



Simulation of genetic rescue from a gene bank using a complex load structure

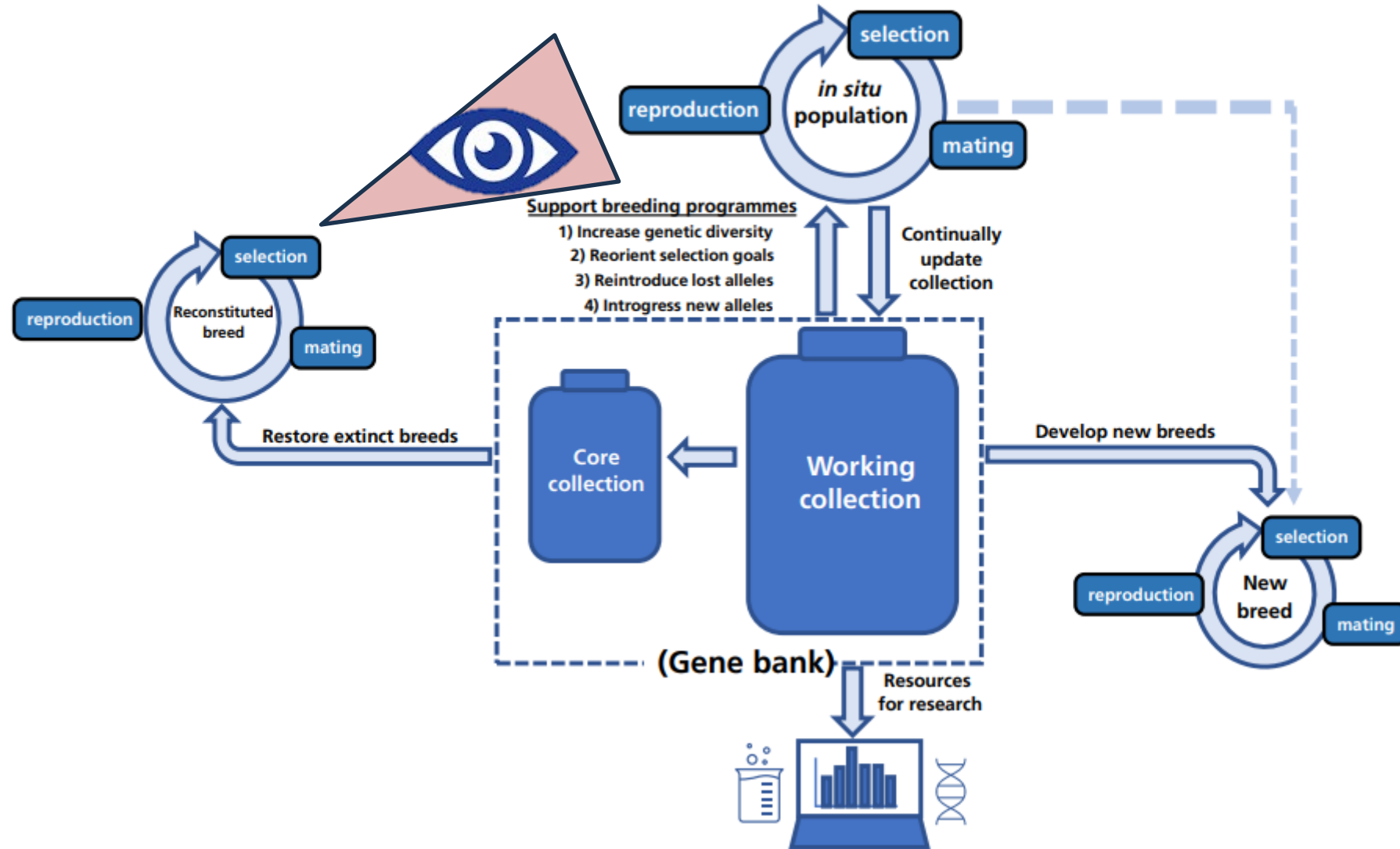
Christian Reimer^{1,2}, Johannes Geibel^{1,2}, Torsten Pook^{2,3}, Steffen Weigend^{1,2}

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Uses of gene bank collection (*ex situ in vitro* cryocollection)



Background

(Farm) animal conservation efforts predominantly focus on

- the maintenance of genetic diversity
- preserving identity of a breed

„A collection of material in a genebank can be used to help maintain genetic diversity, such as by alternating the utilization of parents across generations when using genebank material (e.g. Sonesson *et al.*, 2002) or extending the generation interval. Stored material may also be used to safeguard against the accumulation of deleterious recessive alleles or to redirect a breeding objective.” FAO „INNOVATIONS IN CRYOCONSERVATION OF ANIMAL GENETIC RESOURCES - DRAFT TECHNICAL GUIDELINES

