Understanding Genetic Conditions

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The genetic conditions Herefords Australia can test for are Dilutor (DL), Hypotrichosis (HY), Idiopathic Epilepsy (IE), Maple Syrup Urine Disease (MSUD), Mandibulofacial Dysostosis (MD) and Delayed Blindness (DB). This fact sheet will explain the conditions, and how they are inherited.

When viewing an animal's details online or on a certificate, you may notice these abbreviated letters are followed by another letter that represents the test result or GENEPROB result. They will look this like this:

IEF DLF HYF MSUD49%

_ _ F = (FREE). This result indicates the animal has been tested and found to be free of the causative mutation responsible for the genetic condition. This animal is homozygous free, meaning that it has two copies of the normal variant (or allele) of the gene.

_ _ C = (CARRIER). This result indicates that the sample submitted for this animal has been tested and found to be a carrier of the causative mutation responsible for the indicated genetic condition. This animal is heterozygous for the mutation, meaning that it has one mutant allele and one normal allele. This animal could pass the mutation to approximately half of its progeny.

_ _ A = (AFFECTED). This result indicates that the sample submitted for this animal has been tested and found to possess two copies of the mutant variant of the gene. This animal is homozygous for the mutation responsible for the genetic condition and in the case of HY & IE, will be affected by the genetic condition. In the case of DL, the animal will appear normal but will pass the mutation to 100% of its progeny.

_ _ FU = The animal has not been tested for the causative mutation, but both the sire and dam have either been tested and found to be free or have a status of _ _FU and are expected to be free. (Herefords Australia gives no guarantee as to the animal's genetic condition status.)

_ _ % = (GENEPROB result). This indicates that, based on pedigree information supplied by the breeder of the animal, the animal has a chance to be a carrier but has not been tested. The higher the indicated percentage, the larger the chance the animal may be a carrier. To verify the status of this animal, it is recommended that testing be undertaken prior to using this animal for breeding purposes. For further information on GENEPROB, click HERE.

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In genetics, different variants of the same gene are referred to as alleles. Each individual has two alleles for every gene, one inherited from its father and one inherited from its mother. These two alleles can be the same (homozygous), or they can be different (heterozygous). The DNA test detects three possible outcomes. An individual can be:

1. Homozygous for the normal DNA sequence (both chromosomes have the normal allele)
2. Heterozygous for one normal allele and one mutant allele, or
3. Homozygous for the mutant allele (both chromosomes have the same mutant allele)

There are a number of different modes of inheritance for genetic conditions with the most common being autosomal recessive, where animals need to carry two copies of the mutant allele in order to express the condition. The diagram below shows the inheritance patterns for common recessive conditions.

	Free Bull (AA)	
Free Cow (AA)	Free (AA)	Free (AA)
	Free (AA)	Free (AA)

	Carrier Bull (Aa)	
Free Cow (AA)	Free (AA)	Carrier (Aa)
	Carrier (Aa)	Free (AA)

	Carrier Bull (Aa)	
Carrier Cow (Aa)	Free (AA)	Carrier (Aa)
	Carrier (Aa)	Affected (aa)

Idiopathic Epilepsy (IE)

Age of onset of first seizure can vary, ranging from birth to several months of age. Occurrence and persistence of seizures may be influenced by environmental factors, such as temperature extremes or increased physical activity. During a seizure episode, individuals will typically lie on their side with all limbs extended in a rigid state. Seizure episodes may last from several minutes to more than an hour.

IE is predominantly seen in horned Herefords but can be seen in polled Herefords with horned animals in their pedigree.

IE is caused by a recessive allele, which means that affected calves will only occur when two carrier animals are mated together. IE can lead to death and affected animals have not been known to reproduce.

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Hypotrichosis (HY)

Hypotrichosis is partial to almost complete lack of hair. Affected calves are often born with very short, fine, kinky hair that may fall out leaving bare areas. Calves may be born with abnormal hair that falls out, or are born hairless and develop a short curly coat with age. The abnormal hair can appear over all or parts of the body, often on the poll, brisket, neck and legs.

