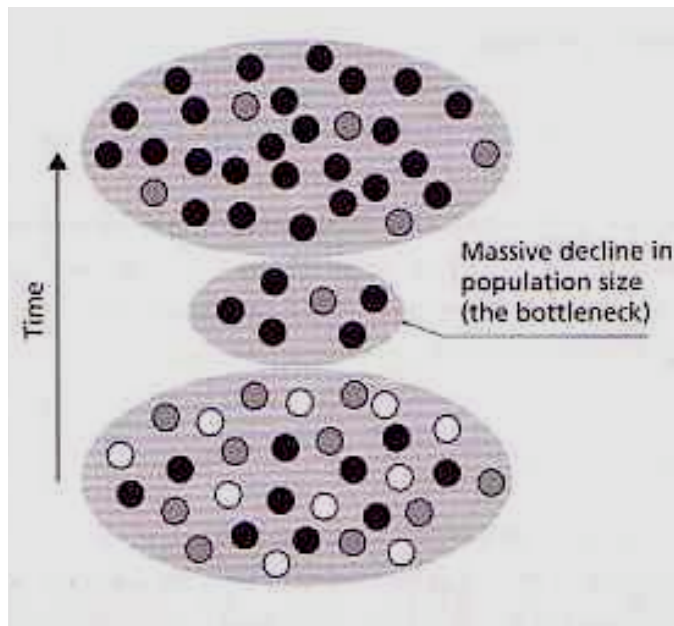


# Lecture 14: Effects of small population sizes

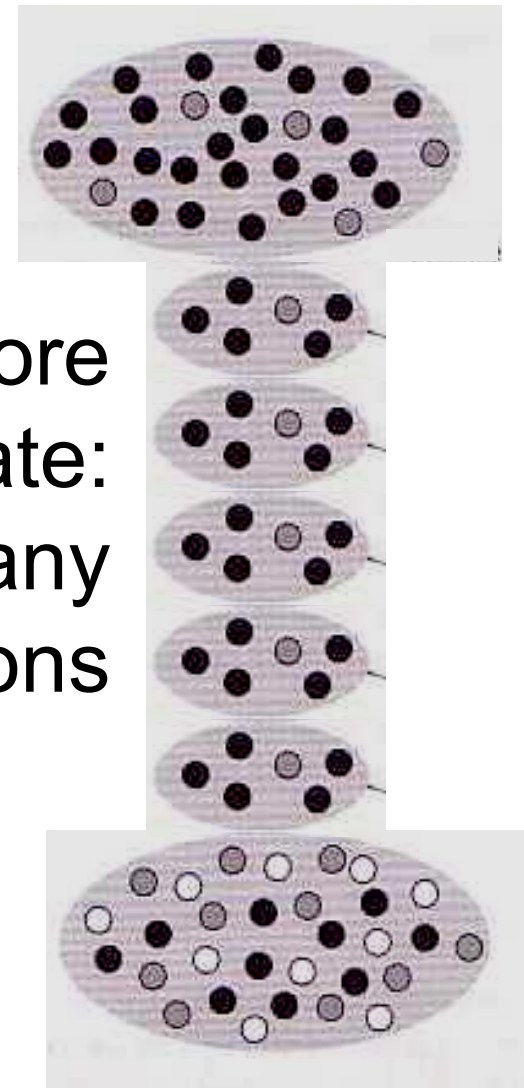
## Overview and genetic drift

**Bottleneck:** periods during which only a few individuals survive to continue the existence of the population

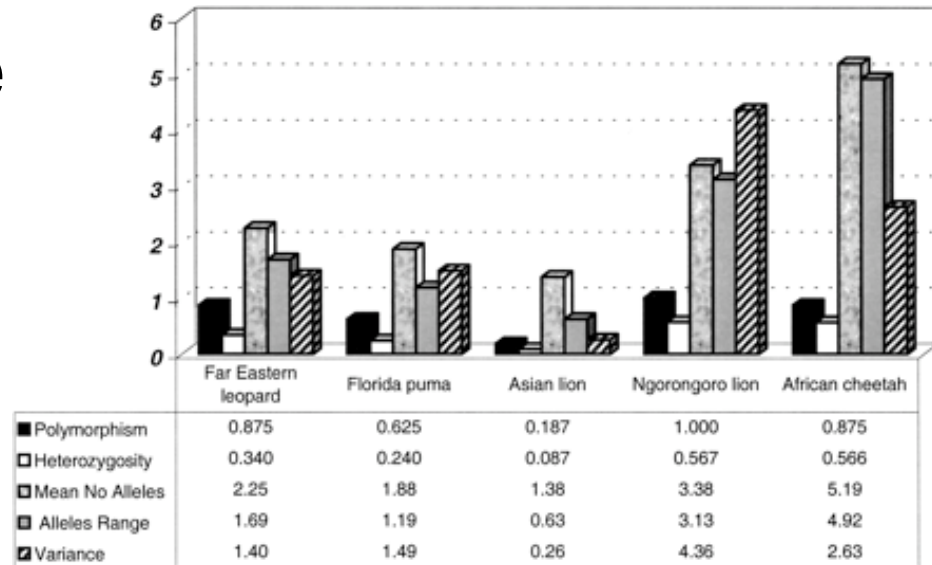


Less accurate

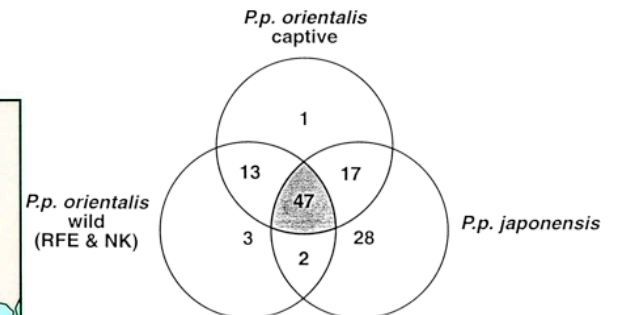
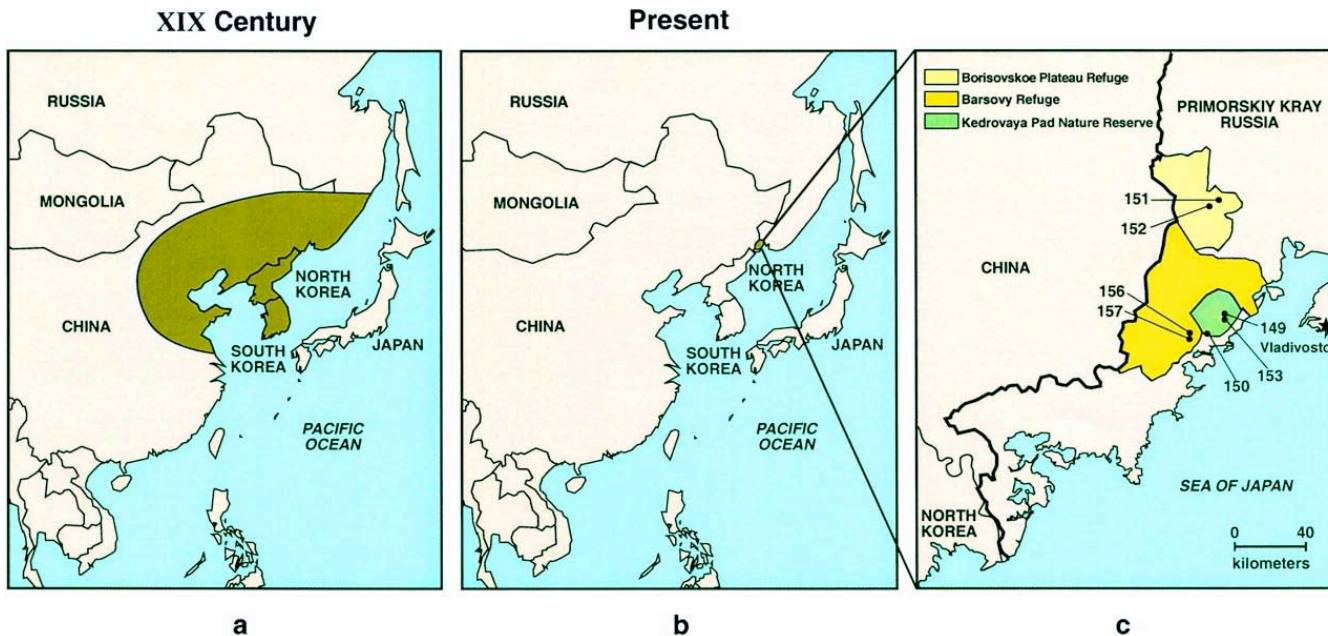
More  
Accurate:  
many  
generations



