

## 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

D = normal allele  
d = affected allele

DD = homozygous normal, unaffected, non-carrier

Dd = heterozygous carrier, unaffected

dd = homozygous, affected

Mate two carriers (Dd)

	1/2 D	1/2 d
1/2 D	1/4 DD	1/4 Dd
1/2 d	1/4 Dd	1/4 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



## Dwarfism

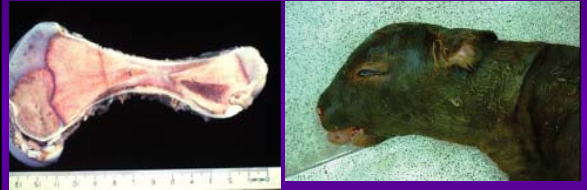
- Not all dwarfs are “genetic dwarfs”
- Dwarf calves can occur when pregnant cows are infected by viruses at key points in embryonic/fetal development
- Veterinary diagnostic labs can classify dwarf calves as genetic/environmental by necropsy

## 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



## Osteopetrosis



## 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 (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



## 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

## 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



## Tibial Hemimelia



## 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-maine bull **Payback** (1992)
- Mutation probably preceded importation of Maines to the US

## Pulmonary Hypoplasia with Anasarca



## 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 6<sup>th</sup> month of gestation, gestation may be shorter than usual, calves may be enormous (> 200 lb.)
- Missense mutation identified, testable

## Protoporphyrria

- 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



## 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 168o**, 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

## Hypotrichosis



## Hypotrichosis

- 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

## 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-grand sire, **Premier Independence K N**, born in 1981

## Fawn Calf Syndrome



## 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

## Inheritance Example

D = normal allele  
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Use clean bulls (DD)

	$\frac{1}{2}$ D	$\frac{1}{2}$ D
$\frac{1}{2}$ D	$\frac{1}{4}$ DD	$\frac{1}{4}$ DD
$\frac{1}{2}$ d	$\frac{1}{4}$ Dd	$\frac{1}{4}$ 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

