

## Policies Regarding Undesirable Genetic Factors

*Effective February 22, 1983*

*Revised July 27, 2011, November 2, 2012,*

*August 19, 2013 March 16, 2014, June 24, 2014*

*June 27, 2018, November 2, 2018*

### Statement of Policy

Every effort should be made within the breed to identify those animals that carry undesirable genetic factors. The American Jersey Cattle Association considers it the responsibility and obligation of each member of the Association and each breeder of Jersey cattle to report to the Executive Secretary any known case of an abnormal Jersey animal. The Executive Secretary shall maintain records of abnormalities and shall make available information from such records in accordance with rules established by the Board of Directors from time to time.

The Board of Directors considers it to be unethical practice to offer for sale an animal, male or female, an embryo or semen from an animal that has been designated a carrier of an undesirable genetic factor without first informing the prospective buyer of this fact. In practice this means that any advertising, descriptive material, or pedigree containing a designated carrier of an undesirable genetic factor shall carry a statement indicating designated carriers.

This statement of policy is made in belief that it is in the best interests of the breed and the breeders of Jersey cattle. It is made in the belief that it will serve the position of those who have taken the more difficult, positive, open approach to this fundamental concept of ethics in the improved breeding of dairy cattle. In the long run, all serve to gain by such a policy, but only to the degree that all cooperate in the acceptance and enforcement of this policy.

### Identification of Undesirable Genetic Factors

In determining what genetic factors are considered to be undesirable in the Jersey breed, the Board of Directors shall consider such evidence as it considers appropriate. The Executive Secretary shall make such investigations of genetic factors occurring in Jersey animals as he or she may believe necessary or advisable and shall report the results of his or her investigations to the Board of Directors. Before recommending that the

### Action On Abnormalities

*Excerpted from*

### Bylaws of The American Jersey Cattle Association

Article IV, Section 4. The Executive Secretary shall make such investigations of genetic factors occurring in Jersey animals as he or she may believe necessary or advisable and shall report the results of his or her investigations to the Board of Directors. Each member and non-member of the Association shall cooperate fully in any such investigation. The Board of Directors shall determine what genetic factors are considered to be undesirable in the Jersey breed and shall take whatever action it may consider appropriate to control and limit such undesirable genetic factors.

The collection, compilation and dissemination of information relating to genetic factors is hereby declared to be in the furtherance of the aims and purposes of the Association, the preservation of the high breeding standards of Jersey cattle, and the protection not only of the members of the Association but also of the public generally; and dissemination of such information is therefore, deemed to be qualified privilege. Accordingly, the Association shall publish and release such information in accordance with procedures established by the Board of Directors. In so doing neither the Association, nor its officers, directors, employees, or agents shall be liable for damages or otherwise.

Board make a determination regarding the existence of an undesirable genetic factor, the Executive Secretary shall consult with at least two experts whose recommendations shall be submitted to the Board.

### Identification of Carrier Animals

When the Board of Directors shall determine that an undesirable genetic factor exists in the Jersey breed, the Board shall take whatever action it may consider appropriate to control and limit the genetic factor. Such action will include procedures to identify animals that are probable carriers of the undesirable genetic factor and to inform persons having an interest in the Jersey breed of the identity of such probable carriers. The procedures for publication of the identity of probable carrier animals, referred to as "designated" carriers, are contained in supplemental statements adopted with respect to each undesirable genetic condition.

For each undesirable genetic condition the Board of Directors shall adopt a separate statement of procedures for designating animals, referred to as "Statement of Designation Procedures," and designate an official report form to be used for reporting affected animals. The Board of Directors shall be responsible for designating animals as carriers of an undesirable genetic factor. When an

animal has been designated as a carrier, the Executive Secretary shall notify the last recorded owner, the breeder, any lessee, and any third-party nominator by email on file at American Jersey Cattle Association (AJCA). If email is not on file, results and notification of undesirable genetic factor status are delivered by regular U.S. Mail to the address on file at AJCA. The undesirable genetic factor status may change with updates in technology and additional genomic testing of related animals.

The Board of Directors may adopt procedures and rules by which a Jersey may be progeny tested for a particular undesirable genetic factor. The rules and procedures for progeny testing are contained in supplemental statements adopted with respect to each undesirable genetic condition.