# Bottleneck: example

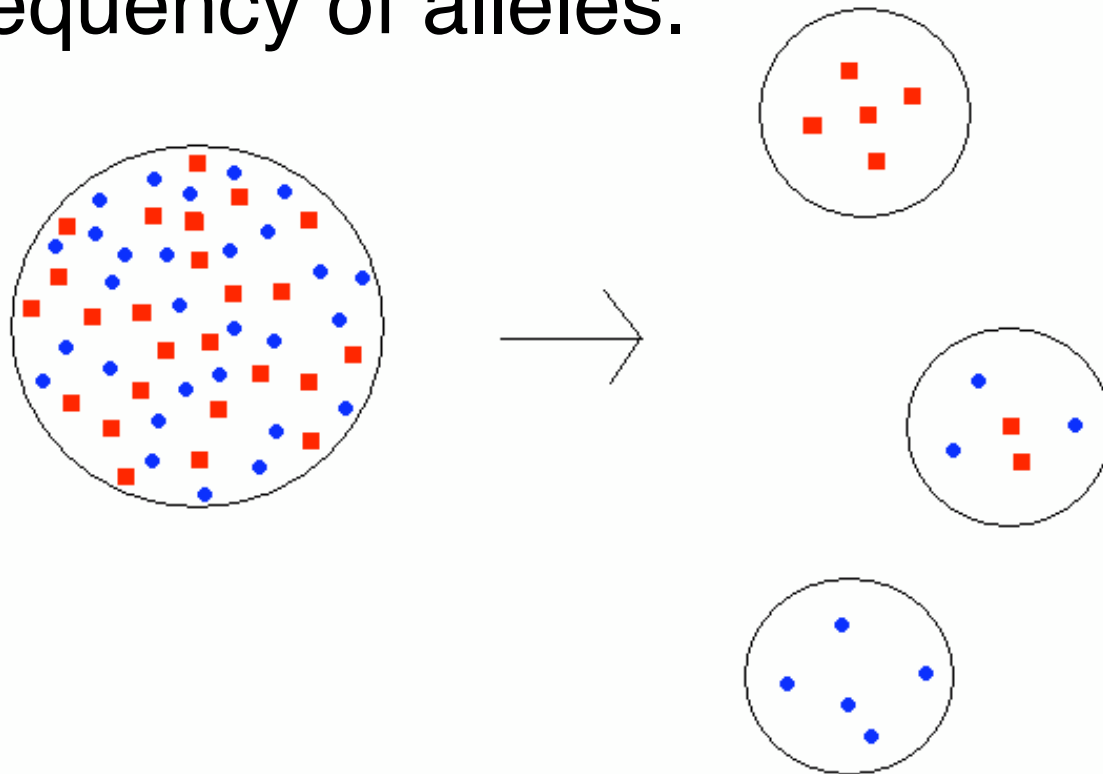


Amur leopard, *Panthera pardus orientalis*, Uphyrkina and colleagues

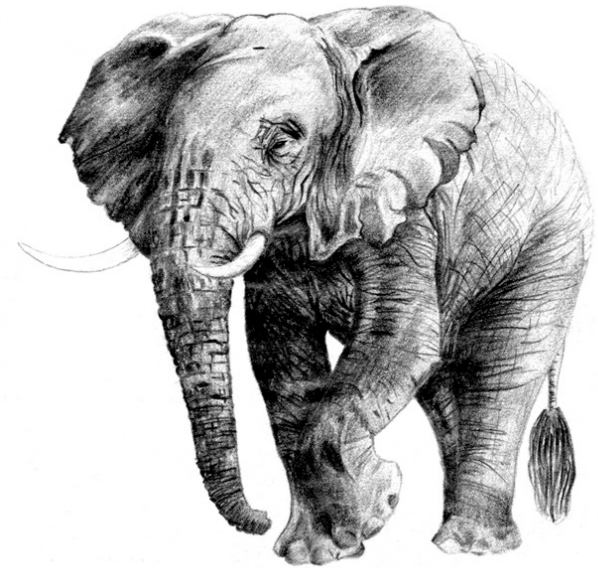
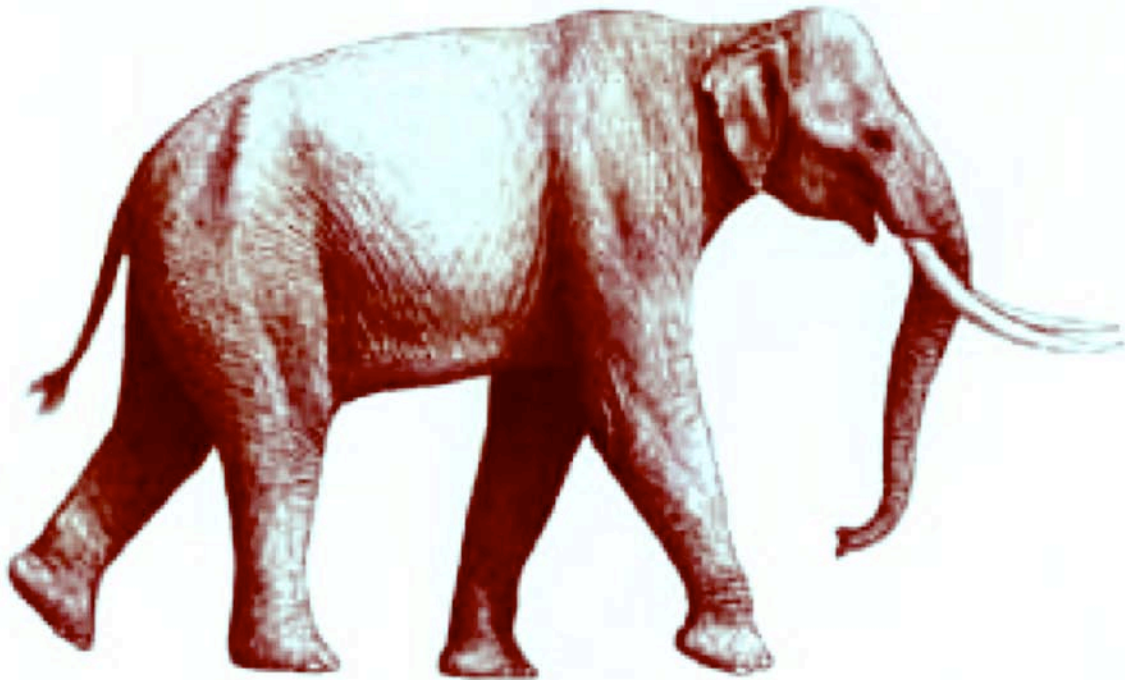


Venn diagram for the number of microsatellite alleles shared or unique from three leopard populations

**Founder effect:** loss of genetic variation when a new population is established by a small number of individuals from a larger founding population. May lead to low