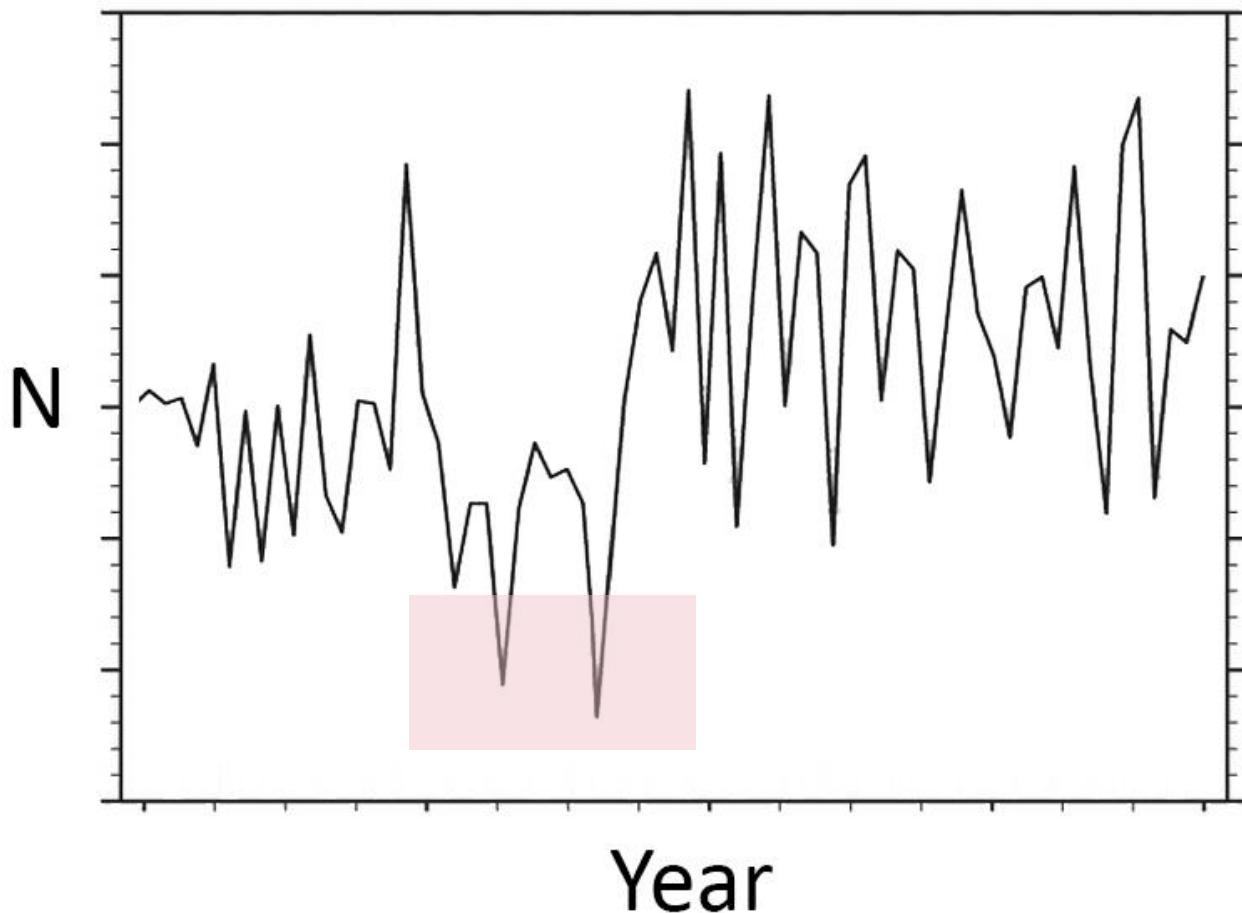
$$\frac{H_1}{H_0} = \left(1 - \frac{1}{2N_e}\right)^t = \left(1 - \frac{1}{1000}\right)^{50} = 0.951$$

~5% of H
lost

Effective Population Size (N_e)

ESTIMATION – DEMOGRAPHIC FACTORS

Fluctuations in N over time (1)

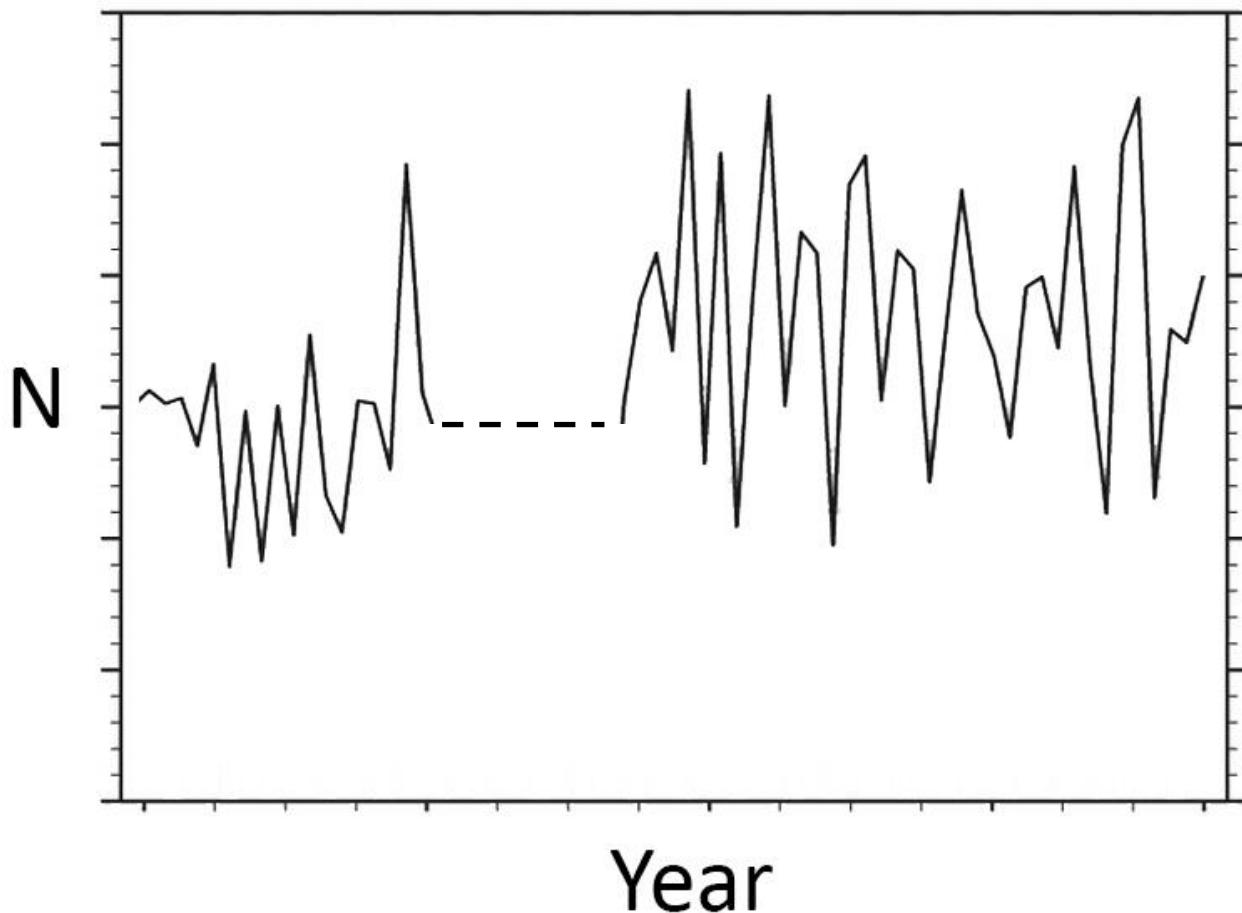


$$N_e = \frac{t}{\sum(\frac{1}{N_{e1...t}})}$$

Effective Population Size (N_e)

ESTIMATION – DEMOGRAPHIC FACTORS

Fluctuations in N_e over time (2)



$$N_e = \frac{t}{\sum(\frac{1}{N_{e1...t}})}$$

Harmonic Mean

ESTIMATION – DEMOGRAPHIC FACTORS

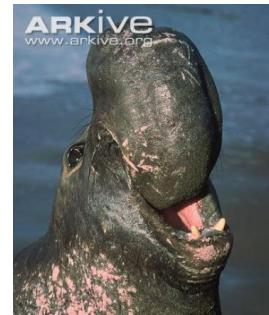
Fluctuations in N over time

Time (t) = # generations

E.g. Northern elephant seals

Assuming historic $N \sim 175,000$,

bottleneck of 20, recovery to 175,000:



$$N_e = \frac{t}{\sum(\frac{1}{N})} = \frac{3}{\left(\frac{1}{175000} + \frac{1}{20} + \frac{1}{175000} \right)} = 60$$

This is the harmonic mean – sensitive to the lowest N value (the **simple average** is actually **116,673**)



Single sample estimators

Table 1 Summary of results comparing performance of effective size estimators on simulated data with true $N_e = 100$

	Microsats; $P_{\text{Crit}} = 0.02$			SNPs; $P_{\text{Crit}} = 0$		
	LDNe	Het Excess	Coancestry	LDNe	Het Excess	Coancestry
Single sample						
Hmean (\hat{N}_e)	106.0	77.5	39.6	102.7	78.4	28.2
Min	82.2	18.2	14.6	84.1	19.5	11.3
Max	139.6	Infinite	Infinite	131.7	Infinite	Infinite
% Infinite	0.0	30.0	17.0	0.0	29.0	11.0
CV ($1/\hat{N}_e$)	0.114	0.974	0.777	0.095	0.992	0.648

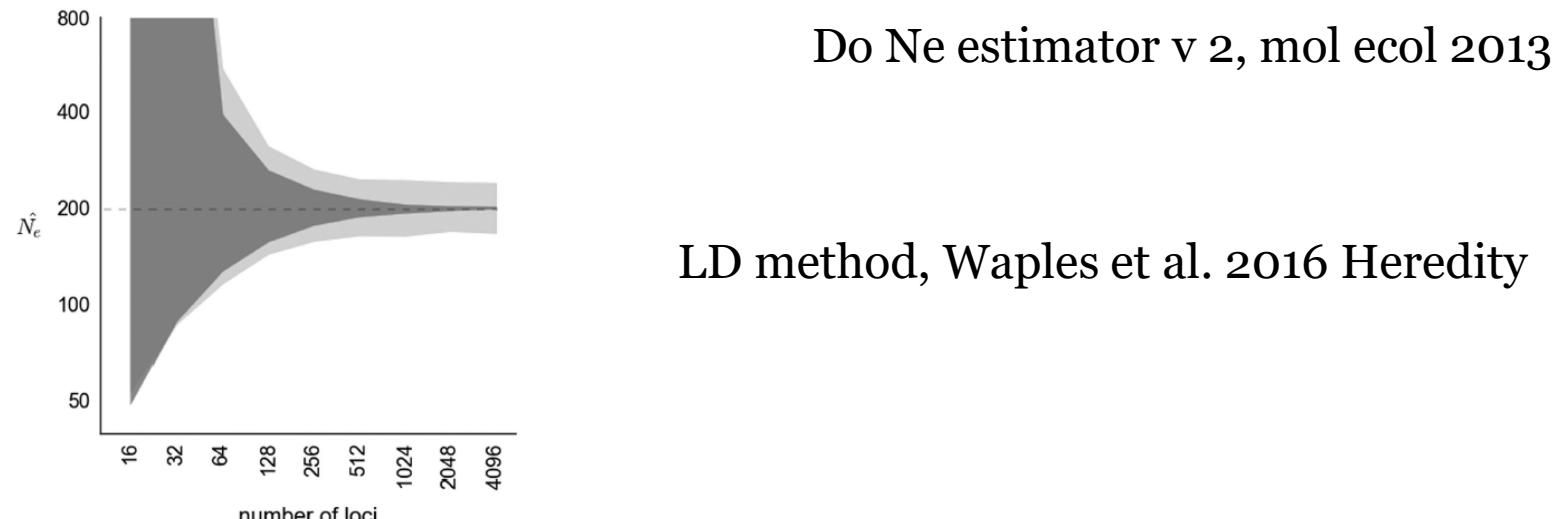
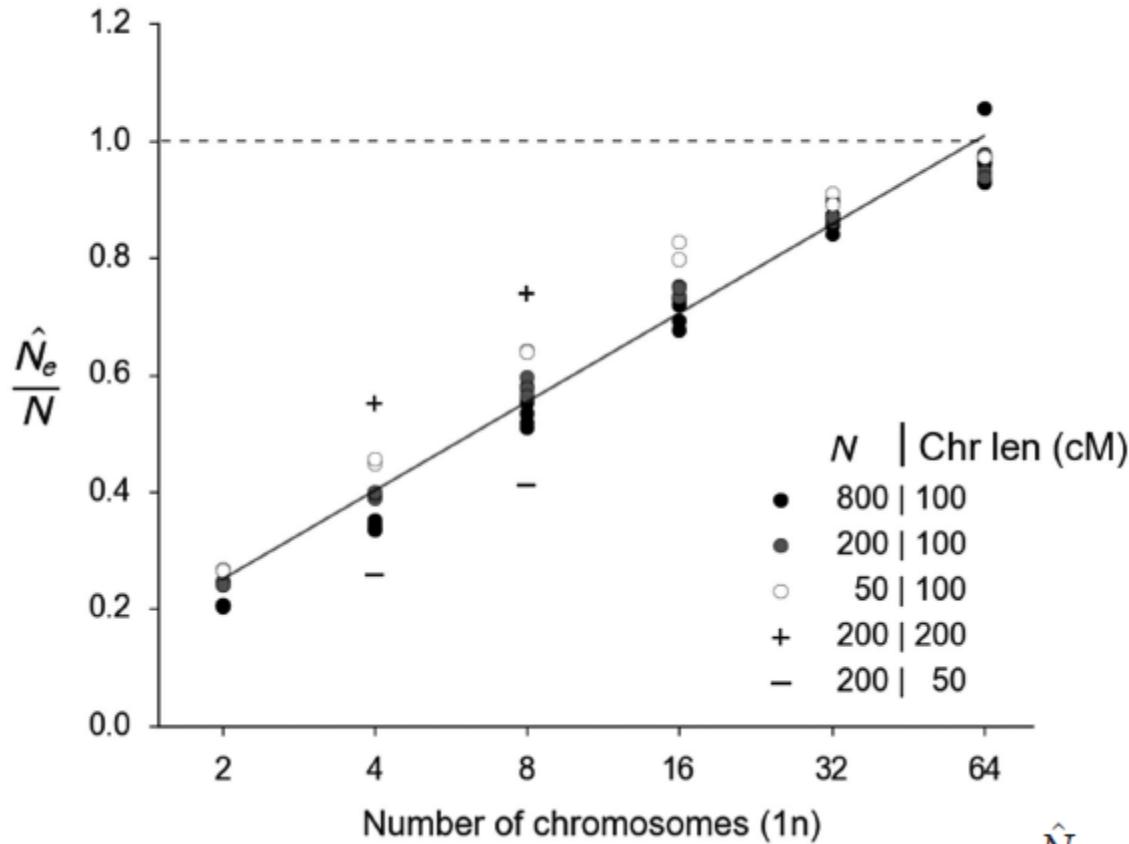


Figure 7 Relationship between theoretical and empirical (light) precision for estimates of N_e . Dark shaded area shows naive 95% CIs (which assume that all pairwise comparisons are independent), light shaded area shows empirical CIs from simulated data. True N_e was 200, sample size was 100 and number of chromosomes was 8. Notice both the x and y axes are log-scaled.

LD method – effects of physical linkage



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Number of chromosomes and their length matter – strong linear relationship allows for adjustment

$$\frac{\hat{N}_e}{N_e} = 0.098 + 0.219 \times \ln(Chr)$$

Effective population size – loss of genetic variation methods..



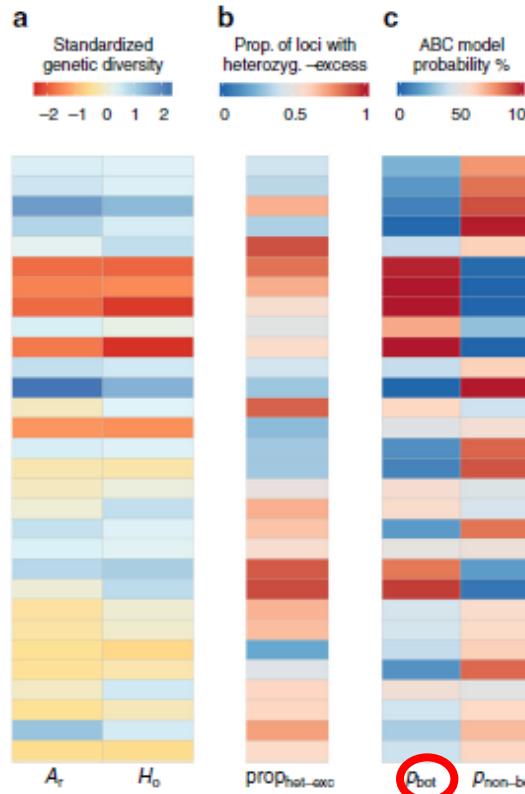
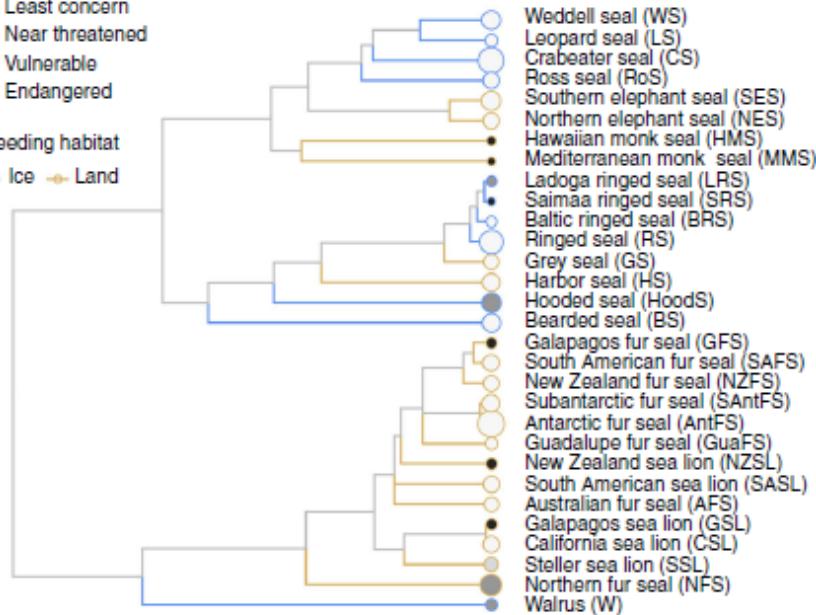
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Global abundance
 $\circ 10^3$ $\circ 10^4$ $\circ 10^5$ $\circ 10^6$

IUCN rating
 \circ Least concern
 \circ Near threatened
 \bullet Vulnerable
 \bullet Endangered

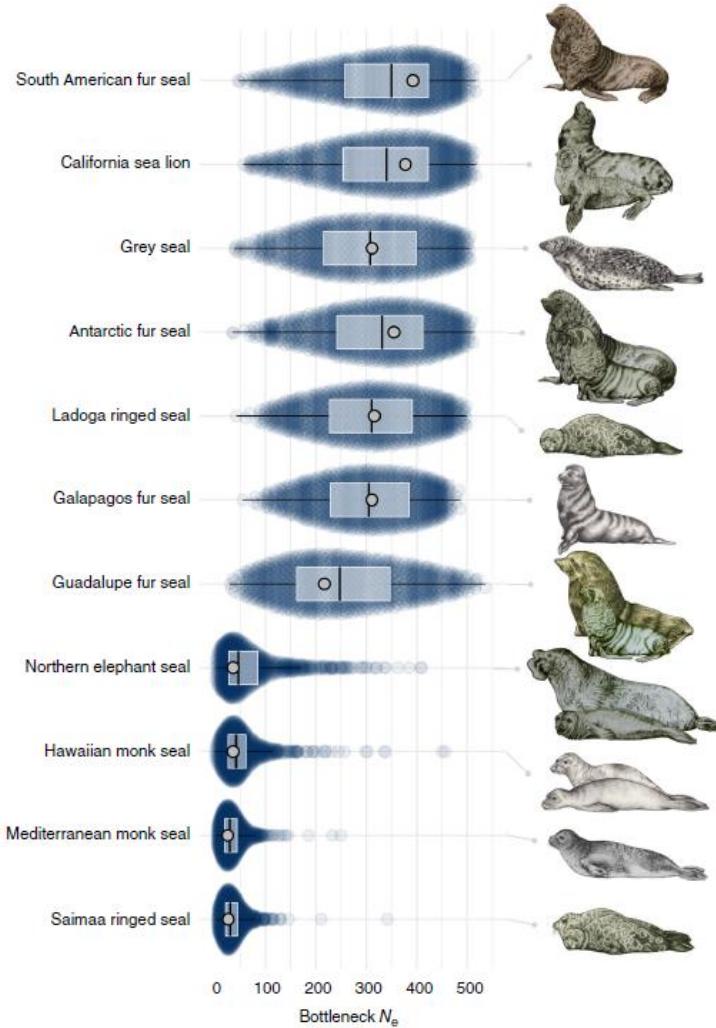
Breeding habitat
 \leftarrow Ice \rightarrow Land



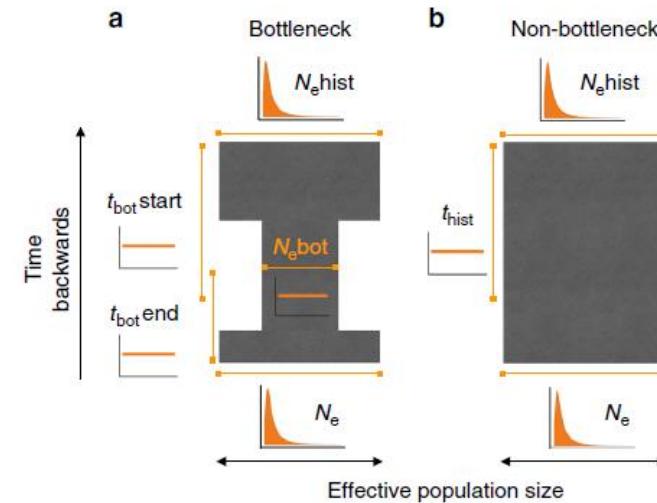
Coalescent based estimate of N_e



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Bottleneck time priors encompass last 4 centuries, before and after bottleneck and N_e 1-500



Endangered

Stoffel et al 2018 Nature Communications



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Case study: Genetic Connectivity of the Australian white ibis

Implications for Management

**Kaytlyn Davis, Dr. Kate Brandis, Dr.
Shannon Smith and Associate
Professor Adam Stow**



A Knowledge Gap in Waterbird Management

- Movement studies
 - Banding and satellite tracking
 - Radio-isotopes (Feather Map of Australia project)
- Few genetic studies
 - Barriers to gene flow?
 - Effective population sizes?
 - Impacts of management?



Why care about gene flow?

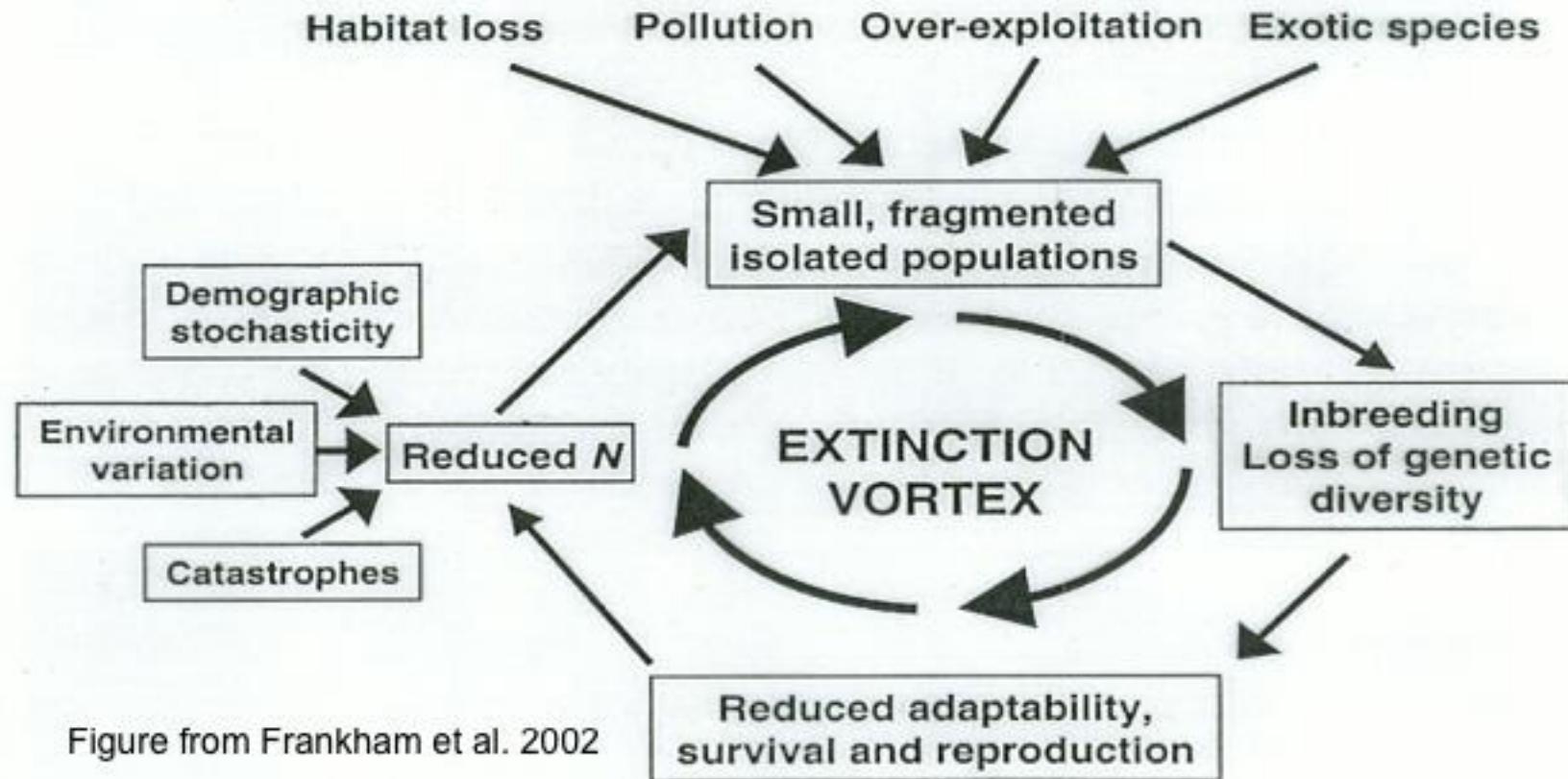


Figure from Frankham et al. 2002

Natural Flow Regime of Australia's Wetlands



‘Boom’ periods

- Sequential flooding
- High productivity
- Recruitment of waterbirds
- Successful breeding

‘Bust’ periods

- Wetland drying
- Low productivity
- Waterbirds disperse to other wetlands

Anthropogenic Processes place added Stress on Waterbirds



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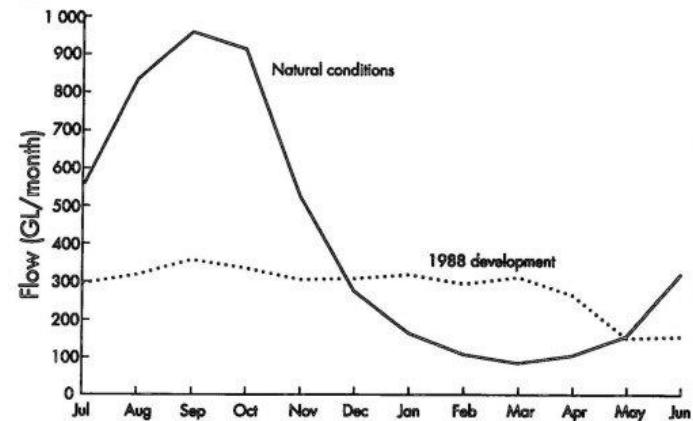
Water diversion and river regulation
for agricultural/commercial uses



Disappearance of drought
refuges and climate change

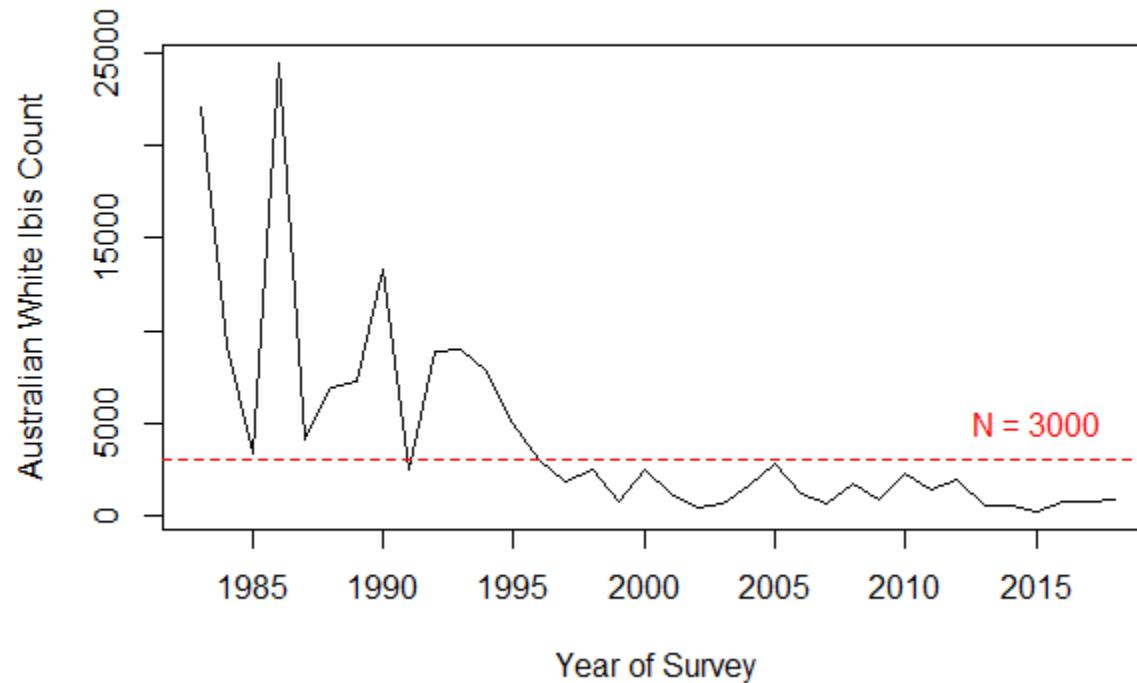


Declines in water quality due to
pollution/land use changes



Altered flow regimes and
flooding onset times/duration

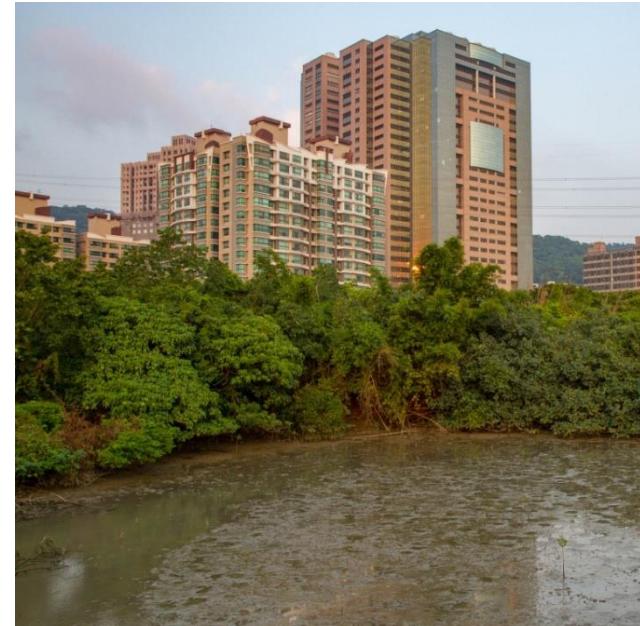
Inland Colonies of the Ibis have Declined Drastically



Source: Australian Aerial Waterbird Survey Data



The Rise of Urban Ibis



Inland Wetlands

Variable

Travel long distances in
search of resources

Urban Wetlands

Regular

Reduced need to travel
long distances



The Rise of Urban Ibis



Inland Wetlands

Variable
Travel long distances in
search of resources



Urban Wetlands

Regular
Reduced need to travel
long distances

Dichotomy of Ibis Management

INLAND



Variable
↓ Flows & flooding
↓ Resources

↓ Breeding opportunities
↓ Abundance
 < 3000

Conservation

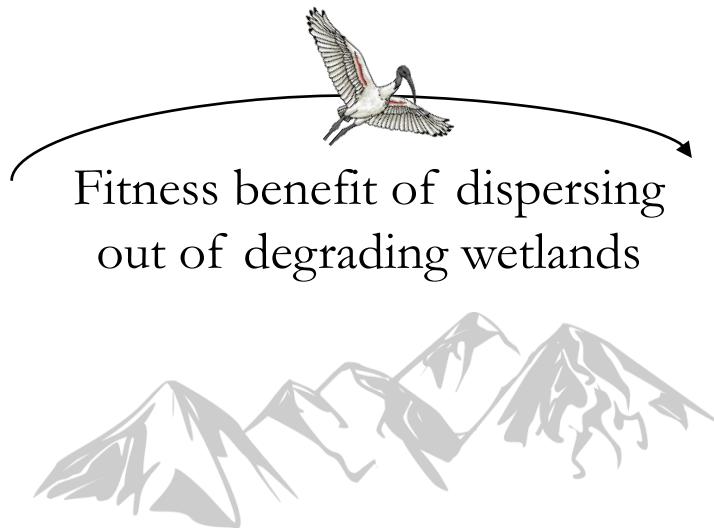
- Controlled release of water (E-water)
- Monitoring

COASTAL

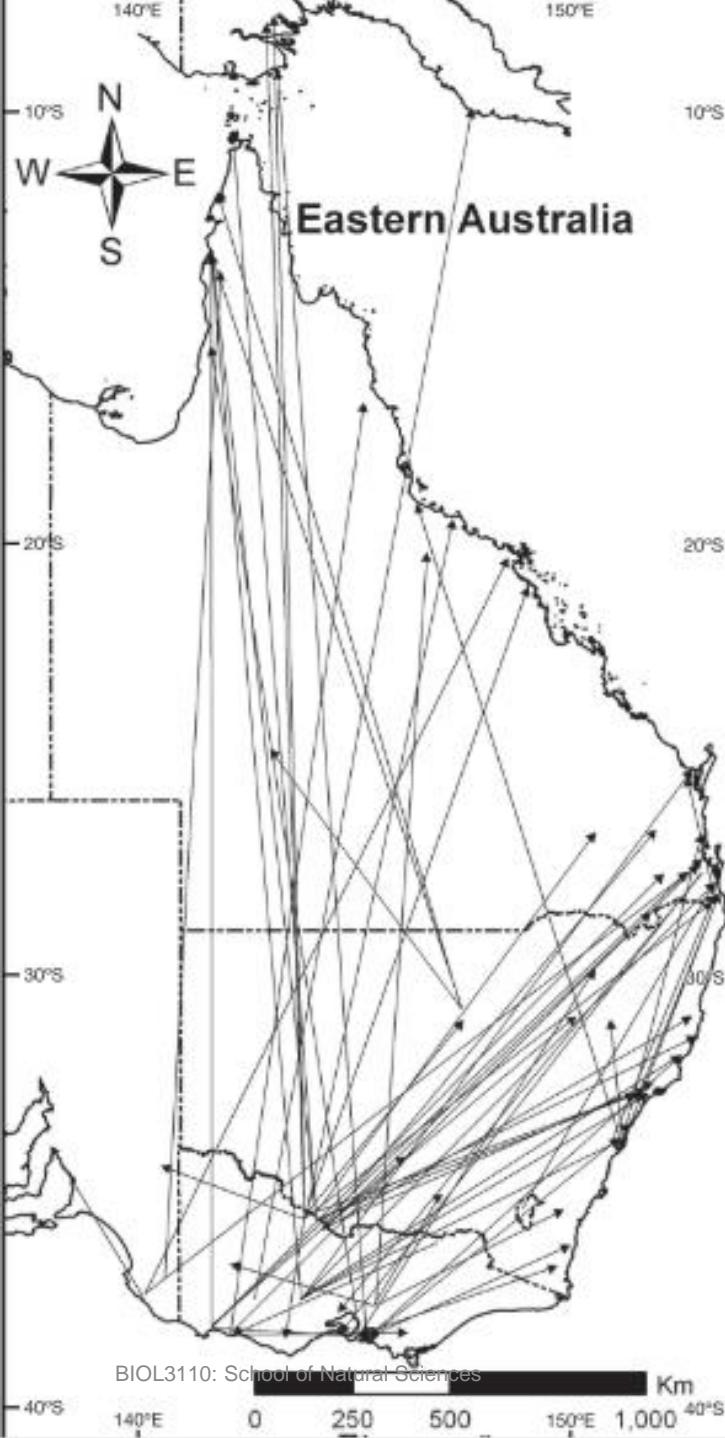


Regular
↑ Permanent water
↑ Resources

↑ Breeding opportunities
↑ Abundance
↑ Sedentariness



Management
• Control numbers
• Management targets



Long-distance dispersal capacity

Likely move to coast to breed

Resource driven

Sedentary and mobile sub-populations?

Less dispersal in urban colonies?

- Impact of wetland stability on gene flow is unknown
- Genetic diversity and demographic stability is unknown



Research themes

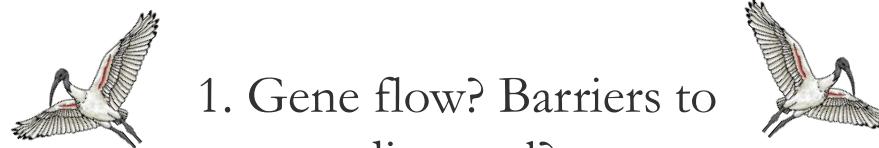
INLAND



Variable

↓
Breeding opportunities
↓
Abundance < 3000

Conservation



1. Gene flow? Barriers to dispersal?

2. Genetic diversity and Effective population size?

3. Will management impact future genetic diversity?

If genetically connected,
local processes may
impact both regions

COASTAL



Regular

↓
↑ Breeding opportunities
↓
↑ Abundance
↑ Sedentariness

Management



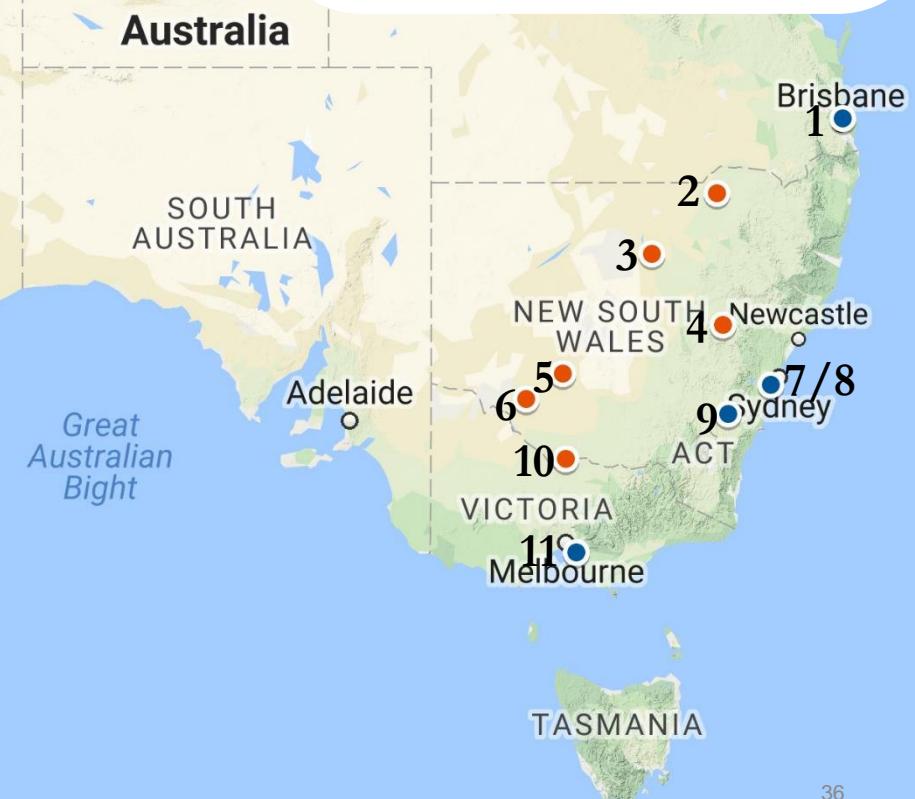
METHODS and RESULTS

Sites and Sampling

- 5 Urban, 6 Inland
- Collaboration with the Feather Map of Australia Project
- Feathers collected non-invasively



1. Boondall Wetlands
2. Gwydir Wetlands
3. Macquarie Marshes
4. Putta Bucca Wetlands
5. Booligal
6. Murrumbidgee
7. Centennial Parklands
8. Lucas Heights (foraging site)
9. Goulburn Wetlands
10. Barmah-Millewa Forest Wetlands
11. River Gum Creek Reserve



Feather Map of Australia Project

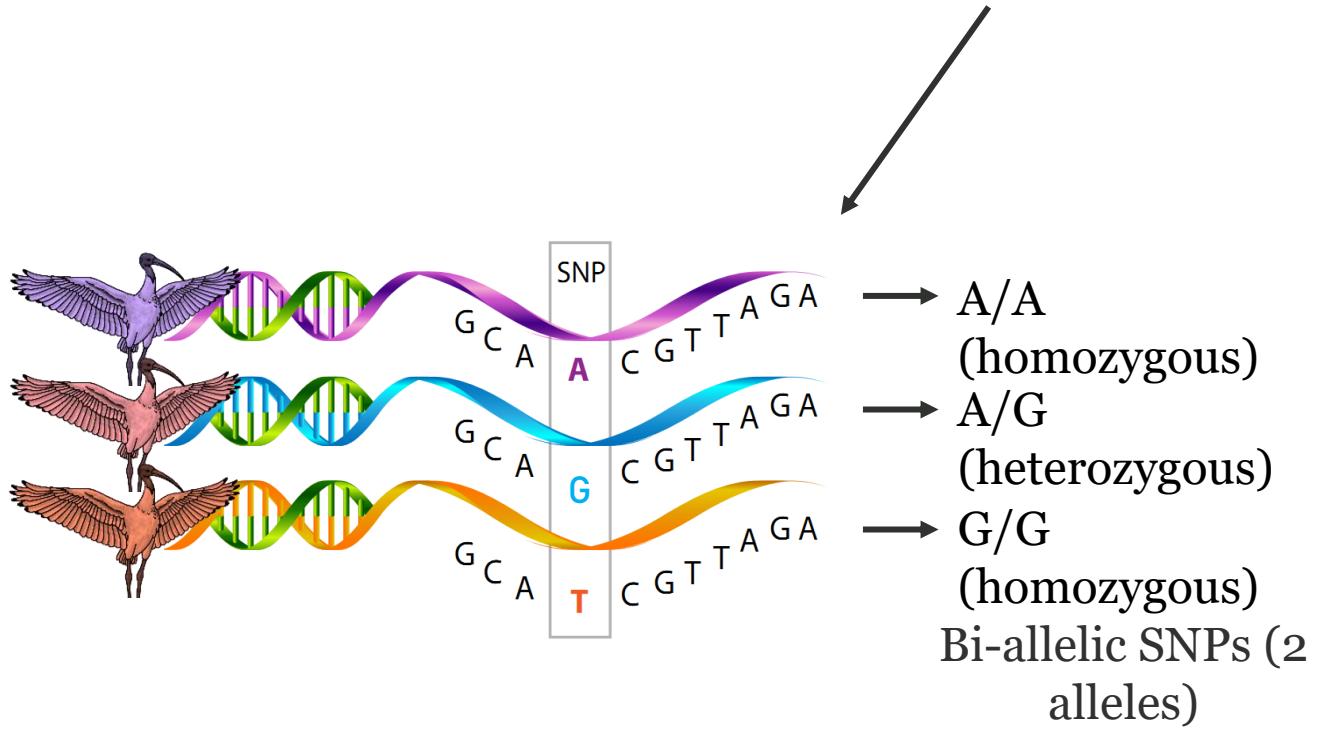
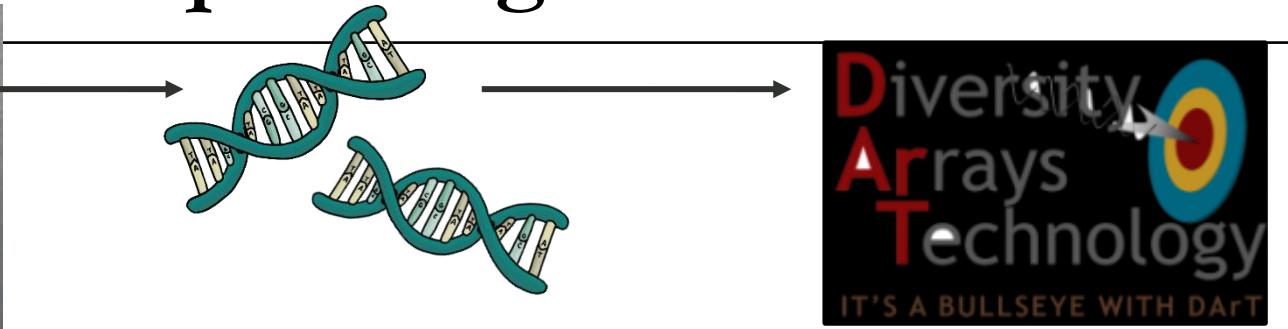
- ANSTO, University of NSW
- Australia wide citizen science project
- Feathers analysed using high resolution x-ray fluorescence
 - Changes in chemical composition over time
 - Different isotopic signatures for different wetlands
- Track waterbird movement over entire lifetime



DNA Extraction and Sequencing



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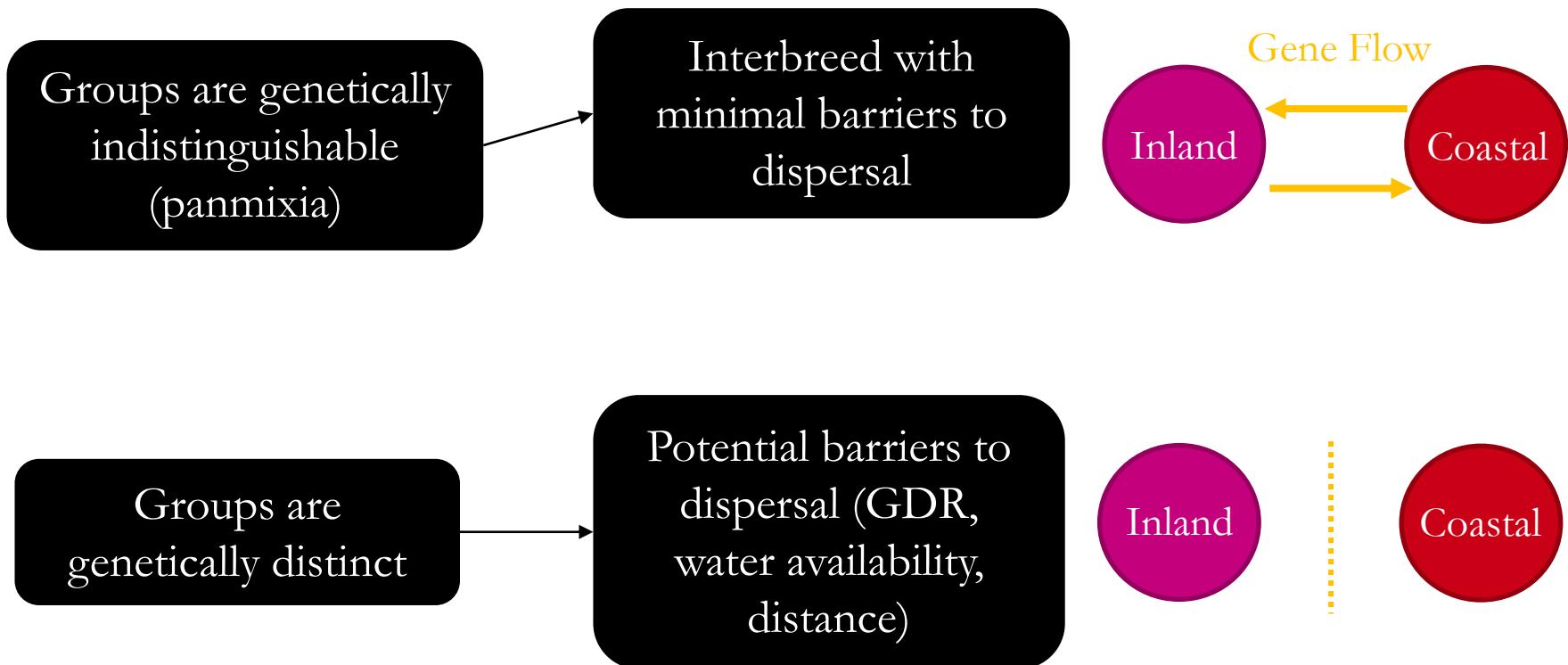


Methods



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Are there barriers to gene flow?



Results

Inland and Coastal colonies are not genetically distinct

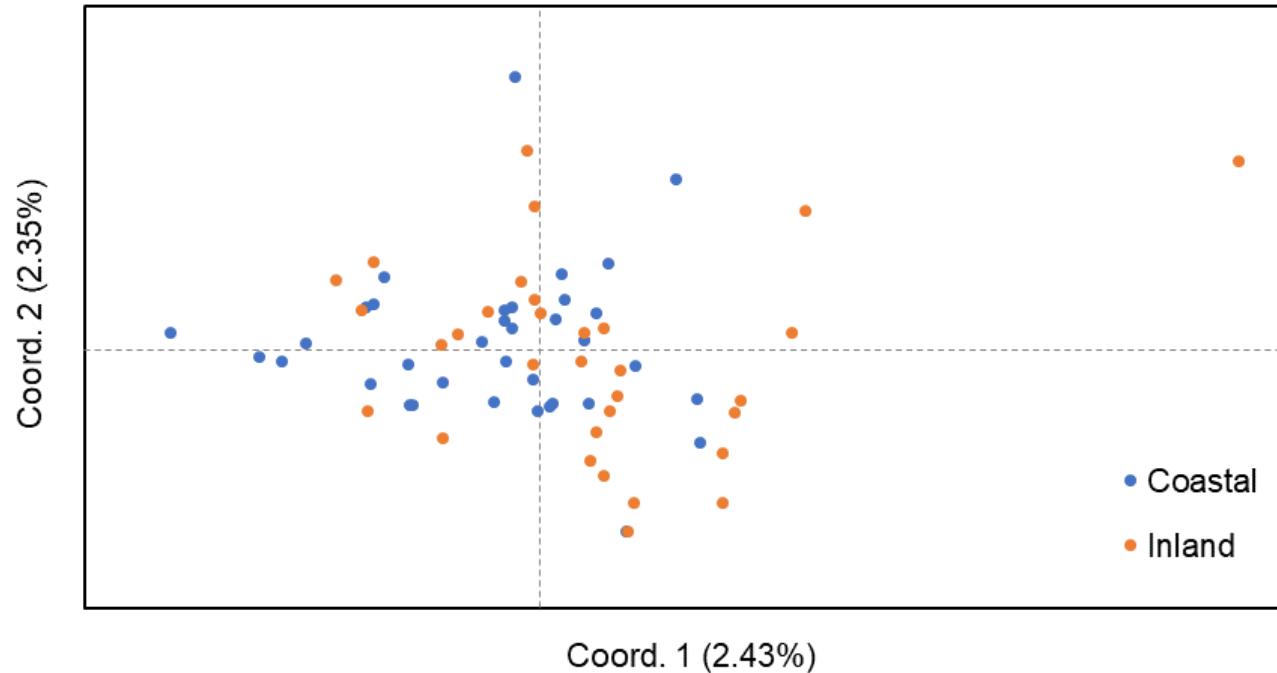
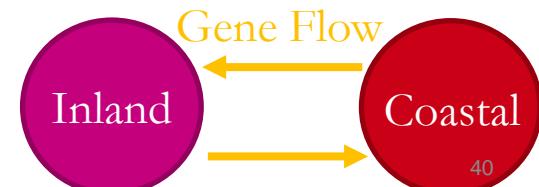


Figure 1: Plot of the 1st and 2nd principle coordinates identified by a principle coordinates analysis (PCoA)



Results

The GDR isn't a barrier to gene flow

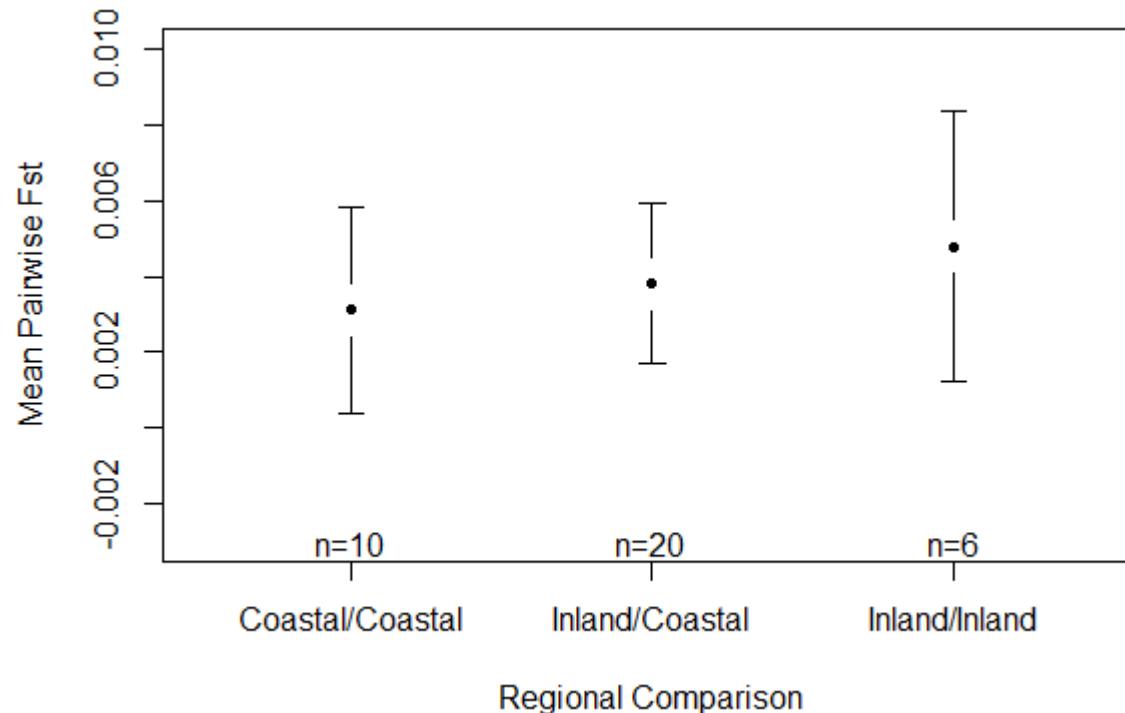


Figure 3: Mean pairwise F_{ST} grouped by region.



