Modelling breeding strategies against gonadal hypoplasia in Swedish Mountain Cattle

A simulation study

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Modelling breeding strategies against gonadal hypoplasia in Swedish Mountain Cattle. A simulation study

Modellering av avelsstrategier mot könskörtelhypoplasi hos svensk fjällko. En simuleringsstudie

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Abstract

Recent developments in the field of genetics have enabled the easy identification of individuals carrying known genetic defects, even in the absence of phenotypic expression. One such defect, known as gonadal hypoplasia, has affected the Swedish Mountain Cattle since the start of the 20th century. Gonadal hypoplasia is a defect characterized by small, underdeveloped gonads, often resulting in impaired reproductive function and fertility. The defect has been associated with homozygosity of a ~500 kb region on bovine chromosome 29. This region, known as the Cs_{29} allele, has also been linked to the colour-sided phenotype which is characteristic of the breed. This correlation together with earlier decisions to favour the colour-sided animals likely explains both the initial increase in gonadal hypoplasia and its continued presence within the breed today.

The aim of this study was thus to evaluate potential breeding strategies against hereditary gonadal hypoplasia to ascertain how the frequency of the disease-associated allele, and the number of hypoplastic animals can be decreased while maintaining genetic diversity of this vulnerable local breed. To achieve this, the project focused on three objectives. 1) Develop a simulation of a deleterious genetic variant under balancing selection, resembling the Cs_{29} allele in Swedish Mountain Cattle. 2) Simulate and evaluate alternative breeding strategies against gonadal hypoplasia, using phenotypic and genetic testing. 3) Evaluate the impact of the various breeding strategies on the effective population size (Ne) in the simulations.

The results show that, compared to the current policy of phenotypic selection, strategies utilising genetic information to facilitate selection against the defect appear better suited to reduce both the frequency of the deleterious allele and the frequency of affected animals. However, the results also highlight clear trade-offs between reducing the prevalence of gonadal hypoplasia and two other critical aspects: namely preserving the breed's genetic diversity and limiting the amount of genetic testing required to achieve the reduction.

Keywords: Gonadal hypoplasia, Swedish Mountain Cattle, Recessive genetic disorder, Stochastic simulations

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1. Introduction

During the early 20th century, when the Swedish Mountain Cattle was still a numerically large breed, there was an increase in a genetic defect that causes gonadal hypoplasia. Gonadal hypoplasia in the Swedish Mountain Cattle effects both genders and is characterized by reduced ovarian and testicular size, leading to decreased fertility or even infertility in severe cases.

Today the disorder is known to be associated with the Cs_{29} allele consisting of a large translocation between chromosome 6 and chromosome 29. At the time the cause of the increase of individuals displaying gonadal hypoplasia was unknown. However subsequent research has linked the translocated allele to colour-sidedness, suggesting that the increase of gonadal hypoplasia during the early 20th century was driven by the selection for white coat colour as was part of the early breeding goal.

Despite not knowing the underlying cause of the increased in gonadal hypoplasia a phenotypic control program was implemented during the 1940-50s which initially managed to decrease the frequency of the defect. Since then, however the Swedish Mountain Cattle breed has decreased in size and a key goal for breeding activities has therefore been to maintain the genetic diversity of the breed. Even though the control program for gonadal hypoplasia has been in effect since its start the frequency of the deleterious Cs_{29} allele is still relatively high within the Swedish Mountain Cattle population.

With resent advances in genetic research and technology, it is now in addition to traditional phenotypic evaluations also possible to identify affected animals and carriers of the Cs_{29} allele through DNA testing. Given these technological advancements and the current classification of the breed as vulnerable, it is of significant interest to explore how breeding strategies should be designed to reduce the frequency of affected animals while simultaneously preserving the genetic diversity of the Swedish Mountain Cattle population.

1.1 Research aim

The project aims to evaluate potential breeding strategies against hereditary gonadal hypoplasia in Swedish Mountain Cattle to ascertain how the frequency of the disease-associated allele, and the number of hypoplastic animals can be decreased while maintaining genetic diversity of this vulnerable local breed. To achieve this, the project focuses on three primary objectives.

1) Develop a simulation of a deleterious genetic variant under balancing selection in an effectively small population, resembling the Cs_{29} allele associated with gonadal hypoplasia in Swedish Mountain Cattle.

2) Simulate different potential breeding strategies against gonadal hypoplasia, using phenotypic and genetic testing, and evaluate the effect on the frequency of the risk allele and the proportion of hypoplastic animals within the Swedish Mountain Cattle population.

3) Evaluate the impact of the various breeding strategies on the effective population size (N_e) in the simulations.

2. Literature review

2.1 The Swedish Mountain Cattle

The Swedish Mountain Cattle is a traditional dairy-type cattle breed of Norden Sweden. The breed originates from the eastern landrace cattle that had been kept in the area for centuries prior to the establishment of the breed at the end of the 19th century (Svensk Fjällrasavel 2016). The Swedish Mountain Cattle and their ancestors have been shaped by traditional Nordic transhumant farming systems, involving seasonal migration between summer highland grazing areas and sheltered winter dwellings (*Svensk Fjällrasavel* n.d.a). As a result, the Swedish Mountain Cattle of today is describes as a rather small agile breed with excellent foraging behaviour well-suited to rough terrain and pasture-based resource-scarce production systems (Svensk Fjällrasavel 2016). Although the breed is sometimes confused with another native Swedish landrace called Fjällnära cattle they are in fact despite similarities in both appearance and ancestry considered as two distinct breeds.

Cows of the Swedish Mountain Cattle breed typically weigh between 400 and 450 kg, with a wither height of around 120 to 130 cm (Svensk Fjällrasavel 2016). Even though the breed is typically recognized by its mostly white coat with black, red, or grey markings, the Swedish Mountain Cattle exhibit substantial phenotypic variation in coat colour and patterning. Figure 1 illustrates the characteristic black and white colour-sided phenotype commonly seen in Swedish Mountain Cattle. However, there are also more pigmented individuals with a broader range of coat expressions found within the breed. All animals are almost exclusively polled, and this absence of horns has been a defining characteristic of the breed since it was first established (Svenska Kulliga Boskapens Förening 1922).

Figure 1. A Swedish Mountain Cattle cow displaying the characteristic black and white colour-side coat pattern. This specific animal is also homozygous for the Cs_{29} *allele.*

According to Växa’s Cattle Statistics 2025, a total of 424 cows were registered in the official milk recording scheme in 2024 under the breed category SKB, 1261 fewer than twenty years earlier (Växa 2025). The SKB category mainly includes cows of the Swedish Mountain Cattle breed, but also encompasses other native polled breeds, such as the Swedish Red Polled.

Compared to more commercial dairy breeds, the Swedish Mountain Cattle have a moderate milk yield. Although no separate production data is available for the breed alone, the average yield for the broader SKB group was in 2024 reported as 5,305 kg ECM in Växa’s official statistics (Växa 2025).

2.1.1 Breed history

The late 1800s saw the start of organized breeding in the northern parts of Sweden and in 1891 government grants and official regulations were issued to support official cattle evaluations (Nilsson 2007). Prior to this, at the end of the 19th century, the native cattle population of northern Sweden had amounted to around 400 000 animals (NordGen n.d.).

In 1893, the first breed standard for the Swedish Mountain Cattle was established (Nilsson 2007). The standard mainly dictated the breed's desired physical appearance, declaring that animals should be polled and have a predominantly white coat with sparse red or black spotting on the flanks as well as coloured ears (Svenska Kulliga Boskapens Förening 1922). This decision to favour colour-sided animals resulted in an immediate shortage of breeding bulls which up until this point had included animals with a wide range of both coat colours and patterns (Funkquist 1913; Nilsson 2007). It also allowed the Cs$_{29}$ translocation today known to be associated with both the colour-sided phenotype and gonadal hypoplasia to spread within the breed (Settergren 1962; Durkin et al. 2012; Venhoranta et al. 2013).

It is unknown exactly when the issue of hereditary gonadal hypoplasia first became apparent to breed officials but it would not be until 1935 that a 17-year long investigation launched which would later link gonadal hypoplasia to the colour-sided phenotype favoured by the 1893 breed standard (Lagerlof & Settergren 1953; Settergren 1962).

The more immediate issue in the late 19th century however was the lack of bulls conforming to the newly established breed standard (Nilsson 2007). To combat what in hindsight can be regarded as the first major genetic bottleneck of the breed, state-founded competitions referred to as "Breeding Center Competitions" were issued in 1903 (*Svensk Fjällrasavel* n.d.b). The aim of the competitions, a practise that would continue until 1939, was to incentivize farmers to select and develop more animals conforming to breed standard (Nilsson 2007). Only herds with ten or more cows of the Swedish Mountain Cattle breed with complete pedigree documentation were allowed to participate in the competitions

(Juhlin-Dannfelt 1923). Each competition took place over a two year period during which milk yield, milk fat content, feed intake, breeding records, and the sale of breeding animals were all recorded. The herds with the best performance were subsequently named "Breeding Centres" a title that came with both a monetary award and a commitment to produce purebred bulls approved by the Breeding Center Board (Juhlin-Dannfelt 1923). Line breeding was predominantly practiced during this time and consequently cow families and bull lines from herds awarded the "Breeding Centre" title had a larger genetic impact on the breed.

In 1937 preliminary results of the 1935 inquest identified gonadal hypoplasia as a hereditary defect, leading to new regulations mandating that all bulls must have two normally developed testicles to be included in the herd book and eligible for breeding (Svenska Kulliga Boskapens Förening 1938). This however was not the only challenge for the Swedish Mountain Cattle breed association in the late 1930s (*Svensk Fjällrasavel* n.d.c). Partly due to economic difficulties, the association merged with that of another landrace, the Swedish Red Polled, in 1938 effectively forming a new breed association and officially establishing the Swedish Polled breed (SKB) (ibid). In practice however, red- and white-coated animals continued to be managed separately, effectively maintaining them as two distinct breeds albeit under the same organizational framework (Nilsson 2007).

By 1943 the inquest into gonadal hypoplasia had conclusively determined that the defect was heritable, following an autosomal recessive pattern of inheritance (Lagerlof & Settergren 1953). As a result, additional regulations regarding herd book registration were introduced stipulating that, in addition to the bull itself, its dam also had to be free from hypoplasia with two normally developed ovaries for the bull to be eligible for breeding (*Svensk Fjällrasavel* n.d.b). In 1950 the regulation was further extended to also include the bull's maternal grandmother.

The next major population decline and genetic bottleneck for the Swedish Mountain Cattle arose as a result two overlapping events in the agricultural history of northern Sweden (Nilsson 2007). These included the introduction of artificial insemination in 1954 and the continuation of the agricultural rationalization during the second half of the 20th century which together fundamentally transformed the conditions for local farmers (Nilsson 2007). The political desire to make Swedish agriculture more efficient resulted in size rationalization of farms and the replacement of local breeds such as the Swedish Mountain Cattle with more high producing breeds like the Holstein and the Swedish red (ibid). Together with the access to artificial insemination the reduction of the Mountain Cattle population in the 1960s effectively centralized most breeding decisions to one single bull station (*Svensk Fjällrasavel* n.d.c).

To counteract the declining numbers of both Swedish Mountain cattle and Swedish Red Polled in the late 1960s, the SKB breed association began to allow crossbreeding (ibid). Despite considerable protests, particularly from supporters of preserving the Swedish Mountain Cattle as a distinct breed, crossbreeding with other breeds, such as Swedish Red and Swedish Holstein, officially became part of the SKB breeding program in 1973 (Nilsson 2007; *Svensk Fjällrasavel* n.d.c). Fortunately, for the Swedish Mountain Cattle, efforts to preserve genetic material by collecting and storing semen samples from AI bulls had already begun during the 1960s (*Svensk Fjällrasavel* n.d.d).

Even though semen collected from older bulls was used to manage inbreeding, by 1990 the breed's genetic base had become narrow, with most animals being closely related through a handful of influential sires (*Svensk Fjällrasavel* n.d.d). In response, *The World Wide Fund for Nature* (WWF) together with a group of dedicated farmers launched the *Save the Swedish Mountain Cattle* (PRF) initiative in 1993 (ibid). That same year, PRF successfully persuaded the SKB breed association to align the breeding objectives for the Swedish Mountain cattle with broader conservation goals (Nilsson 2007). As part of this shift, a group mating system was introduced, and PRF continued to expand the collection of semen from genetically valuable bulls, laying the foundation for a gene bank to safeguard the breed long-term. In 1995, the breed officially separated from SKB and formed its own association, Svensk Fjällrasavel. The following year, responsibility for the gene bank was transferred to the Swedish Board of Agriculture.

2.1.2 New breeding strategy established in 2016

In 2016 a new breeding goal and strategy for the Swedish mountain cattle was approved by the breeding organization Svensk Fjällrasavel (Svensk Fjällrasavel 2016). Since the start of Project Save the Swedish Mountain Cattle (PRF) in the main focus of the breeding efforts had been to maintain the genetic diversity accomplished by the project (Nilsson 2007). In 2016, however, the goal of broadening the breeds genetic base was considered fulfilled and in the new strategy the attention partially shifted towards improving production traits (*Svensk Fjällrasavel* n.d.e).

Up until this point, the development of traits such as milk yield had practically come to a standstill, as efforts to maintain genetic diversity greatly limited the possibility of selective breeding (Nilsson 2007). To strengthen the position of Swedish Mountain Cattle relative to other breeds the 2016 breeding strategy therefore sought to improve milk quality by increasing the content of milk proteins favourable for cheese-making (Svensk Fjällrasavel 2016). These include κ-casein type B, β-casein types A2 and B, and β-lactoglobulin type B, the latter being a whey protein rather than a casein protein (Svensk Fjällrasavel, 2016).

The composition of bovine milk proteins is genetically determined and varies both within and between breeds. The genes encoding κ-casein (CSN3) and β-casein (CSN2) are located on bovine chromosome 6 (BTA6), while the gene encoding β-lactoglobulin (LGB) is located on chromosome 11 (BTA11) (Caroli et al. 2009). These genes are highly polymorphic, with multiple allelic variants at each locus corresponding to specific milk protein variants such as κ-casein A or B, β-casein A1, A2, or B, and β-lactoglobulin A or B (Farrell et al. 2004).

Casein proteins are particularly important for cheese production, as they coagulate in the presence of enzymes or acids to form curds, the basis of most cheeses. Certain allelic variants, notably κ-casein type B, are associated with superior processing properties, such as better rennet coagulation and higher cheese yield (Caroli et al. 2009). Similarly, the B variant of β-lactoglobulin, although not a casein, has been linked to superior cheese-making performance compared to the A variant (Meza-Nieto et al. 2013). The effect of different variants β-casein are however, in this context, more disputed (Vigolo et al. 2023).

Compared to the Swedish Red Polled and the Swedish red (SRB) the Swedish Mountain Cattle reportedly have a notably higher frequency (71%) of the preferred κ-casein type B allelic variant (Poulsen et al. 2017). Both the functional advantages of these milk proteins in traditional alpine pasture-based dairy systems and their high allelic frequency in the breed are mentioned in the 2016 breeding strategy as key reasons for prioritizing the improvement of milk protein content and composition (Svensk Fjällrasavel 2016). Thanks to modern molecular techniques, the genetic regions responsible for these desired variants can be detected through genetic testing, permitting the selection of animals carrying the preferred genotype to breeding.

Together with the revised breeding goal, a new breeding structure was also implemented in 2016, replacing the previous group mating system (*Svensk Fjällrasavel* n.d.f). Under this new system, a limited number of “Elite AI sires” (tjurfäder) are selected annually by the breeding organisation Svensk Fjällrasavel (ibid). These bulls are then promoted for use during specific insemination periods throughout the year. Since 2019, and at the time of writing, the system has operated with only two elite AI sires per year due to the limited availability of eligible bull calves (Svensk Fjällrasavel 2021).

The elite AI sires are chosen in advance, and it is up to individual cow owners to decide whether they want to inseminate using the suggested bulls or not (*Svensk Fjällrasavel* n.d.f). Although most of the actual breeding decisions are independently made by the animal owners, a breeding committee appointed by Svensk Fjällrasavel, is responsible for identifying potential bull dams and work with the owners to ensure that they are inseminated with the most suitable bull (ibid). To be considered as bull dams, cows must be part of a herd actively involved in milk production and registered in Växa Sverige’s Kokontrollen® database (Svensk Fjällrasavel 2016).

Bull calves born from elite AI sires and cows of interest are to be reported to the breeding committee, provided they are free from gonadal hypoplasia. Hair samples are then submitted by the farmer for DNA analysis, including testing for milk protein genotypes (*Svensk Fjällrasavel* n.d.f). Based on this genetic information, along with other selection criteria, the goal is to identify 3 to 4 new potential elite AI sires annually to be purchased by VikingGenetics and later used in the breeding program (ibid).

