Open Access Article

Breed Distribution and Allele Frequencies of Base Coat Color, Dilution, and White Patterning Variants across 28 Horse Breeds

by Felipe Avila [1], Shayne S. Hughes [1], K. Gary Magdesian [2], Maria Cecilia Torres Penedo [1] and Rebecca R. Bellone [1,3,*]

UC Davis Veterinary Genetics Laboratory · Follow
September 16, 2022 ·

VGL Research: New publication by VGL researchers sheds light on breed distribution and allele frequencies of coat color variants in horses.

The recent publication consists of the largest study to date investigating pigmentation variants in horses! Twenty-nine genetic variants that determine base coat color, dilution and white patterning were investigated in 11,281 horses from 28 breeds. The results of this study will help guide breed-specific recommendation on genetic testing for coat color in the horse.

For more on the results read our news story at https://vgl.ucdavis.edu/.../new-publication-vgl...

The full article can be found here: https://www.mdpi.com/2073-4425/13/9/1641/htm

UCDAVIS VETERINARY MEDICINE
Veterinary Genetics Laboratory

VGL researchers shed light on breed distribution and allele frequencies of equine coat color variants in new publication

Abstract

Since domestication, horses have been selectively bred for various coat colors and white spotting patterns. To investigate breed distribution, allele frequencies, and potential lethal variants for recommendations on genetic testing, 29 variants within 14 genes were investigated in 11,281 horses from 28 breeds. The recessive chestnut e^a allele in melanocortin 1 receptor (*MC1R*) (p.D84N) was identified in four breeds: Knabstrupper, Paint Horse, Percheron, and Quarter Horse. After filtering for relatedness, e^a allele frequency in Knabstruppers was estimated at 0.035, thus illustrating the importance of testing for mate selection for base coat color. The Rocky Mountain Horse breed had the highest allele frequency for two of the dilution variants under investigation ($Z_{a.f.}$ = 0.32 and $Ch_{a.f.}$ = 0.026); marker-assisted selection in this breed could aid in the production of horses with desirable dilute coats with less severe ocular anomalies caused by the silver (*Z*) allele. With regard to white patterning, nine horses homozygous for the paired box 3 (*PAX3*) splashed white 2 (*SW2*) allele (p.C70Y) and six horses homozygous for the KIT proto-oncogene, receptor tyrosine kinase (*KIT*) sabino 1 (*SB1*) allele (ECA3g.79544206A>T) were identified, thus determining they are rare and confirming that homozygosity for *SW2* is not embryonic lethal. The *KIT* dominant white 20 (*W20*) allele (p.R682H) was identified in all but three breeds: Arabian (n = 151), Icelandic Horse (n = 66), and Norwegian Fjord Horse (n = 90). The role of *W20* in pigmentation across breeds is not well understood; given the different selection regimes of the breeds investigated, these data provide justification for further evaluating the functional role of this allele in pigmentation. Here, we present the largest dataset reported for coat color variants in horses to date, and these data highlight the importance of breed-specific studies to inform on the proper use of marker-assisted selection and to develop hypotheses related to pigmentation for further testing in horses.

Keywords: pigmentation; white spotting; genetic testing; equine

Outline

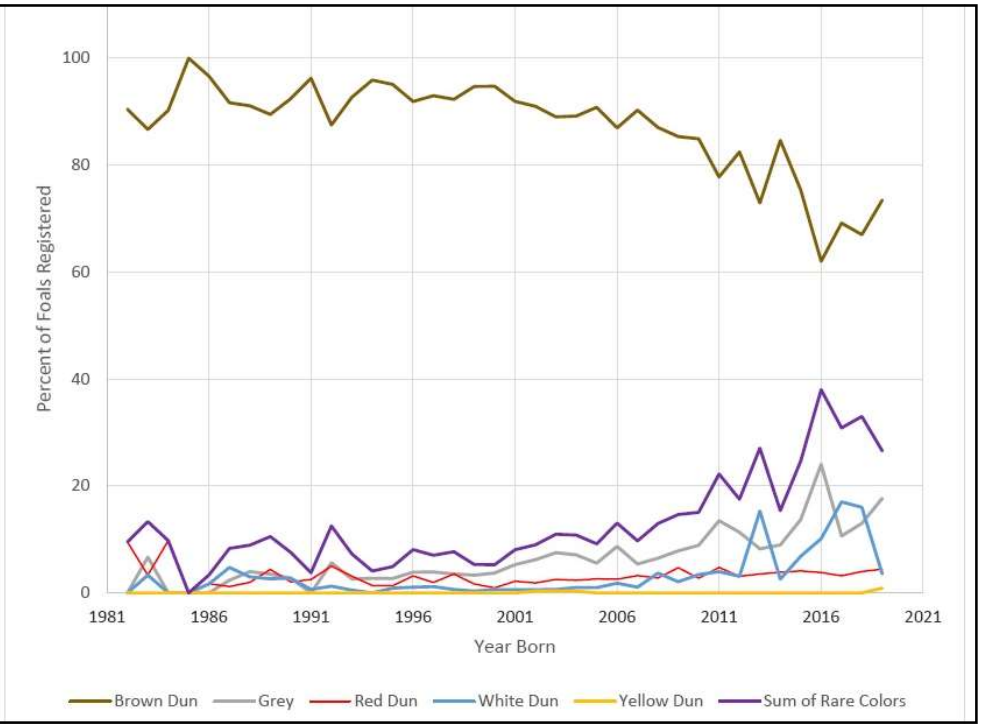
- Population trends
- Colors of Fjords
- Genetic basis of inheritance
- Color testing
- Interpreting color tests
- Kvit
- White marks
- Foal colors
- Other color variations

White dun, Brown dun, Grey, Yellow dun, Red dun

5 Colours of Fjords, photos by Marissa Orsel

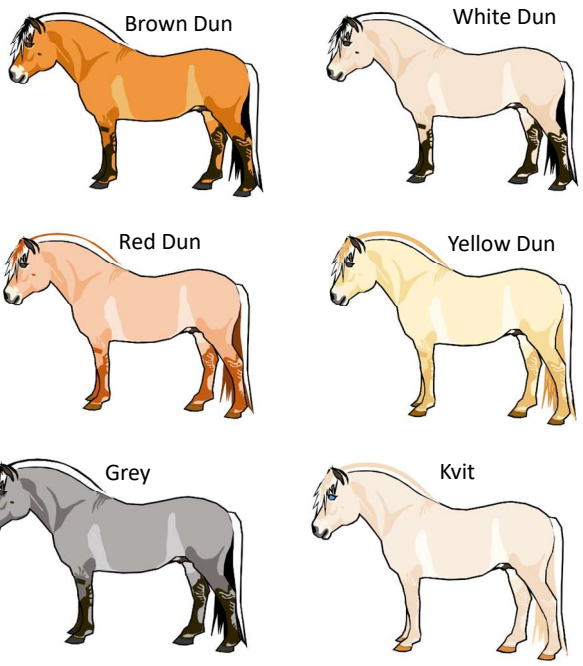
Percent of NFHR Fjord foals by color registered from 1982 to 2019

