

# Genetic studies in wool colour in Swedish sheep breeds

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# Genetics of black wool in sheep

- There are two genes that is known to be involved in black wool colour in sheep, *ASIP* (A lociús) and *MC1R* (*E locus*)
- In *ASIP* there is two different mutations causing black wool colour, one deletion of five basepairs  $D_5$  (five "DNA letters" is missing) in the beginning of the gene and in the end of the gene a T have been replaced by an A (g.5172T>A ).
- To get black wool the sheep must have inherited a mutation in *ASIP* from both the father and the mother, recessive inheritance.
- In *MC1R*, there are also two different mutations that gives black wool. But here it is enough to inherit a mutation from one parent to get black wool, dominant inheritance.





# Studies of the genes *ASIP* and *MC1R* in Swedish sheep

- *ASIP* and *MC1R* sequenced in Swedish finewool sheep, Gotland sheep, Gute sheep, Helsinge sheep, Klövsjö sheep, Roslag sheep, Värmland sheep
- Rochus CM, Westberg Sunesson K, Jonas E, Mikko S, Johansson AM. 2019. Mutations in *ASIP* and *MC1R*: Dominant black and recessive black alleles segregate in native Swedish sheep populations. *Animal Genetics* 50 (6): 712-717 <https://doi.org/10.1111/age.12837>



## Black wool in Gute sheep

- Some black Gute sheep are homozygous for  $D_5$  and some are homozygous for A
- Some black Gute sheep have probably inherited  $D_5$  from one parent and A from the other parent
- None of the mutations in *MC1R* is present



Photo: Ingrid Gosselman

## Black wool in Roslag sheep

- All black Roslag sheep we have investigated has the genotype  $D_5D_5$
- None has the allele *A* in the end of the gene
- None of the mutations in *MC1R* is present



Photo: Stephan Noll

## Black wool in Klövvsjö sheep



Photo: Ingrid Gosselman

- Both the alleles for black colour in ASIP are present in the breed
- This sheep on the photo is homozygous AA
- None of the mutations in *MC1R* is present



## Black wool in Helsinge sheep and Värmland sheep

- Both the alleles for black colour in *ASIP* are present in the breeds Värmland sheep and Helsinge sheep.
- The genotypes for these alleles does however dont show a clear relationship with the phenottpe in these two breeds. There is for example one dark greysheep that is AA and one black that is heterozygous for D<sub>5</sub> but does not have any A allele. Maybe the colour phenotyping have not been accurate or maybe there is some other gene involved
- None of the mutations in *MC1R* is present



## Black wool in Swedish finewool sheep

- Both mutations in *MC1R* is present
- Both mutations in *ASIP* is present
- Some sheep are black due to *ASIP* and some due to *MC1R*



Photo: Anna M Johansson

## Additional sequencing of *MC1R* later

- *MC1R* was sequenced in black sheep of the breeds Rya sheep, Gestrike sheep, Svärdsjö sheep
- The mutations in *MC1R* was not present in any of these breeds, but the number of sheep was low from each breed