In addition to the elite AI sires, other Swedish Mountain Cattle bulls, approximately 30 at the time of writing, are available for artificial insemination through Växa under the category "other AI bulls" (*Svensk Fjällrasavel* n.d.g). A limited number of these, as well as some of the elite AI sires, are also offered as X-sorted semen (Svensk Fjällrasavel 2022). Nevertheless, a significant proportion of all Swedish Mountain Cattle bulls used in breeding still consist of naturally mated herd bulls, for which documentation and genetic data are more limited and less accessible.

2.2 Manifestation and genetic basis of gonadal hypoplasia in Swedish mountain cattle

Gonadal hypoplasia is a defect characterized by small, underdeveloped gonads, often resulting in impaired reproductive function and fertility. Various types of gonadal hypoplasia have been observed across multiple species, with the form affecting the Swedish Mountain Cattle being hereditary and congenital (Eriksson 1943).

Among Swedish Mountain Cattle the majority, 82%, of cases of gonadal hypoplasia present as left-sided, exclusively affecting the left testicle or ovary, while right-sided and bilateral presentation only account for 3% and 15% of all cases respectively (Eriksson 1943). Similar proportions have been reported by Lagerlöf & Settergren (1953) who found that 87.1% of cases was left-sided, 4.3% right-sided, and 9.6% bilateral.

In addition to the localization the severity of the defect, with regards to the number of germ cells present, also vary among cases (Eriksson 1943; Settergren 1997). Total gonadal hypoplasia indicates a complete lack of germ cells in the affected gonads while partial cases are defined by a reduced count and or quality of germ cells compared to normally developed gonads (Settergren 1997).

Depending on both localization and severity of the defect the impact of gonadal hypoplasia among Swedish Mountain Cattle ranges from impaired fertility to sterility, with the latter being more common in severe bilateral cases (Eriksson 1943; Settergren 1964). While the underlying cause of the variation in the manifestation of the defect remains unclear, the genetic basis of hereditary gonadal hypoplasia was recently described by Venhoranta et al. (2013).

Although a candidate gene variant associated to the defect was first identified in 2013, the hereditary nature of gonadal hypoplasia had been recognized much earlier by Eriksson (1943). The findings by Eriksson (1943) were a result from the investigation into gonadal hypoplasia launched in response to the rising number of infertile and hypoplastic animals within the breed during the early 20th century (Lagerlof & Settergren 1953). Through analysis of pedigree and phenotype records, as well as controlled breeding experiments, Eriksson (1943) concluded that the gonadal hypoplasia observed in Swedish Mountain Cattle was a genetic defect with incomplete penetrance, inherited in a recessive autosomal pattern.

Penetrance, in the context of a genetic defect such as gonadal hypoplasia, refers to the probability of individuals with the defect associated genotype expressing the defective phenotype (Coleman & Tsongalis 2017). The investigation by Eriksson (1943) estimated the penetrance of gonadal hypoplasia within the Swedish Mountain Cattle breed to be 0.43 for bulls and 0.56 for cows respectively.

During the clinical examinations of cases of gonadal hypoplasia, a correlation between the defect and coat colour was noted (Settergren 1954, cited in Settergren 1962). Further investigation showed that affected Swedish Mountain cows had a higher proportion of white on the body and ears compared to cows with normal ovarian development (Settergren 1962). While Settergren's work in 1954 and 1962 only noted a general correlation between coat colour and gonadal hypoplasia, it is in fact the colour-sided phenotype that is specifically associated with the defect (Venhoranta et al. 2013).

The association between these two seemingly unrelated traits, gonadal hypoplasia and colour-sidedness, can be attributed to a shared genetic basis, namely the *KIT* gene (Durkin et al. 2012; Venhoranta et al. 2013). The *KIT* gene encodes a type III receptor protein belonging to the tyrosine kinase family which, among other functions, plays a crucial role in the survival, proliferation and migration of melanocyte precursors and primordial germ cells during embryogenesis (Nishikawa et al. 1991; Buehr et al. 1993; Venhoranta et al. 2013).

Mutations of *KIT* causes disruptions in the migration or survival of melanocyte precursors (Rawls & Johnson 2003). These precursors later differentiate into melanin-producing melanocytes and such disruption can then lead to altered coat pigmentation patterns (Sjaastad et al. 2016). Besides the colour-sided phenotype in cattle, mutations involving the *KIT* gene have been shown to be responsible for the dominant white coat colour in pigs (Moller et al. 1996) and several white coat colour variations in horses (Haase et al. 2009). Similarly, primordial germ cells give rise to spermatozoa and oocytes (Sjaastad et al. 2016). Impaired development or migration of primordial germ cells due to *KIT* mutations can thus result in underdeveloped or altered gonads and conditions such as gonadal hypoplasia in cattle (Venhoranta et al. 2013).

As previously mentioned, Venhoranta et al. (2013) were the first to identify the genetic basis of gonadal hypoplasia in Swedish Mountain Cattle, demonstrating a strong association between the defect and homozygosity for a ~500 kb region on bovine chromosome 29 (BTA29). This specific region on BTA29, called the Cs_{29} allele, had previously been linked to the colour-sided phenotype by Durkin et al. (2012). The allele is the result of a duplication and subsequent translocation of a chromosome segment, including the *KIT* gene, from bovine chromosome 6 (BTA6) to BTA29 (Durkin et al. 2012; Venhoranta et al. 2013).

Alongside Cs_{29} Durkin et al. (2012) also identified a second chromosome region on BTA6 correlated to colour-sidedness. This allele, called Cs_{6}, is the result of a duplication and translocation of a fragment of the Cs_{29} allele from BTA29 back to BTA6 and is not associated to gonadal hypoplasia (Durkin et al. 2012; Venhoranta et al. 2013).

In 1935, prior to the implementation of control measures excluding hypoplastic animals from breeding, K. J. Eriksson reported the proportion of affected animals to be around 35% and the frequency of the deleterious Cs_{29} allele to be 0.72 (Eriksson, cited in Lauvergne 1970). Although no historical estimates exist for the prevalence of the non-deleterious allele Cs_{6}, Julia Hinken, Tytti Vanhala, Marta Gòdia, Martin Johnsson, Anna M Johansson (in prep) recently estimated its frequency in the current Swedish Mountain Cattle population to be approximately 0.35. It is important to note that Julia Hinken, Tytti Vanhala, Marta Gòdia, Martin Johnsson, Anna M Johansson (in prep) could not differentiate between the number of copies present of the Cs_{6} allele and thus the estimated frequency may not be wholly representative for the actual population. The same study also reported a current frequency of 0.44 for the hypoplasia-associated Cs_{29} allele.

2.3 Conservation of local and endangered livestock breeds

Over the past century, agricultural intensification and globalization have significantly reduced both the number and diversity of traditional local livestock breeds worldwide (FAO 2007). This loss of genetic resources is not only a direct threat to biodiversity but also reduces the future capacity of global food systems to adapt to challenges, such as shifts in production conditions driven by climate change (ibid). In 2007, Sweden, along with many other countries, adopted the Global Plan of Action for Animal Genetic Resources (GPA), thereby recognizing the need and assuming responsibility for the conservation of local and endangered livestock breeds (Jordbruksverket 2023).

Locally adapted breeds often possess traits that have evolved in response to specific environmental conditions and traditional management systems (Hoffmann 2013). These traits may, among others, include resistance to diseases, efficient utilization of low quality feed and resilience to extreme climates such as heat or drought, characteristics that are all likely to become increasingly valuable as climate change alters the prerequisites for animal production globally (Naskar et al., 2012). The conservation of local livestock breeds is also essential for preserving cultural heritages and traditions as these breeds, and their associated husbandry practices, are often closely linked to and have shaped many local customs and practices (FAO 2015; Ovaska et al. 2021).

The Food and Agriculture Organization of the United Nations (FAO) risk categorization system for livestock breeds includes the following seven degrees of endangerment: *Not at risk*, *Vulnerable*, *Endangered*, *Critical*, *Maintained*, *Extinct* or *Unknown* (FAO 2013). Per the definition by Gandini et al. (2004) the degree of endangerment describe the risk that a breed will become extinct and or lose genetic diversity at a detrimental rate within a specified time period. It is primarily the population size of a breed, in terms of the number of breeding females and males, that determines its risk of endangerment (FAO 2013; White et al. 2024).

At the time of writing the Swedish Mountain Cattle is classified as *Vulnerable* (FAO n.d.). The reported population size of the Swedish Mountain Cattle varies considerably between different sources. While FAO's Domestic Animal Diversity Information System (DAD-IS) reported a total of 5,770 breeding females and 2,032 breeding males in 2021, data from the breed association Svensk Fjällrasavel, together with the Nordic Genetic Resource Center and Växa's Cattle Statistics, indicate that the actual number is much lower. These sources instead report the number of actively breeding cows of the Swedish Mountain Cattle breed to be between 400 and 600 today.

These discrepancies in reported numbers not only pose challenges for conservation planning, but they also showcase the importance of considering factors other than just census population size when assessing the risk status of a breed. One such additional aspect, which is also touched upon in FAO's guidelines regarding determining risk status, is the genetic variation of the breed (FAO 2013). While there are several ways to measure and estimate a breed's genetic variation, the most commonly used parameters to express this variation are inbreeding coefficients (F) and effective population size (N_e). The classic definition of effective population size is the number of breeding individuals in an ideal population experiencing the same rate of drift as the population of interest (Wright 1931).

Due to factors affecting real, non-ideal populations such as non-random mating and unequal sex ratio the effective population size is often smaller than the census size of a population. This is also the case for the Swedish mountain cattle as Adepoju et al. (2024) recently estimated the breeds current effective population size to 132.

In addition to its use in FAO's breed risk status assessment, effective population size can also serve as an independent indicator of a population's long-term viability and its risk of experiencing negative effects due to insufficient genetic diversity. However, there is no clear consensus regarding the minimum effective population size required to avoid adverse effects. For instance, Soulé and Wilcox (1980) proposed short- and long-term thresholds of 50 and 500, whereas Frankham et al. (2014) suggest that the effective population size should not fall below 100 and 1,000 to reduce the risk of negative effects.

Regardless of the specific thresholds, comparisons between the current effective population size of the Swedish Mountain Cattle and historic estimates provide valuable insights into how breeding decisions and structural changes in Swedish agriculture have affected the breeds genetic diversity. According to estimates by Adepoju et al. (2024), the Swedish Mountain Cattle population experienced a drastic decline approximately 20 generations, or around 100 years ago. Prior to this drop, which coincides with known historical events, the same authors estimated that the effective population size of the Swedish Mountain Cattle was around 7 349.

3. Methods

3.1 Simulations

The present study utilised stochastic simulations to model and evaluate breeding strategies against a recessive deleterious allele with incomplete penetrance under balancing selection in a population modelled after the Swedish Mountain Cattle. Simulations and subsequent analysis of simulation outcomes were conducted with R version 4.4.2 (2024-10-31 ucrt) (R Core Team 2024) using the packages AlphaSimR version 1.6.1 (Gaynor et al. 2021), GeneticsPed version 1.68.0 (Gorjanc et al. 2024) and tidyverse (Wickham et al. 2019).

AlphaSimR is a package specifically developed for stochastic simulations of complex plant and animal breeding programmes (Gaynor 2018). In the current study it was used to model a population and breeding program representative of the Swedish Mountain Cattle as well as alternative selection strategies against a deleterious variant mimicking the hypoplasia associated Cs_{29} allele.

There are several aspects of stochastic simulations that make them especially useful when studying genetic systems such as breeding programs. Unlike deterministic approaches stochastic simulations can model random processes and variables that may influence a population, such as random genetic drift and the random recombination of alleles during meiotic recombination (Ewens 2012; Hassanpour et al. 2023). Stochastic simulations are also more flexible and able to model more complex breeding structures including multiple stages of selection spanning several generations (Hassanpour et al. 2023).

However, due to the inherent random element of stochastic simulations the outcome will also vary between replicate runs of the same simulation. Consequently, results from stochastic simulations are not exact but instead convey the average outcome across multiple simulation runs, along with the observed variability between them. In five relatively recent studies where stochastic simulations were used to evaluate breeding strategies the number of simulation replicates varied, ranging from 10 to 100 between the different studies (Salimi et al. 2014; Windig & Oldenbroek 2015; Upperman et al. 2019; Ablondi et al. 2022; Martin et al. 2025). In this study, due to limitations in time and computational resources, the number of replicates was kept relatively low with only ten irritations conducted of each simulation scenario. Each simulation replicate consisted of a total of 40 discrete generations: 20 generations representing a retrospective burn-in phase, followed by 20 generations during which the alternative breeding against gonadal hypoplasia was implemented.

3.1.1 Genetic parameters used in the simulations

Haplotypes of the founder population were created with the Markovian Coalescent Simulator software, MaCS, within the AlphaSimR package using the runMacs2 function. The function utilizes pre-existing in-program population histories or user supplied variables. Typically, these variables include the historic effective population size (histNe), the current effective population size (N_e), and the number of generations between the two (histGen). When used as originally intended, the function simulates the development of the population from the historic effective population size to the current effective population size with the founder haplotypes generated representing the genetic variation at the current effective population size.

In this study however the use of the runMacs2 function was adapted to instead generate the haplotypes of the population corresponding to the historic estimate of the effective population size rather than those of the current population. This was achieved by setting both histNe and histGen to NULL and entering the historic effective population size of 7349, as estimated by Adepoju et al. (2024), in the N_e slot. The haplotypes of biallelic variants created by the Coalescent Simulator software were then used to form the genotypes of the first simulated generation consisting of 2000 individuals with an equal sex distribution. In total two chromosomes segments were simulated, each with a physical length of 1 Mbp and a genetic length of 1 cM to mimic the recombination rate observed in the cattle genome. By minimizing both the number and size of simulated chromosome segments while still being representative of the two loci of interest as well as the recombination rate of the cattle genome, computational demands and data storage requirements were minimized.

3.1.2 Deleterious allele representing Cs_{29}

After generating the founder population an allele representing Cs_{29} was randomly selected from all variants on chromosome one with an allele frequency of between 0.43 and 0.45. The frequency span was set to encompass the current frequency of the Cs_{29} allele most recently estimated to 0.44 by Julia Hinken, Tytti Vanhala, Marta Gòdia, Martin Johnsson, Anna M Johansson (in prep). Using the current frequency estimate to select a representative genetic variant in what is essentially a historical population was deemed as acceptable as the primary purpose of the first 20 simulated generations was not to reconstruct the actual historical development of the population, but rather to establish a genetically and demographically realistic starting point from which alternative strategies could then be implemented and evaluated.

In all simulations the deleterious allele was handled as pleiotropic, with a dominant effect on colour-sidedness and a recessive effect on gonadal hypoplasia. To further mimic the actual Cs_{29} allele the penetrance of the hypoplasia phenotype was set in accordance with Eriksson 1943 to 0.43 and 0.57 for males and females respectively.

3.1.3 Non-deleterious allele representing Cs_6

The variant representing the Cs_6 allele, associated with the desirable colour-sided trait but not with gonadal hypoplasia, was selected from all variants on chromosome two in the same manner as the deleterious allele. Julia Hinken, Tytti Vanhala, Marta Gòdia, Martin Johnsson, Anna M Johansson (in prep) estimated the allele frequency of Cs_6 among the Swedish mountain cattle to be 0.35. To mimic this the frequency range used in the simulation from which a representative variant was randomly select was therefore set to 0.34 to 0.36.

To date, the interaction between the Cs_6 and Cs_{29} alleles regarding their effect on the colour-sided phenotype has not yet been fully determined. In the present study both alleles were therefore assumed to have a dominant epistasis effect on coat pattern and colouration. Thus, the presence of either allele at their respective locus result in the same colour-sided phenotype, regardless of the number of copies present. As mentioned in the literature review modern frequency estimates of Cs_6 involve some amount of uncertainty due to the inability to distinguish between carriers and homozygous animals. However, as no historic frequency records are available the current frequency estimate by Julia Hinken, Tytti Vanhala, Marta Gòdia, Martin Johnsson, Anna M Johansson (in prep) was used to select a representative genetic variant even though it is reasonable to assume that the historic frequency differed from that observed in the current population.

3.1.4 Baseline selection

To establish a genetically and demographically realistic starting point from which alternative strategies could be implemented selection mimicking the current management strategy of gonadal hypoplasia in the breed, was simulated during the first 20 discrete generations, corresponding roughly to 100 years of selection. In each generation 90 bulls and a maximum 1000 cows were selected from 1000 male and female candidates respectively and randomly mated to produce approximately 2000 offspring.

