

Polled gene test for Australian cattle breeds

Technical Report, April 2011



TECHNICAL NOTE

Marker Test for Polled. Beef CRC (project funded by MLA)

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Background

The inheritance of horned or polled in cattle has long been known to be under the control of only a few genomic regions, and the most important of these, the **polled locus**, has been mapped to chromosome 1 by a number of groups. It is not known how many genetic variants, or **alleles**, exist at the polled locus, but it is convenient to group them into those that are associated with horns and those that are associated with polled. Animals inherit one allele from their sire and one from their dam, and so an animal may carry two horned alleles at the polled locus (in which case the animal's **genotype** is **homozygous horned**), one horned allele and one polled allele at the polled locus (the animal's genotype is **heterozygous**), or two polled alleles at the polled locus (the animal's genotype is **homozygous polled**, often referred to as 'true polled'). In *Bos taurus* cattle breeds, polled is usually dominant to horned, so heterozygous animals generally have no horns. Therefore, without additional information it is not possible to determine whether a polled bull is heterozygous or homozygous polled. Additional information that may resolve this ambiguity includes trait or **phenotype** records on relations (such as on progeny), or a marker test. Marker tests are available for use in British and European cattle breeds but, until the CRC research project, no marker test was available for *B. indicus* breeds and composites.

The CRC Marker

The marker discovered by the CRC, like other tests on the market, is a **linked marker**. That is, we can't directly measure whatever it is that causes horns, so we measure something that is located close by on the genome. Mostly, but not always, the close by measurement is a good predictor of the unknown underlying genotype at the polled locus. Most alleles at the marker are almost always associated with the same allele (either polled or horned) at the polled locus. But some alleles at the marker show associations with both polled and horned alleles at the polled locus. For these **ambiguous marker alleles** the test cannot return an unambiguous result. The frequency of ambiguous marker alleles varies between breeds: in breeds where ambiguous marker alleles are rare the test works very well. In breeds where ambiguous marker alleles are common the test is unable to clearly predict genotype at the polled locus in a significant percentage of animals.

Data

The association between the CRC marker and polled was discovered in Brahman, and subsequently tested in a range of breeds as documented in earlier CRC publications. Animals for the experimental population were chosen to give good representation of polled, scurred and horned. Therefore, they are not a random sample. In this report we also include new data from two sources. First, bulls being genotyped with the 50K Bovine SNP chip as part of the National Beef Genomics Database project were also tested for the CRC polled marker. These bulls were selected to be representative of widely used genetics in the breed with high accuracy EBVs, taking no account of polled phenotype, so for the purposes of this study the bulls were essentially a random sample. Second, samples submitted by breeders for testing. These are most definitely not a random sample from the breed, but are animals for which the breeder saw benefit in obtaining marker test results. Consequently most of the samples are from polled animals, where the breeder is unable to distinguish between homozygous polled and heterozygous.

There are advantages and disadvantages to the three sources of data. Animals in the experimental population have a good representation of accurately recorded phenotypes, but are not a random sample and may come from a limited range of studs. The industry sires, being essentially random with respect to polled phenotype, give a good indication of the diversity of marker alleles in the breed, but for some breeds there are very few polled animals represented. The breeder submitted samples are representative of what would be submitted for a commercial test, with mainly polled animals. However, they may not be representative of the wider breed, and give no information about marker alleles associated with horns. In the analyses that follow the data from the three sources are merged, but summary information on the three populations appear in Table 1.

Breed	Experimental Population			Industry Sires			Breeder Submitted		
	Horned	Scurred	Polled	Horned	Scurred	Polled	Horned	Scurred	Polled
Brahman	180	136	86	84	0	5	9	8	116
Brangus	32	27	37	0	0	0	0	0	0
Charolais	0	0	0	57	3	36	0	0	0
Droughtmaster	33	22	39	24	16	60	0	0	0
Hereford	71	27	96	105	0	109	0	0	0
Limousin	22	1	29	64	5	79	0	1	142
Santa Gertrudis	102	31	52	13	1	7	0	1	40
Shorthorn	0	0	0	3	0	105	0	0	0
Tropical Composite	116	47	84	0	0	0	0	0	0
Simmental	0	0	0	60	1	36	0	0	0

Table 1. Counts of animals tested from each group (experimental population, industry sire or breeder submitted) for each phenotype (horned, scurred and polled).

