

Translocations associated with gonadal hypoplasia and colour-sidedness are common in northern Swedish cattle breeds

Anna M Johansson, Swedish University of Agricultural Sciences

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Gonadal hypoplasia in Swedish cattle

- Problem with fertility known for about 100 years in Fjäll cattle (Swedish mountain cattle). The frequency has been reduced but it is still present despite breeding efforts för many years
- Reduced size of testes/ovaries, often only on one side (mostly the left side)
- Some affected animals are completely sterile whereas others only have reduced fertility
- Recessive trait with reduced penetrance
- Association with a lot of white in the coat, known for many years

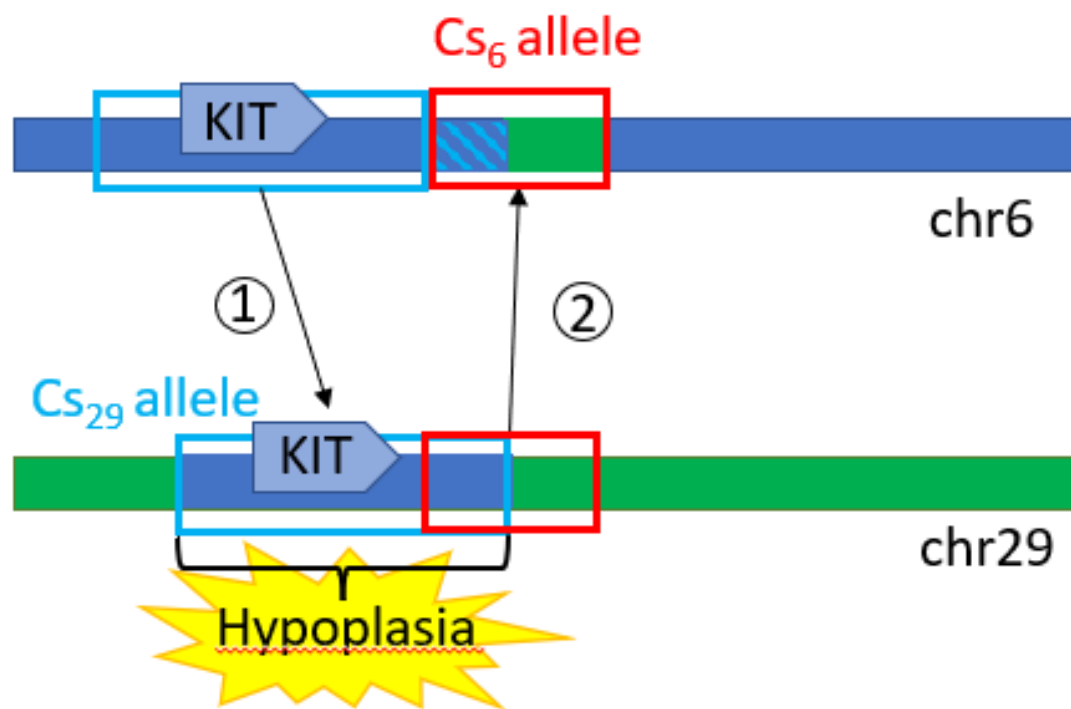


Translocations

- Two translocations involving chromosomes 6 and 29
- The translocation called Cs29 was shown to be causative in northern finncattle and Fjäll cattle by Venhoranta et al. (2013)
- Cs29 and Cs6 both cause colour sidedness but gonadal hypoplasia only from Cs29
- The KIT gene is in the region affected by these translocations



Chromosomes 6 och 29



Colour sidedness

- They have colour on the ears and the nose and some colour on the sides of the body. White along the back and on the belly
- Heterozygotes for colour sidedness have larger areas of colour on the sides than homozygotes do
- The cow on the photo is homozygous for Cs_{29}



