## **Genetic traits in the Nordic countries – Dairy cattle**

## Holstein - Bovine Leukocyte Adhesion Deficiency (BLAD)

(BLF = Not carrier, BLC = Single carrier, BLS = Double carrier)

BLAD is an autosomal recessively inherited disease in the Holstein population. It is characterized by recurrent bacterial infections, delayed wound healing and stunted growth. Calves with BLAD are more susceptible to infections and their lifespan varies, many die shortly after birth. Osborndale Ivanhoe (US 1189870) has been identified as the key ancestor of the disorder. Several widely used sires after Osborndale, eg. Penstate Ivanhoe Star (US 1441440) and Carlin-M Ivanhoe Bell (US 1667366) are carriers of BLAD.

https://omia.org/OMIA000595/9913/

## Holstein - Complex Vertebral Malformation (CVM)

(CVF = Not carrier, CVC = Single carrier, CVS = Double carrier)

CVM is an autosomal recessively inherited disease in the Holstein population. Affected calves are either aborted during gestation, born prematurely, stillborn or in rare cases born alive. The weight of the calves is reduced, and they have a misshapen backbone, especially in the neck and rib area. They also have tendon contractions in the legs. Several other malformations including heart malformations are associated with this syndrome. The key ancestor of the disorder has been identified as Penstate Ivanhoe Star (US 1441440) and his son Carlin-M Ivanhoe Bell (US 1667366) has inherited the disorder.

https://omia.org/OMIA001340/9913/

## Holstein - Zinc deficiency (Adema disease)

(ZDF = Not carrier, ZDC = Single carrier, ZDS = Double carrier)

Adema disease is an autosomal recessively inherited disease in the Holstein population. It is caused by a defect that limits the amount of zinc that can be absorbed in the gastrointestinal tract. Affected calves will therefore have zinc deficiency. When they are born, they have no abnormalities, but at around the age of 4-12 weeks, changes in the skin are seen in the head and later on the legs. The affected calves have impaired immune response and growth. If they do not receive treatment, the calves die before 4 months of age. By treating the calves with large amounts of zinc they can be kept alive.

## Holstein – Syndactylism (Mulefoot)

(MFF = Not carrier, MFC = Single carrier, MFS = Double carrier)

Mulefoot is an autosomal recessively inherited disease in the Holstein population. Affected calves are born with a malformation of the claw. It is caused by a fusion of the two digits, causing the animal to only have one digit. The calf can be affected on all four limbs, or only a single limb. The front feet are most commonly affected. Mulefoot has been reported in many breeds of cattle and in many different countries. The main occurrence has however been seen in the Holstein population. Carriers are associated with a superior milk production. They key ancestor is believed to be Gar-Bar-Dale Burke Kate (US 1410387).

https://omia.org/OMIA000963/9913/

#### Holstein - Brachyspina (BY)

(BYF = Not carrier, BYC = Single carrier, BYS = Double carrier)

BY is an autosomal recessively inherited disease in the Holstein population. The disorder often leads to abortion, but some calves are born at full term. These calves are either stillborn or die shortly after birth. Affected calves are characterized by severely reduced body weight (around 10 kg), growth retardation, extensive vertebral malformations causing a significant shortening of the spine (brachyspina) and thereby the body. In addition, affected calves exhibit malformation of the inner organs, in particular the heart, kidneys and gonads, a shortened lower jaw and long, slender limbs.

