学位論文の要旨

Abstract of Thesis

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学位論文題目 Title of Thesis（学位論文題目が英語の場合は和訳を付記）

Study on genetic diversity and characteristics of Kazakhstan original horses
（カザフスタン固有のウマにおける遺伝的多様性と特徴に関する研究）

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Horse is the domestic animal that have had the greatest impact on human civilization and history as a means of transportation, locomotion, and agriculture. They have also played an important role in warfare. Domestication of horse is believed to have occurred in Central Asia about 5,500 years ago. In particular, a region of current northern Kazakhstan is a most possible domestication center of horse where the wild horses called Tarpan inhabited in the Eurasian continent at that time, was first raised as domestic animals under human control for meat and milk production and means for transportation and locomotion. After that, the domesticated horses spread widely around various parts of the Eurasian continent to form local populations of horses. While current world wide horse populations are mainly come from modern horse breeds that have been established in European countries after Medieval Ages, the descendants of early domesticated horses have still been survived as local native horse populations. Therefore, genetic investigation of these native horse populations is valuable to know the process of horse domestication and spread. Particularly, since Central Asia including Kazakhstan is hypothesized as domestication center of horse, genetic investigation of native horse population in Kazakhstan is important to understand the origin, and process of domestication and spread of the horse.

In this study, haplotypes of mitochondrial DNA and Y-chromosome markers which can provide information for genetic features of maternal and paternal lineages, respectively, were investigated in the native horses and crossbred horse breed of Kazakhstan. In addition, since selective breeding for the traits of horse such as coat color and physical performance has been carried out in domestic horses after domestication, investigation of the genes associated with these traits could also provide useful information about breeding history of horse. Therefore, the allele frequencies of genes associated with these traits were also investigated in this study.

Firstly, haplotypes of mtDNA D-loop region in 50 horses of two different populations of Kazakhstan native horse, namely Adaeve and Zhabe horses were determined. These horses are local native horse of Kazakhstan that are used mainly for meat and milk production. Zhabe is relatively large horse being adapted for cold climate of northern region of Kazakhstan. Adaeve is middle size horse being adapted for dry climate of western region of Kazakhstan. As a result,

total 16 haplotypes with 21 segregated sites which fall into 10 out of the 17 major haplogroups in the horses were observed. Most of these haplotypes were ancient haplotypes survive in modern horse breeds and some of them are widely spread among various primitive breeds of Eurasian continent. These findings indicated high genetic diversity and unique composition of these Kazakhstan native horse population in maternal lineage. Next, four SNPs on Y chromosome in 9 male Adaeve and 9 Zhabe horses were investigated to classify the Y chromosomal haplotypes. The result indicates that 16 of the 18 males possess haplogroup 2 and two possess haplogroup 4 that are mainly present in European and Middle East type horses and no haplotypes observed in Asian native horse was observed. These findings indicate that Y chromosome of Adaeve and Zhabe horses originated from limited number of stallions that results in low genetic diversity of paternal lineage. These findings suggested that the Kazakhstan local horses have high genetic diversity with ancestral genetic features in maternal lineage but low genetic diversity with haplotypes different from those of other Asian native horses in paternal lineage.

Genotypes of the genes associated with coat color (*ASIP, MATP, MC1R, TBX3*) and body composition (*LCORL, MHGA2, LASP1, ZFAT*) were then investigated. As a result, the genotypes the coat color related genes indicated that there is horse with Dun coat colors know as ancestral coat color observed in wild horse, in addition to basic coat color, namely bay, black, and chestnuts. Also, the results indicated that considerable part of these horses have maintained the primitive ancestral feature of coat color represented by dorsal stripe. For body composition and locomotion traits, the minor alleles of *LCORL, ZFAT, LASP1* associated with bigger body size were present at low frequencies in the Adaeve and Zhabe populations, suggesting a possibility that these two populations have not been under strong selection pressure for particular body composition trait.

Next, haplotypes of mtDNA D-loop region and and Y chromosomal haplotype were investigated in 22 Kushum horses. Kushum is a relatively new breed of horses in Kazakhstan established in the middle of the 20th century through a cross between mares of Kazakhstan local horse and stallions of Thoroughbred, Trotter, and Russian Don breeds to supply military horses before World War II. As a result, total 10 haplotypes with 19 segregated sites were observed. Seven of these 10 haplotypes are same as previously reported haplotypes, which are ancient haplotypes survive in modern breeds and widely spread among various primitive breeds of Eurasian continent. These findings indicate that theses haplotypes are likely reflect high genetic diversity of local horse population. The results of genotyping four SNPs on Y chromosome indicate that the haplotypes of Kushum could be originated from Trotter and/or Russian Don. The observed mtDNA and Y chromosomal haplotype distributions are in concordant with the documented origins of this breed and with high genetic diversity in maternal lineage and low genetic diversity in paternal lineage.

Genotyping results of the genes associated with coat color (*ASIP, MATP, MC1R*) and locomotion traits (*MSTN,* and *DMRT3*) in the 22 Kushum horses indicated that the coat colors estimated from the genotypes are bay, chestnut and black suggesting that this breed has been selected to exclude the coat colors other than basic coat colors during past selective breeding. For the locomotion traits, allele frequencies of *MSTN* showed higher frequency of the allele associated with higher endurance performance and *DMRT3* gene showed presence of the allele associated with ambling gait in Kushum horse even frequency is lower than the allele associated

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with normal gait. The endurance performance for long distance locomotion and ambling gait might have been required for this breed as military horses. Therefore, the allele frequencies of the genes associated with locomotion traits might reflect such historical demand for the breeding of Kushum horse as military purpose.

As results of this study, at least 16 haplotypes of mitochondrial DNA were revealed to present in Kazakhstan native horses indicating extremely high maternal genetic diversity. Furthermore, a considerable part of these haplotypes were ancient type haplotypes that are observed in wild horse population before domestication. On the other hand, only 2two Y chromosomal haplotypes were observed in Kazakhstan native horses indicating low paternal diversity. Furthermore, these haplotypes are mainly present in European and Middle East type horses but not in Asian native horse. These high maternal and low paternal diversity of the native horses in Kazakhstan did not differ much from the those of Kushum horses established from cross between native horses and exotic breeds.

In general, worldwide horse population shows high genetic diversity in maternal lineage and low genetic diversity in paternal lineage. Therefore, it has been hypothesized that after the first horse domestication occurred in Central Asia, these domesticated horses spread around the various regions of Eurasian continent and then the stallions of domesticated horses were again crossed with mares of wild horse inhabiting these regions to form new local horse populations that resulted in high maternal diversity and relatively low paternal diversity of the current population of horse. However, recent finding of Y chromosomal haplotypes indicated that Asian native horse populations have Y chromosomal haplotypes that are different from those of European and Middle Eastern horse populations suggesting that Asian native horses have paternal origin different from European and Middle Eastern horse populations. On the premise that the genetic feature of current population of Kazakhstan local horse reflect those of ancestral early domesticated horse population, the present findings of the Y chromosomal haplotype of Kazakhstan native horses suggest that the Asian native horses did not originate from the early domestic horse population in Central Asia and, therefore have a different domestication center. The present findings of high genetic diversity of maternal lineage with ancestral type mtDNA haplotypes, low genetic diversity of paternal lineage with European and Middle East type Y chromosomal haplotypes and presence of ancestral type alleles of coat color gene could be important for understanding the process of domestication and spreading of horses in Eurasian continent.