Identification of Tyrosinase Gene Polymorphisms Associated with Albinism in Swamp and Riverine Types of Water Buffaloes (*Bubalus bubalis* Linn.) in the Philippines

Melinda R. Bacual[1,2,†], E.N. Bacual[1], Y.C. Dela Cruz[1], M.A. Garcia[1], M.G.Q. Diaz[2] and E.B. Flores[1,†,*]

[1]*Animal Breeding and Genomics Section Philippine Carabao Center National Headquarters Science City of Muñoz, Nueva Ecija 3120, Philippines*

[2]*Institute of Biological Sciences, University of the Philippines – Los Baños, Laguna, 4030, Philippines*

Abstract: *Background*: Albinism is a genetic condition marked by a lack of melanin in the skin, hair, and eyes, leading to increased sensitivity to light and susceptibility to skin cancer. Oculocutaneous albinism in buffaloes is caused by a G>A mutation in the tyrosinase gene, which introduces a premature stop codon, rendering the enzyme inactive. Despite efforts to prevent genetic defects, albinism persists because it is an autosomal recessive trait. This study used capillary sequencing to analyze the tyrosinase gene in local buffaloes.

Methods: One hundred forty-eight (148) buffaloes were sampled for genomic DNA extraction, followed by PCR amplification of the reported region of the tyrosinase gene with G>A mutation associated with albinism. The PCR products were subjected to Sanger chain termination sequencing. Genotypic frequencies were computed manually, and phenotypic association was done descriptively.

Results: The proportion of phenotypically albino-looking buffaloes was 4.76% of the sampled animals and are homozygous for the ***A*** allele of the G>A mutation at position 1494 of the tyrosinase gene. These were all riverine-type buffaloes. Phenotypically white but with pigmented irises were all swamp buffaloes and comprised 4.17% of the sampled animals. All swamp buffaloes sampled, including the phenotypically white with pigmented irises, were homozygous for the ***G*** allele of the G>A mutation at position 1494, suggesting these are not similar cases of oculocutaneous albinism.

Conclusions: The study established baseline data on the prevalence of albinism and identified new mutations in the tyrosinase gene for further research on their effects on color phenotypes and production potential.

Keywords: Albinism, tyrosinase gene, mutation, polymerase chain reaction (PCR), DNA sequencing, buffaloes, carabao.

INTRODUCTION

Over the years, the demand for food sustainability and security has intensified due to the continuous increase in the world population. In the Philippines alone, the projected population is expected to reach up to 126 million by 2030 [1], resulting in an annual addition of approximately 1.2 million Filipino individuals. Both the government and the private sectors are now focused on strengthening the country's local plants and animals as food sources in the upcoming years to address this issue. The use of genetic and reproductive biotechnologies would enhance the improvement and dissemination of superior genetics to the population. Marker-assisted breeding and genome-wide association analysis have become crucial tools for identifying important genes associated with the desired traits, which serve as the foundation for the selection processes [2, 3].

For instance, water buffaloes (*Bubalus bubalis*) are specifically promoted and propagated in the country as a source of meat, milk, dairy products, draft power, and hide. They have been important animals in the agricultural fields and have become an integral part of Filipino traditions [4]. Dairy breeds have also been continuously improved through genetic improvement programs to increase milk, fat, and protein yield potential with every calf born [5]. Meanwhile, the swamp types are being studied for their utilization in providing good quality meat. Several important measures, such as thermotolerance [6] and improved buffalo pregnancy by reproduction-assisted techniques [7], have also been established. Concerns regarding the build-up of inbreeding have been raised, especially in areas where some popular bulls are heavily used in artificial insemination programs and the interest in propagating white buffaloes that are assumed to be albinos. As albinism has an autosomal recessive inheritance, the occurrence of genetic defects or abnormalities that could have a lethal effect on the local buffalo population is one avenue to look from.

*Address correspondence to this author at the Animal Breeding and Genomics Section Philippine Carabao Center National Headquarters Science City of Muñoz, Nueva Ecija 3120, Philippines; Tel: +63 9985507809;
E-mail: esterflrs@gmail.com

†The authors share an equal contribution.

ISSN: 1927-5196 / E-ISSN: 1927-520X/24

A genetic defect arises from abnormalities in either the entire or a portion of a species' deoxyribonucleic acid (DNA) sequence. It can result from the mutation of a single gene, mutations in multiple genes, environmental factors, or damage to chromosomes [8]. Various economically significant genetic defects in water buffaloes have already been identified, including musculoskeletal and gastrointestinal defects, craniofacial malformations, and sexual development and skin defects such as freemartinism, acantholytic mechanobullous dermatosis and albinism [9, 10]. Hence, there is a careful need to identify possible genes and polymorphisms affected by these aforementioned methods and investigate their effect on the water buffaloes.