Selection occurred in two steps. Fist all animals that expressed the gonadal hypoplastic phenotype were removed from the pool of selection candidates. This resembles the regulation for registration in the herdbook mandating that all animals must be examined and declared free from gonadal hypoplasia to be registered. However, to simplify the simulations, no restrictions were applied regarding the hypoplasia status of the animal's dam and maternal granddam, as is otherwise the case in the current Swedish Mountain Cattle breeding scheme.

Following the removal of affected animals, 90 bulls were selected based on sample probabilities for the nine genotypes, determined by their phenotypic expression of coat colour and pattern. Sample probabilities for colour-sided and non-colour-sided bulls were estimated using Hedrick's (Hedrick 2015) model of balancing selection on a recessive detrimental allele and are presented in appendix 1. The probabilities resemble the preference towards selection of colour-sided animals for breeding, a remnant from the 1893 breed standard. While this selection affects both sexes in the actual population the sample probability calculations were based on the assumption that selection only occurs on bulls.

The colour-sided phenotype was assumed to be identical for all individuals with at least one copy of the Cs_{29} or Cs_6 allele. Consequently, the only genotype with a lower probability of being selected were the homozygous for the wild-type allele at both loci, as animals with this genotype did not express the preferred colour-sided phenotype.

After selection the 90 bulls were then randomly divided into three groups of 50, 25 and 15 animals respectively called the low-, medium- and high- fecundity group. The group proportions were chosen to roughly reflect the number herd bulls, AI bulls and elite AI sires used in breeding within the Swedish Mountain Cattle breed each generation to give an effective population size similar to that of the current population. Each group was subsequently assigned a specified number of offspring per bull to mimic the differing fecundity between the groups.

For the low offspring group, representing herd bulls, the number of offspring per sire was sampled from a Poisson distribution with a lambda of 1, plus 1 to ensure at least one offspring per sire. Of the remaining offspring 40% were assigned to the medium offspring group representing AI bulls and 60% to the high offspring group representing elite AI sires. The average number of offspring per sire for these groups were then calculated by dividing the respective total number of offspring per group by the number of sires in each group rounding up to the nearest integer. Sires assigned to produce more than one offspring were repeated according to the number of offspring in the final list of selected sires.

Next, 1000 dams were selected from the pool of gonadal hypoplasia-free candidates using the same selection probabilities previously used for the bulls. However, unlike the bulls, individual dams were allowed to be selected more than once. This adjustment was made to account for the possibility that there may be fewer than 1000 gonadal hypoplasia-free females available for selection in some generations. The number of offspring per dam was adjusted to account for variations in the total number of offspring per generation and like the final list of sires, the IDs of dams assigned more than one offspring were repeated in the final list of dams. This list was then randomly sorted to ensure that the order of individual dams in the list was random.

Finally, mating of selected sires and dams was simulated using the makeCross2 function within the AlphaSimR package. The function generates offspring from two populations, in this case the selected sires and dams, using a user-supplied crossing plan. The cross plan was created by generating a matrix from the final lists of selected sires and dams. The mating pairs were assigned randomly due to the randomized sequential order of the final list of dams and there were no restrictions preventing the mating of closely related individuals, such as full or half siblings.

Validation Baseline scenario

Before constructing alternative selection scenarios, the *Baseline* simulation was validated to ensure consistency with the current phenotypic management strategy for gonadal hypoplasia and its impact on the actual Swedish Mountain Cattle population. For this, effective population size (N_e) was chosen as a validation metric.

The goal was to obtain an average effective population size of the simulated population in generation 21-40, across ten replicates, close to that of the actual population ($N_e \sim 136$). Simulation parameters that were adjusted to achieve this were, the total number of bulls selected for breeding, the distribution of the selected bulls across the three fecundity groups as well as the allotment of offspring between these three groups.

Since the runMacs2 function was not used as originally intended to simulate a decline from a historical N_e to the present, it did not generate a demographic bottleneck by itself. Therefore, it was necessary to manually ensure that the applied selection regime led to a realistic loss of genetic variation and an N_e reflecting that of the current mountain cattle population. Outlined in the previous section is the version of the *Baseline* scenario resulting in the most realistic N_e across ten replicates of 137 ± 12.5.

3.1.5 Selection scenarios for genetic management of gonadal hypoplasia

In total, six alternative selection scenarios aiming to reduce the frequency of both the deleterious allele and the frequency of affected animals was simulated. Initially, baseline selection as described above, was applied across all scenarios during the first 20 generations to establish a genetically and demographically stable starting population. Following this burn-in phase, each alternative selection strategy was then implemented from generation 21 to 40. Presented below are all alternative scenarios that were tested, including a continuation of the current strategy. In scenarios where genetic testing was applied, it was assumed to be completely accurate and error-free.

Scenario 1, Baseline

In the *Baseline* scenario the baseline selection, as outlined in the previous section, was implemented across all 40 generations representing what would be a continuation of the current management of the Cs_{29} allele within the Swedish Mountain Cattle breeding program. A schematic overview of the baseline selection scenario is provided in Appendix 3

Scenario 2, Pre-test homozygous excluded

Genetic testing of bulls prior to selection to exclude those homozygous for the deleterious allele. Under this scenario, including four sub scenarios, 25%, 50%, 75%, and 100% of all unaffected bulls in each generation were selected for genetic testing to identify those homozygous for the deleterious allele. The identified homozygous bulls were subsequently removed from the pool of breeding candidates before 90 bulls were selected for breeding using the same sample probabilities as described in the *Baseline* scenario. Division of selected bulls into fecundity groups, allocation of offspring, selection of dams as well as mating to produce offspring was carried out as described in the baseline scenario.

Scenario 3, Pre-test carriers excluded

Genetic testing of bulls prior to selection to exclude carriers of the deleterious allele. The procedure was identical to that described in *Scenario 2*, with genetic testing of varying proportions of unaffected bulls per generation in four distinct sub-scenarios. However, in this third scenario both homozygous bulls and carriers of the deleterious allele were excluded from breeding prior to selection and mating. Both scenario 2 and 3 corresponds to scenarios were bull owners collect and submit DNA samples so that the genotype at the deleterious locus of either 25%, 50%, 75% or 100% of all unaffected bulls in each generation are known. The genetic information is then used to exclude either homozygous or carrier bulls from breeding but does otherwise not affect the selection. For a schematic summary of scenario 2 and 3, see Appendix 4.

Scenario 4, Post-test AI bulls homozygous excluded

Genetic testing of all bulls in the medium and high fecundity groups following selection to exclude those homozygous for the deleterious allele from breeding. Under this scenario 90 bulls where selected and divided into fecundity groups as described in the *Baseline* scenario. Following this all 40 bulls belonging to either the medium or high fecundity group were genetically tested and all individuals homozygous for the deleterious allele were excluded from breeding without replacement. Despite this, the medium and high fecundity groups still received the same proportion of the total number of offspring as in the *Baseline* scenario. Consequently, the number of offspring per remaining bull in these groups could differ from those in the *Baseline* scenario depending on the number of homozygous bulls in each generation.

Scenario 5, Post-test AI bulls carriers excluded

Genetic testing of all bulls in the medium and high fecundity groups following selection to exclude carriers of the deleterious allele from breeding. The procedure was identical to that of scenario 4, except that both homozygous bulls and carriers of the deleterious allele in the medium and high fecundity groups were excluded from breeding. This exclusion had the same potential consequences on the number of offspring per remaining bull in these groups as in the previous scenario. In contrast to the prior scenarios, both scenario 4 and 5 represents scenarios where genetic testing is not carried out by bull owners but instead by the breeding companies targeting only animals intended for use in artificial insemination following the initial selection. Appendix 5 contains a schematic representation summarizing the selection implemented in scenario 5.

Scenario 6, Post-test AI bulls carriers excluded with exemptions

Scenario 6 was developed based on the preliminary analysis of results from scenarios 1–5 and represents a refined version the 5th scenario *Post-test AI bulls carriers excluded*. Scenario 5 was particularly interesting to expand upon because it both effectively reduced the frequency of the deleterious allele while simultaneously being feasible in terms of genetic testing efforts. However, it also had the most detrimental effect on inbreeding, resulting in an effective population size of under 100.

Scenario 6 was developed in an attempt to maintain the effective reduction in frequency of the deleterious allele from scenario 5 while mitigating the decrease in N_e. It was theorised that inbreeding could be diminished by exempting a few bulls from the high fecundity from the criterion regarding genotype at the Cs_{29} locus. In total two sub-scenarios, *Post-test AI bulls carriers 1 exempt* and *Post-test AI bulls carriers 5 exempt*, were simulated each allowing either one or five Elit AI sires per generation to breed regardless of if they carried the deleterious allele or not.

In both sub-scenarios selection and division of bulls in to three fecundity groups was conducted in the same manner as in scenario 5. However, prior to genotyping the medium and high fecundity groups either one or five of the 15 Elit AI Sires was randomly selected and allowed to breed regardless of genotype. All remaining bulls in the two groups were subsequently genetically tested as described in scenario 5 and those found to carry the deleterious allele were removed without replacement. Distribution of offspring, selection of dams as well as mating to produce offspring was then conducted like in all other scenarios.

In line with the previous scenario, responsibility for conducting necessary genetic testing in *Post-test AI bulls carriers excluded with exemptions* falls on the breeding companies. Although, exempted Elit AI sires were chosen at random in the simulations scenario 6 represents a situation wherein breeding companies could select bulls to exempt based on other desirable traits for breeding despite their hypoplasia genotype.

3.1.6 Simulation data collection

In each replicate of all scenarios the data variables presented below in table 1 were collected.

Table 1. Data variables collected.

Variable	Description
Pedigree	Pedigree information, including each individual's ID as well as the ID of their sire and dam, was recorded using the setTrackPed function in AlphaSimR. Pedigree information was later used to calculate inbreeding coefficients and effective population size.
Deleterious allele frequency	The frequency of the deleterious allele was calculated at the end of each generation based on genotype data extracted from chromosome 1 using the pullSegSiteGeno function in AlphaSimR.
Non-deleterious allele frequency	The frequency of the non-deleterious allele was calculated at the end of each generation using genotype data extracted from chromosome 2 via the pullSegSiteGeno function in AlphaSimR.
Affected status	Binary data indicating whether each individual expressed the hypoplastic phenotype (TRUE = hypoplastic; FALSE = unaffected). This variable was recorded for all animals in each generation.
Generation data	Data organized by generation including all individuals belonging to each generation, their unique ID, sex and parental identity (dam and sire ID).
Offspring number	Data organized by generation including the number of offspring generated each generation.

3.1.7 Calculation of inbreeding coefficients and effective population size

In the current study effective population size was calculated from pedigree data using a method based on inbreeding rate between two successive generations. To facilitate a later comparison of each selection scenarios impact on N_e, calculations were focused on generation 21 to 40 when alternative selection was implemented.

To start, inbreeding coefficients of all animals in the ten replicates of each scenario were calculated using the inbreeding function within the Genetics ped package. By adding information regarding generation affiliation off all individuals the average inbreeding coefficient, $\bar{F}_t$, of each generation (t) was then computed. Next, equation 1 was used to obtain the average inbreeding rate of each generation ($\Delta\bar{F}_t$). The mean inbreeding rate per generation across gen 21-40, $\Delta\bar{F}_{t_{21-40}}$, of the respective selection scenario was then calculated as described in equation 2. Lastly, using equation 3, the average effective population size of each scenario was calculated.

$$\Delta\bar{F}_t = \frac{\bar{F}_t - \bar{F}_{t-1}}{1 - \bar{F}_{t-1}} \tag{1}$$

$$\Delta\bar{F}_{t_{21-40}} = \frac{1}{20}\sum_{t=21}^{40} \Delta\bar{F}_t \tag{2}$$

$$N_e = \frac{1}{2 \times \Delta\bar{F}} \tag{3}$$

4. Results

Simulation outcomes were evaluated to assess the effectiveness of the various selection scenarios in reducing the frequency of the deleterious allele associated with gonadal hypoplasia in Swedish Mountain Cattle, while preserving genetic diversity. Scenario performance was assessed based on changes in, deleterious allele frequency, frequency of hypoplastic individuals, effective population size, and the average number of gene tests conducted per generation.

Results are presented by outcome metric, beginning with the frequency of the deleterious allele, followed by the frequency of affected animals, effective population size, and lastly the gene testing effort needed. Table 2 presented below provides an overview of the average outcome of the first three of the aforementioned metrics in all six selection scenarios including sub-scenarios. Data regarding the frequency of the non-deleterious allele associated to the colour-sided phenotype was obtained in a similar manner as for the deleterious allele and the resulting allele frequencies in generation 40 are summarized in appendix 2.

Table 2. Average frequency of deleterious allele and affected animals in generation 40 (Freq. ± SD) and average effective population size (Ne ± SD) across generations 21 to 40

Scenario	Freq. del. ± SD	Freq. aff ± SD	Ne ± SD
Baseline	0.392 ± 0.131	0.0875 ± 0.0526	137 ± 12.5
Pre-test 25% homozygous	0.282 ± 0.076	0.0399 ± 0.0231	136 ± 9.23
Pre-test 25% carriers	0.201 ± 0.094	0.0250 ± 0.0219	133 ± 16.9
Pre-test 50% homozygous	0.252 ± 0.094	0.0366 ± 0.0262	138 ± 10.3
Pre-test 50% carriers	0.090 ± 0.076	0.0039 ± 0.0052	137 ± 17.0
Pre-test 75% homozygous	0.256 ± 0.087	0.0332 ± 0.0192	134 ± 10.5
Pre-test 75% carriers	0.068 ± 0.074	0.0028 ± 0.0043	144 ± 19.0
Pre-test 100% homozygous	0.225 ± 0.095	0.0278 ± 0.0195	131 ± 9.94
Pre-test 100% carriers	0.020 ± 0.047	0.0000 ± 0.0000	138 ± 20.3
Post-test AI bulls homozygous	0.270 ± 0.070	0.0387 ± 0.0207	125 ± 9.69
Post-test AI bulls carriers	0.028 ± 0.041	0.0003 ± 0.0008	91.3 ± 16.1
Post-test AI bulls carriers 1 ex.	0.045 ± 0.057	0.0016 ± 0.0025	93.1 ± 16.1
Post-test AI bulls carriers 5 ex.	0.064 ± 0.069	0.0038 ± 0.0063	94.5 ± 16.9

4.1 Frequency of deleterious allele

All simulated scenarios led to a reduction in average frequency of the deleterious allele, but the effect was generally larger when carriers were removed from breeding compared to the corresponding scenario removing only homozygous bulls. Figure 2 provides an overview of the trajectory of the average allele frequency across 40 generations for all simulated scenarios. Variation in the frequency of the deleterious allele was observed both within and between the different selection scenarios. The standard deviation of the average allele frequency in generation 40 across replicates was, bar one exception, higher in scenarios where homozygous bulls were removed from breeding, indicating greater between-replicate variation in the final generation in these scenarios.

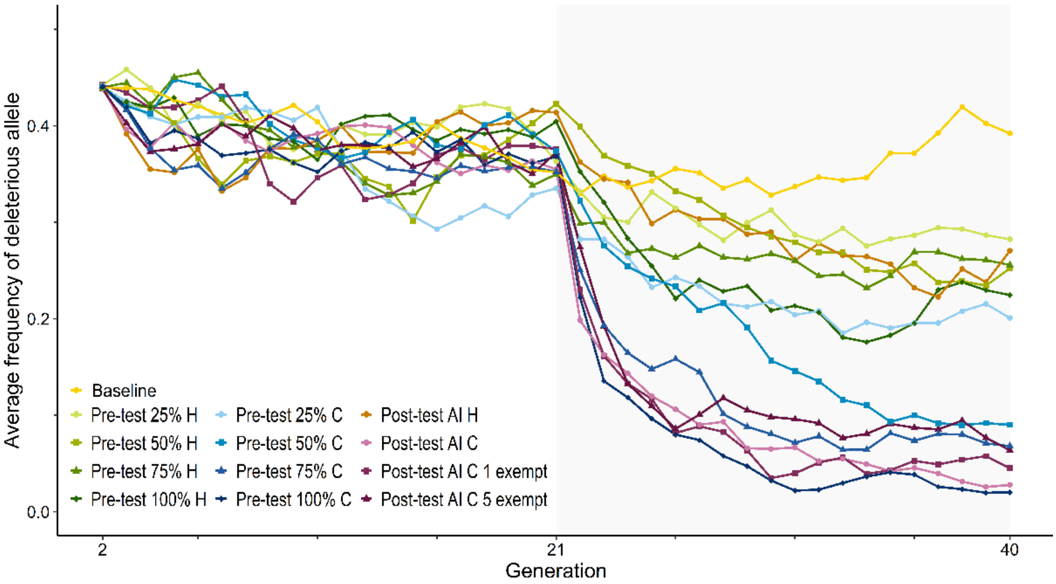

Figure 2. Average frequency of deleterious allele across all simulated scenarios. The grey background marks the period (generation 21-40) when alternative breeding strategies were implemented in all scenarios except the Baseline scenario.

