**APPENDIX D**

**SAES-422**

**Format for Multistate Research Activity**

**Accomplishments Report**

**Project/Activity Number: S1094**

**Project/Activity Title: Genomic tools to improve equine health, wellbeing, and performance**

**Period Covered: 3/2024-2/2025 (“Year 3”)**

**Date of This Report: 2/19/2025**

**Annual Meeting Date(s): 1/12/2025**

**Participants:** An in-person business meeting was held on 1/12/25 in conjunction with Plant & Animal Genome (PAG) 32 in San Diego, CA. All attendees at the PAG Horse Genome Workshop were invited to participate, regardless of membership status in S1094. The workshop itself was held in two sessions, on 1/11 and 1/12. The following individuals attended and presented station reports: Miller, Don (dm96@cornell.edu) – Cornell University (also on behalf of Doug Antczac and Mandi deMestre); Brooks, Samantha (samantha.brooks@ufl.edu) - University of Florida; Laura Patterson-Rosa (laura.patterson@liu.edu) – Long Island University; Staiger, Ann (Elizabeth.Staiger@tamuk.edu) - Texas A&M University – Kingsville; Durward-Akhurst, Sian (durwa004@umn.edu) - University of Minnesota; Petersen, Jessica (jessica.petersen@unl.edu) - University of Nebraska; Delledonne, Massimo (massimo.delledonne@univr.it) – University of Verona; Kalbfleisch, Ted (ted.kalbfleisch@uky.edu) - University of Kentucky (also on behalf of Ernie Bailey and Jamie MacLeod); Mienaltowski, Mike (mjmienaltowski@UCDAVIS.EDU) - University of California-Davis; Norton, Elaine (elainenorton@arizona.edu) - University of Arizona; Bellone, Rebecca ( rbellone@ucdavis.edu) – UC Davis; Mikko, Sofia (sofia.mikko@slu.se) – Swedish University of Agricultural Sciences (also on behalf of Gabriella Lindgren); Ryder, Oliver (oryder@ucsd.edu) – UCSD/San Diego Zoo; Brian Davis (bwdavis@tamu.edu) – Texas A&M University (also on behalf of Terje Raudsepp, Rytis Juras, and Gus Cothran); Finno, Carrie (cjfinno@gmail.com) - University of California-Davis; McCoy, Annette (mccoya@illinois.edu) - University of Illinois. Station reports were sent in writing from Guest, Debbie (djguest@rvc.ac.uk) – Royal Veterinary College (also on behalf Androniki Psifidi and Richard Piercy); Lyons, Leslie (lyonsla@missouri.edu) - University of Missouri; McCue, Molly (mccu0173@umn.edu) - University of Minnesota.

**Brief summary of minutes of annual meeting**: The meeting was called to order by Annette McCoy on behalf of herself and the other co-coordinators (Mike Mienaltowski, Elaine Norton). Angelica Van Goor, National Program Leader for S1094, could not attend, but her contact information was provided and the update that she sent in advance of the meeting was reviewed. This update will be made available to S1094 members. The S1094 multistate project objectives were reviewed, as was the procedure for joining S1094 via Appendix E in NIMSS or by directly contacting Cindy Morley (cmorley@uark.edu). Meeting participants were encouraged to recruit their colleagues for multistate membership. We are particularly interested in engaging members of the equine industry and extension officers. Station reports were presented by all participants. Activities completed in the past year were reviewed. The results of our equine owner stakeholder survey were presented by Mike Mienaltowski (see more details below). Upcoming events relevant to the participants were announced, with a call for volunteers for 1) development of a stakeholder needs assessment aimed at the scientific community, and 2) development of a workshop for the 2025 Equine Science Society meeting. The group voted for the next S1094 Annual Meeting to be held in conjunction with Plant & Animal Genome (PAG) 33 meeting, January 9-14, 2026 in San Diego, CA. A call for contributors to the multistate project newsletter was made, and the floor was opened for other announcements. The meeting was then adjourned.

**Accomplishments:** Three co-coordinators continue to serve the S1094 community: Annette McCoy (University of Illinois), Mike Mienaltowski (University of California-Davis), and Elaine Norton (University of Arizona). Newsletters were produced in 2024 for dissemination to the Horse Genome Workshop listserv (maintained by the co-coordinators) and the general public via member contacts. Each newsletter highlighted meetings and community efforts. In collaboration with Dr. Samantha Brooks (University of Florida), the co-coordinators developed a **stakeholder survey aimed at horse owners**. Distribution of this survey was facilitated via members (particularly Drs. Molly McCue [University of Minnesota] and Laura Patterson-Rosa [Long Island University]) and extension/industry partners. Preliminary results were presented to the international horse genome community at the 14th Dorothy Russell Havemeyer Equine Genome Mapping Workshop in Caen, Normandy, France in May 2024. Final results were presented at the January 2025 annual meeting and are summarized below:

