Introductory Models, Effective Population Size

Models

- Intentional simplification of complex relationships
  - Eliminate extraneous detail, focus on key parameters
  - Appropriate and useful first approximations
- Evaluate fit of data to model
  - Poor fit may implicate violation of model assumptions
  - Refining of models tells us which parameters most important
- Population genetics relies heavily on mathematical models
  - Specify the mathematical relationships among parameters that characterize a population

Random Mating

- One of the most important models in population genetics
- Frequency of mating pairs determined by genotype frequencies

Male Genotype Frequency	Female Genotype Frequency		
	A_1A_1 (P_F)	A_1A_2 (H_F)	A_2A_2 (Q_F)
A_1A_1 (P_M)			
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A_1A_1 (P_M)	P_MP_F	P_MH_F	P_MQ_F
A_1A_2 (H_M)	H_MP_F	H_MH_F	H_MQ_F
A_2A_2 (Q_M)	Q_MP_F	Q_MH_F	Q_MQ_F

Random Mating

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- Also called ‘panmictic’ model

Non-overlapping Generations

Birth
Reproduction
Death

→

Birth
Reproduction
Death

→

Birth
Reproduction
Death

Generation t -2 | Generation t -1 | Generation t

Hardy-Weinberg Model

- Both models convenient first approximations for complex populations
- What happens when we combine them?
- What are consequences of random mating in a non-overlapping generation model?

Godfrey Harold Hardy

Wilhelm Weinberg

HW Model Assumptions

- Discrete generations
- Random mating
- Sexual reproduction
- Diploid
- Bi-allelic locus
- Allele frequencies equal in males, females
- Large population size
- No migration
- No mutation
- No selection

Hardy-Weinberg Principle

- One of first major principles in population genetics
- Describes relationship between genotype frequency and allele frequency
  - Equilibrium state
- Autosomal locus will alleles A, a
  - Frequencies of A, a: p, q
- Genotypes AA, Aa, aa

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Hardy-Weinberg Principle

Frequency(A) = p
Frequency(a) = q

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Mating
AA x AA
AA x Aa
AA x aa
Aa x Aa
Aa x aa
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$$p' =$$

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$$p' = P' + \frac{1}{2}H'$$

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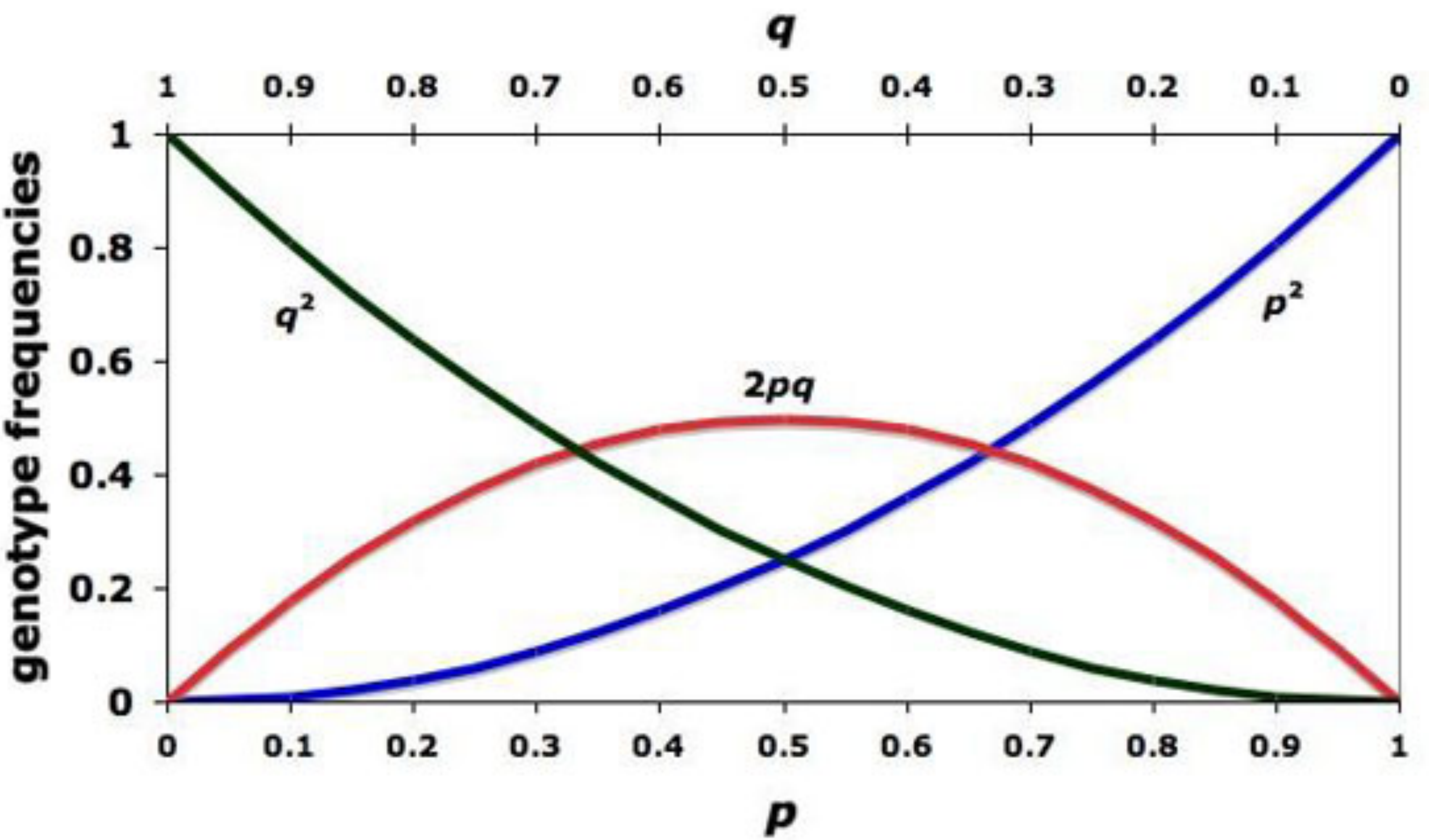
$$q' = Q' + \frac{1}{2}H' = q^2 + \frac{1}{2}2pq = q(q+p) = q$$

- Allele frequency unchanged across generations
  - Mendelian inheritance itself preserves variation
- HWE achieved in ONE generation
  - Equal allele frequencies in males & females, discrete generations

HWE Genotype Frequencies



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  - HW frequencies: p^2, $2pq$, q^2
- Once at HWE, allele & genotype freq constant

Random Genetic Drift

- Hardy-Weinberg equilibrium predicts:
  - 1) Allele frequencies remain constant
  - 2) Genotype frequencies predicted by allele frequencies
- HW model assumes infinite population size
- With finite population size, allele frequencies change over time due to sampling
- Random genetic drift: stochastic change in allele frequencies due to finite sampling of gametes

Random Genetic Drift

- Haploid population of size N
- Two alleles: A, a
- At generation t
  - Frequency of A is p
- Frequency of a is $q = (1-p)$

What is frequency of A at generation t + 1?

Probability(X) =

ways X can happen/Total Possible Outcomes

1. You roll a die. What is the probability that you roll a 4?

2. You roll a die. What is the probability that you roll an even number?

3. You roll a die twice. What is the probability that you roll a 4 twice?

4. You flip a fair coin. What is the probability you flip a head?

5. You flip a fair coin twice. What is the probability you flip a head twice?

6. You flip a fair coin and it flips heads. What is the probability if you flip it again, it will flip heads?

7. You flip a fair coin twice. What is the probability you flip at least one head?

8. A bag of marbles contains 36 blue marbles and 14 red marbles. If you pull out one marble, what is the probability that it is blue?

9. A bag of marbles contains 36 blue marbles and 14 red marbles. If you pull out one marble, what is the probability that it is NOT blue?

10. A population contains 36 AA individuals and 14 aa individuals. If you pull out genotype at random, what is the probability that it is AA?

11. A randomly mating population contains 36 AA individuals, 50 Aa individuals and 14 aa individuals. These individuals each produce an infinite number of gametes. If you reach into the gamete pool and pull out a single allele, what is the probability that it is A?