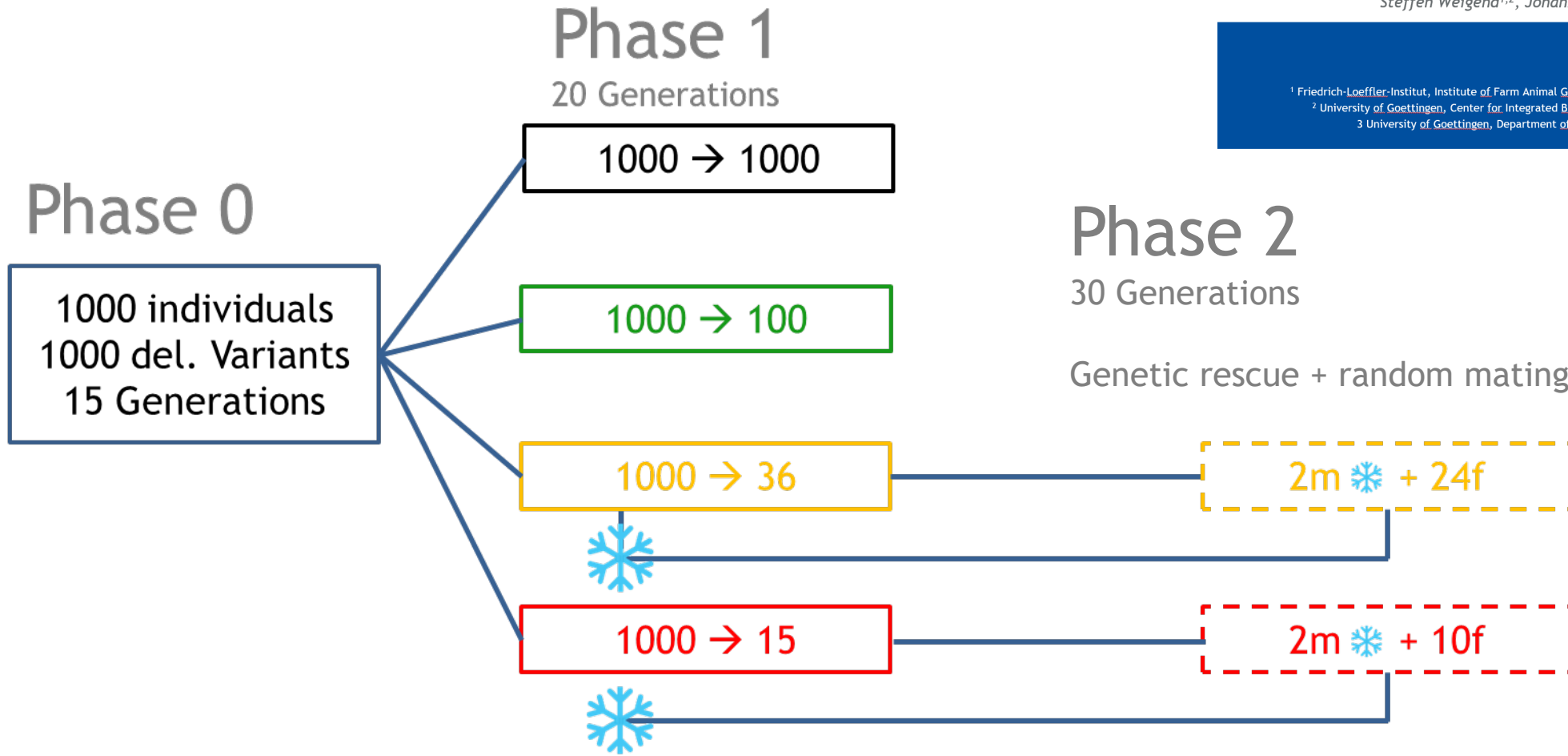
BUT what if the „Savior“ carries deleterious mutations?

Genetic rescue of small populations in the presence of deleterious variation

Christian Reimer^{1,2}

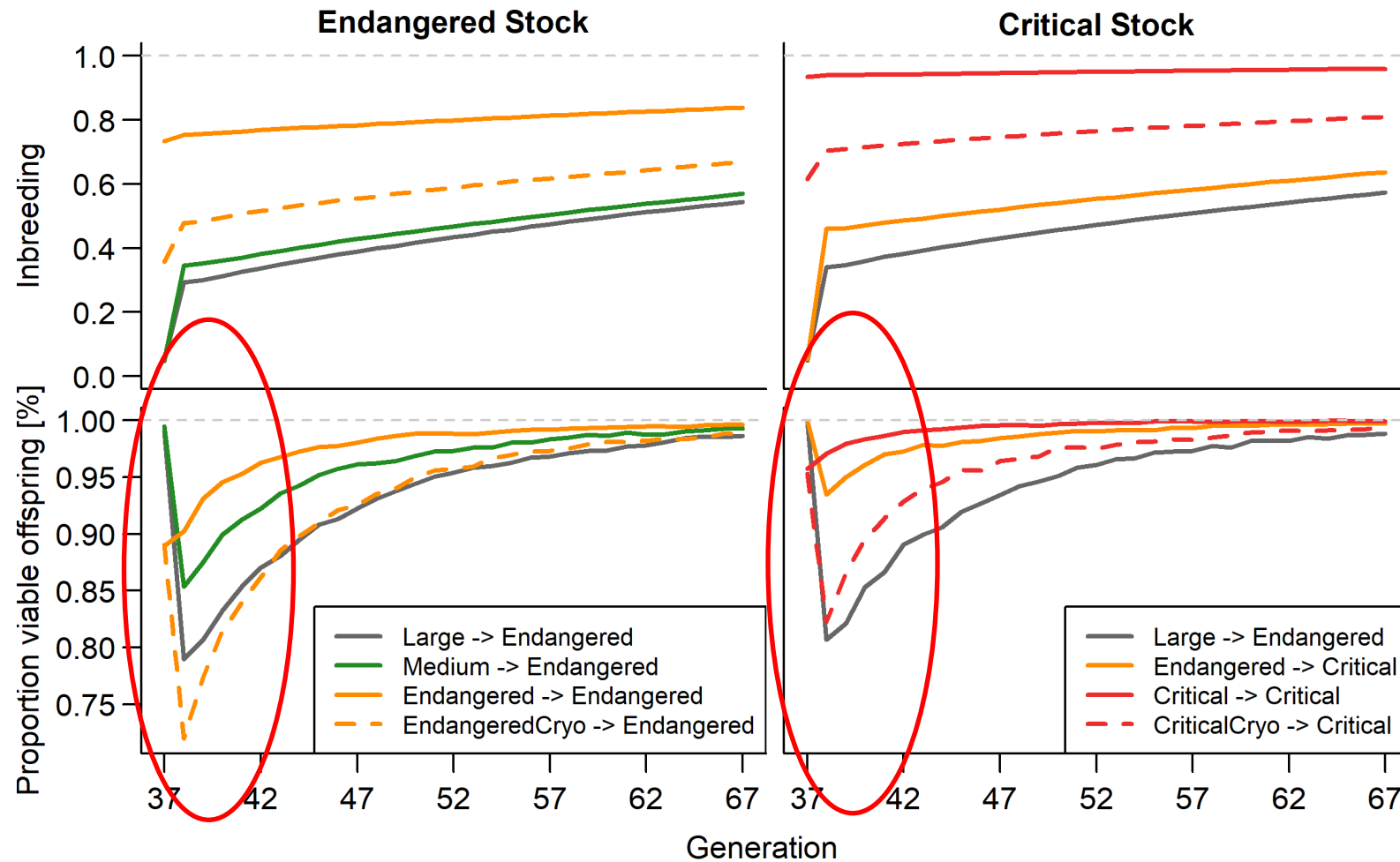
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Simulation study

