



Genotyping of Ukrainian Large White pigs by intronic polymorphism in *HMGA1* gene

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TV: Investigation; Visualisation; Writing — original draft.

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Authors declare no conflict of interests.

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The use of animals in this study was approved by the Scientific Council of the Institute of Pig Breeding and Agro-Industrial Production (protocol no. 1 from 08.01.2024) and was conducted in accordance with the European Convention for the Protection of Vertebrate Animals Used for Experimental and Other Scientific Purposes (Strasbourg, 18 March 1986).

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The *HMGA1* (High Mobility Group AT-hook 1) gene is considered a candidate locus for growth- and fatness-related traits in pigs due to its involvement in transcriptional regulation and energy metabolism and its localization within a quantitative trait locus (QTL) region on *Sus scrofa* chromosome 7. The aim of this study was to characterize a previously described intronic polymorphism of the *HMGA1* gene in the population of Ukrainian Large White pig breed and to evaluate its population genetic parameters for potential application in marker-assisted selection. An *in silico* analysis based on the Sscrofa11.1 reference genome was performed to define the genomic localization of the amplified fragment, describe the polymorphism according to Ensembl nomenclature, and identify its rsID in the European Variation Archive. Genotyping of 30 Ukrainian Large White pigs was carried out using the polymerase chain reaction–restriction fragment length polymorphism (PCR–RFLP) method with the restriction endonuclease *NaeI*. The polymorphic site was localized at position 7:30,326,531 and identified as rs80981303 (c.220-170C>T). Population analysis revealed genotype frequencies of 0.10 (CC), 0.40 (CT), and 0.50 (TT), with allele frequencies of 0.30 for C and 0.70 for T. The distribution of genotypes did not deviate from Hardy–Weinberg equilibrium ($\chi^2 = 0.068$, $p = 0.794$). The polymorphic information content value (PIC=0.3318) indicated that the locus is sufficiently informative. The obtained results confirm the polymorphic nature of the *HMGA1* gene in the Ukrainian Large White pig breed and support the potential utility of rs80981303 as a genetic marker. Further association studies with productive traits could substantiate its practical application in pig breeding programs.

Key words: HMGA1 gene, pig, Ukrainian Large White breed, intronic polymorphism, PCR–RFLP, population genetics, marker-assisted selection



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Introduction

Single nucleotide polymorphisms (SNPs) represent common genetic variants distributed throughout the porcine genome and constitute a valuable source of information for dissecting the genetic basis of economically important traits in pigs [17]. High-density

SNP genotyping and genome-wide association studies (GWAS) have substantially contributed to the identification of quantitative trait loci (QTLs) associated with growth, fatness, carcass composition, and meat quality traits in diverse pig populations, thereby enhancing the prospects of marker-assisted and genomic selection in swine breeding programs [20–21, 29]. The Pig QTL

database [13–14] currently catalogs multiple QTLs for growth and fatness attributes, highlighting the complex nature of the relationships between genes and productivity traits and the importance of considering animal genotypes to ensure efficient livestock production.

Among the candidate genes for animal productivity, the *HMGA1* (High Mobility Group AT-hook 1) gene has emerged as a locus of interest in porcine genetics [6, 9–12, 16, 18–19, 22–24, 27, 29, 31, 34–35]. The *HMGA1* gene encodes a non-histone chromatin structural protein that functions in transcriptional regulation through modulation of chromatin architecture, with documented roles in cell proliferation, differentiation, and metabolic regulation in mammals [4, 15, 30]. Although most functional characterizations of *HMGA1* have been conducted in human and model systems [4, 8, 15, 30], its genomic position on *Sus scrofa* chromosome 7 (SSC7) coincides with loci repeatedly implicated in backfat and growth traits in GWAS and QTL mapping studies, rendering it a biologically plausible candidate gene for performance traits in pigs [6, 9–10, 16, 22–23, 27, 29, 31, 34–35].

Genome-wide association studies in multiple pig populations have identified *HMGA1* among candidate genes associated with external and carcass traits. In large-scale analyses of growth traits, SNPs mapping near or within the *HMGA1* locus have been linked with body size and body length [10, 16, 22, 29, 31], indicating a potential role in skeletal and overall growth dynamics. *HMGA1* also exhibited associations with backfat thickness, supporting its relevance for fat deposition traits and carcass composition [6, 9]. Expression profiling and polymorphism analyses demonstrated that *HMGA1* mRNA is widely expressed across tissues, and specific coding and 3'-UTR SNPs were significantly correlated with backfat thickness in pigs [11]. Furthermore, genome scans have implicated *HMGA1* as a candidate influencing the size of forelimb bones, suggesting its multifunctional involvement in both growth and carcass structure [34–35].