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- Randomly select 1 individual to be parent

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  - Pr(0A) =

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- Randomly select 1 individual to be parent
  - Pr(1A) = p
  - Pr(0A) = $(1 – p)$

Random Genetic Drift

In generation t, freq(A) = p, freq(a) = (1-p)

- Randomly select 1 individual to be parent
  - Pr(1A) = *p*
  - Pr(0A) = (1 – *p*)
- Randomly select 2 individuals to be parents

Random Genetic Drift

In generation t, freq(A) = p, freq(a) = (1-p)

- Randomly select 1 individual to be parent
  - Pr(1A) = p
  - Pr(0A) = $(1 – p)$
- Randomly select 2 individuals to be parents
  - Pr(2A) =

Random Genetic Drift

In generation t, freq(A) = p, freq(a) = (1-p)

- Randomly select 1 individual to be parent
  - Pr(1A) = p
  - Pr(0A) = $(1 - p)$
- Randomly select 2 individuals to be parents
  - Pr(2A) = p^2

Random Genetic Drift

In generation t, freq(A) = p, freq(a) = (1-p)

- Randomly select 1 individual to be parent
  - Pr(1A) = p
  - Pr(0A) = $(1 - p)$
- Randomly select 2 individuals to be parents
  - Pr(2A) = p^2
  - Pr(0A) =

Random Genetic Drift

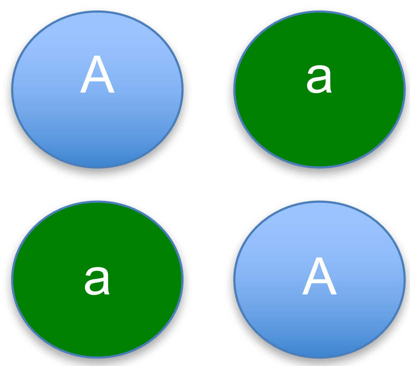
In generation t, freq(A) = p, freq(a) = (1-p)

- Randomly select 1 individual to be parent
  - Pr(1A) = p
  - Pr(0A) = $(1 – p)$
- Randomly select 2 individuals to be parents
  - Pr(2A) = p^2
  - Pr(0A) = $(1 – p)^2$

Random Genetic Drift

In generation t, freq(A) = p, freq(a) = (1-p)

- Randomly select 1 individual to be parent
  - Pr(1A) = p
  - Pr(0A) = $(1 - p)$
- Randomly select 2 individuals to be parents
  - Pr(2A) = p^2
  - Pr(1A) =
  - Pr(0A) = $(1 - p)^2$



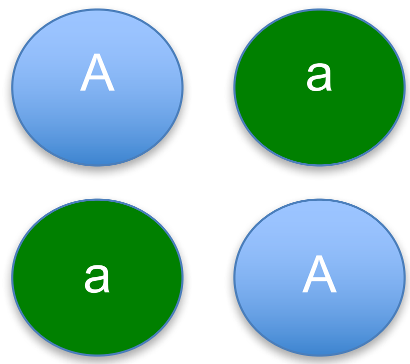
Pr = $p(1 - p)$

Pr = $(1 - p)p$

Random Genetic Drift

In generation t, freq(A) = p, freq(a) = (1-p)

- Randomly select 1 individual to be parent
  - – $Pr(1A) = p$
  - – $Pr(0A) = (1 - p)$
- Randomly select 2 individuals to be parents
  - – $Pr(2A) = p^2$
  - – $Pr(1A) = 2p(1-p)$
  - – $Pr(0A) = (1 - p)^2$



Pr = p(1 – p)

Pr = (1 – p)p

Random Genetic Drift

In generation t, freq(A) = p, freq(a) = (1-p)

- Randomly select 1 individual to be parent
  - $\Pr(1A) = p$
  - $\Pr(0A) = (1 - p)$
- Randomly select 2 individuals to be parents
  - $\Pr(2A) = p^2$
  - $\Pr(1A) = 2p(1-p)$
  - $\Pr(0A) = (1 - p)^2$
- Randomly select 3 individuals to be parents

Random Genetic Drift

In generation t, freq(A) = p, freq(a) = (1-p)

- Randomly select 1 individual to be parent
  - Pr(1A) = p
  - Pr(0A) = $(1 – p)$
- Randomly select 2 individuals to be parents
  - Pr(2A) = p^2
  - Pr(1A) = $2p(1-p)$
  - Pr(0A) = $(1 – p)^2$
- Randomly select 3 individuals to be parents
  - Pr(3A) =
  - Pr(2A) =
  - Pr(1A) =
  - Pr(0A) =

Random Genetic Drift

In generation t, freq(A) = p, freq(a) = (1-p)

- Randomly select 1 individual to be parent
  - $Pr(1A) = p$
  - $Pr(0A) = (1 - p)$
- Randomly select 2 individuals to be parents
  - $Pr(2A) = p^2$
  - $Pr(1A) = 2p(1-p)$
  - $Pr(0A) = (1 - p)^2$
- Randomly select 3 individuals to be parents
  - $Pr(3A) = p^3$
  - $Pr(2A) = 3p^2(1 - p)$
  - $Pr(1A) = 3p(1 - p)^2$
  - $Pr(0A) = (1 - p)^3$

Random Genetic Drift

In generation t, freq(A) = p, freq(a) = (1-p)

- Randomly select N individuals to be parents

$$\Pr(j\ alleles\ of\ type\ A) = \binom{N}{j} p^{j}(1-p)^{N-j}$$

What is frequency of A at generation t + 1?

Random Genetic Drift

Frequency of A	Generation(t+1)
0/N	$\binom{N}{0}(p_t)^0(1-p_t)^{N-0}$
1/N	$\binom{N}{1}(p_t)^1(1-p_t)^{N-1}$
2/N	$\binom{N}{2}(p_t)^2(1-p_t)^{N-2}$
.	
.	
.	
N/N	$\binom{N}{N}(p_t)^N(1-p_t)^0$

Random Genetic Drift

Frequency of A	Generation(t+1)	Generation (t+2)
0/N	$\binom{N}{0}(p_t)^0(1-p_t)^{N-0}$	$\binom{N}{0}(p_{t+1})^0(1-p_{t+1})^{N-0}$
1/N	$\binom{N}{1}(p_t)^1(1-p_t)^{N-1}$	$\binom{N}{1}(p_{t+1})^1(1-p_{t+1})^{N-1}$
2/N	$\binom{N}{2}(p_t)^2(1-p_t)^{N-2}$	$\binom{N}{2}(p_{t+1})^2(1-p_{t+1})^{N-2}$
.		
.		
.		
N/N	$\binom{N}{N}(p_t)^N(1-p_t)^0$	$\binom{N}{N}(p_{t+1})^N(1-p_{t+1})^0$

Random Genetic Drift

Frequency of A	Generation(t+1)	Generation (t+2)	Generation(t+3)
0/N	$\binom{N}{0}(p_t)^0(1-p_t)^{N-0}$	$\binom{N}{0}(p_{t+1})^0(1-p_{t+1})^{N-0}$	$\binom{N}{0}(p_{t+2})^0(1-p_{t+2})^{N-0}$
1/N	$\binom{N}{1}(p_t)^1(1-p_t)^{N-1}$	$\binom{N}{1}(p_{t+1})^1(1-p_{t+1})^{N-1}$	$\binom{N}{1}(p_{t+2})^1(1-p_{t+2})^{N-1}$
2/N	$\binom{N}{2}(p_t)^2(1-p_t)^{N-2}$	$\binom{N}{2}(p_{t+1})^2(1-p_{t+1})^{N-2}$	$\binom{N}{2}(p_{t+2})^2(1-p_{t+2})^{N-2}$
.			
.			
.			
N/N	$\binom{N}{N}(p_t)^N(1-p_t)^0$	$\binom{N}{N}(p_{t+1})^N(1-p_{t+1})^0$	$\binom{N}{N}(p_{t+2})^N(1-p_{t+2})^0$