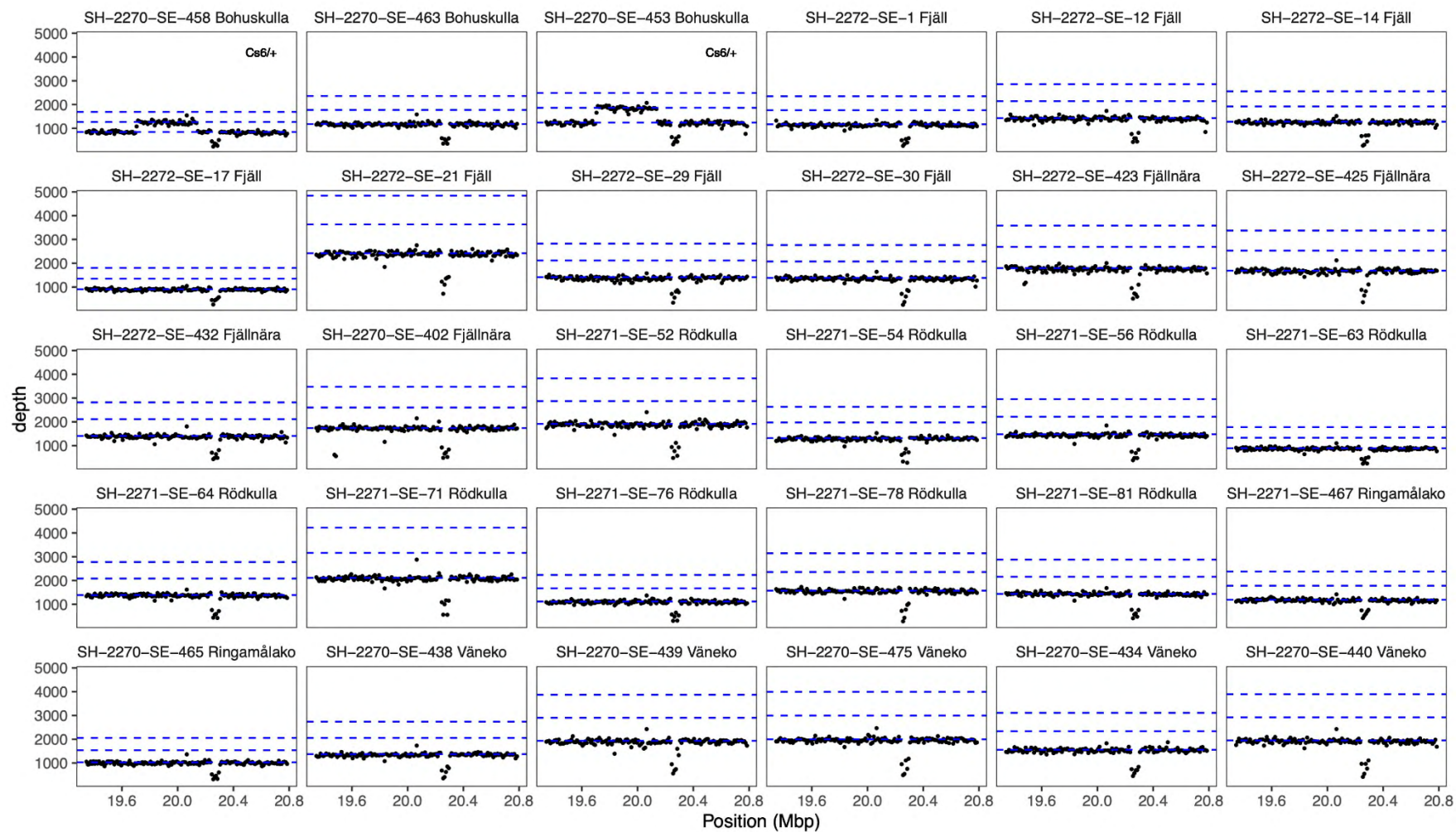
Swedish mountain cattle (Fjäll cattle)

