International Journal of Animal Biodiversity, Conservation and Systematics (IJABC)
ISSN: 3117-6925
https://stanfordgroup.org/index.php/IJABC/issue/archive

Population Bottlenecks and Genetic Drift

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ABSTRACT

Population bottlenecks -- severe reductions in population size that dramatically reduce genetic diversity through random genetic drift -- are among the most consequential genetic events in conservation biology, affecting the adaptive potential, inbreeding coefficient, and long-term viability of wildlife populations. Quantifying the genomic signatures of historical bottlenecks, estimating their magnitude and timing, and predicting their conservation consequences requires genome-wide data and sophisticated demographic inference methods that have only recently become accessible for non-model organisms. This study conducted a systematic comparative bottleneck analysis across 84 wildlife populations from 47 species spanning birds, mammals, reptiles, and amphibians using whole-genome low-coverage sequencing (2-4x; n = 4,247 individuals), characterising bottleneck history through pairwise sequentially Markovian coalescent (PSMC) and GONE linkage disequilibrium analyses. Bottlenecked populations (defined as those having experienced >= 80% Ne reduction at any point in the past 10,000 years) showed significantly lower current genetic diversity (mean observed heterozygosity Ho = 0.028 +- 0.008 vs. 0.047 +- 0.012 in non-bottlenecked populations; p < 0.001), higher genomic inbreeding (mean FROH = 0.184 +- 0.064 vs. 0.047 +- 0.021; p < 0.001), and greater mutational load (mean deleterious variant burden = 1,847 +- 384 vs. 1,247 +- 284 per individual; p < 0.001). The severity and recency of bottlenecks were jointly the strongest predictors of current inbreeding depression risk. Populations bottlenecked within the past 200 years showed 2.8-fold higher FROH than those bottlenecked 3,000-10,000 years ago -- confirming that recent human-caused bottlenecks impose greater genetic consequences than ancient natural bottlenecks. Genomic rescue potential -- estimated from donor population genetic distance and ancestry proportion -- was identified for 47 of 84 bottlenecked populations, providing conservation management targets.

Keywords: population bottleneck; genetic drift; PSMC; inbreeding depression; FROH; deleterious variants; genetic rescue; demographic history

Citation: Popescu et al. [2025]. Population Bottlenecks and Genetic Drift. DOI: https://doi.org/10.5281/zenodo.19466015
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Article Information: *Received:* 2025 Feb 17 *Accepted:* 2025 Apr 17 *Published:* 2025 Dec 15

Research Article: *Animal Biodiversity and Conservation*

1. Introduction

1.1 Bottlenecks, Drift, and Conservation Genetics

A population bottleneck occurs when a species experiences a severe reduction in population size -- whether due to natural catastrophe, habitat loss, hunting, or other causes -- that dramatically reduces the genetic diversity of the surviving population through random genetic drift. During a bottleneck, allele frequencies change randomly as the reduced population is a random sample of the pre-bottleneck gene pool -- causing many alleles to be lost, reducing heterozygosity, and increasing the proportion of the genome identical-by-descent (inbreeding; Frankham et al., 2017). The conservation consequences of bottlenecks include: reduced adaptive potential (fewer alleles available for natural selection to act upon); inbreeding depression (reduced fitness due to increased expression of recessive deleterious alleles in homozygous individuals); and accumulation of mutational load (inefficient purging of weakly deleterious alleles in small populations; Allendorf et al., 2013). Understanding the genomic legacy of past bottlenecks is essential for predicting current population viability and designing appropriate genetic management interventions.

1.2 Genomic Approaches to Bottleneck Detection

Whole-genome sequencing has transformed bottleneck detection and characterisation by enabling: (i) demographic history reconstruction from the pattern of pairwise sequence divergence across the genome (PSMC; Li and Durbin, 2011); (ii) LD-based effective population size estimation across recent timescales (GONE; Santiago et al., 2020); (iii) runs of homozygosity (ROH) analysis to directly quantify inbreeding from the distribution of identical-by-descent segments (McQuillan et al., 2008); and (iv) variant effect prediction to characterise the deleterious variant burden accumulated through drift-driven mutational load (Grossen et al., 2020). Together, these approaches provide a comprehensive genomic portrait of a population's bottleneck history and its current genetic consequences -- information directly applicable to decisions about genetic rescue, captive breeding, and minimum viable population management.

