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ERROR RATE FOR IMPUTATION FROM BovineSNP50 TO BovineHD

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INTRODUCTION

- From 2007 onwards, large numbers of cattle have been genotyped with 50k-chips.
- Since 2010, higher density chips (e.g. Illumina BovineHD) are available.
- No need to re-genotype all animals on HD-chip, if imputation of 50k genotypes to HD is possible with low error rates.

OBJECTIVE

To investigate the error rate for imputation from BovineSNP50 genotypes to BovineHD.

GENOTYPES AND IMPUTATION

- BovineHD genotypes were obtained for 548 high impact bulls from the Eurogenomics reference population.
- Genotypes for all but the BovineSNP50 markers were masked in 60 validation animals.
- Validation animals were randomly chosen from the animals that did not have descendants with HD-genotype.
- The reference set for imputation consisted of the remaining set of 488 animals.
- Beagle 3.3 and DAGPHASE were used for imputation.

- Results were obtained for a subset of 6 BTA: 1, 6, 11, 14, 20, 29.
- Each analysis was repeated 4 times.
- Allelic imputation error:

$$\text{error (\%)} = 100 * \frac{n_{\text{imputed}} \neq \text{observed}}{n_{\text{imputed.and.observed}}}$$

RESULTS

- Imputation error was 0.67% on average, and ranged from 0.56% to 0.82% (Beagle 3.3, Figure 1).
- Imputation error was lower with higher allele frequency (Figure 2).
- Adding linkage information using DAGPHASE did not improve imputation results.

Figure 1. Average imputation error (%), per chromosome and averaged across six chromosomes

