The type of bottleneck matters: insights into the deleterious variation landscape of small managed populations

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IMAGE
Innovative Management of Animal Genetic Resources

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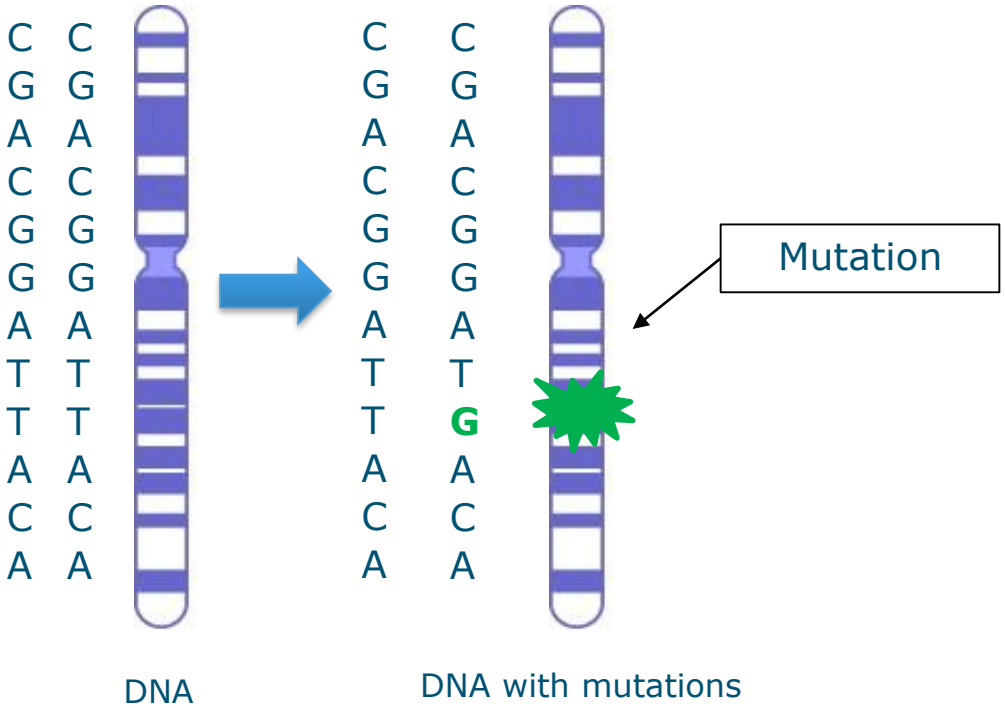
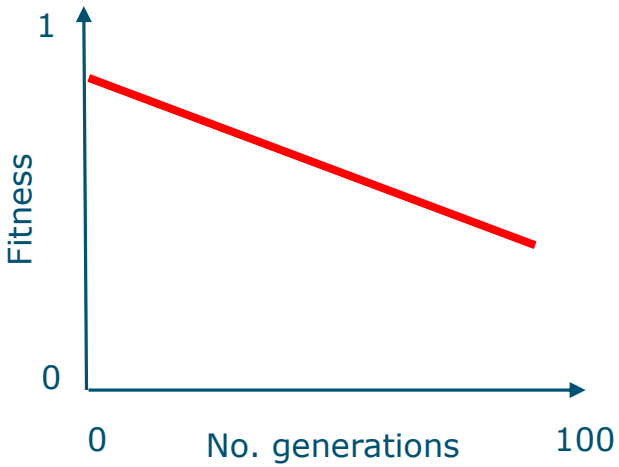
Mutations

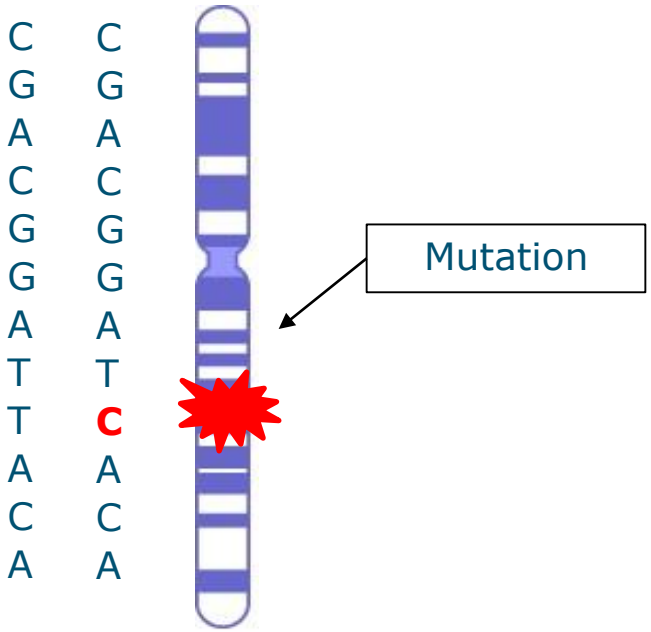
- A **mutation** is a change in the normal DNA sequence
- They are usually **neutral** (no effect on the fitness of the organism)



