

## Haplotypes Affecting Fertility in the Ayrshire Breed

Although the main benefit of DNA genotyping is for the estimation of genomic evaluations for improved genetic selection and faster rates of genetic progress, there are also other significant benefits. Of course, one is the confirmation of parents for accurate recording of pedigrees by national breed association like Ayrshire Canada but another is the potential opportunity to identify and manage genetic anomalies in the breed.

Canadian Dairy Network (CDN) introduced official genomic evaluations for Ayrshires in December 2012 and a by-product of the genotyping in North America was the discovery of a "Haplotype Affecting Fertility", labelled as AH1 (i.e.: Ayrshire Haplotype 1). The CDN web site identifies animals that are AH1 carriers and provides a Carrier Probability value ranging from 1% to 99% for every Ayrshire animal in the database. More recently, a second haplotype affecting fertility was discovered in the Ayrshire breed, which is labelled as AH2 for Ayrshire Haplotype 2.

### Impact on Reproductive Performance

Both AH1 and AH2 reflect the expression of two different genes that are genetically recessive and lethal at some stage of embryonic or fetal development for animals that inherit a copy from both parents (i.e.: homozygous carriers). This outcome is proven by the fact that no genotyped Ayrshire has been found to carry two copies of the given haplotype. Therefore, the propagation of these undesirable genes in the breed occurs solely by the use of carrier sires to breed carrier females and when this occurs, the resulting progeny has a 25% chance of being homozygous, leading to early embryonic death or a stillbirth calf.

Given the relative size of the Ayrshire population in Canada compared to the United States, research was carried out at the University of Guelph to estimate the effect of AH1 and AH2 on female fertility and stillbirth rate. Table 1 presents the results of this analysis showing the effect of 56-day non-return rate and stillbirth rate for mating combinations for which the sire and maternal grandsire were both carriers compared to mating combinations when both were known to be free of the given haplotype affecting fertility.

Table 1: Estimated Effect of Haplotypes Affecting Fertility in Canadian Ayrshires on 56-day Non-Return Rate and Stillbirth Rate		
Haplotype:	AH1	AH2
Oldest Known Carrier Sire (Birth Year)	Selwood Betty's Commander (1953)	Oak Ridge Flashy Kellogg (1961) or Oak-Ridge Lightning (1958)
Effect on 56-day Non-Return Rate:		
- Heifers	n. s.*	-5.1%
- Cows	n. s.*	-4.0%
Effect on Stillbirth Rate:		
- First Calving	+2.3%	n. s.*
- Later Calvings	+2.0%	n. s.*

\* - not significantly different than zero so no effect was observed.

For AH1, the analysis found no significant effect for its impact on 56-day non-return rate as a measure of female fertility but stillbirth rates were approximately 2% higher regardless of

