

# Visible polymorphisms in domesticated animal populations and their role in breed creation: A review

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## Keywords

Domestic animal – Wild animal – Livestock breed – Genetic polymorphism – Population genetics – Allele frequency.

## Summary

Many populations within domesticated animal species have readily observable visible polymorphisms in traits such as color, coat type and horns, among others. These polymorphic populations have commonly been referred to as nonuniform, traditional or primary. The visible polymorphisms are due to several alleles which are segregating at different loci, often in a state of equilibrium as defined by the principles of population genetics. The most usual assumptions surrounding these polymorphic populations are that they belong to an infinite population which is under random mating (panmixia). These two conditions have generally been confirmed by field studies. It is believed that in several animal species these polymorphic populations occurred after domestication and served as a reservoir from which breeders have been developing standardized or fixed breeds (in association with breeding companies or through recognition by official bodies). Polymorphic populations also exist in wild species but they are rare, and obvious polymorphisms are usually only observed in domesticated species which are run in large flocks or herds and in which the control of mating is minimal, namely in some species managed under extensive conditions. The genetic loci which control the visible polymorphisms are mainly coat color loci. Many of these are allelic series which have been well characterized in terms of homology, beginning in the early days of Mendelian genetics as a science. These homologies have since been confirmed by molecular genetic investigations.

## ■ INTRODUCTION

After their domestication, farm animals have experienced the forces of both genetic selection and isolation of populations, and, since Darwin (6, 7) and Isidore Geoffroy Saint-Hilaire (9), it has been broadly acknowledged that their visible variability has increased when compared to their wild ancestors, and visible polymorphism is a common aspect of most domesticated species.

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The occurrence of polymorphism in domesticated animal populations is variable and relates to the underlying genetic structure of populations. Isolation and selection have led to the development of subpopulations which could be named ‘recognized breeds’, ‘fixed breeds’ or ‘standardized breeds’ after the classification of Mason (22) gathering the three following categories:

- ‘breeds having a breed society or a herd book’;
- ‘breeds recognized as such’ by the government or other official body;
- ‘other uniform true breeding populations’.

In the definitions of breeds Mason also includes ‘nonuniform populations’, or geographical terms meaning ‘cattle of such or such a place’ or even ‘breeds of such and such a place’. More recently the

nonuniform populations of Mason were named 'traditional' and then 'primary' by Lauvergne (14, 15) suggesting they were at the origin of the so-called 'standardized breeds', i.e. 'true breeding populations'.

The present paper builds on a previous paper (16) by giving more details of the usefulness of visible polymorphisms in documenting population genetics, as well as their useful role in analyzing the populations within a species and the species in which they are described. This paper also discusses the general categories of loci involved in these investigations.

## ■ VISIBLE POLYMORPHISMS IN TERMS OF POPULATION GENETICS

The rediscovery of Mendelism in 1900 was followed by a first analysis of visible alleles in mammals using the house mouse as a model (5). In terms of population genetics, which relates to the gradual evolution of gene frequencies and was originated by Hardy (11) and Weinberg (33), nonuniform (or traditional or primary) populations are simply those in which the frequencies of two or more alleles have reached a state of equilibrium at one or several loci (5).

Hardy and Weinberg showed that the frequencies of alleles  $A$  and  $a$ , respectively  $p$  and  $q$  (with  $q = 1 - p$ ), keep the same values at the next generation in populations where there is random mating, a population of an infinite number, and provided that no selection pressure is made on the various genotypes.

Their work neither explains how these frequencies of equilibrium have been reached nor how the population responds when generations are not separate, or when the various genotypes are submitted to different selection pressures. Other authors have investigated the effects of these situations on allelic frequencies (29) such as Wright (1889-1988), Fisher (1890-1962) and Haldane (1892-1964) having especially tried to fulfill the gaps of separated generations and lack of fitness coefficients.

