

# Runs of homozygosity and levels of inbreeding in cattle breeds

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# In this study we used:

- Brown Swiss cattle, 463 bulls
- Norwegian red cattle, 498 bulls
- Simmental cattle, 502 bulls
- Tyrol Grey, 215 bulls

# ...on which we tried to

- predict levels of inbreeding
- compare levels of autozygosity
- explain their inbreeding history

# Genomic inbreeding coefficient

**Level of  
homozygosity**

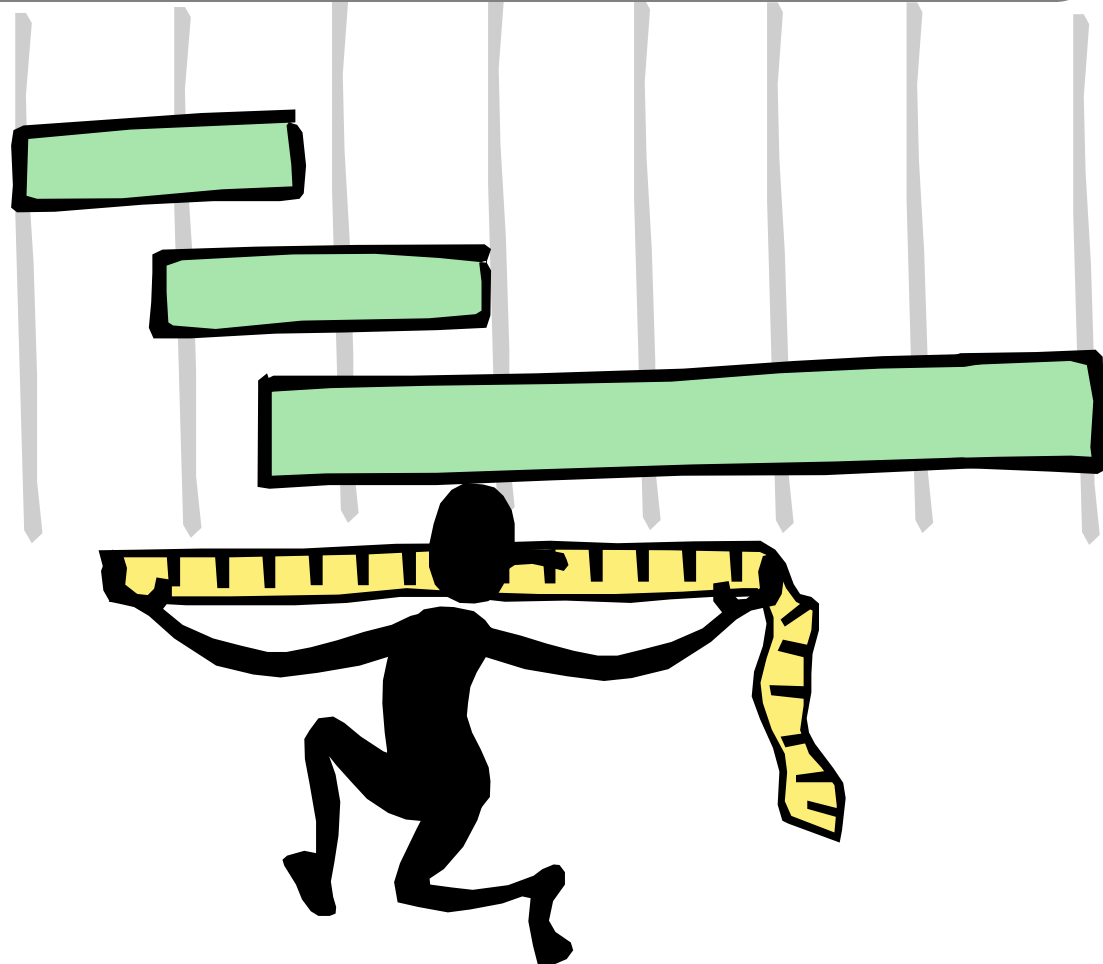
$$F_{\text{hom}} : \frac{H_e - H_o}{H_e}$$

