# BIOL3110 Evolutionary and Conservation Genetics

## LECTURE 2: GENETICS AND EXTINCTION

# Genetics & Extinction

MACQUARIE University

## 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

SCIENCE DIRECT®

Biological Conservation 126 (2005) 131–140

BIOLOGICAL CONSERVATION

www.elsevier.com/locate/biocon

Review

Genetics and extinction

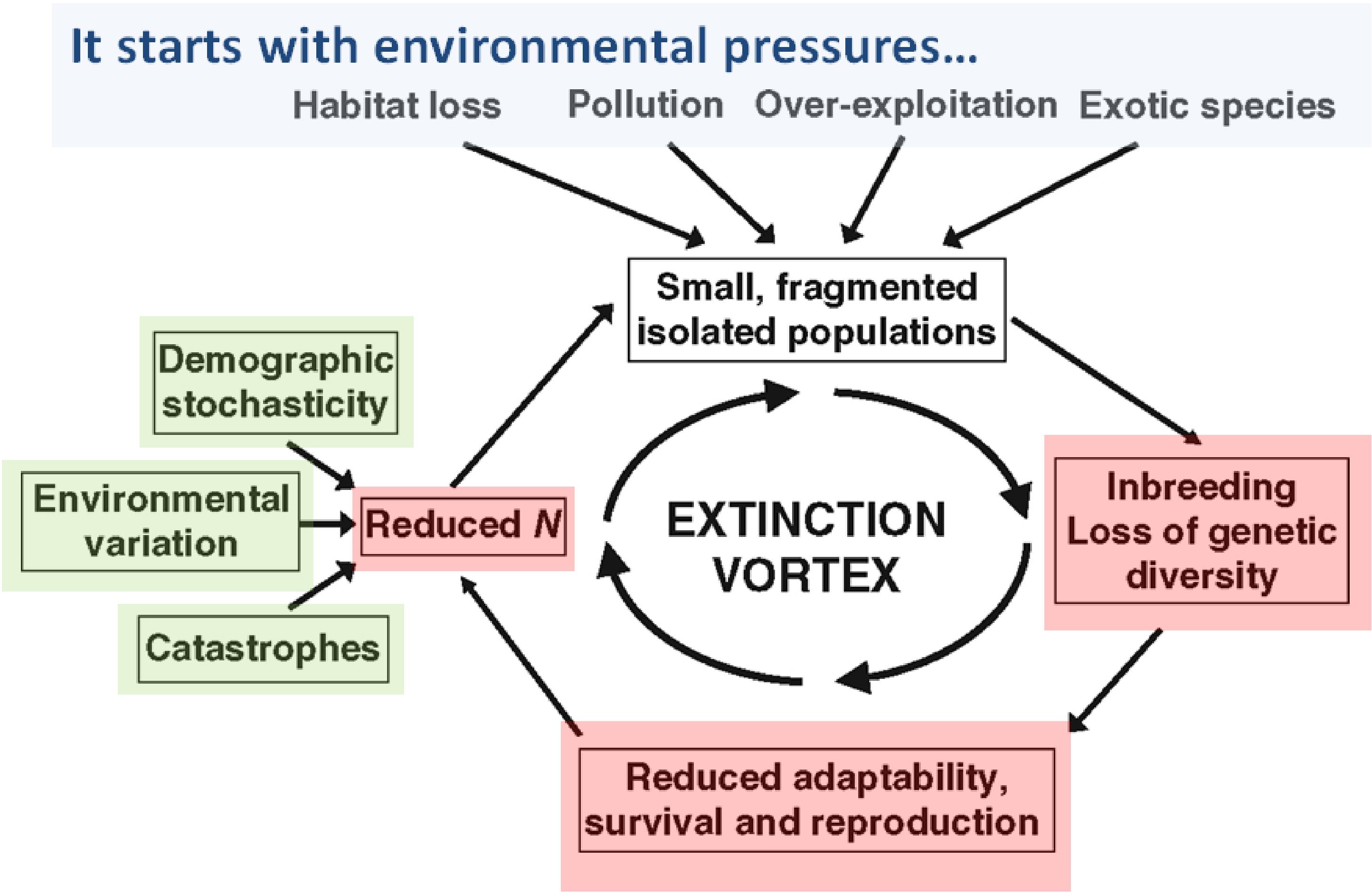
Richard Frankham *

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

Received 2 March 2005
Available online 20 June 2005

# Genetics & Extinction

## 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 ancestory

(or the average expected "genetic overlap" due to shared ancestory)

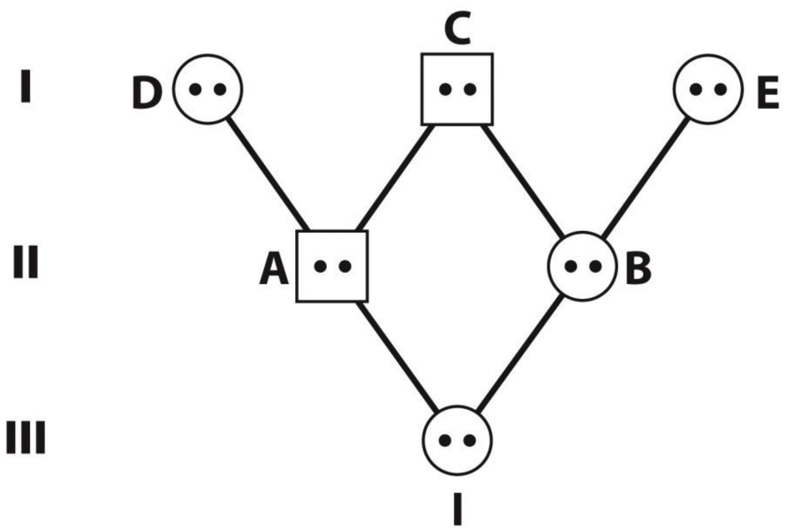
Reaches **0.986** after 20Gen of full-sib matings (~genetic "isoline")

