J. Appl. Genet. 38(2), 1997, pp. 195-204

Review article

Inheritance of colours in horses

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Abstract. The inheritance of colour in horses had been investigated for many years but since Adalsteinsson's findings on basic and diluted colours, made in the seventies, opinions have changed considerably. Many authors supported this hypothesis by detailed breeding and laboratory experiences. This article contains a review of the most important publications concerning the mode of inheritance of all known horse colours. Some of the papers were published long ago but are still valid. The role of different loci determining horse colour is discussed in relation to other mammals and to the current knowledge on melanogenesis. The paper presents references for 14 loci responsible for horse coat colour.

Key words: coat colours, eumelanin, horse, pheomelanin.

Introduction

Basic genes determining the colour of a horse's coat and skin are homologous to those of many mammalian species though certain alleles are often specific (WRIGHT 1917, SEARLE 1968, PROTA, SEARLE 1978, ADALSTEINSSON et al. 1995, KRÄUSSLICH 1996). Albino C-locus is responsible for the intensity of melanin pigmentation. Allele C enables the production of normal amounts of tyrosinase. Together with peroxidase and possibly catalase, tyrosinase catalyzes the initial events of melanogenesis, i.e. the hydroxylation of tyrosine to dopa and its oxydation to dopaquinone which, in turn, can be transformed into melanins (PROTA, SEARLE 1978, PROTA 1988, 1992). Recessive allele c

Received: September 1996.

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strongly reduces tyrosinase production leading to full albinism in mammals other than horses.

There are two main chemical types of melanins: eumelanins and pheomelanins with trichochromes. Brown B-locus determines if the insoluble nitrogenous eumelanins are black or brown. In melanocytes of the skin, eyes and hair follicles, black ellipsoidal eumelanosomes are formed in the presence of dominant allele B, while recessive allele b, when homozygous, produces eumelanosomes which are smaller, round and of chocolate colour (WOOLF, SWAFFORD 1988). Studies carried out on rodents by ITO et al. (1984) have shown that the difference in colour of eumelanosomes results only from the quantity of eumelanin. Hair of a black mouse contains 2-3 times more eumelanin than that of a brown mouse. Yellow to reddish brown pheomelanins contain sulphur in addition to nitrogen, though, on the other hand, they consist of pigments of different chemical nature (PROTA 1988). Pheomelanosomes are of round shape, a little smaller than brown eumelanosomes (WOOLF, SWAFFORD 1988).

Chestnut, bay and black colours

ADALSTEINSSON (1974) published the results of his studies on the colours of Icelandic ponies showing that chestnut colour is due to recessive ee genotype and not, as it was previously thought, to bb genotype. Moreover, he denied the existence of allele b in horses. Later studies made on mice by LAMOREUX and MAYER (1975), among others gave evidence of the responsibility of Extension E-locus for a kind of melanogenesis. Linkage of this locus with Serum Esterase, Roan and Tobiano loci also supported Adalsteinsson's hypothesis (ANDERSSON, SANDBERG 1982). SPONENBERG et al. (1988) proved, using high performance liquid chromatography (HPLC), that black and mouse areas of horse coat are eumelanic, lighter reddish to yellow areas are pheomelanic and dark reddish areas are characterised by mixed melanin type. By homology to rodents, black horses should posses allele E, whereas chestnut individuals should be of ee genotype. If the chestnut colour resulted from allele b, the lighter hair would not contain pheomelanin, but brown eumelanin. Mixed types of melanins found among others in dark chestnuts, were suggested to be due to the weaker action of allele e in horses. Energy dispersive X-ray spectroscopy of melanosomes performed by WOOLF and SWAFFORD (1988) documented the presence of pheomelanosomes in melanocytes of hair follicles and black ellipsoidal eumelanosomes in the melanocytes of skin in chestnut horses. The latter indicates the action of allele B and simultaneously contradicts the dependence of pheomelanogenesis in hair follicles from recessive allele b. In black horses, both in the skin and in the hair follicles, black eumelanosomes

were present, whereas brown eumelanosomes were not found in any of the examined horses.

