#### **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

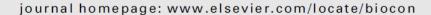


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#### **Biological Conservation**





#### Perspective

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



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## 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



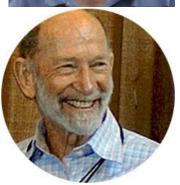


## How large must population be to retain their genetic health?

## How large must populations be to retain genetic 'health'? 50/500

Goal	$N_{ m e}$	References
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<sub>e</sub> = 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<sub>e</sub> ~ 70 required to avoid 10% ID (Caballero et al. 2016)

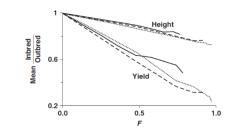
#### Likely an underestimate

- 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%</li>



- 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

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

#### What are $N_{\rm e}$ in practice?

#### What are $N_e$ of TH sp in practice?

- Captive pops of TH sp
   Av N<sub>e</sub> ~ 33
- Wild TH sp: IUCN criterion D
  - CE *N*<sub>e</sub> ≤ 8
  - EN  $N_{\rm e} \le 39$
  - VU  $N_{\rm e} \le 156$
- 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 <sub>e</sub>
USA 475 sp (vert, inv & 17 delisted sp	pl) 2,400 2,360	~ 240 ~ 236









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

#### Retaining evolutionary potential

(Franklin (1980)

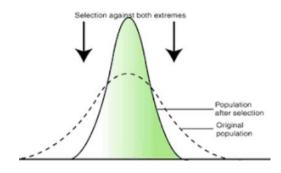
#### Assumptions:

- 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)

$$\Delta V_{A} = V_{m} - V_{A} / (2N_{e}) = 0$$

$$N_{\rm e} = V_{\rm A} / 2 V_{\rm m}$$

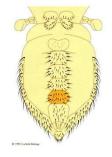
Substituting  $V_{\rm m} \sim 10^{-3} V_{\rm E}$  per generation

$$N_{\rm e} = V_{\rm A} / [2 \times 10^{-3} \times V_{\rm E}] = 500 V_{\rm A} / V_{\rm E}$$

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

$$N_{\rm e} = 500$$





### What has changed since 1980: evolutionary potential?

Adjusting for 90% harmful mutations (Lande 1995)

$$V_{\rm m} = 10^{-4} V_{\rm E}$$
 (~ plant fitness in wild)

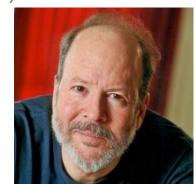
$$N_{\rm e} = V_{\rm A} / [2 \times 10^{-4} \times V_{\rm E}] = 5000 V_{\rm A} / V_{\rm E}$$

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

$$N_{\rm e} = 5,000$$

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

$$N_{\rm e} = 1,250$$



### What has changed since 1980? evolutionary potential

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

 $N_{\rm e} = 500$  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

(Falconer & Mackay 1996; Bataillon & Kirkpatrick 2000)

Lethals

$$N_e \ge 1$$
K

(Nei 1968; Hedrick 2002)

Balancing selection

Heterozygote advantage N<sub>e</sub> ≥ 1K

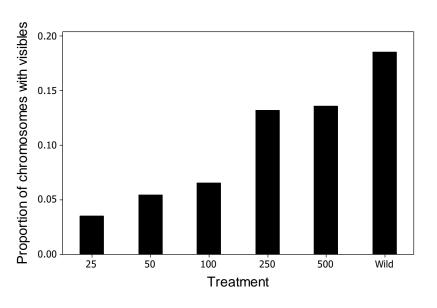
(Robertson 1962)

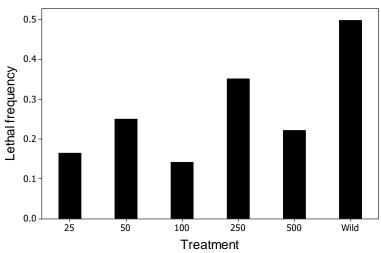
Frequency dependent selection  $N_e \ge 5K$ 

(Roff 1998)

 $N_{\rm e} = 500$  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.  $N_e \ge 1000$  required

N<sub>e</sub> 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

- $N_{\rm e} = 10^5 10^6$  to retain
- No thr sp this large, nor are many nonthr sp (including humans)

#### What happens if $N_{\rm e}$ < 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 regenerating GD by mutation?

(Lande & Barrowclough 1987)

GD	Regeneration (G)	
Quantitative confirmed in empirical s	10 <sup>2</sup> -10 <sup>3</sup> studies	
Single-locus	10 <sup>5</sup> -10 <sup>7</sup>	

