

## Genetic traits in the Nordic countries – Beef cattle

### **Beef 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 Charolais population. For other breeds than Normande the phenotypic effect of the mutation is unclear.

<https://omia.org/OMIA002029/9913/>

### **Aberdeen Angus - Arthrogyposis Multiplex congenita (Curly Calf)**

*(A1F = Not carrier, A1C = Single carrier, A1S = Double carrier)*

Curly calf is an autosomal recessively inherited disease identified in the Aberdeen Angus population. Affected calves are stillborn and severely malformed. The calves have inhibited growth and muscle development. The spine is bent and twisted and in some cases the limbs are rigid, which causes calving difficulties. The key ancestor is the American bull GAR Precision 1680.

<https://omia.org/OMIA002135/9913/>

### **Aberdeen Angus - Neuropathic Hydrocephalus**

*(NHF = Not carrier, NHC = Single carrier, NHS = Double carrier)*

Neuropathic Hydrocephalus is an autosomal recessively inherited disease identified in the Aberdeen Angus population. Affected calves are aborted near to term and have a low birth weight. Some calves may also be aborted earlier in gestation. The skull is markedly enlarged, and the bones of skull are malformed. The cranial cavity is filled with fluid and no brain tissue is evident. <https://omia.org/OMIA000487/9913/>

### **Aberdeen Angus - Development duplication**

*(DDF = Not carrier, DDC = Single carrier, DDS = Double carrier)*

Development duplication is an autosomal recessively inherited disease identified in the Aberdeen Angus population. Affected calves can be born with extra limbs, or parts of an extra limb (polymelia). In some cases, calves may be conjoined (conjoined twins). Because of these malformations, there is a higher frequency of dystocia and a high calf mortality. There is also significantly higher embryonic mortality (spontaneous abortion). Some animals survive and function normally with their extra limb either attached or surgically removed. <https://omia.org/OMIA002103/9913/>

### **Aberdeen Angus – Contractural arachnodactyly (Fawn calf syndrome)**

*(CAF = Not carrier, CAC = Single carrier, CAS = Double carrier)*

Fawn calf syndrome is an autosomal recessively inherited disease identified in the Aberdeen Angus population. Affected calves are usually born alive at a normal birth weight but are malformed. The severity of the disease varies and affected calves appear to be relatively normal at 4-6 months of age. At birth, the upper limb joints are affected by contractures which reduce the range of angular movement. This causes

affected calves to assume an abnormal crouched posture, resembling a deer fawn. Affected calves are reported to be taller and more slender, also as they grow older. This results in poor slaughter results.

<https://omia.org/OMIA001511/9913/>

### ***Blonde d'Aquitaine - Axonopathy (Congenital Neurodegeneration)***

*(AXF = Not carrier, AXC = Single carrier, AXS = Double carrier)*

Axonopathy is an autosomal recessively inherited disease identified in the Blonde d'Aquitaine population. Affected calves are born without symptoms, but often have a wide stance. At around a month of age the calves progressively begin to lose control of their hind legs and become unable to stand. Affected calves are usually euthanized by 10 months of age. <https://omia.org/OMIA001106/9913/>

### ***Blonde d'Aquitaine and Limousine - Protoporfyrria***

*(PRF = Not carrier, PRC = Single carrier, PRS = Double carrier)*

Protoporfyrria is found in both human and cattle, where human is effect if they carry one defect gen, while it is an autosomal recessively inherited disease for cattle, so they shall have two defect gens to be affected. It has been identified in the Blonde d'Aquitaine and Limousine population. The calves are effect from birth, and they are very sensitive for light. Animals can also get rashes and sores on the skin.

<https://omia.org/OMIA000836/9913/>

### ***Charolais - Anhidrotic Ectodermal Dysplasia (AED)***

*(ADF = Not carrier, ADC = Single carrier, ADS = Double carrier)*

AED is an autosomal recessively inherited disease. It has been identified in the French Charolais population. Affected calves are born with hypotrichosis (sparseness of hair) and hypodontia (no teeth). Additionally, they are born with sweat glands that are not functional. In general, affected calves do not live longer than 4-5 months after birth, many die or are euthanized earlier. The calves have problems eating and suffer from hypo- or hyperthermia as their sweat glands are not functional. They also have respiratory distress as they have defects to the bronchial glands. The key ancestor is believed to be the French Charolais sire Invincible (FRA 1893105503). <https://omia.org/OMIA002128/9913/>

### ***Charolais – Ataxia***

*(PAF = Not carrier, PAC = Single carrier, PAS = Double carrier)*

Ataxia is an autosomal recessively inherited disease. It has been identified in the French Charolais population. Affected calves show progressive ataxia symptoms, i.e. unsteady gait and stiff hind limbs. The symptoms grow progressively worse, until the animal remains in a lying position. The first symptoms typically occur at around 18-24 months, but some cases were observed as early as 6 months of age and up to 5 years of age. The progressiveness of the symptoms varies from a few weeks to more than 18 months.

