



WHFF 5505bi Summary of the WHFF Monogenetic Traits Working Group Meeting on June 19, 2024

By Lindsey Worden (Chair)

Attendees: Lindsey Worden (chair), Alex Barenco, Laszlo Bognar, Chiara Franzoni Migliorati, Christa Kuhn, Linda Markle, Tom Lawlor

The WHFF Monogenetic Traits WG had a meeting on June 19, 2024. This had been postponed from a planned meeting in May that had to be rescheduled due to low attendance.

We had perfect attendance of all Working Group members on the call and enjoyed welcoming new member Chiara Franzoni Migliorati to the group.

Adding Traits to the WHFF Genetic Trait Master List

The group reviewed results of a survey that was conducted of WHFF member herdbooks earlier this year, asking whether or not they would be in favor of adding Early Onset Muscle Weakness Syndrome (MW) or BLIRD to the WHFF Master List.

After a good amount of discussion, the WG makes the following recommendation to the WHFF Council: Elevate MW to the Master List and keep BLIRD on the "under investigation list"

The WG recognizes that BLIRD is widely discussed in Europe and many survey respondents indicated that they felt BLIRD should be elevated to the Master List. It is not that the science is not trusted, but BLIRD appears to be more of a polygenic genetic and not monogenetic. It is a recent recessive mutation causing a decrease in the robustness or vigor of animals. Lower survival rates and decreased production have been observed in homozygous affected animals. However, the proportion of genetic variance explained by BLIRD is quite small. Additionally, given its correlation with health and production, it's anticipated that the frequency of BLIRD will decrease, as some of its detrimental effects are already included in our genetic evaluations for health and production.

The group agreed that going forward, we need to have a more formal survey sent out about new conditions that asks about the technical details that are outlined in the document "Process for Evaluating New Genetic Traits" (<https://whff.info/wp-content/uploads/2024/01/Process-for-evaluating-Genetic-Traits.pdf>). Future survey questions would include things such as:

1. Is a direct gene test available for the condition in your country?
 - a. If yes, please list labs that offer testing.



2. What is the frequency of the defective variant in your country's Holstein population?
3. Other questions that would give the WG a better understanding of how the condition is showing up in each respective country.

Archiving Traits from the Master List

After a brief discussion, the WG reached consensus that it made sense recommend to WHFF that both Factor XI and Citrullinemia be removed from the Master List.

The WG feels it is important these traits are not simply "deleted" from herdbooks and removed from history. In order to help with this, the WG recommends creating two new categories that would be included on the Genetic Traits and Carrier Codes document (<https://whff.info/wp-content/uploads/2024/01/Genetic-Traits-and-Carrier-Codes-v4.pdf>)

1. Genetic Conditions under Investigation – this would appear above the currently listed "Suggested Genetic Conditions Under Investigation" and BLIRD would be the only condition under the newly created category. This category would include items that had been reviewed by the WHFF Monogenetic Traits WG and deemed to be important for herdbooks to record information on, but not to the level of being included on the Master List.
2. Archived Genetic Conditions - traits that had formerly been on the Master List but have been reduced to such a low frequency that they no longer qualify, based on WHFF review.

If these conditions are archived (and for any archived conditions in the future), it is recommended that herdbooks would mark any test results that they have for the condition as non-publishable, so they do not appear on documents, but that they do not delete any information from their herdbook.

Discussion on Clone Labelling

After the November 2023 meeting, WHFF had asked the WG to review current policies related to naming clones. The document existing WHFF Guidelines for Registering Clones (<https://whff.info/wp-content/uploads/2023/01/Guidelines-for-Registering-Clones.pdf>) was shared and reviewed. While the members recognize that some countries have regulations around registering clones or descendants of clones, we think the current guidelines provide sufficient information for each country to do the recording that is required. The WG does not recommend making any changes to the WHFF guidelines around clones at this point in time.

Update on Potential ICAR DNA WG Collaboration

Information was shared with the WG related to some recent meetings that have been held jointly between some members of the ICAR DNA WG and some members of this WG. The



two groups intend to have more integration and work more closely together going forward, and it is expected that joint meetings will continue this summer.

Summary of recommendations for WHFF

1. Create two new categories that would be included on the Genetic Traits and Carrier Codes document (<https://whff.info/wp-content/uploads/2024/01/Genetic-Traits-and-Carrier-Codes-v4.pdf>)
 - a. Genetic Conditions under Investigation – this would appear above the currently listed “Suggested Genetic Conditions Under Investigation” and BLIRD would be the only condition under the newly created category. This category would include items that had been reviewed by the WHFF Monogenetic Traits WG and deemed to be important for herdbooks to record information on, but not to the level of being included on the Master List.
 - b. Archived Genetic Conditions - traits that had formerly been on the Master List but have been reduced to such a low frequency that they no longer qualify, based on WHFF review.
2. Elevate MW to the Master List and keep BLIRD on the “under investigation list”
3. Move Factor XI and Citrullinemia to the newly created category of Archived Genetic Conditions.