Albinism is a heritable genetic condition characterized by the partial or complete absence of pigmentation in the skin, hair, and eyes, resulting in a distinctive white or pale appearance [11]. While observed across various species, its occurrence is relatively infrequent compared to the normal types. In humans, the prevalence is approximately one in 40,000 births, whereas in animals, it ranges from one in 10,000 to one in a million individuals, a condition considered rare [12, 13]. At the molecular level, it is due to mutations in the genes associated with melanogenesis. Key genes implicated in this process include melanocortin 1 receptor (MC1R), agouti-signaling protein (ASIP), microphthalmia transcription factor (MITF), tyrosine kinase receptor (KIT), and the tyrosinase gene family (TYR, TYRP1, TYRP2). Among these, tyrosinase (TYR) plays a direct and critical role in the synthesis of the two melanin types, eumelanin, and pheomelanin, acting as the rate-limiting enzyme in the catalysis of tyrosine into L-dihydroxyphenylalanine (DOPA) and the subsequent oxidation of DOPA to dopaquinone, the final metabolite for melanin production. As a result, any disruption in tyrosinase biosynthesis not only halts melanin production but also compromises melanin-associated functions, such as pigmentation, photoprotection, immunity, antioxidation, and thermal regulation [14,15]. Consequently, albinism is frequently associated with extreme photophobia, low immunity, poor thermoregulation, and a range of eye, skin, and health issues, significantly affecting the affected individuals. Hence, understanding the etiology of albinism and the diverse genetic controls associated with this condition is crucial, particularly in the context of farm animals [16].

Albinism in water buffaloes was first reported back in 1925 [17]. However, the molecular mechanism underlying this condition was not clear until the report of Damé *et al*. [18] naming the variation in the exon 5 of the tyrosinase gene as the causative mutation for albinism in riverine (RV) buffaloes. This polymorphism was not yet studied in the local swamp type (PC) buffaloes hence, in this study, the exon 5 of the tyrosinase gene was investigated through molecular techniques and determined its association with albinism and other visibly distinct features of pigmentation in the local breeds of water buffaloes in the Philippines.

MATERIALS AND METHODS

Ethics Statement

Collection of blood, frozen semen, and other appropriate tissue samples followed the guidelines on animal use and care as implemented by the Philippine Carabao Center's Research Ethics Committee.

Sample Collection

Epidemiological data was extracted from existing records at the Philippine Carabao Center. Frozen semen, blood, or tissue samples were available to extract the genomic DNA of albino and non-albino animals. The present study targeted a total of 148 semen samples representing the population of the donor bulls used for artificial insemination of buffalo heifers and cows in all of the PCC herds across the Philippines. Of the 148 semen samples, nine samples were of swamp type (PC), and 139 were of riverine (RV) type. Of the 139 riverine bulls, 2 were phenotypically albino, which already produced albino calves. Additional buffalo families from farmers with produced albino offspring were also used to investigate the tyrosinase mutation further.

Genomic Deoxyribonucleic Acid (DNA) Extraction

Genomic DNA from the collected whole blood samples was extracted using a high-salt concentration procedure using the Promega Wizard® Genomic DNA Purification Kit (Promega Corporation, USA), following the manufacturer's protocol with slight modifications. DNA from semen, tissue, and hair samples was extracted using the Qiagen DNEasy™ Blood and Tissue Kit (Qiagen, Germany) with slight modifications per the PCC Molecular Genetics Laboratory protocol.

Determination of DNA Quality and Quantity

Total genomic DNA was assessed using an agarose gel electrophoresis and quantified in a

NanoDrop™ 2000 Spectrophotometer (Thermo Fisher Scientific, USA). A total of 120 ml 2% agarose gel with 2.4 µl GelRed nucleic acid stain was prepared, and 2 µl of isolated genomic DNA with 2 µl 2X loading dye was run in the gel. Mupid-exU electrophoresis was used to run the samples for 50 minutes at 100 volts. The gel was visualized under UV light, and the integrity of the isolated gDNA was checked. Invitrogen™ 100 bp ladder (Thermo Fisher Scientific, USA) was used as the molecular ladder. On the other hand, one microliter of the isolated gDNA was used to measure DNA concentration and purity. DNA samples with concentrations ranging from 30-100 ng/µl were used for succeeding analyses.

Polymerase Chain Reaction (PCR)

The ***G*** to ***A*** mutation of the tyrosinase gene was investigated through the primers designed by Damé *et al*. [18]. The primer sequences, presented in Table **1**, targeted the 377 bp region located in exon 5 of the tyrosinase gene. PCR reactions were prepared and optimized in a final volume of 10 µL composed of 1X GoTaq® PCR buffer, 2 mM MgCl2, 0.15 mM dNTPs (iNtRON Biotechnology, South Korea), 0.3 µM each of the forward and reverse primers, 0.05 unit of GoTaq® Flexi DNA Polymerase (Promega Corporation, USA) and 1 µL of template DNA. PCR reactions were performed in a Swift™ MaxPro (Esco Lifesciences Group, Singapore) thermal cycler with an initial denaturation of 95°C for 10 minutes followed by 30 cycles of denaturation at 95°C for 15 seconds, annealing at 65 °C for 1 minute and extension at 72°C for 1 min. A final extension step at 72°C for 10 minutes was added and was stored at 4˚C until further analysis.

