Analysis of Genetic Variation in the Mountain Pleasure Horse

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The preservation of genetic variability is the key tenant of biological conservation. Within domestic animal breeds, loss of genetic variability is usually due to small population size (genetic drift) and/or inbreeding. Both processes can lead to the accumulation of deleterious recessive alleles and result in a loss of fitness and vigor or specific genetic diseases. Additionally, intense selection for specific characteristics can lead to genetic fixation of the genes in the region surrounding the area coding for the trait. Again, this can lead to the increase of deleterious genes if such genes are in this chromosomal region. An understanding of how genetic variation within a breed is changing can lead to the development of management strategies to reduce the rate of loss of variation. This study was undertaken to examine levels of genetic variation (estimated as observed heterozygosity) over time in the Mountain Pleasure horse breed.

The Mountain Pleasure Horses have a landrace origin. They are a part of a greater group of Appalachian mountain horses which have contributed to the development of a number of important gaited American breeds including the American Saddlebred, Tennessee Walking Horse, Mountain Pleasure Horse, and Rocky Mountain Horse.

Mountain Pleasure Horses trace their ancestry back more than 170 years. Their lineage is diverse with a geographic area of origin centered on the area of West Liberty, Kentucky. The Mountain Pleasure Horse Association (MPHA) was founded in 1987 and officially organized in 1989 to conserve the land race origins of their breed.

MPHA stallion registry books were closed in 1991 with 100 registered stallions. MPHA mare registry books were closed the following year with 400 mares on the books. The MPHA registry books have remained closed since 1992.

In 1995 there were approximately 1,500 Mountain Pleasure Horses. The foal crop for that year was 150. By 2006 there were 2,828 Mountain Pleasure Horses, a two fold increase in herd size in ten years. As of 2006 the MPHA identified 1,798 mares and 973 stallions and geldings. MPHA foal crop in 2006 was 57.

Genetic variability estimates were based upon data from Blood Typing. All typing was done at the Equine Parentage Testing and Research Laboratory at the University of Kentucky. To examine changes in genetic variation in the breed over time we calculated average variability levels of all individuals born in the same year (an age or year of birth cohort) and compared those to values for individuals born in different years. We were able to examine 23 cohorts covering the years 1979 through 2001. The 1979 cohort actually includes all individuals born in 1979 or before due to the small number of individuals from this time period that have been Blood Typed.

Genetic variation was calculated as observed heterozygosity (*Ho*) which is the actual number of loci that are heterozygous per individual. Only the 10 biochemical loci typed were used for this estimate because it is not possible to unambiguously determine the genotype of a blood group locus in all cases.

Ho was averaged over each year of birth cohort and regression analysis was used to determine the rate and direction of change in *Ho* through time. In addition to *Ho*, variability measures calculated for each cohort were expected heterozygosity (*He*) which is the proportion of heterozygous loci predicted based upon allele frequencies and Hardy-Weinberg Equilibrium Theory, effective number of alleles (*Ae*) which is a measure of allelic diversity, mean number of alleles (*Na*) which is the average number of alleles per locus, and *Fis* which is an estimate of inbreeding base upon a ratio of *Ho* to *He (*Fis=1-*Ho/He)*.

Results

Measure of genetic variation from Blood Typing data for each year of birth cohort is given in Table 1. For the Blood Typing data, there was no significant change in *Ho* through time. Also, there was no indication of differences in variation for any particular locus across cohorts.