RESULTS

Brahman

Data were from 402 animals in the experimental population (4 studs), 89 industry sires (25 studs) and 133 breeder submitted samples sires from 16 studs. In total 38 studs are represented.

	Horned	Scurred	Polled
PP	1	1	84
PH	24	119	96
PA	2	2	9
HH	235	15	4
HA	9	7	10
AA	2	0	4

Table 2. Counts of Brahman cattle with marker genotype PP, PH, PA, HH, HA or AA where P indicates a marker allele that is generally associated with a polled allele at the polled locus, H indicates a marker allele that is generally associated with a horned allele at the polled locus, and A indicates an ambiguous marker allele, that may be associated with either a polled or a horned allele at the polled locus. The phenotypes (horned, scurred or polled) are based on the animal's own trait record, no progeny information is included in this summary.

Ambiguous alleles are uncommon (frequency 4%) and in 89% of polled animals the test returns an unambiguous result. In 98% of these the result is consistent with the observed phenotype; only 4 polled animals are identified as homozygous horned by the test. Ambiguous alleles appear to be associated with both polled and horn. Heterozygous animals are most often scurred, frequently polled and occasionally horned. Animals with scurs are generally heterozygous.

Brangus

Data were from 96 animals in the experimental population (1 stud).

	Horned	Scurred	Polled
PP	0	0	5
PH	8	14	9
PA	1	5	16
HH	16	0	0
HA	7	8	2
AA	0	0	5

Table 3. Counts of Brangus cattle with marker genotype PP, PH, PA, HH, HA or AA where P indicates a marker allele that is generally associated with a polled allele at the polled locus, H indicates a marker allele that is generally associated with a horned allele at the polled locus, and A indicates an ambiguous marker allele, that may be associated with either a polled or a horned allele at the polled locus. The phenotypes (horned, scurred or polled) are based on the animal's own trait record, no progeny information is included in this summary.

Ambiguous alleles are common (frequency 26%) and in only 38% of polled animals does the test return an unambiguous result. Ambiguous alleles appear to be associated with both polled and horn. Heterozygous animals are most often scurred but also may be polled or horned. In all scurred animals where there are no ambiguous alleles the genotype is heterozygous.

Charolais

Data were from 96 industry sires from 38 studs.

	Horned	Scurred	Polled
PP	1	1	13
PH	1	1	13
PA	7	0	8
HH	7	0	0
HA	23	1	1
AA	18	0	1

Table 4. Counts of Charolais cattle with marker genotype PP, PH, PA, HH, HA or AA where P indicates a marker allele that is generally associated with a polled allele at the polled locus, H indicates a marker allele that is generally associated with a horned allele at the polled locus, and A indicates an ambiguous marker allele, that may be associated with either a polled or a horned allele at the polled locus. The phenotypes (horned, scurred or polled) are based on the animal's own trait record, no progeny information is included in this summary.

Although ambiguous alleles are common (frequency 41%) they occur mostly in horned animals that are unlikely to be submitted for testing. In 72% of polled animals the test returns an unambiguous result and in 100% of these the result is consistent with the observed phenotype. The number of Charolais tested is relatively small, but in this sample ambiguous alleles appear to be mainly associated with horns. If this trend is confirmed by additional data then some alleles currently classified as ambiguous may be declared to be associated with horns in Charolais. Heterozygous animals are generally polled, scurs are uncommon.

Droughtmaster

Data were from 94 experimental animals (2 studs) and 100 industry sires (40 studs).

	Horned	Scurred	Polled
PP	2	2	34
PH	8	24	37
PA	0	3	15
HH	34	4	1
HA	11	5	7
AA	2	0	5

Table 5. Counts of Droughtmaster cattle with marker genotype PP, PH, PA, HH, HA or AA where P indicates a marker allele that is generally associated with a polled allele at the polled locus, H indicates a marker allele that is generally associated with a horned allele at the polled locus, and A indicates an ambiguous marker allele, that may be associated with either a polled or a horned allele at the polled locus. The phenotypes (horned, scurred or polled) are based on the animal's own trait record, no progeny information is included in this summary.

