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Coat color genetics in farm animals

Modul no.2: Animal Genetics

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LECTURE CONTENT

- The importance of coat color
- Coat color and domestication
- Pigment synthesis
- Major coat-color-associated genes
- Basic cattle colors
- Pleiotropic effect of pigmentation genes

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THE IMPORTANCE OF COAT COLOR

- The syntheses of pigment do have several functions. Coat color is of importance for mating, for protection against predators, and as an adaptation to the environment.

Photo: A. Andersson, 2020

- In fur animals, the coat color is an important production trait.

- In other species such as cattle and sheep, the coat color is an important breed characteristic.

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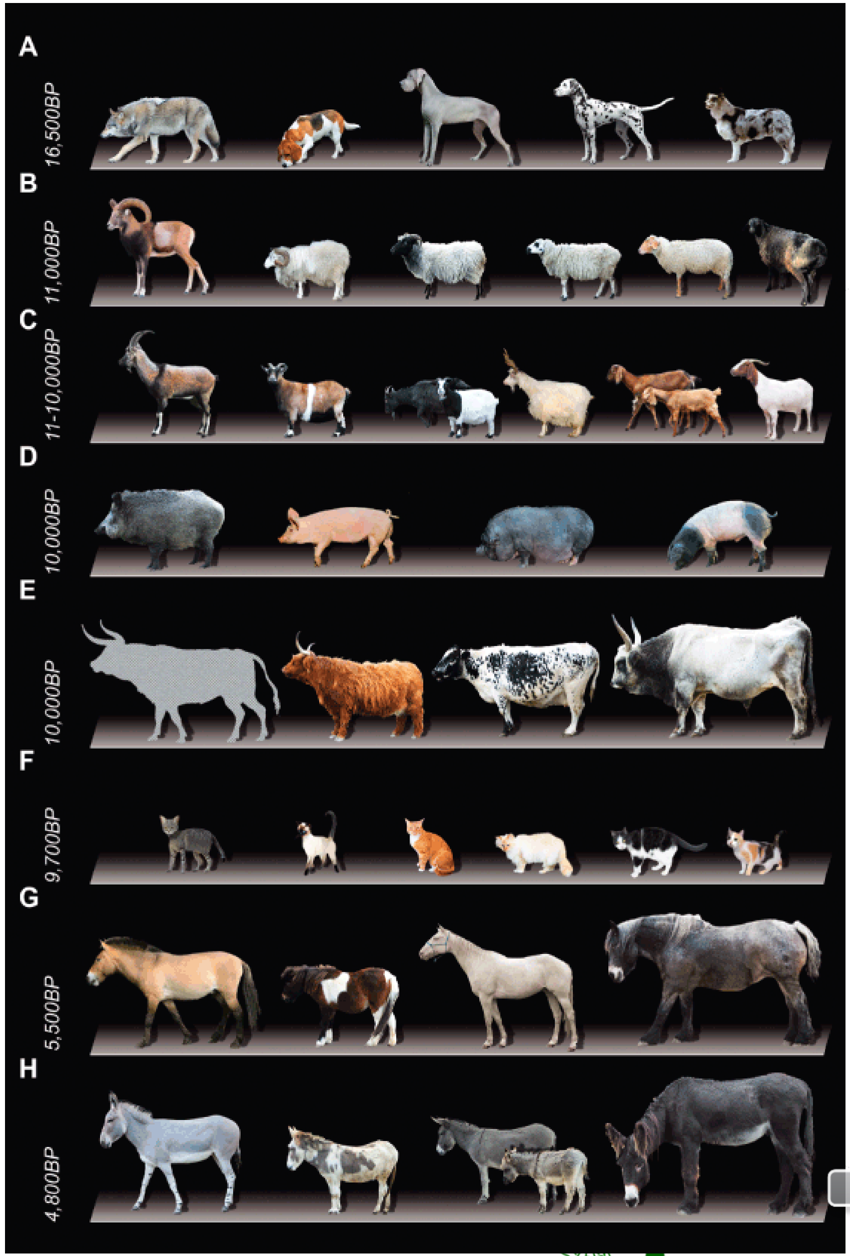
COAT COLOR AND DOMESTICATION