Mutations

- However, sometimes a change in the normal DNA sequence can negatively impact an organism fitness **(deleterious)**





DNA with mutations

Mutation-selection balance

+ genetic load
+ frequency harmful mutations
+ risk of inbreeding depression

- genetic diversity
- population survival

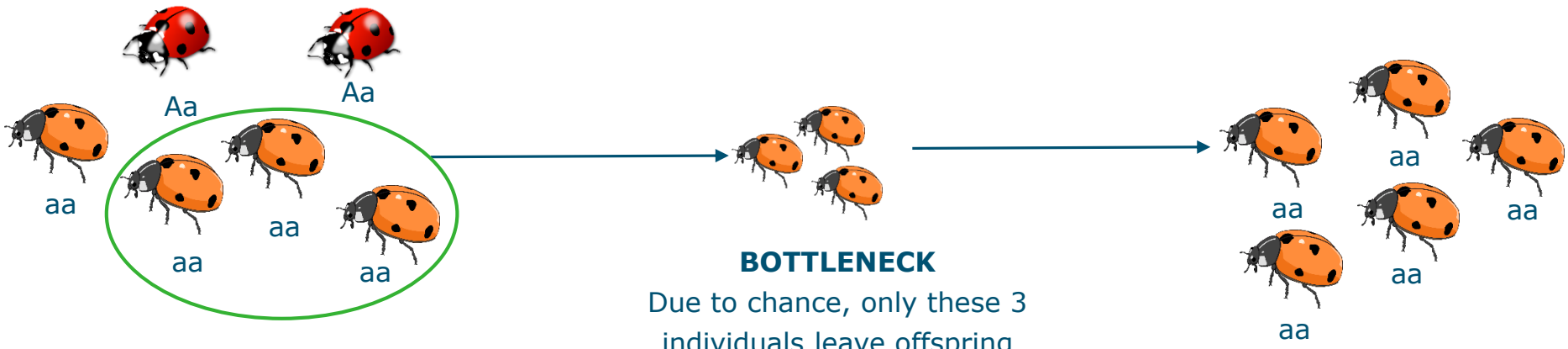
(Kimura et al., 1963)