Ambiguous alleles are relatively uncommon (frequency 14%) and in 73% of polled animals the test returns an unambiguous result. In 99% of these the result is consistent with the observed phenotype; only one polled animal is identified as homozygous horned by the test. Heterozygous animals are most often polled, but also frequently scurred or horned. The majority of scurred animals are heterozygous.

Hereford

Data were from 194 experimental animals (4 studs) and 214 industry sires (13 studs).

	Horned	Scurred	Polled
PP	0	0	102
PH	5	17	44
PA	0	7	49
HH	95	1	2
HA	60	1	4
AA	16	1	4

Table 6. Counts of Hereford cattle with marker genotype PP, PH, PA, HH, HA or AA where P indicates a marker allele that is generally associated with a polled allele at the polled locus, H indicates a marker allele that is generally associated with a horned allele at the polled locus, and A indicates an ambiguous marker allele, that may be associated with either a polled or a horned allele at the polled locus. The phenotypes (horned, scurred or polled) are based on the animal's own trait record, no progeny information is included in this summary.

Ambiguous alleles are relatively common (frequency 20%) and in 72% of polled animals the test returns an unambiguous result. In 99% of these the result is consistent with the observed phenotype; only 2 polled animals are identified as homozygous horned by the test. Ambiguous alleles appear to be generally associated with horns, and if this trend is confirmed by additional data then some alleles currently classified as ambiguous may be declared to be associated with horns in Hereford. Heterozygous animals are most often polled, but also frequently scurred or horned. All but one of the scurred animals with no ambiguous alleles are heterozygous.

Limousin

Data were from 52 experimental animals (1 stud), 148 industry sires from a wide range of studs and 143 breeder submitted samples (22 studs).

	Horned	Scurred	Polled
PP	0	0	59
PH	1	2	38
PA	3	2	102
HH	21	0	0
HA	40	1	11
AA	21	2	40

Table 7. Counts of Limousin cattle with marker genotype PP, PH, PA, HH, HA or AA where P indicates a marker allele that is generally associated with a polled allele at the polled locus, H indicates a marker allele that is generally associated with a horned allele at the polled locus, and A indicates an ambiguous marker allele, that may be associated with either a polled or a horned allele at the polled locus. The phenotypes (horned, scurred or polled) are based on the animal's own trait record, no progeny information is included in this summary.

Ambiguous alleles are common (frequency 42%), and in only 39% of polled animals is an unambiguous result returned by the test. However, in all of these the result is consistent with the observed phenotype; no polled animals are identified as homozygous horned by the test. Ambiguous alleles appear to be associated with both horns and poll. Heterozygous animals are almost always polled, and scurs are uncommon.

Santa Gertrudis

Data were from 185 experimental animals from 2 studs, 21 industry sires (2 studs) and 41 breeder submitted samples (2 studs). In total 5 studs are represented.

	Horned	Scurred	Polled
PP	1	0	15
PH	10	23	60
PA	1	7	16
HH	73	1	1
HA	29	1	6
AA	1	1	1

Table 8. Counts of Santa Gertrudis cattle with marker genotype PP, PH, PA, HH, HA or AA where P indicates a marker allele that is generally associated with a polled allele at the polled locus, H indicates a marker allele that is generally associated with a horned allele at the polled locus, and A indicates an ambiguous marker allele, that may be associated with either a polled or a horned allele at the polled locus. The phenotypes (horned, scurred or polled) are based on the animal's own trait record, no progeny information is included in this summary.

Ambiguous alleles are relatively rare (frequency 13%), and in 77% of polled animals an unambiguous result is returned by the test. In 99% of these the result is consistent with the observed phenotype; only one polled animal was identified as homozygous horned by the test. Ambiguous alleles appear to be generally associated with horns, and if this trend is confirmed by additional data then some alleles currently classified as ambiguous may be declared to be associated with horns in Santa Gertrudis. Heterozygous animals are most often polled, but also frequently scurred or horned. All but one of the scurred animals with no ambiguous alleles are heterozygous.