| 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 |

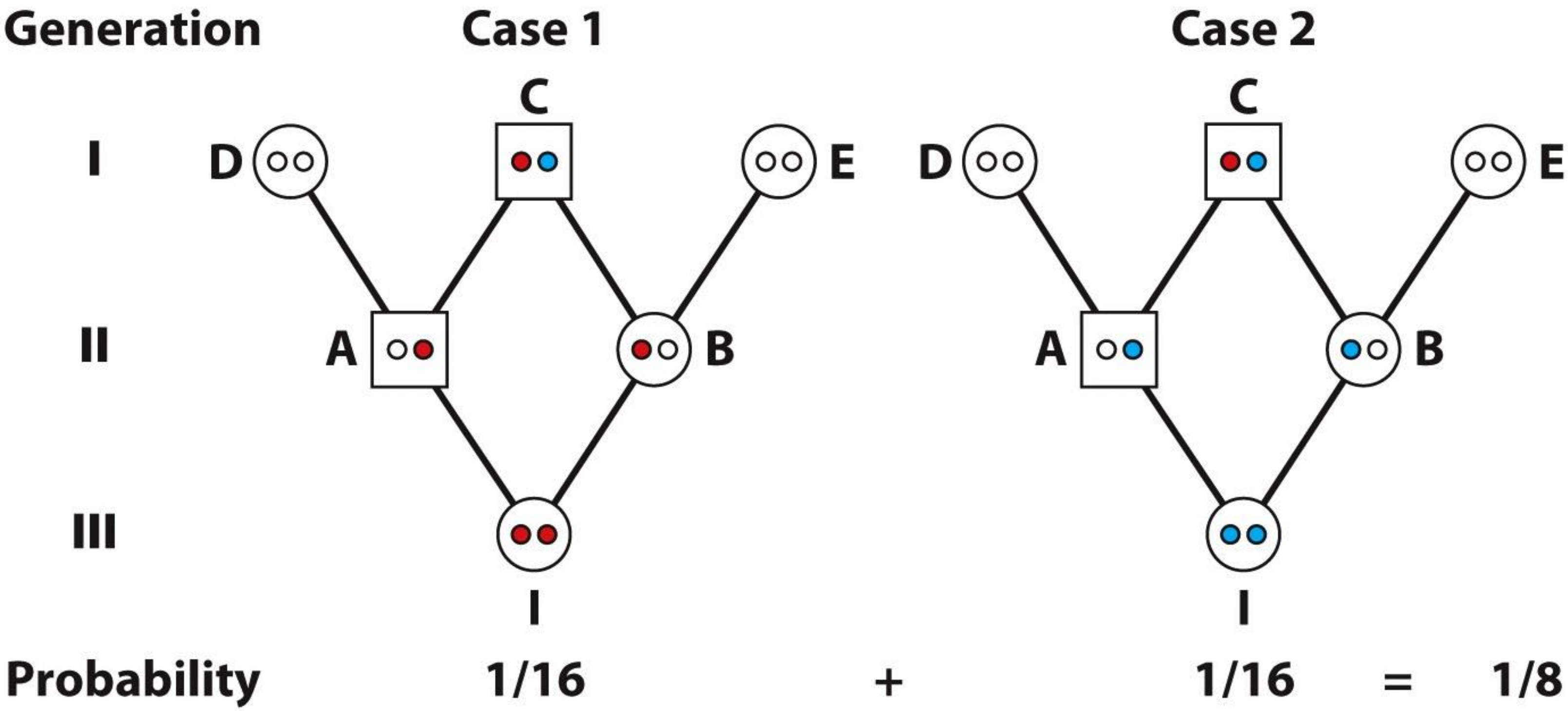
**FOR POPULATIONS:**

- 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
   - $(1/2)^4$ X 2 = 1/8, so $F$ = 1/8


