

# Genetic and Genomic Evaluation of Health Traits in Cattle Breeding: A Review

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This review aims to highlight the importance of genetic evaluation of health traits. In the past, cattle breeding primarily focused on milk production traits, but nowadays, there is a shift towards considering health traits and longevity. Cows are mainly affected by four diseases: mastitis, claw diseases, metabolic diseases, and reproductive diseases. These ailments negatively impact milk production, impair reproduction, and incur high treatment costs. In 2012, the ICAR implemented guidelines for the genetic improvement of health traits, establishing a system for recording diseases and their key indicators. This system includes a comprehensive key (>900 diagnoses), a reduced key (60 to 100 diagnoses), and a simple key (10 diagnoses). Genetic evaluation of health traits started in the mid-70s in Nordic countries. Previously, Czechoslovakia had a system for evaluating and classifying the overall health of breeding cows and bulls. In Slovakia, genetic evaluations for milk production traits, somatic cell score, calving ease, and longevity are performed. However, these traits are not included in the selection index along with health traits, for which genetic evaluations are not made. Two selection indexes are currently used: the Slovak Holstein Index, which takes into account milk production traits and type traits at 60% and 40% respectively, and the Slovak Production Index, which takes into account milk production in kilograms, fat production in kilograms, and protein production in kilograms.

**Keywords:** health traits, genetic evaluation, genomic evaluation

## 1 Introduction

Long-term breeding efforts focused on increasing milk production have reduced fertility, increased health problems, and higher culling levels, ultimately leading to a shorter lifespan (Siatka et al., 2020). It is possible to influence the health of animals genetically, either directly by measuring the incidence of disease and selecting for it or indirectly by selecting for genetically correlated traits (Kašná, 2019b). Genetic evaluation of health has a long tradition in certain countries; it has been integrated into breeding programs in Scandinavian countries since the mid-1970s. Austria and Germany commenced genetic evaluation in 2006, while France initiated genetic evaluation for clinical mastitis in 2010 (Vukasinovic et al., 2022).

### 1.1 Longevity, Culling of Cows on the Farms

Longevity refers to the length of time cows remain in the herd. It is a complex trait influenced by various internal factors such as lactation, health, conformation,

and reproductive performance, as well as external factors such as milk price, nutrition, management, and heifer replacement (Hu et al., 2021). Dairy cow longevity is also connected with the economic performance of the farm, the environmental footprint of the milk industry, and the welfare of animals (Dallago et al., 2021). The longevity of a cow can be measured by lifespan (the time from birth to culling), by the length of productive life (the time from first calving to culling), and by survival to a certain age or the number of calvings or lactations (Sawa & Bogucki, 2010). The natural lifespan of dairy cows is approximately 20 years, while the average productive lifespan varies between 2.5 to 4 years (De Vries & Marcondes, 2020). Rostellato et al. (2022) observed a slightly decreasing length of productive life for Holstein Italian cows over the last 15 years. In Canada and Tunisia, the length of productive life is 3.1 years and 3.41 years, respectively (Sdiri et al., 2023; Warner et al., 2022). In Slovakia, the average length of life for cows is 7.1 years, and the length of productive life is 4.3 years (Mézsáros

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et al., 2006). A similar length of productive life is 898 days (2.46 years) was observed for the Holstein breed, compared to 985 days (2.69 years) for Slovak Spotted cattle in the study by Strapáková et al. (2019). Culling is a process in which cows are removed from herds. It is influenced by different internal (health, milk yield, reproductive status) and external factors (milk yield, land availability, price, and replacement of heifers). Voluntary culling aims to improve the breeding and functional value of the herd; involuntary culling is connected with the prevalence of diseases and sudden deaths of animals (Dziekiewicz-Mrugasiewicz & Wierzbicka, 2020). Rilanto et al. (2020) observed that cows were mostly culled due to claw diseases (26.4%), mastitis (22.6%), metabolic diseases (18.1%), and reproductive problems (12.5%). Similarly, Boujenane (2017) found that the most common reasons for culling were reproductive disorders (36.0%), death (12.7%), mastitis (7.7%), and lameness (3.7%). In the study by Armengol and Fraile (2018), culling rates ranged between (2.4%) for locomotion disorders (7.2%) for metabolic diseases, and (30.2%) for reproductive disorders.

