



Investigations of the variations of horn phenotypes and the genetic architecture of scurs

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Content

- Introduction
- Phenotyping of different horn phenotypes
 - Material and methods
 - Results and discussion
- Genome-wide association studies of the *scurs* locus
 - Material and methods
 - Results and discussion
- Conclusion and outlook





Horn Phenotypes

Introduction

Material & Methods

Results & Discussion



Horns

- Typical and diverse feature of cattle
- Permanent
- Firmly attached to the skull



Polled

- Genetically hornless
- Complete absence of horns or any corneous growths



Scurs

- Incomplete developed horns
- Loosely attached to the skull by soft tissue



Genetic Background

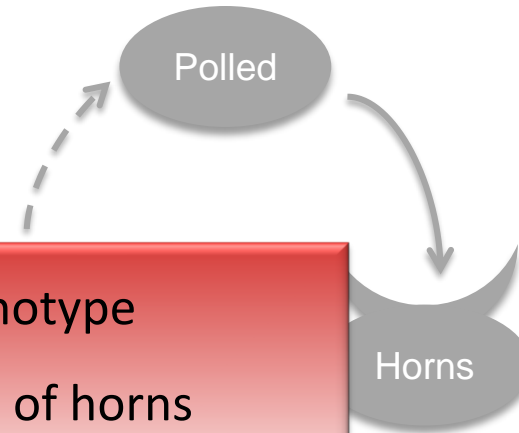
Introduction

Material & Methods

Results & Discussion

Inheritance of Horns – 3-Loci Model

- The *POLLED* locus has been identified on chromosome 1
 - Allelic heterogeneity – Celtic (*Pc*) and Friesian (*Pf*) allele
 - Direct gene test for polledness is available



Objectives

- Examination of the scurred phenotype
- Investigations of the inheritance of horns
- Genetic analysis of the scurs locus

	<i>Sc/Sc</i>	<i>Sc/sc</i>	<i>sc/sc</i>
<i>P/P</i>	P	P	P
<i>P/p</i>	S	S	P
<i>p/p</i>	H	H	H

P: polled (non-scurred), S: scurred, H: horned



Scurs Phenotype

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Results & Discussion

- Selection of farms that breed for polledness and do not dehorn calves routinely
- Examination of bulls of breeding companies
- Phenotyping was carried out by the same person
- Horn area was shaved