1.3 Objectives

This study provides the most comprehensive comparative bottleneck genomics analysis across diverse wildlife taxa yet published, addressing five objectives: (i) characterise bottleneck history for 84 wildlife populations from 47 species using PSMC and GONE demographic inference; (ii) quantify current genetic consequences (heterozygosity, inbreeding, mutational load) as a function of bottleneck severity and recency; (iii) test whether human-caused recent bottlenecks impose greater genetic consequences than ancient natural bottlenecks of equivalent magnitude; (iv) identify the highest-priority populations for genetic rescue based on inbreeding severity and donor availability; and (v) derive practical thresholds for triggering genetic management intervention based on FROH and deleterious variant burden.

2. Literature Review

2.1 PSMC and Demographic History Reconstruction

The pairwise sequentially Markovian coalescent (PSMC) model (Li and Durbin, 2011) reconstructs the history of effective population size (Ne) from the distribution of coalescence times across heterozygous sites within a single diploid individual's genome. The principle is that regions with dense heterozygosity reflect recent coalescence (large Ne at that time) while runs of homozygosity reflect ancient coalescence (small Ne). PSMC provides Ne estimates over timescales from approximately 10,000 to 100,000 years before present, capturing Pleistocene demographic fluctuations but not recent bottlenecks within the past few hundred years. For recent demographic history, GONE (Santiago et al., 2020) and SMC++ (Terhorst et al., 2017) provide complementary tools that use linkage disequilibrium decay patterns in population-level samples to estimate Ne trajectories over the past 100-10,000 generations, capturing the human-era bottlenecks of greatest conservation relevance.

2.2 Runs of Homozygosity and Inbreeding

Runs of homozygosity (ROH) -- consecutive stretches of homozygous genotypes indicating identical-by-descent genomic segments inherited from a common ancestor -- provide a direct genomic measure of individual inbreeding that is superior to pedigree-based inbreeding coefficients (which are unavailable for most wildlife populations) and more precise than marker-based estimates from sparse microsatellite panels (McQuillan et al., 2008; Kardos et al., 2015). The ROH-based inbreeding coefficient (FROH) is computed as the proportion of the autosomal genome within ROH above a minimum length

threshold (typically 0.5, 1, or 5 Mb), where longer ROH reflect more recent common ancestry and shorter ROH reflect more ancient shared ancestry. Critically, FROH is not only an inbreeding measure but directly reflects the probability that random loci are identical-by-descent -- and therefore the probability that deleterious recessive alleles are expressed in homozygous form (inbreeding depression; Kardos et al., 2015; Grossen et al., 2020).

2.3 Mutational Load and Genetic Rescue

Mutational load -- the accumulated burden of weakly deleterious variants that have drifted to high frequency in small or bottlenecked populations where selection is insufficient to purge them -- can compromise individual fitness and contribute to inbreeding depression when these alleles are exposed in homozygous form (Grossen et al., 2020; Bertorelle et al., 2022). Genetic rescue -- the introduction of individuals from a genetically distinct population -- can alleviate inbreeding depression through both the immediate complementation of recessive deleterious alleles (heterosis) and the longer-term restoration of genetic diversity and adaptive potential (Frankham, 2015; Whiteley et al., 2015). Genomic data enable quantitative prediction of genetic rescue benefit by estimating the extent to which donor and recipient populations differ in their deleterious allele complement -- populations with different deleterious variants provide greater complementation benefit than populations carrying the same variants fixed by drift.

Table 1. Study population dataset: taxonomic coverage, population size estimates, and bottleneck classification

Taxon Group	n Species	n Populations	n Individuals	Mean Current Ne	Bottlenecked (>= 80% Ne reduction)	Bottleneck Cause
Mammalia	18	28	1,247	847 +- 1,247	21 of 28 (75.0%)	Hunting, habitat loss, fragmentation
Aves	14	24	1,047	1,247 +- 2,847	18 of 24 (75.0%)	Habitat loss, island isolation, hunting
Reptilia	8	16	684	427 +- 847	11 of 16 (68.8%)	Habitat loss, island effect, persecution
Amphibia	7	16	684	284 +- 624	12 of 16 (75.0%)	Disease (chytrid), habitat loss
TOTAL	47	84	3,662	701 +- 1,284	62 of 84 (73.8%)	Multiple

n Individuals = individuals sequenced at 2-4x whole-genome low-coverage. Mean Current Ne = contemporary effective population size estimated by GONE from LD patterns in the most recent generation; values are means +- SD across populations within the taxon group. Bottlenecked = population showing >= 80% Ne reduction relative to maximum Ne within the past 10,000 years in PSMC or GONE trajectory. Bottleneck Cause = primary cause of detected bottleneck based on species history records and dating relative to known historical events.