The *Baseline* scenario, mimicking a continuation the current gonadal hypoplasia management strategy, was the least effective in reducing the frequency of the deleterious allele, resulting in an average allele frequency of 0.392 ± 0.131 in generation 40 (Table 2). Figure 3 shows the frequency of the deleterious allele in all ten replicates of the *Baseline* scenario over 40 generations. The results show a considerable variation in the frequency of the deleterious allele between the different simulation replicates, which may be attributed to genetic drift. From Table 2, it can be seen that the variation between replicates was also numerically largest for the *Baseline* scenario, with a standard deviation of ± 0.131 in generation 40. Despite this the average frequency of the deleterious allele appears relatively stable over time, suggesting that the population may be approaching a state of genetic equilibrium.

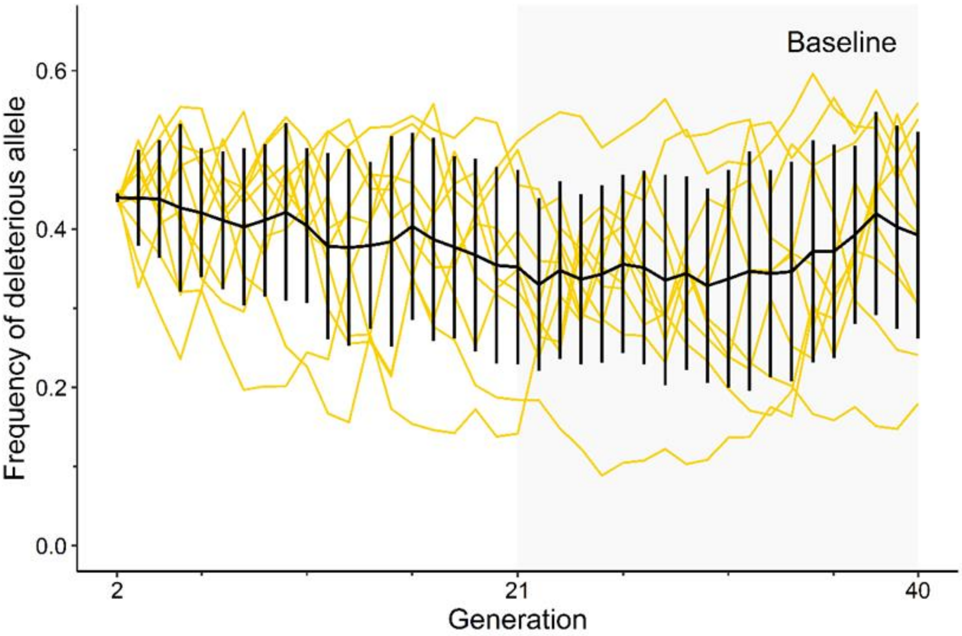

Figure 3. Frequency of deleterious allele across 40 generations in scenario 1, Baseline. The black line represents the mean frequency across ten replicates, with error bars indicating the standard deviation. Yellow lines illustrate individual replicates. The grey background marks the period (generation 21-40) when alternative breeding strategies were implemented in all scenarios except the Baseline.

In contrast to the *Baseline* scenario, all *Pre-test Homozygous Excluded* sub-scenarios exhibit a decline in the frequency of the deleterious allele after generation 21, although to varying degrees. The results of the four sub-scenarios where 25%, 50%, 75%, and 100% of unaffected bulls were genetically tested are depicted in Figure 4.

Unsurprisingly testing all unaffected bulls to exclude those homozygous for the deleterious allele resulted in the largest reduction in allele frequency among the sub-scenarios while testing 25% had the least effect on the frequency of the deleterious allele. In terms of reduction of the deleterious allele, testing either 50% or 75% of unaffected bulls fell between the extremes of 100% and 25% testing. While the two intermediate scenarios resulted in similar average allele frequencies after 40 generations, 50% testing showed a slightly larger and more consistent decline compared to testing 75% of the unaffected bulls (Figure 4). The between replicate variation in generation 40, expressed as the standard deviation of each respective mean, was highest when all unaffected bulls were tested followed by testing 50%, 75% and 25% in descending order (Table 2).

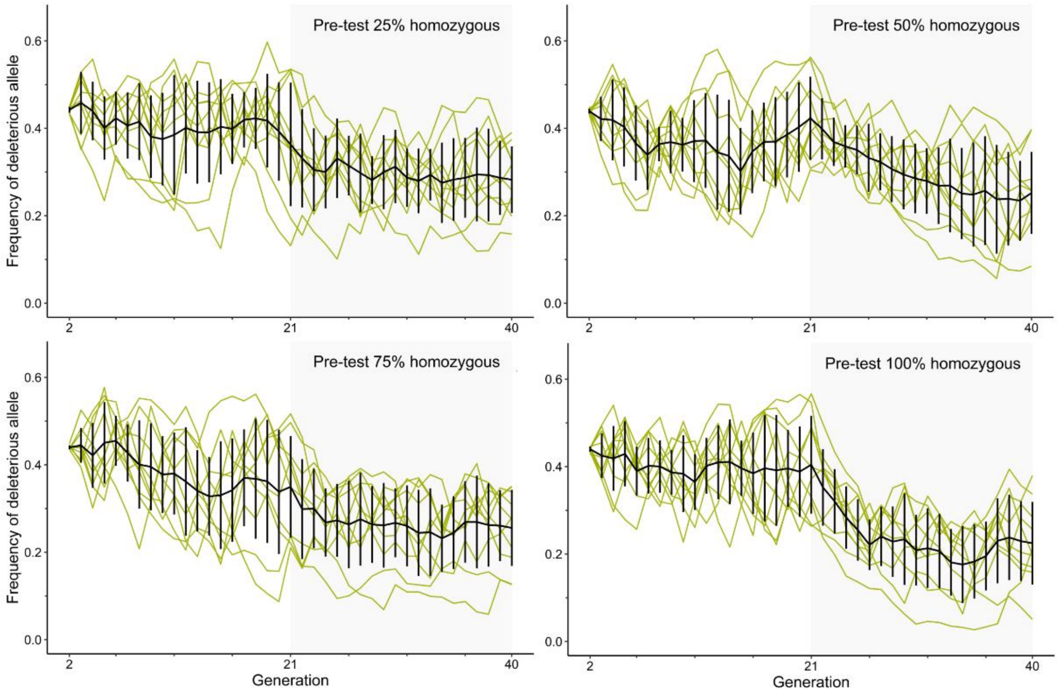

Figure 4. Frequency of the deleterious allele across 40 generations in scenario 2, Pre-test Homozygous Excluded. The figure includes graphs for each sub scenario. The black line represents the mean frequency across ten replicates, with error bars indicating the standard deviation. Green lines illustrate individual replicates. The grey background marks the period (generations 21-40) when the Pre-test Homozygous Excluded selection was implemented.

Similar to scenario 2, the third scenario, *Pre-test carriers excluded*, involved testing varying proportions of unaffected bulls. However, unlike Scenario 2, this approach excluded not only homozygous individuals but also carriers of the deleterious allele from breeding. As shown in Figure 2 and Table 2, testing all unaffected bulls to remove carriers of the deleterious allele was the sub-scenario with the largest and most consistent reduction of the deleterious allele across all simulated scenarios. Like the prior scenario all *Pre-test carriers excluded* sub-scenarios showed a decline in the frequency of the deleterious allele starting at generation 21 (Figure 5). However, unlike in the previous scenario, all four sub-scenarios followed a consistent trend where in a larger proportion of tested bulls result in a larger reduction in the frequency of the deleterious allele.

When comparing the four *Pre-test carriers excluded* sub-scenarios, the second most effective in reducing the average frequency of the deleterious allele, after 100% testing, was to test 75% of unaffected bulls, followed by testing 50% and 25% respectively (Table 2). Although *Pre-test 25% carriers excluded*, by this measure, was the least effective among scenario 3 it still achieved a greater reduction in the allele frequency than all levels of testing in scenario 2 where homozygous were removed rather than carriers (Table 2).

Across scenario 3 the between replicate variation in the last generation was lowest when all unaffected bulls were tested to remove carriers followed by the sub-scenarios testing 75%, 50% and 25% of unaffected bulls respectively (Table 2). In pairwise comparison of scenarios 2 and 3 the between replicate variation in generation 40 was higher when removing homozygous bulls compared to the corresponding sub-scenario excluding carriers in all cases except one. The exception being genetic testing of 25% of unaffected bulls where the variation in the last generation was higher when removing carriers rather than homozygous bulls (Table 2).

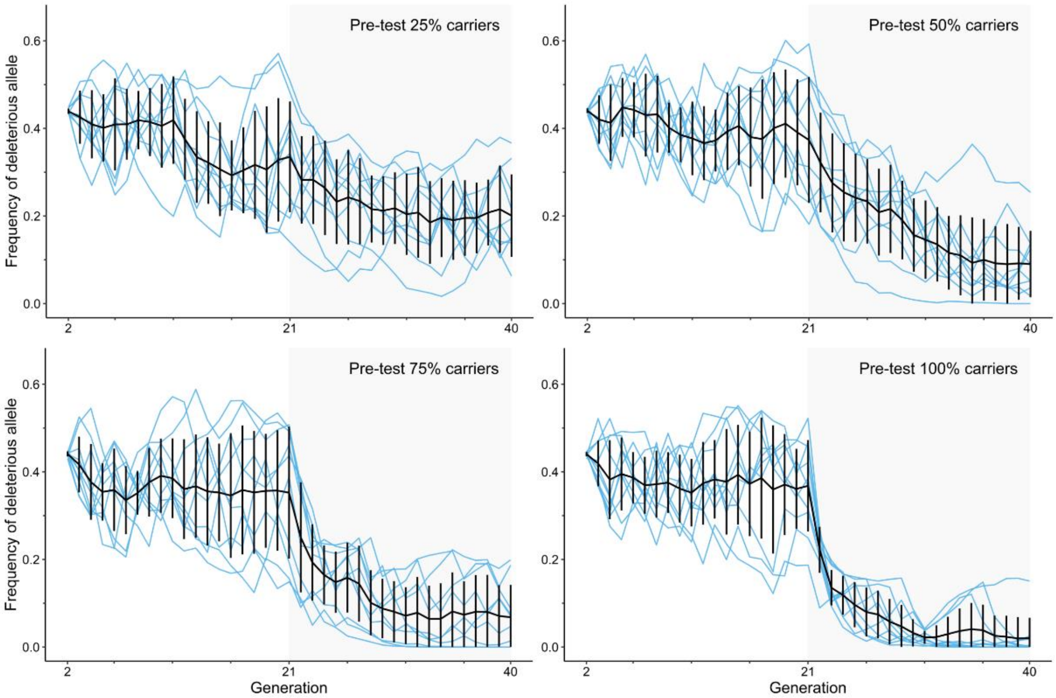

Figure 5. Frequency of deleterious allele across 40 generations in scenario 3, Pre-test Carriers Excluded. The figure includes graphs for each sub-scenario. The black line represents the mean frequency across ten replicates, with error bars indicating the standard deviation. Blue lines illustrate individual replicates. The grey background marks the period (generations 21-40) when the Pre-test Carriers Excluded selection was implemented. Negative error bars are trimmed when the average allele frequency is low to avoid impossible negative values.

Out of all scenarios involving genetic testing after selection, Scenario 4, *Post-test AI bulls homozygous excluded,* exhibited the least reduction in frequency of the deleterious allele (Figure 6). As shown in figure 2 the trajectory of the allele frequency of *Post-test AI bulls homozygous excluded* was instead more similar to that observed from *Pre-test 25% Homozygous Excluded* and both scenarios also resulted in an average final allele frequency and between replicate variation of comparable magnitude (Table 2).

In line with previous comparisons, excluding carriers, as in scenario 5 *Post-test AI bulls carriers excluded*, resulted in a lower final frequency of the deleterious allele compared to only excluding homozygous bulls, as was done in the otherwise comparable Scenario 4. *Post-test AI bulls carriers excluded,* in fact *resulted* in the largest and most consistent decline in frequency of the deleterious allele out of the post-test scenarios (Figure 5). The allele frequency trajectory of *Post-test AI bulls carriers excluded,* resembled that previously observed in Scenario 3 (Figure 4) when testing 100% of the unaffected bulls to remove all carriers. When compared across all scenarios, Scenario 5 was the second most effective strategy in reducing the frequency of the deleterious allele but had the lowest between replicate variation in the final generation out of all six simulated scenarios (Table 2).

Unsurprisingly both *Post-test AI bulls carriers excluded with exemptions* sub-scenarios exhibited a decline in the frequency of the deleterious allele similar to that of Scenario 5, although both slightly less steep and consistent (Figure 5). Of the two sub-scenarios, exempting 1 elite AI sire from selection against carriers resulted in both a lower final allele frequency and between replicate variation then when 5 elite AI sires were exempt (Table 2). Compared across all simulated scenarios *Post-test AI bulls carriers excluded 1 exempt* displayed the third lowest final frequency of the deleterious allele (0.045 ± 0.057) while *Post-test AI bulls carriers excluded 5 exempt* with an allele frequency of 0.064 ± 0.069 in generation 40 placed fifth in this regard.

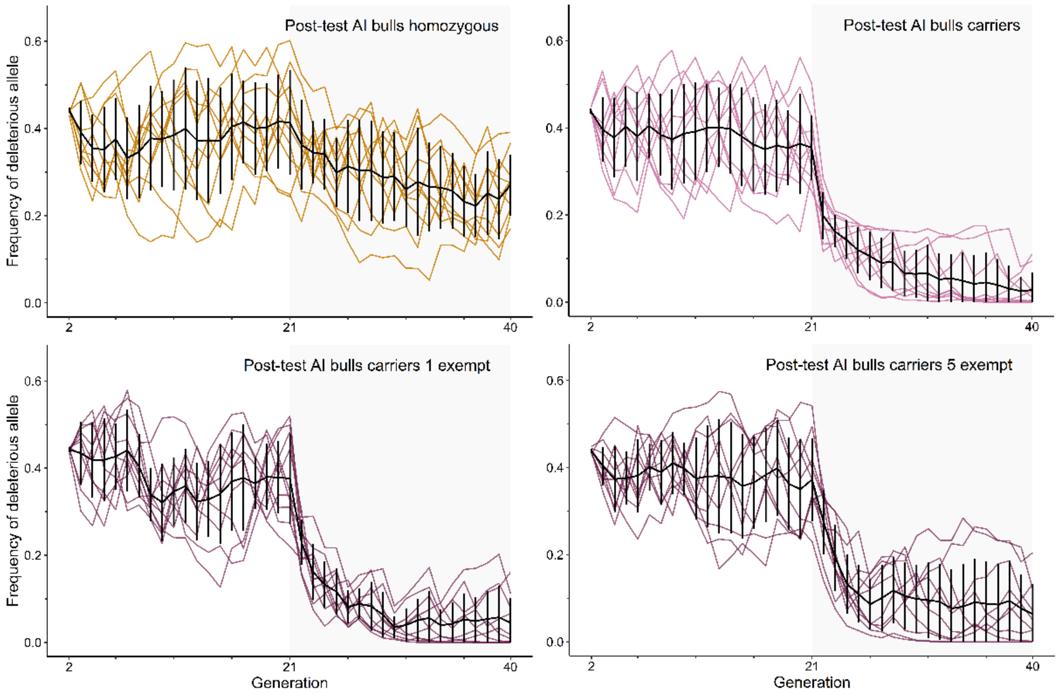

Figure 6. Frequency of deleterious allele across 40 generations in all post selection gene testing scenarios. The black line represents the mean frequency across ten replicates,

with error bars indicating the standard deviation. The coloured lines illustrate individual replicates: Orange for Scenario 4, Light Purple for Scenario 5, and Dark Purple for Scenario 6. The grey background marks the period (generations 21-40) when the different selection scenarios were implemented. Negative error bars are trimmed when the average allele frequency is low to avoid impossible negative values.

4.2 Frequency of hypoplastic animals

All simulated scenarios led to a decline in the frequency of affected animal per generation. In line with the previous outcome metric, scenarios removing carrier bulls from breeding led to a steeper and larger decline in frequency of affected animals compared to the corresponding scenario focused on selection against homozygous bulls. Figure 7 provides an overview of this decline across 40 generations for all simulated scenarios while Table 2 shows the frequency of affected animals in the last generation. As illustrated in table 2 variation, expressed as standard deviation of the mean, in the frequency of affected animal in generation 40 existed both between different scenarios as well as among the replicates of the same scenario. When comparing otherwise equivalent scenarios and sub-scenarios excluding homozygous bulls from breeding generally exhibited a larger between replicate variation in the final generation compared to when carriers were removed. It should be noted that while the average frequency of affected animals in generation 40 differed across scenarios, many exhibited substantial overlap of their standard deviation. In fact, all scenarios except *Pre-test 100% Carriers Excluded* fell within the standard deviation of at least one other scenario (Table 2).