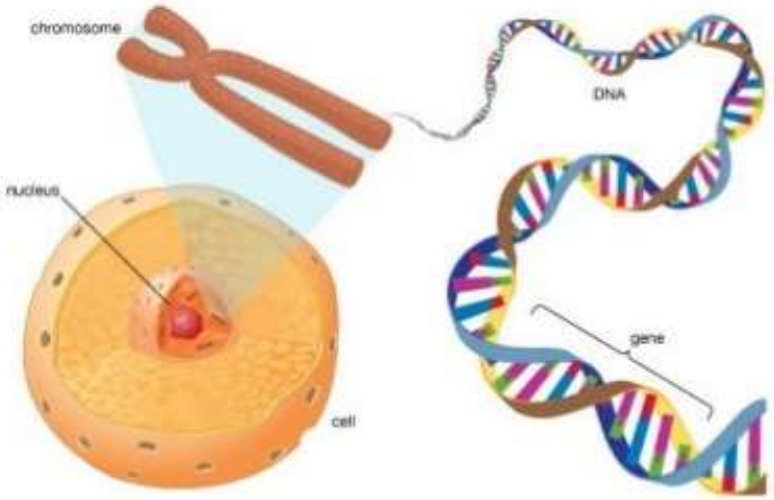
Graphic and Article by Cherrie Nolden, The Fjord Herald, Issue# 134, Spring 2020



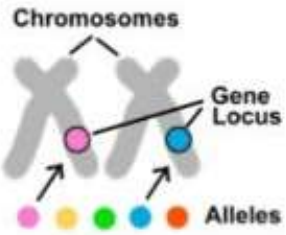
Fjord Color Genetics

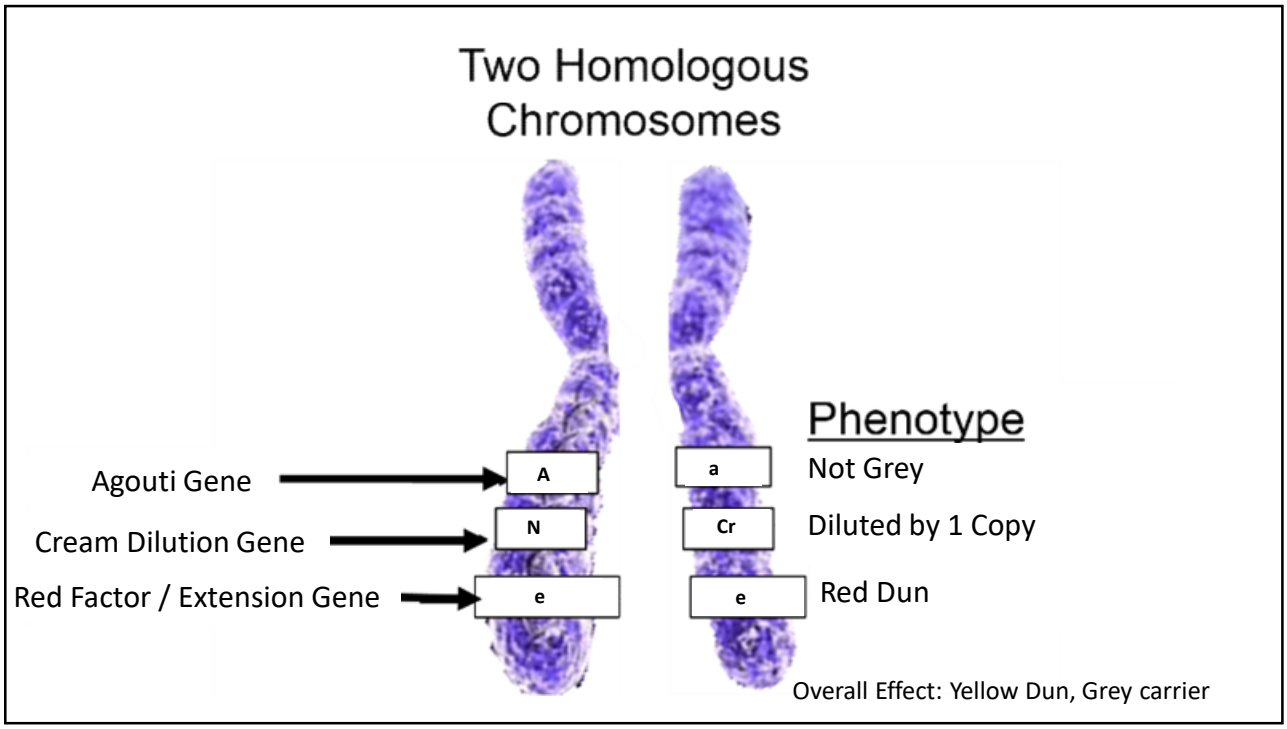
- 5 accepted colors (Kvit is not)
  - 9 colors, really
  - Norwegian names for colors
    - 2 Grey colors lumped under one name
    - 3 Kvit colors lumped together
- All Fjords are Dun
  - D/D nearly ubiquitously
  - Rarely D/nd1 in Australia, Europe
- 3 color genes create the colors
  - Agouti
  - Red Factor / Extension
  - Cream Dilution





Genes are 'sections' of the DNA and can exist on each strand in alternative forms called alleles. Alleles may be dominant or recessive and give rise to differences in inherited characteristics:





Genetic Terminology

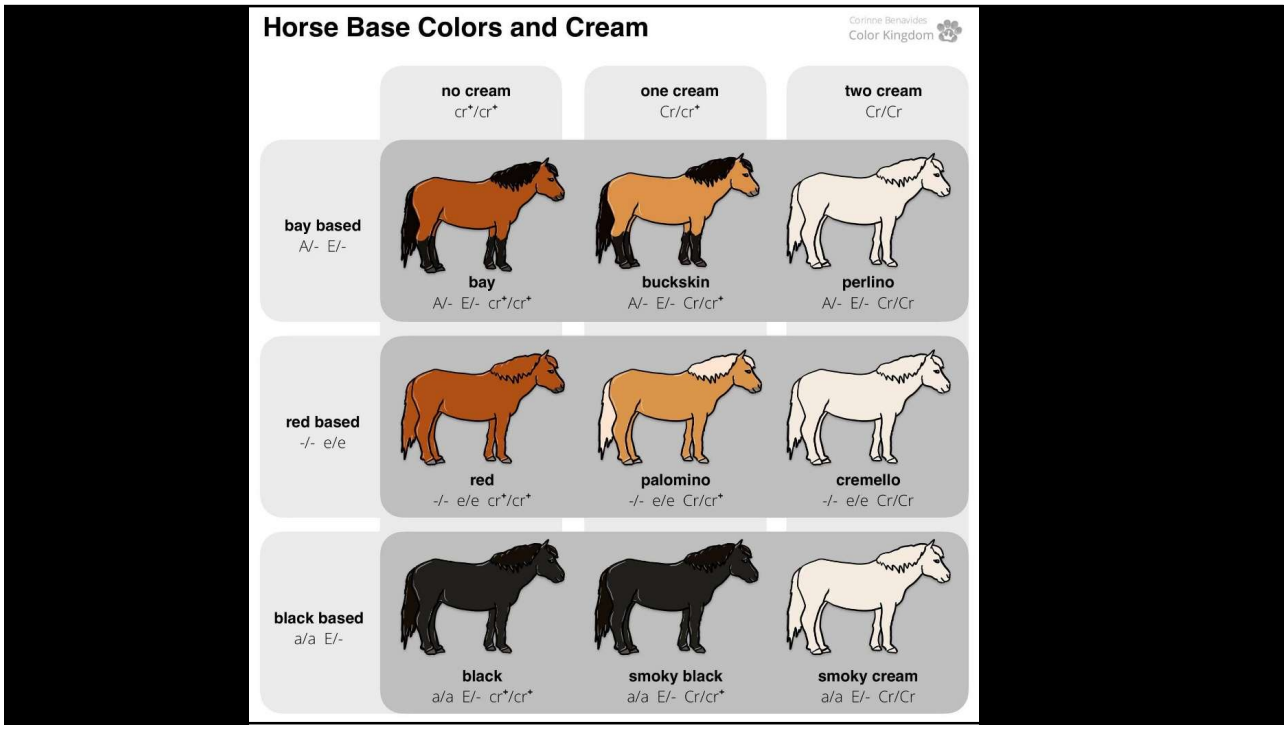
- Trait – any characteristic that can be passed from parent to offspring
- Heredity – passing traits from parent to offspring
- Genetics – study of heredity
- Monohybrid cross – cross involving a single trait e.g. Agouti color
- Dihybrid cross – cross involving two traits e.g. Agouti and Red Factor colors
- Punnett Square – grid used to help solve simple genetics problems

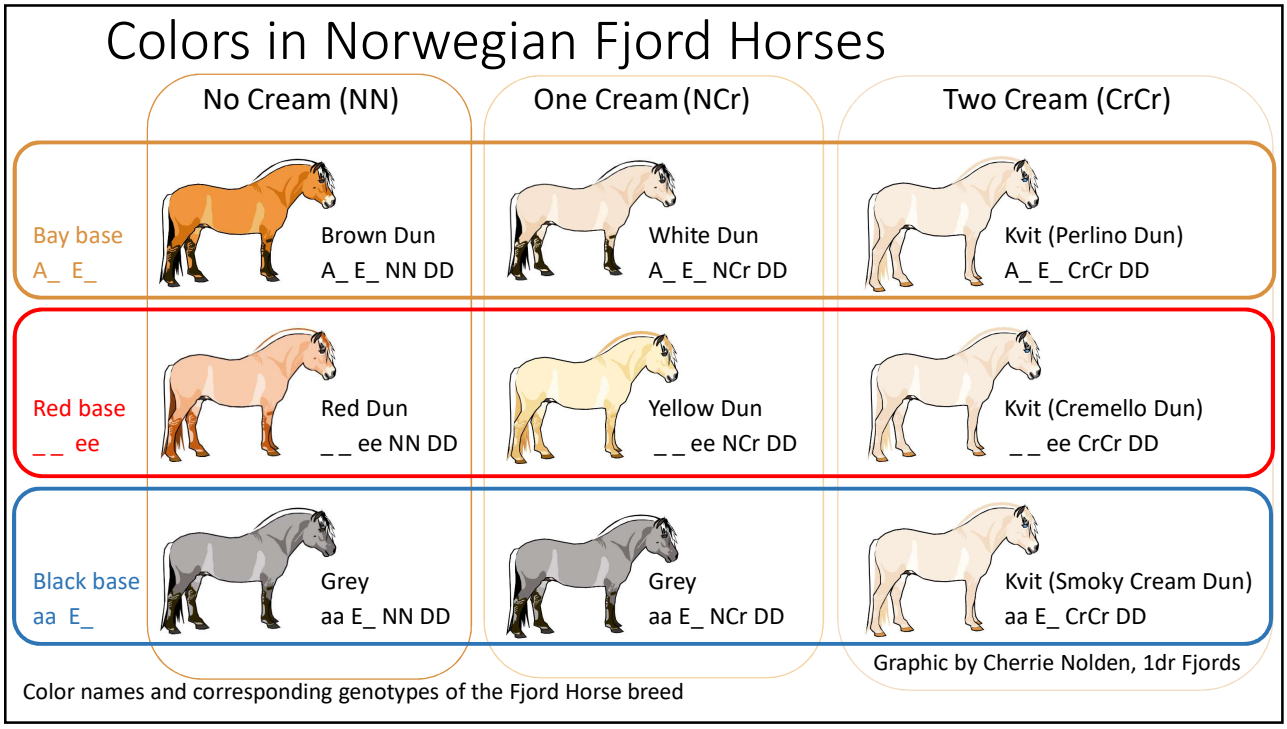
More Gene Terminology

- Alleles – two forms of a gene (dominant and recessive)
- Dominant – stronger of two genes expressed in a hybrid; represented by a capital letter (A, Agouti gene; E, Red Factor gene)
- Recessive – gene that shows up less often in a cross; represented by a lowercase letter (a, Agouti gene; e Red Factor gene)
  - One copy from each parent is required to produce the recessive color
- Incompletely Dominant – gene that has a dose-based effect
  - Cream Dilution gene in Fjords
  - 2 alleles: N (no dilution), Cr (dilution of red base color)
  - Not recessive or dominant
  - Only 1 copy, from 1 parent, is required to produce the rare color

3 Color Genes, 6 Alleles, with Descriptions

Gene	Alleles	Phenotype with Description
Agouti	AA	Brown dun, does not carry Grey
	Aa	Brown dun, carries Grey
	aa	Grey (unless also ee; then Red dun)
Red Factor / Extension	EE	Brown dun, does not carry Red dun
	Ee	Brown dun, carries Red dun
	ee	Red dun (could be genetically Grey if also aa)
Cream Dilution	NN	Brown dun, Red dun or Grey
	NCr	White dun (if also A_), Yellow dun (if also ee), or Grey if aa E_
	CrCr	Kvit (blue eyes, cream skin and hooves, very dilute color)