Despite these associations, the precise causal variants within the *HMGA1* gene that affect phenotypic variation in growth and fatness traits remain incompletely resolved. The integration of functional polymorphisms with well-characterized phenotypes offers a promising approach to improve selection decisions. In domestic pig populations characterized by different breeding histories and trait emphases, the variability of *HMGA1* allelic forms suggests opportunities for selection targeting body size, fat distribution, and carcass composition.

In the context of Ukrainian pig breeding, including indigenous and commercial populations such as the Ukrainian Large White, identifying and validating molecular markers linked to economically significant traits is of particularly interest for enhancing meat productivity and preserving local genetic resources. The *HMGA1* gene, with its demonstrated associations with growth and fatness metrics, presents a candidate marker for incorporation into marker-assisted selection (MAS) panels alongside other well-established markers such as *IGF2*,

MC4R, and *RYR1* [2]. Such integration has the potential to refine genetic evaluation frameworks and support breeding programs tailored to both production efficiency and adaptive performance.

Therefore, this study aimed to characterize a previously described intronic polymorphism of the *HMGA1* gene, identifiable by the polymerase chain reaction–restriction fragment length polymorphism (PCR–RFLP) method, in the Ukrainian Large White pig population and to evaluate its allelic and genotypic distribution with respect to its potential application in genetic improvement strategies.

Materials and Methods

To determine the precise genomic localization of the studied polymorphism in the *HMGA1* gene within the Sscrofa11.1 reference genome [32], to describe this variant according to Ensembl [7] nomenclature, and to identify its rsID in the European Variation Archive (EVA) [3], a multi-step *in silico* analysis was performed. First, the positions of the primers described in [19] were mapped to *Sus scrofa* chromosome 7 using the BLAST tool [1] implemented in the Ensembl genome browser. The chromosomal distance between the primer binding sites was then calculated and compared with the expected amplicon length. Based on the sizes of the restriction fragments obtained after digestion, the putative position of the polymorphic site within the amplified fragment was inferred. Subsequently, the presence of the corresponding restriction site was verified, and the registered polymorphism in EVA matching this genomic position and its allelic variants was identified.

For laboratory analyses, bristle samples were collected from 30 Ukrainian Large White pigs maintained at the State Enterprise “Research Farm “Stepne” (Poltava region, Ukraine). All experimental procedures were carried out in the Genetics Laboratory of the Institute of Pig Breeding and Agroindustrial Production of the National Academy of Agrarian Sciences of Ukraine.

Genomic DNA was extracted from bristles using *Chelex 100* reagent according to the method described in [28]. Isolated DNA samples were stored at –20 °C until further analysis. Genotyping was performed using the polymerase chain reaction (PCR) technique, following approaches described in [5, 33]. PCR was conducted in a total reaction volume of 25 µl using reagents from New England Biolabs (USA) in 0.5 ml *Eppendorf* microcentrifuge tubes (*Eppendorf*, Germany) on a *Biometra* thermal cycler (Germany). Structure of primers [19] is shown in table 1. Amplification parameters, including annealing temperature and number of cycles, were optimized experimentally based on primer characteristics and data from previous studies [19, 24].

Restriction fragment length polymorphism (RFLP) analysis was performed using the restriction endonuclease *NaeI* [19]. PCR products and restriction fragments were separated by electrophoresis in 0.2 % agarose gel. Fragment sizes were determined using the pBR322/MspI

Table 1. Primers structure, GC content and melting temperature

Primers	GC, %	T _m , °C		
		Calculated	Kim et al., 2004 [19]	Makgahlela et al., 2009 [24]
5'-AGAAGGAGCCCAGCGAAGT-3'	57.9	65.7	62	60
5'-ACAGTGCTCACCCAATGGC-3'	57.9	66.7		

molecular weight marker. Gel images were documented under ultraviolet illumination using a Canon digital camera.

Population genetic analysis was conducted using the *GenAlEx* 6.5 software package [25–26]. Allele frequencies, observed and expected genotype frequencies, observed and expected heterozygosity, and polymorphic information content (PIC) were calculated. Deviation from Hardy-Weinberg equilibrium was assessed using the chi-square (χ^2) test.