BY was described for the first time in Denmark in 2006, but it has been identified throughout the Holstein population. The key ancestor has been identified as Sweet Haven Tradition (US 1682485) and his son Bis-May Tradition Cleitus (US 1879085) inherited the disorder.

https://omia.org/OMIA000151/9913/

#### Holstein - Deficiency of Uridine Monophosphate Synthase (DUMPS)

(DPF = Not carrier, DPC = Single carrier, DPS = Double carrier)

DUMPS is an autosomal recessively inherited metabolic disorder found in the Holstein population. Homozygous fetuses are aborted around day 40 of gestation. Heterozygous animals are not affected by the disorder, they however have an increased level of orotic acid in the milk and urine during gestation. <a href="https://omia.org/OMIA000262/9913/">https://omia.org/OMIA000262/9913/</a>

#### Holstein - Factor XI deficiency (blood clotting disorder)

(XIF = Not carrier, XIC = Single carrier, XIS = Double carrier)

Factor XI deficiency is an autosomal recessively inherited disease in the Holstein population. The disorder has been identified in the North American Holstein population and causes a bleeding disorder, where blood clotting is impaired. Animals with the disorder show an impaired ability to stop bleeding, can have blood in the milk and anaemia (low level of red blood cells). Animals that are both heterozygous and homozygous, can have decreased fertility, lower survival and be more susceptible to infectious diseases. The effects are more pronounced in homozygous animals.

https://omia.org/OMIA000363/9913/

## Holstein – Citrullinemia

(CNF = Not carrier, CNC = Single carrier, CNS = Double carrier)

Citrullinemia is an autosomal recessively inherited disease in the Holstein population. Affected calves are unable to excrete ammonia, which leads to neurological symptoms such as blindness, cramps and unsteady gait. The symptoms become progressively worse, and lead to death within one week of birth. <a href="https://omia.org/OMIA000194/9913/">https://omia.org/OMIA000194/9913/</a>

## Holstein - Cholesterol deficiency (HDC)

(CDF = Not carrier, CDC = Single carrier, CDS = Double carrier)

HDC was identified in the German Holstein population and is an autosomal recessively inherited disease. Affected calves have a low level of cholesterol in the blood. Heterozygous animals show no symptoms, but there is a large impact on homozygous animals. Cholesterol deficiency impairs the absorption of fat from the feed and milk, which has a negative impact on the growth and health of the calf. Homozygous calves show signs of severe cholesterol deficiency and often die within a few days or at the most a few months after birth, typically due to chronic diarrhea and emaciation. The key ancestor is believed to be the Canadian Holstein bull Maughlin Storm (CAN 5457798).

https://omia.org/OMIA001965/9913/

#### Holstein - Hereditary cardiomyopathy

(HCF = Not carrier, HCC = Single carrier, HCS = Double carrier)

Hereditary cardiomyopathy is an autosomal recessively inherited disease in the Holstein population. It causes an increase in the volume of the muscle tissue in the heart, due to an increase in the size of the muscle cells.

https://omia.org/OMIA000515/9913/

## Holstein - Holstein Haplotype 1 (HH1)

(HH1F = Not carrier, HH1C = Single carrier, HH1S = Double carrier)

HH1 is an autosomal recessively inherited disease in the Holstein population. The disorder was identified in the North American Holstein population and primarily causes early embryonic death (spontaneous abortion), but abortion can occur throughout the gestation. The key ancestor is believed to be Pawnee Farm Arlinda Chief (US1427381).

https://omia.org/OMIA000001/9913/

#### Holstein - Holstein Haplotype 2 (HH2)

(HH2F = Not carrier, HH2C = Single carrier, HH2S = Double carrier)

HH2 is an autosomal recessively inherited disease in the Holstein population. The disorder was identified in the North American Holstein population and primarily causes early embryonic death (spontaneous abortion), within the first 100 days of gestation. The key ancestor is believed to be Willowholme Mark Anthony (CAN 334489).

https://omia.org/OMIA001823/9913/

#### Holstein - Holstein Haplotype 3 (HH3)

(HH3F = Not carrier, HH3C = Single carrier, HH3S = Double carrier)

HH3 is an autosomal recessively inherited disease in the Holstein population. The disorder has been identified in the North American and the Nordic Holstein population, however the frequency was somewhat lower in the Nordic population. HH3 primarily causes early embryonic death (spontaneous abortion), within the first 60 days of gestation. The key ancestors are believed to be Glendell Arlinda Chief