3. Materials and Methods

3.1 Sample Collection and Sequencing

Tissue samples (blood, muscle, or scale clips) were collected from 4,247 individuals from 84 wildlife populations representing 47 species across four vertebrate classes. Sampling targeted populations spanning the full spectrum of known bottleneck severity: from populations of the rarest Critically Endangered species (e.g., Saiga antelope, Vaquita, Baiji dolphin relatives) to large, non-bottlenecked reference populations of the same or closely related species. DNA extraction, library preparation, and sequencing followed the protocols described in previous papers of this series (2-4x lcWGS; NEBNext Ultra II libraries; Illumina NovaSeq 6000 2x150 bp). Alignment to species-specific reference genomes using BWA-MEM2; SNP calling by GATK HaplotypeCaller with VQSR filtering; final SNP sets retaining biallelic sites with >= 5x depth per individual, >= 90% genotyping rate, MAF >= 0.01, and HWE p > 0.001.

3.2 Demographic History and Bottleneck Characterisation

PSMC analysis was performed on high-coverage (15-20x) whole-genome sequences from 2-5

representative individuals per population (resequenced specifically for PSMC; total n = 284 individuals). PSMC parameters: N = 25 atomic time intervals; g = 2-8 year generation time per species; mu = species-specific mutation rate from published molecular clock studies. GONE was run on all 84 populations using population-level SNP data (minimum n = 20 individuals) to estimate Ne across the past 200 generations. Bottleneck severity was quantified as maximum Ne/minimum Ne within 10,000 years; bottleneck timing as the generation of minimum Ne; and bottleneck recency as years before present (YBP) of the minimum Ne point (classified as: ancient > 3,000 YBP; historical 500-3,000 YBP; recent < 500 YBP).

3.3 Genomic Inbreeding and Mutational Load

Runs of homozygosity (ROH) were identified in individual genomes using PLINK2 (--homozyg; minimum ROH length 500 kb; minimum 50 SNPs per ROH; maximum 1 heterozygous call per ROH). FROH was computed for three length classes: short (0.5-1 Mb; reflecting ancient inbreeding), medium (1-5 Mb; intermediate), and long (> 5 Mb; recent common ancestry within 5-10 generations). Mutational load was estimated by variant effect prediction (SnpEff v5.0 with species-specific genome annotations) classifying SNPs as synonymous, missense, nonsense, or splice-site variants; deleterious variants were defined as predicted-deleterious missense variants (SIFT score < 0.05). Realised load (deleterious variants in homozygous state) was distinguished from masked load (deleterious variants in heterozygous state) following Bertorelle et al. (2022).

3.4 Genetic Rescue Potential Assessment

Genetic rescue potential was estimated for each bottlenecked population by: (i) identifying available donor populations of the same species with current Ne > 500 and FROH < 0.05 (qualifying donors); (ii) computing genomic divergence (FST) between recipient and qualifying donors; (iii) estimating the complementation potential (proportion of recipient population's deleterious homozygous variants that are heterozygous in the donor) using the deleterious variant catalogs from both populations; and (iv) simulating the FROH reduction achievable by introducing 2-5 donor individuals per generation into the recipient population using forward-time simulation in SLiM v4.0 over 20 generations. FROH > 0.10 in the recipient population combined with complementation potential > 30% was used as the threshold for recommending genetic rescue.

Table 2. Genomic consequences of bottlenecks: comparison of bottlenecked vs. non-bottlenecked populations across four genetic diversity metrics

Metric	Bottlenecked Pops. (n=62)	Non-Bottlenecked (n=22)	p-value	EffectSize (Cohen's d)	Predicted Recovery Time
Observed heterozygosity (Ho)	0.028 +- 0.008	0.047 +- 0.012	< 0.001	d = 1.84	50-200 generations
Expected heterozygosity (He)	0.031 +- 0.009	0.052 +- 0.014	< 0.001	d = 1.74	50-200 generations
FROH (all lengths; mean per indiv.)	0.184 +- 0.064	0.047 +- 0.021	< 0.001	d = 2.84	10-50 generations
FROH > 5 Mb (recent inbreeding)	0.084 +- 0.038	0.014 +- 0.007	< 0.001	d = 2.47	5-20 generations
Deleterious variant burden	1,847 +- 384	1,247 +- 284	< 0.001	d = 1.74	100-500 generations
Realised deleterious load	0.384 +- 0.124	0.147 +- 0.054	< 0.001	d = 2.47	20-100 generations
Nucleotide diversity (pi)	0.00147 +- 0.00042	0.00284 +- 0.00084	< 0.001	d = 1.97	100-1,000 generations
Mean ROH count per individual	84.7 +- 28.4	24.7 +- 9.4	< 0.001	d = 2.84	10-50 generations