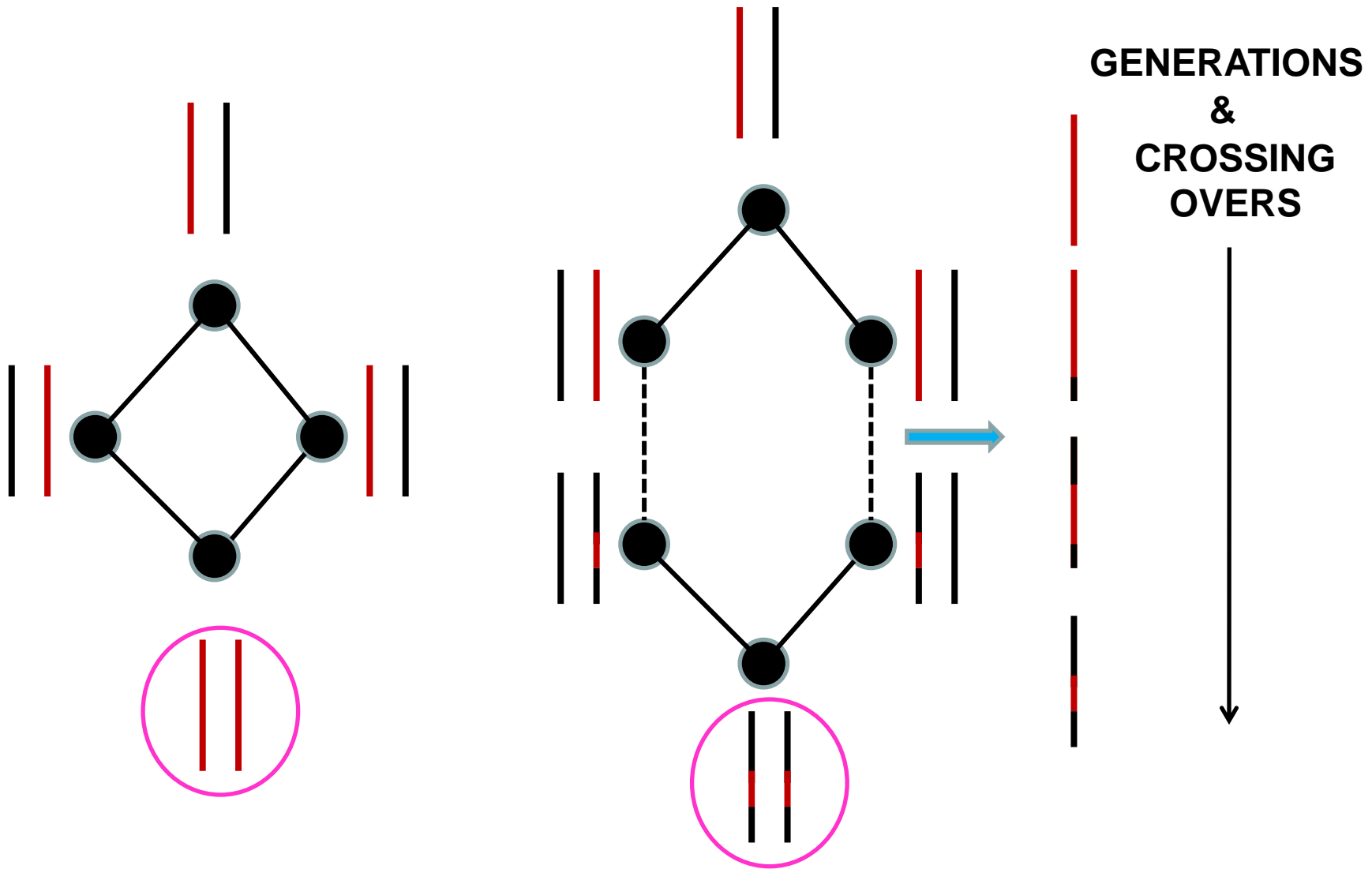
**Runs of  
homozygosity**

$$F_{\text{ROH}_k} : \frac{\sum_k \text{length}(\text{ROH}_k)}{L}$$

# Runs of homozygosity (ROH)

Observable  
inbreeding?