Horse Base Colors and Cream
Corinne Benavides
Color Kingdom
no cream
cr+/cr+
one cream
Cr/cr+
two cream
Cr/Cr
bay based
A/- E/-
bay
A/- E/- cr+/cr+
buckskin
A/- E/- Cr/cr+
perlino
A/- E/- Cr/Cr
red based
-/- e/e
red
-/- e/e cr+/cr+
palomino
-/- e/e Cr/cr+
cremello
-/- e/e Cr/Cr
black based
a/a E/-
black
a/a E/- cr+/cr+
smoky black
a/a E/- Cr/cr+
smoky cream
a/a E/- Cr/Cr


Colors in Norwegian Fjord Horses
No Cream (NN)
One Cream (NCr)
Two Cream (CrCr)
Bay base
A_ E_
Brown Dun
A_ E_ NN DD
White Dun
A_ E_ NCr DD
Kvit (Perlino Dun)
A_ E_ CrCr DD
Red base
_ _ ee
Red Dun
_ _ ee NN DD
Yellow Dun
_ _ ee NCr DD
Kvit (Cremello Dun)
_ _ ee CrCr DD
Black base
aa E_
Grey
aa E_ NN DD
Grey
aa E_ NCr DD
Kvit (Smoky Cream Dun)
aa E_ CrCr DD
Graphic by Cherrie Nolden, 1dr Fjords
Color names and corresponding genotypes of the Fjord Horse breed

Outline

- Population trends
- Colors of Fjords
- Genetic basis of inheritance
- Color testing
- Interpreting color tests
- Kvit
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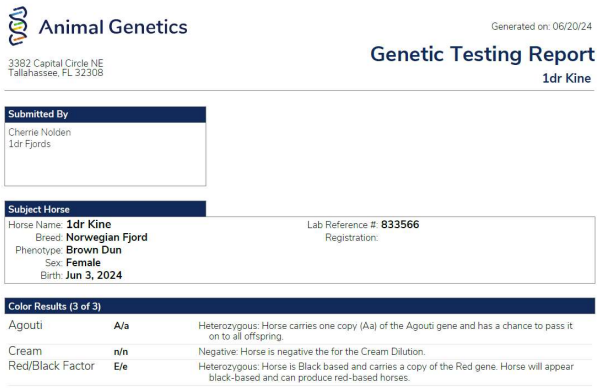
- Other color variations

White dun, Brown dun, Grey, Yellow dun, Red dun

5 Colours of Fjords, photos by Marissa Orsel

Color Testing

- Animal Genetics
  - $40 for the base color test (Agouti and Red Factor together)
  - $25 for Cream Dilution
- University of KY
  - $25 each for the 3 color genes
- University of California-Davis VGL
  - $85 for Agouti, Red Factor and Cream Dilution
- Etalon Diagnostics
  - $149 for all colors, health and temperament tests



Animal Genetics

3382 Capital Circle NE
Tallahassee, FL 32308

Generated on: 06/20/24

Genetic Testing Report

1dr Kine

Submitted By

Cherrie Nolden
1dr Fjords

Subject Horse

Horse Name: **1dr Kine**
Breed: **Norwegian Fjord**
Phenotype: **Brown Dun**
Sex: **Female**
Birth: **Jun 3, 2024**

Lab Reference #: **833566**
Registration:

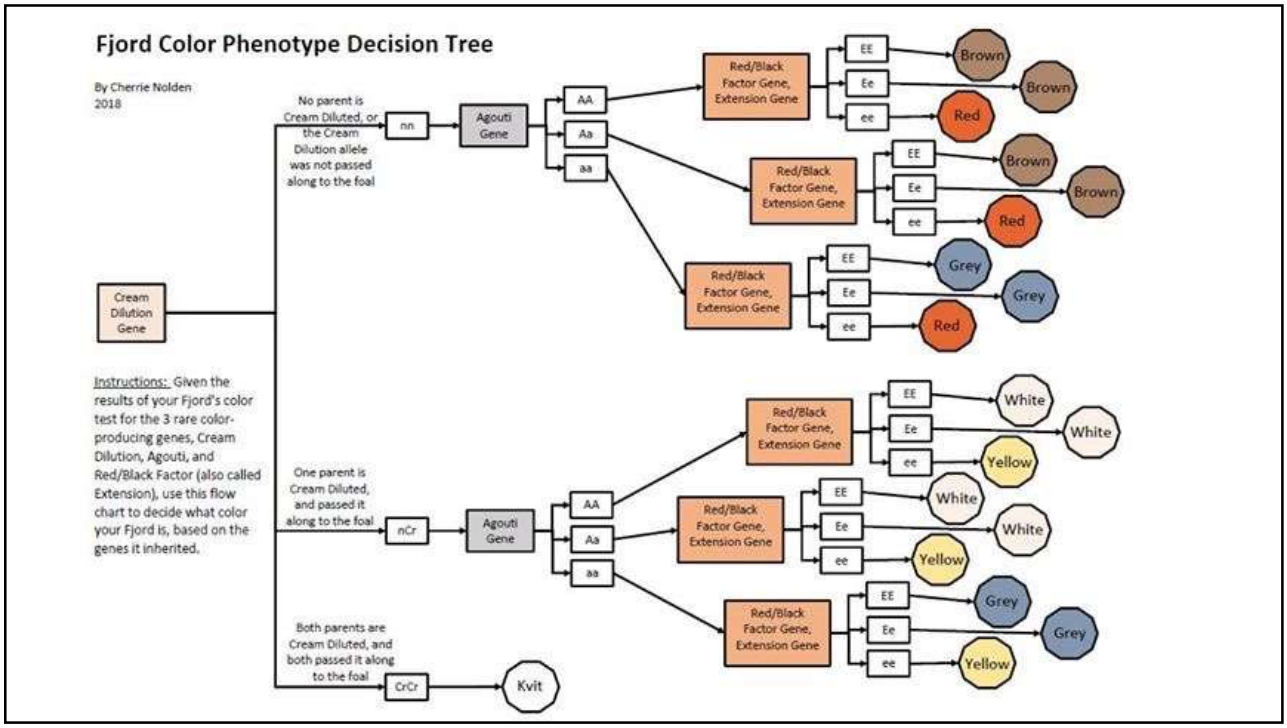
Color Results (3 of 3)

Agouti	**A/a**	Heterozygous: Horse carries one copy (Aa) of the Agouti gene and has a chance to pass it on to all offspring.
Cream	**n/n**	Negative: Horse is negative the for the Cream Dilution.
Red/Black Factor	**E/e**	Heterozygous: Horse is Black based and carries a copy of the Red gene. Horse will appear black-based and can produce red-based horses.