* 412 respondents from 44 states; 60% identified as horse owners and 15% as horse breeders. Nearly 80% of respondents own 10 or fewer horses (62% five or fewer). This was of great interest to us, as many surveys from the horse industry seem to primarily reflect large-scale breeders/trainers.
* A variety of breeds/disciplines were represented, led by sport horses (33%), stock horses (30%), and Arabians (12%). Racing horses were underrepresented (5%), reflecting the demographics of the respondents.
* Interestingly, three-quarters of the respondents self-identified as having at least a solid understanding of genetics. They identified their primary sources of genetics knowledge as universities and breed/discipline organizations. To a lesser extent, veterinarians and horse owner community websites/social media were identified as sources of genetics information. Most owners were interested in knowing about genetic diversity levels in their breed of interest, and most were interested in genetic testing for performance genes. Owners of racing horses were particularly interested in performance gene tests.
* When asked about health concerns of top priority, there were many shared conditions identified across breed/discipline groups. These included arthritis, colic/intestinal disorders, laminitis, metabolic diseases, tendon/ligament injuries, navicular syndrome, and ulcers. Behavior/temperament was also identified as a top priority across groups.

These results will be shared as an abstract at the 2025 Equine Sciences Symposium. A manuscript is in preparation.

More than a dozen graduate students and two dozen undergraduate students completed their training under the guidance of S1094 members in 2024. Public education and stakeholder outreach efforts by S1094 members are highlighted under Aim 3 accomplishments below.

**A major ongoing collaborative effort among S1094 members is the** ***Equid Pangenome International Consortium (EPIC)***. Led by researchers from the University of Kentucky, University of Minnesota, and Texas A&M University, current contributing members are located at 14 institutions around the world. The aims of this consortium are to develop haplotype phased T2T assemblies from crossbred trios that can be incorporated into an equine pangenome reference. The Thoroughbred T2T assembly was made publicly available this year, and the toolboxes and containerized pipelines being designed by EPIC members are freely available. This effort will completely revolutionize the way that researchers handle genomic data in the horse.

**The FAANG data set, which was a major collaborative effort across the S1094 community** led by researchers at the University of California-Davis and University of Nebraska was concluded with all data publicly available. The University of Kentucky also hosts an interactive website for these data. The “adopt-a-tissue” initiative helped drive the success of this project, allowing researchers from around the world to contribute financially to the goals of the project even though they were not part of the original project funding proposal.

Additional accomplishments reported by stations specific to the aims of S1094 are detailed below. Though reported by individual stations, activities marked with an asterisk (\*) reflect collaborations involving other S1094 stations or stakeholder groups. Publications, research presentations, and grants awarded are detailed in the accompanying appendices.

**Aim 1: Improve detection, curation and annotation of pan-genomic variability for genetic selection, as well as stewardship of genetic diversity, across horse breeds and exotic or feral populations.**

* (Mississippi State) \*Launch of a project to identify phenotypical characteristics of U.S. horse breeds to facilitate breeding practices that promote breed preservation. This project is being carried out in collaboration with the Livestock Conservancy, partnering with breed organizations including the Palamino Horse Breeders Heritage Foundation, Palamino Breeders of America Association, and Mountain Pleasure Horse Association.
* (University of Verona) \*We successfully sequenced and assembled the genomes of four horse breeds (Selle Français, Hungarian Sport Horse, Westphalian and Arabian) using deep (30x) ultra-long Nanopore sequencing reads (>1 Mb) from Oxford Nanopore Technologies. These assemblies represent a pivotal advancement in the detection and annotation of pan-genomic variability across horse breeds, significantly enhancing the genetic diversity represented in available reference genomes: the Thoroughbred (EquCab3.0), available since 2018, and the recently assembled Finnish Horse (EquCab\_Finn). Compared to EquCab3.0 (genome size: 2.5 Gb; Ncontig: 10,986; N50contig: 1.5 Mb), our assemblies resulted markedly more contiguous and less fragmented (genome sizes: 2.5 Gb, 2.5 Gb, 2.4 Gb, and 2.4 Gb; Ncontig: 172, 148, 178, and 190; N50contig: 53.6 Mb, 58.6 Mb, 56.1 Mb, and 59.9 Mb, for the Selle Français, Hungarian Sport, Westphalian and Arabian horse, respectively), underscoring their utility for accurate curation of genomic variability and stewardship of genetic diversity. These high-quality assemblies are now being integrated into the first equine pangenome graph as part of the Equid Pangenome International Consortium (EPIC).
* (University of Minnesota) \*Mapped >900 equine genomes to the new Thoroughbred T2T reference. The plan will be to use these genomes to improve annotation and investigate population genomics in this population.
* (University of Minnesota) \*I am part of the Equine Pangenome group that is working towards the first equine pangenome.
* (University of Minnesota) Published on the genetic burden and frequency of disease-causing variants in a population of 534 horses.
* (UC-Davis) \*Provided all FAANG datasets and WGS from multiple breeds to assist with pangenome annotation.
* (University of Missouri) \*We have several horse genomes that we have sequenced at the University of Missouri and then provide to collaborators to help find causal DNA variants. One disease, suspected hematochromatosis, is in a mule, hence the develop of the mule genome will be most beneficial to this project! We have supplied several short read genomes for diversity inclusion.
* (University of Nebraska) \*Participated in the successful PanGenome grant effort. Contributed samples/data from Clydesdales for reference genome assembly. Continued work on genetic diversity of breeds (Thoroughbred, Clydesdale, Shire).
* (Royal Vet College) We developed and used a polygenic risk score for catastrophic fracture risk in Thoroughbred horses to develop in vitro cell culture models to compare bone forming osteoblasts derived from horses at high and low risk of fracture. This approach identified a functional SNP upstream of the Collagen Type III gene which is associated with catastrophic fracture risk in Thoroughbred horses (Palomino Lago et al 2024). It also identified 112 genes that are differentially expressed in osteoblasts in high and low risk horses (Palomino Lago et al 2025), which suggested that processes regulating the extracellular matrix may be involved.

