The detection of putative recessive lethal haplotypes in Irish sheep populations

R. McAuley1,2, N. McHugh1, T. Pabiou3 and D.C Purfield2

1Teagasc, Animal & Grassland Research Centre, Moorepark, Fermoy, Co. Cork, Ireland, 2Munster Technological University - Cork Campus, Bishopstown, Cork, Ireland, 3Sheep Ireland, Link Road, Ballincollig, Co. Cork, Ireland

In livestock populations, recessive lethal alleles are a known contributor to poor reproductive performance due to embryonic death in homozygous individuals. Despite their lethal effect in the recessive form, these alleles may be maintained at high frequencies among carrier animals because of their positive pleiotropic effects on economically important traits. Although several such recessive alleles have been identified in cattle and pig populations, limited studies have been completed in sheep, and none within Irish sheep populations. Genotype data for 69,034 animals from 5 major Irish sheep breeds genotyped on a variety of panels was available for this study. Only animals and single nucleotide polymorphisms (SNPs) with a call rate > 90%, and a minor allele frequency >0.01 were retained. Non-autosomal SNPs and SNPs that did not adhere to Mendelian inheritance patterns were discarded. Haplotype phasing and genotype imputation of all panels to the Ovine SNP50 density was completed using FImpute V2.2. Following imputation, 43,951 SNPs remained across 66,996 animals, which included 32,256 verified progeny-sire-dam and 2,089 verified progeny-sire-maternal grandsire trios. To identify haplotypes with homozygous deficiency, a sliding haplotype window of 10 SNPs was used the scan the genome of all genotyped animals. A window of 10 SNPs was chosen due to the high level of haplotype diversity with homozygosity rates above 0.05. Haplotype lethality was determined using a chi-square test between the number of observed and expected haplotypes. A number of haplotypes showed a significant deficiency of homozygote animals across the five different Irish sheep populations. For example, one candidate haplotype in the Charollais population was observed in the homozygous form significantly less than was expected ($p<1x10^{-4}$). Comparison of at risk-matings (between carriers) and safe matings, demonstrated that this haplotype was associated with a decrease in the average number of lambs born ($p<1x10^{-5}$). This finding can be incorporated into Irish sheep breeding programs to avoid carrier-by-carrier matings to reduce lamb mortality.