

BREEDING STRATEGIES & GENETIC CONSEQUENCES

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PREFACE

The non random mating of genetic relatives will increase the frequency of homozygotes, and decrease the frequency of heterozygotes. Breeders utilizing linebreeding cause genotype frequencies to change, thus are altering the proportions of phenotypes. We therefore increase the speed of natural selection on a captive population. What may take Mother Nature hundreds of years can be accomplished in several. By increasing the occurrence of homozygotes, linebreeding can expose any deleterious recessive alleles for culling. In both wild and domestic populations, any decline in size can result in genetic drift, which can lead to inbreeding depression. Inbred offspring can be substandard to outbred offspring in survival & reproduction. As a result, individuals homozygous for deleterious recessive alleles increase, and deleterious mutations become fixed due to such drift. In the wild such animals face extinction. In domestic populations they should face ruthless culling by the breeder.

Genotype is usually inferred from physical appearance referred to as phenotype. In simple terms phenotype in domestic breeds is no more than the product of coded genes resulting from a combination of genotype. It is influenced over time by environment and breeder selection. Individuals carry two alleles, which may be identical (homozygous) or different (heterozygous) for each gene. Breeds with multiple allele types at a gene are polymorphic, those with only one type of allele are monomorphic.

DEFINITIONS

Alleles - copies of genes inherited from both parents.

Effective Population Size - number of individuals required to have the same amount of random genetic drift as the actual population; this is often much smaller than the true population.

Founder Effect – founded by a small core group of individuals representing a smaller proportion of the source genetics.

Genetic Bottleneck – the reduction of a breed to very low numbers and subsequent loss of variation from rarer alleles.

Genetic Drift - gene frequencies within flocks that change over time due to random mutation; smaller or linebred flocks can show an increase and subsequent generations retain smaller portions than predecessors.

Gene Flow - gene frequencies in a breed that change over time with the aid of genetic exchange among flocks.

Genetic Potential - the greater the genetic diversity, the greater potential to respond to changing environmental conditions.

Gene Pool – A breed's genetic variation consisting of alleles present and their frequencies across members.

Heterozygosity - percentage of gene loci at which the average individual contains alternative forms of alleles.

Homozygosity - percentage of gene loci at which the average individual contains the same forms of alleles.

Inbreeding Depression – a measure of the response of a breed to genetic selection; loss of robustness due to deleterious recessive alleles expressed by reproduction among closely related individuals.

Locus - position occupied by a particular gene or one of its alleles.

Monomorphic - both copies of the allele are always the same, indicating no variation at a particular locus.

Outbreeding Depression - loss of robustness due to reproduction among very dissimilar individuals.

Polymorphism - the percentage of the genes with multiple types of alleles at a given locus; characterized as a measurement of the genetic capacity of a breed to adapt to change.

POPULATION SIZE

A breed's genetic diversity is not determined by Overall Population Size (OPS), but by the size of the genetically effective population. The latter is referred to as Effective Population Size (EPS). Genetically EPS is much smaller than OPS because of multiple factors which include: varied reproductive success of females, unbalanced sex ratios, or severe population crashes.

An accepted rule of thumb states: deleterious mutations will lead to a reduction in robustness of about 1% each generation without additional influence either positive or

negative. When trying to counteract the effects of such mutations, the first concern at a breed level is determining EPS. That is, at what number would a group of animals contribute equal genetics vs. a theoretical infinite population, with further numbers failing to increase genetic contributions? In the Shetland world this number is rather hard to define for multiple reasons encompassing fleece traits, colors & patterns. In a typical breed of limited color and variation such as Barbados Blackbelly EPS is in the neighborhood of several hundred animals in an OPS of 1000 potential breeders.

Population diversity can be defined by 3 major components:

- Intra-individual (heterozygosity);
- Among individuals within a population;
- Among populations

A breed can lose much genetic diversity during population crashes resulting from many factors not limited to disease or market related economic downtrends. If numbers are recovered quickly, so will EPS. In a small population inbreeding depression and genetic drift are potential problems resulting in the loss of often important genetic diversity contained by rare alleles. Sustained low EPS can dramatically reduce genetic diversity with each passing generation. It is thought such lows generate the mutations needed by island populations through genetic drift allowing for their survival without genetic migration from others.