Transitions between states are random, but defined by a probability

Transitions have no memory beyond previous step

Random Genetic Drift

Frequency of A	Generation(t+1)	Generation (t+2)	Generation(t+3)
0/N	$\binom{N}{0}(p_t)^0(1-p_t)^{N-0}$	$\binom{N}{0}(p_{t+1})^0(1-p_{t+1})^{N-0}$	$\binom{N}{0}(p_{t+2})^0(1-p_{t+2})^{N-0}$
1/N	$\binom{N}{1}(p_t)^1(1-p_t)^{N-1}$	$\binom{N}{1}(p_{t+1})^1(1-p_{t+1})^{N-1}$	$\binom{N}{1}(p_{t+2})^1(1-p_{t+2})^{N-1}$
2/N	$\binom{N}{2}(p_t)^2(1-p_t)^{N-2}$	$\binom{N}{2}(p_{t+1})^2(1-p_{t+1})^{N-2}$	$\binom{N}{2}(p_{t+2})^2(1-p_{t+2})^{N-2}$
.			
.			
.			
N/N	$\binom{N}{N}(p_t)^N(1-p_t)^0$	$\binom{N}{N}(p_{t+1})^N(1-p_{t+1})^0$	$\binom{N}{N}(p_{t+2})^N(1-p_{t+2})^0$

Transitions between states are random, but defined by a probability

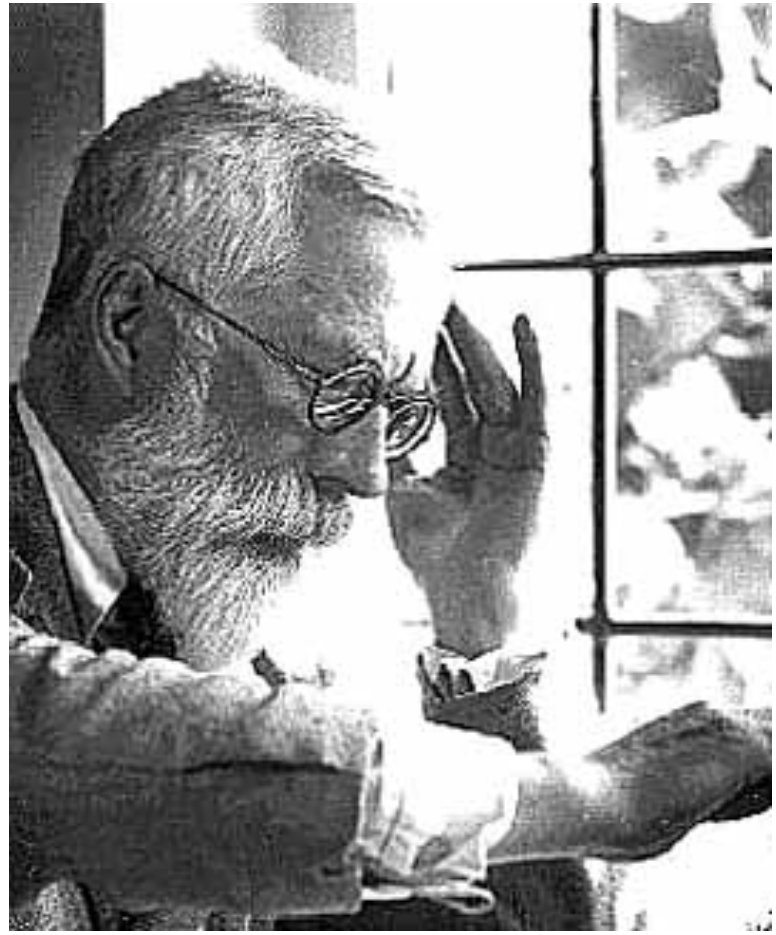
Transitions have no memory beyond previous step

Diploid Model

- N diploid individuals
  - 2N alleles in population
- Two alleles: A, a (frequencies *p*, *q*)
- Randomly draw 2N gametes to found next generation

$$\Pr(j\ alleles\ of\ type\ A) = \binom{2N}{j} p^{j}(1\text{-}p)2^{N\text{-}j}$$

Wright-Fisher Model



Wright-Fisher Model

- Assumptions:
  - N diploid organisms (2N alleles)
  - Infinite gametes
  - Discrete Generations
  - Random mating
  - No mutation
  - No selection

$$P_{ij} = \binom{2N}{j} \left(\frac{i}{2N}\right)^{j} \left(1 - \frac{i}{2N}\right)^{2N-j} = \binom{2N}{j} p^{j} q^{2N-j}$$

Time, Probability of fixation

- Alleles are eventually fixed or lost
- 2N alleles
  - Each equally likely to fix (selectively equivalent)
  - Pr(fixation) =

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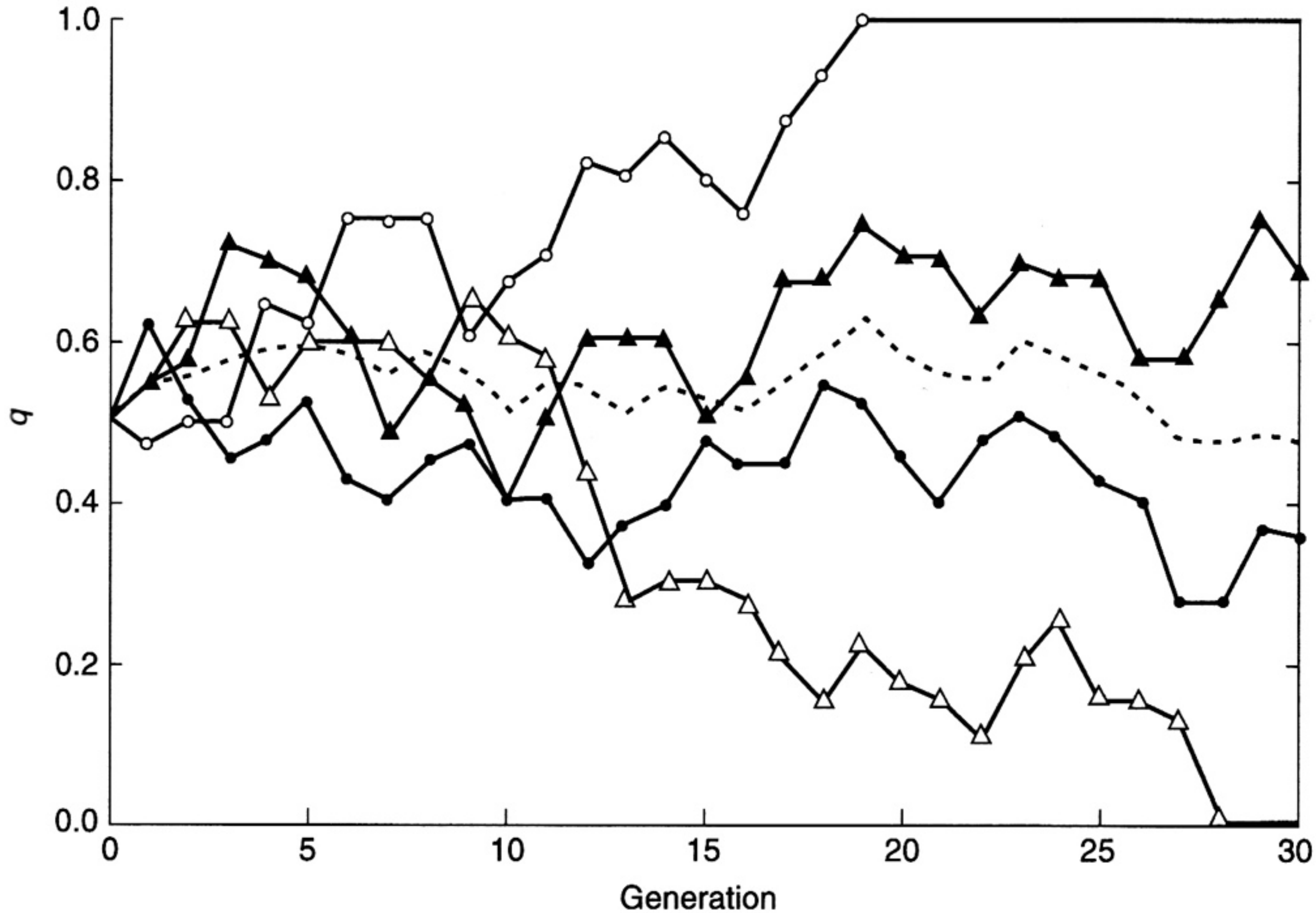
Time, Probability of fixation

