Repro Tracks

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Testing embryos for genetic defects

Arthrogryposis multiplex (AM) and neuropathic hydrocephalus (NH) have threatened Angus breeders with the possible loss of valuable genetics from cows that are carriers of the defects. Breeding carrier cows (AMC or NHC) to non-carrier sires (AMF or NHF) results in one-half the offspring being "free" of the defective gene. Superovulation and embryo transfer (ET) are effective methods of producing AMF or NHF offspring from valuable carrier cows. This process would be more efficient and less costly if the embryos that are AMF or NHF could be identified prior to transfer.

Breeder question:

I am using ET in an effort to "capture" some of the exceptional genetics of a few of my donor cows that tested "positive" as carriers of AM or NH. I know that if I breed a carrier donor to a sire that is AMF or NHF, one-half of the embryos will still be carriers of the defect. Is there any way to test the embryos and not transfer the ones that will be carriers?

Response: Testing freshly collected embryos for AM and NH status is possible if a biopsy of the embryo can be collected. An ET company in the Midwest has worked with Jonathan Beever at the University of Illinois to develop and verify the procedure. They originally sent Beever biopsies from 25 embryos collected after mating an AMC donor and an AMF sire. Carrier status was able to be determined from 92% (23 of 25) of the biopsy samples. Thirteen were determined to be AMF and 10 were determined to be AMC. None of the embryos from the original experimental group were transferred.

Although the original experiment demonstrated that embryo biopsies could be tested for AM or NH, an ET practitioner would only be able to discard carrier embryos and transfer those fresh embryos that were AMF or NHF if a "chute-side" test for the defects were available. The chute-side test would allow immediate determination of each embryo's carrier status before it was to be transferred. Unfortunately, no such test is available. To date it has taken five to 14 days to get results of genetic testing after collecting embryo biopsies.

An alternative to chute-side testing and disposal of carrier embryos is to biopsy and transfer all embryos while waiting on the test results. Later, when results are available, recipients that received carrier embryos could be treated to terminate the pregnancy. When the ET company working with Beever transferred 23 fresh embryos after collecting a biopsy, 17 resulted in pregnancies (74%). It turned out nine pregnancies were from AMF embryos and eight were from AMC embryos. Those recipients carrying AMC embryos had the pregnancy terminated and were resynchronized for another transfer. This system requires a surplus of recipients and does not avoid the costs associated with embryo collection and transfer, but it does result in only maintaining pregnant recipients that will produce calves free of the genes for genetic deformities.

Finally, the ET company doing this research is performing the biopsy and then freezing the embryos. This allows them to store the frozen embryos while they await the genetic testing results. They intend to thaw and transfer only those embryos that test AMF and NHF. This work will be completed later this fall. Although this process has the potential to be more efficient, exposing the embryos to two "insults" (biopsy followed by freeze/thaw) may result in lower pregnancy rates than when only one procedure (biopsy or freeze/thaw) is performed before the embryos are transferred.

In addition to the normal ET costs, the cost of the procedure for genetic testing of biopsies from embryos has been $50 for the initial test (AM or NH) and an additional $3 per biopsy for each additional test. The sex of the embryos can be determined from the biopsy as well as the carrier status for the two genetic defects.

Breeder question:

We have had a large proportion of our embryo donor cows test positive for AM (34%) and NH (23%). Over the past two years, our calving rates following ET have also decreased by 10% to 15%. Is it possible that the incidence of calves with AM or NH resulted in more abortions and that has been the cause of our lower ET calving rates?

Response: Numerous calves with hydrocephalus detected at an ultrasound pregnancy diagnosis have been reported to be lost during the last two-thirds of pregnancy. Hence, based on field reports, it is likely that when an NHC sire and an NHC dam are mated the chance of spontaneous pregnancy termination is increased.

If you were breeding NHC donors with semen from NHC sires, it is possible that the incidence of embryonic loss in your herd increased, and therefore calving rate decreased. Calves that are homozygous for the recessive allele controlling NH, and should exhibit the symptoms of hydrocephalus at birth, have been reported to experience a higher rate of embryonic loss. Those losses could have been responsible for a decrease in your calving rates.

Conversely, matings that result in homozygous recessive AM calves may not decrease pregnancy rate or calving rate. Although there is no conclusive scientific study, it is believed that most homozygous recessive AM calves are carried to term. Although the AM defect is lethal, calves are believed to be born alive (or at full term) in most cases.

Reducing or eliminating the incidence of genetic defects, especially NH, may enhance the pregnancy and calving rates in some herds where the frequency of NH carrier animals has been high and the use of NH carrier sires occurred in the past. However, it is imperative to remember there are many factors other than the possible occurrence of genetic defects that influence pregnancy rate in a herd.

Editor's Note: *Bill Beal is a beef cattle reproductive physiologist at Virginia Tech. He conducts research involving estrus synchronization, artificial insemination, ET and the use of ultrasound technology. If you have questions or comments related to the reproductive management of cows or bulls, contact him at wbeal@vt.edu.*