# Illumina Bovine SNP50™ Beadchip

- We exclude** :
- SNP with  $gc\_score < 0.2$
  - SNP with  $MAF < 0.01$
  - SNP with  $> 0.05$  of missing genotypes
  - animals  $> 0.05$  of missing genotypes

**Pedigree data  
(6.46 – 9.02 CGE)**



$F_{pedT}$  &  $F_{ped5}$

**36273 SNP**



$F_{hom}$



**Five ROH lengths ( $k$ ):  
>1Mb, >2Mb, >4Mb,  
>8Mb, >16Mb**

**ROH lengths ( $k$ ):**



$F_{ROH1}$ ,  $F_{ROH2}$ ,  
 $F_{ROH4}$ ,  $F_{ROH8}$ ,  
 $F_{ROH16}$



# What are inbreeding levels?

$F_{pedT}$	$F_{ped5}$		$F_{ROH8}$	$F_{ROH16}$
4.1 (2.2)	2.1 (1.8)		6.6 (3.1)	3.3 (2.4)
1.4 (1.3)	0.9 (1.2)	≈	1.6 (1.6)	0.8 (1.2)
2.1 (1.4)	1.0 (1.2)		3.3 (2.7)	1.5 (1.6)
2.4 (2.2)	1.6 (2.0)		3.0 (2.6)	1.6 (2.0)

**Brown Swiss**

**Simmental**

**Norwegian Red**

**Tyrol Grey**

**Mean(SD) %**

# ...but ancient inbreeding?

$F_{ROH1}$

$F_{ROH2}$

$F_{ROH4}$

14.2(3.6)

11.5 (3.6)

9.3(3.5)

8.5 (2.0)

5.2 (1.9)

3.0 (1.7)

9.6 (2.5)

7.2 (2.5)

5.3 (2.4)

7.8 (2.7)

5.3 (2.7)

4.1 (2.7)

**Brown Swiss**

**Simmental**

**Norwegian Red**

**Tyrol Grey**

**Mean(SD) %**

# Correlations?

	$F_{pedT}$	$F_{ped5}$
$F_{ped5}$	0.94 – 0.98	–
$F_{ROH1}$	0.60 – 0.71	0.49 – 0.70
$F_{ROH2}$	0.60 – 0.71	0.49 – 0.70
$F_{ROH4}$	0.62 – 0.72	0.52 – 0.72
$F_{ROH8}$	0.61 – 0.71	0.53 – 0.71
$F_{ROH16}$	0.51 – 0.69	0.50 – 0.71

