Réseau laitier canadien
Canadian Dairy Network

Understanding Genetic Recessives in Holsteins

Within each breed association, there are various genetic recessives officially recognized for testing and reporting. This process provides important information for potential buyers of animals, semen or embryos and allows the breed association to monitor the degree of presence of each recessive within its domestic population with the goal of full elimination. For the Holstein breed, Holstein Canada (www.holstein.ca) and the World Holstein-Friesian Federation (www.whff.info) officially recognize six genetic recessives for which carriers must be reported on pedigrees and other similar official documents including those available via the Internet. Given that these genetic recessives are presented using abbreviated codes based on the English name for the associated gene, it is important that producers understand the meaning of each code and the potential impact in their herd.

Recognized Genetic Recessives

The six genetic recessives that are officially recognized by Holstein Canada are listed in Table 1 with their associated gene and expression codes. For each disorder, a two-letter code has been established that is harmonized across all Holstein associations globally. It is important to remember these codes to identify the particular anomaly since tested animals will have the letter "C" added if they are identified as a carrier or the letter "F" added if they are shown to be a non-carrier (i.e.: free of the undesired gene).

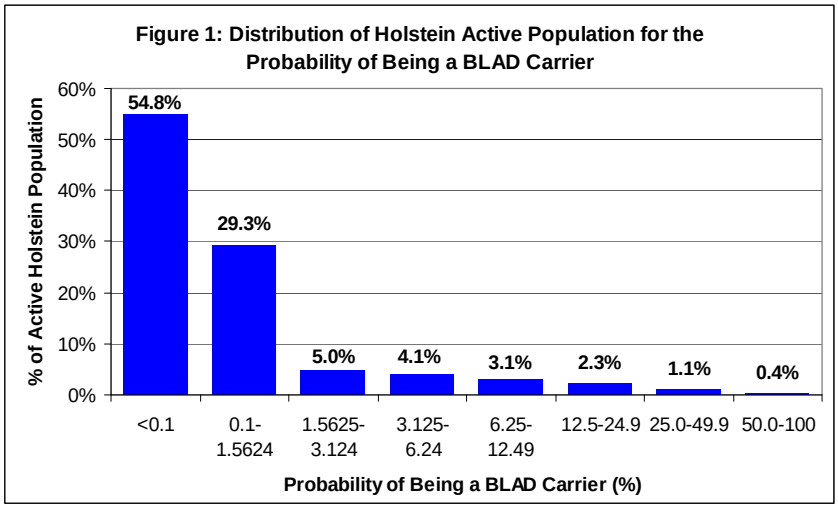
Table 1: Genetic Recessives and Gene Codes for Holsteins

Genetic Recessive	Gene Code	Gene and Expression Codes[1]
BLAD *(Bovine Leucocyte Adhesion Deficiency)*	BL	BLC = Tested carrier of BLAD BLF = Tested non-carrier of BLAD
CVM *(Complex Vertebral Malformation)*	CV	CVC = Tested carrier of CVM CVF = Tested non-carrier of CVM
DUMPS *(Deficiency of Uridine Monophosphate Synthase)*	DP	DPC = Tested carrier of DUMPS DPF = Tested non-carrier of DUMPS
Mulefoot *(Syndactylism)*	MF	MFC = Tested carrier of Mulefoot MFF = Tested non-carrier of Mulefoot
Factor XI *(Bovine Factor Eleven Deficiency)*	XI	XIC = Tested carrier of Factor XI XIF = Tested non-carrier of Factor XI
Citrullinaemia *(Bovine Citrullinaemia)*	CN	CNC = Tested carrier of Citrullinaemia CNF = Tested non-carrier of Citrullinaemia

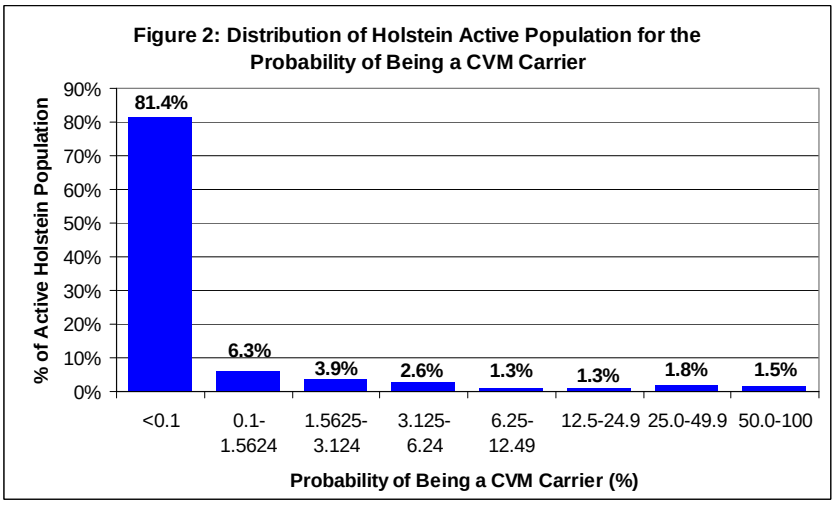
*Note 1: **C** = Carrier, **F** = Tested and Non-carrier (i.e.: Free)*