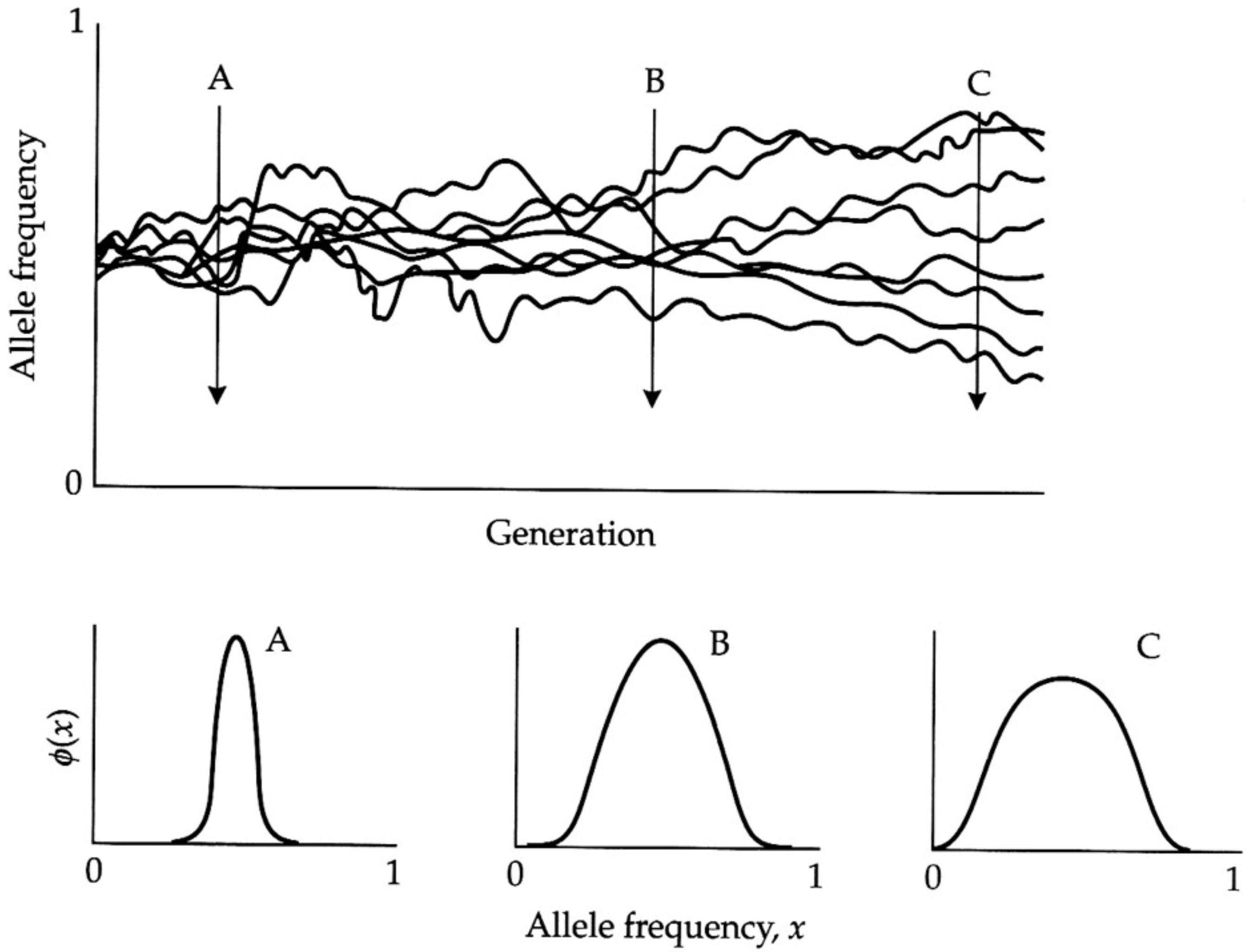
- Alleles are eventually fixed or lost
- 2N alleles
  - Each equally likely to fix (selectively equivalent)
  - Pr(fixation) = 1/2N
  - If *i* copies of allele, Pr(fixation) = *i*/2N
- Pr(fixation) = *p*

Probabilities of fixation, loss

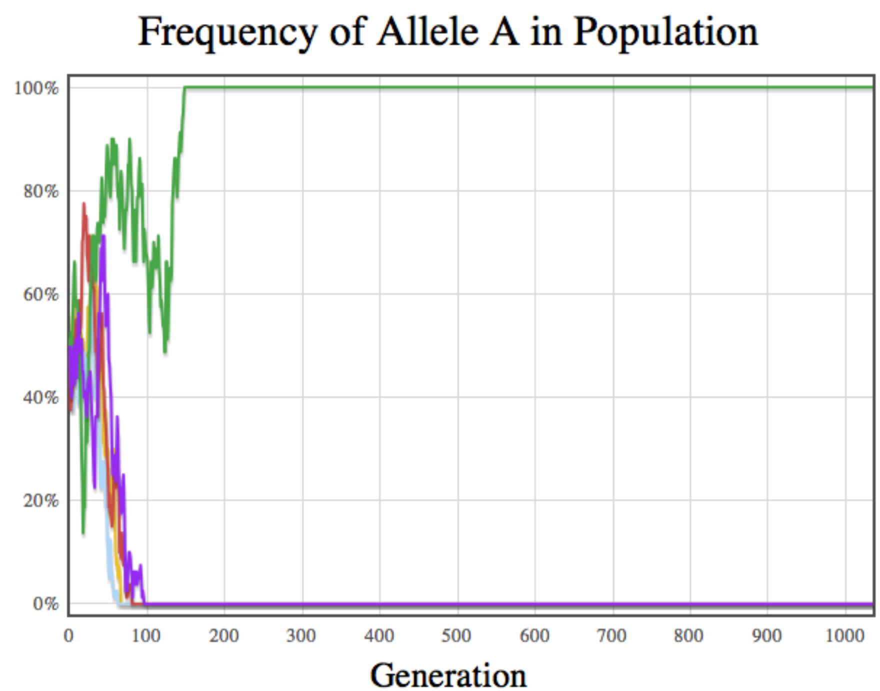
- Alleles are eventually fixed or lost
- 2N alleles
  - Each equally likely to fix (selectively equivalent)
  - Pr(fixation) = 1/2N
  - If i copies of allele, Pr(fixation) = $i/2N$
- Pr(fixation) = p
- Pr(loss) = $1-p$