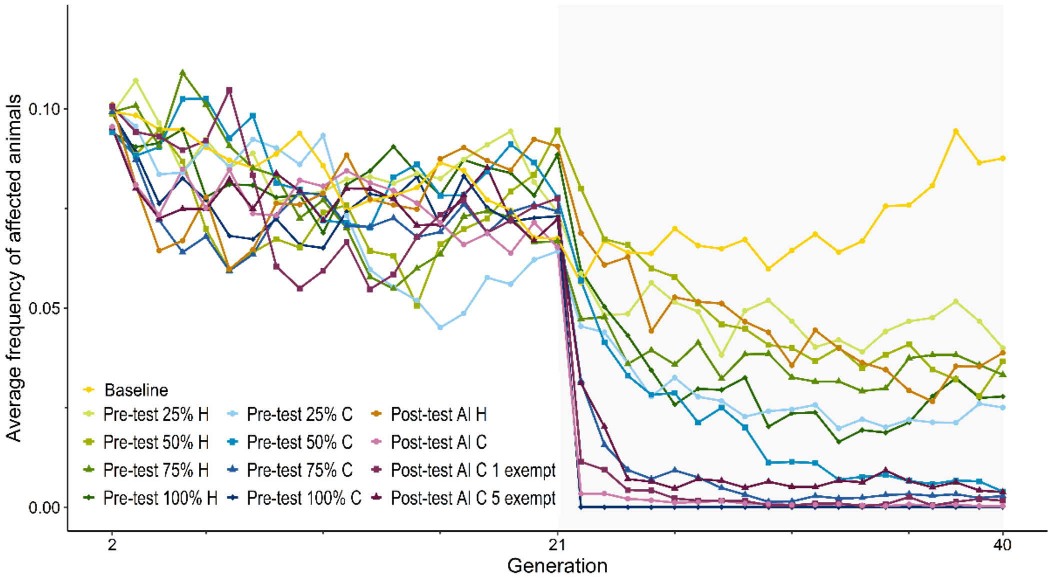

Figure 7. Average frequency of affected animals across all simulated scenarios. The grey background marks the period (generation 21-40) when alternative breeding strategies were implemented in all scenarios except the Baseline scenario.

To no surprise the *Baseline* scenario resulted in the least decline out of all scenarios, with an average of 8.75 % ± 5.26 % affected animals in the last generation (Table 2). As illustrated by Figure 8 there was a considerable between replicate variation in this scenario, however none of the replicates reached an affected frequency of zero in any generation. Despite the *Baseline* scenario displaying the largest between replicate variation across all scenarios in the last generation, as is made evident by Table 2, the average frequency of affected animals appears relatively stable over time with only a slight decline.

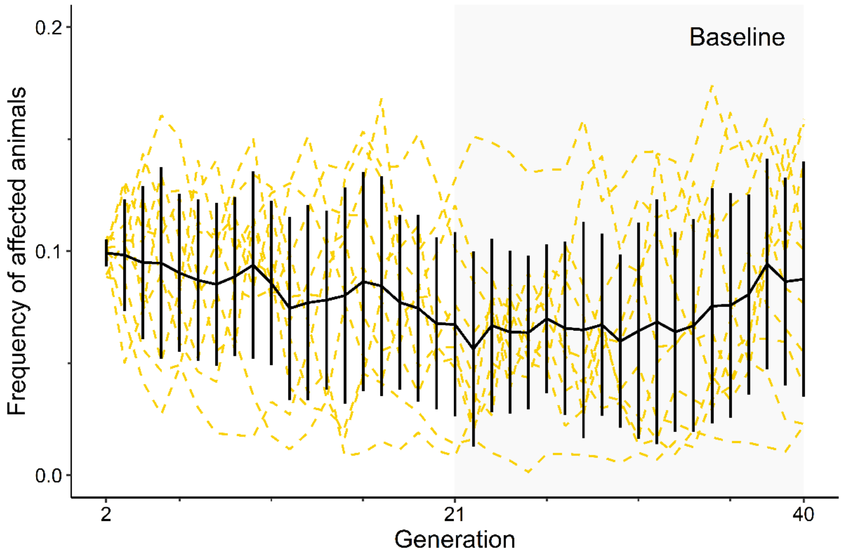

Figure 8. Average frequency of affected animals across 40 generations in scenario 1, Baseline. The black line represents the mean frequency across ten replicates, with error bars indicating the standard deviation. Yellow lines illustrate individual replicates. The grey background marks the period (generation 21-40) when alternative breeding strategies were implemented in all scenarios except the Baseline.

Compared to the *Baseline* scenario, *Pre-test Homozygous Excluded* displayed a reduction of affected animals after generation 21 with all four sub-scenarios resulting in an affected percentage of around 3-4% in the last generation. Figure 9 provides a display of the frequency trajectory in each sub-scenario.

Among the sub-scenarios the two with the lowest and highest final allele frequencies, respectively, *Pre-test 100% Homozygous Excluded* and *Pre-test 25% Homozygous Excluded,* also exhibited the lowest and highest frequencies of affected animals in the final generation (Table 2). Surprisingly, although *Pre-test 50% Homozygous Excluded* led to a lower final deleterious allele frequency than *Pre-test 75% Homozygous Excluded*, the proportion of affected animals in generation 40 was slightly higher in the first of the two intermediate scenarios, (Table 2). However out of the two, the decline in frequency was still more consistent when testing 50% of unaffected bulls (Figure 9). Similarly to the

Baseline scenario no replicate in either of the four *Pre-test Homozygous Excluded* sub-scenarios reached an affected frequency of zero (Figure 9).

The between replicate variation in generation 40 across the four sub-scenarios did not reflect their ranking based on average frequency of affected animals. Instead, testing 50% resulted in the largest between replicate variation followed by 25%, 100% and 75% in descending order, with the two latter being close to equal (Table 2).

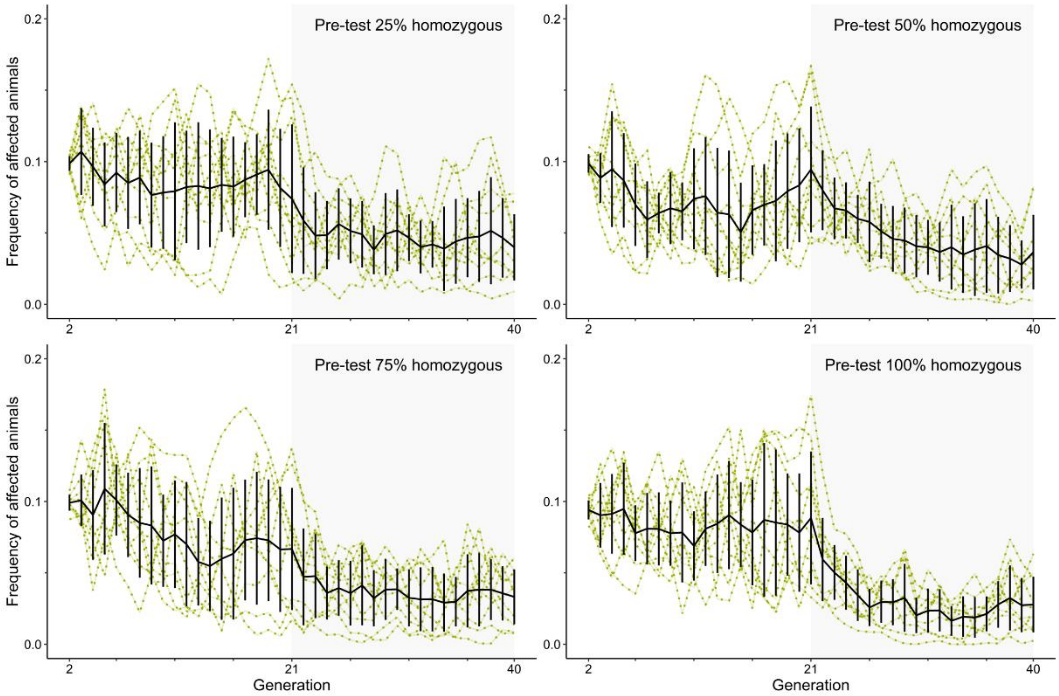

Figure 9. Frequency of affected animals across 40 generations in scenario 2 (Pre-test Homozygous Excluded). The figure includes graphs for each sub scenario. The black line represents the mean frequency across ten replicates, with error bars indicating the standard deviation. Green lines illustrate individual replicates. The grey background marks the period (generations 21-40) when the Pre-test Homozygous Excluded selection was implemented.

Pre-test Carriers Excluded and more specifically its sub-scenario Pre-test 100% Carriers Excluded was the only one out of all simulated scenarios to achieve an affected frequency of zero both on average and in all 10 replicates. As can be seen from figure 10 below this drop also occurred almost immediately after the implementation of the selection. While none of the remaining three sub-scenarios manged to reach an average frequency of zero both Pre-test 50% Carriers Excluded and Pre-test 75% Carriers Excluded included specific replicates that did (Figure 10). More replicates reached zero in the Pre-test 75% sub-scenario and those that did also generally did so after fewer generations compared to Pre-test 50%. Consequently, Pre-test 75% Carriers Excluded was, after Pre-test 100% Carriers Excluded, the second most effective of the four sub-scenarios in reducing the frequency of affected animals, resulting in both a steeper and more consistent decline in frequency compared to Pre-test 50% Carriers Excluded (Figure 10). Testing 25% of unaffected bulls to remove carriers did not decline to a similar degree resulting in an average of 2.50 ± 2.19% affected animals in the last generation. While this was lower than the average observed in Pre-test Homozygous Excluded, the rather large between replicate variation in generation 40 meant that Pre-test 25% Carriers Excluded did not consistently outperform scenario 2 in this regard (Table 2). However, all three remaining Pre-test Carriers Excluded sub-scenarios resulted in a lower final average frequency of affected animals compared to their corresponding Scenario 3 sub-scenarios excluding homozygous bulls under the same level of genetic testing (Figure 7).

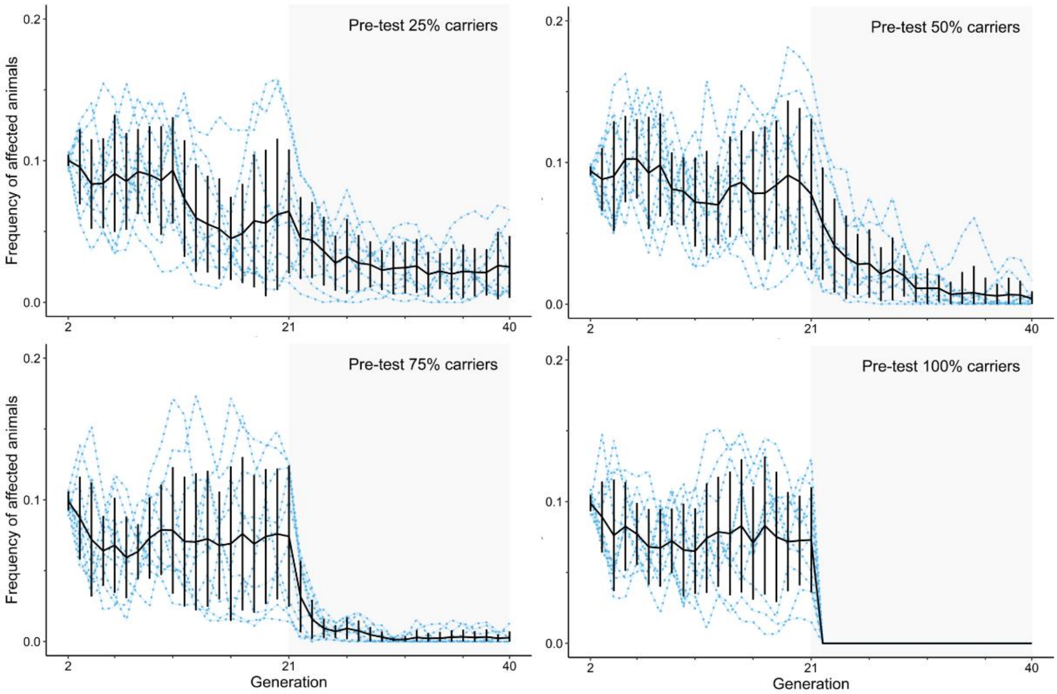

Figure 10. Frequency of affected animals across 40 generations in scenario 3, Pre-test Carriers Excluded. The figure includes graphs for each sub-scenario. The black line represents the mean frequency across ten replicates, with error bars indicating the standard deviation. Blue lines illustrate individual replicates. The grey background marks the period (generations 21-40) when the Pre-test Carriers Excluded selection was implemented. Negative error bars are trimmed when the average frequency of affected animals is low to avoid impossible negative values.

Of all scenarios involving genetic testing of elite AI sires selection against homozygotes, scenario 4, exhibited the smallest and least consistent decline in the frequency of affected animals (Figure 11). In fact, when compared across all selection scenarios the effect on the frequency of affected animals when excluding homozygote elite AI sires was most comparable to that observed when testing

25% of unaffected bulls to remove homozygotes prior to selection in scenario 2 (Figure 7).

In contrast to exclusion of homozygotes, selection against carriers of the deleterious allele among elite AI sires resulted an immediate and drastic reduction in frequency of affected animals (Figure 11). Across all simulated scenarios, this fifth scenario resulted in the second lowest final frequency of affected animals (Figure 7). The only scenario with a lower final frequency was testing 100% of unaffected bulls prior to selection to remove carriers, which led to zero affected animals shortly after generation 21 in all ten replicates. While selection against carriers among elite AI sires on average never reached an affected frequency of zero several of the simulation replicates in the scenario did (Figure11).

Allowing either one or five elite AI sires to breed regardless of genotype resulted in a slightly higher final frequency of affected animals compared to when the entire group were subjected to the selection as in scenario 5 (Table 2). Of the two sub- scenarios, exempting one elite sire each generation unsurprisingly lead to a more immediate and stable decline as well as a lower final average frequency of affected animals compared to when 5 were exempt from selection. Although neither of the two sub-scenarios on average reached zero affected animals, both included individual replicates that did (Figure 11). Compared across all simulated scenarios selection against carriers among elite AI sires allowing one exemption displayed the third lowest final frequency of affected animals, while the sub-scenario exempting 5 elite AI sires per generation placed fifth in this regard (Table 2).

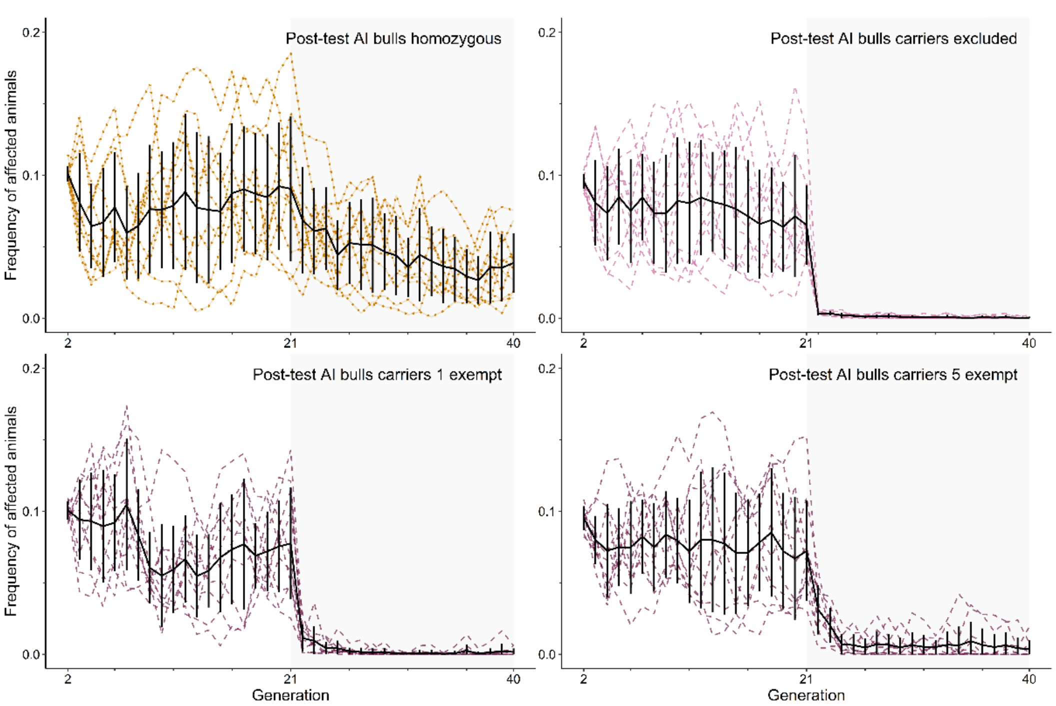

Figure 11. Frequency of affected animals across 40 generations in all post selection gene testing scenarios. The black line represents the mean frequency across ten replicates, with error bars indicating the standard deviation. The coloured lines illustrate individual replicates: Orange for Scenario 4, Light Purple for Scenario 5, and Dark Purple for Scenario 6. The grey background marks the period (generations 21-40) when the different selection scenarios were implemented. Negative error bars are trimmed when the average frequency of affected is low to avoid impossible negative values.