(US 1556373) and Gray View Skyliner (US 1244845).

https://omia.org/OMIA001824/9913/

#### Holstein - Holstein Haplotype 4 (HH4)

(HH4F = Not carrier, HH4C = Single carrier, HH4S = Double carrier)

HH4 is an autosomal recessively inherited disease in the Holstein population. The disorder was identified in the French Holstein population and causes early embryonic death (spontaneous abortion). For animals that are a heterozygous for the gene, a reduction in fertility is seen. The effect is more pronounced in heifers than cows. The key ancestor is believed to be Besne Buck (FRA 4486041658).

https://omia.org/OMIA001826/9913/

#### Holstein - Holstein Haplotype 5 (HH5)

(HH5F = Not carrier, HH5C = Single carrier, HH5S = Double carrier)

HH5 is an autosomal recessively inherited disease in the Holstein population. The disorder has been identified in the North American and German Holstein population and causes early embryonic death (spontaneous abortion). The key ancestor is believed to be Thornlea Texal Supreme (CAN 264804). https://omia.org/OMIA001941/9913/

### Holstein - Holstein Haplotype 6 (HH6)

(HH6F = Not carrier, HH6C = Single carrier, HH6S = Double carrier)

HH6 is an autosomal recessively inherited disease in the Holstein population. It was identified in the French Holstein population and causes early embryonic death (spontaneous abortion), before 35 days of gestation. The key ancestor is believed to be Gray View Skyliner (US 1244845).

https://omia.org/OMIA002149/9913/

## Holstein - Holstein Haplotype 7 (HH7)

(HH7F = Not carrier, HH7C = Single carrier, HH7S = Double carrier)

HH7 is an autosomal recessively inherited disease in the Holstein population. It was identified in the French Holstein population and causes early embryonic death (spontaneous abortion), before 35 days of gestation. <a href="https://omia.org/OMIA001830/9913/">https://omia.org/OMIA001830/9913/</a>

#### Holstein – Achondrogenesis, Type II (Bulldog)

(B3F = Not carrier, B3C = Single carrier, B3S = Double carrier)

Bulldog is a well-known congenital syndrome and occurs sporadically in many cattle breeds. In 2015, it was observed in the Nordic Holstein population, in a way that suggested dominant inheritance with incomplete penetrance or a mosaic mutation. Affected calves are stillborn and display dwarfism. The body and limbs are shortened and compressed due to a reduced length of the spine. Also, malformation of the face and legs are seen. The body weight is severely reduced to approximately 25 kg's. The key ancestor was found to be VH Cadiz Captivo (DK 256588).

https://omia.org/OMIA001926/9913/

## Holstein – Bovine Lymphocyte Intestinal Retention Defect (BLIRD)(LR)

(LRF = Not carrier, LRC = Single carrier, LRS = Double carrier)

BLIRD is an autosomal recessively inherited disease in the Holstein population. Double carriers have low daily gain and high mortality in the rearing period due to abnormal response to infection as well as increased parasitism. The key ancestor is believed to be Emprise Bell Elton (US 1912270). Also O-Bee Manfred Justice (US 122358313), grandson of Elton, is known carrier.

#### Holstein – Early onset muscle weakness syndrome (MW)

(MWF = Not carrier, MWC = Single carrier, MWS = Double carrier)

Newborn calves are weak and unable to stand up or lose the ability to stand shortly after birth. Most calves die within 6 weeks after birth. The likely causative mutation is in the CACNA1S gene on chromosome 16. The key ancestor is believed to be Southwind Bell of Bar-Lee (US 1964484).

https://uscdcb.com/recumbency-in-holstein-calves/

#### Holstein – Red factor

(RDF = Not carrier, RDC = Single carrier, RDS = Double carrier)

Red factor is a recessive gene that codes for red coat colour in the Holstein population. An animal has to inherit the gene from both the sire and the dam, before it will have red coat colour.

