AUSTRALIAN BRAHMAN BREEDERS' ASSOCIATION LTD

POMPES DISEASE

~ COMMONLY ASKED QUESTIONS ~

WHAT IS POMPES DISEASE?

Pompes disease is a lethal inherited genetic disorder which has been diagnosed in Brahman and Shorthorn cattle as well as a number of other species of animals.

The disease is inherited as a recessive i.e. only calves that inherit a mutant gene from both parents will die of the disease. The disease occurs because these affected calves lack activity of the essential enzyme acidic a-glucosidase (AAG). As a result of this deficiency, glycogen builds up inside muscle and nerve cells, interfering with normal tissue function.

WHAT ARE THE SYMPTOMS?

The clinical signs of the disease vary greatly and reflect the tissue most affected by the accumulation of glycogen. In the majority of cases, muscle function is compromised and affected calves suffer from progressive muscular weakness.

The disease is progressive with the signs becoming most obvious when the animal is stressed such as during weaning, under poor nutrition or mustering. In the latter stages of the disease, calves/weaners experience increasing difficulty in rising and may be found lying on their side and paddling in an attempt to rise, even before they have lost a lot of body condition. Calves typically die between 6 and 12 months of age.

In many instances affected animals are found dead in hollows and creeks and appear to have died of "misadventure". Other affected calves may die of a sudden heart attack as glycogen accumulation affects the heart. In others the nervous system may be affected and some calves may be blind including very young calves.

CAN A NORMAL ANIMAL CATCH POMPES DISEASE LATER IN LIFE?

No. Each animal carries two copies of the acidic a-glucosidase genes. Genetically normal animals carry 2 normal copies of the gene. Carrier animals carry 1 normal copy and one defective (mutant) copy. Affected animals carry 2 defective (mutant) copies.

An animal's status as far as Pompes disease is concerned is established at conception and can not change.

CAN CARRIER ANIMALS BREED NORMAL PROGENY?

Yes. The mating of two carrier animals will, on average produce 25% normal progeny, 50% carrier progeny and 25% affected progeny.

The method of inheritance is illustrated in the diagram below.

If a carrier animal is joined to a normal animal you can expect, on average that 50% of the progeny will be normal and 50% will be carriers. **None** will be affected.

ARE THERE ANY DIFFERENCES BETWEEN A NORMAL ANIMAL WHO HAS A CARRIER PARENT AND A NORMAL ANIMAL WITH TWO NORMAL PARENTS?

Absolutely not. The Pompes disease status is determined at conception and normal means the animal has both copies of the normal gene. It is in no way "tainted" or likely to produce carrier progeny in the future.

WILL NORMAL ANIMALS ALWAYS PRODUCE NORMAL PROGENY?

Yes. Provided the normal status has been correctly diagnosed then normal animals mated to normal animals will always produce normal progeny. The only qualification to that is the remote possibility of another mutation occurring at some time on the future. However, a future mutation is just as likely to produce some other defect as it is Pompes disease.

IF AN EMBRYO CALF HAS A NORMAL SIRE AND DAM, CAN A CARRIER RECIPIENT AFFECT THE CALF?

No. Pompes disease status is established at conception and is not contagious in anyway either from a recipient dam or any other animal.

WILL A CARRIER EVER BECOME NORMAL?

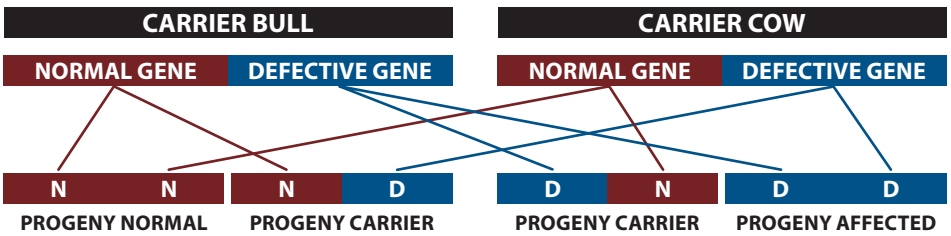
No. Again provided the animal has been correctly diagnosed, an animal's Pompes disease status will remain unchanged all its life.

