

# Changes to evaluation system (December 2011)

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## Genomic evaluations using 6,909 markers

By Paul VanRaden, George Wiggans, Tabatha Cooper, Curt Van Tassell, and Tad Sonstegard

Genotypes from the [BovineLD Genotyping BeadChip](#), a 6,909-marker panel, were included as an additional data source for genomic evaluations in December 2011. The BovineLD panel was developed by [Illumina](#) (San Diego, CA) in cooperation with ARS-USDA's Animal Improvement Programs and [Bovine Functional Genomics](#) Laboratories, France's National Institute for Agricultural Research (INRA) and National Association of Livestock and Artificial Insemination Cooperatives (UNCEIA), and Australia's [Department of Primary Industries Victoria](#). Of the 6,909 markers, 6,836 are used in evaluation after removing 35 that did not meet standard edits and markers on mitochondria and the Y chromosome. The BovineLD panel includes 2,162 markers from the previous [Illumina Bovine3K Beadchip](#), which included 2,900 markers, and is intended to replace the 3K chip. Animals with 3K genotypes will continue to have genomic evaluations updated every 4 months using phenotypic and genomic information from new animals to estimate marker effects. BovineLD genotypes began arriving in October and were included in evaluations released in the November monthly update. The chip field contains "LD" for those animals in the distributed XML files, and the chip number in formats 38 and 105 is "06" because chip numbers "04" and "05" are assigned to the [Illumina BovineHD](#) (777K) and the [Affymetrix](#) (Santa Clara, CA) [Axiom Genome-Wide BOS 1 Array](#) (648K) chips, respectively (see reference note 162).

Because more markers are used and genotype calls are more accurate with BovineLD than with Bovine3K, the percentage of missing genotypes that can be filled correctly is improved. Imputation accuracy averaged 98.9% for Holsteins, 98.3% for Jerseys, and 97.9% for Brown Swiss from BovineLD as compared with 95.9%, 94.6%, and 93.9%, respectively, from Bovine3K. For Holsteins with BovineLD genotypes, imputation accuracy averaged 99.3% if both parents had BovineSNP50 genotypes, 98.6% if 1 parent had a BovineSNP50 genotype, and 96.6% if neither parent was genotyped as compared with 96.7%, 95.2%, and 93.0%, respectively, for Bovine3K genotypes. Corresponding reliabilities also were higher with BovineLD than with Bovine3K. For example, a calf with a parent-average reliability of 35% and a BovineSNP50 genomic reliability of 70% would instead have a 63% Bovine3K reliability or a 68% BovineLD reliability.

Reliability formulas may be further adjusted after more actual BovineLD genotypes are examined. Previously, the fraction of correctly imputed markers was the same constant for all animals with Bovine3K genotypes. With a mixture of Bovine3K and BovineLD genotypes, the fraction is set to the product

$$[n_{\text{call}}/(n_{\text{call}} + 120)] \times [n_{50K}/(n_{50K} + 50)],$$

where  $n_{\text{call}}$  is the number of called markers and  $n_{50K}$  is the number of BovineSNP50 genotypes from which to impute in the animal's breed. For example, with 6,800 called markers for a Jersey from a population with 6,000 Jersey BovineSNP50 genotypes, the imputed markers would be set to 97.5% correct and the overall accuracy to 97.9% when including the observed markers. Evaluations for many animals should be nearly as accurate with BovineLD as with BovineSNP50 genotypes.

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## Maternal grandsire detection and discovery

By George Wiggans, Tabatha Cooper, Paul VanRaden, Jeff O'Connell, and Lillian Bacheller

As genotypes are received, the maternal grandsire (**MGS**) identification is checked, and those that are missing or possibly incorrect have an alternative MGS proposed. For unknown or suspect MGSs, all genotyped males that are 4.5 years older than the animal are checked, and those deemed most likely to be the possible MGS are collected. Those potential MGSs are reported as part of the parentage validation record along with a conflict percentage that reflects the likelihood that the proposed bull is the true MGS. For example, a conflict percentage of 10 indicates that the animal and potential MGS have opposite homozygous genotypes for 10% of the single-nucleotide polymorphism (**SNP**) markers. If the sire is genotyped and homozygous at a SNP, the animal's heterozygous SNP can be used in determining a possible MGS. In most cases, more than one potential MGS are given. The difference in conflict percentages between the first and subsequent suggestions indicates the likelihood that the true MGS has been suggested in each case. A subset of genotypes (2,451) with confirmed MGSs was used to evaluate the accuracy of the SNP MGS method. The true MGS was listed as the first MGS suggestion for 2,402 genotypes (98%); however, that percentage likely will be lower for the full production population, which does not have all MGSs genotyped. The SNP MGS method is based on comparing one SNP at a time. Records with proposed MGS from the SNP method are distributed when the genotype is loaded and monthly if parentage status changes.

Following imputation, the MGS is checked again using a haplotype MGS method that compares the animal's maternal haplotype to its MGS haplotypes. If the number of haplotype matches is lower than expected, males older than the dam are checked to see if any bull has more haplotypes in common. This type of MGS detection is more precise than the SNP method because it can account for the sire's contribution at all SNP, not just those that are homozygous. The haplotype MGS method results are reported monthly in conjunction with the official genomic evaluation. The statistic "percentage match" (expected = 45%) is also given. The accuracy of the haplotype MGS method was tested by replacing correct MGSs with incorrect ones or by setting the MGS to missing (4,131 Holsteins, 552 Jerseys, and 142 Brown Swiss). For Holsteins and Brown Swiss, 97% of MGSs were correctly identified; for Jerseys, 95% were correct. Modifying the edits to decrease the likelihood of an incorrect MGS resulted in 92, 96, and 90% accuracy for Holsteins, Brown Swiss, and Jerseys, respectively. Within Holsteins, the success rate in finding a MGS was 86% for imputed dams, 98% for 50K-genotyped animals, and 94% for 3K-genotyped animals. The MGS analyses extend parentage validation so that incorrect or missing MGSs can be detected and corrected for most animals.