- Simulation of different rescue scenarios



Phase 2

30 Generations

Genetic rescue + random mating

Material & Methods

Stochastic simulations with MoBPS

Ensembl Pig map with 18 autosomes, 60k SNPs

Two traits:

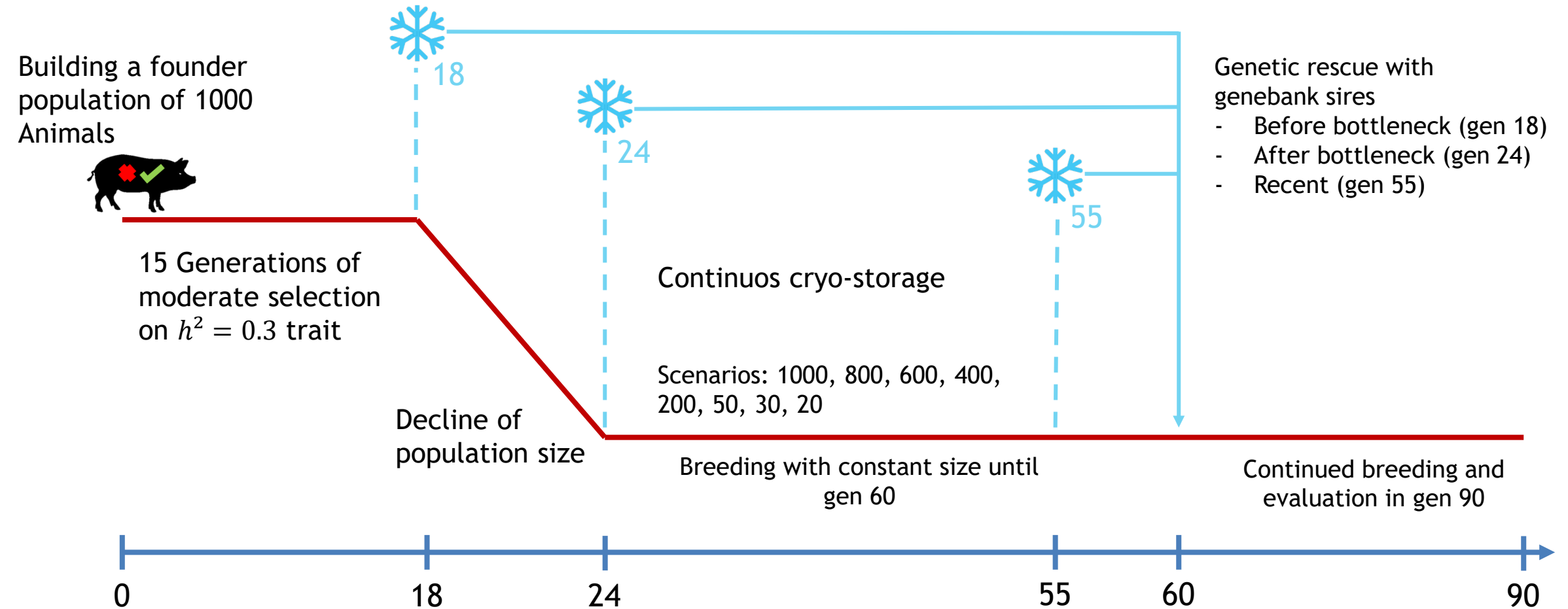
- 6-month weight, moderate selection
- Genetic load trait with effect on survival
 - 1000 strongly deleterious loci
 - 2000 low to moderate effect loci

