UNIVERSITÄT KASSEL | ÖKOLOGISCHE AGRAR WISSENSCHAFTEN

Genome-wide diversity and chromosomal inbreeding in German White-headed Mutton Sheep

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Introduction

History & Importance of German White-headed Mutton (GWM)

- GWM was developed in the middle of 19th century

- Crosses between
  - Local marsh sheep & English breeds
    e.g. Cotswold & Leicester Longwools

- Introduction of Texel (TXL) & Berrichone Du Cher (BDC)

- Mainly found in Schleswig-Holstein & Lower Saxony
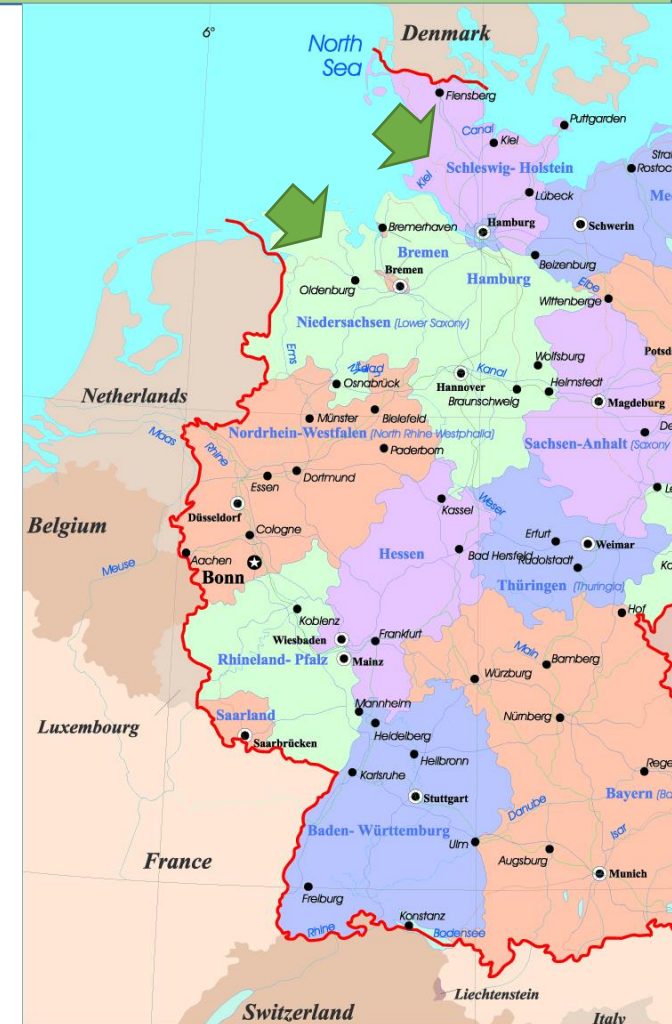
Introduction

History & Importance of German White-headed Mutton (GWM)

- **Some Characteristics**
  - Pooled and uniformly white
  - Mature weight, female: 80 - 90 kg
    - male: > 100 kg
  - Fleece weight > 4 kg

- **Monitoring population (red list)**
  - 2018 herdbook numbers
    - 1,704 ewes
    - 106 rams

- **Most flock owners are hobby breeders**

Introduction

Aims of study

- To quantify genome-wide diversity in GWM from the application of different runs of homozygosity (ROH) measures.

- To investigate historical relatedness of GWM to other breeds.

Materials and methods

Data1

- 48 GWM individuals genotyped with Illumina Ovinesnp50 BeadChip

Data before & after quality filtering

	Before	After
Number of animals	48	46
Number of SNPs	52,413	40,753

Autosomal SNPs
Individual call rate: > 90%
SNP call rate: > 90%
MAF: > 5%
HWE: $P \leq 0.0001$

- 31/46 individuals had pedigree information

- Born between 2009 and 2015

Materials and methods

Data2 (plus other breeds)

Breeds	Acronym	Number
Berrichone du Cher	**BDC**	**19**
German Texel	**GTX**	**46**
Border Leicester	**BRL**	**48**
New Zealand Romney	ROM	24
Spanish Merino (Andalusia)	SMA	7
Spanish Merino (Estremadura)	SME	13
Suffolk	SUF	19
East Friesian White	EFW	9
Mouton Charollais	CHL	24
		209

