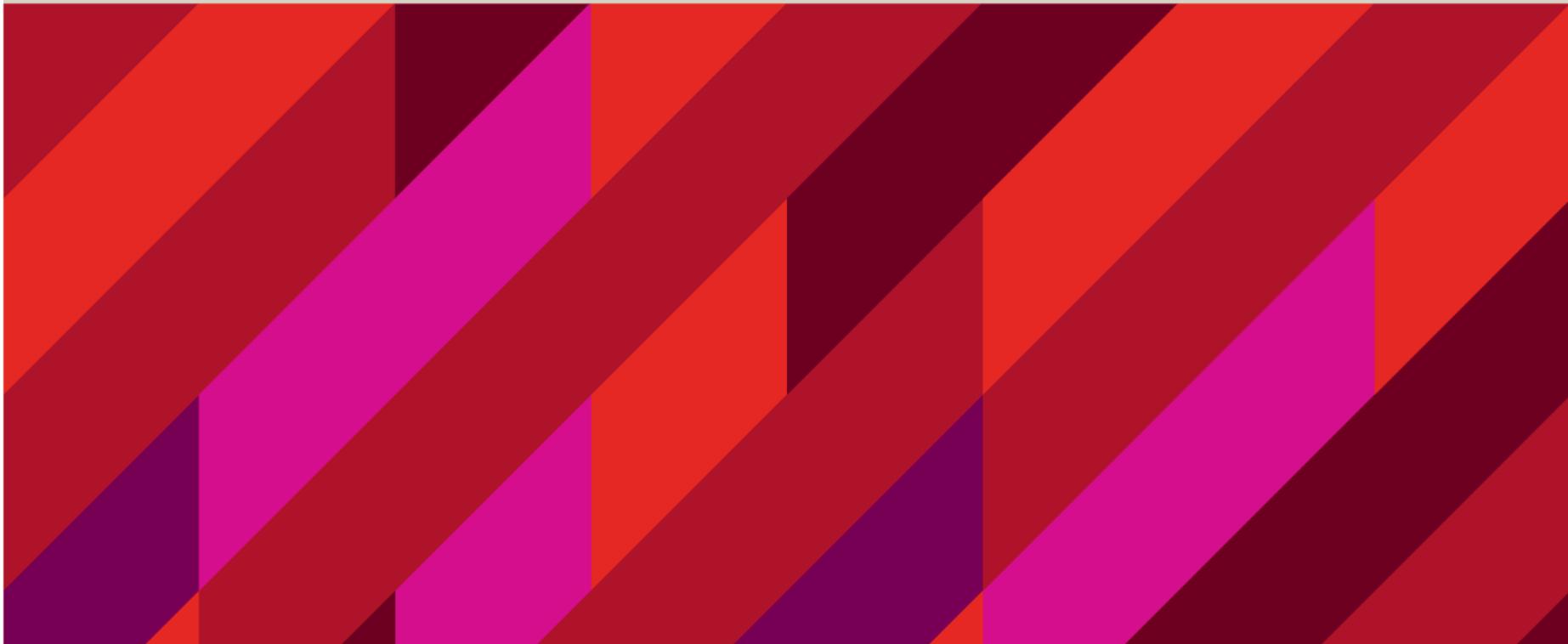




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# BIOL3110 Conservation & Ecological Genetics

## 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 Wang<sup>1</sup>, E Santiago<sup>2</sup> and A Caballero<sup>3</sup>

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 the 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

### Have you listened to the *Heredity* podcast?

The *Heredity* podcast features interviews with the people behind the science and a digest of breaking news. Recent episodes have included:



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## ORIGINAL ARTICLE

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

A-K Mueller, N Chakarov<sup>2</sup>, 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

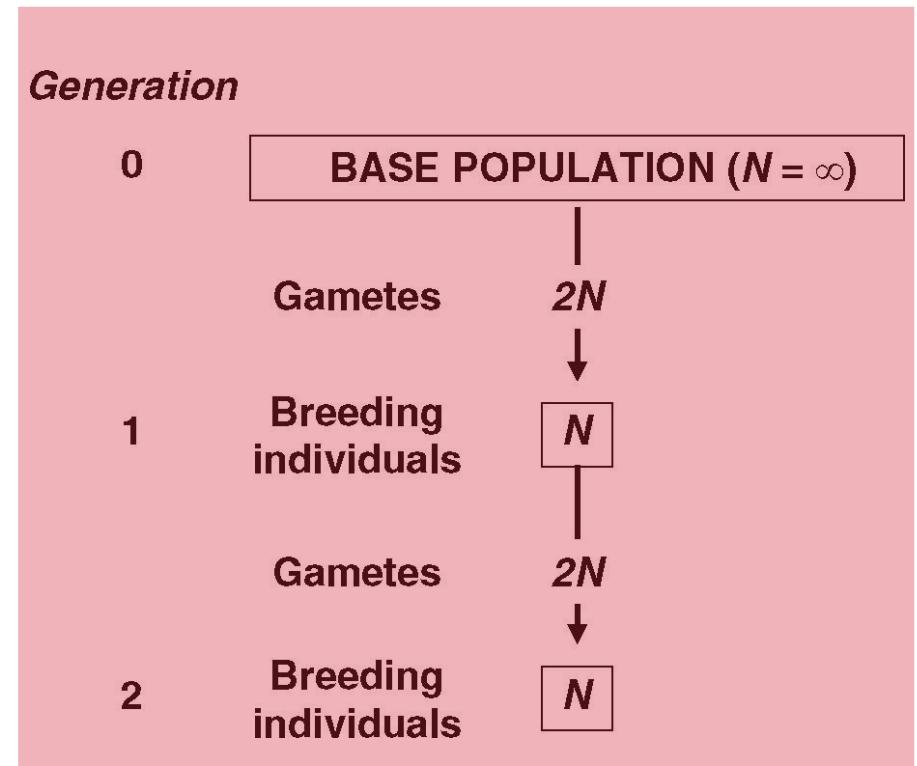
# Effective Population Size ( $N_e$ )

## THE IDEALIZED POPULATION

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

A given level of  $F$

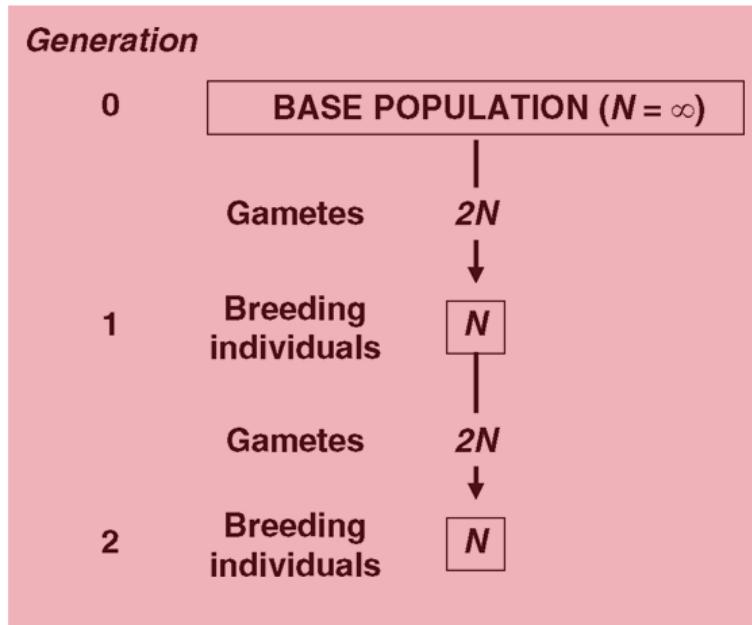
A given dynamic of  $V_G$



# Effective Population Size ( $N_e$ )

## THE IDEALIZED POPULATION

As  $N$  departs from  $\infty$  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:

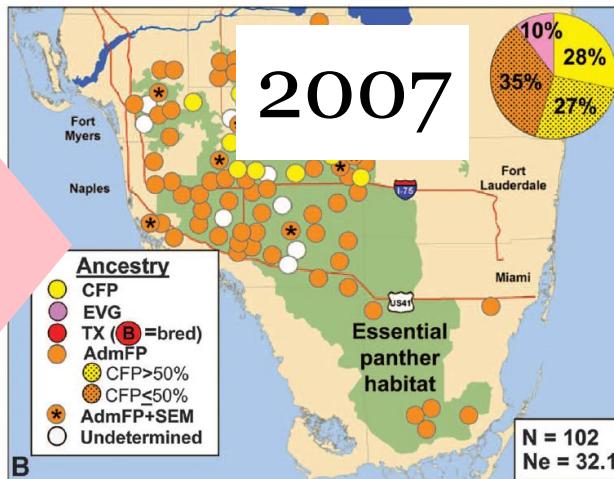
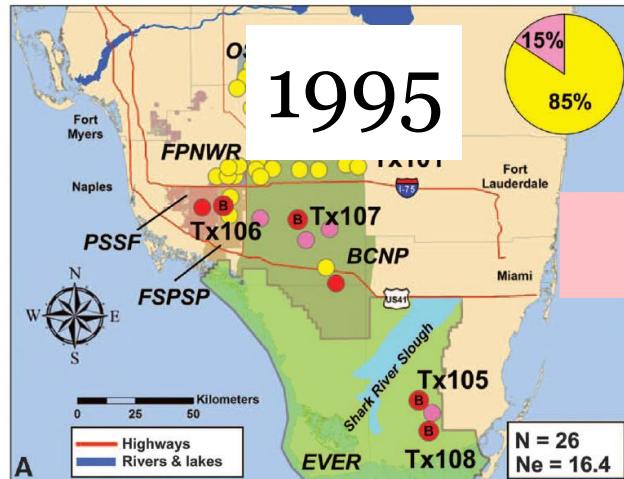
$N = 2000$  but  $N_e = 85$

