2023-08-02 WHFF Monogenetic Traits WG Meeting notes

🗓️ Date

Aug 3, 2023

👥 Participants

- @Lindsey Worden
- Alex Barenco
- Laszlo Bognar
- Linda Markle
- Jiri Motycka
- Tom Lawlor

Not present:

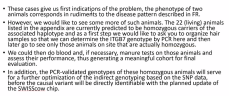
- Christa Kuehn

🗣️ Discussion topics

Item	Supporting Doc	Notes

Review Tom Lawlor's draft document for categorization of traits and discuss/finalize	 **WHFF Guidelin… 23.docx** 03 Aug 2023, 02:48 PM	• Everyone is general comfortable with the document's contents.
		• Two changes were made to the section on "Criteria to be used in determining if a new genetic trait should be added to the WHFF Master List" on page two.
		  ◦ Modified point 6 to specify that the trait should be a "monogenetic trait"
		  ◦ Added point 8 - "There must be publicly available documentation published in a peer-reviewed scientific journal that is available on the trait."
		• **ACTION**: The WG submits this document for WHFF Council adoption (see attachment - WHFF Guidelines for Interpreting New Evidence on Potential Monogenic Traits_Final_August2023.docx).
		  ◦ The WG requests that this information be included in the next WHFF newsletter that is sent out after its adoption, and that article should ask herdbooks to share this information with interested organizations (such as AI companies) within their regions.

Review document from WHFF web site (Request for Investigation of Newly Observed and Previously Unknown Genetic Trait) and make suggestions for improvement and distribution/promotion	 **WHFF-New-Ge… 23.docx** 03 Aug 2023, 02:58 PM	• Group reviewed the document ahead of the call
		• Proposed edits:
		  ◦ Define who should be completing this form; it should be herdbook or AI company, or similar authority
		  ▪ This form should not be used by individual breeders to report a single abnormal calf born on their farm.
		  ◦ Add question about whether or not a name has been established for the condition.
		• **ACTION**: The WG submits a modified document for WHFF Council adoption (see attachment - WHFF-New-Genetic-Trait-Reporting-Process_V2_August2023.docx).

		◦ The WG suggests that this topic be covered at some point during the World Conference in November ▪ Discussion on how to describe the potential new genetic trait and how to present this information to WHFF for consideration and review.
Update on BLIRD		• Coding has been approved by WHFF Council - “LR” • Alex presented some slides from the University of Berne (just for the WG, not public information yet) ◦ Three homozygous females observed, confirmed by PCR testing ▪ Two had some level of symptoms (loose manure and stunted growth compared to herdmates, poor production in the lactating cow), the third was normal. ▪ Researcher on vacation, will resume further research when they are back • Laszlo reported talking to French scientists; found frequencies similar to France in Holland. Calculated frequency is reported to be higher than 5%. • WG is still looking for any scientific papers, publications, documentation - more technical information needed to answer open questions. Only documentation that has been found so far is press releases. • **ACTION:** At this point, the WG opinion is that BLIRD best fits the categorization of being a “genetic condition under investigation.” Based on the information available, it does yet not meet the criteria for inclusion on the master list. More scientific description and information is needed. The WG does encourage herdbooks and other organizations who have test results for this condition to use the WHFF-approved coding of LR (LRF, LRC, LRH). ◦ Additional commentary from the WG: When we think about these traits, we like to have a phenotype that we can observe, and we work backwards to the biological function that broke down to create the phenotype. There is no physiological function that they are describing and proposing as a simple monogenetic defect. As of now, we have no technical description or explanation of the biological function that they think this gene is affecting that is causing this phenotype.
Update on Early Onset Muscle Weakness Syndrome		• Lindsey and Tom provided an update on this trait from the Holstein USA perspective. • HAUSA is using the following to describe this condition at this time: ◦ **Naming:** Early Onset Muscle Weakness Syndrome OR Early Onset Skeletal Muscle Weakness Syndrome. ◦ **Haplotype Codes (to be publicly available this fall)** ▪ Codes of 0 to 4 will be used to denote an animal’s haplotype status. • 0 Non-carrier: free of HMW • 1 Carrier: haplotype confirmed with pedigree information • 2 Homozygous: confirmed on both sides of pedigree • 3 Suspect carrier: haplotype origin could not be confirmed from pedigree • 4 Suspect homozygous: probable carrier and may be homozygous; origin of haplotypes could not be confirmed from pedigree ◦ **Direct test codes**: follow WHFF naming convention of MWF, MWC and MWH • Holstein USA is NOT requesting it to be added to the WHFF Master List at this point in time. As of today, lingering questions remain that we do feel that are

		moving toward resolution. Would feel more comfortable bringing this to this WG within three months (by November). ◦ We do have a phenotype, but it is occurring at a time when not much recording is typically done on the farm, and easily explained by other common ailments. ◦ We do have homozygous animals who are normal/healthy (including a proven AI sire with several thousand offspring); penetrance is not 100% ▪ Low incidence of homozygous affected; testing volume is low for unthrifty young calves. ▪ More research is needed to determine if environment is a factor or additional genes at play. ◦ HAUSA is encouraging breeders to do direct gene test and share those results with the breed organization. Those results are being shared with USDA used to improve haplotypes calls which need refinement before public release. ◦ **ACTION**: At this point, the WG opinion is that Early Onset Muscle Weakness Syndrome best fits the categorization of being a "genetic condition under investigation." Based on the information available, it does yet not meet the criteria for inclusion on the master list. The WG recommends that if herdbooks are recording direct test results for the CACNA1S mutation that they use the proposed three-letter coding (MWF, MWC and MWH).
Next meeting?		The Working Group will meet promptly when we have something to react to for either BLIRD or MW. WG members will continue to monitor information on both conditions within their home regions and share information as soon as it becomes available, and a meeting to discuss can be scheduled.

✅ Action items

- [ ] The WG submits this document for WHFF Council adoption (see attachment - WHFF Guidelines for Interpreting New Evidence on Potential Monogenic Traits_Final_August2023.docx).
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- [ ] The Working Group will meet promptly when we have something to react to for either BLIRD or MW.
- [ ] Lindsey to ask Suzanne - is there follow up or info we need to share with the ICAR DNA group related to MW or BLIRD?