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Development of a Compact SNP Panel for Hereditary Disease Screening in Kazakh Horse Breeds through Integration of OMIA and Genome-Wide Genotyping Data

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ABSTRACT

Genetic monitoring of hereditary disease-associated variants is becoming an increasingly important component of modern horse breeding. However, information on the distribution of disease-associated loci in Kazakh horse breeds remains limited. Integrating genome-wide SNP data with curated disease databases may provide a practical approach to population-level genetic screening. To develop a compact SNP panel for monitoring hereditary disease-associated variation in Adai and Kostanai horses using genome-wide SNP genotypes and disease loci obtained from the Online Mendelian Inheritance in Animals (OMIA) database. A total of 341 horses (291 Adai and 50 Kostanai) genotyped with a high-density equine SNP array were included in the study. After quality-control filtering, we retained 74,210 SNPs for analysis. Disease-associated loci were extracted from OMIA and mapped to SNP-array markers. When direct representation was unavailable, we identified and incorporated linkage disequilibrium proxies into the analysis. Differences in risk-allele frequencies between breeds were evaluated using Fisher's exact test with false discovery rate correction. Individual burden scores were calculated to assess cumulative disease-associated variation. Thirty-five hereditary disease loci were identified, including 26 loci represented directly on the SNP array and eight represented by LD proxies. The final dataset contained 33 polymorphic disease-associated SNP markers. Significant differences between breeds were detected for loci linked to congenital stationary night blindness (GRM6), glycogen storage disease type IV (GBE1), and polysaccharide storage myopathy type 1 (GYS1). Despite these differences, we observed no significant difference in the cumulative burden of disease-associated alleles between populations. Based on biological relevance, marker coverage, and population frequencies, we developed a candidate screening panel of eight markers (GRM6, GBE1, GYS1, EDNRB, RAPGEF5, SEL1L, PLOD1, and ACAN). Integration of OMIA-derived disease loci with genome-wide SNP genotypes enabled the development of a compact SNP panel suitable for monitoring hereditary disease-associated variation in Kazakh horse breeds. The proposed panel may support genetic monitoring, breeding management, and future validation studies of disease-associated variants in local horse populations.

Keywords: *Horse genetics, SNP panel, Genetic monitoring, Disease-associated variants, OMIA, Adai horse*

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INTRODUCTION

Molecular genetic testing has become an important component of contemporary horse breeding. Initially developed to diagnose individual inherited disorders, genetic testing is increasingly used to support breeding decisions, identify carriers of deleterious alleles, and reduce the transmission of harmful variants within breeding populations (Bellone, 2020). As the number of characterized disease-associated variants continues to expand, attention has shifted from single-mutation assays to approaches that can simultaneously evaluate multiple loci.

Progress in equine genomics has provided the foundation for this transition. The publication of the horse reference genome and subsequent improvements in genome assembly quality greatly facilitated the discovery and characterization of genetic variation across breeds (Wade *et al*., 2009; Kalbfleisch *et al*., 2018). High-density SNP arrays further expanded genomic analyses by enabling simultaneous interrogation of tens of thousands of markers across the genome (McCue *et al*., 2012; Martin & Anderson, 2024). These platforms have been widely used to study population structure, genetic diversity, selection signatures, and economically important traits in horses (Petersen *et al*., 2013).

At the same time, researchers have genetically characterized a growing number of hereditary disorders. Disease-associated variants have been reported in genes involved in muscle metabolism, connective tissue development, vision, neurological function, skeletal growth, and gastrointestinal physiology. The Online Mendelian Inheritance in Animals (OMIA) database continuously curates information on these variants and remains the principal repository of inherited traits and genetic disorders in domestic animal species (Nicholas,

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2003; Morin *et al*., 2023). The growing availability of validated disease-associated loci creates opportunities to develop screening tools that can evaluate multiple variants in a single assay.