Shorthorn

Data were from 108 industry sires from 39 studs.

	Horned	Scurred	Polled
PP	0	0	12
PH	0	0	24
PA	0	0	17
HH	3	0	0
HA	0	0	35
AA	0	0	17

Table 9. Counts of Shorthorn cattle with marker genotype PP, PH, PA, HH, HA or AA where P indicates a marker allele that is generally associated with a polled allele at the polled locus, H indicates a marker allele that is generally associated with a horned allele at the polled locus, and A indicates an ambiguous marker allele, that may be associated with either a polled or a horned allele at the polled locus. The phenotypes (horned, scurred or polled) are based on the animal's own trait record, no progeny information is included in this summary.

Ambiguous alleles are common (frequency 40%) and in only 34% of polled animals does the test return an unambiguous result. However, in all of these the result is consistent with the observed phenotype; no polled animals are identified as homozygous horned by the test. It may be that some of the alleles declared to be ambiguous are seldom associated with horns in the Shorthorn breed, but with only 3 horned animals in the sample there is no power to estimate a Shorthorn specific association frequency. All of the animals known to be heterozygous are polled, no scurs were recorded.

Simmental

Data were from 97 industry sires representing 10 studs.

	Horned	Scurred	Polled
PP	1	0	8
PH	2	1	12
PA	2	0	14
HH	17	0	0
HA	24	0	0
AA	14	0	2

Table 10. Counts of Simmental cattle with marker genotype PP, PH, PA, HH, HA or AA where P indicates a marker allele that is generally associated with a polled allele at the polled locus, H indicates a marker allele that is generally associated with a horned allele at the polled locus, and A indicates an ambiguous marker allele, that may be associated with either a polled or a horned allele at the polled locus. The phenotypes (horned, scurred or polled) are based on the animal's own trait record, no progeny information is included in this summary.

Ambiguous alleles are common (frequency 37%) but more frequent in horned animals, so in 56% of polled animals the test returns an unambiguous result. In all of these the result is consistent with the observed phenotype; no polled animals are identified as homozygous horned by the test. Ambiguous alleles appear to be almost always associated with horns in Simmental: no heterozygous HA animals are polled while 24 have horns. If this trend is confirmed by additional data then some alleles currently classified as ambiguous may be declared to be associated with horns in Simmental. Most heterozygous animals are polled, scurs are uncommon.

Tropical Composite

Data were from 247 animals in the experimental population (all from one property).

	Horned	Scurred	Polled
PP	9	8	43
PH	24	21	18
PA	6	13	21
HH	42	3	1
HA	18	1	0
AA	17	1	1

Table 11. Counts of Tropical Composite cattle with marker genotype PP, PH, PA, HH, HA or AA where P indicates a marker allele that is generally associated with a polled allele at the polled locus, H indicates a marker allele that is generally associated with a horned allele at the polled locus, and A indicates an ambiguous marker allele, that may be associated with either a polled or a horned allele at the polled locus. The phenotypes (horned, scurred or polled) are based on the animal's own trait record, no progeny information is included in this summary.

Ambiguous alleles are relatively common (frequency 20%) and in 74% of polled animals the test returns an unambiguous result. However, in 98% of these the result is consistent with the observed phenotype; only one polled animal is identified as homozygous horned by the test. Data are from only one property, but in this sample ambiguous alleles appear to be generally associated with horns. If this trend is confirmed by additional data then some alleles currently classified as ambiguous may be declared to be associated with horns in Tropical Composite. The proportion of horned animals declared to be homozygous polled is relatively high, so it is likely that a marker allele we associate with polled is in this breed sometimes associated with horns. Alternatively, there may be other genes apart from those in the polled locus region affecting horns in Tropical Composite cattle. Heterozygous animals are most often horned, but also commonly scurred or polled. Scurred animals are generally heterozygous, but scurs also appear in homozygous PP animals, and sometimes in homozygous HH animals.