Generation
Case 1
Case 2
C
I
D
E
II
A
B
III
I
Probability
1/16
+
1/16
=
1/8

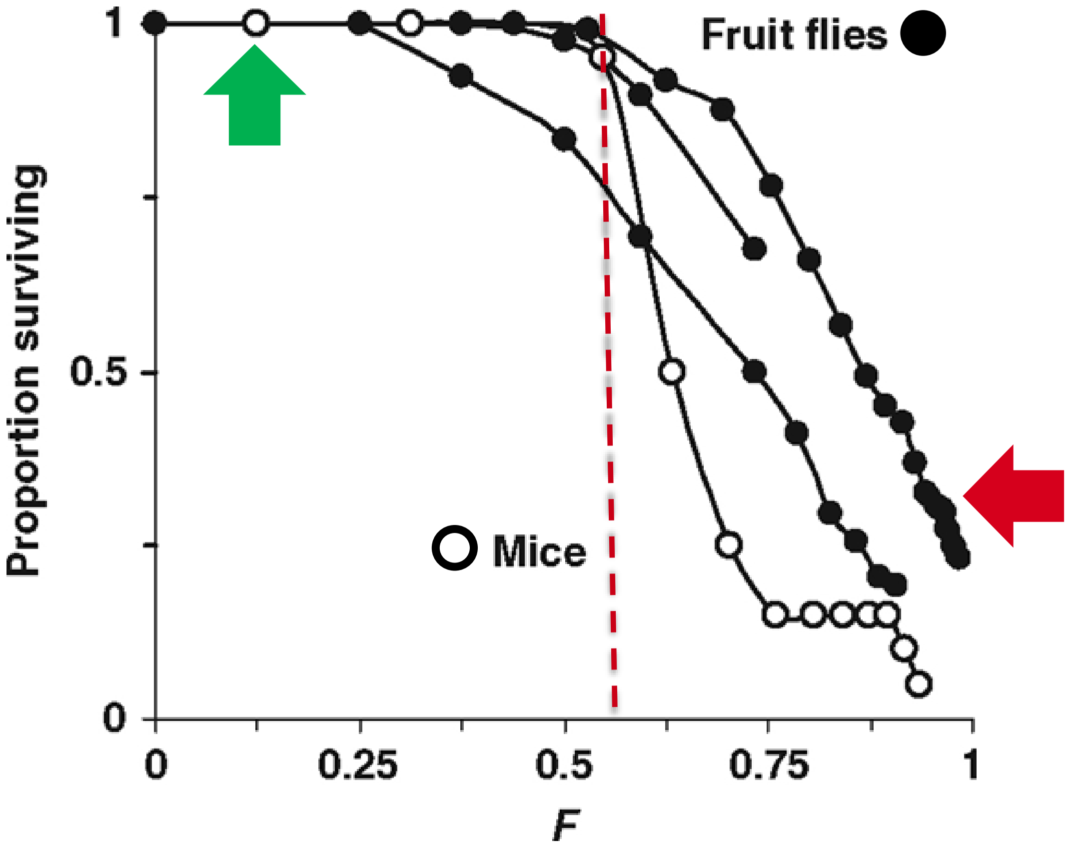
# Genetics & Extinction

## 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

MACQUARIE University

## INBREEDING CAUSES EXTINCTION IN THE WILD

### DIRECT EVIDENCE

Inbreeding depression, revealed in phenotypes, directly related to extinction

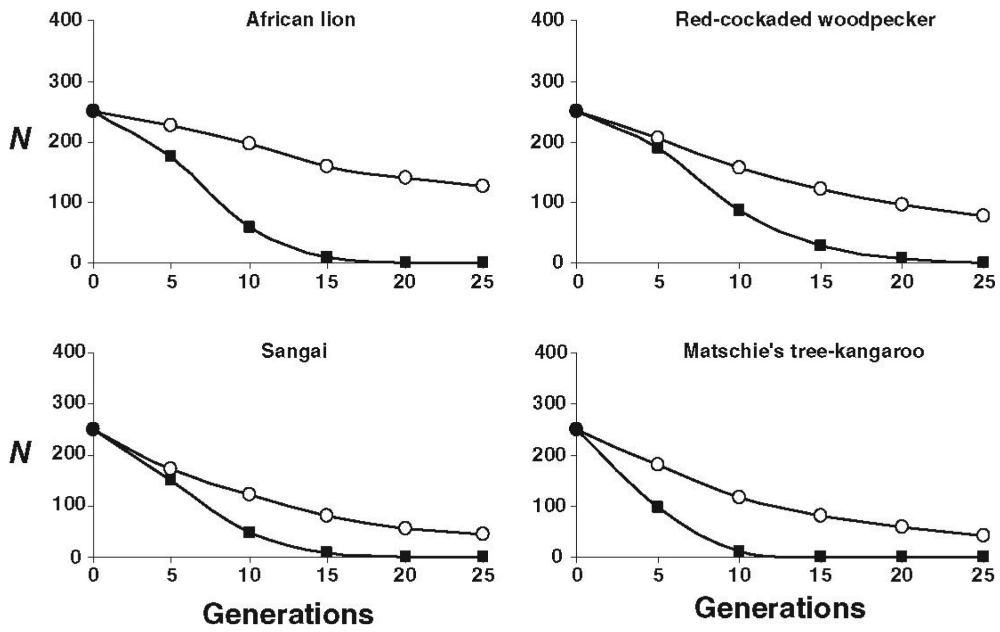
(e.g. observed fitness & recruitment in Fla panther & Japanese quail)

Pr(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…

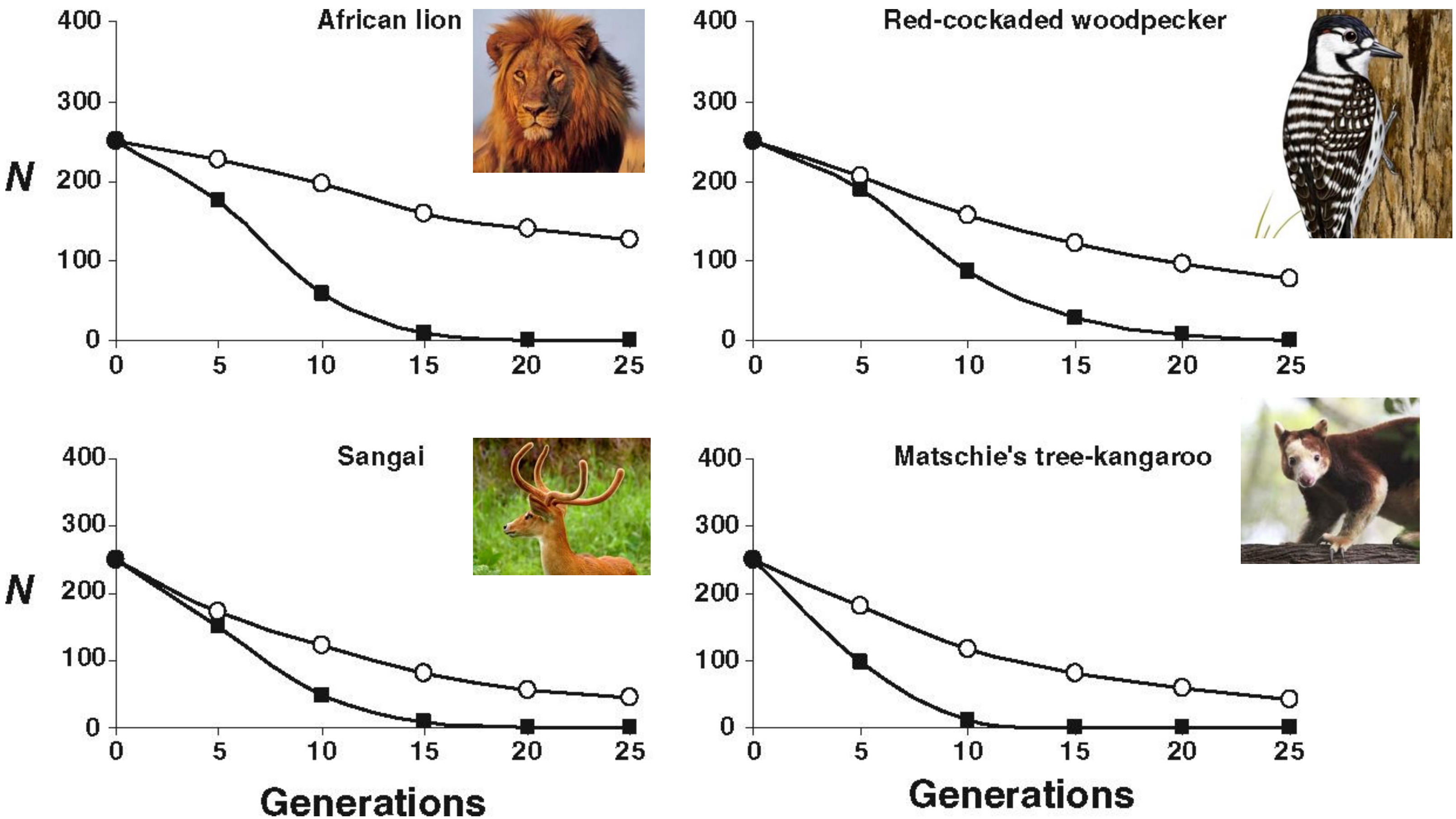


# Genetics & Extinction

## 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 | WiI | WeI |
|---|---|---|---|---|---|---|---|---|
| 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 | + | − | − | − | − | − | − |