Genetic drift is the major driving force in small populations

Parent population

Surviving population

Next generation



BOTTLENECK

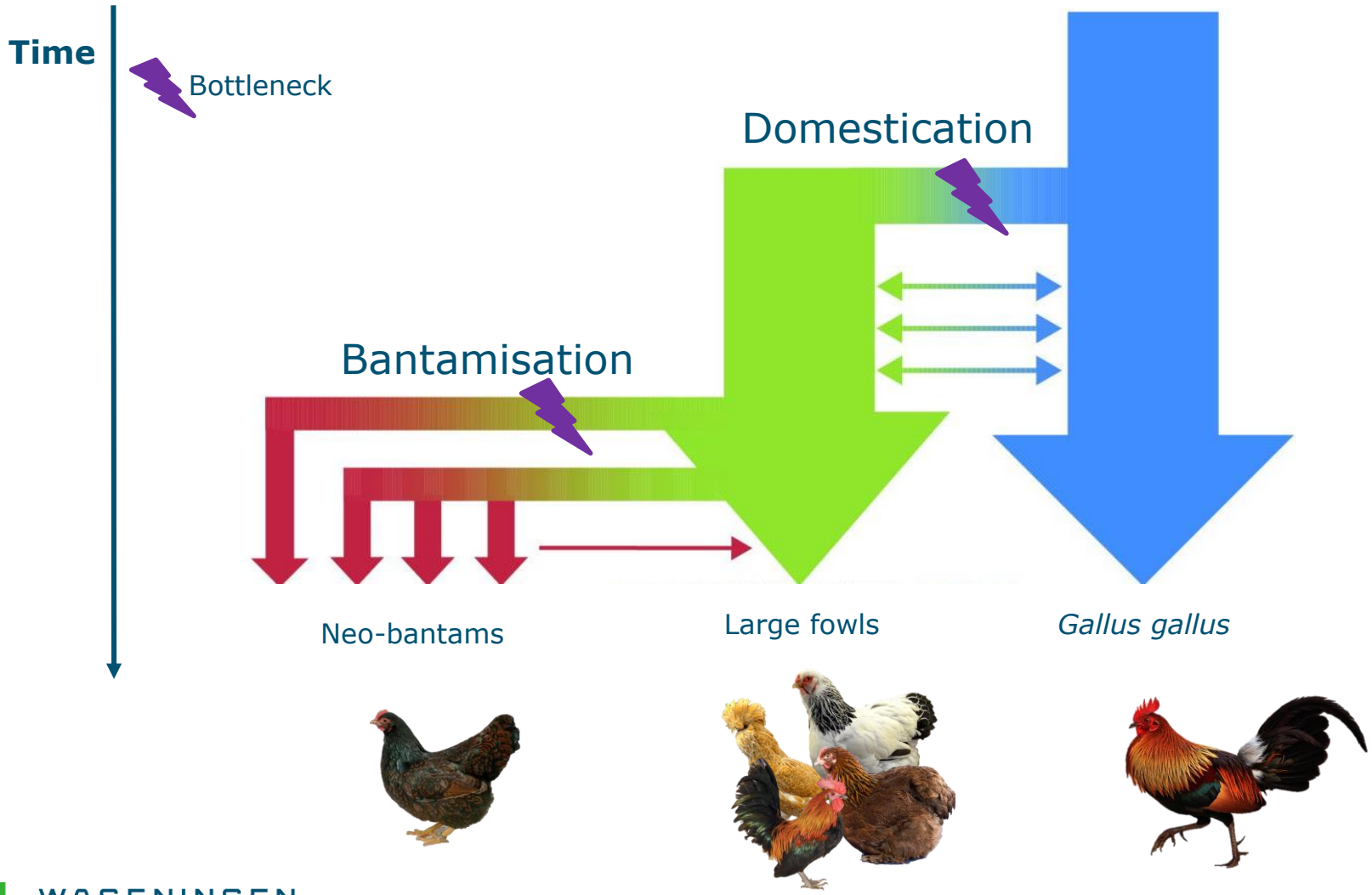
Due to chance, only these 3 individuals leave offspring

Freq. of A = 0.3
Freq. of a = 0.7

Freq. of A = 0
Freq. of a = 1

What kind of bottleneck?

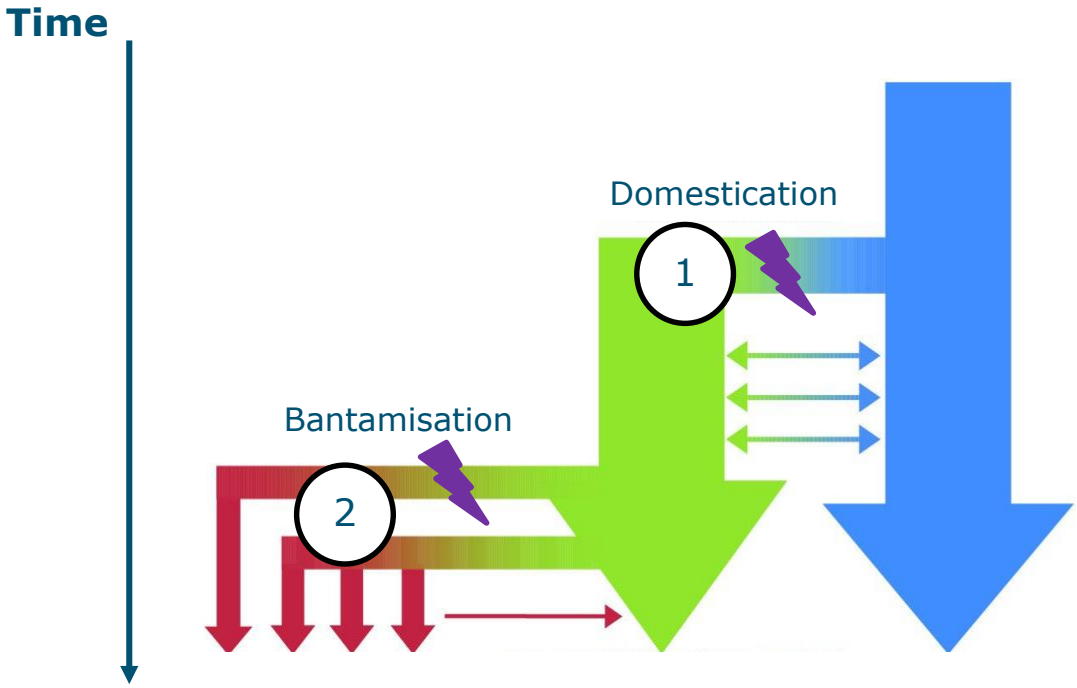
Domesticated species are an excellent model to study the consequences of population bottlenecks on genetic and deleterious variation



