



Genetic Condition Under Investigation: Early Onset Muscle Weakness Syndrome

*This document is related to the condition previously referred to as
Calf Recumbency.*

Last year, farmers in several states contacted researchers at Pennsylvania State University about a suspected genetic condition characterized by a calf's inability to stand up or remain standing if assisted, a condition that has been referred to up to this point as calf recumbency. As the industry takes steps to further understand this condition, Holstein Association USA is now describing this genetic condition under investigation as **Early Onset Muscle Weakness Syndrome** (abbreviated MW).

What We Know Today

According to current research, this disorder usually begins within the first two months of life, where calves exhibit muscle weakness, become unthrifty and succumb to secondary health problems, such as pneumonia, or are euthanized. No known treatments are available at this time. There is some confusion about this condition due to inconsistency in describing its severity, reports of eventual recovery and most importantly, the underreporting of deaths. More genotyped heifers with pedigree data and livability records are needed to develop a clearer picture.

Despite these difficulties, there is evidence to suggest that the genetic condition is due to a single recessive mutation in the CACNA1S gene. Researchers believe that they have narrowed down the putative source of the mutation and its location. It occurs within a haplotype that is common among a large portion of the Holstein breed, which makes it difficult to differentiate a mutated haplotype from the normal haplotype.

More information will be made available as the new data is reviewed and analyzed. In the meantime, extra attention should be placed upon your mating program, avoiding close inbreeding, and staying away from using a bull on a cow with a similar pedigree. The negative effect of this undesirable genetic condition is believed to occur only when the mutated gene is inherited from both parents.

At this time, Holstein Association USA has NOT declared this disorder as an officially recognized undesirable genetic condition and is referring to it as a "Genetic Condition Under Investigation." As such, any available test results will not be displayed on Official Holstein Pedigrees at this time. A determination on that step will be made later this summer as more research is done to better understand the condition.

Available Testing Options

Two genotyping labs, Genetic Visions (www.geneticvisions.com) and Feanix Bio (www.feanixbio.com), are offering a commercially available direct gene test for the CACNA1S mutation associated with this condition. Interested breeders may contact the labs directly, or the HAUSA Genetic Services department for more details.

Testing of AI bulls and elite heifers has started. Direct gene test results have been published by some AI organizations on their websites. Holstein Association USA is endeavoring to aggregate all direct test results that are available, so they may be shared with USDA to aid in improving the accuracy of a haplotype test.

At the time of this writing, the results of the haplotype test are showing an unacceptably high rate of false positives. Once the accuracy of the haplotype test has been improved through the inclusion of direct test results, HAUSA will request that the haplotype for this condition be made available through normal distribution channels.

To assist breeders in determining if bulls they have used or may be interested in using have been tested, HAUSA will be publishing a list of bulls with NAAB codes who we have direct gene test results on file for on our web site. This list will be updated periodically as new results are received.

Data Collection Initiative Ongoing

Holstein Association USA requests that any Holstein breeder or organization who has direct test results for this mutation, for males or females, to share those with HAUSA so that data may be aggregated and shared to USDA to improve the haplotype test and conduct internal research to better understand the genetic condition.

Direct test results for any Holstein male or female can be emailed to LabResults@holstein.com, and at a minimum must contain the animal's registration number, clearly labelled direct test result, and an indication of which lab performed the test. Organizations wishing to transmit genetic condition results data to HAUSA on an ongoing basis may contact the email address above to request preferred file format specifications.

What to Do If You Think You Have an Affected Calf

Holstein Association USA has maintained a file of animals reported to have been born with abnormalities, using the Abnormality Report Form (https://www.holsteinusa.com/pdf/forms_apps/abnormality_report.pdf). If you believe you have an affected calf, please complete this form, and email it to

LabResults@holstein.com, or you may contact the HAUSA Genetic Services department at 800-952-5200.

Outstanding Questions

Holstein Association USA has identified a number of questions that need to be answered to gain a better understanding of this condition. If you believe you have information that would help in any of these areas, please contact LabResults@holstein.com.

- A clear phenotypic description of the genetic condition under investigation needs to be outlined, to avoid misclassifying animals who are affected who are actually not.
- We would like to determine if there is an effect on early embryonic death.
- We still don't have a true picture of percent penetrance from controlled research setting of carrier Sire X carrier MGS matings.
- We still don't have an explanation of why some affected homozygous animals are able to survive. There is a theory that different herd management can have an impact on its penetrance, yet we don't have any advice on what management practices impact whether or not calves are affected.

We appreciate the industry's interest in this condition and cooperation on the investigation and will provide updates as more information becomes available.

References

- Dechow, C.D., E. Frye, and F.P. Maunsell. 2022. Identification of a putative haplotype associated with recumbency in Holstein calves. *JDS Comm.* 3:412–415. <https://doi.org/10.3168/jdsc.2022-0224>.
- Dechow, C. 2023. Mutation sometimes leads to calf recumbency. *Hoard's Dairyman*.
- [Joint Industry Statement: Recumbency in Holstein Calves](#) (April 3, 2023)