- In contrast to their wild ancestors, domesticated species are often characterized by a substantial allelic variability of coat-color-associated genes.
- Recent studies demonstrate that the selection for coat-color phenotypes started at the beginning of domestication.
- Characteristic of modern domesticated animals is their large phenotypic variation, which is not found in any of their wild ancestors.
- Wild species are usually uniform in phenotype and show species-specific colors and patterns. By contrast, domesticated species are highly variable in both colors and color patterns.



Cieslak et al. (2011)

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PIGMENT SYNTHESIS

- Although to date more than 300 genetic loci and more than 150 identified coat-color-associated genes have been discovered, which influence pigmentation in various ways (Cieslak et al., 2011)
- The process of pigment cell development is crucial for the determination of mammalian coat coloration. The pigmentation progress is subdivided into three major stages: **melanocyte development**; **pigment production**; and **pigment distribution**.
- Melanocytes can produce the two different pigments - **eumelanin** (dark brownish to black) and **pheomelanin** (yellowish to reddish).

MAJOR COAT-COLOR-ASSOCIATED GENES

ASIP – agouti signalling protein

MC1R – melanocortin 1 receptor

CBD103 - *beta defensin 103B*

KITL - *KIT ligand*

MATP (SLC45A2) - *solute carrier family 45 member 2*

MITF - *microphthalmia-associated transcription factor*

MLPH - *melanophilin*

MYO5A - *myosin VA*

PMEL17 (SILV) - *silver homolog*

POMC - *propiomelanocortin*

P-protein (OCA2) - *oculocutaneous albinism II*

SLC36A1 - *solute carrier family 36 member 1*

STX17 - *syntaxin 17*

TRPM1 - *transient receptor potential cation channel subfamily M member 1*

TYR - *tyrosinase*

TYRP1 - *tyrosinase-related protein 1*

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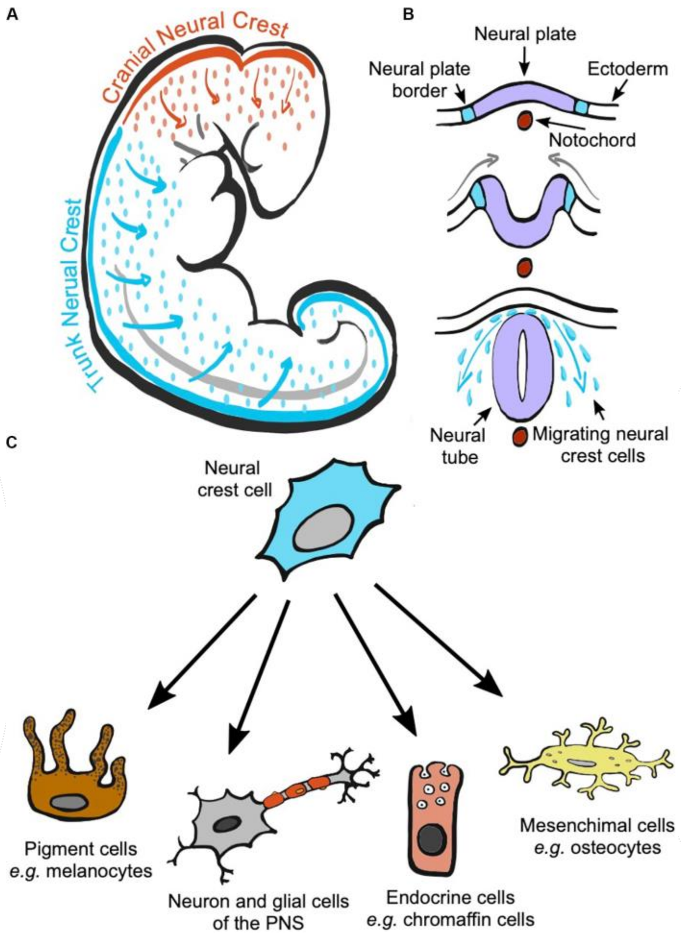
- During vertebrate embryogenesis, neural crest cells arise along the dorsal neural tube, and some differentiate into melanoblasts (precursors of melanocytes), which migrate ventrally along the body.
- Melanoblasts typically enter the epidermis, where some remain, while others localize to the hair follicles and differentiate into melanocytes.
- These melanocytes produce pigment.
- Once pigment is produced, it is packaged into melanosomes and transferred to keratinocytes of developing hair (or epidermal cells).