$N_e/N \sim 4\%$



# Effective Population Size ( $N_e$ )

OFTEN OF INTEREST IS  $N_e/N$



$$N = 26$$

$$N_e = 16.4$$

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

$$N = 102$$

$$N_e = 32.1$$

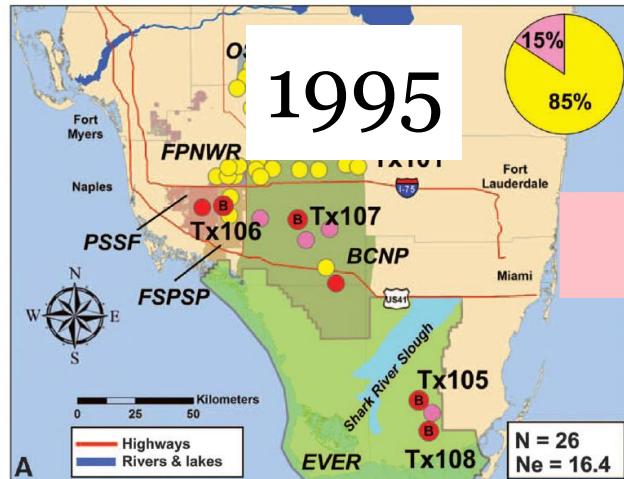
$$N_e/N = \mathbf{0.31}$$

# Effective Population Size ( $N_e$ )

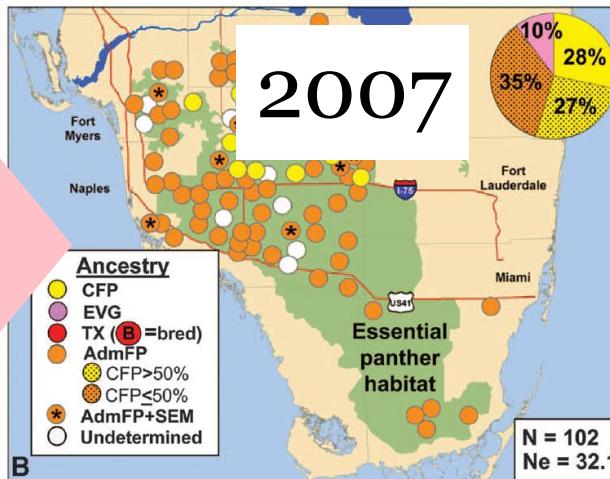


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OFTEN OF INTEREST IS  $N_e/N$



1995



2007



$$\begin{aligned}N &= 26 \\N_e &= 16.4 \\N_e/N &= \mathbf{0.63}\end{aligned}$$

$$\begin{aligned}N &= 102 \\N_e &= 32.1 \\N_e/N &= \mathbf{0.31}\end{aligned}$$

**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;

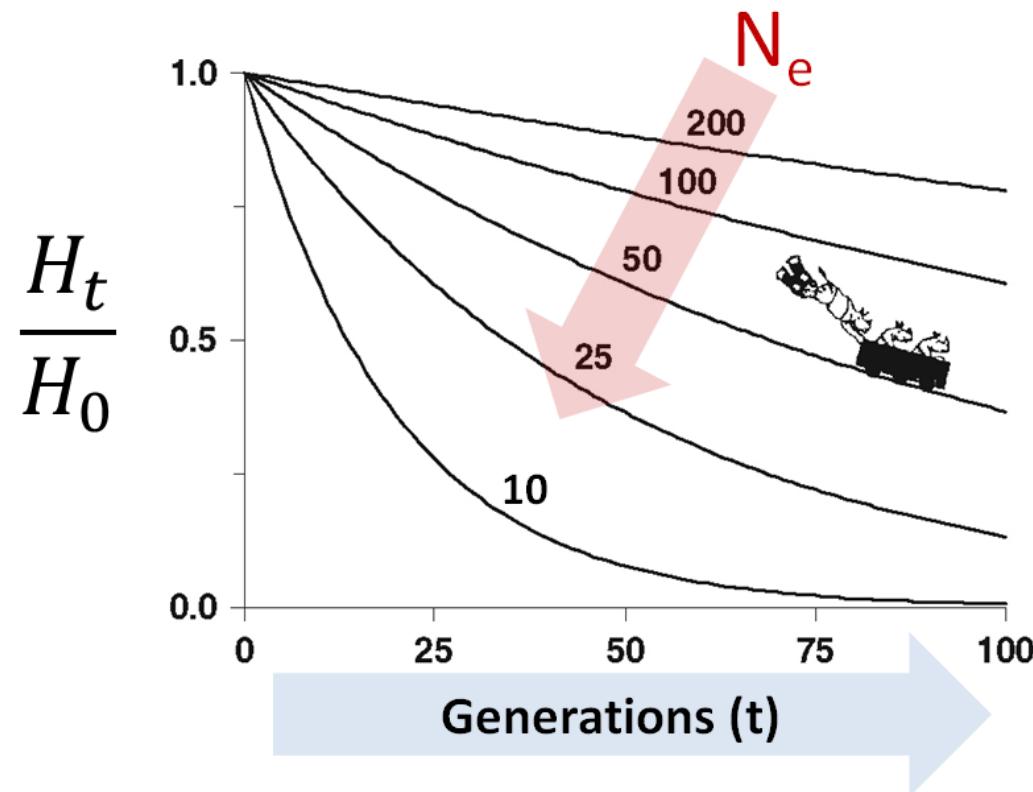
# Effective Population Size ( $N_e$ )



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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...

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Where:  $t$  = number of generations

~0.1% of  
 $H$  lost

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

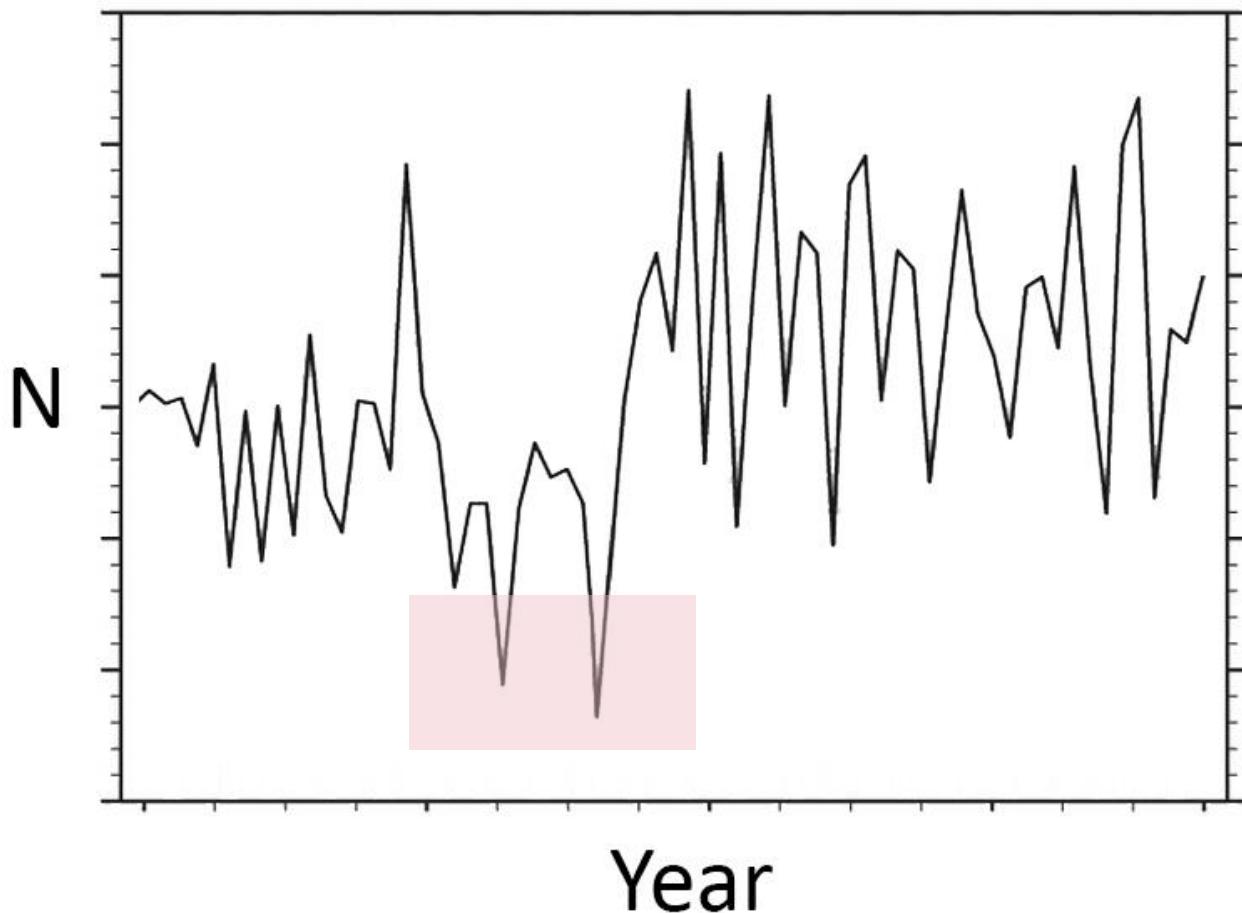
$$\frac{H_1}{H_0} = \left(1 - \frac{1}{2N_e}\right)^t = \left(1 - \frac{1}{1000}\right)^{50} = 0.951$$