Research questions

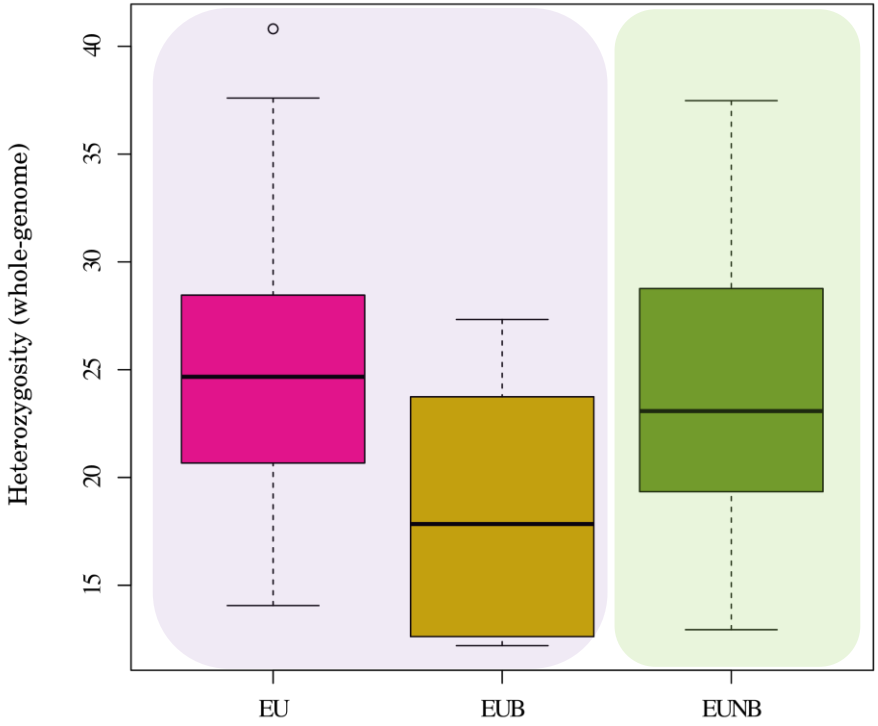
Traditional fancy breeds of chicken were used as a model species to investigate the **consequences** of two types **of bottlenecks on deleterious variation:**

1. Domestication bottleneck
2. Bantamisation bottleneck

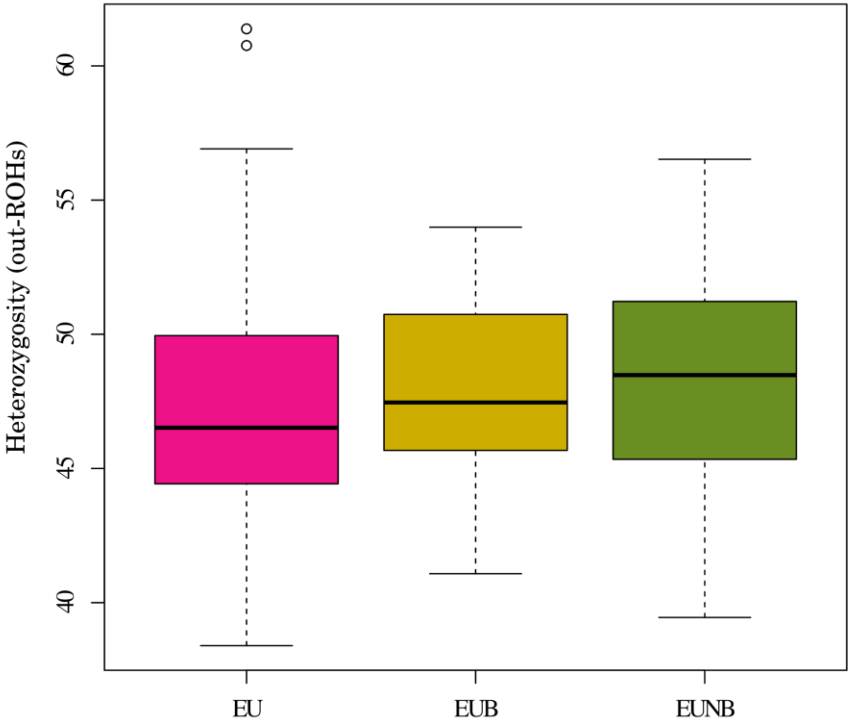


Recently bottlenecked breeds have higher genetic diversity but, ...