Available PCR products were then loaded on a 2% agarose gel and checked for amplification. One hundred base pair DNA marker ladder (Invitrogen™, Thermo Fisher Scientific, USA) was used for standard sizing of PCR products.

Sequencing and Analysis

PCR products of all sampled animals were purified using the ExoSAP-IT™ Express PCR Product Cleanup Reagent (Applied Biosystems™, Thermo Fisher Scientific, USA) followed by the sequencing reaction using BigDye™ Terminator v3.1 Cycle Sequencing Kit (Applied Biosystems™, Thermo Fisher Scientific, USA) and standard ethanol precipitation. These products were then transferred in a genetic analyzer barcoded plates for sequencing using the ABI 3500xl Genetic Analyzer (Applied Biosystems™, Thermo Fisher Scientific, USA) wherein the rapid sequencing analysis template was used to run the sequencing analysis. A double pass sequencing reaction (forward and reverse) per sample was prepared to cover the 377 bp region of the tyrosinase gene.

The resulting sequences were assessed and analyzed with Geneious Prime® Software version 2023.2.1 (Biomatters, Inc., New Zealand), which included quality control, trimming, contig assembly, and sequence alignment. Identities of the contiguous sequences were checked using the Basic Local Alignment Tool (BLAST) and compared to the TYR gene of *B. bubalis* available in the National Center for Biotechnology Information (NCBI) with accession number JN887462.1. Genotypic frequencies were computed manually and phenotypic association was done descriptively.

RESULTS AND DISCUSSION

Characterization of the Sampling Population

The domestic water buffaloes were typically large ruminants with a distinctive dark appearance. They are of two types: the riverine and the swamp types. The riverine buffaloes mainly were black or gray, extending from their short, curved horns to their hooves and tails. Some distinct breeds, like Nili-Ravi, would typically have white hairs on the forehead, muzzle, tail, and legs and would have walled bluish-iris [19]. On the other hand, the swamp types display a variety of skin pigmentation ranging from black to grey, brown to reddish brown, pinkish to white, and some appear lined and spotted. To date, there is no clear information about these pigmentation patterns [20]. However, several genes and gene products were suspected of

Table 1: TYR Primer Details (Damé *et al*., 2012)

PRIMER	5' – 3'	LENGTH	T_M	% GC	PDT SIZE
Forward:	TAGAACAAGCACAACGAATCTGGG	24	61.33	45.83	377 bp
Reverse:	AAAGCAAGCACAGGTGGCTTCTAC	24	63.84	50.00	

causing these pigment variations. Some of which were syndromic in nature. In this study, the tyrosinase gene was investigated to be causing the albinistic white phenotype in the local breeds of riverine and swamp-type water buffaloes.

The total number of animals studied was 168, of which eight (4.76%) appeared to be phenotypically albino due to their total white/pinkish appearance, including a non-pigmented iris that looked bluish/pinkish (Figure **1A-D**). Seven (4.17%) appeared to be only white with colored/pigmented black irises (Figure **1E-F**), and the other 153 animals (91.07%) were black, brown, or grey.

Of the classified animals, all eight albinos were either riverine or crossbred (CB), as shown in Table **2**. All seven white animals were either swamp or crossbred, and the black phenotype was observed across various water buffalo types. Figure **1** shows the difference between an albino and a white carabao, which differed in the color of the eyes. These observed phenotypes are typically based on previous reports, whereas the swamp types display various pigmentation, but the irises remain pigmented [17, 20].

Tyrosinase Gene Polymorphism

The 377 bp fragment of Exon 5 of the tyrosinase gene, located in *B. bubalis* chromosome 5 were sequenced to determine the presence of polymorphisms across the different types of water buffaloes. All 168 individuals sampled were sequenced and results revealed at least 99.7% homology to the deposited *B. bubalis* tyrosinase gene sequences in the NCBI database. Figure **2** shows the amplified 377 bp fragment of the tyrosinase gene in water buffaloes.

