

across chromosomes were calculated as ratios of correctly imputed genotypes to overall imputed genotypes. Imputation accuracy for the 3 methods ranged from 76.31% to 93.91%. The CFI had slightly higher imputation accuracy (93.91%) than PFI (93.56%) and both methods were substantially more accurate than PBG (76.31%). Noticeably most chromosomes that showed either high or low imputation accuracies were the same chromosomes that had high and low average linkage disequilibrium (defined here as the correlation between pairs of adjacent SNP within chromosomes less than 5 MB apart). This suggested that choosing sets of SNP with high levels of average linkage disequilibrium would improve imputation accuracy. Results clearly indicated that FImpute software (population or combined family-population) were more suitable than Beagle for genotype imputation in this Thai multibreed population. Perhaps additional increments in imputation accuracy could be achieved by discarding SNP with low levels of average linkage disequilibrium, and by increasing the completeness of pedigree information.

**Key Words:** imputation accuracy, linkage disequilibrium, multibreed dairy cattle

**W92 Identification of copy number variation in Brazilian synthetic dairy cattle breed.** T. C. S. Chud<sup>1</sup>, M. V. G. B. da Silva<sup>2</sup>, A. S. Carmo<sup>2</sup>, T. B. R. Silva<sup>\*1</sup>, G. A. Oliveira Junior<sup>3</sup>, F. S. Baldi Rey<sup>1</sup>, and D. P. Munari<sup>1</sup>, <sup>1</sup>Univ Estadual Paulista "Júlio de Mesquita Filho," Jaboticabal, SP, Brazil, <sup>2</sup>Embrapa - Brazilian Corporation of Agricultural Research, Juiz de Fora, MG, Brazil, <sup>3</sup>Universidade de São Paulo, Pirassununga, SP, Brazil.

Copy number variation (CNV) refers to genomic segments that present a type of structural variation, such as duplications or deletions. CNVs have been observed as an important source of genetic and phenotypic variation for production traits and animal health. The aim of this work was to identify CNVs in a synthetic breed (Gyr × Holstein) dairy cattle population (Girolando cattle). The data set contained 417 animals genotyped with the Illumina 50K SNP panel (~54,609 SNPs). An algorithm based on the Hidden Markov Model was implemented using PennCNV software (Wang et al., 2007) for CNV identification. PennCNV perl script was used to eliminate calls from low quality samples, based on the standard deviation of LRR (<0.30), the BAF drift (<0.01) and waviness factor (less than 0.05). The final data set was composed of 384 animals. Gene content of cattle CNV was assessed using Ensembl genes. We used the PANTHER classification system for testing the hypothesis ( $P < 0.05$ ) that the GO terms of the molecular function, biological process, and pathway terms were under or overrepresented in the CNV. An account of 1,986 CNVs were found along the genome, of which 84% were duplications and 16% were deletions. The chromosomes BTA3, BTA17 and BTA23 presented higher frequencies (10.52%, 11.53%, 8.30%, respectively) of CNV. Chromosomes that showed lower frequency of CNV (<1%) were BTA27, BTA14 and BTA29. A total of 861 genes were found within these regions and they are involved in biological processes, such as development (105 genes), growth (2 genes), immune system (83 genes), metabolism (343 genes) and reproduction (12 genes). This study showed evidences of structural variations in the genome of Girolando cattle and the genes found in CNV may be involved in the expression of production and animal health traits.

**Key Words:** genomics, single nucleotide polymorphism, structural variation

**W93 Linkage disequilibrium in a Thai dairy cattle population with different Holstein fractions.** Thawee Laodim<sup>1</sup>, Skorn Koona-wotrittriron<sup>\*1</sup>, Mauricio A. Elzo<sup>2</sup>, and Thanathip Suwanasopee<sup>1</sup>,