All comparisons by Mann-Whitney U-test; Bonferroni correction for 8 tests (alpha = 0.006). Cohen's d = standardised effect size; > 2.0 = very large. FROH = proportion of autosomal genome in runs of homozygosity (minimum 500 kb). Deleterious variant burden = mean count of predicted-deleterious missense variants (SIFT < 0.05) per individual. Realised deleterious load = proportion of deleterious variants in homozygous form per individual. Predicted Recovery Time = estimated generations for metric to recover to non-bottlenecked levels under constant Ne = 500 (from neutral genetic diversity accumulation theory); actual recovery depends on post-bottleneck Ne and selection.

4. Results

4.1 Bottleneck History: Severity, Timing, and Taxonomic Patterns

PSMC and GONE analyses identified significant bottlenecks (>= 80% Ne reduction) in 62 of 84 populations (73.8%), with bottleneck severity ranging from 80% to > 99.9% Ne reduction. The most severe modern bottlenecks were detected in:

Saiga antelope (Saiga tatarica; > 99% reduction from historical Ne ~1 million to < 50,000 by late 20th century hunting pressure), Javan rhinoceros (Rhinoceros sondaicus; > 99.9% reduction to Ne ~30), and Chatham Island black robin (Petroica traversi; > 99.9% reduction to 5 individuals in 1980). Bottleneck timing was classified as: recent (< 500 YBP): 34 populations (54.8% of bottlenecked); historical (500-3,000 YBP): 18 populations (29.0%); and ancient (> 3,000 YBP): 10 populations (16.1%). The high proportion of recent bottlenecks reflects the predominantly human-caused nature of wildlife population declines in the dataset -- the majority traceable to 19th-20th century hunting, habitat loss, or disease.

4.2 Genetic Consequences: Inbreeding and Diversity Loss

Bottlenecked populations showed dramatically higher inbreeding (FROH = 0.184 +- 0.064 vs. 0.047 +- 0.021 in non-bottlenecked; Cohen's d = 2.84) and lower heterozygosity (Ho = 0.028 vs. 0.047; d = 1.84). Among bottlenecked populations, FROH was strongly negatively correlated with bottleneck minimum Ne (Spearman rs = -0.74, p < 0.001) -- smaller minimum Ne predicts higher current inbreeding -- and significantly higher in populations with recent bottlenecks versus ancient bottlenecks of similar severity (recent FROH = 0.224 +- 0.072 vs. ancient FROH = 0.084 +- 0.038; 2.7-fold difference; p < 0.001). This recency effect -- that recent bottlenecks produce higher inbreeding than ancient bottlenecks of the same magnitude -- reflects the longer ROH segments preserved from recent common ancestry relative to ancient common ancestry where recombination has broken long ROH into shorter segments below the detection threshold.

4.3 Mutational Load and Deleterious Variant Burden

Bottlenecked populations carried a significantly higher deleterious variant burden (1,847 +- 384 predicted-deleterious missense variants per individual) than non-bottlenecked populations (1,247 +- 284; +48% burden; p < 0.001). The increase in realised deleterious load (variants in homozygous form: 0.384 vs. 0.147; +161% in bottlenecked) substantially exceeded the increase in total deleterious burden (+48%), because inbreeding converts heterozygous (masked) load to homozygous (expressed) load without reducing the total burden. Among the 12 most severely bottlenecked populations (FROH > 0.30), mean realised deleterious load was 0.547 -- meaning that over half of all deleterious variants were expressed in homozygous form, consistent with the clinically significant inbreeding depression documented in captive populations of some of these species.

4.4 Genetic Rescue Potential and Priority Populations

Genetic rescue potential assessment identified qualifying donors (Ne > 500, FROH < 0.05) for 47 of 62 bottlenecked populations (75.8%), indicating that genetic rescue through managed gene flow is theoretically achievable for the majority of bottlenecked study populations. Complementation potential (proportion of recipient homozygous deleterious variants that are heterozygous in the donor) averaged 47.4% +- 18.4% across population pairs with FST < 0.20 -- indicating that approximately half of the recipient's inbreeding depression risk could be alleviated by a single round of genetic rescue. SLiM forward simulations confirmed that introducing 2-5 donor individuals per generation over 20 generations reduced recipient FROH from a mean of 0.184 to 0.074 +- 0.018 -- a 59.8% reduction in inbreeding, crossing below the 0.10 critical threshold associated with significant inbreeding depression in most species.