genetic variation or, by chance, to an unusually high or low frequency of alleles.

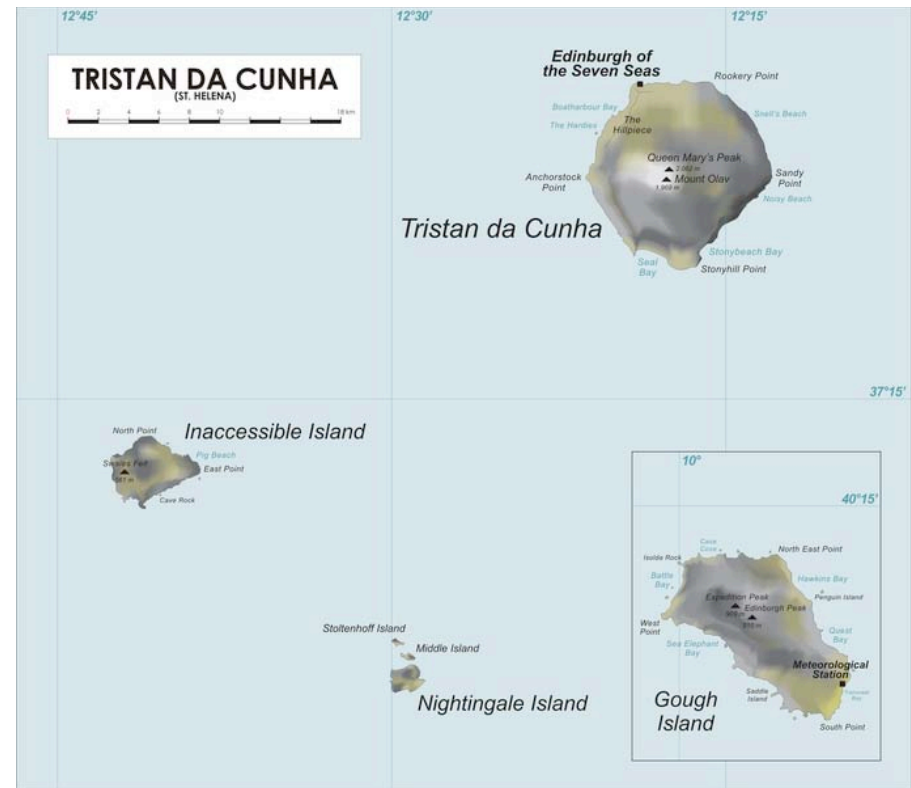
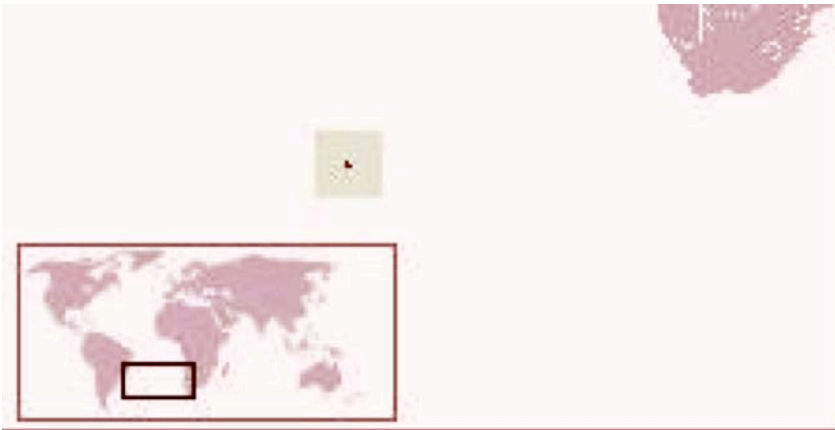


**Founder effect**, possible example:  
expansion of range of *Loxodonta africana*  
after *Elephas (recki) iolensis* becomes  
extinct in the African savannas





**Founder effect:** example, island of Tristan da Cunha, founded by 7 families. Currently 271 residents, suffer from asthma and glaucoma due to endogamy.