Whole genome



Excluding ROHs



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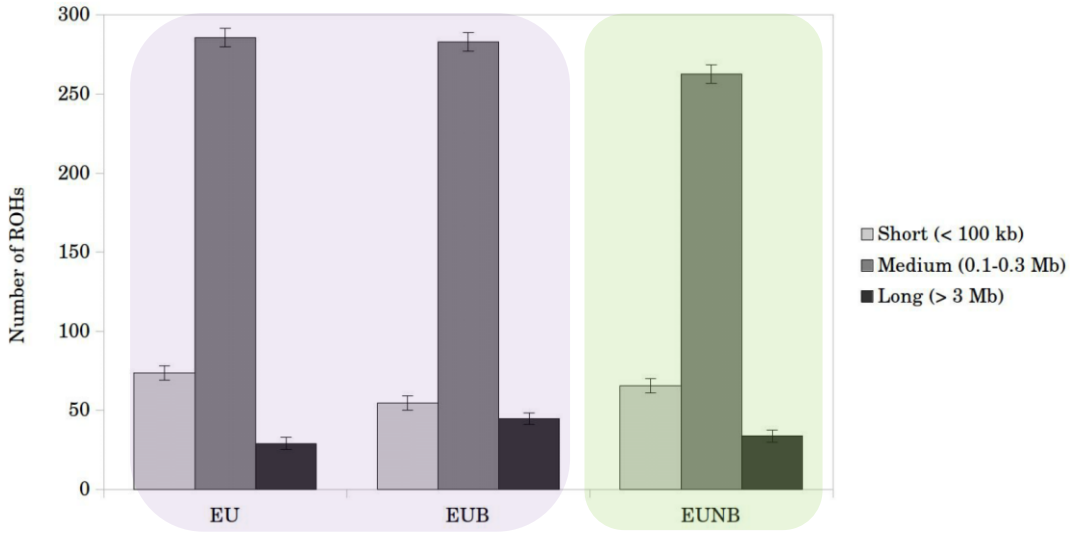
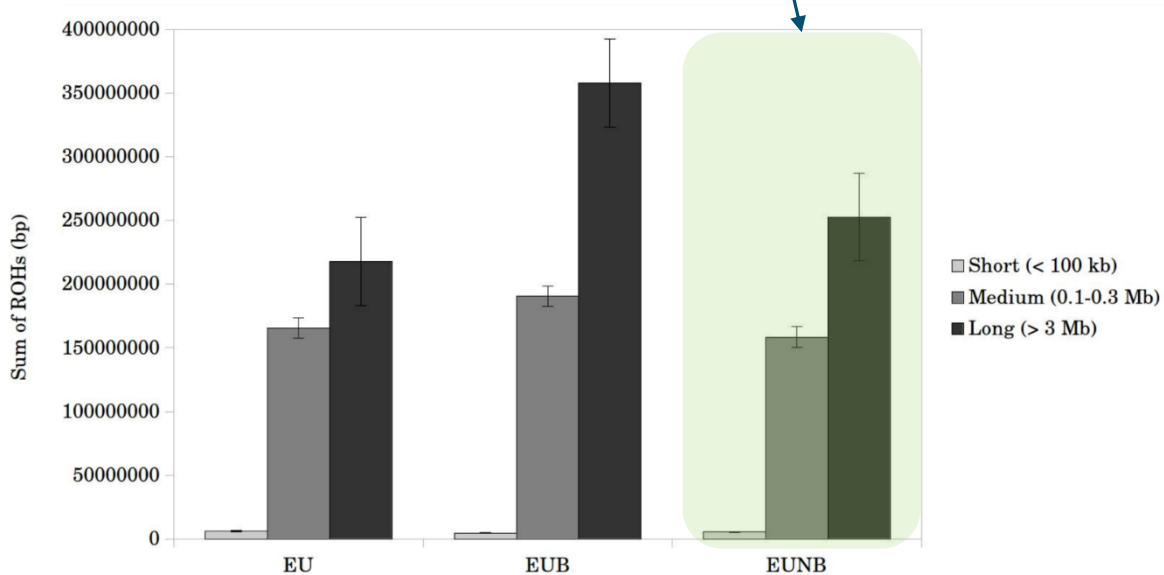
… Are much more inbred!



Up to 70% of the genome covered by ROHs!