10 repetitions



*MoBPS; Pook et al.
2020, G3*

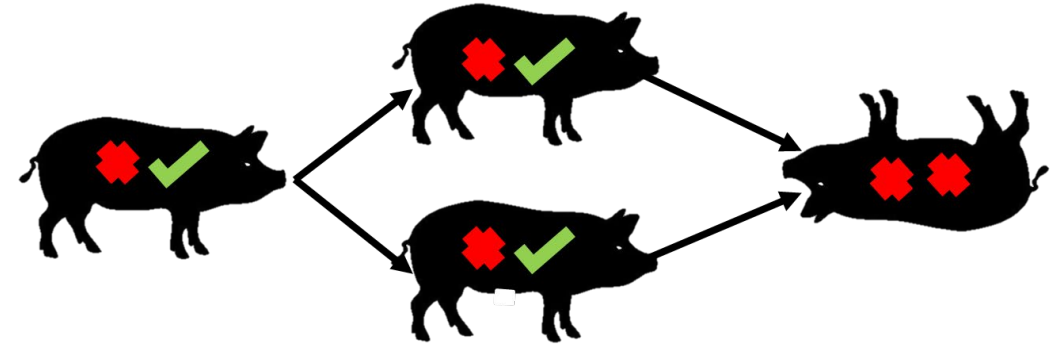
M&M - Simulation study



Modeling of Load

Strongly deleterious loci

- 1000 loci, randomly distributed
- MAF: mean 0.01 ± 0.005
- Effect size: -100 for recessive homozygotes



Low to moderate effect loci

- 2000 loci, randomly distributed
- Recessive homozygotes with negative effect
- Effects sampled from beta-distribution range between -0.00001 and -0.15

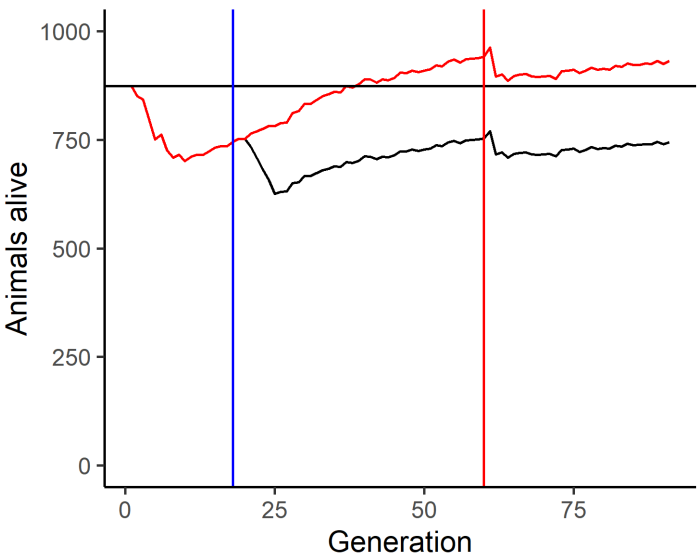
Animals are culled below a threshold of -100 composite load

Results

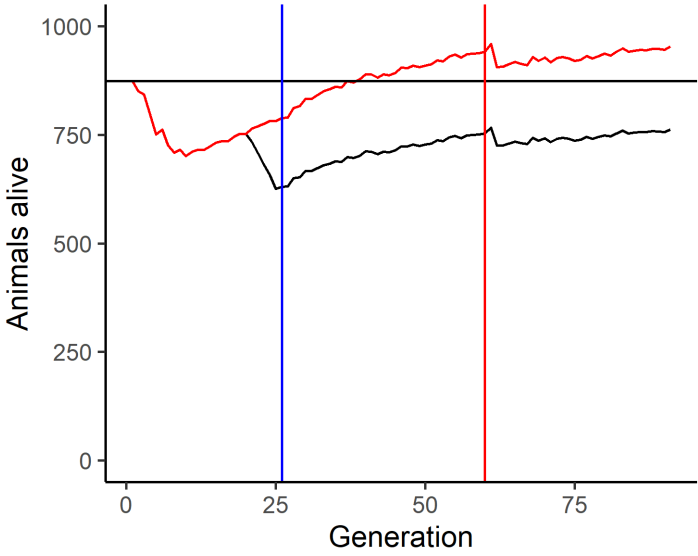
Results - surviving animals

Semen for rescue in Gen 60 taken from:

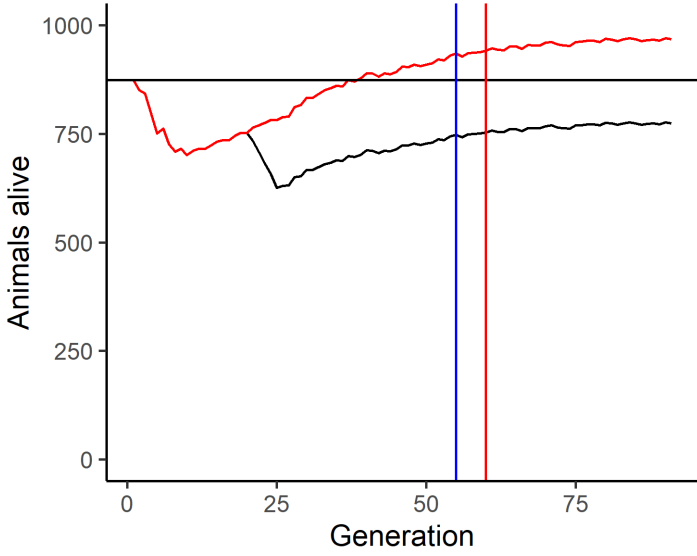
Generation 18



Generation 24



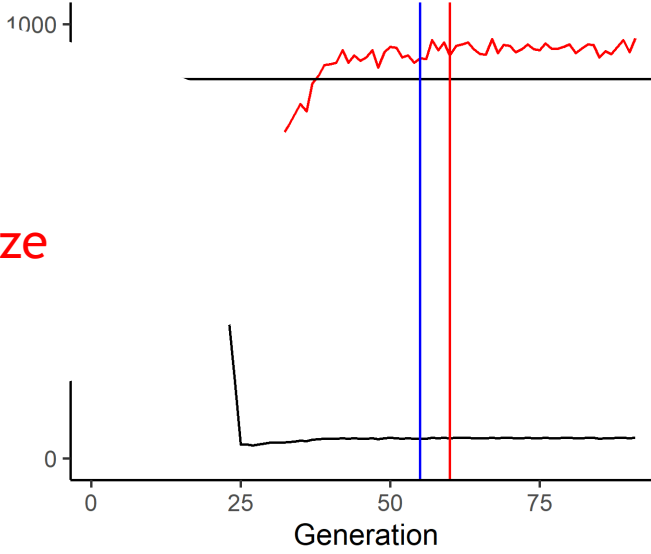
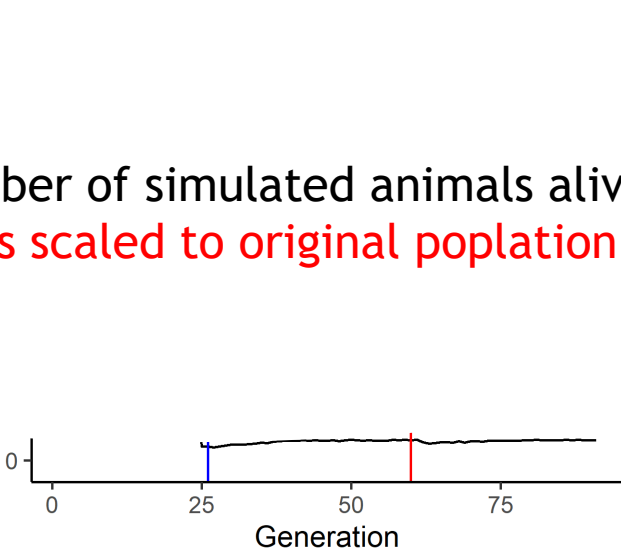
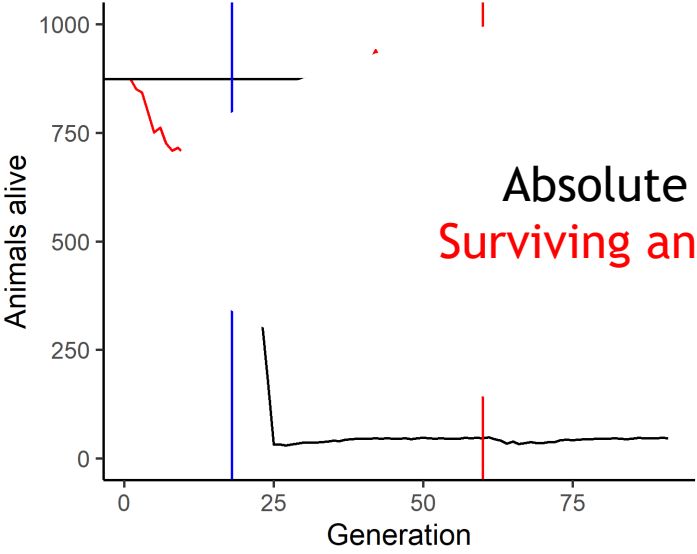
Generation 55



Absolute number of simulated animals alive
Surviving animals scaled to original population size

1000 → 800

1000 → 50

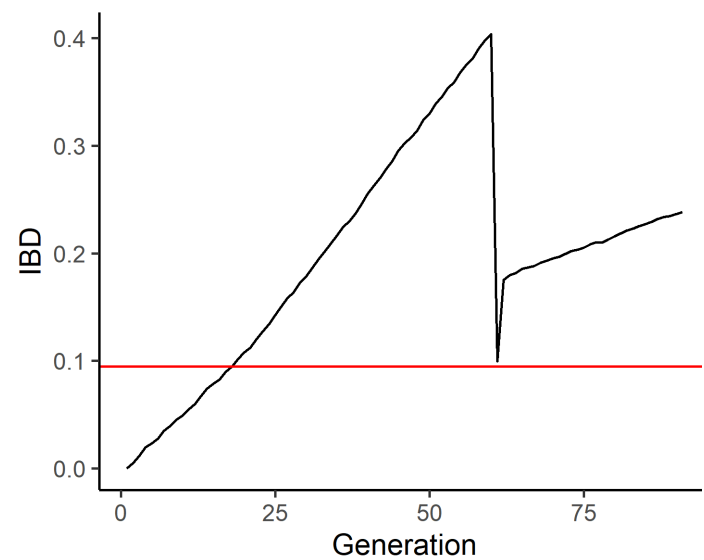


Results - Inbreeding

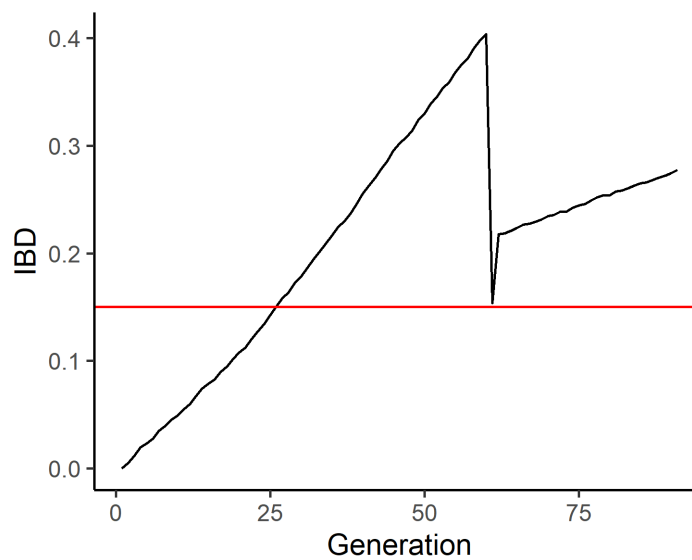
Semen for rescue in Gen 60 taken from:

1000 → 800

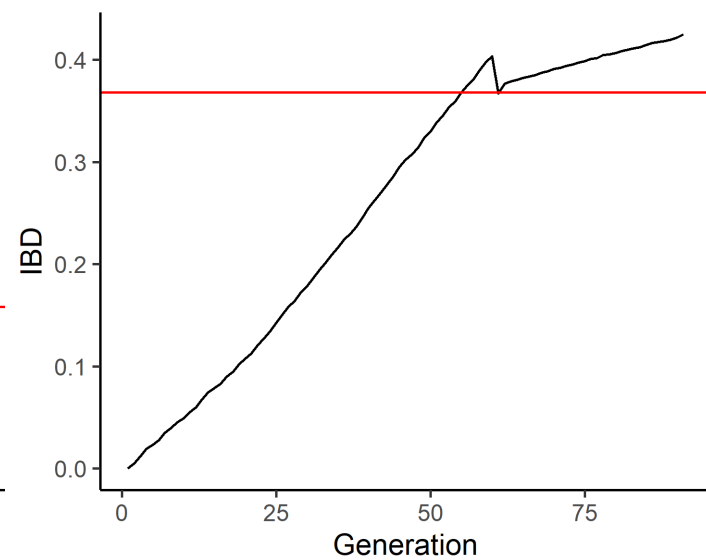
Generation 18



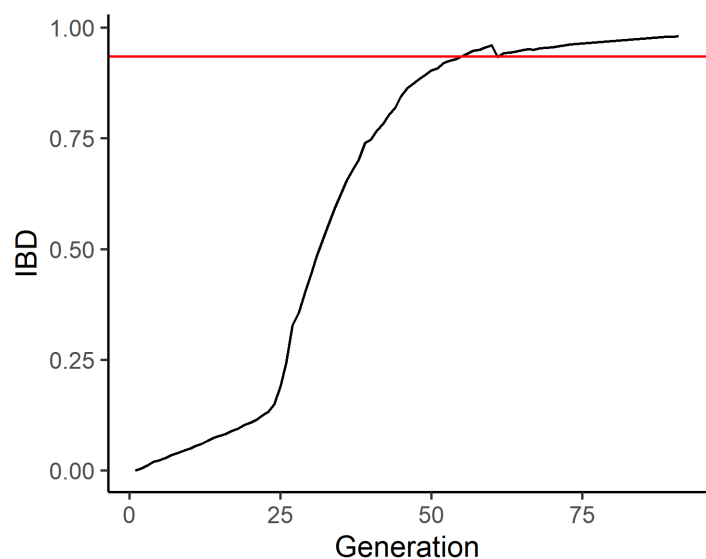
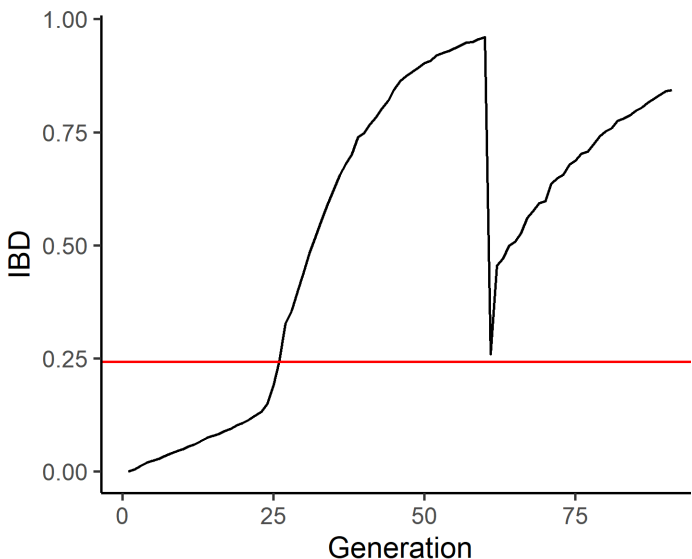
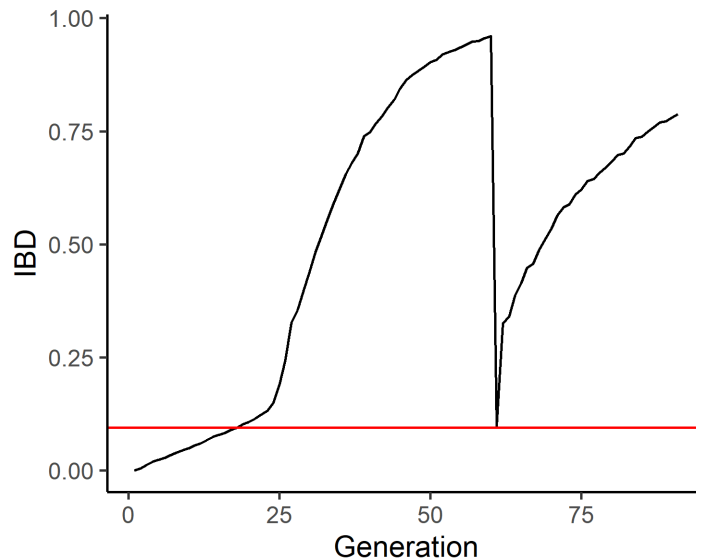
Generation 24