~5% of  $H$   
lost

# Effective Population Size ( $N_e$ )

## ESTIMATION – DEMOGRAPHIC FACTORS

### Fluctuations in $N$ over time (1)

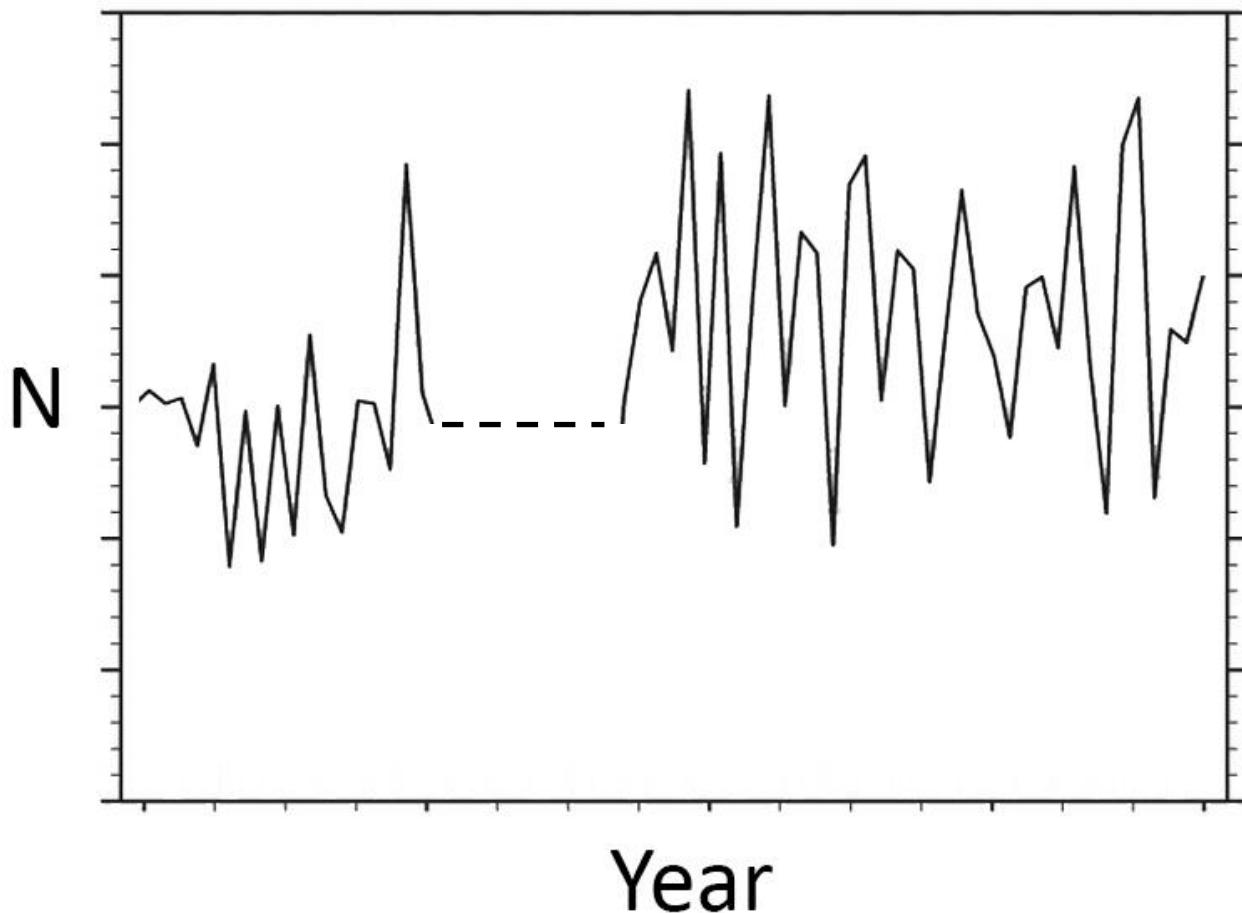


$$N_e = \frac{t}{\sum(\frac{1}{N_{e1...t}})}$$

# 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:



$$N_e = \frac{t}{\sum(\frac{1}{N_1 \dots t})}$$

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

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

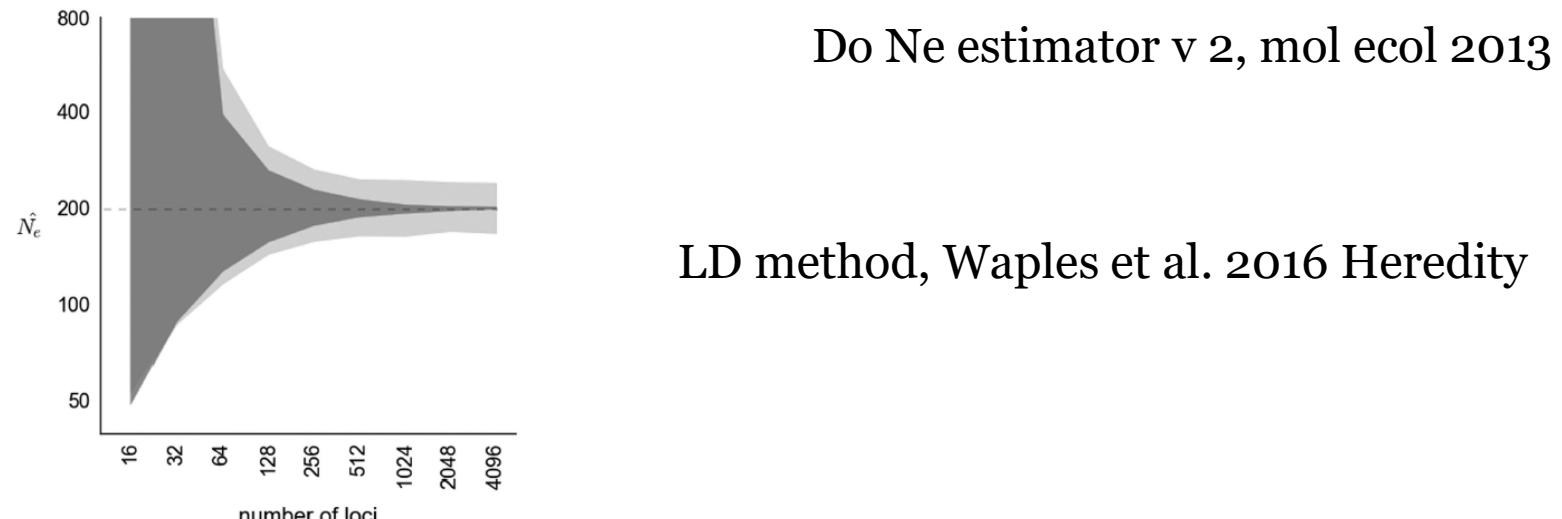
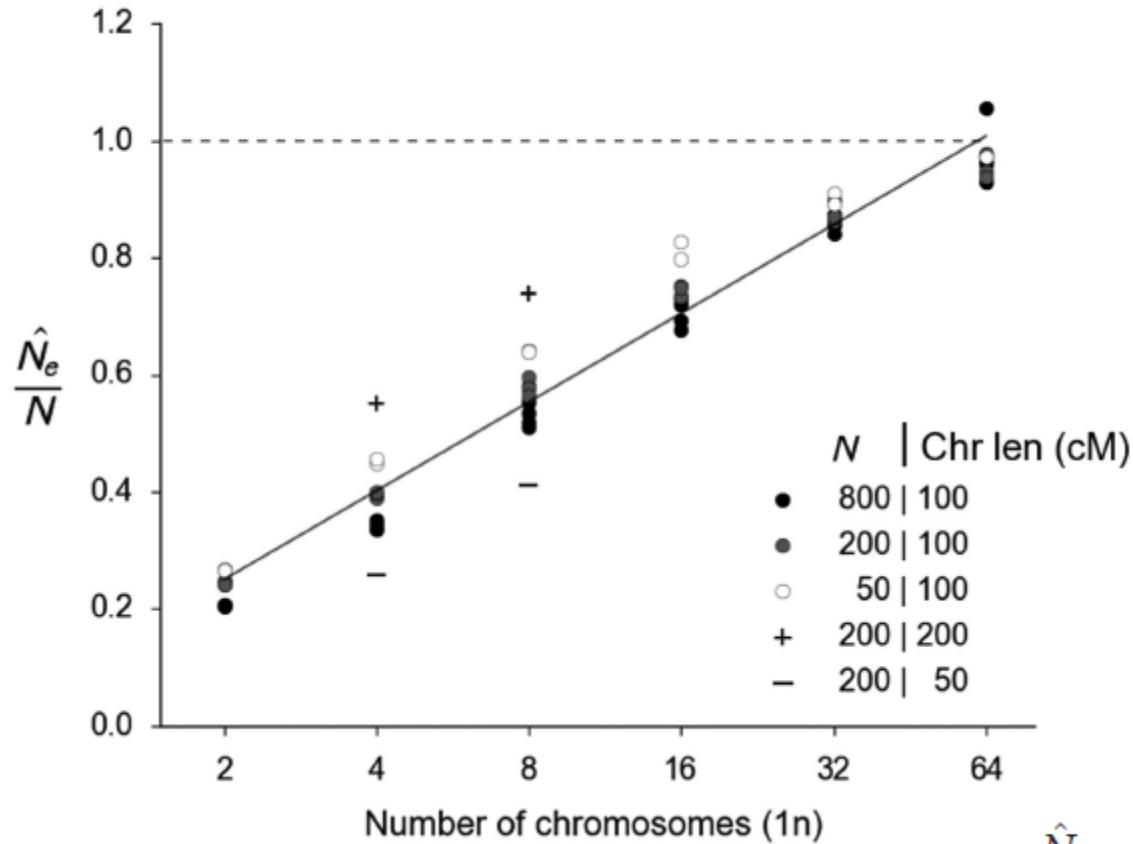


Figure 7 Relationship between theoretical and empirical (light) precision for estimates of  $N_e$ . Dark shaded area shows naive 95% CIs (which assume that all pairwise comparisons are independent), light shaded area shows empirical CIs from simulated data. True  $N_e$  was 200, sample size was 100 and 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..



