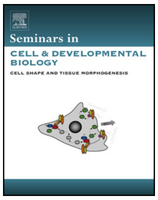
Seminars in Cell & Developmental Biology 24 (2013) 587–593

Contents lists available at SciVerse ScienceDirect

Seminars in Cell & Developmental Biology

journal homepage: www.elsevier.com/locate/semcdb



Review

The role of humans in facilitating and sustaining coat colour variation in domestic animals

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ARTICLE INFO

Article history:
Available online 5 April 2013

Keywords:
MC1R
Selection
Animal breeding

ABSTRACT

Though the process of domestication results in a wide variety of novel phenotypic and behavioural traits, coat colour variation is one of the few characteristics that distinguishes all domestic animals from their wild progenitors. A number of recent reviews have discussed and synthesised the hundreds of genes known to underlie specific coat colour patterns in a wide range of domestic animals. This review expands upon those studies by asking how what is known about the causative mutations associated with variable coat colours, can be used to address three specific questions related to the appearance of non wild-type coat colours in domestic animals. Firstly, is it possible that coat colour variation resulted as a by-product of an initial selection for tameness during the early phases of domestication? Secondly, how soon after the process began did domestic animals display coat colour variation? Lastly, what evidence is there that intentional human selection, rather than drift, is primarily responsible for the wide range of modern coat colours? By considering the presence and absence of coat colour genes within the context of the different pathways animals travelled from wild to captive populations, we conclude that coat colour variability was probably not a pleiotropic effect of the selection for tameness, that coat colours most likely appeared very soon after the domestication process began, and that humans have been actively selecting for colour novelty and thus allowing for the proliferation of new mutations in coat colour genes.

Contents

1. Introduction

Darwin was the first to recognize that a wide range of domestic animals share multiple phenotypic characteristics, the most obvious of which is a wide variety of coat colours [1,2]. Numerous plants also possess colour varieties that differentiate them from their wild ancestors, but in animals, the only trait other than variable coat colours that occurs ubiquitously across domestic animals are dwarf and giant varieties [3]. The commonality of variable coat colours is interesting in light of the fact that the process of domestication likely began no more than 15,000 years ago and occurred independently in both the Old and New Worlds [4].

Domestication significantly altered the genetic, behavioural, and phenotypic characteristics of all the organisms involved, and numerous studies have revealed a great deal about the genetic basis of domestic phenotypes including, in many cases, the causative mutations themselves [5]. Since coat colour patterns often follow a Mendelian inheritance mode, they have been among the first traits to be systematically analysed at the molecular level [6] and the functional genomics of coat colour variation in domestic animals has been studied in depth [7]. Though several recent reviews have focussed on the genetic basis of coat colour variation [7–10], a

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http://dx.doi.org/10.1016/j.semcdb.2013.03.015

number of outstanding questions related to the role that humans played in generating the diversity of domestic animal coat colours remain unanswered.

Firstly, does the ubiquity of coat colour variation result from direct selection by humans, or are variably coat colours a consequence of selection on other behavioural traits including tameness [3]? Secondly, because most studies of domestic animal coat colour have been carried out on modern samples, is it possible that coat colour variation is just a recent phenomenon, and if not, how soon after the domestication process began did coat colour diversity begin to increase? Lastly, to what degree are humans primarily responsible for driving and maintaining coat colour variants that differentiate wild and domestic populations?

1.1. *The genetic basis of coat colour*

Animal colouration is modified through both pigmentation and nanostructure and has many functions including communication, camouflage, predatory avoidance, photoprotection, microbial resistance and thermoregulation. There are two main groups of genes affecting mammalian coat and skin colour: those that act on the pigment synthesis and those that modify the melanocytes, the pigment producing cells [8].

The process of pigment cell development plays a key role in determining coat colour and more than 300 genes have been identified that have an effect on pigmentation either directly or indirectly [11]. Most of these genes act on either the production or the regulation of two pigments, pheomelanin and eumelanin [8]. Both MC1R and ASIP act like switches, while KIT and TYR (and other associated genes) are important in regulation of melanin production (Fig. 1). A brief description of the key genes and the roles they play is described below.

The KIT gene encodes the mast cell growth factor receptor and with its ligand MGF, it plays a key role in the growth and differentiation of melanocytes, hematopoietic cells, and germ cells [12]. Since this gene plays a central role in melanogenesis and melanoblast migration and proliferation, mutations in this gene can have serious consequences. Pleiotropic effects including anaemia, sterility, premature ovarian failure, and chronic gastric ulceration have been identified in a number of different species. In particular, the negative consequences of possessing a white coat as result of a modified KIT gene are generally greater than mutations in other genes that affect coat colour [12–14].