Within the targeted sequence, a total of four sites varied across different water buffalo types. These variations were found at nucleotide positions G1494A, T1499C, G1568A, and G1720A, respectively (Table **3**, Figure **3**), when mapped to reference JN887462.1 deposited in the NCBI GenBank™. The G>A mutation at position 1494 was first reported by Damé *et al*. [18] to be causing complete albinism in riverine buffaloes, that is, the absence of visible pigmentation in the skin, hair, and eyes. This mutation converts the tryptophan (W) residue into a stop codon, leading to the truncation of the signal peptide responsible for delivering the gene product into the melanosome. This primarily disrupts the catalysis of tyrosine into the two melanin products, hence the total white appearance. This is also called oculocutaneous albinism (OCA) type I in humans [13]. The riverine buffaloes are polymorphic at position G1494A, wherein the seven (7) ***AA*** and one (1) ***GA*** genotypes were all phenotypically albino. While there were two crossbred albinos, these were also sired by riverine bulls. At nucleotide position T1499C, the

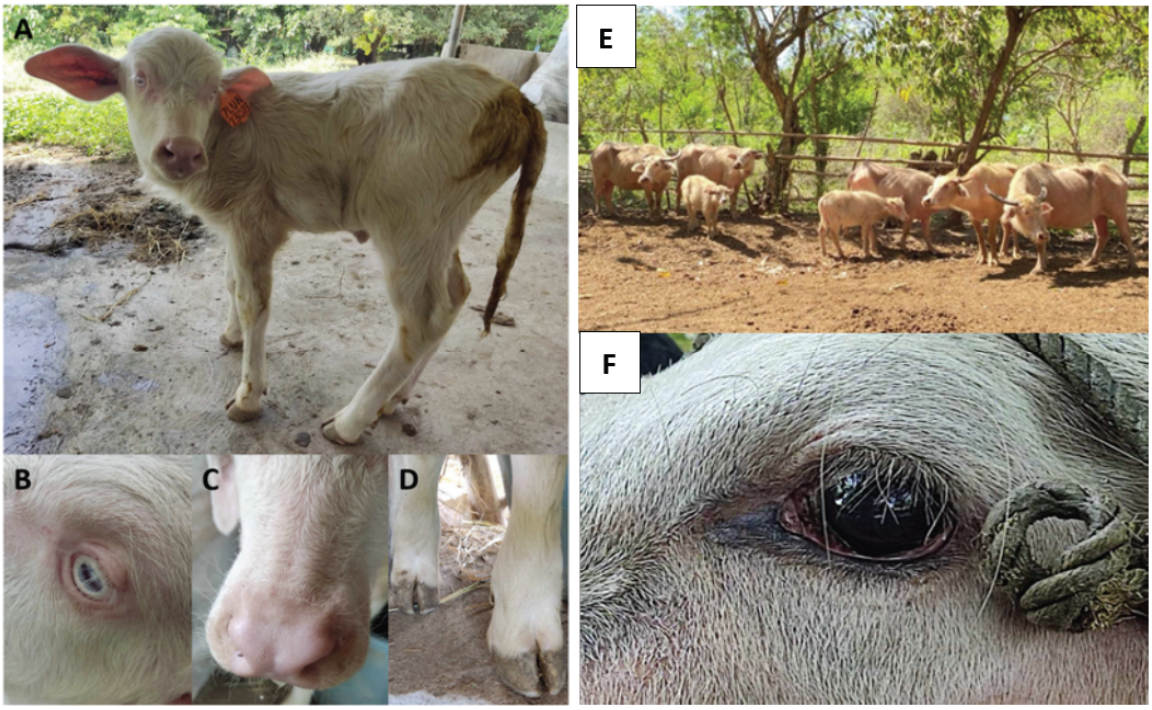

Figure 1: A crossbred calf (**A**) indicating signs of complete albinism in the eyes (**B**), muzzle (**C**), and hooves (**D**) and a herd of white swamp buffaloes (**E**) characterized by having white hair, pink skin, and pigmented iris (**F**).

Table 2: The Proportion of Albino, White, and Dark-Haired Buffaloes according to Breed from Sampled Animals

PHENOTYPE	PC	CB	RV	TOTAL	PERCENTAGE, %
Albino – pink skin, white hair, pink/non-pigmented iris.		2	6	8	4.8
White–pink skin, white hair, pigmented iris	5	2		7	4.2
Jet Black – typical of the riverine type		4	139	143	85
Dark brown/Gray – typical swamp phenotype with chevron	10			10	6
TOTAL	15	8	145	168	100

PC – swamp-type Philippine carabao, CB – crossbred between swamp and riverine-type buffaloes, RV – riverine-type buffalo.