Phenotypes and Corresponding Genotypes

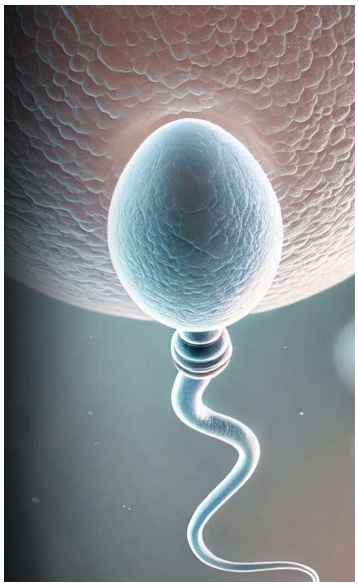
The genotypes below are the possibilities for the three variable hair color genes in Fjords.

Phenotype	Genotypes
Brown Dun	AAEENN, AAEeNN, AaEENN, AaEeNN
Grey	aaEENN, aaEeNN, aaEECrN, aaEeCrN
Red Dun	AAeeNN, AaeeNN, aaeeNN
White Dun	AAEECrN, AAEeCrN, AaEECrN, AaEeCrN
Yellow Dun	AAeeCrN, AaeeCrN, aaeeCrN
Kvit	AAEECrCr, AAEeCrCr, AaEECrCr, AaEeCrCr, aaEECrCr, aaEeCrCr, AAeeCrCr, AaeeCrCr, aaeeCrCr



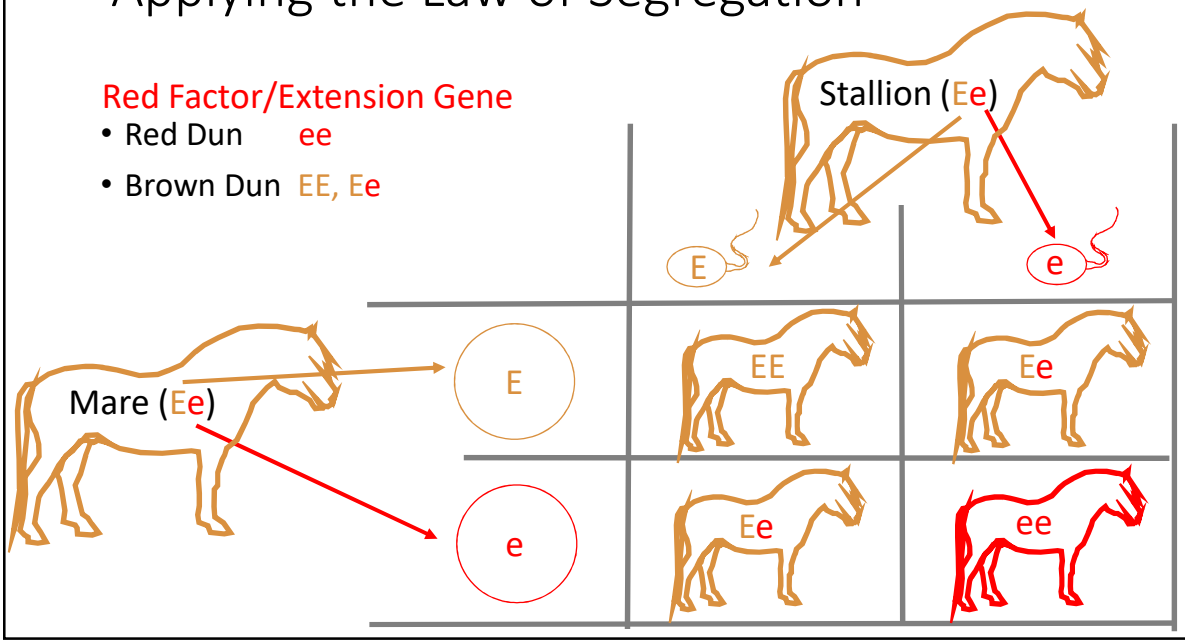
Law of Segregation

- During the formation of gametes (eggs or sperm), the two alleles responsible for a trait separate from each other.
  - Sire: Aa, produces 'A' semen and 'a' semen, at a 50-50 ratio A:a
  - Dam 1: aa, produces 100% 'a' eggs
  - Dam 2: AA, produces 100% 'A' eggs
  - Dam 3: Aa, produces 'A' eggs and 'a' eggs, at a 50-50 ratio A:a
- Alleles for a trait are then "recombined" at fertilization.
  - The genotype of the offspring depends on which semen fertilizes which egg
- Punnett Squares help predict likely color combinations of foals