The policy of the Association is to identify and designate Jersey animals as carriers of undesirable genetic factors when (1) genomic detection based on DNA analyses and/or (2) documentation of their own progeny is sufficient to accomplish designation.

### Publication and Release of Information

#### *Male and Female Animals*

The Executive Secretary shall maintain

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a record of all animals that have been designated carriers of an undesirable genetic factor, and designation shall be noted on all advertising, descriptive material, or pedigrees published by the Association containing reference to a designated carrier. The Association shall also notify the recorded owner, the breeder, any lessee, and any third-party nominator if the animal carries an undesirable genetic factor.

Except as provided in this statement of policies, no information concerning the genetic condition of any animal shall be released by the Association without approval of the Board of Directors.

#### *Male Animals Only*

When the Association receives an official report of an affected animal, the Executive Secretary shall so inform the last recorded owner, the breeder, and the lessee, if any, of the sire of such an animal by regular mail; and thereafter he or she shall routinely inform the breeder, the owner, and the lessee of that sire of the receipt of any additional reports and supporting documentation required by the applicable Statement of Designation Procedures and of such additional information as he/she shall deem appropriate.

When the Association receives an official report that an animal is affected with an undesirable genetic condition and that report is accompanied by the documentation required by the applicable Statement of Designation Procedures and if the sire of the animal described in the report is the son or grandson of a designated carrier or an affected animal, the Executive Secretary shall prepare a statement containing information as he or she, in his or her sole discretion, shall deem appropriate with respect to such sire. Such statement shall be furnished to the last recorded owner, the breeder, and the lessee, if any, of the sire and to any AI organization that is currently offering for sale semen obtained from the sire. The statement shall also be furnished to any other person who requests information from the Association concerning the sire.

The identification of bulls designated as carriers of undesirable genetic factors shall be published in the *Jersey Journal* from time to time in such manner as the Executive Secretary shall determine.

#### **Sales**

For each undesirable genetic condition

the Board of Directors shall establish policies for the marketing of carrier animals and their progeny in all sales sponsored or managed by the Association or Jersey Marketing Service.

#### **Limber Legs (LL)**

##### **Statement of Designation Procedures**

*Designated November 1972*

##### *Description of Condition*

The affected calf has little or no control over movement of legs and is unable to stand. The calf's legs lack normal muscling, appear loose at the joints, and can be flexed, extended and rotated without difficulty or discomfort to the calf. Usually the legs can be crossed above the dorsal side of the neck without discomfort to the calf.

##### *Requirements for Designation of Limber Legs Carrier*

The Board of Directors will not designate an animal a carrier of Limber Legs if the Board considers that there is a reasonable doubt that the animal is a carrier. The determination as to reasonable doubt depends upon the quality and amount of available evidence which will vary in each case.

Usually, the Board will designate a carrier bull without the concurrence of the breeder, last recorded owner, and the lessee, if any, upon receipt of two or more official reports that calves sired by that bull are affected with Limber Legs if (1) the calves reported on were born in two separate herds and (2) each report is accompanied by the written statement of a veterinarian or other person (other than the person signing the official report) who, in the opinion of the Executive Secretary, is qualified to identify the Limber Legs condition describing the condition of the calf and stating whether, in his or her opinion, the calf is affected with Limber Legs. However, with the concurrence of the breeder, the last recorded owner, and the lessee, if any, the Board will usually designate a carrier bull upon receipt of one or more official report(s) that a calf sired by that bull is affected with Limber Legs if each report is accompanied by a written statement by a veterinarian or other qualified person and if a parent or any grandparent of the bull has been designated a Limber Legs carrier.

Usually the Board will designate a carrier cow upon receipt of one official report that a calf delivered by that cow is affected with Limber Legs if the report

is accompanied by the requisite written statement of a veterinarian or other qualified person.

An extended (six generation) pedigree of the affected animal will be prepared to see if the animal traces to designated carrier animals.

With respect to all sales sponsored or managed by the Association or Jersey Marketing Service, a bull shall not be accepted unless the probability of his being a carrier of Limber Legs, based on his relationship to affected animals or designated carriers, is less than 12.5%.