Most currently available genetic tests in horses target individual mutations and are designed for specific diagnostic purposes. Although highly effective for confirming particular genetic conditions, this approach becomes less practical when the objective is population-level monitoring of numerous disease-associated variants. An alternative strategy is to integrate information from existing SNP genotyping datasets with loci cataloged in OMIA. This approach allows researchers to repurpose previously generated genomic data for genetic screening without additional large-scale laboratory testing. Whole-genome sequencing studies have demonstrated that horse populations harbor substantial numbers of potentially deleterious variants, emphasizing the importance of monitoring disease-associated alleles in breeding programs (Jagannathan *et al*., 2018).

SNP-based screening is particularly relevant for local horse populations that remain poorly characterized for disease-associated genetic variation. In such populations, routine genetic testing is uncommon, and information regarding the distribution of risk alleles is often limited. Existing genome-wide SNP datasets therefore represent a valuable resource for identifying informative markers and establishing the basis for future genetic monitoring programs.

Kazakhstan possesses several indigenous horse populations that represent an important component of the country's animal genetic resources. Among them, the Adai horse is recognized for its adaptation to arid environments and extensive management systems. In contrast, the Kostanai breed was developed through systematic crossbreeding involving local horses and imported riding breeds. Previous genetic studies have demonstrated considerable diversity within Kazakh horse populations and highlighted their distinct evolutionary history (Gemingguli *et al*., 2016; Librado *et al*., 2021; Kariamal & Angadi, 2023). More recent investigations based on SNP genotyping and whole-genome sequencing have expanded knowledge of genomic variation in Kazakh horses and provided valuable resources for population-level analyses (Assanbayev *et al*., 2024; Pozharskiy *et al*., 2024; Beishova *et al*., 2025). However, information on hereditary disease-associated variants in these populations remains scarce.

To date, no study has systematically integrated OMIA-derived disease loci with genome-wide SNP genotypes to evaluate disease-associated variants in Adai and Kostanai horses. Furthermore, no compact SNP panel has been proposed for routine monitoring of hereditary disease-associated variation in these breeds. Developing such a panel would increase the utility of existing genomic datasets and provide a practical tool for genetic management of local horse populations.

The objectives of the present study were therefore to (i) evaluate the representation of known equine hereditary disease loci within a high-density SNP dataset, (ii) characterize the distribution of disease-associated variants in Adai and Kostanai horses, and (iii) develop a compact SNP panel suitable for population-level monitoring of hereditary disease-associated variation in Kazakh horse breeds.

MATERIALS AND METHODS

Animals, genotyping and quality control

The study used genome-wide SNP genotypes from two horse breeds maintained in Kazakhstan. After quality-control procedures, the final dataset included 341 unrelated animals: 291 Adai and 50 Kostanai horses. We genotyped the horses using a high-density equine SNP array with approximately 80,000 markers distributed across the genome.

Quality control was conducted using PLINK v1.9 and PLINK v2.0. Closely related individuals were identified using the KING algorithm and removed prior to analysis. Markers showing substantial deviation from Hardy–Weinberg equilibrium ($P < 1 \times 10^{-6}$) or poor genotyping performance were excluded. After filtering, 74,210 SNPs remained for downstream analyses. The overall genotyping rate was 99.94%.

Identification and mapping of disease-associated loci

We obtained information on hereditary disorders and disease-associated variants in horses from the Online Mendelian Inheritance in Animals (OMIA) database (Nicholas, 2003). Phenotypes related exclusively to coat color, morphology, body size, gait, performance, or other non-pathological traits were excluded. Only loci associated with inherited disorders or clinically relevant disease phenotypes were retained.

Disease-associated variants were extracted together with their genomic coordinates and gene annotations. Coordinates were converted to the EquCab3.0 reference assembly when necessary. The resulting set of loci was compared with SNP positions present on the genotyping array.

Direct matches between OMIA variants and array markers were identified using genomic coordinates and marker annotations. When direct representation was unavailable, linkage disequilibrium (LD) proxies were searched within a ±500 kb genomic window using genome-wide genotype data. Candidate proxies were retained when pairwise linkage disequilibrium exceeded $r^2 = 0.60$.

This procedure produced a dataset of disease-associated SNPs and validated LD proxies **(Figure 1)**.