Results

Relatedness remains similar over large distances

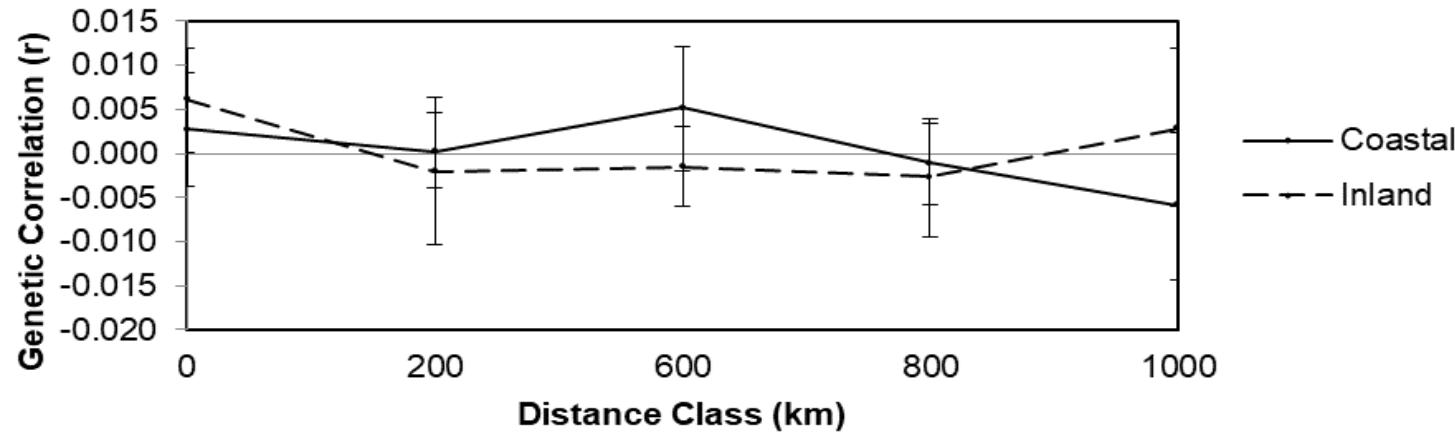
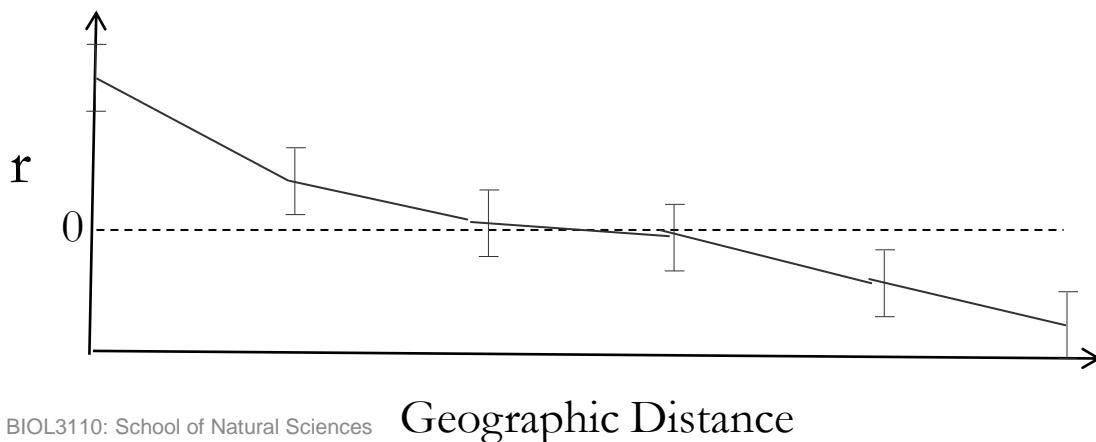


Figure 2: Mean genetic correlation (r) at different distance class bins

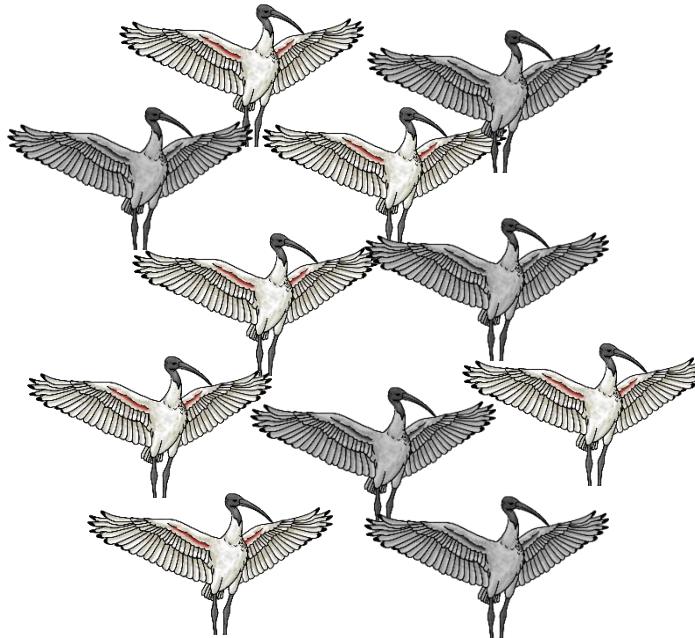


Methods

Effective Population Size

- N_e is the size of an idealised population that would contain an equivalent amount of genetic variation present in the study population

Not all individuals contribute genetic material equally to the next generation



Results

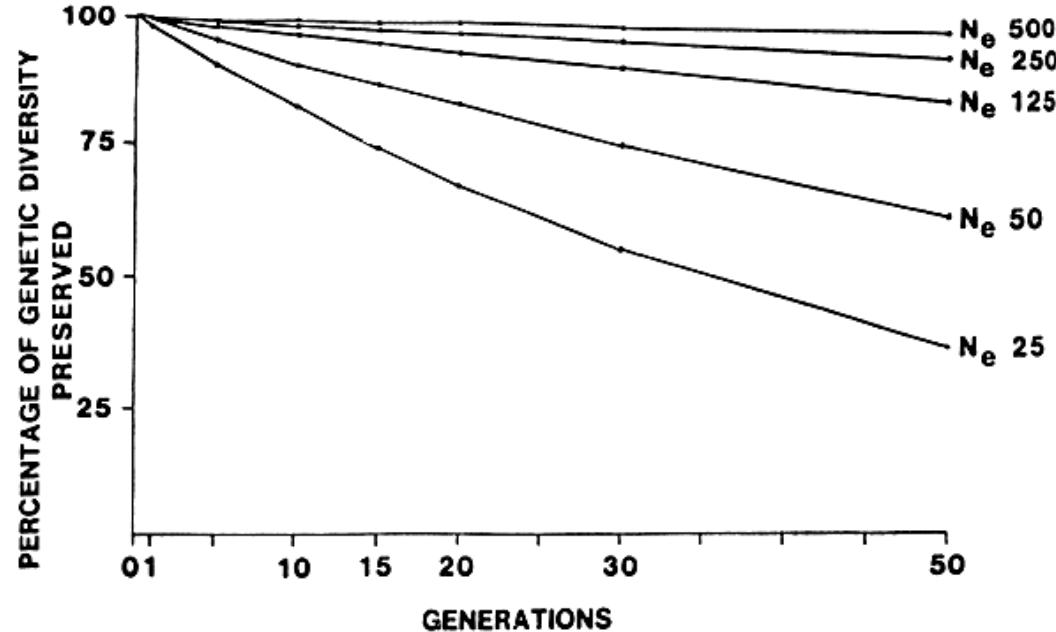


Large effective size

- Bias adjusted \hat{N}_e : 3448
 - Jack-knifed 95% CI: 1202, Infinite
 - Parametric 95% CI: 2138, 8820
- $N_e \geq 100$ to avoid inbreeding depression
- $N_e \geq 1000$ to maintain evolutionary potential

Methods

Forward Simulations



- Predict the impact of urban management on future genetic diversity
- Various annual reductions in effective size tested

Genetic diversity is maintained even in extreme culling scenarios

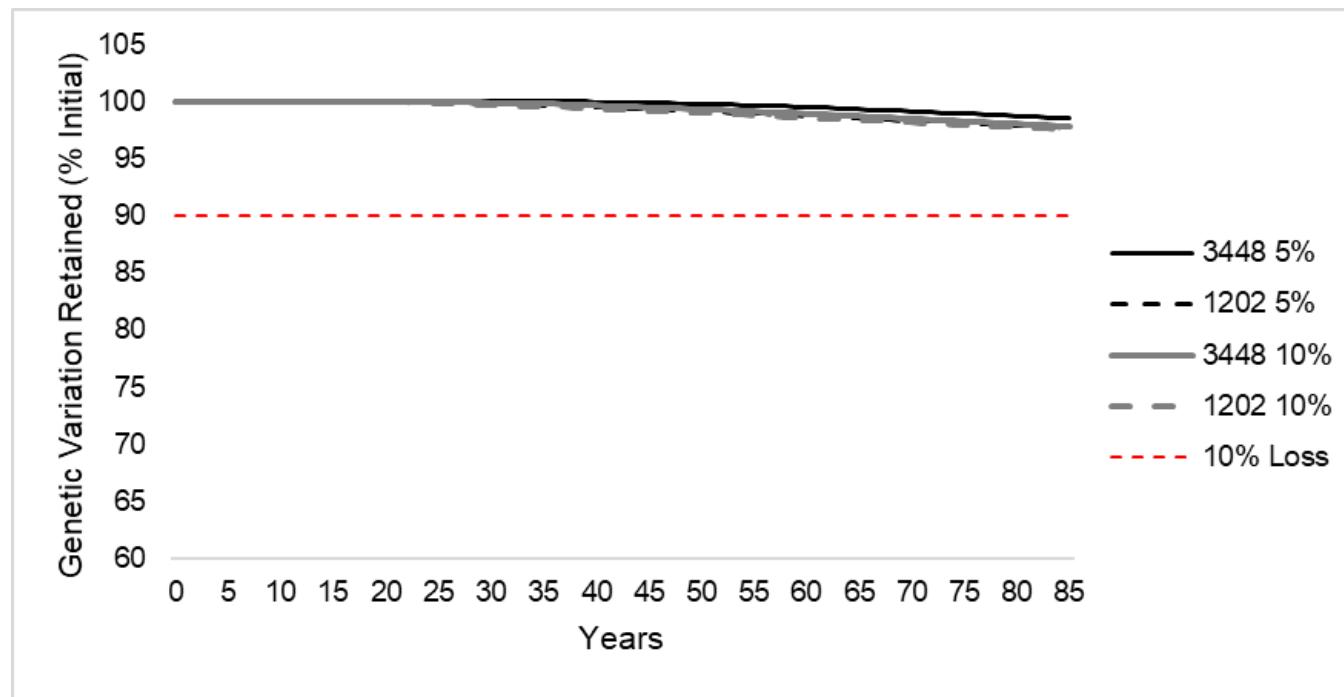


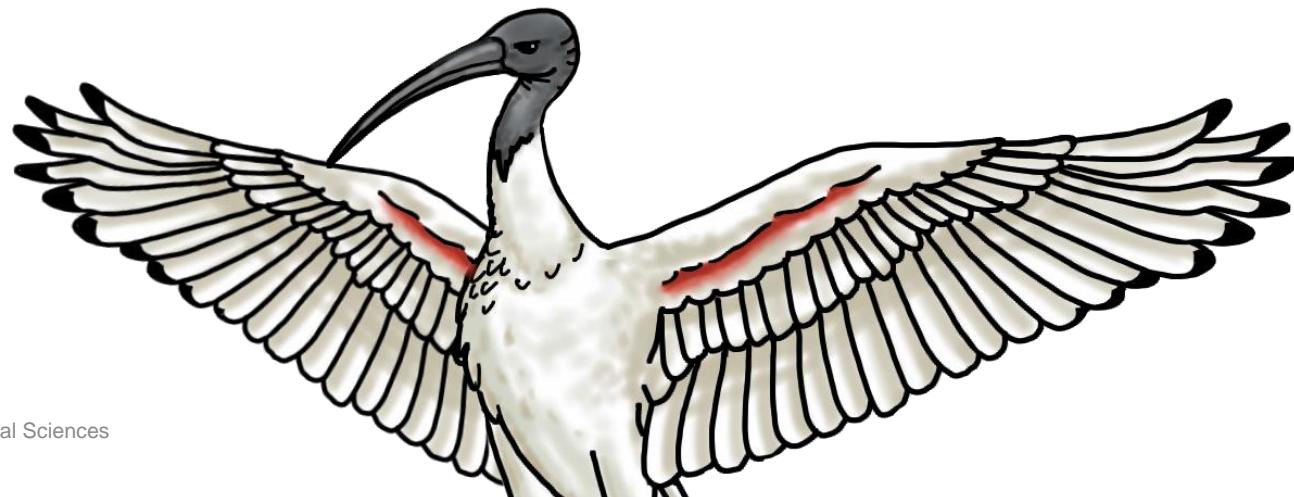
Figure 4: Retained diversity after 85 years, sustaining effective size at 650



IMPLICATIONS FOR MANAGEMENT

Evidence for coastal and inland ibis interbreeding

- No evidence of genetic partitioning
- High gene flow between urban centres and deteriorating inland wetlands
- Baseline to observe changes over time
 - Increasing urbanisation + sedentariness
 - Translocations to rehabilitated inland wetlands
 - Changes to management practices



Large Effective Sizes

- Well above the recommended sizes to avoid inbreeding depression and maintain evolutionary potential in the long term
- Low risk for the ibis



Management Impacts

- Under current management targets genetic diversity is unlikely to be negatively impacted
- High gene flow suggests urban management could impact declining inland colonies in other ways...



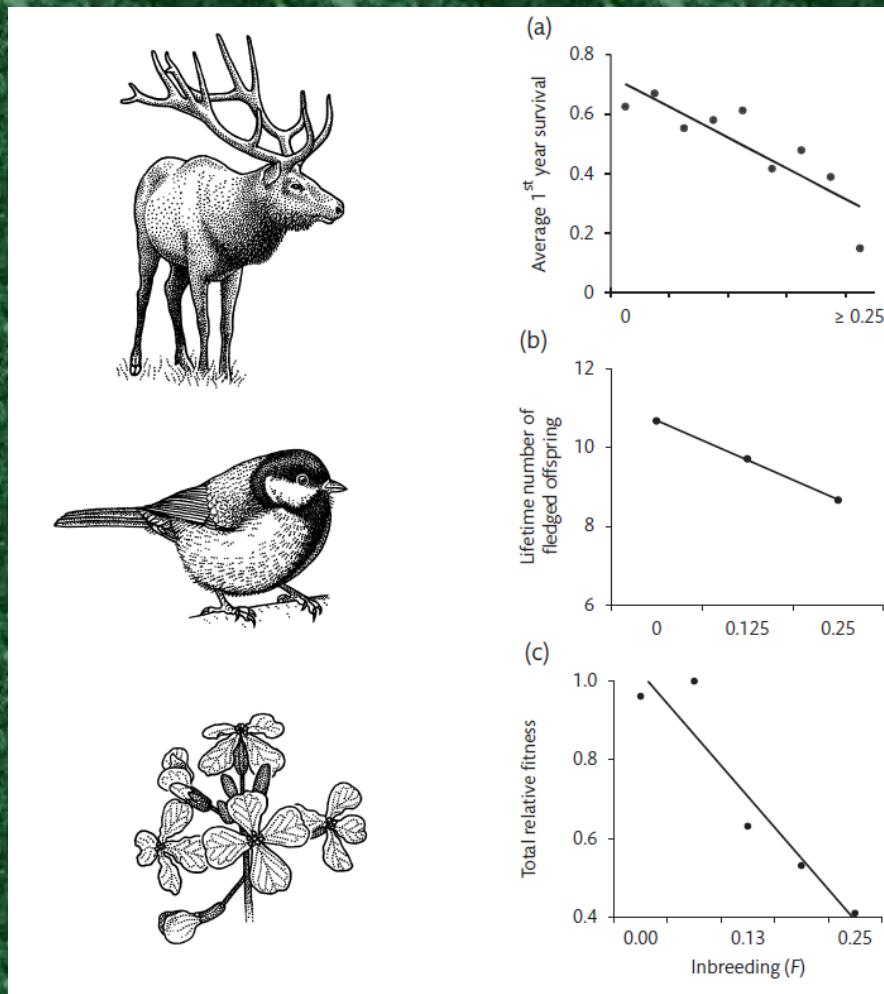


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Next – Inbreeding Depression (Dick Frankham)



Inbreeding depression



Assumed background:

Inbreeding

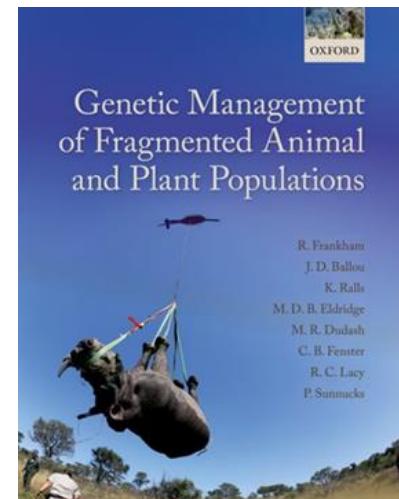
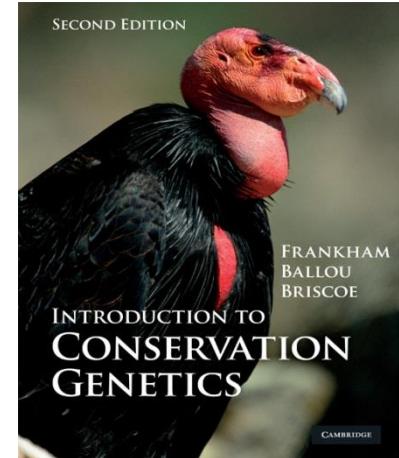
- Inbreeding is the mating of individuals related by descent
- Inbreeding increases homozygosity and exposes harmful recessives
- Inbreeding occurs through either non-random mating or small pop size
- Inbreeding is unavoidable in small isolated populations and accumulates over time
- Inbreeding coefficient (F) is Pr that 2 alleles in an indiv are i.b.d

This lecture: Inbreeding depression

- What is inbreeding depression (ID)?
- Why is ID important in conservation?
- How large are its effects?
- What are its characteristics?
- How does ID and its characteristics arise?
- How does it vary and change?
- How do we detect & measure it?
- What can we do about ID?

References: Text Ch 13

Frankham et al. (2017) GMFAPP Ch3



What is inbreeding depression?

Inbreeding depression is a reduction in mean due to inbreeding, especially for reproductive fitness characters

Inbreeding depression

Effects of inbreeding by selfing (I) versus outcrossing (O) on several characters in 57 species of plants (Darwin 1876)

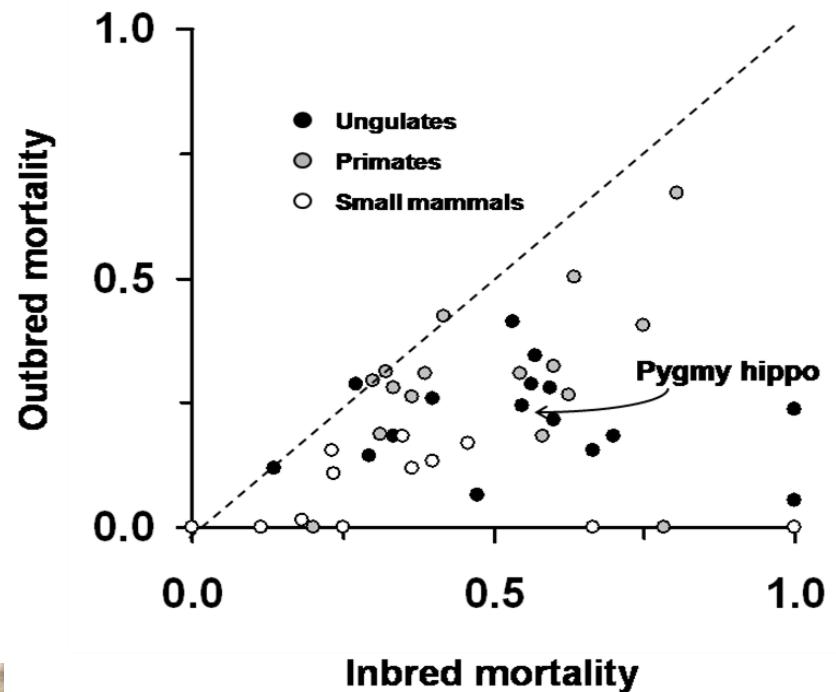
Characters	Species	O > I	O < I	Similar	Difference (O – I)
Height	54	57	8	18	13%
Weight	8	8	1	2	
Flowering time					
	32	44	9	5	
Seed production					
	23	26	2	5	41%

Inbreeding depression

Similar conclusions for laboratory and domestic animals and plants & humans

Inbreeding depression in captive mammals

- 41/44 mammal populations showed inbreeding depression
- Juvenile mortality 32% higher in progeny of full-sibs



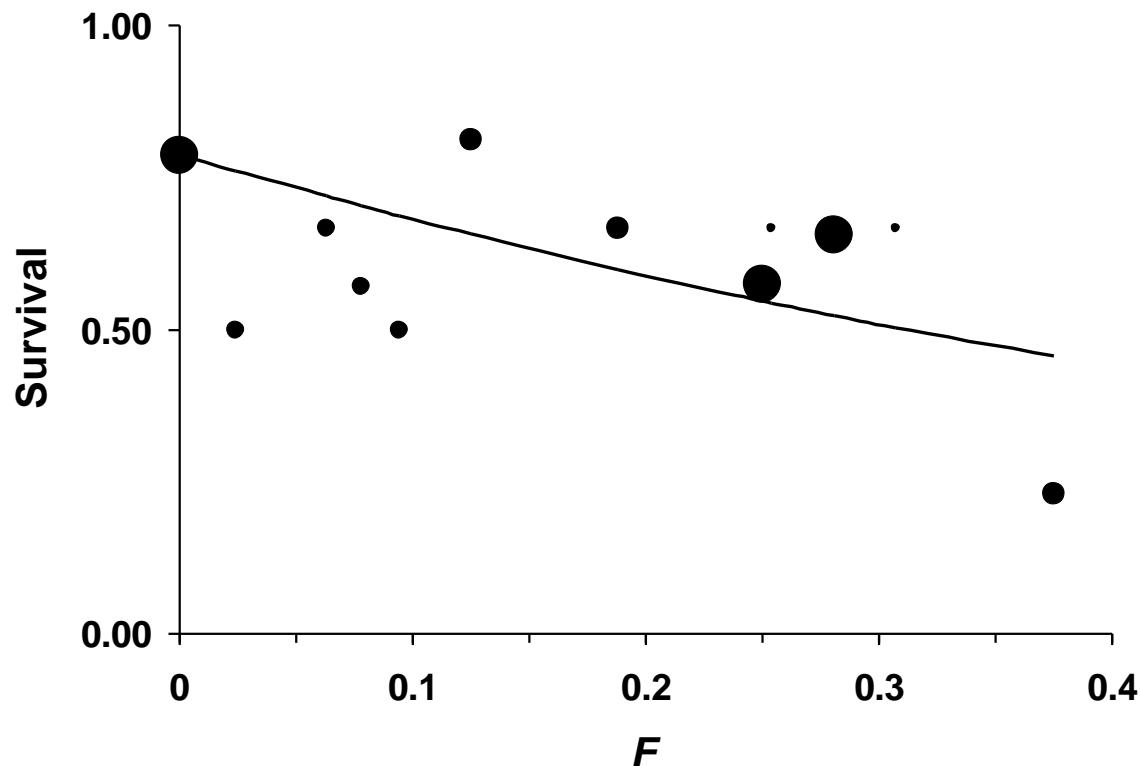
(Ralls & Ballou 83)



Pygmy hippos

Inbreds : 55% mortality
Outbreds : 25% mortality

Inbreeding depression

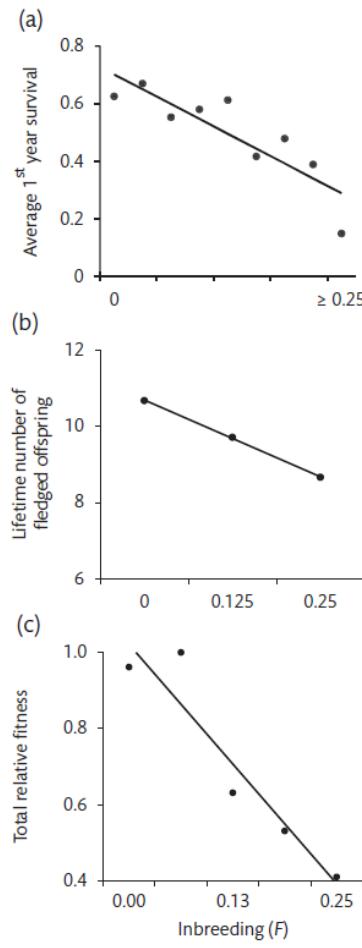
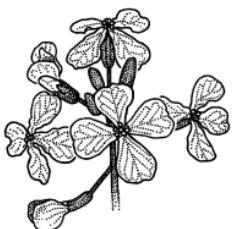
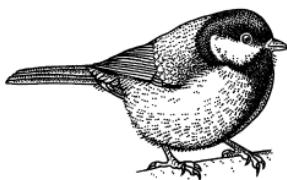


Inbreeding depression in the wild

- 141/157 inbreeding harmful (90%)
91.2% cases harmful for fitness
- 2 equal
- 14 inbreds best

(Crnokrak & Roff 99)

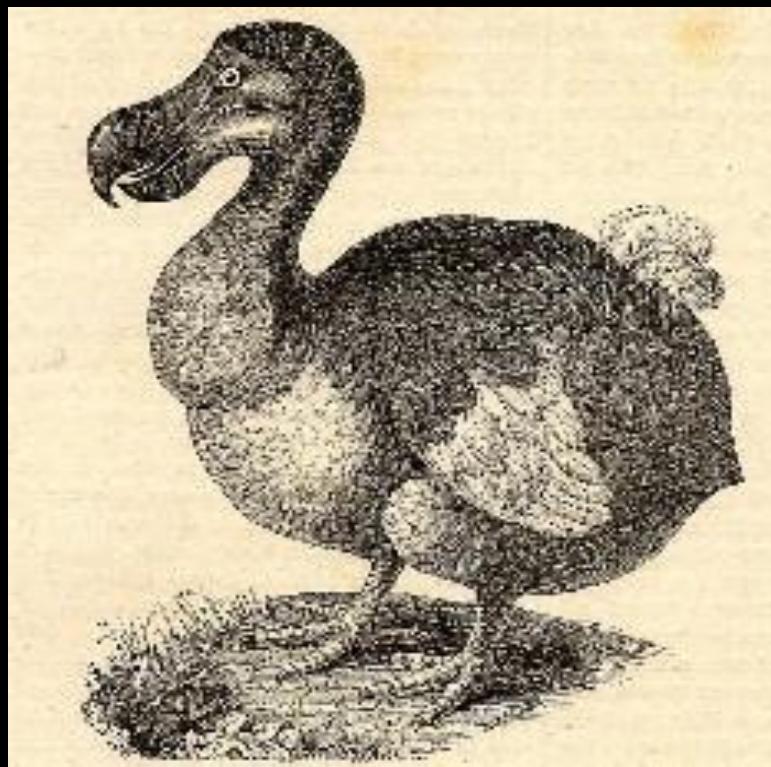
ID is ubiquitous



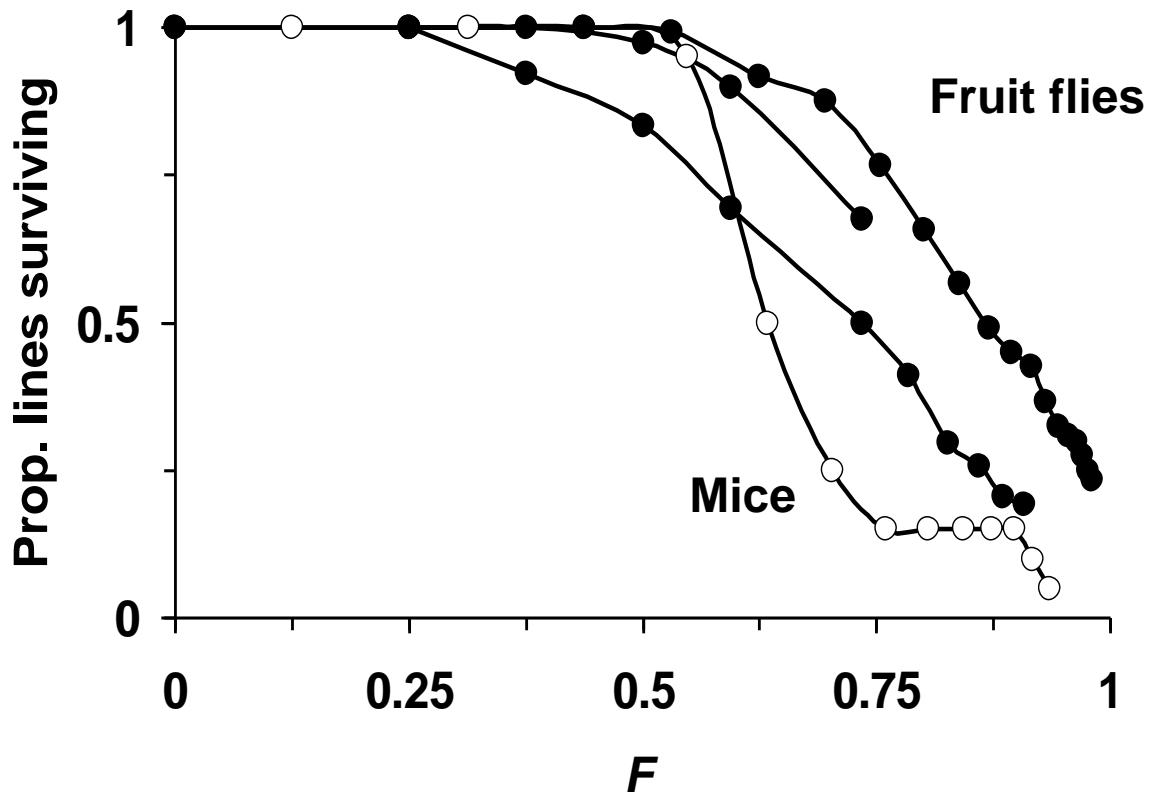
Why is ID important in conservation?

Inbreeding reduces reproductive fitness and adversely affects population persistence i.e. it increases the risk of extinctions

Inbreeding & extinction

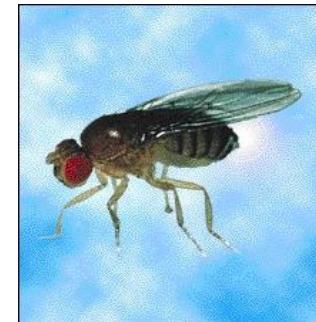


Inbreeding causes extinctions in small captive populations



Fruit flies

Mice



(Frankham 1995 CB)

Extinctions due to inbreeding in wild populations

- Butterflies I > O

(Saccheri et al. 98)

(Niemenen et al. 01)



- Plants I > O

(Newman & Pilson 97)

(Vilas et al. 2006)



Extinctions due to inbreeding in wild populations

- Butterflies
- Plants
- Are these general, or exceptional cases?

Computer projections for real species: +/- inbreeding depression



Computer projections for real species: +/- inbreeding depression

Predict without bias



Computer projections for real species: +/- inbreeding depression

Predict without bias

30 vertebrate species using VORTEX

- *ID* as found in the wild (6 haploid lethal equivalents)
- Included natural selection (purging): 50% lethals
- Isolated random mating populations

Computer projections for real species: +/- inbreeding depression

Predict without bias

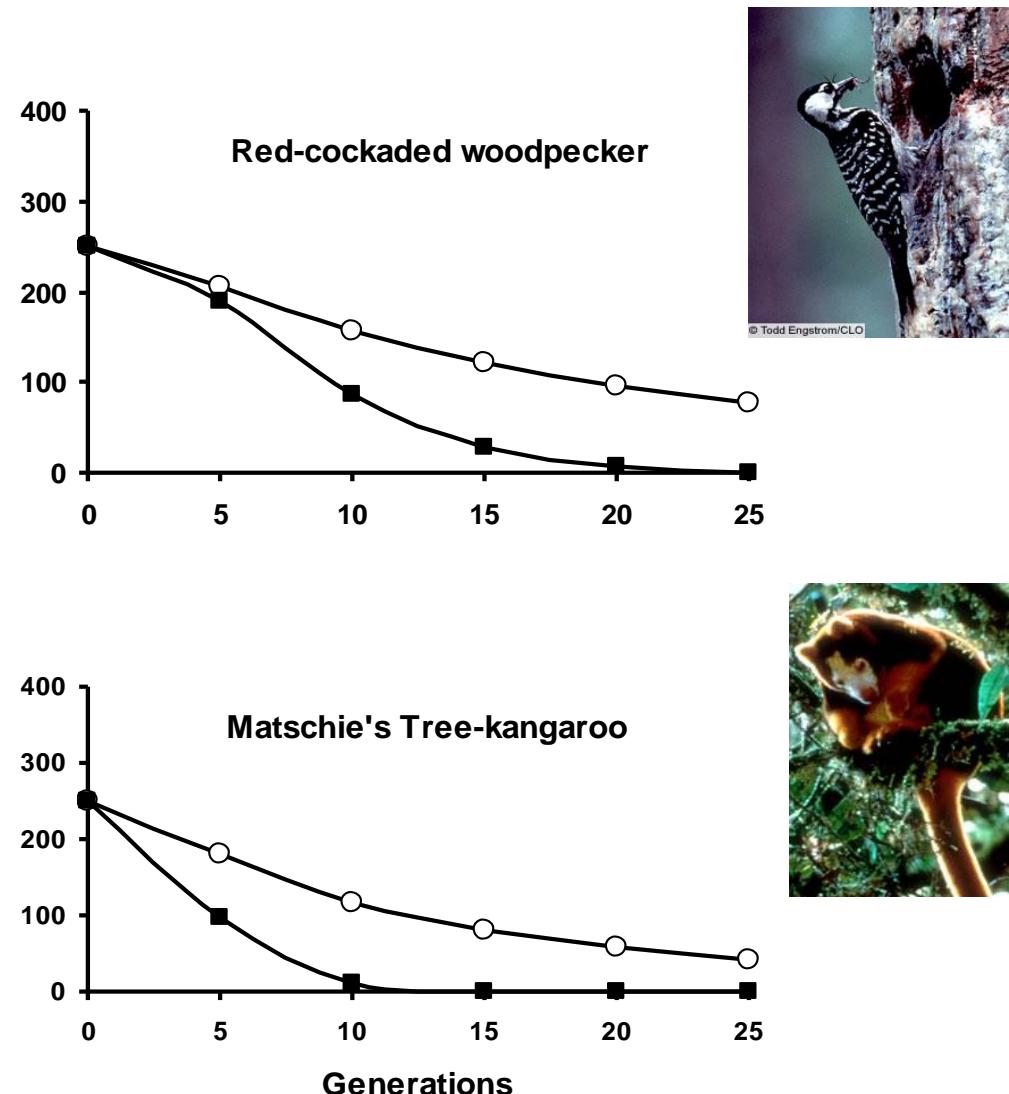
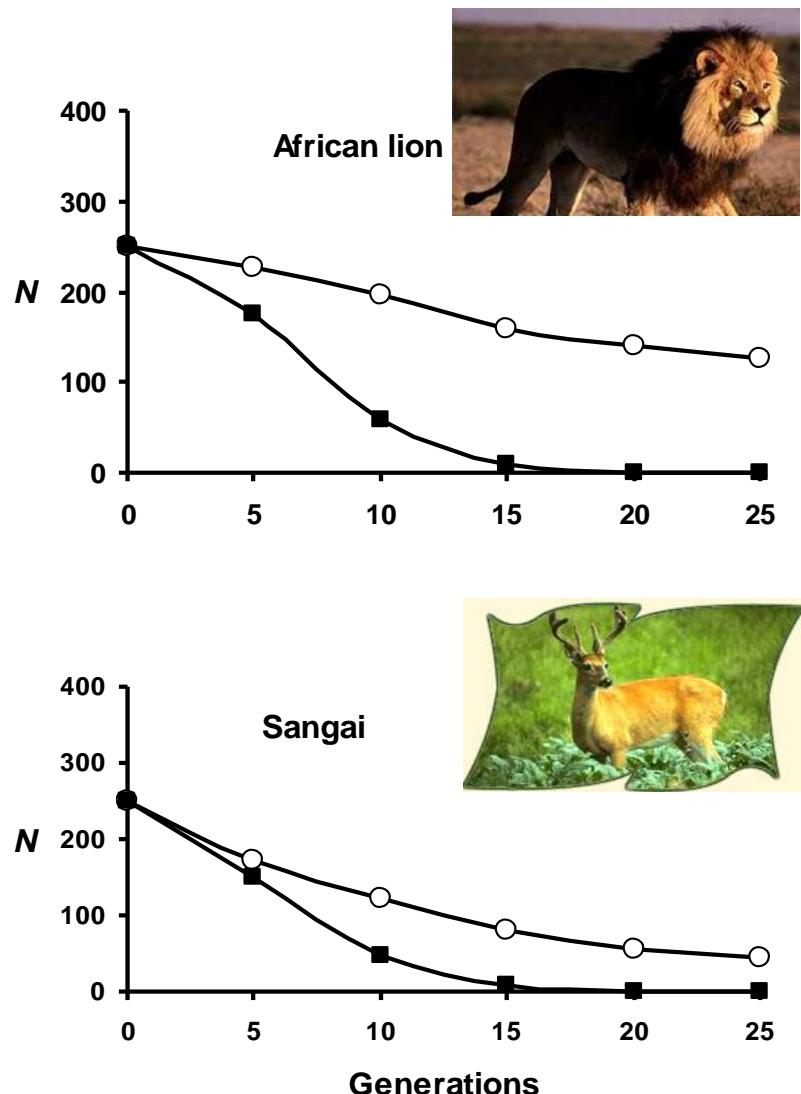
30 threatened species using VORTEX

- ID as found in the wild (12 lethal equivalents)
- purging: 50% lethals

Regimes based on IUCN categories:

- $N_{\text{initial}} = 50, 250 \text{ and } 1,000$ (~ CE, EN & VU)

1000 reps & all threatening factors included



Findings

- Inbreeding depression markedly decreased median times to extinction
 - 31% for $K = 100$
 - 39% for $K = 500$
 - 41% for $K = 2,000$
- Impacts were similar across major vertebrate taxa

(O'Grady et al. 2006 BC)

Results for butterflies and plants are not special cases:

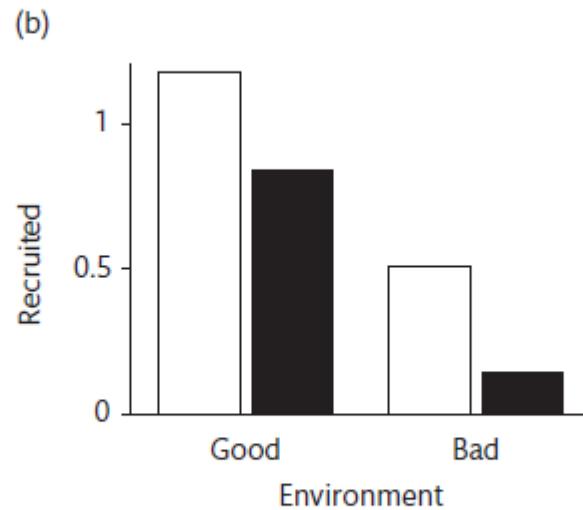
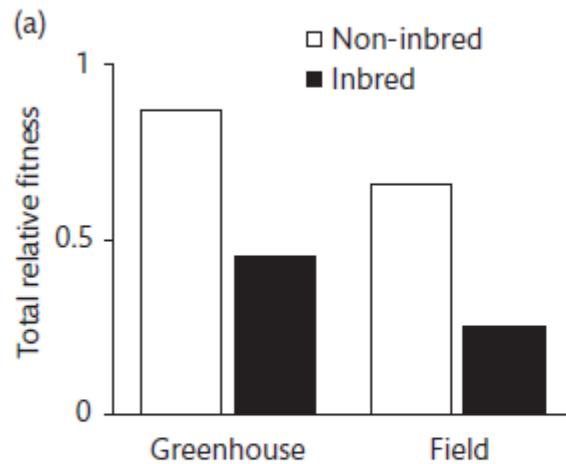
inbreeding generally increases extinction risk

What are the characteristics of ID?

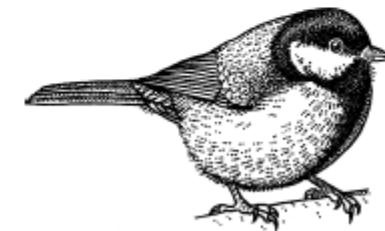
Characteristics of inbreeding depression

- Ubiquitous
- Natural outbreeders > inbreeders
- No ID in haploids or asexuals
- Fitness characters > those peripheral to fitness
- All components of reproductive fitness subject to ID
- Total fitness > its components
- Stressful/wild > benign/greenhouse or lab
- ID is highly variable (has a large stochastic components)

ID is greater in more stressful environments



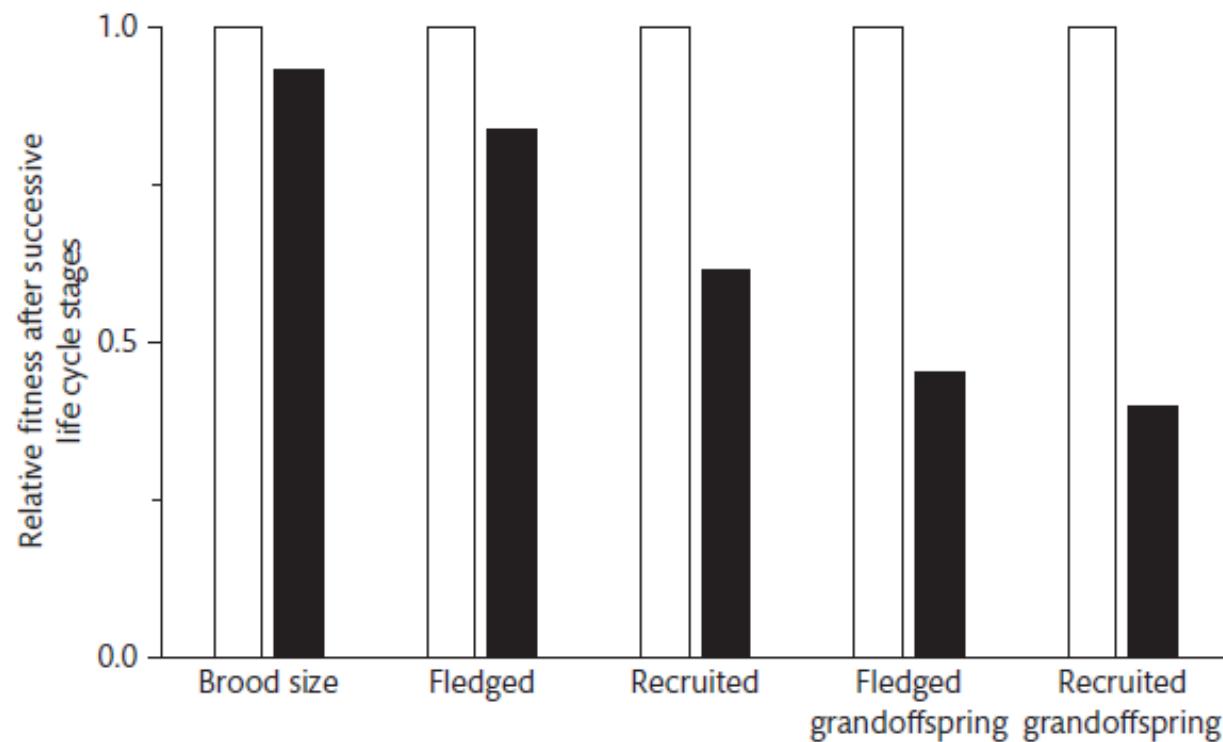
Rose pink plant (USA)



Great tit (UK)

$ID_{\text{stressful}} > ID_{\text{benign}}$ by 69%

Inbreeding affects all fitness components and accumulates across the life cycle



Devastating effects of FS inbreeding on total fitness in wild

Common name	ID %
Red deer	99
Collared flycatcher	94 ^a
Great tit	55
Song sparrow	79
Takahe	88
Deerhorn clarkia	100 ^a
Rose pink plant	38 ^a
Wild radish	56 ^a

How does ID and its characteristics arise?

Genetic basis of ID

- populations contain a load of harmful recessive alleles
- inbreeding increases the frequency of homozygotes and exposes recessive alleles in the phenotype
- this is occurring throughout the genome (~ 20K loci in vertebrates)

Impact of inbreeding on the mean of a population: single locus model

Genotype	Value	Genotype frequencies		Genotype frequency x value	
		Random mating	Inbred	Random mating	Inbred
A ₁ A ₁	a	p^2	$p^2 + Fpq$		
A ₁ A ₂	d	$2pq$	$2pq(1 - F)$		
A ₂ A ₂	$-a$	q^2	$q^2 + Fpq$		

Impact of inbreeding on the mean of a population: single locus model

Genotype	Value	Genotype frequencies		Genotype frequency x value	
		Random mating	Inbred	Random mating	Inbred
A ₁ A ₁	a	p^2	$p^2 + Fpq$	p^2a	$p^2a + Fpqa$
A ₁ A ₂	d	$2pq$	$2pq(1 - F)$	$2pqd$	$2pq(1 - F)d$
A ₂ A ₂	$-a$	q^2	$q^2 + Fpq$	$-q^2a$	$-q^2a - Fpqa$

Impact of inbreeding on the mean of a population: single locus model

Genotype	Value	Genotype frequencies		Genotype frequency x value	
		Random mating	Inbred	Random mating	Inbred
A ₁ A ₁	a	p ²	p ² + Fpq	p ² a	p ² a + Fpqa
A ₁ A ₂	d	2pq	2pq(1 - F)	2pqd	2pq(1 - F)d
A ₂ A ₂	-a	q ²	q ² + Fpq	-q ² a	-q ² a - Fpqa
		Means		$M_o = a(p - q) + 2pqd$	
				$M_F = a(p - q) + 2pqd - 2pqdF$	

$$M_F = M_o - 2pqdF$$

$$ID = 2pqdF$$

What do the terms mean?

$$ID = \sum 2pqdF$$

\sum = sum over all loci with segregating alleles
that are harmful when homozygous

more for total fitness than for components

What do the terms mean?

$2pq$ heterozygosity for harmful alleles

- no ID in haploids

- less with selfing

$2pqF$ loss of heterozygosity

- No ID in asexual species

- less ID in selfing species

- less ID in haplodiploids

What does the d term mean?

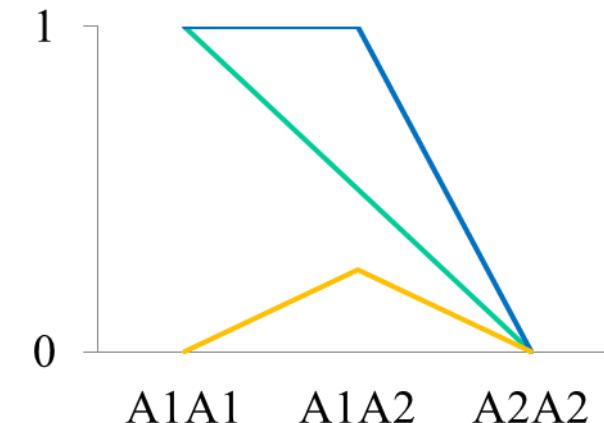
d = dominance of harmful alleles

ID when $d = +ve$ recessive on average or heterozygote adv

- directional dominance (as for fitness)

- not for stabilising seln (non-fitness)

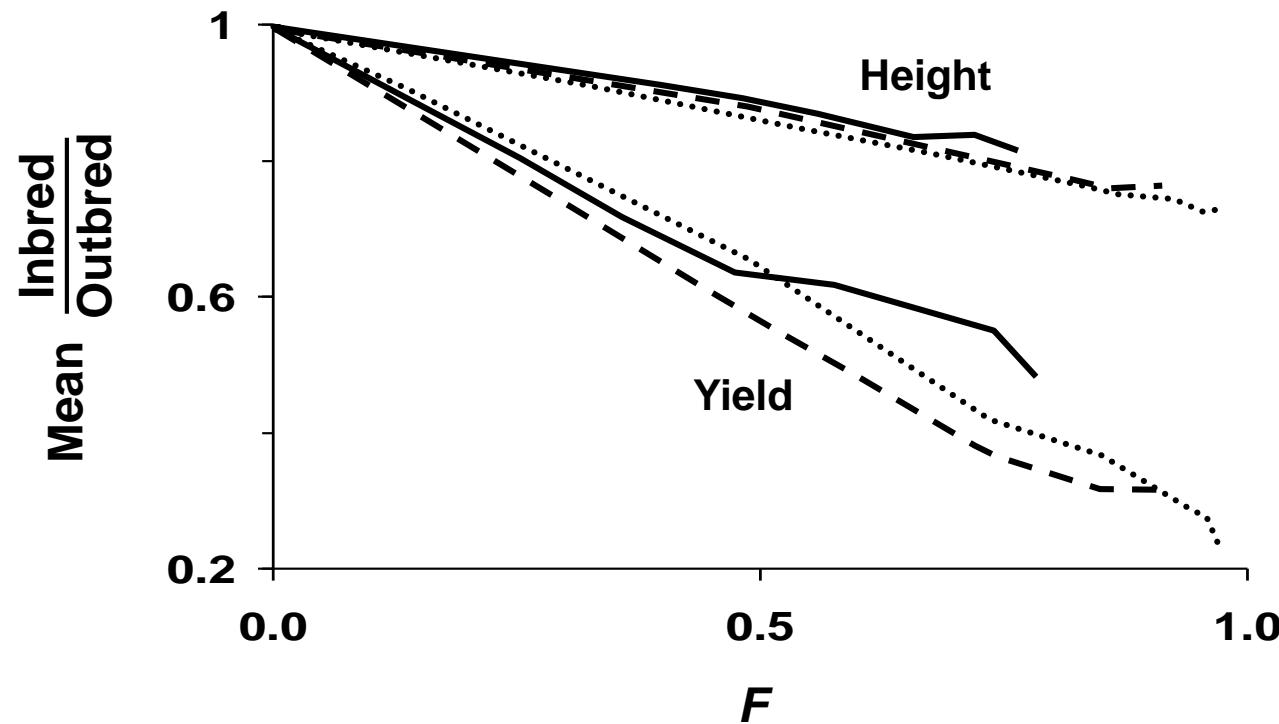
no ID when $d = 0$ hets intermediate



What does this equation predict?

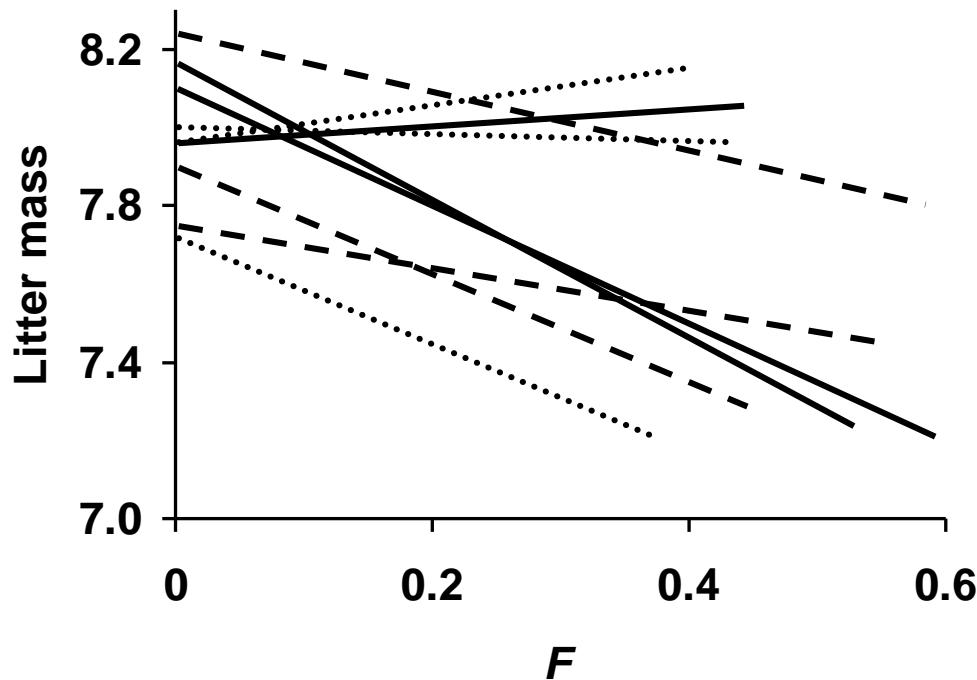
$$ID = \sum 2pqdF$$

predicts linear decline in fitness with F



Introduction to Conservation Genetics 2
Fig 13.4 ID in maize.ppt

How does ID vary and change?



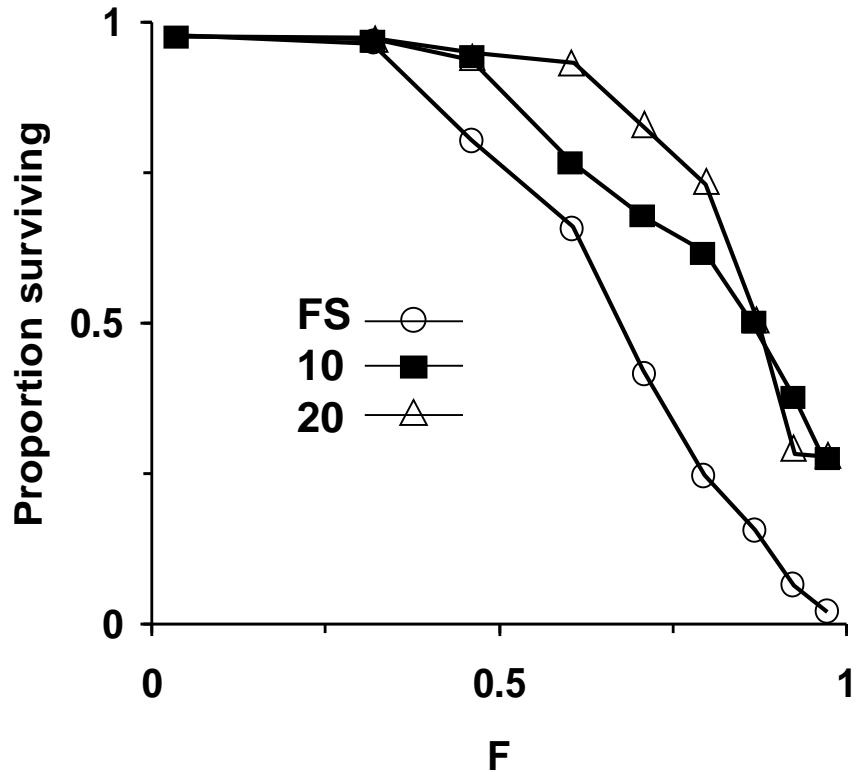
Introduction to Conservation Genetics 2
Fig 13.3 Inbreeding in *Peromyscus*.ppt

How does natural selection affect ID (purgling)?

