



# Inheritance patterns of the Polled and Scur genes in South African beef cattle breeds

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The genetic mechanisms and inheritance patterns of the polled and scur phenotypes in local South African beef cattle breeds

Industry Sector: Cattle And Small Stock

Research Focus Area: Animal Health And Welfare

Research Institute: Department Of Animal & Wildlife Sciences, University Of Pretoria

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## Executive Summary

### Introduction

It is standard practice to dehorn cattle at a young age by means of physical dehorning, but in most cases without the appropriate pain relief. The practice of dehorning has increasingly become a welfare concern and alternatives to dehorning are advocated worldwide. Breeding genetically polled cattle is a long-term, non-invasive and welfare friendly alternative to dehorning. Identification of genetically polled animals through a diagnostic test would therefore be advantageous, but a specific commercial diagnostic test for the polled phenotype is not currently available in South Africa. The DNA tests that are available internationally are applicable to European *Bos taurus* breeds, which can give inconclusive results for indigenous South African and Sanga cattle breeds. Furthermore, the commercial diagnostic tests available for Taurine breeds can not identify carriers of either the scur gene or the African horn gene.

Over the past two decades commercial beef producers and feedlots in South Africa have indicated a preference for polled breeds, due to increased awareness of animal welfare and market preferences. In

South Africa there are a number of polled breeds of European descent such as the Hereford, Angus, Charolais and Limousin, as well as a few local breeds, including the South African Bonsmara, Tuli and Drakensberger, that introgressed the polled gene. The Bonsmara breed requested research on the identification of homozygous polled bulls and the first research project in South Africa was performed at the Department of Animal and Wildlife Sciences, UP (Schmullian, 2006). This study was based on three Bonsmara families and the available microsatellite markers at that time were used. The study by Schmullian (2006) found linkage between the polled phenotype in the South African Bonsmara and alleles of nine microsatellite markers located on BTA1. Since the completion of this research project, the Bovine genome sequence has been completed in 2009 with high through-put molecular technology (Bovine HapMap Consortium, 2009), providing genomic information and high-density SNP chips.

The majority of previous research on the *POLLED* locus and polledness has been performed in European breeds, which does not provide a basis for identification of the causative mutation for polledness or scurs in indigenous South African cattle breeds. These breeds are genetically distinct from the European *Bos taurus* breeds (Makina *et al.*, 2014) and besides the two main types of cattle, *Bos taurus* and *Bos indicus*, indigenous African cattle, such as the Sanga, are also found in South Africa.

## Objective Statement

This study focused on local South African beef cattle breeds to gain an understanding of the genetic basis and inheritance of the *Polled* and *Scur* genes by using pedigree data from phenotyped animals, as well as high density SNP data. The availability of DNA and high through-put molecular technology holds the potential to provide insight on the genetic mechanisms of polled and scurred animals with higher precision, compared to the microsatellite markers that were previously available.

## Project Aims

1. To evaluate whether the Celtic mutation on the POLL locus is the causative mutation for polledness in Bonsmara and Drakensberger
2. To perform a genome wide association study of the Polled and Scur genes based on phenotypic data and genotypic data from the GGP Bovine 150K SNP bead chip
3. To apply sequence data available from the Bovine Genomics Program to finemap the suspected regions for the Polled and Scur genes

## Results

A total of 890 Bonsmara and 224 Drakensberger animals were screened for their status for the Celtic mutation at the *POLLED* locus using a PCR-based diagnostic test. It was possible to distinguish between heterozygous and homozygous polled individuals, but scurs could not be identified on a genotypic level based on the Celtic variant. The majority of animals screened, tested heterozygous polled, with homozygous polled animals occurring at a relatively low frequency. Based on the results of this Celtic screening, a total of 217 Bonsmaras (including homozygous polled, heterozygous clean polled and scurred animals) were genotyped using the GGP Bovine 150K SNP bead chip. Additional genotypes from the Bovine Genomics Program (BGP) were also included in this study.

Haplotype analysis of the *POLLED* locus revealed a reduced genetic diversity around the Celtic allele, with only two haploblocks (1.0-2.2 Mb) observed in the homozygous polled animals investigated. One of these haploblocks of six SNPs encompassed the Celtic mutation and presented only three distinct alleles with major differences in terms of frequencies. This result suggests that the Celtic allele was introgressed in the Bonsmara breed from a very limited number of founders. A low haplotype diversity combined with an intense selection on the polled phenotype in the Bonsmara breed can result in the selection of deleterious alleles linked with the Celtic mutation through a hitchhiking mechanism. Therefore, it is of primary importance to maintain some genetic variability around the Celtic allele.