Results and Discussion

According to the results of the BLAST search performed within the *Sus scrofa* reference genome, the forward primer with the sequence 5'-AGAAGGAGCCCAGCGAAGT-3' was localized on chromosome 7 at positions 7:30,325,937–30,325,955, while the reverse primer with the sequence 5'-ACAGTGCTCACCCAATGGC-3' was mapped to positions 7:30,326,640–30,326,658. This primer pair spans exon 4 and the majority of intron 4 of the *HMGA1* gene (intron numbering according to the transcript ENSSSCT00000098878.1). Consequently, the actual length of the amplified fragment is 722 bp. In earlier studies [19, 24], the amplification of a 700 bp fragment using this primer system was reported; however, in the present work, the precise amplicon size was defined based on the current reference genome assembly.

AGAAGGAGCCCAGCGAAGTGCCAACACCTAAAAGACCTCGGGGCCGACCA
AAGGGGAGCAAAACAAGGGCGCCGCAAGACCCGGTGAGGCTTGAAGG
GGTGGCTCCTGGTGGAGGGAAGTGGGAAGTAACCCCGTCCCTGCAAGC
AGCTGAGGGAGGTCTGGGAAGGGTGGGCTTGTCTGATTCTCTGCATGC
CCTTCTCTGGTACGTGGGCGCGATGGGTCTTGGCTAGTTGAGGAAAGTG
GGTGATGGCCGAGGCCTAAGTCTAGGGCCTTGTCTTGGCCAGGACACT
GGGAAGTCAAGTCAGATGTCCAGAGCTTCTGGTCTGGAGGGAGGCC
AGTTGGGCAGAAATGGAGGGCTGTTCCCGCTGGGCTGAGATGTACCTCCC
CCCCAACCCAGGCCCGCTGGGTCTGAGGGTGGGGAGCAGGCAAGGCC
AGATCTACAGTGGCATGGCCTTTGGAGAAGTTGTTTGTATTTATTTT
ATTTTCTAAGACACGACTCATATCCTCTGAGTCACGGGTGAAGGAGGG
AGTGGGGCGGTGTGTGTATGTTGGGGTGGGGGCGGTGTGGCCGCGCCA
GTCATCCCCAGCTGGACTCCGGTGGGCTGCTGGGCTGAGAGTCCCGGCT
GCCCTCCCTGCTTGGCTCGCCCTCCAGGGCACTGGTCACTGCGGGCA
CCCGCCATTGGGTGAGCACTGT

Fig. 1. Fragment of the *HMGA1* gene (genomic location: 7:30,325,937–30,326,658) amplified in this study. Primer binding sites are underlined. Exon 4 (genomic location: 7:30,325,939–30,326,022) is highlighted in green. The polymorphic site rs80981303 (c.220-170C>T, genomic location: 7:30,326,531) is highlighted in blue.

Previous publications indicated that the analyzed polymorphism in the *HMGA1* gene is located within a recognition site of the restriction endonuclease *NaeI* (GCC/GGC). In the presence of the C allele, digestion was expected to yield two fragments of approximately 580 bp and 120 bp, whereas in the presence of the T allele, restriction does not occur and a single fragment of 700 bp is visualized by electrophoresis. Taking into account the refined amplicon size of 722 bp derived from the reference genome, cleavage at the *NaeI* site generates fragments of 595 bp and 127 bp. Accordingly, the TT genotype is represented by a single band corresponding to the undigested fragment, the CC genotype by two bands of 595 bp and 127 bp, and the CT genotype by three bands corresponding to the undigested and digested fragments.

The polymorphic site was localized on chromosome 7 at position 7:30,326,531 in the Sscrofa11.1 reference genome. According to Ensembl nomenclature, this variant is designated as c.220-170C>T relative to the transcript ENSSSCT00000098878.1. A single polymorphism registered at this genomic position is present in the EVA and is assigned the identifier rs80981303.

The amplified fragment of the *HMGA1* gene, including the positions of the primers and the polymorphic site, is shown in fig. 1.

During optimization of the PCR amplification conditions, several melting temperatures were evaluated. The calculated melting temperatures of the forward and reverse primers were 65.7 °C and 66.7 °C, respectively. Previous studies recommended melting temperatures of 62 °C [19] and 60 °C [24]. In the present study, an melting temperature of 60 °C was found to provide optimal amplification efficiency and specificity. The final PCR cycling program is presented in table 2.

The observed sizes of the PCR products and restriction fragments were consistent with the expected fragment lengths, as confirmed by electrophoretic analysis (fig. 2).

The results of the population genetic analysis based on genotyping of Ukrainian Large White pigs are summarized in Table 3. In total, 3 animals with the CC genotype, 15 animals with the TT genotype, and 12 animals with the heterozygous CT genotype were identified. Accordingly, the observed genotype frequencies were 0.10 for CC homozygotes, 0.50 for TT homozygotes, and 0.40 for CT heterozygotes. The calculated allele frequencies were 0.30 for the C allele and 0.70 for the T allele.