### 1.2 Genetic Parameters of Health Traits

Mastitis is a low heritable trait. According to Costa et al. (2019) heritability for mastitis ranges from 0.02 to 0.05–0.08 (Shabalina et al., 2020); for clinical mastitis, from 0.04–0.047 (Zavdilová et al., 2020) to 0.10 (Krupová et al., 2019) and for subclinical mastitis 0.10 (Uribe et al., 2022). Strapáková et al. (2016) reported a heritability of 0.212 for somatic cell count (SCC) from the Slovak national genetic evaluation. Mastitis negatively influences various production traits and health traits. Ajose et al. (2022) reported that cows affected by mastitis had shorter lactation periods, by about 57 days, and produced around 375 kg less milk during one lactation. There exists a relationship between mastitis and other health traits. Various genetic correlations have been observed between mastitis and different health traits. The lowest genetic correlation, 0.20, was observed between clinical mastitis and retained placenta (Koeck et al., 2015). For clinical mastitis and milk yield, lower correlations of 0.26 were found for Normande and Holstein breeds compared to 0.30 for the Montbeliarde breed (Govignon-Gion et al., 2016). Higher correlations were reported between mastitis and fertility (0.55) (Abdelsayed et al., 2017), somatic cell count and mastitis (0.62) (Pritchard et al., 2013). The highest correlation was found between clinical mastitis and longevity (0.63) (Pfeiffer et al., 2015).

Claw diseases can be divided into two groups: non-infectious (white line disease, sole ulcer, sole hemorrhage, interdigital hyperplasia) and infectious (digital dermatitis, interdigital dermatitis, heel erosion, and interdigital

phlegmon) (Garvey, 2023). For digital dermatitis, a low coefficient of heritability ranging from 0.06 to 0.23 for interdigital phlegmon, from 0.01 to 0.18, for sole ulcer from 0.04 to 0.18, for interdigital hyperplasia from 0.01 to 0.12 and white line disease, from 0.01 to 0.13 were reported (Gernand et al., 2012; Charfeddine et al., 2018; Oliveira Junior et al., 2021; Ødegård et al., 2013; Pérez-Cabal and Charfeddine, 2015; Van der Spek et al., 2015). Claw conformation traits were found to be moderately heritable. The highest heritability observed was 0.34 for claw diagonal in Holsteins and 0.37 for both claw width and claw function area in the Slovak Spotted breed, as reported in the study by Chalupková et al. (2023). Krpálková et al. (2019) reported that milk production decreased by around 1.8 kg per day in first-lactation cows affected by claw disorders, with an increase in SCC of 58,000. In second-lactation cows, milk reduction increased to 2.6 kg, but SCC decreased to 45,000. Vlček and Kasarda (2016) reported that first-lactation Holstein cows affected by interdigital dermatitis produced 4 kg more fat and 4 kg more protein but 209 kg less milk. First-lactation Holstein affected by digital dermatitis produced 9 kg more fat and 28 kg more protein. Additionally, a Holstein affected with sole ulcers produced 63 kg more fat and 42 kg more protein. Both negative and positive genetic correlations have been identified among different groups of claw diseases. For instance, high positive correlations of 0.86 (Malchoidi et al., 2020) and 0.81 (Croué et al., 2017) were found between non-infectious sole ulcer and sole haemorrhage diffused. In the study by Malchoidi et al. (2020), a negative correlation of -0.26 was observed between non-infectious white line disease and infectious interdigital dermatitis.

Low heritabilities were reported for metabolic disorders. For milk fever, heritabilities ranged from 0.01 (Kašná et al., 2019a) to 0.10 (Gonzalez-Peña et al., 2020); for ketosis from 0.06 (Vukasinovic et al., 2017) to 0.13 (Shabalina et al., 2020); and for abomasum dislocation, from 0.04 (Oliveira Junior et al., 2021) to 0.08 (Vukasinovic et al., 2017). Milk production decreases from 1.1 to 2.9 kg per day for cows affected by milk fever. Jamrozik et al. (2016) found a higher positive genetic correlation between ketosis and displaced abomasum in first-lactation cows, 0.62 compared to later-lactation cows, 0.58. However, Koeck et al. (2012) reported a higher positive genetic correlation between ketosis and displaced abomasum 0.64. In the study by Parker Gaddis (2014), a negative low correlation was found between ketosis and mastitis -0.20 and displaced abomasum and ketosis -0.25.

The coefficient of heritability for reproductive diseases ranges from 0.03 (Hardie et al., 2022) for retained placenta, 0.02 (Kašná et al., 2023) to 0.31 (Abdelharith, 2019) for metritis, 0.02 (Kašná et al., 2023) to 0.12 (Gonzalez-Peña

et al., 2020) for endometritis, 0.02 (Koeck et al., 2012) to 0.03 (Kašná et al., 2023) for clinical endometritis, and 0.02 (Kašná et al., 2023) for ovarian cysts. High positive genetic correlation was observed between retained placenta and metritis 0.79 (Neuenschwander et al., 2013) and 0.69 (Guarini et al., 2019). Negative correlation between ovarian cysts and metritis -0.24 (Parker Gaddis et al., 2014). An almost perfect correlation was observed between chronic metritis and endometritis 0.99 (Koeck et al., 2012).

