

Introduction to Special Issue

Animal Pigmentation Genetics in Ecology, Evolution, and Domestication

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Animal coloration is a complex, multifunctional trait that biologists have intensely investigated to unravel its proximate mechanisms and ultimate evolutionary drivers. Pigmentation is known to play a significant role in several aspects of fitness, including mate choice, camouflage/crypsis, defense/warning signaling, thermoregulation, communication, and immunity. Pigmentation genetics has been addressed in an extensive literature over more than a century, with some of the first studies being published in *Journal of Heredity* (e.g., Wright 1917, 1918), which has remained an important outlet for discoveries in this field. Throughout this period, some aspects of animal coloration genetics have become well understood, as discussed in several recent reviews (e.g., Hubbard et al. 2010; Cuthill et al. 2017; Alvarado 2020; Caro and Mallarino 2020; Orteu and Jiggins 2020; Eizirik and Trindade 2021). At the same time, many outstanding questions remain even on those aspects, whereas for other topics, the field remains in its infancy, with major questions still wide-open and myriad opportunities for novel research. In this context, this Special Issue brings together a suite of studies addressing a variety of themes related to animal coloration genetics. The articles that comprise it include a review on diverse aspects of avian coloration and 8 primary research articles targeting a variety of topics in fish, gray wolves, goats, horses, and 2 different groups of birds.

Price-Waldman and Stoddard (2021) provide a comprehensive review of bird coloration that covers a range of fascinating aspects, from recent advances in plumage pigmentation genetics to current research on the colors of bare parts and eggs. The plumage pigmentation component covers several aspects, including a focus on the genetic mechanisms associated with colors produced by carotenoid and psittacofulvin pigments. There is also a focus on structural colors (which are shaped by nanoscale structures and their interaction with light), their physical and genetic underpinnings, and their interactions with pigment-based coloration. Other topics include the genetics of plumage pattern formation (i.e., markings such as stripes, bars, and patches) across the body and within individual

feathers; the genetic bases of coloration in bare parts (e.g., skin, bills, eyes) and eggshells; the genetic and evolutionary links between the avian visual system and body coloration; and the relevance of color phenotypes in bird speciation processes.

Two other articles in this issue also focus on the comparative genetics of pigmentation in bird systems. First, Rubenstein et al. (2021) employed functional genomic tools to investigate the organization and development of melanosomes in the superb starling, which produces both iridescent and noniridescent feathers. They found that melanosomes present in iridescent feathers were saturated with eumelanin and were energetically inexpensive to produce, whereas non-iridescent feathers contained phaeomelanosomes and were likely more costly. The study allowed the characterization of the different gene networks involved in structural and nonstructural colors in this species and explored the genetic and functional relationships between pigment-driven and self-assembly processes that influence bird coloration. Next, Janssen et al. (2021) investigated the genetic basis of plumage polymorphism in skuas (*Stercorarius*). Since it had been previously shown that polymorphic ventral plumage coloration in 2 skua species (*S. parasiticus* and *S. pomarinus*) was driven by convergent variation at the *MC1R* gene, this study tested whether this was also the case in a third species (*S. maccormicki*) that exhibits similar phenotypes. The results indicated that *MC1R* coding variants were not implicated in this case (and neither was another color-related locus, *TYRP1*), indicating that this polymorphism evolved in this species independently, with a different genetic basis.

Domestic species offer a unique opportunity to explore the evolutionary history of a phenotype under artificial selection. For example, ancient DNA studies revealed that most early horses were dark bays with dun dilution (Ludwig et al. 2009; Imsland et al. 2016). Following their domestication 5500 years ago, people selected horses for a wide range of coat color variants. In this Issue, the genetic bases for 3 types of variation are reported. Ciesiak et al. (2021) describe incomplete penetrance for the Dun dilution gene in

Polish Primitive Horses (Konik), confounding accurate color identification by breeders. Hammons et al. (2021) observed variation in the shade of red hair pigment and mapped its genetic basis to a region harboring a candidate gene, *SALL1*. Finally, Rosa et al. (2021) reported 2 additional variants of *KIT* responsible for white spotting in horses, bringing the total number to 32 known spotting variants at the *KIT* locus.

Each component of a coloration phenotype has evolved in response to selective pressures under diverse ecologies, developmental constraints, and compartmentalization. Similarly, the sources of genetic variants add a further layer of complexity to the evolution of these traits. In some cases, the movement of pigmentation-affecting alleles via gene flow and subsequent adaptive introgression can produce novel coloration phenotypes. In the case of domestic species, the movement of functionally relevant genes has often been revealed through coat color phenotypes. In this issue, Henkel et al. (2021) discovered that color variants in the domestic Valais goat are influenced by the introgression of 2 different alleles of the *ASIP* gene, which compound the effects of a previously reported introgressed allele of the *TYRP1* locus. These rare color variants had an unknown molecular basis as the introgression occurred prior to a revision of the breed standard, when 4 color phenotypes were already segregating.

In cases where a novel mutation generates a distinct phenotype in a domestic species' genome, introgression to a wild relative could have potential fitness consequences under natural selection. Johnston et al. (2021) developed a novel phenotyping system to explore the hypothesized pleiotropic fitness effects on viral susceptibility based on a 3 bp deletion in the *CBD103* gene that influences coat color in canines. They established the first cell lines derived from gray wolf keratinocytes from the recessive gray and dominant black coat color phenotypes and assessed expression differences of each line in response to experimental exposure to *TLR3* signaling and the canine distemper virus. Although they did not find evidence for pleiotropy, these cell lines are an innovative approach toward exploring the genetic architecture for fitness-related pigmentation traits. The coat color phenotype has a known fitness tradeoff in gray wolves and has a well-documented evolutionary history in canines.

Overall, melanogenesis is known to also be orchestrated by the complex interaction of dozens of molecules, including hormones and multiple genes. To further understand this system, Wang et al. (2021) used Nile tilapia as a developmental genetics model system to perform targeted mutagenesis of genes involved in melanogenesis. They leveraged a study design to screen for diversity in pigment cell types and map the timing at which they appear during development. The contribution of each gene is not equal, since some may exert a simple yet high level of control, such as those that control pigment-type switching. Alternatively, genes and their products can serve as partners in multigenic epistatic interactions. Both mechanisms are active in the production of complex coloration patterns from simple ones.

Jointly, the articles that comprise this Special Issue provide a variety of novel data, analyses, and insights that contribute to the advancement of the field of animal coloration genetics. Moreover, they raise several new questions and open up additional avenues for future research on this topic. We are thus confident that this collection

of studies will foster increased interest in the many research fronts that comprise the effort to understand the genetic bases and evolutionary significance of coloration phenotypes in animals.

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