<https://omia.org/OMIA000527/9913/>

### ***Dexter – Chondrodysplasia, BD1 (Bulldog)***

*(B1F = Not carrier, B1C = Single carrier, B1S = Double carrier)*

Bulldog is an autosomal incomplete dominant inherited disease identified in the Dexter population. It can be caused by two mutations (BD1 and BD2) in the ACAN gene. Bulldog causes embryonic death

(spontaneous abortion) within the first 7 months of gestation. Affected calves are severely malformed and display extreme dwarfism, a short spine and large “bulldog-like” heads.

<https://omia.org/OMIA001271/9913/>

### **Dexter – Chondrodysplasia, BD2 (Bulldog)**

*(B2F = Not carrier, B2C = Single carrier, B2S = Double carrier)*

Bulldog is an autosomal incomplete dominant inherited disease identified in the Dexter population. It can be caused by two mutations (BD1 and BD2) in the ACAN gene. Bulldog causes embryonic death (spontaneous abortion) within the first 7 months of gestation. Affected calves are severely malformed and display extreme dwarfism, a short spine and large “bulldog-like” heads.

<https://omia.org/OMIA001271/9913/>

### **Hereford – Hypotrichosis**

*(HYF = Not carrier, HYC = Single carrier, HYS = Double carrier)*

Hypotrichosis is an autosomal recessively inherited disease identified in the Hereford population. Affected calves are born with partial or complete lack of hair. As they mature their hair coat is curly or fuzzy in texture. Adult animals may periodically display a “patchy” haircoat, with bare patches.

<https://omia.org/OMIA002114/9913/>

### **Hereford - Idiopathic Epilepsy (IE)**

*(IEF = Not carrier, IEC = Single carrier, IES = Double carrier)*

IE is an autosomal recessively inherited disease identified in the Hereford population. The lifespan of affected calves varies from a few days to several months. They experience seizures, of which the duration can vary from a few minutes to more than an hour. During the seizure, the calf lays on its side with stiff legs. <https://omia.org/OMIA000344/9913/>

### **Hereford – Maple Syrup Urine Disease (MSUD)**

*(MSU1F = Not carrier, MSU1C = Single carrier, MSU1S = Double carrier)*

MSUD is an autosomal recessive defect reported in Hereford. Homozygotic carrier calves have a defect in an enzyme that breaks down complex amino acids in the diet. The resulting build-up of these amino acids in the body causes lethal brain damage. The clinical characters of the disease are characterised by the rapid onset of progressive neurological disease, resulting in death within a few days of birth. Different mutations segregates within different breeds. Results for the Hereford variant is named MSU1 in test results.

<https://omia.org/OMIA000627/9913/>

### **Hereford – Mandibulofacial dysostosis (MD)**

*(MDF = Not carrier, MDC = Single carrier, MDS = Double carrier)*

MD is an autosomal recessive defect reported in Hereford. Homozygotic carrier calves have a variably shortened and/or asymmetric lower mandible. The unique and consistent hallmarks of the condition include unusual bilateral skin tags just behind the corner of the mouth. These tags are attached to an unusual bone formation. <https://omia.org/OMIA002288/9913/>

### **Highland cattle - Crop ear**

*(CEF = Not carrier, CEC = Single carrier, CES = Double carrier)*

Crop ear or Notch ear is an autosomal dominantly inherited disease. It has been identified in the Highland cattle population. Affected calves are born with malformation of the ears. There are two categories of severity. Animals in category 1 have mild to moderate notches in the ear and the ear cartilage appears normal or is mildly deformed. In category 2 the animals have clearly shortened ears, large notches and prominent and slightly twisted upper edges of the ear cartilage. <https://omia.org/OMIA000317/9913/>

