GENETIC CONDITIONS IN BROWN SWISS CATTLE

A. Genetic Recessive Abnormalities

The following four abnormalities found in Brown Swiss cattle have been determined to be inherited characteristics caused by simple recessive genes and declared to be undesirable genetic recessive factors: Weaver (Bovine Progressive Degenerative Myeloencephalopathy), SMA (Spinal Muscular Atrophy), Spiderleg (Syndrome of Arachnomelia and Arthrogryposis [SAA]) and Spinal Dysmyelination (SDM). These abnormalities occur as a result of defects in the genetic code which have been passed on to offspring as recessives. There is no danger of other animals "catching" them. Recessives can only be passed on through genetic inheritance and can only be controlled through careful breeding management.

Weaver

Bovine Progressive Degenerative Myelo-encephalopathy (myelo destruction of the spinal cord, encephalopathy changes in the brain) causes a noticeable weaving gait of the affected animal, and this brought about the nickname "Weaver". Dr. Horst Leipold of Kansas State University initially suspected as the original cause to be a genetic mutation in a Brown Swiss mating in the 1920's or 1930's. Prevalence of this recessive was compounded by inbreeding and the early use of artificial insemination.

There are four basic criteria used for clinical diagnosis. Pathological examination of the brain and spinal cord is the only means of verification.



The onset of bilateral (both sides) hind leg weakness and ataxia (inability to coordinate movements) normally between six to eighteen months of age (around puberty).



Deficient proprioceptive reflexes (nerve impulses from brain not received by muscles in hind legs), but normal motor and sensory reflexes with no other neurological abnormality. (In other words, the animal can't seem to find its hind legs).



Absence of clinically significant skeletal or muscular abnormality.

Adherence to familial relationship (common to a breed). (So far, this has only been found in Brown Swiss cattle.)

What to Look For:



Symptoms generally appear from 6 to 18 months of age. Symptoms are caused from degeneration of nerve passages in the spinal cord and brain which prevent the transfer of nerve impulses from the brain to the leg muscles. Any deterioration of the central nervous system is permanent and irreversible.

Symptoms include:

- Lack of coordination
- Hind legs seem to be overstepping too high
- Front legs are held far apart to maintain balance
- When standing, the animal sways slightly
- Hind feet are close together and may cross
- Animal often tries to stand against something
- Staggering and/or swaying, an erratic gait
- Stumbling and falling, especially when excited
- Loss of control of pelvic limbs

The animal eventually suffers from malnutrition, so muscle atrophy (wasting away) is apparent. It usually takes up to two or three years to die. During this time, the animal should not be kept on concrete as it could fall unexpectedly. It is best to keep the animal in an open lot or on dirt. When the animal can no longer eat or drink, it should be put down by a veterinarian.

To have an animal clinically verified as a Weaver, the spinal cord and cerebellum (intact) must be obtained as soon as possible after death and forwarded to an approved laboratory to have the tissue analyzed before it deteriorates. Degeneration of the spinal cord is most evident in the thoracic (chest) area. This spinal cerebellar degeneration is comparable to Lou Gehrig's disease in humans.

Pictures 1-3 show a Weaver-affected animal



1 Hind feet close or cross while front feet are apart



2 Animal sways or weaves as if intoxicated



3 Animal eventually falls in advanced stages

Spinal Muscular Atrophy (SMA)

Spinal Muscular Atrophy is a condition very easy to miss since affected animals usually die of a secondary illness. Most cases have been reported since the mid-1980's. SMA is similar to Vitamin E or Selenium deficiency and some SMA calves are mistakenly thought to be Weavers. When there is no improvement with treatment for white muscle disease, SMA seems likely. SMA affects the nervous system characterized by skeletal muscle atrophy, decreased spinal reflexes, and motor weakness.