Thus, locus B in horses does not control colour differentiation, but is responsible for the production of solely black eumelanosomes. Probably, in melanocytes of the skin and eyes only eumelanogenesis may take place, hence their black colour results from the action of this gene. Some authors give examples of horses not typically coloured, in which a presence of allele b is suggested (SPONENBERG, BEAVER 1983, SPONENBERG et al. 1988). Yet, the existence of brown eumelanin in horses has not been documented in laboratory conditions.

Locus E determines if eumelanogenesis or pheomelanogenesis occurs in melanocytes of hair follicles. Allele E enables eumelanogenesis. Allele e, when homozygous, causes pheomelanogenesis – chestnut colour (__ee) appears.

Agouti A-locus is the second factor influencing the differentiation of hair colour in horses. Genotype ee suppresses the action of locus A by virtue of a recessive epistasis (WOOLF, SWAFFORD 1988). In the presence of dominant allele E, allele A reduces the occurrence of the eumelanogenesis to the mane, tail, parts of the head and the bottom areas of the limbs – so called "points". Then, in hair follicles of most of the body areas, pheomelanogenesis takes place – a horse becomes bay coloured (A_E_) (ADALSTEINSSON 1974). Evidence has been given by WOOLF and SWAFFORD (1988) who found black eumelanosomes in the base of the mane and pheomelanosomes in hair follicles of yellow-red body areas in light bay horses. Recessive genotype aa does not limit the eumelanogenesis, which takes place in all melanocytes of hair bulbs – black colour (aaE_) is formed.

The agouti pattern in horses differs from that of rodents as hair is not striped. WOOLF and SWAFFORD (1988) noticed similarity between the effects of allele A in horses and allele a^t in mice. Both alleles cause pheomelanogenesis in hair bulbs of certain body areas, in contrast to allele A in mammals other than horses, where it results in the alternate secretion of two pigments from a melanocyte.

The hypothese on the existence of allele A^+ determining bay colour with dorsal stripe (ODRIOZOLA 1951) and allele a^t producing seal brown colour (ODRIOZOLA 1951, ADALSTEINSSON 1974, 1975) have not been confirmed. Another hypothesis on allele E^D occurring in horses, as in rabbits and some other mammals, and causing a kind of black phenotype called "jet black", put forth by CASTLE (1951), has also been called into question (ODRIOZOLA 1951, WOOLF 1992). However, black horses are assumed not to be a uniform group,

for in addition to typical aaE_ genotypes they include some dark bay A_E_ individuals which, because of their dark tint, are identified as black (NEBE 1984, WOOLF, SWAFFORD 1988, WOOLF 1992).

Genetic basis of variations of the discussed colours is not clearly explained.

Diluted C^{cr} colours

As mentioned before, recessive allele c does not exist in horses. Instead, incompletely dominant "cremello dilution" allele C^{cr} occurs. When heterozygous, it causes certain diminution of tyrosinase production (ODRIOZOLA 1951, ADALSTEINSSON 1974). In consequence, eumelanogenesis is continued, whereas the second step of melanogenesis, i.e. pheomelanogenesis, is reduced. Then, eumelanic body areas remain black and pheomelanic parts are diluted. SPONENBERG and BEAVER (1983) suggest that eumelanic areas may also be a little diluted.

Thus, in the presence of chestnut or bay genes, diluted CC^{cr} colours appear: palomino (__eeCC^{cr}) which has been called "isabel" in Poland and some other countries, and buckskin (A_E_CC^{cr}) phenotypically similar to dun, respectively. The latter colour has not been known in Poland so far. The effect of gene C^{cr} is not always visible in dark bay and seal brown horses since, as it has already been shown, they have little amount of pheomelanin usually mixed with eumelanin (SPONENBERG et al. 1988). Probably, in typically eumelanic black and mouse colours, the action of allele C^{cr} is not conspicuous at all (ADALSTEINSSON 1974), though SPONENBERG and BEAVER (1983) postulate that due to this gene black horses can become "smoky black".

It is not well defined why the mane and tail in palominos are more susceptible to dilution than other hair. GUERTS (1977) attributes this phenomenon to different structure of long hair. The origin of hazel eyes and pink skin sporadically occurring in palominos is also unknown. SPONENBERG and BEAVER (1983) suspect it may result from the action of b allele.