The expected heterozygosity slightly exceeded the observed heterozygosity (0.42 *versus* 0.40), indicating a near-equilibrium population structure for this locus. No statistically significant deviation from Hardy-Weinberg equilibrium was detected according to the χ^2 test ($\chi^2 = 0.068$, $P = 0.794$). The PIC value was 0.3318, which is considered relatively high for a biallelic locus and suggests that this polymorphism is informative and potentially suitable for use in directional selection within the Ukrainian Large White pig population.

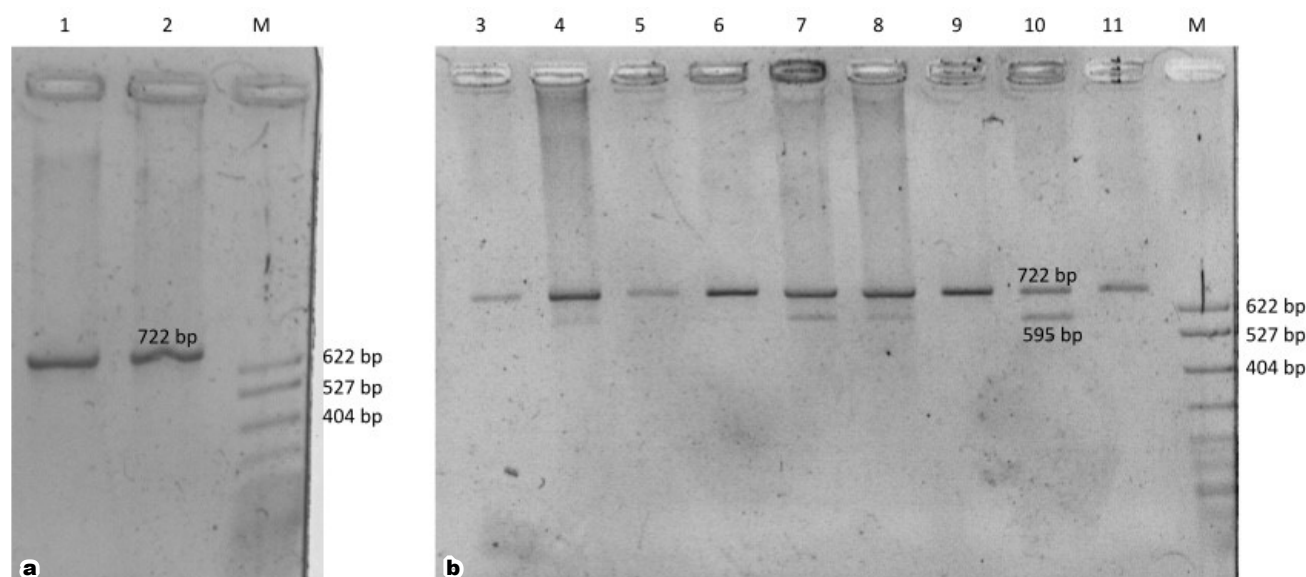


Fig. 2. Electropherograms of the *HMGA1* gene amplicon (a) and *NaeI* restriction fragments (b)

Note. 1, 2 — amplicon (722 bp); 4, 7, 8, and 10 — CT genotypes (722 bp and 595 bp); 3, 5, 6, 9, and 11 — genotypes TT (722 bp), M — pBR322/*MspI* molecular weight marker.

Comparison of the obtained results with previously published data demonstrates substantial variability in allele and genotype frequencies of the *HMGA1* polymorphism among pig breeds and populations. In the study by Kim et al. [19], nearly equal frequencies of CC and TT homozygotes were reported in the Large White breed, with a predominance of heterozygous CT animals. In contrast, a higher frequency of TT homozygotes was observed in Landrace and Duroc pigs. Conversely, in a later study by Kim et al. [18], the Duroc population exhibited an opposite pattern, with genotype frequencies of 0.43, 0.45, and 0.12 for CC, CT, and TT genotypes, respectively, indicating a predominance of CC homozygotes over TT homozygotes.

These discrepancies confirm that the distribution of *HMGA1* alleles and genotypes may vary considerably not only between breeds but also among populations within the same breed, reflecting differences in breeding history, selection pressure, and genetic background. Importantly, Kim et al. [19] demonstrated that in Landrace and Large White pigs, carriers of the C allele exhibited increased backfat thickness, with the highest values observed in CC homozygotes; the differences between genotypes were statistically significant ($P < 0.05$). In the present study, the frequency of the minor C allele was 0.30, indicating that animals carrying the potentially favorable genotype were relatively less common. Nevertheless, this minor allele frequency provides sufficient genetic variation to enable effective directional selection, allowing for an increase in the frequency of the target allele over successive generations.