**Aim 2: Apply and improve genomic resources to increase our understanding of equine performance and disease.**

* (University of Verona) We are working on an imputation panel tailored for steeplechase horses to enable cost-effective genomic characterization at a population scale. Using 30x Illumina whole-genome sequencing (WGS) data from 35 sport horses representing different breeds, we evaluated six state-of-the-art genotype imputation tools: Beagle, GLIMPSE2, Impute5, loimpute, Minimac3, and Quilt2. Quilt2 achieved the best performance, with >95% accuracy in imputing more than 600,000 genomic variants on three chromosomes in two independent horses. This represents a significant improvement, with >25% and >15% increase in imputation accuracy compared to the panels currently available through Animal-ImputeDB and AGIDB, respectively. These findings directly enhance the utility of imputation for population-scale studies, paving the way for improved insights into equine performance traits and disease mechanisms.
* (University of Minnesota) Developed 90 equine simulated genomes, which have been made publicly available on the European Nucleotide Archive.
* (University of Minnesota) \*Generated IsoSeq data on 9 equine tissues. These will be used to improve annotation of the equine genome.
* (University of Minnesota) Investigation of the genetics of exercise associated sudden death and atrial fibrillation in Standardbred and Thoroughbred racehorses.
* (UC-Davis) \*Currently developing large allele frequency database that incorporates all public equine WGS data for SNP variants and small indels.
* (UC-Davis) Using genomics, we examined the cytoprotective properties of ascorbic acid for stressed tendon and peritenon cells, and we determined the correlations between mare’s milk microbiota and foal age to a healthy equine gastrointestinal microbiota. Moreover, we examined the detrimental effects of anti-inflammatory diclofenac on tendon cells.
* (University of Nebraska) Continued work to understand the genetics of shivers in draft horses.
* (University of Nebraska/UC-Davis) Studying chronic progressive lymphedema; we have collected samples, transcriptome and genotype data for this project.
* (University of Arizona) Identification of gene by environment interaction contributing to equine metabolic syndrome. For this project, we have completed collection of the EMS phenotype data from 690 Arabian horses. Using the date, we were able to complete a large-epidemiological evaluation of EMS across the Arabian breed and within their subpopulations. Additionally, we gathered genomic and epigenetic data, which will be analyzed this year to identify potential interactions between genetic predisposition and environmental factors contributing to EMS.
* (University of Arizona) Insulin dysregulation: placental changes and foal health For this project, we have started phenotype and genotype collection on 45 foals (15 born to foals with EMS, 15 insulin sensitive mare, and 15 insulin sensitive-obese mares). This includes data for transcriptomics on the foals.
* (University of Illinois) Ongoing project investigating molecular markers of post-traumatic osteoarthritis in an equine fetlock osteochondral fragment model.
* (University of Illinois) Ongoing project investigating muscle gene expression response to growth and exercise in foals.
* (University of Illinois/University of Minnesota) Ongoing projects investigating osteochondrosis dissecans, gait, and performance in Standardbred racehorses.

**Aim 3: Expand the availability of genetic diagnostic testing and education on its use.**

* (University of Florida) Several lay presentations/podcasts/articles:

Articles:

* + Ciosek, J., Wickens, C. and Brooks, S.A. (2024) 'A Primer on Genetic Testing for Horse Owners and Breeders: VM261, 4/2024', EDIS, 2024(3).
	+ Interviewed in: “How Coat Color and Genetics Influence Equine Behavior” by Sarah Welk Baynum, The Horse, (USA National, March 8, 2024), https://thehorse.com/1125641/how-coat-color-and-genetics-influence-equine-behavior/
	+ Interviewed in: “Handling and Training Donkeys” by Haylie Kerstetter, The Horse, (USA National, August 8, 2024), https://thehorse.com/1122463/handling-and-training-donkeys/

Presentations:

* + Kentucky Equine Research: 27th Equine Health and Nutrition Conference, Ocala FL, February 5-6, 2024 – “Should You Sweat It? Anhidrosis in the Equine Athlete”
	+ Equine Affaire, Columbus OH, April 11th 2024 – “DNA & Genetic Testing Isn’t Just for Breeders: Get a Leg Up on Genetics, Tips for Horse Owners”
	+ Equine Affaire, Columbus OH, April 12th 2024 – “Galloping into the Future: What’s Next for Equine Genomics, Recent Discoveries and Cutting Edge Applications”
	+ 2024 Al Khamsa Annual Meeting and Convention, Online, June 15th 2024 – “DNA 101: Genetic Profiling, what is it?”
	+ American Trakehner Association, 2024 Annual Meeting, Ocala FL, December 20th 2024

Podcasts

* + Guest appearance on Scoop & Scale by Dr. Claire Thunes, “Ep. 23: Is How My Horse Sweats Normal?”, Podcast, May 28, 2024.
	+ Guest appearance on Tack Box Talk by Kris Hiney, “Sane and Sound: The story of an equine geneticists goal to build a better horse”, Podcast, June 10nd 2024.
	+ Guest appearance on Tack Box Talk by Kris Hiney, “Genetics Part 2: The story of fragile foals and muscle disorders”, Podcast, July 22nd 2024.
* (University of Minnesota/UC-Davis/University of Arizona)
	+ ACVIM 1-hour panel: A Panel on Clinical Applications for Complex Genetic Diseases (Drs. Carrie Finno, Sian Durward-Akhurst, Elaine Norton and Lauren Hughes) (~25 attendees, board-certified equine internists and veterinary students, residents and technicians)
* (University of Minnesota) Presentations for the horse/veterinary community:
	+ Horseracing Safety Integrity Authority and International Federation of Horseracing Authorities – discussed opportunities for genetics to reduce the frequency of exercising associated sudden death in Thoroughbred racehorses.
	+ Minnesota Trotting Association – talked about arrhythmias and the genetics or arrhythmias in racing Standardbreds.
* (UC-Davis) Provided location of genetic mutation for Equine Juvenile Spinocerebellar Ataxia (EJSCA) to UC Davis Veterinary Genetics Laboratory for 2024 testing.
* (UC-Davis) Educational presentations on genetics:
	+ American College of Veterinary Internal Medicine Forum, Minneapolis, MN; 45-minute presentation: Ongoing Investigation of Genetic Diseases in Horses (~50 attendees, including board-certified equine internists and veterinary students, residents and technicians)
	+ Updated American Association of Equine Practitioner’s Statement on Genetic Defects
	+ American Association of Equine Practitioner conference, Orlando, FL; 1.5 hr session: Kester News Hour – included current studies on genetic testing (~3500 attendees, including veterinarians, veterinary students and veterinary technicians)
	+ California residents and horse enthusiasts at 2024 UC Davis Horse Day on October 12, 2024.
* (University of Nebraska/UC-Davis) Conducted a study focused on validation of a commercial genetic test (test is not valid and should not be offered commercially).
* (University of Nebraska) Stakeholder outreach sessions:
	+ American Shire Association: approval to conduct a population-level survey of genetic variation, continued interaction with individual breeders
	+ Presented to UNL Extension event, “UNL Race Nebraska and Horsemen’s Health Update.” Genetic Diversity in Thoroughbreds: What do we Know?
* (Royal Veterinary College) Invited speaker at the International Federation of Horseracing Authorities (IFHA) Global Summit on Equine Safety June 2024.
* (University of Arizona) Arizona Arabian Association: Did a presentation on EMS and nutrition to 58 members
* (University of Illinois) Presentation on genetic tools and testing at the American Association of Equine Practitioners, ~100 attendees

**Aim 4: Create platforms for broad sharing of data, technology, and resources to enhance continued development and application of genomics tools in the industry.**

* (University of Florida) We are continuing to develop tools to enable high-throughput and precise locomotor phenotyping and hope to have several packaged and pipelines published in 2025. A preliminary version of our review dashboard is publicly available on BioRxiv and GitHub: <https://www.biorxiv.org/content/10.1101/2023.10.30.564795v2.full>
* (University of Minnesota) \*Development of containerized pipelines for analysis of whole-genome sequencing data, RNA sequencing data, and pangenome map creation.

**Short-term Outcomes:** The members of S1094 leverage community resources in the development of new genetic tests that can then be offered commercially once validated. A recent example is the addition in November 2024 of an updated test for gray alleles offered by the University of California-Davis Veterinary Genetics Lab. This now includes a novel gray allele that was recently found to be correlated to increased risk of gray horse melanoma (Rubin et al. *Nature Communications* 15:7510, 29 Aug 2024) – work that was a collaborative effort between researchers at the University of Uppsala and S1094 members at UC-Davis. Education bulletins are released by the VGL to share updates with stakeholders. One of the major goals of the Horse Genome Workshop community is to make the tools and resources that have been developed easily accessible to the broader community. A web portal which provides resources for accessing equine FAANG data was recently launched, supported by the University of Kentucky. This portal can be freely accessed at: [www.equinegenomics.uky.edu](http://www.equinegenomics.uky.edu).

