Genetic stability in the Icelandic horse breed

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Summary

Despite the Icelandic horse enjoying great popularity worldwide, the breed’s gene pool is small. This is because of a millennium of isolation on Iceland, population crashes caused by natural disasters and selective breeding. Populations with small effective population sizes are considered to be more at risk of selection pressures such as disease and environmental change. By analysing historic and modern mitochondrial DNA sequences and nuclear coat colour genes, we examined real-time population dynamics in the Icelandic horse over the last 150 years. Despite the small gene pool of this breed, we found that the effective population size and genetic profile of the Icelandic horse have remained stable over the studied time period.

Keywords coat colour genetics, effective population size, historic DNA, Icelandic horse, mitochondrial DNA, selective breeding.

The Icelandic horse is an ancient breed descended from horses imported by the original Scandinavian settlers of Iceland (Hendricks 1995). The breed’s gene pool has been effectively closed for a millennium. Since the middle of the 20th century, the Icelandic horse has been one of the world’s most popular breeds in terms of numbers of pedigree horses (approximately 300 000 registered individuals today; International Federation of Icelandic Horse Associations 2010). The majority of the breed now lives outside Iceland, with breeding societies in existence in 19 countries.

Nevertheless, a series of natural disasters, including a devastating famine in the 1790s, as well as the long isolation of the breed and strict selective breeding, have dramatically reduced the effective population size (\(N_e\)) and genetic diversity (Hendricks 1995). This is cause for concern, because populations with low diversity levels are thought to have greater difficulty surviving selection pressures such as disease (Cunningham et al. 2001). In fact, the Icelandic horse has such low resistance to infectious disease that it is illegal to re-import Icelandic horses to Iceland, to prevent the introduction of exotic pathogens to the stock (Hendricks 1995).

Because the Icelandic horse’s gene pool is small and the breed continues to be selectively bred, it is crucial to monitor changes in the diversity of the breed in case conservative action becomes necessary. To determine whether the breed diversity has changed significantly in the recent past, we compared both mitochondrial and nuclear genetic diversity in extant Icelandic horses to the diversity in the specimens represented in the 1860s Krabbe collection of Icelandic horse skulls. Specifically, we studied the non-coding mitochondrial D-loop to examine shifts in neutral diversity. We also analysed three nuclear coding SNPs determining coat colour (in the genes ASIP, SLC45A2, formerly known as MATP, and MC1R). These markers are plausible candidate genes for tracking diachronic change within the Icelandic horse, because breeders are actively attempting to maintain coat colour diversity and preserve rare phenotypes (United States Icelandic Horse Conference 2010).

Whole genomic DNA was extracted using published protocols from 3 to 20 hair roots from 21 extant, unrelated, pedigree Icelandic horses of known ancestry (McGahern et al. 2006) and from 46 historic Icelandic horses from the Krabbe collection (Museum for Natural History, Berlin) according to Campana et al. (2010a). See Appendix S1 for methodological details.

Four hundred and forty base pairs of the horse mitochondrial D-loop were amplified and sequenced. An additional eight modern Icelandic mtDNA sequences of unknown parentage were downloaded from GenBank (AJ413717–AJ413723, AF072988). The Icelandic D-loop sequences were truncated to 247 bp and compared with 1381 publicly available sequences using median-joining networks and neighbour-joining trees to place the haplotypes in the context of a larger phylogenetic context.

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of worldwide horse mtDNA variation (Table S2). Differentiation in mtDNA haplotype frequencies between breeds was tested using the exact test of population differentiation (Raymond & Rousset 1995). Icelandic horse population dynamics over time were modelled in BEAST 1.6.1 (Drummond et al. 2005). \( N_e \) estimates were derived by dividing BEAST’s \( N_e \) estimates by 12, the horse generation time (Sokalov & Orlov 1986). Finally, we assayed coding SNPs in three genes determining horse coat colour (ASIP, SLC45A2 and MC1R) following Campana et al. (2010b).

The 440 bp mtDNA sequence was obtained for all 21 modern Icelandic samples and 35 of the 46 (76%) historic specimens (Table S1). Partial sequences were also obtained for the historic specimens ZM1382, ZM1387 and ZM1395. D-loop sequences have been deposited in GenBank (HQ153701–HQ153760).

We identified 15 D-loop haplotypes in the Icelandic horse (Fig. 1a). The exact test of population differentiation revealed significant \((P < 0.00001)\) differences in the D-loop haplotype frequencies between the modern and historic Icelandic samples (Table S3). Nevertheless, the modern and historic Icelandic populations clustered together in the neighbour-joining tree as expected and were distinct from other breeds (Fig. 1b).

The Icelandic horse \( N_e \) remained approximately constant over the last 150 years, with a median value of approximately 1000 individuals and a 95% confidence interval of 80–38 000 individuals (Fig. 2).

Genotypes for all coat colour SNPs were obtained for all modern Icelandic samples. Repeatable ASIP, SLC45A2 and MC1R results were obtained for 7, 28 and 12 of the historic Icelandic horses, respectively (Table S1). No statistically significant differences were found between the frequencies of the alleles or genotypes in the historic and modern populations for any of the coat colour SNPs (Fisher’s exact tests, \( P > 0.05 \) for all tests). No significant deviations from Hardy–Weinberg equilibrium were
identified for any of the coat colour SNPs (chi-square tests, \( P > 0.05 \) for all tests).

The Icelandic horse has remained genetically stable in the recent past. \( N_e \) has remained constant over the past 150 years (Fig. 2). Moreover, despite finding statistically significant evidence for population change within the last 150 years, there is a high degree of concordance between the mtDNA profiles of historic and modern populations (Fig. 1a). This differentiation is probably because of allelic drift rather than a selection effect. Using a conservative estimate of \( N_e = 1000 \) and a generation time of 12 years, we cannot refute the null hypothesis that genetic drift was the primary cause of allelic frequency shifts within the Icelandic horse population (Appendix S1). Nevertheless, given our small sample sizes, our data may not reflect the complete worldwide Icelandic horse diversity.

Although our sample sizes are limited, we find no evidence to suggest that the frequency of Icelandic coat colour alleles has significantly changed over the last 150 years. Coat colour, however, is determined by a large number of genes, of which only a fraction were analysed here. Further research is required to determine whether allelic frequencies have changed for other coat colour genes.

In summary, despite the small gene pool of the Icelandic horse and an active selective breeding programme, the genetic profile of the breed has remained stable over the last 150 years. Unlike numerous other horse breeds, the diversity of the Icelandic horse appears to be secure.

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References


Supporting information

Additional supporting information may be found in the online version of this article.

Appendix S1 Supplementary methods.

Table S1 Icelandic samples used in the genetic analyses.

Table S2 Horse breeds and populations for which mtDNA sequences were compared to place the historic and modern Icelandic mtDNA sequences in a worldwide context.

Table S3 D-loop haplotypes frequencies in the historic and modern Icelandic horse.

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