The present study provided a detailed characterization of a previously described intronic polymorphism of the *HMGA1* gene in the Ukrainian Large White pig population. Using *in silico* analysis of the Sscrofa11.1 reference

genome, the exact genomic localization of the amplified fragment and the polymorphic site was defined, and the variant was identified as rs80981303 (c.220-170C>T) according to Ensembl and European Variation Archive nomenclature. Refinement of the amplicon length and restriction fragment sizes allowed for a precise interpretation of PCR-RFLP genotyping results.

Table 2. PCR program used for amplification of the *HMGA1* gene fragment

Gene	PCR amplification program		
	Temperature, °C	Time, s	Number of cycles
<i>HMGA1</i>	95°C	120	1
	95	30	31
	60	30	
	68	40	
	68	300	1

Table 3. Population genetic parameters of Ukrainian Large White pigs based on the rs80981303 (c.220-170C>T) polymorphism in the *HMGA1* gene

	Frequencies		Ho	He	χ^2	p-value	PIC
	Observed	Expected					
Alleles:							
C	0.30						
T	0.70						
Genotypes:			0.40	0.42	0.068	0.794	0.3318
CC (n=3)	0.10	0.09					
CT (n=12)	0.40	0.42					
TT (n=15)	0.50	0.49					

Note. Ho — observed heterozygosity, He — expected heterozygosity.

Population genetic analysis revealed that the studied locus is polymorphic in Ukrainian Large White pigs, with allele frequencies of 0.30 for the C allele and 0.70 for the T allele. The genotype distribution did not deviate from Hardy–Weinberg equilibrium. The PIC value equal to 0.3318 demonstrates that the rs80981303 polymorphism is sufficiently informative and suitable for use in genetic studies and breeding applications.

Further studies should focus on evaluating the population genetic characteristics of this *HMGA1* gene polymorphism in larger and samples, as well as on its combined analysis with other polymorphisms within *HMGA1* and additional candidate genes. Moreover, association studies are necessary to assess the relationships between this polymorphism and economically important productive traits in Ukrainian pig breeds. Such investigations will be essential to substantiate the practical applicability of *HMGA1* as a molecular marker for implementation in marker-assisted selection programs in pig breeding.

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Генотипування свиней української великої білої породи за інтронним поліморфізмом гена *HMGA1*

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Ген *HMGA1* (High Mobility Group AT-hook 1) вважається локусом-кандидатом для ознак, пов'язаних із ростом і вгодованістю у свиней, завдяки його участі в регуляції транскрипції та енергетичному метаболізмі та його локалізації в області на локусу кількісних ознак (QTL) на хромосомі 7 *Sus scrofa*. Метою цього дослідження було охарактеризувати раніше описаний інтронний поліморфізм гена *HMGA1* в українській популяції великої білої породи свиней та оцінити його популяційно-генетичні параметри для можливого застосування в маркер-асоційованій селекції. Аналіз *in silico* на основі референтного генома *Sscrofa11.1* був проведений для визначення геномної локалізації ампліфікованого фрагмента, опису поліморфізму відповідно до номенклатури Ensembl та ідентифікації його rsID в European Variation Archive. Генотипування 30 свиней української великої білої породи проводили методом полімеразної ланцюгової реакції–поліморфізму довжини рестрикційних фрагментів (ПЛР–ПДРФ) з ендонуклеазою рестрикції *NaeI*. Поліморфний сайт був локалізований у позиції 7:30,326,531 та ідентифікований як rs80981303 (с.220-170C>T). Популяційний аналіз виявив частоти генотипів 0,10 (CC), 0,40 (CT) і 0,50 (TT), з частотами алелів C — 0,30 і T — 0,70. Розподіл генотипів не відхилявся від рівноваги Харді–Вайнберга ($\chi^2 = 0,068$, $P=0,794$). Значення інформаційного змісту поліморфізму (PIC=0,3318) вказувало на достатню інформативність локусу. Отримані результати підтверджують поліморфність гена *HMGA1* у свиней української великої білої породи та підтверджують потенційну корисність rs80981303 як генетичного маркера. Подальші дослідження асоціації з продуктивними ознаками можуть обґрунтувати його практичне застосування в програмах розведення свиней.

Ключові слова: ген *HMGA1*, свиня, українська велика біла порода, інтронний поліморфізм, ПЛР–ПДРФ, популяційна генетика, маркер-асоційована селекція