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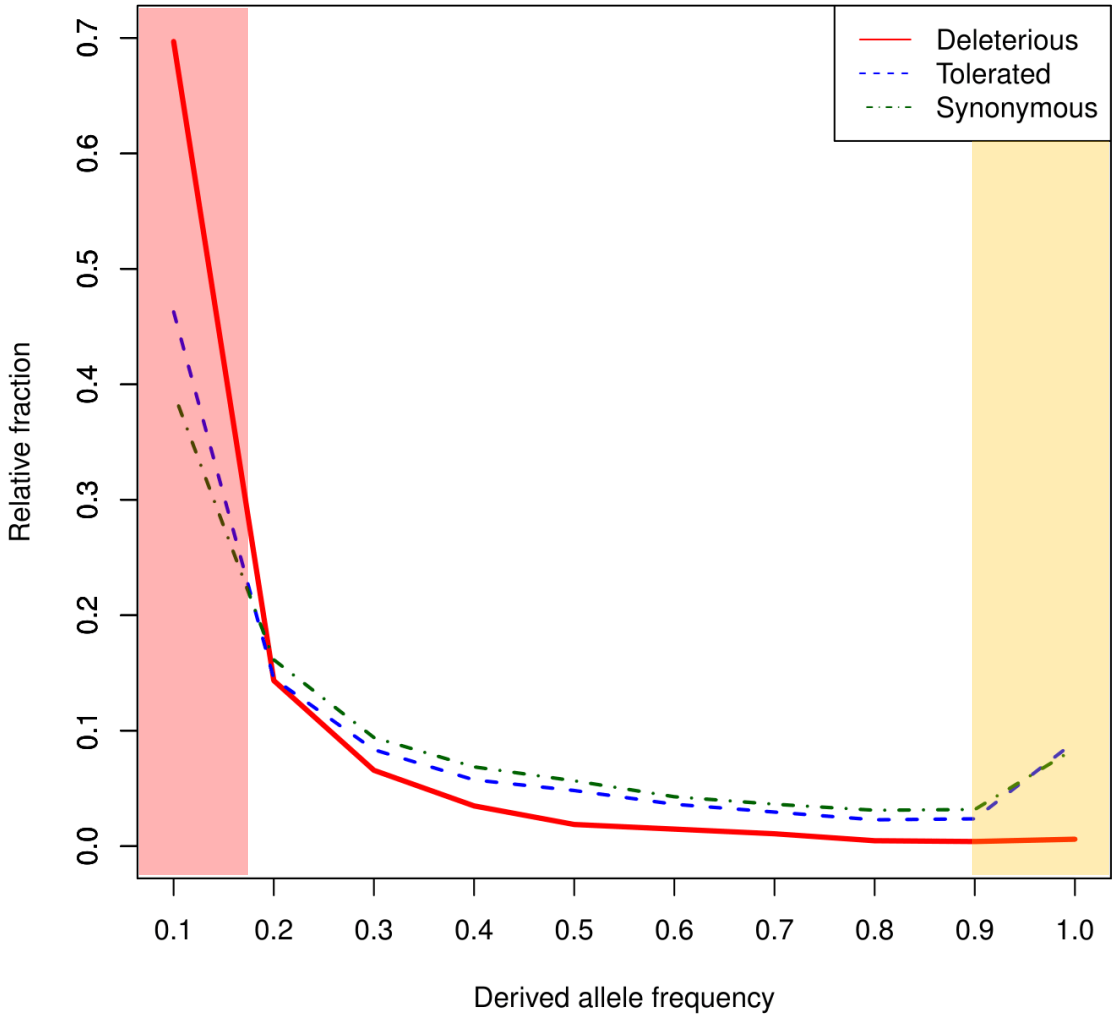
Purifying selection removes deleterious alleles from the population