Reduction in inbreeding depression caused by natural selection reducing frequency of harmful recessive alleles

Purging

- ID may be reduced by selection against harmful recessives under inbreeding
- Harmful recessives can be purged, but not loci with het advantage
- In small populations some harmful alleles become fixed through drift
- Purging more effective in large populations with some inbreeding (e.g. selfing)



Introduction to Conservation Genetics 2
Fig 13.5 F and Population Survival

How we detect and measure ID?

How do we detect ID?

- Compare inbreds & outbreds contemporaneously in same environment
- Compare I & O with same controls
- Regression of fitness on F



Measuring ID using delta (δ)

$$\delta = 1 - \frac{\text{fitness inbred offspring}}{\text{fitness of outbred offspring}}$$

Example: rose pink plant

Total fitness O = 0.99; I = 0.25*

$$\delta = 1 - \frac{0.25}{0.99} = 0.747$$



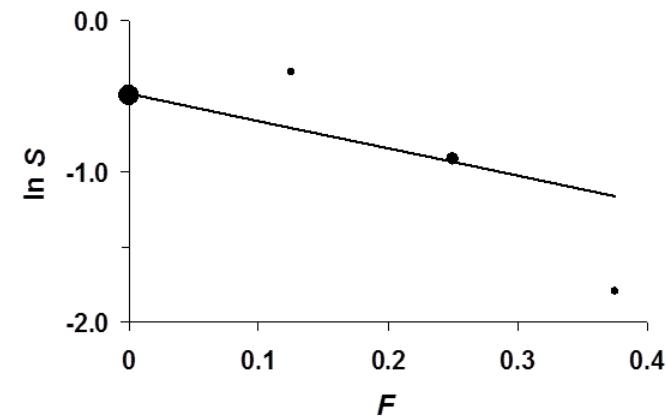
Measuring ID using lethal equivalents (B)

Regress $\ln(\text{survival})$ on F

$$S_l = e^{A - BF}$$

$$\ln S = A - BF$$

$$= -0.48 - 1.80 F \text{ (Okapi)}$$



B = haploid lethal equivalents = 1.80

$2B$ = diploid lethal equivalents = 3.60



Extent of inbreeding depression (lethal equivalents)

- Captive mammals juvenile survival $B = 1.57$
FS have 32% lower juv survival ($1 - e^{-FB}$)
- Verts & plants total fitness in wild $B \sim 6.85$
FS have 82% lower total fitness than outbreds
- Why do we need LE values?
 - To predict ID for any F values.

Extent of inbreeding depression for total fitness in the wild (lethal equivalents)

Common name	Genus and species	δ %	L.E.
Red deer	<i>Cervus elaphus</i>	99	18.7
Collared flycatcher	<i>Ficedula albicollis</i>	94 ^a	7.5 ^a
Great tit	<i>Parus major</i>	55	3.2
Song sparrow	<i>Melospiza melodia</i>	79	6.2
Takahe	<i>Porphyrio hochstetteri</i>	88	8.0
Deerhorn clarkia	<i>Clarkia pulchella</i>	100 ^a	39.2 ^a
Rose pink plant	<i>Sabatia angularis</i>	38 ^a	1.9 ^a
Wild radish	<i>Raphanus sativus</i>	58 ^a	3.3 ^a

What can we do about inbreeding depression?



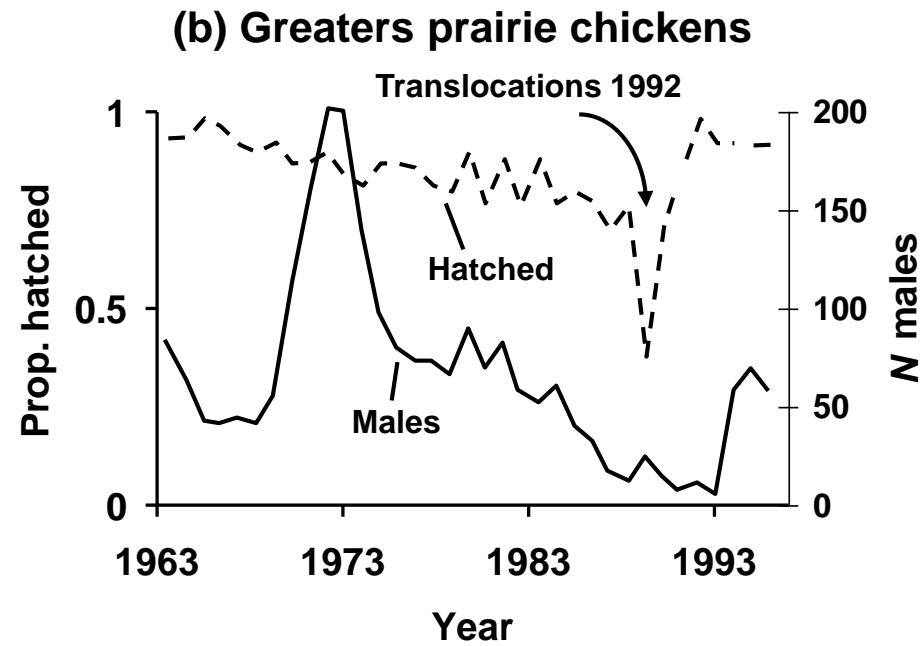
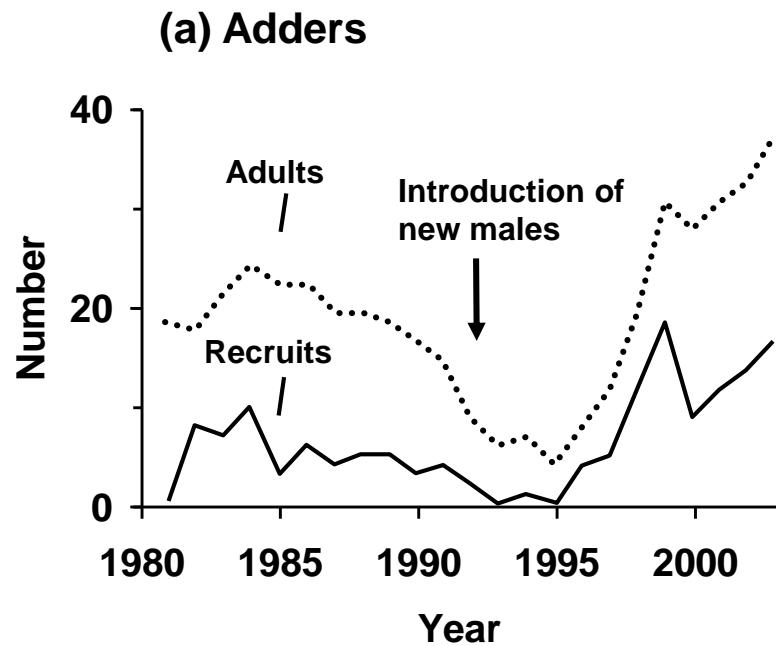
Genetic rescues of small inbred populations by outcrossing



SPECIES
SPOTLIGHT...



Genetic Rescue



Genetic rescue meta-analysis: How consistent? How large?

SPECIES
SPOTLIGHT...



Consistency of genetic rescue:

(Frankham 2015)

- Data: $145 +: 2 =: 9 - (156)$
- χ^2 test of $145: 9$ for equality = 139.1 ***
- **Highly consistently beneficial effects**

Magnitude of GR: composite fitness in outbreeding species

- Data: 67 cf
 - Median benefits
Wild 165%: Captive 51%
(likely underestimates)
Benefits persisted across generation for outbreeders
- Have been very few genetic rescues. Why?

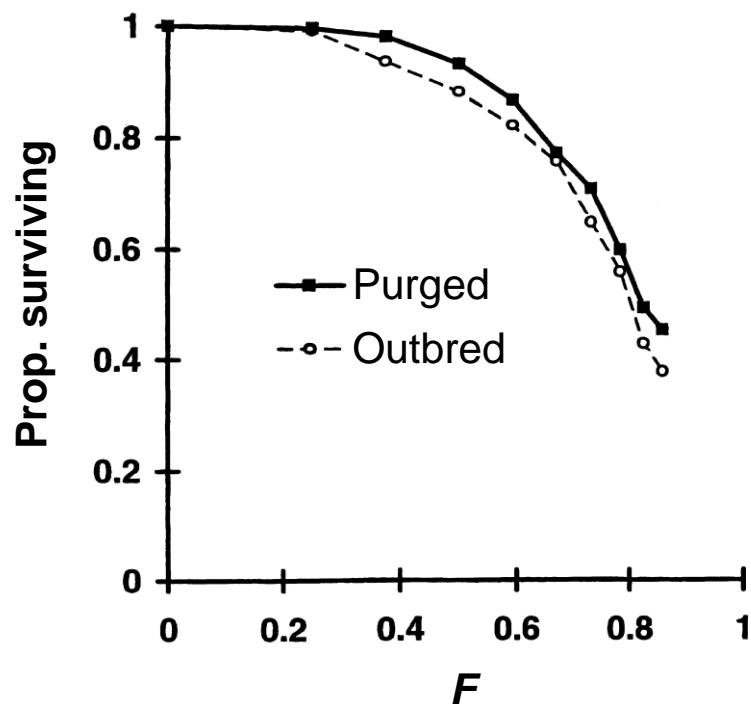
Messages

- Inbreeding reduces reproductive fitness (ID) and increases extinction risks
- This arises because inbreeding makes harmful alleles homozygous
- ID increases with F , is greater in more stressful environments & is greater in outbreeding than inbreeding species
- Natural selection can reduce ID by removing harmful recessive alleles
- Populations can usually be rescued from ID by outcrossing (genetic rescue)

Questions?

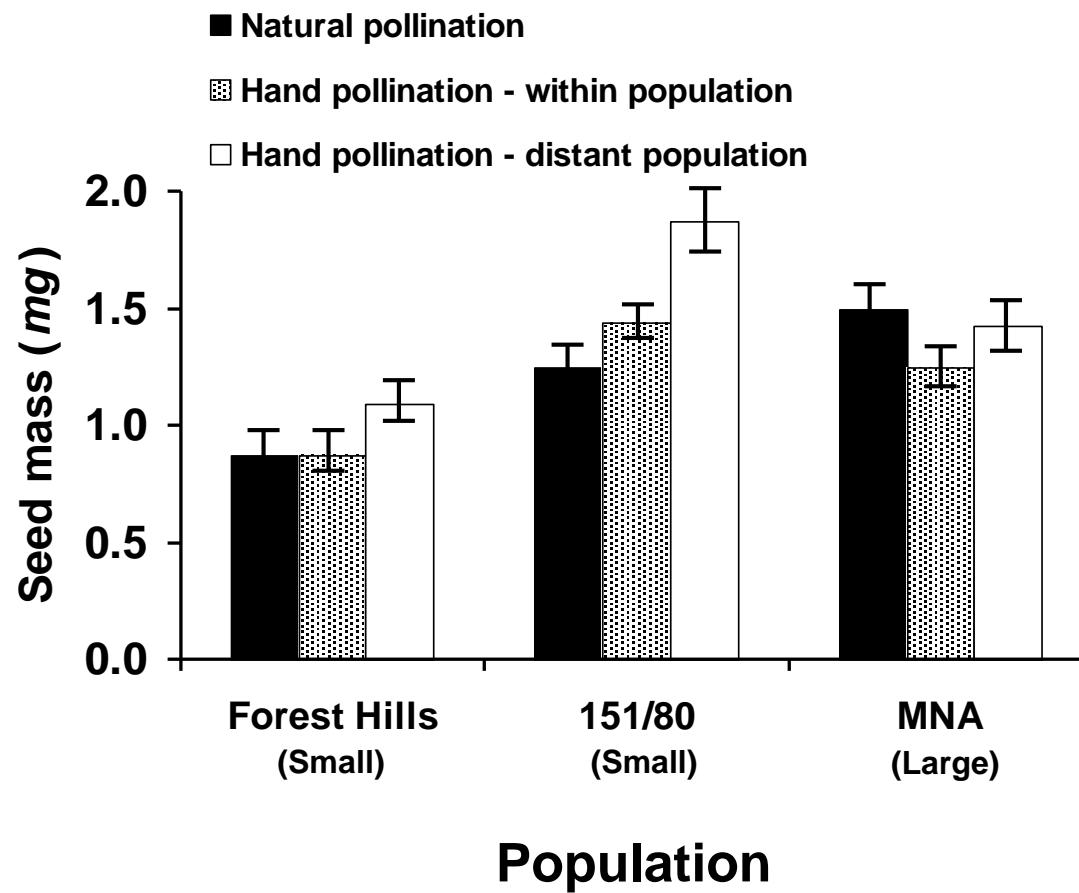
Variables affecting magnitude of genetic rescue

- Inbreeding (ΔF)
- Stressful > benign env
- Br system (outcrossing > selfing)
- Immigrants outbred > inbred
- Major taxa (vert, invert & plants) ns
- (ploidy; purging)

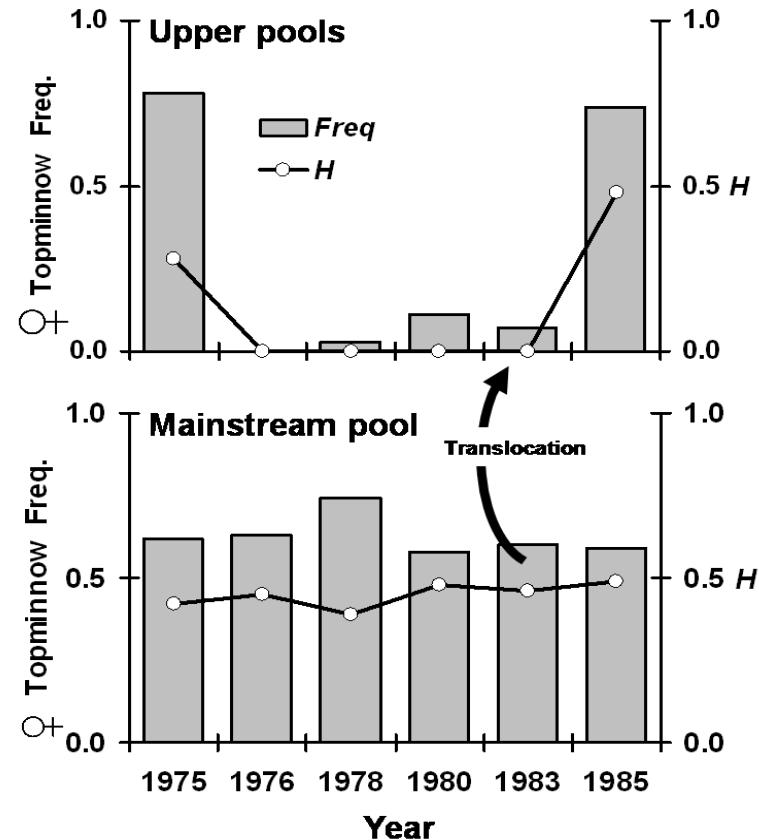
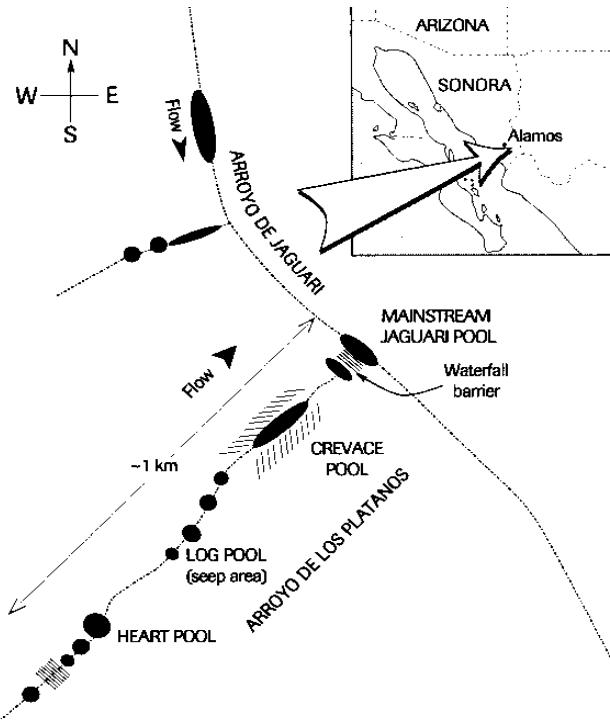


Introduction to Conservation Genetics 2
Fig 13.6 Purged and outbred.ppt

Alleviating inbreeding depression by outcrossing (scarlet gilia)



Case study: Inbreeding depression & rescue in desert topminnow fish (Vrjenhoek, 1994)



BIOL3110

Genetically viable populations

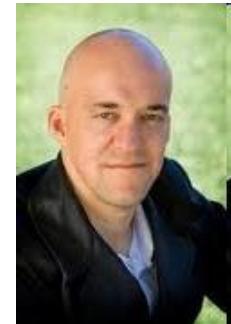
R. Frankham



This lecture: Genetically viable populations

- Why do we need to define the size of genetically viable populations?
- How large to they need to be?
 - Avoid fitness loss in short term?
 - Retain evolutionary potential in perpetuity?
 - Quantitative genetic variation
 - Single locus diversity
 - Avoiding accumulation of harmful mutations?
- How large are populations in practice?
- Captive populations: a compromise
- Fallacy of small isolated surviving populations
- Reference: Text Ch15*
- + Frankham et al. (2014) Biological Conservation 170, 56-63

Reference



Biological Conservation 170 (2014) 56–63



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journal homepage: www.elsevier.com/locate/biocon



Perspective

Genetics in conservation management: Revised recommendations for the 50/500 rules, Red List criteria and population viability analyses



CrossMark

Richard Frankham ^{a,b,*}, Corey J.A. Bradshaw ^c, Barry W. Brook ^c

Why do we need to define the size of genetically viable populations?

- Resources for conservation are limited
- Opportunistic funding
- *Crisis discipline*: require decisions to be made promptly with limited information
- Need for rules of thumb e.g. IUCN Red List, 50/500 etc

Resources limited

- Captivity
 - 2K species require captive breeding
 - Space in zoos for ~ 1K species
- Wild
 - Severe shortage of habitat & \$
 - Even the largest reserves are too small for large species

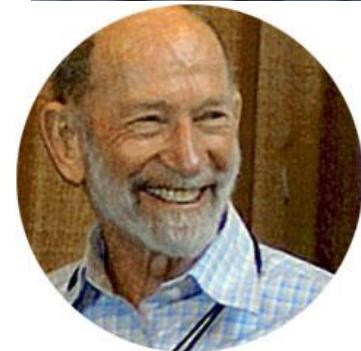


How large must population be
to retain their genetic health?

How large must populations be to retain genetic ‘health’?

50/500

Goal	N_e	References
1. Retain fitness in short term by avoiding ID	50	Franklin (1980); Soule (1980)
2. Retain evolutionary potential in perpetuity	500	Franklin (1980)



1. How large must isolated populations be to retain fitness in the short-term by avoiding ID?

Retaining reproductive fitness: avoiding ID in short-term

(Franklin 1980; Soulé 1980)

- Opinion of animal breeders
 - short term $N_e = 50 \sim F = 5\%$



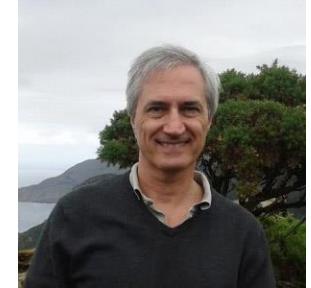
What evidence has accumulated since 1980? avoiding ID in short-term

Exptal data

- House flies lab
 - 14% ID for $N_e = 90$ for 5G* (Bryant et al. 99)
- Plants wild
 - $N_e = 50$ results in 16% ID over 5 gens ($F = 5\%$)
(3.4 haploid LE)
 - $\Delta F = 4\%$ lowered fitness by 79% & increased extns from 25% to 69% (Newman & Pilson 97)
- Vertebrates wild (7.6 haploid LE)
 - $N_e = 50$ results in 32% ID over 5 gens ($F = 5\%$)
(O'Grady et al. 2006; Frankham et al. 2022)



Realistic simulations



Simulations

- $N_e \sim 70$ required to avoid 10% ID
(Caballero et al. 2016)

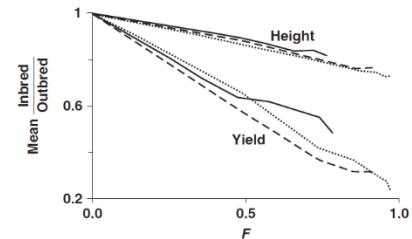
Likely an underestimate

- assumed 6 LE
- actual median 7.6 in vertebrates (Frankham et al. 2022)
- maternal ID not included in half of estimates

Revised guidelines for retaining fitness in wild

(Frankham et al. 2014)

- Specify short term = 5G
- Linear decline in fitness with F , so can't totally avoid ID keep to <10%
- With 7.6 LE $N_e = 179$ required, but allowing for some mild purging
- In plants with LE = 3.4, $N_e = 79$ needed



Recommend

$N_e \geq 100$ over 5G required to keep ID < 10% in wild

What are N_e in practice?

What are N_e of TH sp in practice?

- Captive pops of TH sp
Av $N_e \sim 33$
- Wild TH sp: IUCN criterion D
 - CE $N_e \leq 8$
 - EN $N_e \leq 39$
 - VU $N_e \leq 156$
- In many thr sp N_e is too small to prevent ID

N for de-listing thr species: wild

Species	N for de-listing	N_e
USA 475 sp (vert, inv & pl)	2,400	~ 240
17 delisted sp	2,360	~ 236



2. How large must isolated populations be to retain the ability to evolve in perpetuity?

Retaining evolutionary potential

(Franklin (1980))

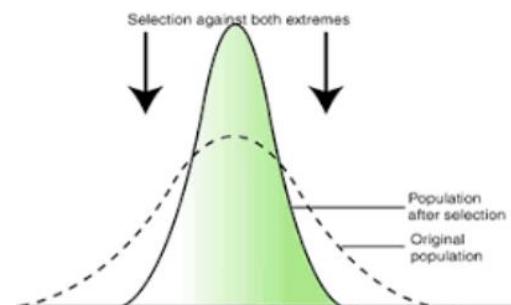


Assumptions:

1. Quantitative genetic variation
2. Heterozygosity not allelic diversity
3. Characters peripheral to fitness

Retaining evolutionary potential

- Quantitative genetic variation
- Heterozygosity not allelic diversity
- Equilibrium: mutation & drift
 - $N_e = 500$ (Franklin 1980)
- Equilibrium: mutation, drift & stabilising selection (Lande & Barrowclough 1987)
 - $N_e \sim 500$



Retaining evolutionary potential: derivation

Mutation-drift equilibrium (Franklin 1980)

$$\Delta V_A = V_m - V_A / (2N_e) = 0$$

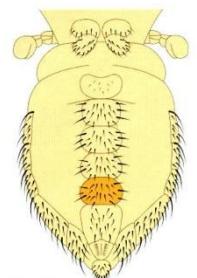
$$N_e = V_A / 2V_m$$

Substituting $V_m \sim 10^{-3} V_E$ per generation

$$N_e = V_A / [2 \times 10^{-3} \times V_E] = 500 V_A / V_E$$

With $V_A / V_E \sim \frac{h^2}{1 - h^2} = 1$, $(h^2 = 0.5)$

$$N_e = 500$$



What has changed since 1980: evolutionary potential?

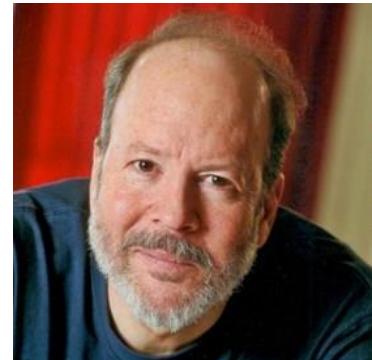
Adjusting for 90% harmful mutations (Lande 1995)

$$V_m = 10^{-4} V_E \text{ (~ plant fitness in wild)}$$

$$N_e = V_A / [2 \times 10^{-4} \times V_E] = 5000 V_A / V_E$$

Assuming $V_A/V_E = 1$ ($h^2 = 0.5$),

$$N_e = 5,000$$



With $V_A / V_E = 1/4$, ($h^2 = 0.2$) (Franklin & Frankham 1998)

$$N_e = 1,250$$

What has changed since 1980? evolutionary potential

- Other quantitative genetic models
 - $N_e > 10K$ (Keightley & Hill 1987)
 - $N_e = 10K \sim \infty$, 1K close (Weber & Diggins 1990)
 - $N_e \geq 1K$ (Lynch & Lande 1998)
 - N_e a few K sufficient (Willi et al. 2006)

$N_e = 500$ inadequate

What has changed since 1980? evolutionary potential

However, we should consider

Quantitative genetic variation for total fitness,
not for peripheral traits

Retaining evolution potential for fitness: theory

- Mildly harmful

$$N_e \geq 1K$$

(Falconer & Mackay 1996; Bataillon & Kirkpatrick 2000)

- Lethals

$$N_e \geq 1K$$

(Nei 1968; Hedrick 2002)

- Balancing selection

Heterozygote advantage $N_e \geq 1K$

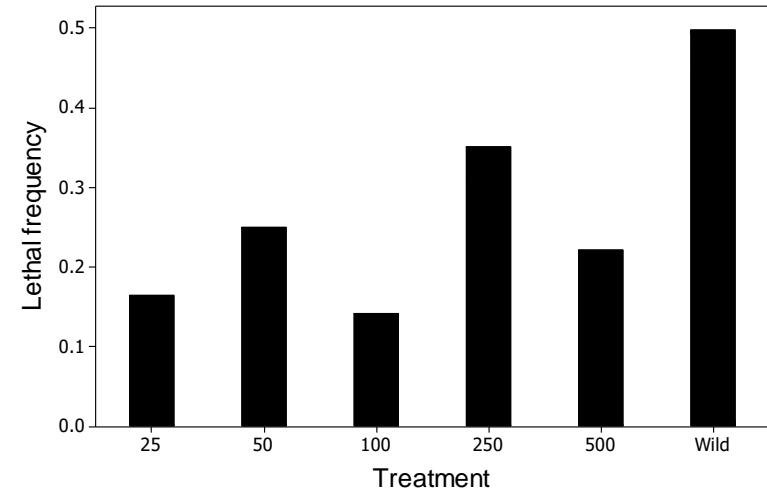
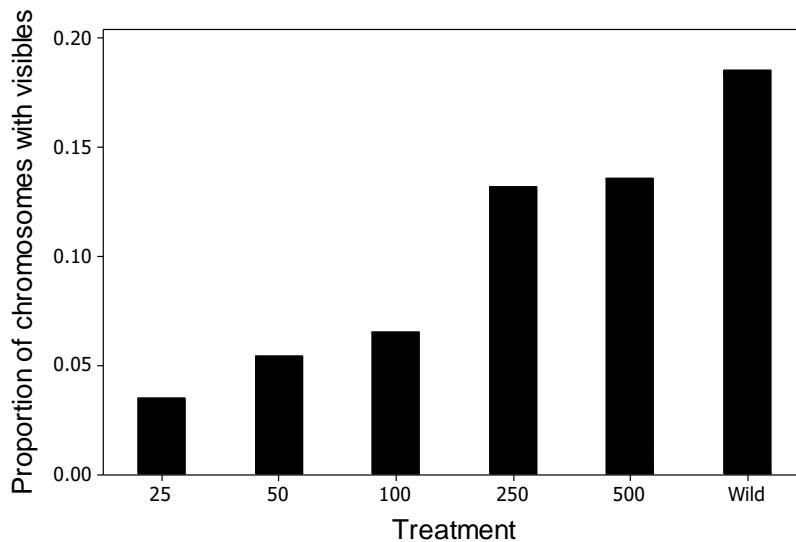
(Robertson 1962)

Frequency dependent selection $N_e \geq 5K$

(Roff 1998)

$N_e = 500$ inadequate

Retaining evolution potential for fitness: Empirical data



Revisions: retaining evolutionary potential in perpetuity

(Frankham et al. 2014)

1. Specify retain QGV for total fitness
2. $N_e \geq 1000$ required

N_e in most thr species are too small
to avoid loss evol potential

3. How large do populations
need to be to retain single
locus genetic diversity in
perpetuity?

Retaining single loci GD

Lande & Barrowclough 1987

- Why are we concerned about indiv loci?
 - MHC in vertebrates
 - SI alleles in plants
 - Sex locus in Hymenoptera
- $N_e = 10^5\text{-}10^6$ to retain
- No thr sp this large, nor are many non-thr sp (including humans)

What happens if $N_e < 1000$?

- Extinction? Not necessarily soon
- Slow and continuous genetic deterioration
- ‘Fragility’
- Higher risk of eventual extinction, especially with catastrophic environmental change

Can we just wait for genetic diversity to be regenerated by mutation?

How long does this take?

It takes many generations to regenerate GD by mutation?

(Lande & Barrowclough 1987)

GD

Regeneration (G)

Quantitative $10^2\text{-}10^3$
confirmed in empirical studies

Single-locus $10^5\text{-}10^7$

Must preserved GD, not rely
on mutation to regenerate it in
eukaryotes

4. Avoid accumulation of harmful mutations

- Chance fixation of harmful alleles is elevated in small populations
- Can lead to extinctions “mutational meltdown”

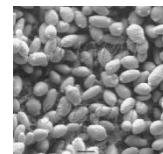
Avoiding mutational accumulation: Theory

- Outbreeders
 - $N_e < 12$ (Charlesworth et al. 93)
 - $N_e < 100$ (Lynch et al. 95)
 - $N_e < 1000$ (Lande 95)
 - Depends on effects of harmful mutations
 - (Garcia-Dorado 2003)
- Asexuals
 - Worse

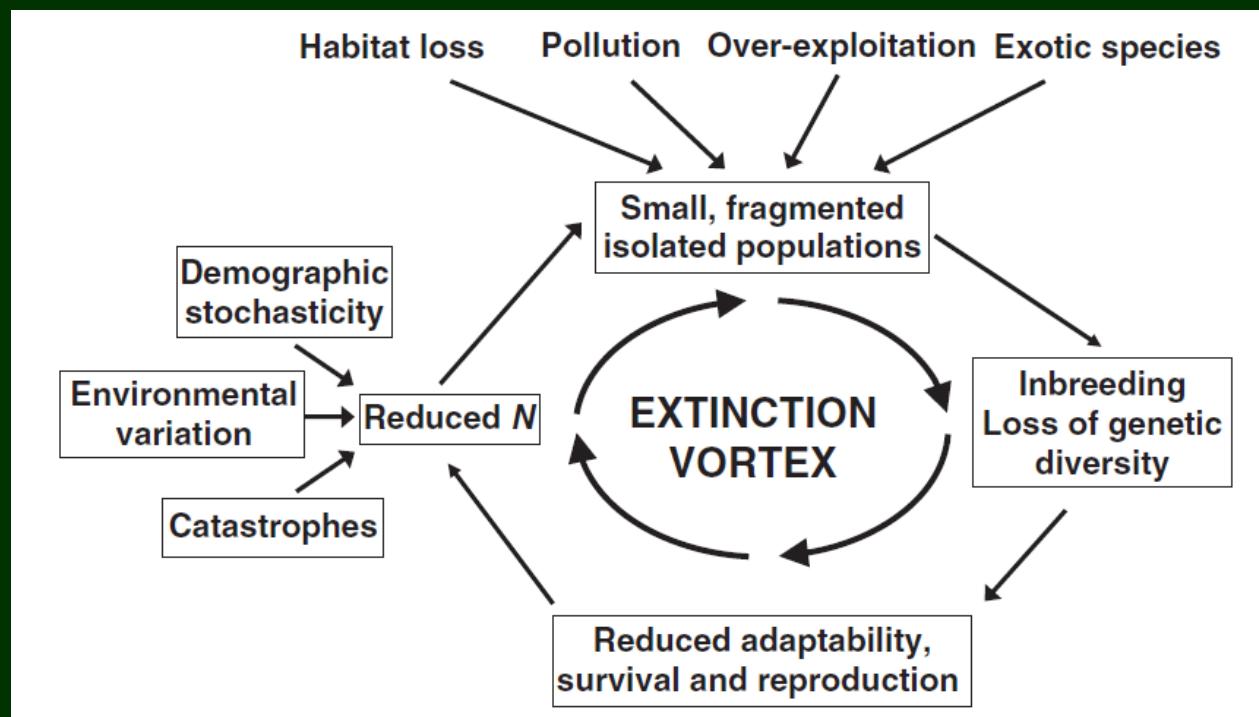
Avoiding mutation accumulation

Empirical data

- No mut accum in *Dros* 45-50G N_e 25-500
(Gilligan et al 1997)
- No mut accum in 2900G asex yeast N_e 250
(Zeyl et al 2001)
- Nematodes $N_e = 1$ lost <1% fitness/G
(Vassilieva et al. 2000; Estes et al. 2004)
- Conclusion:
 - Appears to be a minor threat << ID



What population sizes are required to cope with all threats (MVP)?



Sizes required for long-term viability to cope with different threats

Threat	N_e	N
Theory (Frankham et al. 2014; Nunney & Campbell 1993)		
Retaining QGV	1K	~10K
Demographic stochasticity		10s – 100
Environmental stochasticity		1K+
Catastrophes		1K+
Empirical data (Reed et al. 2003; Traill et al. 2007; Harcourt et al. 2002)		
PVA for 100 vertebrate species: 99% persistence for 40 G		>6K
PVA for 212 species: 99% probability of persistence for 40 G		4.2K
Primates in Sunda Islands		>16K

What are the population size targets for threatened species in captivity?

Goal: Retain 90% of genetic diversity for 100 yrs



How was this arrived at?

- Tradeoff between # species conserved & how well each is conserved
- Scenario: human pop will peak and decline within 100-200yrs & release habitat for reintro of thr sp

How large do the species need to be to meet this target?

Aim: Retain 90% of genetic diversity for 100 yrs

Required N_e depends on generation length (L)

$$H_t/H_0 = [1 - 1/(2N_e)]^t \sim e^{-t/2N_e} = 0.9$$

let $t = 100 / L$

$$0.9 = e^{-100/2LN_e}$$

take ln & rearrange

$$N_e \sim 475 / L$$

Endangered species in captivity

$$N_e \sim 475 / L$$

Examples:

Elephant $L = 35$

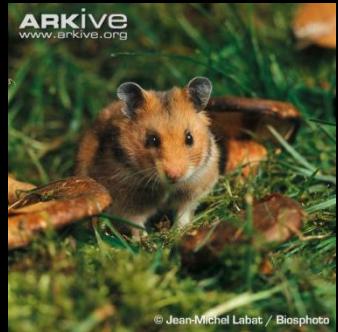
$$N_e = 475/35 = 14$$

White-footed mouse $L = 0.27$

$$N_e = 475/0.27 = 1759$$



Fallacy of small wild isolated surviving populations

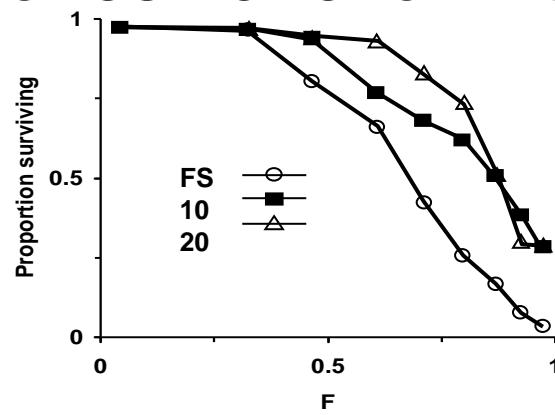


Fallacy of small wild surviving populations

- Small/bottlenecked surviving populations
 - Mauritius kestrel, golden hamster, Catham Is black robin, Seychelles warbler, Mauritius pink pigeon, Socorro Is red-tailed hawk, N elephant seal, California Islands foxes, Chillingham cattle
- Fallacy to argue from a few & generalise
 - ‘Grandad smoked 30 a day & lived to 80, so smoking does not contribute to cancer’

Fallacy of small wild surviving populations

- Selected sample – most small isolated pops go extinct in long-term
- Some highly inbred ($F \sim 1$) populations of mice, guinea pigs, *Drosophila* & plants persist
 - All surviving ones have low fitness when tested



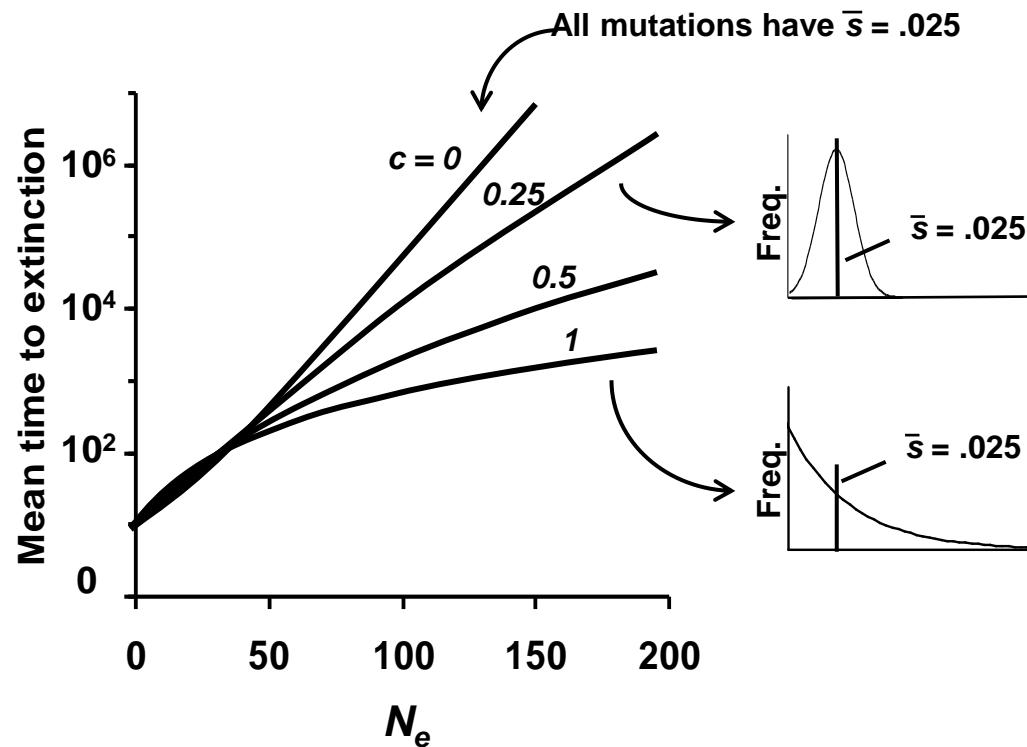
Fallacy of small wild surviving populations

- Claims of no fitness declines have been based on no firm evidence (Ch cattle & Ca Is foxes)
- Most small persisting wild population have improved environments (M kestrel, NE seal)
- Some have not been totally isolated (IR gray wolf, Ca Island foxes)
- Some have gone extinct & been re-established? (Cape Verde kite & several NZ populations)

Messages

- Resources limited for threatened species
- Need to define the minimum N to retain genetic ‘health’
- To retain wild fitness & minimise ID for 5 gens requires $N_e \geq 100$
- To permanently retain evol potential requires $N_e \geq 1000$
- Current population sizes of thr species too small to avoid genetic deterioration
- Captive populations of thr species typically managed to retain 90% GD for 100 years

Questions?



Introduction to Conservation Genetics
Fig 15.2 N_e vs $t(E)$.ppt

Translating from N_e to N

(Frankham 2021)

N_e/N ratios from meta-analyses

- Only multigenerational estimates for ~ 47 species in 2021
 - Average ~ 1/10
(vary according to life-history)

$$N \sim 10 \times N_e$$

BIOL3110

Genetic management of captive populations & reintroductions

R Frankham

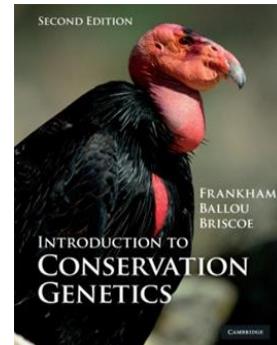


Genetic management of captive pops & reintroductions

Outline

- Why do species have to be captive bred?
- How do captive populations deteriorate genetically?
- How many founders should be used to initiated captive populations
- How should captive populations be genetically managed?
- How should reintroductions be genetically managed?

Reference: ICG2 textbook Ch19 & 20



Why captive breed?

- 2K species of terrestrial vertebrates cannot survive in the wild



How many TH species are being captively breeding?

- 987 threatened & near-thr vertebrate species
- Few invertebrates
- Many threatened plants
 - 105K sp, 41% of thr sp
 - Kew Gardens 2.7K species



Center for Plant Conservation, N Am 1.5 K sp

What is the aim of captive breeding programs?

Save species from extinction & eventual to return them to the wild

but many species will spend long periods in captivity

Scenario

How do populations genetically deteriorate in captivity?

- Inbreeding depression
- Loss of genetic diversity
- Mutational accumulation
- Genetic adaptation to captivity

What are the objective of genetic management of captive populations?

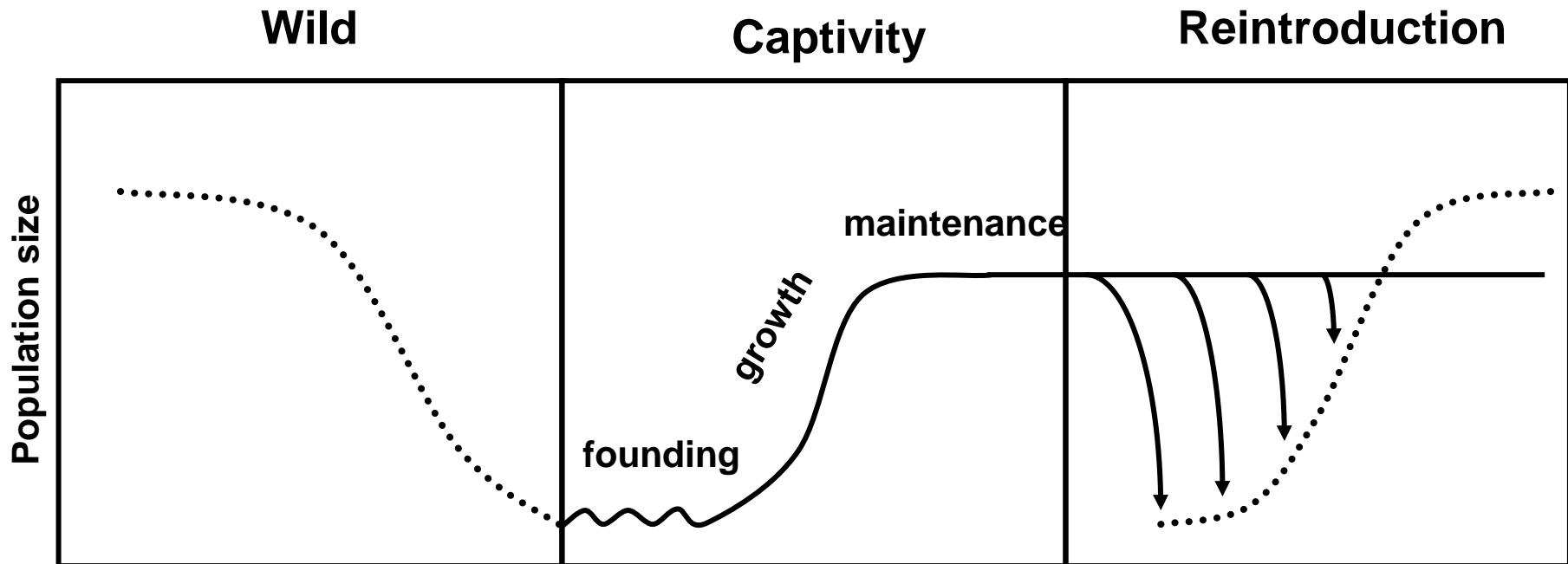
‘Freeze’ evolution

- retain genetic diversity
 - minimizing mean kinship (mk)
 - effective in minimizing inbreeding
- minimize adaptation to captivity

What is the targets set for captive genetic management?

- Retain 90% GD for 100 years
- Trade-off of genetic deterioration for more species maintained

Context



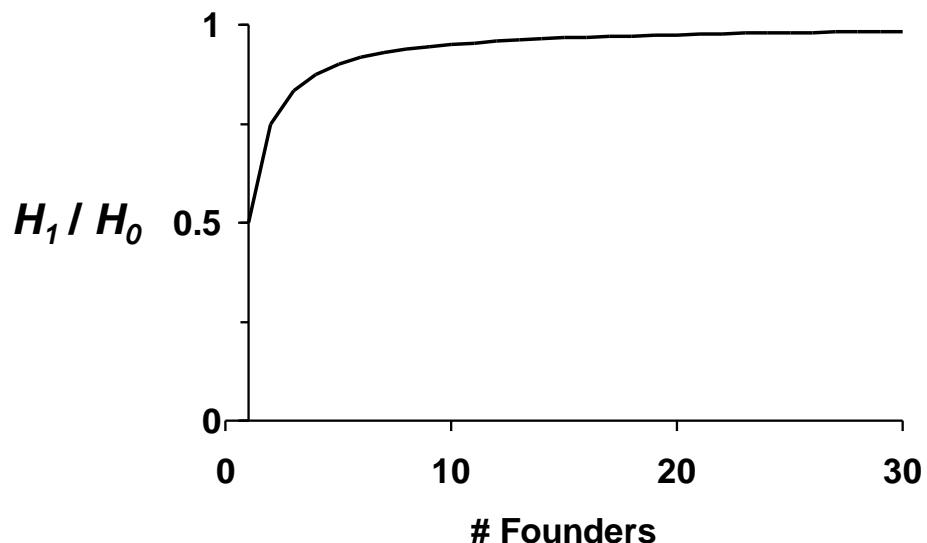
Primer on Conservation Genetics 2
Fig 19.1 Six stages of captive reintro.ppt

How are captive populations genetically managed?



How many founders should be used to initiate captive populations?

- Minimum 20-30 contributing individuals



How many founders have actually been used?

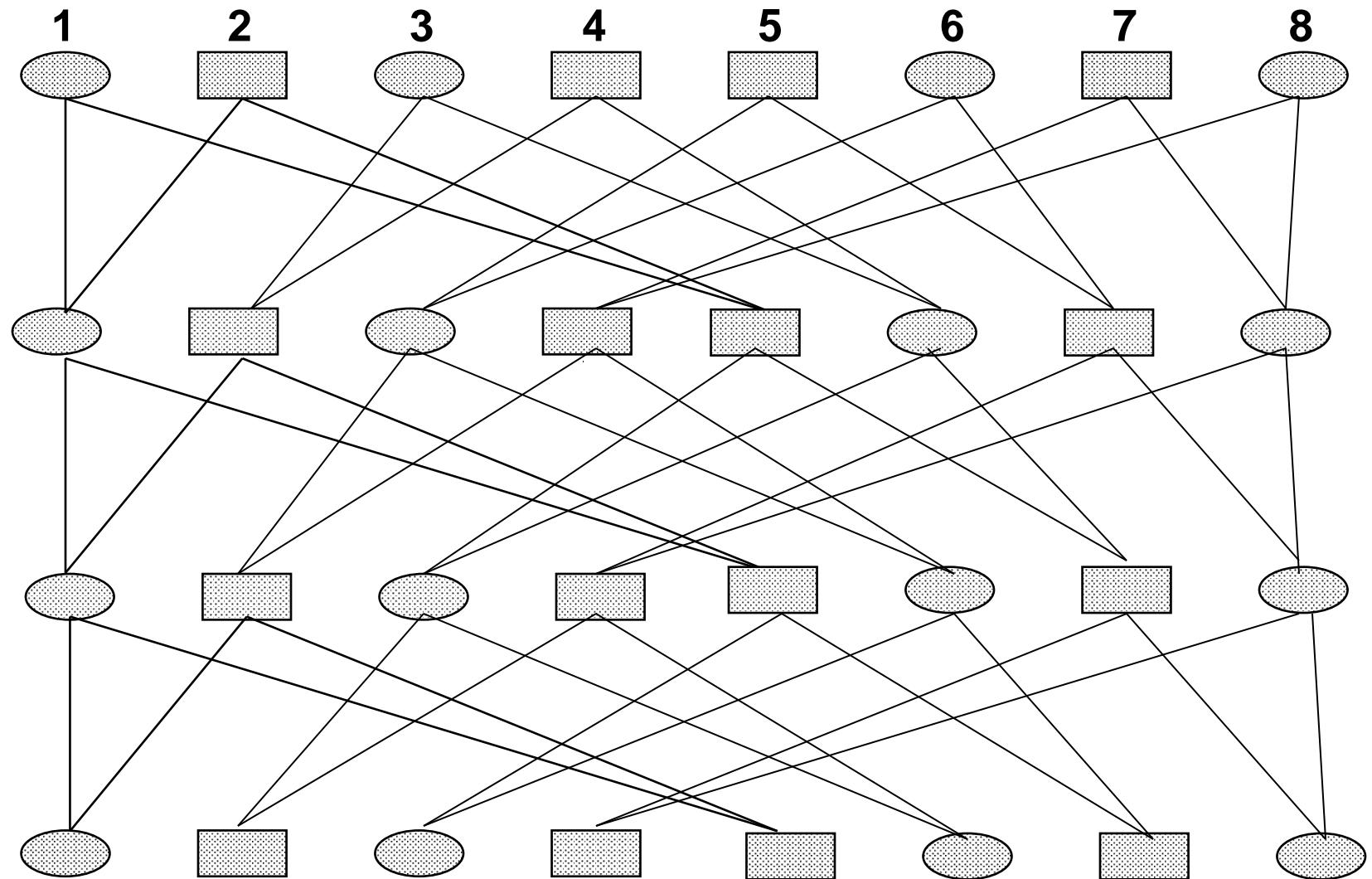
Species	Number of founders
Mammals	
Arabian oryx	10
Black-footed ferret	7
European bison	13
Golden lion tamarin	48
Indian rhinoceroses	17
Przewalski's horse	12 (~ 4 domestic horses)
Red ruffed lemur	7
Siberian tiger	25
Snow leopard	7
Speke's gazelle	4
Birds	
California condor	14
Guam rail	9
Lord Howe Island woodhen	6
Mauritius pink pigeon	6
Plains pigeon	21
Puerto Rican parrot	13
Invertebrates	
British field cricket	12

What is done in the growth phase?

- Increase numbers
- Little genetic management (avoid close inbreeding - FS & OP)

What is done in the maintenance phase?

- Genetic management
 - Retain GD & minimise inbreeding
 - Minimise mk



Maximum avoidance of inbreeding

What are the key features of maximum avoidance of inbreeding?

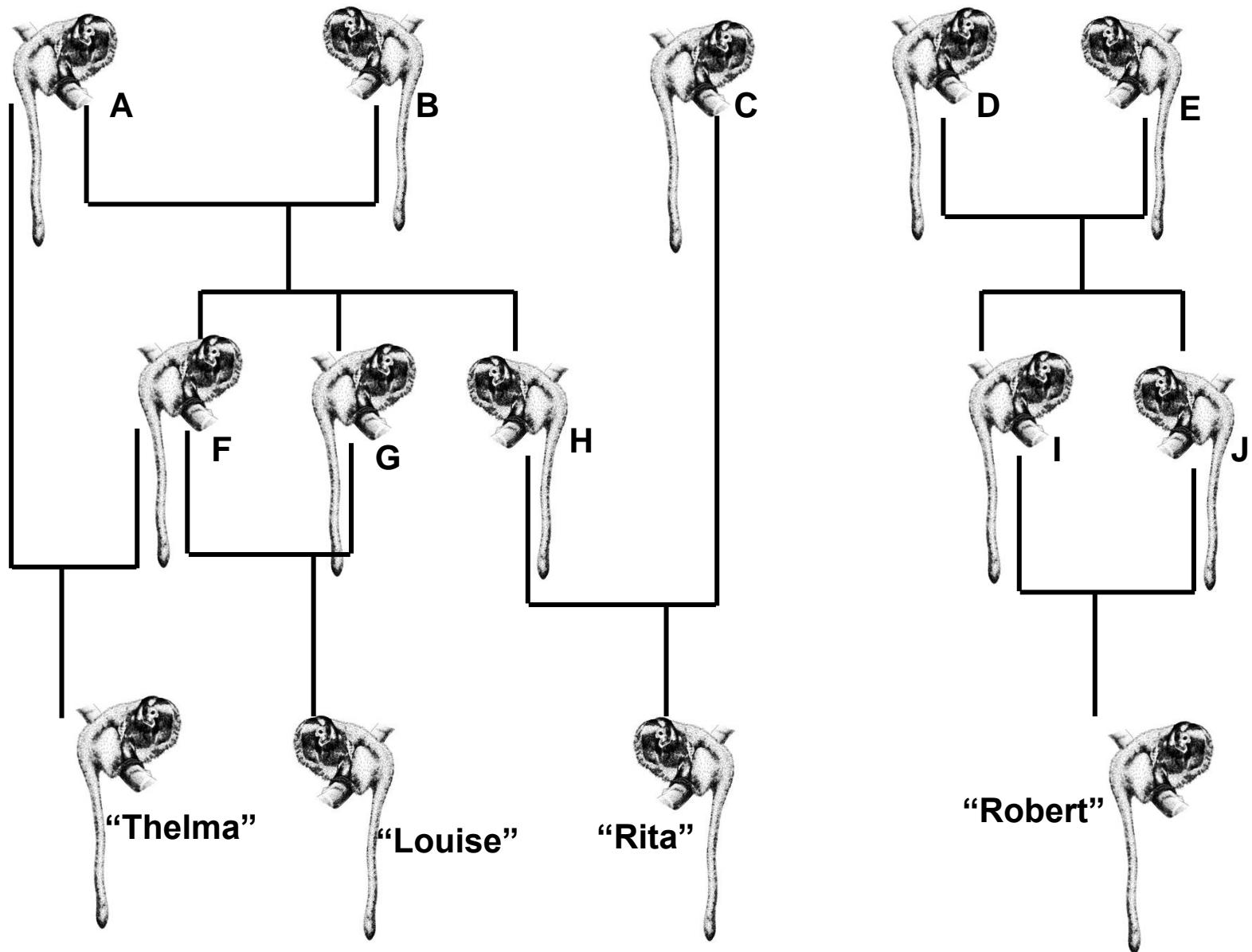
- Unrelated founders
- Equalising founder representation
- Equalising family sizes
- Equalising sex ratios
- $N_e \sim 2N$
- Actual captive pops $N_e \sim 0.3 N$

What is kinship?

- $k_{ij} = F$ of hypothetical offspring of i & j
- Probability that 2 alleles, one from each individual, are identical by descent (recent copies of the same segment of DNA)
- Range 0-1
- Generally determined from pedigrees

How is mean kinship minimised?

