



Update on Haplotypes Including JNS for Jerseys

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- Genetic Conditions

The era of genomic selection has brought forth several benefits. Perhaps the first that comes to mind is the advanced rate of genetic improvement for all traits including those that are hard to measure and/or those with a lower heritability. Two other significant benefits, however, include the ability to confirm parentage with full certainty as well as the identification and management of genetically undesirable characteristics. These undesired genetic defects are normally controlled by a single gene that is recessive rather than dominate in nature. For this reason, the best way to control their spread in a breed or in your herd is to avoid mating carrier animals together.



Gene Tests Versus Haplotype Results

Within each breed there are now thousands, even millions, of animals that have been genotyped. On a routine basis, the Council on Dairy Cattle Breeding (CDCB) in United States analyses all such genotypes to try to identify any new genetically recessive defects within each breed. In addition, researchers can also investigate the DNA profiles of calves born on farm that have similar physical abnormalities in an attempt to identify the gene responsible and the list of carrier animals within the breed. In reality, however, it is not always possible to identify the causative gene responsible for every genetic defect observed in the population. When the gene is known, a gene test can then be developed and offered to test animals and identify those that are carriers or free. That said, even when the causative gene is not known, analysis of all genotypes in the breed may be able to identify a short section of the genome, known as a haplotype, that is expected to include the causative gene. While very valuable in controlling the spread of undesirable genes, identifying carrier animals based on haplotypes is not as accurate as results from a specific gene test.

On the [LactanetGen.ca](https://lactanetgen.ca) website, any known gene test result is presented under each animal's name on its "Genetic Evaluation Summary" and "Pedigree" pages. These include results for polled and beta casein (i.e.: A2) testing for all breeds but other gene test results specific within each breed, such as Brachyspina (BY) and CVM in Holsteins. Since haplotype results are not 100% accurate, they are presented as carrier probability values ranging from 1% (likely free) to 99% (likely a carrier) and appear only on the animal's "Pedigree" page.

New Genetic Recessive Abnormality in Jerseys

A new undesirable genetic trait, known as Jersey Neuropathy with Splayed Forelimbs (JNS), was recently identified in the Jersey breed. JNS is a recessive genetic anomaly and when inherited from both parents results in a calf that is born alive and appears healthy but they cannot stand. Affected calves exhibit neurologic symptoms with muscle stiffness in the head and neck and they also have front legs that are rigid and spread out in front of their body.

At the present time, no gene test is available to identify carriers with complete certainty but a high level of accuracy can be achieved by the analysis of haplotypes for genotyped Jerseys. The CDCB in United States has expanded its current analysis of haplotypes to include JNS and results are routinely shared with Lactanet Canada for all genotyped Jerseys. Based on those results, carrier probabilities for the JNS haplotype will be published on the Lactanet website effective the April 2021 genetic evaluation release. For each Jersey animal, the JNS carrier probability will be displayed on its Pedigree page and the Advanced Group Query tool will also allow users to easily identify and filter Jersey sires that are deemed to be either Carrier or Free based the haplotype analysis. Ultimately, by knowing which Jerseys are carriers, or likely to be one based on their pedigree, Canadian Jersey breeders can avoid mating carriers together and minimize the potential impact of JNS calf losses in

their herd. As shown in Figure 1, the estimated percentage of JNS carriers has been steady at 7 to 8% for Canadian Jerseys born since 2017, which means that it can easily be managed going forward using the tools provided by Lactanet.

Key Sires Contributing to JNS in Canadian Jerseys

Based on an analysis of all genotyped Jerseys conducted by CDCB, the oldest genotyped animals that were identified as carriers of the JNS haplotype are Woodstock Berretta Leslie (JEUSAF3906889, born in January 1995) and her daughter, Woodstock Alf Leslie (JEUSAF3980701, born in December 1996). In terms of how JNS infiltrated the Canadian Jersey population, however, it appears this mainly happened via the high-ranking grandson of Woodstock Berretta Leslie, namely Tollenaars Impuls Legal 233-ET, born in 2004. Table 1 provides a list of those sires that have been identified as a carrier of the JNS haplotype and have at least 50 registered daughters born in Canada. Other than Woodstock LLV Lieutenant-ET, who shares the same maternal granddam as Legal, all other sires in Table 1 are direct descendants of Legal.

Table 1: Sires With at Least 50 Registered Daughters Born in Canada and Identified as Carriers of the JNS Haplotype