**Outputs:** Data from several collaborative projects are being generated and made publicly available in repositories. These include whole genome sequencing, RNAseq, and ChIPseq, all generated as part of the equine FAANG initiative (led by the University of California-Davis and University of Nebraska). As part of the ongoing community pangenome project (led by the University of Kentucky) new shotgun sequencing from horses and non-caballine individuals have been generated and are publicly available. A new telomere-to-telomere (T2T) Thoroughbred reference has recently been completed. All of these resources are being leveraged to update annotation for the reference genome EquCab3.0. Containerized workflows for use with an array of equine genomics data have been developed (led by the University of Minnesota and University of Kentucky) and are available for use. A list of publications from S1094 contributors is included in this report.

**Activities:**

1. **14th Dorothy Russell Havemeyer Equine Genome Mapping Workshop, May 12-15, 2024, Caen, Normandy, France**

Members of the equine genetics community from around the world gathered to share research findings and strengthen collaborative relationships at this workshop, which has been held every 2-3 years since the 1990s.

1. **Equine Genome Workshop, Plant and Animal Genome (PAG) 31, January 11-12, 2025, San Diego, CA.**

The meeting was chaired by chaired by Dr. Ann Staiger (Texas A&M University – Kingsville) and co-chaired by Dr. Sian Duward-Akhurst (University of Minnesota). More than 50 scientists and trainees from 29 different universities and organizations from around the world were given the opportunity to hear about research advances from the past year from 16 diverse speakers. This work highlighted the varied applications of genomic and transcriptomic tools and resources developed by the Equine Genome community for investigation of horse physiology. The results of the S1094 equine owner stakeholder survey were presented and discussed.

1. **Workshop *Practical Pangenomics: Containerized Workflows for Building Graphs and Variant Discovery in Livestock and Companion Animals* Plant and Animal Genome (PAG) 32, January 13, 2024, San Diego, CA.** This workshop was a collaborative effort between S1094 members at the University of Minnesota, University of Kentucky, and Texas A&M University highlighting the development of pangenomes for agricultural animal species (using the horse an example) and associated interactive tools. This workshop was open to researchers across species and had a robust attendance.

**Milestones:** We continue to work towards several milestones for this project:

1. *Engagement of researchers at ~20 stations across the country for multistate participation.* Current S1094 members are being asked to actively recruit new participants, with a particular interest in engaging members of the equine industry and extension agents. We are slowly increasing the number of official participants, with continued robust interactions with researchers who have not yet been able to join. To date, S1094 includes 18 researchers at 14 stations across the United States and three researchers at two European institutions. These members engage in robust collaborative efforts with individuals who are part of the larger equine genetics community but are not yet part of S1094.
2. *Generation of summary stations’ strengths and collaborative interests ahead of summer workshop meeting.* This information was solicited via the call for station reports in January 2025.
3. *Collaborative summer workshop* A workshop focused on equine scientists has been accepted for the Equine Sciences Society meeting in June 2025. This goal of this interactive workshop is to provide information about available genetics resources created by the S1094 community, and to facilitate collaborations between equine genetics experts and scientists in related fields.
4. *Stakeholder needs assessments.* We will be launching two additional stakeholder needs assessments in 2025. The first will be aimed at (non-genetics) researchers, while the second will be aimed at veterinarians. The launches are planned to coincide with the Equine Sciences Society meeting in June (researcher survey) and the American Association of Equine Practitioners meeting in December (veterinarian survey). A summary of results will be shared with the S1094 community and the broader Equine Genome Workshop group, while stakeholder-specific data will be shared with the relevant community.
5. *Renew website with lists of educational materials for stakeholders and genomics resources for researchers.* A newly redesigned website for the International Horse Genome Workshop includes reports from meetings, identification of participants, and links to community tools. The website can be found at: https://horsegenomeworkshop.com/.

**Impacts:** A major focus currently is recruitment to the multistate group, with particular interest in involving extension officers and industry stakeholders. The current members of S1094 have leveraged the resources developed by the equine genomics community for projects that have been awarded grants in the past year totaling more than $3.6 million (complete list in Appendix A). Multistate members provide research and outreach presentations (Appendix B). T. Raudsepp (Texas A&M) continues to operate the only equine clinical cytogenetics service in the United States.

**Indicators:** The most tangible evidence of industry stakeholder buy-in to community efforts is the widespread adoption of genetic testing among several breed organizations, and the ongoing interest in highly accurate parentage testing. Multistate members working in direct support of these efforts are at the University of California-Davis, University of Kentucky, and Texas A&M University.

**Publications:**

Culwell, J., North, E., Nicodemus, M.C., Cavinder, C., Williams, T. (2024) PSIV-9 Pedigree tracing to determine the origin of the golden coat phenotype within the Golden American Saddlebred horse. Journal of Animal Science, 102 (3), 549–550. <https://doi.org/10.1093/jas/skae234.617>.