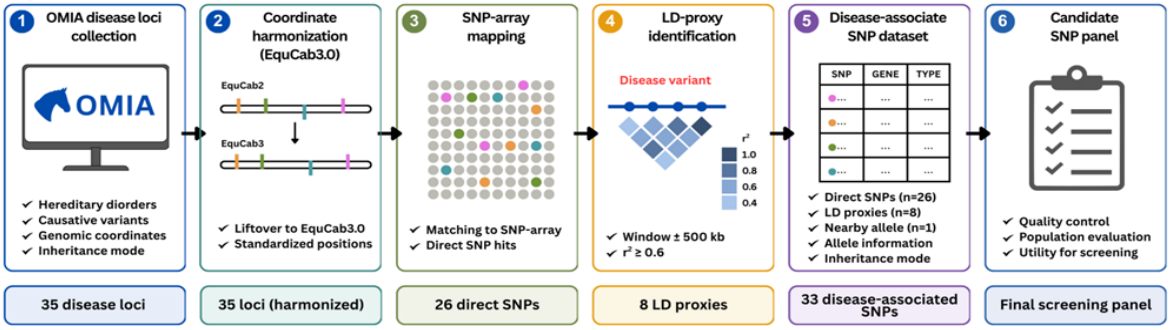


Figure 1. Workflow used for development of the disease-associated SNP panel.

Analysis of disease-associated variants

Risk alleles were assigned according to OMIA annotations and the original publications describing the corresponding variants. Allele frequencies were calculated separately for Adai and Kostanai horses.

Differences in risk-allele frequencies between breeds were evaluated using Fisher's exact test. To account for multiple testing, p-values were adjusted using the Benjamini–Hochberg false discovery rate (FDR) procedure. Markers with FDR-adjusted P-values < 0.05 were considered statistically significant.

To assess cumulative disease-associated variation, we calculated an individual burden score across all disease-associated SNPs. Burden values were compared between breeds using the Mann–Whitney U test. In addition, disease-associated loci were grouped into broad pathology categories derived from OMIA annotations, and aggregate differences were assessed using Stouffer's method.

Carrier frequencies were estimated from genotype distributions according to the reported mode of inheritance. Because clinical and phenotypic information was unavailable, genotype classes were interpreted only in terms of the presence of disease-associated alleles and were not considered evidence of disease status.

Development of the candidate SNP panel

Markers were considered for inclusion in the final screening panel according to their association with a documented hereditary disorder, representation by a direct SNP or high-confidence LD proxy, occurrence within the studied populations, carrier frequency, and biological relevance. The final panel was designed as a population-level tool for monitoring disease-associated genetic variation rather than as a diagnostic assay for individual disorders.

Because phenotypic information was unavailable, all analyses were restricted to disease-associated alleles and genotypes, and no inference regarding disease prevalence or clinical status was made.

All analyses were performed using PLINK v1.9, PLINK v2.0, and custom Python scripts developed for data processing, statistical analyses, and visualization.

RESULTS AND DISCUSSION

Coverage of OMIA loci and construction of the disease-associated SNP dataset

A total of 329 equine phenotypes available in the OMIA database were initially evaluated. After excluding coat-color traits, morphological characteristics, performance traits, and other non-pathological phenotypes, 35 hereditary disease loci remained eligible for analysis.

Comparing OMIA-derived variants with SNP-array markers showed extensive coverage of disease-associated genomic regions. Twenty-six loci were represented directly by markers present on the array, whereas eight loci required the use of linkage disequilibrium (LD) proxies. One additional locus was represented through a nearby marker corresponding to a reported structural variant. As a result, all selected OMIA loci were represented within the analytical framework.

Using LD proxies substantially increased the number of loci available for analysis. Several clinically relevant variants, including loci associated with polysaccharide storage myopathy type 1 (GYS1), glycogen storage disease type IV (GBE1), overo lethal white foal syndrome (EDNRB), and congenital stationary night blindness (GRM6), were captured through markers with strong linkage to the reported disease-associated variants.