The onset of SMA occurs in a calf's first few weeks of life, usually between three to six weeks of age. In the earliest observed stages, the calves show loss of strength and balance in the rear legs. They appear to be healthy except for their stumbling and eventual inability to stand. Though well-fed and alert, SMA calves will become weaker and lose flesh.



What to look for: SMA symptoms generally appear from three to six weeks of age. There is no cure for this condition.

Symptoms include:

- Loss of balance in rear legs
- Difficulty in standing
- Calves lie with front legs outstretched
- Wasting away of muscle mass
- Eventual inability to stand
- Labored breathing

As the condition progresses, SMA calves develop motor problems in the front end. They typically show signs of labored breathing. At this point, the chest muscles have begun to atrophy. Once the calf can no longer get up, death usually occurs within a couple of days. Usually the cause of death is pneumonia by six to eight weeks of age. The association may wish to have the brain and spinal column of the affected animal sent to an approved laboratory for clinical confirmation.

Pictures 4-5 show an SMA-affected calf



4 Shows calf in typical position with front legs outstretched and upright on sternum



5 Shows weakness of hind legs and wasting away of muscle mass

Spinal Dysmyelination (SDM)

Spinal Dysmyelination is a relatively new abnormality found in Brown Swiss cattle. First diagnosed in Germany in 1993, this defect has some similarities with SMA, but appears to be a different abnormality. While it also affects calves, SDM seems to have a quicker onset time than SMA. While SMA calves show symptoms from 2 to 12 weeks after birth, SDM shows its affects immediately upon birth of the calf. The calf cannot stand at all after birth, and it will have spastic movement in its rear legs although they seem normal otherwise. SDM is also believed to be passed on as a genetic recessive. This means that two carriers must be mated for the condition to occur and then it will occur only once in every four offspring.

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What to look for:

SDM symptoms generally appear at birth. There is no cure for this condition.

Symptoms include:

- Inability to stand immediately after birth
- Spastic rear legs
- Calf otherwise alert

Pictures 6-8 show SDM-affected calf



6 Animal shows lateral position, opisthotonos, and extended limbs.



7 Calf has absence of limb movement and support.



8 Calf shows almost normal appearance when placed in sternal position.

Spiderleg (SAA)

Syndrome of Arachnomelia and Arthrogryposis (SAA), is due to an inherited neuromuscular dysfunction early in gestation. Calves are born with both muscular and skeletal deformity. Although only a few cases have been confirmed in the United States, there have been many cases in Europe stemming from the use of one bull family's genetics.



What to look for: Affected animals are self-evident.

Symptoms include:

- Spiderleg affected calves are usually stillborn or die during birth, although some live for a few hours
- Legs are abnormally long and thin and are curved or broken and bent grotesquely
- Joints may be hypermobile or frozen (requiring fetal dissection in the uterus to aid in birth)
- Fragile bones *Note:* Spontaneous fractures during birth could result in injuries to the dam's uterus and genital passage.
- Small and underdeveloped muscles
- Short, crooked or taut, overstretched tendons
- Short skull with dent in the frontal bone
- Short lower jaw
- Defects in vertebral column (twists)
- Large blood vessel aneurysms

Pictures 9-12 show Spiderleg-affected calves & fetuses



9 A newborn Brown Swiss calf afflicted with Spiderleg

Note arrows

indicating

fractures in long

bones of leg ►



10 X-ray of hind leg of Brown Swiss calf with Spiderleg



11 Stillborn Spiderleg-affected calf

TReporting Abnormalities

In order to maintain a breed relatively free of undesirable genetic factors and to provide valuable information for making breeding decisions, it is recommended that every Brown Swiss breeder report the occurrence of any abnormal animal to the National Association office at Beloit, Wisconsin, (608) 365-4474. The following guidelines are established for such reporting:

A. All animals should be reported to the Brown Swiss office by telephone as soon as they are discovered. Please be prepared to provide as much of the following information as possible:

- A description of the animal and its abnormality
- Identification of the animal, such as registration number and name, tattoo number, etc.
- Date of birth
- Sire and dam registration number and name
- Whether or not the animal itself and its sire and dam are alive

B. Based on the information received in the original report, the Brown Swiss office may request that blood samples be obtained from the abnormal animal and the sire and dam. These blood samples will be sent to an approved serology laboratory for parentage verification. The fees for such parentage verification will be paid by the national Brown Swiss Association.

