

PHA (Pulmonary Hypoplasia with Anasarca)

#### **Genetic Terms and Definitions:**

Phenotype: The characteristics of an animal that can be seen and/or measured.
Gene: A segment of DNA on the chromosome that codes for a specific trait and determines how that trait will develop.
Allele: A version of a gene. Alleles occur in pairs, one being inherited from the dam and one from the sire.
Dominant Allele: A dominant allele will always express itself and will suppress the expression of a recessive allele.
Recessive Allele: A recessive allele can only be expressed if a dominant allele is not present.
Heterozygous: The two alleles in a gene pair are not alike.
Homozygous: The two alleles in a gene pair are alike.
Pulmonary: referring to the lungs
Hypoplasia: incomplete formation
Anasarca: a general accumulation of serum fluids in various tissues and body cavities
PHA-affected animal: an animal that inherits the PHA gene from both parents, thus being homozygous for the gene.

#### **Definition:**

PHA is a genetic defect caused by a recessive gene that leads to abortion or the birth of a dead calf in its homozygous state. Associated dystocia can also lead to death of the cow. PHA is characterized by incomplete development of the lungs and severe subcutaneous fluid accumulation. It has an autosomal recessive mode of inheritance, meaning two mutated genes are inherited, one from each parent. Animals that are heterozygous carriers of the gene are not affected by it at all.

#### **History**:

PHA occurs in several cattle breeds, including Dexter, Maine-Anjou Shorthorn and related composite cattle. Research by Dr. Jon Beever at the University of Illinois identified the mutations responsible for PHA in cattle. The affected gene is the same for the different breeds, but the specific mutation associated with PHA in Dexter cattle is different from the Maine-Anjou/Shorthorn mutation.

The first publicly-identified PHA-carrier in the Dexter breed was Trillium Chabotte, ADCA #3168. His semen was tested in Australia, and the test result was reported through Dexter Cattle Australia, Inc. on December 5, 2008. This bull was born in Canada in 1985. He was collected for artificial insemination, and his semen was used in Canada and exported to Australia.

At a later date, PHA was traced back to Woodmagic Wheatear, an English cow born in the mid 70's. Wheatear's son born in 1982, Aldebaran Priapus, ADCA #2544, was found to be a PHA-carrier. He was also collected for AI and his semen was used widely in both Canada and the US. Priapus has 50 offspring registered in the ADCA, including Trillium Chabotte, so most PHA-carriers in the US trace their lineage back through Priapus.

The PHA gene confers risk with no potential for benefit. Most breeders who discovered they had PHA-carriers have worked diligently to eliminate it from their herds. They've done this by breeding PHA-carriers only to non-carriers and testing offspring. Most of them culled PHA-carrier bulls because of the risk of passing the gene through numerous cows. When they had excellent PHAcarrier cows, they bred for a non-carrier replacement heifer, then culled the carrier dam. With careful genetic testing and responsible breeding, PHA can be completely eliminated.

#### **Genetics**:

In order to be PHA-affected, a calf must inherit a PHA gene from both its sire and dam. PHA-affected calves (those that are homozygous for the gene) are either aborted or stillborn. Because of the anasarca that is associated with this condition, the PHA-affected calf may also swell tremendously with fluid, making delivery difficult and potentially endangering the life of the cow. Animals that are heterozygous for the PHA gene are not affected by it and are called PHA-carriers.

# American Dexter Cattle Association

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### **Responsible Breeding With PHA:**

Breeding two PHA-carriers together affords a 25% chance of a PHA-affected calf, a 50% chance of a PHA-carrier, and 25% chance of a non-carrier of PHA. It is, therefore, considered **strongly inadvisable** to breed two PHA-carriers together. Breeding a PHA-carrier with a non-carrier affords a 50% chance of producing a PHA-carrier, a 50% chance of producing a non-carrier, and 0% chance of producing a PHA-affected calf. This would be responsible breeding practice. Breeding two non-carriers of PHA will never produce a PHA-carrier or a PHA-affected calf.

## **Genetic Testing:**

Both ADCA-approved labs offer testing for PHA. This only detects the specific type of PHA found in Dexter cattle.

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