Generation 55



1000 → 50

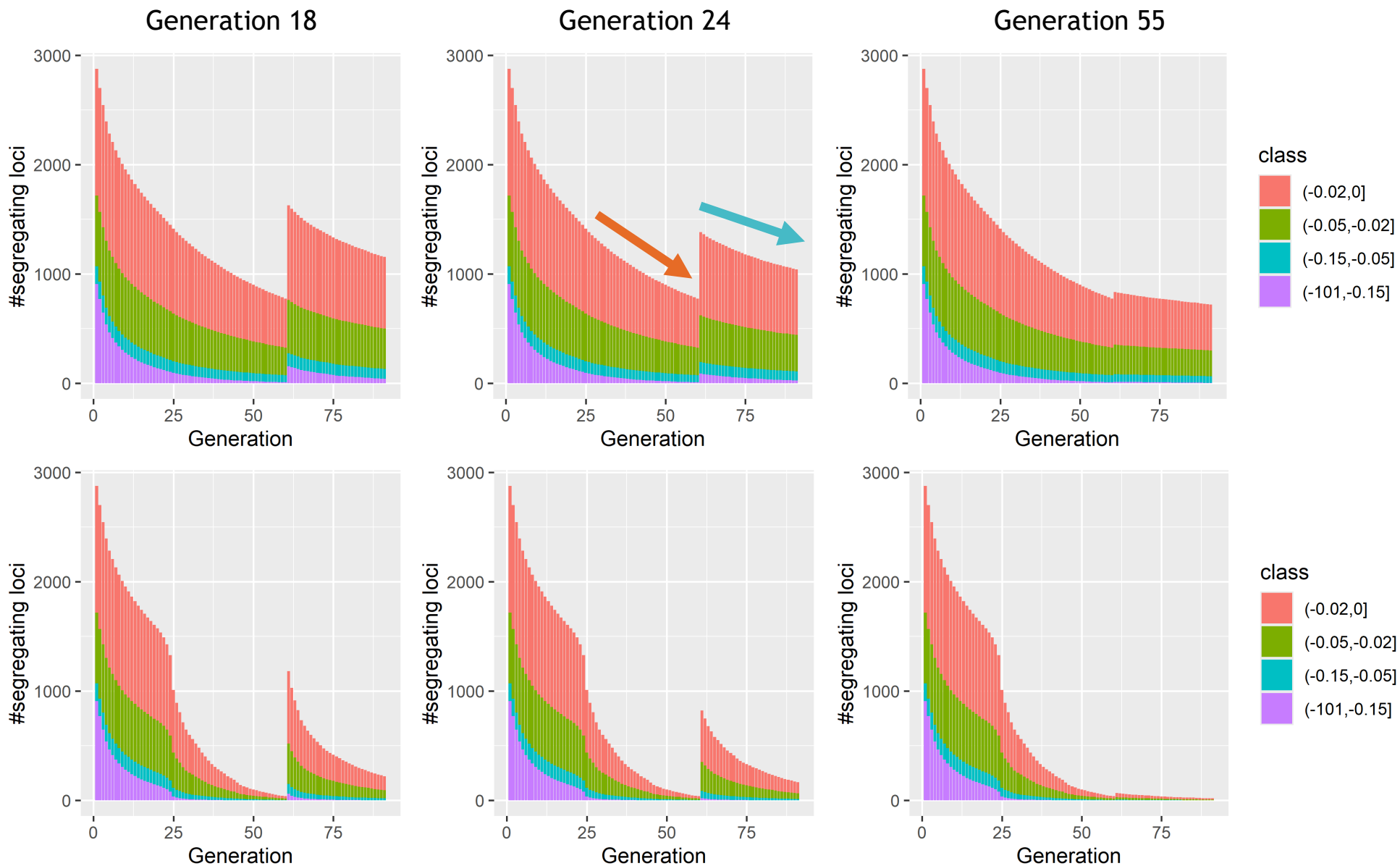


Results - Segregating deleterious loci

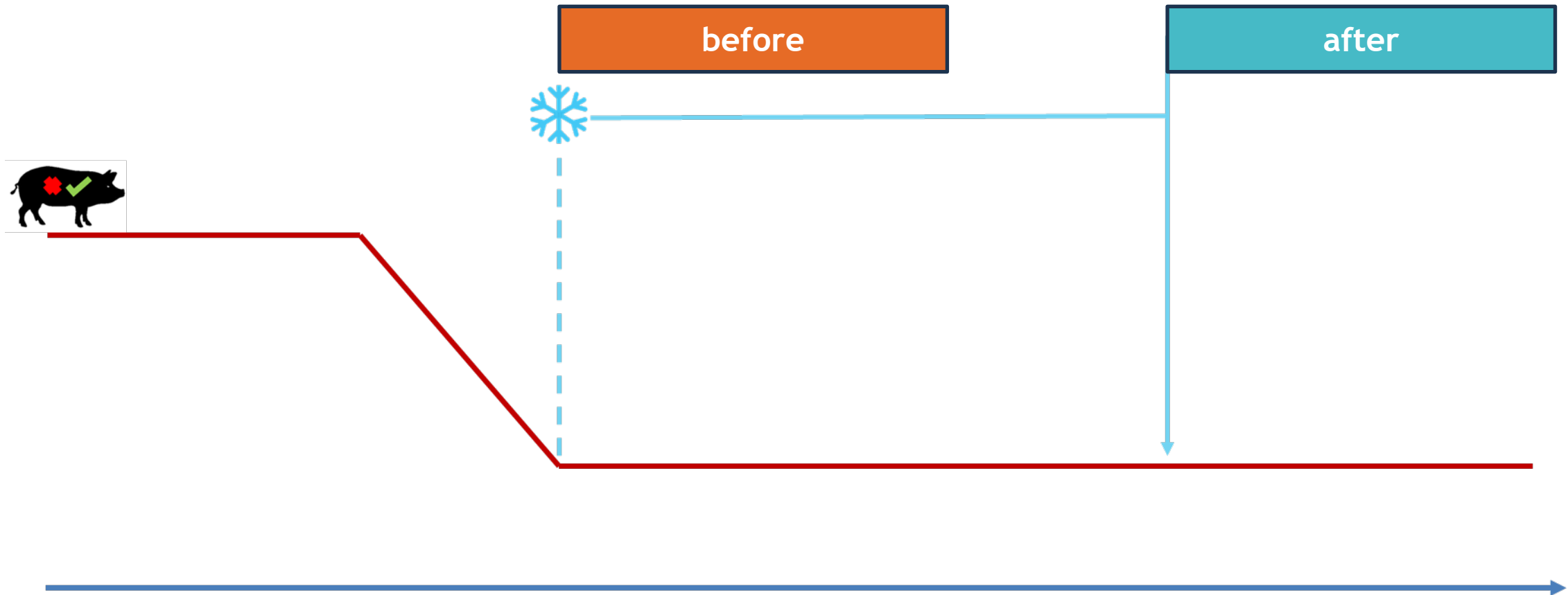
Semen for rescue in Gen 60 taken from:

1000 → 800

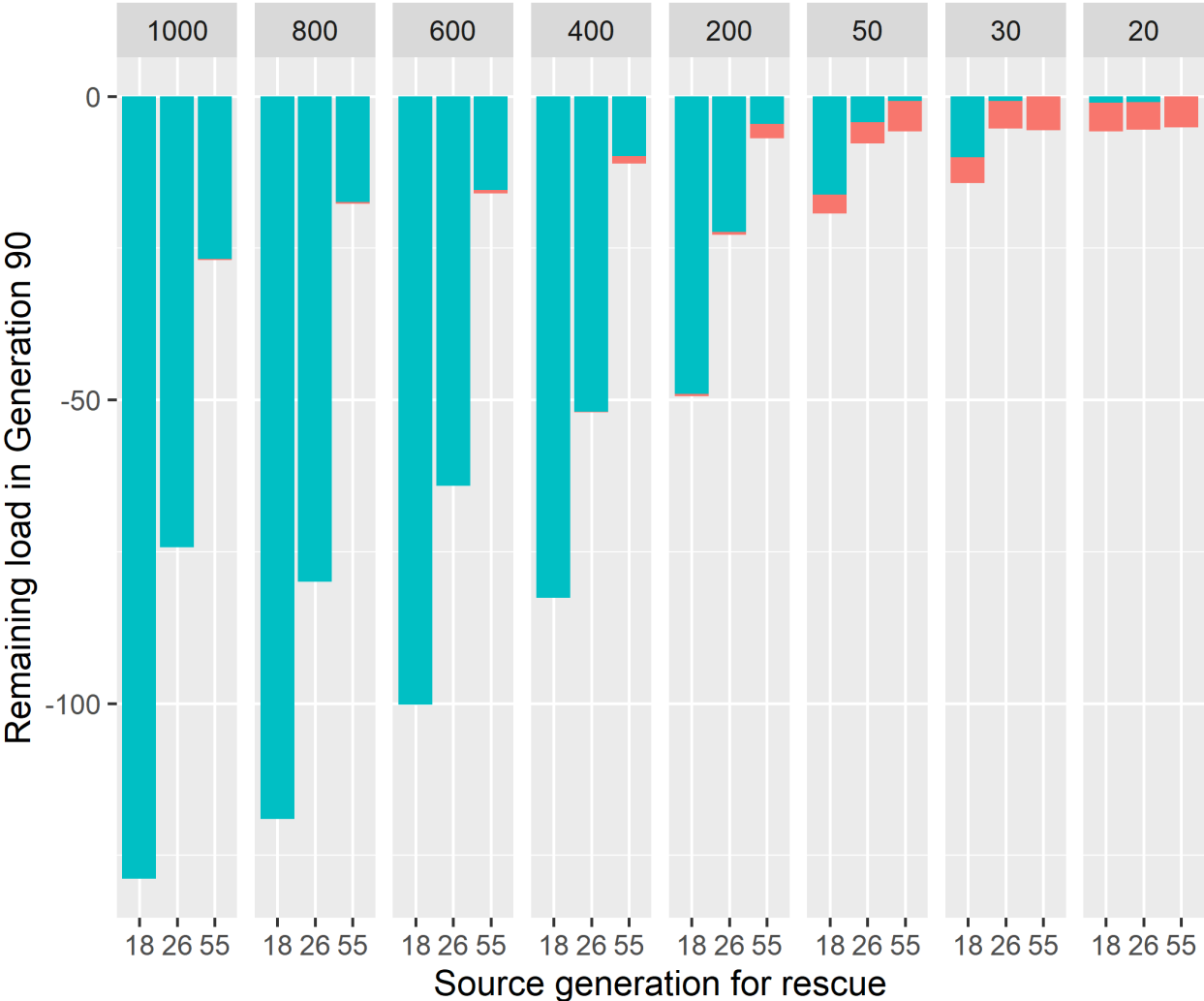
1000 → 50



Results - Segregating deleterious loci



Results - remaining load 30 generations after rescue



Fixed = monomorphic for deleterious allele

Free = deleterious allele variable

Conclusion

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Rescue with more diverse population increases diversity, and deleterious load

But

Reintroducing load compromises viability

Purging efficient against all effect classes

But

Purging reintroduced load in a deminished population is less efficient or impossible

It appears advisable to use semen obtained after a bottleneck

But

Limited knowledge of load structure leads to limited informative value of simulations

Thank you for your attention!



Results - Segregating deleterious loci