Recessive Description, Source and Occurrence

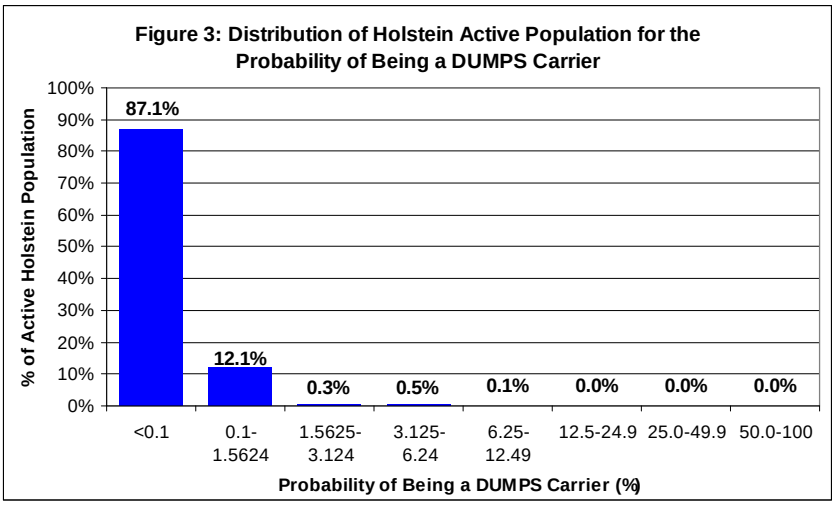
BLAD is a lethal condition that is visibly expressed when the animal has received the gene from both parents. Affected calves have stunted growth, recurrent infections such as pneumonia, slow wound healing and die within the first year after birth. Osborndale Ivanhoe is the common sire in the pedigree of all affected calves. Since various proven sires that are known carriers of BLAD have been used to some degree within the Canadian Holstein population since the early 1990s, an analysis at Canadian Dairy Network (CDN) based on gene transmission probabilities (Figure 1) estimated that 1.28% of the current active Holstein female population are carriers of the BLAD gene (gene frequency of 0.64%), which spiked near 5% for heifers born in 1992 (Figure 5), but has steadily decreased since then due to strict testing programs for young sires entering A.I. in Canada.



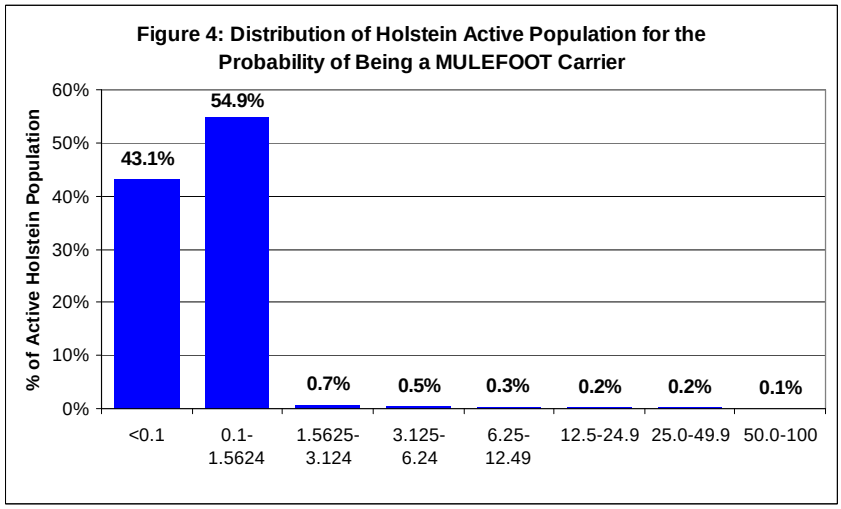
CVM is also a lethal condition that occurs when both parents transmit the undesirable gene to the resulting progeny. In this case, however, expression occurs during pregnancy, which normally leads to embryonic death, abortion or the birth of a stillbirth calf with neck and leg deformities and often heart abnormalities. While Carlin-M Ivanhoe Bell is likely the most well-known CVM carrier, his sire, Penstate Ivanhoe Star, is considered to be the original source of this gene. Since most global A.I. companies immediately tested their battery of sires for potential carriers when the CVM gene was discovered in 2000, the CDN analysis on gene frequencies (Figure 2) estimated that 1.67% of the active Holstein female population are carriers of CVM (gene frequency of 0.83%), but the brief increase to near 2.5% for heifers born in 2001 or 2002 (Figure 5) was controlled by the immediate reaction of Canadian A.I. to eliminate carriers from their young sire testing programs.

