

Lecture 16 – Genetic Diversity and Population Viability

I. Initial definitions:

A. An *allele* is a DNA variant that occupies a given *locus* (position) on a chromosome (plural of locus is 'loci'). A locus with more than one allele is said to be *polymorphic*. Since mammals are diploid, we have two alleles at each nuclear locus.

We'll track allele frequencies in a population. They are calculated as the total number of copies of an allele divided by (for mammals) $2N$. 13 samples, 26 gene copies:

frequency of *A*: $14/26 = 0.53$

frequency of *a*: $1.00 - 0.53 = 0.47$

B. Genetic diversity is most commonly indexed by Heterozygosity.

Individual H_I - proportion of loci at which an individual is heterozygous. $2/4$: $H_I = 0.5$

Population H_P - average heterozygosity of an individual in a population. $6/13$: $H_P = 0.46$

I've shown the H_P for a single locus, usually we have data for several loci and we average across loci.

The second definition, H_P , is often the value of interest, especially in studies of rare and endangered species, because it is an index of the genetic diversity within that group.

II. Genetic diversity as a factor influencing population viability.

1. Population genetics theory predicts that a population bottleneck (where the population is reduced to just a few individuals) will result in loss of genetic diversity within a population.

This loss of genetic variation is the result of:

Sampling Effect - The chance of a small sample containing all variants is small.

Genetic drift - Random loss of low frequency alleles will occur faster in small pops.

Inbreeding - When a population is small, most of the individuals will be related to some degree.

Furthermore, this reduction in genetic diversity will be more pronounced the longer the population remains very small, that is, if the bottleneck is extended for many generations.

Classic mammalian examples:

- a. *Mirounga angustirostis*, northern elephant seal, experienced a bottleneck during the 1800's; some estimates are that fewer than 30 animals survived. Allozyme studies revealed heterozygosity values of zero. In the southern elephant seal *M. leonia*, there are normal values, ca. 4% (Reviewed in Sanvito et al. 2013. Conservation Genet. Resour. 5:93). Populations have rebounded, but genetic diversity remains very low (Abadio-Cordoso et al. 2017).
- b. *Acinonyx*, cheetah, experienced non-anthropogenic bottlenecks 10,000 years ago. There is virtually no genetic variation in cheetahs. 155 allozyme loci exhibited no heterozygosity. Skin grafts are accepted from unrelated individuals suggesting that MHC loci are monomorphic. This is incredible because these loci are usually under strong selection to maintain polymorphism (see e.g., Terrell et al. 2016. Biol. Conserv. 200:192.).

A common observation is that many rare and endangered species exhibit very low levels of genetic diversity.

2. The role of genetic diversity in assessing population viability.

The classical view in conservation is that population viability is intimately linked with genetic diversity, especially as measured by heterozygosity.

The idea is that genetically homogeneous species will:

- a) be very susceptible to disease and parasites.

Gibson (2021 – pdf on course website) reviews the association between genetic diversity and disease susceptibility.

The well-publicized spread of and extremely high mortality of Devil Facial Tumor Disease (DFTD) in *Sarcophilus* was probably permitted by the low levels of genetic diversity that characterized pre-disease populations (Miller et al. 2011. PNAS 108.).

- b) be subject to inbreeding depression; this is well documented in lab studies.

Decrease in fitness associated with mating with close relatives.

manifest as - decreased growth rates
 - lower survivorship to maturity
 - decreased sperm viability.
 - fluctuating asymmetry - breakdown in bilateral symmetry
 - decreased ability to raise offspring (e.g., Huisman et al. 2016. PNAS, 113:3585 for *Cervus elephas*).

c) experience decreased evolutionary potential.

Because rapid adaptation can only occur if there is standing genetic variation, low levels of variation will impinge on the ability of a population to adapt to future environmental fluctuations.

Even though *Sarcophilus* have reduced genetic diversity, they have retained alleles that confer resistance and these seem to be sweeping through post-disease populations (Epstein et al. 2016. Nature Comm. 7.).

Because of these issues, biologists spend a great deal of time and effort trying to maintain genetic variability, through (for example) maintenance of dispersal corridors for gene flow (to maintain connectivity), based on the **50/500** rule, and attempts of genetic rescue.