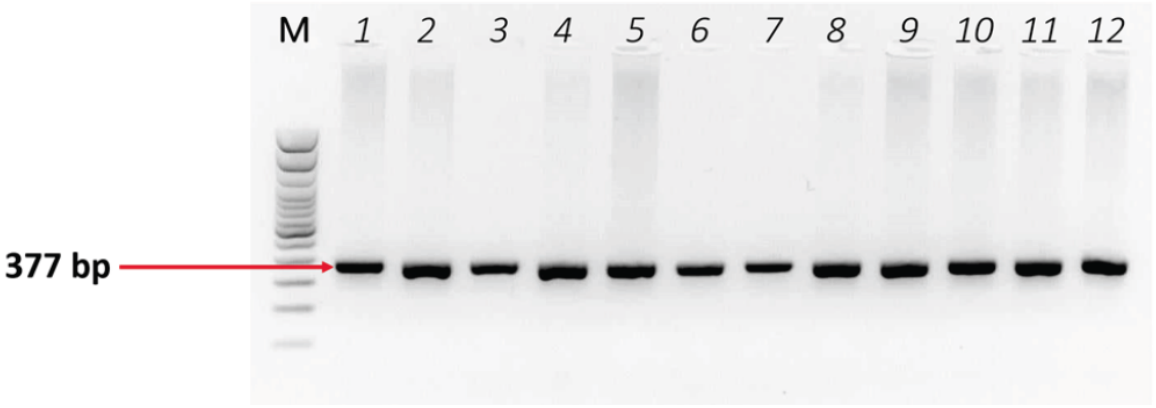

Figure 2: Optimized PCR amplification of the 377 bp-fragment of the tyrosinase gene in buffaloes: Lane M - 100 bp DNA ladder; Lanes 1–12, amplified products.

riverine buffaloes are non-polymorphic with homozygous ***TT*** genotype.

On the other hand, the swamp buffaloes are homozygous ***GG*** at nucleotide position G1494A and polymorphic at position T1499C (Table **3**). The swamp buffaloes that are phenotypically white with pigmented iris are all homozygous ***GG*** at nucleotide position G1494A, and it is highly indicative that these are not genetically albinos. The white buffaloes described herein are similar phenotypically to the Levin report [17]. The polymorphism at the four (4) nucleotide positions could not establish a specific genotype that is consistent with the occurrence of white buffaloes. The results of this study suggest that other genes may be involved or influencing the phenotypically white swamp buffaloes.

In this study, it is clear that the mutation at nucleotide position G1494A of the tyrosinase gene with the ***AA*** genotype determines the occurrence of albinism and supports the findings of Damé *et al*. [18]. The presence of a single phenotypically albino buffalo, which is heterozygous ***GA*** and ***TC*** genotypes at positions G1494A and T1499C, respectively, suggests a manner of inheritance other than autosomal recessive might be possible. While there were other riverine buffaloes with ***GA*** genotype at position G1494A that are not phenotypically albino, these have ***TT*** genotype at position T1499C, suggesting heterozygosity is required at this position to produce phenotypically albino animals if the genotype at position G1494A is ***GA***. This information will contribute to a better understanding of the pattern of inheritance of albinism. Further study with a bigger data set is warranted. It would appear that ***the A*** allele at position G1494A is not present in the local swamp buffalo population, but due to a crossbreeding program being conducted, the ***A*** allele might eventually be introgressed into the population. Identifying the causative gene and developing a screening test is needed as it is possible that in the future, a buffalo might inherit the mutant allele for both albinism and white buffalo. The genotype and allele frequencies of the four polymorphic sites in the tyrosinase gene that were investigated from the sampled animals are shown in Table **4**. The genotype frequency for the AA, GA, and GG genotypes at nucleotide position G1494A were 4.2%, 10.7%, and 85.1%, respectively. The frequency of the GA genotype in this study is higher than those reported by Bernardino et a. [21], wherein they sampled phenotypically normal Brazilian riverine buffaloes. In

Table 3: Polymorphism of the Tyrosinase Gene at the Four Nucleotide Positions according to Breed and Phenotype

PHENOTYPE	BREED	TYROSINASE GENE NUCLEOTIDE POSITION				N
		G1494A	*T1499C*	*G1568A*	*G1720A*	
ALBINO	RV	GA	TC	GG	GG	1
ALBINO	RV, CB	AA	TT	GG	GG	7
WHITE	PC	GG	TC	GG	GG	2
WHITE	PC, CB	GG	TT	GG	GG	5
Black/Gray	PC	GG	TT	GG	GG	1
Dark Brown	PC	GG	TT	GG	GG	3
Dark Brown	PC	GG	TC	GG	GG	4
Dark Brown	PC	GG	CC	GG	GG	2
Jet Black	RV	GG	TT	GG	GG	62
Jet Black	RV, CB	GG	TT	GG	GA	10
Jet Black	RV	GG	TT	GG	AA	2
Jet Black	RV	GG	TT	GA	GG	43
Jet Black	RV	GG	TT	GA	GA	5
Jet Black	RV	GG	TT	AA	GG	4
Jet Black	RV, CB	GA	TT	GG	GG	11
Jet Black	RV, CB	GA	TT	GG	GA	3
Jet Black	RV	GA	TT	GA	GG	3
TOTAL						168

RV – Riverine type buffalo, PC – swamp-type buffalo or carabao, CB – crossbred (a cross between riverine and swamp-type buffaloes).