The final dataset consisted of 33 polymorphic disease-associated SNP markers representing a broad spectrum of hereditary conditions affecting neurological, metabolic, developmental, connective tissue, hematological, endocrine, and sensory systems.

Distribution of disease-associated variants in Adai and Kostanai horses

Risk-allele frequencies were compared between breeds for all 33 disease-associated SNP markers. The overall distribution of disease-associated variation was similar in the two populations. The mean risk-allele frequency was 0.159 in Adai horses and 0.162 in Kostanai horses. Likewise, the average individual burden score was 0.405 and 0.411, respectively. No significant difference in cumulative burden was detected between breeds (Mann–Whitney P = 0.358).

Figure 2 summarizes differences in risk-allele frequencies for individual markers. Most loci showed comparable frequencies in the two populations; however, three markers remained significant after false discovery rate correction **(Figure 2; Table 1)**. These markers were associated with congenital stationary night blindness (GRM6), glycogen storage disease type IV (GBE1), and polysaccharide storage myopathy type 1 (GYS1).

Markers linked to GRM6 and GBE1 occurred more frequently in Adai horses, whereas the marker associated with GYS1 was more common in the Kostanai population. **Table 1** presents

detailed estimates of frequency differences and adjusted p-values.

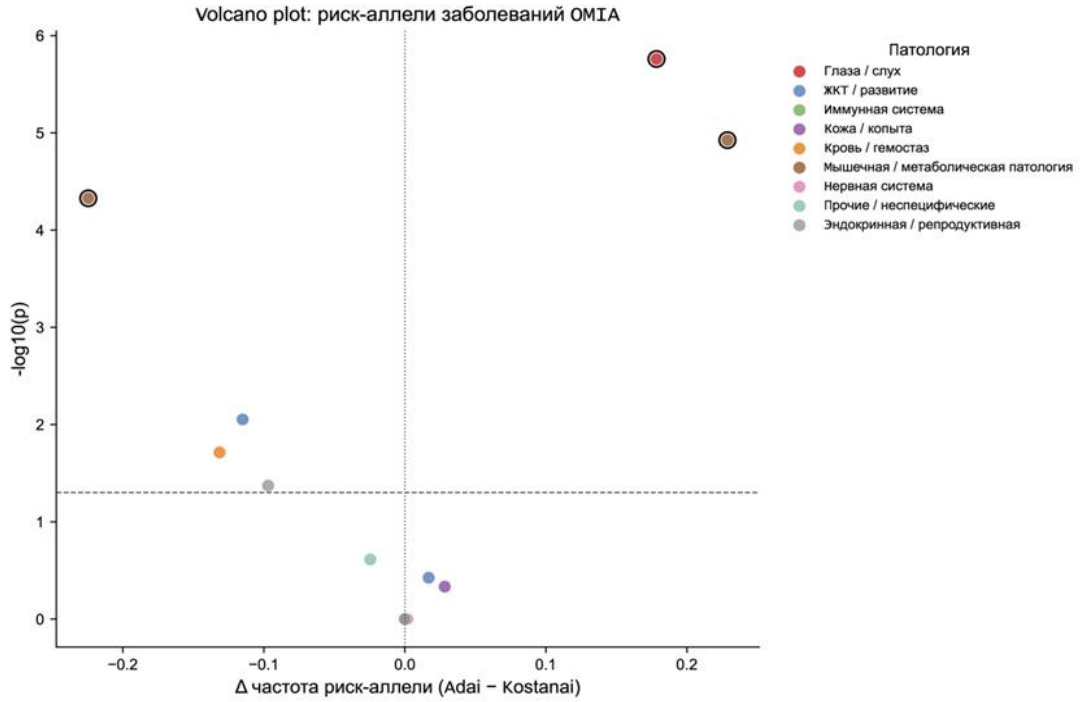


Figure 2. Volcano plot of disease-associated SNP markers showing differences in risk-allele frequencies between Adai and Kostanai horses. Distribution of disease-associated SNP markers according to the magnitude of allele-frequency differences between breeds and their statistical significance. Markers remaining significant after false discovery rate correction are highlighted and annotated.