- Mean k for every individual with everyone else including self
- Choose individuals with lowest mean k as parents of next generation
- Maximizes retention of GD
 $\sim \min F$
- It becomes MAI regime in time



Kinships

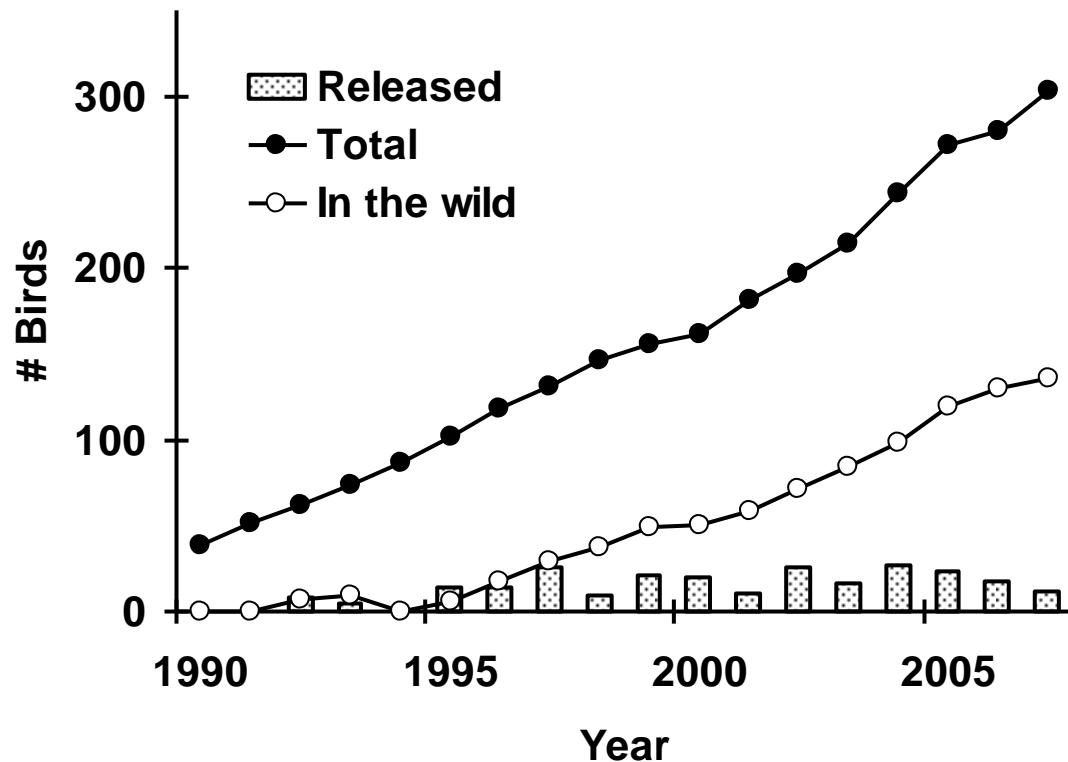
	Thelma	Louise	Rita	Robert	Mean kinship
Thelma	5/8	5/16	1/8	0	0.266
Louise	5/16	5/8	1/8	0	0.266
Rita	1/8	1/8	½	0	0.187
Robert	0	0	0	5/8	0.156

Choose Robert, then Rita as parents

Case study: California condor



- Declined due to DDT & lead poisoning
- Captive pop: 14 founders – 3 from 1 family
- Growth with little G mgmt
- G management by min mk (genomes now)
- N total > 446 , ~ 276 wild
- Inherited disease
 - chondrodystrophy (dwarfs 3%) - recessive
 - G management (prev, now carrier detect) ²¹

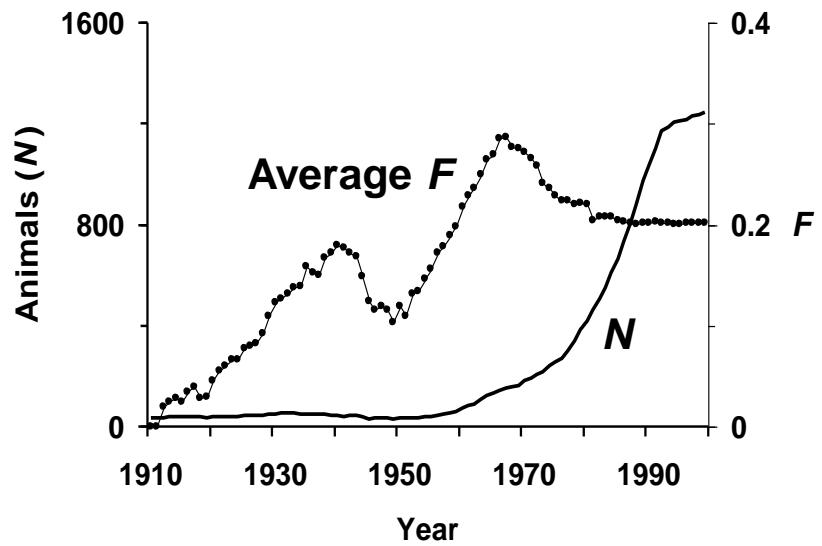


Introduction to Conservation Genetics 2
Fig 20.7 Census of condors

Case study: Przewalski's horse



- Decline – hunting & competition
- Captive 12 founders + up to **4 domestic horses**
- G Managed to
 - minimize domestic horse genes
 - equalize founder rep & min inbreeding
 - Problem: pedigree errors
- Reintroduced ~ 500 in wild (~ $\frac{1}{4}$)
- Genome: distinct from domestic horses
 - Low GD, inbred, 0-31% dom horse genes²³



How successful is captive breeding?

Many species have been successfully captive bred

A few not successful e.g hairy-nosed wombats

How much used is made of assisted reproductive technologies?

- Artificial insemination (5 thr sp)
- Cryopreservation (plants)
- Cloning/nuclear transplantation
 - Plants (widespread)
 - Few thr animals (BF ferret & Prz horse – added founders)



- Available for very few animal sp, but **½ way** improving

Attempts to resurrect extinct species

- Considered nigh impossible in 2010
- Great advances in stem cell technology since then
- Still very low probability of success, but
- Efforts underway in mammoth, passenger pigeon, thylacine, gastric brooding frogs

Reintroductions



Genetic issues in reintroductions

Inbreeding and loss of genetic diversity
Consequences of genetic adaptation to captivity

- How can we minimise it?

Genetic adaptation to captivity



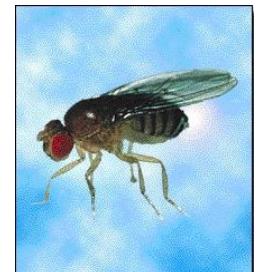
Genetic adaptation to captivity is unavoidable

- Rats > 3x (25G)
- Turkeys
- Fish – many species
- Biocontrol insects
- Butterfly 13x (100-150G)
- *Drosophila* 2x & x3
- Plants ~ 2x

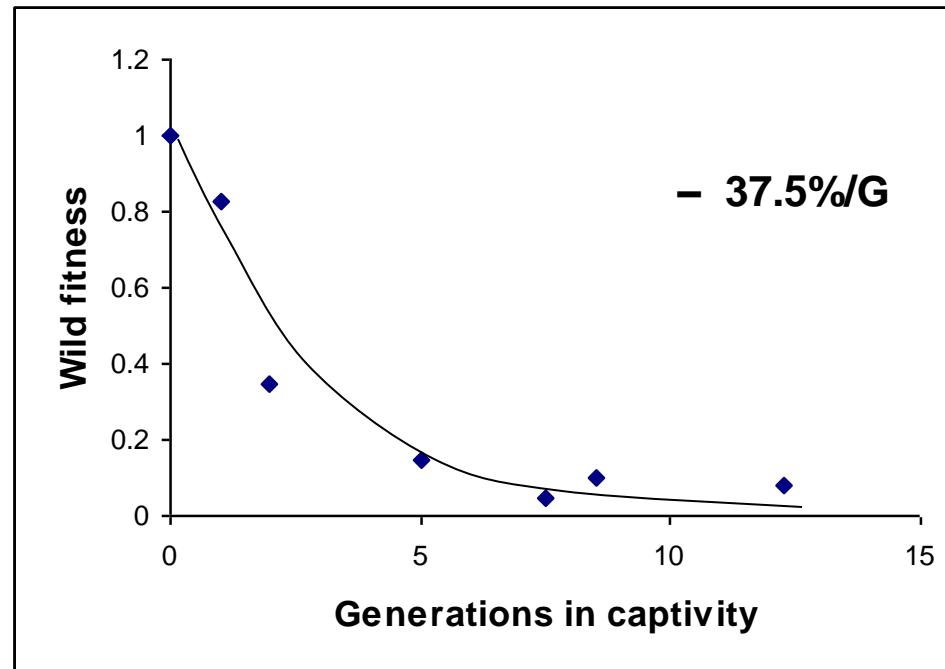


Genetic adaptations to captivity are overwhelmingly harmful following reintroduction

- Biocontrol insects
- Fish – many species
- Rabbits & birds
- *Drosophila* populations



Harmful impact of GADC on fitness of reintroduced salmonids



Conservation implications of GADC

- Captive populations are undergoing ‘genetic revolutions’, not having their evolution ‘frozen’
- Need to manage captive populations to explicitly minimize genetic adaptation to captivity

How can we minimize GADC?

Minimize

- generations in captivity
 - Seed storage in plants
- selection (**EFS halves**)
- N_e & genetic diversity (**fragmentation**)

Minimizing GADC: Does fragmenting population work?

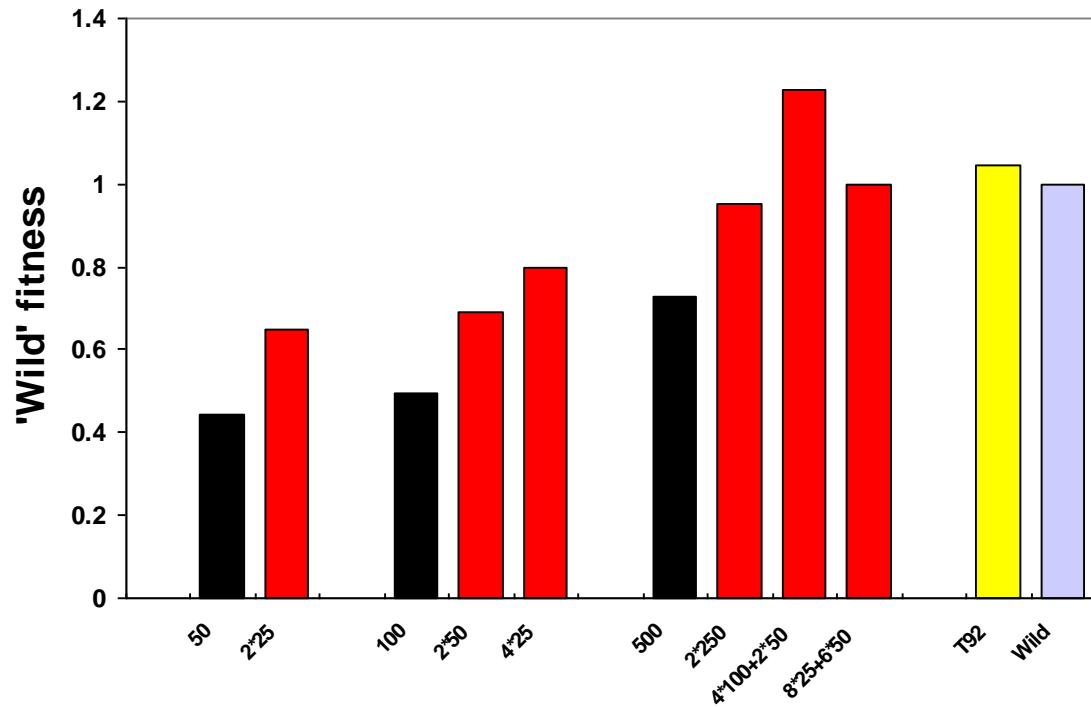
Test of fragmentation strategy: SL v SS

- 50 v 2x25
- 100 v 2x50 v 4x25
- 500 v 2x250 v
- 50 gens & pooled SS
- ‘wild’ fitness
- genetic diversity



Fitness of SL v SS (pooled)

(Margan et al. 98)



Fragmentation reduces harmful
fitness effects of GADC in
reintroduced populations

Other benefits of fragmenting captive populations

- Retains more genetic diversity
- Reduces inbreeding
- Captive populations already fragmented
 - Reduces costs of translocations
 - Reduced risk of disease transfer

What problems are associated with fragmentation of populations

- Inbreeding depression
- Solution: cross replicates after inbreeding has accumulated to say $F \sim 0.2$ cross & fragment again

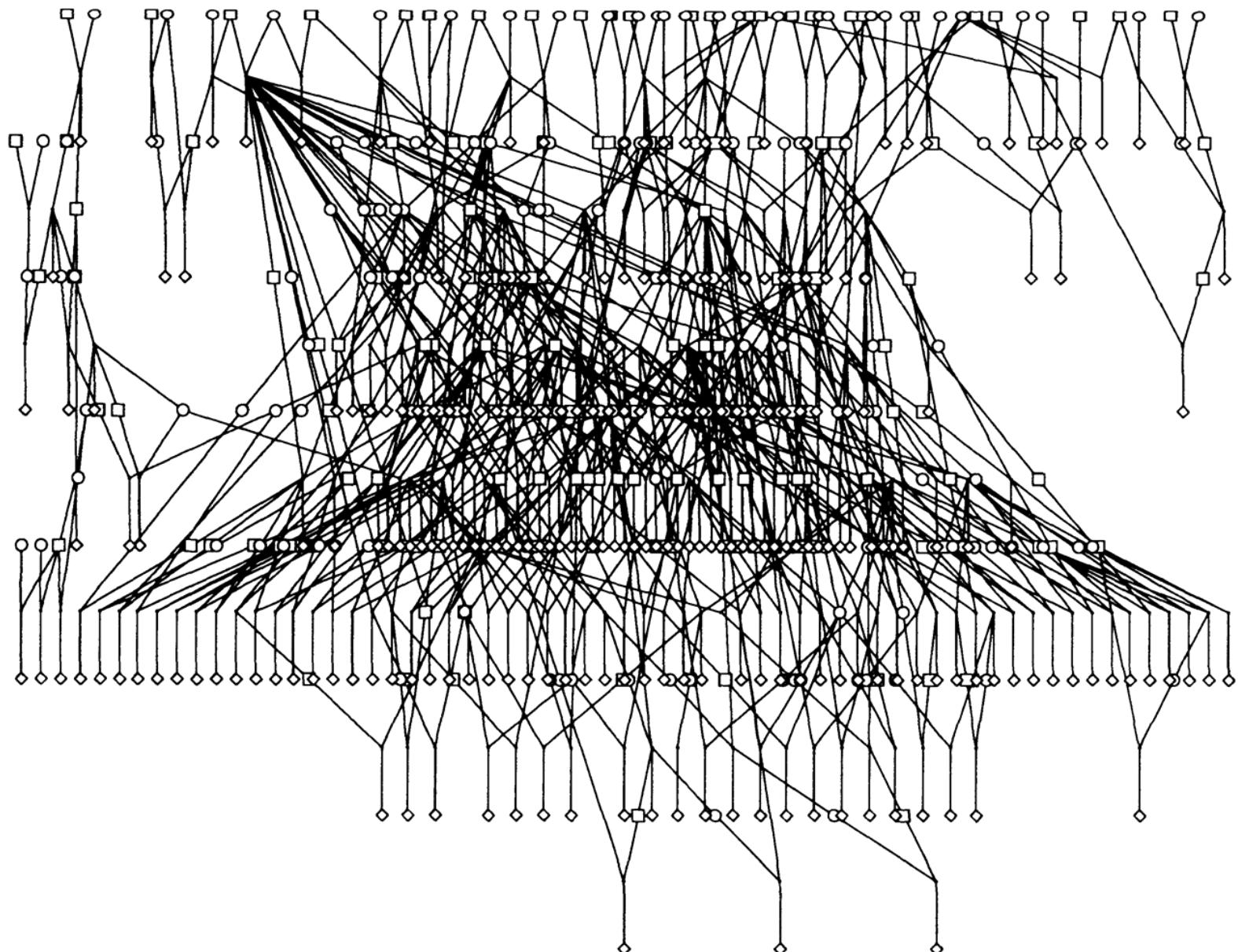
How are reintroductions genetically managed?

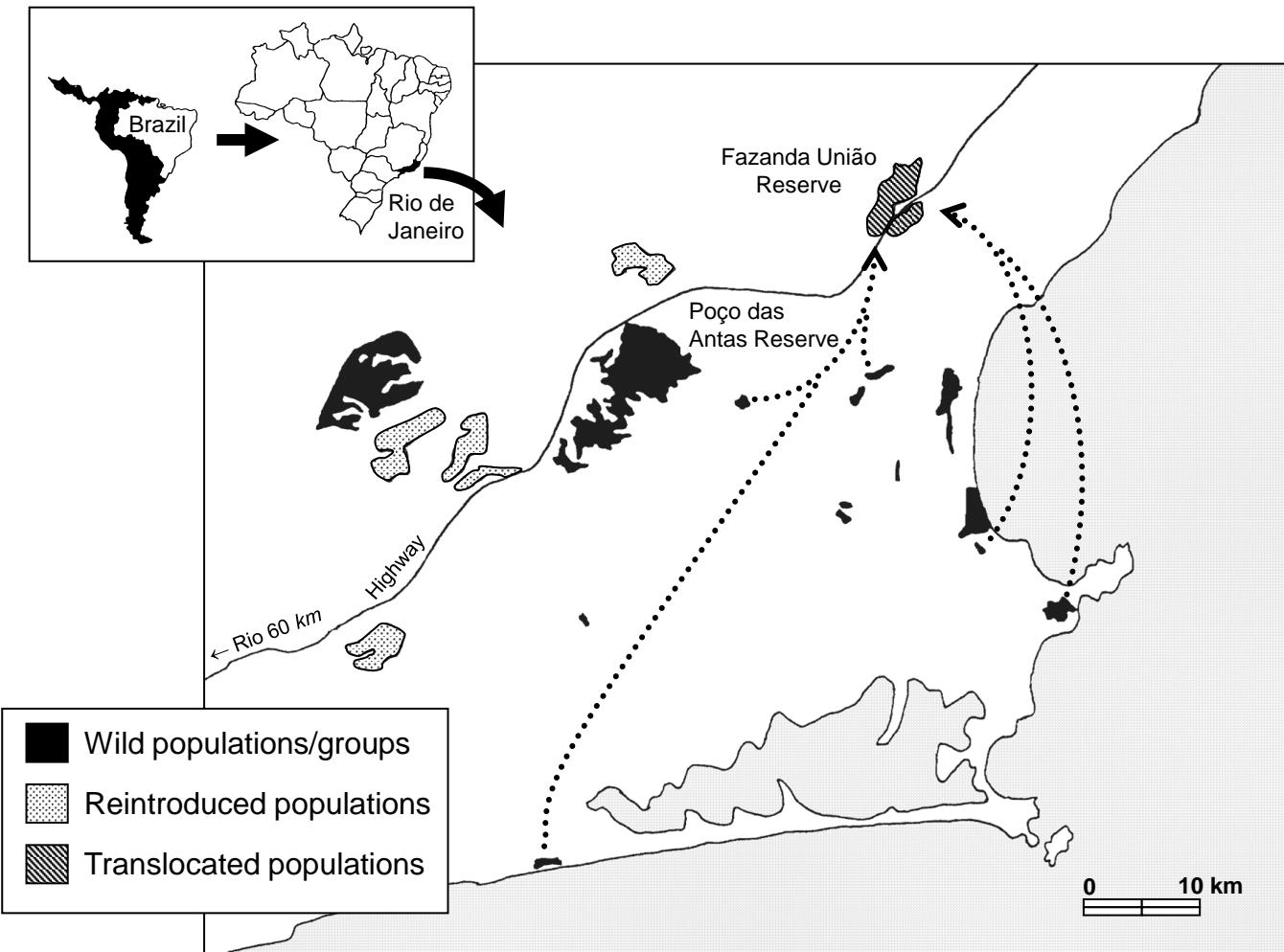
- Captivity less risky than wild
- Founded from animals of low genetic value in captive population
- As released population establishes, release animals to eventually transfer all genetic diversity from captivity to wild

Case study: Golden lion tamarin

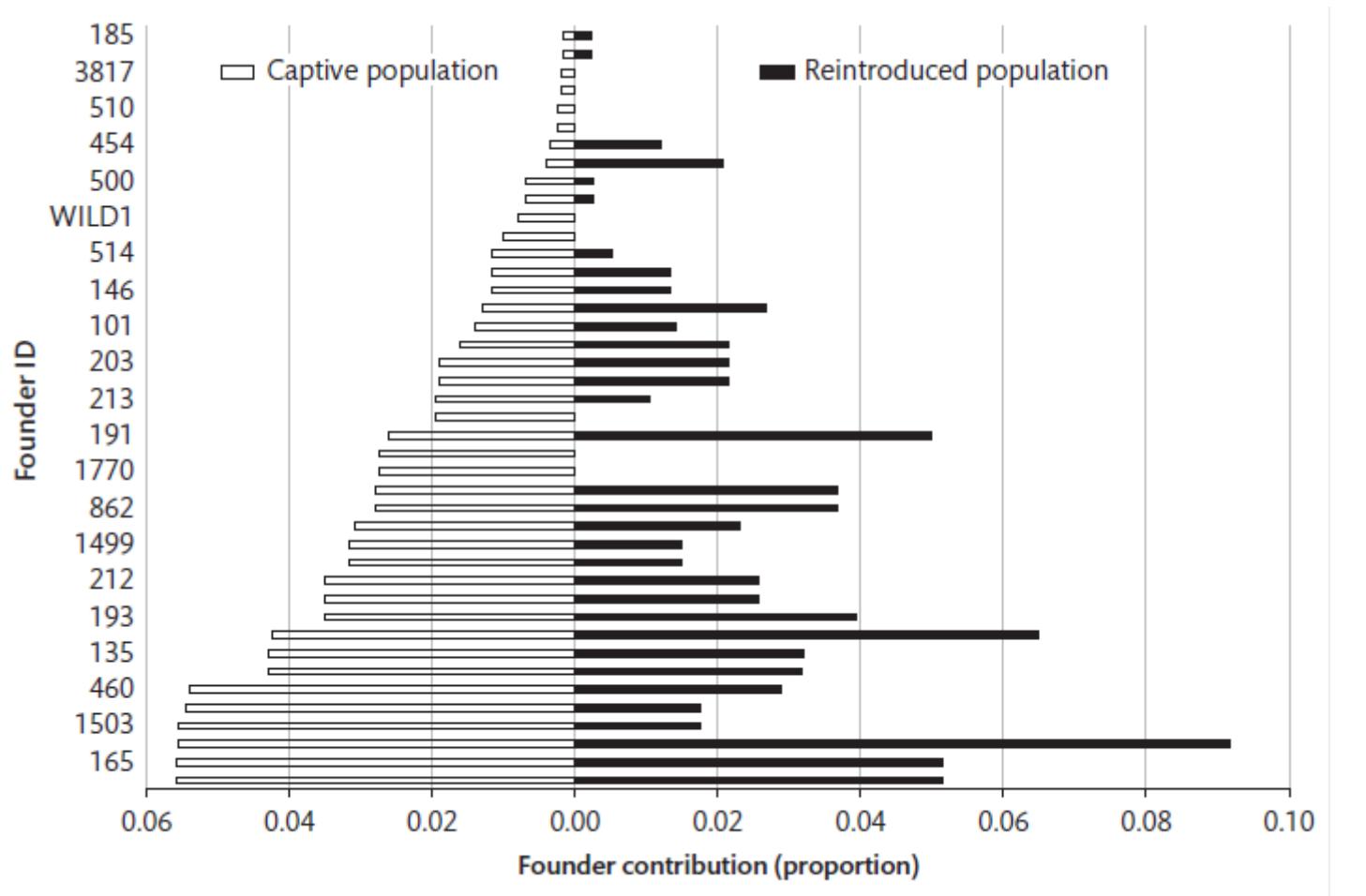


- Decline due to loss 98% habitat
- 242 captive founders, 48 still contributing
- 2/3 originally from 1 pr *
- Min mk devised to manage it ($F = 6.1\%$ before management & 2.6% now)
- Successfully reintroduced *
 - ~ 1600 in wild over 11 pops
- Fragmented distribution
 - low GD in small populations
 - corridors built





Founder representation in captive and reintroduced population



Case study: Lord Howe Island woodhen



- Decline to ~30 due to introduced pigs
- Pigs eliminated
- Captive breeding – 3 prs founders
- Reintroduction - 82 birds
 - recovery to ~220, now 160
 - No captives left
 - More inbred
 - Low GD
 - Genomics
- Example of how not to manage genetically
- Single population only & no routine monitoring

Case study: Mauna Kea silversword



- Decline to 41 plants – grazing
- Control of herbivores
- Outplanted 1500 plants
- All from 2 maternal founders
 - Self-incompatible
 - 20% seed set
 - 60% seed set with outcross
 - Trying to outcross outplanted plants
 - Problem of few founders & low pr flowering in any year
- Now: 8K plants in wild from 6 founders

How successful are reintroductions?

- 67% (Wolf et al. 1996)
 - 53% for threatened sp
- 44% (Beck et al. 1994)
- 26% successful, 27% failures, 47% unknown (Fischer & Lindenmayer 2000)

Summary – captive breeding

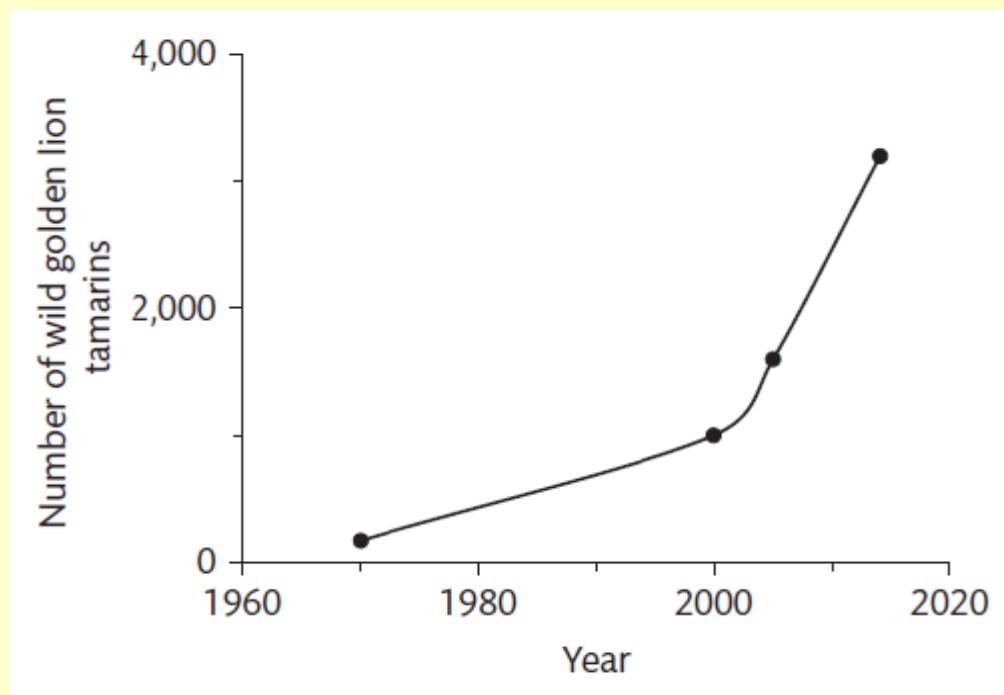
- Many species have to be captive bred to save them from extinction
- Captive populations deteriorate genetically due to
 - inbreeding depression
 - loss of genetic diversity
- Captive populations should be initiated with a minimum of 20-30 contributing founders
- Minimizing mean kinship recommended for managing endangered species in captivity

Summary - reintroductions

- Captive populations can provide individuals for reintroductions into wild
- Genetic adaptations to captivity are harmful when populations are reintroduced
- Genetic adaption can be minimized by
 - minimizing generations (seed storage in plants)
 - minimizing selection
 - fragmenting captive populations
- Begin reintroduction with genetically surplus individuals from the captive population
- Once successful, augment wild pops until they contain the all GD found in captives

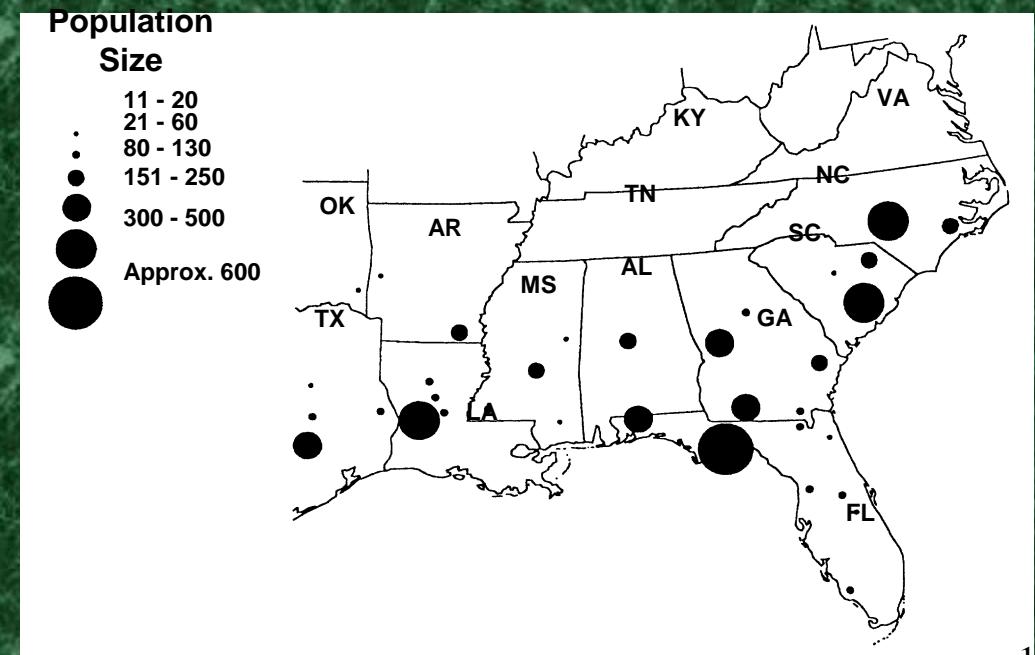
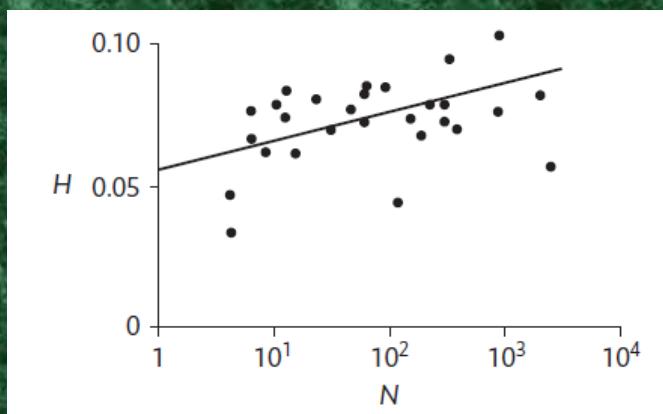
Questions?

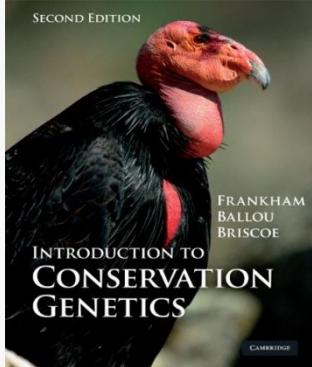
Total numbers of tamarins in the wild



BIOL3110

Genetic management of wild populations





Genetic management of wild populations

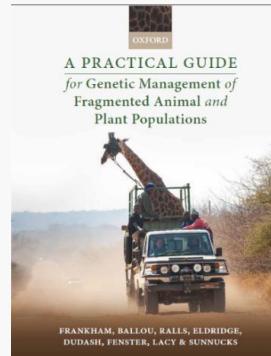
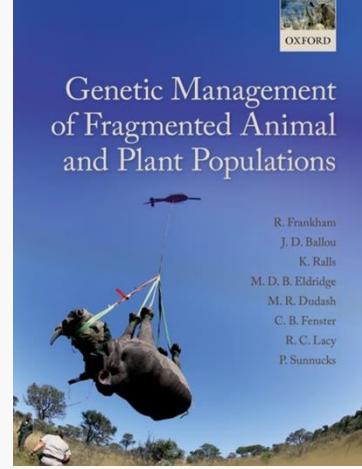
Outline

- What genetic issues are faced by wild pops?
- How should we manage fragmented pops?
- How can we manage sp with 1 population?
- What management is required under global climate change?
- How does genetic management differ for species with diverse breeding systems & modes of inheritance?

References:

ICG2 Ch17

Frankham et al. 2017 Genetic management of fragmented animal and plant₂ populations



What genetic problems are faced by wild populations?

- Taxonomic uncertainties & OD
- Inbreeding & reduced fitness
- Loss of genetic diversity & reduced ability to evolve
- Fragmentation & inadequate gene flow
- Consequences of global climate change
- Consequences of different mating systems & inheritance modes
- Genetic management of translocations
- Genetic effects of selective harvesting
- Genetic swamping from related species

What are the genetic goals of wild management?

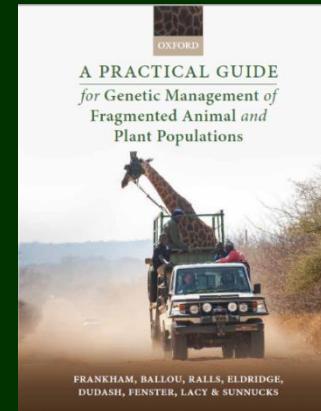
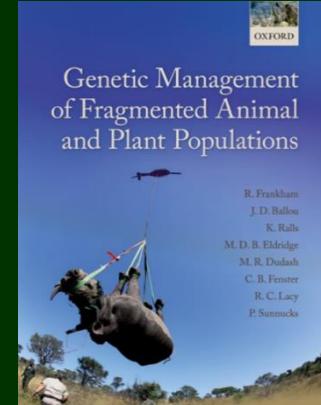
- Minimise extinction risk
 - Maintain reproductive fitness
 - Conserve genetic diversity and the ability to evolve
- Maintain self-sustaining species

What genetic management can be applied to species with multiple population fragments?



Why is fragmentation a problem?

- Inadequate gene flow
- No gene flow
 - Relevant N_e is that of the fragment, not species
 - Inbreeding, loss GD, reduced fitness & Ev potential worse than for single popn of same total size
- One of the major, largely unaddressed issue in conservation biology (our books)



Vast magnitude of the fragmentation problem

- ~ 1.4m pop fragments of thr species with genetic problems
- ~ 150m total pop fragments with G problems
- ~ 34 cases of conservation management to reverse the fragmentation problems

How should we genetically manage fragmented populations?



Genetic management

- Increase N to slow rate of deterioration
 - increase habitat size or quality
 - re-establish extinct populations
- Genetic rescue - re-establishing gene flow into inbred pops –
 - outcrossing
 - corridors
 - Rarely being done due largely to fears of OD
 - Outcrossing can be beneficial or harmful

What is outbreeding depression?

- Harmful effects on fitness that are sometimes found upon crossing of populations within or across species (= reproductive isolations)
 - Observed in F_1 or later generations
 - Occurrence predictable

The causes of outbreeding depression are known

Different species



Fixed chromosomal differences



Adapted to different environments



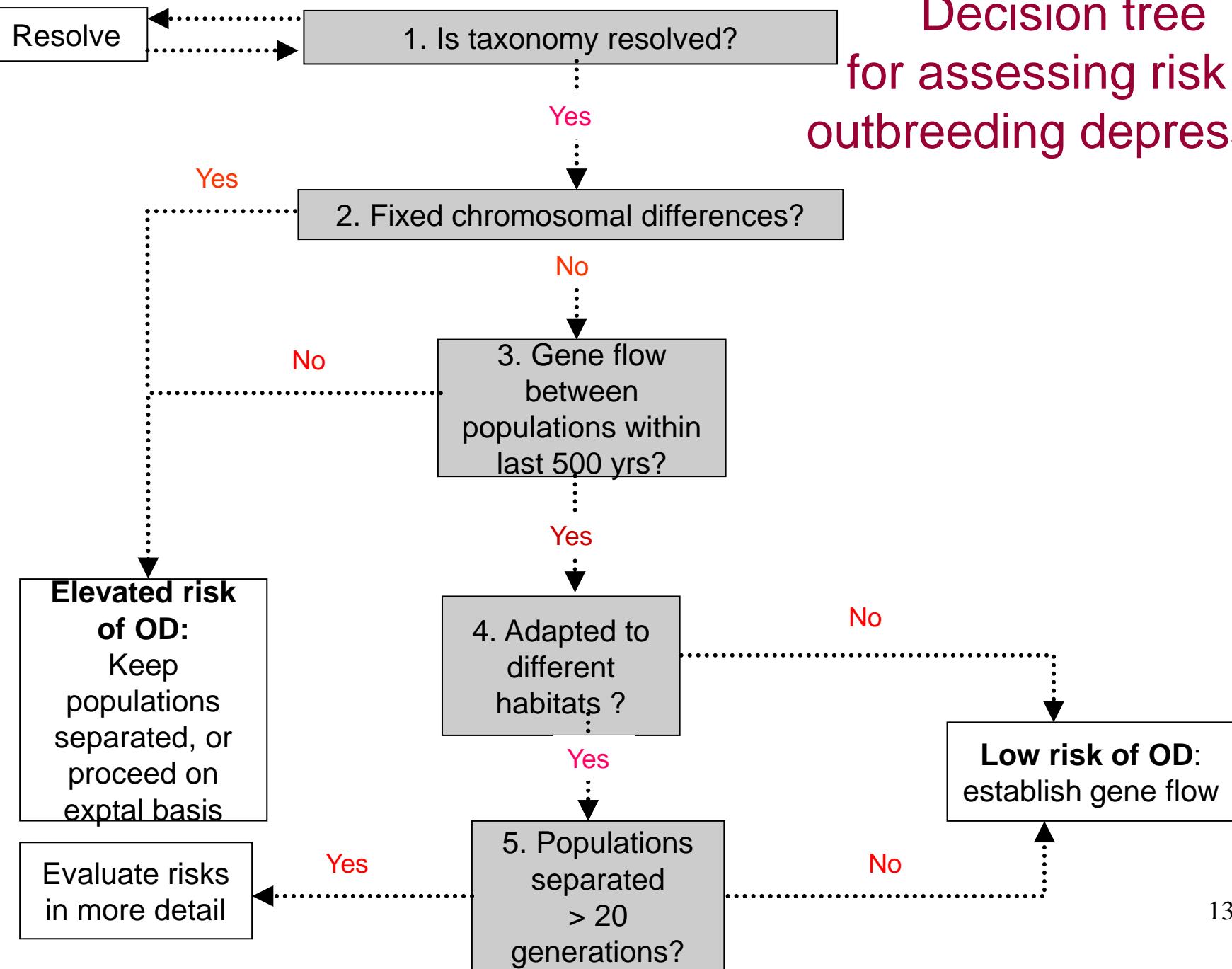
Long isolation



We used this information to
predict the risk of outbreeding
depression

(Frankham et al. 2011 CB)

Decision tree for assessing risk of outbreeding depression



The decision tree works

- Preliminary evaluation: identified high risk cases
- Meta-analysis: screened for low risk: 156 cases, $145 +: 2 =: 9 -$
 - OD modest at worst ($\leq 14\%$)
 - (1 true OD)
- Highly effective

Genetic rescue of inbred pops by outcrossing



SPECIES
SPOTLIGHT...



Large and consistent benefits of outcrossing on fitness

(Frankham 2015 ME)

- Inbred & screened to have low risk of OD
 - 92.9% beneficial
 - 148% increase in fitness in wild, 45% in captivity
- Benefits persist over gens in outbreeding species
 - F_1 (91%), F_2 (100%) & F_3 (94%)
- Recommend outcrossing of inbred population, provided risk of OD is low

Recovery from inbreeding & restoration of genetic diversity following crossing of inbred populations

Population	Genotype	F	Heterozygosity
Inbred a	A ₁ A ₁	1	0
Inbred b	A ₂ A ₂	1	0
F ₁ (a x b)	A ₁ A ₂	0	1
F ₂ (F ₁ x F ₁)	¼ A ₁ A ₁ : ½ A ₁ A ₂ : ¼ A ₂ A ₂	½	½



Case study: Florida panther

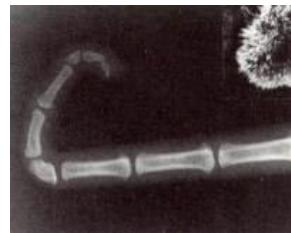


Taxonomy

Population bottleneck (min $N = 6$)

Low genetic diversity & ID – undescended testis, sperm, disease & kinked tails

Sub-species	Allozymes	DNA fingerprint
	H	H
Florida (authentic)	1.8	10.4
Western US	4.3	46.9
Other felids	3-8	45.9



Action: Outcrossed to Texas indiv - recovery

F1 benefits: survival: adults +26%, pups +99%
litter size +6.8%

Overall F₁ fitness benefit 168%

Unrelated individuals used for genetic rescue can be:

1. Outbred from within same species
2. Inbred, but genetically distinct

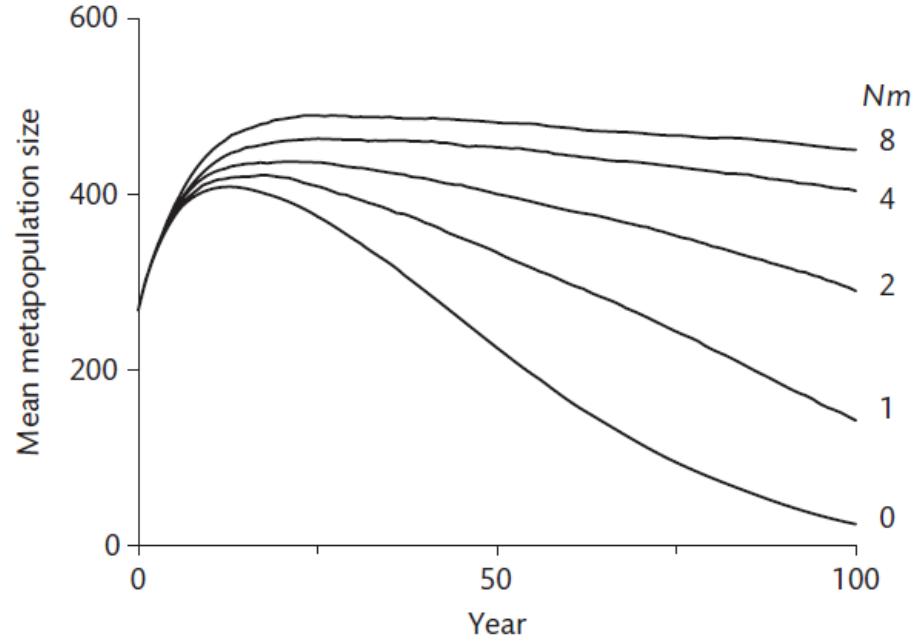
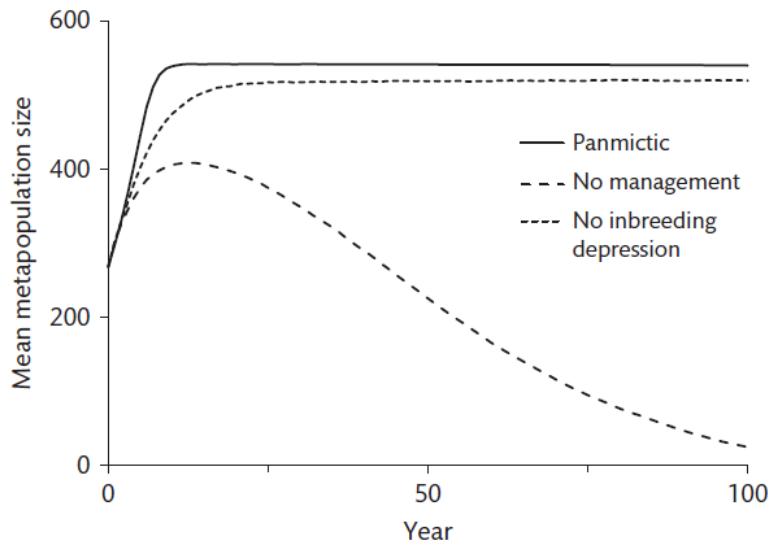
Choosing donor popn for crossing

- No genetic data
 - Any is better than none
 - Large N better than small
 - Longer isolation better than shorter
 - Several pops better than one
- Extensive genetic data
 - Lowest mean kinship with recipient

How much gene flow is desirable?

- Any better than none
 - 1 contributing migrant/gen – too few
 - ~ 5/gen needed
- Computer modelling
- Minimizing mean kinship

Computer simulation: Allegheny woodrat



(Frankham et al. 2017)

Managed translocations	Mean probability of sub-population extinction	Final metapopulation N	Mean inbreeding (F)
$Nm = 0$	0.81	26	0.37
$Nm = 1$	0.18	144	0.24
$Nm = 2$	0.02	288	0.18
$Nm = 4$	0.00	404	0.14
$Nm = 8$	0.00	450	0.11
Panmixia	0.00	540	0.09

Kinship

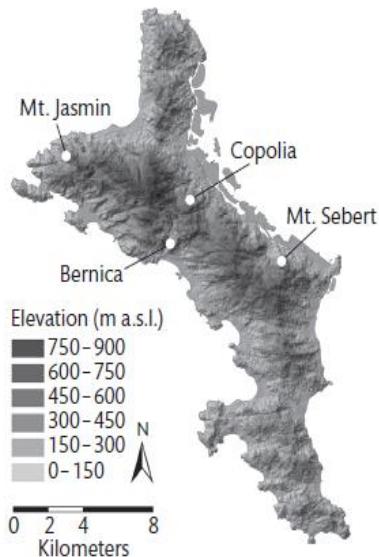
Kinship for 2 individuals is the F of offspring (if they had them)

Kinships in wild pops without pedigrees can be estimated using (many) genetic markers

Managing by minimizing mean kinship

- Favours populations (and indivs) with lowest kinship to all others
- retains maximal genetic diversity
- effective in minimizing inbreeding

Managing by mean kinship: CR jellyfish tree (Seychelles)



	Mt Jasmin	Copolia	Mt Sebert	Bernica
Mt Jasmin (2)	0.57	0.07	0.02	-0.02
Copolia (3)		0.17	0.09	-0.02
Mt Sebert (7)			0.41	-0.04
Bernica (78)				0.004

Cross of Bernica x Mt Sebert resulted in
151% improvement in fitness

What management can be applied to species with single population & low GD?

Few genetic remedies to reverse



Reverse cause of decline & increase N

Ban hunting

Indian rhino

N elephant seal

National Park & cattle removed

N hairy-nosed wombat

Predator proof fence

Bilby & N hairy-nosed wombat

Ban DDT

Mauritius kestrel

Bald eagle

Peregrine falcon

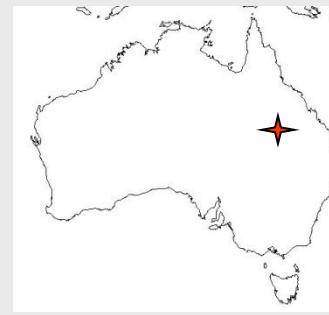
Exterminate introduced predators

Lord Howe Island woodhen

Improve habitat

Red-cockaded woodpecker

Translocations to establish new pops, or re-establish extinct ones



Case study

Wollemi pine

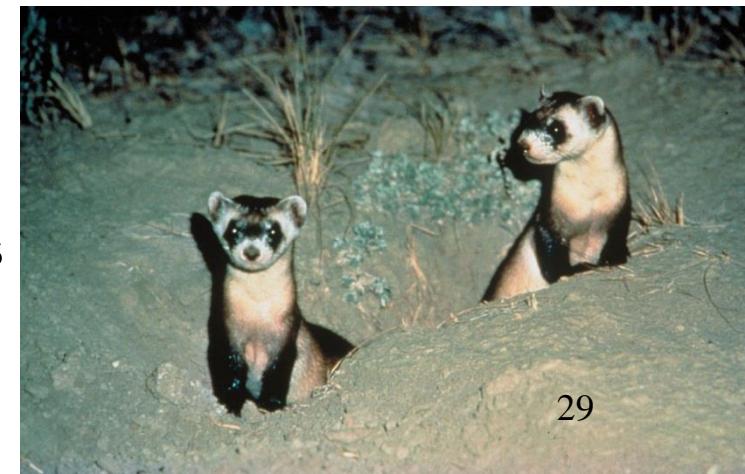
- < 100 adults in 3 pops in Wollemi National Park, NSW
- all individuals genetically identical for nuc (1 genetic pop)
- likely to be ‘fragile’
 - Susceptible to fungal disease
- Management:
 - Location secret
 - Restricted access & site hygiene
 - Fire management
 - Cuttings from all adults
 - *Ex situ* conservation & sale
 - Research (genetics & fungi)



Case study

Black-footed ferrets

- Extinct in wild (eliminating prey + disease)
- Captive populations (18 founders)
- Low genetic diversity
- likely to be ‘fragile’
- Risk management:
 - 10 reintroduced pops ($N = 1500$)
 - Actually reintroduced into 24 locations
 - Only 4 self sustaining
 - avoid catastrophes



Future genetic improvement

- Use gene editing to selectively “cure” major ID defects or add useful genetic variants
- Gene editing has already been used experimentally to cure several genetic diseases (e.g. DuMD dogs, mice & human cells, + others)
- Gene transfer to confer disease R in Am Chestnut

Managing under global climate change

Options:

- Adapt *in situ*
 - Improve ability to adapt
 - Adding genetic diversity by outcrossing
 - Move

Need for gene flow even more pressing

How is genetic management altered with diverse mating systems or modes of inheritance?



Frequencies of diverse mating systems & modes of inheritance

- Asexual/clonal (obligate rare, partial common)
- Selfing (10-15% plants habitual & 40% mixed)
- Self-incompatible (~ $\frac{1}{2}$ Angiosperms)
- Haplo-diploids (Hymenoptera ~ 15% animals)
- Polyploids (most plants & some animals)

Genetic management of asexual species

- No inbreeding depression or OD
- Accumulation of harmful mutations is a threat
- Many individuals may have identical genotypes
- Genetic diversity among clones
- Examples: some lizards, fish & plants
 - *Haloragodendron lucasii* (N. Sydney)
 - 53 plants = 7 clones
- Management:
 - Identify clones
 - Conserve clones
 - If 1 clones, often ‘fragile’



Genetic management of selfing species

- Less heterozygosity within pops
 - Greater differentiation among pops
 - Less inbreeding depression
 - Possibly more OD
 - Lesser genetic rescue
-
- G Management:
 - Conserve distinct populations



Self-incompatible species

- SI alleles are lost in small populations
- Loss of SI alleles decreases proportion of ovules fertilised
- Loss of SI alleles decreases fitness
- Also suffer ID, more susceptible to low N
- Re-establish gene flow & augment GD
- Increase N



Haplo-diploids ($\text{♂ } n / \text{♀ } 2n$)

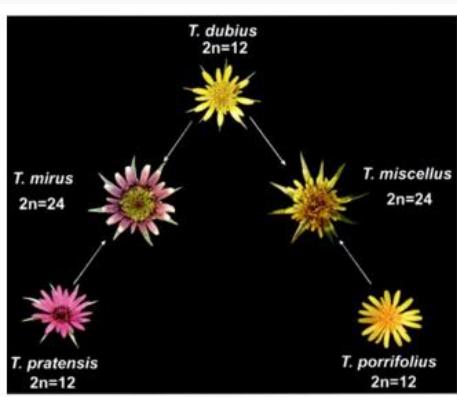


- Highly susceptible to loss of CSD alleles
- $N_e = \frac{3}{4}$ diploid
 - Eusocial sp have sterile workers, $N_e \sim 2-4$ # colonies
- Low susceptibility to ID
- OD more likely
- G Management
 - retain & augment genetic diversity (outcross)
 - Increase N

Genetic management of autopolyploids

- Less affected by small N
 - Slower loss of genetic diversity than diploids
 - Slower incr in inbreeding (homozygosity) and less inbreeding depression
- G management
 - As for diploids, but less concern about N
 - Avoid crossing diff ploidies e.g. $4n \times 2n$





Genetic management of allopolyploids

- Often behave as diploids (amphidiploids)
 - Most loci duplicated
 - Likely less susceptible to ID than diploids
 - May have better ability to evolve
- G management
 - Similar to diploids
 - Avoid crossing diff ploidies e.g. 4n x 2n

Very little practical genetic management is being done in wild pops

1. Taxonomic uncertainties & ESU – lots
2. Inbreeding & loss of genetic diversity – very little except diagnosis
3. Fragmented populations & G rescue – very little - corridors or outcrossing
4. Diverse breeding systems & inheritance modes– very little

Field is in its infancy

Messages

- Genetic management of wild populations involves
 - resolving any taxonomic uncertainties
 - increasing N
 - genetically rescuing small inbred population fragments suffering ID & low genetic diversity
 - modifying genetic management regimes for species that are not outbreeding diploids

Questions?

How should we manage
genetically to cope with global
climate change?

Selective harvest

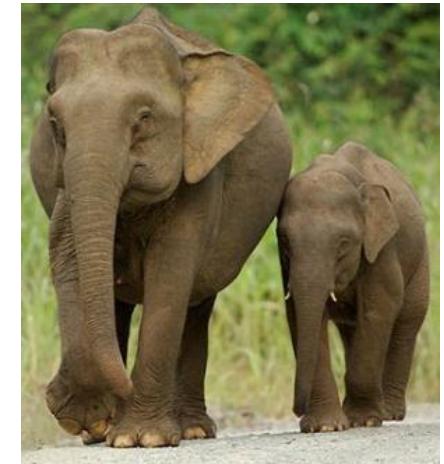


What are the genetic consequences of selective harvest?