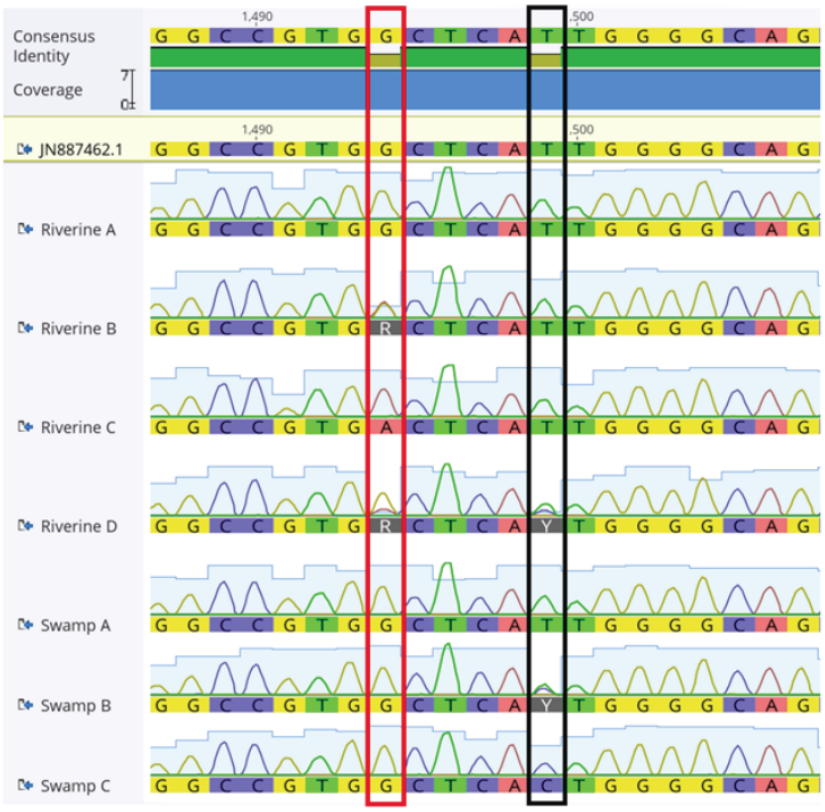

Figure 3: Nucleotide alignments showing point mutations at positions 1494 (red) and 1499 (black) in riverine and swamp water buffaloes.

Table 4: Genotype and Allele Frequencies at the Four Polymorphic Nucleotide Positions in the Tyrosinase Gene

GENOTYPE AT NUCLEOTIDE POSITION	FREQUENCY			
	GENOTYPE		ALLELE	
G1494A	**n**	**%**	**A**	**G**
AA	7	4.2	0.10	0.90
GA	18	10.7		
GG	143	85.1		
TOTAL	168			
T1499C	**n**	**%**	**C**	**T**
CC	2	1.2	0.03	0.97
TC	7	4.2		
TT	159	94.6		
TOTAL	168			
G1568A	**n**	**%**	**A**	**G**
AA	4	2.4	0.18	0.82
GA	51	30.4		
GG	113	67.3		
TOTAL	168			
G1720A	**n**	**%**	**A**	**G**
AA	2	1.2	0.07	0.93
GA	18	10.7		
GG	148	88.1		
TOTAL	168			

contrast, this study purposely sampled semen donor bulls and animals reported to be phenotypically albino and their families.

The presence and use of heterozygous GA genotyped bulls at nucleotide position G1494A could increase the production of carrier females in the population. Concern was raised regarding the reproductive performance of albino carrier cows, especially regarding the male/female sex ratio. There appear to be proportionately fewer female calves born from albino carriers (P=0.0409) compared to non-albino cows based on past calving/reproductive records of a single herd (Table **5**). The albino carrier cows were not genotyped but assumed as carriers as these were sired by a homozygous ***AA*** albino bull and subsequently mated to different albino carrier bulls (bulls were genotyped to be heterozygous ***GA***). The status of the non-carrier cows was also assumed based on pedigree records as these were not genotyped, and data gathered was from past performance. The calves born were mating with heterozygous ***GA*** bulls; thus, there is no possibility of producing homozygous ***AA*** calves. The disproportionately fewer female calves born from mating heterozygous individuals suggest that female embryos could be lost due to early embryonic death. Further study is warranted to understand the disproportionate sex ratio, including test mating and early pregnancy detection, to determine if there is early embryonic death occurring.