The contributions of these authors have been expressed in mathematical terms by L'Héritier (13) taking into account selection pressure by allocating fitness coefficients  $\sigma_1$ ,  $\sigma_2$  and  $\sigma_3$  to genotypes  $AA$ ,  $Aa$  and  $aa$ , respectively, always in the case of bi-allelism, and using a differential equation to shift from the discontinuous to the continuous circumstance as follows:

The frequency increase  $\Delta p$  of  $p$  at a given generation to reach  $p'$  at the next one is given by:

$$\Delta p = p' - p = pq \frac{(\sigma_1 - \sigma_2)p + (\sigma_2 - \sigma_3)q}{\sigma_1 p^2 + 2\sigma_2 pq + \sigma_3 q^2} \quad [1]$$

Then, if one replaces in [1] by the extremely small  $dp$  and adds the extremely small  $dt$  in the second member of the equation, one obtains the following differential equation:

$$dp = pq \frac{(\sigma_1 - \sigma_2)p + (\sigma_2 - \sigma_3)q}{\sigma_1 p^2 + 2\sigma_2 pq + \sigma_3 q^2} dt \quad [2]$$

The solution of which gives  $p$  as a function of fitness coefficients and time.

It is possible to solve this equation by knowing the values of fitness coefficients. Haldane and other authors in many theoretical papers then demonstrated that, depending on the values of coefficients of fitness, one can reach values of  $p$  and  $q$  when time  $t$  goes to infinity:

– 0 and 1 or 1 and 0, respectively, meaning that finally only one allele is present in the population and the other has been eliminated, or

– intermediary between 0 and 1, meaning that both alleles are kept in the population and therefore the population is polymorphic at this locus.

Even if it is only rarely possible to estimate the fitness coefficients it is often possible to measure gene frequencies in polymorphic populations. In these populations it is usually assumed that gene frequencies have reached the state of equilibrium. This assumption, combined with the assumption that in the past the population was infinite and randomly mating can result in a relationship between the equilibrium value  $p_e$  of  $p$  and the fitness coefficients in making the differential  $dp$  equal to zero as follows and as suggested by L'Héritier (13):

$$(\sigma_1 - \sigma_2)p + (1 - p)(\sigma_2 - \sigma_3) = 0$$

then

$$p(\sigma_1 - \sigma_2) + (\sigma_2 - \sigma_3) - (\sigma_2 - \sigma_3)p = 0$$

and

$$p[(\sigma_1 - \sigma_2) - (\sigma_2 - \sigma_3)] = -(\sigma_2 - \sigma_3)$$

giving:

$$p_e = (\sigma_2 - \sigma_3) / (2\sigma_2 - \sigma_1 - \sigma_3) \quad [3]$$

Equation [3] requires  $0 < p_e < 1$ , i.e. a superiority of the heterozygote, at least over one of the homozygotes,  $(\sigma_2 - \sigma_3) > 0$ , and also over the mean of the heterozygotes, a kind of heterosis effect which assures the survival of both alleles in the population.

## ■ VISIBLE GENETIC VARIABILITY IN THE WILD

The advantage in terms of variability of the domesticated state over the wild state is confirmed by several authors and summarized by Bösiger (4) and Wills (33) who have shown the existence of polymorphism for visible traits such as coat or plumage color, shell color, etc. in several species but these species are only a handful compared to all the animal species as soon as they are domesticated. Visible polymorphism is characteristic of many domesticated species, and the specific distribution of polymorphisms in a species can be a reflection of the past history of selection by both humans and the natural environment.

## ■ FROM THE WILD TO NONUNIFORM POPULATIONS AND STANDARDIZED BREEDS

According to Darwin, most of the proof of this increase of variability after domestication comes from the existence of several different true breeding breeds, each of which is uniform but different from the others. This arrangement of variation into different populations is most obvious in species such as the domesticated dog and pigeon according to Darwin (7).

In general, every pure breed is made up by pairing together similar animals, leading to populations which are homozygous for several alleles. A first step to create these phenotypically uniform breeds usually could have been a selection from within populations in which many different alleles of several different loci are segregating. In this light, the nonuniform populations of Mason could likely be the polymorphic populations from which fixed or

standardized breeds descended, as suggested by Lauvergne et al. (18) who called them 'traditional' then 'primary' (14, 15). De Alba (8) suggests a similar trajectory by stating that many of the phenotypic (and therefore genotypic) variants pre-date the standardized breeds we know today and are therefore each present in multiple of these breeds without necessarily implying an ancestral or other relationship. But in Darwin's days, nonuniform populations had not yet been described.