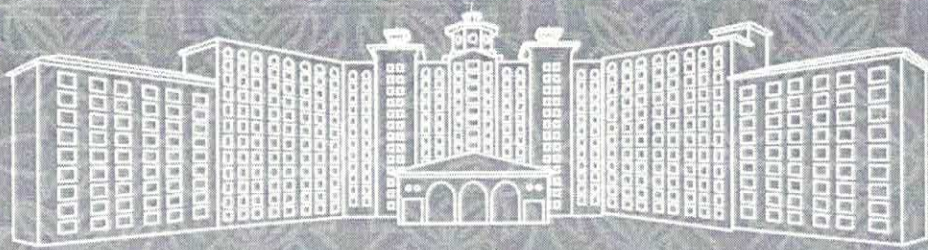
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Linkage disequilibrium (LD) is important for gene mapping, accuracy of genomic prediction, and understanding of recombination biology in dairy cattle populations. The level of LD can vary among populations depending on their genetic structure, selection and recombination rates. The objective of this study was to estimate and compare levels of LD in dairy cattle with different Holstein fractions under tropical conditions. Blood samples of 2,643 dairy cattle (89 bulls and 2,554 cows) from 304 farms located in Central, Northern, Western and Southern Thailand were extracted for DNA. The DNA samples were genotyped with one of 4 GeneSeek Genomic Profiler BeadChips (9K, 20K, 26K, or 80K). Only SNPs from autosomes in common among the 4 chips were considered. In addition, SNPs with a minor allele frequency (MAF) lower than 0.01 and a call rate lower than 90% were excluded. This resulted in a set of 7,123 SNPs used in this study. Animals were classified into 7 groups based on their Holstein fraction (HF): HF <75%, 75% ≤ HF <80%, 80% ≤ HF <85%, 85% ≤ HF <90%, 90% ≤ HF <95%, 95% ≤ HF <100%, and purebred HF. Distribution of MAF and estimation of LD were done using Haploview. All HF groups had similar patterns of MAF across autosomes (fraction of SNPs increased with an increase in MAF). However, means of MAF across autosomes differed among HF groups and it tended to decrease with an increase in H fraction (from 0.376 for HF <75% to 0.362 for purebred HF). Conversely, the mean  $r^2$  across autosomes tended to increase as HF increased from 0.081 for HF <75% to 0.109 for purebred HF. Results from this study will be useful for genome wide association studies and for genomic prediction and selection of crossbred Holstein cattle in tropical regions.

**Key Words:** linkage disequilibrium, Holstein, tropics

**W94 Improving the genotyping-by-sequencing (GBS) approach for the identification of SNPs associated with Johne's disease.** Émilie Constant<sup>1,2</sup>, Eveline M. Ibeagha-Awemu<sup>1</sup>, Filippo Miglior<sup>3,4</sup>, Gilles Robitaille<sup>5</sup>, and Nathalie Bissonnette<sup>\*1,2</sup>, <sup>1</sup>Dairy & Swine Research and Development Centre Agriculture and Agri-Food Canada, Sherbrooke, Quebec, Canada, <sup>2</sup>Department of Biology, Université de Sherbrooke, Sherbrooke, Quebec, Canada, <sup>3</sup>Canadian Dairy Network, Guelph, Ontario, Canada, <sup>4</sup>CGIL, University of Guelph, Guelph, Ontario, Canada, <sup>5</sup>Food Research and Development Centre, Agriculture and Agri-Food Canada, Saint-Hyacinthe, Québec, Canada.

Bovine paratuberculosis is a disease caused by *Mycobacterium avium* ssp. *paratuberculosis* (MAP). Most infected cows are culled before they reach clinical stage, leading to the premature slaughter of many animals and significant economic losses. Several genetic variants have been reported associated with host susceptibility to MAP. The objective of this study was to validate a whole genome genotyping-by-sequencing (GBS) method to identify single nucleotide polymorphisms (SNPs) associated with bovine paratuberculosis. Animals were selected from 10 farms in the province of Quebec. Fecal and blood samples were collected to identify 24 MAP infectious status by fecal culture and serum ELISA and 24 healthy cows. Two GBS methods were compared: a conventional (restriction enzymes *Pst*I and *Msp*I at 5'/3' used to construct DNA libraries) method (CM) and CM with more selective primers to reduce the complexity of the libraries (RM). DNA was extracted from isolated peripheral blood monocyte cells. Multiplexed libraries (48 libraries per lane) were subjected to 100-bp single-end sequencing on an Illumina HiSeq 2000 system. Reads that passed all filtering criteria were mapped to the bovine genome (Bta\_4.6.1). The SNPs were called using the Universal Network-Enabled Analysis Kit and 30,266 and 82,593 passed



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