Standardised Labeling for Genetic Trait Coding

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on behalf of the WHFF Registration Working Group

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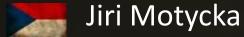
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World Holstein Friesian Federation Objectives

- *Harmonise* technical and administrative matters with regard to the Holstein Breed

- *Represent* the common interest of breeders worldwide in developing and promoting the Holstein Breed

- *Exchange Information* on important issues concerning the breed

- *Assist* emerging herdbook associations

- *Co-operate* with Research Centres and other recognised International Organisations involved in animal improvement
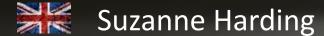
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Assigned Tasks

- Harmonize a process for the categorization / recognition of major recessives.

- Standardize labeling nomenclature and codes – tested carrier, tested free and not conclusive.

- Publishing haplotypes and standardize the way they are reported.

Why Standardise?

- Easy for all organisations to reference the same Genetic Traits

- Useful on farm when making breeding decisions
  - Farmers can choose to minimise the impact or increase the likelihood of the effect

- Recommended to report on official documents

- Harmonization of codes and nomenclature is imperative for overall accuracy and international data exchange.

Genetic Trait Names

Gene Name	Description	Gene and Expression Code
BLAD	**Bovine Leukocyte Adhesion Deficiency** (deficiency of a normally occurring protein needed for white blood cells or leukocytes, which are body's infection fighters)	**BLC** = tested carrier of BLAD **BLF** = tested non-carrier of BLAD
Mule foot	**Mule-Foot** (toes of foot are joined, giving animal a single hoof, instead of cloven ones)	**MFC** = tested carrier of Mule foot **MFF** = tested non-carrier of Mule foot
DUMPS	**Deficiency of Uridine Monophosphate Synthase** (one of many enzymes contributing to normal metabolic processes)	**DPC** = tested carrier of DUMPS **DPF** = tested non-carrier of DUMPS
CVM	**Complex Vertebral Malformation** (causes still-born calves, abortions, and early embryonic losses)	**CVC** = tested carrier of CVM **CVF** = tested non-carrier of CVM
Factor X1	**Factor X1** (blood clotting disorder)	**XIC** = tested carrier of Factor X1 **XIF** = tested non-carrier of Factor X1
CIT	**Citrullinemia** (accumulation of ammonia and other toxics in blood in baby calves)	**CNC** = tested carrier of Citrullinemia **CNF** = tested non-carrier of Citrullinemia
	Brachyspina (causes abortion and	**BYC** = tested carrier of Brachyspina

Genetic Trait Names

Coat Colour Carrier Gene	Description	Gene and Expression Code
Red	Red gene	RDC = carrier of red gene RDF = tested non-carrier of red gene
Red	Variant Red gene	VRR = not tested/determined by lineage VRS = tested true (homozygous) VRC = VRC =tested carrier (heterozygous) VRF = tested free
Black/Red	Black/red gene	BRC = carrier of black / red gene
Black	Black gene	BKC = carrier of black gene

Gene Test Differences

- Direct gene test:
  - reliability: very close to 100%, excluding technical errors / issues

  - are marker-based tests

  - result from presence of mutated allele

- Indirect gene test:
  - reliability: very high, can be as high as 98%

  - risk of false positive / false negative results

  - does not detect causal allele; are looking for alleles in close proximity to causal nucleotide / genome variation

An Example of a direct test code

- Cholesterol Deficiency

  o Originally indirect test

  o Causal mutation found so direct test available

  o Naming can be completed

An Example of an indirect test code

- Haplotypes?

  o Only indirect test available for HH2 - Still looking for causal mutation

  o HH1, HH3, HH4 now have direct tests

  o HH5 recently detected

Expression Codes

- For many years WHFF has adopted two Alpha characters assigned for monogenetic inherited traits

- New proposal will facilitate the differentiation between direct and indirect traits

- Codes to be used following the WHFF two (alpha) characters

- No previously named traits will be renamed

- Naming of traits will continue to evolve as research continues

Expression Codes

- Codes to be used following the WHFF two (alpha) characters assigned for the monogenetic trait.

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.

An Example of naming a direct test code

- 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)

An Example of naming an indirect test code

- Currently there are no common codings of indirect tests using the WHFF recommended nomenclature

- Each country has named using their own coding

Reporting Procedure

- Industry partner advises WHFF that there is a newly discovered Genetic Trait

- Fill in 'Request for information' form
  - Describe new genetic trait
  - What is the evidence
  - Who is reporting
  - When was it first observed?
  - Which animal / family was it observed in
  - Additional information
  - Contact details

- Send back to WHFF (worldholstein@gmail.com)

- Four weeks later the WG will deliver the standardised label for coding.

BUT…..

- Practical problem of informing all herd books of genetic codes

- Proposal from WHFF President Jos Buiting to ICAR for better dissemination of Bulls genetic codes

- New procedure:
    - Every herd book sends genetic codes for all bulls to Interbull when send evaluation data
    - Interbull can then send this data back to members
    - Could link in with plans to add data to the Interbull data exchange

- Procedure standardized Internationally

- ICAR / Interbull considering proposal

Conclusions

- Important to promote nomenclature to scientific community

- Talk to ICAR with regard to proposing new Guidelines

- Encourage reporting of new genetic traits

- Communicate new genetic traits

- Harmonisation reduces farmer and industry confusion

- Farmer can choose to use when breeding

- WHFF proposal to ICAR for automatic data dissemination

- www.whff.info for full list of Holstein Genetic Traits

Thank you for your Attention.

Any Questions ?