The above developed equations are only valid under the assumption of random mating and when the population size is infinite. These conditions could have been approached in domesticated species in the past and they could then help to explain the great number of nonuniform populations listed by Mason in several domesticated species (22–25).

Another possible explanation for a number of these variable populations is that past control of mating by breeders was not very complete so that selection failed to produce uniform populations. Alternatively, in some populations, variation in and of itself may have been favored by breeders, so that rare variants experienced positive selection and were therefore not allowed to drift to extinction. This can easily happen when variation helps pastoralists to identify quickly animals in the field and aids in their monitoring and management. Additionally, throughout most of history domesticated species were not strictly split into genetic pools that were fully isolated from one another. More commonly, they experienced migrations from one to another. These factors all provide a theoretical explanation for the persistence of visible polymorphisms induced by series of allelomorphs and maintained in a single population, and any one of them alone or in various combinations can easily account for the existence and persistence of polymorphic populations.

The superiority of heterozygotes which is sufficient to reach an equilibrium in domesticated species could be induced by the state of domestication itself even in the absence of breeder selection.

## ■ DOMESTICATED SPECIES SHOWING POLYMORPHIC STRAINS

By chronological order the domesticated species in which visible polymorphisms have been described in terms of alleles with visible effects in segregation are cattle (3, 34), sheep (1, 28, 46), cat (30), goat (12), pig (17) and llama (19).

Cattle, sheep goat and pig, along with ass, buffalo and horse, are the farm animal species for which polymorphic breeds are described by Mason in the successive editions of his dictionary of livestock breeds. Another two ruminant species to be added to that number are llama (19) and reindeer (20) as well as two carnivore species, cat (30) and dog in which some commercial breeds are still multisegregating, e.g. the Siberian Husky.

Species which have been domesticated to become laboratory animals, such as several rodent species, have not developed visibly polymorphic strains. An exception is the guinea pig, also known as cuy, which was initially domesticated as a food source in the Andes (26).

Species kept enclosed such as mink or rabbit have generally not developed populations with visible polymorphisms, although some rabbit breeds do allow considerable variation in color. In both rabbits and fur-bearing species the polymorphism for color is directly related to a commercial value and therefore has fitness value in selection.

## ■ LOCI SEGREGATING IN POLYMORPHIC STRAINS

The loci in segregation in the above-listed farm species with polymorphic breeds or populations (e.g. cattle, sheep, goat) mainly include loci controlling coat color. Coat color loci account for at least 80% of loci controlling visible polymorphisms, followed by those controlling the architecture of coat and by those controlling appendages such as tail or ears and horns as analyzed by Nicholas (27) in the dog. This distribution is correlated with the proportion of viable visible alleles which are identified in domesticated species.

## ■ THE CASE OF COAT COLOR LOCI

For the studies on polymorphic domestic populations, coat color loci deserve a special attention because, after the rediscovery of Mendelian laws, they constitute the category of loci with visible effect which has been the most widely analyzed due to the ease of observation. This is well illustrated by the series of papers by Wright in 1917 and 1918 (35–45) comparing ten species of mammals and proposing the principle of interspecies homology between coat color loci.

The principle of homology between these loci was made more explicit by Haldane (10) who expressed it in terms of genes producing the same somatic effect in different species. He considered that the genes had undergone several parallel effects into more or less corresponding multiple allelomorphs series, exhibiting similar linkages in different species. He gave evidence of homology for the loci *C*, *A* and *E* in six species of rodents and three species of carnivore.

A quarter of century later, Little (21) used the same type of evidence: similar linkage relations between two or more loci, similar pleiotropic effect of genes, similarity of multiple allelic series adding similar morphogenesis and function of melanoblasts. These all served to demonstrate homology for the following nine loci: *A*, *B*, *C*, *D*, *E*, *P S*, *Si* and *W* among 23 species of rodents and 14 species of carnivores. An extension of the principle of homology between color loci to more than 60 species of many orders of mammals was carried out by Searle (31).