### 1.3 Genetic Evaluation of Health in Cattle

Approaches for the improvement of health traits can be based on direct health data (direct observation) or indirect health data (correlated with diseases) (Stock et al., 2013). Each country has a different system for disease recording. In Nordic countries, health trait data for cattle has been collected for decades. Austria has a national uniform system for recording, while Germany created regional systems that differed, but they followed a list of diagnoses provided by ICAR. In Italy, electronically recording veterinary treatments has been mandatory since 2019 (De Monte et al., 2020). In Slovakia, recording every veterinary treatment in a diary is also mandatory, usually in paper form complemented by software form. Collecting phenotypic data is an essential component of genetic progress, and data should be collected consistently over time (Parker Gaddis et al., 2020). The use of health data requires the standardisation of diagnoses. In 2012, the International Committee for Animal Recording (ICAR) approved guidelines for the Recording, Evaluation and Genetic Improvement of Health Traits. A system consisting of a comprehensive key (>900 diagnoses), a reduced key (60 to 100 diagnoses) and a simple key (10 diagnoses) has also been described (Egger-Danner et al., 2015). Diseases can be defined in various ways. For example, clinical mastitis or claw diseases are often defined as binary traits, but they can also be categorised based on the number of cases (Heringstad et al., 2018; Vazquez et al., 2009).

In national systems, linear models are commonly applied to traits that follow a normal distribution, whereas non-linear models are utilised for traits with no normal distribution. However, linear models are favoured for international evaluations due to their simpler computational demands (VanRaden et al., 2010). Animal models have become the international base for estimating breeding values (Dash et al., 2014). It enables the evaluation of all animals within the herd. Animal models accommodate repeated records, multiple traits, non-additive genetic effects, litter effects, and fixed and random environmental effects, whether fixed or random (Henderson, 1988). To analyse binary or categorical

traits, threshold models developed in 1983 by Gianola and Foulley are used (Weigel et al., 2017). Despite their long-standing existence, the usage of threshold models has been limited due to the difficulty of integrating random effects (Rekaya et al., 2001). However, breeding values (EBV) estimated from these models tend to be more precise than those obtained by fitting binary or categorical phenotypes with a linear model (Weigel et al., 2017).

Genetic evaluation for disease resistance has its limitations. Interactions between phenotype and environment can exist, complicating statistical analysis. The measured phenotype may contain some errors, and it may not be measurable in both genders. Performance for adult cattle may not be predictable from data collected during the calf stage. Between some traits, there may be antagonistic or unfavourable genetic correlations, which they cannot be easily resolved (Berry et al., 2011). In many countries in Europe and North America, breeding values for health traits are estimated from health records or antibiotic usage records. However, using breeding values from other countries is not optimal due to genotype and environmental interactions. There are variations among countries in terms of production systems, incidence rates of diseases, genetic control, and the economic importance of diseases (Abdelsayed et al., 2017). Evaluation for mastitis resistance is realised in several countries (USA, UK, Germany and Luxemburg, Czech Republic), claw diseases (Czech Republic, Nordic countries, Canada, Netherlands), metabolic disorders (Canada, Denmark, Finland, Sweden), reproductive diseases (Austria, Nordic countries) (Heringstad et al., 2018; Krupová 2024; Pryce et al., 2016; Zavadilová et al., 2021). In Slovakia, genetic parameters for functional traits (calving ease) and somatic cell count are routinely estimated (Strapák et al., 2004; Strapák et al., 2011).

Selection indices play a crucial role in numerous breeding programs. Developed in the 1940s, they integrate information from various traits into a single numerical value, aiding in the ranking of animals. These indices predict the economic merit of animals by leveraging their genetic potential (Fessenden et al., 2020). There are various selection indexes that take health traits into account. In the UK, the Profitable Lifetime Index (£PLI) is utilised to promote milk yield, prioritise fertility and longevity, improve functional type and udder health, reduce lameness and calving performance and reduce maintenance costs (Ahdb, 2024). The UK also introduced in the 2021 selection index Healthy Cow, which considers 10 health traits, including length of productive life, calf survival, fertility, SCC, mastitis, functional type, and calving ease (Cogent, 2021). Selection index Total Nordic Merit, used in Nordic countries, combines 90 traits into