Preliminary results of the GWAS study, based on 150k SNP chip data, indicated significant association for the scurs phenotype with three SNPs on BTA5, which contradicts previous findings that mapped the *SCURS* locus to BTA19. For the *POLLED* locus, preliminary results show significant association between the polled phenotype and BTA1, as expected. Of interest is another significant association with the polled phenotype that were observed on BTA28, which was not reported in previous studies. The significant SNPs that were identified in the GWAS analysis of the *POLLED* and *SCURS* loci will be annotated to identify candidate genes and to investigate the potential significant physiological pathways of these SNPs.

## Conclusion

The *POLLED* Celtic variant was validated as the causative mutation of polledness in three South African beef cattle breeds and can be used as an efficient diagnostic test for polledness. This study also highlighted the current difficulties and limitations of accurate phenotypic recording of the horn status. It also confirmed that scurs cannot be identified on a genotypic level with the Celtic screening. Preliminary results of genotypic SNP data indicated significant association for the scurs phenotype on BTA5 in the Bonsmara, but these results need to be further investigated.

The following scientific output were achieved for the project:

Grobler, R., Visser, C. & van Marle-Köster, E., 2017. Accelerating selection for polledness in the South African Bonsmara using DNA technology. 50<sup>th</sup> South African Society for Animal Science (SASAS) Congress, Port Elizabeth, 18 – 21 September 2017.

Grobler, R., van-Marle-Köster, E., Visser, C. & Capitan, A., 2018. Haplotype variation at the *POLLED* locus in the South African Bonsmara cattle breed. World Congress on Genetics Applied to Livestock Production (WCGALP) 11-16 February, Auckland.

Grobler, R., Visser, C., Capitan, A. & van Marle-Köster, E., 2018. Validation of the *POLLED* Celtic variant in South African Bonsmara and Drakensberger beef cattle breeds. *Livestock Science*. 217, 136-139.

## Popular Article

### Introduction

## Identifikasie Van Poena Status In Die Bonsmara Met Behulp Van DNA Tegnologie

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### Inleiding

Die voordele van poenskop beeste vir die kommersiële vleisbeesindustrie is alombekend en wêreldwyd word die druk vir meer menslike praktyke in terme van dierebehandeling hoër, as gevolg van die impak op diere-welsyn. Alhoewel die opsie daar is om kalwers te onthoring, dui navorsing aan dat dit 'n pynvolle prosedure is, ten spyte van die tegniek of voorsorgmaatreëls wat gebruik word. Behalwe dat die onthoring van kalwers nie welsynsvriendelik is nie, is dit ook tydrowend en arbeidsintensief, en veroorsaak dit ook stres wat die groei van die kalf negatief kan affekteer. Hierdie faktore het dan ook 'n verdere ekonomiese implikasie vir die boer. Dit is ook 'n opsie om poenskop beeste te selekteer gebaseer op fenotipiese rekords, maar hierdie proses is egter tydrowend en oneffektief, en veroorsaak stadige genetiese vordering. Deur gebruik te maak van DNA tegnologie om poenskop diere te identifiseer, kan seleksie vinniger en meer effektief plaasvind en genetiese vordering sal ook vinniger toeneem. Verder is dit ook 'n welsynsvriendelike alternatief, so wel as 'n langtermyn oplossing vir die onthoring van kalwers.

### Poenskop oorerwing

Die Poena geen is outosomaal dominant en indien teenwoordig, sal die uitdrukking van die horing fenotipe onderduk word. Daar is twee allele teenwoordig by die Poena geen, P en p, en diere wat die dominante P alleel dra is fenotipies poenskop. Homosigotiese poenskop diere dra twee dominante P allele (PP), terwyl heterosigotiese poenskop (Pp) diere een dominante P alleel dra en een horing alleel. Dus, diere wat twee p allele dra het dan die horing fenotipe (pp). As gevolg van dominansie, kan daar nie onderskei word tussen die homosigoot en heterosigoot poena fenotipe nie. Dus is dit nodig vir 'n genetiese toets om draers van die poena en horing allele te identifiseer.