Source: WIDDE, 2019

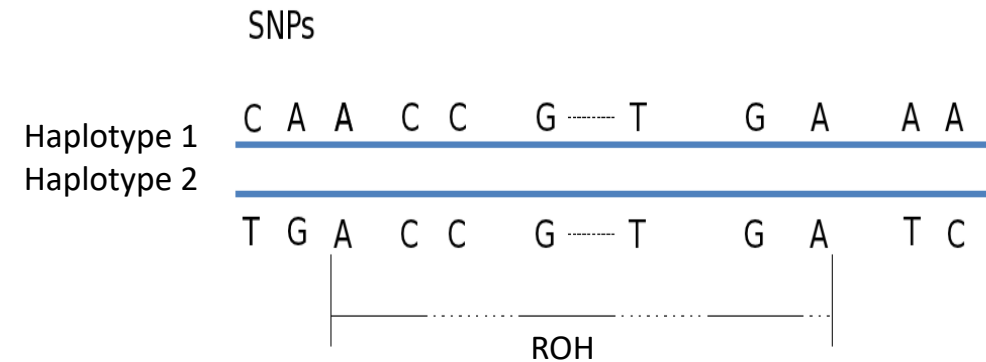| --indep 50 5 2 |
|---|
| 255 samples 16,852 SNPs |

Materials and methods

Computations

- ROH definition

    - 50 SNP-window
    - 1 het. SNP
    - 2 missing genotypes
    - Density of 1 SNP per 100 kb
    - Maximum gap of 1000 kb

Individual is homozygous across all sites

SNPs

Haplotype 1   C A A   C C   G ----- T   G A   A A
Haplotype 2   T G A   C C   G ----- T   G A   T C

ROH

- ROH were detected in 2 ways:

    - For entire autosome to calculate an individual's total inbreeding coefficient
        - F_{ROH_L}
        - F_{ROH_N}
    - For each chromosome to calculate an individual's chromosomal inbreeding
        - F_{ROH_KK}
        - F_{ROH_KA}

Materials and methods

Computations

Total F$_{ROH}$ was calculated in 2 ways:

(1) Based on the **L**ength of ROH (Mb) as:

$$F_{ROH\_L} = \frac{\sum L_{ROH}}{L_{AUTO}}$$

Following McQuillan *et al.*, 2008

(2) Based of **N**umber of SNPs in ROH as:

$$F_{ROH\_N} = \frac{ROH\_SNP}{N\_SNP}$$

Chromosomal F$_{ROH}$ was based on the ROH lenght measure and calculated either

(3) Relative to specific **c**hromosome length as:

$$F_{ROH\_KK} = \frac{\sum L_{ROH\_K}}{L_{AUTO\_K}}$$

(4) Relative to entire **a**utosomal length

$$F_{ROH\_KA} = \frac{\sum L_{ROH\_K}}{L_{AUTO}}$$

Materials and methods

Other diversity parameters

- **Genetic diversity indices**

  - Pedigree-based inbreeding (F_{PED}) using ENDOG (Gutiérrez JP, Goyache F, 2005)

  - Observed (H_o) & Expected (H_e) Heterozygosity using PLINK (Purcell S *et al.*, 2007)

  - LD-based Effective Population Size (Ne_{LD}) using *SNeP* (Barbato *et al.*, 2015)

- **Population structure**

  - Multidimensional scaling (MDS) analysis

  - Admixture analysis (Alexander et al, 2009)

Results and discussion

Inbreeding approaches

The distribution of inbreeding coefficients from different computational approaches in GWM (n = 31)

