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Research Article

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Genome-Wide Association Study of Body Measurements in Saburai Goat

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ABSTRACT

Body measurements are crucial indicators of growth performance and productivity in livestock. This study investigates the genetic basis of key body measurements in Saburai goats, a breed developed in Lampung Province, Indonesia, through grading up between Ettawa Grade does and Boer bucks. Genome-Wide Association Studies (GWAS) were employed to identify genetic markers associated with shoulder height, body length, and chest girth, which are important traits for growth and meat production. Phenotypic data from 100 Saburai goats were collected, and genotyping was conducted using the Illumina Goat SNP52 BeadChip. Statistical analyses using a mixed model identified significant genetic markers, including SNPs on chromosomes 6, 14 and 19, associated with traits such as chest girth, body length, and shoulder height. Several annotated genes, such as HDAC5 and CSN1S1, were implicated, highlighting their potential roles in growth and development. The results provide valuable insights into the genetic architecture of these traits, facilitating marker-assisted selection to enhance breeding programs. This research underscores the utility of integrating phenotypic and genotypic data to accelerate genetic improvement in livestock, offering a foundation for developing more productive and resilient Saburai goats. The findings also contribute to broader genomic studies in small ruminants, emphasizing the role of GWAS in modern livestock breeding strategies.

Key words: Genome-wide association study (GWAS), Saburai goat, Body measurements, Genetic markers, Breeding improvement.

INTRODUCTION

Body measurements in livestock play a critical role in assessing growth performance and animal productivity. In Saburai goats (the result of grading up between Ettawa Grade does and Boer buck with composition of 25% Ettawa Grade and 75% Boer goat) (Dakhlan et al. 2021a; Dakhlan et al. 2021b; Dakhlan et al. 2022), a new breed created in Lampung Province, Indonesia, known for its adaptability and meat production, identifying genetic factors that influence these physical traits is essential for improving breeding programs and enhancing productivity. Traditional breeding methods have achieved significant advancements, however, modern genomic approaches offer the potential for more precise selection strategies.

A Genome-Wide Association Study (GWAS) is a powerful tool used to identify genetic variants associated with specific traits by scanning the entire genome (Visscher et al., 2012). In livestock, GWAS has been widely employed to explore complex traits such as growth rate, milk yield, and body conformation (Wiggans et al. 2017; Keogh et al. 2021; Davoudi et al. 2024; Shangguan et al. 2024). By linking specific genetic markers, such as Single Nucleotide Polymorphisms (SNPs), with phenotypic traits, GWAS provides insights into the genetic architecture of important characteristics (Hayes et al. 2009; Weigel et al. 2024) such as in Saburai goats.

This study focuses on applying GWAS to investigate the genetic basis of body measurements in Saburai goats. Traits such as body length, chest girth, and shoulder height are critical indicators of growth and productivity in goats (Rahmatalla et al. 2018; Moaeen-ud-Din et al. 2022). Understanding the genetic variations that contribute to these traits can help breeders make informed decisions, improve the selection process and accelerate the genetic improvement of Saburai goats (Wakchaure et al. 2015; Magoro et al. 2022; Sharma et al. 2024).

The findings from this research have the potential to not only enhance breeding strategies but also contribute to the broader understanding of the genetic factors that influence growth-related traits in small ruminants.

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MATERIALS AND METHODS

Ethical approval

Ethical approval for the study was obtained from Faculty of Agriculture University of Lampung and all procedures were conducted in accordance with the relevant animal welfare regulations.

Animal population

A total of 100 Saburai goats were used in the study, including both males and females, with ages ranging from 1-1.5 years old. The animals were sourced from the Saburai goat breeding center in Tanggamus regency, Lampung province, Indonesia. The pedigree for each goat was traced back at least two generations.

Phenotypic data collection

Body measurements were recorded for each goat, including shoulder height (cm), body length (cm) and chest girth (cm). Standard livestock measurement techniques were followed, using a measuring tape and stick, as outlined in the protocols by Dakhlan et al. (2020).

