GENETIC DEFECTS AND CONDITIONS

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In the Shorthorn breed, like almost all genetic populations, human and animal, genetic conditions or defects show up from time to time. The terms condition and defect tend to be interchangeable although, generally defect is attached to lethal outcomes.

There are 4 conditions/defects that can be prevalent in Shorthorn populations. Most lethal outcomes can be avoided and exposure can be managed by ensuring that the sire and dam both do not carry the gene and if one does, it is important to be aware of the negative or positive status of the animal it will be mated with.

Australian Shorthorns are currently working together with Weatherbys Scientific to provide genetic testing opportunities for members.

TH (TIBIAL HEMIMELIA)

TH is a lethal genetic defect when an animal inherits two copies of the recessive gene (sire and dam). Calves are born with severe deformities including twisted back legs with fused joints, large abdominal hernias and/or skull deformities. If calves survive the birthing process, they cannot stand and do not survive long.

PHA (PULMONARY HYPOPLASIA WITH ANASARCA)

PHA is a lethal genetic mutation when an animal inherits two copies of the recessive gene (sire and dam). Calves are born dead with under-developed lungs (pulmonary hypoplasia) and extreme swelling caused by excessive fluid retention (Anasarca). This is a potentially a doubly lethal defect as the cow often cannot survive trying to birth the calf.

DS (DIGITAL SUBLUXATION)

DS is not a lethal condition from a genetic standpoint. Animals that carry two copies of the undesirable gene (sire & dam) are known as Homozygotes and are identified in a number of registries worldwide by the symbol 'DSH'.

Most cattle that are DSH show some outward signs of the genetic condition, usually malformation of the rear pastern (ankle area). It is important to know that ALL progeny of DSH animals will be at least carriers of the condition and Carriers will be recognized with the symbol DSC in each registry. Although the original mutation happened in completely separate populations, the DS condition sits on the same chromosome as the PHA condition and as a result, it appears that a PHA Carrier animal mated with a DS Carrier can result in the same physical deformities as mating two DS Carriers.

MYOSTATION (ALSO CALLED DOUBLE MUSCLE)

An animal classified as a Myostatin Carrier does not actually have two muscles in place of one normal muscle. However, due to a genetic abnormality, the affected animal has a greater number of muscle fibres which can result in a much larger than normal muscle mass appearance.

This condition is common in many mammals and is due to the production of “myostatin”, which is a protein found in the gene that regulates muscle growth. In normal animals, myostatin tells the muscles to stop growing when they reach a certain size however, the defective gene lacks the myostatin protein and therefore allows the muscle to keep growing.

Nine Myostatin mutations (also called variants) of the gene have been identified and some are more common than others in certain beef breeds. Six of the nine are classified as “disruptive” as they may cause conditions along with extra muscling that are not advantageous such as increased birth weights and calving difficulties. The other three variants are referred to as “missense” or nondisruptive. The nine identified variants are:

Disruptive Variants

E226X, nt419, nt821, C313Y, E291X, 0204X

Non-Disruptive Variants

D128N, F94L, nt821, S105C

The Myostatin condition is not classified as a simple recessive gene defect, like TH for example, where the carrier is an animal having no physical affect, but ‘carries’ the defect (essentially hidden) and can pass it on to offspring. The Myostatin condition affects all that are not “0” for variants. However, that physical affect could be virtually undetectable all the way to obviously affected or to the point of looking deformed. Typically, it is expected that those with only one variant would show much less increased muscularity compared to those with two variants.

Test results will typically be presented as in the following examples:

0

No variants present

1, E226X

1 copy of the variant present

2, E226X

2 copies of the variant present

1, E226X; 1, nt419

1 copy of each of 2 different variants present
Predictable breeding results from mating sires and dams who do and do not have a myostatin variant present in their DNA can be grouped as follows:

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0 x 1, V

50% have 0 copies; 50% have 1 copy

1,V x 1,V

25% have 0 copies, 50% have 1 copy; 25% have 2 copies

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0 x 2,V

100% have 1 copy

1,V x 2,V

50% have 1 copy; 50% have 2 copies

2,V x 2,V

100% have 2 copies

As with most genetic conditions there are often additional positive and negative associated traits. Positive Myostatin condition characteristics can include: leaner meat (less intramuscular fat), more tenderness, larger rib eye measurement and higher carcass yield. Negative characteristics may include: heavier birth weights, calving difficulties, thinner bones and less fat cover.

For reasons associated with the positive and negative characteristics noted above, breeders may specifically choose to include Myostatin positive animals in their herd and manage them with careful breeding programs. Or breeders may choose to bypass the necessary management and discourage the inclusion of Myostatin positive animals in their herd.

Both programs are valid and acceptable and the availability of genetic testing for the presence of the condition ensures management either way is accessible to all members.