

## World Holstein-Friesian Federation (WHFF)

### Harmonization and Exchange of Genetic Recessives

The World Holstein Friesian Federation (WHFF) has reviewed the recording of Genetic Recessives prevalent in the Holstein breed today as a positive approach and with a view of harmonization and exchange.

Since there is an increasing concentration of breeding within bloodlines and continued widespread use of a limited number of sires, it is fundamental to the advancement of the breed that all available information, both desirable and undesirable, be made known to breeders, to potential purchasers of animals or to purchasers of artificial service from sires.

The full disclosure of named genetic defects / recessives in the Holstein population is useful information in making breeding decisions on farm, allows the breed an opportunity to minimize the impact of any associated problems and to breed around / eliminate it from the breed.

Animals of any breed or species may possess undesirable inherited factors which may or may not exhibit themselves in the animal itself, but which may be transmitted to its offspring. Transmission of such a factor may take place for a considerable period of time from one generation to another before the factor manifests itself in an identifiable form. By that time, the factor can be widely seeded as a recessive gene in many herds and thousands of animals without breeders being aware of the potential loss that they might suffer. Such a situation might have an adverse effect upon their breeding programs and the future of their herds.

It is to be expected, that due to improved data collection and progress in genetic investigation of traits an increasing number of genetic recessives will be detected and/or published. Many of them will be restricted in their value to limited consumer markets or environments.

#### **Initial Phase of Discovery and Research of a New Recessive**

The first step in the recognition process of a new recessive is the identification of the condition. Once the condition is reported a series of facts should be gathered so that an initial evaluation of the condition can be made.

Has evidence been provided that establishes the presence of the condition in the cattle population?

- Is the mutation within germinal or somatic cells?
- What breed or breeds are involved?
- Is the condition lethal and at what time in life is the condition expressed?
- What proportion of the population may be affected?
- Are similar conditions known in other species?
- Are tests currently available to detect the condition and if so what do they cost?

Are the tests definitive and if not what are their limitations?

## **Secondary Phase of Research**

The secondary phase of research begins when some sort of diagnostic test becomes or is available. Preliminary testing of individuals is a necessary part of the research process and will allow the researchers to more clearly:

- Establish the physiology of the condition
- Determine the mode of inheritance
- Determine the economic consequences
- Establish an accurate, rapid and inexpensive diagnostic test, if needed

Recommend procedures for developing controls, if needed.

### ***Classification of the Condition***

- Class 1 Of lethal proportions with manifestation and diagnosis occurring just prior to entering productive life
- Class 2 Of lethal proportions, but identified at birth or early in life
- Class 3 Lethal in the early embryonic stages or not lethal but seriously impairs production or reproduction
- Class 4 Not lethal but has a small effect on production or reproduction; economically insignificant
- Class 5 No known impairment on production or reproduction

### **Public Disclosure with Discretion**

Once the preliminary testing of individuals is completed and some facts about the gene frequency and economic implications have been established, results from all of the preliminary testing should be released to all bull controllers and the appropriate breed organization or designee. This is a very sensitive period and great care should be taken so that control programs are implemented without creating undue alarm throughout the industry.

It seems responsible to keep test results confidential until research has documented the accuracy of the test and the significance of the condition so that the tests results can be properly interpreted. However, withholding tests results for an extended period of time puts organizations at some liability risk for continuing to market a product that they were aware was defective. Since the chronology of events will vary by condition, the proper timing for releasing test results will be a decision that will need to be made separately for each condition. Complete public disclosure of the condition and identification of carriers should not occur until either the manuscript has been accepted for publication or some expert review has been formalized.

### **Guidelines for Adding to WHFF Master List**

Principally, each country should be free to include as many genetic recessives as desired into their official national pedigrees. However, for official international exchange of pedigrees a master list is necessary for those genetic recessives, which obligatory have to be included into the pedigree when the individual is tested for it.

The phenotypic description of the trait and proof of its inheritance are obligatory requirements for adding a genetic recessive to the WHFF master list. Additionally, the procedure of testing has to be listed. Testing can be done by informative mating, laboratory test of phenotypes or genetic tests of causative genomic variation. Wherever available detection of the causative genetic mutation of a recessive should be preferred.

To enable critical review of this information, publication of these data in a peer reviewed international scientific journal is preferred. Equivalent to this is a publicly available international patent file. For each genetic recessive a catalogue comprising detailed, published description of phenotype, inheritance and detection should be established.

In the case of inconclusive test results, records must be maintained by the responsible breed association (s). It would not be prudent to label an animal a carrier as the result of a test that may not be valid, thus negatively affecting the value of the animal.

The Board of Directors and/or each national Holstein Association reserve the right to define the nature, type and form of the information that is accepted, produced and retained on file by the respective Association.

### **Exchange of Genetic Recessives**

An electronic exchange mechanism is required for the efficient and effective exchange of carrier and tested free animals and the corresponding recessive code to all WHFF herdbooks / societies on a timely and/or or as needed basis. This work is being addressed by the WHFF-EDE Working Group.

Harmonization of codes and nomenclature is imperative for overall accuracy, assignment and a prerequisite for electronic data exchange. With each breed association maintaining its own file instead of having one central master file, the exchange process as proposed is viewed as more practical and less vulnerable to misinterpretation or error. Survey respondents indicate their willingness to the publishing of recessive codes and animals on their respective websites and are willing to transmit electronic files containing the identification of carrier and tested free animals.

However Dr. Christa Kuehn, Germany and WG member, re-opinions that in most businesses global networks are set up, but in Holstein breeding we propose a structure, which may cause extra work and a reduction of data quality due to repetitive bilateral data plausibility checks and updates. Further thought can be given to a central database for those carefully selected defects on the master list, which are mandatory for recording in each population? Localisation of a recessive repository database at Interbull would be in line with a central database on performance traits there and would overcome many problems concerning identification problems for individuals. \*Council has agreed to explore possibilities with Interbull and ICAR.

### **Master List of WHFF Approved Recessives**

The list is in keeping with EDE WG and its Appendix 4. The gene and corresponding expression codes have been modified and expanded in keeping with WHFF General Assembly 2004 encouragement of a coding system for exchange that was simple, expandable and easy to use, interpret and understand.

### **Toward Harmonization (Re-confirmation)**

Gene codes and/or gene expression codes should not be part of an animal's name but rather utilized as a secondary or separate code or field. This approach facilitates the accurate exchange of information and negates the need to rename or amend names initially penned to animals given assignment of singular or multiple recessive codes.

It is recognized that new or changes to named recessives and expression codes might be difficult to implement by all member herdbooks / societies / countries. As such, each herdbook / country reserves the right to define the nature, type and form of information that is accepted, published and retained within its jurisdiction. However, each participating herdbook is expected

to comply with the WHFF gene codes and expressions (appendix 4) for the purposes of international data exchange of recessives.

Additional or new recessives / traits not currently provided for on the master list or pending approval by WHFF Council should not be expression coded nor exchanged prior recognition / consensus of WHFF Council.

Herdbooks / societies / countries with experience or particular interest should be encouraged and invited to initiate or lead submission to WHFF Council for recognition and approval of new recessives / defects (Factor XI, Polled and Bulldog for example) in keeping with steps of the program previously described.