| Parameter	Mean (min - max) %
F_{ROH_L}	3.894 (1.070 – 13.243)
F_{ROH_N}	3.946 (1.121 – 13.305)
$\sum F_{ROH\_KA}$	3.894 (1.070 – 13.243)
$\emptyset F_{ROH\_KK}$	3.764 (0.441 – 14.532)
F_{PED}	2.936 (0.459 – 8.956)
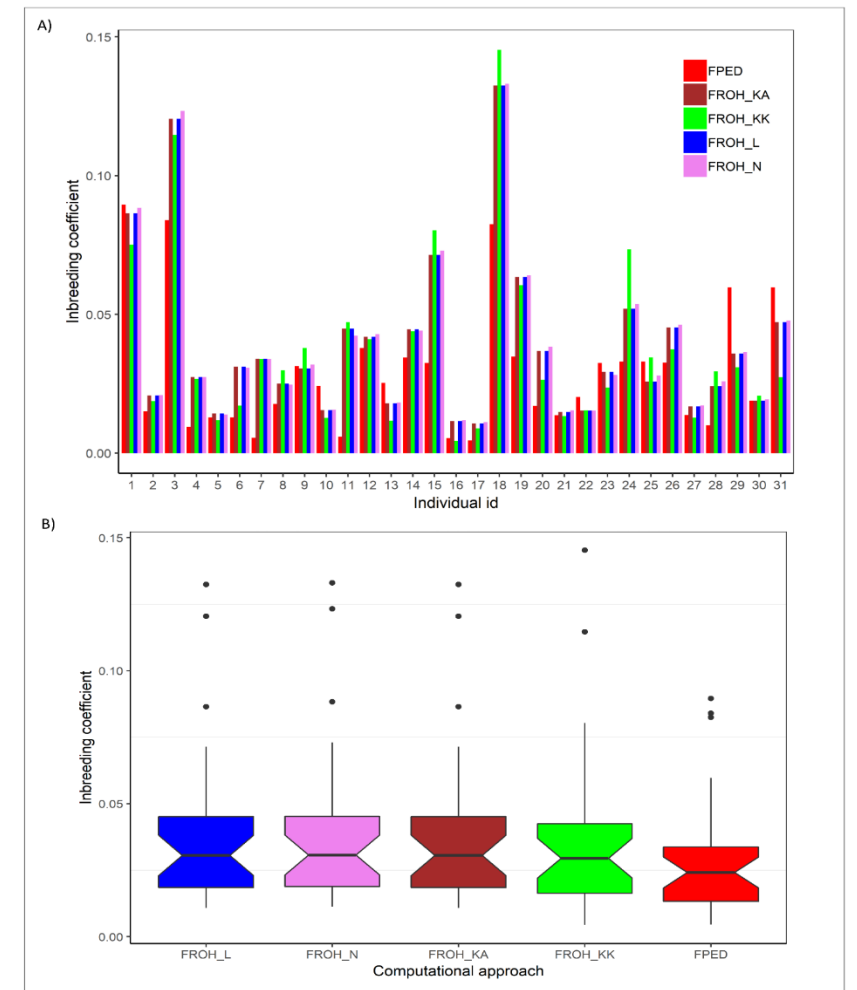
Results and discussion

Inbreeding approaches

- Correlations between inbreeding estimates of different approaches

	F_{ROH_L}	F_{ROH_N}	F_{ROH_KA}	F_{ROH_KK}
F_{PED}	0.8179	0.8235	0.8179	0.7428
F_{ROH_L}	-	0.9995	1.0000	0.9674
F_{ROH_N}		-	0.99945	0.9678
F_{ROH_KA}			-	0.9674
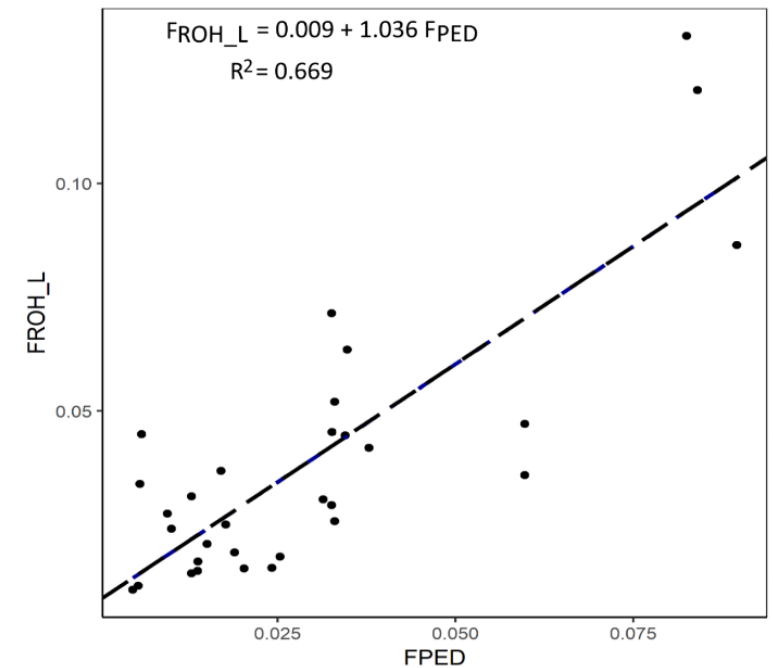
Regression of F_{ROH_L} on F_{PED}

# Results and discussion

Heterozygosity & Effective size

- High level of genetic diversity consistent with relatively low F level in GWM

- Ne estimate at the boarder of critical threshold

Parameter	Value
H_e	0.38
H_o	0.39
$Ne_{LD(5Gen)}$	53

Results and discussion

Population structure (MDS)