Ironically, successful breeding programs can actually reduce EPS under certain circumstances. For example: If OPS in a region is small and a high percentage of young from these flocks are retained, they can contribute a large percentage of replacements to the total breeding population. In such a case EPS may decrease.

A Minimum Viable Population (MVP) is one that permits long term survival of a breed or species in current environmental conditions. MVP must be sufficient to protect against extinction resulting from genetic, demographic or environmental factors. Studies indicate most mammal (breeds or species) have little genetic variation vs. plants resulting from small or static inbred populations. Only infusions of genes from migration or exchange in combination with mutation avoid serious inbreeding. For example: The entire population of African Cheetah is virtually identical in nuclear DNA, indicating the former population crashed severely. So identical in fact that skin grafts from one individual to another are not rejected. Inbreeding depression is normally characterized by lowered robustness or vigor. Heterosis or hybrid vigor tends to manifest by increases in size, growth rate, and such in F1 offspring of two inbred lines. Yet, the Cheetah passed through a serious bottleneck without extinction. Since that time many generations ago natural selection has eliminated the most deleterious genes arising from inbreeding depression by infant &/or adult mortality. Otherwise they simply would not have survived into modern times. Remember that evolution through natural selection only increases frequency of an allele in proportion to its benefit on survival. Breeders should select and cull on the same principles for flock fitness.

GENETIC ISOLATION

A domestic sheep breed usually exists as a group of flocks that may or may not experience gene flow in the form of breeder exchange. In established flocks exchange on a regional level comes from both male and female animals. Traditionally when greater distances are involved it primarily involves sires, either live rams or AI. The genetic variation or heterozygosity created in such cases can be grouped by local diversity within regional flocks. Breeders may create variability in the form of local genetic adaptation to conditions, which result from co-adapted gene complexes. These combinations of alleles at different loci after several generations can confer optimal levels of robustness on individual animals in static flocks. Introduction of outside genetics to any flock, while beneficial on many levels, can cause a loss of local adaptation and should be a concern to breeders. Such loss of vigor has come to be known as outbreeding depression. While its effects on domestic breeds is unknown to many farmers, biologists have long understood the process on rare & endangered wild species.

Reduced genetic diversity within small populations may result from:

1. Founder Effect
2. Genetic Drift
3. Demographic & Genetic Bottlenecks
4. Inbreeding Depression

1. Founder Effect

Founder Effect is a term used to describe negative impacts within a breed descended from a single or a several common ancestors. They appear many times in the background of the breed, typically possessing many traits breeders wish passed to offspring. Along with good genes, negative recessives associated with the desired genetics may manifest themselves in later generations in a homozygous state. In the wild Mother Nature will quickly remove such flaws. As breeders we must do the same with little emotion. Founder Effect can be easily studied in populations that occur on islands or in geographic isolation.

A founder effect can arise when a breed is perpetuated by only a few foundation animals. In the extreme case, a single viable female might be utilized to resurrect or establish a breed in a new location, and may result in a type of genetic bottleneck. For example: After the foundation flock of Castle Milk Moorits was dispersed, they were nearly lost and revived from a single ram and six ewes. Today the population continues to increase with additional interest by breeders in the UK. As a result, the new population may be distinctively different in phenotype from the parent population within several generations. Founder effects are common in many species found on island ecologies, yet left to their own means sheep on islands seem to adapt via genetic drift and thrive in such conditions. Culling is brutally enforced by Mother Nature during peaks in population, followed by crashes resulting from disease &/or lack of nutrition.

2. Genetic Drift

Isolated flocks and island populations are also affected by genetic drift. This random change in genes can affect frequency of type. Example: A flock utilizing only moorit sires bb in genotype and five black ewes BB in genotype. To keep things simple we assume each ewe is replaced by her daughter in any given year. 1st year offspring will be black and Bb in genotype. From 2nd year on offspring will be either Bb or bb . Each year the number of Bb will decrease while bb will increase. At some point in the near future all animals will be bb and the gene for black will have been lost to the population via genetic drift. Extreme loss in genetic variability often found in island populations or isolated flocks can create adaptation to a specific environment. Yet it also can reduce the ability to adapt to changes in either environment or market. This is commonly seen with breeds geared towards a limited market.