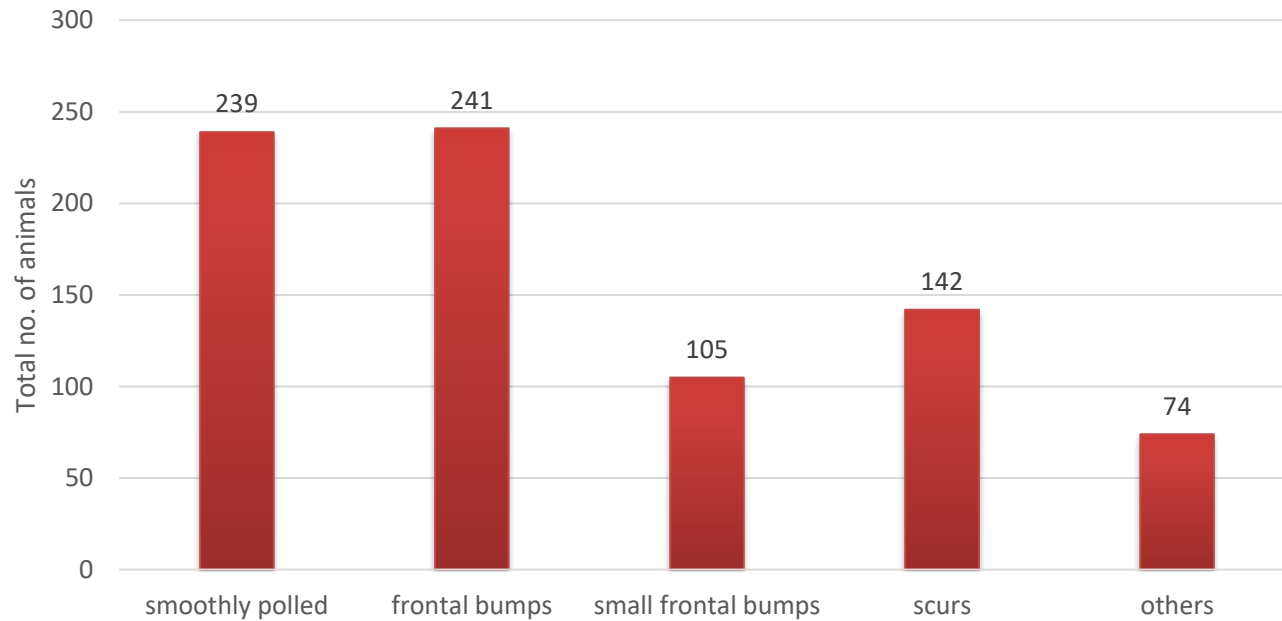
Phenotyping

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Results & Discussion

Observed Horn Phenotypes





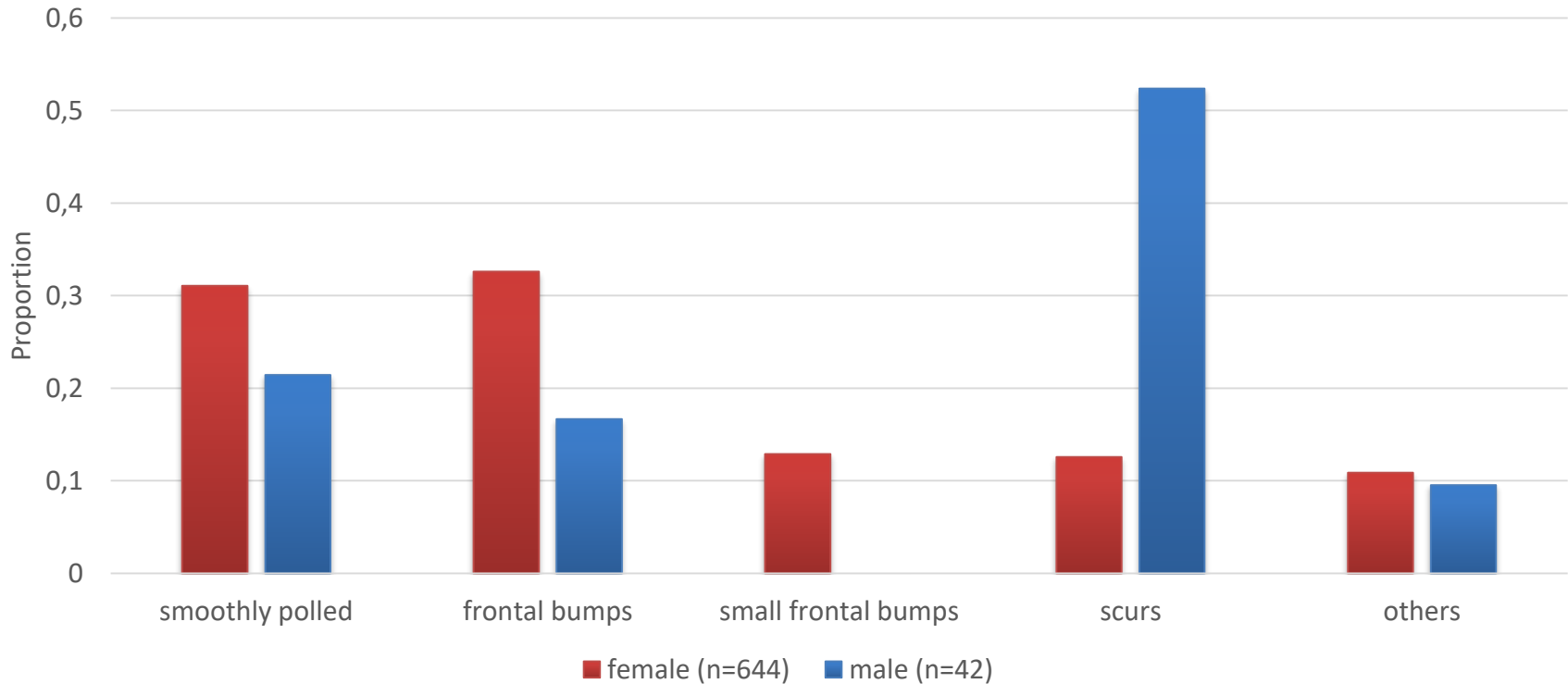
Does the Expression of Scurs Depend on Sex?