Black-footed rock wallaby

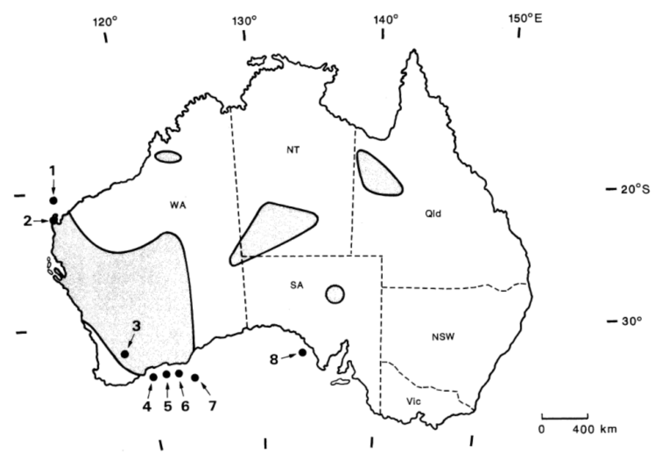
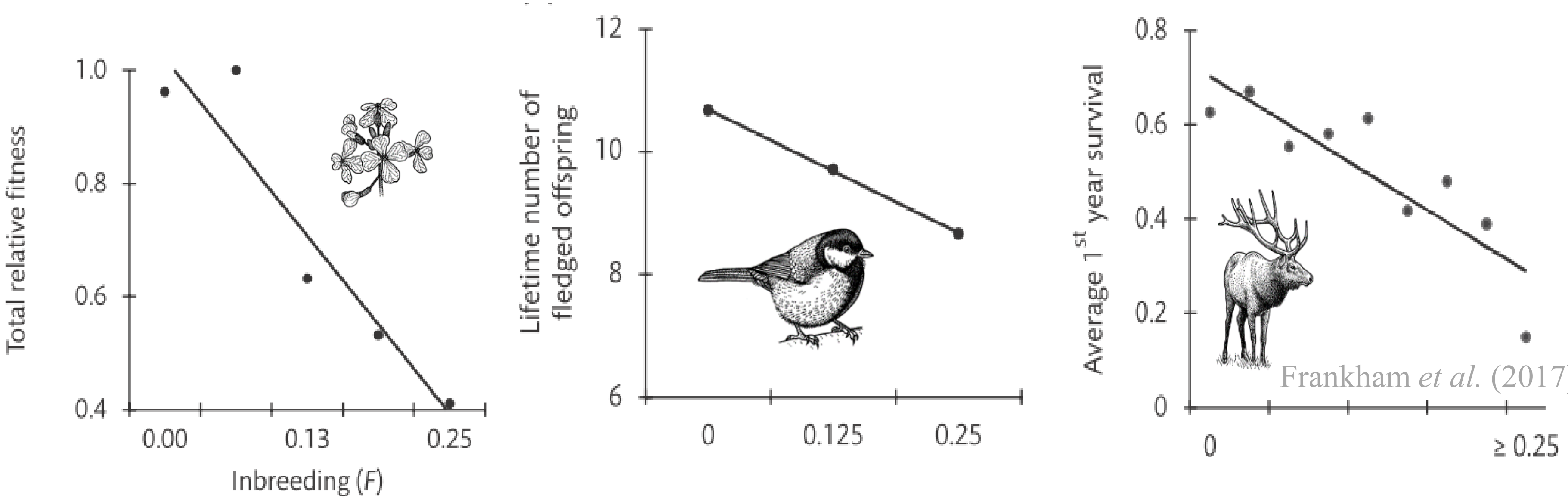



*Figure 1. Distribution of the black-footed rock-wallaby* (Petrogale lateralis) *in Australia. Collection localities are Barrow Island (1), Exmouth (2), Wheatbelt (3), Wilson Island (4), Mondrain Island (5), Westall Island (6), Salisbury Island (7), Pearson Island (8).*

# Small, isolated populations suffer loss of Vg

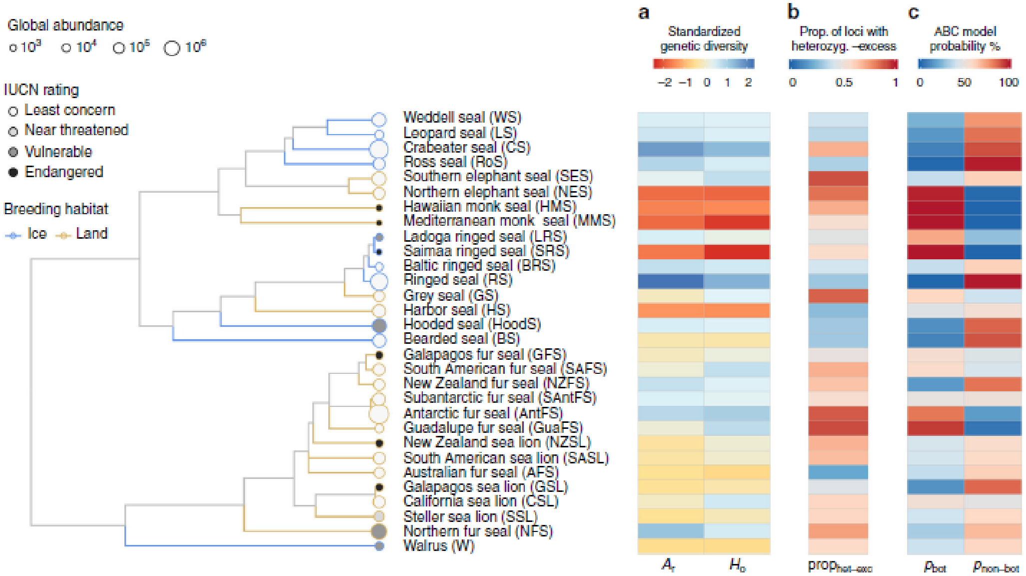
(1) Inbreeding depression seen in virtually all species that usually outbreed




