

# FACT SHEET

## Utilising DNA to manage genetic conditions



### FAST FACTS

- **It is important for breed societies and producers to manage genetic disorders**
- **Genetic conditions are usually rare but where they occur in high frequency, DNA tests offer an important tool for managing them**
- **DNA testing allows seedstock breeders to cull carriers of genetic conditions as well as maximise the bloodline's superior genetics for other traits**
- **DNA testing allows producers to avoid production losses by using only animals that are tested 'free' of the condition**

**Genetic disorders or conditions are usually caused by mutations of a specific gene. They occur in all species and are more prevalent where there are high levels of inbreeding (for a comprehensive listing see <http://omia.angis.org.au/>). Many genetic conditions have a simple recessive inheritance and can be managed with DNA testing if available. Historically, genetic conditions were managed by expensive progeny testing or eradicating all related animals, potentially leading to the loss of superior genetics for production traits. DNA testing for genetic disorders allows producers to carefully manage genetic disorders and minimise production losses.**

#### Recognising genetic conditions

Genetic mutations can have different expressions ranging from poor animal performance, structural unsoundness through to semi-lethal and lethal conditions. In the past three years, two lethal mutations and one semi-lethal mutation have been recognised in several breeds of beef cattle. Numerous other examples have been identified in other cattle breeds over many years. It is important to note that each condition requires its own specific management plan based on symptoms expressed by affected animals.

The incidence of genetic conditions is normally low but the frequency can rise through artificial breeding, which widely disseminates the genetics of a few selected animals. If these animals are carriers of mutations, the condition can spread rapidly before being recognised. Identifying an abnormality as a genetic condition can be difficult, as other factors, such as nutritional deficiencies, toxins, extreme weather conditions and infections can cause abnormalities in calves that mimic genetic disorders. For these reasons, reporting of all abnormalities is important and strongly encouraged.

When large numbers of calves are dying or being born with a similar abnormality, both environmental and genetic causes are suspected. Investigation requires cooperation between many people, and early identification is facilitated by:

- surveillance and reporting of abnormal calves to breed societies or beef technical officers
- submission of photographs for identification of the condition, and samples for DNA testing
- submission of DNA samples and pedigrees from the affected animal's sire and dam

#### DNA test development

Existing genomics technologies allow scientists to compare normal and affected animals across the whole genome and to pinpoint mutations that cause these genetic disorders. Once the mutation has been located, the inheritance patterns need to be determined before the test can be released for diagnostic use.

A genetic condition with simple recessive inheritance will have three possible genotypes as shown in the example (see Figure 1 over page):

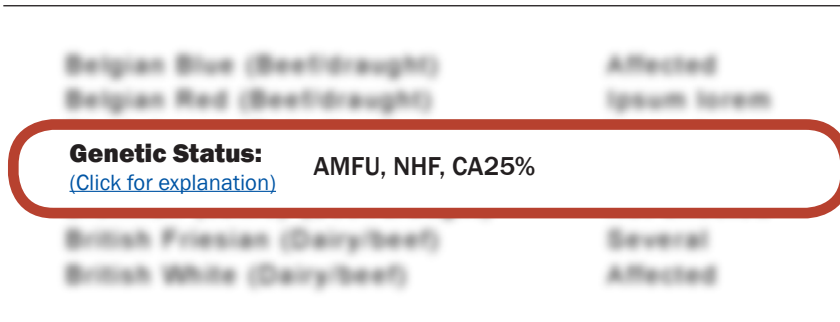
- affected, or abnormal animals that carry two copies of the mutation
- carrier, which looks normal but can transmit the condition to offspring. These animals carry one normal gene and one copy of the mutation
- free, which carry two normal genes and no copies of the mutation

In addition to DNA testing, the software tool, GeneProb, can calculate an untested animal's risk of carrying the mutation based on pedigree information and test results of animals in that pedigree. This can help to focus the testing effort toward those pedigrees with higher risk and provide an interim risk assessment. GeneProb reports two possible results: free untested and %chance of being a carrier (Figure 1).

| Code | Description  |
|------|--|
| AMF  | Tested Free  |
| AMFU | Not tested, based on pedigree expected to be free  |
| AM_% | Not tested, based on pedigree the animal has the indicated percentage chance to be a carrier |
| AMC  | Tested Carrier   |
| AMA  | Tested Affected  |

**Figure 1:** The possible genotypes recorded for the genetic condition AM. DNA testing reports AMA, AMF and AMC while the 'GeneProb' predictions report AMFU and AM\_%.

For genetic conditions, DNA test results and GeneProb predictions are made publicly available on several breed society websites and in sale catalogues (Figure 2) so producers are fully informed and able to avoid spreading deleterious genes.

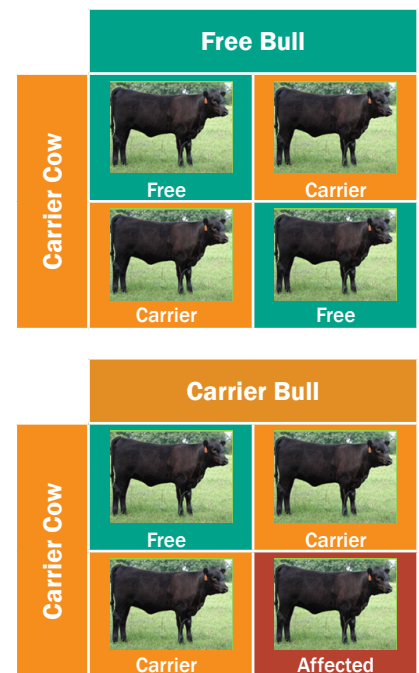


**Figure 2:** For some breed societies an animal's genetic status for known genetic conditions is listed in sale catalogues. Based on GeneProb prediction this animal is listed as having a 25% chance of carrying the recessive genetic disorder.

### Using DNA testing to manage genetic conditions

When a genetic condition causes economic losses it needs to be managed. Simple recessive genetic conditions are easy to manage if a DNA test is available to distinguish between carrier and free animals. When managed, producers are able to continue to realise the production value of the affected blood line as well as avoid production losses and dissemination of the mutation.

Management involves the selection of sires and dams to avoid mating carriers to other carriers. Mating a carrier and a free animal will avoid production losses by producing 100% normal calves, although 50% will carry the gene. By comparison, mating two carriers will produce 75% normal calves and 25% affected calves. Of the normal calves, one third will be free and two thirds will be carriers (Figure 3). Commercial herds that don't keep pedigree information would therefore benefit from using only bulls that are tested free.



**Figure 3:** The genotypes of progeny from matings of carrier cows with free and carrier bulls.

### Further reading

The Beef CRC website (<http://www.beefcrc.com.au/>) and the SBTS/TBTS webinar series (<http://sbts.une.edu.au/Webinars/webinars.html>) have additional information on managing genetic conditions with DNA. In addition your breed society and preferred commercial supplier can advise on testing options.