At the pathology level, most categories showed comparable distributions between breeds. The only category exhibiting a significant aggregate effect was the group comprising eye and sensory disorders, largely driven by variation at the GRM6 locus. Overall, these results indicate that disease-associated variation is largely shared between the two populations, with differentiation concentrated in a limited number of loci rather than distributed across the entire marker set.

Table 1. Disease-associated SNP markers showing significant differences between Adai and Kostanai horses.

Parameter	Adai	Kostanai
Number of horses	291	50
SNPs after quality control	74,210	74,210
Genotyping rate	0.9994	0.9994
Mean disease burden	0.405	0.411
Mean risk-allele frequency	0.159	0.162

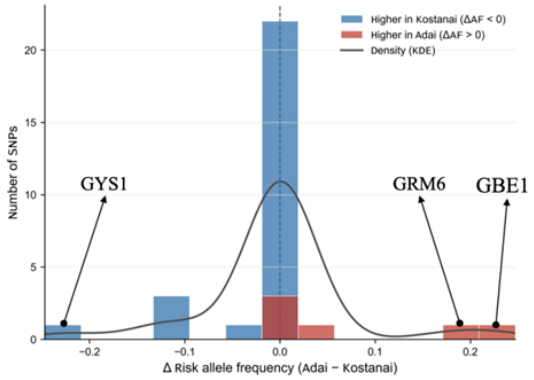
Carrier frequencies and burden of disease-associated alleles

Carrier frequencies varied substantially among loci and disorders **(Figure 3)**. The highest frequencies were observed for markers associated with polysaccharide storage myopathy (GYS1), atypical thrombasthenia (SEL1L), glycogen storage disease type IV (GBE1), hypoparathyroidism (RAPGEF5), and megacolon/overo lethal white foal syndrome (EDNRB).

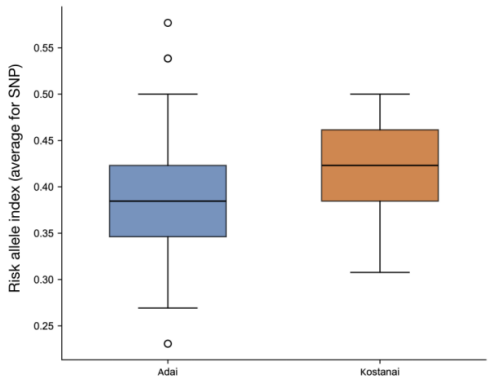
Several markers exhibited pronounced differences between breeds. The largest carrier-frequency differences were observed for GYS1, GRM6, EDNRB, GBE1, and RAPGEF5. In contrast, markers associated with dwarfism (ACAN), cerebellar abiotrophy (MUTYH), and ocular squamous cell carcinoma (DDB2) occurred at very low frequencies in both populations.

The distribution of cumulative burden scores was broadly similar between breeds **(Figure 3)**, supporting the observation that overall levels of disease-associated variation are comparable despite differences at several individual loci.

Because phenotypic information was unavailable, interpret carrier frequencies and homozygous risk genotypes as measures of disease-associated genetic variation rather than estimates of disease occurrence.



a)



b)

Figure 3. Distribution of disease-associated alleles in Adai and Kostanai horses: (a) Differences in risk-allele frequencies among disease-associated SNP markers, and (b) Distribution of individual burden scores in the two populations.

Development of the candidate SNP panel

The final screening panel was constructed using a combination of biological, population-genetic, and practical criteria. Priority was given to markers associated with well-characterized hereditary disorders, loci represented by direct SNPs or high-confidence LD proxies, and variants occurring at detectable frequencies within the studied populations.

The final panel included eight markers in or linked to the genes GRM6, GBE1, GYS1, EDNRB, RAPGEF5, SEL1L, PLOD1, and ACAN **(Table 2)**. Three of these loci (GRM6, GBE1, and GYS1) were included because they showed significant differences between populations, whereas the remaining markers were selected because of their carrier frequencies, biological relevance, and representation of major categories of hereditary disorders.