# Founder effect: Tristan da Cunha Island

(a)

<i>Founding females</i>	<i>mtDNA sequence</i>	<i>N (proportion)</i>
S. W.	ACTTGTTTCG	46 (0.29)
M. W. and M. W.	GTTCGCTTCG	34 (0.21)
E. S. and A. S.	GCTTATCTTG	25 (0.16)
M. L.	ATCTGCCCTA	11 (0.07)
S. P.	GTCTGTCCTG	45 (0.28)
Total		161 (1.0)

(b)

<i>Family</i>	<i>Y-chromosome haplotype</i>	<i>N (proportion)</i>
1	15-12-25-10-14-13	5 (0.066)
2	14-12-24-11-13-13	3 (0.039)
3	14-12-23-11-13-13	9 (0.118)
	14-12-23-10-13-13 (mutant)	4 (0.053)
4	14-12-24-10-13-14	8 (0.105)
	16-12-25-10-11-13 (migrant)	1 (0.013)
5	14-12-23-10-14-13	16 (0.211)
	14-14-22-10-11-13 (from family 7)	3 (0.039)
6	16-13-24-10-11-13	10 (0.132)
	14-12-23-10-14-13 (from family 5)	1 (0.013)
7	14-14-22-10-11-13	14 (0.184)
	14-12-23-10-14-13 (from family 5)	2 (0.026)
Total		76 (1.0)

mutation

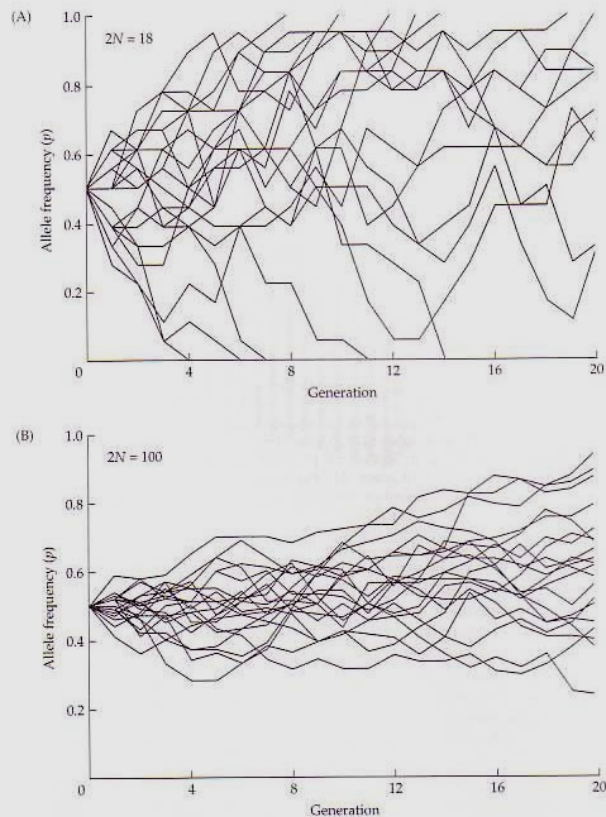
migrant

paternity?

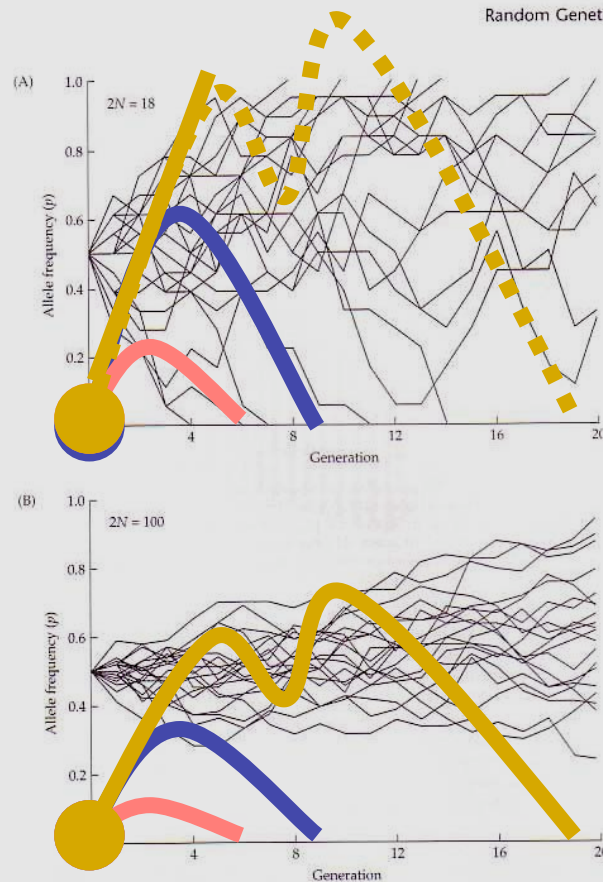
**Genetic drift:** chance changes in allele frequencies caused by finite population sizes.

In small populations, genetic drift may be significant even for loci with sizable selective effects.





**Figure 7.2** Computer simulations of the Wright-Fisher model of random genetic drift. Each line represents a population of size (A)  $2N = 18$  or (B)  $2N = 100$ , simulated for 20 generations. Each generation alleles are sampled with replacement as described in the text. An allele frequency of  $p = 0.5$  in A implies that there are nine copies of the  $A$  allele, and nine copies of the  $a$  allele. In B, an allele frequency of 0.5 implies 50 copies of each allele. Note that the larger population size in B results in smaller oscillations of allele frequency, and a slower rate of fixation.



