

# A genome-wide association analyses of leg conformation traits and stayability in sows

Young-Sin Kim\*, Joon-Ki Hong\*, Eun-Seok Cho\*, Yeon-Ho Kim\*, Ha-Seung Seong\*\*

\*Swine Science Division. National Institute of Animal Science, Rural Development Administration

\*\*Animal Breeding and Genetics Division. National Institute of Animal Science, Rural Development Administration  
e-mail:kys60m@korea.kr

## 돼지 다리 건강에 대한 전장유전체연관 분석

김영신\*, 홍준기\*, 조은석\*, 김연호\*, 성하승\*\*

\*국립축산과학원 양돈과

\*\*국립축산과학원 가축개량평가과

### Abstract

A study was conducted to estimate genetic relationship between sow stayability and leg conformation traits using records of Yorkshire and Landrace sows. The study recorded sow stayability from one parity to the next, with a score of 0 assigned when a sow had no farrowing record, a score of 1 assigned when a sow had one farrowing record, and a score of 2 assigned when a sow had the farrowing record more than once (parity > 1). The heritability of sow stayability and leg conformation traits was estimated using threshold animal models. The study also investigated genetic correlations among stayability and leg conformation traits. The heritability estimates for leg conformation traits were ranged from 0.15 to 0.63, and 0.03 for sow stayability. Genetic correlations between stayability and leg conformation traits were ranged from 0.35 to 0.83. Additionally, we found several genomic regions that might be potential signatures of selection for stayability and leg conformation traits such as AKAP11, EPST11 DNAJC15, and VWA8. The findings of this study showed the genetic relation between leg conformation traits are stayability in sows.

## 1. Introduction

Lameness is a significant issue in pig production that has both welfare and economic implications. Pain and distress are likely associated with lameness. Stress, which is linked to lameness, has been found to negatively impact the reproductive cycle of sows, leading to a lower number of ovulated eggs and an increased risk of re-mating [1]. Lameness has reduced daily weight gain compared to those without lameness, despite being treated with antibiotics for arthritis. Sows with leg weakness tend to exhibit uncontrolled lying-down behavior, which may increase the risk of crushing piglets, resulting in a lower litter size at weaning [2]. Moreover, leg weakness is one of the leading reasons for premature culling of sows, with several studies reporting that 9% to 13% of sows removed from commercial herds in Sweden, Finland, and Denmark were culled due to lameness [3]. Therefore, improving leg quality in pig herds is essential for both animal welfare and profitability in pig production.

## 2. Materials & Methods

### 2.1 Animals, Pedigree and Phenotype Data

The pedigree data of 23,754 animals, including 18,311 Yorkshire and 5,402 Landrace pigs, were collected from 2001 to 2021. The phenotype data for leg conformation was obtained from 6,472 animals, including 5,527 Yorkshire and 945 Landrace pigs. The reproduction data was collected from 11,745 sows (8,862 Yorkshire and 2,883 Landrace pigs).

### 2.2 Genotype Data

The gDNA was extracted from hair root samples and genotyped using the Neogen GGP porcine SNP 80K BeadChip, which includes 76,756 single-nucleotide polymorphisms (SNPs) for 935 animals. The quality control (QC) procedure was conducted for animals and SNP markers as the following criteria: (1) SNPs unmapped to *Sus scrofa* 11.1 or sex chromosomes; (2) SNPs with a call rate less than 90%; (3) SNPs with minor allele frequency less than 0.05; (4) animals with a call rate less than 90%; (6)

animals with Mendelian conflicts. The overall quality control process removed 27,583 SNPs and 29 animals, resulting in a total of 49,173 SNPs for 906 animals for further analyses.

**2.3 Estimates of heritability**

Heritability for leg conformation and stayability were estimated using the multi-trait model in THRGIBBS1F90 [4]. The Gibbs sampler was run a total of 500,000 rounds with single chains, and the first 200,000 rounds were excluded as burn-in rounds, thinning every 100 samples. Consequently, we used 3,000 samples for the subsequent post-Gibbs analysis in POSTGIBBSF90 [5]. Sex (male or female), breed (Yorkshire and Landrace), test year-month-day (106 levels) for leg conformations traits, and herd-year-month (124 levels) for stayability were used as the fixed effects. The birth litter (1,844 levels), and animal (23,777 levels) were used as random effects. Genetic analysis was performed using the animal model as follows:

$$y = Xb + Za + Ul + e,$$

where y is the vector of observation, b is the vector of fixed effects, a is the vector of additive genetic effects, l is the vector of random birth litter, e is the vector for the residuals, and X, Z, and U are the corresponding incidence matrixes.

**2.4 Single-Step Genome-Wide Association Study**

We conducted a genome-wide association study (GWAS) on traits of leg conformation and stayability using the ssGBLUP approach, which utilized all the data of genotype, phenotype, and pedigree in a single step. This method uses a realized relationship matrix which is also called H matrix. We obtained the G weight matrix generated by reciprocals of expected variance of markers as proposed by VanRaden [6] as follows:

$$G = ZDZ'q$$

where Z is the incidence matrix of genetic content that is altered for allele frequencies, D is the diagonal weight matrix of SNPs, and q is a normalizing factor. To identify potential genes related with leg conformation and stayability in pigs, we established a threshold for significant SNPs that had top 1% of the additive genetic variance. We then identified 1-Mb regions that contained significant SNPs in their centers and classified them as quantitative trait loci (QTLs). Using the Sus scrofa genome assembly 11.1 as reference, we annotated the genes within these QTLs.