- Close relationship between GWM and BDC

- Some degree of closeness with GTX

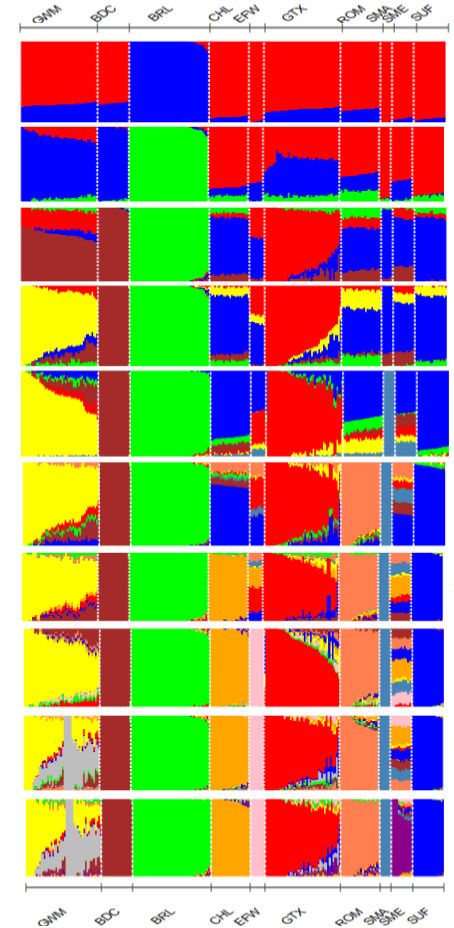- Highlight the improvement of lamb quality
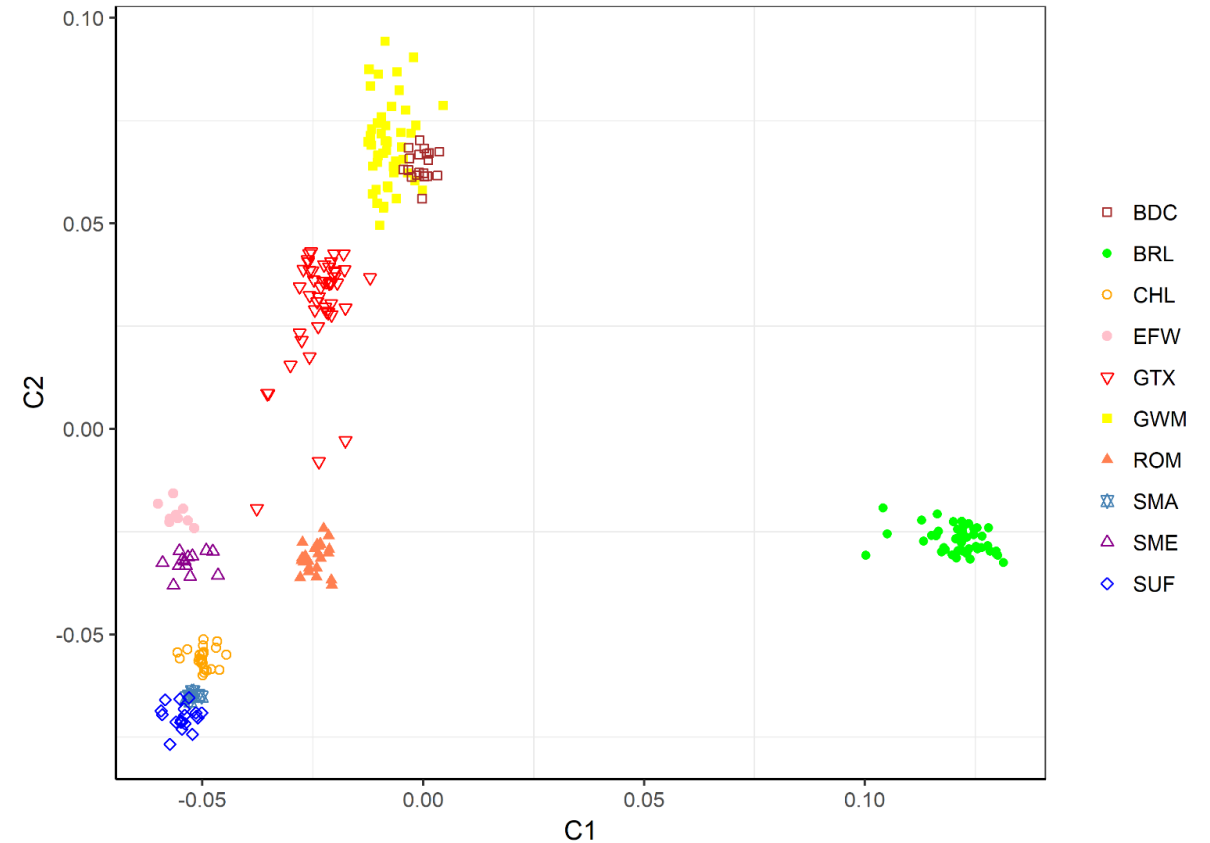
BDC: Berrichone du Cher, **BRL:** Border Leicester, **CHL:** Mouton Charollais, **EFW**: East Friesian White, **GTX**: German Texel, **GWM:** German White-headed Mutton, **ROM:** New Zealand Romney, **SMA**: Spanish Merino (Andalusia), **SME**: Spanish Merino (Estremadura), **SUF:** Suffolk