DNA extraction and genotyping

Blood samples (5mL) were collected from the jugular vein of each goat using sterile syringes and stored in EDTA-coated tubes for genomic DNA extraction. Genomic DNA was extracted using the standard phenol-chloroform protocol (Sambrook and Russell 2006). DNA samples were genotyped using the Illumina Goat SNP52 BeadChip (Illumina Inc., San Diego, CA, USA) and quality control was carried out prior to conducting an association study. SNPs with a minor allele frequency (MAF) <0.05, missing genotypes >10%, or deviations from Hardy-Weinberg equilibrium (P<0.001) were excluded from the analysis. Samples with low call rates (<95%) were also excluded.

Statistical analysis

We utilized a mixed model approach, specifically the animal model method, implemented using the WOMBAT software (Meyer 2007; Meyer 2024), to accurately predict body measurements (shoulder height, body length, and chest girth) and residual data for each goat. The fixed effects in the animal model included birth year (grouped into rainy and dry seasons), sex (male or female), birth type (single, twin, or triplet), and rearing location (Gisting and Sumberejo Districts), with dam age as a covariate. Random effects comprised the individual goat and the dam. Residual data derived from the mixed model analysis were subsequently used for association studies. The mixed model is represented as follows:

$\mathbf{y} = \mathbf{X}\mathbf{b} + \mathbf{Z}_1\mathbf{a} + \mathbf{Z}_2\mathbf{m} + \mathbf{e}$

where y is the vector of observations, b represents the vector of fixed effects, a denotes the vector of additive genetic effects, m signifies the vector of maternal genetic effects and e is the vector of residual effects. X is the incidence matrix corresponding to the fixed effects, while Z_1 and Z_2 are incidence matrices linking the observations to the random effects of the animal (additive genetic) and the dam (maternal genetic), respectively.

Validation of results

Following quality control, 98 goats with 49,874 SNPs

were retained for the association analysis. Residual body measurements were individually regressed against SNPs using a linear model implemented in the R programming environment (R Core Team 2023). SNP annotation was performed using the *Capra hircus* genome reference (assembly ARS1.2) from the National Center for Biotechnology Information (NCBI) (https://www.ncbi.nlm.nih.gov/datasets/genome/GCF_001 704415.2/).

To determine genome-wide significance in the GWAS, the Bonferroni correction was applied, setting the threshold at P<0.05/52KP < 0.05/52KP<0.05/52KP<0.05/52K (Kaler and Purcell 2019; Chen et al. 2021). The Manhattan -log10 plot and P-value distribution for the GWAS results were visualized using the R package *qqman* (Turner 2018).

RESULTS AND DISCUSSION

Descriptive statistics of body measurements

The body measurements of the Saburai goats used in the Genome-Wide Association Study (GWAS) were analyzed to provide basic descriptive statistics, including mean, standard deviation, minimum, and maximum values. The body measurements analyzed included chest girth (CG), body length (BL) and height at the withers or shoulder height (SH). The summary of these measurements is shown in Table 1.

 Table 1: Descriptive statistics of body measurements (cm) in

 Saburai goat

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Trait	Mean±SD	Minimum	Maximum
Shoulder height	62.20 ± 5.85	41.21	69.10
Body length	60.91±7.01	43.17	75.16
Chest girth	66.80±7.94	43.39	75.14

The analysis of body measurements in Saburai goats provides foundational insights into the phenotypic diversity of the population, which is critical for effective breeding and genetic studies such as Genome-Wide Association Studies (GWAS). Table 1 presents the descriptive statistics for SH, BL and CG, showcasing phenotypic performance variability of Saburai goat population.

With a mean of 62.20±5.85cm and a range from 41.21 to 69.10cm, shoulder height demonstrates moderate variability. This trait is essential for assessing the stature and structural soundness of goats. Similar findings in other studies suggest that SH is often influenced by both genetics and growth conditions, such as feed quality and health management (Alade et al. 2008).

