

# MYOSTATIN E226X

## FACT SHEET

Myostatin gene mutations, responsible for the condition known as Double Muscling, take more than one form. In fact there are several known mutations across many beef breeds. Amongst these is the E226X mutation, which is found in Shorthorn cattle. Whilst two copies of the gene may be considered unfavourable, one copy of the gene may also be considered as favourable for its effect in increasing retail beef yield, without having a negative effect on other production traits. For breeders there is a simple DNA test, which allows them to manage the level of the condition within their herd.

This Fact Sheet is designed to answer many of the common questions asked by Shorthorn Beef members.

### WHAT IS MYOSTATIN?

Myostatin (GDF-8) is essential for proper regulation of skeletal muscle mass. Mutations make the myostatin gene inactive, resulting in variation of normal phenotypes, including muscle hyperplasia and muscle hypertrophy (Double Muscling). Myostatin mutated genes are known to be present in many breeds of beef cattle.

### WHAT IS MYOSTATIN E226X?

E226X is an autosomal recessive genetic defect and also a disruptor gene. Similar disruptor genes in homozygous affected animals are known to produce variations from normal phenotypes which may include increased birthweights and dystocia, reduced fat and marbling and hyperplasia (increase in the number of muscle fibres) and hypertrophy (increase in the size of muscle fibres). Also improved beef yield and improved tenderness.

### AREN'T ALL GENETIC DEFECTS BAD?

Genetic defects range from favourable to unfavourable to lethal. It is worth noting that the desirable Poll gene by example, is also a mutated gene, so not all genetic defects are bad. E226X is not a lethal defect and should not be considered as such.

### WHAT IS AN E226X CARRIER?

There are three distinct levels of status that each animal may have. An **E226X Free animal** carries no forms of the mutated gene (Homozygous Free). An **E226X Carrier animal** carries one form of the mutated gene (Heterozygous) and an **E226X Affected animal** carries two forms of the mutated gene (Homozygous Carrier).

### WHY ARE AFFECTED ANIMALS IMPORTANT?

Affected animals are important because regardless of the status of the animal you join an affected animal to, the progeny will always be carriers at least.

### WHAT ARE THE EFFECTS OF E226X?

There are several known Myostatin disruptor genes, of which E226X is one. However, the exact phenotypic responses for E226X are not well documented at this stage. Whilst two forms of a disruptor Myostatin gene mutation are generally considered unfavourable, one form of the gene is often considered to have a positive effect. It is important to understand that the phenotypes normally affected by Myostatin mutations are largely measured within genetic evaluation and therefore any negative effects will be either visual or reported within genetic evaluation traits for each animal, provided that the traits have been correctly collected and reported.

### WHAT HAPPENS IF I JOIN TWO CARRIER ANIMALS?

Inheritance will follow the simple inheritance pattern. When two carrier animals are mated together, the progeny will have a 25% chance of being Homozygous Free, a 50% chance of being Heterozygous and a 25% chance of being a Homozygous Carrier or affected.

### IF I USE A FREE SIRE WILL I GET AFFECTED PROGENY?

If you use a sire with a free status, you will never get affected progeny. Again, following the simple inheritance pattern, if the dam is a carrier, the progeny will have a 50% chance of being Homozygous Free and a 50% chance of being a Heterozygous Carrier. If the dam is an affected animal, the progeny will have a 100% chance of being a Heterozygous Carrier, but will not be Affected.

### IS THERE A TEST FOR E226X?

Yes. A DNA test is readily available to determine the E226X status of an animal. Breeders have the opportunity to test through two methods. First is a standalone DNA test, which costs \$33 inc. GST. The second is to request the E226X testing in the HD50K bundle, in which case it will cost an extra \$11 inc. GST. Simply send a tissue sample to the Shorthorn Beef office, requesting your preferred testing method.

### HOW DOES E226X AFFECT MY HERD?

Remember, there is little research to adequately describe all the variations in phenotype for the E226X mutation. Whilst the double muscle gene tends to conjure certain images, it is known that in breeds that have selected for double muscling, there is both a proliferation of affected animals and also the effects of genetic selection for enhanced muscling traits.

In breeds such as Shorthorn, where a more "balanced" genetic selection has occurred, the effects of E226X may prove to be less pronounced. Certainly, not all well muscled Shorthorns have any copies of the E226X gene.

However, as most effects will be visual or measured for genetic evaluation, where the variations in phenotypes may be considered undesirable, it is unlikely that these animals would have entered the population anyway. Where the variations in phenotype may be considered favourable, then these animals should still be considered favourably.

Importantly, for all breeders, there is a very simple DNA test, which will allow breeders to manage the level of E226X within their own population, depending on their particular requirements. The E226X status of an animal will not currently affect its registration status in any way.



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