

Description of the research topic

General information

Institute: National Research Institute of Animal Production.

Research topic: Exploring the genetic basis of the BEW (blue-eyed white) phenotype in alpacas using the latest genome sequencing techniques.

Scientific discipline: Animal Production and Fisheries.

Name of potential supervisor: Prof. dr hab. Katarzyna Piórkowska.

Background information

The BEW phenotype, a white, blue-eyed alpaca often with hearing impairment, occurs spontaneously when crossbreeding alpacas of different colors. BEWs are undesirable due to the iris color and hearing impairment. The only information regarding the genetic basis concerns a SNP mutation in the KIT gene, although this mutation also occurs in gray and crypto-gray alpacas. Breeders need a useful DNA marker that would enable targeted crossbreeding that would eliminate the BEWs in subsequent generations. The biggest problem is crypto-gray alpacas, which carry the mutation but do not reveal it in their fleece color, and for them, a test for KIT gene mutations would seem essential, however: which alpacas to test?

The scientific objective of the study is to identify DNA markers associated with the BEW phenotype using long- and short-read sequencing techniques to increase breeders' awareness of crossbreeding and to avoid the appearance of undesirable BEW individuals in offspring.

The results will be directly used to develop a DNA marker closely associated with the BEW phenotype, a cost-effective and rapid detection method, and to develop such a service for breeders, which is currently identified as a high-priority need by breeders.

The main question to be addressed in the project:

- Is the BEW phenotype (white, blue-eyed alpaca, often with hearing impairment) associated solely with variants of the KIT gene?
- There is extensive information in the literature about the correlation of variants in the ALX7, DCT, and TYR genes with the occurrence of blue irises. Does this correlation also exist for the BEW phenotype?
- Does the rose grey phenotype (gray-pink alpaca) share many genetic traits with BEW, as breeders suggest?

- Are other genes that play a key role in melanogenesis, such as KITLG, MITF, and ENRD, associated with determining the BEW phenotype?
- Will DNA isolates obtained from buccal swabs be useful for long-read genome sequencing using the PacBio technique?

Information on the methods/description of work

The research will be conducted on groups of alpacas with the BEW phenotype, silver grey (silver-grey alpaca), rose grey (pink-grey alpaca), and white alpacas. The research material will be collected in the form of swabs and blood samples. DNA isolates from swabs will be used for genome sequencing using short-read techniques, while DNA isolates from whole blood will be used for genome sequencing using long-read techniques. The selected characteristics will be used to develop DNA markers that can be used in breeding to avoid producing BEW individuals, enabling monitoring of parental DNA. Special, inexpensive and less labor-intensive identification methods will be developed for the selected BEW-specific markers. The selected genetic traits characteristic of the BEW phenotype will be validated on a group of approximately 400 alpacas with varying fleece colors, including individuals with the BEW phenotype, silver and rose grey alpacas, and white alpacas. The obtained results will be presented at as many scientific conferences as possible, including industry conferences, to increase breeders' awareness of the potential risk of producing alpacas with the BEW phenotype in their offspring. Blue eyes are considered a defect by breeders, and this phenotype is also associated with a high risk of hearing impairment. Transcripts for genes whose variants demonstrate a correlation with the BEW phenotype will be analyzed in hair follicles, taking into account alpacas with different fleece colors.

Additional information

The tests will also involve collecting material from alpacas, in the form of swabs, blood samples, or hair follicles, which will require travel to alpaca farms located in various parts of Poland. Therefore, the candidate should not be afraid of alpacas, as they will be actively involved in collecting this material.

Special requirements:

- Genetic laboratory skills required, including knowledge of basic DNA and RNA isolation methods and PCR techniques.
- Knowledge of Sanger sequencing techniques is a plus.
- Basic knowledge of alpaca breeding in Poland is a plus.
- Good command of English.

Place/name of potential collaborators

At this stage, we don't anticipate hiring a co-supervisor. We will collaborate with various alpaca farms in Poland to obtain material. As part of previous research, we have collected material from 400 alpacas in the form of DNA isolates from swabs, but this database will require various additions at different stages of the research.

References

Jones, M., Sergeant, C., Richardson, M., Groth, D., Brooks, S., & Munyard, K. (2019). A non-synonymous SNP in exon 3 of the KIT gene is responsible for the classic grey phenotype in alpacas (*Vicugna pacos*). *Animal Genetics*, 50(5), 493-500.

Baxter, B. P., Edens, L., & Wear, J. L. (2025). Colour phenotypes, genotypes and colorimetry of unprocessed North American Huacaya alpaca fibre, and comparisons with some other common natural fibres. *Small Ruminant Research*, 250, 107558.

Cerna, M. N. M., Stroupe, S., Kolb, E., Jevit, M., Li, S., Raudsepp, T., & Davis, B. W. (2026). Alpaca chromosome-level reference genome VicPac4 reveals a novel NOR-associated satellite specific to South American camelids. *Genomics*, 111212.