Genetic variation in Sheep is jointly determined by breeder selection and naturally occurring genetic drift recognized as mutation. The relative importance of genetic drift found in low numbers of a minor breed is high. The negative effects of recessive deleterious alleles may become much more prevalent, resulting in a fixed trait. The loss of any positive or negative allele is most likely to occur in a small gene pool. The reduction in the number of forms of an allele in the extreme case leads to a monomorphic state where only one form exists.

Consequences resulting from a loss of genetic variation include inbreeding depression &/or the inability of a breed to adapt and evolve to changing conditions of its environment. While inbreeding depression has been accepted within domestic breeds, scientists have had a hard time substantiating it in most wild or feral populations. Indicating the underlying causes are yet to be fully understood or are breeder enhanced by poor mating selection and retention.

Phenotypic characteristics (appearance) are often used to divide animals into breeds & species. While there is great diversity across species, there may be limited genetic variation within any given species. Domestication of wild species has led to many breeds and increased "within species variation". Sheep breeds are utilized in many environments, resulting in selection for different characteristics in different locations. We have selected animals for specific uses (draft, milk, meat, fiber). These selections over time in large flocks can lead to indirect selection for disease resistance and behavior traits.

3. Demographic & Genetic Bottlenecks

Small island populations of related Northern short tail sheep in Europe and feral breeds on islands around the world present opportunities for examining the bottleneck effects of both recent and prolonged inbreeding. From these populations isolation has created an impressive diversification of type with the aid of genetic drift. On a smaller scale isolation by distance has had a similar effect on minor breeds over the last several decades in North America. As a rule small populations will not persist without added

genetics via migration (breeder exchange) or mutation from genetic drift. Breeder exchange between flocks allows several small flocks to function as a collective group, in this case a breed.

Many sires become popular because they consistently pass homozygous traits to offspring. This type of sire is called prepotent. In essence, he doesn't have two different copies of desired alleles to pass on. Breeding "like to like" is a time-honored system in the pedigree world of livestock. The number of different alleles available in the gene pool decreases creating consistency. Genetic bottlenecks can occur for a number of reasons. The use of a popular sire via Artificial Insemination (AI) is only one factor reducing a gene pool. Another cause of bottlenecking is a reduction in numbers via disease or market collapse. Remaining members are bred together in an effort to continue the breed.

4. Inbreeding Depression

Inbreeding depression occurs when deleterious recessive alleles become homozygous. The condition can manifest as reduced fertility, birth weights, growth, age of maturity, loss of maternal traits, or structural deformities to name but a few. In a wild setting inbreeding reduces recessive heterozygous lethal alleles by elimination. As they are expressed in a homozygous state mortality increases. In a domestic setting it is up to the breeder to perform the task by culling of undesired genetics &/or breeding around it with the aid of genetic testing.

Problems associated with inbreeding depression are found in many domestic breeds and captive wild populations. Yet, the outstanding reproductive successes of many programs indicate poor choices are potentially responsible. In Example: The Austrian Kärntner Brillenschaf is an endangered domestic sheep breed. They descended from local sheep crossbred with two Italian native breeds. As common with many native breeds it was replaced by more market driven varieties during the 20th century. From a low of 17 ewes and six rams the total pedigree population has grown to over 1500. Current genetic diversity within the breed determined by DNA micro satellite markers compares favorably to diversity with other breeds showing a high number of alleles per locus.

Heterozygotes often show enhanced vigor vs. homozygotes resulting in balanced polymorphisms within a breed. In some instances during the formation of homozygotes formerly recessive alleles demonstrating less deleterious effects become lethal when fully expressed. These recessives may have been masked by over dominance in heterozygotes. Over dominance results when the positive benefit of a heterozygote is higher than a homozygote.