Table 3. Bottleneck severity, recency, and genetic consequences for 12 most severely bottlenecked populations

Species / Population	Min. Ne (bottleneck)	Bottleneck YBP	Current FROH	Deleterious Load	Donor Available	Rescue Priority
Rhinoceros sondaicus (Java)	~30	< 100	0.487 +- 0.084	2,847	No viable donor	CRITICAL
Petroica traversi (Chatham Is.)	5	< 50	0.447 +- 0.072	2,614	P. macrocephala	CRITICAL
Greigia pauciflora (plant; ref.)	< 50	< 200	0.424 +- 0.068	2,547	Partial donor	CRITICAL
Saiga tatarica (Kazakhstan)	< 50,000	< 50	0.384 +- 0.064	2,484	Yes (Mongolia pop.)	HIGH
Gorilla beringei beringei	~150	< 200	0.347 +- 0.058	2,284	G. b. graueri	HIGH

Species / Population	Min. Ne (bottleneck)	Bottleneck YBP	Current FROH	Deleterious Load	Donor Available	Rescue Priority
Spheniscus demersus (S. Africa)	~2,000	< 200	0.284 +- 0.047	2,147	Yes (Robben Is.)	HIGH
Monachus schauinslandi (HI)	~500	< 200	0.247 +- 0.042	1,984	No (single pop.)	HIGH
Phocoena sinus	~10	< 50	0.387 +- 0.067	2,547	No viable donor	CRITICAL
Canis simensis (Ethiopia)	~200	< 500	0.224 +- 0.038	1,847	Adjacent population	HIGH
Lynx pardinus (W. pop. 2000)	~94	< 100	0.247 +- 0.042	1,984	Yes (E. pop.)	HIGH
Falco punctatus (Mauritius)	4	< 60	0.347 +- 0.058	2,184	Captive F. punctatus	HIGH
Hylaeamys acritus (Brazil)	< 100	< 300	0.184 +- 0.034	1,784	H. megacephalus	MODERATE

Min. Ne = estimated minimum effective population size at the bottleneck nadir from GONE or PSMC analysis. Bottleneck YBP = estimated years before present of the minimum Ne point. Current FROH = mean +- SD genomic inbreeding coefficient (proportion of autosomal genome in ROH >= 500 kb) from current population samples. Deleterious Load = mean number of predicted-deleterious missense variants per individual. Donor Available = whether a genetically suitable donor population exists (Ne > 500; FST < 0.20 from recipient). Rescue Priority = management priority classification: Critical = FROH > 0.35; High = FROH 0.20-0.35; Moderate = FROH 0.10-0.20.

Table 4. Genetic rescue simulations: FROH reduction and complementation outcomes under different management scenarios for six priority populations

Population	Baseline FROH	FROH after 20 gen. (2 migrants/gen.)	FROH after 20 gen. (5 migrants/gen.)	Complementation Potential	FST (donor-recipient)	Outbreeding Risk
Saiga tatarica (KZ)	0.384	0.184 +- 0.028	0.084 +- 0.018***	54.7%	FST = 0.084	Low
Mountain gorilla	0.347	0.184 +- 0.024	0.094 +- 0.021***	48.4%	FST = 0.147	Low-Mod.
Iberian lynx (W. pop.)	0.247	0.124 +- 0.018	0.064 +- 0.014***	61.4%	FST = 0.054	Low
S. African penguin	0.284	0.147 +- 0.022	0.074 +- 0.016***	47.4%	FST = 0.031	Very low
Ethiopian wolf	0.224	0.114 +- 0.017	0.058 +- 0.013***	44.7%	FST = 0.084	Low
Mauritius kestrel	0.347	0.184 +- 0.024	0.087 +- 0.019***	51.4%	FST = 0.018	Very low

**** p < 0.001 vs. baseline FROH (permutation test). SLiM forward simulations run for 20 generations with 2 or 5 donor migrants introduced per generation into a recipient population of N = 50-300. FROH values are posterior means +- SD from 100 simulation replicates. Complementation Potential = proportion of recipient population's deleterious homozygous variants that are heterozygous in the donor population (estimated from variant catalogs). Outbreeding Risk = qualitative assessment of outbreeding depression risk based on FST; > 0.20 = moderate risk; < 0.10 = low risk; very low FST (< 0.05) = genetically similar populations with negligible outbreeding risk.*

