Standardized Labelling for Genetic Trait Coding

The World Holstein Friesian Federation registration working group reviews the recording of Genetic Traits prevalent in the Holstein breed with emphasis on harmonization and exchange of data. The term 'Genetic Trait' is used to describe a monogenetic inherited trait, i.e., one that is simply inherited by a single gene. Official Genetic traits for the Holstein breed are listed on the WHFF website for easy reference for all International Holstein Association and their respective Herdbooks. When newly observed or previously unknown Genetic Traits are discovered, they should be reported to WHFF for the classification.

The full disclosure of named Genetic Traits in the Holstein population is very useful information when making breeding decisions on the farm. It allows farmers to minimize the impact of any associated problem by breeding around, through careful mating decisions, to eliminate the harmful expression of the genetic trait.

It is strongly recommended that Genetic Traits be reported on breed Herdbook official documents and made available for data exchange. Harmonization of codes and nomenclature is imperative for overall accuracy and international data exchange.

For the most part, genetic traits do not become 'Herdbook official' until the results of a direct genetic test for the causal variant are available. Today, we are also able to acquire genetic trait information from identified regions in the genome associated with a genetic trait, by indirect testing (e.g. via haplotypes). Labelling of these test types (direct and indirect) provides breeders with the information and opportunity to calculate the risk with mating decisions.

WHFF recommended by their Registration Working Group (WG) proposes the following standardized labelling for genetic trait coding to indicate gene name and expression code.

Gene Name

Two (2) Alpha characters are to be assigned to represent the gene name and should be associated with the description of the gene or its primary function. The characters should be proposed by the researcher/countries that discovers the new genetic trait. WHFF will adopt this two Alpha character label, if no severe or technical reason stands against it.

The naming of 'indirect / haplotype discovered traits' relates to the name of the genetic condition (as known) and, in some cases, may change (e.g. missing haplotypes) when the specific phenotype or genetic information of the trait is discovered. Previously labelled traits will not be renamed e.g. HH1.

Expression Codes

To be used following the WHFF adopted two (2) Alpha characters assigned for the monogenetic inherited trait. There are expression codes for direct and indirect gene tests to facilitate the differentiation between both testing types.

Direct Tests		Indirect Tests	
F	Tested Free	0	Tested Free/non-carrier.
C	Tested Carrier / Heterozygous	1	Tested Carrier/Heterozygous/Confirmed with pedigree info.
S	Tested / Homozygous	2	Tested True/Homozygous/Confirmed on both sides of pedigree.
		3	Additional Characteristics e.g. suspect carrier origin could not be confirmed from pedigree.
		4	Additional Characteristics e.g. suspect homozygous origin could not be confirmed from pedigree.
		5	As required should an additional characteristic be identified.

Example of direct test codes below:

Cholesterol Deficiency	**CDF** = tested non-carrier / free of cholesterol deficiency **CDC =** tested carrier of cholesterol deficiency (heterozygous) **CDS** = tested true carrier of cholesterol deficiency (homozygous)

Timing for labelling

When the WHFF is advised by industry partners and/or laboratories of a newly discovered genetic trait, there will be a four week time period before delivery of the standardize label for coding by the WG.

Gene tests differences

To help understand some of the key differences between gene test types, here are some descriptors. Due to the various technical, scientific and constantly evolving means to complete testing, these descriptors are general and to be used as a guide only.

About Direct gene test:

- reliability: very close to 100%, excluding technical errors / issues
- are marker-based tests
- result from presence of mutated allele

About Indirect gene test:

- reliability: very high, can be as high as 98%
- risk of false positive or false negative results
- does not detect causal allele itself; detects proxy of causal allele

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