



# Genetic and statistical analysis of the coat color roan in Icelandic horses

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69<sup>th</sup> Annual Meeting of the European Federation of Animal Science  
Dubrovnik, Croatia, 27<sup>th</sup> to 31<sup>st</sup> August 2018  
Session 71, Abstract 29376 [kvoss@tierzucht.uni-kiel.de](mailto:kvoss@tierzucht.uni-kiel.de)



Islandpferde vom Holttdorfal





# What is Roan?

## Phenotype

- Mixture of white and colored hair
- Seasonal change of color
- Rare in most and not existing in some breeds





# What is Roan?

## Genetic Background

- Dominant mode of inheritance
- Association to *KIT* and 4 more genes (Linkage group II) on chromosome 3
- Causative mutation is unknown
- Marker test available
- Said to be embryonically lethal under homozygous conditions (Hintz and van Vleck, 1979)



# Aim of this project

- Identify the causative mutation for roan in Icelandic horses
- Test the hypothesis whether roan is homozygous lethal



# Data analysis

- *Worldfengur* = database originated from Icelandic farmers' association *Fengur* : >460,000 Icelandic horses

Take a look at:

- Total number of roan registered horses
- Distribution among different coat colors
- Matings of roan x not-roan
- Matings of roan x roan





# Genetic analysis

## Pre-Study

- Sanger-sequencing of 21 coding exons of the *KIT* gene
- 30 cases and 23 controls
- Association analysis

## Whole genome sequencing

- NGS Illumina HiSeq4000 (ø 25 X)
- Two Icelandic horses: Heterozygous (Rnrn) and homozygous (RnRn; Marker tested)
- Focus on variants at ECA3
  - Analysis of variants within Linkage group II
    - Further filtering

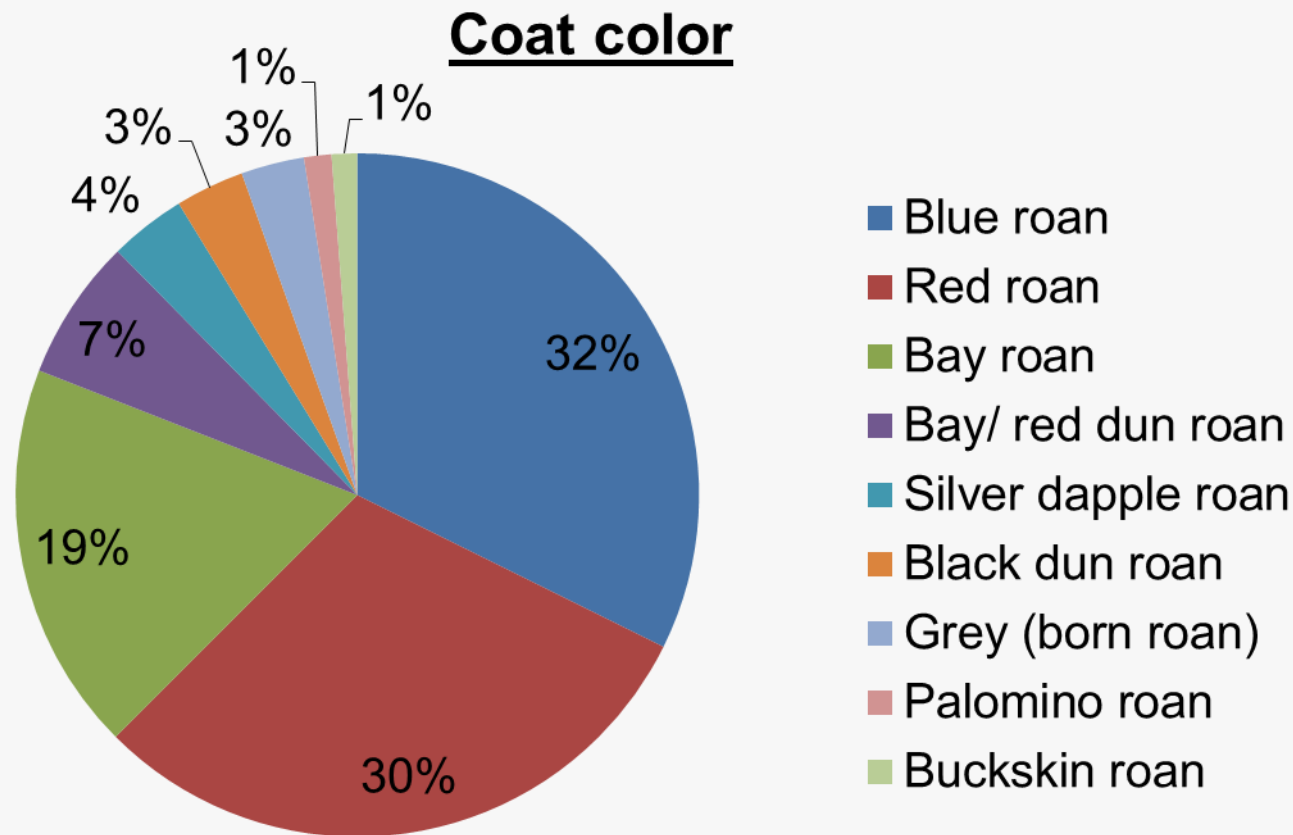


Islandperde Drei-Gleichen



# Descriptive statistics

- 2,240 roan horses → 0.46 % of all registered Icelandic horses





# Descriptive statistics

## Offspring of roan horses

Parent	Total (n)	Roan (Rn)	Percentage (%)
Roan sire	2055	911	<b>44.33</b>
Roan mare	1300	624	<b>48.00</b>
	3355	1535	<b>45.75</b>

roan : not-roan 1:1

## Offspring of roan x roan - matings

Total No. of offspring	Roan	Not-roan	Percentage Rn (%)
59	49	10	<b>83.05</b>

roan : not-roan 3:1

→ Roan not homozygous lethal in Icelandic horses





# Genetic analysis

## Pre-Study

- Identified 16 polymorphisms, 5 located in exons
- Association analysis: significant to highly significant association ( $p < 0.001$ )
- None of the polymorphisms causative for roan phenotype

## Whole genome sequencing

Total number of variations on ECA3

Horse 1 (RnRn)	Horse 2 (Rnrn)
298,525	300,441

- Variations present in both horses: 207,726



# Genetic analysis

## Further filtering:

1. ~~both homozygous~~  
~~both heterozygous~~  
→ 61,857 variants left
2. Horse 1: homozygous → ~~heterozygous~~  
Horse 2: heterozygous → ~~homozygous~~  
→ 56,299 variants left
3. Limit to the Linkage Group II  
→ 3,895 variants left
4. Filtering with Ensembl Variation 93 database  
→ 330 variants left

# Summary and outlook

## Summary

- Roan is rare in Icelandic horses
- Combinations with all basic colors and modifiers
- Not homozygous lethal
- Association to *KIT* gene in Icelandic horses
- After filtering 330 putative variants left

## Outlook

- Further analyses of the variants on ECA3
- Enlarge the number of samples and sequencing of possible candidate variants





Thank you  
for your  
attention

## Acknowledgement

H. WILHELM SCHAUMANN STIFTUNG



**WorldFengur**



The Studbook of Origin for the Icelandic horse