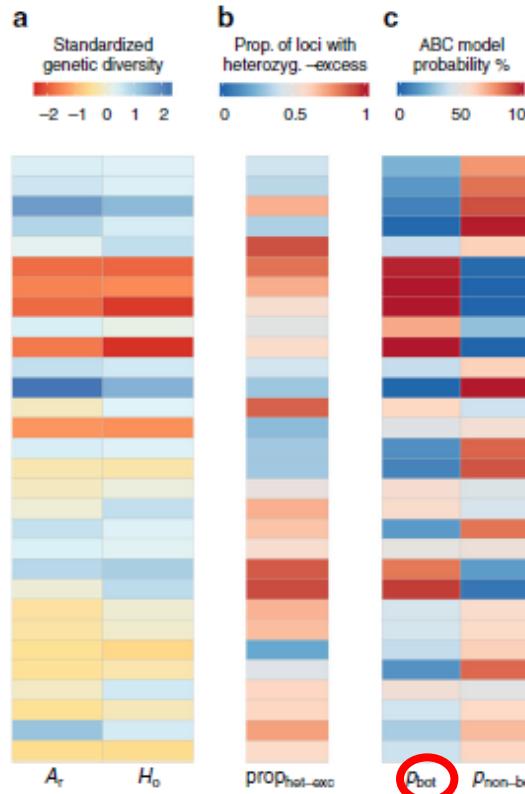
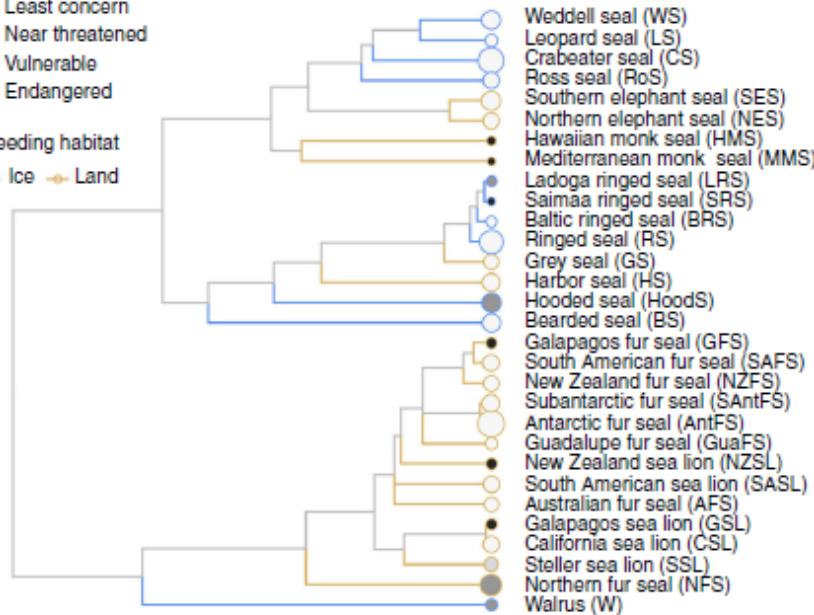
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Global abundance  
 $\circ 10^3$   $\circ 10^4$   $\circ 10^5$   $\circ 10^6$

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

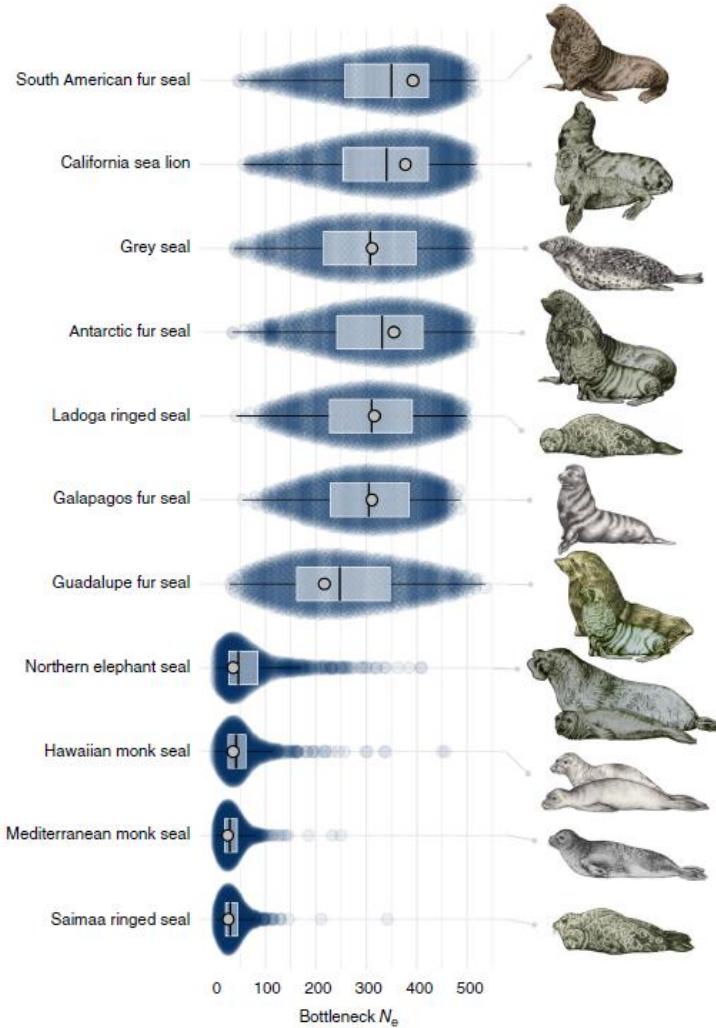
Breeding habitat  
 $\leftarrow$  Ice  $\rightarrow$  Land



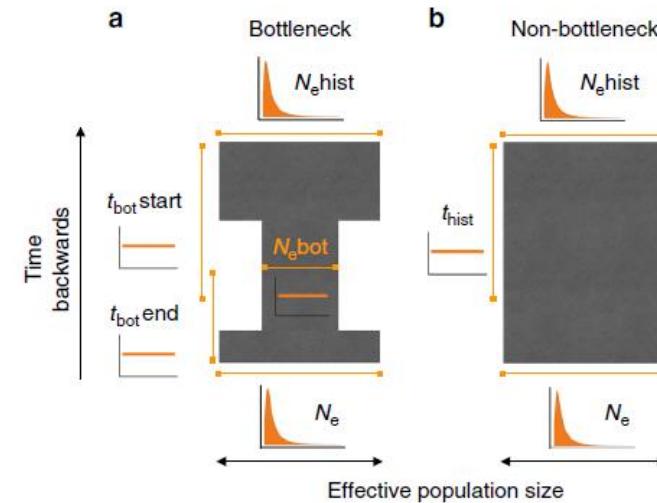
# Coalescent based estimate of $N_e$



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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?

