

## **Summary of Major Items from Monogenetic Traits Working Group Reviewed by WHFF Council at November 20, 2023, Meeting**

1. The Council reviewed and approved the accompanying document, "*WHFF Guidelines for Interpreting New Evidence on Potential Monogenetic Traits.*" Note that the following sentence was added at the end of the third paragraph, at the suggestion of the Council, "However, a primary responsibility of herdbook organizations is to protect the health of the Holstein breed, and the viability of breeders of Holstein cattle around the world."
2. The Council reviewed and approved the accompanying document, "*Request for Investigation of Newly Observed & Previously Unknown Genetic Traits.*" This document should replace the document that is currently on the website, found at the link called "WHFF New Genetic Trait Reporting Process." The placement of the document on the web site should be reviewed so that it is easy to find.
3. The Council reviewed and approved the WG recommendation for a new formal process to be used for evaluating potential new monogenetic traits that may arise in the future, outlined below:

### **WHFF Process for Evaluating Potential New Monogenetic Traits**

1. Individuals or organizations who believe they have identified a potential new monogenetic trait should complete the form on the WHFF web site ("Request for Investigation of Newly Observed and Previously Unknown Genetic Trait"), which will be sent to the Monogenetic Traits Working Group Chair, who will organize a WG meeting.
  2. The WG will meet to review the information provided. If a satisfactory level of detail is provided, the WG will set a time frame for a public comment period (ex: 90 days). At this time, information will also be shared with the appropriate ICAR groups (such as the ICAR DNA Working Group).
  3. If there is a direct gene test already available at the time of initial review, the WG will propose labelling recommendations that will be reviewed quickly by the WHFF Council.
  4. WHFF will publicize information regarding the newly identified Genetic Condition Under Investigation and notify member herdbooks of the public comment period.
  5. At the close of the comment period, the WG will meet again to review comments received and other available documentation and propose recommended action to the WHFF Council regarding the Genetic Condition Under Investigation.
4. The Council reviewed and endorsed the WG recommendation to designate BLIRD as a genetic condition under investigation, and requests that WHFF publicize the following information about BLIRD:

- a. Labelling recommendations to be used by herdbooks who are recording direct gene test results (LRF, LRC, LRS).
  - b. A link to the available pre-print article containing information about the mutation (<https://www.biorxiv.org/content/10.1101/2023.09.22.558782v1>)
  - c. The WG would also like to open a 90-day comment period for feedback on Early Onset Muscle Weakness Syndrome from WHFF members. The 90-day comment period starts at the time that the WHFF membership is informed. At the close of the comment period, the WG will meet again to review comments received and other available documentation and propose recommended action.
  - d. WHFF should also use this opportunity to promote this new process outlined in point 3 of this document.
5. The Council reviewed and approved the WG recommendations for herdbooks that are recording direct gene test results for the CACNA1S mutation that is associated with Early Onset Muscle Weakness Syndrome (MW): MWF (tested free), MWC (tested carrier), and MWS (tested homozygous). The WG would also like to open a 90-day comment period for feedback on Early Onset Muscle Weakness Syndrome from WHFF members. The 90-day comment period starts at the time that the WHFF membership is informed. At the close of the comment period, the WG will meet again to review comments received and other available documentation and propose recommended action.
6. The Council reviewed and approved the WG recommendations for herdbooks that are recording direct gene test results for SLICK hair coat variants: S(variant#)F (tested free); S(variant#)C (tested carrier); S(variant#)S (tested homozygous). For example, coding for SLICK1 variant would be S1F, S1C and S1S. This condition is not being added to the WHFF Master List at this time.
7. The Council reviewed and approved the WG recommendation for a new formal process to be used for periodically evaluating the genetic traits included on the WHFF Master List, and the recommendation to consider archiving of the traits Factor XI and Citrullinemia from the WHFF Master List. The WG would like to open a 90-day comment period to solicit feedback from member herdbooks the idea of archiving/removing either or both of these two traits from the WHFF Master List. The 90-day comment period starts at the time that the WHFF membership is informed. At the close of the comment period, the WG will meet again to review comments received and other available documentation and propose recommended action.
8. Regarding the request from the ICAR DNA WG to develop standards for the regulation of the labs that test for the genetic traits; the WHFF Monogenetic Traits WG would be interested in collaborating with ICAR on this work, given the extensive work they have done to develop other DNA lab accreditation guidelines. The intent would be to collaborate on a special project, not to permanently increase the size of either WG.