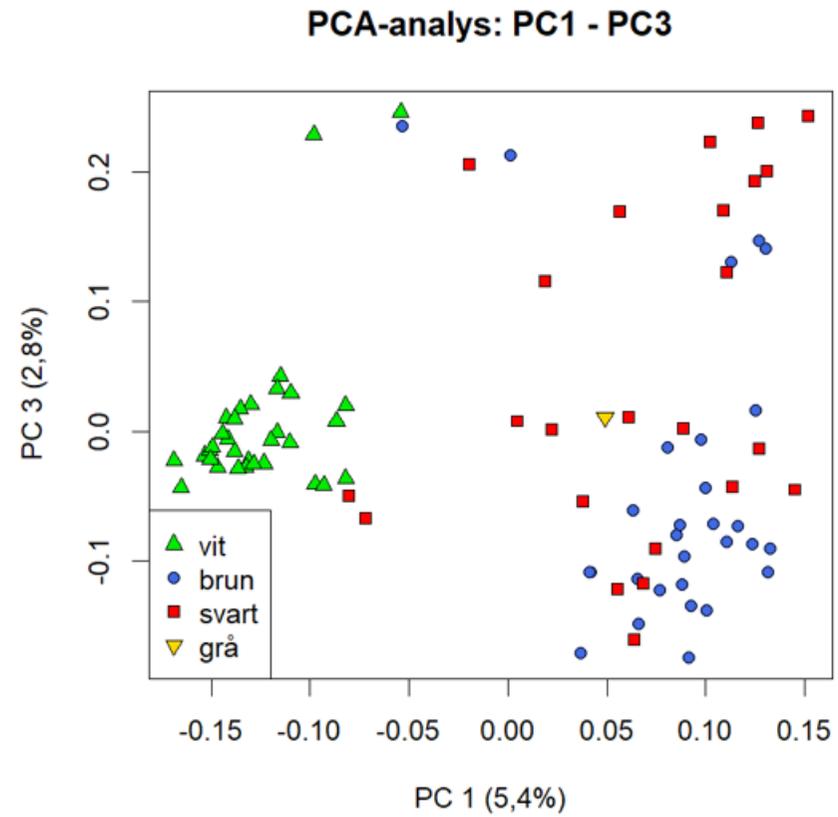
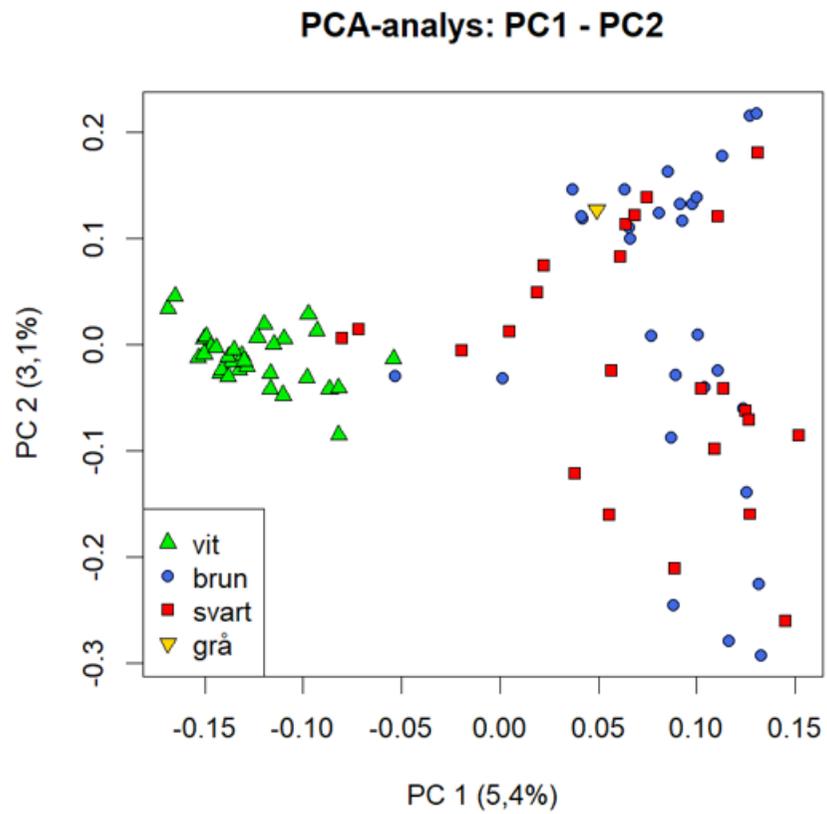


## Master thesis about Rya sheep

- 90 samples genotyped, 87 samples and 29470 SNPs passed quality control. 32 white, 29 brown, 24 black, 1 grey, 1 unknown colour.
- A master thesis by Amanda Hellqvist during spring 2025, supervised by Anna M Johansson
- Good level of heterozygosity compared to other breeds. White Rya sheep had slightly lower heterozygosity.
- Relatively low level of inbreeding on average (0.028), but white sheep had a little bit higher on average (0.045).