In rural communities where there are white swamp buffaloes, there is a greater demand for riverine albino bulls for breeding, with the expectation that the resulting offspring would be albino as well. This may not necessarily be the case unless the white swamp-type buffalo is homozygous ***CC*** at position T1499C or perhaps a small chance if it is of ***TC*** genotype. The use of homozygous albino bulls and heterozygous carriers in an artificial insemination (AI) program would increase the number of heterozygous carriers in the local buffalo population. If indeed homozygous ***AA*** female embryos result in early embryonic death, the government's herd build-up program would be affected by a reduced pregnancy rate or an increase in the re-breeding rate

Table 5: Comparison of Proportion of Male and Female Calves Born between Albino Carrier and Non-Carrier Cows Mated to Albino Carrier Bulls

STATUS	M		F		TOTAL
	n	%	n	%	
Carrier cows	7	87.5	2	22.2	9
Non-carrier cows	34	61.8	21	38.2	55
TOTAL	41	64.1	23	35.9	64

Prob>ChiSq = 0.0409.

among albino-carrier female buffaloes. Identifying carriers of albinism-associated mutations through genetic testing can inform selective breeding programs, helping to avoid the propagation of albinism as a conservative approach until more information regarding disproportionate sex ratio is known. In some cultures, white or albino animals hold special significance and may be preferred for cultural or religious reasons. Understanding the genetic basis allows for informed decisions that balance cultural preferences with animal health and management practices.

CONCLUSIONS

The oculocutaneous albinism gene was investigated in this study, and capillary sequencing was used to analyze, in particular, the G>A mutation in the tyrosinase gene in local buffaloes. Four polymorphic sites in the tyrosinase gene were investigated. The proportion of phenotypically albino-looking buffaloes was 4.76% of the sampled animals and are homozygous for the ***A*** allele at nucleotide position G1494A mutation of the tyrosinase gene. One phenotypically albino bull was heterozygous ***GA*** and ***TC*** at nucleotide position 1494 and 1499, respectively. The result suggests that aside from assumed autosomal recessive, other patterns of inheritance may be possible with oculocutaneous (OCA) albinism. These were all riverine-type buffaloes.

Phenotypically white but with pigmented irises were all swamp buffaloes and comprised 4.17% of the sampled animals. All swamp buffaloes sampled, including the phenotypically white with pigmented irises, were homozygous for the ***G*** allele at nucleotide position G1494A mutation of the tyrosinase gene, suggesting these are not similar cases of oculocutaneous albinism. Further research into identifying the gene/s involved in producing phenotypically white swamp buffaloes is to be considered.

The study established baseline data on the prevalence of albinism and identified new mutations in the tyrosinase gene for further research on their effects on color phenotypes and production potential. Further study involving bigger data set and investigation into the possible early embryonic death of female embryos from heterozygous albino carrier cows is needed to understand the inheritance of OCA albinism.

CONFLICT OF INTEREST

There is no conflict of interest with any financial organization regarding the materials discussed in the manuscript.

ACKNOWLEDGEMENTS

The authors express their gratitude to the dedicated buffalo raisers, farm workers, and staff of the Animal Breeding and Genomics Section, Philippine Carabao Center, for their invaluable support and assistance in conducting this study. The full support of PCC management and the funding provided by DOST-PCAARRD throughout the course of this study are also duly acknowledged. Appreciation was also extended to the DNA Core Sequencing Facility of the Philippine Genome Center.

Special mention is also duly given to Aaron Dela Cruz and Rogelio Antiquera for their assistance during sample preparation and collection. M.R.B would also like to thank the DOST-ASTHRDP-NSC for her graduate scholarship grant.

FUNDING

This study is part of a DOST-PCAARD-funded project being implemented by the author's institution, the Philippine Carabao Center.

REFERENCES

[1] Salas JMIS, Abrigo MRM, Racelis RH. Implications of projected Philippine population growth, age structure change,

and aging: using national transfer accounts results. RePEc: Research Papers in Economics 2012. Discussion Papers DP 2012-30, Philippine Institute for Development Studies. Accessed at https://www.econstor.eu/bitstream/10419/126899/1/pidsdps1230.pdf

[2] Dekkers JC. Commercial application of marker- and gene-assisted selection in livestock: strategies and lessons. J Anim Sci 2004; 82(E-Suppl): E313-328.

[3] Uffelmann E, Huang QQ, Munung NS, de Vries J, Okada Y, Martin AR, Martin HC, Lappalainen T, Posthuma D. Genome-wide association studies. Nat Rev Methods Primers 2021; 1(1): Article 59. https://doi.org/10.1038/s43586-021-00056-9

[4] Pineda PS, Flores EB, Herrera JRV, Low WY. Opportunities and challenges for improving the productivity of swamp buffaloes in Southeastern Asia. Front Genet 2021; 12. https://doi.org/10.3389/fgene.2021.629861

[5] Herrera JRV, Flores EB, Duijvesteijn N, Moghaddar N, Van Der Werf JH. Accuracy of genomic prediction for milk production traits in Philippine dairy buffaloes. Front Genet 2021; 12. https://doi.org/10.3389/fgene.2021.682576

[6] Maylem ERS, Rivera SM, Leoveras MEDC, Venturina EV, Atabay EP, Atabay EC. Thermotolerance identification in water buffalo using heat shock protein 70 (hsp70) and its effect to semen quality in varying environmental conditions. Philipp J Vet Anim Sci 2018; 44(1): 22-31.