CONCLUSIONS

In Brahman and Hereford cattle we have a large sample size from many sources, a good balance between polled and horned, and a clear majority of polled animals tested return an unambiguous result. Few horned animals carry the marker alleles we associate with polled and few polled animals carry the marker alleles we associate with horns. These conclusions also apply to Droughtmaster, however we have fewer records for this breed.

For Charolais and Simmental we have only a relatively small number of records, but from a reasonable number of studs. The proportion of ambiguous alleles is high but there is good evidence that in these breeds most if not all ambiguous alleles could be called as being associated with horns, suggesting that the test may be of value in Charolais and Simmental. In both breeds we require additional records to confirm the associations.

In Santa Gertrudis and Tropical Composite cattle a clear majority of polled animals tested return an unambiguous result, but data are from a limited range of studs so more data are required to confirm that these results can be generalised to the wider breed populations. Additional data may also confirm that alleles currently classed as ambiguous may in fact be reliably used to predict a horn genotype in these breeds. Additional data are also required to establish why alleles that in other breeds are almost always associated with polled are in Tropical Composite cattle often associated with horns.

For Shorthorn the proportion of unambiguous results is relatively low. It may be that alleles currently classed as ambiguous may in fact be reliably used to predict poll genotype in Shorthorn, but confirming this would require more samples from horned animals to supplement the 3 horned samples currently available.

For Brangus and Limousin the proportion of unambiguous results is relatively low, and unless augmented with additional data (for example, additional markers or progeny test data), this test has limited ability to predict genotype.

Scurs In the Charolais, Limousin, Shorthorn and Simmental breeds, heterozygous animals are almost always polled, and scurs are uncommon. This model, with polled dominant to horned, is often assumed for *Bos taurus* animals. In the Brahman, Brangus, Droughtmaster, Hereford and Santa Gertrudis breeds, heterozygous animals are most often polled but also commonly scurred, and sometimes horned. In these breeds scurs are uncommon in other than heterozygous animals. In the Tropical Composite animals tested heterozygous showed all phenotypes, horned, scurred and polled. Scurs also occurred in homozygous animals. These breed differences in the expression of scurs indicate that variation in the prevalence of scurs is due to genetics. In breeds where scurs are common a significant proportion of heterozygous animals will be scurred, and (apart from in Tropical Composite) scurs is an indicator of carrying a horned allele. However, polled animals can also be heterozygous so the absence of scurs is not an indicator of an absence of horned alleles.

PRESENTATION OF RESULTS

Where the result is not consistent with the observed phenotype it could be due to an incorrect phenotype record, to variation in horn status not due to the polled locus, or to a marker allele having an alternative association to that generally the case. Because of this last possibility, we never assume that an association between a marker allele and an allele at the polled locus is complete: we allow a small probability for the alternative association. Consequently results for the marker test are presented as probabilities of being homozygous polled, and that probability is never 100%. For example: if an animal carries two copies of a marker allele that we believe to be associated with a polled allele at the polled locus 95% of the time, then we report that the probability of being genetically homozygous polled as 90% (as $0.95 \times 0.95 = 0.9025$). If instead the animal carried one copy of that marker allele and one copy of an ambiguous marker allele that we believe to be associated with a polled allele at the polled locus only 50% of the time, then we report that the probability of being genetically homozygous polled as 48% (as $0.95 \times 0.5 = 0.475$).

ON-GOING WORK

To improve the test across a range of breeds, research continues on a number of fronts. As additional samples from animals with phenotype records are received, the estimated associations between marker alleles and alleles at the polled locus are refined. Soon we may have breed specific estimates for some marker alleles for some breeds. Currently estimates do not take account of progeny horn phenotypes or of horn phenotypes and marker genotypes on more distantly related animals. Procedures for incorporating this information are under development. Methods are also under development for shifting a herd from horned to polled in an optimal way, minimising loss of progress in other traits. Finally, markers close to the one used in this test are being tested to determine whether they add precision to the current test.

COMMERCIALISATION

Currently the test is being provided by the Animal Genetics Laboratory at The University of Queensland. We have also made the test available to anyone else who wishes to provide a commercial service. The method of reporting of results from these providers may differ from that described here, as may the populations used to validate the test and consequently the estimated associations between marker alleles and alleles at the polled locus.