The rationale for inclusion of each marker is summarized in **Table 2**. Together, the selected loci cover multiple physiological systems, including vision, muscle metabolism, connective tissue integrity, hematological function, growth and development, and endocrine regulation.

The resulting panel was designed as a practical tool for population-level monitoring of disease-associated genetic variation in Kazakh horse breeds.

Table 2. Candidate SNP panel for monitoring hereditary disease-associated variation in Adai and Kostanai horse breeds.

Gene	Disease	Marker	Marker type	Δ risk allele frequency	FDR
GRM6	Congenital stationary night blindness	BIEC2_238897	LD proxy	+0.17	<0.05
GBE1	Glycogen storage disease type IV	BIEC2_682281	LD proxy	+0.25	<0.05
GYS1	Polysaccharide storage myopathy type 1	BIEC2-107192	LD proxy	−0.25	<0.05

This study shows that genome-wide SNP genotyping data can be repurposed to monitor hereditary disease-associated variation by integrating OMIA-derived loci. Although SNP arrays are primarily applied in studies of population structure, genetic diversity, and genomic selection, the growing availability of curated disease databases has expanded their potential utility for genetic health monitoring (Nicholas, 2003; McCue *et al*., 2012). This approach shows how existing genomic resources can be transformed into a practical screening tool without additional targeted genotyping.

A major outcome of the study was the extensive coverage of OMIA-derived loci achieved using the SNP-array dataset. Of the 35 hereditary disease loci retained after OMIA filtering, 26 were represented directly on the array, and eight additional loci were captured through high-confidence LD proxies. This level of coverage is consistent with the marker density and linkage disequilibrium structure previously reported for equine SNP arrays (McCue *et al*., 2012). Recovering disease-associated loci from existing genotyping resources substantially increases the value of archived SNP datasets and provides a cost-effective framework for population-level screening. Similar strategies have been widely adopted in livestock genomics, where dense SNP datasets and imputed markers are increasingly used to evaluate functional variation and genetic diversity simultaneously (Eusebi *et al*., 2019; Castillo-Guevara *et al*., 2025; Chessari *et al*., 2025).

Despite moderate genome-wide differentiation between Adai and Kostanai horses, most disease-associated variants exhibited comparable frequencies in the two populations. Only three loci, linked to GRM6, GBE1, and GYS1, remained significant after multiple-testing correction. Furthermore, cumulative burden analysis revealed no significant difference in overall disease-associated variation between breeds. These findings indicate that most risk alleles are shared between populations and that differentiation is concentrated in a small subset of loci. This pattern is consistent with observations from other domestic animal populations, where breed history and demographic processes often shape the distribution of disease-associated variants more strongly than direct selection acting on the variants themselves (Jagannathan *et al*., 2018).

The strongest differentiation was observed at the GRM6 locus. Variants in GRM6 have been associated with congenital stationary night blindness, a disorder affecting retinal signal transmission in low light (Hack *et al*., 2021). Although congenital stationary night blindness has been most extensively studied in relation to TRPM1-associated forms of the disease, recent work identified a likely causal missense variant in GRM6 in Tennessee Walking Horses (Hack *et al*., 2021). Genetic testing for inherited ocular disorders is increasingly incorporated into breeding programs because visual impairment may directly affect animal welfare and performance (Bellone, 2020). The marked difference observed at the GRM6-associated marker therefore highlights this locus as a priority target for future validation in Kazakh horse populations.

Differences were also detected for loci associated with glycogen metabolism. The GYS1 gene is responsible for polysaccharide storage myopathy type 1 (PSSM1), one of the most extensively investigated inherited muscle disorders in horses (McCue *et al*., 2008). The causative mutation has been reported in numerous breeds and is particularly common in Quarter Horse- and draft-related populations (McCue *et al*., 2008). Functional studies show that the mutant glycogen synthase enzyme has

constitutively elevated activity, resulting in abnormal glycogen accumulation in skeletal muscle (Maile *et al*., 2016). More recent reviews continue to identify PSSM1 as one of the most important inherited metabolic disorders encountered in equine practice (Firshman & Valberg, 2025). Although Kostanai horses show higher frequencies of the disease-associated allele, the lack of phenotypic information prevents any inference about disease occurrence.

