

SMALL POPULATIONS:

OBSERVED GENETIC EFFECTS

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ABSTRACT

It has been demonstrated theoretically that small populations should suffer a reduction in genetic variation. This paper will review studies of the effects of small population size on genetic variation utilizing examples from both laboratory and natural populations. Although many studies have shown that there is a decrease in genetic variation in small populations, other studies have indicated that there may not be a direct relationship between population size and the amount of genetic variation. Implications of these findings for wildlife management will be made.

INTRODUCTION

We have just heard about some of the factors that can lead to changes in gene frequency. Such factors as drift, founder effect, and inbreeding can result in shifts in gene frequency with a resultant lowering of the mean heterozygosity in the population. In my talk I would like to discuss what is known concerning the actual genetic effects of small population size and inbreeding in natural populations of animals.

Before addressing these questions, it would be well to examine briefly how one determines something about the genetic composition of a population. How is it possible for example, to characterize the genetic variation in elk, deer, mountain goats, bighorn sheep, etc. Without a doubt the major breakthrough in this area has been the application of electrophoresis to detect allelic and non-allelic forms of proteins in individuals. The use of electrophoresis has made it possible to identify precisely the number of individuals with different genotypes since it is possible usually to differentiate between homozygotes and heterozygotes. This allows one to monitor the amount and distribution of genetic variation in natural populations and to determine the genetic structure of different populations. Most of the studies which have utilized electrophoresis to characterize natural populations have examined twelve or more different genetic loci. In some cases the specific function of the enzyme may not even be known. However, the enzymes are under genetic control and they provide the investigator with a means of determining the genetic variations

within and among different populations. It is of interest to determine how many loci are polymorphic in the population and how much heterozygosity is shown by individuals in the population. A review of the mean heterozygosity and the proportion of polymorphic loci (Table 1) indicates that the mean heterozygosity tends to be small, usually 1 to 2 percent.

A study of nearly 300 elephants in the Kruger National Park in Africa (Osterhoff and Schoeman 1974) found genetic differentiation in the serum transferrins, albumins, and esterases, but did not find any differences in the hemoglobins and a number of enzymes indicating that these groups of elephants seemed to be relatively homogeneous genetically. Since, in general, the loss of genetic variation may make a population or species less able to adapt to sudden environmental changes, the small amount of genetic variation in this herd of elephants may have profound implications for the proper management of the herd.

An additional example of the types of information which come out of electrophoretic studies is seen in the data on deer in South Carolina (Ramsey et al. 1979). Two adjacent populations of white-tailed deer in South Carolina were found to differ widely in a number of demographic features. The two herds, designated "swamp" and "upland", were found to have different age structures, sex ratios, and reproductive rates. The swamp herd had changing demographic patterns whereas the demographic features of the upland herd tended to remain stable over time. Ramsey and his co-workers also studied the electrophoretic variations of 21 proteins encoded by 22 structural genetic loci. Differences in genotypic frequencies between herds, sexes and age classes were found for the esterase and hemoglobin loci. Correlations between demographic and genetic differences may be useful in identifying populations and in defining management units and practices.

We have seen that inbreeding or consanguinity increases the likelihood of homozygosity among the offspring. This will be true whether inbreeding is a result of some irregular system of mating (within some human cultures it was the preferred social custom) or a result of some irregular system or happening. Inbreeding, per se, is not necessarily good or bad. Its outcome will depend on whether or not the increase homozygosity is good or bad. Studies of laboratory animals, farm animals, zoo animals, and men have shown clearly that inbreeding brings about a decline in characters which are concerned with fitness and survival. Cattle show a 3.2 percent decline in milk yield with a 10 percent increase in inbreeding (Robertson 1954). Sheep show a 5 percent decline in fleece weight (Morley 1954) and poultry show a 6.2 percent decrease in egg production (Shoffner 1948) with a 10 percent increase in inbreeding. Juvenile mortality was higher in inbred animals than in noninbred mammals in several species of small ungulates and other small mammals maintained in zoos (Ralls et al. 1979; Ralls and Ballou 1982).