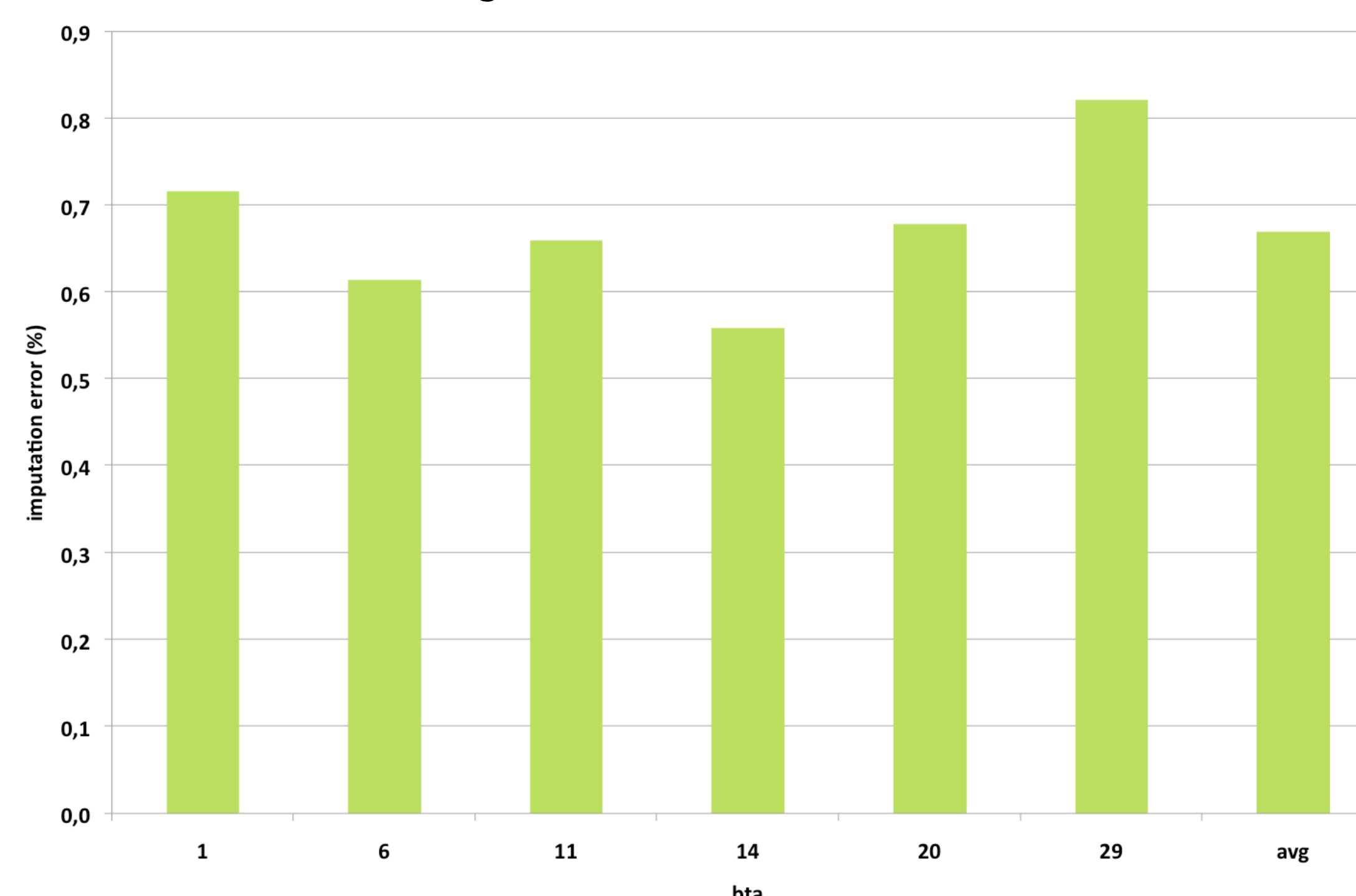
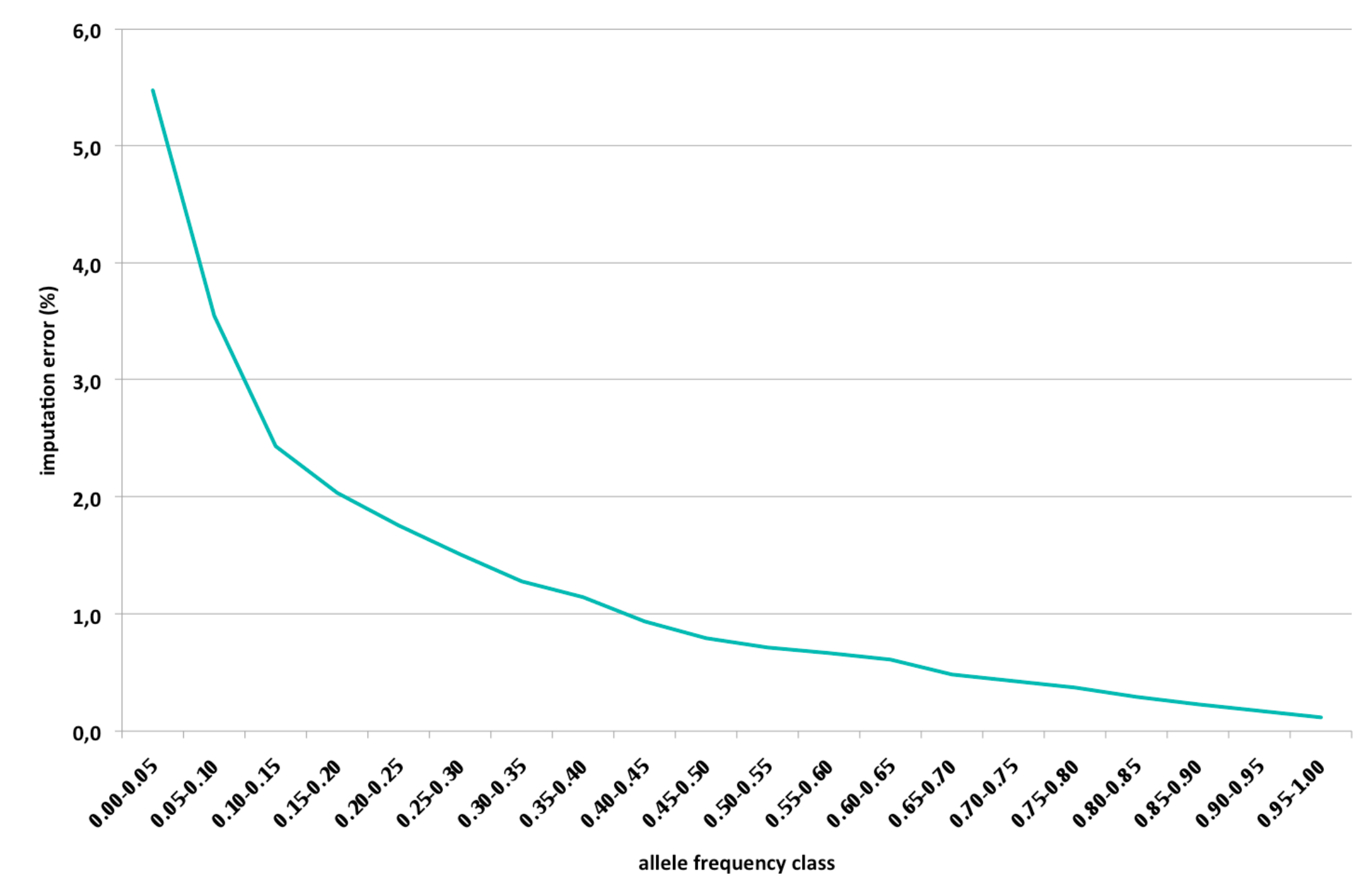


Figure 2. Imputation error (%) per allele frequency class



CONCLUSIONS

- Imputation from BovineSNP50 to BovineHD results in low error rate when using Beagle 3.3.
- With high density genotypes, linkage information does not improve imputation results.