Loss of useful genetic variation from populations

These genetic problems cause extinction

# Genetic variation and endangered species

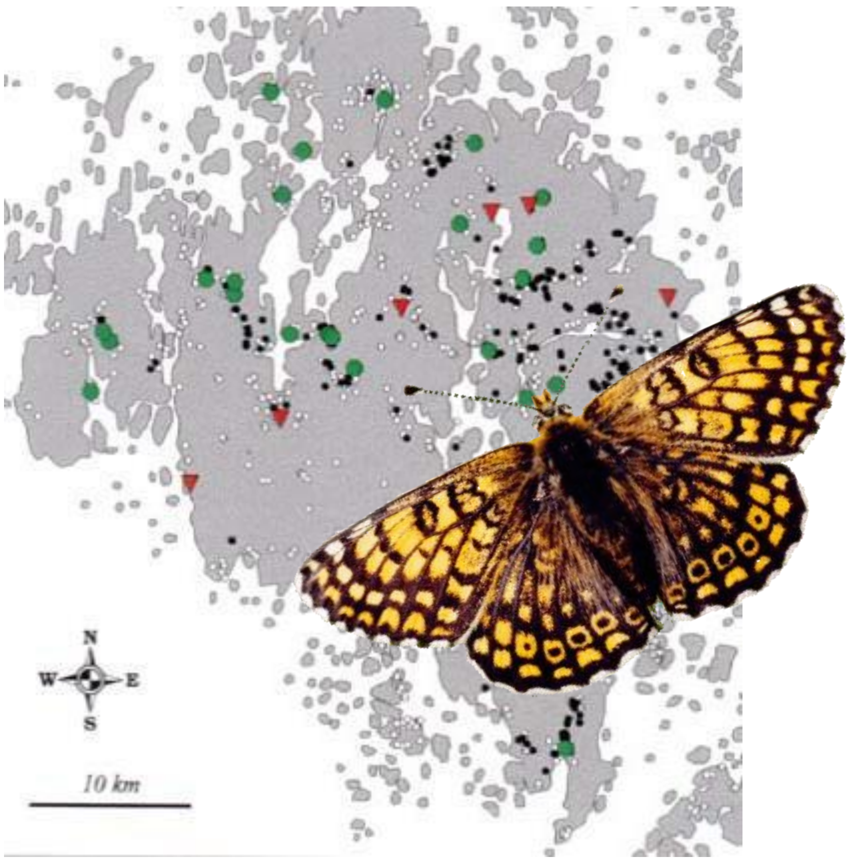



Probability of bottleneck

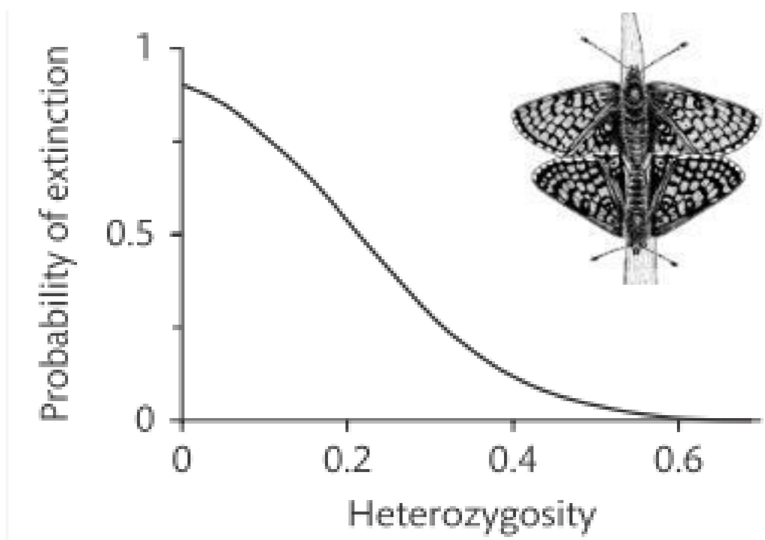
Stoffel et al 2018 Nature Communications

## Glanville fritillary butterfly

Saccheri *et al*. 1998 *Nature*

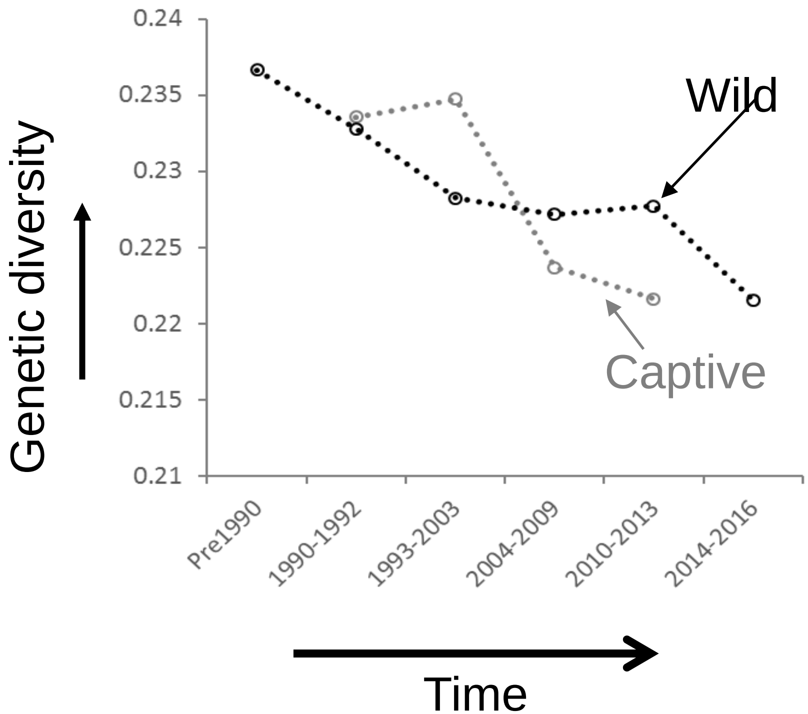



Simple genetic diversity was the best predictor of local extinction

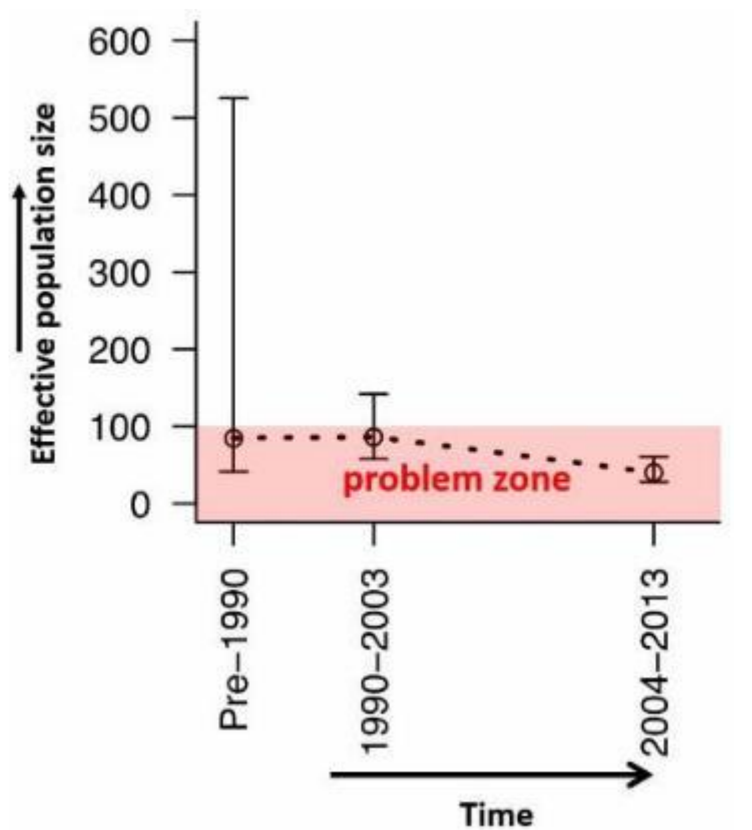



Frankham *et al.* (2017)