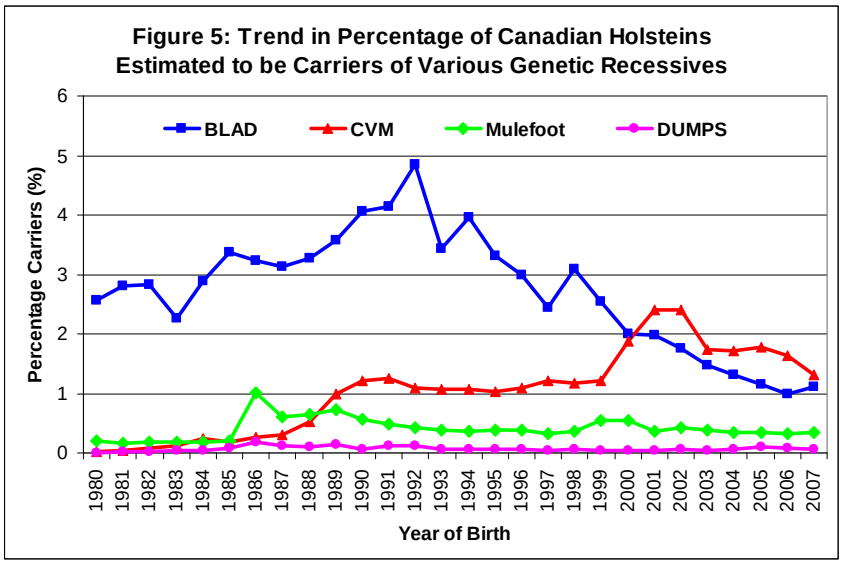


DUMPS also leads to embryonic mortality when the gene is inherited from both parents but, contrary to CVM, the foetus always dies within 40 days after conception. This genetic disorder is caused by the deficiency of a specific enzyme important for the synthesis of DNA. Although only carriers can be found in the population, carrier females may require more breedings per calf born since some of their pregnancies could end in early embryonic death. DUMPS was discovered in the late 1980s with all known carriers tracing back to Skokie Sensation Ned. In Canada, due to the strict testing strategy implemented by A.I. companies, already more than a decade ago, the proportion of the active population that are carriers is estimated at 0.068% (gene frequency of 0.034%), which means that DUMPS has essentially been eliminated from the Holstein breed in Canada (Figures 3 and 5).



Mulefoot is characterized by the fusion of the claws on one or more feet, with the front feet being affected before the rear feet and the right side before the left. As with the other genetic recessives above, affected animals must receive the gene from each parent. In the case of Mulefoot, however, the condition is not lethal and while affected animals have locomotive difficulties, they can live to reach maturity and beyond. In the Holstein breed, Wayne Spring Fond Apollo is commonly considered the original source of Mulefoot but, in fact, he inherited this defect from ancestors further back in his pedigree, namely Raven Burke Ideal and his dam, Raven Burke Elsie. Although no DNA test has yet been developed, it is estimated that a very low percentage (0.364%) of the active Holstein population in Canada are carriers of Mulefoot (Figures 4). The slight spike in gene frequency for heifers born in 1986 resulted from the arrival of second crop daughters of A Hurtgen-Vue Marathon once he was first proven in Canada in 1985, at which time he was not yet known to be a Mulefoot carrier.





Factor XI (eleven) deficiency is a blood clotting disorder whereby affected animals show symptoms similar to haemophiliacs. While not a lethal condition, only animals that inherited the undesirable gene from both parents will express the symptoms. For these animals, haemorrhaging may occur at their birth from the umbilical cord, when being dehorned and/or when giving birth as an adult, which may result in death. Factor XI has only recently been officially recognized as a genetic recessive and therefore animals in Canada have not yet been formally tested for it. The originating source animal of this disorder in the Holstein breed has not yet been definitively identified.

Citrullinaemia has also been recently acknowledged as a genetic recessive in Holsteins and, like others described earlier, it is lethal when the responsible gene is inherited from both parents. Calves that are affected cannot metabolize urea properly, which leads to high levels of ammonia in the plasma and ultimately in the brain. They consequently display neurological symptoms and rapidly deteriorate, leading to death within the first week of life. In Holsteins, Linmack Kriss King, has been identified as the source animal of this condition.

Summary

There are currently six genetic recessives officially recognized in the Holstein breed, which are briefly described in this article. Test results for these anomalies must be disclosed for presentation on official pedigrees and other similar documents. In Canada, data exchange procedures between Holstein Canada and Canadian Dairy Network ensure that their respective web site also displays the appropriate codes for each animal with a genetic recessive test result. Producers and A.I. personnel should be familiar with the various gene and expression codes to avoid unintentional usage of carrier animals in their breeding program. The aggressive testing policies implemented by A.I. companies have been critical for the control and eventual elimination of these disorders.

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