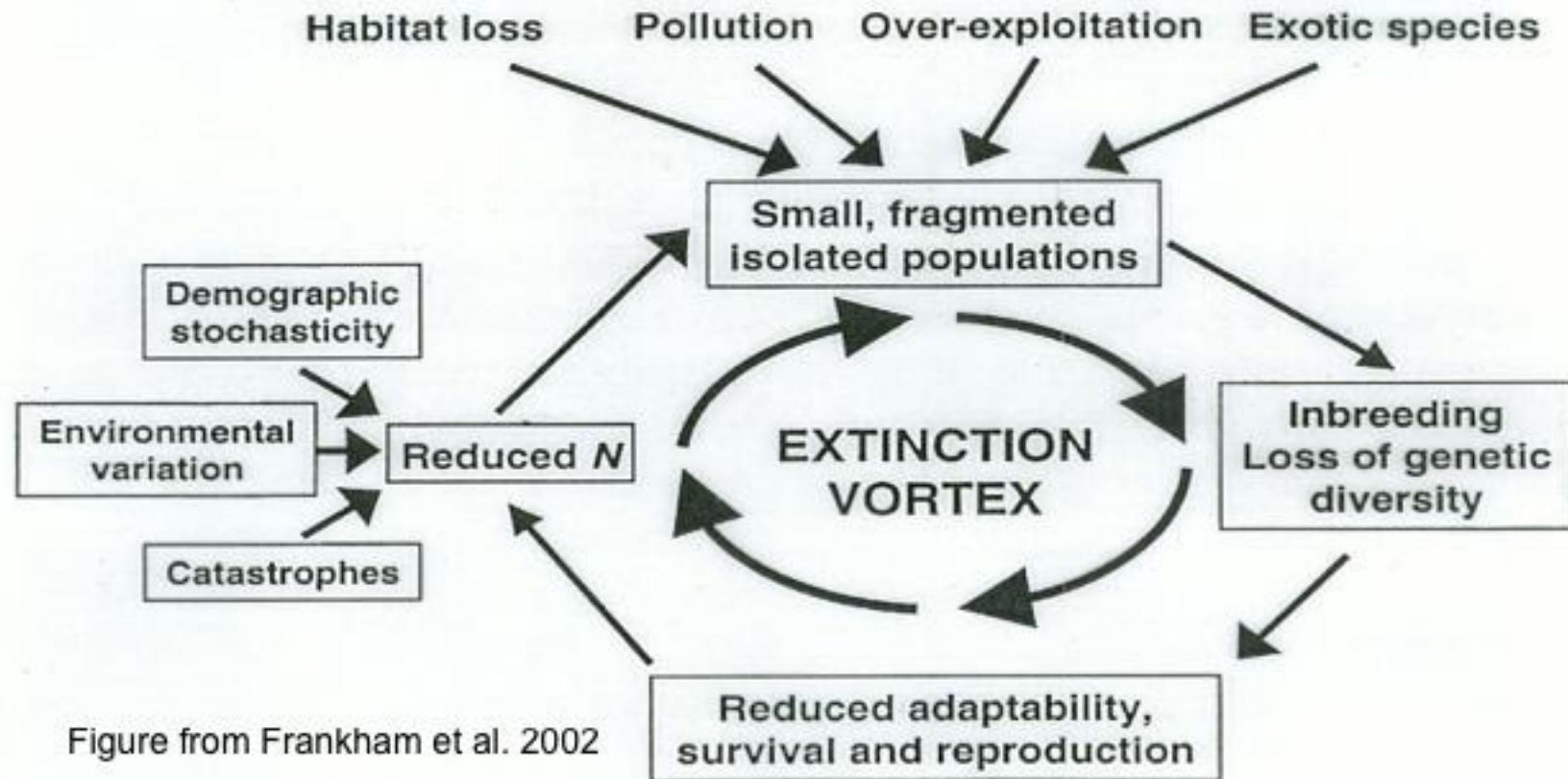


Figure from Frankham et al. 2002

# 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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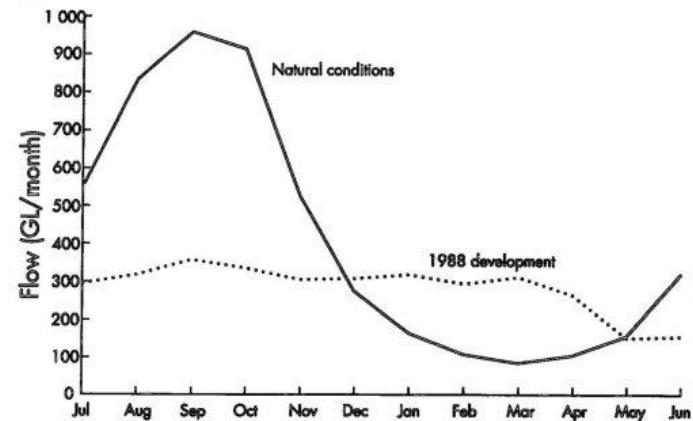
Water diversion and river regulation  
for agricultural/commercial uses



Disappearance of drought  
refuges and climate change



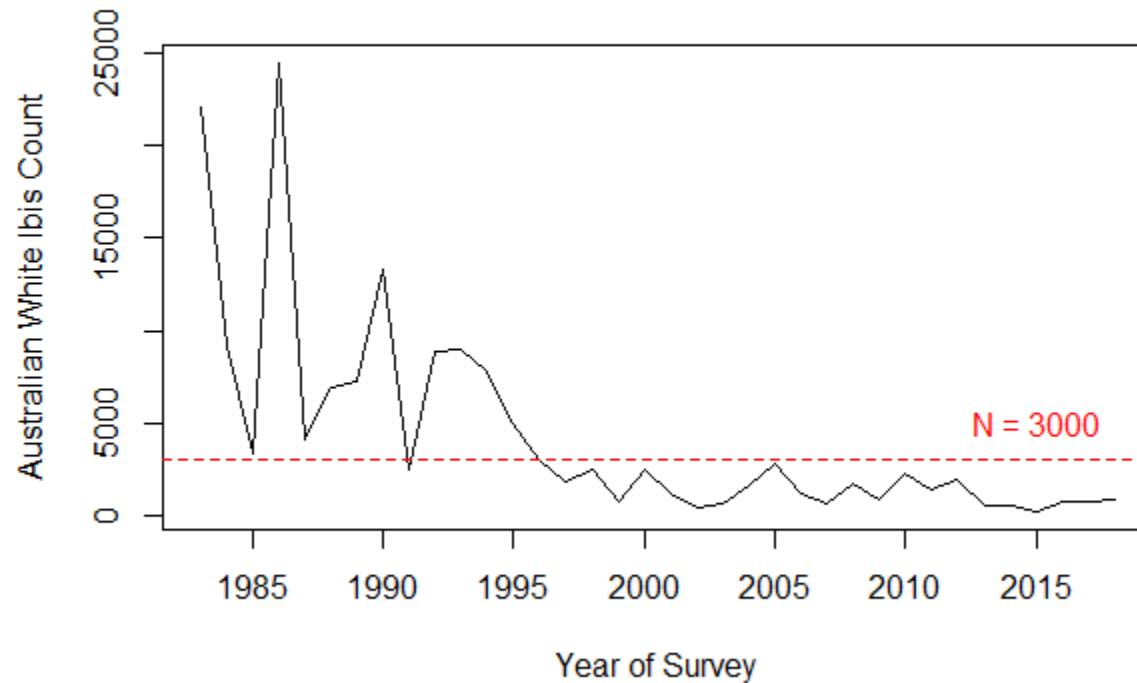
Declines in water quality due to  
pollution/land use changes



Altered flow regimes and  
flooding onset times/duration

# Inland Colonies of the Ibis have Declined Drastically

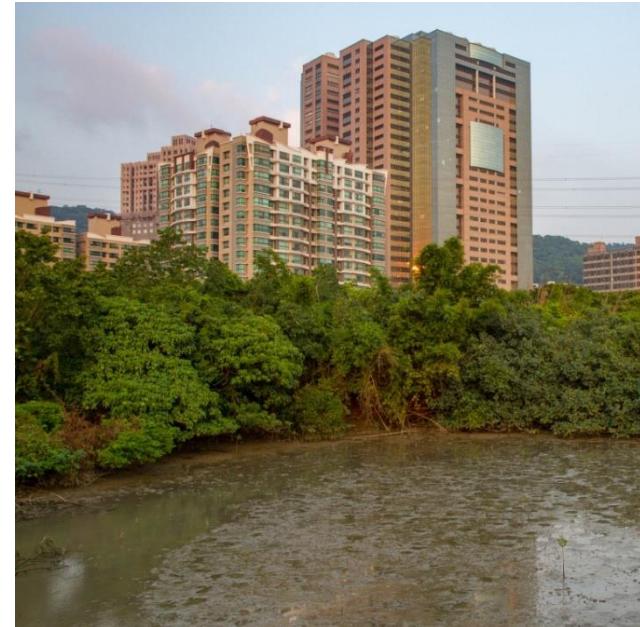
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**Source: Australian Aerial Waterbird Survey Data**



