Genetic Disease and Trait Definitions

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The pages below list information about the validated trait probes on the IDB chip in the following format.

Full Trait Name

Abbreviations: Abbreviations and alternative names for the trait

Royalty Fee: If this trait is free in Ireland or if a Royalty fee is required.

For traits that require a Royalty fee, please contact Weatherbys Ireland for cost and reporting

Genetic Mode: If the trait is recessive, dominant, or additive

Trait Type: If the trait is Lethal, Unwanted, Beneficial, Milk, Muscle, or Coat Colour related

Breeds found in: Breed lineages this trait is known to occur in. Breed specific alleles will be in parentheses.

General: A general description of the trait

Common Ancestor: If carriers of the trait can be traced back to a common ancestor(s)

Image: If available an image of an animal with the trait will be provided

More in depth information on each trait, including clinical, gene, genomic, sequence, and reference information can be found in the Full Disease Mutation Definition document at ICBF.com

Traits are grouped by the following:

1) Lethal or Unwanted: Alleles that either result in mortality or have a negative economic effect

2) Beneficial: Alleles that are economically beneficial

3) Colour: Alleles that affect an animal's coat colour

4) Meat: Alleles that affect meat or muscle quality or quantity

5) Milk: Alleles that affect the quantity of milk produced or the milk components

Traits on the IDB

LETHAL

1. Alpha Mannosidosis
2. Beta Mannosidosis
3. Brachyspina
4. Bulldog Dwarfism
5. Citrullinaemia
6. Congenital Muscular Dystonia 1
7. Congenital Muscular Dystonia 2
8. Complex Vertebral Malformation
9. Deficiency of Uridine Monophosphate Synthase
10. Holstein Haplotype 1
11. Holstein Haplotype 3
12. Holstein Haplotype 4
13. Idiopathic Epilepsy
14. Jersey Haplotype 1
15. Maple Syrup Urine
16. Montbeliarde Haplotype 2
17. Neuropathic Hydrocephalus
18. Osteopetrosis
19. Paunch Calf Syndrome
20. Spinal Muscular Atrophy
21. Tibial Hemimelia Improver

UNWANTED

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2. Congenital Myoclonus
3. Crooked Tail Syndrome
4. Dystrophic Epidermolysis Bullosa
5. Hypotrichosis KRT71
6. Mulefoot
7. Protoporphyria
8. Pseudomyotonia
9. Rat-tail Syndrome
10. RNF11 Growth Retardation
11. STAT1
12. STAT5A

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1. Infectious Bovine Keratoconjunctivitis (Pinkeye)
2. Poll

MEAT

1. Calpain1
2. Calpastatin
3. Myostatin

MILK

1. ABCG2
2. AcylCoA:Diacylglycerol Acyltransferase
3. Growth Hormone
4. Growth Hormone Receptor
5. Casein Beta
6. Casein Kappa
7. Lactoglobulin Beta

COLOUR

1. Dun
2. MC1R
3. PMEL17

LETHAL

Alpha Mannosidosis

Abbreviations: AM 662, AM 967

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Angus (AM_961), Murray Grey (AM_961), Galloway (AM_662)

General: Affected calves are either aborted, born dead, die soon after birth, or die within the first year. Those born alive can show signs of ataxia, head tremor, aggression, and paralysis before death.

Common Ancestor: None identified

Beta Mannosidosis

Abbreviations: BM

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Salers

General: Affected calves are unable to rise with intention tremors, hidebound skin, slightly domed skull, slight underbite or overbite, and narrow eye slits. Calves born with this disorder do not get up after birth and soon die.

Common Ancestor: None identified

Brachyspina

Abbreviations: BY

Genetic Mode: Recessive

Royalty Fee: YES

Trait Type: Lethal

Breeds found in: Holstein-Friesian

General: Affected calves are either aborted in the first 40 days of gestation or stillborn. Stillborn calves are born after a prolonged gestation with reduced body weight, a short neck and body, a hump between the shoulder blades and a deformed lower jaw.