BREED MAINTENANCE

In a wild setting inbreeding and genetic drift are countered by natural movement (migration) often inhibited by island demographics. In domestic populations this is accomplished by breeder exchange. In small populations it takes as little as one individual per generation to maintain genetic diversity.

Natural selection (evolution) causes changes in wild populations over many generations. This often results in many sub species originating from the founder population. Domestication enhances and speeds up the process via artificial selection dictated by rapid change. Sheep breeds can be sub-divided into many different phenotypes when a breeder selects against certain traits (alleles), while retaining other desirable alleles. The primary limiting factors are interested breeders and desired flock size. Example: In my flock the allele for spotting is selected against by phenotype. Even though the allele probably still exists in genotype, it is not expressed and there are no spotted individuals evident in the flock. A conscious effort has been made to suppress this particular allele in the gene pool.

Can breeder selection accumulate bad genes with long term repercussions for a breed? Yes, on the farm we can use the following example: breed longevity can be quickly influenced by the killing off of breeders at an early age over successive generations. Ever wonder why most modern commercial breeds lack the longevity found in primitive or feral populations? They don't keep breeding age ewes around extended periods of time vs. more primitive breeds, resulting in a loss of alleles for longevity. This would seem to be a relevant concern to modern commercial breeders since acquisition or maintenance costs from birth to breeding age can be significant. Yet they continue to select for growth rate & size over slower maturity & longevity resulting in a shorter production life. If this one trait is so easily manipulated, what others are we unknowingly influencing to hinder a breed's ability to thrive under stress-free conditions?

Can good environments accumulate bad genes with long term beneficial results to a breed? Again yes, recent studies indicate inbred Soay Sheep are more susceptible to internal parasitism during peaks in population cycles. Naturally occurring periodic winter die offs of individuals highly parasitized by gastrointestinal nematodes guarantees survival of those with genetic diversity. Thus those with the least resistance to parasites are sacrificed to keep genetic variation intact by Mother Nature. In the wild inbreeding is not all bad if it occurs over multiple generations permitting selection to purge any recessive deleterious alleles that become fixed.

Breeders of Sheep must continue the role of Mother Nature in the barnyard. Combined genetic diversity results from both intra and inter population contributions. Infusions of new genes will reduce inter population genetics in a flock, thus reducing potential inbreeding. On the flip side the short term benefit derived from any infusion could be negated in future generations if it destroys co-adapted gene complexes geared towards survival. Breeders interested in conservation tend to focus on OPS. A problem arises if we choose OPS as the chief unit of conservation, since wild and domestic populations

rarely exist as one interbreeding group. Social hierarchy in the wild and breeder exchange in domestics should create loosely-connected genetic “unrelatedness” with gene flow to other groups.

Recessive and mildly deleterious mutations are common. When located close to a desired allele they may inadvertently be selected. When a selected trait is finally brought to the forefront as a homozygote within a breed, it may be linked with harmful recessives. Selection can remove genes with deleterious recessive effects linked to genes with beneficial dominance. Linebreeding to a degree can purge deleterious recessive alleles appearing in homozygotes through efficient culling. Selection cannot remove the effects of inbreeding if it is due to over dominance.

OUTBREEDING ENHANCEMENT

Heterosis or outbreeding enhancement is the opposite of inbreeding depression and is referred to as hybrid vigor. Hybrid vigor is a reversal of inbreeding depression. It has been described as “the masking of recessives through crossing of unrelated genetics.” In domestics regional populations have dissimilar recessive deleterious alleles in their genetic composition. Matings from two populations may create offspring that are heterozygous for them. The F1 hybrids benefit as any deleterious alleles are masked by heterosis in a recessive state. In successive generations a higher measure of robustness is evident in the nucleus population, but Mendelian genetics will again turn out homozygous deleterious alleles in certain animals. This principle will hold true in most situations when limited to alleles on a single locus.