Perhaps the most extensive and systematic study of inbreeding has been that of populations in Japan (Schull and Neel 1965). Some representative

data are given in Table 2. These results, as well as other effects on mortality including stillbirths, all point in the expected direction from the genetic point of view. The children of first cousins, who are the most inbred compared to the children of second cousins and unrelated parents, show the most detrimental effects on child mortality. Second cousins are less related than first cousins and show less effects of inbreeding or inbreeding depression.

Why should an increase in homozygosity be detrimental or lead to inbreeding depression? First of all, increased homozygosity will increase the chance of detrimental recessive genes, which in effect were hidden or masked in the heterozygote, being expressed (Table 3). As the frequency of the recessive gene, q , goes down, the impact of mating a relative goes up. If a gene has a frequency of 1 in a 1000, for example, the chance of getting a homozygous recessive child is $(1/1000)^2$ or 1 in a million. In matings of second cousins there will be 16.6 times as many affected children compared to 63.4 times as many for first cousin matings and 250.8 times as many for matings between full sibs.

What is actually known concerning inbreeding and its effects in natural populations? VERY LITTLE!!! Even the most basic question of knowing if inbreeding has taken place or is taking place is a most difficult one. One might attempt direct field observations to identify all breeding pairs. As you might well imagine behavioral observations of this sort are difficult at best, and for practical reasons, may be impossible for many species. Nevertheless, this approach has been used with good results for some species including Greenwood et al. (1978) for Great Tits and Packer (1979) for anubis baboons.

Actually, one might be more concerned with determining the genetic results of inbreeding - increased homozygosity or low heterozygosity or loss of genetic variance. Here is where the use of electrophoresis has assumed such a major role.

Theoretical considerations have led us to think automatically that populations with small effective population sizes have low levels of genetic variation when compared to populations with large numbers. However, the relationship between population size and the amount of genetic variation may not necessarily be a simple or direct one.

An interesting paper was published late last year by Parker and associates dealing with genetic variation in mallard ducks. Although there was a large and nearly panmictic population with high dispersal abilities, there was little genetic variation. The ducks showed low variability both in individual heterozygosity and in the proportion of polymorphic loci. The authors examined 20 loci and found only 4 (20%) to be polymorphic. Heterozygosity was only 2.7 percent in the population.

There are few studies on the effects of inbreeding in natural populations. Perhaps the first detailed evidence of inbreeding depression

in a natural population is provided in the study by Greenwood et al. (1978) on great tits near Oxford, England. During the period from 1964-1975 a total of 1,000 matings were observed of which 16 (1.6%) were examples of inbreeding. The 16 matings included 7 mother-son, 9 brother-sister, and 1 aunt-nephew. Laboratory studies have indicated that the timing of reproduction, litter size, infant or fledging mortality, and offspring viability or fertility may all be affected adversely by inbreeding. In the population of great tits there was no evidence that inbreeding pairs produced smaller clutches but nestling mortality was higher for the inbreeders than for the outbreeders, 27.7 percent versus 16.2 percent. The effect of 30 percent is quite similar to that obtained in similar laboratory studies on birds and other organisms.

For mammals there are only limited observations on the effects of inbreeding. These are usually anecdotal in nature as in the study by Packer on anubis baboons (Packer 1979). In these baboons, typically, the males transfer out of one troop into another troop and such behavior is regarded as an example of animals avoiding inbreeding by not staying in their natal troop. It still remains to be determined if the baboons are actually avoiding inbreeding. Nevertheless, Packer did have observations on one male who had bred related females. There seemed to be an effect on infant mortality as only four out of eight inbred progeny survived more than one month compared to 32 out of 38 offspring from matings of transferred males who had mated females from another troop. Packer's behavioral observations indicated that females seemed to prefer "transferred" males and, thus, there appeared to be a genuine tendency for the females to avoid mating related males.

With the current emphasis on Sociobiology one sees a lot of recent papers dealing with inbreeding avoidance being exhibited by many species. Zoologists tend to look for behavioral mechanisms of outbreeding. Are there conditions under which inbreeding might actually increase fitness? Smith (1979) has examined this question in an interesting paper entitled "On selection for inbreeding in polygynous animals". If inbreeding is selected against because it leads to increased homozygosity, then so should small population size, but the range of effective population size for many mammals is 10-100. If inbreeding increases inclusive fitness, it should be selected for. If the costs of inbreeding depression are less than the costs of dispersing, etc., then it should occur. Inbreeding or incest is well documented in some cooperative hunters such as the wolf and coyote. This, cooperation among kin in an inbred unit may outweigh the cost of inbreeding depression. All of this is further complicated by the fact that inbreeding depression is not a fixed amount but may vary depending on the reproductive history of the population and the nature of the selective forces acting upon it.