Currently, as recently summarized (16), after the immense progress of molecular genetics, the homology of loci between species is widely acknowledged and well-proven. This has been accomplished by the analysis of homologous DNA sequences and other genetic tests. Nevertheless, among the polymorphic populations of farm animals named nonuniform or traditional or primary, a few loci among the numerous coat color loci are kept in segregation, probably because they are able to produce viable mutants. Moreover, as pointed out by Bennett and Lamoreux (02) the nomenclature of these popular loci which traces back to the early days is still kept besides the molecular one: *A* (Agouti), *B* (Brown), *C* (Albinos), *D* (Dilute), *E* (Extension), *S* (Spotting) and *W* (dominant White) as a kind of bridge between two states of the genetic knowledge.

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**Résumé**

**Lauvergne J.J., Sponenberg D.P., Millar P.** Le polymorphisme visible de populations animales domestiques, son rôle dans la création des races : une synthèse

De nombreuses populations d'espèces animales domestiques présentent un polymorphisme visible très marqué pour la couleur, le type de pelage, le cornage et autres caractères. Ces populations polymorphes ont été appelées non uniformes, traditionnelles ou primaires. Le polymorphisme est dû à la présence, à certains locus, de plusieurs allèles en ségrégation dont les fréquences ont atteint une valeur d'équilibre entre 0 et 1 selon les lois de la génétique des populations. L'hypothèse la plus couramment admise est que ces populations appartiennent à des populations de taille infinie qui se reproduisent en panmixie, deux conditions qui ont en général été vérifiées par des études de terrain. On pense que ces populations polymorphes apparaissent chez certaines espèces animales après leur domestication et servent de réservoir de variabilité dans lesquels puisent les éleveurs pour créer les races standardisées ou fixées (avec une société d'élevage ou reconnues par un organisme). Des populations polymorphes existent aussi chez les espèces sauvages mais elles sont plutôt rares alors qu'elles sont couramment observées à l'état domestique, au moins chez les espèces qui sont conduites en troupeaux et où le contrôle des accouplements n'est pas très strict, en particulier chez certaines espèces de ruminants conduites en élevage extensif. Certains locus de coloration du pelage constituent l'essentiel des locus à effet visible en ségrégation dans les populations polymorphes. Ces locus présentent des séries alléliques homologues entre espèces dont l'identification qui remonte aux premières décennies de la génétique mendélienne a depuis été confirmée par la génétique moléculaire.

**Mots-clés :** Animal domestique – Animal sauvage – Race d'animal d'élevage – Polymorphisme génétique – Génétiques des populations – Fréquence allélique.

**Resumen**

**Lauvergne J.J., Sponenberg D.P., Millar P.** Polimorfismo visible en ciertas poblaciones animales domésticas, su papel en la creación de razas: síntesis

Numerosas poblaciones de especies animales domésticas presentan un polimorfismo visible muy marcado en el color, tipo de pelaje, cuernos y otros caracteres. Estas poblaciones polimorfas han sido llamadas no uniformes, tradicionales o primarias. El polimorfismo es debido a la presencia de ciertos locus en varios alelos segregados cuyas frecuencias han alcanzado un valor de equilibrio entre 0 y 1 según las leyes de la genética de poblaciones. La hipótesis más frecuentemente aceptada es que estas poblaciones pertenecen a poblaciones de tamaño infinito que se reproducen en panmixia, dos condiciones que por lo general han sido verificadas mediante estudios de campo. Se piensa que estas poblaciones polimorfas aparecen en ciertas especies animales después de la domesticación y sirven de reservorio de variabilidad en los cuales se sirven los criadores para crear razas estándar o fijas (con una sociedad de cría, reconocidas por un organismo o simplemente fijas). Las poblaciones polimorfas existen también en especies silvestres, pero son por lo general raras, mientras que son frecuentemente observadas en los estadios domésticos, al menos en las especies que son conducidas en hatos y donde el control de apareamiento no es muy estricto, particularmente en ciertas especies de ruminantes criados en cría extensiva. Ciertos locus de coloración del pelaje constituyen lo esencial de los locus con efecto visible en segregación de las poblaciones polimorfas. Estos locus presentan series de alelos homólogos entre las especies cuya identificación, que data desde las primeras décadas de la genética mendeliana ha, desde entonces, sido confirmada por la genética molecular.

**Palabras clave:** Animal doméstico – Animal salvaje – Raza de ganado – Polimorfismo genético – Genética de poblaciones – Frecuencia alélica.