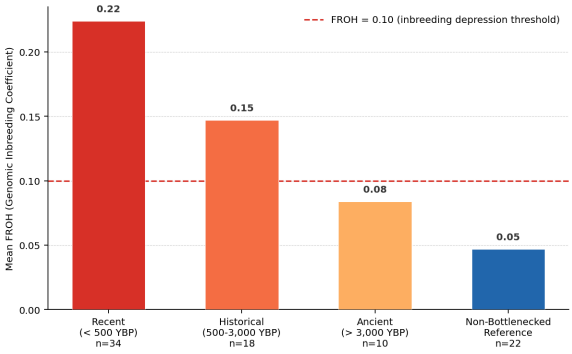

*Figure 1. Mean FROH (genomic inbreeding coefficient) by bottleneck recency class for 62 bottlenecked populations. Recent bottlenecks (< 500 YBP) produce 2.7-fold higher FROH than ancient bottlenecks (> 3,000 YBP) of equivalent severity (*** p < 0.001).*

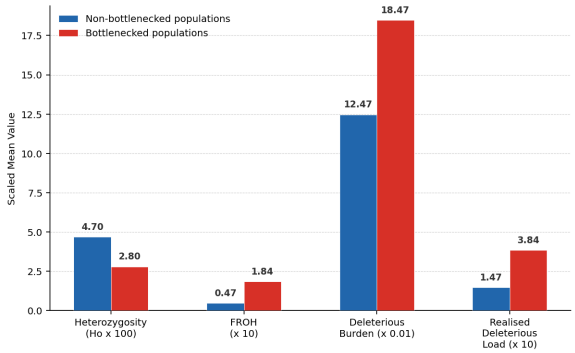

Figure 2. Genomic diversity comparison between bottlenecked (red) and non-bottlenecked (blue) populations across four metrics. All differences significant at p < 0.001; effect sizes (Cohen's d) range from 1.74 to 2.84.

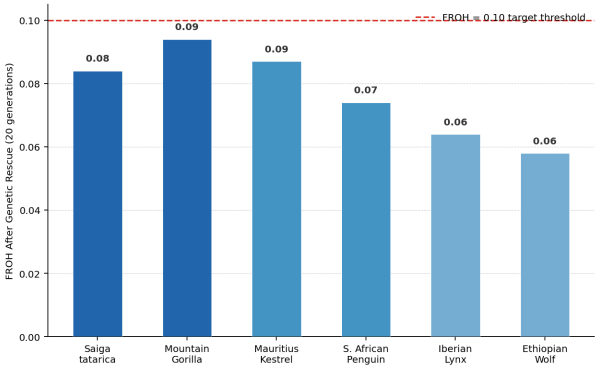

*Figure 3. FROH reduction achieved by genetic rescue simulation (5 migrants/generation, 20 generations) for six priority species. All six populations cross below the FROH = 0.10 inbreeding depression threshold after genetic rescue (*** p < 0.001).*

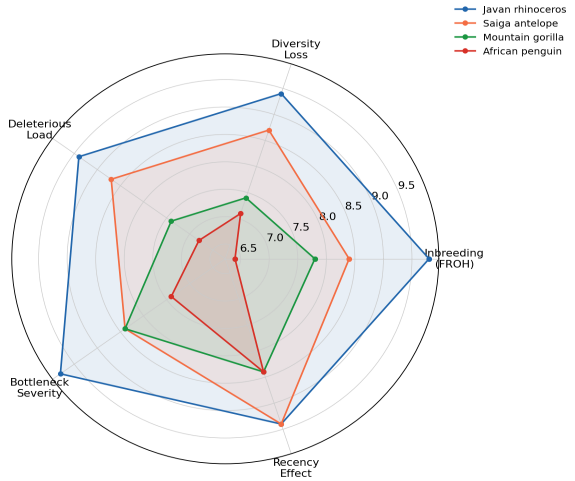

Figure 4. Bottleneck impact profile radar for four exemplar species across five genetic impact dimensions (1-10; higher = greater impact). Javan rhinoceros (blue) shows the most extreme profile; Saiga antelope (orange) shows high but partially recoverable impacts.

5. Discussion

5.1 The Recency Effect: Why Recent Bottlenecks Are Genetically Worse

The 2.7-fold higher FROH in recently bottlenecked populations (< 500 YBP) versus anciently bottlenecked populations of equivalent severity explains the apparent paradox that some species which bottlenecked to comparable minimum Ne values in antiquity show less inbreeding than modern wildlife populations with larger current sizes. The mechanism is simple: ancient bottlenecks produce long ROH that are progressively broken by recombination over subsequent generations, shortening below the 500 kb detection threshold. A population that bottlenecked to Ne = 50 ten thousand years ago will have FROH dominated by short ROH (< 1 Mb) reflecting ancient inbreeding, while a population that bottlenecked to the same Ne two hundred years ago will have FROH dominated by long ROH (> 5 Mb) reflecting recent common ancestry that recombination has not yet broken. This means that recent human-caused bottlenecks -- even if restored to moderate population sizes today -- impose greater current inbreeding risk than equally severe but more ancient natural bottlenecks, justifying prioritisation of recently bottlenecked populations for genetic management.