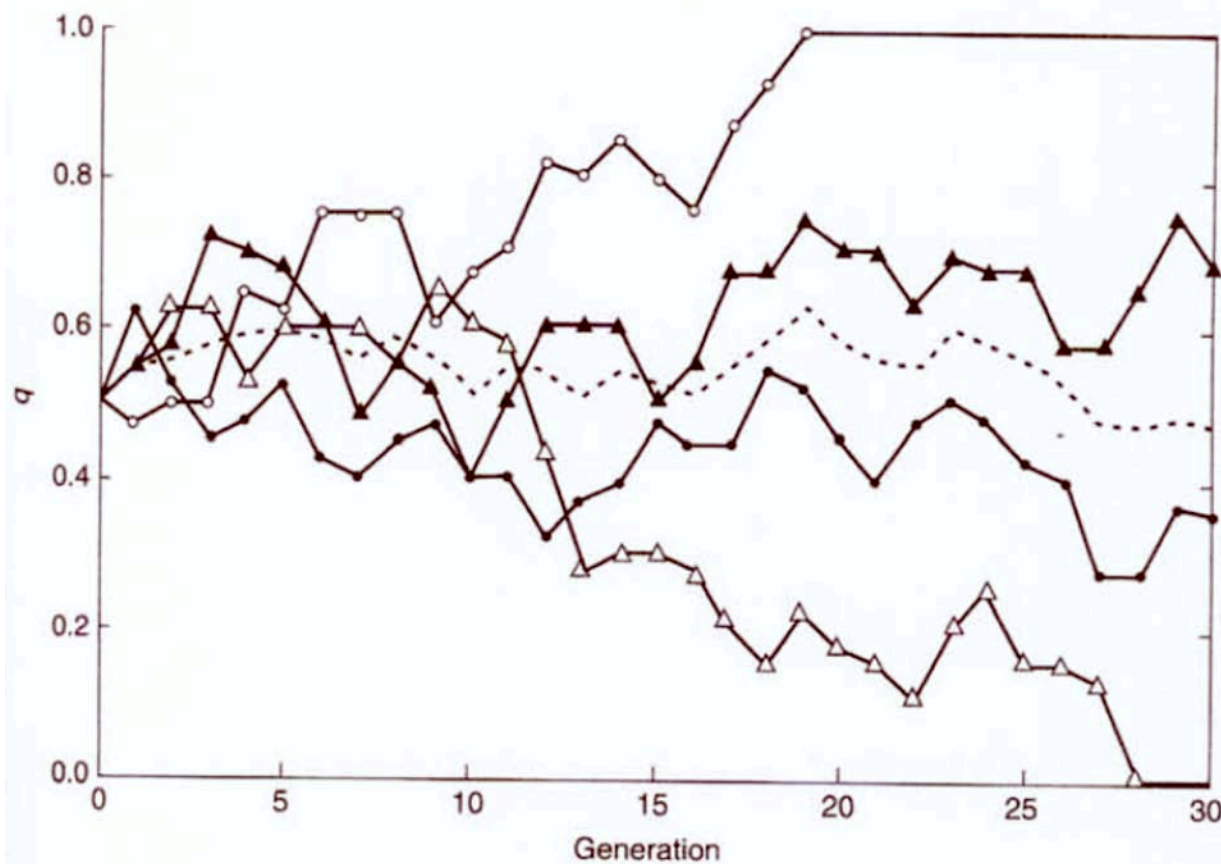
**Figure 7.2** Computer simulations of the Wright-Fisher model of random genetic drift. Each line represents a population of size (A)  $2N = 18$  or (B)  $2N = 100$ , simulated for 20 generations. Each generation alleles are sampled with replacement as described in the text. An allele frequency of  $p = 0.5$  in A implies that there are nine copies of the  $A$  allele, and nine copies of the  $a$  allele. In B, an allele frequency of 0.5 implies 50 copies of each allele. Note that the larger population size in B results in smaller oscillations of allele frequency, and a slower rate of fixation.

Small  
populations  
genetic drift

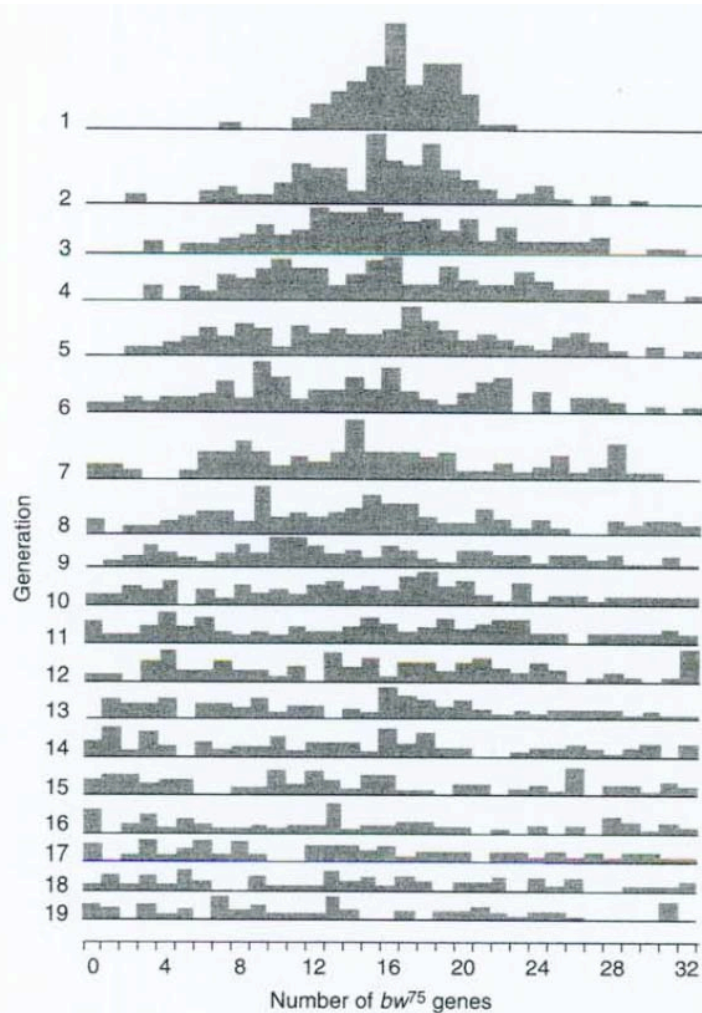
Large  
populations  
selection

Detrimental  
mutations

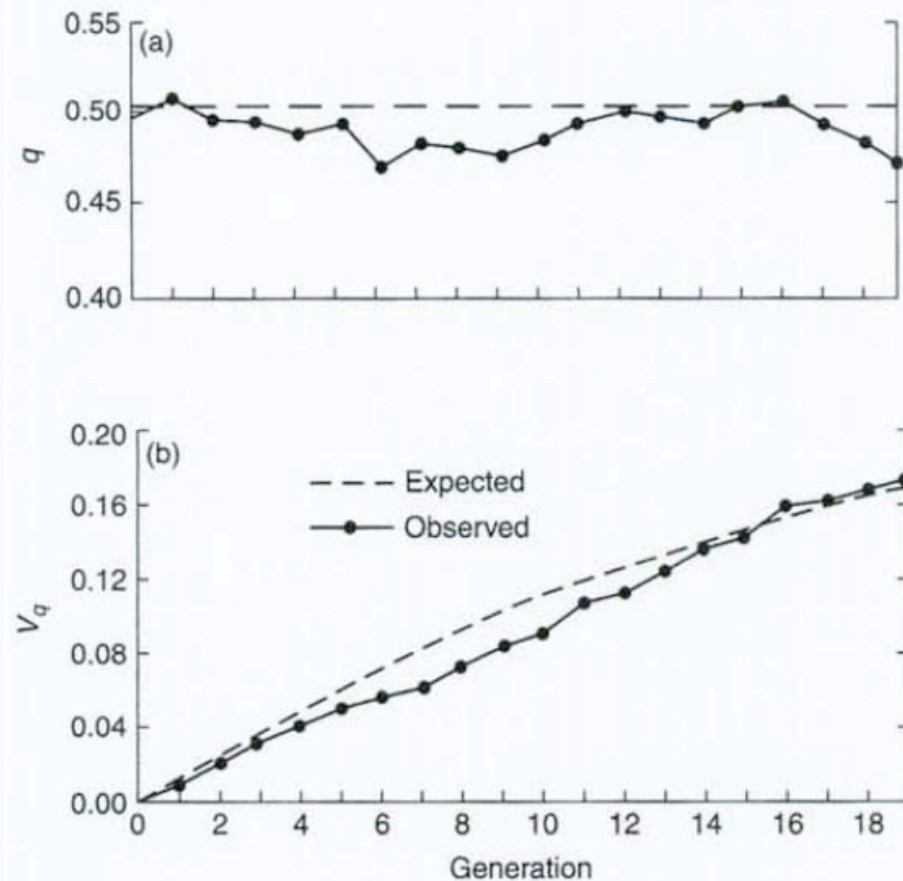
**Genetic drift:** Monte Carlo simulation to imitate allele changes for  $A_2$  in 4 populations of size 20 over 30 generations.