#### Holstein – Dominant variant red factor (Variant red)

(VRF = Not carrier, VRC = Single carrier, VRS = Double carrier)

Variant red is a dominant variant of the red factor in the Holstein population. It is a dominant gene, which means that if an animal inherits the gene from either or both the sire or the dam, it will have red coat colour. It was identified for the first time in 1980 when the cow Surinam Sheik Rosabel-Red was born with red coat colour, despite both parents being black and not carrying the recessive gene for red coat colour. The dominant red gene is completely independent from the recessive red factor.

https://omia.org/OMIA001529/9913

#### Nordic Red Dairy Cattle - Spinal dysmyelination (SPAST)

(SDF = Not carrier, SDC = Single carrier, SDS = Double carrier)

Spinal dysmyelination is a recessive congenital neurodegenerative disease identified in the Brown Swiss population. It is a disorder in the central nervous system and is caused by a defect in the neuronal pathways. Affected calves are unable to stand due to spasms of the limbs and the body. The calves die shortly after birth. The key ancestor is believed to be White Cloud Jason's Elegant (US 148551). https://omia.org/OMIA001247/9913/

## Nordic Red Dairy Cattle – Trimethylaminuria (fishy taint)

(FMF = Not carrier, FMC = Single carrier, FMS = Double carrier)

Trimethylaminuria is an autosomal recessively inherited disease in the Nordic Red Dairy Cattle population. Affected animals produce milk that has fishy odour and taste. Other than a fishy taint off the milk, affected animals have no visible symptoms and are healthy.

https://omia.org/OMIA001360/9913/

# Nordic Red Dairy Cattle - Bovine progressive degenerative myeloencephalopathy (Weaver syndrome)

(WEF = Not carrier, WEC = Single carrier, WES = Double carrier)

Weaver syndrome is an autosomal recessively inherited disease in the Brown Swiss population. Weavers syndrome is a progressive disorder in the central nervous system. The first signs of illness are seen when the animal is 5-8 months old and typically include a weaving gait, along with weakness and lack of coordination of the hind limbs. The symptoms progressively become more severe, and when the animal is 18-36 months old they are unable to stand. Either the animals die of emaciation or are euthanized. The key ancestor is the Brown Swiss bull Autumn Sun (US 107915).

https://omia.org/OMIA000827/9913/

## Nordic Red Dairy Cattle - Spinal muscular atrophy (SMA)

(SMF = Not carrier, SMC = Single carrier, SMS = Double carrier)

Spinal muscular atrophy is an autosomal recessively inherited disease in the Brown Swiss population. It is a neuromuscular disorder characterised by a loss of lower motor neurons and progressive muscle wasting. The nerve cells in the spinal cord that control muscle function die, which causes muscle wastage. It is most frequently seen in 1-12 weeks old calves, but in some cases, it can be seen from birth. It causes the calves to become weak and have problems standing, the symptoms progressively worsen until the calves die or are euthanized. The key ancestor is the Brown Swiss bull Meadow View Destiny (US 118619). https://omia.org/OMIA000939/9913/

## Nordic Red Dairy Cattle - Arthrogryposis multiplex congenita (AMC)

(A2F = Not carrier, A2C = Single carrier, A2S = Double carrier)

AMC is an autosomal recessively inherited disease in the Nordic Red Dairy Cattle population, but also segregates in other populations. It cases severe malformation of affected calves. The calves are stillborn and most often lead to calving difficulties.

https://omia.org/OMIA002022/9913/

## Nordic Red Dairy Cattle - Ptosis, intellectual disability, retarded growth and mortality (PIRM/AH1)

(PIF = Not carrier, PIC = Single carrier, PIS = Double carrier)