The neural crest and melanocytes. A: location of the neural crest in the embryo. B: development of the neural crest.
C: neural crest cells migrate to different cell types. (San-Jose & Roulin, 2020)

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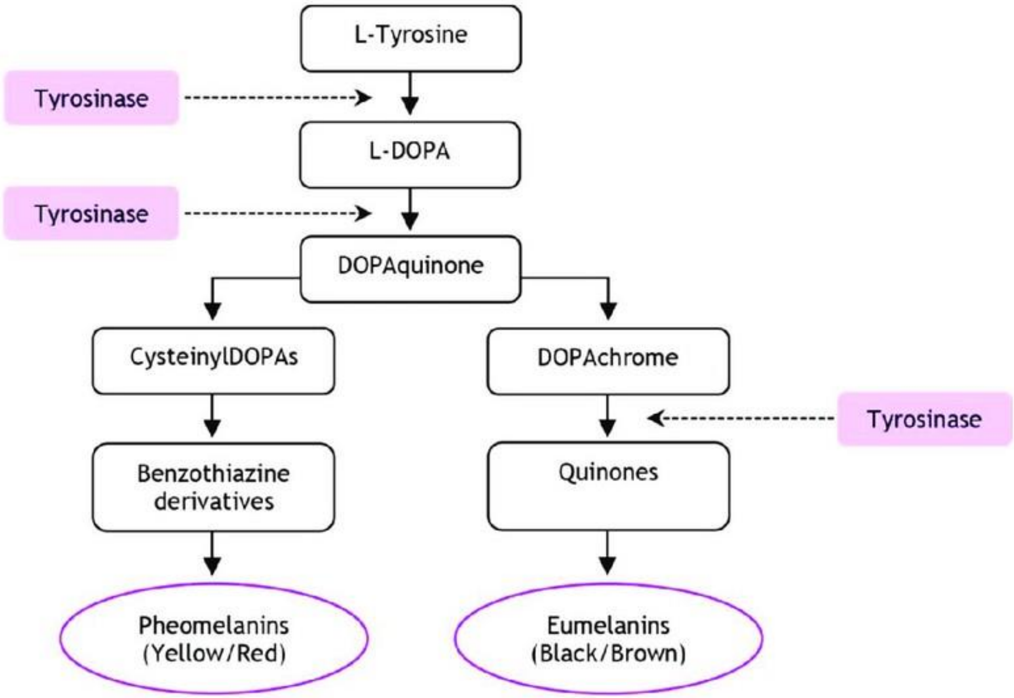
Melanocytes can produce the two different pigments:

eumelanin (dark brownish to black) and

pheomelanin (yellowish to reddish).



Raoufinejad and Rajabi, 2021

Tyrosinase is the rate-limiting enzyme in melanogenesis. There are over 100 alleles of tyrosinase that have been characterized, ranging from null alleles, resulting in the complete absence of pigmentation (**albinism**), to alleles with reduced function that limit the melanin production (e.g. cremello horses).