15 sub-indexes. This index allocates 45% to health and reproduction, 40% to production and efficiency, and 15% to conformation traits and workability. (Vikingsgenetics, 2023). Pro\$ is the selection index used in Canada, which includes conformation traits for functional traits (Alcantara et al., 2022). Semex offers a selection index Immunity +. This index aims to reduce the incidence of diseases such as metritis, mastitis, lameness, retained placenta, ketosis, displaced abomasum in cows and pneumonia, scours, and diarrhoea in calves (Semex, 2024). The Czech selection index incorporates health traits such as mastitis and claw diseases (both infectious and non-infectious) (Krupová et al., 2024). In Czechoslovakia, since the 1960s, a system of health control or health inheritance control has been implemented. Cows and bulls were classified into health groups. For cows, there were four categories (A, B, C, D), and for bulls, there were three categories (A, B, C). Animals categorised as A were considered free from serious diseases and were used in reproduction without restrictions, while health categories C and D indicated that bulls or cows could not be used in reproduction (Pšenica et al., 2007). In Slovakia, the Slovak Holstein Index (SHI) and the Slovak Production Index (SPI) are used for selection nowadays (Kleknerová & Candrák, 2012). The SPI index includes metrics for milk production in kilograms, fat production in kilograms, and protein production in kilograms (Kasarda et al., 2007). The SHI is the first selection index for bulls, which includes milk production traits and type traits. The relative importance of milk production is 60%, and 40% for type traits. The criteria for publishing breeding values are a reliability of 75% for milk traits and 64% for type traits (Strapák et al., 2004).

#### 1.4 New Trend – Genomic Evaluation of Health Traits

Genomic analysis has emerged as a highly effective method for estimating dairy cattle traits previously overlooked in traditional assessments. The implementation of genomic evaluation enhances the genetic merit of animals (Gutierrez-Reinoso et al., 2021). Genetic variance is not only conditioned by the additive effects of quantitative trait loci (QTL) for health and fitness traits, but these traits can also harbour QTL due to dominance or epistasis (Schneider et al., 2023). Estimation of genomic breeding values (GEBV) requires a prediction equation based on DNA markers and single nucleotide polymorphisms (SNPs). Methods for genomic prediction can be divided into two groups: linear and non-linear models, with linear models being commonly used, whether unbiased or biased (Terakado et al., 2021). For individuals with DNA information, accurate GEBV can be estimated before they reach sexual maturity (Scheffers and Weigel, 2012). The accuracy of GEBV depends on the size

of the reference population, the heritability of the traits, and the degree of relatedness between the candidate genes and the reference population. Various models are available, including TABLUP, RRBLUP, GBLUP, ssGBLUP, and ssSNPBLUP (Štrbac et al., 2023). Single-step Best Linear Genomic Prediction (ssGBLUP) is a method that combines phenotypes, pedigree, and SNP information to obtain GEBV. This method reduces the prediction dispersion of GEBVs and increases their accuracy compared to those obtained from multi-step genomic prediction (Pahlavan et al., 2023). In this method, the pedigree-based relationship matrix (A) and the genomic relationship matrix (G) are converted into one matrix (H) (Misztal et al., 2011). Genomic selection allows farmers to identify genetically important animals at a younger age (Scheffers and Weigel, 2012). It is based on information from densely distributed animal genomes. The result is an estimate of the association between each marker and the phenotype (Wientjes et al., 2022). Nowadays, there is a shift from genetic evaluation to genomic evaluation. Since 2014, Canada has utilized genomic evaluation for mastitis. Zoetis, in collaboration with the Holstein Association USA and the University of Georgia in Athens, launched the first commercially available program for genomic evaluation of wellness traits in dairy cattle (Vukasinovic et al., 2017; Gonzalez-Peña et al., 2020). In the Czech Republic, national estimates of GEBV for claw disorders and mastitis are available. The methodology for estimating GEBV for reproductive diseases has been established but is not yet implemented (Krupová et al., 2024). Genomic evaluation of clinical mastitis is routinely performed in Denmark, Sweden, Finland, France, and Switzerland. SCS is evaluated in Switzerland, Belgium, the USA, Japan, Poland, Great Britain, Italy, Germany, Austria, and Slovenia (Interbull, 2023).

## 2 Conclusions

This review discusses the genetic evaluation of health traits in cows, providing readers with a comprehensive understanding of dairy cow health, methods, and estimation of genetic or genomic breeding values and selection indexes. The average production life of cows ranges between 2.5 to 4 years, but the natural lifespan of cows is much higher around 20 years. The main culling reasons are belonging diseases, infertility and low milk production. Genomic evaluation of health traits is nowadays implemented in many countries. Implementation of this type of evaluation requires routine collection of veterinary data. Diseases are evaluated using linear models or threshold models, with the prevalence of cases mostly binary-coded. In Slovakia, bulls and cows were previously classified into health classes, but nowadays, only genetic evaluation is conducted for fitness traits and

somatic cell count. There is currently a shift from genetic evaluation to genomic evaluation. In genomic breeding, values are linked to phenotypic and DNA information, resulting in higher accuracy.

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