### **Limousine – Cleft Palate**

*(CPF = Not carrier, CPC = Single carrier, CPS = Double carrier)*

The palate is a structure separating the oral and nasal cavities and its integrity is essential for feeding and breathing. The total or partial opening of the palate is called a cleft palate and is a common malformation in mammals with environmental or hereditary aetiologies. Generally, it compromises life expectancy in the absence of surgical repair. In the Limousine breed a mutation in the MYH3 gene leads to cleft palate. <https://www.omia.org/OMIA002590/9913/>

### **Simmental – Thrombopathia**

*(TPF = Not carrier, TPC = Single carrier, TPS = Double carrier)*

Thrombopathia is an autosomal recessively inherited disease, identified in the Simmental and Fleckvieh populations. Animals that are double carrier will have reduced ability for blood coagulation, and therefore an increased risk of deadly bleeding after a trauma. <https://omia.org/OMIA002433/9913/>

### **Simmental – 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, Fleckvieh and Simmental 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/>

### **Beef 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.

### **Beef cattle - Dilutor (coat colour dilution)**

*(DLF = Not carrier, DLC = Single carrier, DLS = Double carrier)*

Dilutor causes the coat colour to be diluted in affected calves. If a black cow that carries the gene has a calf, the black coat colour would be diluted to grey. If the affected calf receives the gene from both the sire and the dam (homozygous), the dilution will be stronger compared to a calf that only receives a single gene (heterozygous). <https://omia.org/OMIA001545/9913/>

### **Beef – Red factor**

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

Red factor is a recessive gene that codes for red coat color in several cattle breeds amongst these the Aberdeen Angus breed. An animal has to inherit the gene from both the sire and the dam, before it will have red coat colour.

### **Beef cattle – Myostatin 1 (nt821)**

*(M1F = Not carrier, M1C = Single carrier, M1S = Double carrier)*

Myostatin influences the production of a protein that controls muscle development. Mutation of the gene causes the proteins to be less effective at controlling muscle development. This results in increased muscle mass, or double-muscularity. There are many different mutations of the myostatin gene, that have slightly different expressions.

The nt821 mutation is recessive and is found in the Angus, Belgian Blue, Blonde d'Aquitaine, Charolais and Limousin populations. Heterozygous animals are superior for the carcass traits, compared to calves that do not have the mutation. Homozygous animals will an increased muscle mass, reduced fat content, but also heavier birth weight and therefore a higher risk of calving difficulties. <https://omia.org/OMIA000683/9913/>

### **Beef cattle - Myostatin 2 (nt419)**

*(M2F = Not carrier, M2C = Single carrier, M2S = Double carrier)*

Myostatin influences the production of a protein that controls muscle development. Mutation of the gene causes the proteins to be less effective at controlling muscle development. This results in increased muscle mass, or double-muscularity. There are many different mutations of the myostatin gene, that have slightly different expressions.

The nt419 mutation is recessive and is found in the Maine Anjou population. Homozygous animals will exhibit double muscling (hyperplasia), increased meat tenderness, larger birth weights and therefore a higher risk of calving difficulties. The effects in heterozygous animals are less pronounced.

<https://omia.org/OMIA000683/9913/>

### **Beef cattle - Myostatin 3 (Q204x)**

*(M3F = Not carrier, M3C = Single carrier, M3S = Double carrier)*

Myostatin influences the production of a protein that controls muscle development. Mutation of the gene causes the proteins to be less effective at controlling muscle development. This results in increased muscle

mass, or double-muscularity. There are many different mutations of the myostatin gene, that have slightly different expressions.

The Q204x mutation is a 'partially dominant' mutation of the Myostatin gene and is found in the Blonde d'Aquitaine, Charolais and Limousin populations. Homozygous animals will exhibit double muscling (hyperplasia), reduced fat cover, increased meat tenderness, larger birth weights and therefore a higher risk of calving difficulties. In females, there is a slightly reduced milking ability. The effects in heterozygous animals are less pronounced. <https://omia.org/OMIA000683/9913/>

### **Beef cattle – Myostatin 4 (E226X)**

*(M4F = Not carrier, M4C = Single carrier, M4S = Double carrier)*

Myostatin influences the production of a protein that controls muscle development. Mutation of the gene causes the proteins to be less effective at controlling muscle development. This results in increased muscle mass, or double-muscularity. There are many different mutations of the myostatin gene, that have slightly different expressions.

