

PHA (Pulmonary Hypoplasia with Anasarca) FACT SHEET

This fact sheet addresses the most commonly asked questions about a genetic defect that has recently been confirmed by DNA testing in several breeds of cattle, including Dexters. The questions and answers presented herein are based on the conclusion that Pulmonary Hypoplasia with Anasarca (PHA) is the result of a mutation in a specific gene on cattle chromosome 6. Dr. Jon Beever, of AgriGenomics, Inc., has developed a genetic test that identifies the presence of the mutation in this specific gene.

1. What is Pulmonary Hypoplasia with Anasarca (PHA)?

"Pulmonary" refers to the lungs, and "Hypoplasia" means incomplete formation; thus "Pulmonary Hypoplasia" means incomplete formation of the lungs. "Anasarca" means a general accumulation of serum fluids in various tissues and body cavities; thus anasarca results in swelling and a swollen appearance. In order to be PHA-affected, a calf must inherit a PHA gene from both its sire and dam. Since the PHA gene is recessive, the lethal form of this condition is only expressed when the calf is homozygous for the PHA gene. PHA-affected calves 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.

2. What is a gene? What is the difference between a gene and an allele?

A gene is a sequence of DNA which provides the basic instructions or "code" for a certain trait or function. Genes are the basic units of the hereditary information that is passed from one generation to the next, and they can have alternate versions. The alternate versions of any specific gene are called alleles. Genes occur in pairs. For each pair of alleles, one is inherited from the sire, and one is inherited from the dam. Alleles can be dominant, co-dominant, or recessive relative to each other. The allele for PHA is recessive to its normal, non-mutated allele. The PHA allele does not have the normal sequence and number of base pairs needed to code properly for its functions. Since alleles occur in pairs, an animal that has one PHA allele and one normal allele will not be affected by PHA, but it will be a carrier of PHA. Since the normal allele is dominant, it will code properly for its functions and normal development of the lungs will occur even if the animal is a carrier of the PHA allele.

3. What is a mutation?

A mutation is a spontaneous change in the DNA sequence that makes up a gene. Mutations can range in size from a single DNA building block (a base pair) to a large segment of a chromosome. The PHA mutation in Dexter cattle occurs in the same gene as the PHA mutation in Maine-Anjou and Shorthorns, but it is a different change in the DNA sequence, so it is considered to be a different mutation. It results in the same outcome because the same gene is damaged and cannot perform its intended function.

4. Is PHA in Dexters due to a recent mutation?

We don't believe so, but the ability to conduct a DNA test for the PHA mutation is very recent. The Journal of Anatomy and Physiology in Great Britain, published in 1906, page 293, references the following: "Lesbre and Forgeot describe another achondroplastic calf which was born dead in an extraordinarily anasarctic condition. Dissection showed that both kidneys were absent." Since Lesbre and Forgeot were dealing with "bull-dog" calves that were found only among the pure Dexter-Kerry breed, they may have confused a PHA-affected fetus with a Chondrodysplasia-affected fetus, or it may have been a fetus affected by both conditions.

The first publicly-identified PHA carrier in the Dexter breed is 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. Although Chabotte has only 9 ADCA-registered progeny, according to the ADCA Online Pedigree, as of January 14, 2009, he has more than 3400 descendants in the ADCA registry.

5. What is a PHA-affected calf?

A PHA-affected calf is a calf born dead with underdeveloped lungs (Pulmonary Hypoplasia) and swelling caused by excessive fluid retention (Anasarca). When genetically tested, this animal has two alleles for the PHA mutation, so it is termed homozygous for PHA.

6. What is a PHA carrier?

A PHA carrier is an animal that has one PHA allele and one normal allele, so it is termed heterozygous for PHA. A PHA carrier Dexter appears normal, and there is nothing in its phenotype (appearance) that indicates it is a carrier of the PHA mutation.