Registration Number	Bull's Name	Birth Year	No. Registered Daughters Born in Canada	Relationship to Legal
JEUSAM111389227	WOODSTOCK LLV LIEUTENANT-ET	1999	71	Maternal Cousin
JEUSAM61929249	TOLLENAARS IMPULS LEGAL 233-ET	2004	715	Himself
JEUSAM117222740	ALL LYNNS LEGAL VISIONARY-ET	2010	261	Son
JECANM106797223	GUIMO JOEL ET	2011	2,723	Son
JEUSAM118201001	ROWLEYS 11 VISIONARY CHILI-ET	2012	349	Grandson
JEUSAM71199770	AHLEM NIKON-P	2012	85	Grandson
JEUSAM67089973	SCENIC VIEW MARTINEZ-P-ET	2013	57	Grandson
JE840M3012423929	RIVER VALLEY CECE CHROME-ET	2013	1,175	Grandson
JECANM108820219	TRAILBLAZER CCCJ BARRACUDA ET	2014	163	Grandson
JEUSAM74067487	AHLEM TEXAS WILDCAT 22702-ET	2015	296	Grandson (Maternal line)
JE840M3126479215	RIVER VALLEY CHAR CHANGE UP ET	2015	443	Great Grandson (Maternal line)
JECANM109516964	PERKINS SKYCLASS ET	2015	248	Great Grandson
JEUSAM67650122	FOREST GLEN KALAHARI	2016	154	Great Grandson
JECANM12725636	GRAYCLAY CROWN	2017	89	Great Grandson

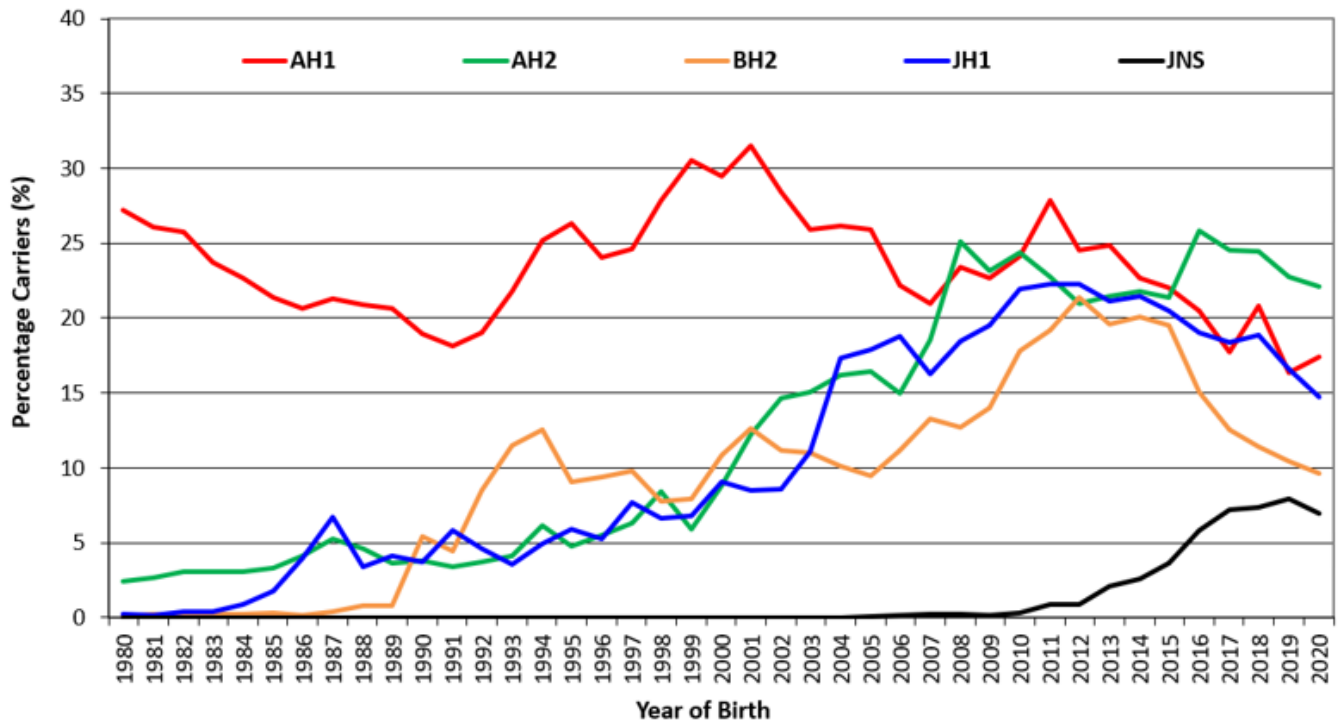
Removal of JH2 and BH1 Results

While the analysis of haplotypes among genotyped animals in a breed is normally a very valuable tool for controlling the spread of undesirable genes, this process is not perfect and results can change over time. In fact, this happened for two haplotypes affecting fertility that were initially identified in the Jersey (JH2) and Brown Swiss (BH1) breeds. As more genotypes accumulated in these breeds and, more importantly, as the accuracy of the reference map for the bovine genome improved, CDCB decided that the analysis and publication of the JH2 and BH1 haplotypes was no longer appropriate, effective 2018. As a consequence, Lactanet will also follow the same approach and therefore JH2 and BH1 will no longer be displayed effective the April 2021 genetic evaluation release. Canadian Jersey and Brown Swiss breeders no longer have to consider these haplotypes when making sire selection and mating decisions.

Haplotype Trends

The trends in percentage carriers for known haplotypes in the Jersey, Ayrshire and Brown Swiss breeds are presented in Figure 1 and similar trends for the Holstein haplotypes are shown in Figure 2.