In closing, I would like to call your attention to a recent paper by Ryman et al. (1981) which appeared late last year in OIKOS. The paper examined the genetic implications of various wildlife management programs. Using stochastic computer simulations, the study estimated the effects of

various hunting policies for moose and white-tailed deer on effective population size and generation interval. Time does not permit me to go into much detail, but for the moose the different hunting regimes caused the generation intervals to range from 4.2 to 9.9 years and ratio of effective population size to actual population size to vary from 0.2 to 0.4. What does it mean? The amount of genetic variation in a population can be influenced considerably in relatively short periods of time by the type of management.

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Table 1. Heterozygosity and Polymorphic Loci in Mammals

Species	Number of loci	Mean Heterozygosity	Proportion of Polymorphic Loci	References
Man	74	0.009	0.31	Nei & Roychoudhury (1974)
Northern Elephant Seal	24	0.000	0.00	Bonnell & Selander (1974)
White-tailed Deer	28	0.10	0.32	Manlove et al. (1976)
Moose	23	0.0006	0.04	Ryman et al. (1977)
Pika	26	0.011	0.15	Glover et al. (1977)
Elk	24	0.012	0.04	Cameron & Vyse (1978)
Raccoon	12	0.014	0.42	Beck & Kennedy (1980)
Sika Deer	19	0.000	0.00	Feldhamer et al. (1982)

Table 2. Effects of Inbreeding in a Japanese Population from Hiroshima (data from Schull and Neel, 1965).

	Relationship of Parents		
	Unrelated	Second Cousins	First Cousins
Number of Livebirths	4089	722	1,651
Number of Infant Deaths (prior to on year of age)	145	32	101
Per Cent of Infant Deaths	3.6	4.4	6.1

Table 3. The Factor Increase in the Frequency of Homozygous Recessives with Matings of Related Parents Relative to Unrelated Parents

Frequency of Recessive Gene, q	Relationship of Parents		
	Second Cousins	First Cousins	Full Sibs
0.1	1.1	1.6	3.2
0.01	2.5	7.2	25.8
0.001	16.6	63.4	250.8

Factor Increase values (F.I.) are obtained from the expression:

$$F.I. = \frac{fq + (1-f)q^2}{q^2}$$

where f is the coefficient of inbreeding of the children (second cousins, $f = 1/64$; first cousins, $f = 1/16$; full sibs, $f = 1/4$). and q is the frequency of the recessive gene.

CONFERENCE DISCUSSION

Q. Could we get information from the animal husbandry people on inbreeding of domestic sheep herds and how could we get information for wild sheep on inbreeding coefficients?

Ans. A couple of good references for domestic sheep are Morley (1954) and Lamberson et al. (1982). There are no data on inbreeding coefficients for wild sheep. If one had accurate estimates of the effective population size in a herd over a number of years and a good knowledge of the behavior of the species, one might be able to come up with a reasonable estimate.

Commentary. In the late 60's and the early 70's, we had a situation in sheep population in Canada and the question arose - how many? How many should we include in a herd? The number of animals never exceeded 80. 1. The degree of heterozygosity was related to heterosis, and the degree of heterosis was related to dominant individuals in that group. All the formulas you have showed assume random mating, a classical assumption. Under natural conditions, if there is a relationship between heterosis and dominance, the largest animals breed. 2. The population that is preyed upon showed a higher degree of fitness due to heterosis alone, you will then have a very heavy selection against heterozygotes which means even in a tiny population, you maintain a high degree of heterozygosity and therefore genetic variants. Obviously, other mechanisms are working; there is improvement of genetic stocks.

Comment: If the population effective size appears to be really small or the population appears to be isolated, it doesn't take much in the way of immigration, one animal coming into that population, to keep genetic variation reasonably high.

Q. Occasional immigration in a population will make a difference. Are we talking about 1 in a generation or 1 in 10 generation or 1 per 100 generation?

Ans. Normally used is 1 in a generation.