Discussion

The Mountain Pleasure Horse breed has a small founding population size and has been maintained with a relatively small population size. From a genetic standpoint, what is important is what is termed effective population size (*Ne*) which basically is the number of unique genomes that contribute to the next generation. Within populations such as horses with long lives and complex population structure *Ne* is not an easy number to calculate. The primary factor involved in determining *Ne* is the ratio of breeding males to breeding females. In horses, as in most domestic species, the proportion of males to females is less than one which will lead to an estimate of *Ne* that is less (and often much less) than the census size. Based upon the simplest estimator of *Ne* which is 4Nm*Nf/(Nm+Nf) where Nm and Nf are the number of males an females, the effective population size of the Mountain Pleasure Horse in 1992 when the registry was closed was 320. This is a small number. Based upon the 2006 numbers of mares and stallions the *Ne* is 2,525. This is unquestionably an over estimate because it does not take into account factors such as degree of inbreeding or variance in reproductive contribution among individuals of the breed which also are significant factors related to effective population size. In fact, if the registry was completely closed in 1992, the *Ne* would not increase from 320 because no new genomes would have been added to the population. Despite this, mean *Ho* is near the mean for domestic horses based upon Blood Typing data (domestic horse mean *Ho* is 0.371 compared to 0.378 for the Mountain Pleasure Horse).

Due to the small population size of this breed, some loss of genetic variation would be expected. This clearly was not seen for the MP. *Ho* for each year of birth cohort fluctuated around the mean with no trend either for an increase or a decrease of *Ho*. The explanation for the lack of change in variability is not clear. The most likely reason is that the breed started off with relatively high genetic diversity and there simply has not been sufficient time for the reduction that would be expected due to the small population size to show up. However, if this is the case, it indicates that the breeding practices within this breed are very sound and that *Ne* has been maintained at near the maximum for the population size. Significant loss of variation can occur within 20 years as shown by a study of Standardbred horses. In this study both pacing and trotting segments of the breed showed loss of heterozygosity based upon Blood Typing data at rates of approximately 5% and 17%, respectively, for the two different gaits. The primary reason for this high rate of loss of genetic variation was a concentration on a very small number of stallions for breeding. Based upon the data available for the present analysis, the mean number of foals per MP stallion over the time period of the study was 4.87 (with a range of 1 to 121). This number is low enough to not have a significant effect on *Ne*. Continued monitoring of the Mountain Pleasure breed is recommended because the total population size is small. It would not take much of a change in breeding practices or selection intensity to effect a change in the preservation of genetic variability.

Table 1. Measures of genetic variability based upon Blood Typing of the Mountain Pleasure Horse.

YOB	*N*	*Ho*	*He*	*Fis*	*NA*	*Ae*
Mountain Pleasure Horse						
Fully registered						
1979	67	0.4060	0.3858	-0.05236	4.6	2.1541
1980	7	0.3629	0.3980	0.08819	2.6	1.9409
1981	15	0.3600	0.4113	0.12473	3.5	2.1645
1982	25	0.3772	0.3751	-0.00560	3.7	2.1630
1983	37	0.3793	0.3870	0.01990	4.4	2.2217
1984	37	0.4213	0.4201	-0.00286	4.7	2.3485
1985	47	0.3611	0.3846	0.06110	4.2	2.0653
1986	43	0.3873	0.3929	0.01425	4.2	2.1064
1987	54	0.3805	0.3874	0.01781	4.8	2.2419
1988	70	0.3422	0.3695	0.07388	4.6	2.2065
1989	72	0.3767	0.3728	-0.01046	4.3	2.1441
1990	83	0.3594	0.3767	0.04593	4.8	2.2278
1991	95	0.4024	0.3988	-0.00903	5.3	2.2523
1992	85	0.3866	0.3779	-0.02302	5.0	2.1732
1993	99	0.3509	0.3761	0.06700	4.8	2.2364
1994	104	0.3704	0.3762	0.01542	5.0	2.1330
1995	95	0.3878	0.3913	0.00894	4.8	2.2481
1996	106	0.3796	0.3884	0.02266	4.7	2.2918
1997	126	0.3683	0.3643	-0.01098	4.7	2.1717
1998	124	0.3631	0.3699	0.01838	5.0	2.2597
1999	110	0.3798	0.3754	-0.01172	4.6	2.2320
2000	814	0.3819	0.3795	-0.00632	5.9	2.2425
2001	398	0.3842	0.3830	-0.00313	5.5	2.2464

Figure 1. Plot of Blood Typing *Ho* vs Year of Birth for fully registered Mountain Pleasure horses.