

# Test On Its Way

Research identifies mutation, source of NH; test is in the pipeline.

by *American Angus Association staff*

**W**hile April's announcement of neuropathic hydrocephalus (NH) as a genetic defect might have felt like rain on the Angus parade, breeders can take heart that April showers bring May flowers.

Jonathan Beever, University of Illinois, announced in early April the spirit-dampening news that the cases of hydrocephalus uncovered while documenting arthrogryposis multiplex (AM) were determined to be genetic. However, that was quickly followed by news that a single recessive gene unrelated to AM was identified as the cause of the mutation, allowing a DNA test to be developed to identify animals as carriers (NHC) or free (NHF) of the genetic abnormality.

Preliminary genotyping of AI bulls in research to formulate the DNA test revealed 91 of 934 bulls (roughly 10%) carried the mutation. But with a test expected to be commercialized in May, producers can begin the process of eliminating the lethal gene from the Angus population, assuring commercial customers of Angus members' commitment to breed purity and the economic livelihood of their customers.

## How we got here

At the encouraging of David Steffen, its lead advisor in matters of genetic abnormalities, the American Angus Association requested in September 2008 producers report any incidences of an abnormality originally called curly calf syndrome. Producers responded with more than just cases of AM, including 15 cases of an extreme form of hydrocephalus.

Steffen notes that with increased reporting of abnormal calves, lower-frequency conditions within populations often become apparent and warrant investigation.

At that point it was unclear whether the condition was genetic or environmental, Beever says. While a pedigree analysis of the calves did show common ancestry, it was unclear whether that was due to genetics or due to a bias in submissions.

In February 2009, at Steffen's request, the Association asked members to report calves displaying hydrocephalus or fawn calf syndrome (see "Report Abnormal Calves," April 2009 *Angus Journal*).

Beever reports that using DNA analysis of the reported calves, they were able to confirm NH as being genetic and caused by a recessive mutation (see "By the Numbers: Visualize a

## What is NH?

David Steffen described the neuropathic hydrocephalus (NH) phenotype, saying calves are born near term and weigh 25-35 pounds (lb.) at birth (see "Report Abnormal Calves," April 2009 *Angus Journal*). The cranium is markedly enlarged (volleyball- to basketball-sized). The bones of the skull are malformed and appear as loosely organized bony plates that fall apart when the cavity is opened. The cranial cavity is filled with fluid, and no recognizable brain tissue is evident. The spinal canal is also dilated, and no observable spinal tissue is found.

simple recessive," April 2009 *Angus Journal*). Further research, he says, identified the specific mutation, allowing development of a DNA-based test to identify carriers of the defect.

Beever notes that while the frequencies of AM (8.5%) and NH (10%) are basically the same, the number of AM calves reported is two to three times higher than that of NH-affected calves. "This means there is a potential for 50% to 70% fetal loss in NH," Beever says.

## Test on the horizon

"Similar to the work that was performed for AM, we have had the opportunity to test more than 1,000 DNA samples for this mutation," Beever notes in the April 9 research update posted to [www.angus.org](http://www.angus.org). All calves submitted with the condition were homozygous (carrying two copies of the gene) for the NH mutation.

"Using DNA from affected calves and their relatives, we have identified the founder of the mutation causing NH," Beever notes in an April 13 update. "All NH-affected calves trace back to the common ancestor, GAR Precision 1680, who is confirmed to be heterozygous for the mutation. However, both parents of GAR Precision 1680 have tested as NHF, thus allowing us to conclude that the mutation originated in this animal."

In that update, Beever notes that several thousand animals have been genotyped for the mutation, including a current list of AI sires submitted by members of the National Association of Animal Breeders (NAAB).

AI organizations requested the Association provide to the membership the identity of and preliminary test results of the 934 Angus AI sires genotyped in the initial research

(available at [www.angus.org](http://www.angus.org)). The Board agreed, but with the caution that the results are preliminary. As of this writing, the test has not yet been validated.

Until the Association can establish a protocol for the direct receipt of results of a fully validated commercialized test from a lab approved by the Association, it will not alter or amend any animal's registration or Performance Pedigree Certificate.

## Preliminary testing

"Based on information we have received, we hope to have a test in May," notes Bryce Schumann, CEO for the American Angus Association. That will depend on the time it takes for validation and licensing of commercial labs to conduct the test and develop a protocol for submission of results to the Association.

The Association's task force on genetic defects will be meeting in the near future to develop policy and rules for handling carriers of the NH gene mutation, Schumann says. It's been a longstanding policy within the Association that once an animal is registered it will remain registered.

Genetic defects are not unique to Angus nor a new phenomenon, Schumann says, noting similar scenarios in 1978 (see "Movin' Forward," page 28). "Today we have a technological advantage allowing us to identify carriers and noncarriers of simple recessive mutations with confidence, rather than identifying animals as suspects and requiring long, drawn-out and expensive progeny testing to prove an animal free of a defect."

The Association Board and staff will continue to move forward to ensure the success of the Angus breed and its members as quickly, accurately and efficiently as they can.

Meanwhile, there are plenty of flowers already blooming in the Angus business, which should not be overshadowed by NH. Commercial bull buyers have continued to demand Angus bulls at sales throughout the Plains states this winter and spring. The AngusSource® program that documents source, age and genetics posted a record month of enrollments in March 2009, and demand for *Certified Angus Beef*® (CAB®) product is reaching record highs as well.

For more information on AM or NH, visit the Association's web site at [www.angus.org](http://www.angus.org).