A key enzyme regulating pigment synthesis is TYR. The TYR gene encodes the enzyme tyrosinase, which is in turn involved in the production of melanin from tyrosine. As a result, mutations in the TYR gene result in unpigmented skin, fur or feathers. Albinism caused by a deficiency of melanin pigment in the skin, hair, and eye, results from mutations in genes involved in the biosynthesis of melanin pigment and has been detected in a number of wild and domestic species [15–17]. In addition, mutations in other regions of the TYR gene can result in TYR activity becoming sensitive to temperature. This type of mutation leads to a distinct coat colour pattern were the colder parts of the body (i.e. ears, tail and legs) are coloured and the rest of the body remains white [18]. This type of pattern is prominent in both Siamese and Burmese cats. The Siamese has a phenotype referred to as “pointed” reflecting the fact that pigment is expressed at the extremities creating a mask on their faces. Burmese cats possess an allelic variant that is less temperature sensitive and they display pigment mainly on the torso rather than the extremities [18].

Most coat colour variation is the result of the ratio between the two pigments: eumelanin and pheomelanin. Both MC1R and ASIP control this ratio by acting like switches. MC1R is a G protein-coupled receptor located in the melanocyte plasma membrane (Fig. 1). The gene is highly conserved among vertebrates and has a relatively simple genetic structure made up only of a single 1 kb exon [8]. The MC1R protein binds to a class of pituitary peptide hormones called melanocortins that regulate the synthesis of both eumelanin and pheomelanin. Darker colours are produced when eumelanin is up-regulated and light colours result when pheomelanin is increased [15].

The Agouti signal peptide (ASIP) is a paracrine signal protein that antagonizes MC1R to switch to eumelanin production [19]. Though there have been more mutations identified in MC1R, they only occur in the coding region [8], whereas mutations affecting ASIP are found in both the coding and regulatory regions [20].

2. Three questions

2.1. *Tameness and domestic coat colours*

Dmitry Belyaev hypothesized that the phenotypes expressed by all domestic animals resulted not from deliberate selection for individual traits, but as a consequence of selection for a behavioural characteristic. In 1959 he began testing his hypothesis by breeding silver foxes, an animal never previously domesticated. He selected against aggression and only allowed the tamest 10% of the population to breed. Belyaev expected that by selecting a specific behavioural trait he would influence the phenotype of subsequent generations, making them more domestic in appearance [21]. The experiment succeeded by producing foxes with drooping ears, upturned tails, shortened snouts and other features that commonly distinguish domestic animals from their wild ancestors.

Beyond demonstrating how readily domestic phenotypes could appear through a selection for a behavioural trait, Belyaev also provided a mechanism to explain how early animal domestication could have began that did not require deliberate human action. For instance, wolves, initially wary of humans may have been attracted to the waste generated by human camps. Only those with a reduced fight or flight distance would have been able to take advantage of the resources available near the human niche, and it was those wolves that were likely to become more intimately involved in human settlements.

This possibility has been formalised and expanded upon by both Vigne [22] and Zeder [4]. Vigne sets out several consecutive phases of the intensification of the relationship between humans and animals that begins with anthropophily and proceeds to commensalism, control in the wild, control of captive animals, extensive breeding, intensive breeding, before ending in pets. Zeder has recently expanded this view by recognising that not all animals entered into a domestic relationship in the same way. Instead, Zeder characterised three separate domestication pathways: a commensal pathway, a prey pathway and a directed pathway [4]. The commensal pathway is similar to Vigne’s model in that the process begins with a habituation phase with no intentional human selection. Animals that were first hunted, and then later more directly managed before humans began actively controlling their breeding followed a prey pathway. The directed pathway, most commonly associated with much more recently domesticated pets (<200 years) skips the earliest phases of the other pathways and begins with the intentional capture of wild animals followed by taming in captivity.

These perspectives are important since they allow for the possibility that the genetic mechanisms and appearance of domestic coat colours may have been different amongst animals that followed separate pathways. In Belyaev’s fox-farm experiment, piebald and spotted coat colours, a trait determined by the incompletely dominant Star mutation, first appeared after only 10 generations [23]. Belyaev only selected for tameness, and given the lack of intentional human action in the early stages of the commensal pathway,