

WHFF 5505ii Monogenetic Traits Survey Survey Results

By Suzanne Harding (SG)

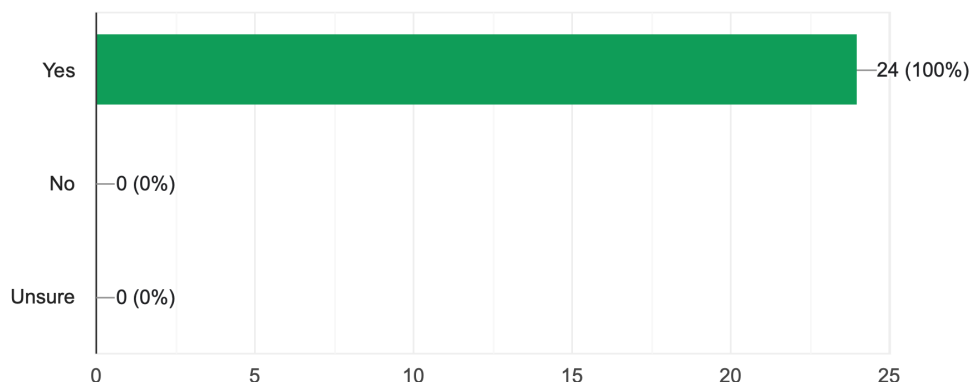
Background

At the 54th WHFF Council meeting it was agreed that a survey should be sent out to the WHFF membership regarding the changes to certain traits on the Master list of Genetic Traits.

There were 24 responses out of 42 WHFF members.

Early Onset Muscle Weakness is being considered as a new trait for the WHFF Master List of Genetic Traits. Do you approve the addition of this trait?

24 responses



Please add any comments about your decision about Early Onset Muscle Weakness.

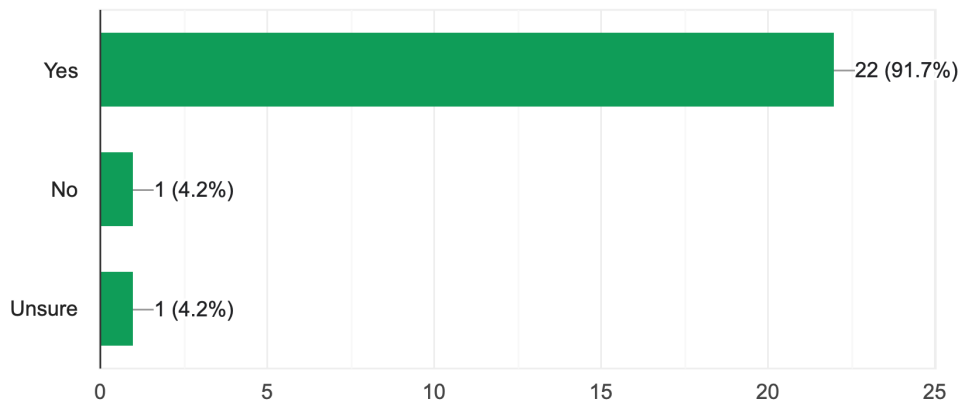
9 responses

- Prevalence in Ireland currently unknown
- None
- we consider it important that the acronyms to be used for tested animals are harmonised and clear for both direct and haplotype outcomes. For haplotype results what are the abbreviations to be used? Are they the same as for the direct test?
- We should be looking to code these as soon as possible including the haplotype version
- We support this direction, we have some uncertainty about if additional labelling will be introduced about unaffected homozygous carriers.
- From a breeding point of view, it is a mutation that has highly negative effects. Therefore it is important to screen the population and identify carriers to avoid economic losses due to this genetic disorder in the herd and eliminate deleterious mutation.
- As of January 2024, over 12,000 gene tests were available for research. Based upon these gene tests, the frequency of the MW deleterious allele in the Holstein breed is 5%. Our most recent year, i.e., heifers born in 2023, showed a penetrance rate of that 54%. The true level of penetrance is most likely higher as calf deaths are hard to track. The World Holstein Friesian Federation (WHFF) defined a "high-penetrance variant as a variant that segregates in a Mendelian pattern and in which 50% or more of the carrier individuals develop features of the condition". We recommend that Early Onset Muscle Weakness be added to the WHFF Master List.
- Current reports indicate that EOMW is of high enough prevalence to be of concern and is present in all major AI studs.

- More traits more possibilities.

BLIRD is being considered as a new trait for the WHFF Master List of Genetic Traits. Do you approve the addition of this trait?

24 responses



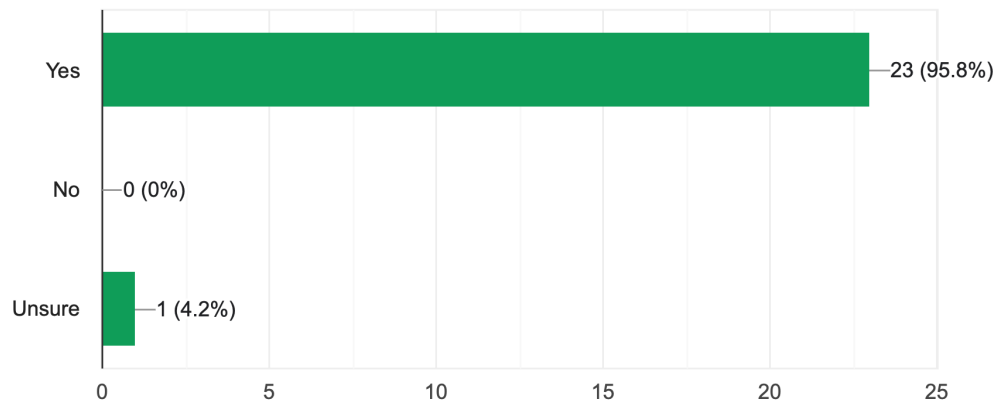
Please add any comments about your decision about BLIRD. (8 responses)