DOES POMPES DISEASE CAUSE INFERTILITY?

No. Pompes disease symptoms become evident and affected animals die before they reach breeding age. There are no symptoms in carrier animals. The loss of calves, however will have an impact on the number of calves weaned compared to cows joined. It will have no affect on the number of calves born.

WHAT DOES E7 AND E13 MEAN?

Pompes disease is caused by a mutation in the gene controlling acidic a-glucosidase. It is possible for more than one mutation to produce the same disease symptoms. This is the case with Pompes disease in Brahman cattle. The mutation at the E7 site is common in the Brahman population and the second E13 is extremely rare. The E7 mutation is present in about 12% of the Brahman



AUSTRALIAN BRAHMAN BREEDERS' ASSOCIATION LTD

POMPES DISEASE

~ COMMONLY ASKED QUESTIONS ~

population. The E13 mutation in Australia is limited to the progeny of one imported animal. All future importations will be screened for both mutations.

HOW ACCURATE IS THE DNA TEST?

Thanks to the work of Dr Peter Healy and his wife Dr Julie Dennis from the Elizabeth Macarthur Agricultural Institute (EMAI) in NSW we have a DNA test for both the E7 and E13 mutations. These tests work on the precise site of the mutation and are therefore 100% accurate in determining the status of animals for the particular mutation in question.

Testing for the E13 mutation is only conducted within the bloodline of the carrier imported animal. All other Pompes disease testing is conducted using the E7 Pompes test. There is no evidence to suggest there are any other mutations causing Pompes disease which are, as yet, undetected. However if they do exist, new tests would be required to be developed.

Because of the possibility of mixing of foetal blood between twins, in the womb, it is possible, although rare, to have a misdiagnosis with twins where blood is used as the sample medium. It is for this reason, as well as convenience, that the preferred sample for testing is hair root. This avoids the problem with twins and is also more convenient to collect, label and transport. Semen is also just as efficient as hair roots.

HOW DO THE CURRENT DNA TESTS COMPARE WITH THE PREVIOUS DNA AND ENZYME TESTS?

The first test which was developed relied on measuring the level of activity of the acidic a-glucosidase enzyme relative to two other enzymes. This produced a guide to the animal's status with animals in the higher enzyme range designated normal and those in the lower range regarded as carriers. It however left a large number of animals in the equivocal range where the status was not possible to determine accurately.

The first generation DNA test developed by the CSIRO and operated by the QLD DPI was a substantial improvement. It was an indirect marker DNA test which means it was operating on a marker on the acidic a-glucosidase gene which was close to but not right on the E7 mutation.

It was 100% accurate in detecting animals carrying the E7 mutation.

Because it did not operate directly on the mutation there was a small proportion (2%) of normal animals which tested as carriers. (The new E7 test has eliminated this inaccuracy).

Comparison of the enzyme results with the DNA test results indicated that an animal with an enzyme level of 100% or more is normal.

WHAT DOES "ABBA APPROVED FOR ARTIFICIAL BREEDING" SIGNIFY?

The Associations policy is to encourage members to reduce the frequency of the Pompes disease gene in the population. The gene can be spread quickly through artificial breeding, therefore animals which are used for artificial breeding are required to be of normal Pompes disease status before their progeny may be registered without testing.

Animals produced by artificial breeding from parents of unknown or carrier Pompes disease status must be tested for Pompes disease before they can be registered. ABBA approved for Artificial breeding therefore means the animal is Pompes normal.

There are currently over 150,000 animals in the Association's records with normal Pompes disease status.

CAN CARRIER ANIMALS BE REGISTERED?

Yes they can, however their carrier status will be noted and all of their progeny must be tested before they can be registered. This allows members with a carrier animal they consider important to their breeding program to develop a bloodline of normal progeny.

HOW DO WE GO ABOUT TESTING?

The Association has arranged with Genetic Disease Laboratory at EMAI a batch testing service to reduce the cost of testing for members.

Tests will be accumulated and sent in batches of 10. The cost is $36.50 per test for this service. Results usually take up to 6 weeks.

An express service is available with samples going straight to the laboratory for a higher fee. Members wishing to undertake testing should contact the Association for specimen advice sheets and sample envelopes.