The mean body length was recorded at 60.91±7.01cm, ranging from 43.17 to 75.16cm. Body length is a critical indicator of growth and meat yield potential. The higher standard deviation relative to shoulder height may indicate greater genetic or environmental influences on this trait, consistent with findings in other goat breeds where body length serves as a proxy for productive capacity (Baker et al. 2017).

Chest girth exhibited the highest mean $(66.80\pm7.94\text{cm})$ and range (43.39-75.14cm). As an indicator of body capacity and health, CG variability highlights potential differences in nutrition and management practices. Studies on small ruminants have similarly emphasized chest girth as a predictor of live weight and overall productivity (Adamu et al. 2020).

The descriptive statistics provide a quantitative foundation for identifying phenotypic traits linked to genetic markers in GWAS. High variability in traits like CG and BL suggests significant opportunities for selective breeding to enhance growth and production traits. Previous research has shown that integrating phenotypic and genotypic data can optimize selection criteria and accelerate genetic improvement in livestock populations (Rexroad et al. 2019; Xu et al. 2020).

These measurements not only serve as benchmarks for evaluating the current performance of Saburai goats but also guide breeders in selecting animals with desirable traits. For instance, larger CG and BL values might be prioritized in meat production systems, while appropriate SH is critical for structural balance and adaptability. Additionally, these results can inform interventions to improve management practices, including feed supplementation and health programs.

Genome-wide association analysis

The GWAS was performed to identify single nucleotide polymorphisms (SNPs) associated with body measurements in Saburai goats. The analysis revealed several SNPs significantly associated with the measured traits, particularly chest girth. The top significant SNPs with a genome-wide significance level for each body measurement trait are listed in Table 2. While annotated gene with the significant SNPs are presented in Table 3. While Manhattan plot generated to visualize the association between the SNPs and body measurements are shown in Fig. 1-3 and the QQ-plot for GWAS result is shown in Fig. 4-6.



Fig. 1: Manhattan plot of genome-wide association analysis for shoulder height of Saburai goat.



Fig. 2: Manhattan plot of genome-wide association analysis for body length of Saburai goat.

The genome-wide association study (GWAS) conducted to identify single nucleotide polymorphisms (SNPs) associated with body measurements in Saburai goats provided significant insights into the genetic

architecture underlying these traits. The identification of SNPs associated with key body measurements such as chest girth, shoulder height and body length underscore the utility of GWAS in livestock breeding programs.



Fig. 3: Manhattan plot of genome-wide association analysis for chest girth of Saburai goat.



Fig. 4: Quantile-quantile plot of genome-wide association analysis for shoulder height of Saburai goat.



Fig. 5: Quantile-quantile plot of genome-wide association analysis for body length of Saburai goat.



Fig. 6: Quantile-quantile plot of genome-wide association analysis for chest girth of Saburai goat.

The analysis revealed that the SNP with the strongest association for chest girth was snp14750-scaffold1594-1124587, located on chromosome 19 (P-value= 1.65×10^{-26}). This finding is consistent with similar studies in other livestock species, where SNPs in the HDAC5 gene have been linked to growth and skeletal traits, suggesting a