Damé et al., 2012

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COAT COLOR PHENOTYPES

- **solid colored** phenotypes (pigmented to white)
- **patterned** phenotypes (e.g. dorsal-ventral pigmentation, white spotted, spotted, striped, leopard)

- The basic coat colorations are defined by the **ratio of the two pigments eumelanin and pheomelanin**. Their ratio is primarily controlled by the **agouti signalling protein (ASIP)** and **melanocortin 1 receptor (MC1R)** ligand-receptor system, which determines the ratio between the two types of melanin.

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BASIC CATTLE COLORS

EXTENSION (E) LOCUS

Three alleles: E^D, $E^+$and *e*

The E^D allele is for dominant black, E^+ is the wild-type allele and is responsible for combinations of red or reddish brown and black and *e* is the recessive red. Cattle that are e/e are red, E^D/. are typically black (as E^D is the dominant allele), E^+/. are „wild type“ red.

The order of dominance is $E^D > E^+ > e$.

SPOTTING (S) LOCUS

Four alleles: S^+(non-spotting), S^H, S^p, and *s*. The S^H (homozygous) is seen in Herefords (S^H/S^H), S^p is responsible for lined back-pattern seen in Pinzgauers and *s* (recessive spotting) makes up for the irregular white spotting seen in breeds like Holstein.

The order of dominance is $S^H = S^p > S^+ > s$.

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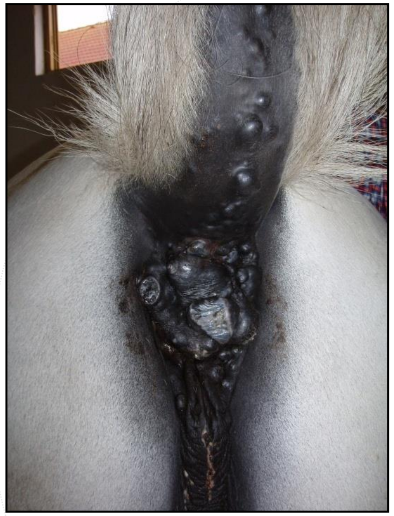
PLEIOTROPIC EFFECT OF PIGMENTATION GENES

Some coat-color phenotypes are associated with or linked to disorders.

e.g.

- Oculocutaneous albinism (vision disorders)
- Overo pattern in horses (homozygote lethal white syndrome)
- Leopard complex in horses (homozygote night blindness)
- Silver dapple dilution in horses (multiple congenital ocular anomalies)
- Greying in horses (melanoma)
- Merle pattern in dogs (homozygote are often deaf and sterile)
- Some KIT genes mutations can affect reproductive performance (mice, cattle) or neither be lethal (horses, hamsters)



https://www.vetlexicon.com/equis/gastrohepatology/articles/intestine-aganglionosis/

https://www.petmojo.com/what-is-a-double-merle-dog/

PLEIOTROPIC EFFECT OF PIGMENTATION GENES

Behaviour

- Although the link between tame behaviour and coat coloration is not well understood, mutations affecting receptor functions could be directly linked to both traits.
- Domestication studies on foxes, rats and minks strongly supported a close relationship between the selection for tameness and a manifestation of novel color variants.
- There is a speculated correlation between pale coat color and lower aggressiveness related to the common synthesis pathway of adrenalin and dopaquinone from DOPA.

- There are several examples highlighting the correlation between coat color and temperament:
  - albinos are less robust,
  - horses with the cream allele are gentle and tame,
  - piebald cattle, white sheep and swine are more sensitive to certain herbs,
  - red Cocker Spaniels are much more nervous than other phenotypes of this breed.

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CONCLUSION

- Artificial selection is the main factor responsible for the large phenotypic variation observed in domesticated animals.
- Selection for color phenotypes started at the beginning of domestication, resulting in a significant increase of polymorphism in coat-color-associated loci.
- The production and ratio of eumelanin/pheomelanin is mainly regulated by the MC1R and ASIP genes.
- Some coat-color phenotypes are associated with or linked to disorders.

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Thank you for your attention!

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