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Alhaddad, H., Powell, B., Del Pinto, L., Sutter, N., Brooks, S. and Alhajeri, B. (2024) 'Geometric morphometrics of face profile across horse breeds and within Arabian horses', Journal of Equine Veterinary Science, 132, 104980.

Ciosek, J., Wickens, C. and Brooks, S.A. (2024) 'A Primer on Genetic Testing for Horse Owners and Breeders: VM261, 4/2024', EDIS, 2024(3).

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McFadden, A., Martin, K., Vierra, M., Robilliard, H., Lundquist, E.W., Everts, R.E., Brooks, S.A. and Lafayette, C. (2024a) 'Three HPS5 mutations associated with depigmentation in diverse horse breeds', Livestock Science, 282, 105454.

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McFadden, A., Vierra, M., Robilliard, H., Martin, K., Brooks, S.A., Everts, R.E. and Lafayette, C. (2024c) 'Population Analysis Identifies 15 Multi-Variant Dominant White Haplotypes in Horses', Animals, 14(3), 517.

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Marlowe, J. L., McCue, M. E. (Lead Author), & Durward-Akhurst, S. A. (Lead Author) Simulated whole genome sequencing data of Equus Caballus as a novel benchmark truth set. Nature Scientific Data. [Submitted:2024]

Palomino Lago, E. (Lead Author), Baird, A., Blott, S., McPhail, R., Ross, A., Durward-Akhurst, S. A., & Guest, D. (Corresponding Author) (2024). A functional single nucleotide polymorphism upstream of the collagen type III gene is associated with catastrophic fracture risk in Thoroughbred horses. Animals, 14(1). doi: 10.3390/ani14010116

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Smith, E. J., Beaumont, R. E., Dudhia, J. & Guest, D. J. Equine Embryonic Stem Cell-Derived Tenocytes are Insensitive to a Combination of Inflammatory Cytokines and Have Distinct Molecular Responses Compared to Primary Tenocytes. Stem Cell Rev Rep, May;20(4):1040-1059. doi:10.1007/s12015-024-10693-8 (2024).

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**Appendix A: Grants and Contracts Received**

Nicodemus, M, Cavinder, C., “Pedigree Tracing to Determine the Origin of the Dilution Gene within Color-Designated Horse Breed Registries Requiring a Golden Coat Color,” Office of Research and Economic Development, Undergraduate Research Program Grant, Mississippi State University, $1,999 (June 2024 – August 2025).

Bailey E (Principal Investigator), Kalbfleisch T (Co-I), Petersen J (Co-I). Comparing Genetic Diversity of North American Thoroughbred Horses from two periods: 2005-2019 and 1965-1984; Koller Endowment. 2023-2024 $135,300

Durward-Akhurst S (Principal Investigator). Genetic mechanisms of sudden cardiac death in racehorses as a model for sudden arrhythmic death syndrome in human athletes. UMN CTSI pre-K award. $50,000

Durward-Akhurst S (Principal Investigator). Investigation of arrhythmic-SCD causing mutations in racehorses. UMN College of Veterinary Medicine Animal Health Grant. $60,000

Guest D (Principal Investigator). Bad memories: do horse tendon cells become primed to inflammation? Horserace Betting Levy Board, £173,000

Guest D (Principal Investigator). Utilising novel therapeutics to reduce inflammation in equine adult tenocytes Petplan Charitable Trust, £10,000.

McCoy AM (Principal Investigator). Effects of Early Exercise on Muscle Expression of

Myokines in Foals. USDA Hatch Funds FY23. 9/2023-9/2024. $17,385.

McCoy AM (Principal Investigator). Molecular markers of early equine post-traumatic osteoarthritis. USDA-AFRI-NIFA. 5/2023-4/2026. $627,000.

McCoy AM (Mentor). Alterations in the peritoneal environment of horses with intestinal disease: a multiomics approach. Morris Animal Foundation. 7/2023-6/2025. $124,956.

McCue ME (Principal Investigator), Durward-Akhurst S (co-I). Mendelian and oligogenic disease mutation discovery: best practices and automated tools. UMN College of Veterinary Medicine Multistate. $70,000

Mieneltowski M (Principal Investigator). Delivery of Exogenous EGR1 mRNA to Improve Tendon Formation. UC Davis Center for Equine Health Research Grant. $28,735

Petersen J (Principal Investigator). Equine genomics for the management of breed diversity, health, and performance. ARD Hatch Enhanced Grant $200,000

Raudsepp T (Collaborator). Genetic Analysis for the Wild Horse and Burro Program (WHBP). DOI-Bureau of Land Management. 2021-2025. $152,187.

Raudsepp T (Principal Investigator). Exploring the Genomic Component of Equine Sex Development and Reproduction. USDA-NIFA2022-08309. 2023 - 2026. $644,320.