7. Can an animal be a non-carrier of Chondrodysplasia and still be a PHA-carrier?

Yes, the two genetic conditions are not related to each other. Two different tests are used to determine the status of these two unrelated genetic conditions. An animal can be a carrier of both mutations, or it can be a carrier of one or neither of them.

8. Can a "bulldog" calf also be a PHA-affected calf?

Yes, a severely affected Chondrodysplasia ("bulldog") calf can also be a PHA-affected calf, but only if it is homozygous for both mutations.

A dead, deformed Dexter fetus that may be difficult if not impossible for a layman breeder to discern which condition, if either or both, lead to its death. Other factors may result in aborted or stillborn calves, so those factors should not be ruled out without a DNA test of the fetal tissues to confirm its PHA and Chondrodysplasia status.

9. If a bull sires a PHA-affected calf, what does that mean?

If a bull sires a DNA-confirmed PHA-affected calf, it means that the bull is a PHA carrier and that the dam of the calf is also a PHA carrier.

10. If a cow has a PHA-affected calf, what does that mean?

If a cow has a DNA-confirmed PHA-affected calf, it means that the cow is a PHA carrier and that the sire of the calf is also a PHA carrier.

11. If I've never had a PHA-affected calf, does that mean that my cows are non-carriers of PHA?

No, a cow's status as a non-carrier of PHA can only be determined by genetically testing the cow.

12. How do I test to determine the PHA status of my Dexters?

The ADCA is actively promoting PHA testing of all Dexter AI bulls to build the initial knowledge base needed to determine the extent or potential extent of the number of PHA carriers that may currently exist and to allow breeders to make informed decisions regarding their own breeding programs. This is a recently developed test, and, at the present time, only semen or blood samples can be tested in the United States. The ADCA hopes that a test procedure that uses tail hair samples will soon be available.

To submit semen or blood samples for testing: [Click here for the instructions and test application form](#)

The instructions and application form may also be obtained from the DNA Testing link on the ADCA website. Please note that semen samples from half of the approximately 75 ADCA AI bulls have been submitted for testing by members of the ADCA's Genetics and Pedigree Committee (GPC) and by others who have volunteered to submit samples. Some of these bulls have already been tested in Australia. Owners of past or present AI bulls who are willing to participate in the testing program are encouraged to contact John Potter of the GPC by email (j.potter@att.net) with the name(s) of the bull(s) which you are volunteering to test. Owners of recently- or currently-advertised AI bulls are being contacted if the GPC does not yet have samples from those bulls.

13. What can happen when a PHA carrier is bred to another PHA carrier?

Every time a PHA carrier bull is bred to a PHA carrier cow there is a 25% probability of producing a dead PHA-affected calf, a 50% probability of producing a live calf that carries the PHA gene, and a 25% probability of producing a live calf that is a non-carrier of the PHA gene.

14. What can happen when a PHA carrier is bred to a PHA non-carrier?

Every time a PHA carrier is bred to a PHA non-carrier there is a 50% probability of producing a PHA carrier calf and a 50% probability of producing a PHA non-carrier calf. You **will not** produce a PHA-affected calf if you breed a PHA carrier to a PHA non-carrier.

15. What are the recommended "best practices" to breed the Dexters in my herd that are confirmed to be PHA carriers and conserve their *desirable* genetic traits?

- As with any trait, PHA can be managed in a sensible manner. Dexters, like all cattle, can be bred in a careful and deliberate program to eliminate any undesirable trait. The ADCA recognizes and respects the concept that there is no need to "throw the baby out with the bath water" with respect to any trait that has been identified in our breed.
- Breed PHA carriers only to PHA non-carriers.
- Test any calves that you produce from PHA carrier animals to determine their PHA status if you intend to retain or sell them as registered breeding stock. Make careful selections based on that knowledge.
- Inform prospective buyers of the PHA status on any registered breeding stock that you offer for sale.
- Retain the offspring that are PHA non-carriers that have the desired traits you wish to propagate within your herd. Gradually replace your PHA carriers as they become old and unproductive with their carefully selected PHA non-carrier progeny.