4.3 Effective population size

None of the simulated scenarios involving genetic testing and removal of homozygous or carrier bulls prior to selection (*Pre-test*) resulted in an average effective population size of less than 130 in generation 21-40 (Table 2). Interestingly, there were no clear association between average N_e and the proportion of unaffected bulls tested in these scenarios. The between replicate variation expressed as the standard deviation of the average N_e were for all *Pre-test* scenarios also of similar magnitude. While individual replicates may have resulted in $N_e < 100$, the lower range of the standard deviation of the average N_e did not drop below 100 for any of the *Pre-test* scenarios (Table 2).

In contrast, all three scenarios implementing selection against carriers among AI bulls and elite AI sires resulted in an average N_e below 100. While the three all yielded an N_e just above 90, not allowing any elite AI sires to breed despite their genotype, as in *Post-test AI bulls carriers excluded,* was most detrimental to the genetic diversity with an N_e of 91.3 ± 16.1 (Table 2). It is worth noting that the SD intervals, which were similar between the scenarios excluding carriers among elite AI sires, all included values at or slightly above 100 (Table 2). Selection against homozygotes among AI bulls and elite AI sires did not have an equally detrimental effect on N_e as when selection targeted carriers. Still the exclusion of homozygous AI bulls and elite AI sires resulted in a smaller average effective population size in the last 20 generations compared to all *Pre-test* scenarios (Table 2).

The same pattern observed from scenarios targeting elite AI sires, where selection against carriers resulted in lower N_e then selection against homozygotes, was not consistently observed among the *Pre-test* scenarios. While selection against carriers rather than homozygotes did result in a smaller affective population size when 25% or 50% of unaffected bulls were tested, excluding homozygote bulls had a more detrimental effect on N_e when genetic testing was conducted on 75% or all unaffected bulls (Table 2).

Contrary to expectations based on the results of the two prior outcome metrics, the *Baseline* scenario, which had the least effect on the frequency of the deleterious allele and the number of affected animals, did not yield the highest average N_e (Table 2).

4.4 Number of genetic tests

The last outcome metric, average number of genetic tests used per generation across generation 21-40, is presented below in Table 3. Aside from the *Baseline* scenario all simulated scenarios involved some amount of genetic testing to facilitate the selection against either carriers or homozygotes of deleterious allele.

Naturally, in all scenarios removing carrier or homozygous prior to selection the number of genetic tests utilised per generation increased in tandem with the proportion of unaffected bulls tested. However, in pairwise comparisons of the sub-scenarios excluding either homozygotes or carriers, those excluding carriers consistently resulted in a higher number of genetic tests per generation compared to those with the same proportion tested excluding homozygotes (Table 3).

Due to the design of the *Post-test* selection scenarios, the number of tests used did not vary, with each of the four scenarios requiring exactly 40 genetic tests per generation (Table 3). Although the last two sub-scenarios exempted either 1 or 5 bulls from the selection against carriers it is assumed that these bulls were still tested to obtain their genotype even if that information was not directly utilized to exclude them from breeding.

Table 3. Average number of genetic tests used per generation, mean ± SD, across generation 21-40.

Scenario	Gene tested per gen
Baseline	Non[a]
Pre-test 25% homozygous	239 ± 2
Pre-test 25% carriers	243 ± 2
Pre-test 50% homozygous	478 ± 7
Pre-test 50% carriers	490 ± 9
Pre-test 75% homozygous	723 ± 6
Pre-test 75% carriers	742 ± 11
Pre-test 100% homozygous	969 ± 14
Pre-test 100% carriers	995 ± 15
Post-test AI bulls homozygous	40[b]
Post-test AI bulls carriers	40[b]
Post-test AI bulls carriers 1gen	40[b]
Post-test AI bulls carriers 5gen	40[b]

a) No genetic testing used in the Baseline scenario, b) Genetic testing targeted only at the 40 AI and elite sires selected each generation

5. Discussion

This study set out to formulate and evaluate breeding strategies aimed at reducing the frequency of gonadal hypoplasia and the associated Cs_{29} allele in Swedish Mountain cattle, while preserving the breed's genetic diversity.

While the current policy of phenotypic selection against gonadal hypoplasia appear to maintain a relatively stable prevalence of the Cs_{29} allele and hypoplastic phenotype, the simulation results show that such an approach is unable to achieve a significant decline of either the allele frequency or the frequency of affected animals. Instead, based on the results of the simulations, strategies utilising genetic information to facilitate selection against the defect appear better suited to reduce both the frequency of the deleterious Cs_{29} allele as well as the frequency of hypoplastic animals in the population.

However, the results also highlight clear trade-offs between reducing the prevalence of the defect and two other critical aspects: namely preserving the breed's genetic diversity and limiting the amount of genetic testing required to achieve the reduction. In the simulations, substantial reductions in the frequency of the deleterious allele, along with corresponding decreases in the number of affected animals, were rarely achieved without compromising one of the other two critical aspects.

This trade-off is well illustrated by the scenario in which all unaffected bulls were genetically tested to exclude carriers of the deleterious allele from selection. Although such widespread testing is unlikely to be realistic in practice it resulted in both the lowest average allele frequency observed and complete elimination of affected animals, while maintaining a relatively high effective population size of 138 ± 20.3. In contrast, subjecting only AI bulls to selection against carriers which achieved a comparable, albeit slightly less prominent, reduction of the defect only required 40 genetic test to be conducted each generation. However, this breeding strategy in turn had the most detrimental impact on the genetic diversity of the simulated population, resulting in an average Ne of 91.3 ± 16.

The following discussion will in part explore to what extent it can be deemed acceptable to compromise on these aspects to achieve a reduction in the frequency of Cs_{29} and gonadal hypoplasia. The timing of the genotyping strategy will also be examined, with focus on how it can be used to reduce hypoplasia while avoiding negative effects on genetic diversity and cost-efficiency.

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5.1 Aims of the selection

In the current study, both the frequency of the deleterious allele and the prevalence of affected animals were used to evaluate the effectiveness of each selection strategy in mitigating the problems associated with gonadal hypoplasia. When comparing these two parameters the frequency of hypoplastic animals was consistently lower than that of the deleterious allele across all simulated scenarios. This is to be expected for a defect following an autosomal and recessive pattern of inheritance where, under random mating, the frequency of the defective genotype theoretically should be equal to the squared frequency of the associated allele.

However, the average frequency of hypoplastic animals where consistently lower than the square root of the average frequency of the Cs_{29} allele. This can be explained by the incomplete penetrance of the hypoplasia genotype resulting in only roughly half of animals carrying two copies of the Cs_{29} allele developing underdeveloped gonads.

While these results in of themselves are unsurprising they raise an interesting question relevant to most breeding programs trying to manage the presence of a recessive deleterious genetic variant. Namely, whether the goal of the selection should be to merely avoid undesirable outcomes in the form of homozygous offspring or to attempt to eliminate the deleterious allele from the population.

Although not the primary focus of the article, this question was addressed in a relatively recent review article by Cole et al. (2025), which focused specifically on management of recessive genetic defects in dairy cattle populations. Based on the available literature, the authors proposed that strategies aimed at avoiding homozygous offspring should be viewed as short-term solutions, whereas the reduction of the deleterious allele represents a more long-term approach.

However, it is worth noting that most of the studies discussed in the review focused on lethal recessive variants, which tend to have a much more immediate and serious impact on both animal welfare and production economics compared to non-lethal but still harmful defects, such as gonadal hypoplasia. Therefore, the same conclusions made by Cole et al. (2025) may not be directly applicable, and the suggested management strategies might need to be adapted when dealing with sublethal conditions.

The main issue with hereditary gonadal hypoplasia is that it can cause infertility, which in turn, if a large enough proportion of the population is affected, may reduce the number of breeding animals and increasing the risk of inbreeding. The problem is particularly serious in a population such as the Swedish mountain cattle, which is already classified as endangered and characterized by both a small census and effective population size.

Following this reasoning, it appears as if aiming to avoid homozygous offspring would be sufficient to render gonadal hypoplasia practically harmless by not allowing it to be expressed in individuals homozygous for the associated Cs_{29}

allele and thus removing the risk of increased inbreeding due to the inability of animals to reproduce due to underdeveloped gonads. However, as highlighted by Cole et al. (2025), while avoiding the production of homozygous offspring may prevent the expression of the defective phenotype, it does not necessarily result in the elimination or even a reduction of the deleterious allele from the population.

In the present study this phenomenon can be observed in the selection scenario where no carrier bulls were allowed to breed, *Pre-test 100% carriers excluded*. As previously mentioned, this strategy resulted in zero animals being afflicted by gonadal hypoplasia but was despite this not able to eliminate the deleterious allele from the population. If the deleterious allele remains in the population, the risk of the defect being expressed remains if efforts to avoid homozygous offspring are not maintained over time. The means that even if the phenotypic defect temporarily disappears from the population, measures such as genetic testing will continue to be necessary.

In contrast a total elimination of the deleterious allele would theoretically mean that no further efforts to manage the presence of the allele would be necessary. While this might sound appealing there are potential issues with this approach. To start, the question should be asked whether it is even practically possible to eliminate a harmful allele from a population and, if possible, what other consequences this would entail.

In the review article by Cole et al. (2025), the authors strongly emphasize the difficulty of eliminating a deleterious allele from a population. Even the most intensive selection strategy in the current study, in which all unaffected bulls were genetically tested to remove all male carriers from breeding, failed to eliminate the deleterious allele from the population. In this specific case, the inability to eliminate the deleterious allele can be explained by the fact that the genetic selection against carriers was only directed at bulls, which meant that the cows could act as a reservoir for the allele. To achieve complete elimination of the allele within the 20 generations simulated, it would likely have been necessary to also include females in the selection against carriers.

Although this is a specific example, it illustrates a general issue: that is that a deleterious allele can remain in a population as long as some subgroup of that population, be it animals in geographically isolated herds or individuals where genetic testing was unsuccessful, carries it.

Even if elimination of the allele was obtainable, excluding all carriers of the deleterious allele from breeding may have detrimental impacts on both inbreeding as well as the genetic gain of other important traits (Cole 2015; Derks & Steensma 2021; Cole et al. 2025). Weather this is the case or not depends on the prevalence of the deleterious allele in the population, if there are enough non-carriers to uphold a genetically sustainable and effective breeding program and if there are any genetic correlation to other traits under selection (Cole et al. 2025).

At the time of writing, there is no evidence supporting the existence of genetic correlations, either favourable or unfavourable, between gonadal hypoplasia and other traits of importance, apart from the colour-sided phenotype. However, the absence of evidence does not prove that no such correlations exist; it could simply reflect that the issue has not yet been thoroughly investigated.

Although homozygosity for the Cs_{29} allele has been definitively linked to gonadal hypoplasia by Venhoranta et al. (2013), the cause of the defect's incomplete penetrance remains unknown. If this underlying mechanism is not fully understood, it cannot be ruled out that additional genetic factors, potentially correlated with other traits, may influence the expression of the defect, and thus be inadvertently affected by selection against it.

Even in the absence of direct genetic correlations between gonadal hypoplasia and other traits under selection, genetic progress may still be negatively impacted by selection against the defect. This is not due to the biological linkage between traits, but rather because excluding individuals based on their Cs_{29} genotype reduces the pool of selection candidates. With fewer animals available for breeding, particularly among bulls, the ability to optimise selection across other traits in the breeding goal becomes more limited.

This may ultimately lead to lower genetic gain for other economically or functionally important traits, especially if carriers of the deleterious allele otherwise possess high genetic merit. However, since this simulation study only included two traits, gonadal hypoplasia and the colour-sided phenotype, it is not possible to determine how the different selection scenarios might potentially affect genetic progress for other traits within the actual breeding goal.

Nevertheless, conclusions can still be drawn about the effects of the different strategies on effective population size. Based on these results, it is possible to speculate on the feasibility of eliminating the deleterious allele even though such a strategy was not directly tested in the study.

Given that the complete exclusion of carrier bulls did not have a particularly detrimental effect on effective population size in the simulations, this may indicate that the current allele frequency is sufficiently low to allow for the theoretical possibility of eradicating the deleterious allele from the population by adding genetic selection on the cows as well.

However, this strategy is unlikely to be feasible in practice. Firstly, genotyping all bulls as in the simulation, let alone all animals, if selection would also be applied to the cows, is both logistically and economically unrealistic. Secondly, it is unlikely to that all potential bulls could be tested and selected in the same controlled manner as was done in the simulation due to the actual population structure.

The number of males is often proportionally small compared to the number of females, as is the case for most dairy breeds where only the females contribute to the primary production. Unlike in the simulations, many male calves are likely either slaughtered or castrated at an early age, before they can be genotyped and considered for breeding. If too few candidate bulls are available for selection, this may in turn have a greater negative impact on effective population size than that observed in the simulation scenario excluding all carrier bulls from breeding.

5.2 Effective population size

Given that the purpose of developing an alternative breeding strategy against gonadal hypoplasia is to reduce the risk of inbreeding associated with infertility, it is essential that such a strategy in of itself does not contribute to the very problem it seeks to prevent. A key part of this study was therefore to evaluate the impact of the simulated scenarios on the genetic diversity expressed as the effective population size. The findings in this study indicate the existence of such a trade-off, as some strategies that successfully reduced the prevalence of the defect also resulted in a drop in effective population size.

This section will therefore not only discuss how different selection scenarios affected the effective population size but also what, if any, loss of genetic diversity could be deemed acceptable in favour of mitigating gonadal hypoplasia.

As mentioned in the literature review multiple recommendations have been proposed concerning threshold values for effective population size. These typically include both short-term guidelines, spanning approximately five generations, and long-term minimum values intended to apply for the next 10 or more generations. The short term recommendations aim to prevent the decreased fitness caused by inbreeding; a phenomenon known as inbreeding depression.

Inbreeding depression results from increased homozygosity which heightens the risk of deleterious recessive traits being expressed while also reducing the fitness for traits where the heterozygous genotype is superior to the homozygous. A minimum effective population size of 50, proposed by (Soulé & Wilcox 1980) was long regarded as the standard recommendation to prevent inbreeding depression in the short term. However, in 2014 Frankham *et al.* challenged this, instead suggesting that the effective population size should not be less than 100 to adequately minimize the risk of inbreeding depression in the next five generations. The same authors also proposed an increase of the long-term recommendation from an effective population size of above 500 to instead above 1000. The goal with the long-term recommendation is to ensure that there is adequate genetic variation within the breed to maintain its evolutionary potential, in other words, enough genetic variation to ensure that the breed it can adapt to potential future changes and challenges (Soulé & Wilcox 1980; Frankham et al. 2014). Although there has been some discourse regarding the different recommendations to air on the side of caution the higher thresholds suggested by

Frankham *et al.* (2014) seems to be the most widely accepted within the field today.

With a current estimated effective population size of 136, the Swedish mountain cattle surpass the short-term criteria but does not come close to meeting either criteria for long-term genetic sustainability. Based on the recommendation by Frankham et al (2014) a slight loss of genetic variation may, in the short term, be acceptable to facilitate stronger selection against gonadal hypoplasia but for the future preservation of the breed no such loss is welcome.

However, the loss of genetic variation due to selection against gonadal hypoplasia should be put in relation to the potential gain that a reduction in the incidence of hypoplasia could entail, if fewer animals are infertile and unable to contribute to the gene pool. As touched upon in the literature overview only a small proportion of gonadal hypoplasia cases in Swedish mountain cattle present as severe and bilateral resulting in infertility. While many hypoplastic animals are physiologically capable of reproducing, they are effectively rendered infertile from a breeding perspective due to current herd book regulations, which prohibit their use in breeding regardless of the defect's severity. In practice, this means they are excluded from contributing genetically to the population, irrespective of their actual reproductive potential.

As expected, given the breed's current effective population size, none of the simulated scenarios achieved an average effective population size sufficient to meet the long-term recommendation of $N_e > 1000$ proposed by Frankham et al. (2014). However, most scenarios did reach an average effective population size exceeding 100, suggesting that the implementation of these selection strategies, at least over 20 generations, does not lead to a detrimental increase in genetic load that would result in inbreeding depression.