Common Ancestor: Sweet Haven Tradition, Bis-May Tradition Cleitus, Rothrock Tradition Leadman

Brachyspina affected calf. Image from Agerholm et al., 2006

Bulldog Dwarfism

Abbreviations: BD1, BD2, Dexter Chondrodysplasia

Genetic Mode: Recessive

Trait Type: Lethal

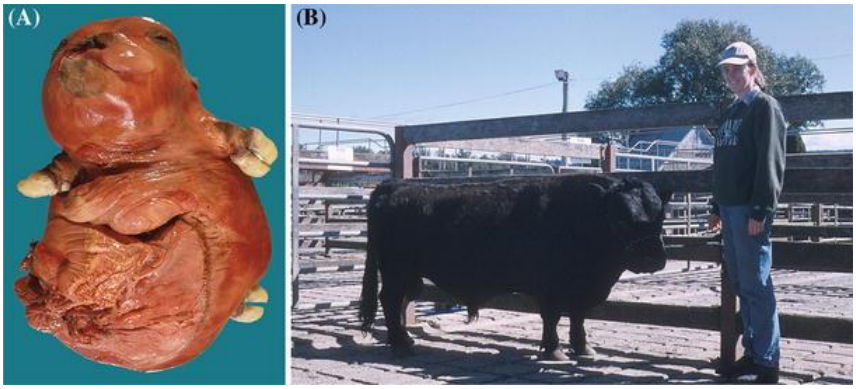
Royalty Fee: No

Breeds found in: Dexter

General: This disease is caused by one of two mutations (BD1 and BD2) in the ACAN gene. Affected animals display extreme dwarfism, die around the seventh month of gestation, and are aborted. Heterozygous animals are born alive and live but have a mild form of dwarfism.

Being homozygous for either mutation or heterozygous for both will cause bulldog dwarfism.

Common Ancestor: None identified



A) BD affected embryo, B) Heterozygous animal exhibiting dwarfism. Images from Cavangh et al., 2007

Citrullinaemia

Abbreviations: CT **Genetic Mode: Recessive**

Royalty Fee: No **Trait Type: Lethal**

Breeds found in: Holstein-Friesian

General: Affected calves are born normal and become depressed within 24 hours. In 3-5 days they develop tongue protrusion, unsteady gait, wander aimlessly, froth at the mouth, will press their head against something solid, develop convulsions, and die.

Common Ancestor: None identified

Congenital Muscular Dystonia 1

Abbreviations: CMD1 **Genetic Mode: Recessive**

Royalty Fee: No **Trait Type: Lethal**

Breeds found in: Belgian Blue, Dutch Improved Red and White

General: Affected calves have episodes of generalized muscle contractures, impaired swallowing, and falling. CMD1 calves usually die within a few weeks as a result of respiratory complications.

Common Ancestor: None identified

Congenital Muscular Dystonia 2

Abbreviations: CMD2, Startle Disease **Genetic Mode: Recessive**

Royalty Fee: No **Trait Type: Lethal**

Breeds found in: Belgian Blue

General: Affected calves show episodes of generalized muscle contractures and sever muscle twitching. Affected calves typically die within a few hours to days after birth.

Common Ancestor: None identified

Image of a CMD2 affected calf. Image from Harvey et al., 2008.

Complex Vertebral Malformation

Abbreviations: CVM

Genetic Mode: Recessive

Royalty Fee: YES

Trait Type: Lethal

Breeds found in: Holstein-Friesian

General: Affected calves are usually aborted during gestation, some are born alive but die soon after. Animals have a shortened neck and curved spine, they can have abnormal ribs, contracted joints, and contracted and rotated fetlocks.

Common Ancestor: Carlin-M Ivanhoe Bell and Pennstate Ivanhoe

CVM affected calf. Image from Thomsen et al., 2006

Deficiency of Uridine Monophosphate Synthase

Abbreviations: DUMPS

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Holstein, Friesian, Wagyu

General: Affected calves are aborted around day 40 of pregnancy. The affected embryos often are resorbed during the first two-month of gestation, leading to more services per calving and longer than normal calving intervals.

Common Ancestor: None identified

Holstein Haplotype 1

Abbreviations: HH1

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Holstein-Friesian

General: Affected calves are aborted during in pregnancy.

Common Ancestor: Pawnee Farm Arlinda Chief

Holstein Haplotype 3

Abbreviations: HH3

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Holstein-Friesian

General: Affected calves homozygous are aborted before day 60 of gestation.

