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Gredler, B.; Sargolzaei, M.; Bapst, B.; Bieber, A.; Simianer, H. and Seefried, F.R. (2013): Accuracy of 54K to HD genotype imputation in Brown Swiss cattle. Plant and Animal Genome XXI Congress, San Diego, USA, 11.-16.1.2013

ABSTRACT

Imputation of genotypes can be used to reduce the implementation costs of genomic selection. In this study, we evaluated the accuracy of genotype imputation from Illumina 54k to Illumina High Density (HD) in Brown Swiss cattle. Genotype data comprised 6,106 54k and 880 HD genotyped bulls and cows of Brown Swiss and Original Braunvieh cattle. Genotype data was checked for parentage conflicts and SNP were excluded if MAF was below 0.5% and SNP call rate was lower than 90%. The final data set included 39,004 SNP for the 54k and 627,306 SNP for the HD chip. HD genotypes of animals born between 2004 and 2008 (n=365) were masked to mimic animals genotyped with the 54k chip. Methods used for imputation were FImpute and Findhap V2. Both programs use pedigree information for imputation. The accuracy of imputation was assessed by the correlation (r) between true and imputed genotypes, the percentage of correctly and incorrectly imputed genotypes. Both programs gave high imputation accuracy with FImpute outperforming Findhap. Accuracy of imputation increased with increasing relationship between the HD genotyped reference population and 54k genotyped imputation candidates. Average r for FImpute and Findhap were 0.992 and 0.988 when both parents of the 54k genotyped candidate were HD genotyped, respectively. Correlations were lower when no direct relatives were HD genotyped (0.971 and 0.918 for FImpute and Findhap, respectively). Accuracy of imputation highly depended on MAF of the imputed SNP. For FImpute, average r ranged between 0.89 (MAF <0.025) and 0.99 (MAF between 0.4 and 0.5).