C. Based on the information received in this initial report, the Brown Swiss Association may request that such abnormal animals or their brain and spinal cord be sent to an approved laboratory for examination and verification of the abnormality.

B. Haplotypes

A haplotype is a combination of alleles (DNA sequences) at adjacent locations on a chromosome that are inherited together.

BH1 – Bovine Haplotype 1

Discontinued Effective December 2018

BH1, a fertility factor previously identified in Brown Swiss and thought to impact early embryonic development and conception, has been shown to be insignificant and has been discontinued.



12 98-day old Spiderleg-affected fetus

BH2 – Bovine Haplotype 2

A haplotype impacting stillbirth (**BH2**) has been identified by Swiss and Austrian researchers. When a calf inherits BH2 from both the sire and dam as a homozygous recessive, it may be stillborn or die soon after birth. In Europe, two calves that died shortly after birth have been discovered as homozygous for BH2.

A list of BH2 carriers derived from monthly genomic results is available on the BSCBA website under genetics and will be updated with each sire evaluation run. Making efforts to avoid inbreeding when selecting matings will also assist in avoiding known and other potential genetic defects that have not yet been discovered.

A gene test for BH2 was added with the 80k SNP set used for genomic testing effective December 2018.

C. Polled

Polled (hornless) is expressed as a dominant gene over the more prevalent horned gene. This "dominance" allows a breeder to obtain polled animals more quickly through selection. A heterozygous (one gene copy) polled animal mated with another heterozygous polled animal will result in 25% horned offspring, 50% heterozygous polled and 25% homozygous polled offspring. Whereas, a homozygous (two gene copies) polled animal mated to another homozygous polled animal will result in 100% homozygous polled offspring.

With concern about animal welfare practices, genetic selection for the polled gene is expected to become more popular in the dairy industry. Currently, there are polled Brown Swiss bulls available, some of which are listed in the Brown Swiss Performance Summary.

A test is available for identifying polled animals, whether heterozygous or homozygous. To order a testing kit, you may contact UC Davis Veterinary Genetics Laboratory via their website at <u>https://www.vgl.ucdavis.edu/services/cattle.php</u> or by phone at 530-752-2211.

Designations:

BSCBA keeps a record of polled animals through registrations with the designations listed below and has provided this list to AIPL researchers.

NP – Observed Naturally Polled. The designations for polled animals prior to 2012.

P – **Observed Polled.** The current designation for observed Polled. If any of a polled sire's offspring are horned, the bull is heterozygous polled - one parent supplied a polled gene to the animal..

PP – Homozygous Polled. Both parents supplied a polled gene to the animal. (In order for an animal to be labeled homozygous polled, UC-Davis Poll testing results must be provided to BSCBA confirming the animal carries two copies of the polled gene.)

D. Genetic and Genomic Testing

<u>Weaver</u>

After much genetic research, a DNA test is available to determine Weaver carrier status with high reliability. Carrier status results are published only when a confidence level of 90% or above is reached. Before this test was available, not transmitting the Weaver gene meant limiting the Brown Swiss genetic base and the genetic progress of the breed. While not conclusive or informative for every affected lineage, this test has now become a valuable tool to re-open many "closed" pedigree lines which represent some high indexing production and type in the Brown Swiss breed. Through use of this test and its results, a breeder can now select for the most profitable characteristics in Brown Swiss even though the Weaver gene may be present in the bloodlines.