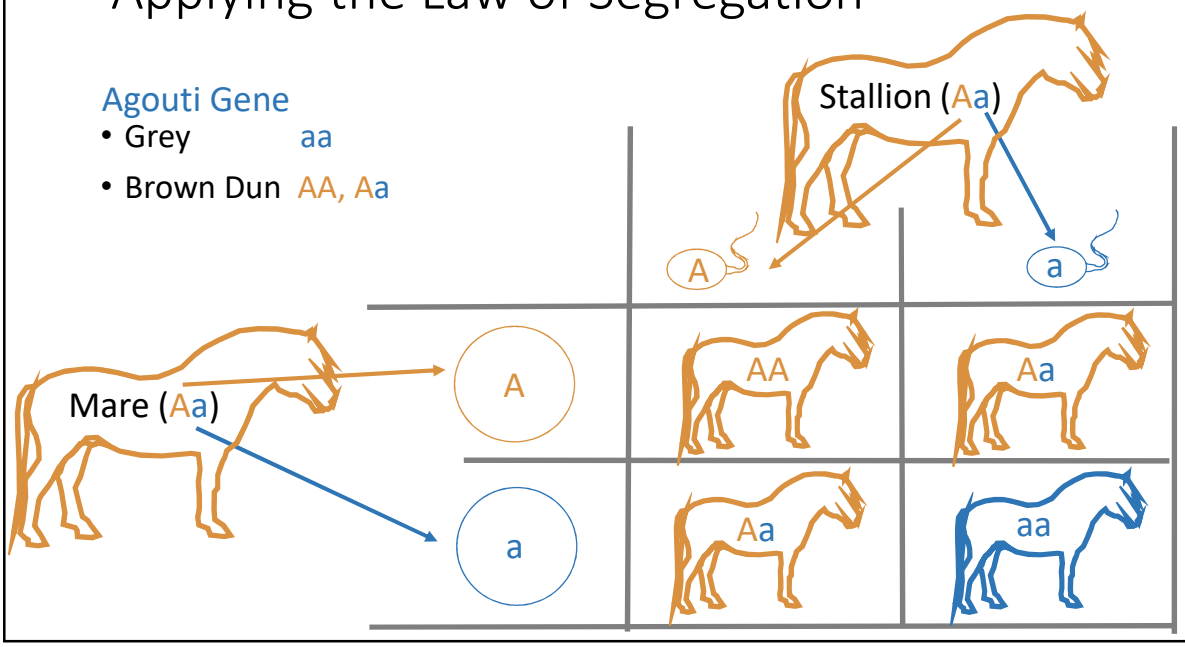
Applying the Law of Segregation

Red Factor/Extension Gene
- Red Dun ee
- Brown Dun EE, Ee



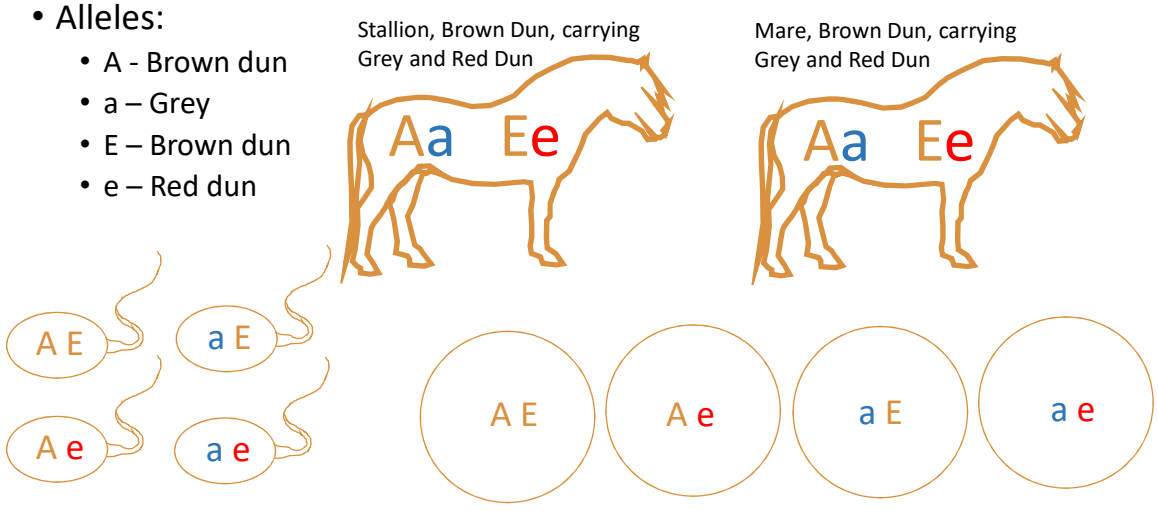
Applying the Law of Segregation

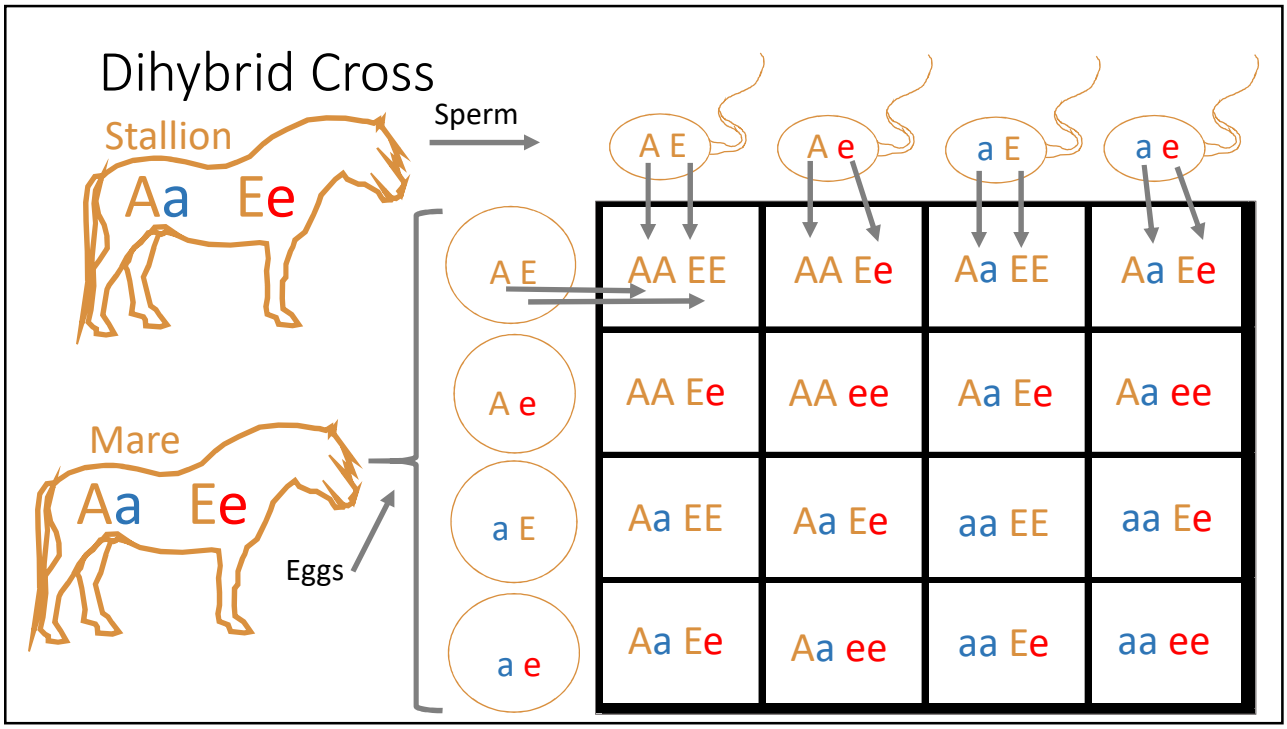
Agouti Gene
- Grey aa
- Brown Dun AA, Aa



Dihybrid Cross

- Traits: Agouti Gene & Red Factor Gene
- Alleles:
  - A - Brown dun
  - a – Grey
  - E – Brown dun
  - e – Red dun





Dihybrid Cross (Aa Ee x Aa Ee)
Parents carry Grey, Red

AA EE	AA Ee	Aa EE	Aa Ee
AA Ee	AA ee	Aa Ee	Aa ee
Aa EE	Aa Ee	aa EE	aa Ee
Aa Ee	Aa ee	aa Ee	aa ee

Phenotypes:

- Brown Dun
- Red Dun
- Grey

Count, %

- Brown Dun:
  - 9 of 16,
  - 56% of offspring
- Red Dun:
  - 4 of 16,
  - 25% of offspring
- Grey:
  - 3 of 16,
  - 19% of offspring

