

# ajava

Asian Journal of Animal and Veterinary Advances



Academic  
Journals Inc.

[www.academicjournals.com](http://www.academicjournals.com)

## Val126Ile Mutation Within Third Transmembrane Domain of the Melanocortin 1 Receptor (MC1R) is Associated with a Pigmentation of Plumage Color in Chicken

<sup>1</sup>Dongyep Oh, <sup>2</sup>KiEun Hyeong and <sup>3</sup>Yoonseok Lee

<sup>1</sup>Gyeongsangbuk-do Livestock Research Institute, Yeongju, 750-871, Gyeongsangbuk-do, Korea

<sup>2</sup>School of Biotechnology, Yeungnam University, Gyeonsan, 712-749, Korea

<sup>3</sup>Research Institute of Eco-friendly Animal Science, Institute of Green Bio Science and Technology, Seoul National University, Pyeongchang-gun, Kangwon-do, 232-916, Korea

*Corresponding Author: Yoonseok Lee, Research Institute of Eco-friendly Animal Science, Institute of Green Bio Science and Technology, Seoul National University, Pyeongchang-gun, Kangwon-do, 232-916, Korea*

### INTRODUCTION

Plumage color in chickens depend on the pigmentation and the balance of pheomelanin and eumelanin in the melanocyte. Mutation within MC1R gene is associated with the pigmentation of plumage color in chicken (Guo *et al.*, 2010; Kerje *et al.*, 2003; Takeuchi *et al.*, 1996; Zhang *et al.*, 2013). MC1R gene encodes seven-span transmembrane G protein-coupled receptors which is located on the plasma membrane of melanocyte and plays a major role in regulation of eumelanin (brown and black) and pheomelanin (red, yellow and white) plumage pigmentation. In general, stimulation of the receptor by its endogenous agonist  $\alpha$ -melanocyte stimulating hormone ( $\alpha$ MSH) results in increased eumelanin pigment while binding with the inverse agonist agouti protein results in decreased eumelanin and increased pheomelanin pigment. Particularly, among seven transmembrane domain in MC1R gene, mutation of the first, second and third transmembrane domain contributed to the regulation of production of eumelanin and pheomelanin pigment. In previous studies, the Ile58Val and Glu92Lys mutation of the first and second transmembrane domain in MC1R gene are associated with plumage pigmentation in chicken (Guo *et al.*, 2010; Zhang *et al.*, 2013). Therefore, the aim of this study was to discover nonsynonymous SNP within the third transmembrane domain of MC1R and evaluate relationship between mutation of the third transmembrane domain in MC1R and plumage pigmentation in 3 chicken breed.

**Sample/SNP discovery/genotyping:** Genomic DNAs were extracted from Black silky (n = 96), White Leghorn (n = 96) and Golden duckwing Araucana (n = 96) with different plumage colors (Table 1). In particular, Golden duckwing Araucana is a breed of chicken originating in Chile that lay blue eggs. It had yellow and blue plumage color with tail and ear-muffs. We discovered G834A SNP from reference sequence data (GenBank accession No. D78272.1) reported by Kerje *et al.* (2003). This SNP was a non-synonymous SNP (dbSNP accession No. rs316651020) and substituted from valine to isoleucine in the 126th amino acid position. Primer sequence was designed using the Primer3 program and their information was shown in Table 1. For genotyping of G834A SNP within MC1R gene, SBE method was performed according to the protocol of SNaPshot ddNTP Primer Extension Kit (Applied Biosystem, Foster City, CA) (Vreeland *et al.*, 2002). The allele and genotype frequency of G834A SNP were calculated based on genotyping.

Table 1: Number of animal and plumage color in 4 chicken breed used in this study

Breed (Code)	Plumage color	No. of animals
Black silky (O)	Black	96
White Leghorn (W)	White	96
Golden duckwing Araucana (GA)	Blue and yellow	96
Total		384

Table 2: Genotype and allele frequency of G834A variation within MC1R in 3 chicken breed

Nucleotide	Amino acid	Genotype and allele	Genotype and allele frequency (No. of animals)		
			O	GA	W
G834A	Val	GG	1.000 (96)	0.250 (24)	
	Val/Ile	GA		0.750 (72)	0.167 (16)
	Ile	AA			0.833 (80)
	Val	G	1.000	0.625	0.083
	Ile	A		0.375	0.917

Val: Valine, Ile: Isoleucine

**Genotype and allele frequency/comments:** Table 2 showed the allele and genotype frequency of G834A SNP in Black silky (O), Golden duckwing Araucana (GA) and White Leghorn (W). The GG genotype frequency of G834A SNP in black silky (black) was 1.000 and the AA genotype frequency in White Leghorn (white) was 0.833. Also, the GA genotype frequency of G834A SNP in Araucana (blue/yellow) was 0.750. The G834A is a nons-synonymous SNP which substitutes the 126th amino acid from valine to isoleucine and located in the third transmembrane domain of MC1R receptor. The mutation (Val126Ile) of this domain was linked Ile58Val and Glu92Lys mutation which is associated with plumage pigmentation in chicken (Guo *et al.*, 2010; Kerje *et al.*, 2003; Zhang *et al.*, 2013). Therefore, this result suggested that the G834SNP could play a important role in regulation of eumelanin and pheomelanin production.

## REFERENCES

- Guo, X.L., X.L. Li, Y. Li, Z.L. Gu and C.S. Zheng *et al.*, 2010. Genetic variation of chicken MC1R gene in different plumage colour populations. *Br. Poult. Sci.*, 51: 734-739.
- Kerje, S., J. Lind, K. Schutz, P. Jensen and L. Andersson, 2003. Melanocortin 1-receptor (MC1R) mutations are associated with plumage colour in chicken. *Anim. Genet.*, 34: 241-248.
- Takeuchi, S., H. Suzuki, S. Hirose, M. Yabuuchi, C. Sato, H. Yamamoto and S. Takahashi, 1996. Molecular cloning and sequence analysis of the chick melanocortin 1 receptor gene. *Biochim. Biophys. Acta*, 1306: 122-126.
- Vreeland, W.N., R.J. Meagher and A.E. Barron, 2002. Multiplexed, high-throughput genotyping by single-base extension and end-labeled free-solution electrophoresis. *Anal. Chem.*, 74: 4328-4333.
- Zhang, X.H., Y.Z. Pang, S.J. Zhao, H.W. Xu and Y.L. Li *et al.*, 2013. The relationship of plumage colours with *MC1R* (Melanocortin 1 Receptor) and *ASIP* (Agouti Signaling Protein) in Japanese quail (*Coturnix coturnix Japonica*). *Br. Poult. Sci.*, 54: 306-311.