Raudsepp T (Principal Investigator). Genomics of Thoroughbred stallion subfertility. Grayson-Jockey Club Research Foundation. 2023- 2025. $77,371.

Raudsepp T (Principal Investigator). Exploring the Genomic Component of Reproductive Health in Mares: Molecular Signatures for X-Monosomy-Like Gonadal Dysplasia. American Quarter Horse Research Foundation. 2023- 2024. $39,021.

Raudsepp T (Co-PI). Understanding the co-evolution of phylogenomic signal, gene linkage, and recombination rate through comparative genomics. NSF-DEB #2150664. 07/01/2022-06/30/2025. $1,200,000. (includes horse and equids)

Brooks SA; USDA-HATCH- MULTISTATE, Hatch Multistate Award, 2024, $ 1,000

Brooks SA; NATL INST OF HLTH NIAMS (R01AR085334), Sept/2024 - Aug/2027, $5,470,940 Psychometric Testing of Behavioral Metrics of Osteoarthritic Pain in Horses (AWD17480)

Kalbfleisch, T. (PD), Durward-Akhurst SA (co-PD); Functionally Annotated Equine Pan Genome with Infrastructure for an Accessible, Integrative, Community Genomics Resource; UNIVERSITY OF KENTUCKY

Durward-Akhurst, SA (PI) Identification of putative exercise-induced pulmonary hemorrhage causing variants in Ontario's Thoroughbred racehorses; UNIVERSITY OF GUELPH; March 1, 2024 - June 30, 2024

Durward-Akhurst, SA (PI) A multi-omics approach to prediction of exercise associated sudden death in Thoroughbred racehorses; Horseracing Integrity and Safety Authority

Durward-Akhurst, SA (co-PI), McCue ME (co-PI) A robust pipeline to functionally annotate non-traditional model pangenomes; University of Minnesota Informatics Institute Medium Seed Grant; 2024 – Present

Kurandina, R (Trainee), Durward-Akhurst, Sian A. (Mentor) Investigating ECGs and human causative variants in a spontaneous athletic sudden cardiac death animal model; UMN Clinical and Translational Science Institute APReP

Durward-Akhurst, Sian A. (PI), Dudley, S (Primary Mentor), Talkachova, A (Mentor), Rendahl, A (Mentor) The horse as a naturally occurring model of arrhythmias and autopsy negative sudden arrhythmic death; NIH NCATS K12

Grayson Jockey Club Foundation Finno (PI) March 2023-Feb 2025

Role: Principal Investigator $97,365

Validation of Biomarkers for Equine Neurodegeneration

The goal of this study is to validate previously identified biomarkers for equine neurodegeneration in serum and cerebrospinal fluid to assist with phenotyping for genetic studies.

The Foundation for the Horse Fellowship Ryan (student) Dec 2023- Dec 2024

Role: Mentor $20,000

Hepatic gene expression in equine neuroaxonal dystrophy

This Young Investigators Research Grant was awarded to Dr. Stephanie Ryan, a 3rd-year PhD student in my laboratory, to support her graduate training.

UC Davis Center for Equine Health 23-02 Finno (PI) March 2024-March 2025

Role: Principal Investigator $40,000

Long-range sequencing for equine neuroaxonal dystrophy (eNAD)

The goal of this study is to use long-range whole-genome sequencing in horses with equine neuroaxonal dystrophy to identify a causative genetic mutation.

Morris Animal Foundation D24EQ-056 Finno (PI) March 2024-Feb 2026

Role: Principal Investigator $97,107

Unraveling the Genetic Etiology of Equine Neuroaxonal Dystrophy in Quarter Horses and Warmbloods

The goal of this study is to interrogate the liver transcriptome of horses with equine neuroaxonal dystrophy to identify changes in gene expression related to hepatic vitamin E processing and storage.

Mienaltowski MJ (PI) Internal UC Davis Center for Equine Health Award: Evaluation of Mechanical Stimulation within an In Vitro Three‐Dimensional Equine Tendon Construct Model ($26,843)

Mienaltowski MJ (PI) Internal UC Davis Center for Equine Health Award: Mitigating the Adverse Effects of Diclofenac on Tendon Cells with Vitamin C ($39,272)

UC Davis Center for Equine Health – Drs. Bannasch, Affolter, Petersen.

Investigation of genetic susceptibility for chronic progressive lymphedema in draft horses

“Insulin dysregulation: placental changes and foal health.” Grayson-Jockey Club Research Foundation, Inc. April 2024-April 2026. PI: Elaine Norton. $115,003.