Introduction

Material & Methods

Results & Discussion

Comparison of Horn Phenotypes of Male and Female Cattle



Long and Gregory (1978):

Expression of scurs in double heterozygous (P/p ; Sc/sc) animals depends on sex

- Females do not express scurs
- Males express scurs



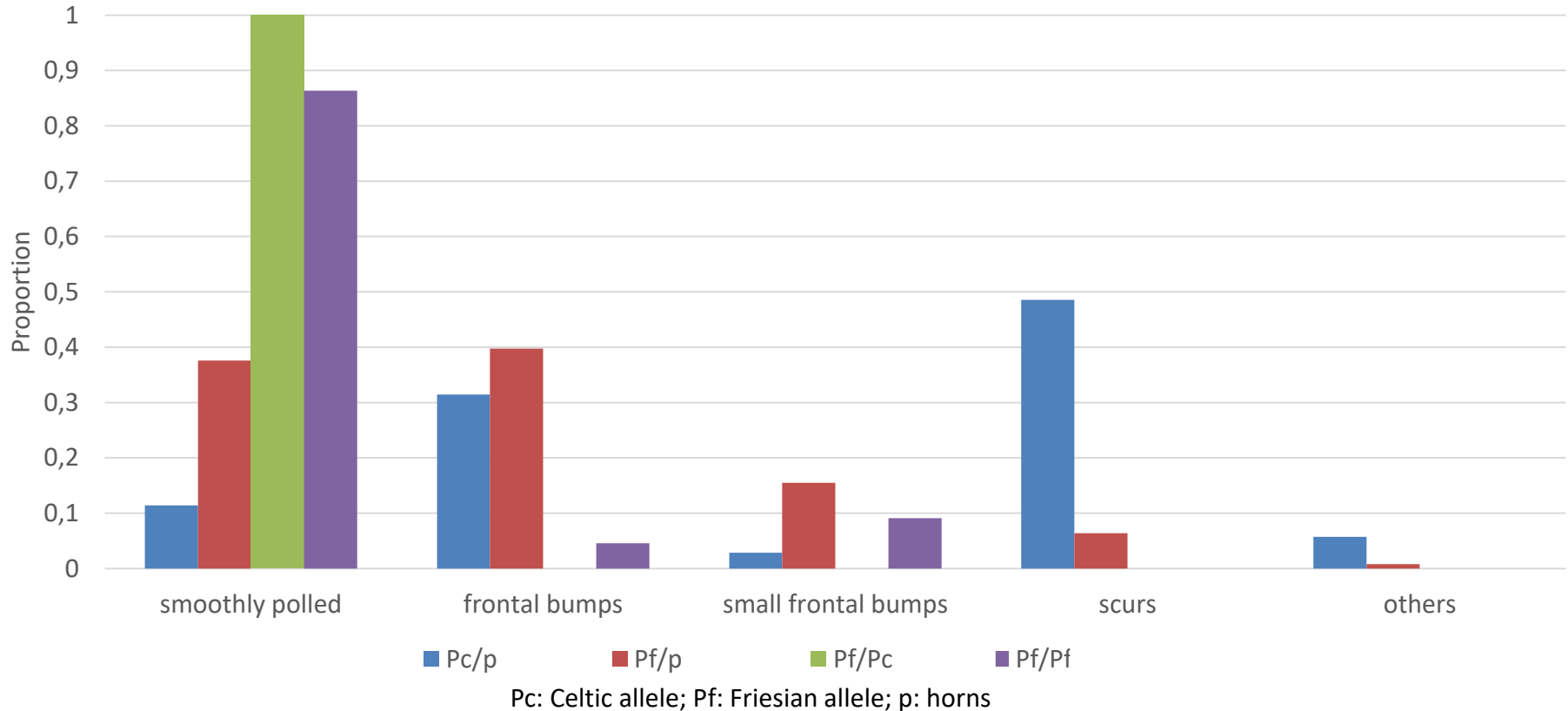
Expression of Scurs and Horn Genotype

Introduction

Material & Methods

Results & Discussion

Horn Phenotypes by Genetic Horn Status



Wiedemar et al. (2014)

Scurs are not expressed in homozygous polled animals



Experimental Setup for GWAS

Introduction

Material & Methods

Results & Discussion

Requirements:

- Horn genotype: Pf/p
- Sex: female
- Phenotype
 - Smoothly polled
 - Scurs
 - Frontal bumps
- Age > 700d (except for scurs)

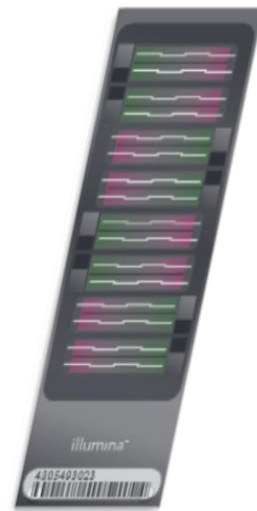
Hornstatus	No. of animals
Pc/p	55
Pf/p	568
Pf/Pc	8
Pf/Pf	27

Results of the direct gene test for polledness
(Pc: Celtic allele; Pf: Friesian allele; p: horns)

240 animals were genotyped

- BovineHD Genotyping BeadChip (777,962 SNP Markers)

Phenotype	No. of Animals	Data Set 1	Data Set 2
smoothly polled	77	Control	Control
scurs	31	Case	Case
frontal bumps	132	--	Case





GWAS – Quality Control

Introduction

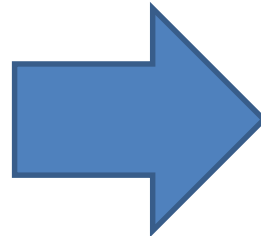
Material & Methods

Results & Discussion

Statistical analysis (PLINK! -- v1.07)

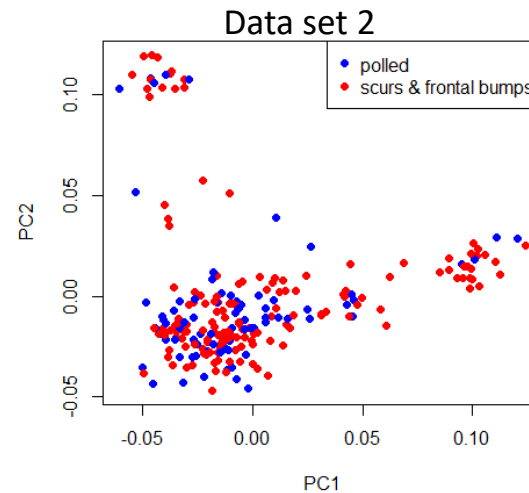
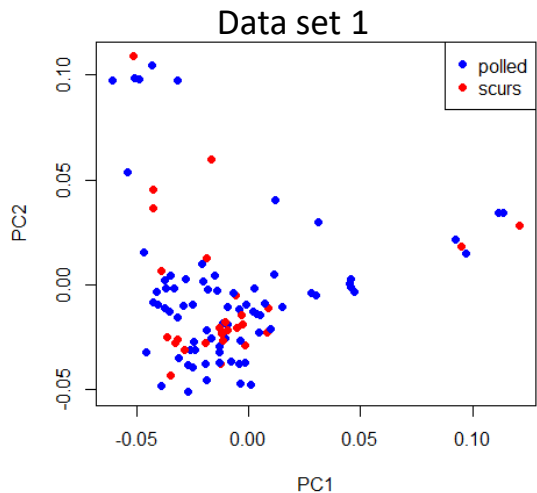
Quality control

- GC-score > 0.2
- MAF > 0.05
- Missingness per marker < 0.05
- Missingness per individual < 0.05



- 8 Animals excluded due to high missingness
- 218,377 Markers excluded due to high missingness or homozygosity

Principal components plot for stratifications (substructures in the population)