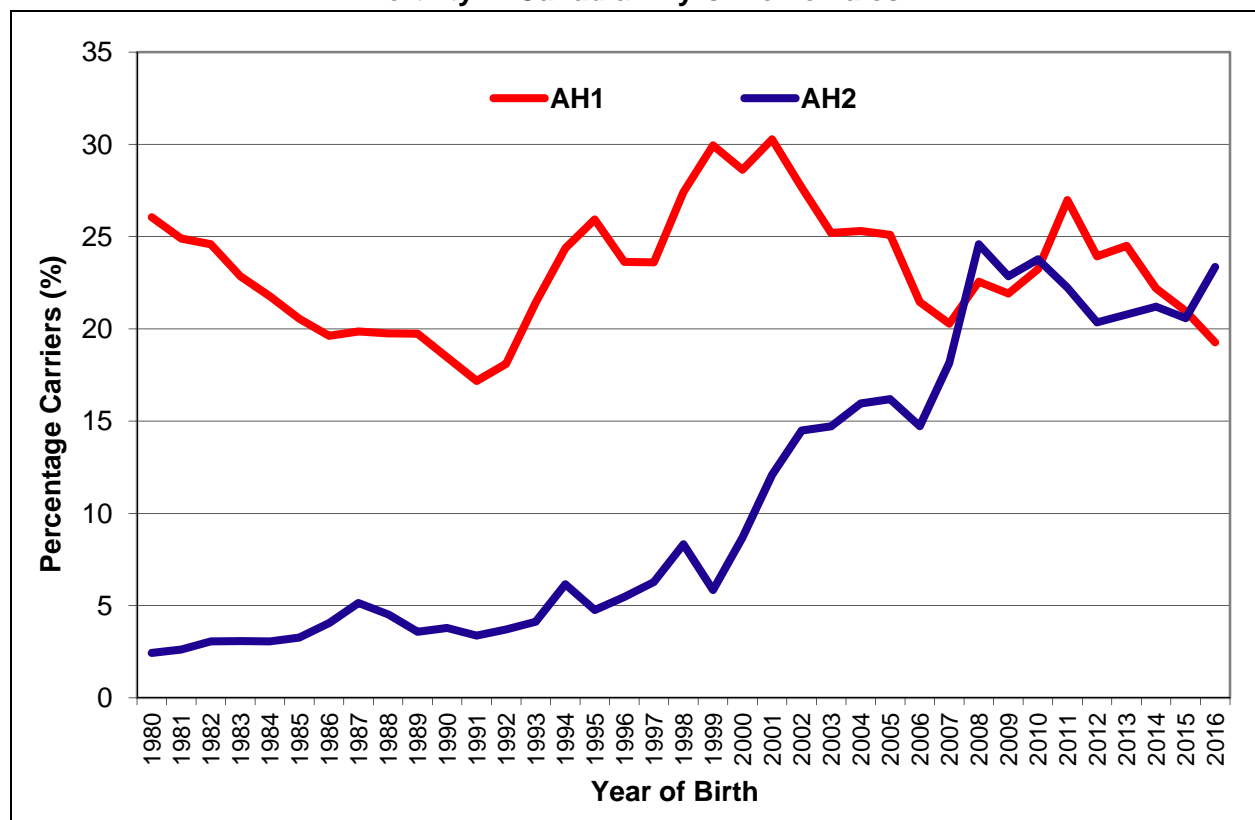
whether the dam was calving for the first time or for a subsequent calving. For AH2, no significant effect was found on stillbirth rates but the impact on non-return rate was a reduction of 5.1% in heifers and 4.0% in cows. These results suggests that AH2 causes early embryonic mortality while the effect of AH1 occur much later during the pregnancy for calves that are homozygous for the gene associated with this haplotype.

### Frequency and Management

For all genotyped Ayrshires, CDN can identify those that are carriers of each haplotype, AH1 and AH2. Given the complete pedigree data also available at CDN, the likelihood that any non-genotyped Ayrshire is a carrier of each haplotype is calculated. For AH1, CDN has been displaying each animal's Carrier Probability for years, as part of their Pedigree page on the CDN web site. In addition, the Advanced Group Query allows users to list bulls depending on whether they are known to be a carrier of AH1 or not. In the near future, these features of the CDN web site will be expanded to include the AH2 status for genotyped Ayrshires and Carrier Probability values for non-genotyped animals.

Using the preliminary AH2 results available at CDN, based on the research carried out in the United States that identified this newest haplotype affecting fertility, Figure 1 shows the trend in the average carrier probability for AH1 and AH2 for Canadian Ayrshire females born since 1980. This graph shows that the frequency of AH1, which traces back to Selwood Betty's Commander born 1953, has varied from 17 to 30 percent for the past 35 years. For AH2, however, the frequency in Canadian Ayrshires was below 10% up to 2000 but has increased to surpass 20% for heifers born since 2008. Four popular sires that produced more than a 1,000 milk-recorded daughters during that time period, who are AH2 carriers, are Blackaddar B B Kellogg, Woodland View Pardner ET, Des Champois Poker-ET and Duo Star Normandin.

**Figure 1: Trend in Percentage Carriers for the AH1 and AH2 Haplotypes Affecting Fertility in Canadian Ayrshire Females**



Therefore, there is value for the Ayrshire breed to manage both haplotypes and the underlying genes responsible for their impact on reproductive performance. The ideal approach to achieve this goal is to minimize the frequency that carrier sires are mated to carrier females. Genotyping your heifers and cows is the only way to know those that are carriers of AH1 and/or AH2 and based on these results CDN provides the probability of being a carrier for each non-genotyped Ayrshire. Knowledge of these Carrier Probabilities should be used to identify those that should not be mated to known carrier sires available in A.I. This strategy has already been adopted by some breeders for AH1, which has led to a lower average carrier probability for heifers born since 2011 (Figure 1).

## **Summary**

Continued genotyping of Ayrshires in Canada and United States has resulted in official genomic evaluation provided by CDN and increased rates of genetic progress of traits of importance. A secondary benefit of this genotyping has been the discovery of two genetic anomalies negatively affecting reproductive performance, which have existed unknowingly for decades. Now that AH1 and AH2 are known and it is possible to accurately know carriers by genotyping, Ayrshire breeders have all the tools needed available from CDN to manage these anomalies and gradually reduce their impact on the breed going forward.

Author: Brian Van Doormaal, General Manager, CDN

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