**3. Results**

**3.1 Phenotypic statistics and genetic parameters**

Descriptive statistics of the 5 leg conformation traits and stayability are shown in Table 1. Among heritability value for leg conformation traits, the joint angle had the lowest value with  $0.15 \pm 0.03$ , and hoof width had the highest value with  $0.63 \pm 0.02$ . The stayability showed the low heritability of  $0.03 \pm 0.01$ .

[Table 1] Descriptive statistics of leg conformation and stayability traits and heritability

Traits	Number	Average	Heritability
Leg width	6,265	4.80±0.42	0.27±0.05
Leg angle	6,265	4.72±0.68	0.35±0.04
Joint angle	6,265	4.44±0.81	0.15±0.03
Hoof width	6,265	4.62±0.65	0.63±0.02
Overall score	6,265	8.58±1.49	0.38±0.04
Stayability	11,745	1.30±0.90	0.03±0.01

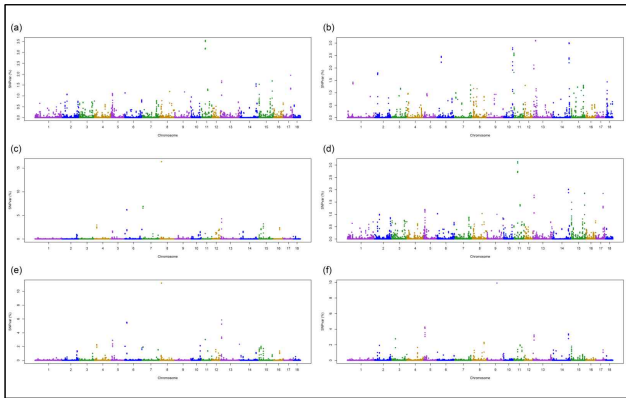
As shown in Table 2, the stayability had the high genetic correlations with leg width ( $0.83 \pm 0.07$ ), Hoof width ( $0.83 \pm 0.07$ ), and overall score ( $0.69 \pm 0.08$ ). Also, the genetic correlation between stayability and joint angle was favorable ( $0.48 \pm 0.11$ ). Among leg conformation traits, the genetic correlation was the lowest ( $0.37 \pm 0.07$ ) between leg angle and leg width, and the highest correlation was observed between hoof width and leg width with  $0.99 \pm 0.00$ .

[Table 2] Genetic correlation (above diagonal) between leg conformation traits and stayability.

Traits	Stability	Leg width	Leg angle	Joint angle	hoof width	Overall score
Stability	—	0.83 ±0.07	0.35 ±0.10	0.48 ±0.11	0.83 ±0.07	0.69 ±0.08
Leg width		—	0.37 ±0.07	0.63 ±0.07	0.99 ±0.00	0.81 ±0.03
Leg angle			—	0.59 ±0.09	0.38 ±0.07	0.71 ±0.06
Joint angle				—	0.64 ±0.07	0.93 ±0.02
Hoof width					—	0.83 ±0.03
Overall score						—

### 3.2 GWAS

Among the SNPs which had the top 1% of the variance explained by SNPs within 0.4 Mb-sized windows, we have located several genes that might be associated with leg conformation traits or stayability (Fig. 1). A QTL in SSC11 (24.04-25.07 Mb), we identified the AKAP11, EPSTI1, DNAJC15, and VWA8 genes which are previously reported to have association with bone density (EFO\_0003923), heel bone mineral density (EFO\_0009270), and calcium measurement (EFO\_0004838).



[Fig. 1] Results of single-step GWAS for leg conformation and stayability traits. SNPvar (%) is the variance explained by SNPs within 0.4 Mb-sized windows: (a) leg width, (b) leg angle; (c) joint angle (d) hoof width, (e) Overall score, and (f) stayability.

### References

- [1] Fitzgerald, Robert F., et al. "The effect of hoof abnormalities on sow behavior and performance." *Livestock Science* 145.1-3: 230-238. 2012.
- [2] Pluym, L. M., et al. "Prevalence of lameness and claw lesions during different stages in the reproductive cycle of sows and the impact on reproduction results." *Animal* 7.7: 1174-1181. 2013.
- [3] Heinonen, Mari, et al. "Lameness and fertility of sows and gilts in randomly selected loose housed herds in Finland." *Veterinary Record* 159.12: 383-387. 2006.
- [4] Tsuruta, S., and I. Misztal. "THRGIBBS1F90 for estimation of variance components with threshold and linear models." *Threshold* 3.4 2006.
- [5] Aguilar, Ignacio, et al. "BLUPF90 suite of programs for animal breeding with focus on genomics." *Proceedings of the world congress on genetics applied to livestock production*. 2018.
- [6] VanRaden, Paul M. "Efficient methods to compute genomic predictions." *Journal of dairy science* 91.11: 4414-4423. 2008.