An animal affected by HY is more vulnerable to skin infections, pests, sunburn, cold stress and have a decreased economic value.

HY has been reported in the Hereford breed for many decades and is non-lethal.

HY is caused by a recessive allele, which means that affected calves will only occur when two carrier animals are mated together.

Maple Syrup Urine Disease (MSUD)

Calves affected by MSUD have a defect in an enzyme that breaks down complex amino acids in the diet. The resulting build-up of these amino acids in the body causes lethal brain damage.

Affected calves display severe neurological signs within the first week of life. Typically, affected calves are dull and will become recumbent by 2 to 4 days of age. Affected calves will enter a state of terminal opisthotonus, where severe muscle spasms will cause their head, neck and spinal column to present in an “arching” position. Maple Syrup Urine Disease is fatal, with affected animals tending to die as calves.

This disease is also found in humans and is named for the smell of urine observed in human babies — the smell is not always noted in calves.

Maple Syrup Urine Disease is caused by a recessive allele, which means that affected calves will only occur when two carrier animals are mated together.

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Dilutor (DL)

Dilutor is a coat colour abnormality and will not cause any issues with calf performance. DL can be found when a Hereford bull that is a carrier of the DL gene is mated to a black cow. The gene causing DL is the dominant gene; therefore, baldy calves that are expected to have a black coat are born with grey or smoky coats because the DL gene is dominant over the black gene.

In addition to the change in colour, the dark areas can show signs of hair loss, while the areas with white hair appear normal.

Affected calves are susceptible to cold, stress and poor growth rate, especially in the first year.

The condition will never be expressed in purebred Hereford animals, meaning that carrier animals may appear normal but possess either one or two copies of the undesirable gene.

Mandibulofacial Dysostosis (MD)

In June 2020, Herefords Australia were informed of Mandibulofacial Dysostosis. MD was discovered by the American Hereford Association in collaboration with the University of Nebraska-Lincoln.

MD may result in a facial malformation, which can include cleft palate, brachygraphic (short jaw) and camplygnathia (crooked jaw or face). Affected calves could display bilateral skin tags, attached to an unusual bone formation, just behind the corner of the mouth. Additional skin tags may be present near and/or below the ears. The calves' ears are sometimes small and floppy.

Jaw muscles are underdeveloped, and calves may have an elongated oral opening giving them the appearance of an exaggerated smile. Affected calves have a sucking reflex but will not suckle vigorously. Calves that display additional cleft palate and/or shortened or crooked jaws will be further debilitated. While affected calves are typically born alive, they are not able to thrive.

MD is caused by a recessive allele, which means that affected calves will only occur when two carrier animals are mated together.

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Delayed Blindness (DB)

Delayed Blindness (DB), sometimes known as CLN, is caused by retinal degeneration within the eye. Cattle affected by DB are not born blind, but vision loss is noticeable near or just after one year of age. Initial clinical signs may include the animal having difficulty navigating their surroundings, bumping into stationary objects, and slowly navigating unfamiliar terrain. These cattle will also lack a menace responsive (reaction to something being moved toward the eye). Cattle are remarkable at adapting; if in a stable environment vison loss may be severe when first noticed. Unless secondarily injured, the eye appears normal to the casual observer. A detailed ophthalmologic evaluation of the retina in the back of the eye can confirm retinal degeneration. Diagnosis can also be established by genetic testing.

DB is caused by a recessive allele, which means that affected calves will only occur when two carrier animals are mated together.

Further information on managing genetic conditions

There is no 'one size fits all' strategy to managing genetic conditions. Before embarking on a management strategy producers should consider:

- The economic impact of the condition.
- The frequency of the condition within the herd.
- The availability and cost of DNA tests.
- Research the genetic condition status of any animals being brought into the herd.
- Their legal obligations about disclosing the carrier status of sale animals.
- The relevant breed society regulations.

Click here for the ABRI Extension Services Fact Sheet on Managing Genetic Conditions for further information.

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