All simulated strategies involving pre-selection against carriers or homozygotes of the deleterious allele met the lower threshold for effective population size of above 100. However, considerable variation in the resulting effective population size was observed not only in the pre-test selection scenarios but across all scenarios, with standard deviations large relative to each mean. This indicates that although reproduction was influenced by preferences for the colour-sided phenotype and the various selection strategies against gonadal hypoplasia, several sources of stochastic variation remained. Such variability underscores the importance of continuous monitoring of genetic diversity, regardless of which management strategy is implemented to avoid unintended erosion of genetic variation.

Surprisingly, no clear correlation was observed between the resulting average effective population size and the intensity of pre-selection, i.e., the proportion of unaffected bulls that were genetically tested. Nor was there a consistent difference in effective population size between strategies targeting carriers versus homozygotes under otherwise comparable conditions in the scenarios where genetic testing was implemented as the first stage of selection.

Two possible explanations may account for these unexpected findings. First, the initial frequency of the deleterious allele may have been sufficiently low so that even the most intensive pre-selection strategy, where all unaffected bulls were genotyped and carriers removed, still left a large enough pool of selection candidates to avoid a detrimental increase in inbreeding.

This mechanism has previously been demonstrated in simulation studies of genetic defects in dogs, where the impact of carrier exclusion on genetic diversity was shown to depend strongly on the initial frequency of the deleterious allele (Leroy et al., 2012).

Alternatively, the absence of a detectable effect of the pre-selection intensity or selection target on the effective population size in the current study may be due to a compensatory mechanism. Because while all pre-selection strategies produced similar effective population sizes, the scenarios involving broader testing and/or selection against carriers consistently resulted in a lower frequency of hypoplastic animals unable or prohibited from contributing genetically to the population. This reduction may have helped preserve genetic diversity in the population in the more intensive selection scenarios by increasing the pool of hypoplasia free animals available for breeding, thereby offsetting the inbreeding pressure introduced by the selection itself.

However, in the scenarios where selection was limited to AI bulls already designated for breeding, the effective population size displayed a clearer connection to the selection target of each scenario. The post-test selection scenario where all carriers were excluded had a more detrimental impact on genetic diversity, resulting in an effective population size below the recommended threshold of 100. In contrast, the post-test selection scenario, where only homozygous AI bulls were removed from breeding, resulted in a larger effective population size, closer to those observed in the pre-selection scenarios. The fact that selection against carriers rather than homozygotes resulted in a lower effective population size, despite yielding a more substantial reduction of hypoplastic animals, indicate that the gain in terms of fewer hypoplastic animals does not outweigh the cost of not allowing any carriers of the deleterious allele among the bulls used in artificial insemination.

Although the observed differences in effective population size may appear to be caused by whether selection targets carriers or homozygotes; it is more likely that the variation stems from how large a proportion of bulls are excluded. Since carriers of the deleterious allele make up a larger share of the candidate population than homozygous bulls selection against carriers leaves fewer individuals available for breeding. This is likely the real reason behind the higher inbreeding rates and lower effective population size observed when selection targeted carriers rather than homozygous AI bulls.

Further support for this comes from the last two simulations, which were, for all intents and purposes, the same as the scenario requiring all AI sires to be non-carriers, except that either one or five elite AI sires were each generation exempted from the non-carrier requirement. While both scenarios resulted in an effective population size below 100, they had a less detrimental impact on genetic diversity compared to the scenario where all AI sires hade to be non-carriers. The inbreeding rate also declined relative to the number of bulls exempted per generation. This was despite these scenarios resulting in a slightly higher proportion of hypoplastic animals which, based on previous reasoning, would be expected to negatively affect the genetic diversity. This further supports the notion that the most influential factor on effective population size is the number of influential sires contributing to the next generation. This conclusion could also help explain the similar effective population sizes observed across all pre-test selection scenarios, as these consistently allowed for the selection of 90 bulls each generation due to the relatively low deleterious allele frequency.

Another noteworthy, yet disappointing, finding was the limited positive impact on the resulting effective population size achieved by allowing either one or five elite AI sires to breed despite their carrier status. Since much literature regarding management of genetic defects in dairy cattle, as reviewed by Cole et al. (2025), indicates that complete exclusion of carriers often leads to increased inbreeding and thus advocates a limited use of carriers, there was hope that the exemptions applied in this study would have a more significant effect than what was observed.

There are several explanations as to why such an effect was not observed here. In the last simulation scenario, the elite AI sires exempt from the non-carrier requirements were chosen at random as no other trait than the colour-sided phenotype impacted the selection probabilities in the simulations. Therefore, it is possible that non-carrier bulls that would have been selected for breeding regardless were exempt from the rule, while carrier or homozygous bulls for the deleterious allele were excluded. This may have led to fewer AI bulls contributing to the next generation than if the exemptions had been limited to elite AI sires that had otherwise failed to meet the non-carrier selection criteria. While this may partly explain why the effect on inbreeding was less pronounced than expected, targeting exemptions solely at carrier or homozygous elite AI sires would likely have resulted in a smaller reduction of the Cs_{29} allele and the number of hypoplastic animals in the population.

If implemented as an actual strategy to combat gonadal hypoplasia, additional factors would likely influence which elite AI sires were exempt from selection, such as whether they also possessed other desirable traits. Alternatively, sires with a low average kinship to the population could be selected as elite AI sires despite their hypoplasia genotype to further minimize the inbreeding rate.

As the results of this study indicate that the number of breeding males is crucial for the rate of inbreeding it would be of interest to investigate the possibility of a strategy where selection still targets AI bulls but where the bulls not meeting the non-carrier criteria are replaced, keeping the number of influential sires stable over generations. Such a strategy would potentially, based on the results of this study, mitigate the negative impact on the genetic diversity while maintaining the same effective reduction of the deleterious allele and of hypoplastic animals. However, to have other eligible bulls available to replace those failing to meet the selection criteria against carriers, a larger number of candidate bulls than the estimated 40 influential sires needed per generation in this study would need to be selected and genetically tested. Although effective in theory, such a strategy would likely lead to higher costs due to increased genotyping efforts, while its practical feasibility remains questionable. As noted in the literature review, identifying even two new elite AI sires per year is already considered challenging. This raises doubts about the likelihood of producers being able, or even willing, to retain a larger number of intact bulls solely for the purpose of replacing potential carrier bulls. This concern is particularly relevant given the population structure of dairy breeds such as the Swedish mountain cattle, in which males do not contribute directly to production and are therefore often culled or castrated at an early age.

5.3 Genotyping strategy

That gene-assisted selection is an effective tool for reducing the prevalence of gonadal hypoplasia is clearly supported by the findings of this study. However, the results also show that the timing of the genetic testing, as well as which subset of the population is tested, can have a significant impact on the genetic diversity of the breed. A central question therefore arises: should genetic testing be conducted prior to the selection of bulls for breeding, thereby applying the genetic criteria early in the selection process, or should it be implemented after selection based on other merits, when testing can be limited to bulls intended for use in artificial insemination?

If considering only the reduction of hypoplasia prevalence and preservation of the genetic variation of the Swedish mountain cattle the results from this study suggest that genetic testing of the unaffected bull population prior to selection would be the most rational choice. However, the simulation results also demonstrate that this type of strategy is resource intensive. With even the most limited pre-test scenario requiring approximately six times as many genetic tests be conducted per generation as the strategies where gene-assisted selection against hypoplasia only targeted AI bulls and elite AI sires.

Importantly, it is not only the number of tests that raises concerns regarding the pre-test strategies, but also who would be responsible for carrying out these tests. If gene-assisted selection is to be conducted on a large scale prior to selection, the burden of collecting and submitting genetic samples would likely fall on individual bull owners, similarly to the practices for milk protein genotyping in the current breeding program. Without financial support for bull owners to perform the required testing, the practicality of such a strategy seems questionable, especially considering that bulls would need to be kept until an age at which they can be tested for hypoplasia and subsequently considered for breeding.

Given these practical limitations focus naturally shifts towards the strategies where the gen- assisted selection targets only the most genetically influential bulls, namely those used in artificial insemination. In such scenarios, the responsibility for conducting genetic tests would shift from individual bull owners to the AI companies which, combined with the lower number of tests required, makes these strategies appear more feasible. Unfortunately, none of the simulated scenarios in this study targeting AI bulls and elite AI sires proved optimal, either resulting in a non-sustainable decline in effective population size when selection targeted carriers or in an unsatisfactory decline of hypoplasia and the associated allele when homozygotes were excluded.

This may suggest that a more balanced strategy than those simulated in this study could be needed to better resolve the observed goal-conflicts between reducing the prevalence of gonadal hypoplasia, preserving genetic diversity, and keeping the associated costs at a manageable level. One such approach could involve planed mate allocation where genetic information at the Cs_{29} locus is used to avoid at-risk matings while still allowing carriers of the Cs_{29} allele among the AI bulls and elite sires to reduce inbreeding.

The suitability of planned mating strategies for managing deleterious recessive genetic variants in cattle populations has been demonstrated in several simulation studies (Van Eenennaam & Kinghorn 2014; Upperman et al. 2019; Bengtsson et al. 2022). While these studies addressed the presence of multiple recessive deleterious and or lethal recessive alleles, the findings can still be relevant for management of gonadal hypoplasia in Swedish mountain cattle. Both Van Eenennaam & Kinghorn (2014) and Upperman et al. (2019) concluded that using mate allocation to avoid homozygous offspring, given a perfect knowledge of the deleterious genotype, was always preferable compared to strict culling or exclusion of all carriers from breeding, as such strategies often had a detrimental impact on both inbreeding rates and overall profitability.

This is consistent with the findings of the present study, where complete exclusion of carrier bulls either resulted in very high costs or led to an unacceptable reduction in the breed’s genetic diversity. However, as shown in this study, genotyping the entire male or female population, let alone both, is likely neither economically nor practically feasible regarding the Swedish mountain cattle. Thus, a universally applied mate allocation strategy against gonadal hypoplasia which requires that the genotypes of each mating pair are known seams fare fetched.

A more realistic alternative could be to focus genotyping efforts on AI bulls and elite AI sires and to publish their carrier status. This would provide individual farmers with the necessary information to make informed breeding decision, whether or not they can or wish to genotype their cows. With the carrier status of AI bulls made public cow owner not interested in genotyping their herd could simply select non-carrier bulls and still be confident that their calves will not be affected by gonadal hypoplasia. For farmers willing to genotype their cows, this strategy would also allow for the continued use of carrier bulls that possess other desirable traits or contribute valuable genetic diversity.

In theory, a strategy as that described above could provide a more balanced approach by mitigating the adverse effects on inbreeding associated with the complete exclusion of carrier AI bulls, while still ensuring an effective reduction in the incidence of hypoplastic animals. A note of caution is however warranted, as this particular strategy was not simulated in the present study nor in the literature mentioned. Thus, although both this study's results and the literature suggest that such an approach could be promising, further research should be conducted prior to any implementation of such a strategy.

5.4 Methodological limitations and concerns

While AlphaSimR enables the modelling of intricate breeding programs, the simulations in this study still rely on a number of assumptions and simplifications that may affect how well the results translate to the actual population. This section will therefore try to identify and discuss such methodological limitations that have not yet been addressed.

Among the most obvious discrepancies are the structure and size of the simulated population. Unlike in the simulations, where the population size was constant and the sex ratio equal, the real mountain cattle population is both smaller and much more female-dominated. The assumption of discrete generations is also unrealistic for a population such as the Swedish mountain cattle but necessary, as simulating overlapping generations is much more complex and time consuming.

Similarly, there were several differences between the simulated baseline selection and the current breeding strategy and management of gonadal hypoplasia it aimed to emulate. While selection intensity is often higher for males than females in dairy breeds due to the population structure, assuming that the preference for colour-sided animals only affect males, as in the simulations, is most likely not entirely accurate. However, this discrepancy was handled by recalculating the selection probabilities of the different genotypes to reflect that selection only occurred among bulls in the simulations.

The sampling probabilities also entailed assumptions regarding the existence of a preference for the colour-sided phenotype, despite no such mention in the current breeding goal. However, that fact that both frequencies of gonadal hypoplasia and the associated Cs_{29} allele remains high despite the ongoing selection against the defect suggest the presence of a balancing selection. Given the shared genetic basis of the two traits, it is reasonable to assume that it is in fact a preference for colour-sidedness that contributes to the persistence of gonadal hypoplasia within the breed.

As for the strength of this preference, estimations were based on the assumption that it, together with the current selection against hypoplasia, is what maintains the Cs_{29} allele at its current state of genetic equilibrium. Since the average frequency of the deleterious allele remained stable in the baseline simulations, this suggests that the estimated strength of the preference combined with the simulated selection against the defect, provides an acceptable approximation of the ongoing balancing selection in the actual population.

Aside from this, other selection parameters such as the number of bulls in the three fecundity groups and the number of offsprings assigned to each bull, may not exactly reflect the actual population. However, without access to accurate data on the use of AI bulls or detailed records of the number of calves born from these bulls it was difficult to construct a more realistic scenario.

An additional oversight when constructing the simulation was that no constraints were applied to ensure that the number of offspring assigned to each dam remained within realistic limits. Upon reviewing the simulation outputs, it was discovered that some dams produced more offspring than would be biologically possible during a five year period. This problem resulted from a failure to account for the fact that, to select the intended 1000 individuals dams, a surplus of females would be needed as a portion would always express gonadal hypoplasia and be excluded as selection candidates. To compensate for the lack of eligible dams, some individuals were assigned more than one offspring, which, in the absence of the aforementioned restrictions, resulted in the observed unrealistic fecundity of certain individuals. This oversight may have affected the results of the simulations, especially with regard to the level of inbreeding and the resulting effective population size.

Another factor that did not fully align with the actual breeding program and also likely affected the resulting effective population size was that no restrictions on relatedness where imposed when assigning mating pairs. As a result, individuals that under real circumstances would not have been allowed to breed due to their level if kinship were still able to do so in the simulations possibly elevating the level of inbreeding.

From the above discussion it is clear that several of the population and baseline selection parameters simulated were not entirely representative of the actual population. However, the fact that they collectively yielded an effective population size close to that of the real population indicates that they, when put all together, provide a reasonable base from which alternative strategies to combat gonadal hypoplasia could be evaluated.

6. Conclusion

This study set out to evaluate potential breeding strategies against gonadal hypoplasia in Swedish Mountain Cattle without compromising the genetic diversity of this already endangered breed.

The most obvious finding to emerge is that utilizing both phenotypic and genetic information to facilitate selection against gonadal hypoplasia is more effective than when selection decisions are based on phenotype data alone.

Focusing gene-assisted selection against gonadal hypoplasia on the most genetically influential bulls appear as the most viable strategy to effectively reduce the prevalence of the defect while keeping the associated costs down. Yet, the results also indicate that such a strategy also carries the greatest risk of jeopardizing the breed's genetic diversity.

Unfortunately, none of the simulated selection strategies targeting influential bulls were able to adequately balance the reduction of gonadal hypoplasia with the preservation of genetic diversity. Nevertheless, the results still provide useful insights for future research and the potential application of gene-assisted selection to manage gonadal hypoplasia.

Given that the number of influential sires proved to be a major determinant of inbreeding levels, excluding all carriers of the Cs_{29} allele from breeding among the influential sires would not be advisable without providing suitable replacements. Instead, alternative approaches such as strategic mate allocation could be explored. To enable the future construction of a more nuanced breeding strategy such as mate allocation against gonadal hypoplasia however, a rational first step could be to initiate genetic testing of all AI bulls and elite AI sires.

At the time of writing, there is no evidence indicating that the frequency of Cs_{29} would differ between the bulls selected for AI use and the general population. However, systematic testing could serve to verify this assumption as well as provide other useful information. Should a discrepancy in allele frequency for example be found, it would not only impact a future selection strategy but could also potentially indicate the existence of a yet undetected correlation to another trait under selection. In addition, if a genetic test for the hypoplasia allele becomes publicly available, publishing the test results for all AI bulls would also allow individual farmers to make informed breeding decisions and eliminate the risk of producing hypoplastic offspring.

Finally, the results of this study show that regardless of which strategy is implemented, continuous monitoring of inbreeding levels in the population is essential. Given the limited size of the breed, genetic diversity could otherwise decline rapidly if too few or too closely related animals are used extensively in breeding.

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Popular science summary

En möjlig genväg för fjällrasen?

