Genetic Defects in Beef Cattle: An Update

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Outline

• How are defects inherited?
• What defects exist in which breeds?
• Working with Commercial Cow-calf producers
• Working with Seedstock producers

Inheritance

• All the defects we will discuss today are inherited as simple recessives
• All affected calves must have two copies of the defective allele (homozygous)
• Any sire or dam that produces an affected calf must be a carrier of the defective allele (heterozygous)
• Any carrier will transmit the defect half the time

Inheritance Example

\[
\begin{array}{ccc}
\frac{1}{2} D & \frac{1}{2} d \\
\frac{1}{4} D D & \frac{1}{4} D d \\
\frac{1}{4} d D & \frac{1}{4} dd \\
\end{array}
\]

D = normal allele
d = affected allele
DD = homozygous normal, unaffected, non-carrier
Dd = heterozygous carrier, unaffected
dd = homozygous, affected

Mate two carriers (Dd)

Inheritance

• For many of these defects, the causative mutations have been identified, and diagnostic DNA tests are available
• Allows identification of carriers
• Identifies homozygous normal descendants of carrier ancestors

Genetic Defects

• Almost every beef breed has some identified genetic defect
• Some defects can cause embryonic loss
• Spontaneous mutations are rare, but do occur
**Dwarfism**

- At least three forms:
  - Snorter dwarf
  - Bulldog dwarf
  - Long-headed dwarf
- Several types were observed in Angus and Hereford in the 1950’s and 1960’s
- Also diagnosed in Brahmans and Dexters

**Snorter Dwarf**

**Reemergence of Dwarfism**

- In 2003, an Angus AI sire born in 1997, High Valley 7D7 of 4G9, was identified as a carrier of long-headed dwarfism
- The mutation was identified at Iowa State University, and is now testable
- Carrier bull was not widely used, carriers are rare, believed to be a spontaneous mutation

**Dwarfism**

- Not all dwarfs are “genetic dwarfs”
- Dwarf calves can occur when pregnant cows are infected by viruses at key points in embryonic/fetal development
- Veterinary diagnostic labs can classify dwarf calves as genetic/environmental by necropsy
**Osteopetrosis (OS)**
- Commonly known as Marble Bone
- Recently diagnosed in Red Angus
- Past occurrences in Angus, other breeds
- Several prominent Red Angus AI sires were identified as carriers

**Idiopathic Epilepsy (IE)**
- Diagnosed in Herefords (mostly horned) in 2003
- All carriers trace to HH Advance P242, born in 1982
- Almost all trace to his widely-used grandson, HH Advance 9012Y, born in 1989

**Tibial Hemimelia (TH)**
- Diagnosed in Shorthorns in 2000
- Also in crosses, mostly “show steer” bulls
- Most trace to Deerpark Improver, Irish import born in 1972
- Later a different mutation in the same gene was found in TKA Outcast, born in 2001

**Osteopetrosis**
- Calves aborted 10 to 30 days early
- Calves have short lower jaw
- Bones contain no marrow cavity, are fragile and easily broken
- Deletion in a gene identified on chromosome 4
- DNA test for Red Angus commercially available

**Idiopathic Epilepsy**
- Affected calves have seizures, often stress induced
  - Last for minutes, sometimes an hour
- Affected calves appear normal when not seizing
- Mutation identified, DNA test commercially available

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Tibial Hemimelia
- Calves born dead or die shortly after birth
- Twisted legs, fused joints, hernia, hole in skull, etc.
- DNA tests for both mutations available
- Some theorize that carriers may have been indirectly selected

Pulmonary Hypoplasia with Anasarca (PHA)
- Found in Maine-Anjou, some Chianina and Shorthorn, also in Dexter
- Most trace to Maine bulls Draft Pick (1989), Stinger (1985) or the Chi-mainne bull Payback (1992)
- Mutation probably preceded importation of Maines to the US

Pulmonary Hypoplasia with Anasarca
- Calves born dead or die shortly after birth
- Water-logged appearance, small lungs, no lymph glands
- Fetus begins to accumulate fluid by 6th month of gestation, gestation may be shorter than usual, calves may be enormous (> 200 lb.)
- Missense mutation identified, testable

Protoporphorgia
- Found in Limousin cattle
- Calves are sensitive to light
- Calves develop hair loss, sores, fail to thrive
- DNA test available since the mid-1990’s
- Defect largely removed from the population, potential carriers must be tested in order to be registered
**Beta-Mannosidosis**
- Found in Salers
- Calves exhibit weakness, incoordination, head-swaying, poor suckling reflex, other nervous system disorders,
- DNA test available since 1999
- Defect practically eliminated from the population

**Arthrogryposis Multiplex (AM)**
- Known as Curly Calf Syndrome, recognized as a genetic defect in Angus in 2008
- Most carriers trace to Rito 9J9 of B156 7T26, born in 1979
- Angus composites (SimAngus, LimFlex, Gelbvieh Balancer, Chiangus, Maine-Anjou, Salers, etc.) may also be carriers

**Arthrogryposis Multiplex**
- Born dead or die shortly after birth
- Small in size, diminished muscling, fixed joints, limbs twisted, spine twisted
- HUGE mutation (23,000 base pairs)
- Mutation discovered after only 2 months of research
- DNA test commercially available

**Neuropathic Hydrocephalus (NH)**
- Diagnosed in Angus in 2008, also in Angus crosses
- Affected calves were submitted to diagnostic labs as potential AM calves
- All carriers trace to G A R Precision 1680, born in 1990
- He is the original, spontaneous mutant, his sire and dam are not carriers

**Neuropathic Hydrocephalus**
Neuropathic Hydrocephalus

- Calves born dead, full term
- 25-35 lb. birth weights
- Basketball-sized head, full of fluid, no brain
- Single base pair mutation
- DNA test available

Hypotrichosis

- Hairlessness
- Found in Herefords (mostly polled), recognized decades ago, also found in Charolais, Simmental, dairy breeds
- Many current carriers trace to JR Nick the Butler P183, born in 1982, but many other carriers are unrelated

Any More?

- Fawn Calf Syndrome
  - Found in Angus, first reported in Australia
  - Subtle, non-lethal, calves appear hunched or crouched, less extension of upper limb joints
  - DNA test being developed
  - Some reported calves trace to Bon View Bando 598, born in 1988, or his great-grandsire, Premier Independence KN, born in 1981

Fawn Calf Syndrome

- Some calves are practically slick, hairless
- Others have extremely curly hair
- Not lethal, but calves often succumb to elements
- Mutation recently identified
- DNA tests will be available soon
Working with Commercial Cow-Calf Producers
- Testing is expensive (average $35/sample)
- Look at bull pedigrees to see if they are potential sires
- Test results of bulls’ ancestors may be available online
- Probably don’t need to test their bulls unless they have seen affected calves

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Use clean bulls (DD)

Working with Commercial Cow-Calf Producers
- Many have cows that are carriers for one or more defects
- As long as sires are not carriers, they will never have an affected calf
- Usually, all commercial producers need to do is to buy clean bulls from now on
- If they have had some affected calves, they might test the bulls they wish to keep

Working with Seedstock Producers
- Recognize this is an emotional issue
- A high percentage of seedstock suppliers have unknowingly sold some carrier bulls in the past few years
- Seedstock breeders will likely want to eliminate most carriers ASAP
- Most will test all animals that are potential carriers

Working with Seedstock Producers
- Most seedstock producers should be in better shape this year, as they should be able to test for most defects by weaning
- Association policies vary as to mandatory testing and registration of carriers
- Breed associations are a good source of information on the defects found in their breed