- Evolutionary change (& possibly reduced fitness)
- Reduce N_e
 - Distorted sex-ratios
 - Change in breeding system (higher selfing)

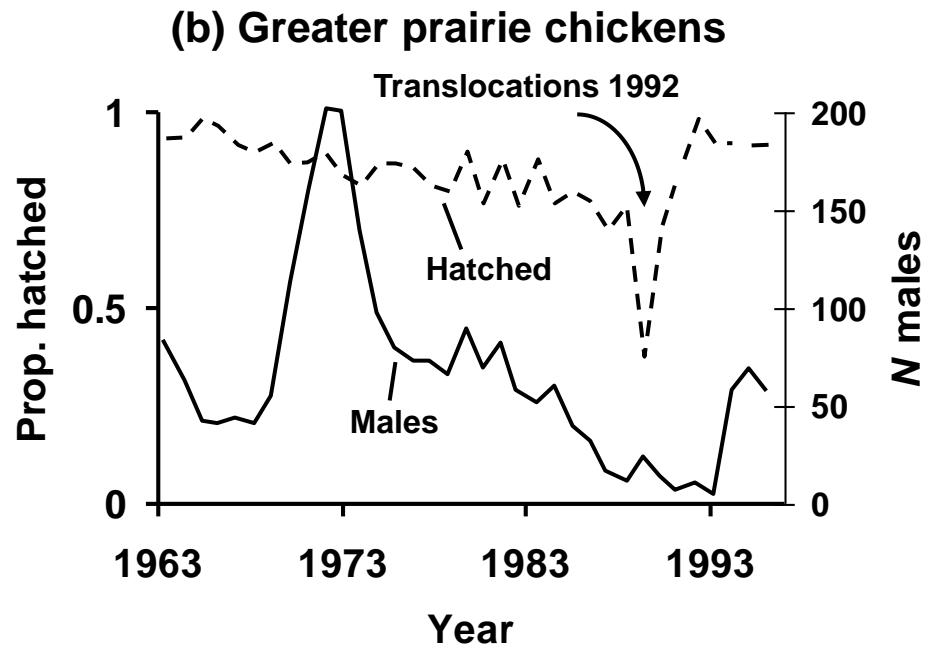
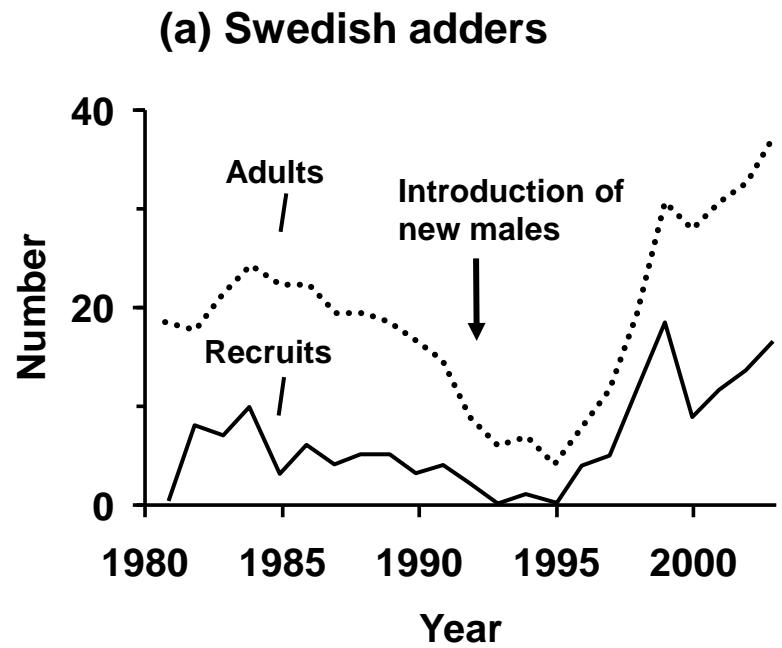
Distortion of sex-ratio by poaching in Asian elephants

- Ivory poaching
- Periyar Reserve, India
 - 6 males : 605 females
 - $N_e = 24$
 - Females that do not breed early, remain barren

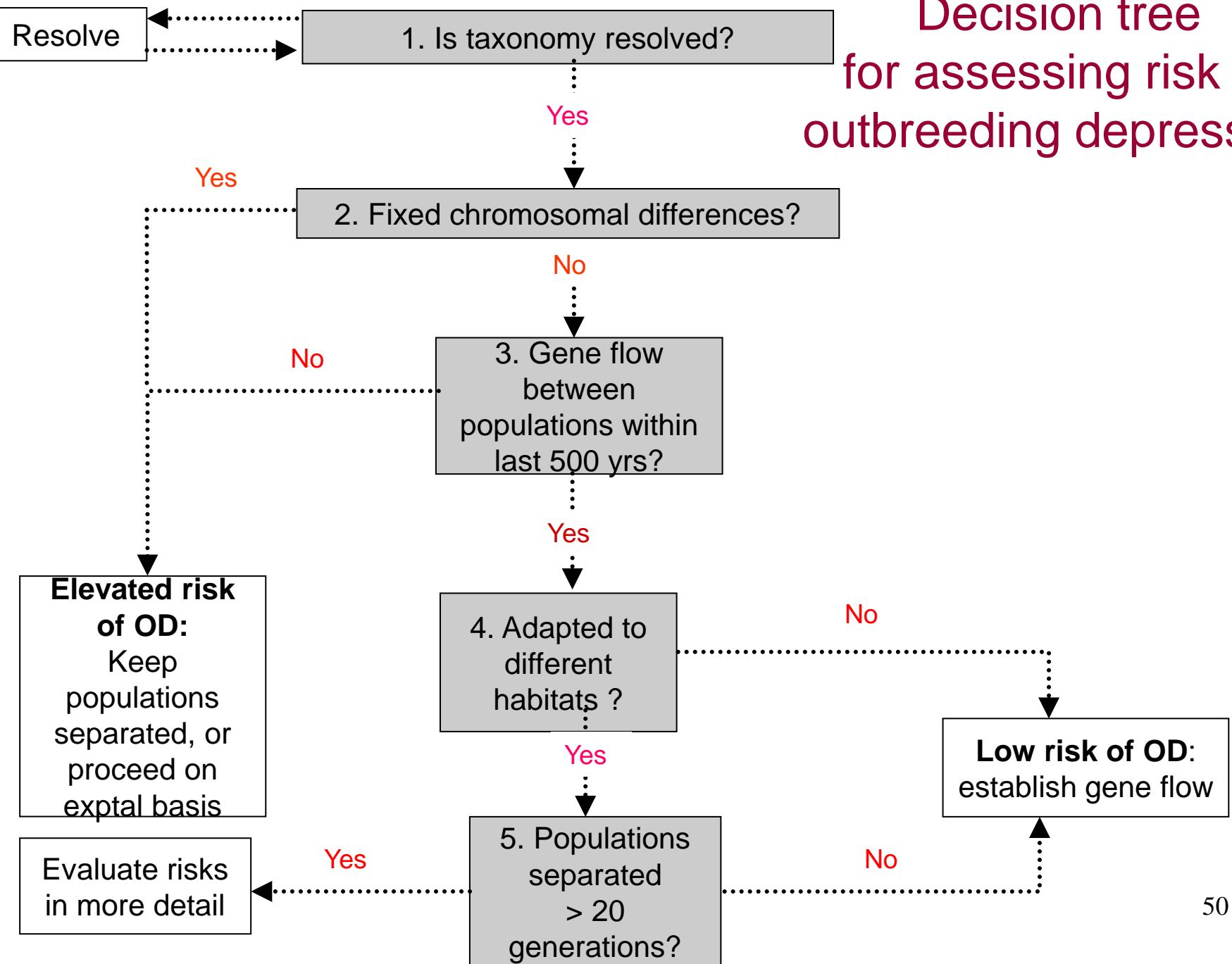


What can be done to avoid the adverse effect of selective harvest?

- Alter harvest regime
- Keep some pops without harvest
(e.g. marine protected areas)
- Problem: often illegal & difficult to police



Decision tree for assessing risk of outbreeding depression

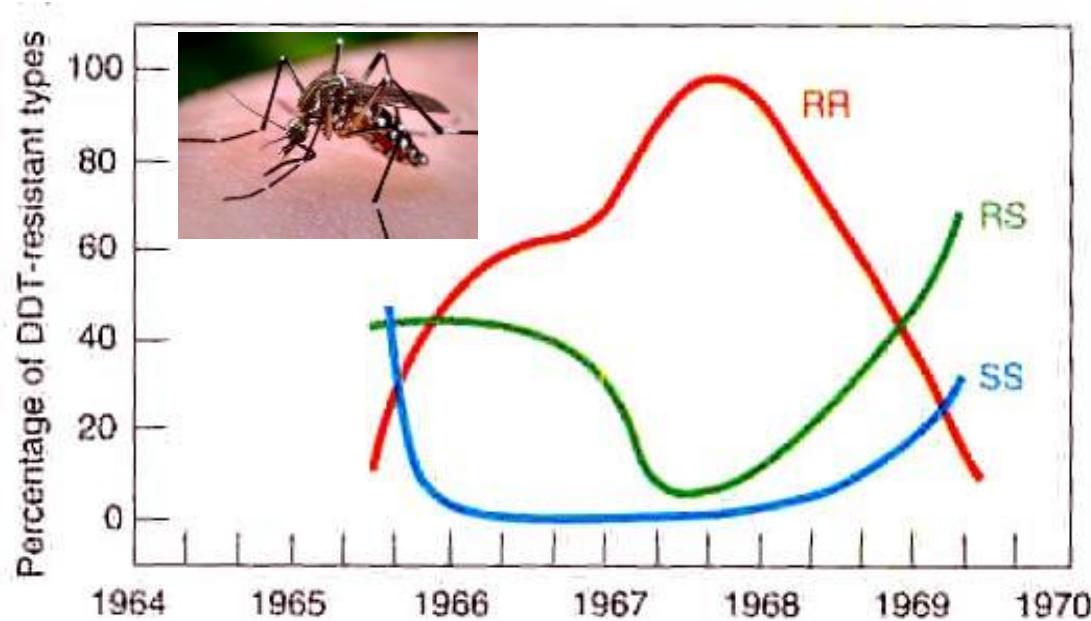


Selection and Adaptive Potential

Adam Stow

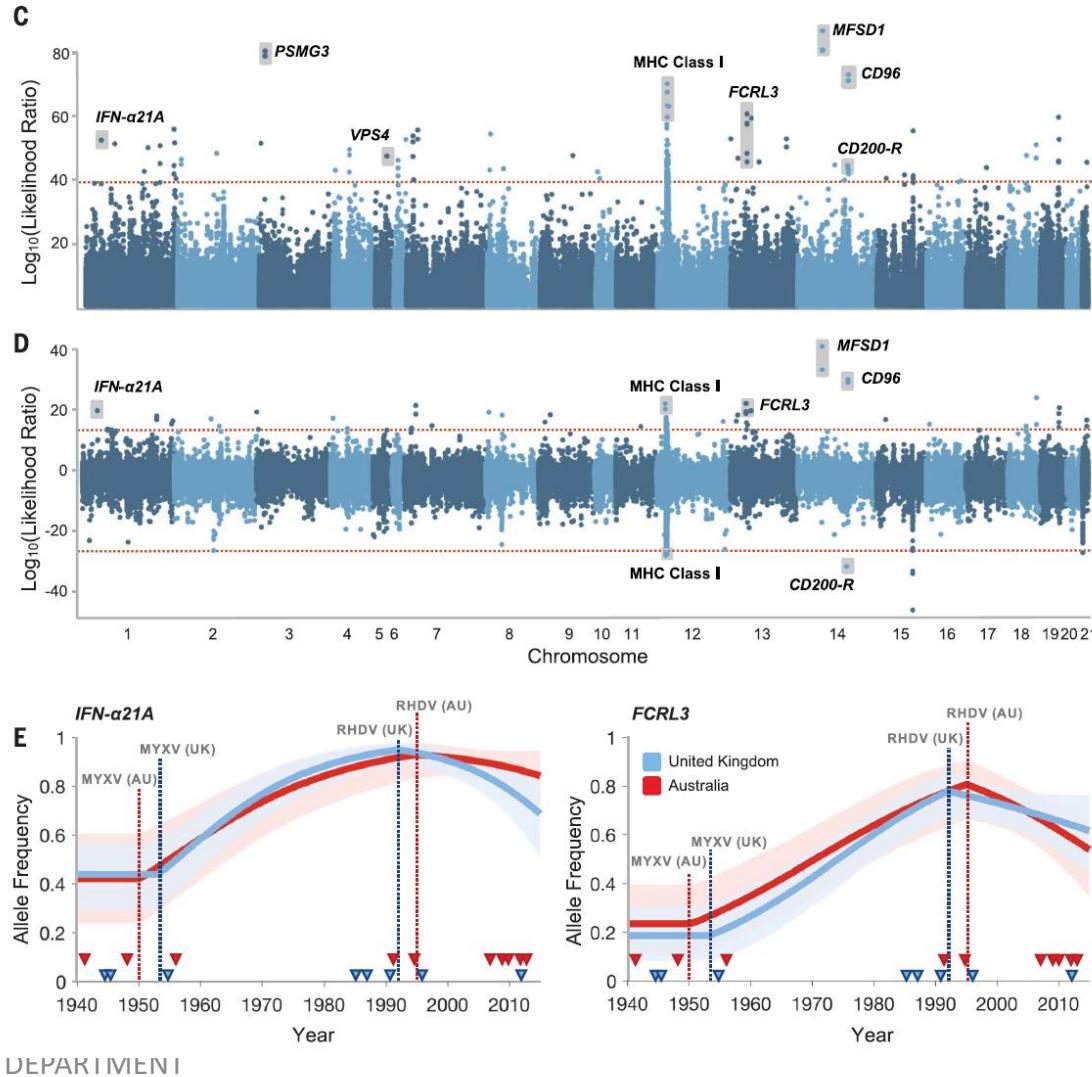
What is adaptive potential?

The ability of a population to respond adaptively to a new selective pressure



How genotype frequencies among populations of *A. aegypti* mosquito larvae change in response to Insecticide. (a) Mosquitoes and larvae. (b) Changing proportions of resistance genotypes of *A. aegypti* (larvae) under selection with DDT, and after selection was relaxed, in a suburb of Bangkok, Thailand.

Enough standing genetic variation for evolution



Alleles selected for
by myxoma virus

Decrease in
Myxoma virulence

Can we use genomics to estimate adaptive potential?

Using genomics to characterize evolutionary potential for conservation of wild populations

Katherine A. Harrisson, Alexandra Pavlova, Marina Telonis-Scott and Paul Sunnucks

Evolutionary Applications

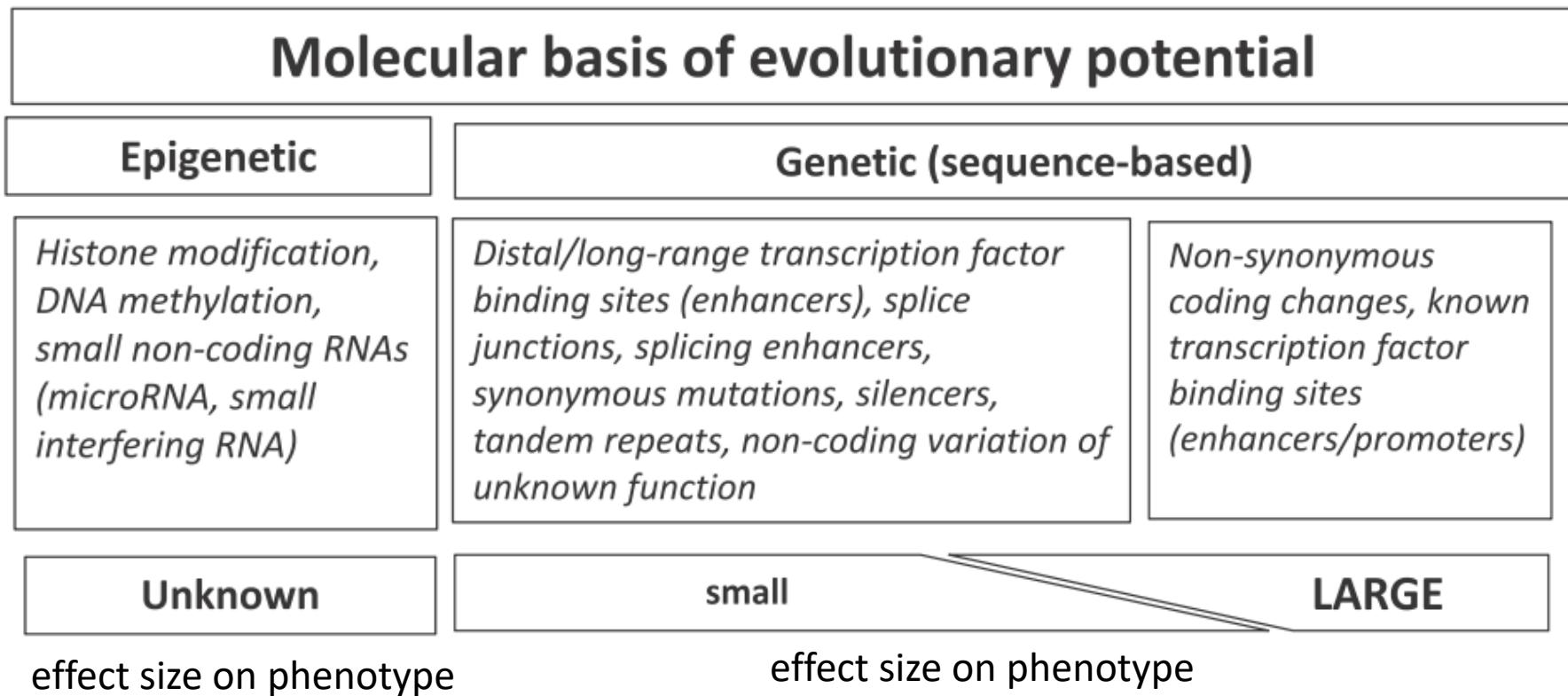
doi:10.1111/eva.12149

We could try and get estimates from genes of known function....but,

Given the current levels of understanding of how genomes work in non-model organisms screening genome wide diversity will usually give a better estimate of evolutionary potential.

Two distinct components of evolutionary potential

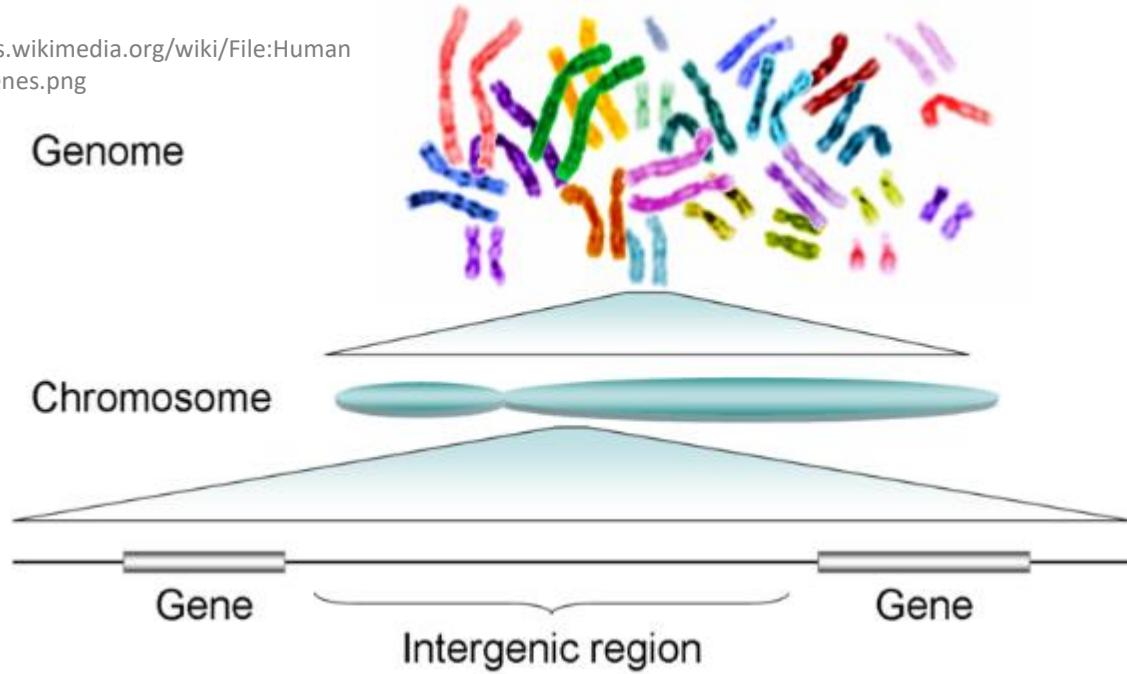
- **Genetic** (DNA-sequence-based)
 - **Epigenetic** (non-DNA-sequence-based)



Adaptive variation has been found in parts of the genome previously thought non-functional

Figure:

http://commons.wikimedia.org/wiki/File:Human_genome_to_genes.png



Comparing rat and mouse genomes, a great number of evolutionarily conserved sequences (i.e probably have a function) were intergenic, far from known genes

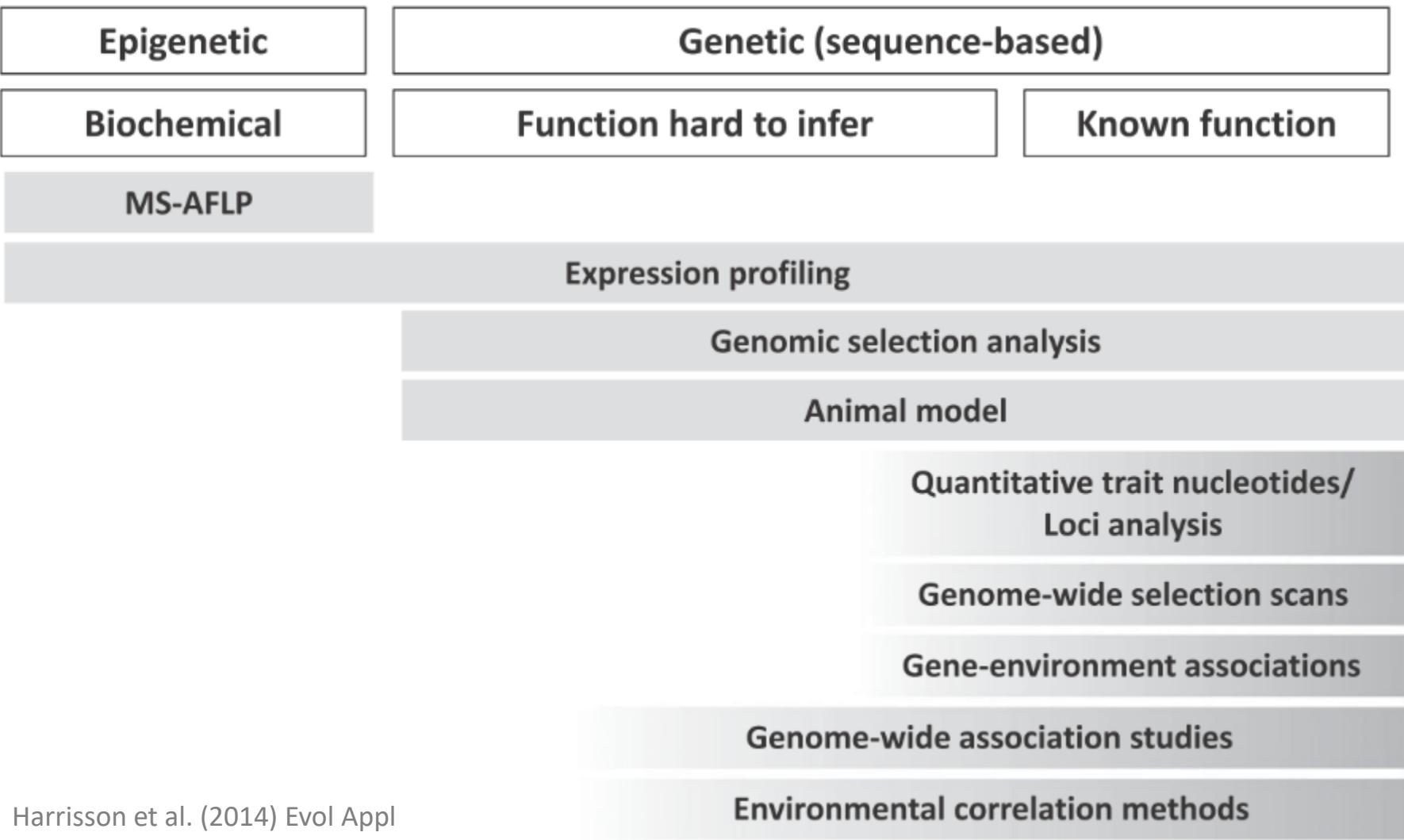
How much of the genome has a function?

- In humans only ~ 1.5% of sequence codes for amino acids
- at least ~ 10–15% of sequence is evolutionarily conserved in a way that suggests it is potentially under selection and has a function

To better understand evolutionary potential and what drives localised adaptation it's useful to find parts of the genome influenced by selection.

How is it done, and how effective is it?

Methods to infer selection are complex, numerous, emerging



Three main recent insights into evolutionary potential
summarized by Harrisson et al. 2014:

- (1) Rapid adaptive evolution is driven predominantly by changes in gene expression
- (2) Most traits are polygenic (controlled by lots of genes of small effect)
- (3) Most recent adaptation is due to subtle shifts of allele frequencies

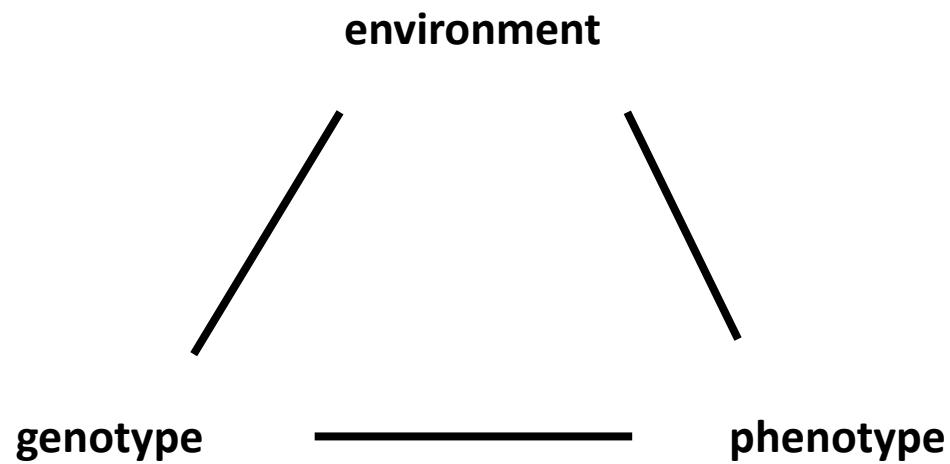
Detecting environmental selection using genomic data

Goals

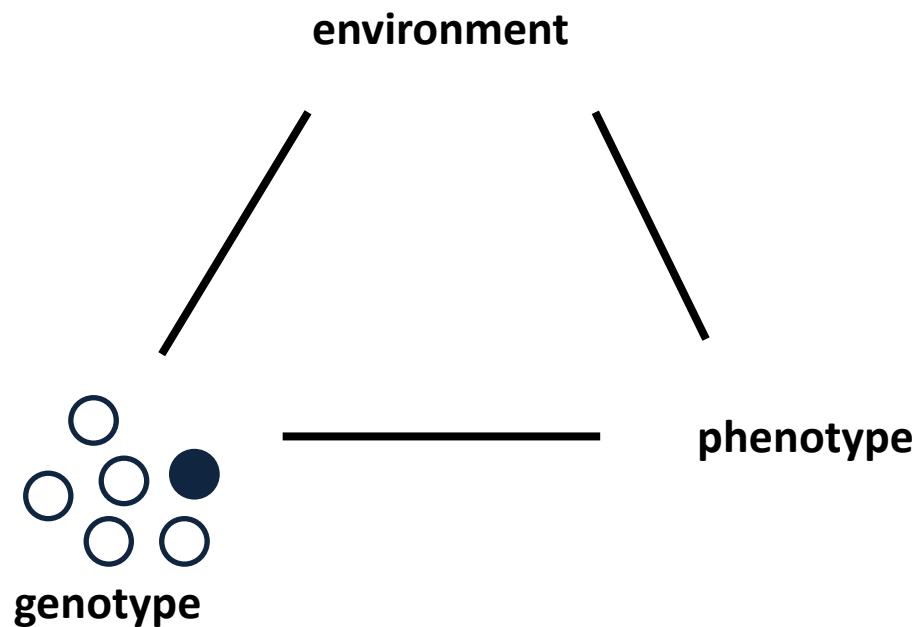
environmental variables that shape adaptive variation

gene variants that drive local adaptation

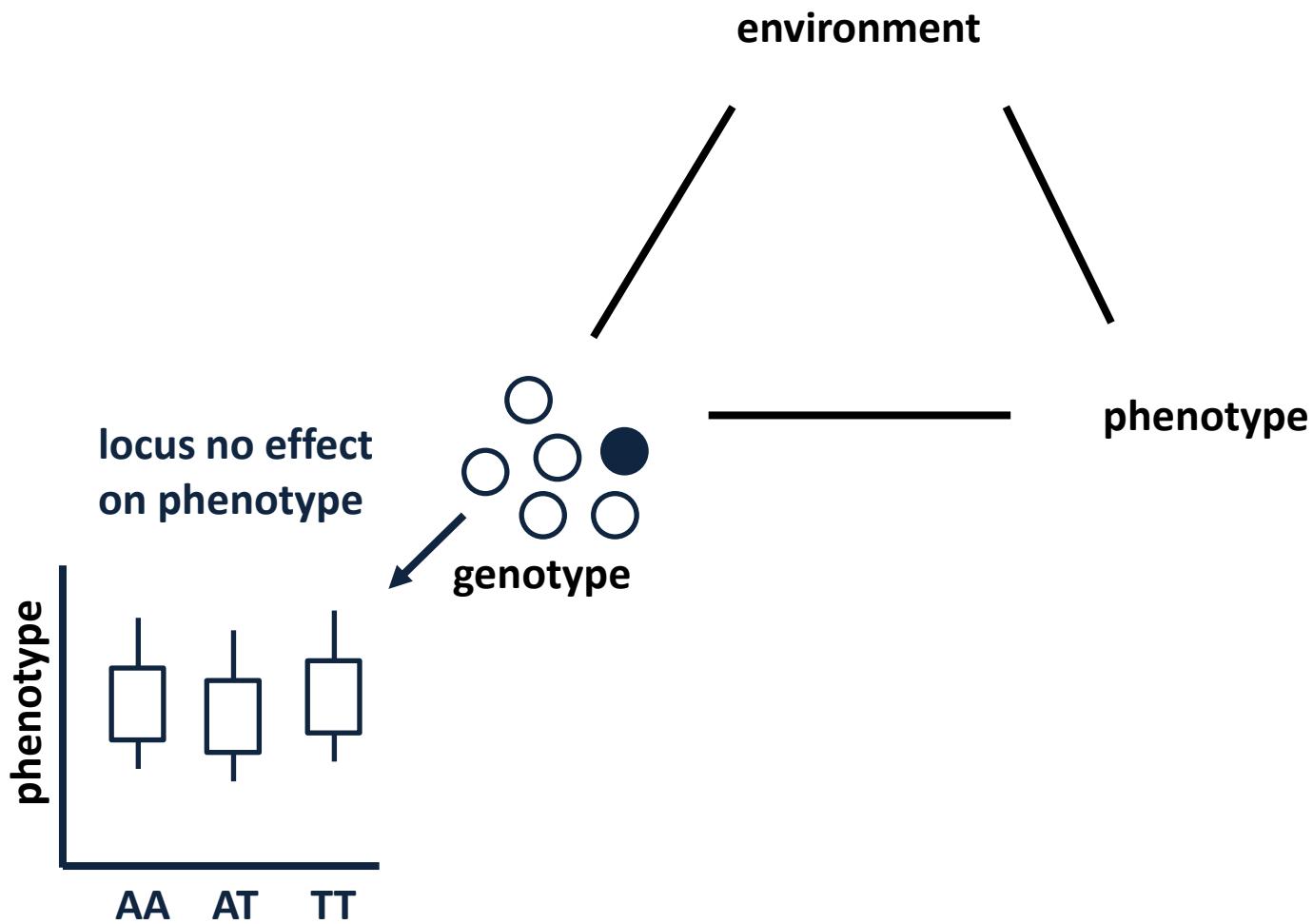
Adaptation: process and patterns



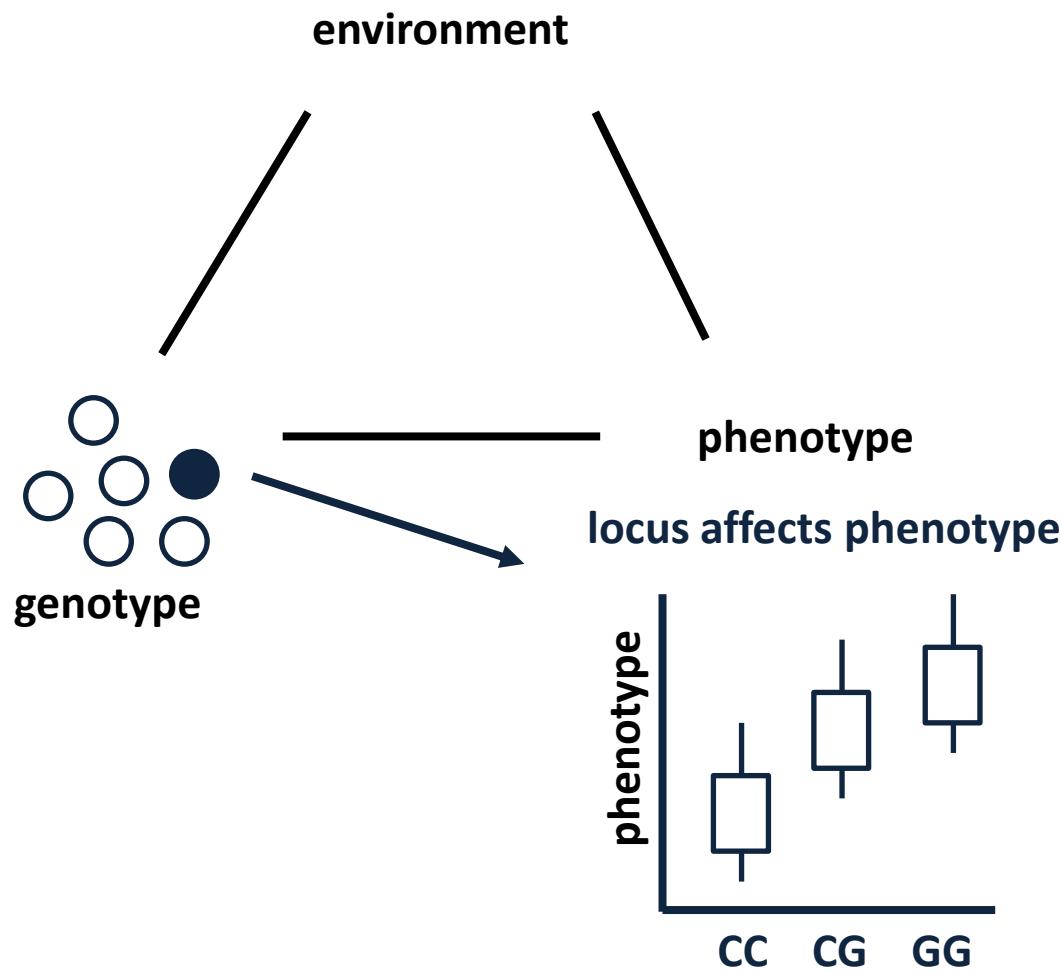
Adaptation: process and patterns



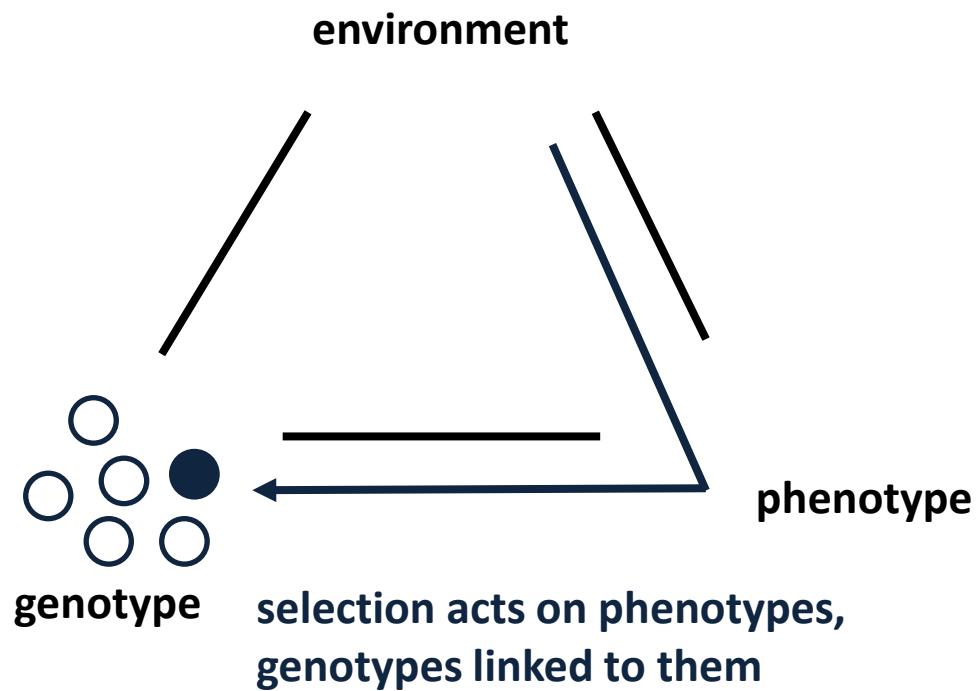
Adaptation: process and patterns



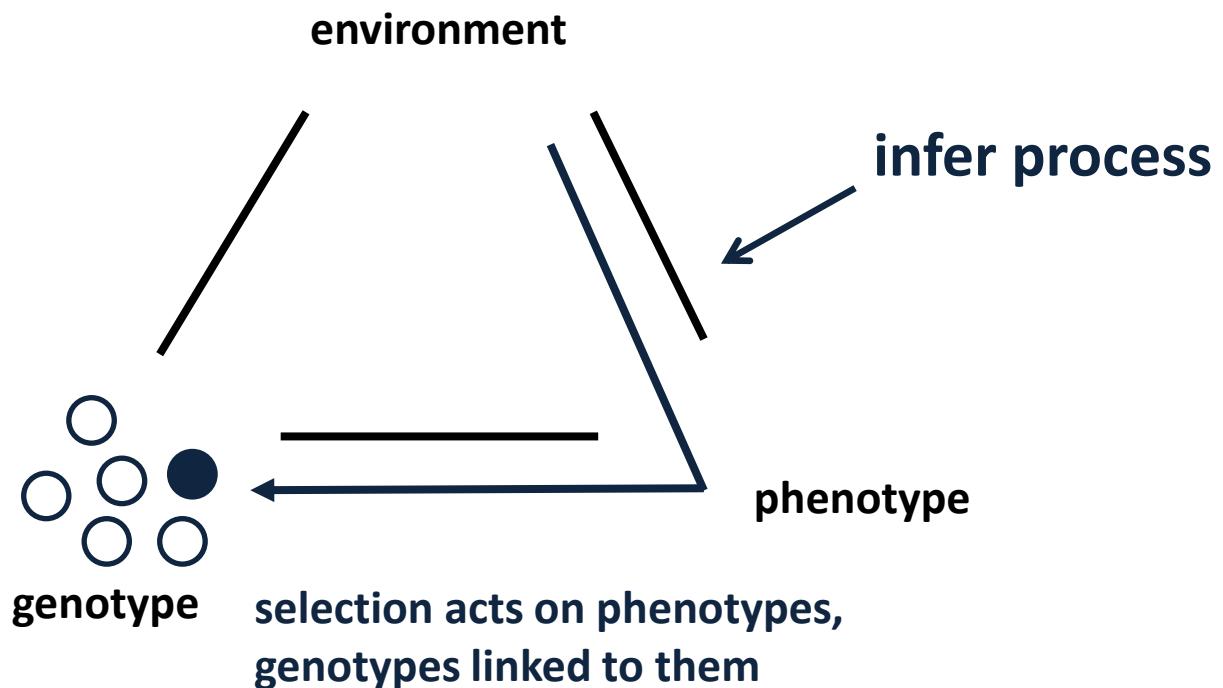
Adaptation: process and patterns



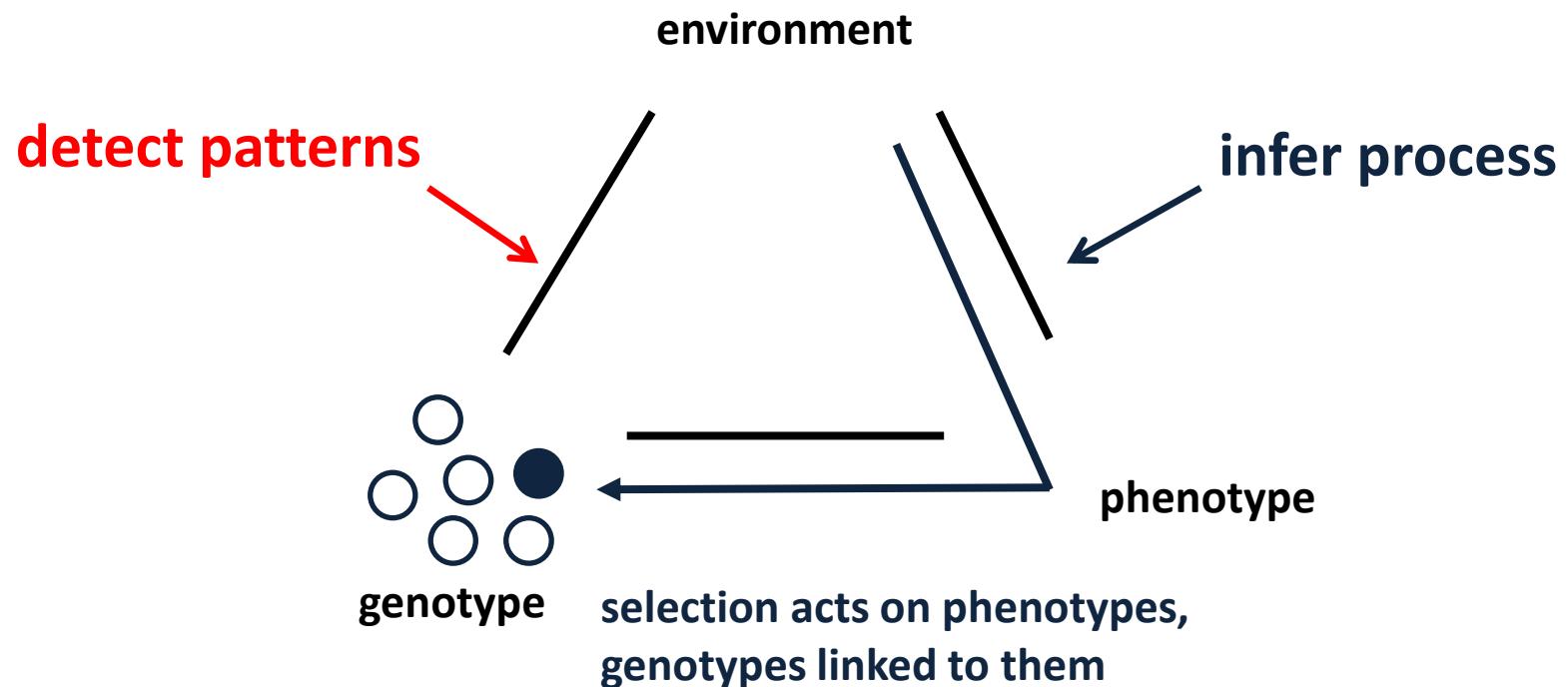
Adaptation: process and patterns



Adaptation: process and patterns



Adaptation: process and patterns



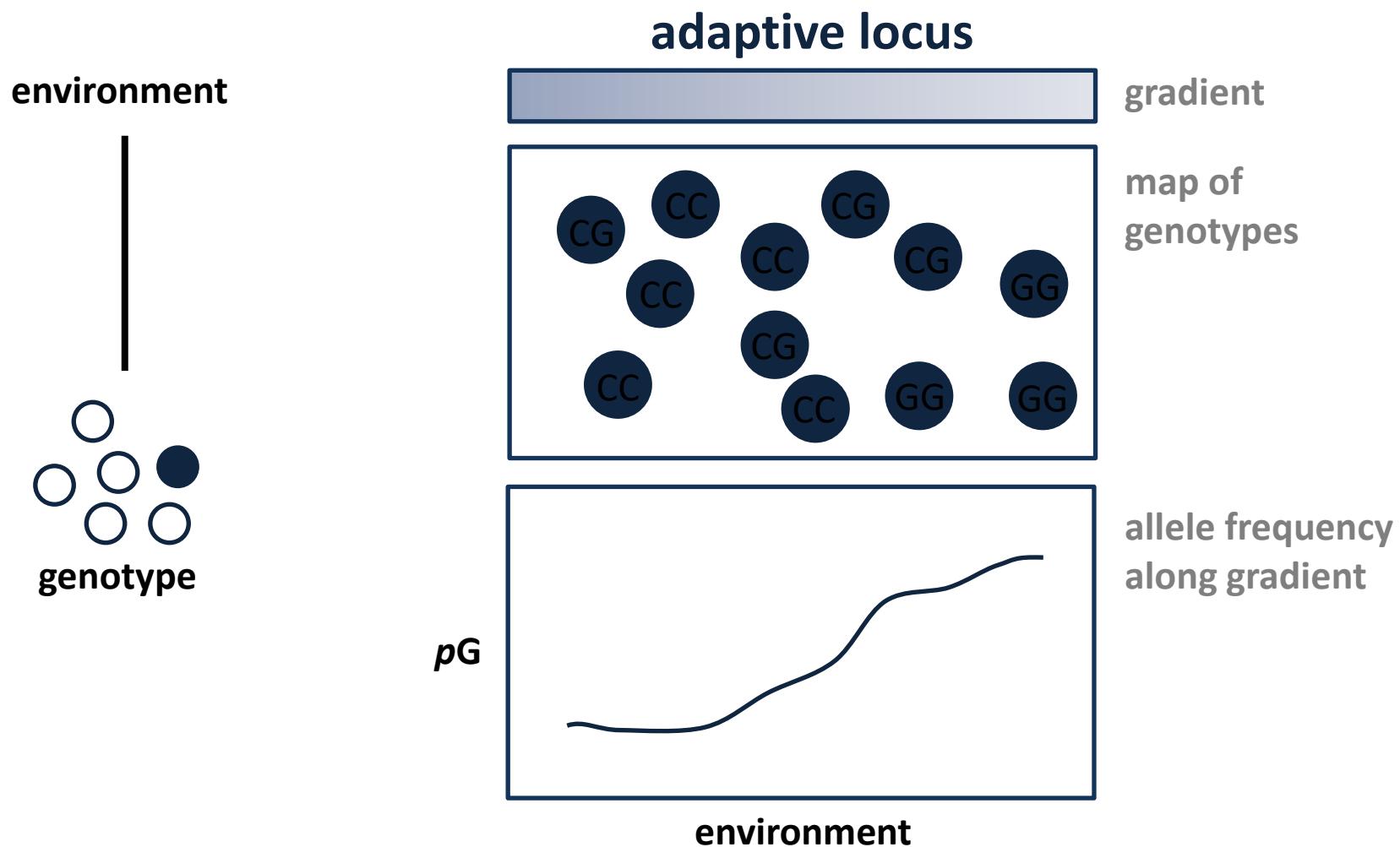
Adaptation: process and patterns

environment

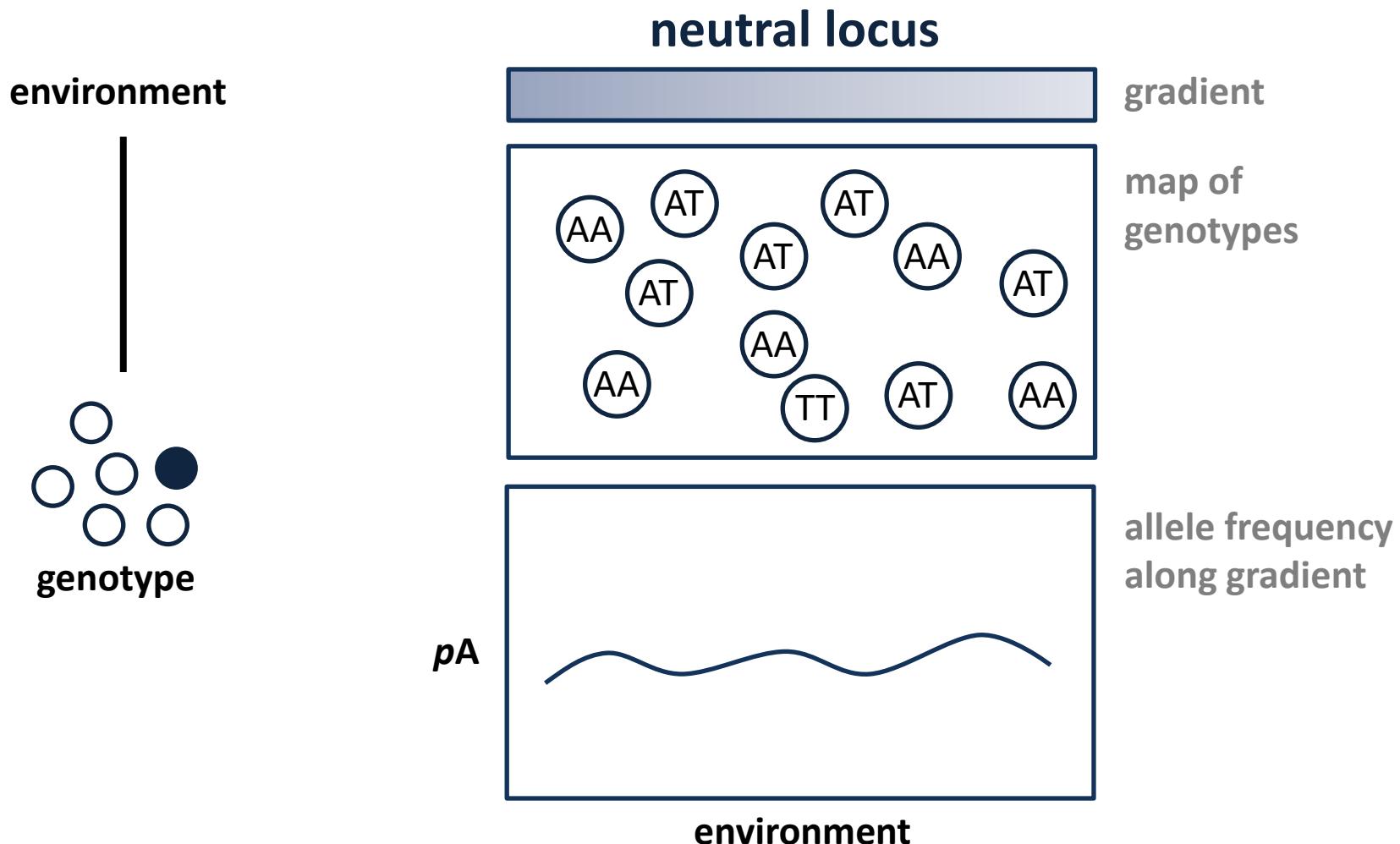


genotype

Adaptation: process and patterns

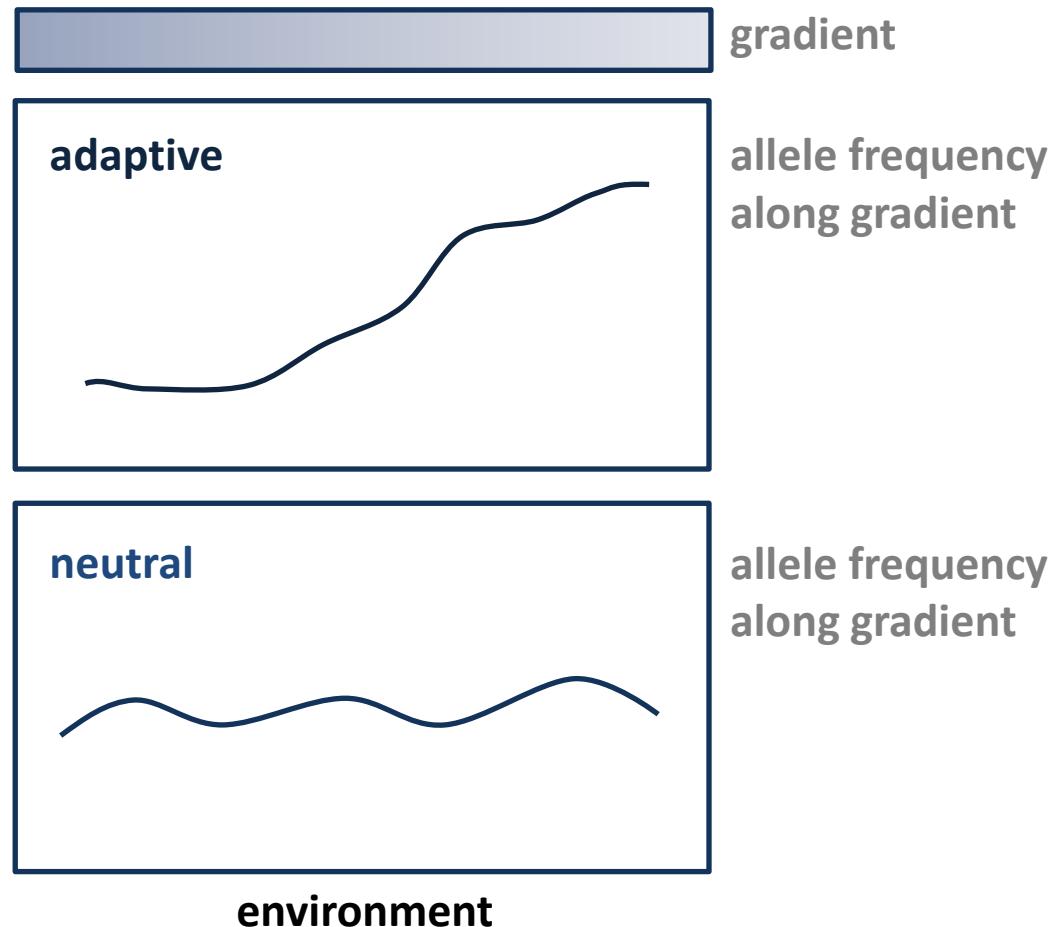
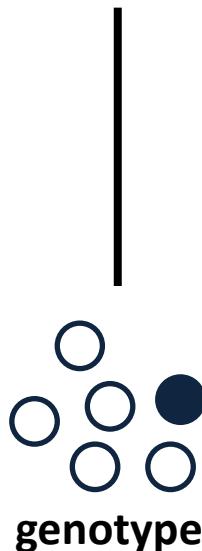


Adaptation: process and patterns

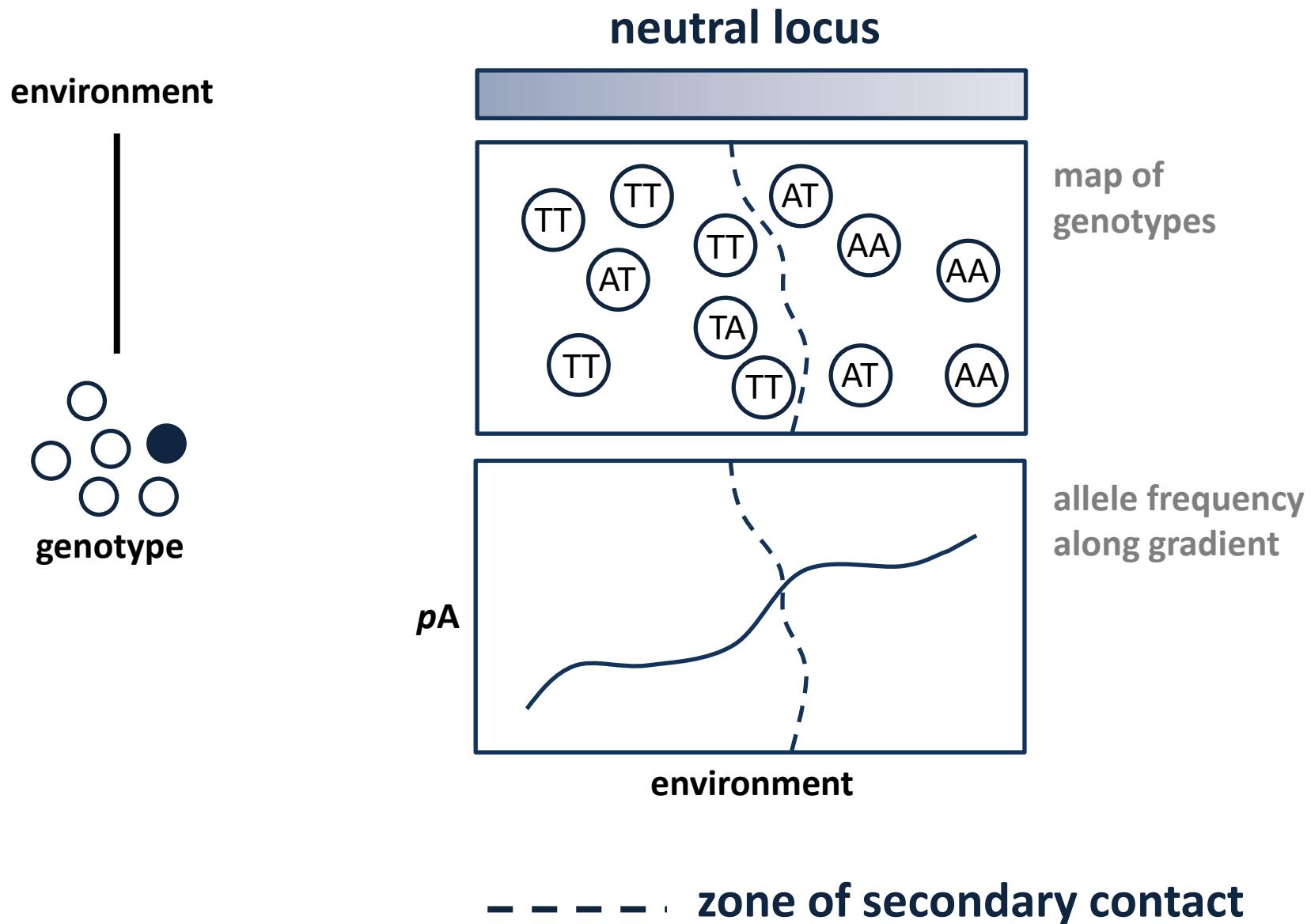


Adaptation: process and patterns

environment

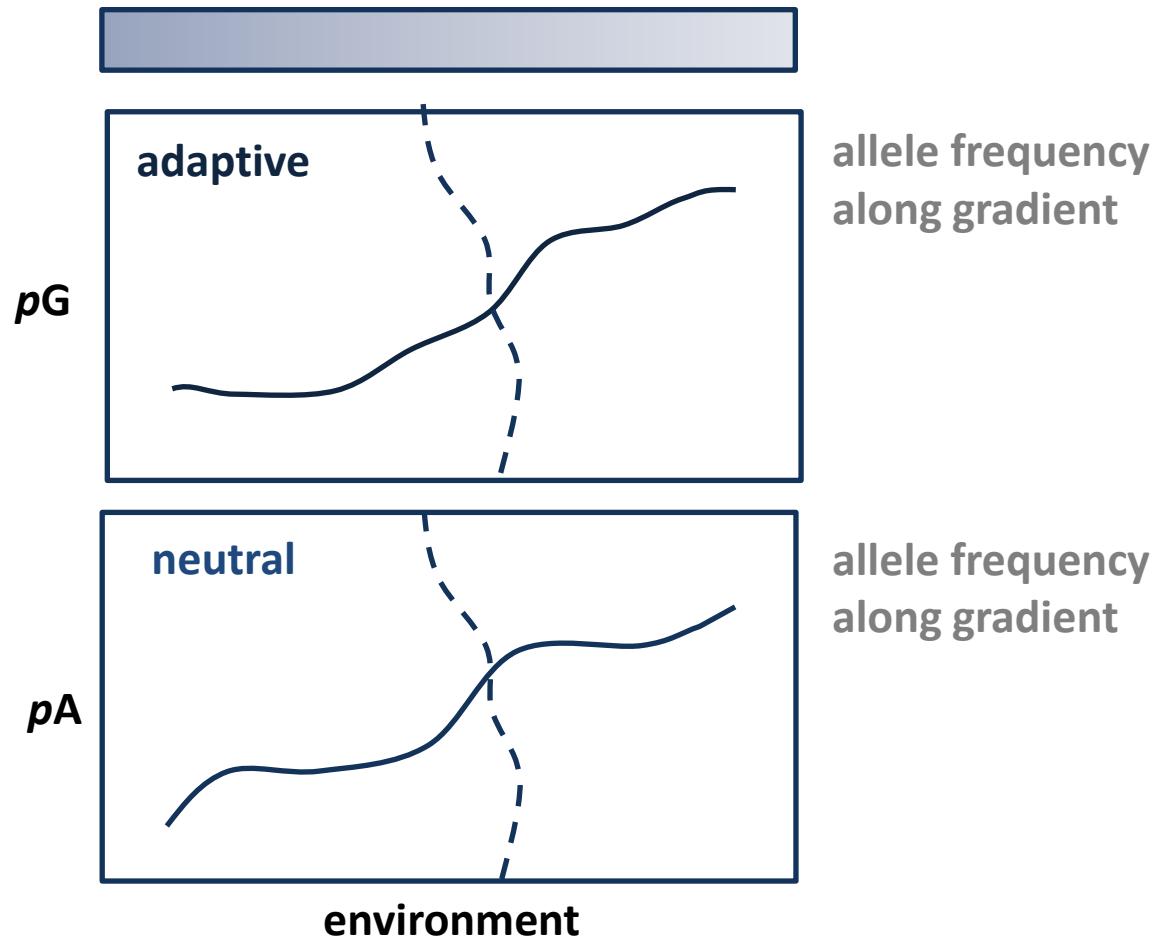


Adaptation: process and patterns



Key challenge: distinguishing from neutral variation

environment



— — — · zone of secondary contact

Data

environment



Sample	Pop	Lat	Lon	RF	Temp
S1	P1	-15.1	132.3	328.4	23.6
S2	P1	-15.1	132.3	328.4	23.6
S3	P1	-15.1	132.3	328.4	23.6
S4	P1	-15.1	132.3	328.4	23.6
S5	P2	-15.7	135.4	312.8	21.2
S6	P2	-15.7	135.4	312.8	21.2
S7	P2	-15.7	135.4	312.8	21.2

Data

environment



Sample	Pop	Lat	Lon	RF	Temp
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S6	P2	-15.7	135.4	312.8	21.2
S7	P2	-15.7	135.4	312.8	21.2

sampling

Data

environment



Sample

S1
S2
S3
S4
S5
S6
S7

Sample	Lat	Lon
S1	-15.1	132.3
S2	-15.2	132.5
S3	-15.4	132.1
S4	-15.4	132.7
S5	-15.5	133.6
S6	-15.7	134.2
S7	-15.7	135.4

RF	Temp
328.4	23.6
329.2	23.3
326.3	24.0
324.4	22.4
318.6	22.6
314.8	21.8
312.8	21.2

measure these
(map datum)

how to get
these?

