

# AG2PI SEED GRANT - PROJECT FINAL REPORT

PROJECT NAME	A genetic data portal to enable discovery of deleterious genetic variants in farmed animals
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PROJECT PRINCIPAL INVESTIGATOR	TODAY'S DATE	PROJECT START DATE	DATE OF COMPLETION
Theodore S. Kalbfleisch	10/31/2023	06/01/2022	05/31/2023
TEAM MEMBERS (co-PI, co-I, personnel)	COLLABORATORS		
	Dr. Fiona McCarthy, Dr. Elaine Norton, Dr. Jennifer Janes, Dr. Jessica Petersen, Dr. David Steffen, Dr. Brenda Murdoch, Dr. Darren Hagen,		

## ACCOMPLISHMENTS

Please provide a short summary of the conclusions (both successes and failures) made from your project. Include a description of how this project will provide benefits to the agricultural genome to phenome community and, possibly, to a broader audience. You should include both qualitative and quantitative details, as necessary, to support your conclusions. Include a short accomplishment statement in non-technical language and do not include names.

The objective of this project was the identification of recessive lethal alleles from aborted fetuses with no recognizable pathology. The foremost result of this project is the demonstration that idiopathic abortions should be treated as a cohort.

We demonstrated that analysis of individual animals resulted in too many candidate alleles for subsequent analysis. Treating all animals as a disease cohort produced a common homozygous haplotype shared by several animals such as the 5/17 horse fetuses that were homozygous for the LCORL locus, where no homozygotes appeared in a sample of 183 healthy adult Thoroughbreds with a 16% minor allele frequency, and could potentially explain as many as 2% of IAs in North American Thoroughbreds.

Our project has produced a workflow and software capable of comparing the Idiopathic Abortion (IA) cohort to a healthy cohort of animals identifying genotypes in the IA cohort that do not appear in the healthy.

## Products

Please list any products from this project. This may include (but not limited to) publication, concept/white paper, workshop, conference presentation, website, publicly available data or pipelines, etc. Reminder: you are required to make your products available to the broader stakeholder community using standard USDA practices, open source, FAIR, or other models. Metrics may include number of participants or times accessed, etc. Include links to recordings, DOI, etc. when possible. For presentations and posters, provide authors, date, location and presentation title.

ACTIVITY / PRODUCT	DESCRIPTION (include URL, if applicable)	OUTCOME / METRICS
Poster Presentation PAG	A Genetic Data Portal to Enable Discovery of Deleterious Genetic Variants in Farmed Animals	First poster presentation for student Xiomara Arias
Poster Presentation AG2PI Meeting KC	Discovery of Deleterious Genetic Variants in Farmed Animals	AG2PI presentation
Podium Presentation	Discovery of Deleterious Genetic Variants in Farmed Animals	Podium Presentation at ISAG
Data Portal	<a href="#">Idiopathic Abortion Sequence and Variant Data</a>	Sequence and variant data from Idiopathic samples including a VCF file with filtered against a healthy population of animals. Instructions to use the data can be found at <a href="https://youtu.be/1GGLC9DTYng">https://youtu.be/1GGLC9DTYng</a>

## Audience

With whom has this work been targeted to and shared? Please describe how this project and its products have been disseminated to a community of interest. Include any outreach activity or information sharing as well as training or professional development opportunities provided in this project.

This work has been communicated to researchers in the animal genomics community via poster and podium presentations at the KC AG2PI meeting, ISAG in Cape Town, South Africa, and PAG. It has likewise been communicated to producers via departmental seminars

## CONTINUATION OF WORK

### Next steps

How do you/your team plan to continue moving this project forward? Include how AG2PI can assist in your forward momentum.

Once we have validated our findings, the results will be submitted for publication in a peer reviewed journal. We are currently working with the University of Kentucky Veterinary Diagnostic Lab to set up policy and funding that would guide and enable routine sequencing of IA samples. Additional opportunities for funding, and presentation by AG2PI would be appreciated.

## Outreach

In what ways are you able to stay engaged with AG2PI? (check boxes as appropriate)

- Will present at a field day
- Will lead a training workshop
- Would like to participate in any future AG2PI conference
- Work with AG2PI on a news release on project conclusions
- Will continue attending AG2PI events
- Other (please explain)

Would like to establish a service at UK that sequences idiopathic abortions to build out a database of genotypes found only in these samples.