For example, the Florida panther (*Puma concolor coryi*) had been reduced to ca. 30 individuals, but Culver et al. (2008. Animal Conservation, 11:104), in a population genetic study, estimated that there were as few as 6 breeding individuals. In the early 1990's, these were exhibiting rather extreme inbreeding depression (e.g., low sperm count, malformed sperm, abdominal testes, heart deformations). In 1995, eight females from Texas were introduced to the population in an attempt to inject greater genetic diversity into the population. It seems to have been successful. Hybrid kittens have a 3-fold higher survival rate than the purebred kits (Pimm et al., 2006; Animal Conservation) and by 2010 the population had grown to 100 - 180 individuals (Johnson et al., 2010. Science, 329:1641). It's still growing and it's becoming clearer that this can be taken as a success story for genetic rescue (Hostetler et al. 2012. J. Animal/ Ecol. 82:608; Ochoa. 2017. Dissertation. <http://arizona.openrepository.com/arizona/handle/10150/624483>).

A relatively recent meta-analysis of >150 cases suggests that genetic rescue is routinely beneficial (Frankham 2015. Mol. Ecol. 24:2610).

So, the notion that genetic diversity is important for population viability has become central to conservation genetics.

3) Nevertheless, the classical view has been challenged consistently on several grounds, so there are some caveats:

a) Low levels of variability may not lead to extinction (e.g., Reed 2010. Animal Conserv., 13:448).

The poster children of low genetic diversity are persisting.

In cheetahs, genetic diversity has been low for 10,000 years and the species hasn't gone extinct.

“The most notable and still poorly understood feature of cheetah evolutionary history is how the cheetah has persisted in spite of remarkably low levels of genetic variation.” (Schmidt-Kuntzel et al. 2018).

Similarly, elephant seals numbers have rebounded in the face of low genetic diversity (Abadia-Cardosa et al. 2017).

- b) Furthermore, some species can withstand a great deal of inbreeding without showing any sign of inbreeding depression (see Rall et al. 1998, for an ageing review).

Canis lupus - and other canids such as the bush dog *Speothos*.

Leontopithecus rosalia (Golden lion tamarin) exhibits great deal of inbreeding with little evidence of inbreeding depression.

- c) Inbreeding can increase the fitness of a population in the long run if population survives a bout of inbreeding. This can happen via two avenues.

- i. It is possible to increase the variation available to respond to selection, especially for polygenic traits, traits such as height that are governed by several genes.

$$V_P = V_G + V_E + V_{G \times E}.$$

$$V_G = V_A + V_{Epi} + V_D.$$

This has been demonstrated empirically with Houseflies, but the theory has been developed that applies to all organisms, including mammals and involves conversion of non-additive genetic variation (e.g., epistasis and dominance effects) to additive genetic variation. This increases heritability and permits adaptive response to selection. See papers on course website: Naciri-Graven & Goudet (2003) and Mularo et al. (2022).

- ii. Decrease the genetic load.

Genetic Load - The cumulative fitness-decreasing effect of deleterious recessive alleles that can't be purged by selection. These hide from selection in heterozygotes.

If a deleterious allele is recessive, its effects won't be manifest in the heterozygote, Aa.

In an out-bred population, most of the deleterious alleles will be present in heterozygotes and won't be removed by selection.

In inbred populations, deleterious recessive alleles will be expressed more frequently as homozygotes, and therefore can be purged by selection. This will decrease the genetic load within a group.

Population that survives a bottleneck may be more viable than the original pre-bottleneck population (Garcia-Dorado. 2015. Heredity, 115).

Example – *Panthera tigris* populations in India (Khan et al. 2021. PNAS, 118).

- d) The alleles that are almost always lost are very low frequency alleles that are likely to be lost due to drift anyway; future adaptation is not likely to be compromised.
- e) In even the most genetically depauperate species, the cheetah, the danger of extinction is much higher from non-genetic threats such as habitat destruction (Purchase et al. 2007). Conservation dollars may be better-spent habitat preservation than on the preservation of genetic variability.

A similar example has been demonstrated in arctic foxes, *Vulpes (Alopex) lagopus*.

Some of these arguments are compelling, especially because many of them have been made by theoretical population geneticists.

A couple years ago a pair of papers encapsulated these two different views:

Teixeira and Huber. 2021. The inflated significance of neutral genetic diversity in conservation genetics. *PNAS*, 18:10. e2015096118.

DeWoody et al. 2021. The long-standing significance of genetic diversity in conservation. *Molecular Ecology*, 30:4141-4154.

At the very least, these arguments should cause us to consider very carefully the way we allocate limited conservation resources, and not always assume any single issue (genetics, demography, habitat, etc.) will *a priori* be the most important to address.