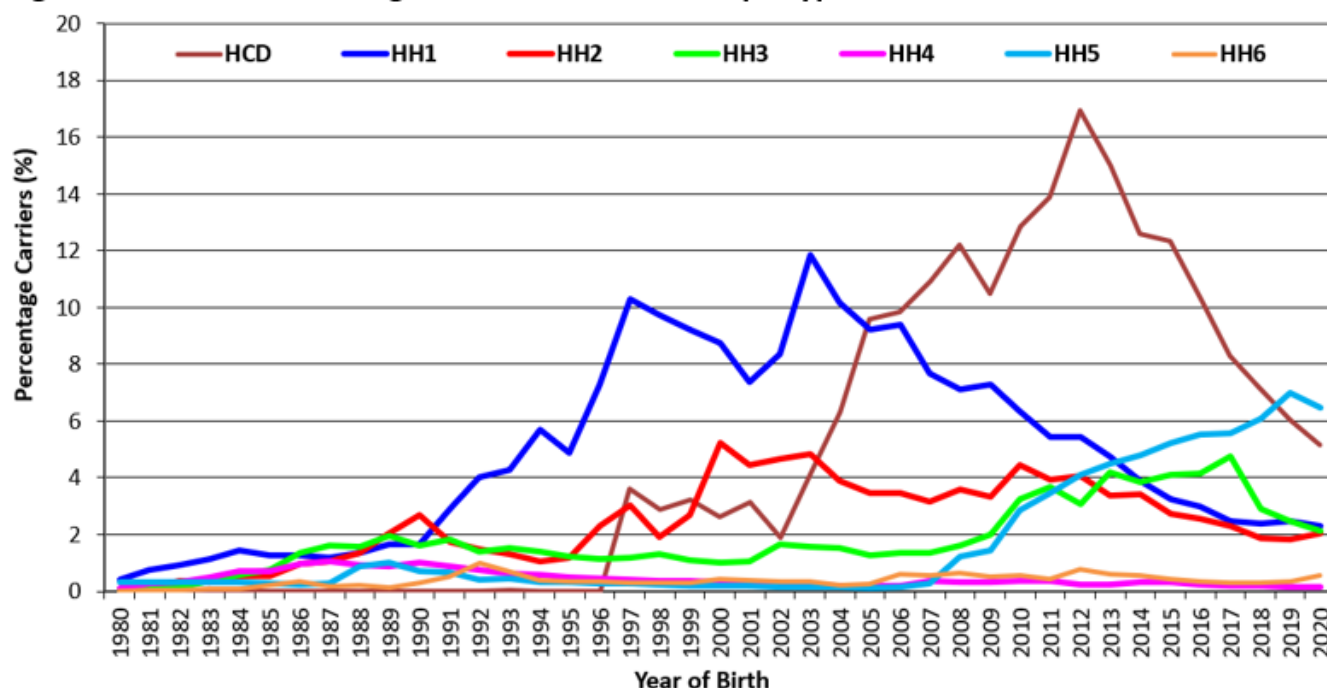
Figure 1: Trend in Percentage Carriers for Known Haplotypes in Canadian Jersey, Ayrshire and Brown Swiss Females



For all haplotypes that have been known for a several years, there has been a downward trend in the percentage of carriers among registered females born in Canada. More specifically, for heifers born in 2020, the carrier percentages are estimated to be 17.4% and 22.1% for AH1 and AH2, respectively, 9.6% for BH2 and 14.7% for JH1. For JNS, as the newest haplotype discovered, the percentage carriers for Jersey heifers born in 2020 is 7.0%.

In the Holstein breed, significant decreases in the percentage of carriers have been realized for the two that had the highest frequencies, namely HCD, now at 5.2% for heifers born in 2020, and HH1, which is now at 2.3% for 2020. HH2 and HH3 also have estimated frequencies near 2% while HH4 and HH6 have extremely low frequencies in Canadian Holsteins at 0.1% and 0.6% for heifers born in 2020. Surprisingly, HH5 is still experiencing an upward trend in the estimated percentage of carriers, reaching 6.5% for heifers born in 2020.

Figure 2: Trend in Percentage Carriers for Known Haplotypes in Canadian Holstein Females



Summary

A key benefit of the genomic era and the accumulation of genotypes, now approaching a total of 5 million in North America, is the ability to discover new genetic recessive traits within each breed. Even without a gene test available, the analysis of haplotypes is tremendously valuable for identifying animals that are most likely to be carriers and those that are expected to be free. Based on haplotype results received from CDCB in United States, Lactanet can provide carrier probabilities for all animals in its database within each breed. In recent months, a new genetic abnormality was identified in the Jersey breed, known as Jersey Neuropathy with Splayed Forelimbs (JNS), and Lactanet will be introducing new JNS haplotype results effective April 2021. At the same time, two previous haplotypes affecting fertility, namely the JH2 in Jersey and BH1 in Brown Swiss, will no longer be published due to analysis by CDCB clearly indicating it is not an accurate haplotype to analyze. In general, Lactanet's publication policy of displaying carrier probability results for all animals on their Pedigree page, accompanied by its website filtering tools, have led to reduced frequencies of essentially all known haplotypes over time.

By Brian Van Doormaal