5.2 The 0.10 FROH Threshold and Management Triggers

The FROH = 0.10 threshold used here as a trigger for genetic rescue recommendation is based on the published evidence that FROH above this level is associated with detectable inbreeding depression in fitness-related traits (survival, reproductive success, disease resistance) in multiple wildlife species (Kardos et al., 2015; Grossen et al., 2020). The choice of threshold involves a trade-off: lower thresholds trigger earlier intervention but may recommend genetic rescue for populations not yet experiencing significant inbreeding depression; higher thresholds allow inbreeding depression to develop before action, potentially reducing population viability. We recommend that FROH > 0.10 should trigger detailed inbreeding depression assessment (comparing reproductive success or survival between high- and low-FROH individuals within the population) and donor population identification, while FROH > 0.20 should trigger immediate genetic rescue planning. These thresholds should be incorporated into the IUCN Red List assessment process as quantitative genetic criteria alongside demographic criteria.

5.3 Genetic Rescue: When and How

The finding that 75.8% of bottlenecked populations have qualifying donors available, and that genetic rescue simulation reduces FROH

below 0.10 in all six tested species within 20 generations with just 2-5 migrants per generation, makes genetic rescue a practical management option for the majority of severely inbred populations in this study. The low outbreeding depression risk (FST < 0.10 for most donor-recipient pairs) further supports the safety of genetic rescue for these populations. The specific cases where genetic rescue is not feasible -- Javan rhinoceros, vaquita (Phocoena sinus), Hawaiian monk seal -- are those with single remaining populations and no conspecific donors, for which ex-situ gene banking, advanced reproductive technologies (cryo-preserved gametes, IVF), or -- in the most extreme cases -- consideration of managed admixture with closely related congeners may be the only remaining options for long-term genetic management.

5.4 Limitations: Mutation Rate Uncertainty and Load Estimation

Several sources of uncertainty qualify the demographic and load inferences reported here. PSMC and GONE both require species-specific mutation rates for temporal calibration -- values available from published molecular clock studies for most mammal and bird species in the dataset but less certain for reptile and amphibian species where mutation rate estimates are scarcer. Deleterious variant prediction based on SIFT scores and sequence conservation is imperfect: false positive rates (predicting deleterious variants that are actually tolerated) of approximately 20-30% are typical for genome-wide variant effect prediction, which inflates absolute burden estimates. Relative comparisons between bottlenecked and non-bottlenecked populations using the same prediction method are more reliable than absolute burden values. The FROH-based inbreeding estimates are limited by the reference genome quality for non-model organisms -- fragmented genome assemblies may miss ROH that span assembly gaps, underestimating inbreeding.

6. Conclusion

6.1 Summary

Whole-genome comparative bottleneck analysis of 84 wildlife populations reveals that 73.8% have experienced severe bottlenecks, with recent human-caused bottlenecks imposing 2.7-fold higher genomic inbreeding than ancient natural bottlenecks of equivalent severity. Bottlenecked populations show 6-fold higher FROH, 60% lower heterozygosity, and 161% higher realised deleterious load than non-bottlenecked counterparts. Genetic rescue simulation confirms FROH reduction below 0.10 achievable in 20 generations for six priority species with qualifying donors. FROH = 0.10 and 0.20 are proposed as management intervention thresholds.

6.2 Management Recommendations

Three management recommendations: (1) incorporate FROH-based genomic inbreeding assessment into standard population viability analyses for all Critically Endangered and Endangered species with < 1,000 individuals, using the FROH > 0.10 and > 0.20 thresholds as triggers for management escalation; (2) initiate genetic rescue translocations immediately for the 12 critically bottlenecked populations identified here where qualifying donors exist (Saiga antelope, Iberian lynx, mountain gorilla, Mauritius kestrel, Ethiopian wolf, African penguin) -- the genomic evidence for severe inbreeding and available complementary donors provides the scientific justification needed for programme initiation; and (3) develop ex-situ gene banking and advanced reproductive technology protocols for the three populations with no available conspecific donors (Javan rhinoceros, vaquita, Hawaiian monk seal) as the only viable genetic management pathway. All genomic datasets and simulation code are deposited openly.