Brown Swiss

Norwegian Red

Simmental

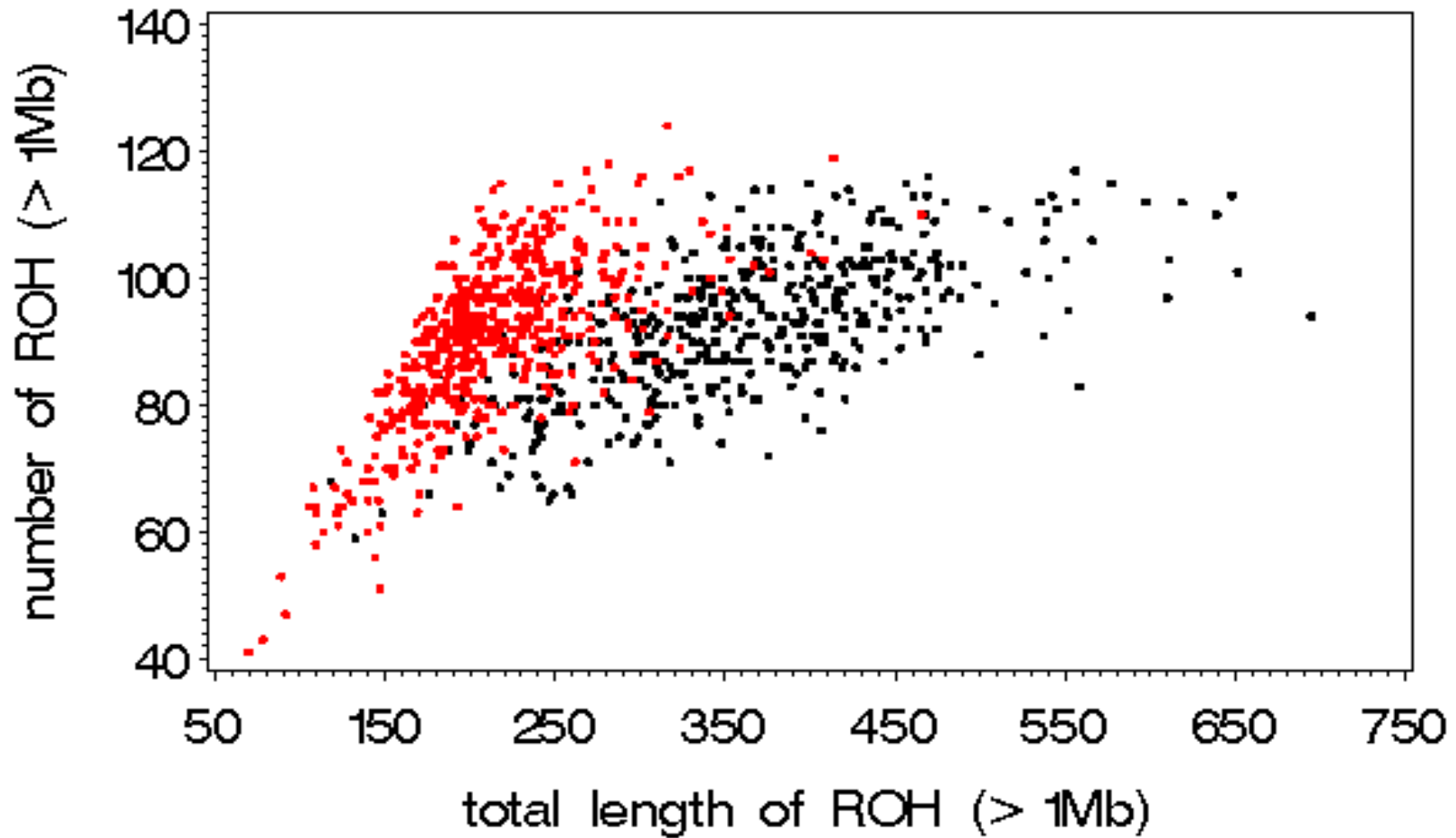
Tyrol Grey

# ...level of homozygosity?

		$F_{\text{hom}}$	
		$F_{\text{pedT}}$	$F_{\text{ped5}}$
<b>Inbreeding</b>	$F_{\text{hom}}$ Mean(SD)%	<b>0.56</b> – <b>0.63</b>	<b>0.49</b> – <b>0.64</b>
	Brown Swiss	<b>13.8</b> (3.6)	<b>0.84</b> – <b>0.93</b>
	Simmental	<b>6.6</b> (2.7)	<b>0.80</b> – <b>0.92</b>
	Norwegian Red	<b>6.6</b> (2.6)	<b>0.76</b> – <b>0.91</b>
	Tyrol Grey	<b>7.2</b> (3.6)	<b>0.69</b> – <b>0.87</b>
		$F_{\text{ROH16}}$	<b>0.61</b> – <b>0.81</b>

**Correlations**

# ROH structure



**Brown Swiss**

**Simmental**

# Conclusions about ROH & $F_{ROH}$

- Useful especially if pedigree is missing or incorrect
- Demonstrated as optimal way for identification of ancient inbreeding in humans (Keller et.al. 2011)
- ROH give information about level of inbreeding and its age
- Observational approach: most likely provide more accurate information than probabilistic approaches

# Acknowledgments

We are very grateful to owners of the genotype data for providing them :

**Geno** – breeding organization for Norwegian Red

**AGOEF** – Association of Austrian Fleckvieh Breeders

**ArGe Braunvieh**

**ArGe Grauvieh**

# Thank you for your attention



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