# Genetic drift: Classic experiment (Buri, 1956), *brown* locus with 2 neutral alleles for eye color in *Drosophila*, $p = q = 0.5$ , $n=16$



**Figure 6.3.** The distribution of  $bw^{75}$  alleles over time in populations of size 16 for the segregating replicates in an experiment of Buri (after Buri, 1956).



# Genetic drift, probability transition matrix:

Computes the probability of  $i$   $A_2$  alleles in generation  $t+1$ , given  $j$   $A_2$  alleles in generation  $t$

**TABLE 6.2** A probability transition matrix for a population of size two ( $2N = 4$ ), where the values indicate the probability of  $iA_2$  alleles in generation  $t + 1$ , given  $jA_2$  alleles in generation  $t$ .

<i>Generation <math>t + 1</math></i>	<i>Generation <math>t</math></i>				
	0	1	2	3	4
0	1	0.3164	0.0625	0.0039	0
1	0	0.4219	0.25	0.0469	0
2	0	0.2109	0.375	0.2109	0
3	0	0.0469	0.25	0.4219	0
4	0	0.0039	0.0625	0.3164	1



# Genetic drift, probability transition matrix: populations of size 2 ( $2N = 4$ ) where initially $p_0 = q_0 = 0.5$

**TABLE 6.3** The distribution of allele frequencies and heterozygosity over generations for populations of size two ( $2N = 4$ ) when  $q_0 = 0.5$ .

Number of $A_2$ alleles	Generation					
	0	1	2	3	4	$\infty$
0	0	0.0625	0.1660	0.2490	0.3117	0.5
1	0	0.25	0.2109	0.1604	0.1205	0.0
2	1	0.375	0.2461	0.1813	0.1356	0.0
3	0	0.25	0.2109	0.1604	0.1205	0.0
4	0	0.0625	0.1660	0.2490	0.3117	0.5
$q_t$	0.5	0.5	0.5	0.5	0.5	0.5
$H_t$	0.5	0.375	0.2812	0.2109	0.1582	0.0



# Genetic drift, probability transition matrix: populations of size 2 ( $2N = 4$ ), $q_0 = 0.5$ or $0.25$

**TABLE 6.3** The distribution of allele frequencies and heterozygosity over generations for populations of size two ( $2N = 4$ ) when  $q_0 = 0.5$ .

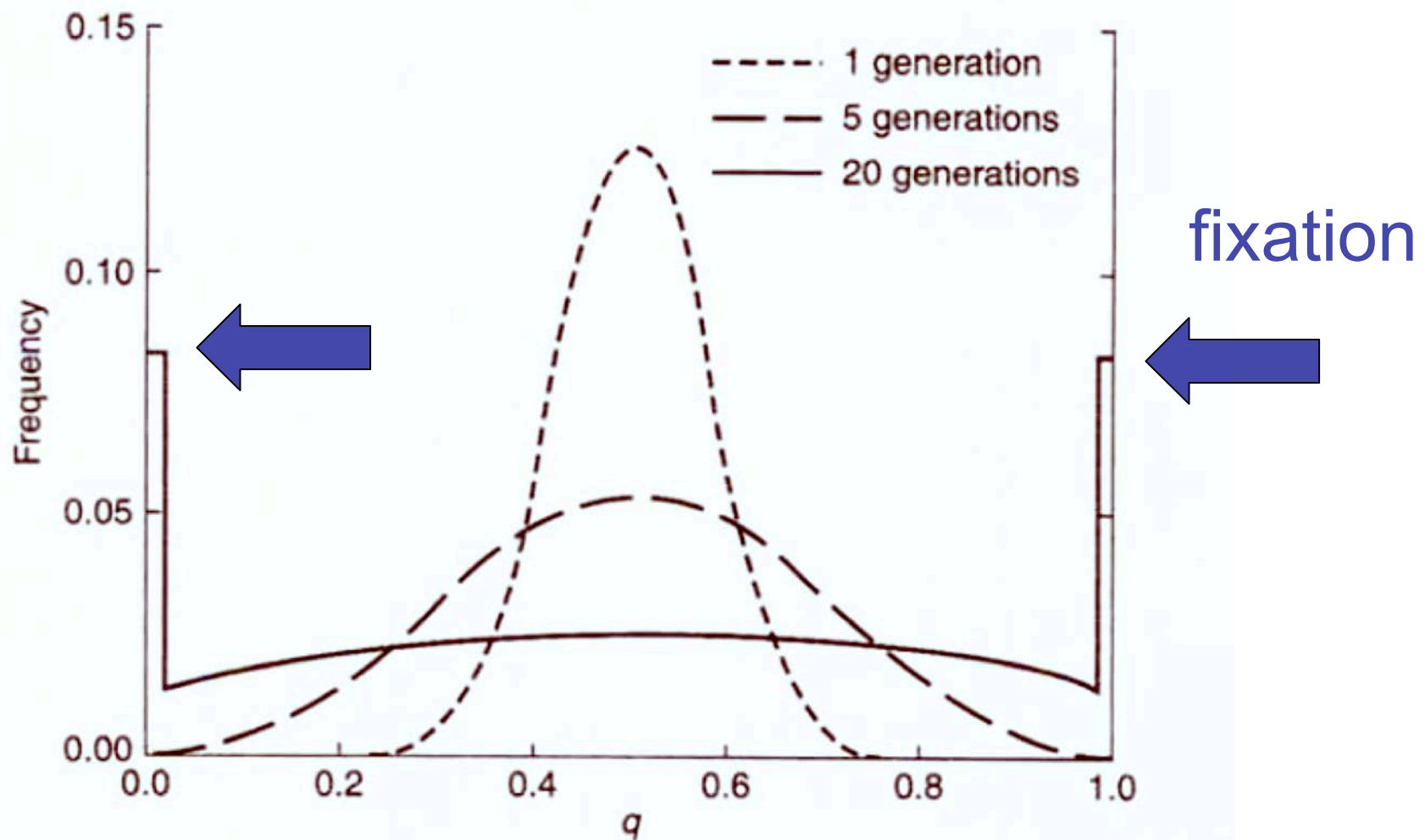
Number of $A_2$ alleles	Generation						
	0	1	2	3	4	...	$\infty$
0	0	0.0625	0.1660	0.2490	0.3117	...	0.5
1	0	0.25	0.2109	0.1604	0.1205	...	0.0
2	1	0.375	0.2461	0.1813	0.1356	...	0.0
3	0	0.25	0.2109	0.1604	0.1205	...	0.0
4	0	0.0625	0.1660	0.2490	0.3117	...	0.5
$q_t$	0.5	0.5	0.5	0.5	0.5	...	0.5
$H_t$	0.5	0.375	0.2812	0.2109	0.1582	...	0.0

**TABLE 6.4** The distribution of allele frequencies and heterozygosity over generations for populations of size two ( $2N = 4$ ) when  $q_0 = 0.25$ .