Data

environment



Sample

S1
S2
S3
S4
S5
S6
S7

Lat Lon

-15.1	132.3
-15.2	132.5
-15.4	132.1
-15.4	132.7
-15.5	133.6
-15.7	134.2
-15.7	135.4

RF Temp

328.4	23.6
329.2	23.3
326.3	24.0
324.4	22.4
318.6	22.6
314.8	21.8
312.8	21.2

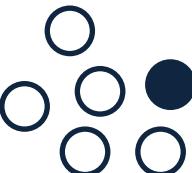
measure these
(map datum)

how to get
these?

WorldClim
(consider local
measurements...)

Data

environment	Sample	Lat	Lon	RF	Temp
	S1	-15.1	132.3	328.4	23.6
	S2	-15.2	132.5	329.2	23.3
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	S4	-15.4	132.7	324.4	22.4
	S5	-15.5	133.6	318.6	22.6
	S6	-15.7	134.2	314.8	21.8
	S7	-15.7	135.4	312.8	21.2



The diagram illustrates the relationship between environment and genotype. On the left, there is a red-bordered box containing the word "environment". A vertical line connects this box to a cluster of seven circles below it. The cluster consists of six white circles and one dark blue circle, representing different genotypes.

environment

genotype

Data

environment



Sample	Lat	Lon	RF	Temp
S1	-15.1	132.3	328.4	23.6
S2	-15.2	132.5	329.2	23.3
S3	-15.4	132.1	326.3	24.0
S4	-15.4	132.7	324.4	22.4
S5	-15.5	133.6	318.6	22.6
S6	-15.7	134.2	314.8	21.8
S7	-15.7	135.4	312.8	21.2

RF Temp

328.4 23.6
329.2 23.3
326.3 24.0
324.4 22.4
318.6 22.6
314.8 21.8
312.8 21.2

many choices

what matters to
your species?

Data

environment



genotype

Sample	Lat	Lon	RF	Temp
S1	-15.1	132.3	328.4	23.6
S2	-15.2	132.5	329.2	23.3
S3	-15.4	132.1	326.3	24.0
S4	-15.4	132.7	324.4	22.4
S5	-15.5	133.6	318.6	22.6
S6	-15.7	134.2	314.8	21.8
S7	-15.7	135.4	312.8	21.2

many choices

all available vs.
hypothesis-driven

what matters to
your species?

kinds of variables
(e.g. aquatic life-stages)

thresholds
(e.g. frost intolerance)

Data

environment



Sample	Lat	Lon	RF	T. (max)	T. (min)
S1	-15.1	132.3	328.4	29.6	23.6
S2	-15.2	132.5	329.2	29.3	23.4
S3	-15.4	132.1	326.3	30.0	25.5
S4	-15.4	132.7	324.4	28.4	22.4
S5	-15.5	133.6	318.6	28.6	24.1
S6	-15.7	134.2	314.8	27.8	20.6
S7	-15.7	135.4	312.8	27.2	22.1

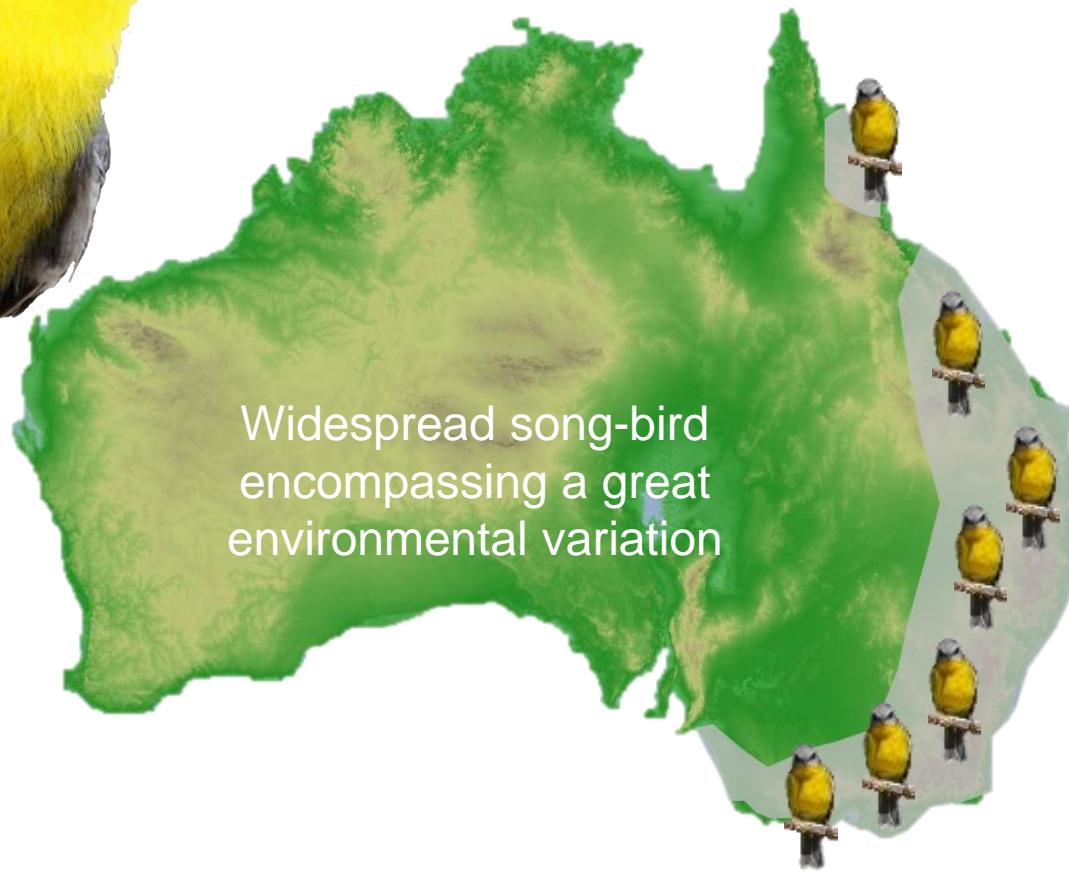
T. (min)



T. (max)

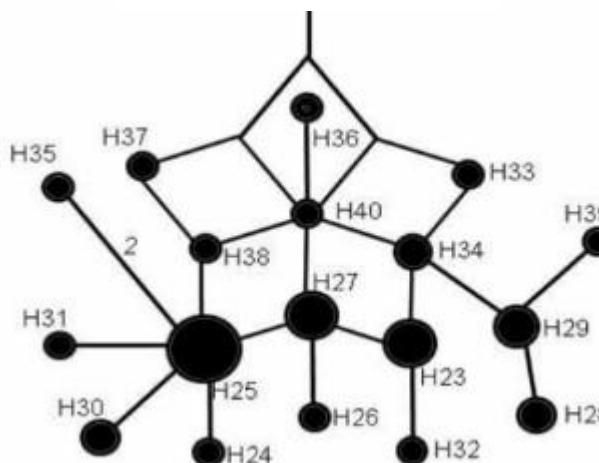
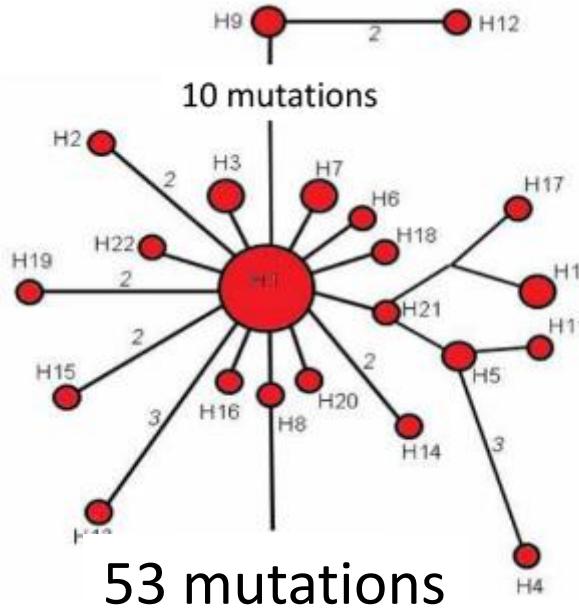
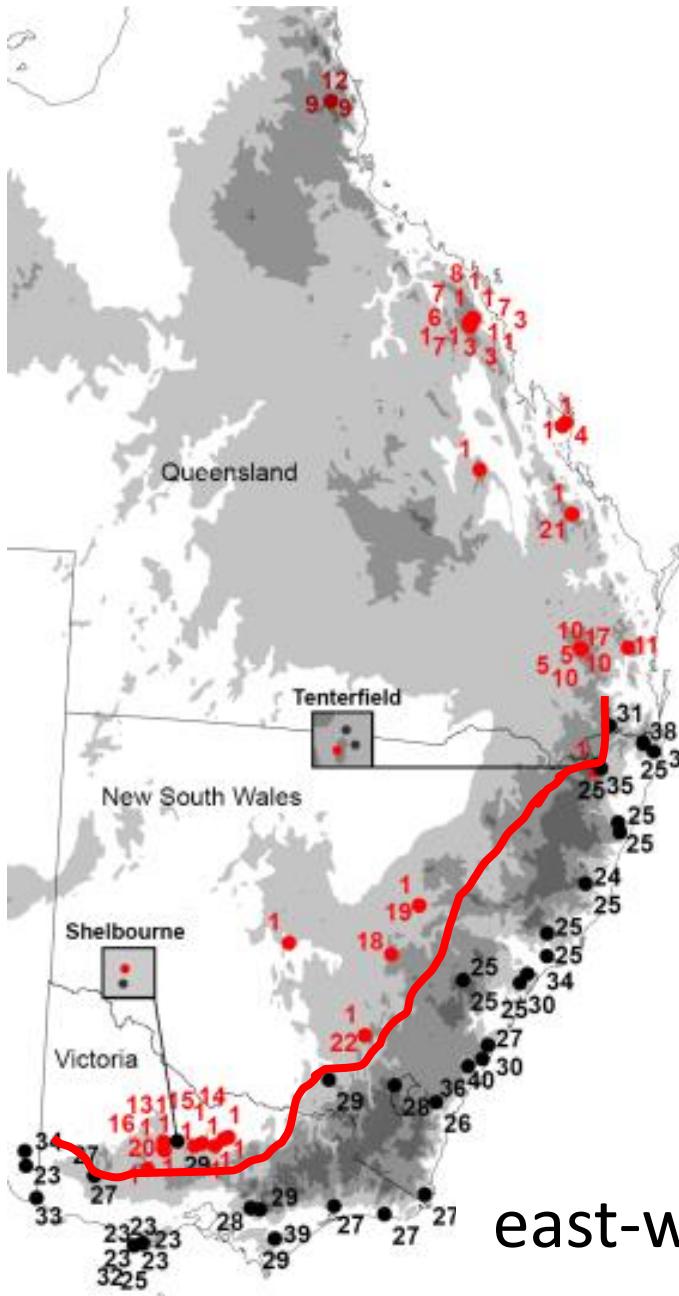
A close-up photograph of an Eastern Yellow Robin perched on a rough, textured tree trunk. The bird has a bright yellow breast and belly, a greyish-blue cap, and a dark grey back.

Eastern Yellow Robin



Widespread song-bird
encompassing a great
environmental variation

Pattern: Mitochondrial DNA sequences

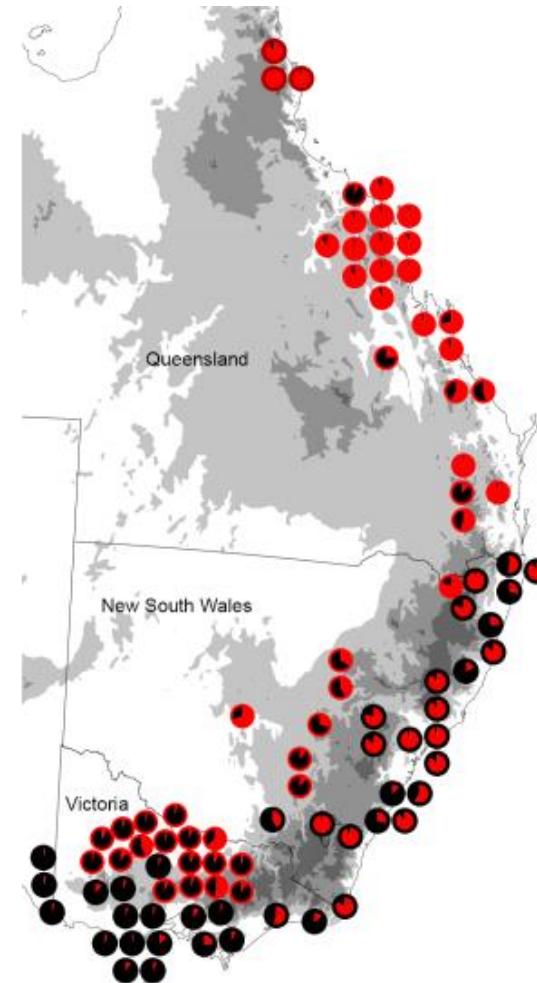


east-west split across Gt. Dividing Range

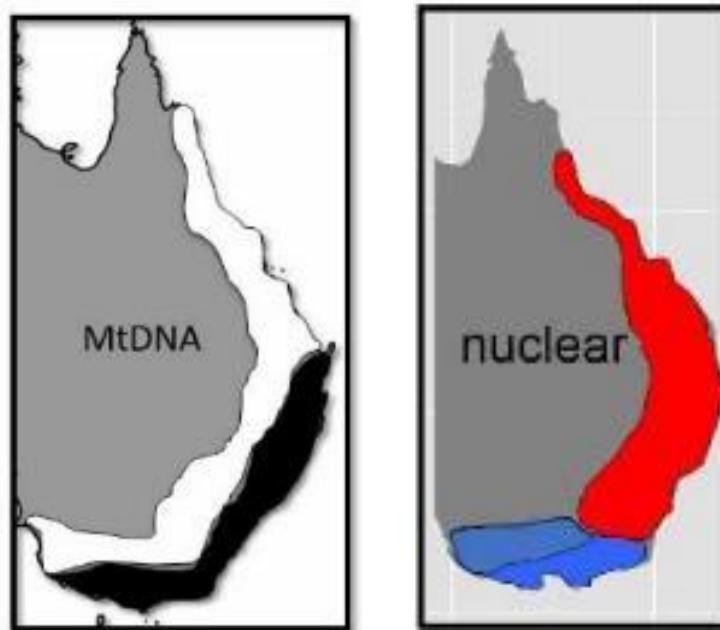


Pattern: Nuclear microsatellites

north-south pattern



Summary 2 genomes, completely different patterns



Having ruled out the boring alternatives, we inferred interesting female-associated selection:

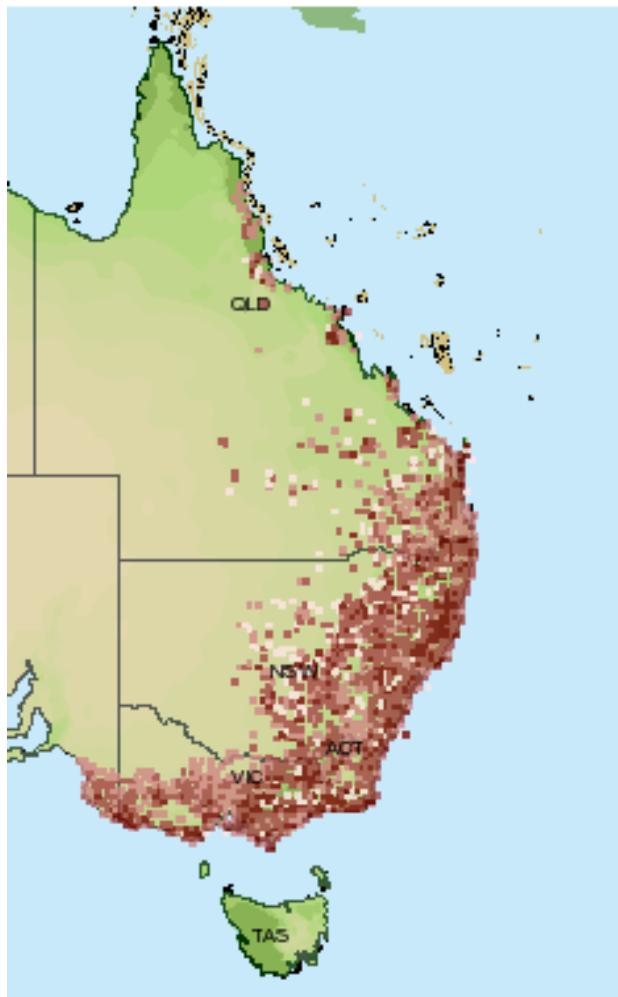
Reasons in Pavlova *et al.* 2013 Perched at the mito-nuclear crossroads: divergent mitochondrial lineages correlate with environment in the face of ongoing nuclear gene flow in an Australian bird. *Evolution* 67, 3412–3428



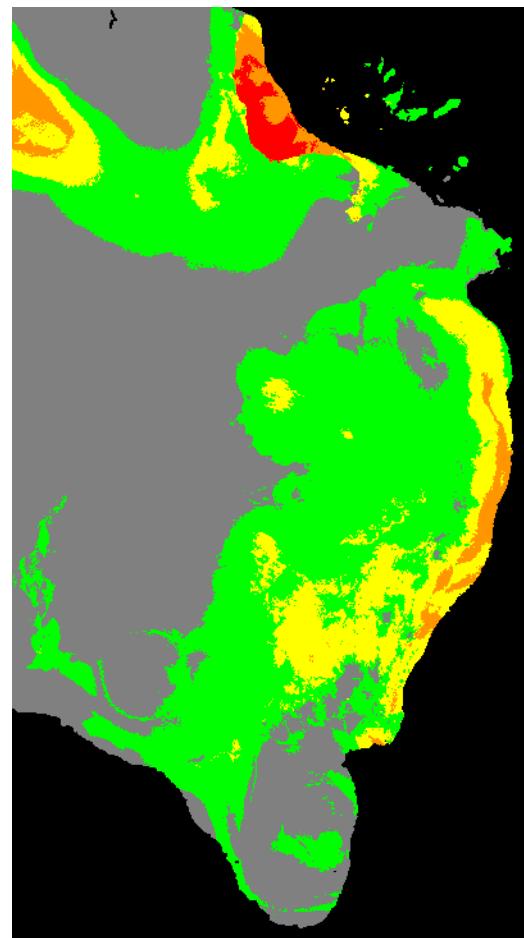
Sasha Pavlova

1. Vicariance: Is/was the Great Dividing Range a barrier? No

Current distribution is continuous



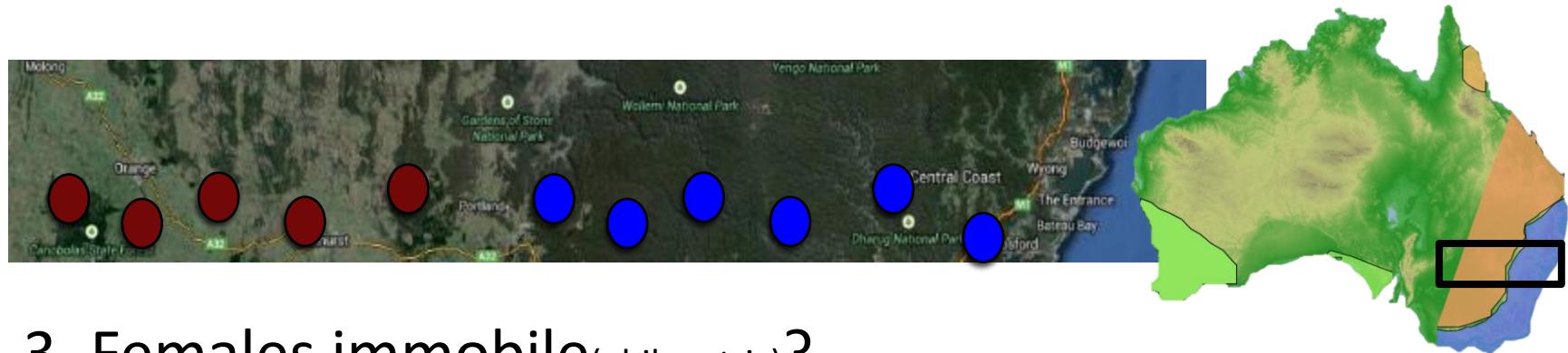
Last glacial maximum – no split over the GDR



2. Chance mtDNA split?

No

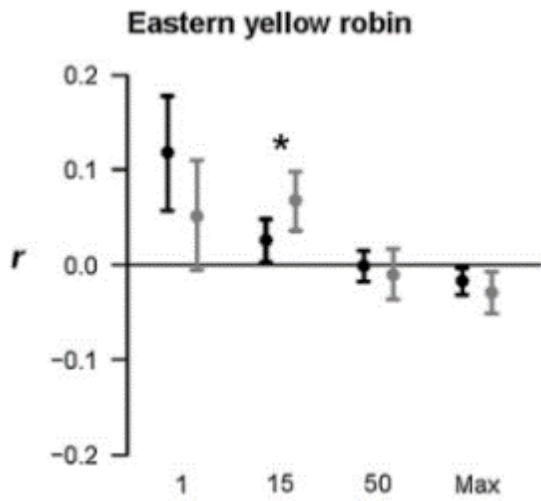
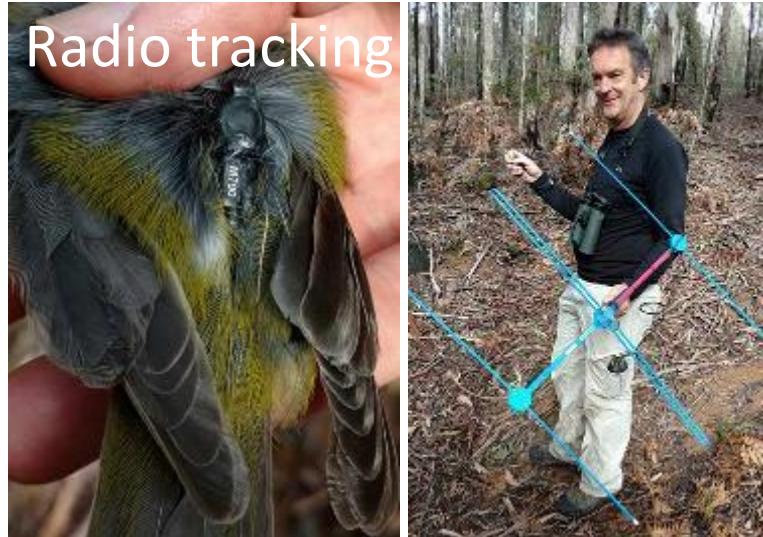
mtDNA lineages remain geographically distinct for >1500 km, even though they are well within the dispersal distance of a robin



3. Females immobile (philopatric)?

No Dispersal is female-biased

Radio tracking

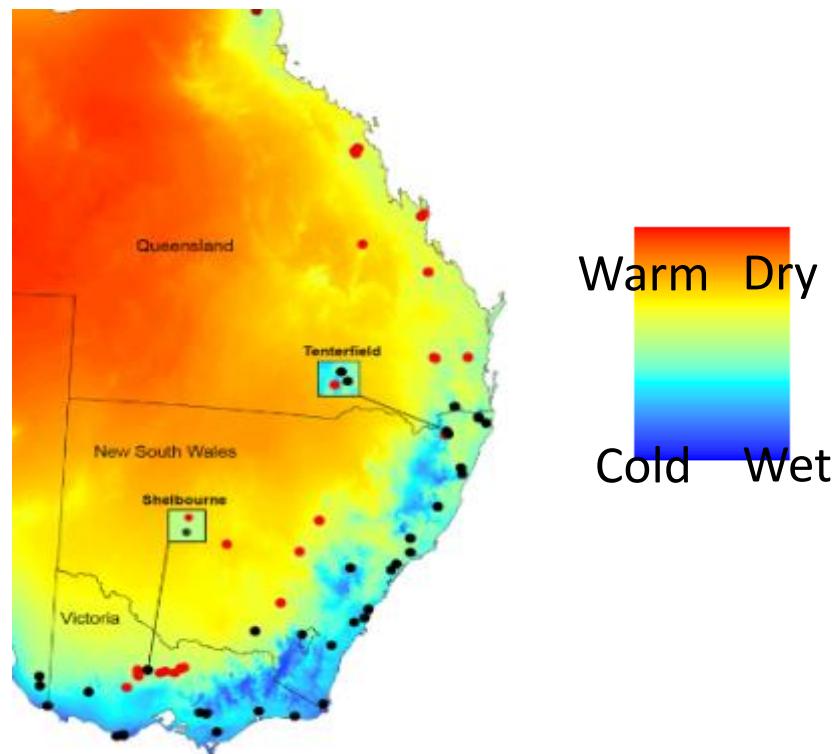


Harrisson et al. (2012) spatial autocorrelation
Female in grey, male in black

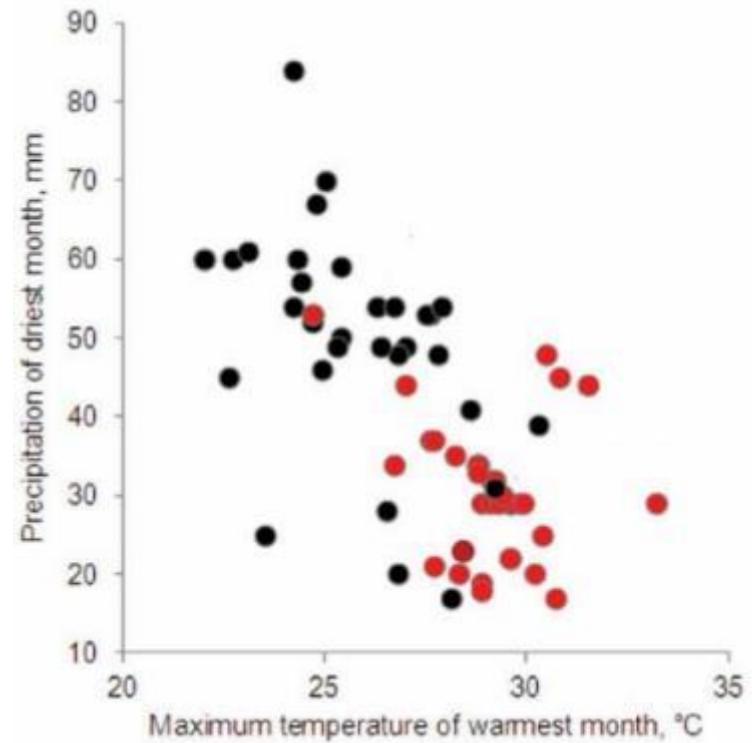
4. Female-associated selection – looks like it

Significant correlation of mtDNA with climate variables

Max. temp of the warmest month



Environmental Association Analysis (EAA)
(also called Genetic–environment associations (GEA): alleles correlate with environments)

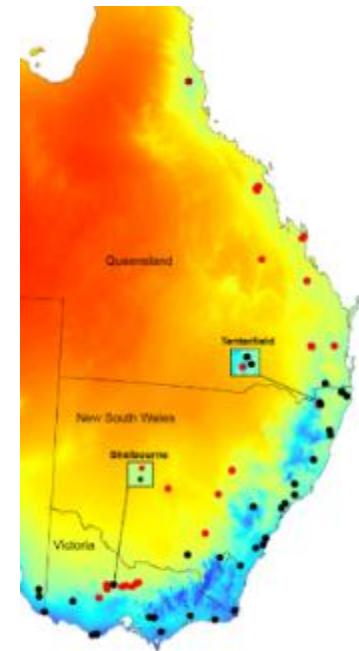


...remains significant after accounting for geography (by dbRDA)

EMPHASIS ...the fact that a potential driver remains significant after accounting for geography (by dbRDA)

Australia's climate is highly correlated with geography:
eg it's warmer in the north and cooler in the south

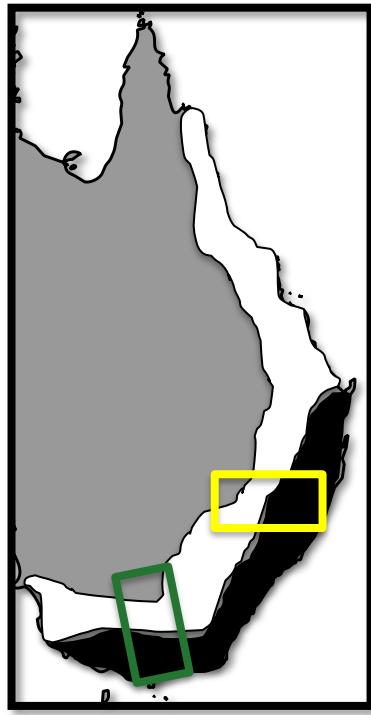
so any N-S pattern will look like it could be driven by climate



Need to test whether association of mtDNA with climate remains significant after accounting for ('partialling-out') the automatic correlation of climate with geography

eg using distance-based redundancy analysis (dbRDA)

Testing for nuclear genes with mitochondrial functioning being 'dragged along' with mtDNA during introgression

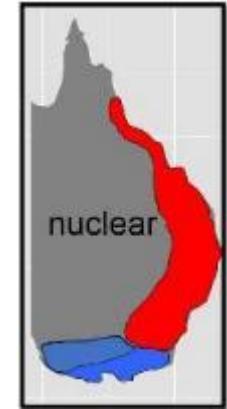


- Two transects across mtDNA split
- True replicates (different nuclear backgrounds)



Northern transect

Southern transect



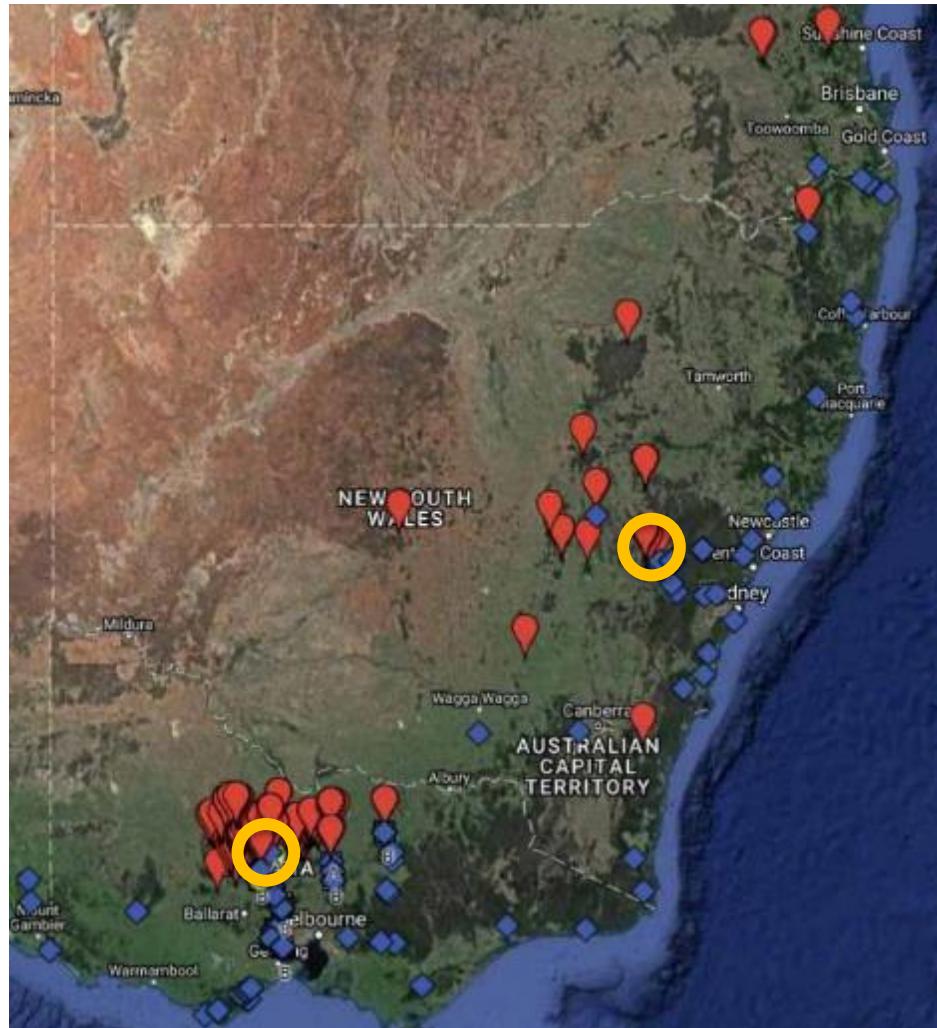
Nuclear genomewide marker scan >64,000 SNPs from short DNA sequences

Finding nuclear genes that differ between mitolineages

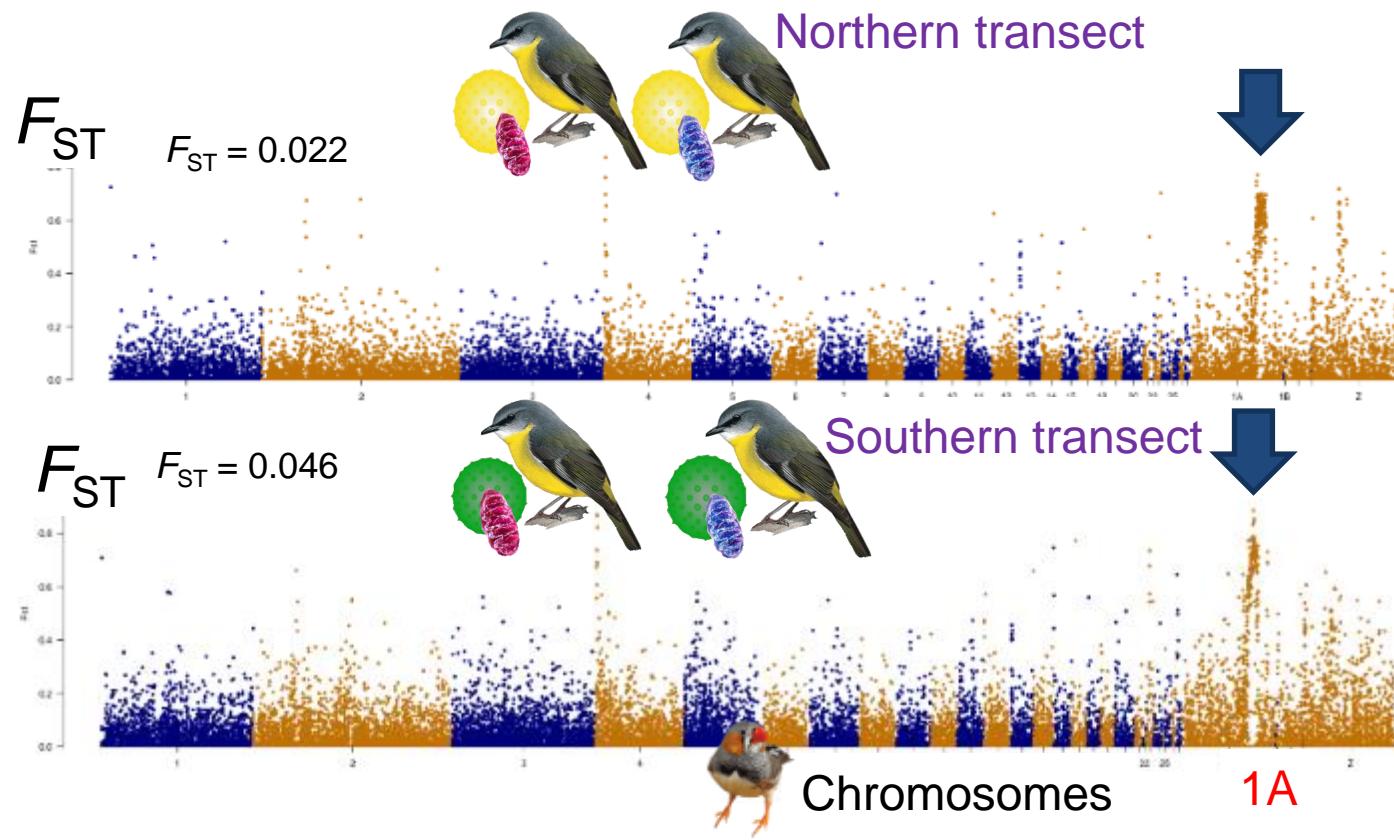
i.e. seeking high F_{ST}
between adjacent inland
and coastal robins

(This is a GWAS -
genome wide
association study)

Morales HE, Pavlova A, Amos JN, Major R, Kilian A, Greening C and Sunnucks P (2018)
Concordant divergence of mitogenomes and a
mitonuclear gene cluster in bird lineages
inhabiting different climates. *Nature Ecology & Evolution* 2, 1258–1267



Genomewide differentiation between inland and coastal mitolineages

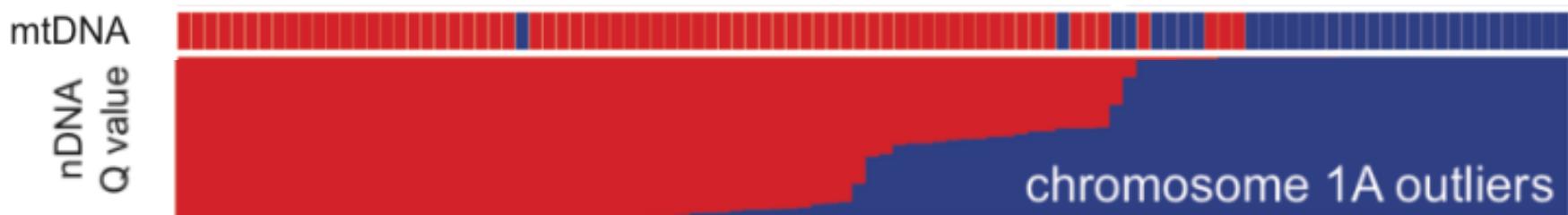


Outliers have average $F_{ST} > 0.48$

This chromosome 1A ‘island of divergence’ is special

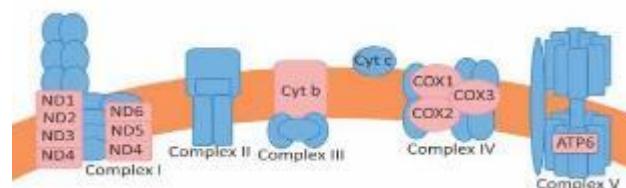


- (1) ~15 million nucleotides long containing ~340 genes
- (2) inherited as if one giant gene with two alleles
- (3) inherited along with mitochondrial DNA, even though the rest of the nuclear genome is not



- (4) Contains 32 genes that have functions in the mitochondrion
(a significant excess)

including 4 OXPHOS genes



Three of these OXPHOS genes are partners of the mtDNA ones also showing signals of positive selection

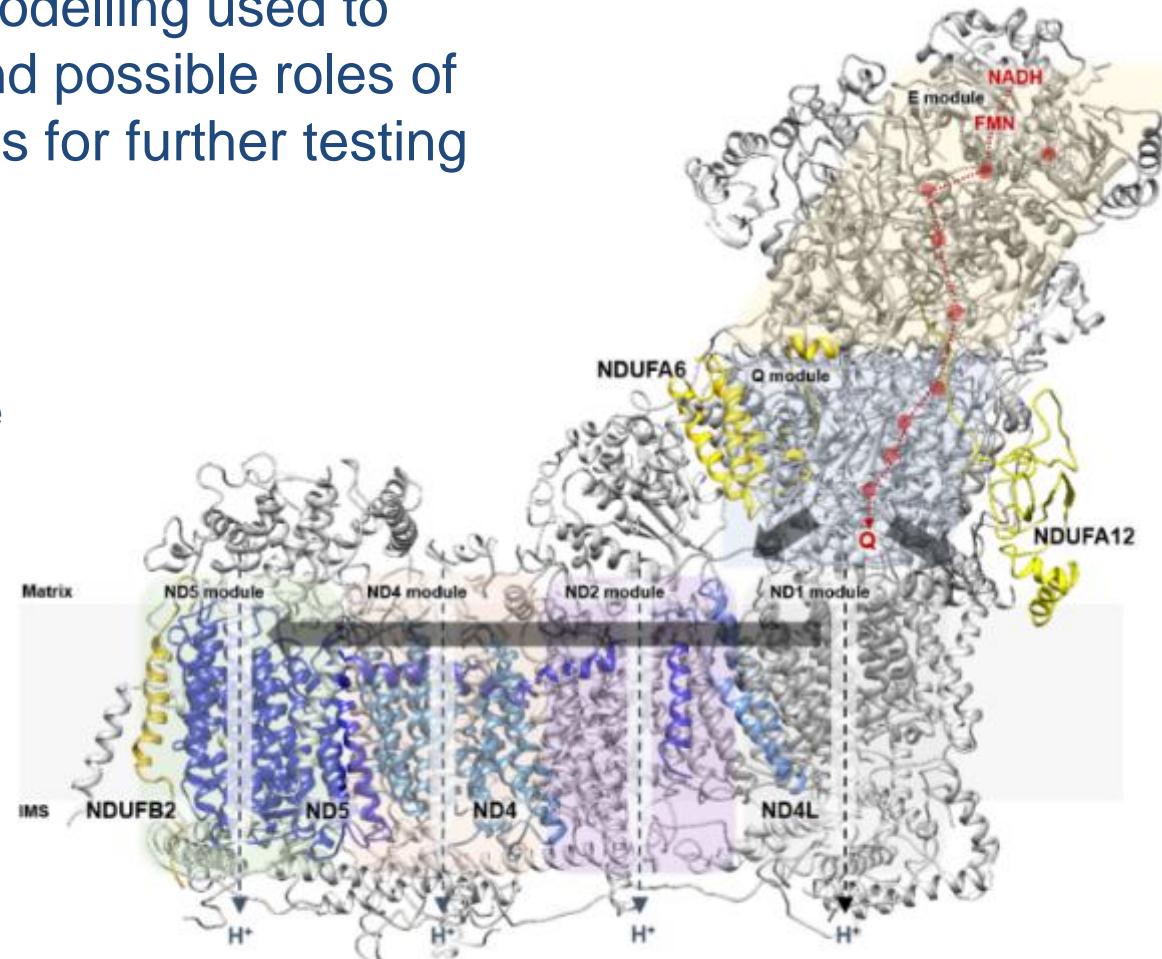


Chris Greening

Protein modelling used to understand possible roles of candidates for further testing

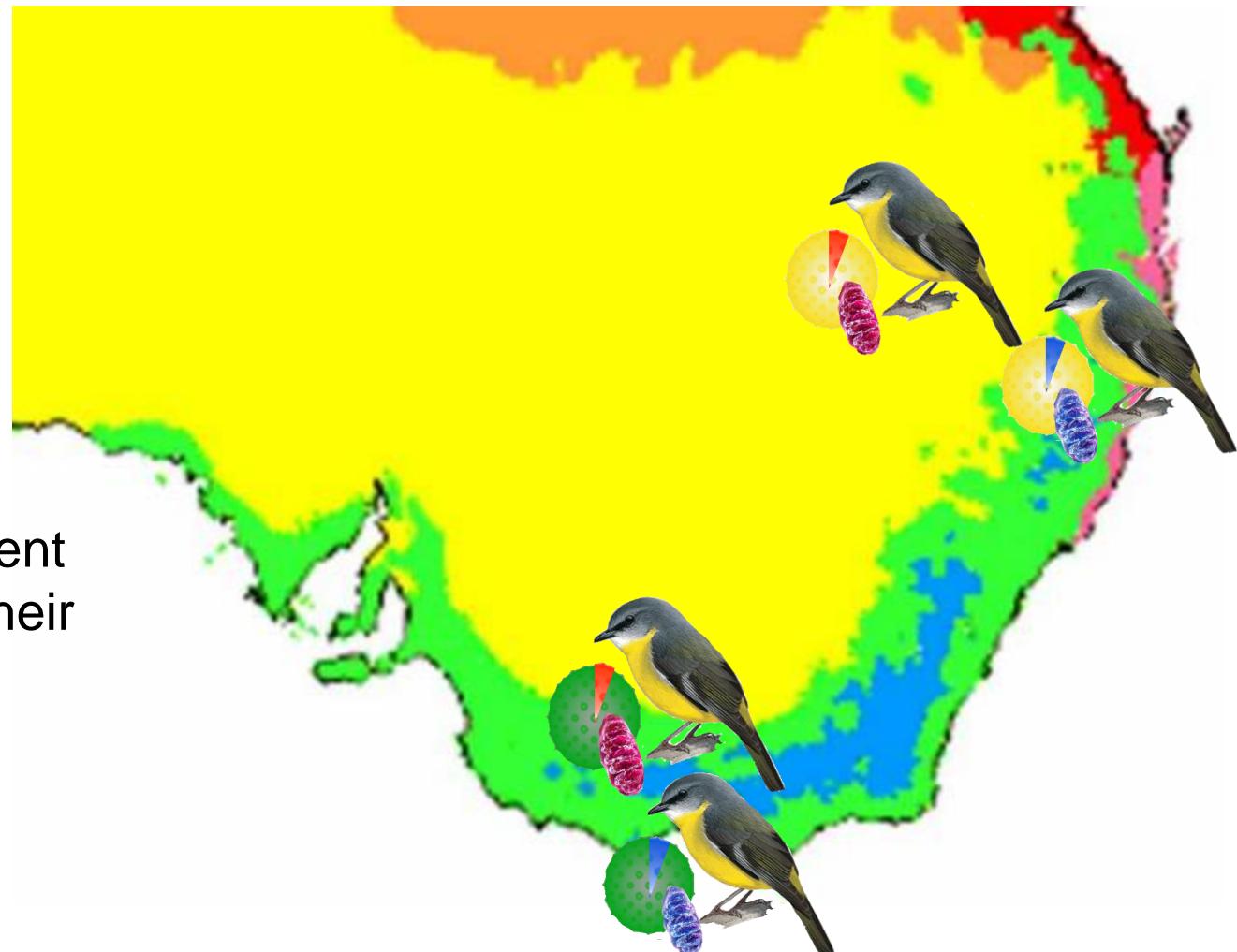
Nuclear mitochondrial gene products from in the 1A island in **YELLOW**

Mitochondrial gene products evidence for positive selection (ND4, ND4L and ND5) in **PURPLE**



Summary

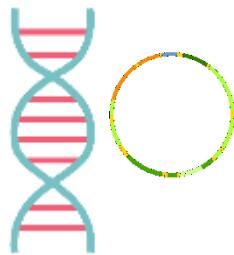
Two distinct mtDNA groups that also have set of putatively co-adapted nuclear genes



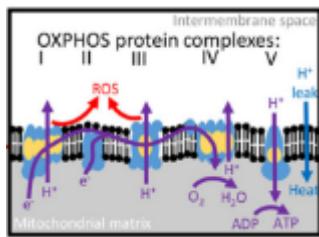
If adaptive, they
should have different
biology suited to their
environments

Local adaptation What to look for?