Common Ancestor: Glendell Arlinda Chief, Gray View Skyliner, Oman

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Holstein Haplotype 4

Abbreviations: HH4

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Holstein-Friesian

General: Affected calves are aborted early in pregnancy.

Common Ancestor: Besne Buck

Idiopathic Epilepsy

Abbreviations: IE

Genetic Mode: Recessive

Royalty Fee: YES

Trait Type: Lethal

Breeds found in: Hereford, Simmental

General: Affected calves are born normal and have no outward appearance of the disorder until they start having seizures. The age of onset of the initial seizures can occur from birth up to several months of age. When seizing the animal will lay on its side with legs straight out. Episodes may last from several minutes to more than an hour.

Common Ancestor: None identified

Idiopathic Epilepsy affected cow appear normal unless having a seizure. Image from Kaiser 2010

Jersey Haplotype 1

Abbreviations: JH1

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Jersey

General: Calves homozygous for the mutation are aborted before day 60.

Common Ancestor: Observer Chocolate Soldier

Maple Syrup Urine

Abbreviations: MSU_SH

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Shorthorn

General: Some affected calves are stillborn, those born alive look normal but exhibit mental disorders within 1 day. Their condition will rapidly deteriorate with ataxia, sweet smelling urine, an inability to walk, and death within 96 hours after birth.

Common Ancestor: None identified

Montbeliarde Haplotype 2

Abbreviations: MH2

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Montbeliarde

General: Affected animals are aborted early in gestation.

Common Ancestor: None identified

Neuropathic Hydrocephalus

Abbreviations: NH, Water Head

Genetic Mode: Recessive

Royalty Fee: YES

Trait Type: Lethal

Breeds found in: Angus

General: Affected calves may be stillborn or die soon after birth. Those born alive might have an enlarged head, they will likely show depression, weakness, poor suckle reflex, droopy ears and head, head tremors, and convulsions.

Common Ancestor: GAR Precision 1680

Neuropathic hydrocephalus affected calf. Image from Kaiser 2010

Osteopetrosis

Abbreviations: OS, Marble Bone Disease

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Angus

General: Affected calves are typically stillborn prematurely (250-275 days of gestation). They often have a small body size, flat skull, impacted molars, shortened lower jaw, protruding tongue, the leg bones are easily broken.

Common Ancestor: None identified

Head of Osteopetrisis affected calf. Image from Meyers et al., 2010

Paunch Calf Syndrome

Abbreviations: PCS

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Romagnola

General: Affected calves are usually stillborn, have abnormal development of multiple organs; facial deformities; and an enlarged distended fluid-filled stomach (hence the name 'Paunch Calf'). Some affected calves also have a protruding tongue and cleft palate.

Common Ancestor: None identified

Affected Paunch Calf Syndrome calf. Image from Toolan et al., 2014

Spinal Muscular Atrophy

Abbreviations: SMA

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Lethal

Breeds found in: Brown Swiss

General: Calves often die of pneumonia by six to eight weeks of age. While born normal SMA affected calves start to show symptoms between three and six weeks of age when they show loss of strength and balance in the rear legs. As the disease progresses they will become weaker, lose flesh, and lose balance in the front legs. Once they show signs of laboured breathing death usually occurs within a couple of days. Usually the cause of death is pneumonia by six to eight weeks of age

Common Ancestor: None identified

Images of SMA affected calves from Brown Swiss Association 2001

Tibial Hemimelia Improver

Abbreviations: TH-Improver

Genetic Mode: Recessive

Royalty Fee: YES

Trait Type: Lethal

Breeds found in: Galloway, Shorthorn

General: Affected animals are born with severe deformities including twisted rear legs with fused joints, large abdominal hernias and/or skull deformities. Affected calves are born dead or die (or are euthanized) shortly after birth.

Common Ancestor: Deerpark Improver

Tibial Hemimelia affected calf. Image from Kaiser 2010

UNWANTED

Bovine Leukocyte Adhesion Deficiency

Abbreviations: BLAD

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Unwanted

Breeds found in: Holstein-Friesian

General: Affected cattle often have severe ulcers in the mouth, tooth loss, chronic pneumonia, and diarrhoea. Affected cattle often die at a young age due to infections.

