



## CITRULLINAEMIA

### Notice of Recessive Gene in the Pedigree:

If the wording "Suspect Citrullinaemia" is written on the top right hand corner of the Registration Certificate. This means your animal within its first 3 generations has an ancestor that has been tested positive for the genetic recessive Citrullinaemia.

As a promoter of Holstein cattle, our aim is to inform you as much as possible when your animal may be affected and what you could do in this situation. Your animal may or may not be a carrier, as per the information outlined below:

### What is a genetic recessive?

Carrier (heterozygous) animals may not show any symptoms of this disorder. This is because the gene associated with this disorder is a recessive gene. An animal requires two copies of the gene for it to become an affected (homozygous) animal. For an animal to be affected, it requires one copy of the gene from its sire and one also from its dam. It is the affected animal that displays the symptoms of the genetic disorder (in this case it is lethal). A carrier animal has only one copy of the affected gene, the second copy being the dominant normal gene.

Although a carrier may not display the symptoms, it still has the possibility of passing on the gene to its progeny.

The following table may help to explain this:

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Normal Gene = N	Faulty Gene = F		
	Progeny: (Possible gene combinations)		Progeny: (Possible gene combinations)
Sire (Carrier) = NF	NN	Sire (Carrier) = NF	NN
Dam (Normal) = NN	NN	Dam (Carrier) = NF	NF (Carrier)
	NF (Carrier)		NF (Carrier)
	NF (Carrier)		FF (Affected-Lethal)

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### Cause and Symptoms:

Citrullinaemia is caused by a deficiency of argininosuccinate synthetase (ASS), one of the enzymes of the urea cycle. It is believed that ammonia builds up in the brain of the affected calves because, due to the ASS deficiency, it is unable to be converted to urea for elimination.

Calves affected with citrullinaemia appear normal at birth, but within 4-6 days it has died. Signs begin with depression and poor feeding, followed by aimless wandering or standing with the head pressed against a wall or fence. Eventual collapse follows within 12 hours after the symptom of blindness, bellowing and paddling of limbs. (From extracts from Elizabeth Macarthur Institute, Camden, NSW.)

**The Test:**

A laboratory test for Citrullinaemia is available at the option and expense of the owner of the animal. If tested positive for Citrullinaemia the animal is a “carrier”. If tested negative, the animal does not carry the recessive gene and is normal.

The test is available via the Queensland University Laboratory. Hair samples can also be sent to Holstein Australia Office who will forward the sample for testing. Please contact Holstein Australia Office for more details (telephone: 03 9835 7600).

**Code on Pedigree and how to read it:**

Once tested, a Citrullinaemia carrier is designated internationally by the letters “CN”, while the animal which has been tested and found to be free of the condition is labelled as “TC”. This TC label is very important, as it designates an animal that has been certified free of the condition, and assists in the control of the disease.

Please Note: Genetic recessive carrier status of a sire or dam may not be known until after its progeny has been registered. In this case, the “Suspect” wording would not be listed on the Registration Certificate of the progeny. It is worthwhile to periodically examine an animal’s pedigree for genetic recessive test results.

**Methods of control through breeding:**

Dairy farmers can avoid making Citrullinaemia-risk matings through breeding decisions. When mating suspect Citrullinaemia or “carrier” cows, use bulls that have been tested free of this genetic recessive, or ensure that the bulls used do not have Citrullinaemia carriers in their pedigree. This will reduce the number of carrier progeny to a minimum.

Citrullinaemia is one of the earlier genetic recessive defects identified in Australia, having been traced to its origins in the AI bull, Linmack Kriss King-CN, GBRM303731. Through stringent AI screening, there has been a reduction in the number of carrier animals within the Holstein population. Holstein Australia and dairy farmers should not become complacent on this defect and Citrullinaemia remains part of the gene alert programme.

Control of Citrullinaemia and other significant genetic disorders in the dairy cattle population depends entirely on self-regulation and cooperation between all those involved in the dairy breeding industry in Australia. Holstein Australia advocates the screening of all AI test sires as well as embryo donors, and the open disclosure and clear publication of BLAD status at sales and in all information, education and marketing material disseminated to dairy farmers. This stimulates awareness and allows farmers to make clearer breeding decisions.