Our studies

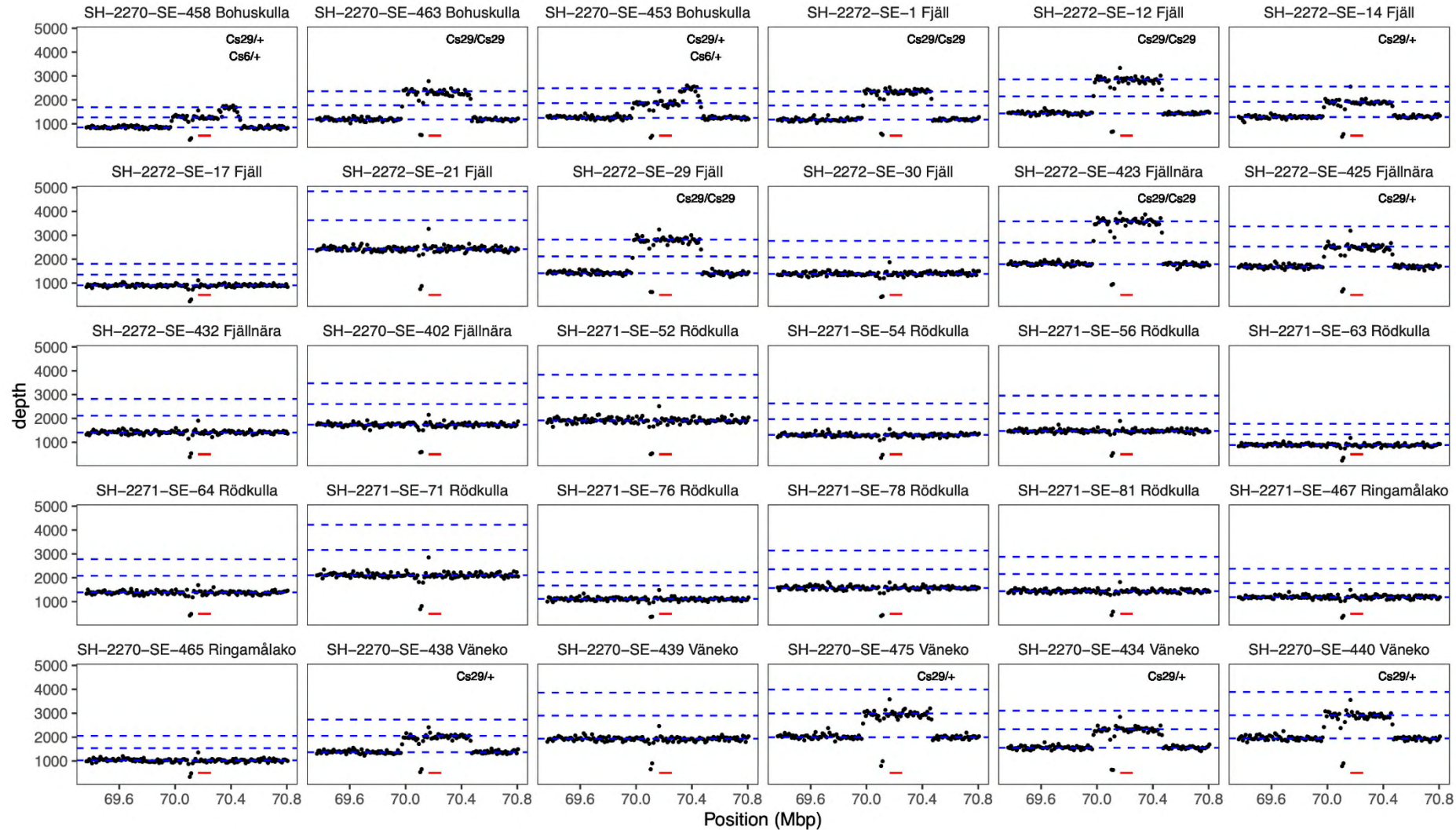
- Pilot study as master thesis by Julia Hinken during autumn 2022 to investigate the frequency of the translocations in Swedish breeds. She used WGS data we already had (from Harish et al. 2024) from 30 Swedish cattle. She also and did PCR genotyping of translocation breakpoints in additional samples.
- Some further WGS analyses by Martin Johnsson
- Funding from Stiftelsen Seydlitz MP bolagen to Anna M Johansson and Martin Johnsson to continue the research. We have collected and genotyped more samples and have also started to collect phenotypes.