In homozygotes $C^{cr}C^{cr}$ both eumelanin and pheomelanin are diluted to pale cremello colour (ADALSTEINSSON 1974, 1978, van VLECK, DAVITT 1977). Some authors call homozygous chestnuts cremello and slightly darker homozygous bays, perlino (GUERTS 1977, van VLECK, DAVITT 1977, SPONENBERG, BEAVER 1983). The question is whether the doubly diluted cremello colour can also appear on eumelanic colours, as it was postulated by ADALSTEINSSON (1975). SPONENBERG and BEAVER (1983) suggest that the homozygous effects on black and mouse horses are probably "silver smoky" and "silver grullo", respectively, both similar to mouse, but with blue eyes.

Diluted D colours

Dilution D-locus sometimes called "dun" factor in horses controls the intensity of eumelanogenesis and pheomelanogenesis at body areas and, to a minimum degree, at points (ADALSTEINSSON 1974, 1975, van VLECK, DAVITT 1977, CRAIG, van VLECK 1985). Dominant allele D reduces pigment synthesis, whereas recessive allele d allows full pigmentation. In the presence of allele D, a chestnut horse becomes red dun (__eeD_), bay becomes yellow dun with a dark mane and tail (A_E_D_) and black becomes mouse (aaE_D_). The action of locus D in horses differs from that of rodents, in which recessive homozygotes (dd) produce clustered pigment granules and of a changed shape (PROTA, SEARLE 1978, SPONENBERG et al. 1988).

Joint effects of D and C^{cr} genes have not been documented sufficiently. ADALSTEINSSON (1974) distinguishes "dun palomino" and "dun buckskin" genotypes without giving any phenotypic description. According to SPONENBERG and BEAVER (1983), the effect of combining these alleles is not additive, i.e. the horse is not lighter than it would be with a single effect alone. Thus, black would be diluted to grullo (mouse), bay to dun with zebra markings and chestnut to palomino with dorsal stripe.

Diluted Z colour

Silver dapple Z-locus (also called S or D^S) reduces eumelanin synthesis on the body and on points. Hence, it affects only eumelanic colours, such as black and dark bay changing them into silver dappled (__E_Z_). Homozygotes ZZ are a little lighter than heterozygotes (CASTLE, SMITH 1953, GUERTS 1977). In Poland silver dappled horses have not been registered so far.

Some silver dappled horses can be confused with light chestnuts. If mated *inter se* or with chestnuts, they can give bay foals (A_E_Zz × A_eezz = A_Eezz) (SPONENBERG, BEAVER 1983). It may lead to a faulty conclusion on the inheritance of chestnut colour (TROMMERSHAUSEN-SMITH et al. 1976).

According to GUERTS (1977), there are some silver dappled horses with light hazel or blue eyes, which cannot be explained in the light of current knowledge on the genetics of the horse.The interaction of alleles Z and G results in the so-called grey-white coat colour noticed in Shetland ponies. The foals are grey or incompletely grey at birth, but turn white very quickly (CASTLE, SMITH 1953, GUERTS 1977).

Colours including white hair

Roan is a result of dominant Rn (or R) allele (HINTZ, van VLECK 1979). Dominant homozygotes (RnRn) are considered to be usually lethal, although

several such horses have been identified (BUTAY 1974, SPONENBERG, BEAVER 1983).

Dominant gene G causes grey colour by reducing the pigment production in hair, progressively with age, as it is in humans (TROMMERSHAUSEN-SMITH 1977a). It is epistatic to all loci except W-locus.

Dominant To (or T) allele causes tobiano colour, currently widespread in Poland. Tobiano horses mated *inter se* sometimes give white foals, which has not been genetically explained (GUERTS 1977).

Overo colour ocurring mainly in North and Latin America, in the light of the latest studies made by BOWLING (1994), is inherited as a dominant trait (O gene). However, this colour is affected by more genes, as sporadically it appears in foals born from matings of "non-overo" parents. The latter caused the fact that for a long time overo was considered recessive, due to at least two loci or a series of alleles at a single locus (TROMMERSHAUSEN-SMITH 1977b, JONES 1979). Overo colour is associated with the so-called "overo white foal syndrome" (TROMMERSHAUSEN-SMITH 1977b, JONES 1979, VONDERFECHT et al. 1983, BOWLING 1994). Overo × overo matings produce overo, solid colour and white foals with only a little amount of pigment in the eyes. Sometimes white foals are born to overo crossed with solid colour parents. Most of white foals develop symptoms of colic a few hours after birth. They fail to pass meconium due to intestinal aganglionosis. It may be attributed to homozygosity of the overo gene (BOWLING 1992). Viable white foals can result from an additive effect of heterozygous To and O genes (BOWLING 1994).