Common Ancestor: Osborndale Ivanhoe

Congenital Myoclonus

Abbreviations: CM

Genetic Mode: Recessive

Royalty Fee: No

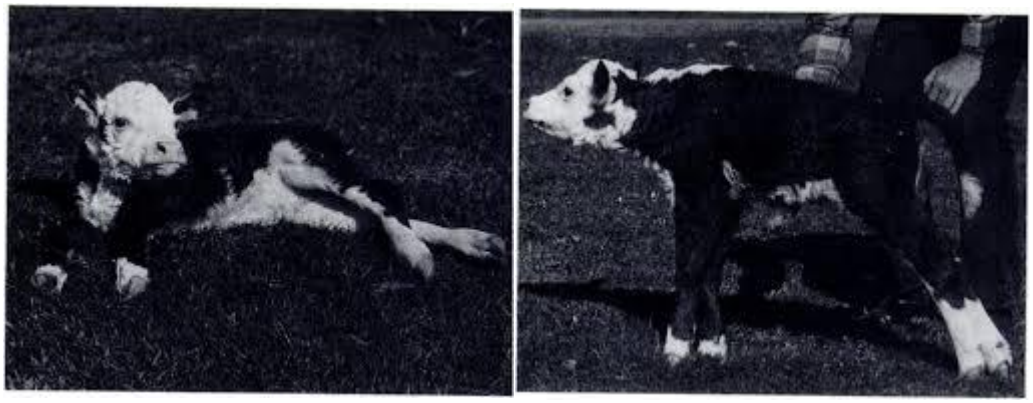
Trait Type: Unwanted

Breeds found in: Hereford

General: Affected animals often appear normal but have spontaneous muscle spasms and whole body rigidity in response to stimulation. When laying down the back legs are often crossed. When assisted to a standing position the handlers touch can cause full body rigidity and a sawhorse position.

While not lethal, affected calves are usually humanly euthanized.

Common Ancestor: None identified



Affected calf with crossed limbs (left) and sawhorse posture (right). Images from Gundlach, A.L, 1990.

Crooked Tail Syndrome

Abbreviations: CTS_AG, CTS_T>C

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Unwanted

Breeds found in: Belgian Blue

General:. There are two mutations that cause the disease: CTS_AG and CTS_T>C. Being homozygous for either mutation or heterozygous for both will cause the disease.

It is not lethal but >25% of affected animals are euthanized on welfare grounds. It causes substantial economic losses due to growth retardation and treatment. Affected animals have a crooked tail, abnormally shaped legs, stocky head, growth retardation, extreme muscularity, and straight hocks.

Heterozygous animals have enhanced muscular development, and are smaller, stockier, and toed-in front legs.

Common Ancestor: None identified

Images of affected CTS animals.
Images from Fasquelle et al., 2009.

Dystrophic Epidermolysis Bullosa

Abbreviations: DEB

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Unwanted

Breeds found in: Rotes Hohenvieh

General: The skin and mucus membranes of affected animals are very fragile making it easy to rip or tear, especially around the muzzle, mouth, fetlocks, and hooves. Some demonstrate a large loss of skin or blisters around the fetlocks and on the muzzle. While not fatal, affected animals are usually humanely euthanized due to the extent of the skin lesions.

Common Ancestor: None identified

Images of lesions found on a DEB affected. Images from Menoud et al., 2012

Hypotrichosis KRT71

Abbreviations: HY_KRT71

Genetic Mode: Recessive

Royalty Fee: YES

Trait Type: Unwanted

Breeds found in: Hereford

General: Affected cattle have partial absence of hair at birth over all or parts of the body: often on the poll, brisket, neck and legs. The hair can be very short, fine, or kinky that may fall out leaving bare spots, and the tail switch can be underdeveloped. Affected animals are more vulnerable to environmental stress, skin infections, pests, sunburn, cold stress, and have a decreased economic value.