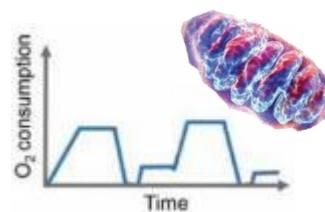
Genomes →



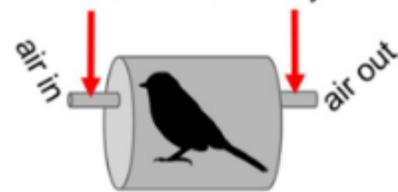
OXPHOS →



mitochondrial →
function



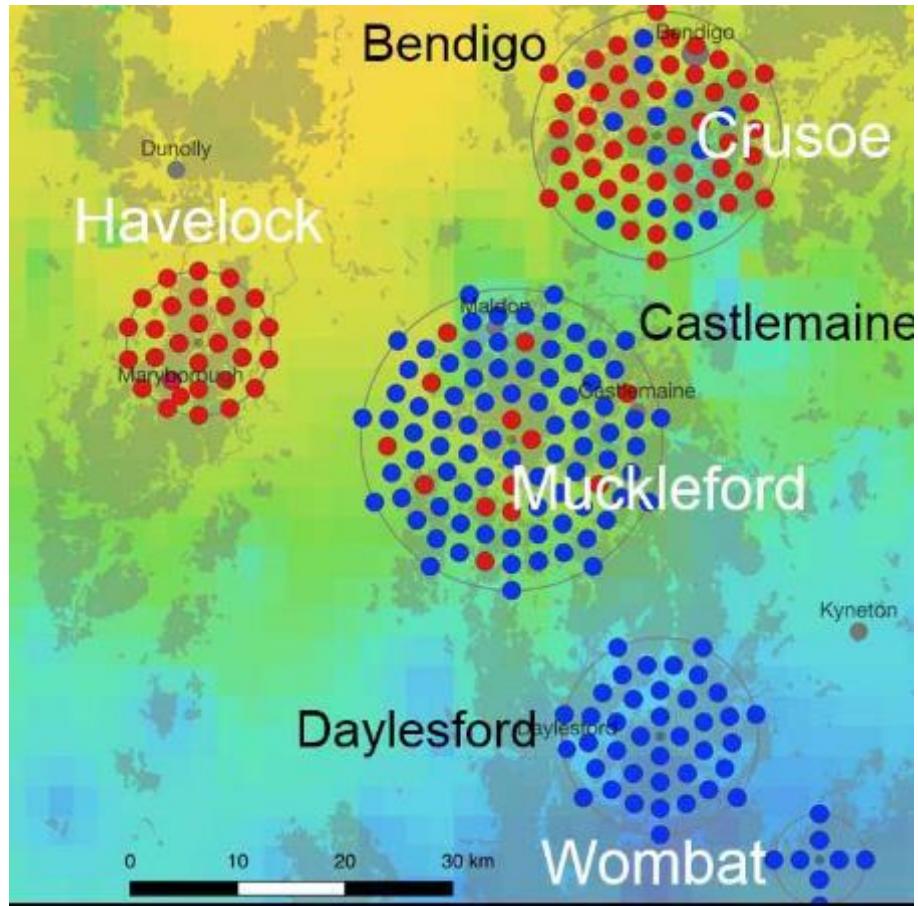
metabolism



Some main items to measure that yield predictions

In a feasible design across an environmental gradient, with lineages side-by-side:

- (1) Mitochondrial function
- (2) Metabolism
- (3) Environments
- (4) Gene flow between the lineages
- (5) Fitness x environment indicators

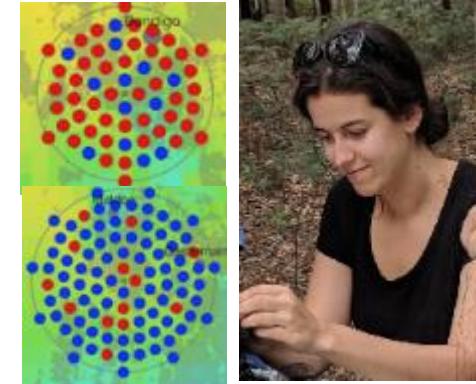


(The lineages should evolve to recognize and not breed with each other
Do females choose a mate of the same mitolineage?



Parentage analysis to find out:

- How often do mitolineages interbreed?
- Are there sex-specific consequences for their offspring?



Hons: Jessica Walters



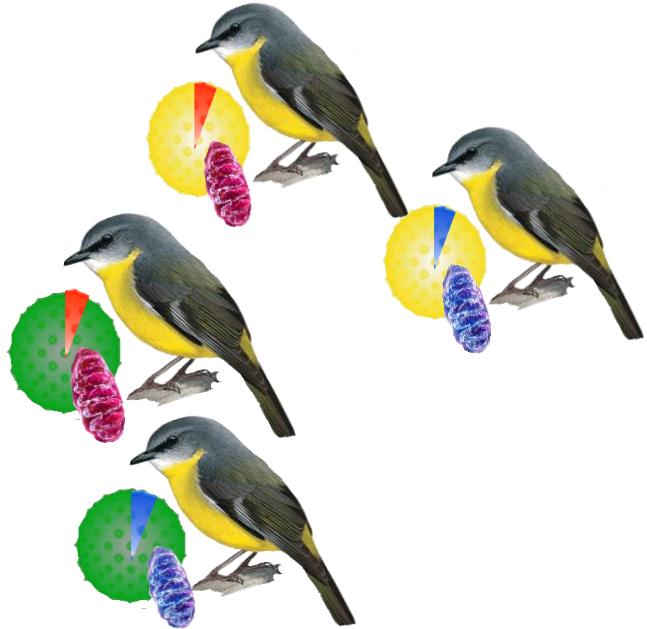
Apparently no preference for mating with own lineage, but...

...admixed parents ♀♂ or ♀♂
have almost no daughters

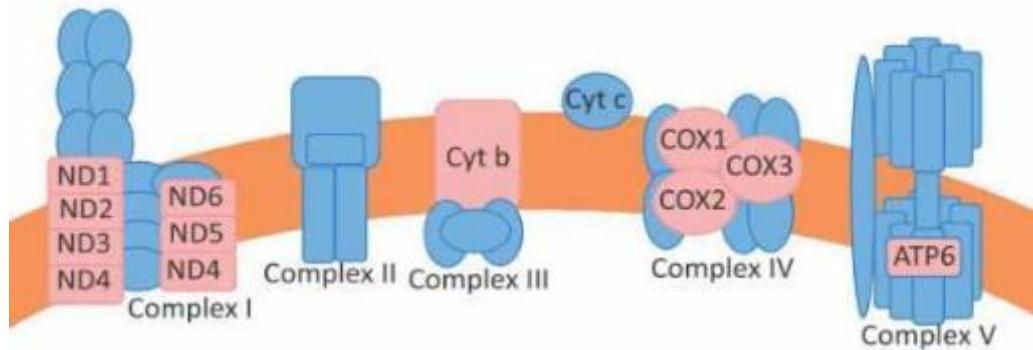
That is, strong selection against hybrid females

Implications

Inland and coastal Eastern yellow robins
may be speciating by climate adaptation



through mitonuclear interactions



Hill (2015) Mol. Biol. Evol. 32(8):1917–1927

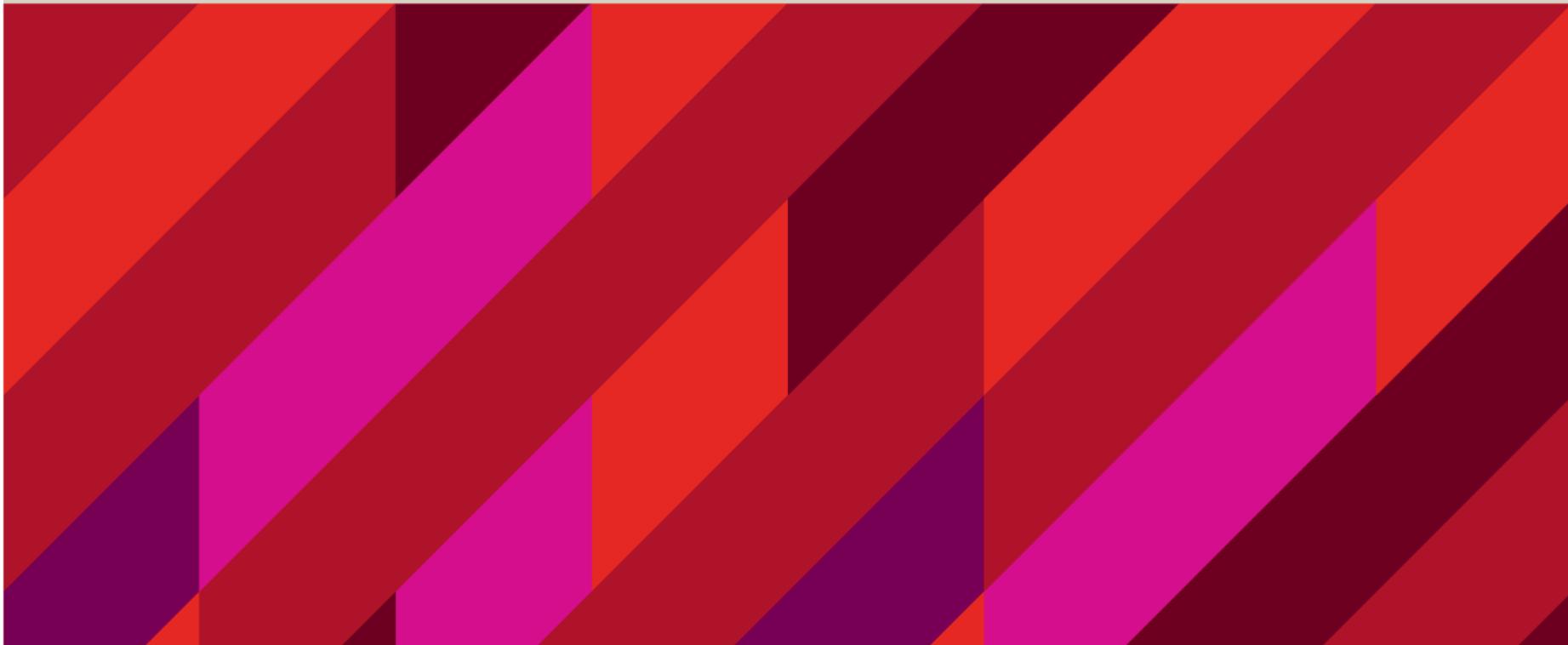
Next lecture – more on
selection/adaptation



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BIOL3110 Conservation & Ecological Genetics

LECTURE 19: ADAPTIVE POTENTIAL 2



Case Study: Research by Jeffrey Good

University of Montana

*Climate change and the evolution of
seasonal camouflage*

Research to understand

- 1) the evolution of seasonal camouflage and**
 - 2) genomic responses to climate change in mammals**
- using population and functional genomic approaches**

SUMMER

WINTER



Snowshoe hare seasonal camouflage



Rapid environmental change (eg climate change) can make these systems break down

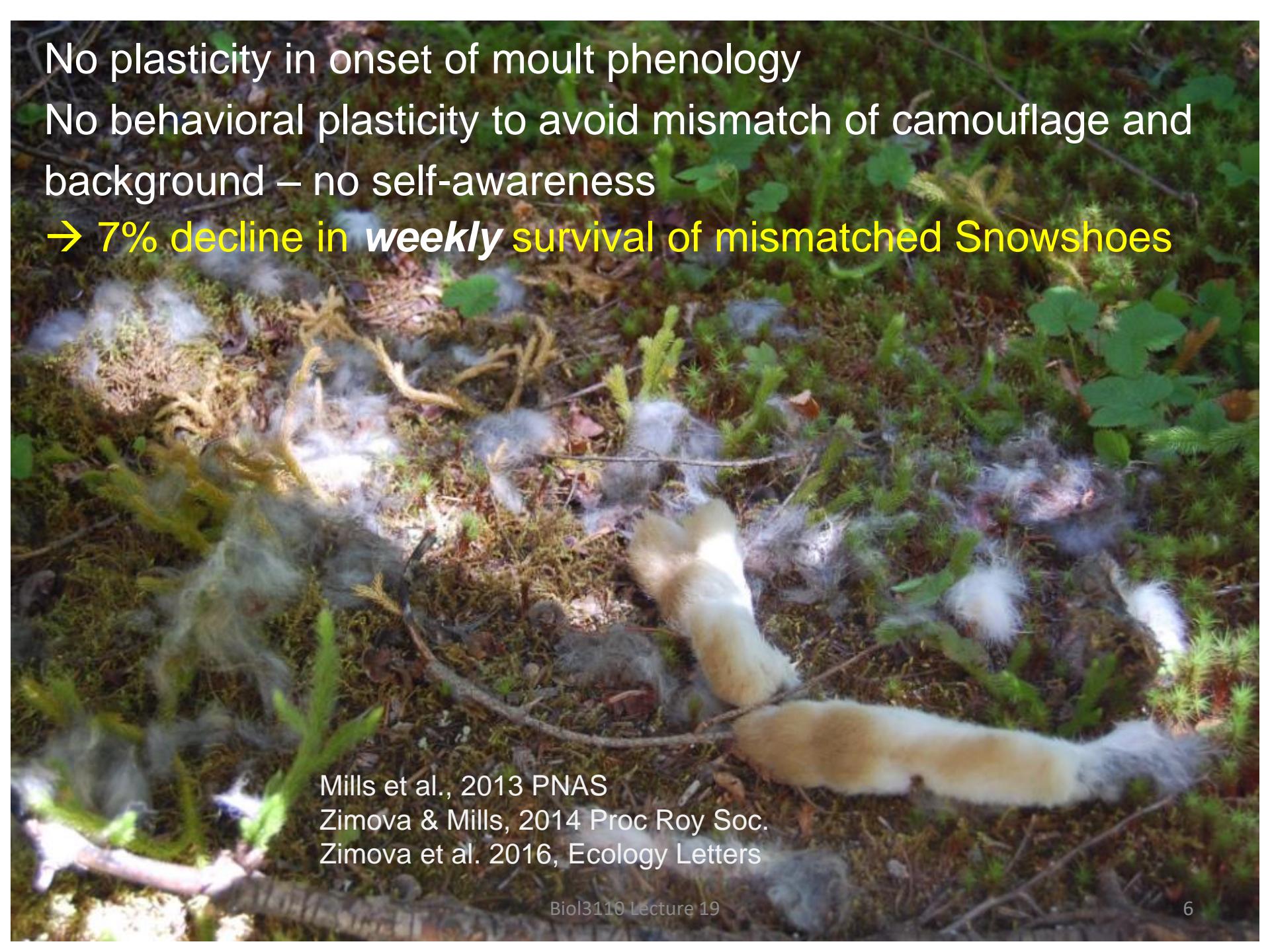
Photoperiod may be an increasingly unreliable predictor of snow cover



No plasticity in onset of moult phenology

No behavioral plasticity to avoid mismatch of camouflage and background – no self-awareness

→ 7% decline in **weekly** survival of mismatched Snowshoes



Mills et al., 2013 PNAS
Zimova & Mills, 2014 Proc Roy Soc.
Zimova et al. 2016, Ecology Letters

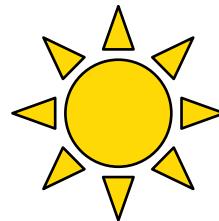
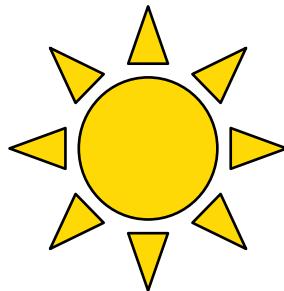
How does seasonal change in phenotype (phenology) happen?

Animals need a cue that warns them of the upcoming season, eg when there will be snow on the ground

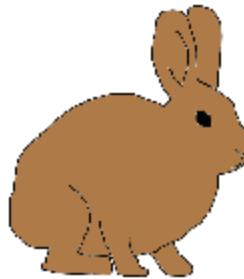
Often use photoperiod (daylength), detected by a molecular receptor system (e.g. melatonin) that communicates with the systems that cause phenotypic seasonal change

e.g. photoperiod connects to systems of circadian rhythm ('clock') genes, controlling expression of many genes, driving changes in phenotype

Coat color change is tied to seasonal signaling molecules



Changes in:
Thermoregulation
Reproduction
Behavior



↑ melatonin



autumn

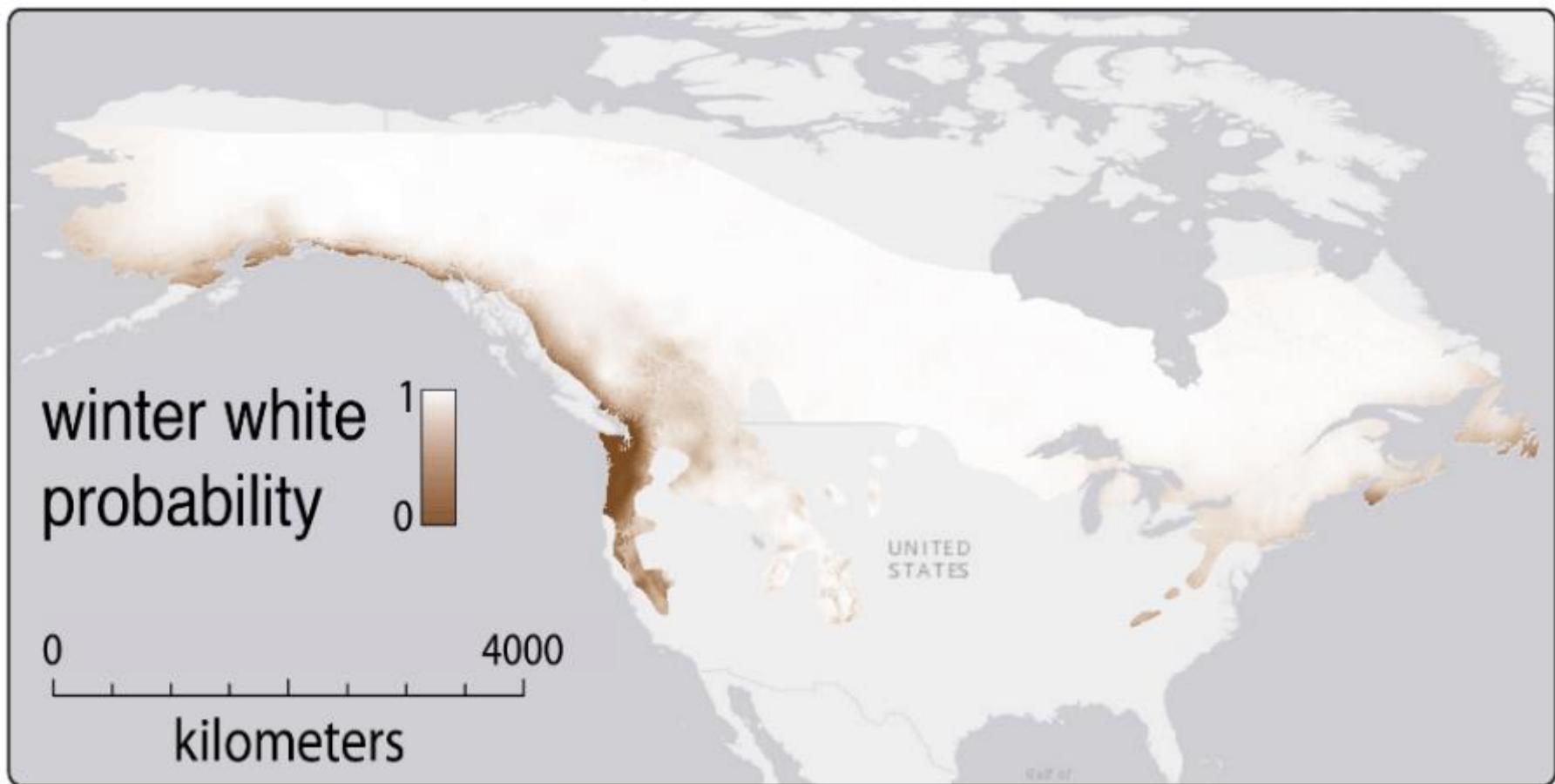


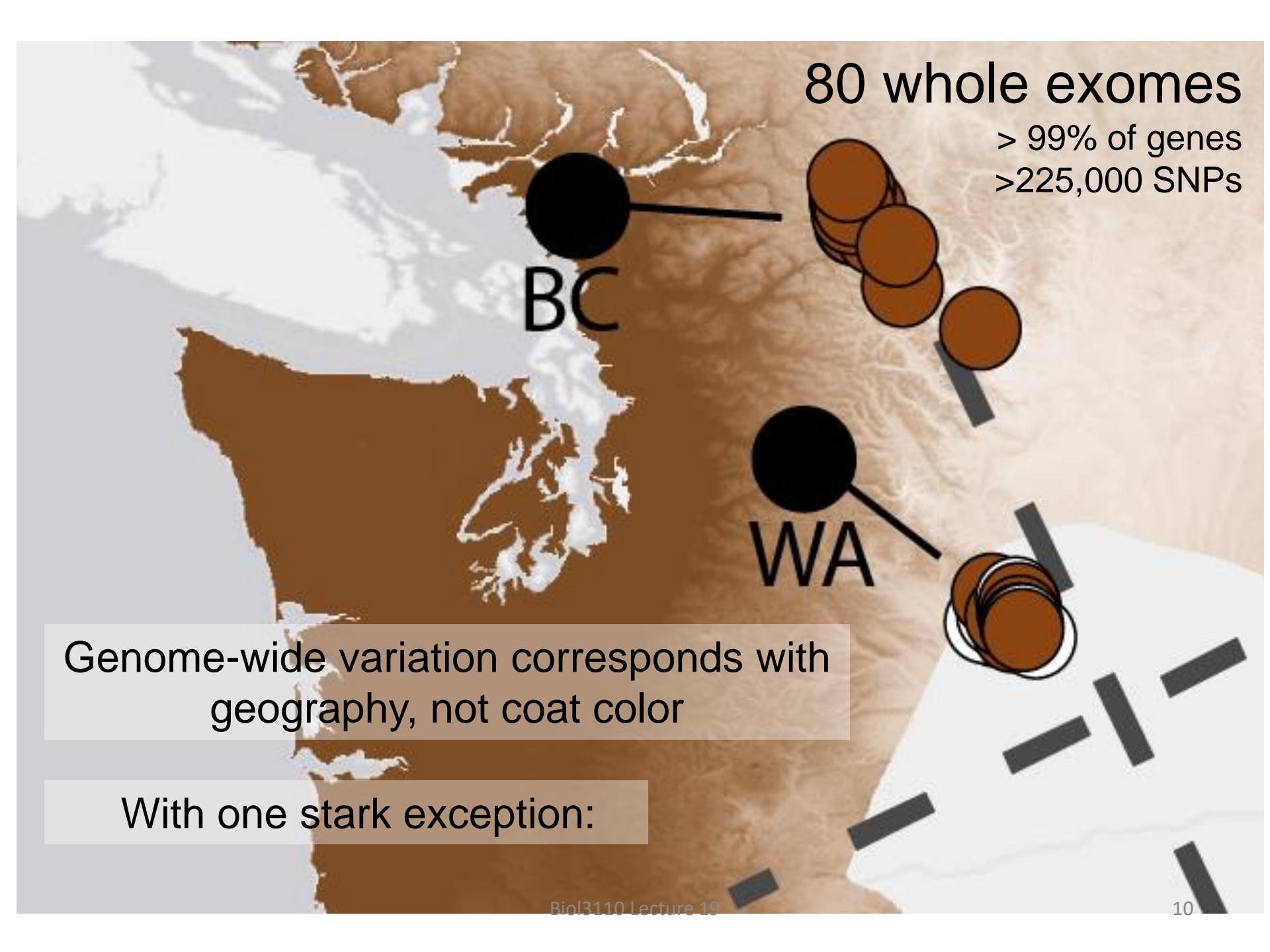
winter



Two components of variation:

- *If* change colour
- *When* to change colour





80 whole exomes

> 99% of genes
>225,000 SNPs

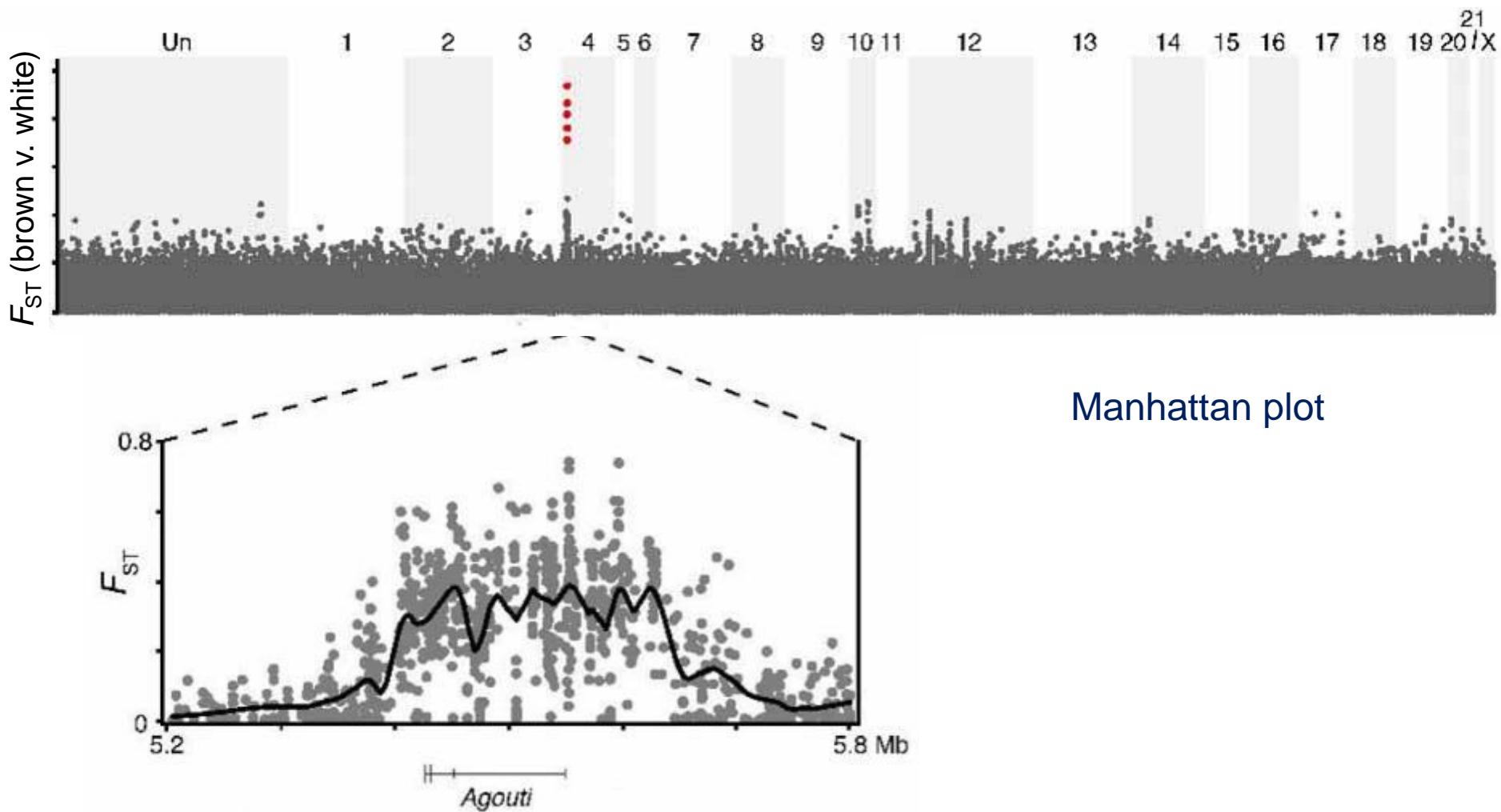
BC

WA

Genome-wide variation corresponds with geography, not coat color

With one stark exception:

Agouti is perfectly associated with winter colour



The *Agouti* gene determines winter coat colour

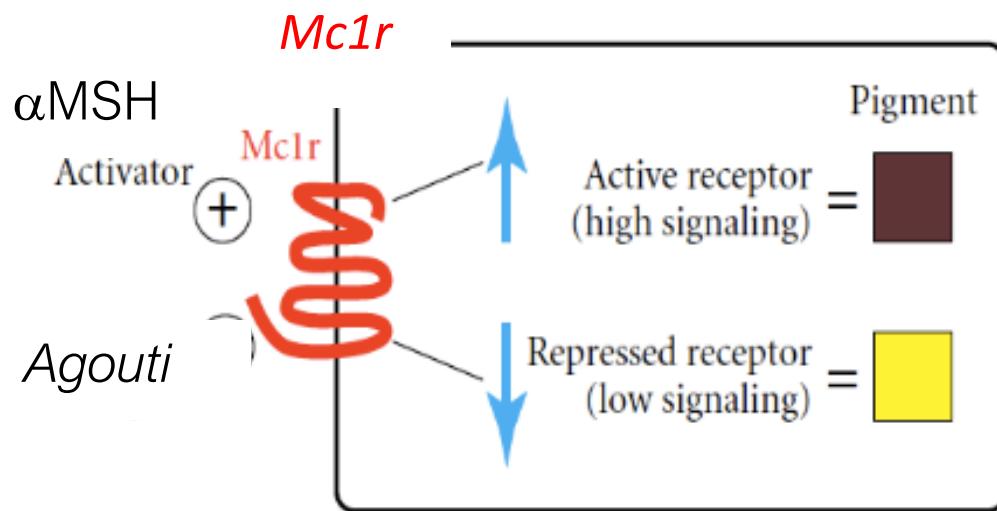


aa
(a recessive)

AA Aa
(A dominant)

How does *Agouti* determine winter colour?

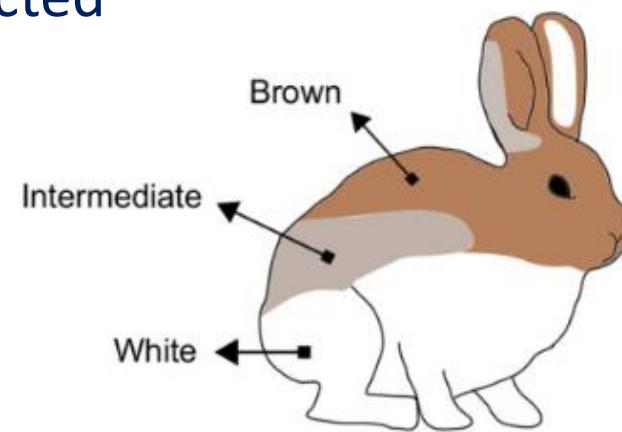
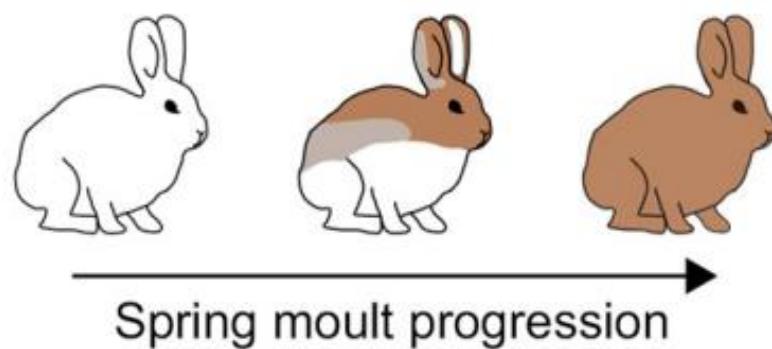
Agouti influences coat colour by reducing the binding of the melanocortin-1 receptor (MC1R), impeding the action of α -melanocyte-stimulating hormone (α MSH) that would cause production of dark eumelanin



From which you would predict that  is associated with seasonal upregulation of *Agouti* – was that true?

Finding genes that have different seasonal expression to do with coat colour

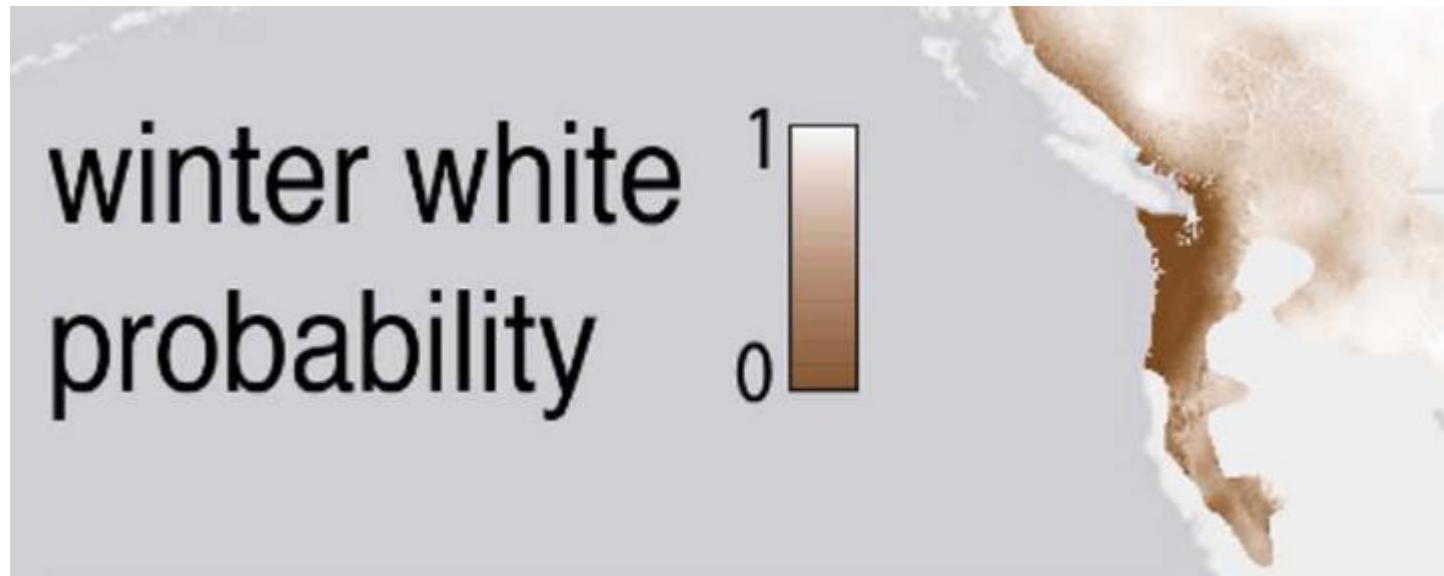
Progressive moult over the body allowed sampling of cells containing expressed genes, so that the changes in gene expression leading to differences in coat colour could be detected



Gene expression tested using RNAseq (RNA sequencing, i.e. transcriptomics) to estimate the abundance of gene transcripts in different-coloured areas of Snowshoes – result: The Agouti Signalling Peptide (ASIP) gene was upregulated in white hares.

How did the warm coastal Pacific Northwest populations evolve to never become white?

(they moult, but from summer brown to winter brown)



Species tree

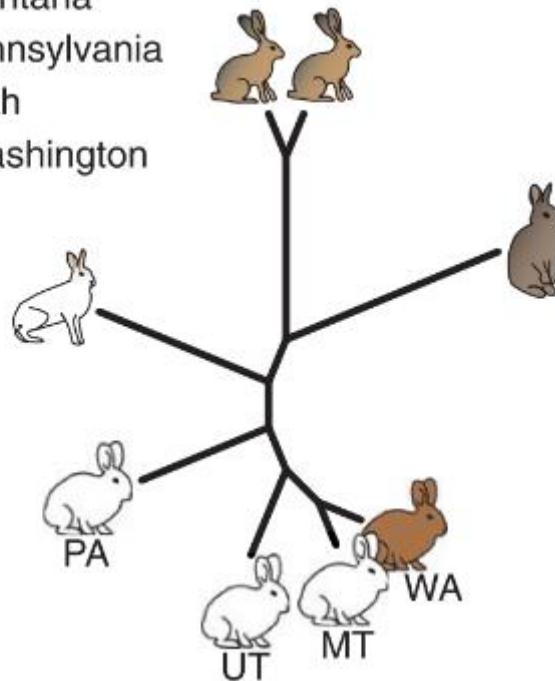
Based on whole genome sequencing

MT=Montana

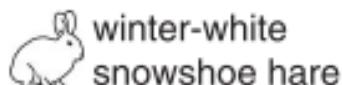
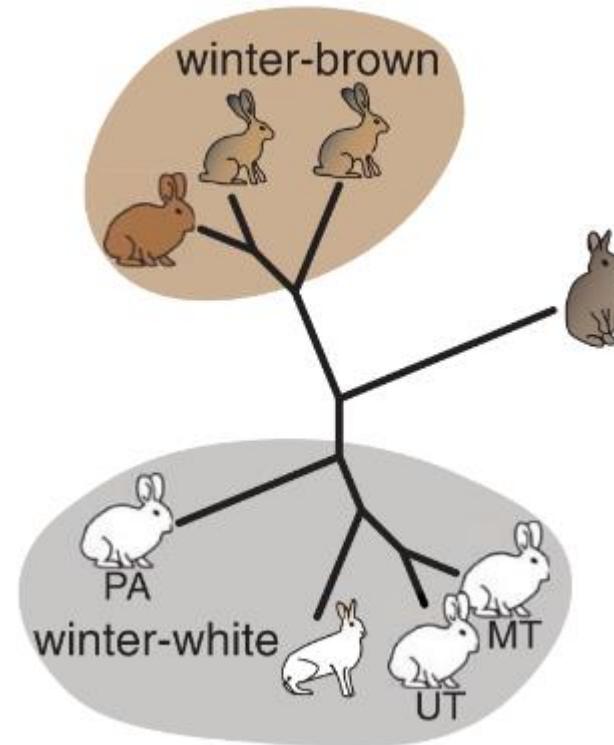
PA=Pennsylvania

UT=Utah

WA=Washington



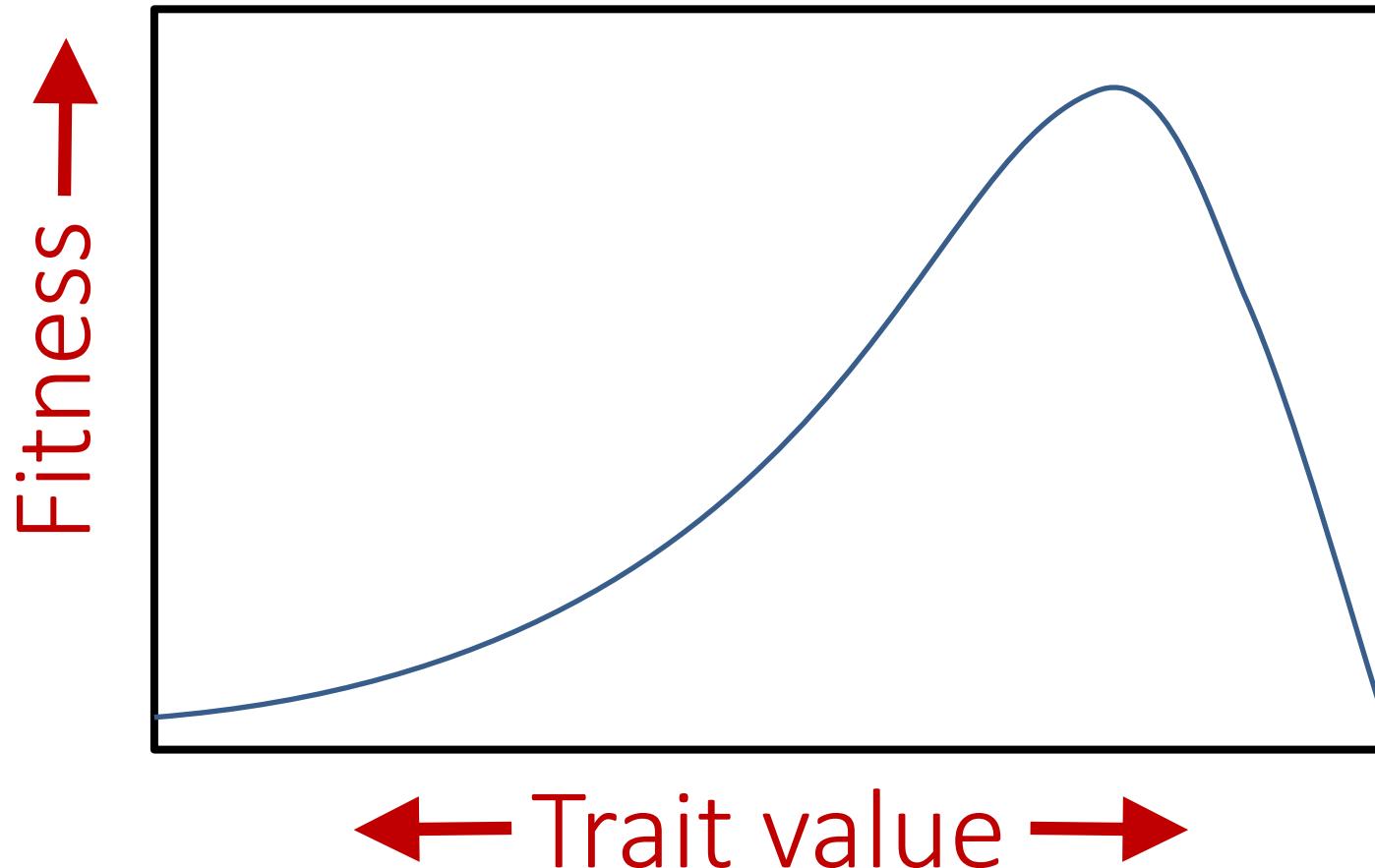
Agouti tree



i.e. the 'brown' gene moved by hybridization from Black-tailed jackrabbit into Pacific Northwest Coast hares - **adaptive introgression**

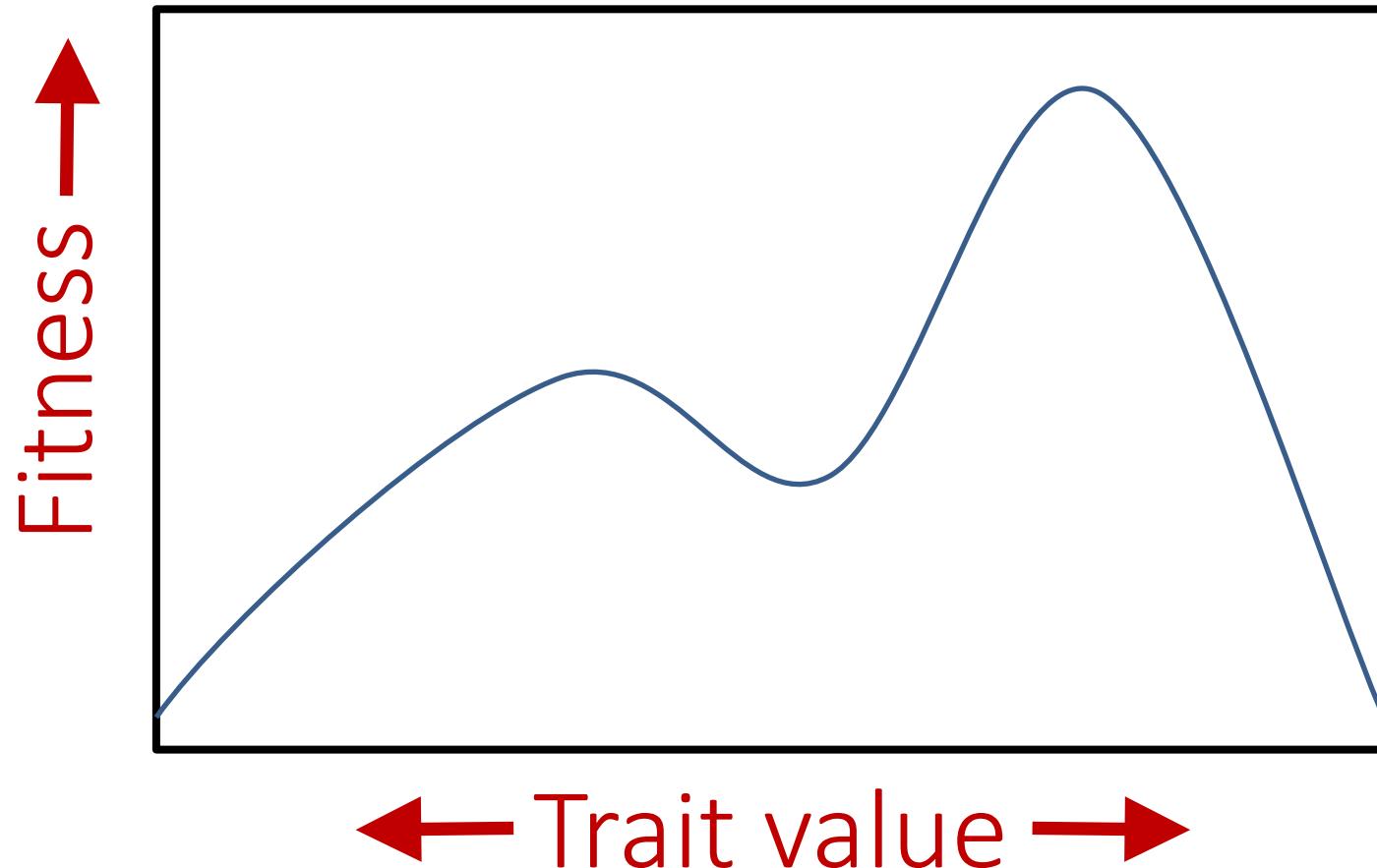
“Fitness landscape” concept

If we think of how a single trait relates to fitness:



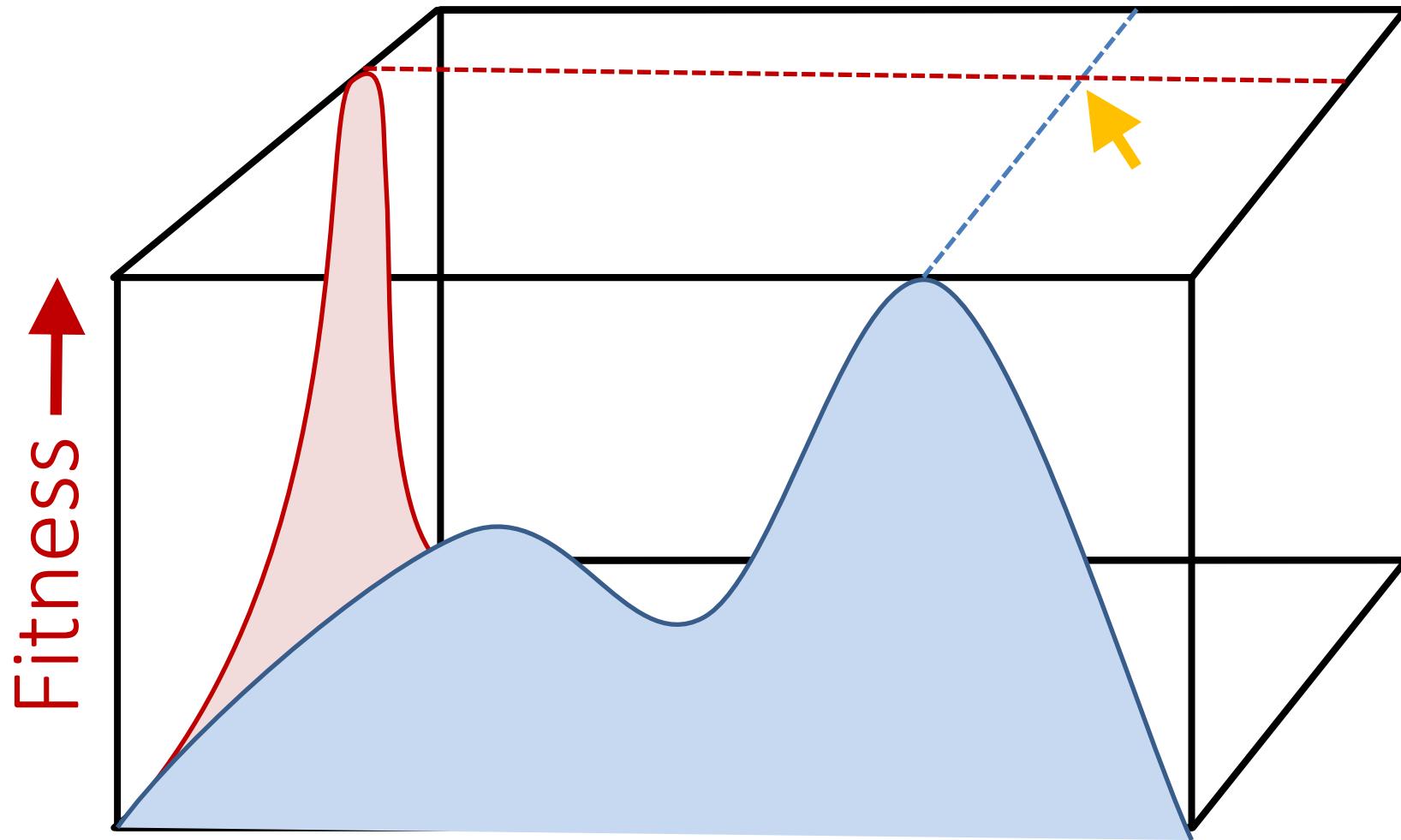
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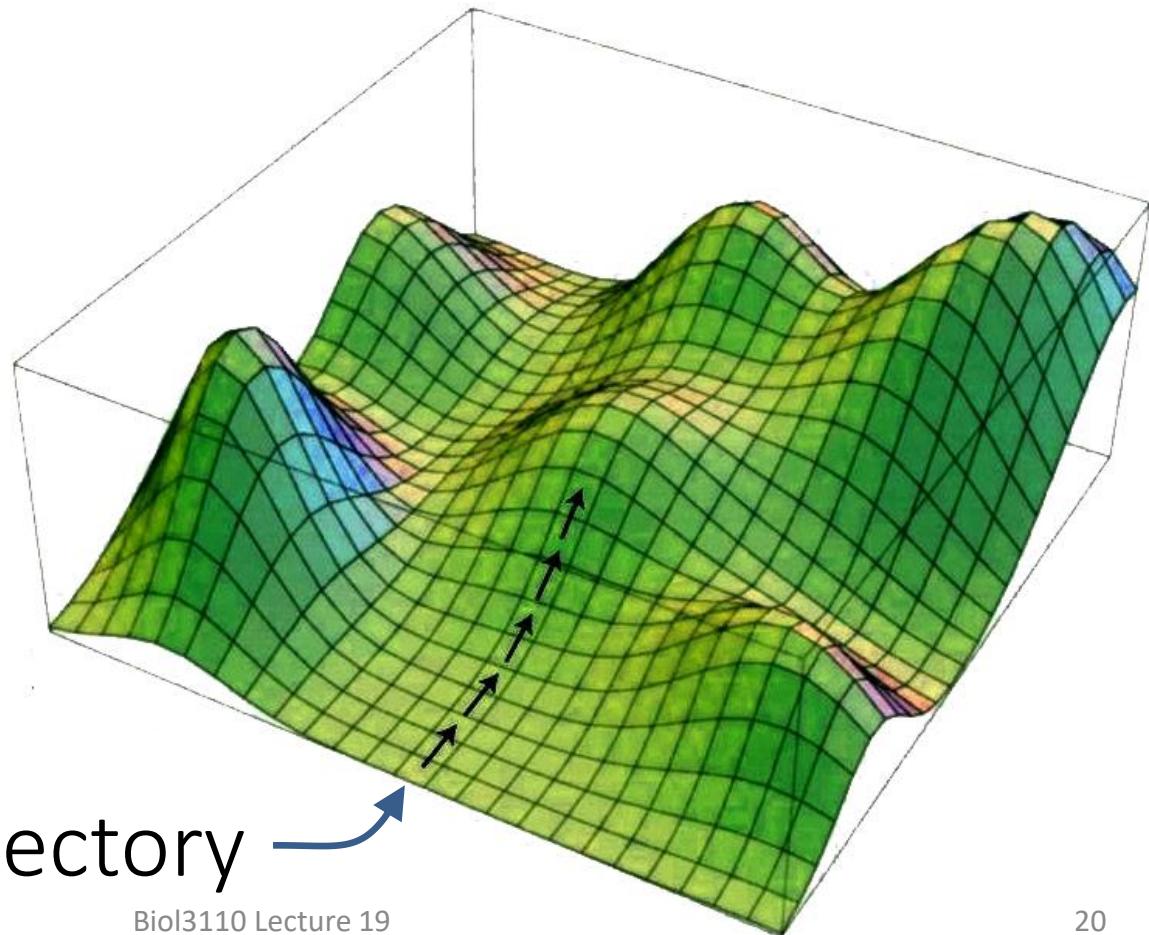
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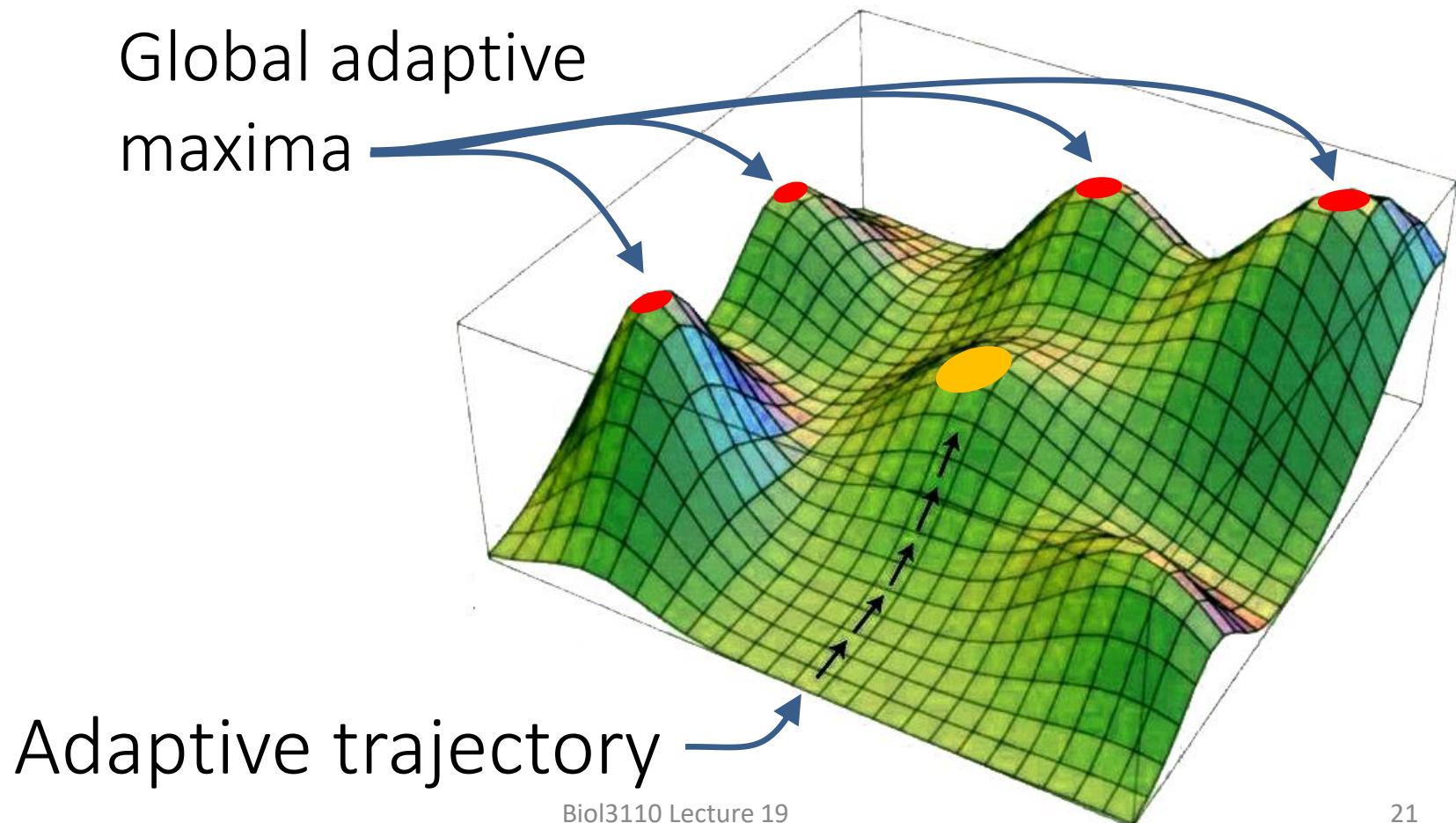
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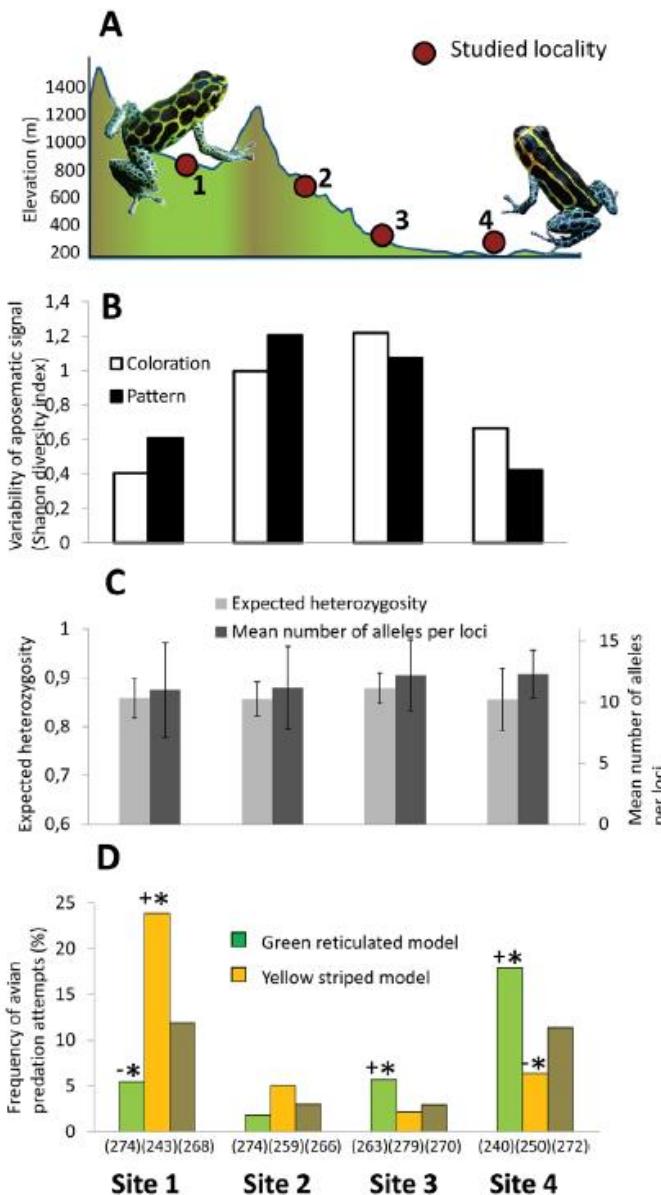
Sewall Wright: Species evolve to localised adaptive “peaks” in an n-dimensional “landscape”