Testing will qualify approximately 50% as non-carriers and give breeders management choices on the other half. An animal that is tested a carrier is no worse than it was assumed to be and its offspring may also be tested. An animal that is tested Weaverfree no longer carries the stigma of the (W) in its pedigree and, thereby, enhances its value and marketability. Descendants from an animal verified Weaver-free will always be Weaver-free unless bred to a carrier.

<u>SMA</u>

GeneControl in Germany originally developed a DNA marker test for SMA to determine SMA carrier status with high reliability. Results with a confidence level of 90% or above were then published. In July 2006, GeneControl found the specific gene for SMA. Animals can now be tested with 100% accuracy for the SMA gene. If you wish to have an animal in your herd gene tested for SMA, please contact the Brown Swiss Association.

SDM and Spiderleg

Because no gene test was available for Spiderleg or SDM, Switzerland and the U.S. have used progeny testing to determine the Spiderleg carrier status of sires. Super-ovulated carrier cows were inseminated with semen from potential carriers and the resulting embryos were implanted in recipients and allowed to gestate for 95-100 days (minimum of 16 embryos needed to get an acceptable reliability), then the fetuses were removed and examined for deformity which would be apparent by that time.

Genomic Testing

Now, in lieu of specific gene tests, the widespread use of genomic testing yields reliable results on haplotypes for Weaver, SMA, SDM, Polled, and BH2. Carrier lists are published on the Brown Swiss website after official genetic evaluation releases three times per year.

E. Declaration of Known Carriers

A registered Brown Swiss animal which has met any of the following conditions shall be declared a known carrier of that genetic defect.

1. One first-generation offspring must be diagnosed by an approved laboratory as abnormal because of a recognized undesirable defect. A second first-generation offspring must be inspected by an Association authorized person and verified to be typical of the undesirable defect. Both affected first-generation offspring must have their parentage verified at a laboratory approved for parentage verification.

2. Three first-generation offspring must be diagnosed by an approved laboratory as abnormal because of the genetic defect.

3. Tested positive by a BSCBA-approved DNA gene test or by genomic testing.

A registered Brown Swiss animal that has completed an approved testing program for a specified recessive factor and has been determined to be a probable carrier or non-carrier of that genetic recessive at the established confidence level shall be so declared.

A registered Brown Swiss animal that has been declared to be a carrier for a specified recessive factor as designated by a foreign Brown Swiss Association and meeting the conditions above shall be declared a known carrier of that genetic defect.

Periodically, a list of known carrier sires will be published on the association website www.brownswissusa.com.

Designation of Abnormality Carrier Status

Name Suffixes are a means to inform breeders of the current status of animals as known carriers or tested probable carriers of genetic abnormalities. Such animals shall be identified as follows:

A. An animal confirmed through diagnosis by an approved laboratory as abnormal due to an inherited undesirable genetic recessive factor or the DNA gene test.

- 1. Confirmed Weaver designated as (CW)
- 2. Confirmed SMA designated as (CM)
- 3. Confirmed Spiderleg designated as (CA)
- 4. Confirmed SDM designated as (CD)

B. Known carriers of undesirable genetic recessive factors1. Known Weaver Carrier designated as (W)

- 2. Known SMA Carrier designated as (M)
- 3. Known Spiderleg Carrier designated as (A)
- 4. Known SDM Carrier designated as (D)

C. An animal determined through genomic testing or an approved marker testing program to be a probable carrier at a confidence level of 90% or above.

- 1. Weaver Tested Carrier designated as (W*)
- 2. SMA Tested Carrier designated as (M*)
- 3. Spiderleg Tested Carrier designated as (A*)
- 4. SDM Tested Carrier designated as (D*)

D. An animal determined through genomic testing or an approved testing program to be a probable non-carrier at a confidence level of 90% or above or the DNA gene test.

- 1. Tested Non-Carrier for Weaver designated as *TW
- 2. Tested Non-Carrier for SMA designated as *TM
- 3. Tested Non-Carrier for Spiderleg designated as *TA
- 4. Tested Non-Carrier for SDM designated as *TD

Note: Name suffixes have been discontinued for Non-carrier status.