Daughters of designated carriers of Limber Legs shall not be accepted in any consignment sale managed by Jersey Marketing Service. Granddaughters of designated carriers shall not be accepted in the National Heifer Sale, the Pot O'Gold Sale or The All American Sale but may be accepted in other consignment sales managed by Jersey Marketing Service.

#### **Rectovaginal Constriction (RVC)**

##### **Statement of Designation Procedures**

*Designated June 1975*

##### *Description of Condition*

Rectovaginal Constriction (RVC) is constriction of the rectum and vagina such that the arm usually may not be inserted normally in the rectum to permit artificial breeding. An episiotomy or Caesarian section is usually required for calving. The condition may be accompanied by hardening of the udder.

##### *Requirements for Designation of Rectovaginal Constriction Carrier*

The Board of Directors will not designate an animal as a carrier of Rectovaginal Constriction (RVC) if the Board considers that there is a reasonable doubt that the animal is a carrier. The determination as to reasonable doubt depends upon the quality and amount of available evidence which will vary in each case.

Usually, the Board will designate a carrier bull without concurrence of the breeder, last recorded owner, and the lessee, if any, upon receipt of two or more official reports that, calves sired by the bull are affected with RVC if (1) the calves reported on were born in two separate herds; (2) the bulls relationship to the calves is supported by blood typing; and (3) each such report is accompanied by the written statement of a veterinarian or other person (other than the person signing the official report) who, in the opinion of

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the Executive Secretary, is qualified to identify the RVC condition describing the condition and stating whether in his or her opinion the calf is affected with RVC. However, with the concurrence of the breeder, the last owner, and the lessee, if any, the Board will usually designate a carrier bull upon receipt of one or more official report(s) that a calf sired by that bull is affected with RVC if each report is accompanied by blood typing to verify parentage and by written statements of a veterinarian or other qualified person and if a parent or grandparent of the bull has been designated an RVC carrier or is affected with RVC.

Usually, the Board will designate a carrier cow upon receipt of one official report that a calf delivered by that cow is affected with RVC if the report is accompanied by blood typing to verify parentage and by the requisite written statement of a veterinarian or other qualified person.

An extended (six generation) pedigree of the affected animal will be prepared to see if the animal traces to affected or designated carrier animals.

With respect to all sales sponsored or managed by the Association or Jersey Marketing Service, a bull shall not be accepted unless the probability of his being a carrier of Rectovaginal Constriction, based on his relationship to affected animals or designated carriers, is less than 12.5%.

Daughters of designated carriers of Rectovaginal Constriction shall not be accepted in any consignment sale managed by Jersey Marketing Service. Granddaughters of designated carriers shall not be accepted in the National Heifer Sale, the Pot O'Gold Sale or The All American Sale but may be accepted in other consignment sales managed by Jersey Marketing Service.

## **Jersey Haplotype 1 (JH1) Statement of Designation Procedures** *Designated June 2011 Revised November 2011, August 2013*

### *Description of Condition*

Jersey Haplotype 1 (JH1) designates a reduction in fertility, specifically early embryonic loss, attributable to a specific haplotype on *Bos taurus* autosome 15 (*BTA15*). Subsequent investigation identified the biological cause of embryo loss as a loss-of-function mutation in the

*CWC15* spliceosome-associated protein homolog gene (Sonstegard et al., 2012, *PLoS ONE*, 8:354872).

### *Determination of JH1 Status*

The Board of Directors will not designate an animal either a carrier or free of JH1 haplotype if the Board considers that there is a reasonable doubt that the animal is a carrier. The determination as to reasonable doubt depends upon the quality and amount of available evidence which will vary in each case.

Official JH1 status is obtained (1) by direct observation of the loss-of-function (LOF) mutation in *CWC15* in the genotype of an animal, or (2) by use of LOF mutation test results of family members to determine if the mutation is contained in the JH1 haplotype of other genotyped animals, and reassessed as additional information is obtained from DNA analyses.

With respect to all sales sponsored by the Association, a bull shall not be accepted unless designated free of Jersey Haplotype 1.