Number of $A_2$ alleles	Generation						
	0	1	2	3	4	...	$\infty$
0	0	0.3164	0.4633	0.5484	0.6038	...	0.75
1	1	0.4219	0.2329	0.1471	0.1003	...	0.0
2	0	0.2109	0.1780	0.1353	0.1017	...	0.0
3	0	0.0469	0.0923	0.0943	0.0805	...	0.0
4	0	0.0039	0.0336	0.0748	0.1137	...	0.25
$q_t$	0.25	0.25	0.25	0.25	0.25	...	0.25
$H_t$	0.375	0.2812	0.2109	0.1582	0.1187	...	0.0

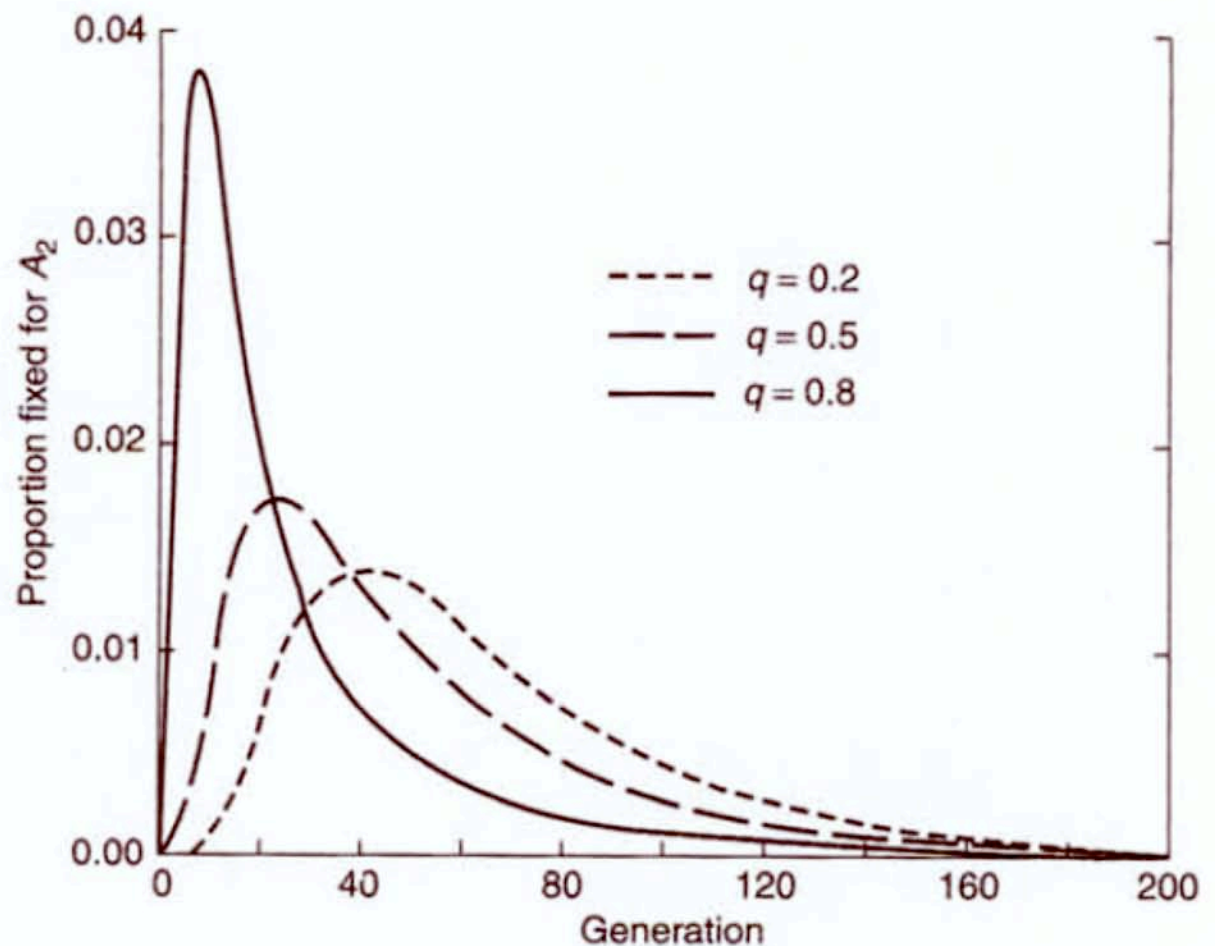
**Probability of fixation of allele  $A_2$  is equal to the initial frequency of allele  $A_2$**

**Probability matrix approach** can calculate the distribution of allele frequencies over time for finite populations of different sizes.



