Réseau laitier canadien
Canadian Dairy Network

Brachyspina: A New Official Genetic Recessive

In accordance with the official recognition by the World Holstein Friesian Federation (WHFF) of Brachyspina as a recessive genetic defect in the Holstein breed, both Holstein Canada and Holstein USA have recently announced their approval to publish testing results on pedigrees and other official documents. First discovered in 2007 and described by researchers in the Netherlands and Denmark, Brachyspina is a single autosomal defect, therefore the condition is only expressed when an animal is homozygous recessive for the detrimental gene. In other words, the lethal condition only occurs in one-quarter of the matings when both parents are carriers. Animals receiving the undesirable gene from both parents will normally die early during pregnancy (similar to CVM). In rare cases, the calf survives pregnancy and is born dead with a shortened spinal cord, long legs and abnormal organs.

Transmission and Testing

Research has shown that the lethal condition traces back to a single source, namely Sweet-Haven Tradition. Tradition was born in the USA in 1974 but never had his semen available for purchase in Canada. Most of the transmission of the undesirable gene therefore occurred through semen sales of his sons, grandsons, etc. A genetic marker test for Brachyspina was developed by researchers in the Netherlands and Belgium. Testing services for this genetic recessive are now publicly available via a laboratory in the Netherlands. In Canada, animals can be tested at $85 per sample by contacting Holstein Canada to order the required testing kit. Holstein Canada will use the officially recognized WHFF codes to designate Brachyspina tested-free (BYF) and tested-carrier (BYC) animals. Holstein USA plans to report different two-letter codes for tested-free (TY) and tested-carrier (BY) animals. At this time, semen from some bulls that are carriers of the undesirable Brachyspina gene may be available for sale in Canada.

Probability of Being a Carrier

Canadian Dairy Network (CDN) has been gathering Brachyspina test results (both BYF and BYC) from various AI companies. To date, most testing has been conducted on males but the availability of testing services through Holstein Canada will facilitate female testing in Canada. Starting with Tradition as the source animal and using all known Holstein pedigree data, CDN traced the undesirable gene through all known descendants of Tradition (and any additional progeny that could be deduced). The probability of being a carrier of the Brachyspina gene was estimated for all animals that do not have a test result at CDN. Using the computed carrier probabilities, CDN estimated the frequency of carriers in the Canadian Holstein active population to be less than 5% at 4.4%. Based on similar analyses in the Netherlands and United States, it was estimated that 8% and 6% of their respective Holstein populations are carriers of Brachyspina.

Expected Rates of Early Embryonic Death

When two carrier animals are mated, the resulting progeny born alive have a two-thirds chance of also being a carrier and a one-third probability of being totally free. For

matings whereby the progeny received both recessive alleles, they will either die during pregnancy or be stillborn. Based on all known Holstein pedigree data at CDN, an analysis was conducted to examine the overall impact of Tradition in Canada. Of the 7.6 million registered Holsteins born in Canada since 1980, nearly 2.2 million can be traced back to Tradition, some of which are now 11th generation descendants. Among all registered females born in Canada in 2010, 84% can be traced back to Tradition. Specific to the current population of active Holstein cows and heifers in Canada, 76% could be mapped back to having at least one connection to Sweet-Haven Tradition.

Probability of being a carrier in the Canadian Holstein active female population has slowly increased to a current average of 4.4%. Therefore, we would expect that of 1.1 million active females in Canada, approximately 48,400 are carriers of the Brachyspina defect. Among the progeny proven AI sires born since 2002, it is estimated that 6.3% are carriers. If animals are mated at random (i.e., test results are not publicly available), roughly 3,000 matings in the current active population are expected to be between carriers. Since there is a 25% chance of inheriting the lethal condition (homozygous recessive) when two carriers are mated, it is therefore expected that 1 in every 750 pregnancies resulting from random mating would result in early embryonic death. Given the official publication of Brachyspina testing results by Holstein Canada and CDN, known carriers will be easily identified. When a known carrier sire is mated randomly to non-tested cows and heifers in the Canadian active population, roughly 1 pregnancy out of 100 is expected to lead to early embryonic death or a stillborn calf due to Brachyspina.

Industry Direction

Holstein Canada has declared Brachyspina to be an officially recognized genetic recessive defect in the Holstein breed and plans to publish all test results on official documents and its website. The probability of being a carrier as calculated by CDN will be extremely useful to identify highest risk females and males that should be targeted for testing. It is highly recommended that all AI organizations publish any Brachyspina test results (BYF or BYC), especially for their active sires. Industry partners are encouraged to increase awareness of this undesirable gene and its known mode of transmission through descendants of Tradition using heightened communication to producers. While this recessive should not cause panic, an improved understanding is warranted to control its further spread in the Holstein breed for years to come.

CDN plans to investigate potential depression in fertility and reproductive performance for animals that carry this lethal gene. Preliminary research results in the Netherlands reported that sires carrying the BY gene had lower fertility than non-carrier sires. Embryonic deaths for any reason can cause depression in fertility and reproductive performance in future lactations. These relationships should be investigated in Canadian Holsteins when a sufficient number of carriers and non-carriers of Brachyspina can be accurately identified. It is important that results for tested animals be made publicly available to avoid close mating of carriers. Top genetic sires and cows identified as being a carrier of the defect can still be used as long as caution is given to the pedigree of the potential mate.

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