# Genetic variation and fitness




Inbred to a degree that is usually harmful

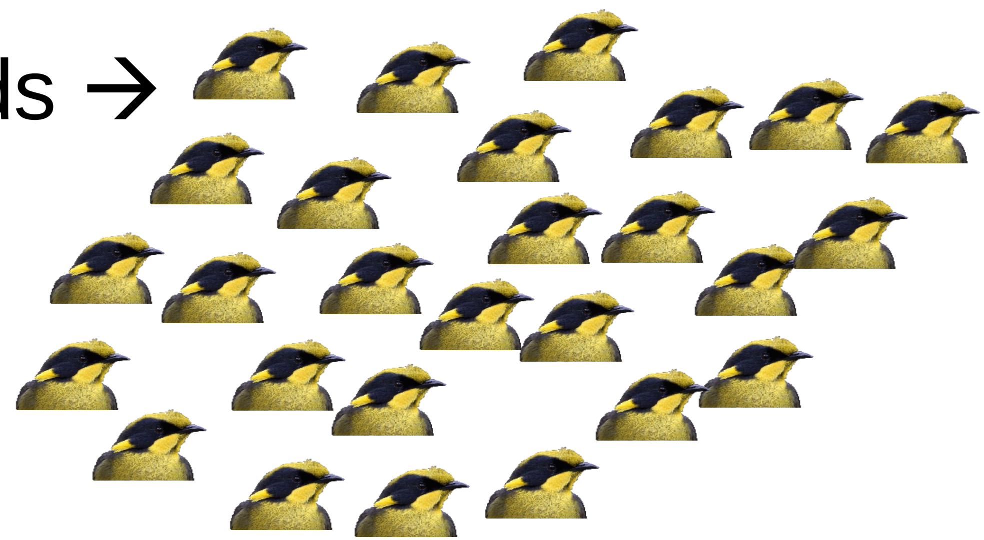



Harrisson et al. 2016
*Molecular Ecology*

# More-inbred birds have far fewer offspring in their lifetimes

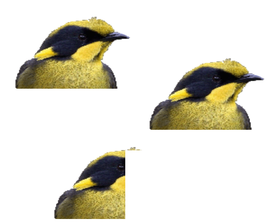
The most outbred birds →

27 fledglings



The most inbred →

2.5 fledglings



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

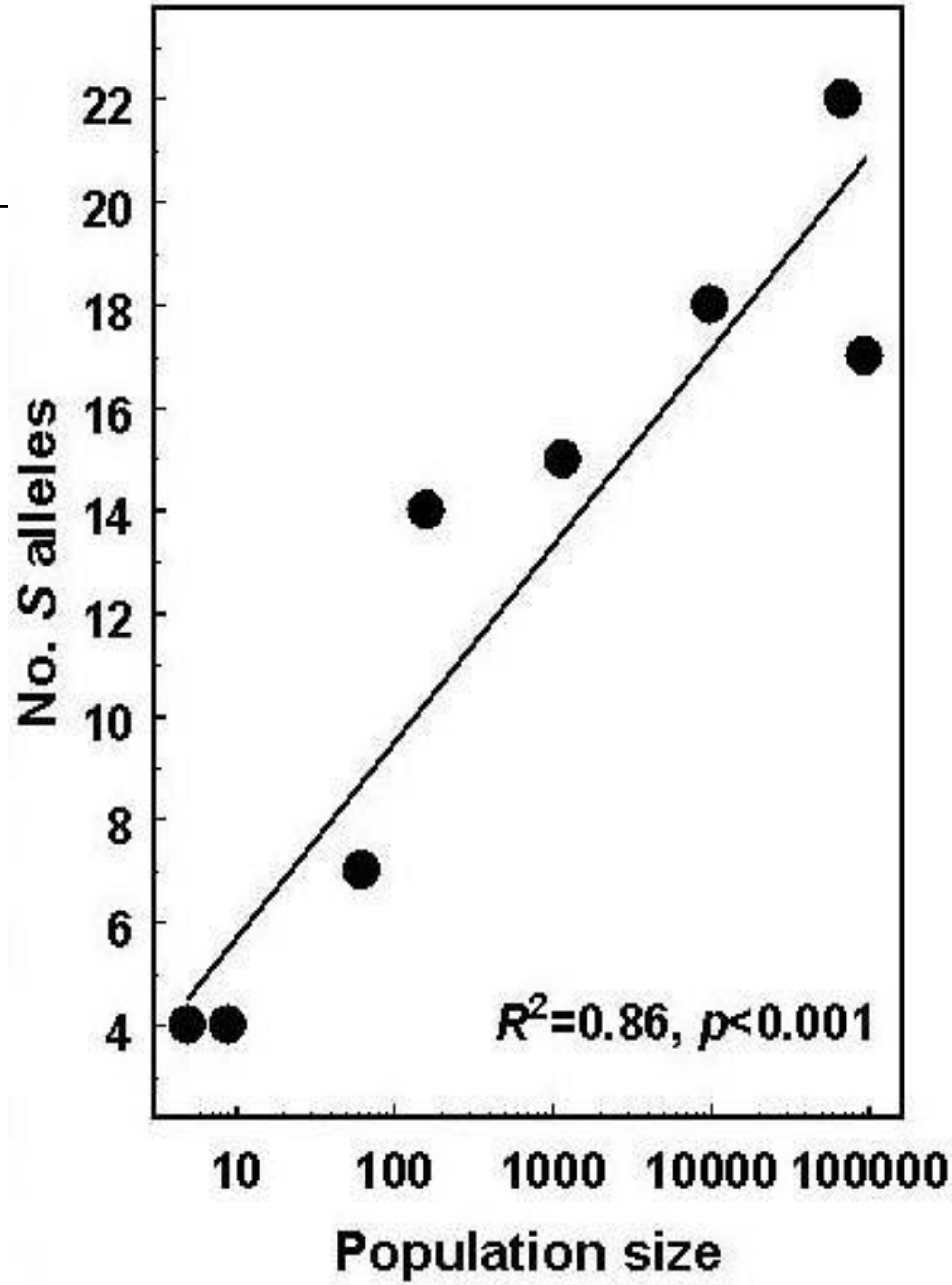
# An example of how connectivity has been influenced the mating system of a plant