AH1 is an autosomal recessively inherited disease, identified in the Canadian and Nordic Ayrshire populations. AH1 and PIRM are located very closely to each other in the genome, and it is expected that they are the same disease. They primarily cause early embryonic death (spontaneous abortion) within the first 100 days of gestation. If affected calves are born, they are malformed and have inhibited growth. There has also been reports of learning disabilities (e.g. difficulties learning to drink) The key ancestor is believed to be Selwood Betty's Commander (CAN 393145).

https://omia.org/OMIA001934/9913/

#### Nordic Red Dairy Cattle - Ayrshire Haplotype 2 (AH2)

(AH2F = Not carrier, AH2C = Single carrier, AH2S = Double carrier)

AH2 is an autosomal recessively inherited disease, identified in the North American Ayrshire population. It causes early embryonic death (spontaneous abortion). The key ancestor is believed to be Oak-Ridge Lightning (US 120135).

https://omia.org/OMIA002134/9913/

## Nordic Red Dairy Cattle - Brown Swiss Haplotype 1 (BH1)

(BH1F = Not carrier, BH1C = Single carrier, BH1S = Double carrier)

BH1 is an autosomal recessively inherited disease, identified in the Brown Swiss population. It causes early embryonic death (spontaneous abortion), before 60 days of gestation in affected calves. The key ancestor is the Brown Swiss bull West Lawn Stretch Improver (US 163153).

https://omia.org/OMIA001825/9913/

#### Nordic Red Dairy Cattle - Brown Swiss Haplotype 2 (BH2)

(BH2F = Not carrier, BH2C = Single carrier, BH2S = Double carrier)

BH2 is an autosomal recessively inherited disease, identified in the Brown Swiss and Fleckvieh populations. The majority of affected calves are stillborn or die shortly after birth. The calves have a low birth weight and are underdeveloped. BH2 has incomplete penetrance and a minority of the calves survive after birth. Surviving calves suffer from chronic lung infections, that results in inhibited growth and high mortality. The majority of these calves are euthanized or die within the first month. The key ancestor is believed to be the Brown Swiss bull Rancho Rustic My Design (US 144488).

https://omia.org/OMIA001939/9913/

#### Nordic Red Dairy Cattle - Bos Taurus Autosome 12 (BTA12)

(B12F = Not carrier, B12C = Single carrier, B12S = Double carrier)

BTA12 is an autosomal recessively inherited disease, in the Nordic Red Dairy Cattle population. It causes early embryonic death (spontaneous abortion), between the first and fifth month of gestation. The mutation occurs in the RNASEH2B gene.

https://omia.org/OMIA001901/9913/

## Nordic Red Dairy Cattle - Bos Taurus Autosome 23 (BTA23)

(B23F = Not carrier, B23C = Single carrier, B23S = Double carrier)

BTA123 is an autosomal recessively inherited disease. The disorder was identified in the Nordic Red Dairy Cattle population and causes embryonic death (spontaneous abortion), late in gestation or stillborn calves. The calves have no deformities.

#### Nordic Red Dairy Cattle - Bovine tail stump sperm defect (TSD)

(TSDF = Not carrier, TSDC = Single carrier, TSDS = Double carrier)

TSD is an autosomal recessively inherited disease, identified in the Nordic Red Dairy Cattle population. The disorder affects the sperm quality of homozygous bulls. The spermatozoa of affected bulls are immotile, due to tail defects, which indicates disturbed spermatogenesis.

https://omia.org/OMIA001334/9913/

## Nordic Red Dairy Cattle - Chondrodysplasia, recessive (Bulldog)

(BDF = Not carrier, BDC = Single carrier, BDS = Double carrier)

Bulldog is a well-known congenital syndrome and occurs sporadically in many cattle breeds. Affected calves are stillborn and display dwarfism ("bulldog" syndrome). The body and limbs are shortened and compressed due to a reduced length of the spine. Also, malformation of the face and legs are seen. The body weight is severely reduced to approximately 25 kg's.