Table 2: Significant SNPs Associated with Body Measurements in Saburai Goat

SNP ID	Chromosome	Position (bp)	Trait	P-value
snp14750-scaffold1594-1124587	19	43563475	Shoulder height	1.24×10^-11
snp53157-scaffold8-343582	14	1836428	Shoulder height	3.15×10^-11
CSN1S1-E-allele-1-2	6	85995008	Body length	3.85×10^-8
snp14750-scaffold1594-1124587	19	43563475	Body length	2.87×10^-19
snp29825-scaffold323-3014209	7	41699831	Body length	8.57×10^-8
snp38148-scaffold477-592558	2	124720482	Body length	4.69×10^-7
snp42099-scaffold55-2633854	3	59028955	Body length	4.07×10^-8
snp462-scaffold1011-1266531	9	19529989	Body length	1.29×10^-10
snp53157-scaffold8-343582	14	1836428	Body length	4.84×10^-15
CSN1S1-E-allele-1-2	6	85995008	Chest girth	1.36×10^-10
snp11085-scaffold14-482393	14	21520797	Chest girth	3.71×10^-11
snp14750-scaffold1594-1124587	19	43563475	Chest girth	1.65×10^-26
snp17841-scaffold185-1452068	11	72428670	Chest girth	8.24×10^-7
snp26621-scaffold278-501036	26	46053499	Chest girth	8.39×10^-9
snp29825-scaffold323-3014209	7	41699831	Chest girth	2.71×10^-9
snp38148-scaffold477-592558	2	124720482	Chest girth	7.18×10^-10
snp40114-scaffold511-3740135	6	61559529	Chest girth	7.31×10^-7
snp414-scaffold1010-28033	1	137123894	Chest girth	2.24×10^-7
snp42099-scaffold55-2633854	3	59028955	Chest girth	4.05×10^-9
snp462-scaffold1011-1266531	9	19529989	Chest girth	1.51×10^-14
snp52855-scaffold793-6368	11	48387776	Chest girth	9.23×10^-7
snp53157-scaffold8-343582	14	1836428	Chest girth	7.36×10^-18
snp5331-scaffold1184-194029	14	14654159	Chest girth	1.57×10^-9

Table 3: Significant SNPs and annotated gene

Table 5. Significant Sivi 8 and annotated gene					
SNP ID	Chromosome	Position (bp)	Range position	Gene name	
snp14750-scaffold1594-1124587	19	43563475	43553898-43590779	HDAC5	
snp414-scaffold1010-28033	1	137123894	1282061-1376560	SYNJ1	
snp38148-scaffold477-592558	2	124720482	124500873-124854226	ZNF804A	
snp42099-scaffold55-2633854	3	59028955	58458809-58770853	ADGRL2	
			60586459-60586530	TRNAC-ACA	
snp40114-scaffold511-3740135	6	61559529	61544757-61574269	BEND4	
CSN1S1-E-allele-1-2	6	85995008	85978463-85995270	CSN1S1	
snp29825-scaffold323-3014209	7	41699831	41642015-41708827	ITK	
snp462-scaffold1011-1266531	9	19529989	19489278-19567440	LOC106502469	
snp52855-scaffold793-6368	11	48387776	48350620-48437096	POLR1A	
snp17841-scaffold185-1452068	11	72428670	72403105-72492661	DPYSL5	
snp53157-scaffold8-343582	14	1836428	1699168-1707602	LOC106502869	
snp5331-scaffold1184-194029	14	14654159	14508816-14632179	SDC2	
			14668587-15236122	CPQ	
snp11085-scaffold14-482393	14	21520797	21215837-21817751	RIMS2	
snp26621-scaffold278-501036	26	46053499	45822146-46732977	PCDH15	

conserved genetic mechanism across species (Bradley et al. 2015; Klymenko et al. 2020; Wang et al. 2021). This SNP is particularly noteworthy, as its association with chest girth may align with findings from studies in other livestock species linking SNPs within the HDAC5 gene to growth-related traits. The exceptionally strong association observed here suggests that HDAC5 could play a pivotal role in regulating phenotypic variation in chest girth among Saburai goats, warranting further investigation and cross-species comparisons. This SNP is within the annotated gene HDAC5 (histone deacetylase 5), a gene previously implicated in various biological processes, including skeletal muscle development and growth regulation. Other significant SNPs include snp53157-scaffold8-343582 on chromosome 14 and snp462-scaffold1011-1266531 on chromosome 9. emphasizing a polygenic basis for chest girth.