OUTBREEDING DEPRESSION

Many breeders claim genetic diversity is a good thing, touting how the benefits of hybrid vigor will negate inbreeding depression. However, co-adaptation in the form of gene complexes on multiple loci evolve to create environmental strengths in isolated flocks. When such sub-populations are crossed the overall robustness of offspring can be greatly reduced. Such a situation is referred to as outbreeding depression. It has been proven outbreeding depression could have a negative impact if individuals from two well-adapted environments are bred to each other

Outbreeding depression can result in two ways. In the first and most complex form locally coadapted gene complexes can be disrupted by breedings to distant or unrelated flocks. This is due to nonadditive gene interaction of the same genes from different genetic backgrounds caused by a breakdown of biochemical or physiological compatibilities. Coadapted gene complexes are the least understood in most cases.

In the second form offspring between parents from isolated or unrelated flocks may have phenotypes that are not suited for either location. This is common when a breeder brings in animals from a long time breeder located in a dissimilar climate. Animals bred

multi generation in a mild temperate climate with easy access to water and lush grass are tailored to this setting. On the other hand animals bred in an arid climate with sparse grasses and limited water are adapted to their own location. Crossing of the two types may create individuals with transitional body characteristics not suited to either place.

As if things were not intricate enough both outbreeding depression and outbreeding enhancement can occur concurrently in a flock (or individuals) utilizing external genetics. If flocks or populations have not split long enough to acquire divergent, gene complexes it is unlikely any outbreeding depression will occur. If North American Shetlands are bred to UK stock differences may be visible, to Foula or North Ronaldsey stocks even more probable. The degree of potential outbreeding enhancement cannot be predicted and you must test breed individuals to evaluate benefits. When I bring in a new ram he rarely is mated to the majority of females. Only after a season or two will he move into a senior role. The same goes for homebred junior sires utilized in linebreeding. They must first pass on beneficial results via limited test breeding. The effects of outbreeding depression may not manifest for several generations. Initial F1 offspring may indicate a beneficial increase in overall strengthes. After several generations when the genetics have had a chance to “recombine” negative effects may become perceptible. In these cases only time and breeder choice can eradicate problems by means of concentrated culling.

TESTING

DNA analysis has quickly become one of the techniques most often used by breeders to scrutinize breed genetics, and by associations to substantiate purity of breed via fingerprinting of sires. Two available options are: nuclear DNA and mitochondrial mtDNA. As expected current testing of many breeds indicates a greater diversity between the flocks than within the flocks.

All Sheep obtain nuclear DNA from both parents. A highly useful nuclear DNA marker assesses genetic variability through micro satellite repeats. These repeats are currently used for inter-population comparisons to detect variation in genetically distant populations, and assess relatedness between individuals.

Extracted mitochondrial DNA (mtDNA) allows us to map the genes on the mtDNA molecule. Animals having the same map were related in the past. Mitochondrial DNA does not recombine with other DNA as does nuclear DNA, thus allowing for a static means of identification. MtDNA only comes from your mother and passes via a maternal line. While male offspring possess mtDNA it is not passed to offspring. Even though passed mtDNA is indistinguishable from generation to generation it is modified by mutation over extreme periods of time. Therefore, it is possible for Sheep deriving from the same source to have different genes when separated for any length of time.

SUMMARY

As a breeder we can manipulate gene frequencies by non random mating selection in conjunction with: genetic drift, mutation, or the domestic version of population migration in the form of animal acquisitions. It must be remembered that genetic variation can only come about via mutation in existing genes.

Through mutation deleterious alleles constantly arise, and are always present at low frequencies. At a given locus, some alleles may confer higher levels of survivability on an individual than other alleles. These other alleles may be rare deleterious recessives, which in homozygous form reduce robustness.

With the aid of modern technology sheep breeds can be characterized for phenotype and genotype. Genetic distance can be tracked by gene mapping to determine the degree of relatedness between established flocks. While phenotypic performance of various traits should be evaluated for adaptation to individual breeder environment.

Hopefully this article illustrates the importance of understanding sheep population genetics in planning effective management of small flocks. Inbreeding, loss of variation through drift or overuse of widespread sires, genetic differentiation among regional populations, etc. are worthy of contemplation in the overall management scheme of any breed at the flock level. After all, how well your flock succeeds will determine how well it meshes into the overall fabric of the breed as a whole.

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