Results and discussion

Population structure (Admixture)
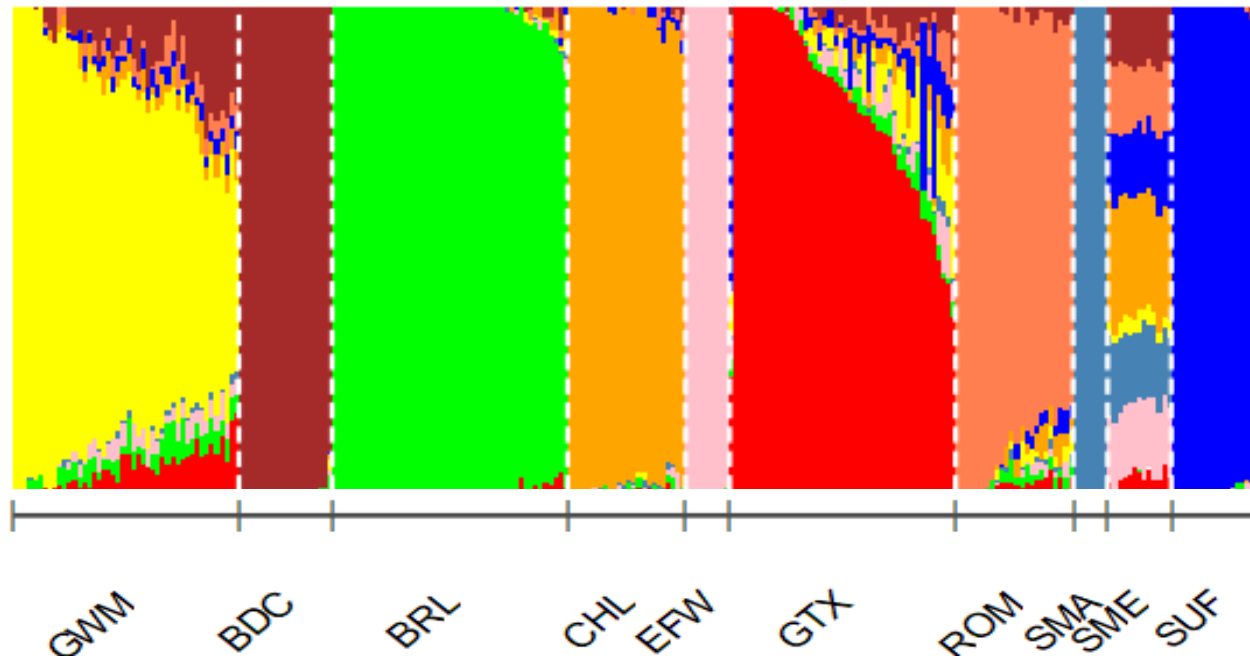
Admixture results representing K values from 2 to 11, with K = 9 having the lowest cross validation error

GWM: German White-headed Mutton, **BDC**: Berrichone du Cher, **BRL:** Border Leicester, **CHL:** Mouton Charollais,
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Conclusion

- Use of the "*number of SNPs in ROH*" and "*length of ROH*" in the computation of inbreeding coefficient produce comparable results.

- Total inbreeding can be elegantly partitioned into values for specific chromosomes following appropriate measures.

- GWM is more closely related to BDC and GTX and still harbours genetics of English descent.

- Genetic diversity is high in GMW, however, the Ne estimate suggests the implementation of a close monitoring system.

Thank you for your attention