Note: Name Suffixes enclosed in parentheses indicate an animal as a carrier or affected with the abnormality. Name Suffixes without parentheses indicate a non-carrier animal. Name Suffixes with an asterisk indicate the results from an approved marker testing program at a confidence level of 90% or above. Name Suffixes without an asterisk indicate carrier status information is from diagnosis of affected animals or a DNA gene test.

Genetic Testing Indicators

Abnormality & Haplotype indicators listed on Registration Certificates & Pedigrees:

WT, MT, DT, PT, BH2T – Indicate Tested Non-carrier for Weaver, SMA, SDM, or Polled, BH2 genes or haplotypes.

WC, MC, DC, PC, BH1C, BH2C – Indicate Tested Carrier for Weaver, SMA, SDM, or Polled, BH2 genes or haplotypes.

NOTE: The following <u>haplotype indicators</u> are listed on some database reports:

BHWC or BHWT (Weaver carrier or tested free); BHMC or BHMT (SMA carrier or tested free); BHDC or BHDT (SDM carrier or tested free); BHPC or BHPT (Polled carrier or tested free); BH2C or BH2T (BH2 carrier or tested free)

Ethics

The Association considers it an unethical practice to offer for sale a registered Brown Swiss animal, semen from an animal, or an embryo from a parent which has been determined through an approved testing program to be a probable carrier or is a known carrier of an undesirable genetic recessive factor, unless such fact is made known to the potential buyer. Identifying such an animal in the pedigree or stating such fact in advertising such animal, semen, or embryo for sale shall be deemed as sufficient notice.

F. Genetics

It is possible for a genetic recessive to be present for many generations without being detected. Due to ET flushing, many cows are having increased numbers of offspring and labeling of the dams is also important.

Animals on the known carrier list still have a place in some breeding programs. Wholesale elimination of carriers is not practical. The decision of whether or not to use a known carrier sire depends on whether the benefits to be gained outweigh the possible consequences of getting carrier or affected offspring from such a mating. Through careful breeding, the number of affected animals can be controlled; and through the use of non-carrier sires, the practical effect of the abnormality can be eliminated over a period of time.

Keeping breeders informed of the genetic nature of recessives and how to breed around them is the key to control of any abnormality in a breed. Making use of genetic and specific pedigree information to make wise breeding choices is essential not only to individual progressive breeding management but also to breed progress as a whole.

Cows have 30 pairs of chromosomes which carry all of the cow's genes. Half of an individual animal's genetic material (30 chromosomes) is received from each parent. Each parent then is only transmitting half of their own genetic material to an offspring. "The luck of the draw" can mean the difference between a carrier or non-carrier of a genetic recessive. Using blue and brown eyes as an example, brown eyes are a dominant factor over blue eyes (a recessive factor). If one parent transmits the gene for brown eyes and the other parent the gene for blue, the offspring will have brown eyes. It would take both parents transmitting the gene for blue eyes (recessive) before an offspring will have blue eyes. The parents may or may not have blue eyes themselves, but would have to be carriers of the recessive gene for blue eyes in order to transmit it to an offspring. In the case of Weaver, SMA, Spiderleg, or SDM, the normal gene is dominant and the abnormal gene is recessive. It would take both parents to be carriers in order to produce an affected offspring. In such case, the odds would be a 25% chance of an affected offspring (homozygous for abnormal gene), a 50% chance of carrier offspring (heterozygous), and a 25% chance of clean offspring (homozygous for normal gene). If only one parent is a carrier, no affected offspring will result; however, there is a 50% chance the abnormal gene will be transmitted to the offspring. It is very important to understand these odds when making breeding choices. (See diagram below).

Don't be stymied by genetic recessives. Take full advantage of the top genetics in your herd by making informed breeding decisions.

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