MACQUARIE University

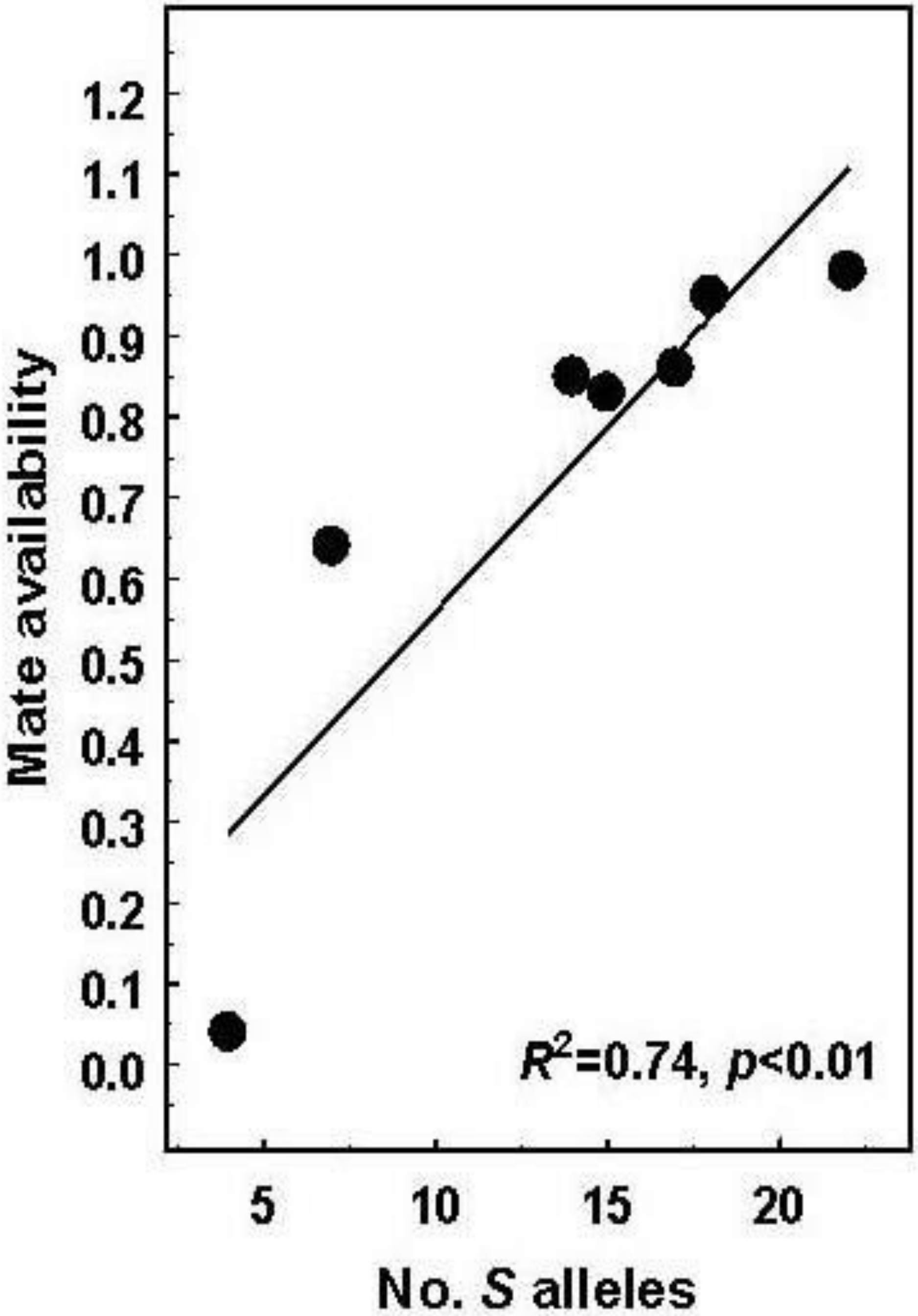
- Button wrinklewort, 24 sites in 2 groups 500Km apart
- Largely self-incompatible i.e. cannot mate with self or very similar individuals – simple genetic control (SI locus)

Andrew Young
Button wrinklewort *Rutidosis leptorrhychoides*

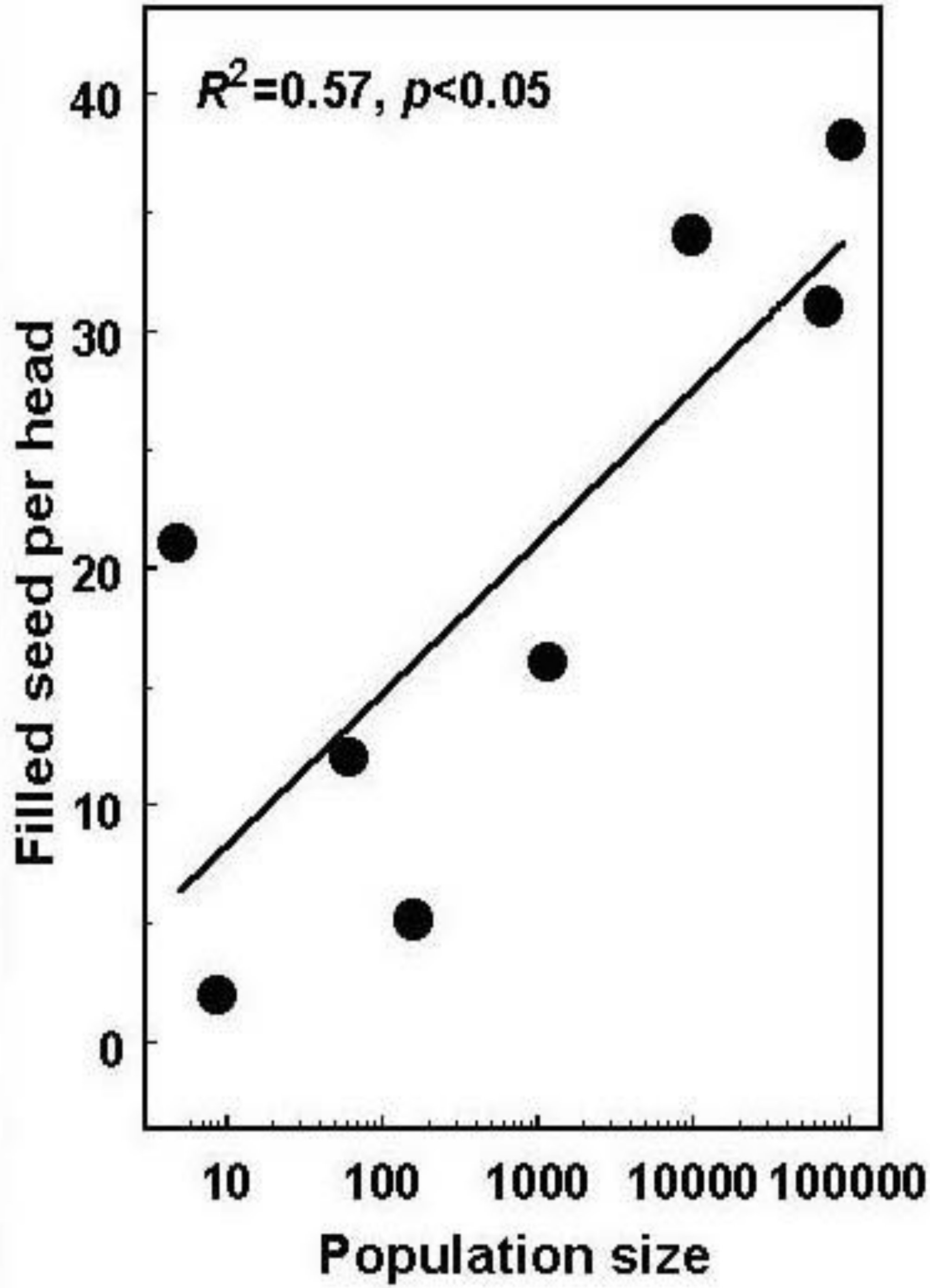
- genetic erosion - SI alleles lost



• lost SI alleles make many mates incompatible



• mate limitation reduces female success in small populations



# Genetics & Extinction

## GENETIC VARIANCE

### GENETIC VARIANCE or DIVERSITY

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

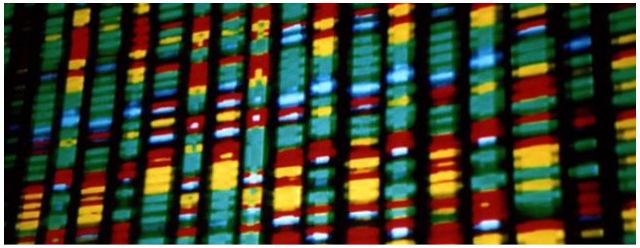
### HOW WE MEASURE IT

**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

### WHY IS VG SO IMPORTANT?

(1) It informs the **potential for inbreeding depression**;