Common Ancestor: None identified

Calf with hypotrichosis affected legs. (Photo kindly provided by Dr. Johnathan Beever, University of Illinois)

Mulefoot

Abbreviations: Syndactyly, MF_R1740X, MF_P1647L, MF_NG1621KC, MF_G1199S, MF_G907R, MF_G81S

Royalty Fee: No

Genetic Mode: Recessive

Trait Type: Unwanted

Breeds found in: Angus (MF_R1740X), Charolais, Holstein (MF_NG1621KC), Simmental (MF_G907R, MF_G81S), Simmental x Charolais x Holstein crossbred (MF_G1199S)

General: Also called Syndactyly which means "joined finger, the cloven hoof is fused together. Affected cattle can have 1-4 fused hooves, show varying degrees of lameness, have a high-step gait, and may walk slowly.

Common Ancestor: None identified

Photo of an animal affected by Mulefoot. Image from Duchesne et al., 2006

Protoporphyria

Abbreviations: Proto

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Unwanted

Breeds found in: Limousin, Blond de'Aquitaine

General: Protoporphyria causes extreme photosensitivity. Affected animals have hair loss and ulcers develop on skin exposed to sunlight, especially the ears, lips, nose and udder. Soon after birth affected animals often lick their lips and nose due to the pain/itchiness of developing ulcers. Affected animals are very reluctant to leave shade. Their teeth, bones, and urine can also have a reddish brown discoloration.

While not lethal affected animals often fail to thrive and are sold to slaughter before reaching optimal slaughter weight.

Common Ancestor: None identified

Examples of skin ulceration on a Protoporphyria affected calf. Image from McAloon et al., 2015

Pseudomyotonia

Abbreviations: PMT_164, PMT_211, PMT_284

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Unwanted

Breeds found in: Chininia (PMT_164), Romagnola (PMT_211, PMT_284)

General: Affected animals are characterized by having muscle contractions when startled or move faster than a slow walk. When contractions occur the animals will have an uncoordinated gait, sometimes 'bunny hopping' on their back feet. a lifelong history of exercise-induced (more intense than a walk) muscle contractions. Under prolonged stimulation the muscles become so stiff the animals can fall over. The contractions stop once the stimulation is removed and they are able to move normally again.

Common Ancestor: None identified

Pseudomyotonia affected animal. Image from Drogemuller et al., 2008

Rat-tail Syndrome

Abbreviations: PMEL17_50_52delTTC, PMEL17_3del

Genetic Mode: Semi-Dominant

Trait Type: Unwanted

Royalty Fee: No

Breeds found in: Crossbred: Red Colour x Black Colour. Often 1 Simmental parent

General: Rat-Tail refers to a phenotype that has is deficient of hair on the tail switch and other parts of the body has short, curled, and crimped hair. Rat-tail animals have lower average daily gain in the winter months. This trait can occur from the mating of a black or black pied parent with a red coloured parent, particularly Simmental, when the red coloured parent carries the PMEL17_50_52delTTC allele.

If a calf from such a cross inherits **only 1** PMEL17_50_52delTTC allele it will be rat-tailed. If the animal is homozygous for the PMEL17 3bp deletion then it will be light grey coloured and not rat-tailed. If it inherits no PMEL17_3del alleles it will be black and not rat-tailed.

Common Ancestor: None identified

Left is a heterozygous PMEL17_3del calf with Rat-Tail. Centre is a homozygous PMEL17_3del calf. Right is a normal (no PMEL17_3del alleles) calf. Images from HECHT, B. C. 2006

RNF11 Growth Retardation

Abbreviations: RNF11

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Unwanted

Breeds found in: Belgian Blue

General: Affected animals appear normal at birth but suffer from severely stunted growth at 6 months, they have a narrow skull and very hairy head. Approximate one-third of affected animals will die from infections before 6 months of age due to a compromised resistance to pathogens.

Common Ancestor: Galopeur des Hayons

RNF11 affected (front) and normal (back) calf of the same age. Image from Sartelet et al., 2012

STAT1

Abbreviations: STAT1

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Unwanted

Breeds found in: Multiple Breeds

General: Animals homozygous for the mutation have a decreased embryo survival rate. Animals born alive and heterozygous or homozygous will appear normal.