Afhangende van die poena status van die moeder en vader, word die poena allele in verskillende proporsies oorgedra na die nageslag (Figuur 1). Bv. Wanneer 'n homosigotiese poena (PP) bul, wat dan twee dominante poena allele dra, geteel word met 'n horing koei (pp), is daar 'n 100% kans dat die nageslag fenotipies poenskop sal wees, omdat die nageslag een dominante P alleel kry van die vader en een horing

alleel van die moeder. Maar wanneer n heterosigotiese poena ( $Pp$ ) bul gebruik word, verminder die kans vir poenskop nageslag met 50% (Figuur 1).

**Figuur 1** Die moontlike genotipiese proporsies vir verskillende paringsituasies van horing, hetero- en homosigotiese poenskop individue

Die oorerwing van die poena word verder gekompliseer deur die scurs fenotipe, as gevolg van epistatiese interaksie tussen die Poena en Scurs gene. Scurs is klein horingagtige vergroeiings wat op dieselfde plek as horings op die kop voorkom, maar hierdie abnormale vergroeiings is losweg aan die skedel geheg en is beweeglik (Figuur 2). Scurs is geslagsbeïnvloed en word gevolglik verskillend oorgeërf in manlike en vroulike diere. Dit is waargeneem dat scurs kan voorkom in diere wat heterosigoties poenskop is, en dat scurs meer in manlike diere as in vroulike diere voorkom.

**Figuur 2** Die poenskop (A en B) en variasie van die scurs (C – F) fenotipes soos waargeneem in die Bonsmara

### Die Poena Projek by UP

Die Poena geen is geleë op chromosoom 1 (BTA1) en minstens twee verskillende variante is verantwoordelik vir die poenskop fenotipe in beeste, naamlik die Celtic ( $P_C$ ) en Friesian ( $P_F$ ) variante (Allais-Bonnet et al., 2013). Die Celtic ( $P_C$ ) variant is verantwoordelik vir die poenskop fenotipe in die meeste *Bos taurus* rasse van Europese herkoms, terwyl die Friesian ( $P_F$ ) variant hoofsaaklik voorkom in die Holstein Friesian ras.

In samewerking met 'n navorser van Frankryk (INRA), is Bonsmara diere getoets vir beide die Celtic ( $P_C$ ) en Friesian variante ( $P_F$ ). Dit is vasgestel dat al die Bonsmara diere met n poenskop fenotipe, dra ten minste een alleel van die Celtic variant en geen diere is positief getoets vir die Friesian variant nie. Hierdie bevinding is in lyn met die geskiedenis en ontwikkeling van die Bonsmara vanuit n Europese *Bos taurus* ras. 'n Groter groep Bonsmaras is getoets vir die Celtic variant ( $P_C$ ) en dit is bevestig dat die Celtic variant ( $P_C$ ) van die Poena geen verantwoordelik is vir die poenskop fenotipe in die Suid-Afrikaanse Bonsmara (Grobler et al., 2018).

Deur gebruik te maak van 'n haarmonster, word DNA geëkstraer uit die haarwortels. Die DNA word dan gebruik om die dier te toets vir die Celtic variant ( $P_C$ ) deur gebruik te maak van 'n PCR-gebaseerde diagnostiese toets. Gevolglik kan draers van die  $P_C$  variant geïdentifiseer word en sodoende kan diere ook as homo- of heterosigoties poena geïdentifiseer word op 'n genotipiese vlak.

Bonsmara bulle en koeie, asook sekere kalwers, is uit spesifieke kuddes geselekteer om poenskop diere te identifiseer en tot dusver is n totaal van 890 Bonsmaras getoets vir die Celtic variant ( $P_C$ ) met die bogenoemde diagnostiese toets. 'n Hoë frekwensie poenskop diere is waargeneem, waarvan die meerderheid diere heterosigoties poena getoets het (Figuur 3). Dit beteken dat hierdie diere slegs een  $P_C$  alleel dra, asook een horing alleel, wat dan moontlik oorgedra kan word aan die dier se nageslag. Alhoewel homosigotiese poena diere wel waargeneem is, is dit waargeneem in slegs 12% van die diere wat tot dusver getoets is (Figuur 3). Die Bonsmara diere wat horing getoets het, is by n relatiewe hoë frekwensie van 42% waargeneem (Figuur 3). Dit is tog nodig om te noem dat die meerderheid van die homosigotiese poenas in een kudde waargeneem is wat vir meer as twee dekades al spesifiek selekteer vir die poenskop fenotipe. Alhoewel hierdie toets kan onderskei tussen homo- en heterosigote poenskop diere, kan die toets nie scurs op 'n genotipiese vlak identifiseer nie en verdere navorsing is nodig vir scurs.