# 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

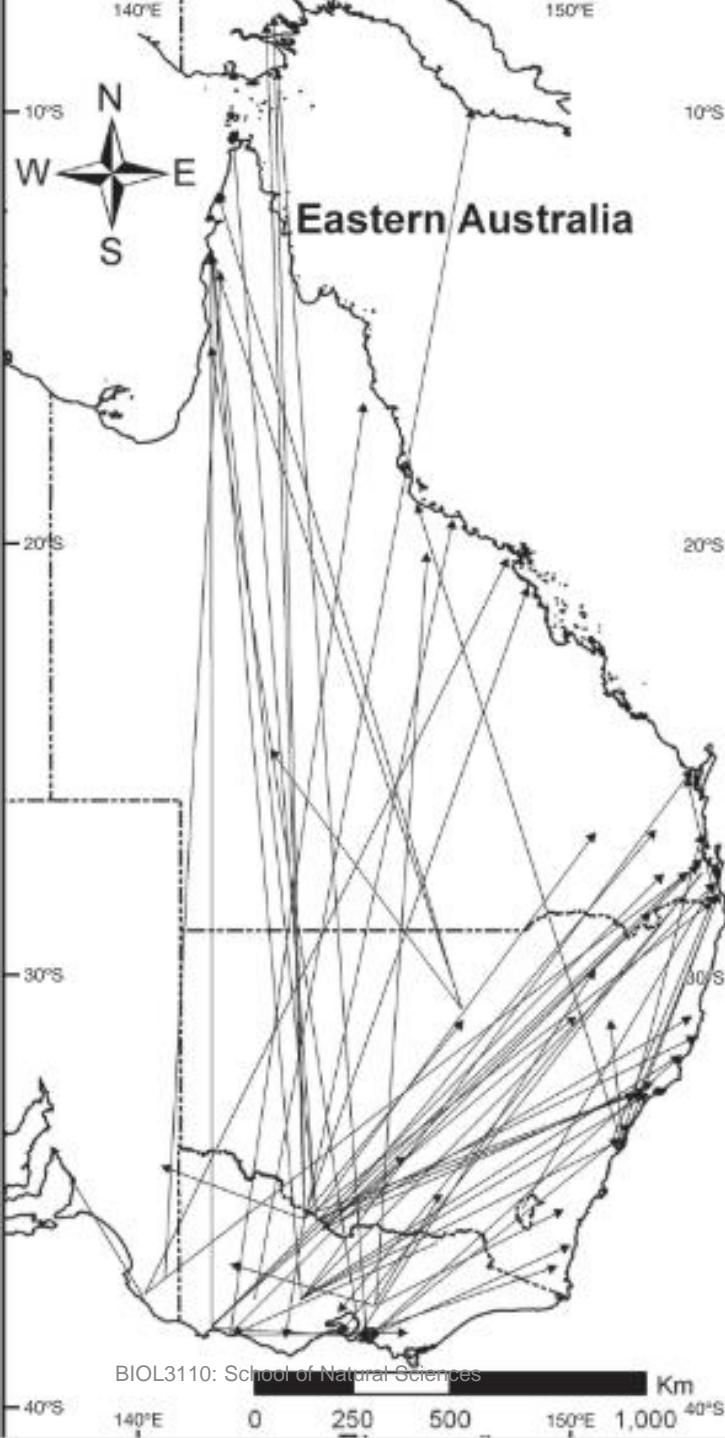


**Regular**  
↑ Permanent water  
↑ Resources

↑ Breeding opportunities  
↑ Abundance  
↑ Sedentariness



**GDR**



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

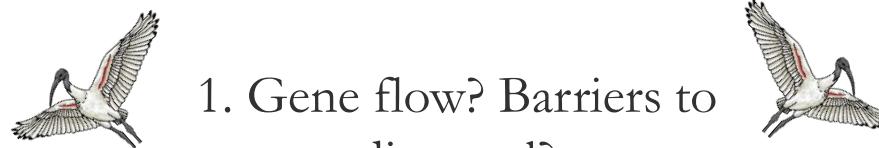
## INLAND



### 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



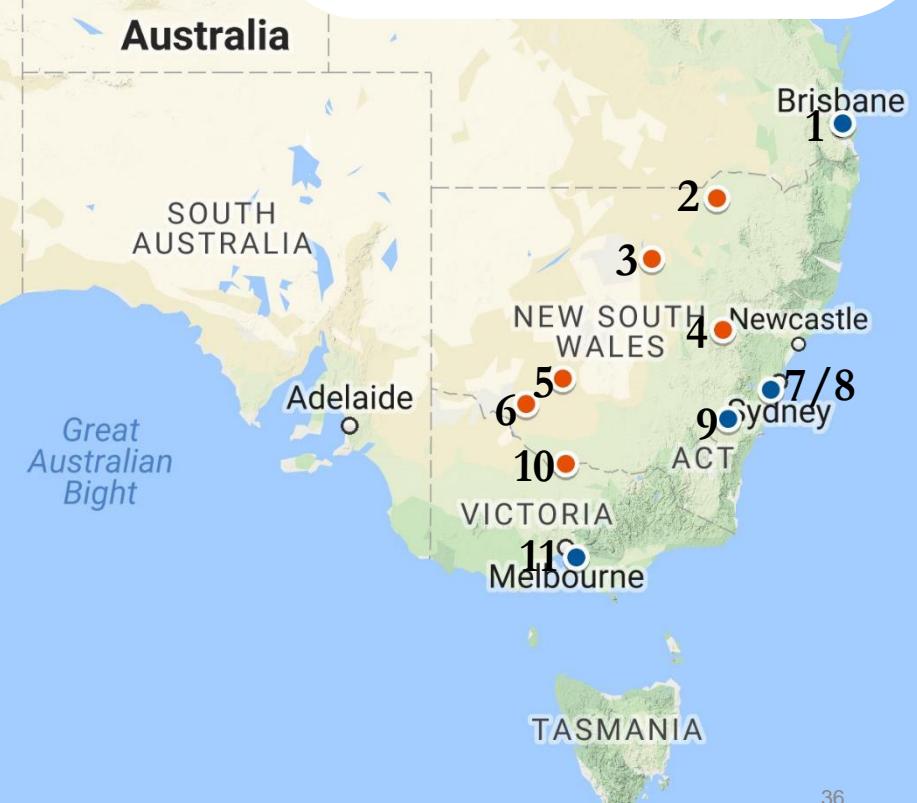
## METHODS and RESULTS

# 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

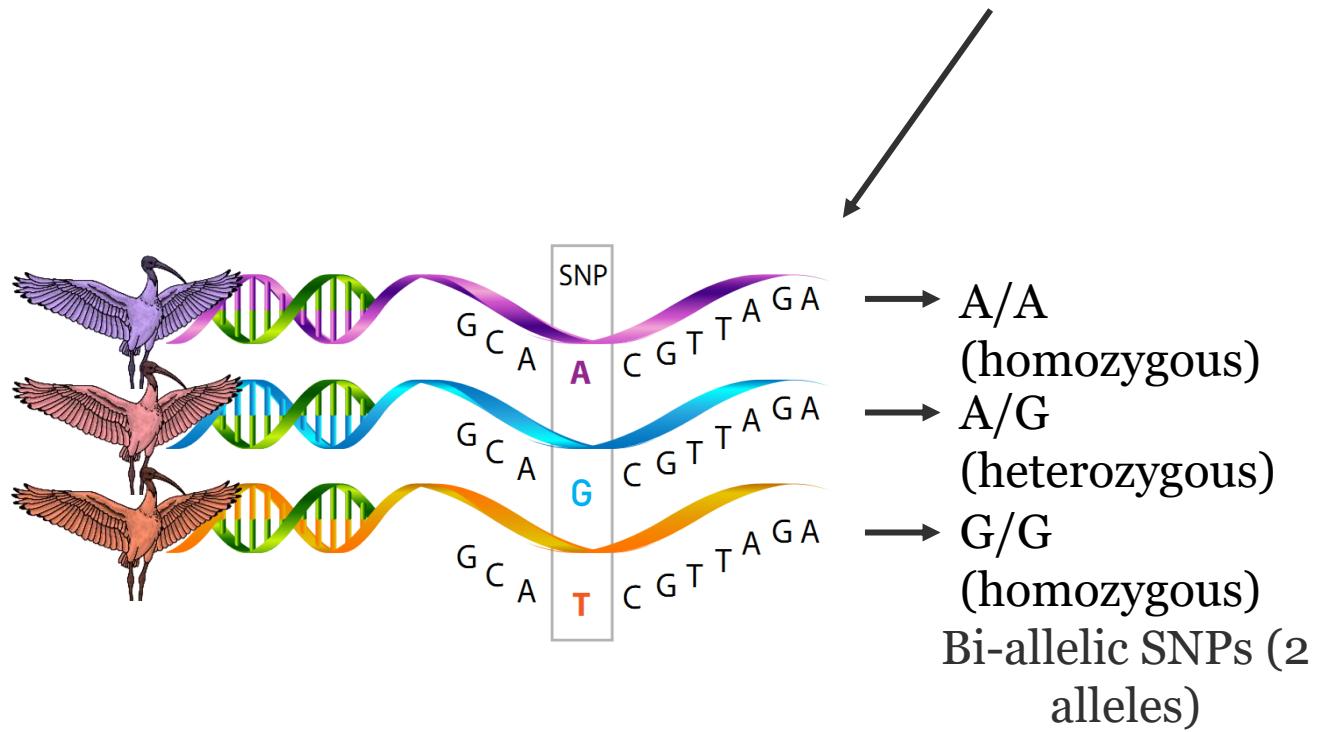
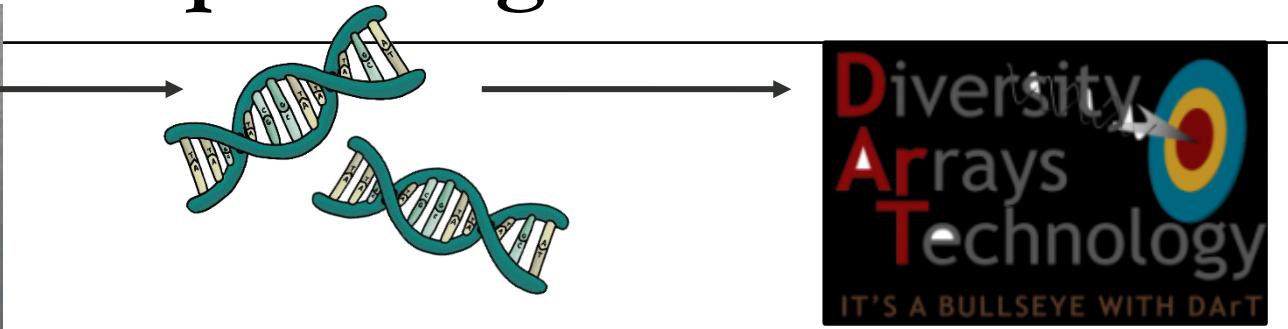
- ANSTO, University of NSW
- Australia wide citizen science project
- Feathers analysed using high resolution x-ray fluorescence
  - Changes in chemical composition over time
  - Different isotopic signatures for different wetlands
- Track waterbird movement over entire lifetime



# DNA Extraction and Sequencing



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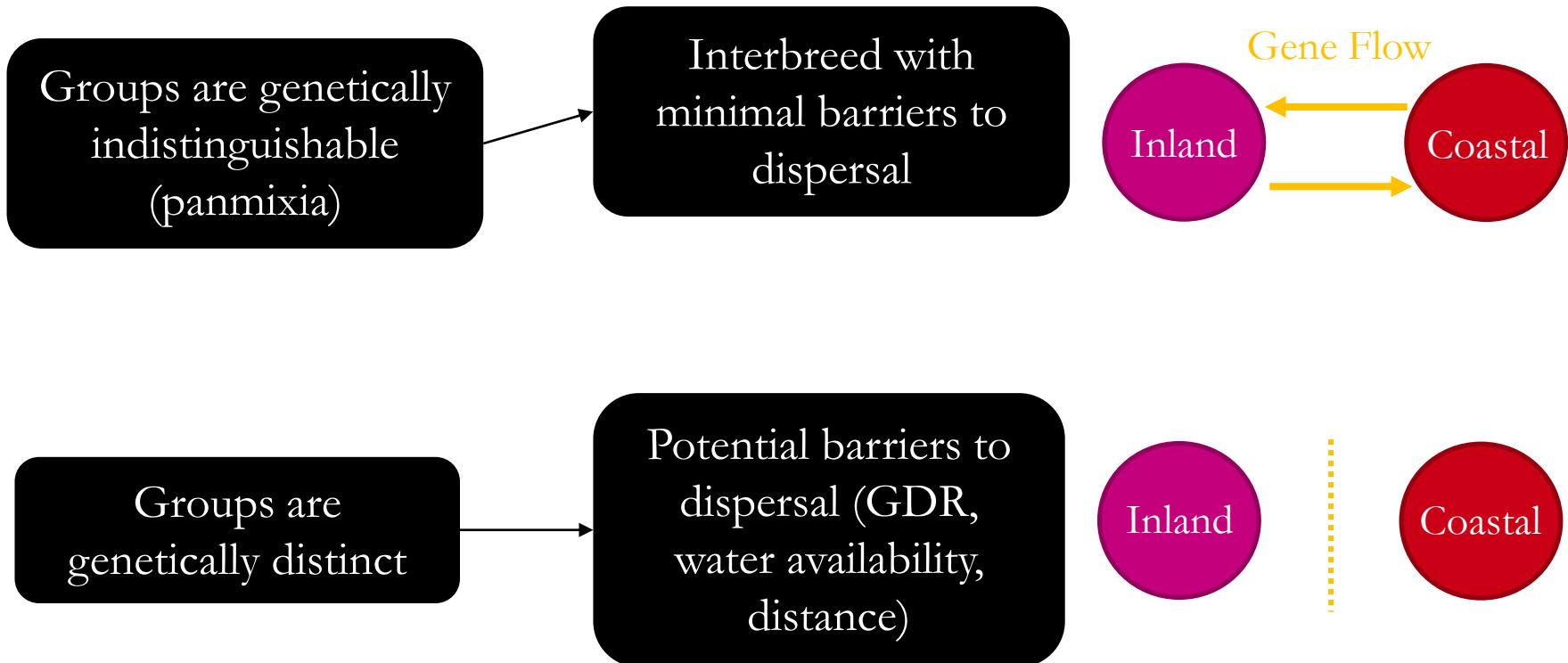


