



WHFF 5407b Monogenetic Traits Working Group Report March 1, 2024 by Lindsey Worden

The WHFF Monogenetic Traits Working Group has not met since our November Council meeting, but there has progress made in moving our initiatives forward.

Updates and new documents approved at our Council meeting in November have been added to the WHFF web site at <https://whff.info/genetic-traits/>. This includes an updated "Request for Investigation of Newly Observed and Previously Unknown Genetic Traits" form; posting of the new "Process for evaluating Genetic Traits" document; and updating of the WHFF Genetic Traits and Carrier Codes page to reflect the newly approved codes.

On January 4, 2024, a survey was sent out to WHFF members inviting comments regarding adding BLIRD and Muscle Weakness to the WHFF Master List, and archiving Factor XI and Citrullinemia. A 90-day comment period is open on the survey, which closes April 3, 2024. A reminder will be sent out to those who received the survey, reminding them of this deadline.

Regarding a collaborative effort with the ICAR DNA WG to establish guidelines for laboratories performing genetic condition testing, Lindsey Worden, László Bognár and Suzanne Harding met with ICAR WG members Romy Morrin-O'Donnell, Brian Van Doormaal and Ezequiel Nicolazzi on February 28 to have an initial discussion on what the goals of such a collaborative effort might be. We are going to have a second call with this joint group in late March and adding in one additional colleague from each group with technical expertise – Dr. Tom Lawlor (WHFF WG) and Matthew McClure (ICAR WG). Key discussion points for our next meeting are:

1. Defining the lines between the ICAR DNA WG and WHFF regarding communication and a harmonized process, as well as defining tasks for this combined group.
2. Performing a technical view of the documents that WHFF has recently developed (primarily "Process for evaluating Genetic Traits") and looking for opportunities to enhance or expand upon that process.
3. Discussing the ICBF/ICAR Genetic Defects document and come up with a plan for who is responsible for maintaining it.

We are planning to have our next WHFF Monogenetic Traits WG meeting sometime in April, after we have the results of the survey, and have had a chance to meet again with the joint ICAR group. We will also plan to discuss clone labelling at that meeting, as requested by the Council at our November meeting.