**Appendix B: Research presentations**

January 2025. Plant and Animal Genome 32, San Diego, CA

Esdaile E, Till BJ, Malvick J, Magdesian KG, Bellone RR. De Novo Variants in Key Genes of Melanocyte Development, KIT and Its Regulators Mitf and PAX3: A Common Cause of White Spotting Patterns in Horses

Ryan S, Finno CJ. Liver Transcriptome in Equine Neuroaxonal Dystrophy

Bacon EK, Velie B. Identifying Potential Candidate Genes Influencing Equine Analgesic Efficacy Using Ranked Pain Responses

Balasubramaniam NP, AbouEl Ela NH, Bailey E, Kalbfleisch, Petersen JL. Comparative Analysis of Inbreeding Coefficients and Genetic Variation in American Thoroughbred Horses Using the T2T and Ecab3 Genome Assemblies

Anderson HC, Stroupe SC, Conley AJ, Caruso C, Cotterman R, Juras R, Davis BW, Raudsepp T. Equine Disorders of Sex Development Involve Mutations in Several Sex Development Key Genes

Wyngarden S, Stein F, Stauhammer CD, Moore A, McCue ME, Durward-Akhurst SA. Identification of Genomic Regions of Interest for Atrial Fibrillation in Standardbred Racehorses.

Mikko S, Picchi F. Genomic Analysis of a Hypermobility Syndrome in Gotland Ponies

Moss A, Han H, Hall TJ, Holtby AR, McGivney BA, Mitchell K, Mullen S, O’Reilly O, Rand A, Salter-Townshend M, Katz LM, Hill EW. Maternal Care Style in Thoroughbred Horses Is Linked to Genetic Variation at the Glucocorticoid Receptor Gene (NR3C1) Locus

Hammack SM, Kersh ME, McCoy AM. Sex-Specific Muscle Transcriptome Response to Exercise in Pre-Pubertal Foals

Miller D, Tallmadge RL, Simon T, Bergstrom TF, Antczak D, Kalbfleisch T. Manual Annotation of the Major Histocompatibility Complex in the Horse T2T Assembly

Cullen JN, Stroupe SC, Durward-Akhurst SA, Martini D, Delledonne M, Petersen JL, Kalbfleisch T, Davis BW, McCue ME. Reining in Reference Bias with the Equine Pangenome

Stroupe SC, Cullen JN, Durward-Akhurst SA, Martini D, Delledonne M, Petersen JL, Kalbfleisch T, McCue ME, Davis BW. Unbridled Insights into the Equine Pangenome

Kalbfleisch T, Li K, Ciosek JL, Johnson LC, Robyn ED, Petersen JL, Durward-Akhurst SA, McCue ME, Cullen JN, Laird-Smith M, Hudson E, AbouEl Ela NH, Balasubramaniam NP, Miller D, Antczak D, Raudsepp T, Adelson DL, Dessaix C, Giulotto E, Cappelletti E, Watson C, Safanova Y, Voss K, Zhu Y, Horin P, Bergstrom, Antonacci R, Davis BW. Progress Toward Annotation of the Mule T2T Assembly

Thibaud-Nissen F, Kodali V, O’Leary NA, Rangwala SH, Schneider VA, Murphy TD. Exploring the Power of the NIH Comparative Genomics Resource for Equine Species and Beyond

Hamsher H, Cullen JN, McCue ME. Herder: An Interactive Application for Investigating Variants and Allele Frequencies in Large WGS Datasets

Mieneltowski MJ, Hernandez S, Nastrini E, Norton E, McCoy AM, Brooks SA. Findings from a U.S. Stakeholder Survey of Horse Genetics Knowledge and Concerns

Others reported by stations

Culwell, J., North, E., Nicodemus, M.C., Williams, T. (2024) Pedigree tracing to determine coat color phenotype found within the Mountain Pleasure Horse Breed. Mississippi State University Undergraduate Research Symposium, Summer Showcase, 2 August 2024, 25.

McCoy AM, Tate NM, McCue ME. Integration of genome-wide association analysis and tissue-specific transcriptomics to prioritize functional and predictive variants for equine osteochondrosis. 14th Dorothy Russell Havemeyer Equine Genome Mapping Workshop, Caen, Normandy, France, May 2024.

Brooks S, McCoy AM, Mienaltowski M, Norton EM. Assessing stakeholder needs to inform outreach efforts and research priorities from the equine genomics community. 14th Dorothy Russell Havemeyer Equine Genome Mapping Workshop, Caen, Normandy, France, May 2024.

Bishop RC, Kemper AM, Clark L, Wilkins PA, McCoy AM. Effect of pasture vs stable management on gastric and fecal microbiota of healthy horses. 14th Dorothy Russell Havemeyer Equine Genome Mapping Workshop, Caen, Normandy, France, May 2024.

Bishop RC, Kemper AM, Clark LV, Wilkins PA, McCoy AM. Stability of gastric fluid and fecal microbial populations in pastured or stabled horses. 14th International Colic Symposium, Edinburgh, United Kingdom, July 2024.

Frake EE, Tate NM, McCoy AM, McCue ME. Relationships between performance traits and osteochondrosis of the hock in Standardbred racehorses. American Society of Animal Science, Calgary, Alberta, Canada, July 2024.

**Authorization**: Submission by an AES or CES director or administrative advisor through

NIMSS constitutes signature authority for this information.

\*Limited to three pages or less exclusive of publications, details may be appended.