# Methods



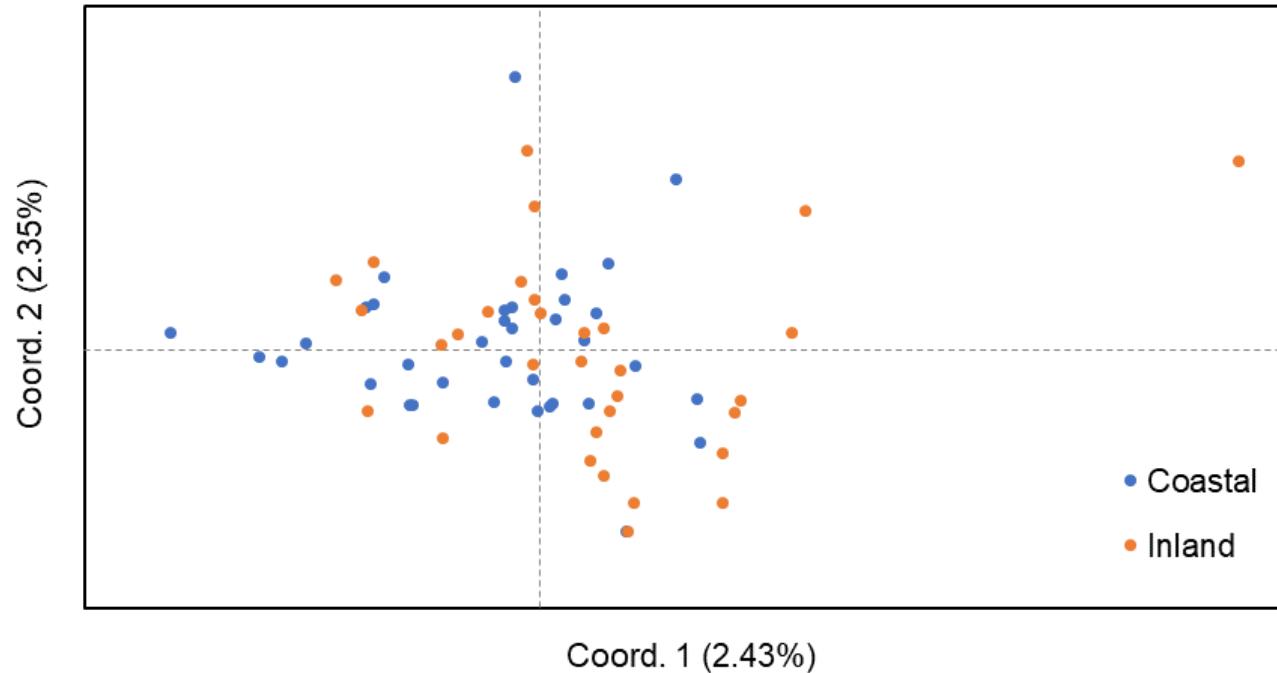
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## Are there barriers to gene flow?

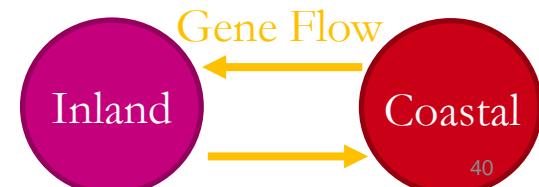


# Results

## Inland and Coastal colonies are not genetically distinct

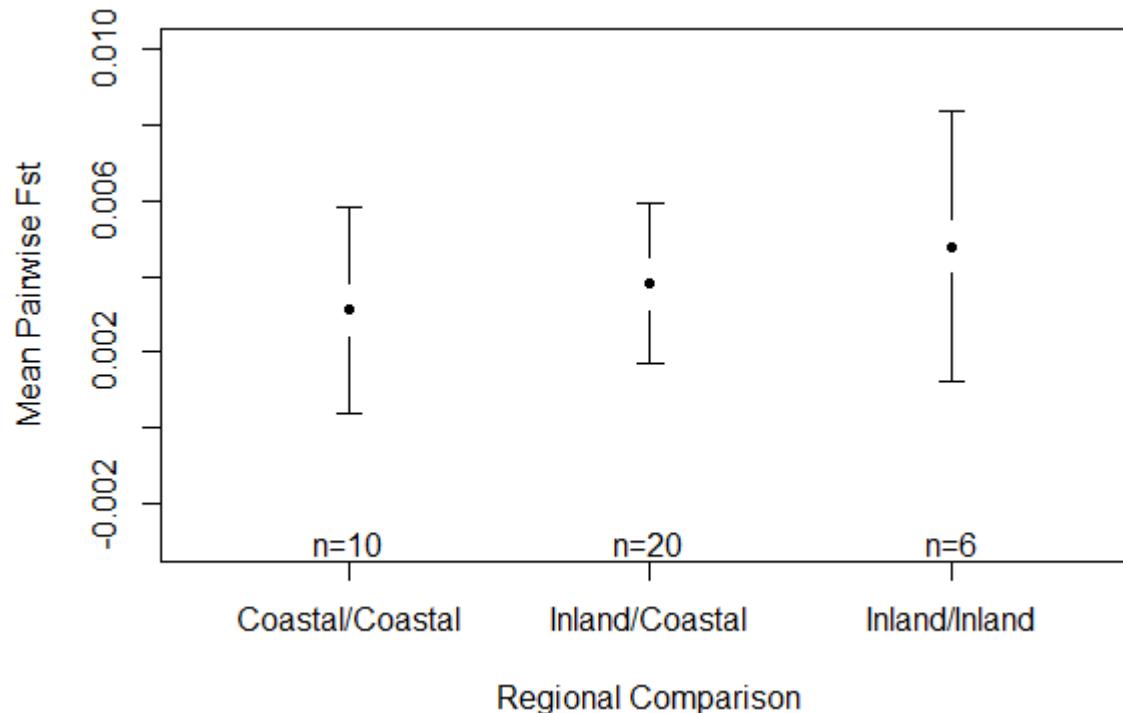


**Figure 1:** Plot of the 1<sup>st</sup> and 2<sup>nd</sup> principle coordinates identified by a principle coordinates analysis (PCoA)