**Figuur 3** Die genotipe frekwensie van die Celtic variant ( $P_C$ ) soos getoets in 890 Bonsmaras

### Implikasies vir SA Bonsmara

Die diagnostiese DNA toets kan effektief gebruik word om heterosigotiese en homosigotiese poena diere te identifiseer op n genotipiese vlak. Hierdie toets kan egter nie gebruik word om scurs op n genotipe vlak te identifiseer nie, omdat beide poenskop en scurs diere genotopies heterosigoties poenskop toets ( $Pp$ ). Dit is dan juis waarom dit belangrik is om die horingstatus van diere vroegtydig en akkuraat aan te teken. Die poenskop fenotipe is maklik om te observeer en verander nie tydens die dier se leeftyd nie. Die scurs fenotipe is egter moeiliker om aan te teken, omdat dit dikwels verwar word met horings of eers later uitgedruk word. Daarom word dit aanbeveel dat beste ondersoek word by n jong ouderdom (gewoonlik tydens speen), asook tussen 18 en 24 maande.

Met behulp van hierdie DNA tegnologie kan die poena status van diere vroegtydig geïdentifiseer word, wat sodoende die genetiese seleksie van poena diere sal vergemaklik, asook versnel. Verder hou dit 'n ekonomiese voordeel vir telers in wanneer gesertifiseerde poenskop bulle bemark kan word. Meer poenskop diere in die mark sal ook 'n voordeel inhou deur arbeidskoste te verlaag en diere welsyn te bevorder omdat die onthoring van diere dan mettertyd nie meer nodig sal wees nie. Die relatiewe hoë frekwensie van horing diere wat waargeneem is bevestig juis die belangrikheid van 'n DNA toets, deurdat telers eers die poena status van diere op 'n genotipiese vlak moet bevestig voordat vermeende poena

diere ingesluit word in 'n paringsprogram. Dit is veral belangrik vir bulle wat vir teelddoeleindes en veilings gebruik gaan word.

## Erkennings

Dankie aan elke boer wat haarmonsters en inligting bygedrae het vir die Poena projek, en ook spesifiek vir Charl Uys vir sy hulp. Dankie aan Prof E. van Marle-Köster en Dr C. Visser; die studiepromotors op die PhD projek. Dank aan RMRD SA en die NRF vir befondsing.

## Verwysings

Allais-Bonnet et al., 2013. PloS ONE. 8, 1-14.

Grobler et al., 2018. Livestock Science. 217, 136-139.

## Footer And Tags And Categorisation

***Please contact the Primary Researcher if you need a copy of the comprehensive report of this project on : – mmakgahlela@arc.agric.za***

- Animal Health and Welfare, Cattle and Small Stock
- ◆ 2019, CSS, MarleKoster, Online, UP
- < Gene expression: Nguni and Bonsmara
- > Gene expression: Nguni and Bonsmara cattle

## DEADLINES for RESEARCHERS 2021

Proposals for 2021: TBC  
Progress reports: 28 Jan 21

Final reports: 29 Jan 21 Final includes comprehensive report and popular article

## COMMITTEE MEETINGS for 2021

RMRDSA CSS Planning - TBC

Project Committee - TBC

Pork Planning - TBC



## Calendar

<span style="float: left;">&lt;</span> <span style="float: right;">&gt;</span> <b>Apr 2021</b>						
Sun	Mon	Tue	Wed	Tur	Fri	Sat
				1	2	3
4	5	6	7	8	9	10
11	12	13	14	15	16	17
18	19	20	21	22	23	24
25	26	27	28	29	30	

## PORK Priority Areas

## **Cattle & Small Stock Programmes**

### **1 Sustainable natural resource utilisation**

### **2 Improvement of Livestock production and forage**

### **3 Management of agricultural risk to create a resilient Red Meat sector**

### **4 Sustainable health and welfare for the Red Meat sector**

### **5 Enhancement of production and processing of Animal Products**

### **6 Consumer and market development of the Red Meat sector**

### **7 Commercialisation of the emerging sector**

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