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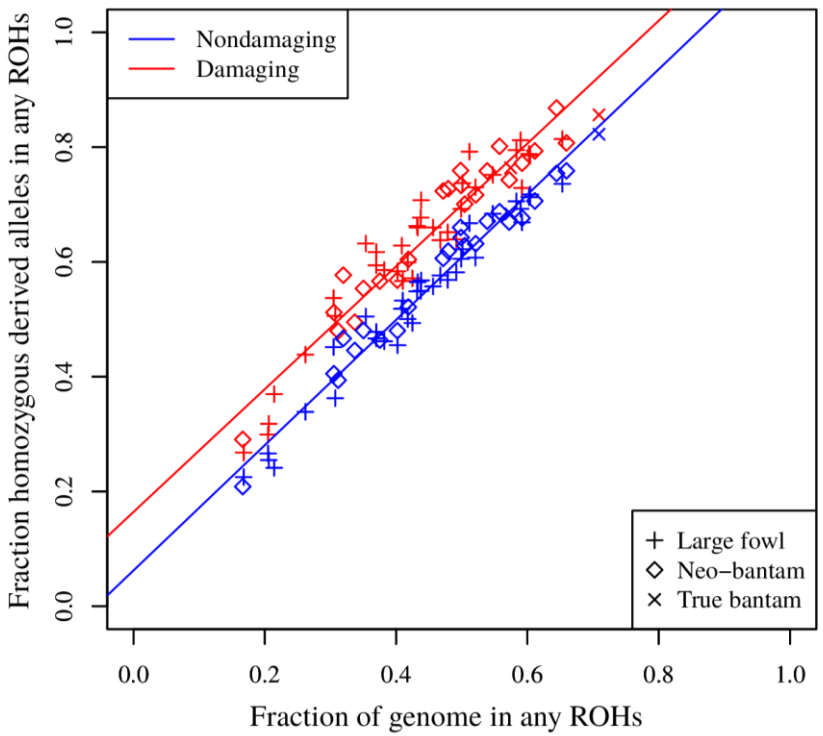
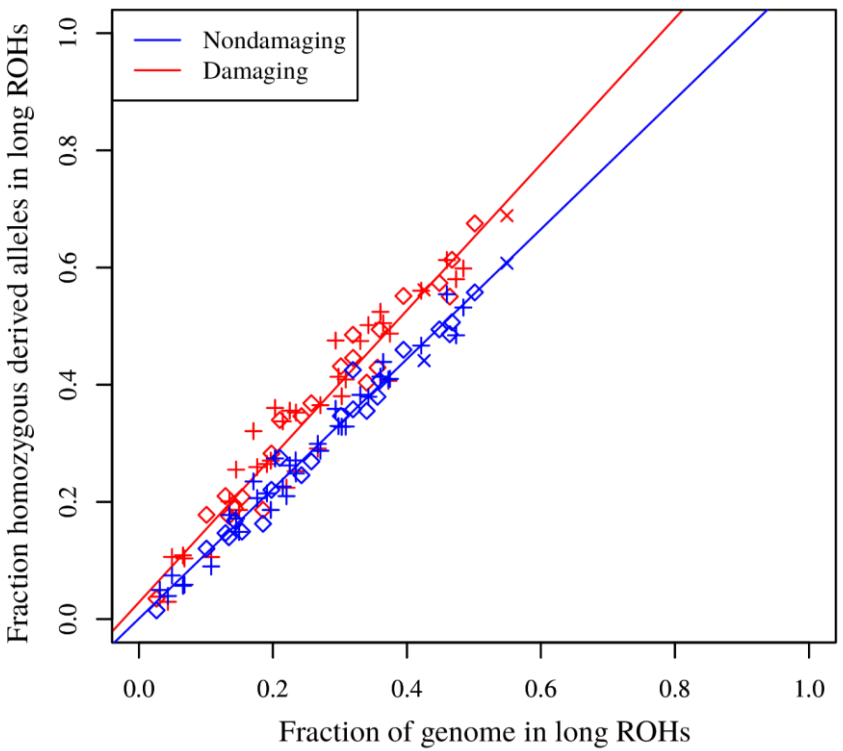
Deleterious alleles are mostly found in ROHs



