Identifying Chromosomal Recombinations in Beef Cattle from Genotyped Parent-Offspring Pairs

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Summary and Implications

This study investigated meiotic recombination in two breeds of cattle by comparing phased SNP haplotypes in sire-offspring pairs. The positions and number of recombination events were identified. The number of recombination events varies between individuals and is a heritable trait. A genome-wide association analysis identified quantitative trait loci (QTL) associated with variation in the number of recombination events. Regions that had more recombination events than expected were identified in both breeds, and many of these hotspots were in common. Recombination is important biologically because it is the mechanism for reassembling paternal and maternal alleles. Recombination impacts the accuracy of imputation, a commonly-used approach to infer the genotypes of some individuals based on genotypes of others.

Introduction

Diploid organisms carry pairs of autosomal chromosomes, one inherited from their sire and the other from their dam. Crossover events can occur at any and every pair of autosomes, leading to gametes with chromosomes that are paternal in origin on one side of the recombination point and maternal in origin on the other side. On average every pair of chromosomes exhibits one meiotic recombination, but some have none and others have two or three. The location of these recombination events can be identified by comparing phased genotypes known as haplotypes, in parent-offspring pairs.

Recombination events tend to occur in hotspots and the number of recombination events varies among individuals. The presence of recombination influences the accuracy of haplotype phasing and the imputation of missing genotypes. The objectives of this study were to locate recombination hotspots, scan the genome for QTL, and identify candidate genes influencing recombination.

Materials and Methods

There were 2,775 Angus and 1,485 Limousin parent-verified sire/offspring pairs genotyped with the Illumina BovineSNP50 BeadChip (Illumina, San Diego, CA). Haplotype phasing was performed with DAGPHASE2.4 (Druet and Georges, 2010), or BEAGLE3.3 (Browning and Browning, 2007) using UMD3.1 assembly SNP coordinates. Recombinations were detected by comparing the two reconstructed chromosomal haplotypes inherited by each offspring with those of their sires. The total numbers of paternal recombination were determined for each offspring by summing up the number of recombinations on each of the 29 autosomes. Narrow sense heritability of genome-wide numbers of recombination was estimated separately for each breed using a repeatability model in ASREML3.0 (Gilmour et al., 2009). The BayesB (Meuwissen et al., 2001) approach for genome-wide association analysis implemented in GenSel software (Fernando and Garrick, 2013) was used to identify genomic regions harboring QTL with large effects on recombination.

Results and Discussion

DAGPHASE was superior to BEAGLE in haplotype phasing, indicating that pedigree information increases the accuracy of haplotype phasing. The estimated genetic length of the 29 bovine autosomes was 3,097 cM, with a genome-wide recombination distance averaging 1.23 cM/Mb. There were 427 and 348 hotspots detected in Angus and Limousin, respectively, of which 166 were in common. The estimated heritabilities of number of recombination were 0.26 in Angus and 0.23 in Limousin. Several common candidate genes (eg. *RECQL4*), which influence the number of genome-wide recombination events were localized to QTL regions detected in the two breeds.

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