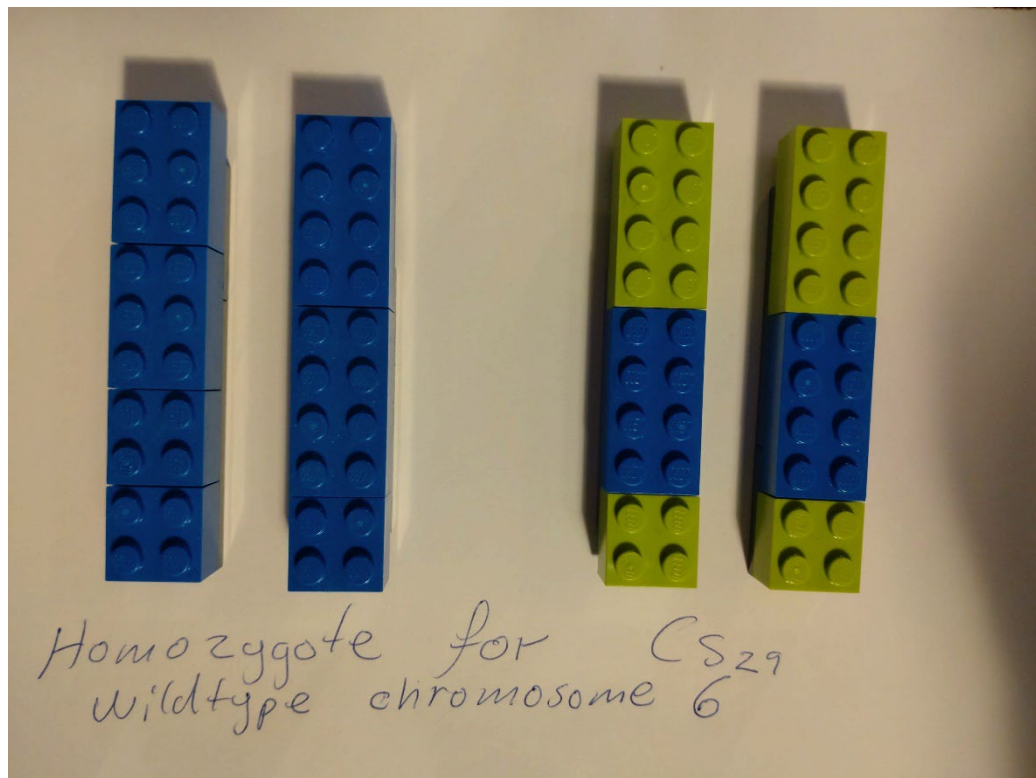
Chromosome 29



Chromosome 6

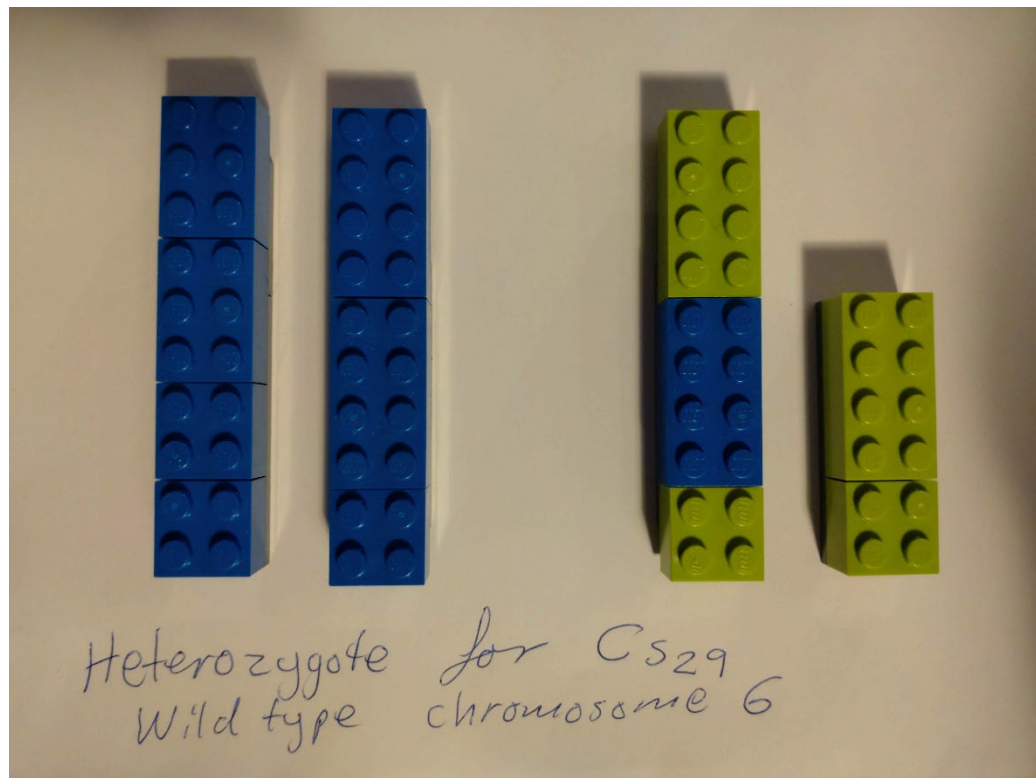


Chromosomes for genotype Cs29/Cs29



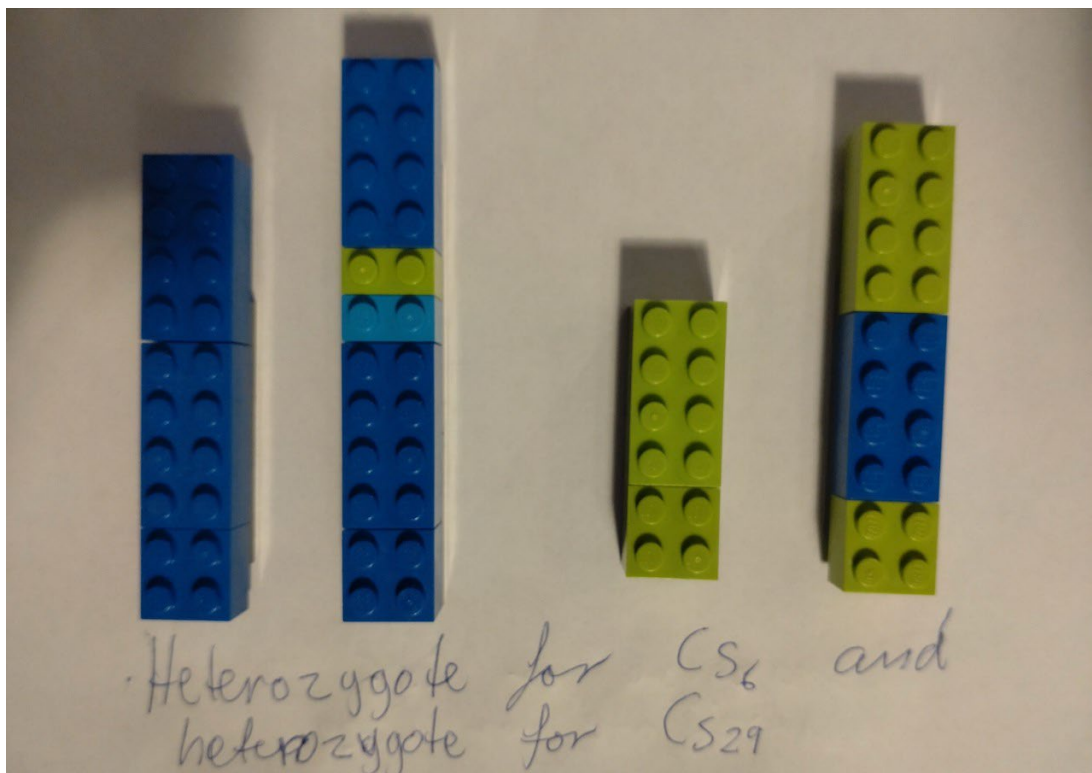
- Four copies of a part of chromosome 6
- Two copies of the whole chromosome 29

Chromosomes for genotype Cs29/+



- Three copies of a region of chromosome 6
- Two copies of the whole chromosome 29

Chromosomes for genotype Cs29/+, Cs6/+



- Four copies of a region on chromosome 6
- Three copies of a region on chromosome 29

Results from PCR genotyping

- We detected the Cs_{29} allele in all five Swedish breeds where colour-sidedness occur (Fjäll cattle, Fjällnära cattle, Bohus polled, Swedish polled, Väne cattle).
- Some bulls that have quite many offspring are homozygous for Cs_{29}
- We also detected Cs_6 in four breeds (Fjäll cattle, Fjällnära cattle, Bohus polled, Swedish polled)



Frequencies of Cs29 based on PCR genotyping

Population	Frequency of Cs29
Bohus Polled (n = 8), sampled in 1990s	0.56
Fjäll (n = 22) mostly sampled in 1990s	0.57
Fjällnära (n = 15), sampled in 1990s	0.27
Swedish Polled (n = 8), born 1994-2004	0.22
Väne (n = 5), sampled in 1990s	0.40
Fjäll (n = 70) sampled in 2023-2024	0.45
Fjällnära (n = 12), sampled 2023-2024	0.17

Future studies

- We are currently collecting phenotypes and samples and doing genotyping using PCR over the translocation breakpoints. This will allow us to investigate the association between gonadal hypoplasia and the translocations and estimate penetrance.
- In the future, long range sequencing of samples with the translocations would be interesting



Thanks to

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- Cattle owners that contributed samples to the study
- Funding from: Stiftelsen Seydlitz MP bolagen