# Results

## The GDR isn't a barrier to gene flow

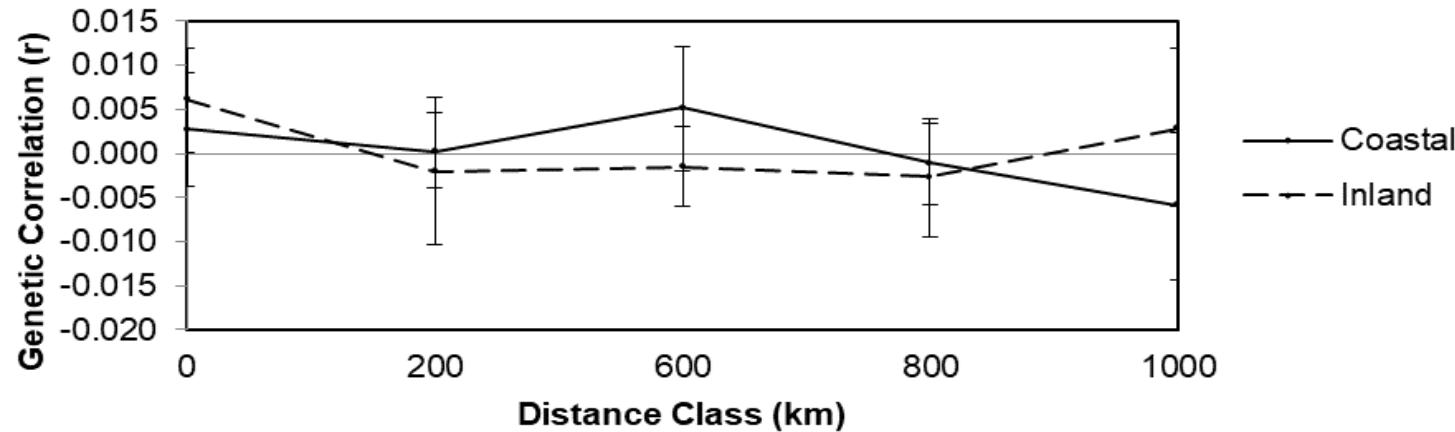


**Figure 3:** Mean pairwise  $F_{ST}$  grouped by region.

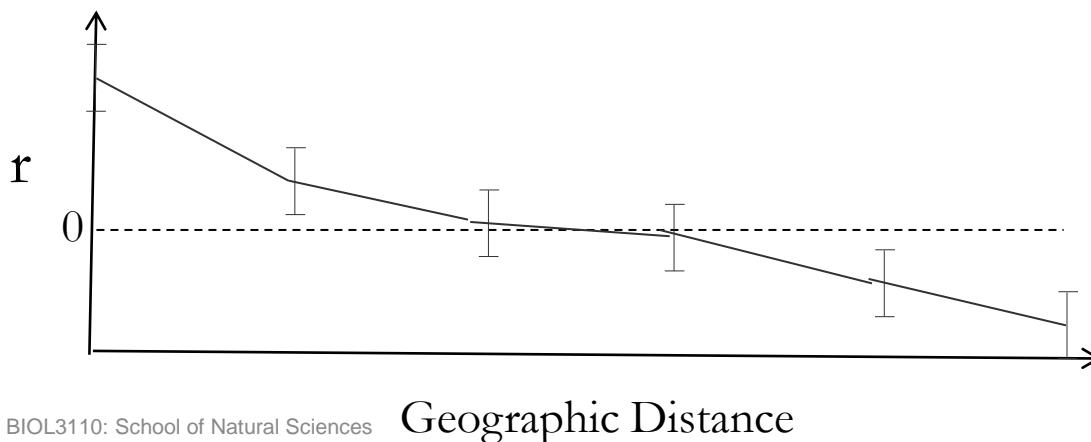


# Results

## Relatedness remains similar over large distances



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

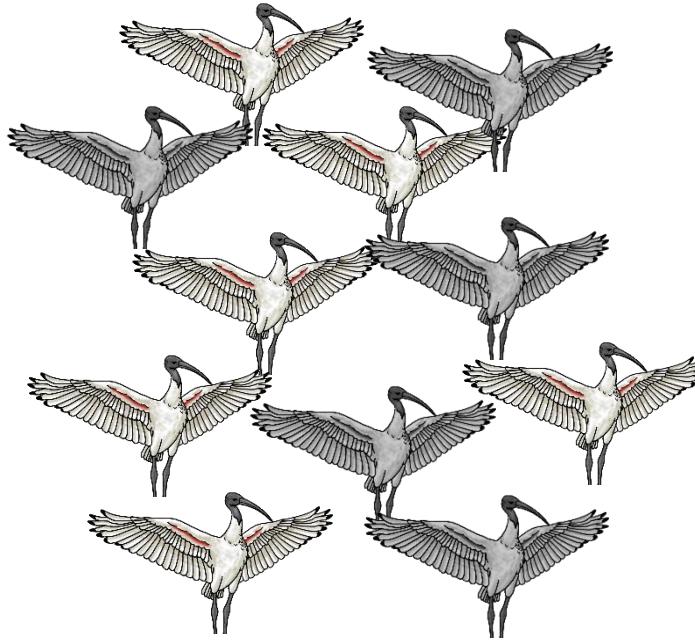


# Methods

## 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



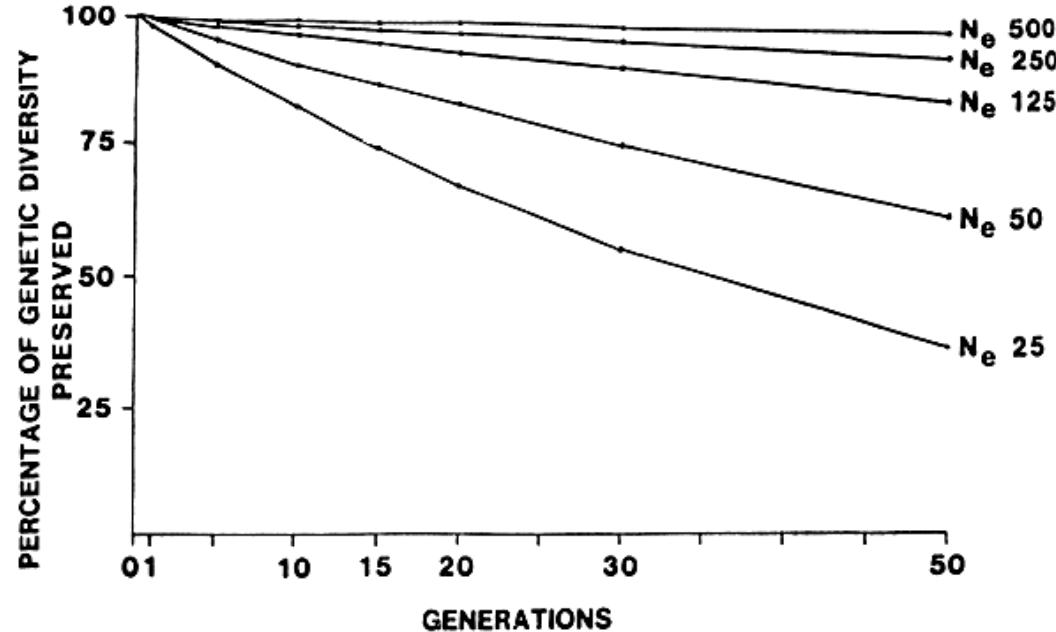
## Large effective size

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- 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

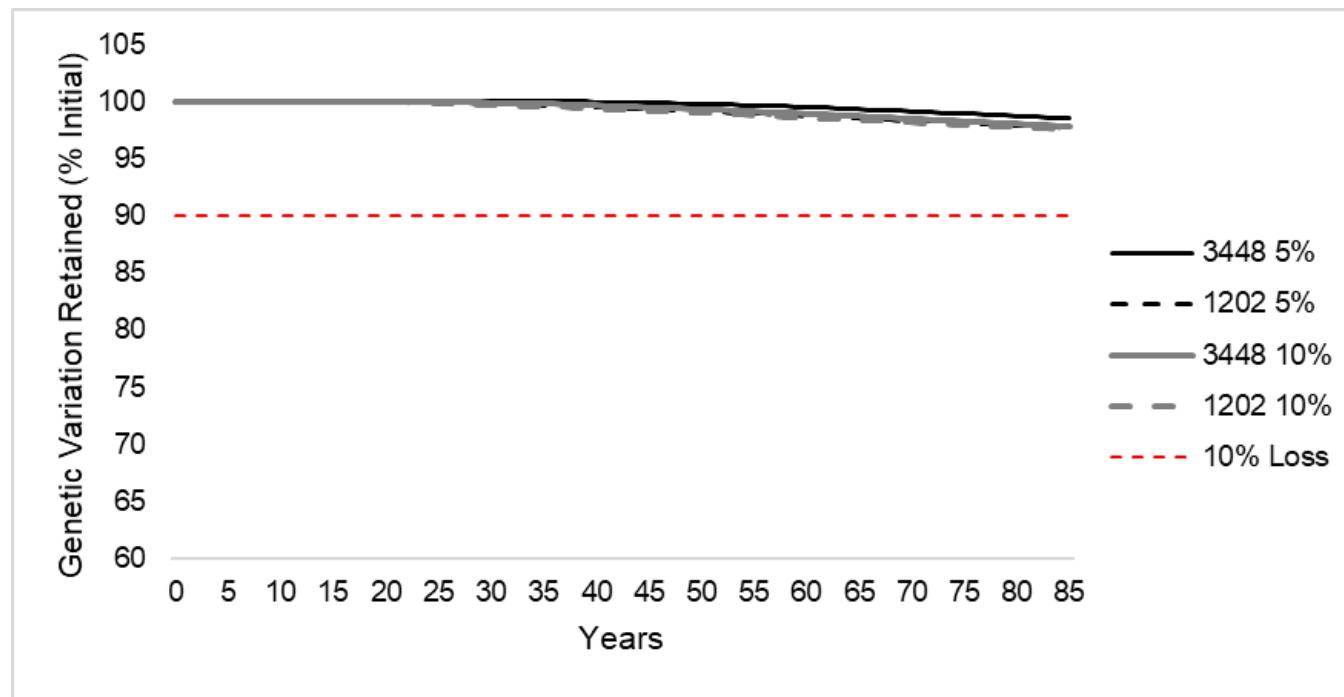
# Methods

## 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



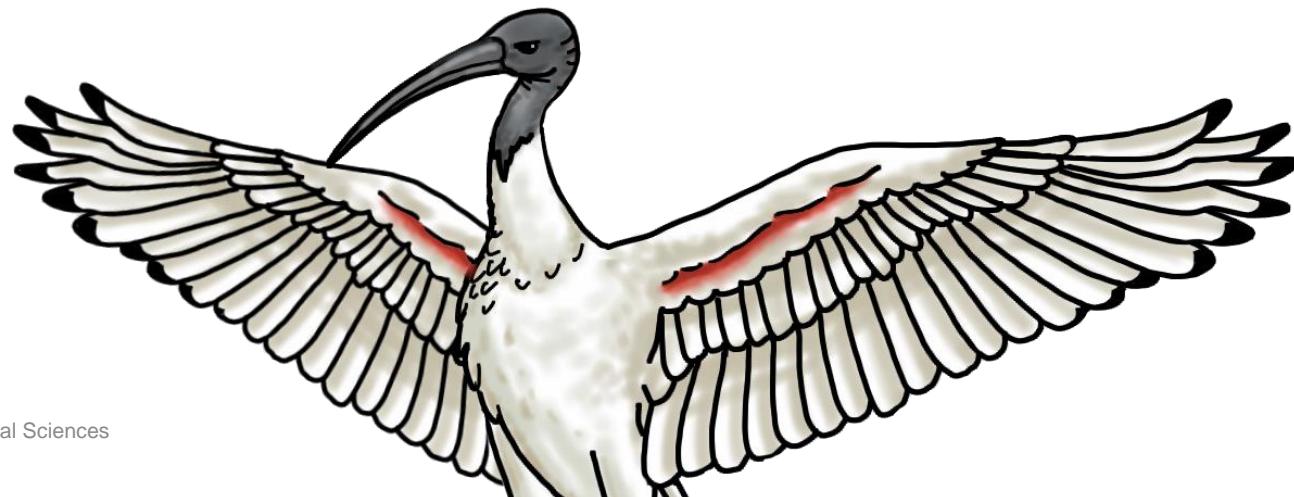
**Figure 4:** Retained diversity after 85 years, sustaining effective size at 650



## 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)