“Fitness landscape” concept

Sewall Wright: Species evolve to localised adaptive “peaks” in an n-dimensional “landscape”



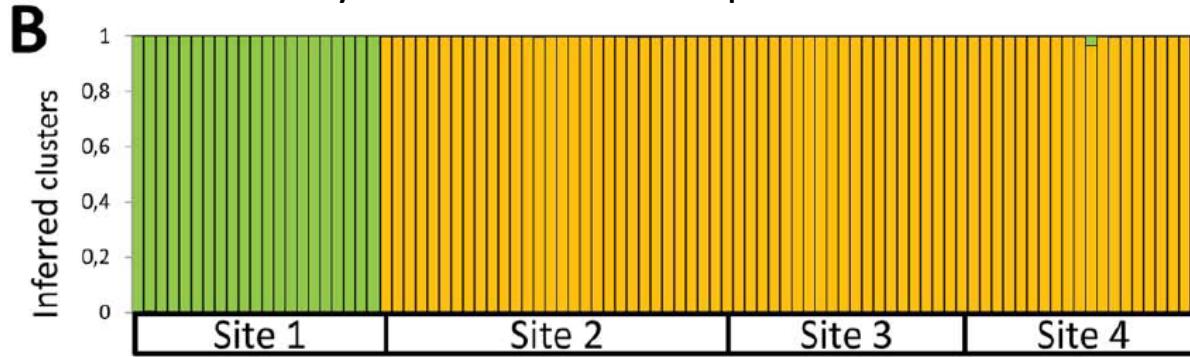


Wrights shifting balance theory –
relaxation of selection + drift can
allow for polymorphisms

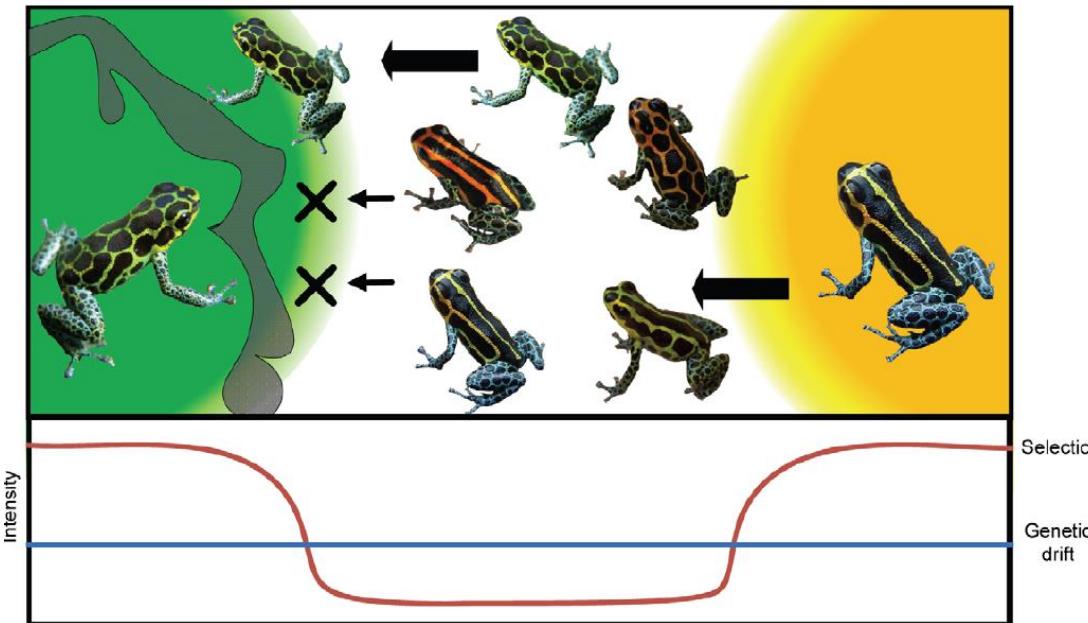
High variability sites 2 & 3

Low rates of predation sites 2 & 3

Hybridisation can't explain the variation



Wrights shifting balance theory – relaxation of selection + drift can allow for polymorphisms that then ‘compete’ across the fitness landscape



Next Lecture

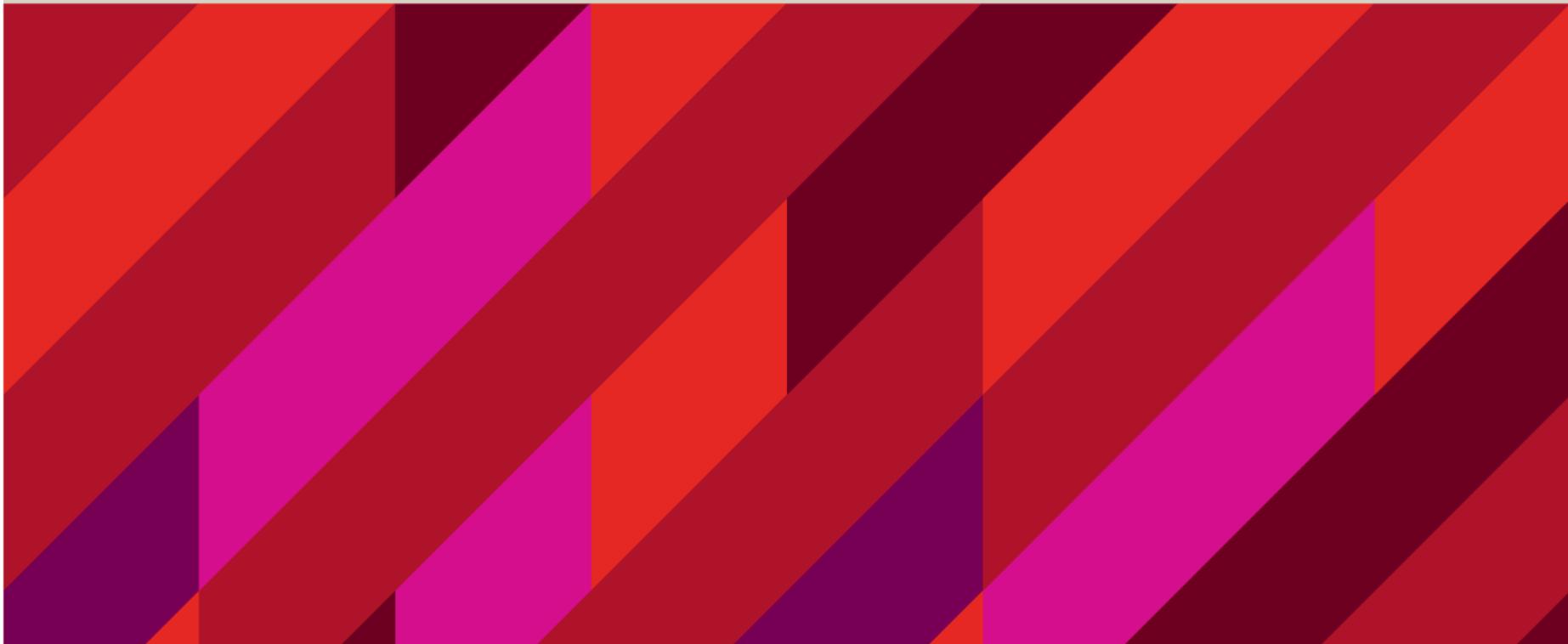
Social influences on connectivity



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Photoperiod may be an increasingly unreliable predictor of snow cover



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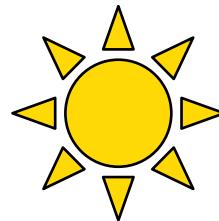
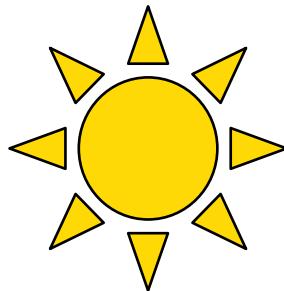
How does seasonal change in phenotype (phenology) happen?

Animals need a cue that warns them of the upcoming season, eg when there will be snow on the ground

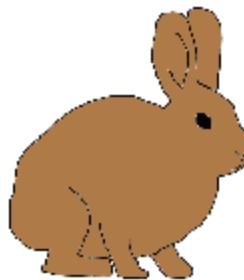
Often use photoperiod (daylength), detected by a molecular receptor system (e.g. melatonin) that communicates with the systems that cause phenotypic seasonal change

e.g. photoperiod connects to systems of circadian rhythm ('clock') genes, controlling expression of many genes, driving changes in phenotype

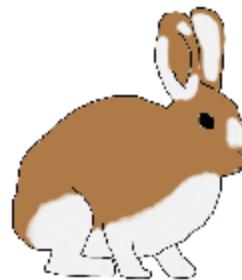
Coat color change is tied to seasonal signaling molecules



Changes in:
Thermoregulation
Reproduction
Behavior



↑ melatonin



autumn

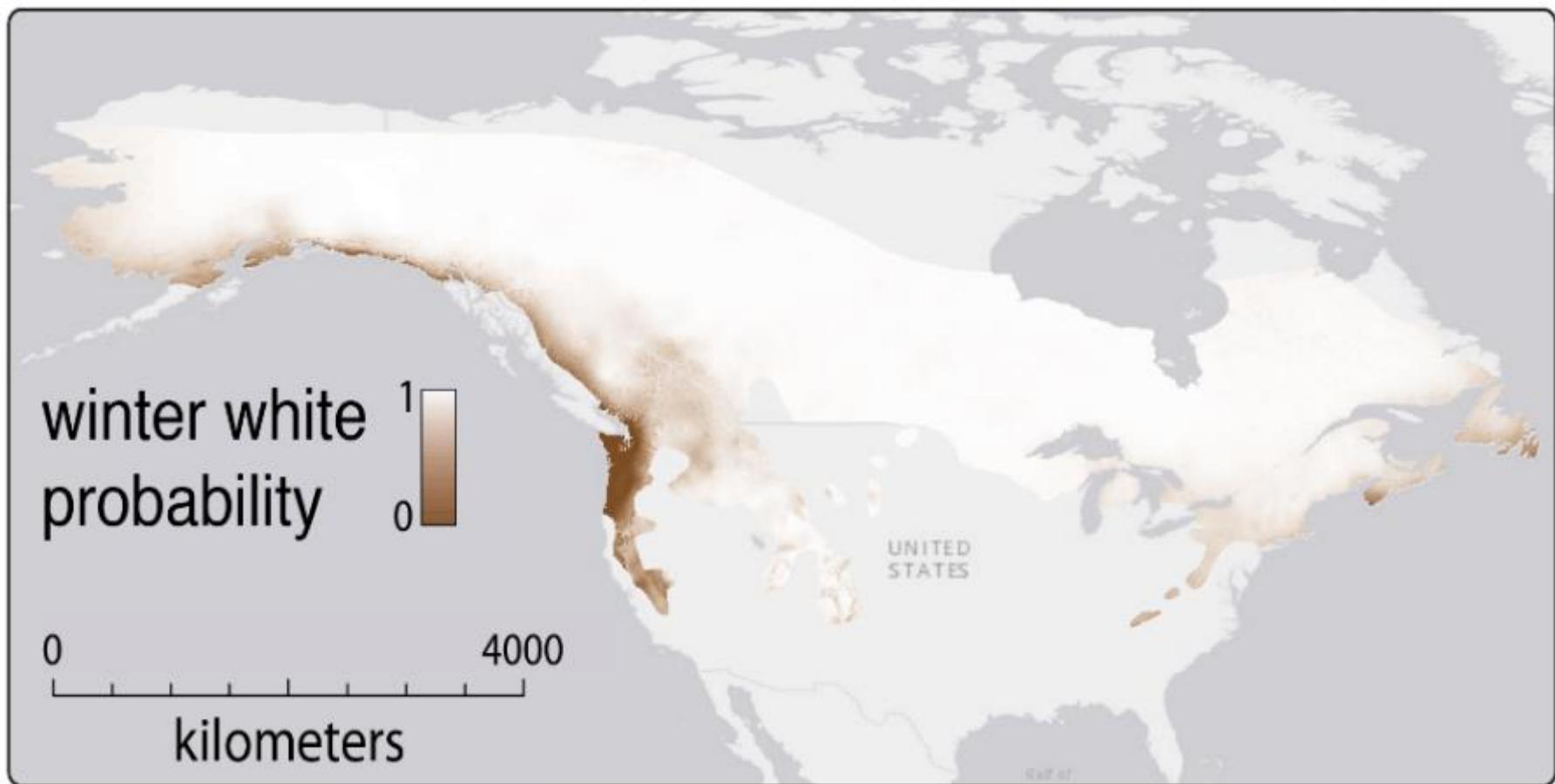


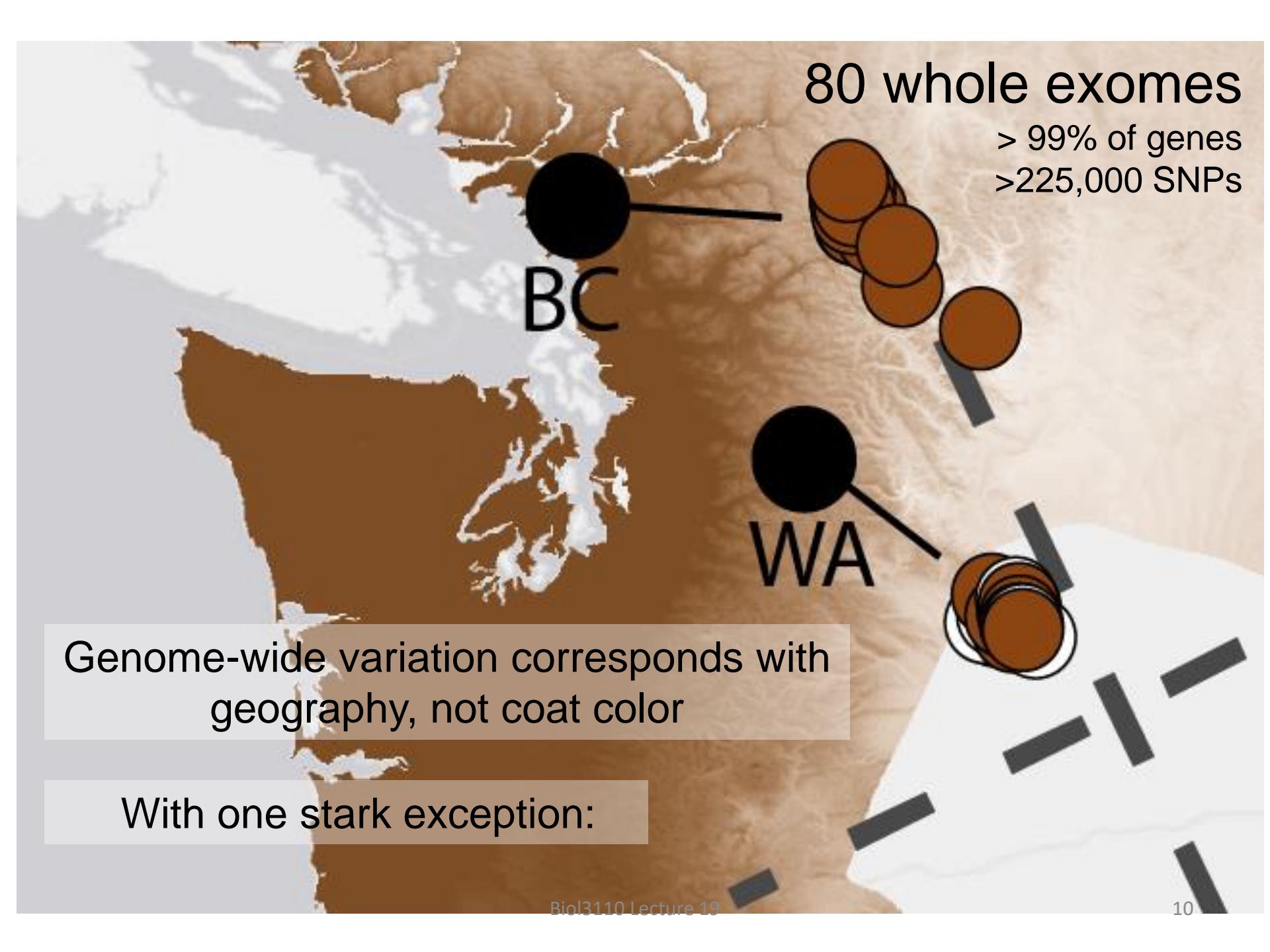
winter



Two components of variation:

- *If* change colour
- *When* to change colour





80 whole exomes

> 99% of genes
>225,000 SNPs

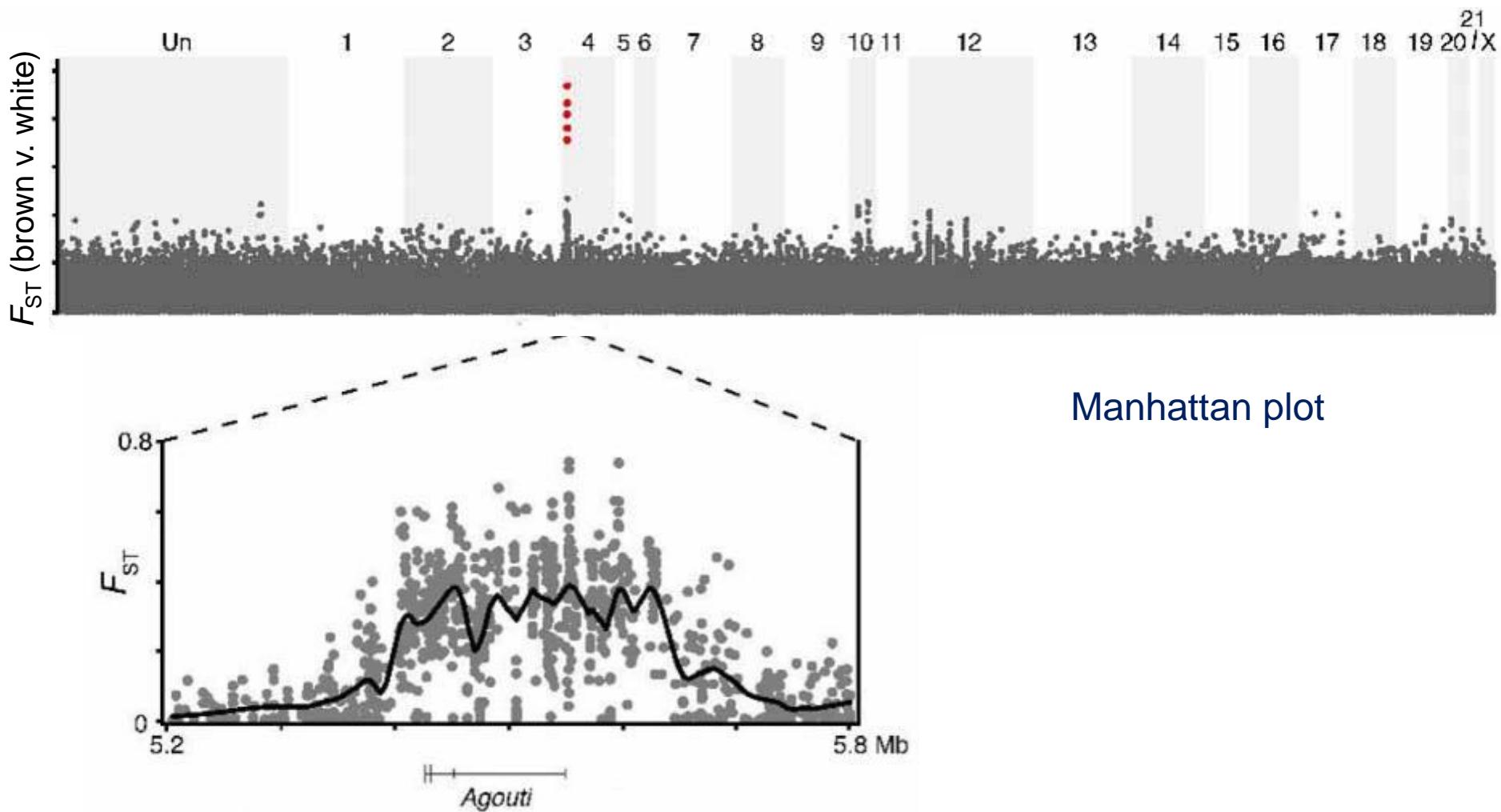
BC

WA

Genome-wide variation corresponds with geography, not coat color

With one stark exception:

Agouti is perfectly associated with winter colour



The *Agouti* gene determines winter coat colour

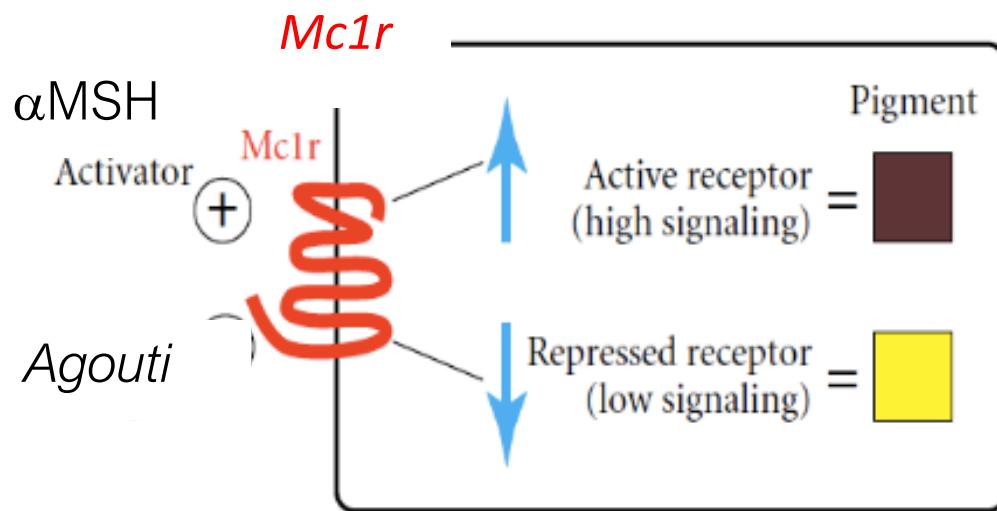


aa
(a recessive)

AA Aa
(A dominant)

How does *Agouti* determine winter colour?

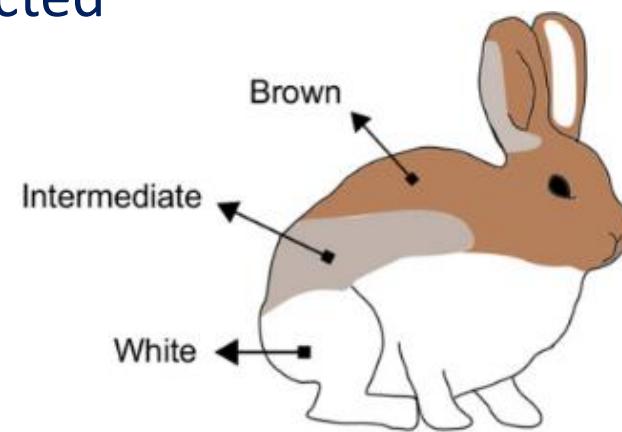
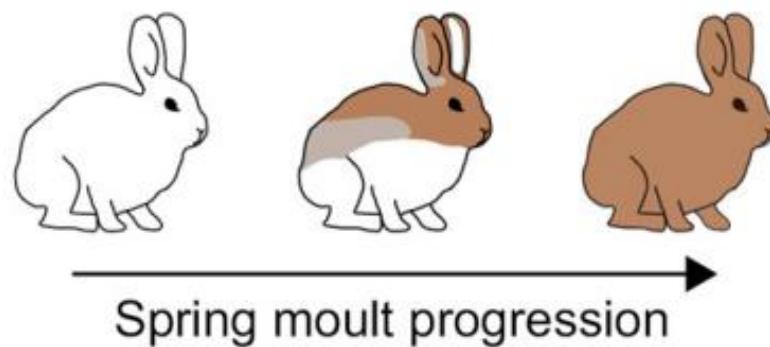
Agouti influences coat colour by reducing the binding of the melanocortin-1 receptor (MC1R), impeding the action of α -melanocyte-stimulating hormone (α MSH) that would cause production of dark eumelanin



From which you would predict that  is associated with seasonal upregulation of *Agouti* – was that true?

Finding genes that have different seasonal expression to do with coat colour

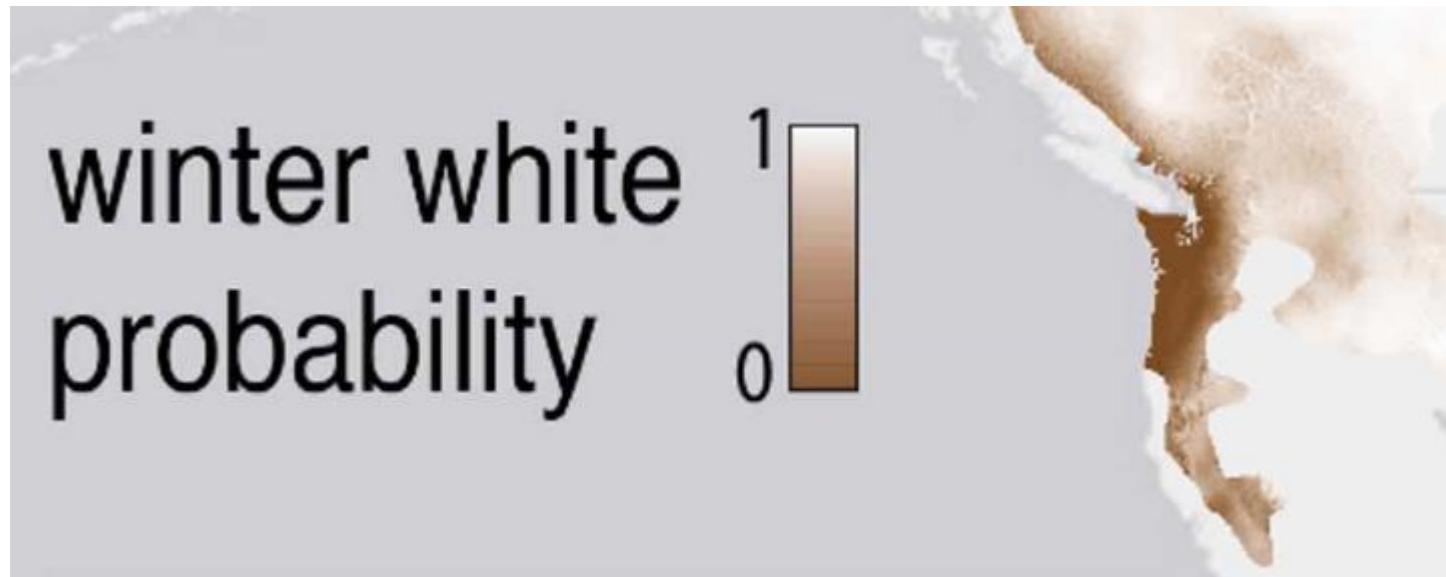
Progressive moult over the body allowed sampling of cells containing expressed genes, so that the changes in gene expression leading to differences in coat colour could be detected



Gene expression tested using RNAseq (RNA sequencing, i.e. transcriptomics) to estimate the abundance of gene transcripts in different-coloured areas of Snowshoes – result: The Agouti Signalling Peptide (ASIP) gene was upregulated in white hares.

How did the warm coastal Pacific Northwest populations evolve to never become white?

(they moult, but from summer brown to winter brown)



Species tree

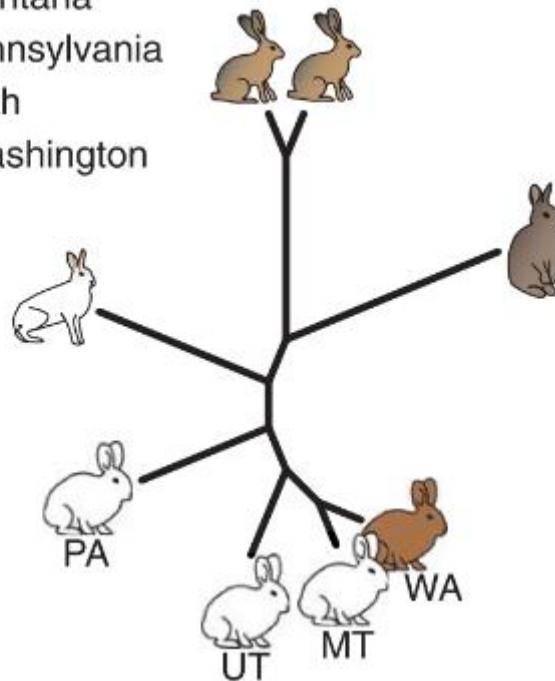
Based on whole genome sequencing

MT=Montana

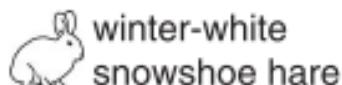
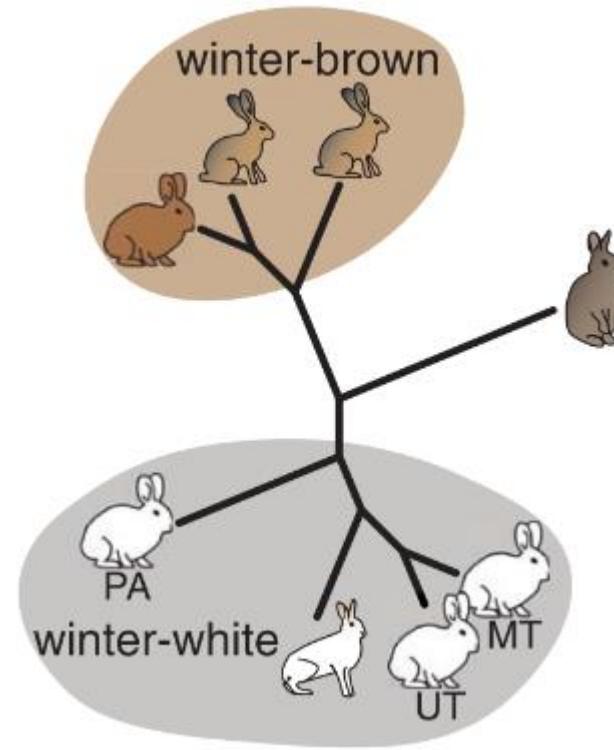
PA=Pennsylvania

UT=Utah

WA=Washington



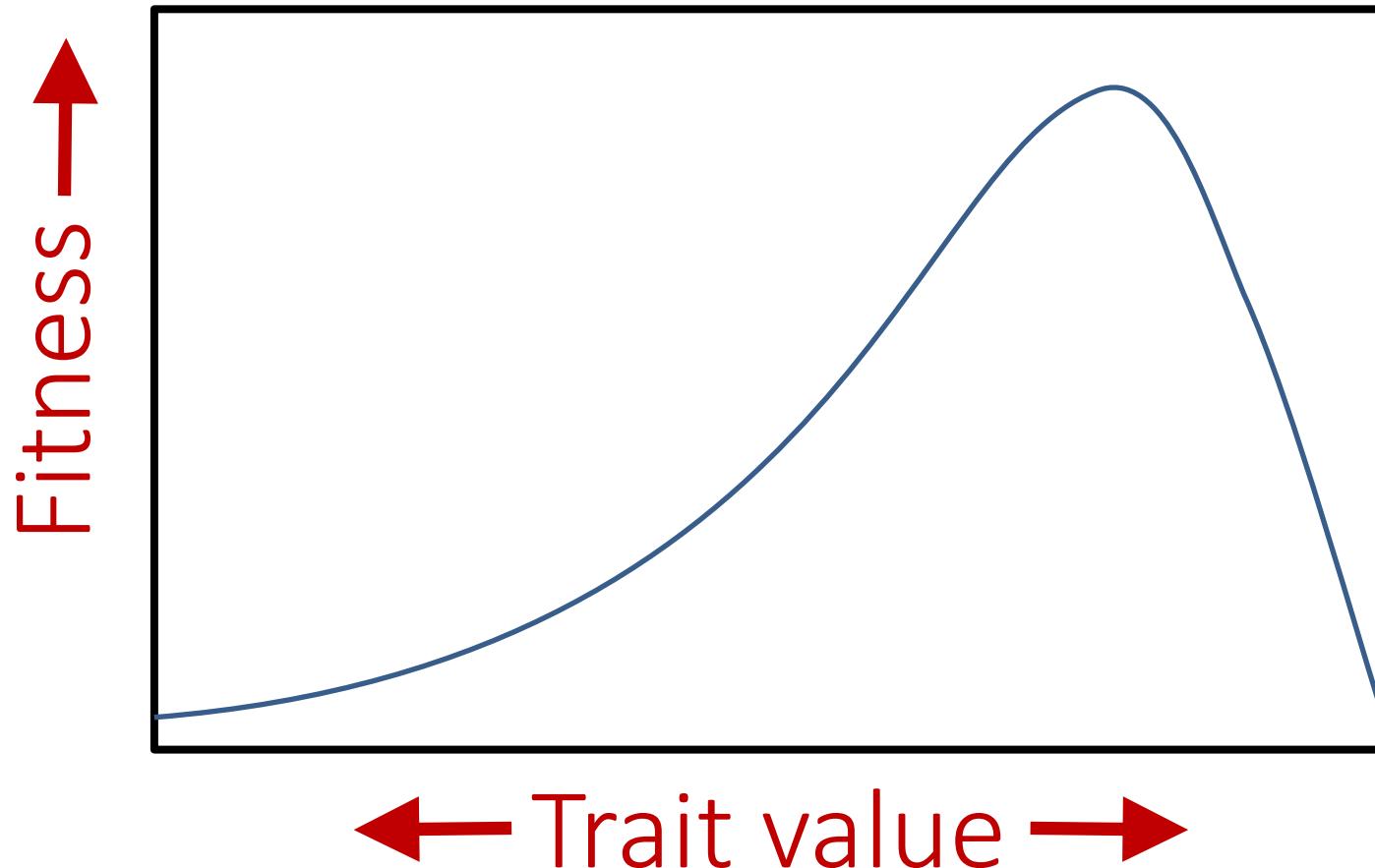
Agouti tree



i.e. the 'brown' gene moved by hybridization from Black-tailed jackrabbit into Pacific Northwest Coast hares - **adaptive introgression**

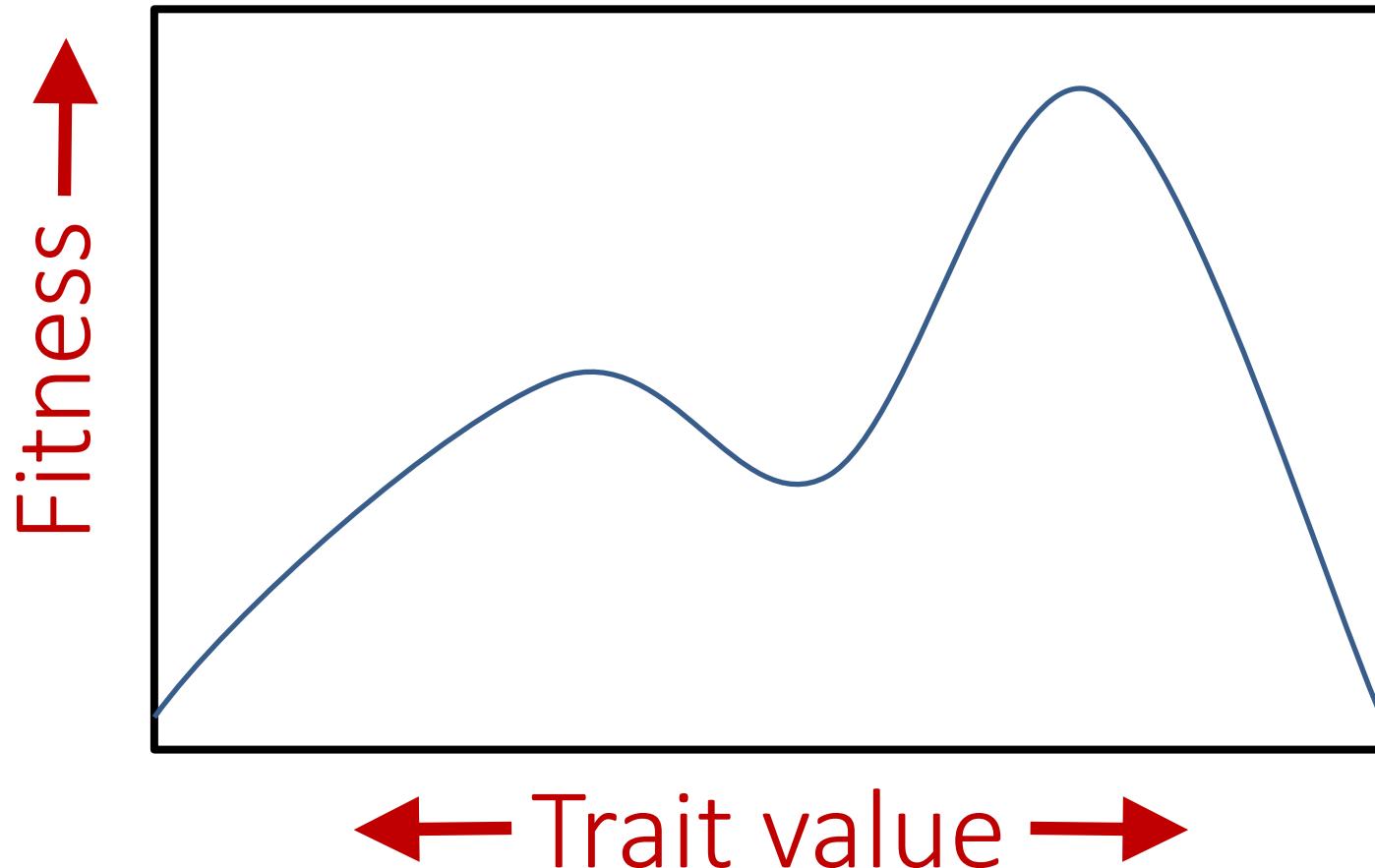
“Fitness landscape” concept

If we think of how a single trait relates to fitness:



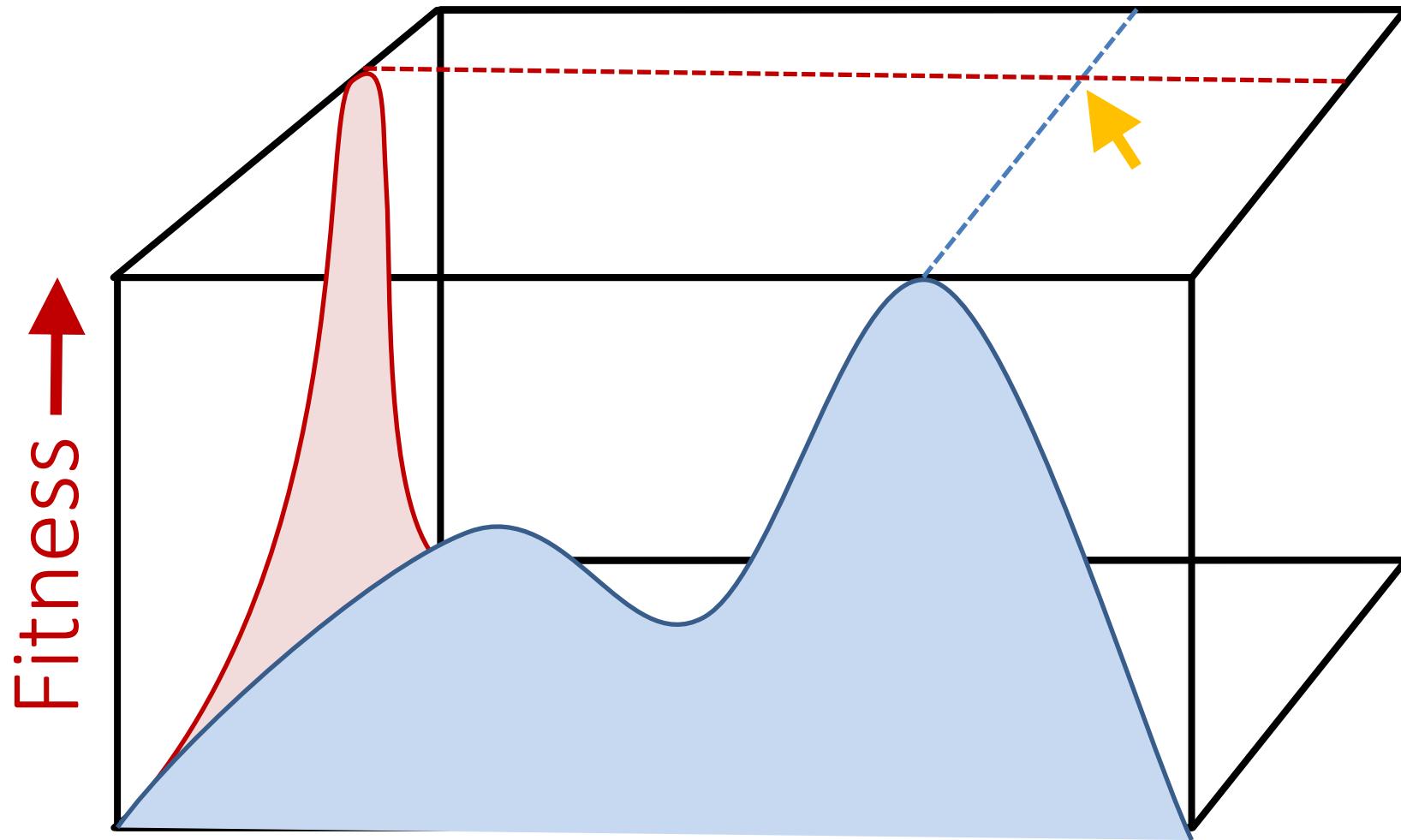
“Fitness landscape” concept

If we think of how a single trait relates to fitness:



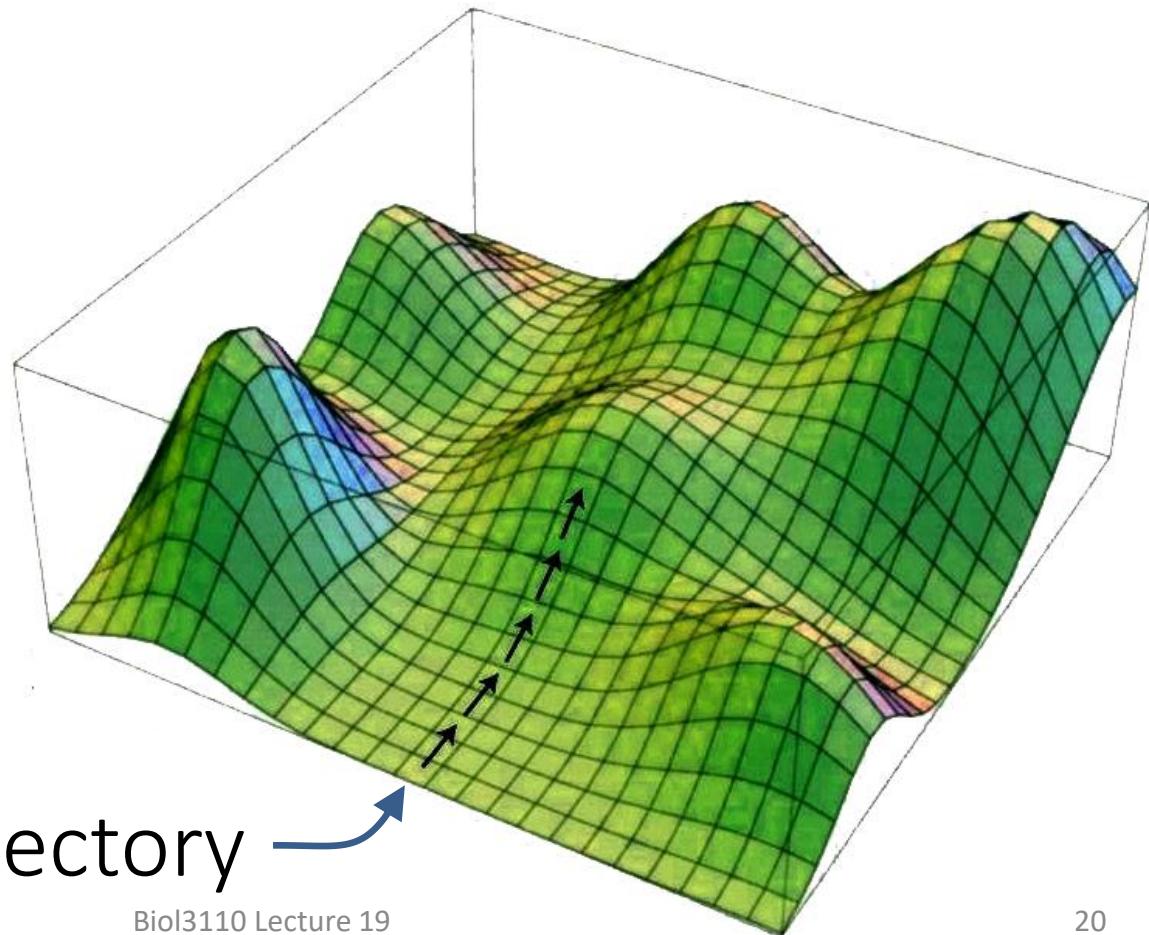
“Fitness landscape” concept

If we think of how **multiple traits** relate to fitness:



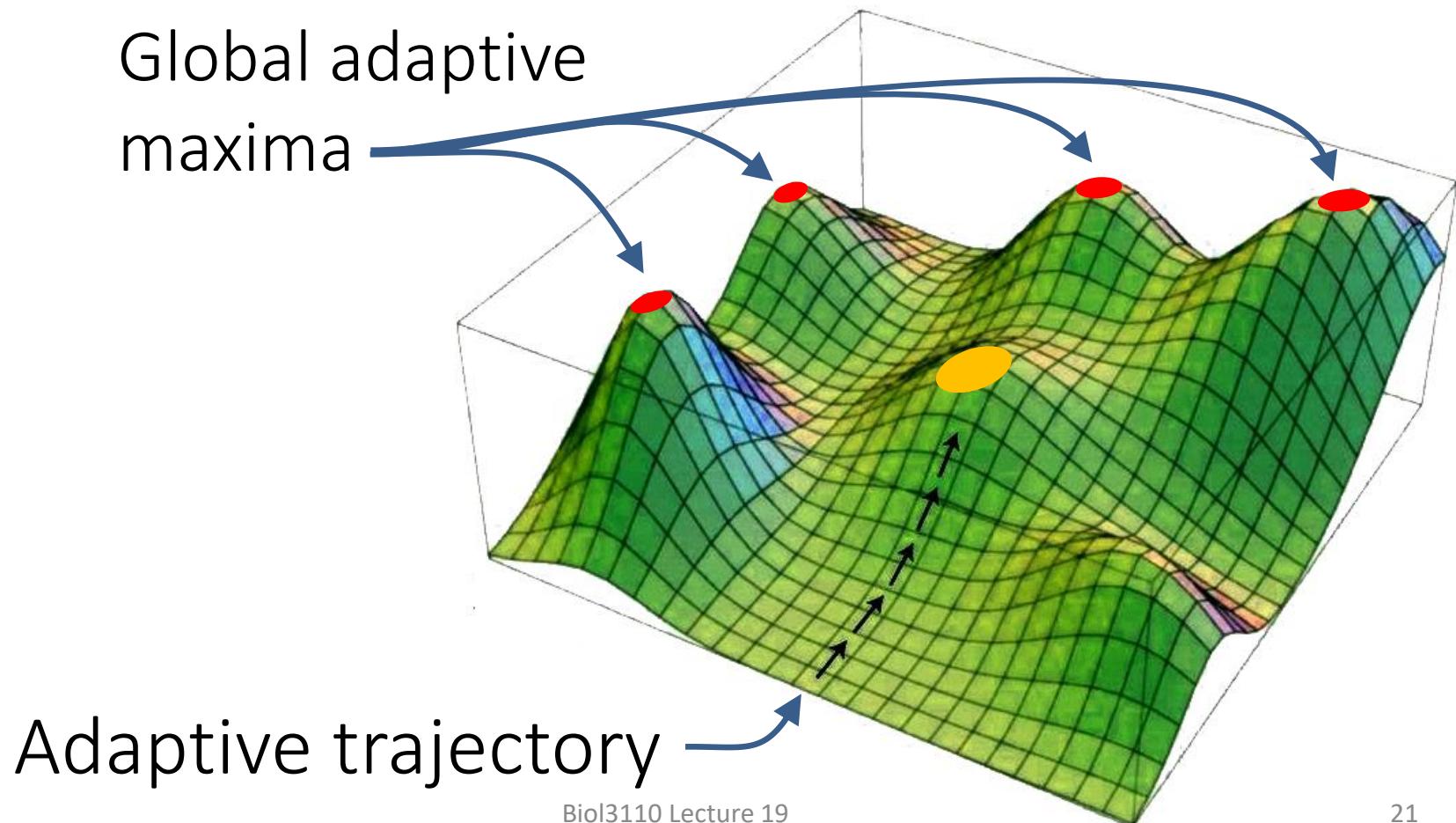
“Fitness landscape” concept

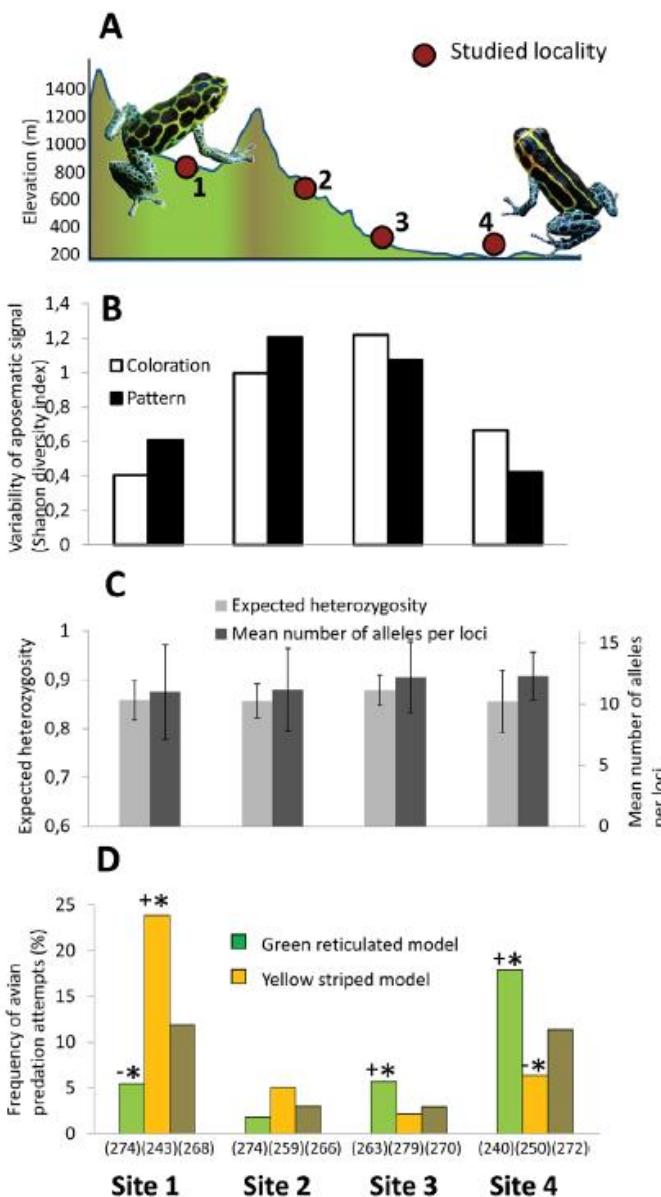
Sewall Wright: Species evolve to localised adaptive “peaks” in an n-dimensional “landscape”



“Fitness landscape” concept

Sewall Wright: Species evolve to localised adaptive “peaks” in an n-dimensional “landscape”





Wrights shifting balance theory –
relaxation of selection + drift can
allow for polymorphisms

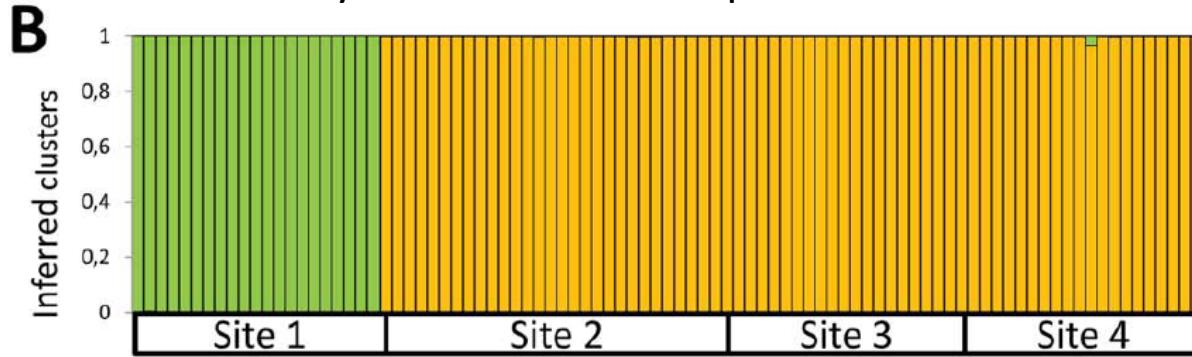


High variability sites 2 & 3



Low rates of predation sites 2 & 3

Hybridisation can't explain the variation



Wrights shifting balance theory – relaxation of selection + drift can allow for polymorphisms that then ‘compete’ across the fitness landscape