Common Ancestor: None identified

STAT5A_13319

Abbreviations: STAT5A_13319

Genetic Mode: Recessive

Royalty Fee: No

Trait Type: Unwanted

Breeds found in: Multiple Breeds

General: Animals homozygous for the mutation have a decreased embryo survival rate. Animals born alive and heterozygous or homozygous will appear normal.

Common Ancestor: None identified

BENEFICIAL

Infectious Bovine Keratoconjunctivitis

Abbreviations: IBK, Pinkeye

Genetic Mode: Additive

Royalty Fee: No

Trait Type: Beneficial

Breeds found in: Multiple Breeds

General: Pinkeye, also called Infectious Bovine Keratoconjunctivitis, is primarily caused by the bacterium Moraxella bovis. With each copy of this allele the animal reduces its risk of pinkeye infection by 8-13%. Thus an animal that is homozygous for the allele will have a 16-26% reduction in pinkeye infection risk. Pinkeye can cause a decrease in weight gain.

Common Ancestor: None identified

Left to Right: Examples of Stage 1, 2, 3, and 4 Pinkeye infections. Images from Whitter et al., 2009

Polled

Abbreviations: Poll_C

Genetic Mode: Dominant

Royalty Fee: No

Trait Type: Beneficial

Breeds found in: Multiple breeds including Augus, Galloway, Speckle Park, Murray Grey, Senepol, and Holstein

General: The poll allele causes animals to have an absence of horns. Besides the lack of horns, genetically polled animals also have a narrower skull, especially noticeable at the poll. Horned and dehorned cattle typically have a flat-looking poll, while genetically polled cattle have more peaked-looking poll. The Poll_C allele is found in animals with Nordic and British lineages.

Common Ancestor: None identified

Angus with polled versus Aubrac with horn phenotype. (Photos from ICBF)

MEAT

Calpain 1

Abbreviations: CAPN1_316, CAPN1_4751, CAPN1_530

Royalty Fee: No

Genetic Mode: Additive

Trait Type: Meat

Breeds found in: Multiple breeds

General: The Calpain 1 gene has alleles (listed below) associated with more tender meat.

CAPN1_316: The 'C' allele

CAPN1_530: The 'G' allele

CAPN1_4751: The 'C' allele

Common Ancestor: None identified

Calpastain

Abbreviations: CAST_282, CAST_2870, CAST_2959

Trait Type: Meat

Royalty Fee: No

Breeds found in: Multiple breeds

Genetic Mode: Additive

General: The Calpastain gene has alleles (listed below) associated with more tender meat.

CAST_282: The 'C' allele

CAST_2870: The 'G' allele

CAST_2959: The 'A' allele

Common Ancestor: None identified

Myostatin

Abbreviations: See below **Genetic Mode: Recessive**

Royalty Fee: YES **Trait Type: Meat**

Breeds found in: Multiple, breed specific mutations listed below

General:. Multiple alleles in the Myostain gene increase muscle mass and some cause double muscling. The effect on calving difficulty, birth weight, and if it causes double muscling depends on the allele.

MYO_821del11: Found in Asturiana, Belgian Blue, Blonde d' Aquitaine, Limousine, Parthenise, Asturiana, South Devon, Santa Gertrudis, Braford, Murray Grey, and Angus lineages. Results in double muscling (hyperplasia), larger birth weights, increased dystocia and meat tenderness

MYO_C313Y: Found in Gasconne, Piedmontese and Parthenise lineages. Results in double muscling (hyperplasia), larger birth weights, increased dystocia and meat tenderness

MYO_E226X: Found in Marchigiana and Maine-Anjou lineages. Results in double muscling (hyperplasia), larger birth weights, increased dystocia and meat tenderness

MYO_E291X: Found in Maine-Anjou and Marchingina lineages. Results in double muscling (hyperplasia), larger birth weights, increased dystocia and meat tenderness

MYO_F94L: Found in Angus and Limousin lineages. Results in increased muscularity and reduced external and intramuscular fat, with no change in birth weight

MYO_Q204X: Found in Blonde d'Aquitaine, Charolaise, Charolais and Limousin lineages. Results in double muscling (hyperplasia), larger birth weights, increased dystocia and meat tenderness

MYO_S105C: Found in Parthenaise lineages. Results in increased muscularity and reduced external and intramuscular fat, with no change in birth weight

Common Ancestor: None identified

Homozygous MYO_nt821 BelgianBlue (left) and homozygous MYO_F94L Limousine (right). Images from ICBF

MILK

ATP-Binding Cassette, Sub-Family G, Member 2

Abbreviations: ABCG2 **Genetic Mode: Additive**

Royalty Fee: No **Trait Type: Milk**

Breeds found in: Holstein, Friesian, Jersey, Brown Swiss, Simmental, and multiple beef breeds

General: Increases milk fat (kg and %), protein (kg and %), and decreases milk volume.

