

What to Know About MD

Learn more about the new novel defect discovered by researchers.



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The American Hereford Association (AHA), in collaboration with Dr. David Steffen and Dr. Jessica Petersen at the University of Nebraska-Lincoln (UNL), has discovered a novel defect in Hereford cattle called **Mandibulofacial Dysostosis (MD)**. This genetic defect is considered autosomal recessive, meaning an affected calf must have two carrier parents.

Overview

The AHA was first contacted in early March 2020 about a calf with facial deformities, and per AHA rules, the case was referred to Dr. Steffen at UNL for diagnosis. Additional calves were reported from two other herds in the following weeks. The following are the findings from Dr. Steffen and Dr. Petersen:

- Four Hereford calves were presented in March and April of 2020 with unusual defects of the face and jaw. Autopsy, evaluation of breeding records and genotyping were a cooperative effort.
- The calves originated from three herds, each in a different state, making an environmental cause unlikely. Autopsy of the calves revealed shared, unique abnormal facial features. Breeding records demonstrated a common sire in the pedigree of both the sire and the dam of each affected calf. The similarity in the defect among the

calves and the pedigree analysis together suggested a new deleterious recessive mutation may exist.

- The anatomic features overlap with a variety of other facial defects and can include cleft palate, brachygnathia (short jaw) and campygnathia (crooked jaw or face). These are variably present in affected calves (Figures 1-3) and are sometimes dramatic (Figure 3). The unique and consistent hallmarks of the condition include unusual bilateral skin tags just behind the corner of the mouth (Figures 1-3). These tags are attached to an unusual bone formation. There may be additional skin tags near and/or below the ears. A ridge of Meckel's cartilage, a structure usually present only during embryonic development, is retained in these calves and attaches to the skin tag. This cartilage is encased in bone as it is followed from the skin tag toward the base of the ear. This bone attaches specifically to the zygomatic process of the temporal bone (just above the articulation of the jaw). The calves' ears are sometimes slightly small and floppy. Muscles of the jaw are underdeveloped, and calves may have an elongate oral opening appearing as an exaggerated smile. The nursing reflex is present, but nursing is not vigorous. Calves with

Figure 1



Figure 2



Figure 3



the additional cleft palate and/or severely shortened or crooked jaws are debilitated in their ability to nurse. Calves with the defect are live born but are not able to thrive.

Utilizing whole genome sequencing, the research team at UNL identified a causative mutation from the animals submitted and the animals directly related to the affected calves. Next, UNL developed a single-marker assay to test additional animals to validate the test and to continue to verify the implicated genetic line. The research team at UNL is working with Neogen® to develop a commercially available test so breeders can submit DNA for testing or can have prior samples on file at the AHA tested. This test will be a standalone test and is not part of any current genotype panels on which animals have been tested. The AHA expects this test will be available in the coming months for breeders to utilize.

Testing options

The research team at UNL is willing to test specific animals utilizing their own test to mitigate risk and to aid in identifying breaks in pedigree relationship transmission from established carrier animals. However, to ensure quality control, only artificially inseminated (AI) permitted bulls which have available semen will be allowed for initial testing. Since this is a single-marker test, parent verification is not possible on the assay developed at UNL, making it imperative parentage confirmation already be completed. For testing efficiency, UNL has requested only semen straws be submitted and samples be submitted through the AHA for batching and verification.

To test herd bulls, breeders need to contact the AHA via phone or email with information on bulls they plan to submit. Semen straws must be shipped to the AHA. UNL will not be able to accept semen submission directly from breeders because this test is completed outside the scope of UNL's regular diagnostic services in cooperation with research. Semen straws must be shipped thawed and with some form of protection with a frozen gel pack

or a frozen water bottle to: American Hereford Association, Attn: Shane Bedwell, 11500 N Ambassador Dr., Suite 410, Kansas City, MO 64153. Place the cold pack inside a ziplocked or similar bag to prevent condensation from damaging the paperwork, samples and shipping containers.

The AHA suggests first testing impacted pedigrees in order to help identify breaks in transmission down the pedigree, but all AI-permitted bulls with semen available may be submitted for testing. Breeders should expect typical turnaround time for genotyping when submitting for MD testing at this time.

To aid in determining which bulls to test, breeders can utilize the potential defect carriers report generated through MyHerd, but understand this tool constantly updates. Therefore, the AHA recommends breeders run the report periodically over the next couple of months as pedigree relationships will

be broken as more animals are tested and identified as either carriers or individuals free from the condition, which could implicate animals in other herds. Breeders can find animals that have been tested for MD at *Herfnet.com*.

The AHA appreciates your cooperation with reporting cases of affected calves in order to identify this novel defect. Truly, it was a team effort between the breeders, UNL and the AHA staff in quickly addressing this genetic condition. Fortunately, with new technology and advancements, breeders can work around this defect to continue to breed carrier animals if they so choose, as opposed to totally culling affected genetic lines. The AHA will update the membership as more information becomes available on the commercial test at Neogen. We appreciate our strong membership and thank breeders in advance for their patience and cooperation. **HW**

Identified carriers

On June 5, the American Hereford Association (AHA) published 11 animals (below) that were identified as a carrier for Mandibulofacial Dysostosis (MD) and are directly involved in the cases submitted to the University of Nebraska-Lincoln for research. These 11 animals have been identified on *Herfnet.com* as Mandibulofacial Dysostosis Carrier (MDC). Please note the following list was last updated June 5.

Animal name	Registration number	Defect
SHF WONDER M326 W18 ET	42991698	MDC
NJW 73S W18 HOMEGROWN 8Y ET	43214852	MDC
NJW 73S W18 HOMETOWN 10Y ET	43214853	MDC
SHF ACCESS Y90	43379349	MDC
K KING 400	43565031	MDC
GO MS ACCESS C81	43584089	MDC
GO MS ACCESS E24	43796704	MDC
GO KING E43	43796809	MDC
RVF SWEET PEA SHES REAL QUIET	43810058	MDC
GO KING F42	43910077	MDC
PERKS CATO RUMMY 8011 ET	43937392	MDC

If a suspect calf is presented, please alert the AHA or Dr. Steffen at the Nebraska Veterinary Diagnostic Center. Both parties can assist with proper diagnosis and parentage verification for this or any other abnormalities you may encounter. If you identify suspect calves in your herd, be alert for testing availability as screening at-risk animals in the breeding population is critical to prevent future losses. **HW**