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## Dr. Samantha A. Brooks Lab Research:

The horse occupies a unique place in our global culture and presents a fascinating opportunity for study. Having catalyzed the rise and fall of civilizations for thousands of years, the horse is now in a process of transition. Post industrial revolution, its role in human culture has adapted to one of icon and companion. This unique history has led to selection by man for an unusual and varied set of traits. Our research focuses on the use of genomics tools to study traits of health, conformation and performance. This work spans not only the mapping and identification of mutations causing traits of interest, but also the use of a variety of nucleic acid based techniques in genomic research. The following is an outline of some of our current projects.

- Several targeted studies are currently underway to identify the basis of Mendelian genetic diseases, as well as complex quantitative trait loci (QTL) for traits like behavior, body size and disease resistance. In particular, genome wide association studies (GWAS) employing a commercially available genotyping chips or genotyping by sequencing have uncovered the genetic cause of several new traits. GWAS, by enabling the use of populations rather than families, force us to better understand the evolutionary and breed history of our horses in order to adeptly design our mapping strategies.
- High-throughput genotyping has also provided the necessary data as well as a strong motivating force to encourage a better understanding of the population genetics of equids. We are currently using datasets from multiple projects to investigate the genetic diversity, subpopulations and admixture of the horse, both within our own group and as part of collaborations.
- 3. Recently, we have also begun to utilize RNA-seq to study the transcriptome of a number of equine conditions. Transcriptome analysis is also allowing us to improve genome annotation, and identify novel variants responsible for our traits of interest. For example, we have just initiated work to use RNA-seq to better understand the evolution of the single-toed body plan of the horse, adaptations to bear the immense force of locomotion and the biological processes behind hoof conditions like laminitis.
- 4. Finally, on a limited basis, are expanding the usage of genotyping array data for clinical cytogenetics, (detection of chromosome aberrations) and surveys of CNVs. In recent years it has become increasingly apparent that structural variation is a frequently overlooked source of genetic polymorphism. Recently we have discovered large deletions and rearrangements in cases of congenital abnormality, as well as more moderate alterations in infertile mares.