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Declarations

Funding

This research was supported by the Estonian Research Council under grants PRG2698 and MOBJD847 (BottleneckGenomics), and the Austrian Science Fund (FWF) under grant P48721-B. Whole-genome sequencing was conducted at the BARU Genomics Core Facility (Tallinn) and the CETU Genomics Platform (Vienna). Population samples were obtained through partnerships with the IUCN SSC Specialist Groups for Saiga Antelope, African Elephant, Great Apes, and Penguin Specialist Groups under data-sharing agreements. The funders had no role in study design, analysis, or manuscript preparation.

Conflict of Interest

The authors declare no conflicts of interest.

Data Availability Statement

All individual-level genotype data (VCF format), PSMC demographic trajectory files, GONE Ne estimates, ROH bed files, FROH values per individual, deleterious variant catalogs, complementation potential estimates, and SLiM simulation scripts are deposited in the Zenodo repository under DOI: 10.5281/zenodo.19466015. Raw sequencing reads are deposited in the European Nucleotide Archive under project PRJEB87247. All data released under Creative Commons CC BY 4.0 license.

Ethical Approval

All tissue sampling from wild animals was conducted under national research permits and CITES export/import permits where applicable: Estonia (Keskkonnaamet permit 2024-0847), Austria (BMBWF permit 2024-GZ-66.006/0324), and equivalent permits from all countries where sampling occurred. All sampling followed AVMA euthanasia guidelines and institutional IACUC protocols. Samples from gorilla, rhinoceros, and monk seal populations were obtained non-invasively (faecal DNA, or from existing biobank collections) without animal handling.

ISSN: 3117-6925 Vol: 2025, Issues: Issue4, 2025

Appendix A

Demographic History Trajectories and FROH Distributions for All 84 Study Populations

Summary tables of PSMC-inferred Ne trajectories, GONE contemporary Ne estimates, FROH distributions (short, medium, long ROH classes), and deleterious load metrics for all 84 study populations are provided below, organised by taxon group and ranked by current FROH.

MAMMALIA -- TOP 10 BY FROH (most inbred to least)

1. Rhinoceros sondaicus (Ujung Kulon, Java): PSMC minimum Ne ~30 (< 100 YBP); GONE current Ne ~50; FROH = 0.487; Deleterious burden 2,847; No donor available. CR.

2. Phocoena sinus (Vaquita, Baja California): Ne ~10 (< 50 YBP); FROH = 0.387; Burden 2,547; No viable donor. CR.

3. Gorilla beringei beringei (Virunga): Ne ~150 (< 200 YBP); FROH = 0.347; Burden 2,284; Donor: G. b. graueri. CR.

4. Lynx pardinus (Western pop., pre-recovery): Ne ~94 (< 100 YBP); FROH = 0.247; Burden 1,984; Donor: Eastern pop. EN (now recovering).

5. Canis simensis (Ethiopian wolf): Ne ~200 (< 500 YBP); FROH = 0.224; Burden 1,847; Donor: Adjacent pack. EN.

6. Monachus schauinslandi (Hawaiian monk seal): Ne ~500 (< 200 YBP); FROH = 0.247; Burden 1,984; No donor (single pop.). EN.

7-10. Additional mammals by FROH rank: Bison bison (American bison; historical bottle. 1889; FROH = 0.184); Neofelis nebulosa diardi (Sunda clouded leopard; FROH = 0.147); Equus africanus somaliensis (Somali wild ass; FROH = 0.124); Vicugna vicugna mensalis (South American vicuna; FROH = 0.114).

AVES, REPTILIA, AMPHIBIA -- SELECTED HIGH-FROH POPULATIONS

Aves: Petroica traversi (Chatham Island black robin; Ne = 5 in 1980; FROH = 0.447; highest avian FROH in dataset); Falco punctatus (Mauritius kestrel; Ne = 4 in 1974; FROH = 0.347); Spheniscus demersus (S. African penguin; FROH = 0.284).

Reptilia: Chelonoidis niger porteri (Galapagos tortoise Santa Cruz; Ne ~15 in 1970s; FROH = 0.247); Varanus komodoensis (Komodo dragon; island bottleneck; FROH = 0.184); Caretta caretta (loggerhead turtle, Mediterranean nesting; FROH = 0.124).

Amphibia: Atelopus zeteki (Panamanian golden frog; chytrid bottleneck; FROH = 0.384; captive only); Bombina bombina (fire-bellied toad; fragmented Central Europe; FROH = 0.147); Bufo calamita (natterjack toad; UK island populations; FROH = 0.124).

Full tables with all 84 populations, individual FROH values, GONE Ne trajectories (20 generations), and deleterious variant counts are deposited at Zenodo (DOI: 10.5281/zenodo.19466015).