Allele frequencies will change randomly over time

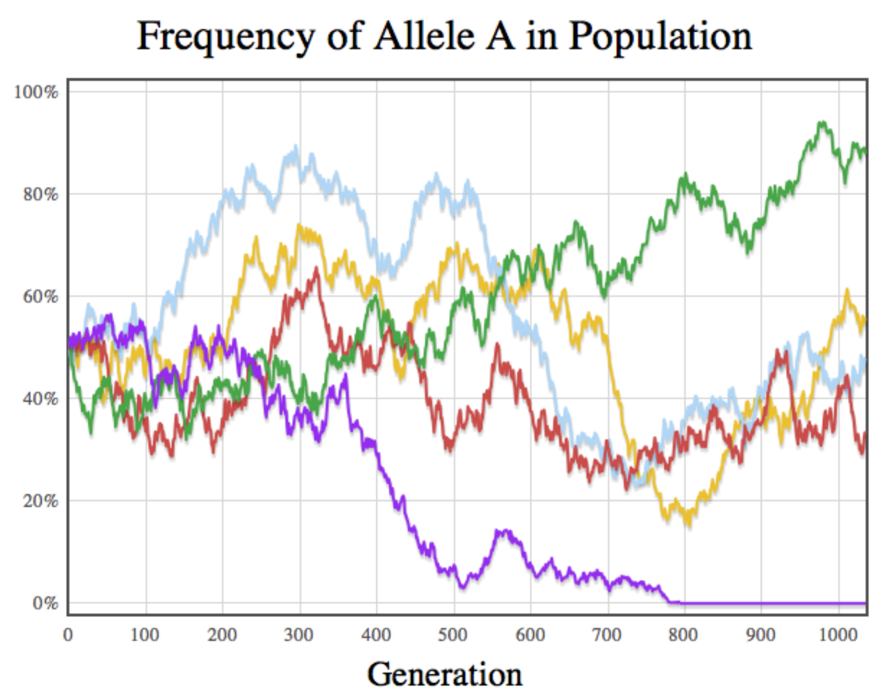



1
A
B
C
Allele frequency
0
Generation
A
B
C
ϕ(x)
0
1
0
1
0
1
Allele frequency, x

N = 40



N = 2000



http://scit.us/redlynx/

Times to fixation, loss

$$t_{fix} = \frac{-4N(1-p)\ln(1-p)}{p}$$

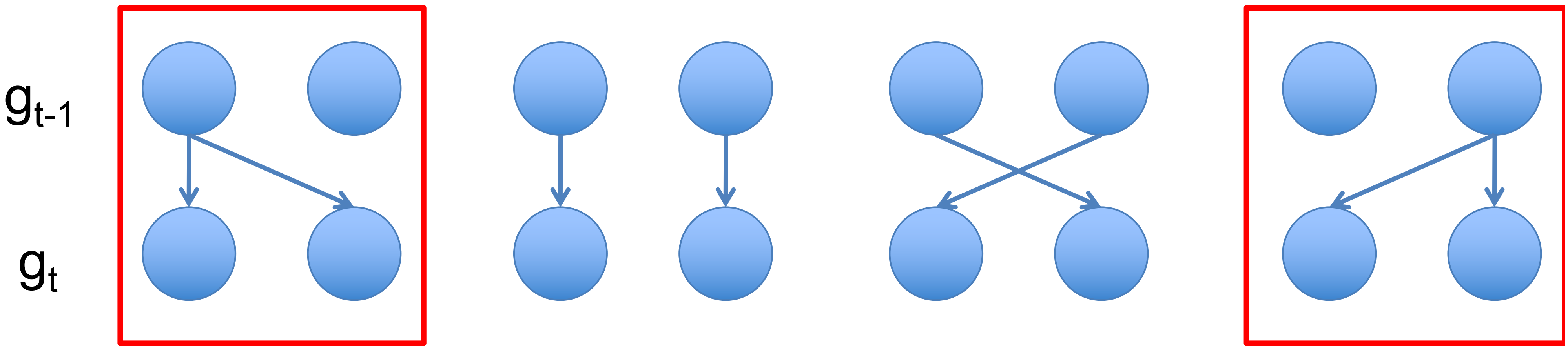
For p = 1/2N, $t_{fix} \approx 4N$

$$t_{loss} = \frac{-4N(p)\ln(p)}{1-p}$$

For p = 1/2N, $t_{loss} \approx 2\ln(2N)$

Decay of Heterozygosity

2N=2



Pr(IBD) = 2/4 = 1/2

Pr(IBD) = 1/2N

Pr(not IBD) = 1 - 1/2N

$$\Pr(IBD_t) = F_t = \frac{1}{2N} + (1 - \frac{1}{2N})F_{t-1}$$

$$\text{If } F_0 = 0, F_t = 1 - (1 - \frac{1}{2N})^t$$

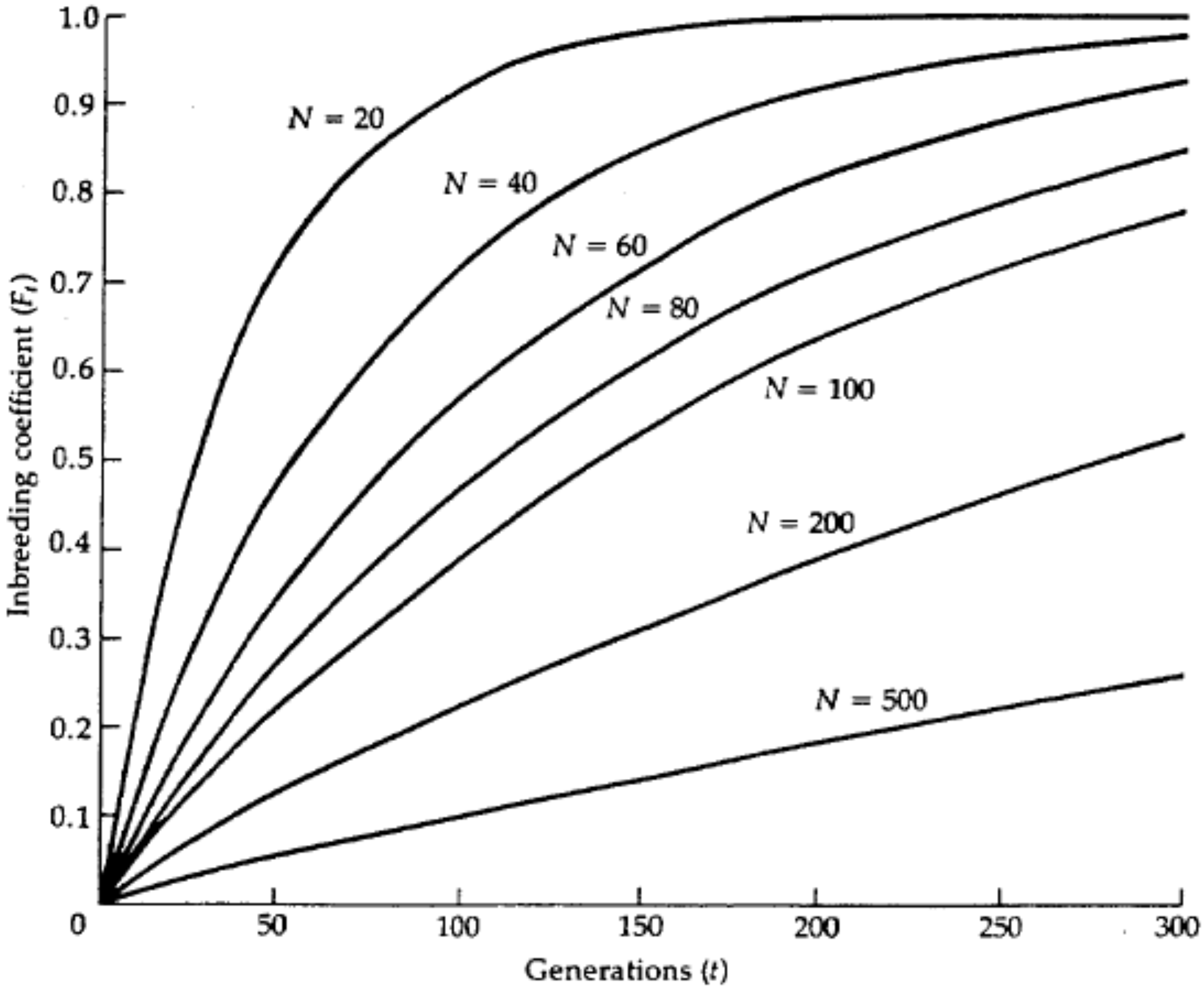


Figure 9. Increase of F_t in ideal populations as a function of time and effective population size N.

Hartl & Clark

Decay of Heterozygosity

$$F_t = 1 - (1 - \frac{1}{2N})^t$$

$$H_t = H_0\left(1 - \frac{1}{2N}\right)^t \approx H_0 e^{-t/2N}$$

Summary of Drift

- Stochastic fluctuations in allele frequencies due to sampling in a finite population
- Described by Wright-Fisher model
- Alleles are ultimately fixed or lost from population
  - Pr(fix) = p; Pr(loss) = 1 – p
  - Time($\text{fix}_{1/2N}$) ≈ 4N; Time($\text{loss}_{1/2N}$) ≈ 2ln(2N)
- H decreases (~1/2N) over time
- Hardy-Weinberg largely still applies
  - Allele frequency changes small
  - Deviation from expected genotype frequencies ~ 1/2N

How does drift come into play in conservation genetics?