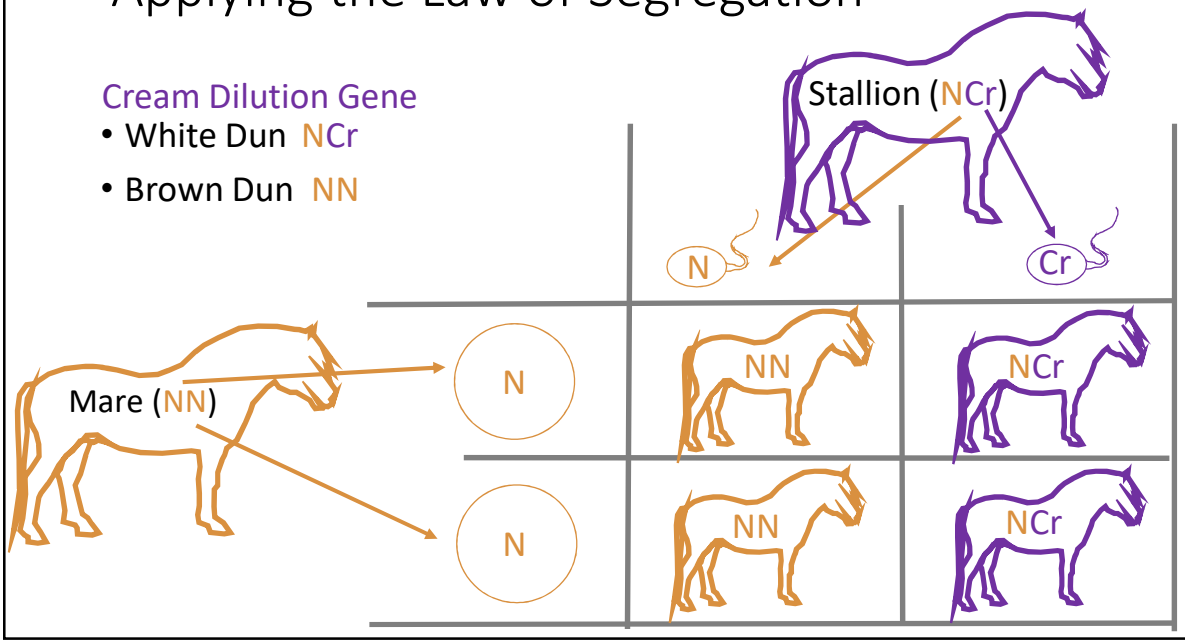
Incomplete Dominance

White dun stallion, Kjor Hunter, 2025

- Cream Dilution gene
- Two alleles: N (No dilution), Cr (Dilution)
- Not recessive/dominant
- Expressed in an additive manner, based on copy number
  - No copies (NN) no dilution of color
  - One copy (NCr) dilutes red-based color slightly, doesn't dilute black points
  - Two copies (CrCr) dilutes red based color to Kvit, dark points lightened
- Does not dilute black-based color (grey unaffected) w 1 copy
- 'Color' produced by just one parent passing along this gene
  - Recessive genes require a copy from each parent to produce rare color

Applying the Law of Segregation

Cream Dilution Gene
- White Dun NCr
- Brown Dun NN



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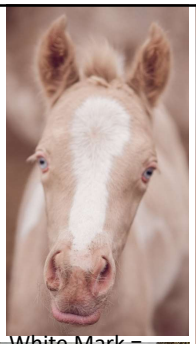
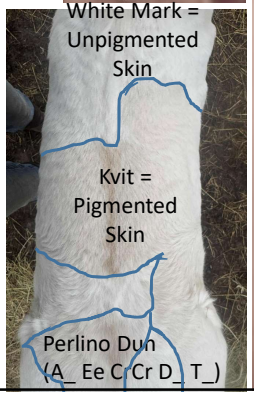
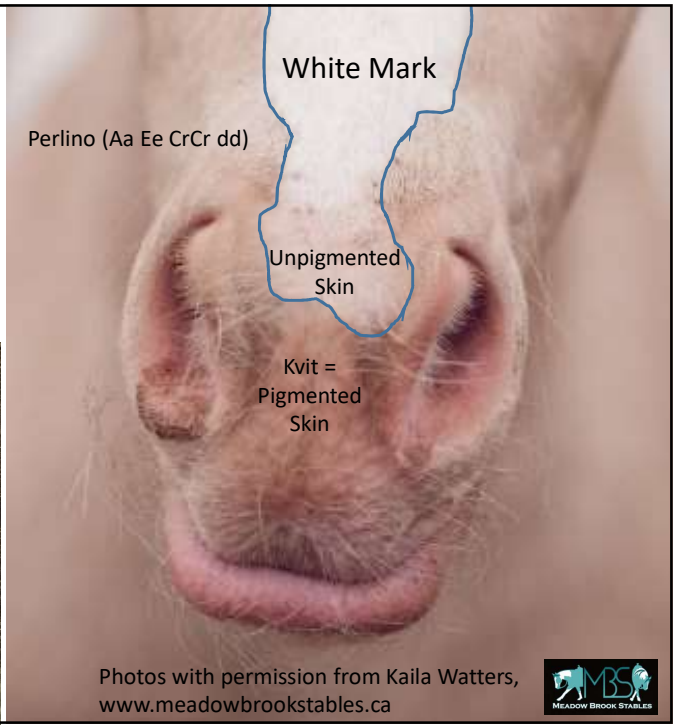
5 Colours of Fjords, photos by Marissa Orsel

Kvit colors

- Is 2 copies of Cream Dilution
  - 1 copy from each parent
- Is not albino
- Eyes stay blue
- Skin contains pigment
- Not associated with health problems
- In Norway:
  - Are registered
  - Stallion won't receive score high enough for breeding license
  - Kvit mares can be bred; no licensing required
  - 25% chance Kvit from parents with 1 copy
  - 50% chance of colt vs filly, so breeders rarely take the chance







Photos with permission from Kaila Watters, www.meadowbrookstables.ca

A- THE BASIS FOR BREEDING AND JUDGING FJORD HORSES

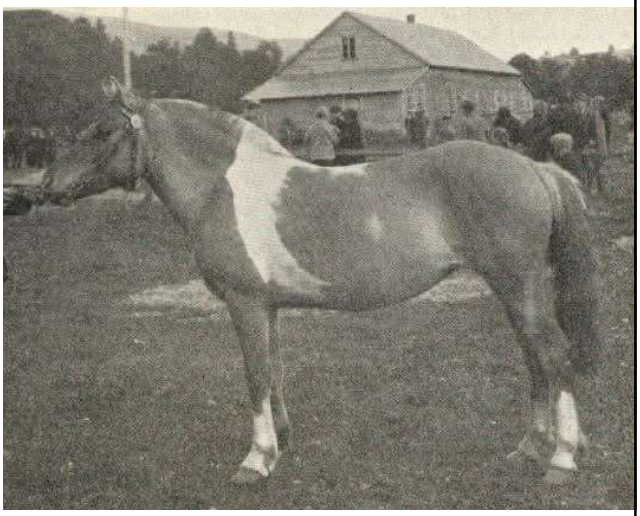
The History of Norwegian Fjord Horse Breeding and the The Beginning of Organized Breeding