The E226X mutation is a mutation of the Myostatin gene and is found in the Maine Anjou population. Homozygous animals will exhibit double muscling (hyperplasia), increased meat tenderness, larger birth weights and therefore a higher risk of calving difficulties. The effects in heterozygous animals are less pronounced. <https://omia.org/OMIA000683/9913/>

### **Beef cattle - Myostatin 5 (3811T)**

*(M5F = Not carrier, M5C = Single carrier, M5S = Double carrier)*

Myostatin influences the production of a protein that controls muscle development. Mutation of the gene causes the proteins to be less effective at controlling muscle development. This results in increased muscle mass, or double-muscularity. There are many different mutations of the myostatin gene, that have slightly different expressions.

The 3811T mutation is a mutation of the Myostatin gene and is found in the Blonde d'Aquitaine population. Homozygous animals will exhibit increased muscle mass. The effects in heterozygous animals are less pronounced. <https://omia.org/OMIA000683/9913/>

### **Beef cattle – Myostatin 6 (C313Y)**

*(M6F = Not carrier, M6C = Single carrier, M6S = Double carrier)*

Myostatin influences the production of a protein that controls muscle development. Mutation of the gene causes the proteins to be less effective at controlling muscle development. This results in increased muscle mass, or double-muscularity. There are many different mutations of the myostatin gene, that have slightly different expressions.

The C313Y mutation is a mutation of the Myostatin gene and is found in the Piedmontese population. Homozygous animals will exhibit double muscling (hyperplasia), increased meat tenderness, larger birth weights and therefore a higher risk of calving difficulties. The effects in heterozygous animals are less pronounced. <https://omia.org/OMIA000683/9913/>

### **Beef cattle – Myostatin 7 (F94L)**

*(M7F = Not carrier, M7C = Single carrier, M7S = Double carrier)*

Myostatin influences the production of a protein that controls muscle development. Mutation of the gene causes the proteins to be less effective at controlling muscle development. This results in increased muscle mass, or double-muscularity. There are many different mutations of the myostatin gene, that have slightly different expressions.

The F94L mutation is a mutation of the Myostatin gene and is found in the Limousin and Charolais populations. The majority of Limousin cattle carry this mutation. Homozygous animals will exhibit increased muscle mass and meat tenderness. They also have reduced external and intramuscular fat. The birth weight is not affected, which means there is no associated increase in calving difficulty. The effects in heterozygous animals are less pronounced. <https://omia.org/OMIA000683/9913/>

### **Beef cattle - Myostatin 8 (E291X)**

*(M8F = Not carrier, M8C = Single carrier, M8S = Double carrier)*

Myostatin influences the production of a protein that controls muscle development. Mutation of the gene causes the proteins to be less effective at controlling muscle development. This results in increased muscle mass, or double-muscularity. There are many different mutations of the myostatin gene, that have slightly different expressions.

The E291X mutation is a mutation of the Myostatin gene and is found in the Limousin population. Homozygous animals will exhibit double muscling (hyperplasia), increased meat tenderness, larger birth weights and therefore a higher risk of calving difficulties. The effects in heterozygous animals are less pronounced. <https://omia.org/OMIA000683/9913/>

### **Beef cattle – Myostatin 9 (D182N)**

*(M9F = Not carrier, M9C = Single carrier, M9S = Double carrier)*

Myostatin influences the production of a protein that controls muscle development. Mutation of the gene causes the proteins to be less effective at controlling muscle development. This results in increased muscle mass, or double-muscularity. There are many different mutations of the myostatin gene, that have slightly different expressions.

The D182N mutation is a mutation of the Myostatin gene. Homozygous animals will exhibit increased muscle mass and meat tenderness. They also have reduced external and intramuscular fat. The birth weight is not affected, which means there is no associated increase in calving difficulty. The effects in heterozygous animals are less pronounced. <https://omia.org/OMIA000683/9913/>

### **Beef cattle – Myostatin 10 (S105C)**

*(M10F = Not carrier, M10C = Single carrier, M10S = Double carrier)*

Myostatin influences the production of a protein that controls muscle development. Mutation of the gene causes the proteins to be less effective at controlling muscle development. This results in increased muscle mass, or double-muscularity. There are many different mutations of the myostatin gene, that have slightly different expressions.

The S105C mutation is a mutation of the Myostatin gene. Homozygous animals will exhibit increased muscle mass and meat tenderness. They also have reduced external and intramuscular fat. The birth weight is not affected, which means there is no associated increase in calving difficulty. The effects in heterozygous animals are less pronounced. <https://omia.org/OMIA000683/9913/>