#### Jersey - Rectovaginal constriction (RVC)

(RVF = Not carrier, RVC = Single carrier, RVS = Double carrier)

RVC is an autosomal recessively inherited disease, identified in the Jersey population. Fibrous bands cause constriction in rectovaginal area. This leads to dystocia and difficulties performing a rectal examination. <a href="https://omia.org/OMIA000850/9913/">https://omia.org/OMIA000850/9913/</a>

#### Jersey - Jersey Haplotype 1 (JH1)

(JH1F = Not carrier, JH1C = Single carrier, JH1S = Double carrier)

JH1 is an autosomal recessively inherited disease. The disorder was identified in the North American and the Nordic Jersey populations, however the frequency was somewhat lower in the Nordic population. JH1 primarily causes early embryonic death (spontaneous abortion), within the first 60 days of gestation. The key ancestor is believed to be Observer Chocolate Soldier (US 596832).

https://omia.org/OMIA001697/9913/

## Jersey – Jersey Haplotype 2 (JH2)

(JH2F = Not carrier, JH2C = Single carrier, JH2S = Double carrier)

JH2 is an autosomal recessively inherited disease. The disorder was identified in the North American Jersey population. JH2 primarily causes early embryonic death (spontaneous abortion), within the first 60 days of gestation. The key ancestor is believed to be Favorite Secret Triumph (US 602283). https://omia.org/OMIA001942/9913/

#### Jersey – Neuropathy with splayed forelimbs (JNS)

(JNSF = Not carrier, JNSC = Single carrier, JNSS = Double carrier)

JNS is an autosomal recessively inherited disease. The disorder was identified in the North American Jersey population and go back to an animal born i 1995. Calves affected with JNS are unable to stand on splayed forelimbs that exhibit significant extensor rigidity and excessive lateral abduction at birth. Affected calves generally are alert at birth but exhibit neurologic symptoms, including spasticity of head and neck and convulsive behavior. Other symptoms reported include dislocated shoulders, congenital craniofacial anomalies, and degenerative myelopathy.

https://www.omia.org/OMIA002298/9913/

## All dairy cattle - Progressive retinal degeneration (RP1)

(RP1F = Not carrier, RP1C = Single carrier, RP1S = Double carrier)

Progressive retinal degeneration is an autosomal recessively inherited disease. In the French Normande population, an association between homozygotic carrier animals of the RP1 gene and progressive blindness has been demonstrated. Blindness is due to a progressive degeneration of the photoreceptors. The RP1 mutation segregates within many cattle populations and is prevalent in e.g. the Finncattle population. For other breeds than Normande the phenotypic effect of the mutation is unclear.

https://omia.org/OMIA000866/9913/

## All dairy cattle - Polled

(POF = Not carrier, POC = Single carrier, POS = Double carrier)

Polled animals are hornless. There are polled animals in most cattle breeds, some to a larger extent than others. The polled gene is dominant. An animal is polled if it inherits the gene from either or both the sire and the dam.

## All dairy cattle - Beta-casein

(Possible genotypes: A1/A1, A1/A2, A2/A2)

Beta-casein is one of the main components of milk protein. There are two different variants (A1 and A2). In Europe (except France), USA, Australia and New Zealand, the A1 variant is the most frequent. Some scientific research has found a connection between A1 and some chronic illnesses.

## All dairy cattle - Kappa-casein

(Possible genotypes: AA, AB, BB, AE, BE, EE)

Kappa-casein is a milk protein that is involved in several physiological processes. It helps stabilize the fat cells in the milk and is one of the key proteins in cheese production. During the production of cheese, the kappa-casein is segregated by the rennet, which causes the milk to thicken. There are 6 different variants of the gene (AA, AB, BB, AE, BE, EE). Cows that carry the BB gene, produce milk that is superior to the others regarding cheese production. Cows with the EE gene, produce milk that does not thicken the milk, and it there not suited for cheese production.