A stallion and six mares of the "original Norwegian breed", the Fjord, were purchased by the state as a base for the stud at Hjerkinn.
Little was known about the genetics of colour. As in that time uls dun was very popular (statistic shows; between 1857 - 79 48,0% of the approved stallions were uls dun and between 1860 - 79 46,8% of the registered mares were uls dun) all this breeding stock was uls dun. Of the 32 foals born on the stud, 15 were white with blue eyes, showing up dramatically what happens by mating within this colour, but without anybody understanding why this happened. So because of this and the fact that the stud was poorly managed it was decided to close down the stud. *)

Fjord Horse International, 'The Red Book' Official Handbook for Fjord Horse Judges, May 3, 2003

White Marks

- W-series, KIT, PAX3 & MITF, EDNRB and/or SOX10 genes
- Multifactorial and polygenic (Woolf, 1990; Rieder et al., 2008)
- 34-35 genes identified now
- New discoveries annually, esp KIT
- Haplotypes
  - W22 is a haplotype containing W22 and W20
- Larger and more frequent w/red base colors
- Extensively marked individuals more likely to produce similar offspring
- Most recessive: impossible to select out, lacking a test
- Some dominant: have been selected out of Fjords
- Some embryonic lethal W22/W22, W13/W13
- Masking alleles possible
- Squamous cell cancer risk, no pigment
- Photosensitivity dermatitis only in white mark areas
- Heavy draft breeds have unique alleles, not described yet
- Fjords have unique alleles, not described yet
  - In Norway:
  - Stallions w/ white marks won't receive breeding license
  - Mares: smaller than 5 cm, no white marked hooves
  - Until tests are available, choose unmarked breeding stock
- Other breeds with Breed Standard of an Unmarked Horse



https://equusferus.photography/2025/01/18/the-inheritance-of-white-facial-and-extremity-markings/

Open Access Article

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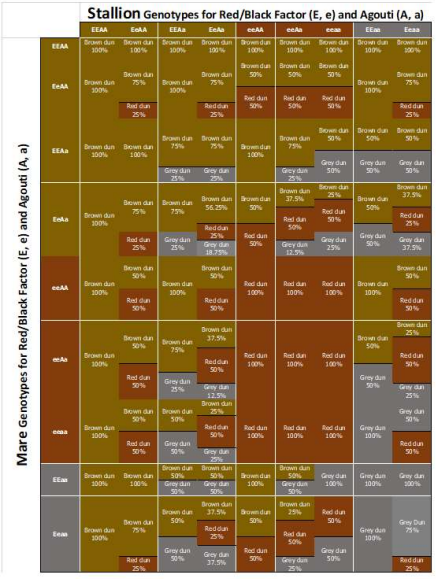
VGL researchers shed light on breed distribution and allele frequencies of equine coat color variants in new publication

Basic Assumptions

- Reduce need for testing
- Red dun parent (ee)
  - All foals inherit one copy of ‘e’
- Grey parent (aa)
  - All foals inherit one copy of ‘a’
- Parent with Cream Dilution (NCr)
  - 50% chance of foal inheriting ‘Cr’
  - 100% if one parent is CrCr
- Order Herald back-issue for an extensive list of ‘if-then’ statements regarding Fjord color



Fjord Color Inheritance and Testing Decision Guide, by Cherrie Nolden. The Fjord Herald, Issue 148, Fall 2023.

Foal Coats

- Born generally lighter than mature color
- Dorsal stripe becomes bolder with age
- Leg stripes pale at birth
- Njal marks present at birth
- Red base colors
  - Cream hooves at birth (not white)
  - Orangeish-purple eyelid color
  - No black hairs
- Grey foals
  - Grey eyeliner around eyes
  - Grey on muzzle is not reliable
- Cream diluted colors
  - Some very white highlights at birth
  - White marks are most visible at day 1-2
- Test, Don't Guess




Other Color Variations in Fjords

- Shade or intensity [1],
- Pangaré expression,
- Extent of dun expression [1],
- Somatic mutation, lines of blaschko, skewed dun,
- Changing hoof horn color in sections,
- Njal marks,
- Leg darkness,
- Splits in the dorsal stripe,
- White hairs in the dark dorsal stripe section,
- Low dorsal stripe expression on the back,
- Fading of dark points in sunlight,
- Appearance of white hairs years after birth with no known injury,
- White spots on the penis,
- Eye color from gold to dark brown [4].



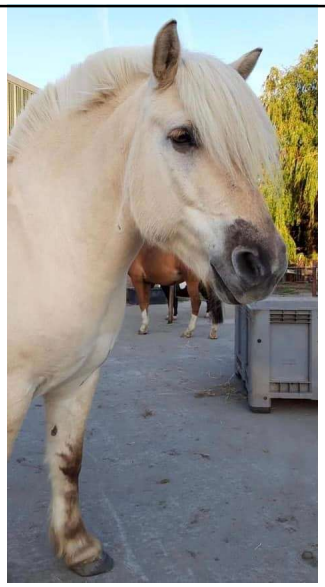



Amigo, Yellow dun

Veronica Schijven-Langenberg

- Dark muzzle, legs
- Eye rings like Grey
- Split dorsal

- Minimal Pangare



Business: 1dr Fjords **Email:** wonderfjords@yahoo.com

Date Received: 1/18/2020

Horse Name	Breed	Rfactor	Agouti	Cream
Amigo	Norwegian Fjord	ee	AA	nCr

Breed Responsibly

- Colors are Fun
- Fjords are SO much more than their color, and they should be!
  - Temperament
  - Structure
  - Movement
  - Genomic diversity
  - Breed standard
  - 3 generation no repeated parent rule for registration

References

- [1] Avila et al., 2022. Breed Distribution and Allele Frequencies of Base Coat Color, Dilution, and White Patterning Variants across 28 Horse Breeds. Genes 2022, 13(9), 1641; https://doi.org/10.3390/genes13091641. https://www.mdpi.com/2073-4425/13/9/1641/htm.
- [2] Lori Mclaughlin. South Steens Wild Horses, in southeast Oregon. "We actually have not seen any issues regarding crems / perls and light & heat. In fact, on this range they have been faring better than the pintos." https://www.facebook.com/story.php?story_fbid=pfbid02Aa7UH3TAeJSELCgKc8NA1bBxtjrGH5tPFMfDW1s9V46Xn2MWr2xn8R4gE8gEo1udl&id=100064763856396&mibextid=cr9u03
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Questions

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