Sedan tidigt 1900-tal har en recessiv genetiskt defekt känd som könskörtel hypoplasi skapat bekymmer för den svenska fjällras kon. Defekten som uttrycker sig i underutvecklade äggstockar eller testiklar, kan hos drabbade djur orsaka både nedsatt fertilitet och i svåra fall total infertilitet. För den ej insatte kan det låta långsökt att dessa problem skulle bero på att man i slutet av 1800-talet beslutade att fjällkor skulle vara vita med små svarta eller röda fläckar. Trots detta är det faktiskt helt sant!

Vad är det då som gör att avel för vita fjällkor resulterar i vissa djur blir infertila? Boven i dramat har lokaliserats till fjällkornas 29:e kromosom och kallas Cs_{29}. Cs_{29} är en recessiv skadlig allel, vilket i praktiken innebär att en individ måste ärva två kopior av allelen för att riskera att drabbas av könskörtelhypoplasi. Att besitta två kopior är dock inte en garanti för att drabbas och i snitt utvecklar endast hälften av dessa djur hypoplasi. Men hur var det då med färgen? Jo, utöver hypoplasi så orsakar även Cs_{29} den eftertraktade vita färgen, för detta krävs dock endast en kopia för att egenskapen ska synas.

Även om man vid tiden inte kunde peka ut Cs_{29} som länken mellan hypoplasi och färgen, fastslogs redan i början av 1900-talet att hypoplasi troligen orsakades av en recessiv genetisk defekt med ofullständig penetrans. Med denna vetskap introducerades snabbt krav på att fjällras tjurar för att användas i avel måste ha två normalt utvecklade testiklar och senare infördes liknande krav även för kor.

Trots att dessa krav har varit i ständig användning sedan de infördes, återfinns hypoplasi-problematiken fortfarande hos dagens fjällkor. Detta, i kombination med att rasen kraftigt minskat i antal under de senaste 100 åren och idag räknas som hotad, gör det än mer angeläget att minska antalet fall av hypoplasi.

Med hjälp av modern teknik är det idag möjligt att genom ett gentest identifiera djur som bär på den skadliga allelen oavsett om de uppvisar fysiska symptom eller inte. Syftet med denna studie var därför att med hjälp av datasimuleringar utvärdera hur dessa gentester skulle kunna användas för att bekämpa könskörtel hypoplasi bland svenska fjällkor utan att rasens genetiska variation minskar.

Glädjande nog visar resultaten att gentest i kombination med fysiska bedömningar är ett effektivare verktyg för att bekämpa hypoplasi jämfört med dagens strategi. Det finns dock uppenbara målkonflikter att ta hänsyn till då en effektiv reduktion av hypoplasi i simuleringarna antingen medförde betydande ekonomiska kostnader eller risk för att rasens genetiska variation minskar. Sammantaget framstår gentest som en lovande metod för att bekämpa hypoplasi. Dock måste en sådan framtida strategi vara nyanserad och kapabel att balansera olika aspekter såsom kostnader, genetisk variation och hälsa. Annars finns risken det som idag tycks vara en genväg för fjällrasen snabbt kan bli en senväg.

Appendix 1

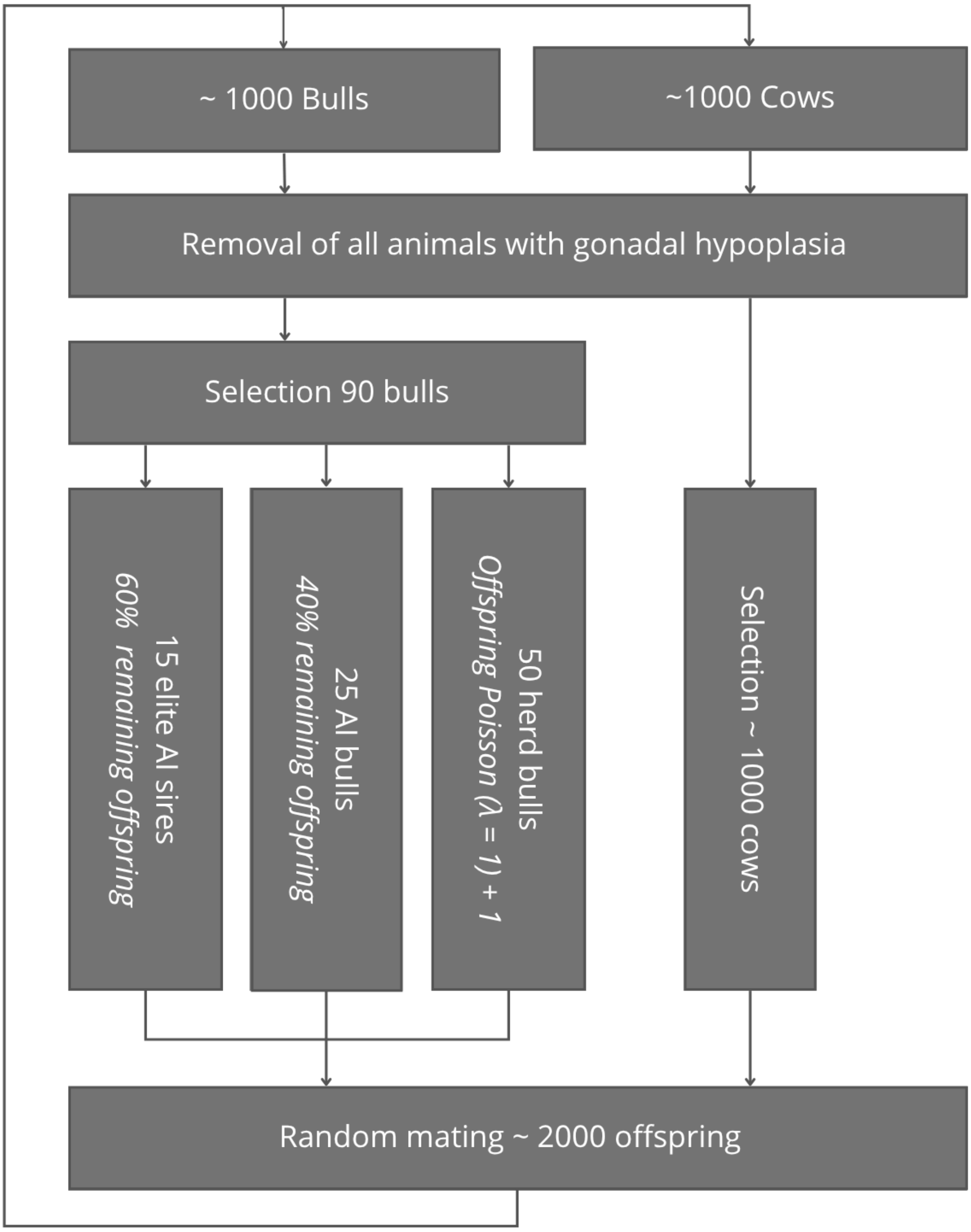
BTA29 / BTA6	**(W/W)**	**(Cs_{29}/W)**	**(Cs_{29}/ Cs_{29})**
(W/W)	0,06112	0,11736	0,11736
(Cs_6 /W)	0,11736	0,11736	0,11736
(Cs_6/ Cs_6)	0,11736	0,11736	0,11736

Appendix 2

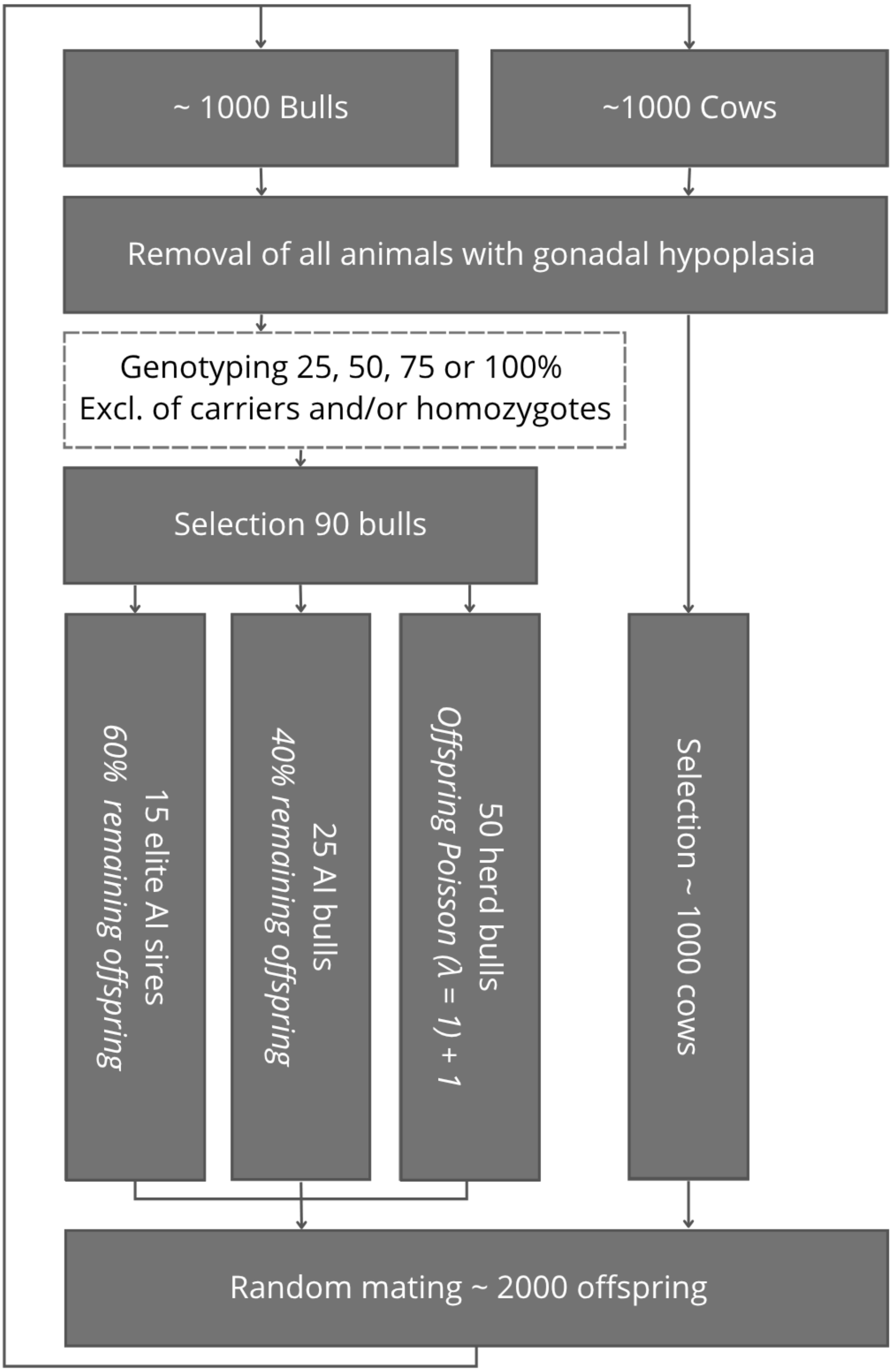
Average frequency of non- deleterious allele in generation 40 (Freq. ± SD) across all simulated scenarios.

Scenario	Freq. non-del. ± SD
Baseline	0.391 ± 0.206
Pre-test 25% homozygous	0.435 ± 0.182
Pre-test 25% carriers	0.322 ± 0.094
Pre-test 50% homozygous	0.448 ± 0.186
Pre-test 50% carriers	0.420 ± 0.255
Pre-test 75% homozygous	0.281 ± 0.153
Pre-test 75% carriers	0.353 ± 0.152
Pre-test 100% homozygous	0.391 ± 0.160
Pre-test 100% carriers	0.327 ± 0.158
Post-test AI bulls homozygous	0.422 ± 0.159
Post-test AI bulls carriers	0.307 ± 0.151
Post-test AI bulls carriers 1gen	0.306 ± 0.165
Post-test AI bulls carriers 5gen	0.292 ± 0.180

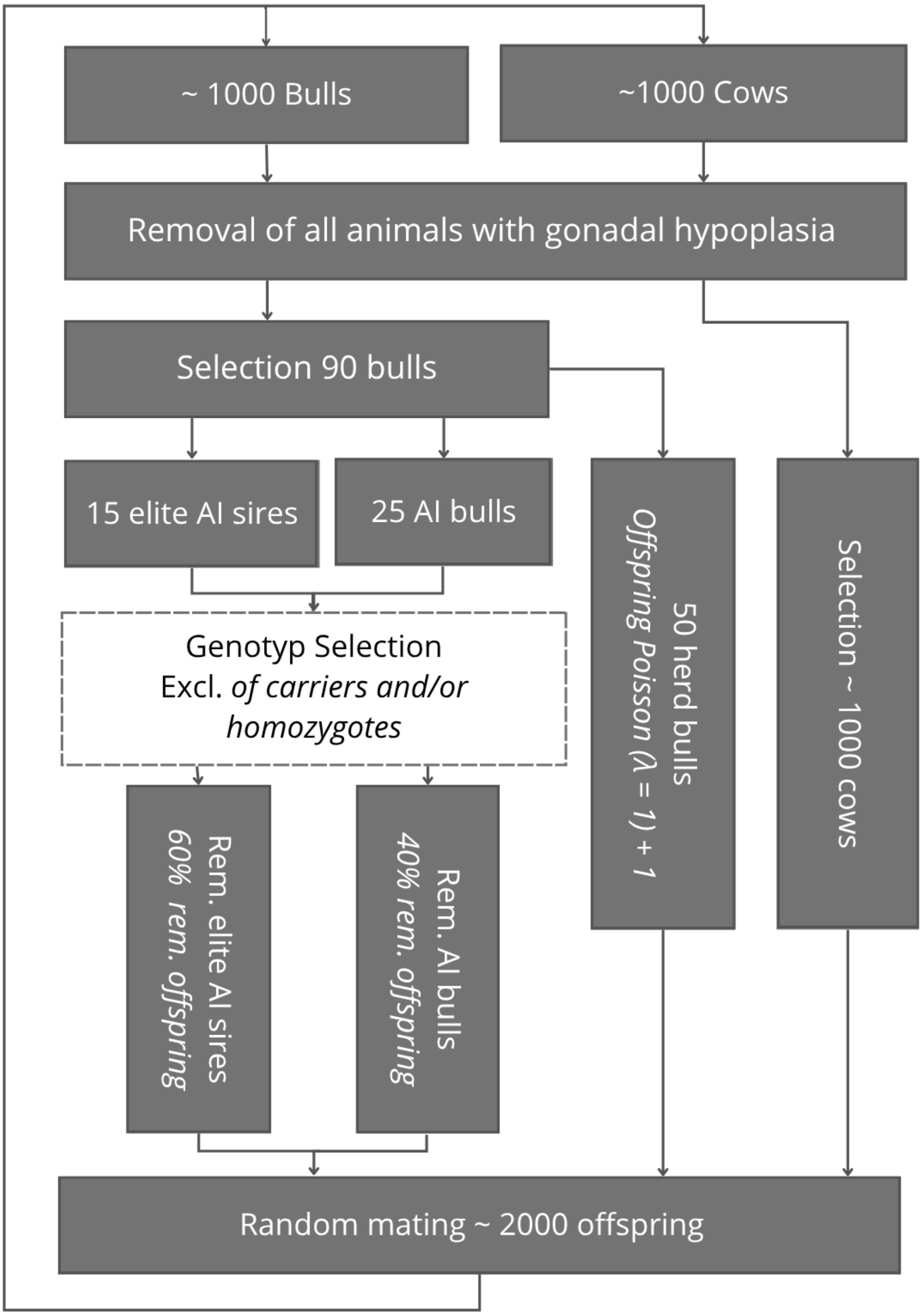
Appendix 3


~ 1000 Bulls
~1000 Cows
Removal of all animals with gonadal hypoplasia
Selection 90 bulls
15 elite AI sires
60% remaining offspring
25 AI bulls
40% remaining offspring
50 herd bulls
Offspring Poisson (λ = 1) + 1
Selection ~ 1000 cows
Random mating ~ 2000 offspring

Appendix 4


~ 1000 Bulls
~1000 Cows
Removal of all animals with gonadal hypoplasia
Genotyping 25, 50, 75 or 100%
Excl. of carriers and/or homozygotes
Selection 90 bulls
15 elite AI sires
60% remaining offspring
25 AI bulls
40% remaining offspring
50 herd bulls
Offspring Poisson (λ = 1) + 1
Selection ~ 1000 cows
Random mating ~ 2000 offspring

Appendix 5


~ 1000 Bulls
~1000 Cows
Removal of all animals with gonadal hypoplasia
Selection 90 bulls
15 elite AI sires
25 AI bulls
50 herd bulls
Offspring Poisson (λ = 1) + 1
Selection ~ 1000 cows
Genotyp Selection
Excl. of carriers and/or homozygotes
Rem. elite AI sires
60% rem. offspring
Rem. AI bulls
40% rem. offspring
Random mating ~ 2000 offspring

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