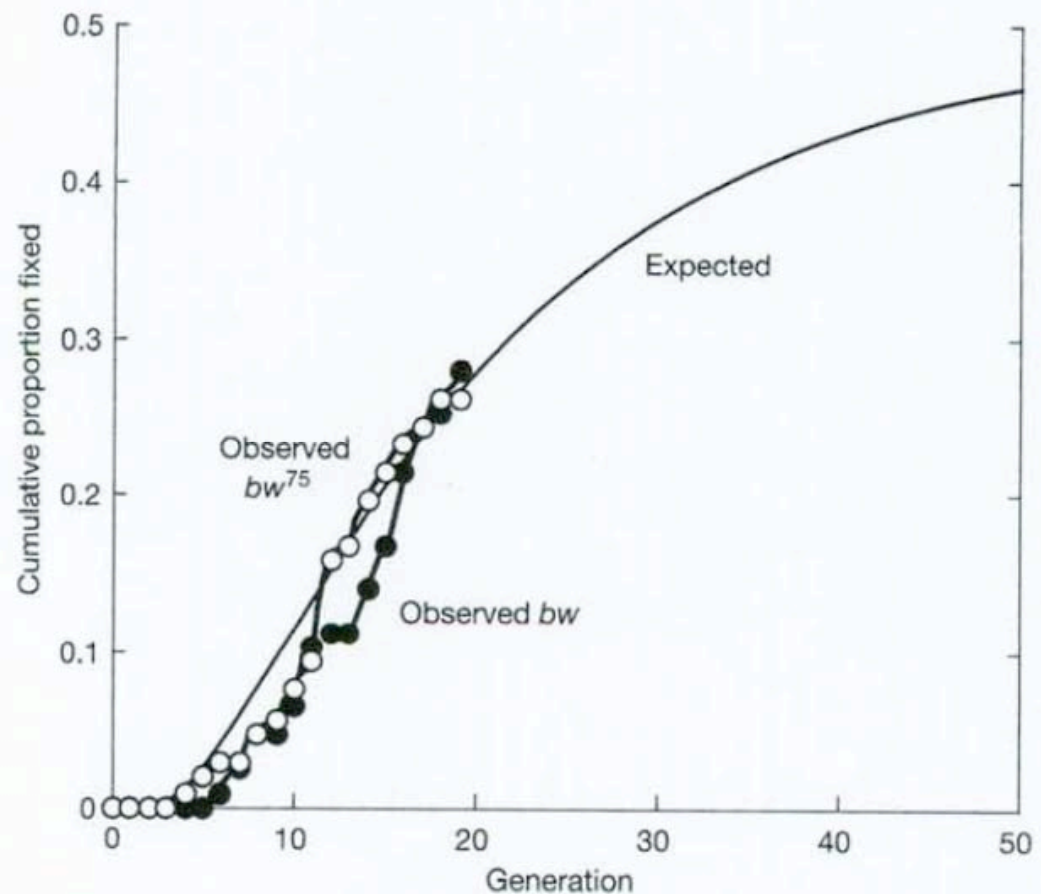
**Mean time for fixation** of an allele depends on population size and initial allele frequency. Where  $q$  is small, the expected time to fixation of a neutral mutant is four times the population size

**Figure 6.6.** The smoothed distribution of populations becoming fixed for  $A_2$  in each generation for three initial allele frequencies when  $N = 20$ .



**Mean time for fixation** of an allele depends on population size and initial allele frequency. Where  $q$  is small, the expected time to fixation of a neutral mutant is four times the population size

**Figure 6.7.** The observed cumulative proportion of fixed populations for alleles  $bw$  (closed circles) and  $bw^{75}$  (open circles) (Buri, 1956). Also given is the expected cumulative proportion from iteration of the transition matrix with  $2N = 18$ .



Effects of small population sizes

Effective population size



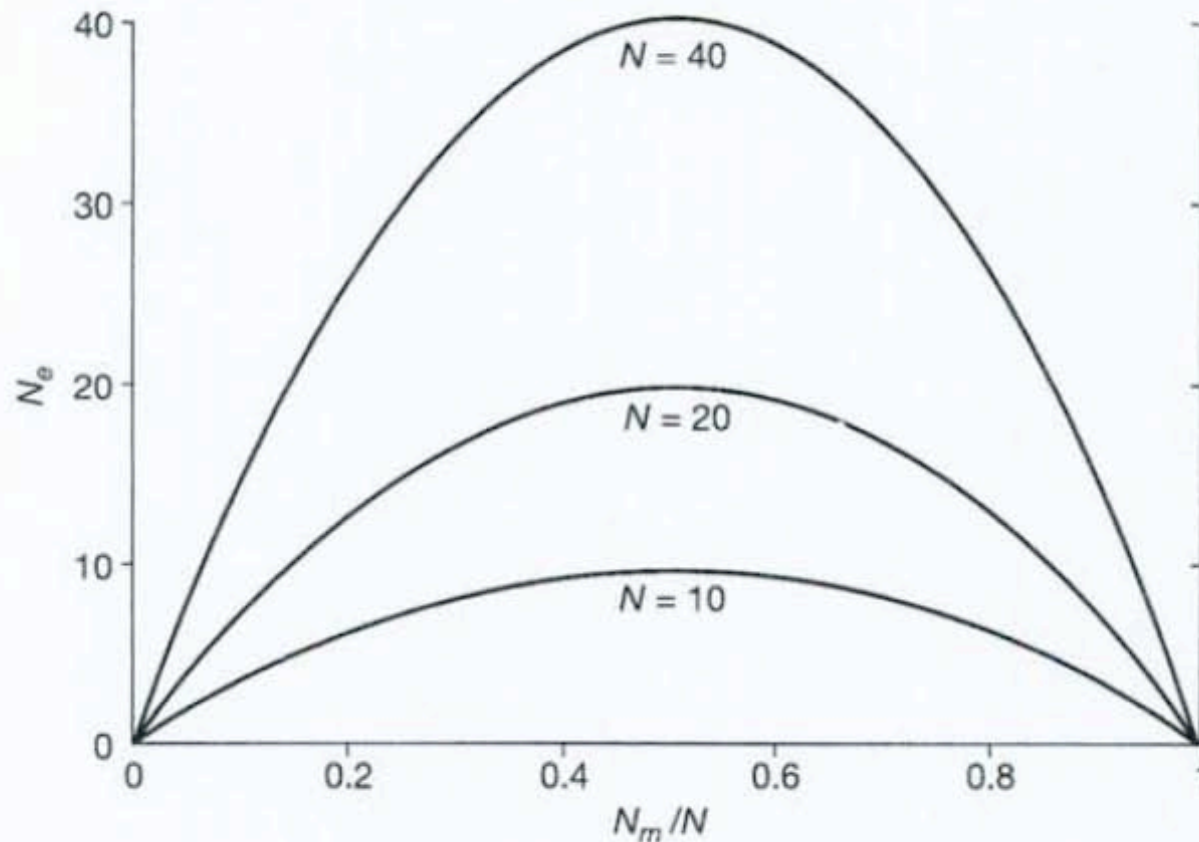
**Effective population size,  $N_e$ :** "the number of breeding individuals in an idealized population that would show the same amount of dispersion of allele frequencies under random genetic drift or the same amount of inbreeding as the population under consideration" ie a population of size  $N$  where all parents have equal expectation of being the parents of any progeny individual.

Usually smaller than the actual or census or breeding population size,  $N$ .

**Effective population size,  $N_e$ :** takes into account variations in the sex ratio of breeding individuals, offspring number per individual, difference in numbers breeding across generations, and other factors

So  $N_e$  is the size of an idealized population that would produce the same amount of inbreeding, allele frequency variance, or heterozygosity loss as the actual population under consideration

**Effective population size,  $N_e$ :** affected by sex ratio of breeding males to breeding females (in many species  $N_m < N_f$ )



**Figure 6.8.** The effective population size as a function of the proportions of males,  $N_m/N$ , for three different total numbers of individuals.

**Effective population size,  $N_e$ :** affected by sex ratio of breeding males to breeding females (in many species  $N_m < N_f$ )