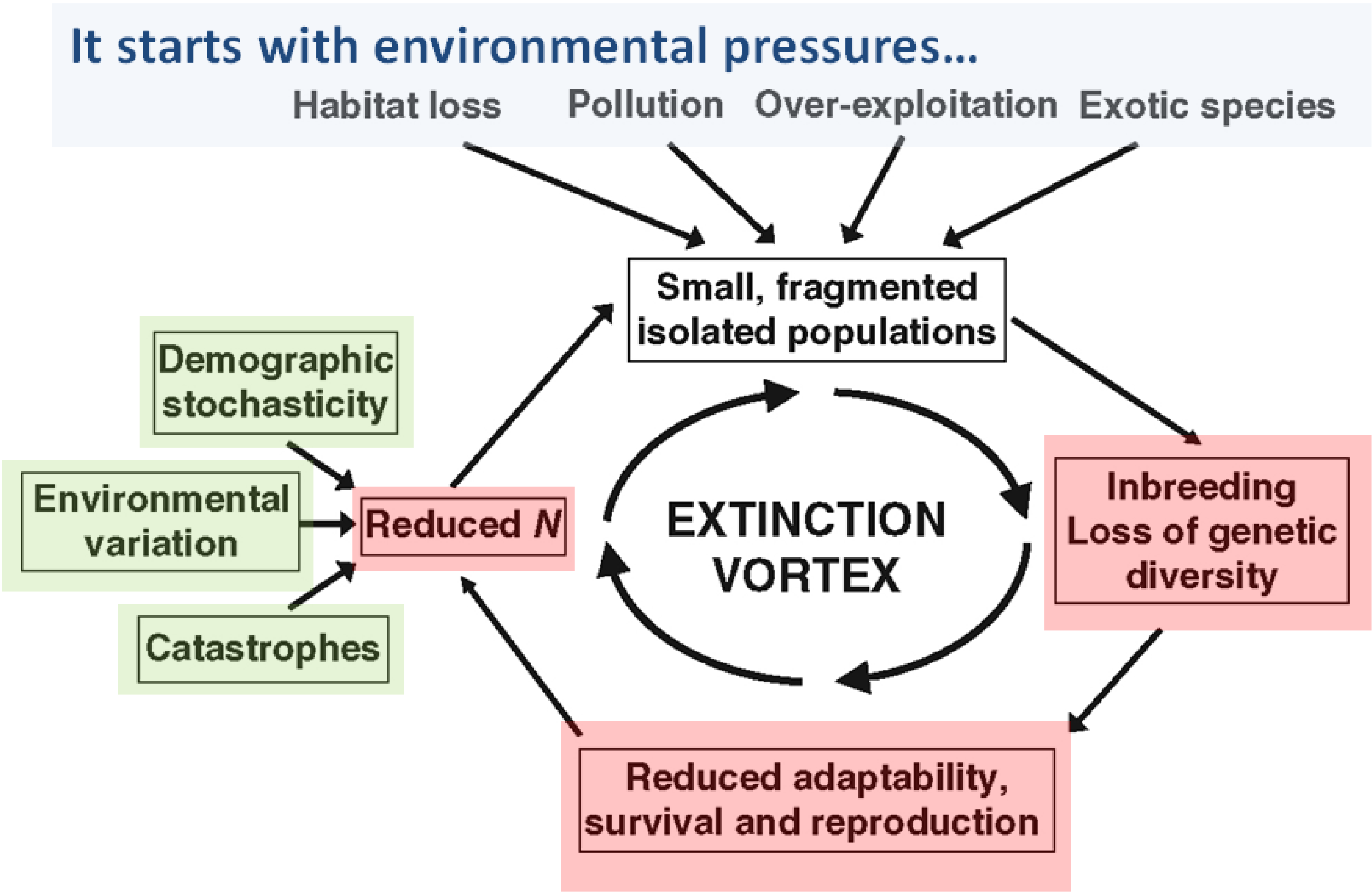
(2) It allows **population persistence** even in extreme environments, buffering:

- 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

EXPLICITLY IN TERMS OF $V_G$



# Genetics & Extinction

## FURTHER READINGS (ON Ilearn)

Available online at www.sciencedirect.com

SCIENCE DIRECT®

BIOLOGICAL CONSERVATION

Biological Conservation 126 (2005) 131–140

www.elsevier.com/locate/biocon

Review

Genetics and extinction

Richard Frankham *

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

BIOLOGICAL CONSERVATION 133 (2006) 42–51

ELSEVIER

available at www.sciencedirect.com

ScienceDirect

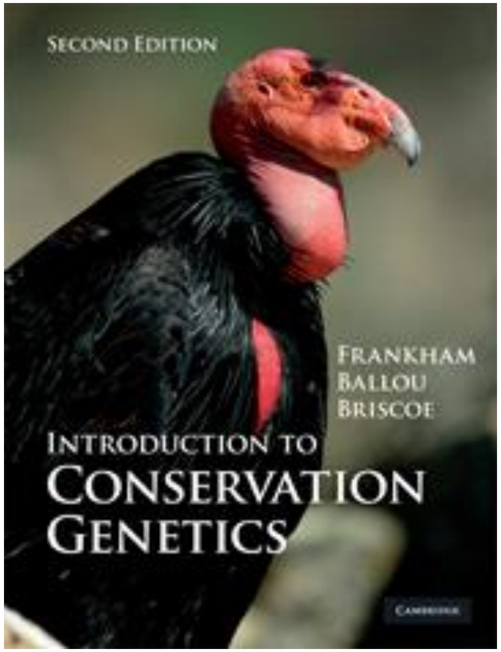
journal homepage: www.elsevier.com/locate/biocon

BIOLOGICAL CONSERVATION

**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]*

[a]Biodiversity and Bioresources Group, Department of Biological Sciences, Macquarie University, Sydney, New South Wales 2109, Australia
[b]School for Environmental Research, Institute of Advanced Studies, Charles Darwin University, Darwin, Northern Territory 0909, Australia
[c]Department of Biological Sciences, Clemson University, SC 29634, USA




Chapter 2

# Next lecture:

$V_G$ in more detail