Common Ancestor: None identified

AcylCoA:Diacylglycerol Acyltransferase

Abbreviations: DGAT1 **Genetic Mode: Additive**

Royalty Fee: No **Trait Type: Milk**

Breeds found in: Holstein, Friesian, Jersey, Brown Swiss, Simmental, and multiple beef breeds

General: Increases fat yield, fat percentage, and protein percentage, while reducing milk yield and protein yield

Common Ancestor: None identified

Growth Hormone

Abbreviations: GH_2141, GH_2291, **Genetic Mode: Recessive**

Royalty Fee: No **Trait Type: Milk**

Breeds found in: Holstein, Friesian, Jersey, Brown Swiss, Simmental, and multiple beef breeds

General: Two alleles in the Growth Hormone gene have an effect on milk traits.

GH_2141: 'G' allele is associated with decreased milk protein yield and fat yield.

GH_2291: 'C' allele is associated with increased milk fat yield, fat percent, and protein percent.

Common Ancestor: None identified

Growth Hormone Receptor

Abbreviations: GHR_F279Y

Genetic Mode: Additive

Royalty Fee: YES

Trait Type: Milk

Breeds found in: Holstein, Friesian, Ayshire, Jersey, Brown Swiss, Simmental, and multiple beef breeds

General: Increases milk, casein, and lactose yield and a decrease in protein yield and in fat yield.

Common Ancestor: None identified

Casein Beta

Abbreviations: CSN2_A1, A2, A3, B, C, D, E, F, G, H1, H2, I

Genetic Mode: Additive

Trait Type: Milk

Royalty Fee: A2 YES, others No

Breeds found in: Multiple dairy and beef breeds

General: Approximately 25-30% of cow's milk is beta-casein (ß-casein). There are several alleles of ß-casein, the most common of which are A1 and A2 – other types include A3, B, C, D, E, F, G, H1, H2, and I are rarer. The A1 allele is associated with increased fat percent and protein percent. The A2 allele has a positive impact milk yield and protein yield and there are some theories that A2 milk is heathier than A1 milk. The B allele is more favourable for rennet coagulation and cheese making. Casein Beta does have an interaction effect with Casein Kappa. For coagulation time and curd firmness having one 'B' allele for each gene produces the best result

Common Ancestor: None identified

Casein Kappa

Abbreviations: CSN3_A, A1, B, B2, C, D, E, F1, F2, G1, G2, H, I , J

Genetic Mode: Additive

Trait Type: Milk

Royalty Fee: No

Breeds found in: Holstein, Friesian, Jersey, Brown Swiss, Simmental, and multiple beef breeds

General: The 'B' allele has a positive effect on coagulation time and cheese yield due to a firmer curd production. The 'G' and 'E' alleles are associated with less favourable coagulation properties. Casein Kappa does have an interaction effect with Casein Beta, for coagulation time and curd firmness having one 'B' allele for each gene produces the best result.

Common Ancestor: None identified

Beta-Lactoglobulin

Abbreviations: LBG_ A, B, C, D, H, I, J, W, -215C>A **Genetic Mode: Additive**

Royalty Fee: No **Trait Type: Milk**

Breeds found in: Holstein, Friesian, Jersey, Brown Swiss, Simmental, and multiple beef breeds

General: Lactoglobulin Beta is the major milk whey protein in cattle and has 8 alleles: A, B, C, D, H, I, J, and W. The 'B' allele is the ancestral allele, other alleles and their corresponding SNPs at various positions within the LGB gene are listed below. The 'B' allele is more favourable for rennet coagulation and the cheese making quality of milk

The -215C>A is associated with lower LGB content in milk which results in lower whey protein percent and casein number percent.