## **Bovine Leukocyte Adhesion Deficiency (BLAD) Statement of Designation Procedures** *Designated March 2014*

### *Description of Condition*

Bovine Leukocyte Adhesion Deficiency (BLAD) is a genetic defect affecting white blood cell function which causes extreme susceptibility to infection. The white blood cells of the affected animal fail to attach to the cells lining the blood vessels, an essential step in their migration to the point of infection to destroy invading pathogens. BLAD carriers have a mutation in the *CD18* gene (Shuster et al., 1992, *Proceedings of the National Academy of Sciences of the United States of America*, 89:9225-9229).

### *Determination of BLAD Status*

The Board of Directors will not designate an animal a carrier of BLAD if the Board considers that there is a reasonable doubt that the animal is a carrier. The determination as to reasonable doubt depends upon the quality and amount of available evidence which will vary in each case.

BLAD carrier status is determined by observation of the mutation at position 383 of the *CD18* gene in the genotype of an animal.

With respect to all sales sponsored

by the Association, no animal shall be accepted that is a designated carrier of Bovine Leukocyte Adhesion Deficiency.

## **Deficiency of Uridine Monophosphate Synthase (DUMPS) Statement of Designation Procedures** *Designated March 2014*

### *Description of Condition*

Deficiency of Uridine Monophosphate Synthase (DUMPS) is a genetic defect that causes failure in the synthesis of DNA and results in early embryonic death of homozygous offspring. DUMPS carriers have a mutation with loss of an Aval site at codon 405 (Schwenger et al., 1993, *Gene Genomics* 16:241-244).

### *Determination of DUMPS Status*

The Board of Directors will not designate an animal a carrier of DUMPS if the Board considers that there is a reasonable doubt that the animal is a carrier. The determination as to reasonable doubt depends upon the quality and amount of available evidence which will vary in each case.

DUMPS carrier status is determined by observation of the mutation and the loss of an Aval site at codon 405 in the genotype of an animal.

With respect to all sales sponsored by the Association, no animal shall be accepted that is a designated carrier of Deficiency of Uridine Monophosphate Synthase.

## **Spinal Muscular Atrophy (SMA) Statement of Designation Procedures** *Designated March 2014*

### *Description of Condition*

Bovine Spinal Muscular Atrophy (SMA) is a neurodegenerative genetic disease that typically occurs at three (3) to four (4) weeks of age as weakness of the rear legs. Terminal stages are marked by severe muscular atrophy, weakness in all four limbs and inability to stand. The causative gene SMN has been mapped at the very distal end of *BTA24* (Krebs et al., 2007, *Proceedings of the National Academy of Sciences of the United States of America*, 104:6746-51).

### *Determination of SMA Status*

The Board of Directors will not designate an animal a carrier of SMA if the Board considers that there is a reasonable doubt that the animal is a carrier. The determination as to reasonable doubt depends upon the quality and amount of

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available evidence which will vary in each case.

SMA carrier status is determined by observation of the causative gene SMN on *BTA24* in the genotype of an animal.

With respect to all sales sponsored by the Association, no animal shall be accepted that is a designated carrier of Spinal Muscular Atrophy.

## **Bovine Spinal Dysmyelination (SDM) Statement of Designation Procedures** *Designated June 2018*

### *Description of Condition*

Bovine Spinal Dysmyelination (SDM) is a neurodegenerative genetic disease manifested immediately at birth. The homozygous recessive affected calf cannot stand with symptoms that include lateral recumbency with slight to moderate opisthotonos, body tremor, and spastic extension of the limbs. Attempts to rise and limb movements are absent; however, the animals remain alert to their surroundings, and spinal reflexes are normal or slightly increased. SDM carriers have an R560Q substitution with loss of function (Thomsen et al., 2010 *May, Neurogenetics*, 11(2): 175–183).

### *Determination of SDM Status*

The Board of Directors will not designate an animal a carrier of SDM if the Board considers that there is a reasonable doubt that the animal is a carrier. The determination as to reasonable doubt depends upon the quality and amount of available evidence which will vary in each case.

SDM carrier status is determined by observation of an R560Q substitution at a position in the ATPase domain of the Spastin protein in *Bos taurus* *BTA11* in the genotype of an animal.

With respect to all sales sponsored by the Association, no animal shall be accepted that is a designated carrier of Bovine Spinal Dysmyelination.

