Genetic Conditions
Technote 11

HIGHLIGHTS

- DataGene reports test results for genetic defects and characteristics such as A1/A2, polled and coat colour.
- DataGene follows an internationally agreed 3/4-letter code system.

Table 1 Traits reported and their corresponding codes

Trait	Description	Free	Heterozygous carrier	Homozygous carrier	Observed carrier	Breed
Genetic defects						
BLAD	Bovine Leukocyte Adhesion Deficiency. A lethal genetic metabolic disease of cattle affecting the animal's immune system.	BLF	BLC		BLA	Holstein
Brachyspina	A genetic defect that causes physical deformities as well as embryonic death. Affected calves have reduced bodyweight and a shortening of the spine with long and thin limbs.	BYF	BYC		BYA	Holstein
Citrullinaemia	A lethal genetic enzyme deficiency interrupting the normal processing of ammonia in the brain. Affected calves appear normal at birth but usually die within 4-6 days.	CNF	CNC		CNA	Holstein
Cholesterol deficiency	A genetic disorder that causes low cholesterol levels and death in calves.	CDF	CDC	CDS	CDA	Holstein
CVM	Complex Vertebral Malformation. A lethal condition that causes early abortion due to foetal death or, if the calf survives to term, it usually dies within a week of birth.	CVF	CVC		CVA	Holstein
DUMPS	Deficiency of Uridine Monophosphate Synthase. A genetic metabolic disease which is lethal at the embryonic stage (at about day 40 of pregnancy).	DPF	DPC		DPA	Holstein
Factor XI	Factor-XI deficiency can cause some cows to have a tendency to bleed. Haemorrhaging or excessive bleeding may be fatal.	XIF	XIC		XIA	Holstein
Early onset muscle weakness	Affected calves are unable to stand or walk. Some calves may lose the ability to suckle. Read more	MWF	MWC	MWS		Holstein
Mulesfoot	Also known as syndactylism - the fusion of the two toes of the foot (resembling a thin mule's foot).	MFF	MFC			Holstein
Jersey neuropathy with splayed forelimb	A recessive genetic condition. Affected calves are unable to stand.	JNSF	JNSC			Jersey
CHRNB1	Arthrogryposis Multiplex Congenita - characterised by multiple joint contractures	AHF	AHC			ADIU Red Group
BHD SDM	Spinal Dismyelination - a condition where the myelin sheath in the spinal cord is malformed or defective	BHDT	BHDC			Brown Swiss
WEAVER	Progressive Degenerative Myeloencephalopathy characterized by hindlimb weakness and ataxia (lack of coordination).	BHWT	BHWC			Brown Swiss
Genetic characteristics						
A1A2 milk	A1/A2 milk refers to the presence or absence of the A2 proteins in the milk.	A11	A12	A22		ALL
Black/red colour carrier (MC1R)	Calves may be born with a red coat but turn almost totally black after a few months of age. This gene is dominant over the true red gene.	BKF	BKC	BKS		Holstein
Dominant red/ variant Red		VRF	VRC	VRS		Holstein
Recessive red carrier	True red coat colour gene.	RDF	RDC	RDS		Holstein

Explaining genetic conditions

To provide breeders with the tools they need to make informed breeding decisions it is important to identify heritable, sometimes undesirable, genes that animals can carry. Genetic defects refer to genetic disorders such as BLAD and CVM whilst genetic characteristics refer to potentially desirable traits including coat colour, polled and A1A2 milk. The following outlines the common theory underlying inheritance of these traits.

Simple inheritance traits

Simple inheritance traits are traits largely controlled by one major gene. These are 'yes' or 'no' characteristics that are either present or absent in an animal. In some cases, simple inheritance traits may involve more than one gene. The pattern of inheritance remains fairly simple. Testing for these genes allows breeders to effectively select for or against the trait/disease. Most of the traits outlined in Table 1 fit into this category.

Multi-gene traits

Multiple gene traits are controlled by a larger number of genes. This group includes many economically important traits such as kilograms of protein and daughter fertility. In these cases, Australian Breeding Values (ABVs) are used to identify superior/inferior animals as it much more difficult to clearly identify animals with the favourable variants of each gene. Multi-gene traits are not discussed in this Tech Note.

Dominant vs recessive?

The different variants of a gene which regulate the expression of a trait are known as alleles. For each gene all cattle inherit one allele from their mother and one from their father.

Where the allele controlling a trait is dominant the animal only requires one copy for that trait to be expressed. Conversely a recessive trait requires both alleles (one from each parent) to be expressed.

A good example of this is coat colour. In the following example both animals are heterozygous black (Bb) i.e. they are black cattle who carry both the black and red alleles. The uppercase 'B' refers to the dominant black allele and the lower case 'b' refers to the recessive red allele.

Figure 1: Example mating between heterozygous coat colour

		Sire Heterozygous black	
		B	**b**
Dam Heterozygous black	**B**	**BB**	**Bb**
	b	**Bb**	**bb**

When these two animals are mated there are three possible outcomes:

- A 25% chance the offspring will be a homozygous black calf (BB)
- A 50% chance of a heterozygous black calf (Bb) and
- A 25% chance of a red calf (bb).

Because black is dominant over red the heterozygous offspring (Bb) will be black. The calf will only have a red coat when it inherits the red coat allele from both parents.

Using codes for breeding decisions

If your herd contains a strong pedigree influence from animals known to carry a genetic defect it is particularly important to keep an eye out for the corresponding code (refer to Table 1). To avoid undesirable matings, use an inbreeding report to avoid joining closely related animals. For cows identified as carriers of a genetic defect, avoid using a bull with a three letter code ending in C which indicates it is a carrier. Where a genetic code ends in 'F' the animal has been tested as free from carrying that trait.

To include desirable genetic characteristics in your breeding objective, look for the relevant 'tested positive' code (e.g. RDC for red carrier).

DataGene records calls and publishes codes for these traits across its range of web-based publications such as DataVat and the Good Bulls Guide App and spreadsheets. The absence of a code means that the animal either does not have a pedigree containing carrier animals, has not been tested for the given trait, or that this information has not been supplied to DataGene.

Haplotypes

A haplotype is a stretch of DNA (containing one or more genes) that is inherited intact or as a block from a single parent. Because they are of genetic origin, haplotypes are breed-specific.

Haplotypes are used to identify a number of genetic conditions where the actual gene causing the condition is yet to be identified.

In the Australian dairy industry, the term haplotype generally refers to genetic conditions that affect fertility. These conditions result in embryonic death (e.g. HH1, HH2, JH1) at varying times through gestation leading to sub optimal reproductive performance. For more information refer to DataGene's tech note: Harmful haplotypes in dairy cattle.

More information

DataGene
Ph 1800 841 848
Email: enquiries@datagene.com.au

www.datagene.com.au

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