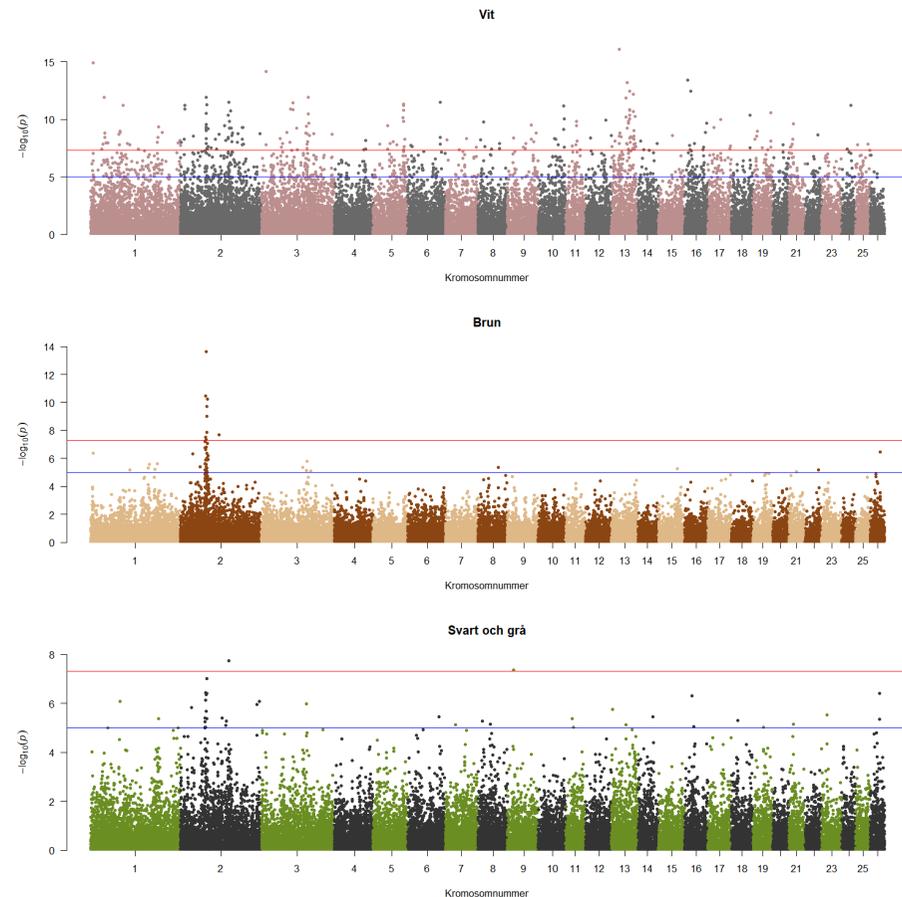


# PCA analysis Rya sheep



# Association to colour

- Most significant SNP for brown colour is close to the gene *TYRP1* on chromosome 2
- *TYRP1* is known for influencing brown colour in other studies
- Some other SNPs close to genes influencing other wool traits





# Ancient DNA in sheep in collaboration with researchers at Uppsala University

- Five high quality ancient sheep genomes (0.54X-16.8X)
- From two islands in the Baltic sea, Gotland and Åland,
- Dating from ~4100 cal BP (Late Neolithic - Early Bronze age) to ~1600 CE (late Medieval/early Vasa periods).
- We compared these genomes to a panel of present day breeds





## Ancient DNA in sheep

- The results show that ancient sheep from Åland and Gotland are genetically similar to present day North European short-tailed sheep.
- The ancient samples were more genetically diverse than present day breeds.
- Larsson MNA, Morell Miranda P, Pan L, Vural KB, Kaptan D, Rodrigues Soares AE, Kivikero H, Kantanen J, Somel M, Özer F, Johansson AM, Storå J, Günter T. Ancient Sheep Genomes reveal four Millennia of North European Short-Tailed Sheep in the Baltic Sea region. *Genome Biology and Evolution*, Volume 16, Issue 6, June 2024, evae114,