## **Holstein Haplotype 1 (HH1) Statement of Designation Procedures** *Designated November 2018*

### *Description of Condition*

Holstein Haplotype 1 (HH1) designates a reduction in fertility attributed to a nonsense mutation in the apoptotic peptidase activating factor 1 (APAF1) on *Bos taurus* autosome 5 (*BTA5*). Because functional APAF1 peptide is required for embryo development, homozygosity for

this allele results in natural spontaneous abortion (Adams et al., *Journal of Dairy Science*, 99: 6693-6701).

### *Determination of HH1 Status*

The Board of Directors will not designate an animal a carrier of HH1 if the Board considers that there is a reasonable doubt that the animal is a carrier. The determination as to reasonable doubt depends upon the quality and amount of available evidence which will vary in each case.

HH1 carrier status is determined by observation of the causative mutation that maps to chromosome *BTA5*.

With respect to all sales sponsored by the Association, no animal shall be accepted that is a designated carrier of Holstein Haplotype 1.

## **Holstein Haplotype 3 (HH3) Statement of Designation Procedures** *Designated November 2018*

### *Description of Condition*

Holstein Haplotype 3 (HH3) designates a reduction in fertility attributed to a non-synonymous SNP (T/C) within exon 24 of the Structural Maintenance of Chromosomes 2 (SMC2) on Chromosome 8 at position 95,410,507 (UMD3.1). The single base pair change renders SMC2 ineffective in its essential role of DNA repair, chromosome condensation and segregation during cell division. Homozygosity results in spontaneous abortion in early gestation (McClure et al., 2014, *PLoS One* 9:e92769).

### *Determination of HH3 Status*

The Board of Directors will not designate an animal a carrier of HH3 if the Board considers that there is a reasonable doubt that the animal is a carrier. The determination as to reasonable doubt depends upon the quality and amount of available evidence which will vary in each case.

HH3 carrier status is determined by observation of the causative mutation that maps to chromosome *BTA8*.

With respect to all sales sponsored by the Association, no animal shall be accepted that is a designated carrier of Holstein Haplotype 3.

## **Holstein Haplotype 4 (HH4) Statement of Designation Procedures** *Designated November 2018*

### *Description of Condition*

Holstein Haplotype 4 (HH4) designates a reduction in fertility attributed to

a missense mutation (g.1277227A.C; UMD 3.1 genome assembly) in the GART gene which encodes glycinamide ribonucleotide transformylase located on *Bos taurus* autosome 1 (*BTA1*). Loss of GART function is expected to cause embryonic death at early stages of embryonic life. Homozygosity results in spontaneous abortion in early gestation (Fritz et al., 2013, *PLoS One*, 8: e65550).

### *Determination of HH4 Status*

The Board of Directors will not designate an animal a carrier of HH4 if the Board considers that there is a reasonable doubt that the animal is a carrier. The determination as to reasonable doubt depends upon the quality and amount of available evidence which will vary in each case.

HH4 carrier status is determined by observation of the causative mutation that maps to *BTA1*.

With respect to all sales sponsored by the Association, no animal shall be accepted that is a designated carrier of Holstein Haplotype 4.

## **Holstein Haplotype 5 (HH5) Statement of Designation Procedures** *Designated November 2018*

### *Description of Condition*

Holstein Haplotype 5 (HH5) designates a reduction in fertility attributed to a deletion of 138kb, spanning position 93,233kb to 93,371kb on *Bos taurus* 9, harboring only dimethyl-adenosine transferase 1 (TFB1M). TFB1M is essential for synthesis and function of the small ribosomal subunit of mitochondria. Homozygosity results in spontaneous abortion in early gestation (Schütz et al., 2016, *PLoS One*, 11(4): e0154602).

### *Determination of HH5 Status*

The Board of Directors will not designate an animal a carrier of HH5 if the Board considers that there is a reasonable doubt that the animal is a carrier. The determination as to reasonable doubt depends upon the quality and amount of available evidence which will vary in each case.

HH5 carrier status is determined by observation of the causative mutation that maps to *BTA9*.

With respect to all sales sponsored by the Association, no animal shall be accepted that is a designated carrier of Holstein Haplotype 5.