And particularly long ROHs



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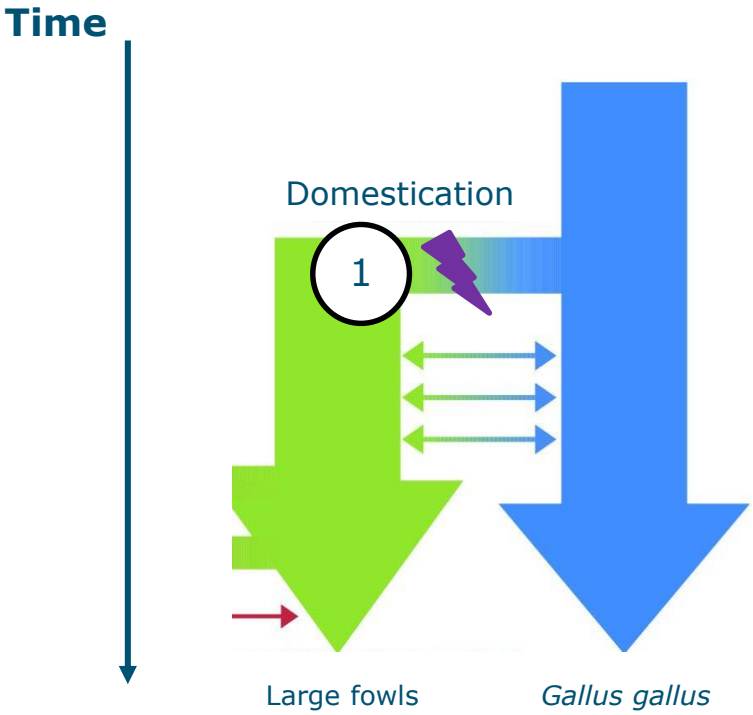
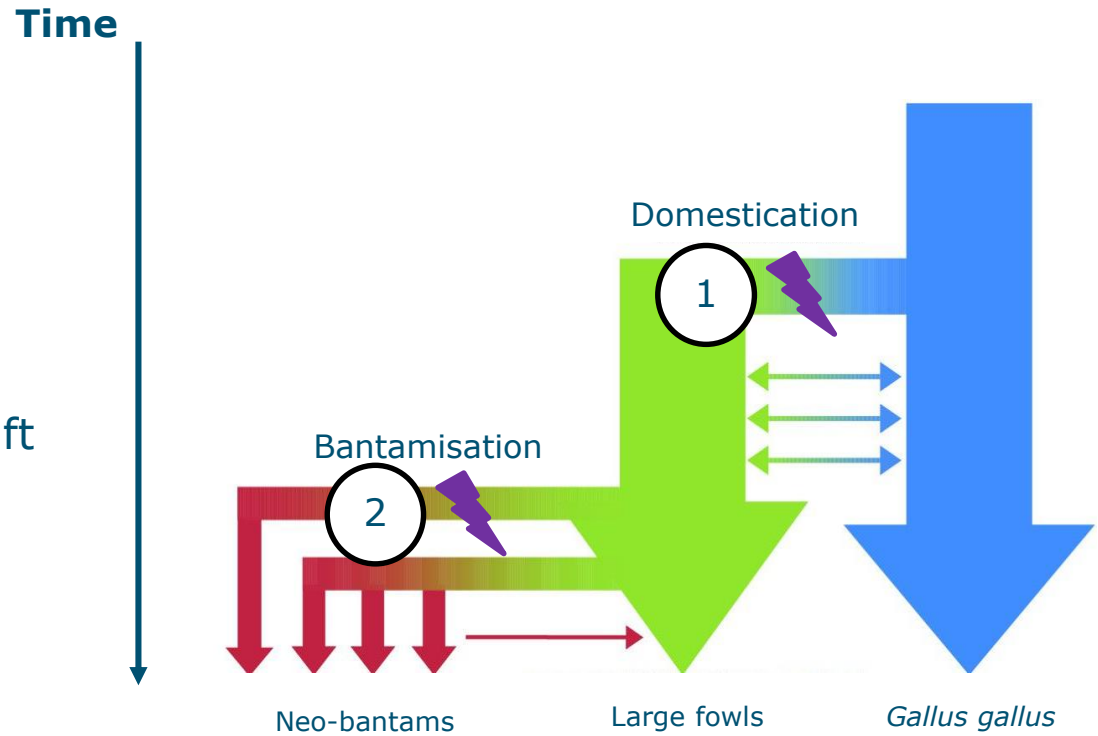
Discussion – domestication bottleneck

- Small population size for a long period of time
- Domestication substantially decreased the effective population size (*Ne*) and efficacy of purifying selection
- Deleterious mutations of especially small effects are expected to have risen in frequency and become fixed



Discussion – bantamisation bottleneck

- Small number of founder individuals
- Weakly deleterious mutations accumulate due to genetic drift → low efficacy of purifying selection



Discussion

- Inbreeding and artificial selection can also affect the mutational load → probability of harmful mutations to become homozygous
- In the long-term, lowered individual fitness (inbreeding depression)
- In small populations the probability of mating between relatives is high

Discussion

- Breeding programs can counterbalance the effects of inbreeding and artifial selection (Derks et al., 2018)
- In traditional breeds, as well as in small populations under natural selection, breeding and conservation programmes are often not in place
- Future breeding preferences?

Conclusions

- The time and nature of population bottleneckes can substantially shape the deleterious variation landscape in small populations
- Recently bottlenecked populations have a higher mutational load

Future perspectives:

- Use genomic information on deleterious variation to establish breeding/conservation programs
- Facilitating purging through inbreeding avoidance
- Information on fitness-related traits

Thank you for your attention!

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IMAGE consortium

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