GWAS - Clustering

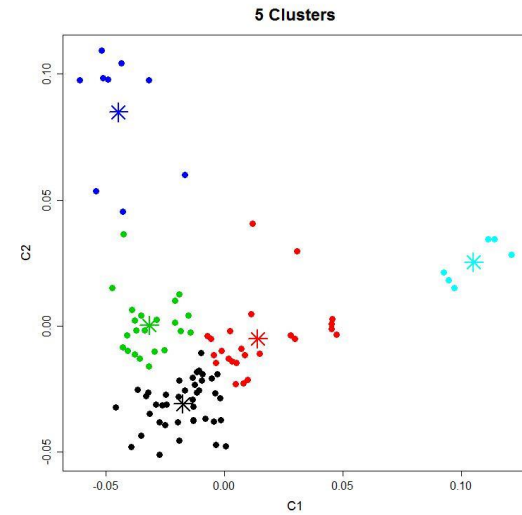
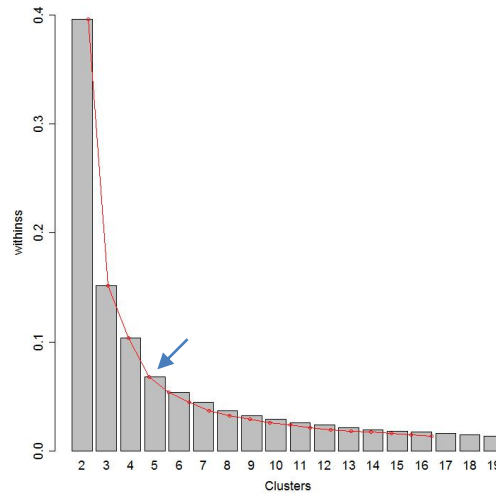
Introduction

Material & Methods

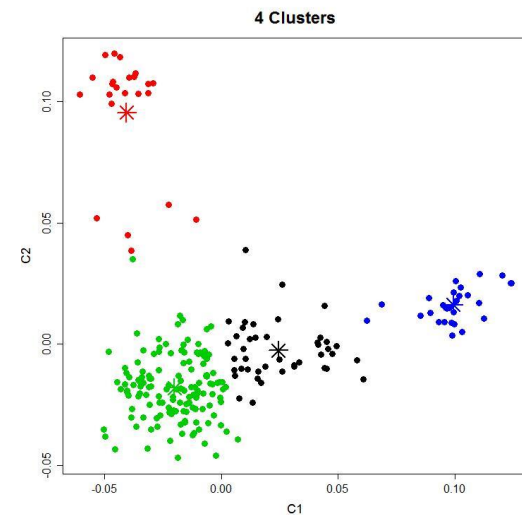
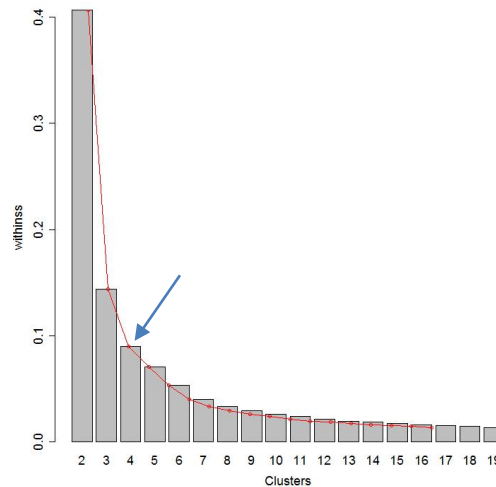
Results & Discussion