## 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

- $N_{\rm e}$  < 12 (Charlesworth et al. 93)
- $N_{\rm e}$  < 100 (Lynch et al. 95)
- $N_{\rm e}$  < 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  $N_{\rm e}$  250 (Zeyl et al 2001)
- Nematodes  $N_e = 1 \text{ lost } <1\% \text{ fitness/G}$

(Vassilieva et al. 2000; Estes et al. 2004)



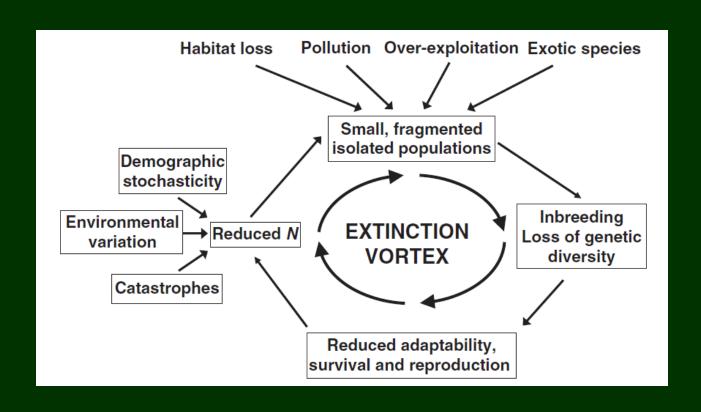
Appears to be a minor threat << ID</li>







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



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

Threat	<b>N</b> <sub>e</sub>	N			
Theory (Frankham et al. 2014; Nunney & Campbell 1993)					
Retaining QGV	1K	~10K			
Demographic stochastic	city	10s – 100			
Environmental stochast	icity	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 4	0 G 4.2K			
Primates in Sunda Islan	ds	>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 thr species need to be to meet this target?

Aim: Retain 90% of genetic diversity for 100 yrs

Required  $N_{\rm e}$  depends on generation length (L)

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

let t = 100 / L

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

take In & rearrange

$$N_0 \sim 475 / L$$

#### Endangered species in captivity

$$N_{\rm e} \sim 475 / L$$

#### **Examples:**

Elephant L = 35

$$N_{\rm e} = 475/35 = 14$$

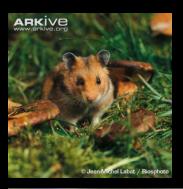
White-footed mouse L = 0.27

$$N_{\rm e} = 475/0.27 = 1759$$





## Fallacy of small wild isolated surviving populations

















### Fallacy of small wild surviving populations

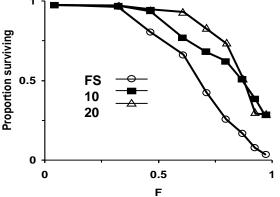
- Small/bottlenecked surviving populations
  - Mauritius kestrel, golden hamster, Catham Is black robin, Seychelles warbler, Mauritius pink pigeon, Socorro Is red-tailed hawk, N elephant seal, California Islands foxes, Chillingham cattle
- Fallacy to argue from a few & generalise
  - 'Grandad smoked 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 ~ 1) populations of mice, guinea pigs, Drosophila & plants persist

All surviving ones have low fitness when

tested



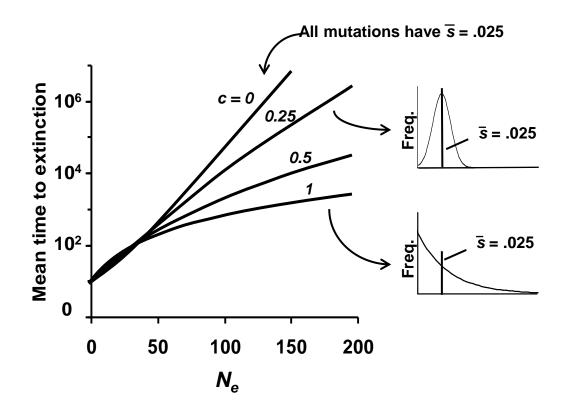
### Fallacy of small wild surviving populations

- 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 reestablished? (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<sub>e</sub> ≥ 100
- To permanently retain evol potential requires
   N<sub>e</sub> ≥ 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?



### Translating from $N_{\rm e}$ to N

(Frankham 2021)

 $N_{\rm e}/N$  ratios from meta-analyses

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

 $N \sim 10 \times N_e$