Habitat loss

- Small, isolated populations are threatened
  - RGD

Habitat loss

- Small, isolated populations are threatened
  - RGD, inbreeding
- Consequences:
  - Reduction in genetic diversity

Genetically depauperate species

Species	Populations (N)	Individuals (N)	Loci (N)	Polymorphic loci (%)	Average heterozygosity	Reference
			Allozyme			
Drosophila	43*	> 100	24	43.1	0.140	*(10)*
Mus musculus	2	87	46	20.5	0.088	*(15)*
Felis catus	1	56	55	22.0	0.076	*(16)*
Homo sapiens	Many	> 100	104	31.7	0.063	*(43–45)*
Acinonyx jubatus	2	55	47	0.0	0.0	




O'Brien et al. 1983

Genetically depauperate species

TABLE 2. Distribution and allele frequency of polymorphic allozyme loci in lions

Population	No. lions scored	Allozyme locus[a] *IDH1*	*TF*	*PT1*	Percent polymorphic loci	Average Heterozygosity[b]
African						
Kruger Park South Africa	15	A = 1.0	a = 0.65 b = 0.35	s = 1.0	7	0.023
Serengeti ecosystem Tanzania, East Africa	27	A = 0.72 B = 0.28	a = 0.52 b = 0.28	s = 1.0	11	0.038
African Zoo lions (Atlas)	18	A = 1.0	a = 0.72 b = 0.28	s = 1.0	7	0.030
Asian						
Indian lions (Gir Forest)	28	B = 1.0	a = 1.0	d = 1.0	0.0	0.0
Indian lions (SSP-studbook)	29	A = 0.72 B = 0.28	a = 0.93 b = 0.07	s = 0.45 d = 0.55	7	0.021

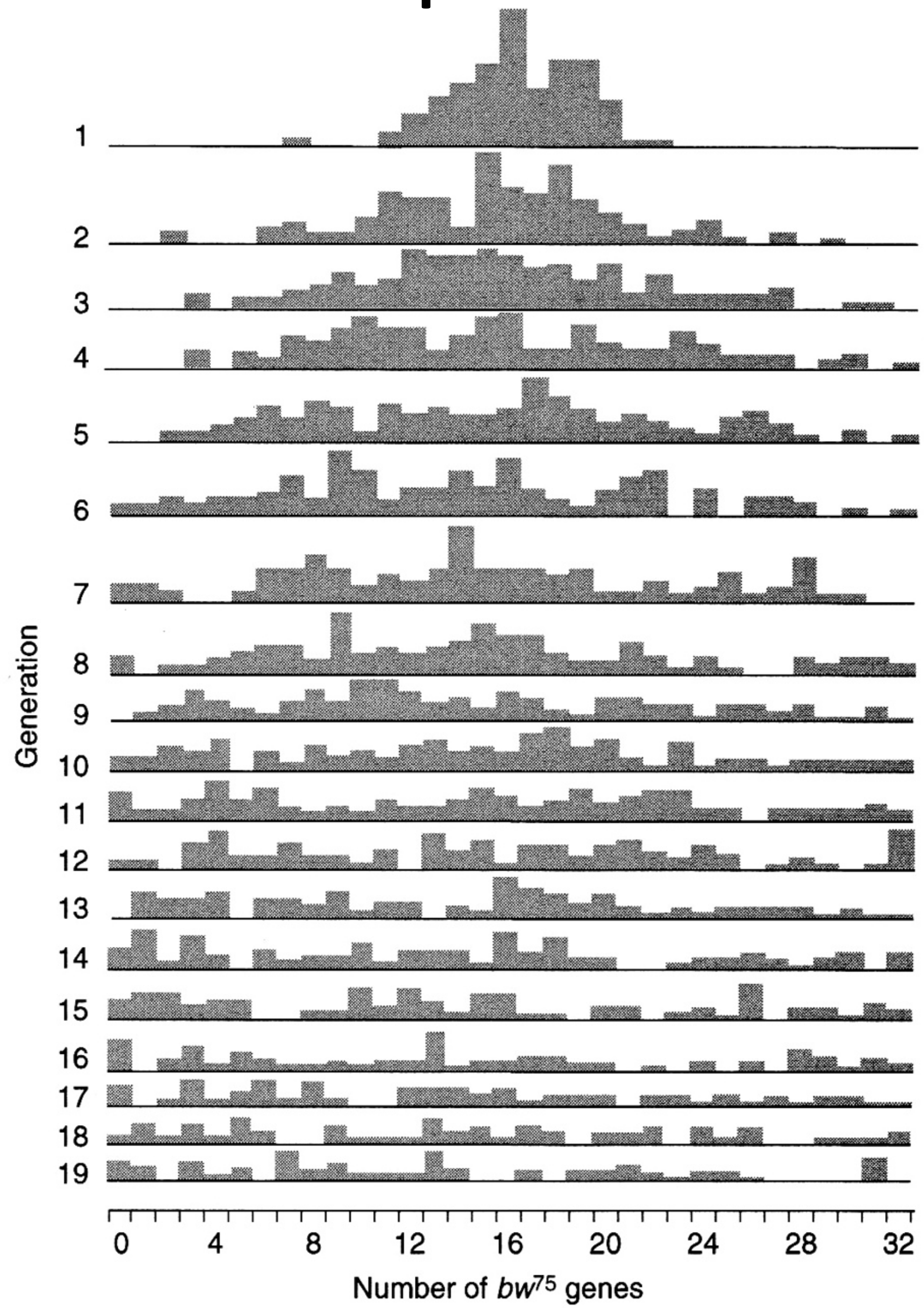
O'Brien et al.

Genetically depauperate species

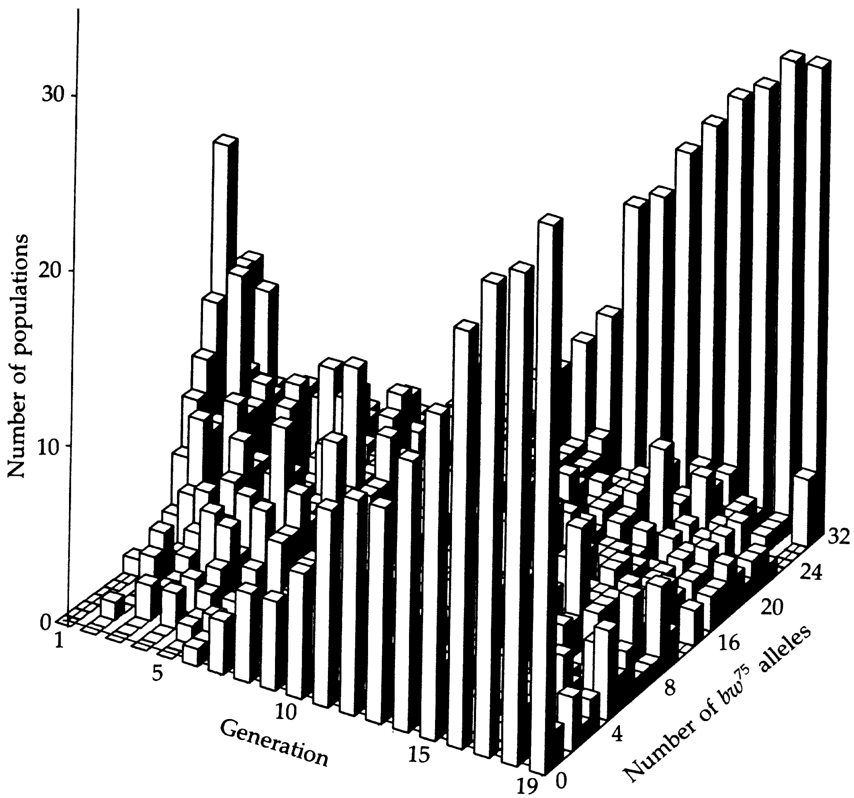
			Microsatellite locus			
Number	**Date**	**Location**	**043**	**090**	**096**	**mtDNA**
Contemporary						
14	1980s	Big Cypress Swamp	122/122	121/121	203/203	A
67	1980s	Big Cypress Swamp	122/122	121/121	203/203	A
71	1980s	Big Cypress Swamp	122/130	121/121	203/203	A
422	1980s	Big Cypress Swamp	122/122	121/121	203/203	A
426	1980s	Big Cypress Swamp	122/130	121/121	203/203	A
428	1980s	Big Cypress Swamp	122/122	121/121	203/203	A
Museum						
777	1890s	Florida	–	–	–	C
778	1890s	Florida	–	–	–	C
779	1890s	Florida	–	–	–	C
780	1890s	Immokolee	122/124	127/127	203/203	C
785	1898	Sebastian	134/134	–	–	A
787	1898	Sebastian	104/126	–	–	–
792	1922	Allen's River	–	–	–	B