K-Means clustering in R

Data Set 1



Data Set 2



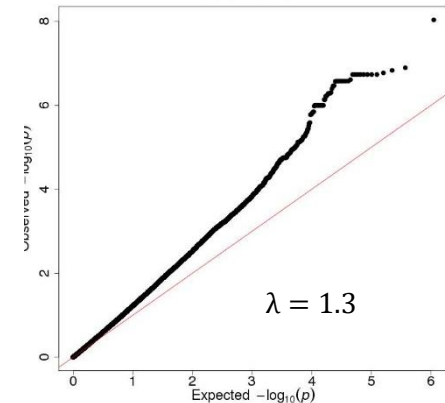
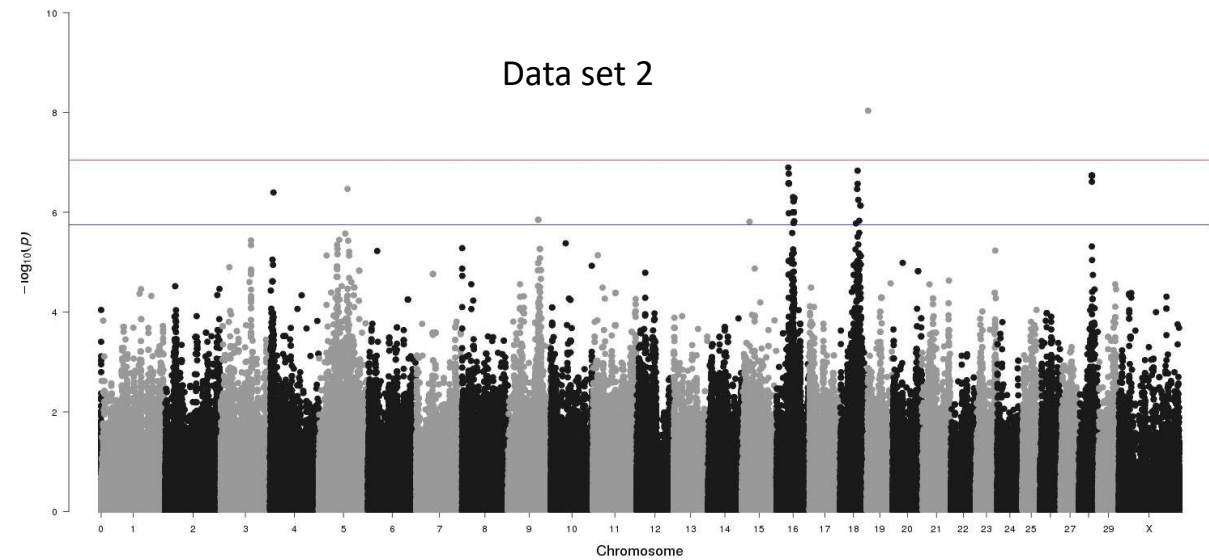
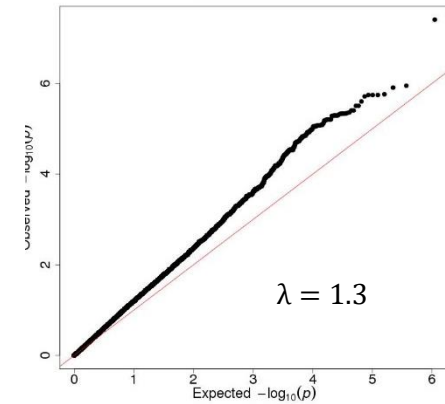
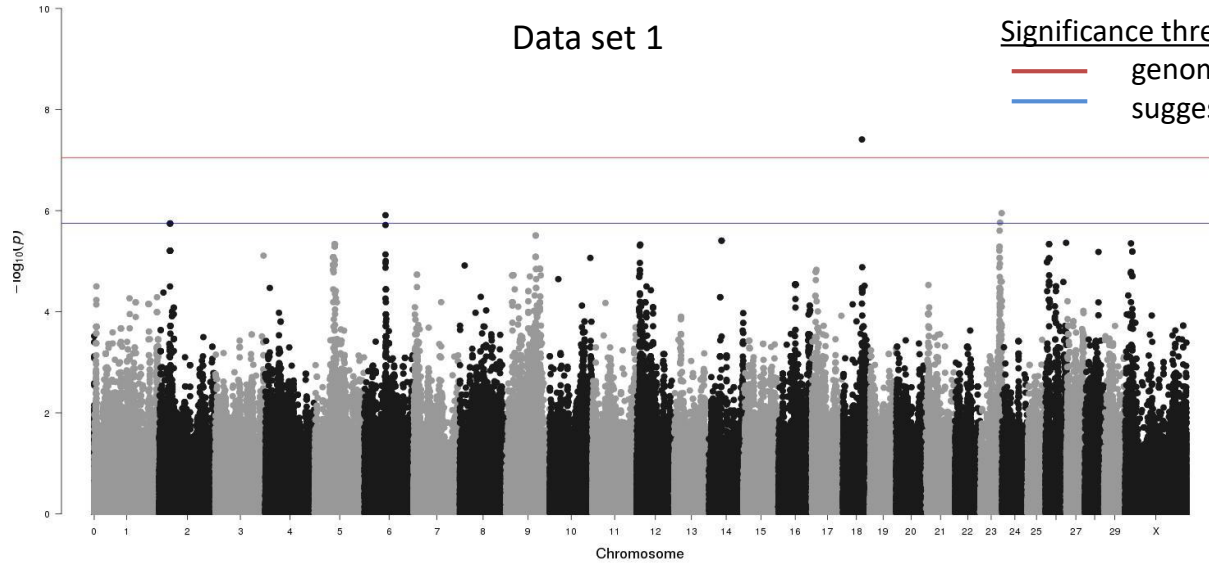


GWAS - Results

Introduction

Material & Methods

Results & Discussion





Conclusion & Outlook

Introduction

Material & Methods

Results & Discussion

- The expression of scurs seems to be dependent on sex, at least in double heterozygous animals
- Scurs do not seem to be expressed in homozygous polled animals
- Suggestive significances for the *scurs* Locus on BTA 2, 6, 16, 18, 23, 28
 - genetic heterogeneity ?
 - more than one *scurs* locus ?