Common Ancestor: None identified

COLOUR

Dun

Abbreviations: DUN

Genetic Mode: Recessive and multi-gene interaction

Royalty Fee: No

Trait Type: Colour

Breeds found in: Dexter

General: The Dun coat colour allele (b) causes dilution of black hair pigment (eumelanin). The resulting hair colour is diluted to shades of dark brown to golden. Red hair pigment (phaeomelanin) is not diluted by this allele. There is an interaction with the MC1R gene as shown below.

Common Ancestor: None identified

MC1R	DUN	Colour
EE	BB	**Black**
EE	Bb	**Black**
EE	bb	Dun
Ee	BB	**Black**
Ee	Bb	**Black**
Ee	bb	Dun
EE+	BB	**Black**
EE+	Bb	**Black**
EE+	bb	Dun
E+E+	BB	Usually Red
E+E+	Bb	Usually Red
E+E+	bb	Usually Red
E+e	BB	Red
E+e	Bb	Red
E+e	bb	Red
ee	BB	Red
ee	Bb	Red
ee	bb	Red

Interaction between the MC1R and DUN alleles and their effect on coat colour

MC1R

Abbreviations: MC1R_Ed, Ebr, E+ , e **Genetic Mode: Recessive**

Royalty Fee: No **Trait Type: Colour**

Breeds found in: Multiple breeds including Angus, Brown Swiss, Holstein, Highland, Jersey

General: The four alleles of the MC1R gene are dominant black (MC1R_Ed), Black/Red (MC1R_Ebr), ancestral red (MC1R_E+) and recessive red (MC1R_e). Dominant black (Ed) is dominant to the other three alleles and animals with Ed are black and white. Black/Red, also known as Telstar, (Ebr) results in red colour at birth which changes to black at a young age. E+E+ cattle can be almost any colour since other genes take over dictating what coat colour pigments are produced. Two copies of the recessive red (e) allele result in red colour. The order of dominance is Ed>Ebr>E+>e.

Common Ancestor: None identified

PMEL17_50_52delTTC

Abbreviations: PMEL17_50_52delTTC, PMEL17_3del, Dilutor 3, Silver Char Dilutor 2,

Genetic Mode: Semi-Dominant

Trait Type: Colour and multi-gene interactions

Royalty Fee: No

Breeds found in: Multiple breeds including Simmental, Highland, Galloway, Hereford, Charolais

General: The PMEL17_50_52delTTC allele causes dilution coat colours such as dun, silver dun, yellow, and cream based on an interaction with the MC1R gene. The resulting colour from the PMEL and MC1R interaction is listed below.

Common Ancestor: None identified

C

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Coat colour	*MC1R*	*PMEL*	*Photo*
Red	E^+/e	+/+	TR
	e/e	+/+	
Yellow	E^+/e	+/del	MR
	e/e	+/del	
White/cream	*e/e*	del/del	BR
	E^+/e	del/del	
Black	E^D/E^D	+/+	TL
	E^D/E^+	+/+	
	E^D/e	+/+	
Dun	E^D/E^D	+/del	ML
	E^D/E^+	+/del	
	E^D/e	+/del	
Silver dun	E^D/E^+	del/del	BL
	E^D/e	del/del	

Photographs, MC1R and PMEL17_50_52delTTC genotypes of Highland cattle exhibiting distinct coat colours. The wild type allele is designated by '+'. Photo location: T=top, M=middle, B=bottom, L=left, R=right. Table and photos adapted from Schmutz & Dreger 2013

PMEL17_64G_A

Abbreviations: PMEL17_64G_A , SD1, Silver Char Dilutor 1

Genetic Mode: Additive

Trait Type: Colour

Royalty Fee: No

Breeds found in: Charolais

General: This allele causes coat colour dilution. Animals that are homozygous 'A' for the PMEL17_64G>A allele are white coloured while heterozygous animals are an intermediate colour: light grey, dark grey, light red, or dark red, brown, or yellow depending on the animal's base coat colour.

Common Ancestor:

Examples of coat colour dilutions from PMEL17_64G>A. Image from Gutierre-Gil et al., 2007.