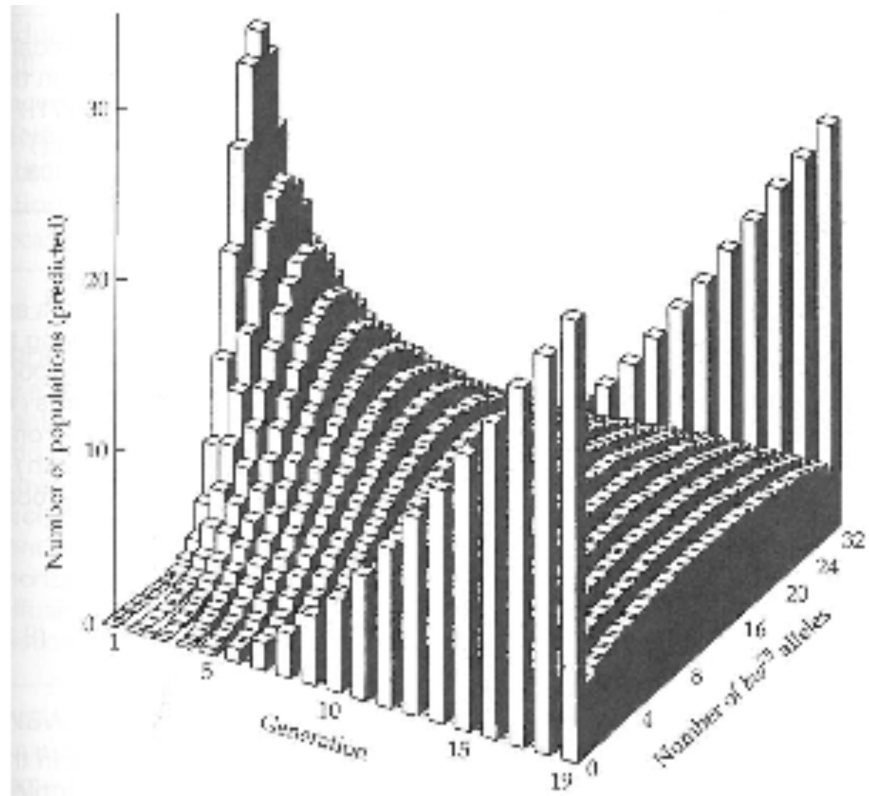
Culver et al. 2008

Effective Population Size



Effective Population Size





Buri 1956

Effective Population Size

$$\mathrm{Var}(p) = \mathrm{Var}\left(\frac{\#A}{2N}\right) = \left(\frac{1}{2N}\right)^2 \mathrm{Var}(\#A)$$

$$= \left(\frac{1}{2N}\right)^2 2Npq$$

$$= \frac{pq}{2N}$$

Effective Population Size

$$\mathrm{Var}(p) = \mathrm{Var}\left(\frac{\#A}{2N}\right) = \left(\frac{1}{2N}\right)^2 \mathrm{Var}(\#A)$$

$$= \left(\frac{1}{2N}\right)^2 2Npq$$

$$= \frac{pq}{2N}$$

- In reality, allele frequency changes > $pq/2N$
  - Fluctuations in population size, $N_m \neq N_f$, *etc.*

Effective Population Size

- Number of individuals in a theoretically ideal population having the same magnitude of drift as the actual population
- Measure 'magnitude' in 3 ways
  - Change in variance of allele frequency (variance N_e)

$$\mathrm{var}(p) = \frac{pq}{2N}$$

$$\overline{\mathrm{var}}(p) = \frac{pq}{2N_e}$$

$$N_e = \frac{pq}{2\overline{\mathrm{var}}(p)}$$

Effective Population Size

- Number of individuals in a theoretically ideal population having the same magnitude of drift as the actual population
- Measure ‘magnitude’ in 3 ways
  - Change in variance of allele frequency (variance N_e)
  - Change in Pr(IBD) (inbreeding N_e)
  - Rate of loss of heterozygosity (eigenvalue N_e)

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- Typically smaller than census size N

Effective Population Size

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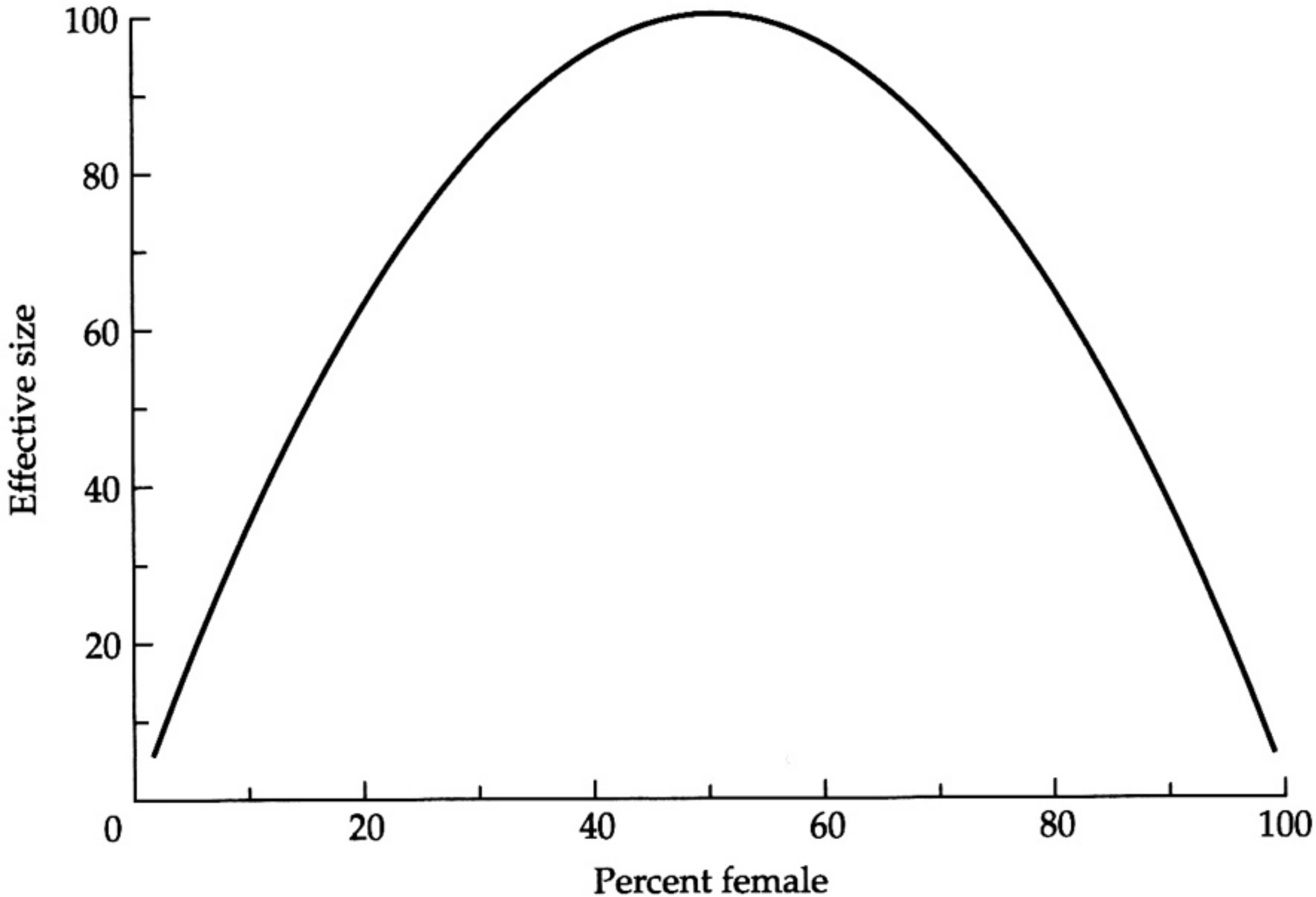
  - Change in Pr(IBD) (inbreeding N_e)
  - Rate of loss of heterozygosity (eigenvalue N_e)
- Typically smaller than census size N
  - Sex ratio
  - Variance in reproductive success
  - Population size changes