Outlook:

- Functional annotation of the significant SNPs (in progress)



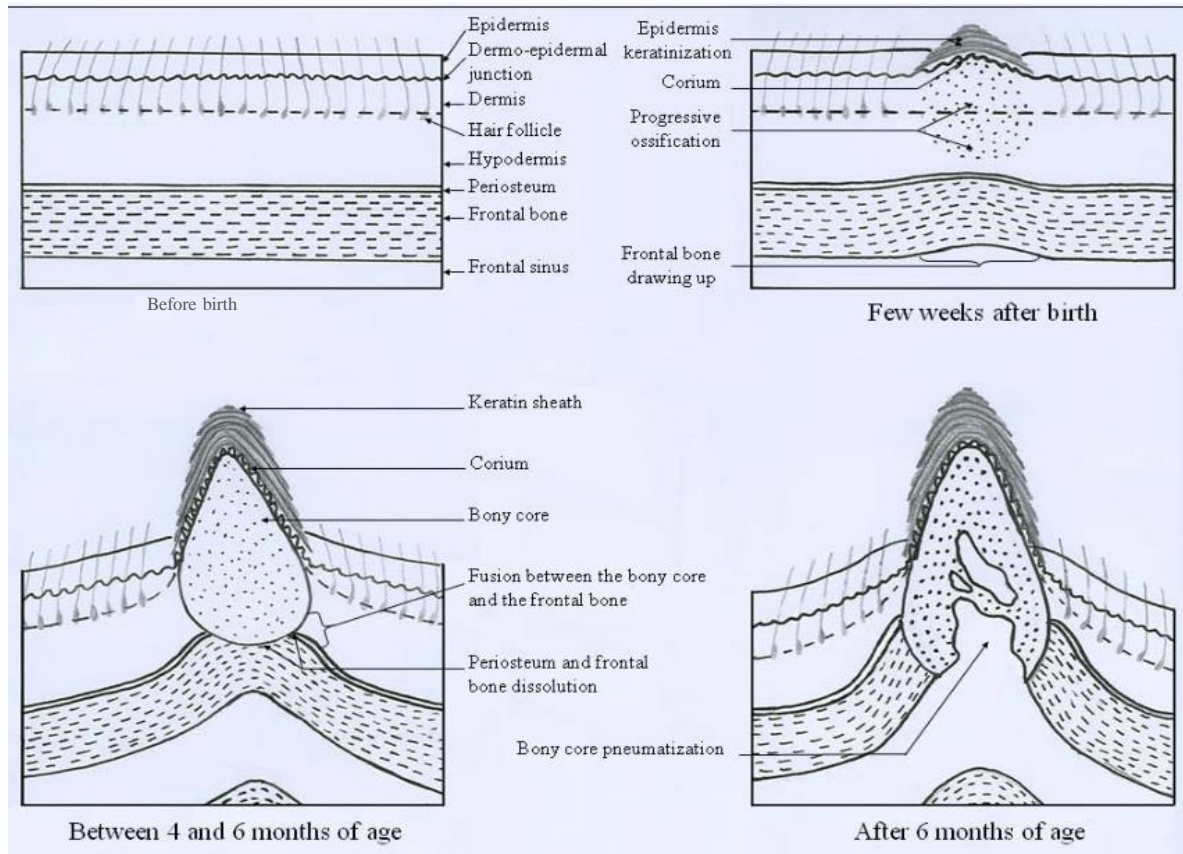
Thank you for your attention!



H. WILHELM SCHAUMANN STIFTUNG

Thanks to the H. Wilhelm Schaumann Stiftung for funding.

Appendix



Horn Ontology

1. Epidermal horn predisposition
2. Separate ossification centre
3. Development of a bony core and keratinisation of the epidermis
4. Fusion to the frontal bone
5. Pneumatisation