A similar interpretive caution applies to the GBE1 locus. Pathogenic variants in GBE1 are associated with glycogen storage disease type IV, a severe recessive disorder characterized by abnormal glycogen branching and often fatal outcomes in affected foals (Ward *et al*., 2004). Identifying the causative mutation enabled DNA-based carrier testing and remains a classic example of successful implementation of molecular diagnostics in horse breeding (Ward *et al*., 2004). Unlike GYS1, the GBE1-linked marker showed a higher risk-allele frequency in Adai horses; this contrast indicates that the locus remains relevant for future monitoring programs, particularly if larger population datasets become available.

An important methodological consideration is the use of linkage disequilibrium proxies. The eight loci included in the analysis were represented by markers linked to, but not identical to, the reported disease-associated variants. Although this approach substantially increased locus coverage, the relationship between a proxy marker and a causal variant depends on local LD structure and may differ among populations. Consequently, frequencies reported for proxy markers should not be interpreted as direct estimates of causal variant frequencies. Similar limitations have been discussed in studies evaluating imputed genotypes and SNP-array-based analyses in livestock populations (Eusebi *et al*., 2019; Chessari *et al*., 2025). Validation through targeted genotyping or sequencing therefore remains necessary before making any breeding recommendations for specific loci.

The present study should also be interpreted in the context of several additional limitations. First, the Kostanai sample size was substantially smaller than the Adai sample, reducing statistical power for rare variants. Second, the analysis was restricted to currently described OMIA loci and therefore could not identify previously unreported disease-associated variants. Third, no clinical or phenotypic information was available. As a result, the study evaluated disease-associated alleles and genotypes rather than confirmed hereditary disorders. The observed frequencies should therefore not be interpreted as estimates of disease prevalence.

Recent genomic studies have highlighted the considerable genetic diversity present within Kazakh horse populations and have demonstrated the value of these breeds as regional genetic resources (Gemingguli *et al*., 2016; Assanbayev *et al*., 2024; Pozharskiy *et al*., 2024; Beishova *et al*., 2025). Incorporating disease-associated markers into existing genomic datasets represents a logical extension of these efforts. From a practical perspective, the proposed panel was intentionally designed as a monitoring tool rather than a diagnostic assay. The selected markers represent multiple categories of hereditary disorders, including neurological, metabolic, connective tissue, developmental, endocrine, and hematological conditions. Such a panel may support routine genetic monitoring, help identify carriers of disease-associated alleles, and contribute to breeding strategies that maintain genetic diversity while limiting the accumulation of potentially deleterious variants.

Overall, the study shows that integrating OMIA-derived information with genome-wide SNP data provides an effective framework for developing compact screening panels in understudied horse populations. The proposed marker set offers a practical starting point for future validation studies and for incorporation of genetic health monitoring into breeding programs for Kazakh horse breeds.

CONCLUSION

Integration of OMIA-derived disease loci with genome-wide SNP genotyping data enabled the development of a compact panel for monitoring hereditary disease-associated variation in Kazakh horse breeds. Of the 35 disease loci identified from OMIA, all were represented through direct SNPs or validated linkage disequilibrium proxies, resulting in a dataset of 33 polymorphic disease-associated markers. Most variants showed similar frequencies in Adai and Kostanai horses, although significant differences were detected at loci linked to GRM6, GBE1, and GYS1. Based on biological relevance, marker coverage, and population frequencies, an eight-marker screening panel was proposed. The panel is intended as a population-level monitoring tool rather than a diagnostic assay and may support genetic management and breeding programs in Kazakh horse populations. Future studies incorporating larger sample sizes, direct genotyping of causal variants, and phenotypic information will be necessary to validate the practical significance of the identified markers.

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CONFLICT OF INTEREST: None

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ETHICS STATEMENT: The study used previously obtained DNA samples from horses of the Adai and Kostanai breeds. The collection of biological material was carried out within the framework of planned selection and breeding work without harming the animals. All work was carried out in accordance

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