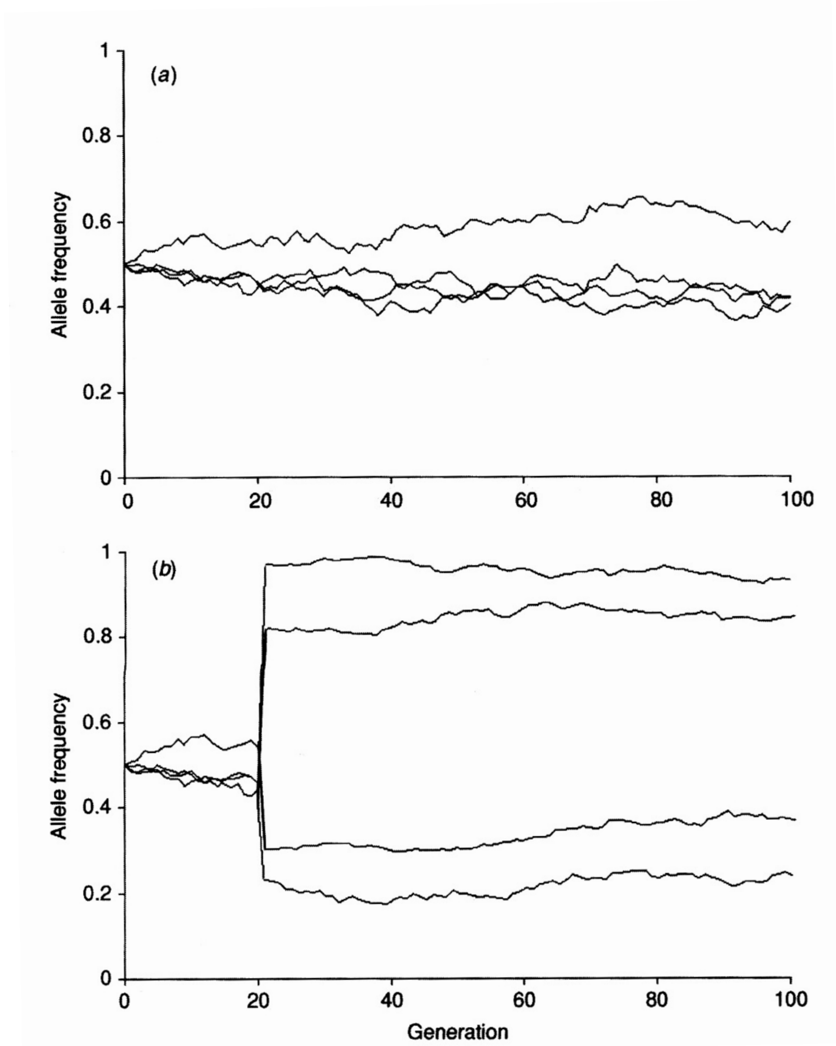
Effective Population Size



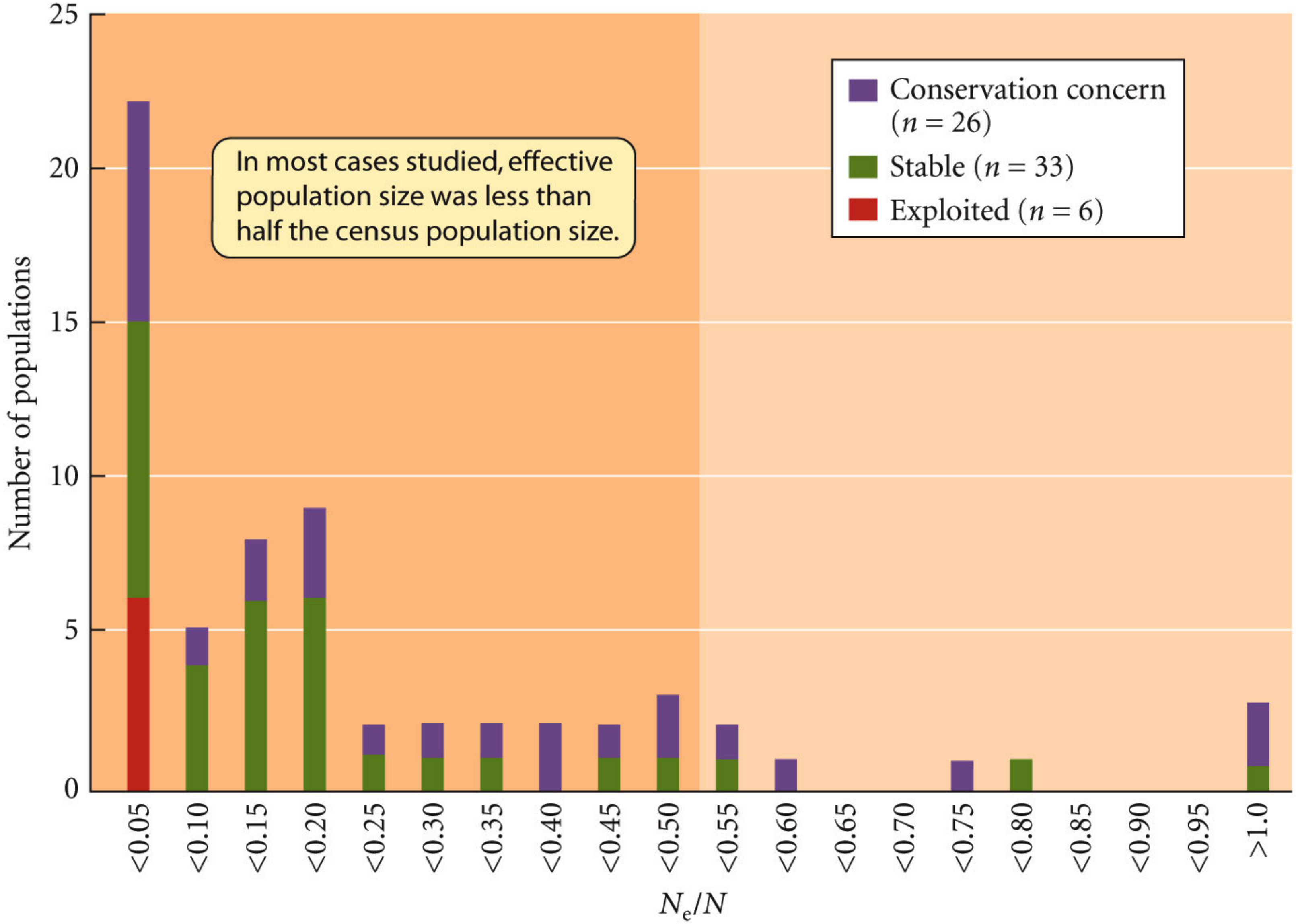
Effective Population Size

No bottleneck

Bottleneck at generation 20



Effective Population Size



***EVOLUTION 2e*, Figure 10.6**

Relating N, N_e

- Population size changes

$$\frac{1}{N_e} = \frac{1}{t}\left(\frac{1}{N_0} + \frac{1}{N_1} + \ldots + \frac{1}{N}\right)$$

Relating N, N_e

- Population size changes

$$\frac{1}{N_e} = \frac{1}{t}\left(\frac{1}{N_0} + \frac{1}{N_1} + \ldots + \frac{1}{N}\right)$$

- Unequal males, females

$$N = N_m + N_f$$

$$N_e = \frac{4N_mN_f}{N_m + N_f}$$