- Prevalence in Ireland currently unknown
- None
- we consider it important that the acronyms to be used for tested animals are harmonised and clear for both direct and haplotype outcomes. For haplotype results what are the abbreviations to be used? are they the same as for the direct test?
- Although we believe this is low frequency in the population
- We hope more analysis and research continues to be brought forward on this trait to provide more insight to its impact.
- From a breeding point of view, it is a mutation that has highly negative effects. Therefore it is important to screen the population and identify carriers to avoid economic losses due to this genetic disorder in the herd and eliminate deleterious mutation.
- The level of the genetic variation explained by BLIRD for heifer livability and milk yield has been calculated to 0.5%. This value does not meet the WHFF criteria for being added to their Master List of Monogenic Traits. It is recommended that BLIRD remain under investigation and that neither WHFF nor Holstein Association, USA declare BLIRD an official undesirable genetic condition. Conclusions: BLIRD 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. It is recommended that BLIRD continue to be treated as a genetic condition under investigation.
- While the BLIRD allele is present in today's bloodlines the impact is questionable. Not implying that BLIRD is not real. My understanding is that BLIRD homozygous animals are smaller and have a weaker immune system. As its not lethal I think this is a defect that should be monitored, breedings recommended to reduce its risk, but I don't think its bad enough for BLIRD testing to be required for importation or registration requirements.



The Working Group are recommending the archiving of Factor XI from the WHFF Master List of Genetic Traits. Do you approve the archiving of this trait?

24 responses

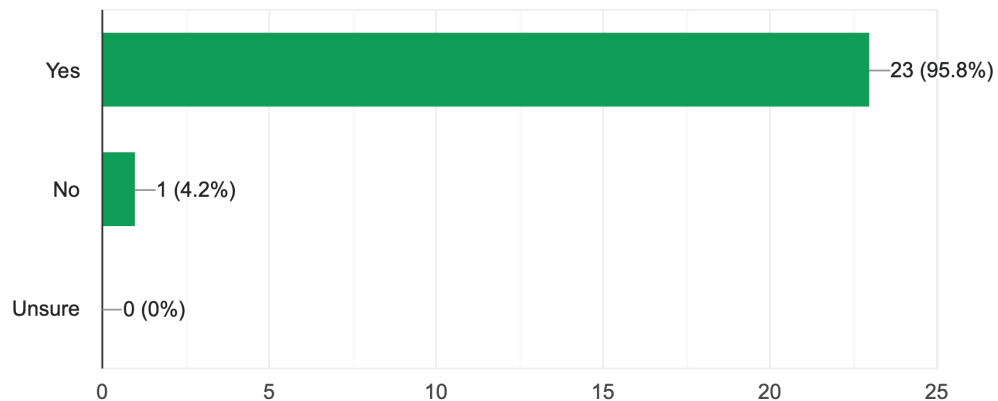


Please add any comments about your decision about Factor XI .
(5 responses)

- None
- We acknowledge this is low frequency now but it may become more in the future
- We support the archiving of this trait, an archived list of Genetic Traits, definitions and rationale for archiving should be made accessible. We would like guidance to be provided to herdbooks for how to handle their existing records for this trait, and how to handle new records for this trait that they may continue to receive.
- The number of carriers is very small (1 carrier was found out of approximately 100,000 tested).
- Via my work with others on the ICAR DNA working group we have been unable to find DNA from confirmed carrier or affected Holstein Factor XI animals. Some labs are using alternative ways to validate their Factor XI tests which are scientifically valid. The labs we have talked to also note that they have not come across any animals that their tests show as Factor XI carriers. I truly wonder if this was a falsely identified genetic defect given the challenge this group has had and we represent/have talked to most of the major global livestock genotyping labs

The Working Group are recommending the archiving of Citrullinemia from the WHFF Master List of Genetic Traits. Do you approve the archiving of this trait?

24 responses



Please add any comments about your decision about Citrullinemia. (5 responses)

- None
- We do still see this from time to time
- We support the archiving of this trait, an archived list of Genetic Traits, definitions and rationale for archiving should be made accessible. We would like guidance to be provided to herdbooks for how to handle their existing records for this trait, and how to handle new records for this trait that they may continue to receive.
- The number of carriers is very small (2 carriers were found out of approximately 100,000 tested).
- I believe that the carrier frequency of Holstein Citrullinemia is low enough to not be of concern any more to any country or Holstein farm

If you have any other thoughts on this topic you would like to share with the WHFF Council, please comment below. (7 responses)

- None
- it would be useful to understand the behaviour of the various countries with regard to bulls carrier of BLIRD or Early Onset Muscle Weakness: are they still admitted to artificial insemination?
- it would be nice if member countries had a harmonised approach to this aspect as well.
- It would be helpful to understand what 'archiving' means in reality
- We accept WG recommendation
- No
- I am sending a write-up on our BLIRD research to Suzanne Harding.
- Glad that WHFF is reviewing this

Summary

1. 24 respondents
2. Early Onset Muscle Weakness - YES 24/24
3. BLIRD - Yes 22/24, NO 1/24, UNSURE 1/24
4. archive Factor XI - YES 23/24, UNSURE 1/24
5. archive Citrullinemia - YES 23/24, NO 1/2