[7] Atabay EP, Atabay EC, Maylem ER, Tilwani RC, Flores EB, Sarabia AS. Improved pregnancy in water buffaloes through synchronization of ovulation and fixed-time artificial insemination technique. Philipp J Vet Med 2019; 56(2): 105.

[8] Gholap PN, Kale DS, Sirothia AR. Genetic diseases in cattle: a review. Research Journal of Animal, Veterinary and Fishery Sciences 2014; 2(2): 24-33.

[9] Damé MCF, Riet-Correa F, Schild AL. Hereditary diseases and congenital defects diagnosed in water buffalo (Bubalus bubalis) in Brazil. Pesqui Vet Bras 2013; 33(7): 831-9. https://doi.org/10.1590/S0100-736X2013000700001

[10] Albarella S, Ciotola F, D'Anza E, Coletta A, Zicarelli L, Peretti V. Congenital malformations in river buffalo (Bubalus bubalis). Animals 2017; 7(2): 9. https://doi.org/10.3390/ani7020009

[11] Sandoval-Castillo J, Mariano-Melendez E, Villavicencio-Garayzar C. New records of albinism in two elasmobranchs: the tiger shark Galeocerdo cuvier and the giant electric ray Narcine entemedor. Cybium 2006; 30(2): 191-192.

[12] De Vasconcelos FTGR, Hauzman E, Henriques LD, Goulart PRK, De Faria Galvão O, Sano RY, Da Silva Souza G, Lynch JW, Silveira L, Ventura DF, Bonci DMO. A novel nonsense mutation in the tyrosinase gene is related to the albinism in a capuchin monkey (Sapajus apella). BMC Genom Data 2017; 18(1). https://doi.org/10.1186/s12863-017-0504-8

[13] Federico JR, Krishnamurthy K. Albinism. [Updated 2023 Aug 14]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2023. Available from: https://www.ncbi.nlm.nih.gov/books/NBK519018/

[14] Cao W, Zhou X, McCallum NC, Hu Z, Ni QZ, Kapoor U, Heil CM, Cay KS, Zand T, Mantanona AJ, Jayaraman A, Dhinojwala A, Deheyn DD, Shawkey MD, Burkart MD, Rinehart JD, Gianneschi NC. Unraveling the structure and function of melanin through synthesis. J Am Chem Soc 2021; 143(7): 2622-37. https://doi.org/10.1021/jacs.0c12322

[15] McNamara ME, Rossi V, Slater TS, Rogers CS, Ducrest AL, Dubey S, Roulin A. Decoding the evolution of melanin in vertebrates. Trends Ecol Evol 2021; 36(5): 430-443. https://doi.org/10.1016/j.tree.2020.12.012

[16] Ren HX, Song TZ, Chen XY, Wang G, Zhou P. The genetics of skin color variation in farm animals. Int J Zool Anim Biol 2(4). https://doi.org/10.23880/IZAB-16000151

[17] Levine CO. Albino water buffaloes. J Hered 1925; 16(2): 66. https://doi.org/10.1093/oxfordjournals.jhered.a102559

[18] Damé MCF, Xavier GM, De Oliveira Filho JP, Borges AS, De Oliveira HN, Riet-Correa F, Schild AL. A nonsense mutation in the tyrosinase gene causes albinism in water buffalo. BMC Genom Data 2012; 13(1). https://doi.org/10.1186/1471-2156-13-62

[19] Dhandapani S, Vohra V, Mukesh M, Kumar S, Mehrara KL, Singh K, Kumari N, Kataria RS. Revealing inheritance of white markings in Nili Ravi buffalo. Indian J Anim Sci 2021; 91(3). https://doi.org/10.56093/ijans.v91i3.114150

[20] Yusnizar Y, Wilbe M, Herlino AO, Sumantri C, Noor RR, Boediono A, Andersson L, Andersson G. Microphthalmia-associated transcription factor mutations are associated with white-spotted coat color in swamp buffalo. Anim Genet 2015; 46(6): 676-682. https://doi.org/10.1111/age.12334

[21] Bernardino PN, Martins AFA, Barbosa JD, Schild AL, Damé MCF, Borges AS, Oliveira-Filho JP. Molecular detection of albinism gene in Brazilian buffalo herds (Bubalus bubalis). Pesqui Vet Bras 2019; 39(3): 175-178. https://doi.org/10.1590/1678-5150-pvb-6185

Received on 20-11-2024 Accepted on 15-12-2024 Published on 31-12-2024

https://doi.org/10.6000/1927-520X.2024.13.20