For shoulder height, two highly significant SNPs were identified: snp14750-scaffold1594-1124587 (P-value=1.24×10^-11) and snp53157-scaffold8-343582 (P-value=3.15×10^-11). Both SNPs also showed associations with other traits, suggesting pleiotropic effects. While the

annotated genes for these loci were not specified in Table 3, their strong statistical significance highlights their potential as candidates for further functional analysis. Future steps should include experimental validation methods such as CRISPR-Cas9 gene editing to confirm the role of these SNPs and genes in trait development. Additionally, pathway analysis using transcriptomics or proteomics could help identify specific biological mechanisms and interactions influenced by these loci (Li et al. 2021; Sanches et al. 2024).

Multiple SNPs were significantly associated with body length, including snp14750-scaffold1594-1124587 on chromosome 19 (P-value= 2.87×10^{-19}) and snp462scaffold1011-1266531 on chromosome 9 (Pvalue= 1.29×10^{-10}). The annotated gene CSN1S1 (casein alpha s1) associated with CSN1S1-E-allele-1-2 on chromosome 6 suggests a potential link between milk protein genes and growth traits in goats. This association aligns with findings in other livestock species, where casein genes have been implicated in both milk production and growth-related traits, highlighting their multifunctional roles (Dettori et al. 2024; Caroli et al. 2004). For instance, studies in dairy cattle and sheep have reported similar dual roles of casein genes, linking them to both lactation and body development. However, some research has suggested that these effects might be influenced by genetic linkage or pleiotropic effects rather than direct causation, emphasizing the need for further functional studies to validate these associations. Conversely, some studies have questioned these links, attributing observed effects to genetic linkage rather than causation. Such insights underscore the need for functional validation and comparative studies across species to elucidate the mechanisms underlying this association. This finding with previous studies demonstrating aligns the multifunctionality of casein genes in livestock.

The functional annotations of significant SNPs revealed genes involved in diverse biological pathways. Several of the identified SNPs were located near annotated genes, potentially involved in growth, development, and metabolism. For instance, HDAC5 is known to regulate transcriptional activity and influence growth-related processes, associated with chest girth, is involved in histone deacetylation and could play a role in regulating genes affecting muscle growth and body structure. ZNF804A is associated with the zinc finger protein family, which plays roles in DNA binding and transcriptional regulation, may have regulatory functions in gene expression, potentially affecting growth traits indirectly. RIMS2 involved in synaptic membrane exocytosis, potentially linking neural regulation with growth. BEND4 and ITK is indicating roles in immune response and cellular signaling, which may indirectly affect growth traits. CSN1S1 on chromosome 6 is known for coding casein alpha s1, which is involved in milk protein synthesis. indicating that this gene might influence body development through nutrient utilization and energy partitioning.

The implications for breeding, the identification of these SNPs and their associated genes provide a foundation for marker-assisted selection (MAS) in Saburai goats. For example, SNPs linked to HDAC5 and CSN1S1 could be used to select for individuals with favorable chest girth and body length traits. Furthermore, the polygenic nature of these traits suggests the need for a genomic selection approach that incorporates multiple markers for optimal genetic improvement.

While the study highlights significant associations, several limitations need to be addressed, for example population size, larger sample sizes are required to validate the identified SNPs and ensure robust estimations of effect sizes. In addition, the causal relationship between the identified SNPs and the traits needs to be confirmed through functional studies. Furthermore, the interaction between genetic factors and environmental influences on body measurements should be investigated.

Future studies should focus on integrating these SNPs into genomic prediction models using methods such as best linear unbiased prediction (BLUP) or genomic best linear unbiased prediction (GBLUP) and exploring their effects in diverse goat populations. Additionally, transcriptomic and proteomic studies could elucidate the mechanisms by which these genes influence growth and body measurements.

Conclusion

This GWAS study has identified several significant

SNPs and associated genes that influence body measurements in Saburai goats. These findings provide valuable insights for genetic improvement programs and underscore the potential of integrating genomic tools into livestock breeding strategies.

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Author's Contribution: AD: responsible for designing the research, drafting the initial manuscript, and conducting statistical analyses. PES: managed methodology and supervision. DK: carried out data collection and did a literature review. All authors approved the final version of the manuscript.

Data Availability Statement: The data supporting this study's findings are available in this article. There is no other supporting data.

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