Sabino colour is due to an incompletely dominant gene Sb (GUERTS 1977, SPONENBERG, BEAVER 1983). Heterozygotes are typically sabino, whereas dominant homozygotes are white with few pigment spots. Sabinos mated *inter se* can also give completely white foals. Sabino colour occurs in America as well as in Europe, among others in descendants of East Prussian horses.

Splashed white colour is determined by recessive allele spl occurring among some northern European breeds of horses (KLEMOLA 1933).

Lp-locus is responsible for the complex of leopard spotting variations consisting of the leopard, few-spot leopard, blanket, blanket with spots, varnish roan (or marble), snowflake, frosted, speckled, and mottled patterns (SPONENBERG 1982, SPONENBERG et al. 1990). The patterns are closely related to one another as all of them may appear in every breed, in which any of the patterns occurs. This takes place even in spite of intense selection against certain leopard variants in some breeds (Noriker, Welsh). A great diversity of patterns is attributed either to modifiers affecting the expression of single dominant allele Lp, or to multiple alleles at the single locus.

Table 1. Genotypes and phenotypes of coat colours in horses

Colours	Genotype – Loci	Phenotype		
	A E D C Others	Colour of body	Colour of points	Other traits
Chestnut	__ ee dd CC	yellowish-reddish-brown	yellowish-reddish-brown	
Bay	A_ E_ dd CC	brown	black	
Black	aa E_ dd CC	black	black	
Palomino	__ ee dd CC^{cr}	gold	flaxen to white	
Buckskin	A_ E_ dd CC^{cr}	yellowish-brown	mixed (dark)	
Cremello	__ ee __ C^{cr} C^{cr} A_ E_ __ C^{cr} C^{cr}	pale cream	pale cream	pink skin, blue eyes
Red dun	__ ee D_ CC	yellowish-reddish-brown	yellowish-reddish-brown	zebra marking
Dun	A_ E_ D_ CC	yellowish-brown	mixed (dark)	zebra marking
Mouse	aa E_ D_ CC	brownish-grey	mixed	zebra marking
Silver dappled	__ E_ dd CC Z_	dark cream to brown	mixed white-brown to white	usually dappled
Roan	__ __ __ C_ Rn_	mixed white	mixed	not progressive pattern
Grey	__ __ __ C_ G_	white	white	progressive pattern
Tobiano	__ __ __ C_ To_	asymmetric white and coloured areas		
Overo	__ __ __ C_ O_	asymmetric white and coloured areas		
Sabino	__ __ __ C_ Sb_	asymmetric white and coloured areas		
Splashed white	__ __ __ C_ splspl	asymmetric white and coloured areas		
Leopard	__ __ __ C_ Lp_	symmetric white and coloured areas		
dominant white	__ __ __ __ Ww	white	white	pink skin, blue eyes

Homozygous leopards tend to have more extensive patterns than heterozygous horses do (SPONENBERG 1982, SPONENBERG et al. 1990). Cases of white foals born to leopard parents (SPONENBERG 1982) and certain linkage of the extension of the pattern with sex (GUERTS 1977) have not been explained.

Dominant white colour results from the action of single dominant allele W. Locus W is epistatic to all loci controlling horse coat colour. Dominant homozygotes WW are lethal at an early embryonic stage, which was proved with a proportion of coloured to white foals born to dominant white parents mated *inter se* (PULOS, HUTT 1969).

Conclusions

14 autosomal genes controlling coat colour in horses have been identified until now. B-Locus can be omitted because of its lack of influence on the differentiation of the known horse colours. All colours including white hair, except the epistatic dominant white, can occur in association with every other colour (SPONENBERG, BEAVER 1983, BOWLING 1992, 1994). Loci Rn, To and E are closely linked in Linkage Group II (TROMMERSHAUSEN-SMITH 1978, ANDERSSON, SANDBERG 1982, SPONENBERG et al. 1984, BOWLING 1987).

Undoubtedly, future results of cytogenetic and molecular studies will verify to a certain extent current knowledge on the inheritance of horse colours.

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